ABSTRACT GUIDE
of the 2017 AANEM Annual Meeting

American Association of Neuromuscular & Electrodiagnostic Medicine
Annual Meeting Abstracts
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Welcome to the 64th Annual Meeting of the
American Association of Neuromuscular of Electrodiagnostic Medicine

Abstracts present basic science, clinical and technical or developing information in one of the following areas: clinical neurophysiology, topics in nerve, topics in muscle, topics in neuromuscular junction, topics in anterior horn cell, musculoskeletal, practice issues, academic topics, pain and therapies. The Program Committee reviews and scores submissions. Abstracts are selected on the basis of their medical and scientific significance, timeliness, quality of data and methodology, adherence to specific format requirements, and other criteria.

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EFFECTS OF HAEMODIALYSIS ON INTRANEURAL BLOOD FLOW IN END-STAGE KIDNEY DISEASE

Adeniyi Borire (Moorebank, NSW), Ria Arnold (Sydney, NSW), Leo Visser (Tilburg, Netherlands), Matthew Kiernan (Sydney, NSW), Arun Krishnan (Sydney, NSW), Neil Simon (Sydney, NSW), Bruce Pussell (Sydney, NSW), Natalie Kwai (Sydney, NSW)

INTRODUCTION/OBJECTIVE: We quantified intraneural blood flow (INBF) in 18 patients with end-stage kidney disease (ESKD) and examined its relationship with nerve size, neuropathy severity, and nerve excitability parameters.

METHODS: Enrolled were 18 ESKD patients on thrice-weekly high-flux hemodialysis and 20 healthy control subjects. Sonographic and electrophysiologic studies were conducted before and after a single session of hemodialysis. Serial measurements of median nerve cross-sectional area (CSA) and intraneural blood flow (INBF) were performed at the same nonentrapment site in the mid-forearm. INBF was quantified by analyzing power Doppler sonograms to obtain the vessel score (VS) and maximum perfusion intensity (MPI). Corresponding median motor nerve excitability studies were performed. Neuropathy severity was quantified using the Total Neuropathy Score (TNS).

RESULTS: Of the ESKD cohort, 89% had neuropathy (TNS>1). ESKD patients had significantly higher prevalence of detectable INBF compared to the control group (39% versus 0%;p<0.0001). There was a strong correlation between baseline MPI and VS (r=0.962, p<0.0001). Both forms of INBF measurement correlated significantly with CSA (MPI: r=0.489, p<0.05; VS: r=0.556, p<0.05) and TNS (MPI: r=0.674, p<0.01; VS: r=0.643, p<0.01). Patients with detectable INBF had larger nerves and more severe neuropathy (p<0.01). INBF parameters were significantly reduced after a session of dialysis (VS: 1.4±0.5 versus 1.0±0.8, p<0.01; MPI: 1.38±0.92 cm/s versus 0.55±0.58 cm/s, p<0.01). A significant relationship was found between intradialytic change in INBF and changes in nerve excitability.

CONCLUSION: This study shows that peripheral nerve hypervascularity is demonstrable in ESKD patients and reversible following hemodialysis. INBF is more likely to be detected in patients with moderate-to-severe neuropathy.

Adeniyi Borire, MD
Golseth Young Investigator Award Recipient

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PREVALENCE OF CARPAL TUNNEL SYNDROME PRESENTING WITH SYMPTOMS IN AN ULNAR NERVE DISTRIBUTION: A PROSPECTIVE STUDY

Berdale Colorado (St. Louis, MO), Daniel Osei (St. Louis, MO)

INTRODUCTION: It is common for patients presenting with symptoms of numbness, tingling and pain primarily in an ulnar nerve distribution to be found to have exclusive CTS on subsequent EDX testing. There has been limited research to date looking at the prevalence of this clinical presentation.

OBJECTIVE: To determine the prevalence of patients with CTS presenting with symptoms primarily in an ulnar nerve distribution.

METHODS: A cohort of adult residents in a metropolitan area were surveyed to assess for severity and localization of hand symptoms using the Katz hand diagram and Boston Carpal Tunnel Questionnaire Symptom Severity Scale (BCTQ-SSS). Thirty volunteers who met our case definition for ulnar neuropathy (BCTQ-SSS score >2 and symptoms localized to the ulnar nerve distribution) were identified and underwent a standardized physical examination, EDX testing, and ultrasound of the ulnar nerve.

RESULTS: Of the 30 volunteers, 3 subjects (10%) were found to have exclusive ulnar neuropathy at the elbow (based on both EDX and ultrasound criteria); 6 subjects (20%) were found to have exclusive ulnar neuropathy at the elbow (based on ultrasound criteria); 2 subjects (7%) were found to have coexistent ulnar neuropathy at the elbow (based on ultrasound criteria) and CTS (based on EDX criteria); 11 subjects (37%) were found to have exclusive CTS (based on EDX criteria); and 8 subjects (27%) were found to have negative EDX and ultrasound testing.

SUMMARY/CONCLUSION: Symptoms in the ulnar nerve distribution are a common presenting symptom in CTS.

Berdale Colorado, MD
Golseth Young Investigator Award Recipient-Runner Up

Berdale Colorado, MD
President’s Research Initiative Award Recipient
3 GENDER AND QUALITY OF LIFE IN MYASTHENIA GRAVIS PATIENTS FROM THE MYASTHENIA GRAVIS FOUNDATION OF AMERICA REGISTRY

Ikjae Lee (Birmingham, AL), Henry Kaminski (Washington DC, DC), Haichang Xin (Birmingham, AL), Gary Cutter (Birmingham, AL)

INTRODUCTION/OBJECTIVE: Myasthenia gravis (MG) is an antibody-mediated autoimmune disorder affecting the neuromuscular junction leading to various degrees of fatigable muscle weakness causing disability and impaired quality of life.

METHODS: The MG patient registry is a patient-driven, nationwide database with MG patients participating across the United States, supported by the Myasthenia Gravis Foundation of America (MGFA). A total 1315 patients aged 18 or older who were diagnosed with MG and registered between 7/1/2013 and 6/30/2016 were included. Patients were grouped into male (n=488) and female (n=827) and basic demographic information, disease related history, and the 15-item MG quality of life questionnaire (MG-QOL15) scores were compared between the groups.

RESULTS: The female group was significantly younger (50.3 years versus 61.6 years), had a younger age at symptom onset (39.8 years versus 54.9 years), and was more likely to have thymoma (13% versus 6%) and thymectomy (38% versus 18%) compared to the male group. The MG-QOL15 score was significantly higher (worse) in the female group compared to the male group (24.5 versus 18.4). Age, gender, years from symptom onset to diagnosis, and thymectomy status were identified as significant factors associated with the MG-QOL15 score. Multivariable analysis adjusted for the above variables demonstrated that the female subgroup that had thymectomy had comparable MG-QOL15 scores with the male group with or without thymectomy (21.3 versus 21.1).

SUMMARY/CONCLUSION: Quality of life reported by MG patients was worse in the female group, but this disparity was improved in thymectomized female subgroup.

Ikjae Lee, MD
Best Abstract Award Recipient

4 ELECTROPHYSIOLOGIC FEATURES OF RADIAL NEUROPATHY IN CHILDHOOD AND ADOLESCENCE

Ioannis Karakis (Atlanta, GA), Sofia Georghiou (Boston, MA), H. Royden Jones (Burlington, MA), Basil Darras (Boston, MA), Peter Kang (Gainesville, FL)

INTRODUCTION: There is dearth of electrophysiological studies on pediatric radial neuropathy (PRN).

OBJECTIVE: To analyze patterns of nerve injury in PRN.

METHODS: Retrospective analysis on 19 children and adolescents with PRN.

RESULTS: Of the 19 participants (mean age: 12 years, range: 1 month-19 years; 56% female), 53% had traumatic etiologies. Weakness in the finger and wrist extensors were the prevailing complaints (82%). Predominant localization was at the posterior interosseous nerve (37%), followed by the radial nerve below the spiral groove (32%), the radial nerve at the spiral groove (26%), and the radial nerve above the spiral groove (5%). Extensor indicis proprius (EIP) compound muscle action potential amplitude was reduced in 86% of cases when tested, with a median axon loss estimate (MAXE) of 78% (interquartile range [IQR] 7). Radial sensory nerve action potential (SNAP) amplitude was reduced in 53% of all cases, and in 83% of cases affecting the main radial trunk with a MAXE of 100% (IQR 56). For cases affecting the main radial trunk, there was a high correlation of EIP MAXE and radial SNAP MAXE (r=0.72, p=0.02). Neurogenic changes were seen in the EIP, extensor digitorum communis, extensor carpi radialis, and brachioradialis in 88%, 94%, 60%, and 44% of cases, respectively. Pathophysiology was demyelinating in 10%, axonal in 58%, and mixed in 32%.

SUMMARY/CONCLUSION: PRN is frequently of traumatic etiology and axonal pathophysiology. Contrary to adults where localization at the spiral groove predominates, pediatric cases are commonly localized at the PIN or distal main radial trunk.

Ioannis Karakis, MD
Best Abstract Award Recipient-Runner Up
EFFECT OF HIP AND KNEE POSITION ON NERVE CONDUCTION IN THE COMMON FIBULAR NERVE.
Peter Broadhurst (Toronto, ON), Lawrence Robinson (Toronto, ON)

OBJECTIVE: To study ways to measure the influence that hip and knee position have on routine fibular motor NCSs.

METHODS: Healthy subjects under age 40 were recruited (n=24) to have fibular NCSs completed in various positions, using hip extension/knee extension as a control position.

RESULTS: A mean increase in conduction velocity of 2.5 m/s across the knee (p=0.020) was seen during hip flexion compared with hip extension. A mean decrease in velocity of 1.6 m/s through the leg segment (p=0.016) was seen during knee flexion compared with knee extension.

SUMMARY/CONCLUSION: This study shows that the optimal position of the leg during fibular nerve studies is with the hip in flexion and knee in extension to more accurately reflect nerve length for velocity calculations. This may have implications for other peripheral nerves with respect to proximal joint position affecting calculated velocity.

Peter Broadhurst, MD, PhD, MSc
Best Abstract Award Recipient-Runner Up

POPULATION-BASED CHRONIC OPIOID THERAPY PREVALENCE AMONG POLYNEUROPATHY PATIENTS AND THE RESULTANT IMPACT ON FUNCTIONAL STATUS AND ADVERSE OUTCOMES
E. Matthew Hoffman (Rochester, MN), James Watson (Rochester, MN), Jennifer St. Sauver (Rochester, MN), Nathan Staff (Rochester, MN), Christopher Klein (Rochester, MN)

INTRODUCTION: Polyneuropathy is a commonly painful neurologic condition managed within general and specialty clinics. Chronic neuropathic pain suffered by patients with polyneuropathy frequently leads to decisions about using chronic opioid therapy. Understanding the impact of chronic opioids on patients with polyneuropathy could influence disease-specific opioid treatment decisions.

OBJECTIVE: To quantify the prevalence and net impact of chronic opioid therapy among polyneuropathy patients.

METHODS: This retrospective population-based cohort study determined chronic opioid exposure (≥90 days) by ambulatory prescriptions given to polyneuropathy patients (n=2892) and control subjects (n=14,435) between 2006 and 2010. Patient-reported functional status, documented adverse outcomes, and mortality through 2016 were compared between polyneuropathy cases on chronic and shorter durations of opioid therapy using multivariate regression methods to calculate odds ratios (OR) and hazard ratios (HR) with 95% CI, while adjusting for comorbidities and other confounders.

RESULTS: Polyneuropathy patients received chronic opioids more often than control subjects (18.8% versus 5.4%), and those on chronic opioids had modestly worse functional status markers than polyneuropathy patients on shorter duration opioids, including increased reliance on gait aids (OR 1.9, 95% CI 1.4-2.6). No functional status markers were improved by chronic opioids. Adverse outcomes were more common among polyneuropathy cases on chronic opioids: depression (adjusted HR 1.5; 95% CI 1.3-1.8), opioid dependence (adjusted HR 2.9, 95% CI 1.5-5.5), and opioid overdose (adjusted HR 5.1, 95% CI 1.6-20.0).

SUMMARY/CONCLUSION: Polyneuropathy diagnosis associated with higher likelihood of being prescribed chronic opioid therapy, which appeared to associate with higher risk of subsequently developing opioid dependency and overdose without demonstrating improved functional status.

E. Matthew Hoffman, DO, PhD
President’s Research Initiative Award Recipient
AFTERDISCHARGES FOLLOWING M WAVES IN PATIENTS WITH VOLTAGE-GATED POTASSIUM CHANNELS ANTIBODIES
Mingsheng Liu (Peking, CN), Jingwen Niu (Beijing, CN), Yuzhou Guan (Beijing, CN), LIYING CUI (Beijing, CN)

INTRODUCTION: Needle EMG is commonly used for monitoring peripheral motor nerve hyperexcitability in patients with voltage-gated potassium channel (VGKC) antibodies.

OBJECTIVE: To explore the correlation between afterdischarges in motor NCSs and clinical motor hyperexcitability in patients with VGKC antibodies.

METHODS: Six patients with positive serum antibodies to contactin-associated protein-like 2 (CASPR2) or/and leucine-rich glioma-inactivated protein 1 (LGI1) were recruited, including 5 with autoimmune encephalitis and 1 with cramp fasciculation syndrome. Needle EMG, NCSs, and F waves were performed, and afterdischarges were assessed.

RESULTS: Five patients had clinical evidence of peripheral motor nerve hyperexcitability (myokymia or cramp), and 4 of them had abnormal spontaneous firing in concentric needle EMG. Prolonged afterdischarges following normal M waves were present in all 6 patients, including the 2 who had no needle EMG evidence of peripheral nerve hyperexcitability. Afterdischarges disappeared after treatment with IV immunoglobulin.

SUMMARY/CONCLUSION: Afterdischarges in motor NCSs might be more sensitive than needle EMG for detecting peripheral motor nerve hyperexcitability, and they could disappear gradually in accordance with clinical improvement and reduction of antibodies.

Mingsheng Liu, MD
President’s Research Initiative Award Recipient

A NOVEL CHLORIDE CHANNEL DNA VARIANT PRODUCING PREGNANCY-INDUCED MYOTONIA
Hani Kushlaf (Cincinnati, OH), John Quinlan (Cincinnati, OH)

INTRODUCTION: Myotonia congenita is a skeletal muscle chloride channelopathy that presents at a young age with painful muscle stiffness. A report of a common mutation causing myotonia congenita presenting as pregnancy-induced myotonia was described.

OBJECTIVE: To report the clinical presentation and diagnostic testing of 2 sisters with pregnancy-induced myotonia caused by a novel chloride channel DNA variant.

METHODS: A 30-year-old female, gestational age 21 weeks, noted painful muscle stiffness that started 9 weeks from gestation. The muscles involved were in the thighs, upper limbs, and jaw. Symptoms worsened with the progression of pregnancy. She had stiffness during a first pregnancy 3 years earlier that resolved following pregnancy. The sister, 27 years old, gestational age 26 weeks, developed muscle stiffness at 11 weeks from gestation. Symptoms started in the hands followed by thighs and jaw. Examination revealed normal strength with bilateral handgrip, and thenar and finger extensor percussion myotonia in both sisters. Electrodiagnosis in the index patient revealed generalized profuse waxing and waning myotonic discharges in the right arm, right leg, and paraspinal muscles. Short exercise testing showed a right ulnar motor amplitude drop of 27% postexercise that returned to normal in 10 seconds. Genetic testing detected a heterozygous variant in the CLCN1 gene (p.Ser183Pro). Testing for myotonic dystrophy type 1 and type 2 and sequencing of the SCN4A gene were negative.

SUMMARY/CONCLUSION: Symptomatic onset of myotonia congenita during pregnancy occurs in rare patients with chloride channel DNA mutations. The described novel chloride channel DNA variant adds to the known chloride channel mutations producing this specific phenotype.

Hani Kushlaf, MD
President’s Research Initiative Award Recipient
MANIPULATING CHRONIC INFLAMMATION TO REDUCE NOCICEPTIVE HYPERREFLEXIA AND PAIN AFFERENT SPROUTING AFTER NEURAL INJURY

Keith Tansey (Jackson, MS), Hyun Joon Lee (Jackson, MS), Jumi Chung (Jackson, MS), A. Arturo Leis (Jackson, MS), Malu Tansey (Atlanta, GA)

INTRODUCTION: Lateral hemisection spinal cord injury (SCI) at T10 produces nociceptive hyperreflexia in the cutaneous trunci muscle (CTM) reflex and central sprouting of nociceptive afferents from dorsal cutaneous nerves (DCNs) 6 weeks after SCI, both above (T7) and below (T13) the level of injury. Iba1+ microglia/macrophages are also increased at T7 and T13 at this time point.

OBJECTIVE: To study whether a selective soluble tumor necrosis factor (TNF) blocker, XPro1595 (Xencor Inc., Monrovia, California), could modulate chronic inflammation and alter neural plasticity in these rostral and caudal spinal segments.

METHODS: Long Evans female rats were subjected to T10 SCI and injected with 10 mg/kg of XPro1595 subcutaneously every third day for 6 weeks. Before the terminal electrophysiological experiments, animals were injected with axon tracers, IB4 and CTB, at their T7 and T13 DCNs. CTM neurograms evoked by segmental DCN stimulation were subsequently recorded to measure reflex sizes. Spinal cords were harvested for immunohistochemistry to quantify IB4+ C fibers and CTB+ A fibers, as well as Iba1+ microglia/macrophages.

RESULTS: Six weeks of XPro1595 treatment reduced the number of Iba1+ microglia/macrophages at T7 to uninjured levels and at T13 to lower than uninjured levels. Treatment also reversed injury-induced nociceptive hyperreflexia, returning T7 DCN evoked CTM reflex sizes to uninjured values and causing relative hyporeflexia in T13 DCN evoked reflexes. XPro1595 treatment also diminished nociceptive afferent sprouting in T7 and T13 DCNs, particularly below the level of injury, and preferentially with A fibers over C fibers.

SUMMARY/CONCLUSION: Reducing chronic inflammation decreases nociceptive hyperreflexia and pain afferent sprouting after neural injury.

Keith Tansey, MD PhD
President’s Research Initiative Award Recipient

MENTORING, EDUCATION AND ENGAGEMENT - AN EMPLOYEE TRAINING TECHNIQUE TO IMPROVE PATIENT EXPERIENCE AND EMPLOYEE SATISFACTION

Tammy Hether (Columbia, MO), Raghav Govindarajan (Columbia, MO)

BACKGROUND: Changes in health care have put patient experience at the forefront.

OBJECTIVE: To evaluate the impact of a novel training technique—Mentoring, Education, and Engagement (MEE)—on employee satisfaction and, in turn, patient experience.

METHOD: This was a prospective study that compared patient experience in the neurophysiology laboratory using the National Research Corporation Picker Catalyst survey. Scores from July 2015 to December 2015 were compared with scores from January 2016 to August 2016 after MEE was introduced. Employee satisfaction survey data using the Gallop online survey tool from December 2015 were compared with employee satisfaction survey data from July 2016.

RESULTS: For the patient question, “Using any number from 0 to 10, where 0 is the worst facility possible and 10 is the best possible facility, what number would you use to rate this outpatient testing facility?” the percentage of patients who gave scores 9/10 improved from 79% to 90% (p<0.05). For the patient question, “Did staff/technologists who performed your tests or procedures treat you with courtesy and respect and explain the procedure?” the percentage of patients who gave scores of 9/10 improved from 73% to 91% (p<0.05). Overall pain score for the procedure using the visual analog scale reduced from 8 to 5 (p<0.05). For the employee satisfaction question, “In the last 7 days, have you received recognition or praise for doing good work?” the mean score increased from 3.00 to 3.91.

CONCLUSION: Patient experience and pain perception is directly related to overall explanation and respect perceived by them during a procedure.

Tammy Hether, R.EEG/EP T., R.NCS T
President’s Research Initiative Award Recipient

Tammy Hether, R.EEG/EP T., R.NCS T
Technologist Member Award Recipient
CERVICALGIA IN FIBROMYALGIA PATIENTS: A SIMPLY PART OF THEIR OVERALL CONDITION OR A COEXISTING CERVICAL RADICULOPATHY?
Alexandre Recchia (Sao Paulo, BR)

INTRODUCTION: Fibromyalgia is a chronic pain condition characterized by widespread pain, stiffness, fatigue, and sleep disturbances. Among the many pain sites present in fibromyalgia patients, noteworthy are the trigger points crowded around the base of the neck and the shoulders that can often overlap with other local symptoms masking an eventual coexistence condition.

OBJECTIVE: To determine the prevalence of cervical radiculopathy (CR) in patients with and without fibromyalgia with a clinical presentation of cervicalgia in a cross-sectional study.

METHODS: The study was conducted with 85 patients diagnosed with fibromyalgia, according to criteria of the American Academy of Rheumatology (fibromyalgia group, or FG), and a second group consisting of 146 patients with no previous history of fibromyalgia or CR (non-fibromyalgia group, or NFG). All patients, in both groups, presented insidious cervical pain and were evaluated with cervical MRI and needle EMG.

RESULTS: The FG included exclusively women (mean age: 38.9±9.3 years) who presented with a prevalence of 14.1% of CR (95% CI 6.7-21%). The NFG was composed of 54 men and 45 women (mean age: 54.3±8.7 years) who presented with a prevalence of 43.1% of CR (95% CI 35-51%). The prevalence ratio (PR) between the NFG and FG groups was 3.05. However, when the prevalence of CR was considered only in women in the NFG group (23 cases), the PR was 1.11.

SUMMARY/CONCLUSION: Coexistence of CR cases observed in the FG, although not numerous, are not negligible when compared to the NFG, especially when considering a more homogeneous sample; therefore, a coexisting CR should never be disregarded.

Alexandre Recchia, MD
President’s Research Initiative Award Recipient

PARSONAGE-TURNER SYNDROME IS NOT A BRACHIAL PLEXITIS
Darryl Sneag (New York, NY), Schneider Rancy (New York, NY), Scott Wolfe (New York, NY), Susan Lee (New York, NY), Steve Lee (New York, NY), Joseph Feinberg (New York, NY)

INTRODUCTION: Parsonage–Turner syndrome (PTS)—commonly known as neuralgic amyotrophy and brachial neuritis—is described etiologically as a brachial plexitis, implying diffuse plexus involvement.

OBJECTIVE: To characterize lesion distribution in PTS using high-resolution MRI.

METHODS: We searched our institution’s MRI database for patients with a clinical diagnosis of PTS. Two radiologists specializing in neurography independently evaluated signal characteristics and morphology of the plexus on high-resolution MRI, including its terminal branches as well as peripheral nerves when involved. An authority in PTS reviewed all charts to confirm the diagnosis of PTS using history, examination, and electromyographic findings of complete/near-complete motor loss.

RESULTS: A total of 24 patients (18 male, 6 female; age: 41±15 years) had ≥1 clinically-involved nerves: suprascapular (15), long thoracic (7), axillary (5), radial (2), anterior interosseous (2), thoracodorsal (1), phrenic (1), musculocutaneous (1), and pronator teres (1). Mean time between symptom onset and initial needle EMG was 5.5±4.6 months; between onset and MRI was 8±6.7 months. On MRI, the plexus proper appeared normal in 21/24 patients; in 3 others, signal hyperintensity was seen immediately proximal to the terminal branch take-off. Focal caliber decreases of terminal branches, suggesting intrinsic constrictions, were detected in 19/24. Hyperintensity and enlargement of involved peripheral nerves were present in all patients, except the phrenic nerve (too small to identify). MRI inter-reader agreement was substantial (Cohen’s kappa=0.833).

SUMMARY/CONCLUSION: MRI findings, corroborated by needle EMG, localize abnormalities to the plexus branches and peripheral nerves, suggesting PTS is a mononeuropathy multiplex rather than a brachial plexitis.

Darryl Sneag, MD
President’s Research Initiative Award Recipient
PAIN AS A MANIFESTATION OF VOLTAGE-GATED POTASSIUM CHANNEL AND N-TYPE CALCIUM CHANNEL AUTOIMMUNITY
A. Arturo Leis (Jackson, MS), Brent Goodman (Scottsdale, AZ)

INTRODUCTION: The spectrum of neurological manifestations associated with voltage-gated potassium channel (VGKC) and N-type calcium channel (NTCC) autoimmunity is broad, and ranges from brain dysfunction to neuromuscular and autonomic dysfunction. Pain is a less recognized manifestation of VGKC and NTCC autoimmunity.

OBJECTIVE: To describe pain characteristics, electrodiagnostic EDX findings, comorbid conditions, and treatment response in VGKC and NTCC autoimmunity.

METHODS: A medical record review of 22 patients with varied neurological symptoms and positive VGKC or NTCC antibodies was conducted. Antibody testing was performed by Mayo Clinic Laboratories, Rochester, Minnesota.

RESULTS: In 13 of 22 patients (9 VGKC, 4 NTCC), pain was severe and a chief complaint. Pain was typically subacute, intractable, unexplained, and difficult to describe, although hyperalgesia (increased response to painful stimuli) and allodynia (pain from stimuli that normally do not provoke pain) were common. Pain was poorly responsive to conventional therapy. In contrast, 6 VGKC patients improved after IV immunoglobulin (IVIg) (4), IV methylprednisolone (1), or thymectomy (1). In NTCC patients, 1 improved with oral prednisone and 1 with rituximab, but 3 failed IVIg. EDX assessment in VGKC autoimmunity showed peripheral nerve hyperexcitability (PNH) (4), normal (3), polyneuropathy (1), or omitted (1). EDX assessment in NTCC cases was normal (2), polyneuropathy (1), or multiple radiculopathies (1). Common comorbidities included dysautonomia (gastroparesis, postural instability, hyperhidrosis, cardiac dysrhythmias, or bladder dysfunction) (7), other autoimmune diseases (6), migraines (3), and fibromyalgia (2).

SUMMARY/CONCLUSION: VGKC and NTCC autoimmunity should be considered in cases of idiopathic intractable pain, particularly when accompanied by autonomic symptoms or other autoimmune diseases. A trial of immunotherapy or immunosuppression is warranted.

A. Arturo Leis, MD
President's Research Initiative Award Recipient

NOVEL UNOBTRUSIVE CARPAL TUNNEL TISSUE MANIPULATION DEVICE TO DECOMPRESS THE MEDIAN NERVE: A PILOT CLINICAL STUDY
Pauline Luong (Los Angeles, CA), Frank King (Mission Viejo, CA), Zong-Ming Li (Cleveland, OH), Matt Dickason (Newport Beach, CA), Matthew Diamond (New York, NY), Jae Son (Los Angeles, CA)

INTRODUCTION: CTS involves chronic compression of the median nerve. A novel unobtrusive carpal tunnel tissue manipulation device (CTMD) was developed for the volar aspect of the wrist to decompress the median nerve by applying negative pressure to the wrist.

OBJECTIVE: To investigate the feasibility, safety, and efficacy of the CTMD to treat CTS in patients with mild to severe CTS.

METHODS: A prospective, single-arm clinical trial was conducted involving 11 patients (64% unilateral, 36% bilateral) after confirmation of CTS by EDX examination. Subjects wore a CTMD on the affected wrist(s) for 8-10 hours daily and completed a Boston Carpal Tunnel Questionnaire (BTCQ) at 0, 2, 4, and 12 weeks post-baseline. The Symptom Severity Scale (SSS) score of the BTCQ was evaluated as the primary outcome measure. Only the SSS scores for the affected wrist of unilateral patients and for the wrist with the higher baseline SSS score of bilateral subjects were analyzed.

RESULTS: Mean SSS score improved by 0.59 points (SD 0.68, p<0.05) after 4 weeks of treatment compared to baseline. Surprisingly, the SSS continued to improve 8 weeks post-treatment, by 0.79 points (SD 0.74, p<0.01) compared to baseline. Interestingly, unilateral subjects’ SSS scores improved more dramatically than bilateral subjects (1.2, SD 0.5, versus 0.09, SD 0.41) at 8-weeks post-treatment.

SUMMARY/CONCLUSION: Participants wearing the CTMD for 4 weeks showed significant improvements in CTS symptom severity, and symptoms continued to improve even after the treatment period. Future studies are necessary to investigate long-term effects and to understand moderating factors, including bilaterality of disease.

Pauline Luong, M Eng
President’s Research Initiative Award Recipient
THE EVOLUTION OF CORTICOSTEROID IN MYASTHENIA GRAVIS AND THE MESSAGE FROM IT
SANKAR BANDYOPADHYAY (Hummelstown, PA)

INTRODUCTION: Many physicians believe that steroids came, saw, and took the world by storm. History speaks otherwise: a staggering course with an ultimate success was the reality.

OBJECTIVE: To study the evolutionary history of the use of corticosteroids in myasthenia gravis (MG).

METHODS: A historical study from published journal articles and books was conducted.

RESULTS: Adrenal gland extracts, used in 1896 in Paris by Marsh, showed disapproving results. Thirty years later, Simon from Alabama added “wind to the sails” with good results from the newly available anterior pituitary extract in 2 MG patients. The euphoria died 5 years later when Schlezinger from New York showed how useless steroids were for 7 patients. Following this seesaw course came a hay period of commercial availability of adrenocorticotropic hormone resulting in “significant partial remission” in 1949. The next year, the world saw the Nobel Prize going to Hench at Mayo Clinic for the use of cortisone. This was shot down the next year by Lee Eaton at Mayo Clinic, based on a small clinical observation. A study from Johns Hopkins the next year completed the funeral process. An epitaph was written by Oslerman in 1958: steroids were too dangerous. . . . It was over. Then, in 1965, the phoenix rose in a New York symposium based on a Swedish study. Supportive studies flooded in. Oslerman, 8 years after writing the “epitaph,” changed the adjective to “gratifying” instead of “dangerous”!

SUMMARY/CONCLUSION: It took nearly 70 years for the mighty steroid to be accepted. It went through death, disappearance, and glorious resurrection. It gives a strong learning message to drug discoverers throughout the world.

DIAGNOSTIC CHALLENGES OF PARAPLEGIA IN CANCER PATIENTS: LEPTOMENINGEAL DISEASE VS INTRATHECAL/RADIATION THERAPY COMPLICATION
Sara Dehbashi (Galveston, TX), Ahmad Daher (Houston, TX), Sudhakar Tummala (Houston, TX)

INTRODUCTION Paraplegia can arise from leptomeningeal disease spread or therapy-induced central nervous system (CNS)/peripheral nervous system (PNS) toxicity leading to diagnostic challenges.

OBJECTIVE: To highlight the importance of biomarkers that distinguish between leptomeningeal disease spread and CNS/PNS toxicity.

CASE REPORTS: Patient 1: A 54-year-old female with metastatic lung cancer had resection of the tumor at T11-12 followed by radiation. She presented 3 years later complaining of progressive bilateral lower extremity weakness and numbness. MRI of the spine was non-revealing while needle EMG/NCSs showed bilateral axonal lumbosacral radiculopathy. Cerebrospinal fluid (CSF) studies were unremarkable with the exception of an elevated myelin basic protein (MBP) level to 19.83 ng/mL (cytology: negative). Patient 2: A 64-year-old male with blastic plasmacytoid dendritic neoplasm plus lumbar microdiscectomy of the lumbosacral spine, who received intrathecal chemotherapy and systemic chemotherapy on diagnosis, developed lower extremity weakness, numbness, and bowel/bladder incontinence 2 months later. Initial imaging did not show any major pathology while needle EMG/NCSs suggested axonal lumbosacral radiculoneuritis. CSF studies were largely unremarkable then, but his symptoms progressed and he eventually became paraplegic. Repeat MRI of the spine showed diffuse myelopathy with dorsal column involvement, and CSF studies showed pleocytosis of 778, elevated protein of 355 g/L, and an MBP level of 449.70 ng/mL that continued to increase on subsequent checks, with repeated negative CSF cytology.

SUMMARY/CONCLUSION: Radiographic changes in CNS therapy-associated toxicity can be delayed or absent, which underscores the need for better biomarkers. In that regard, MBP, a marker of injury to nerve sheath, not typically elevated upon leptomeningeal tumor infiltration, may play an important role.

Sara Dehbashi, MD
Resident and Fellow Member AwardRecipient
A PAUCITY OF IN-USE REINFORCEMENT TECHNIQUES, AND HOW THAT CAN BE AMENDED
Sankar Bandyopadhyay (Hummelsyown, PA)

INTRODUCTION: Personal experience as a clinical mentor—working with medical students, neurology residents, and fellows—marks a limited nature of reinforcement techniques while examining reflexes. Frustration is obvious.

OBJECTIVE: To look for additional techniques for reinforcement in situations where deep tendon reflex (DTRs) are poorly elicited.

METHOD: Literature search, including published articles and books on DTR.

RESULT: Two core principles govern reinforcement: (1) distraction and (2) slight activation of the muscle whose tendon is about to be stretched by the percussion hammer. For the lower extremity, one available method is the Jendrassik maneuver of finger locking. Additional methods not currently in use for the lower extremity include: (1) teeth clenching, which works by distraction and increasing generalized tone, and (2) partial forceful extension of the knee against slight resistance for the knee jerk and partial forced plantar flexion against slight resistance against the examiner's spare hand, or pushing toes against the ground or stepping stool (principle 2). For the upper extremity, there is no commonly used reinforcement technique, as the Jendrassik maneuver cannot be used. Suggested techniques for the upper extremity include: (1) teeth grinding, as a distraction method, and (2) more than usual flexion of the elbow for the biceps jerk, and more than usual extension for the triceps jerk, both against slight resistance of the examiner's non-hammer hand, and passive extension of the wrist with slight intended flexion of the wrist blocked by examiner's resistance for the supinator jerk (principle 2).

CONCLUSION/SUMMARY: Elicitation of DTRs can be greatly improved, if standard techniques are insufficient, by using methods described in literature, but forgotten, due to diminished importance given to the clinical examination.

A PLEA FOR RESURRECTION OF FORGOTTEN DEEP TENDON REFLEXES, AND ITS JUSTIFICATION
Sankar Bandyopadhyay (Hummelstown, PA)

INTRODUCTION: A surprisingly large battery of deep tendon reflexes (DTRs) developed mostly between 1850 and 1950 has become largely extinct. The ones to survive the spoils of time, are the biceps, triceps, supinator, knee, and ankle jerks. A few among the lost were not deserving an evolutionary death, but got ignored by the diminutive adherence to clinical examination.

OBJECTIVE: To look for signs of life in the pile of presumed dead DTRs.

METHOD: Robert Wartenberg's classic book on reflexes, published in 1945, with 465 precious references, was exhumed from the out-of-print graveyard.

RESULTS: The need to bring DTRs back were shown by the following: (1) Babinski's inversion of the supinator jerk, with lack of elbow flexion, but with flexion of wrist and fingers, may suggest a C5-6 lesion; (2) Babinski and Sable's paradoxical triceps reflex, with flexion of the elbow without the expected extension, may suggest a C7 lesion or an UMN lesion such as cerebrovascular accident (CVA), due to relative hyperreflexia in flexor muscles of upper extremity; (3) Rossolimo's reflex, in which hammering the Achilles tendon results in flexion of the toes, especially the lateral ones, may suggest hyperreflexia or an upper motor neuron involvement; in patients without an obvious extensor plantar response, this can be useful; and (4) Guillain and Barré's semitendinosus and semimembranosus reflex, achieved by striking the tendons or the tibia, gives us an L5 reflex. We have a good S1 reflex: the ankle jerk.

CONCLUSION: A search of the lost wisdom, regarding a need for more useful clinical examination, can empower us with a better DTR examination.
ASSOCIATION BETWEEN ALS AND MYASTHENIA GRAVIS
Shuja Sheikh (Wharton, NJ), Abu Nasar (Newark, NJ), Francisco Gomez (Newark, NJ), Nizar Souayah (Westfield, NJ)

INTRODUCTION: Previous studies have demonstrated an association between ALS and autoimmune disorders.

OBJECTIVE: To investigate the association between myasthenia gravis (MG) and ALS.

METHODS: We sought MG associated with ALS utilizing the New York Statewide Planning and Research Cooperation System (SPARCS) database, 1998-2014. Data were analyzed via IBM SPSS software. An ALS group was identified and then comorbidities analyzed.

RESULTS: Total of 7102 ALS patients were reported (mean age: 64.5 years old, 55% male). Prior to the diagnosis of ALS, 0.4% were diagnosed with MG. The mean age of ALS patients with a prior diagnosis of MG is significantly higher than the age of MG patients in the general population and than patients who were diagnosed with ALS prior to the diagnosis of MG (mean age: 71.7±11.4 years, 60.15±19.8 years, and 64.5±16.4, respectively, p<0.05).

SUMMARY/CONCLUSION: Our study demonstrated a trend toward developing ALS at a significantly older age in patients who already have been diagnosed with MG compared to ALS patients without MG and to patients who were diagnosed with ALS prior to being diagnosed with MG. There is work in progress to compare the association between ALS and MG to the association to other comorbid and autoimmune disorders.

Shuja Sheikh, MD
Resident and Fellow Member Award Recipient

CHRONIC GRAFT-VERSUS-HOST DISEASE WITH DERMATOMYOSITIS-LIKE FEATURES IN CHARCOT MARIE TOOTH
Tiffany Lee (Cleveland, OH), Jinny Tavee (Chicago, IL)

INTRODUCTION: Chronic graft-versus-host disease (cGVHD) following allogeneic hematopoietic stem cell transplantation is a multisystemic disorder that rarely includes myositis.

OBJECTIVE: To report the first case of cGVHD with dermatomyositis-like features reported in a patient with Charcot–Marie–Tooth (CMT) disease.

METHODS: A 57-year-old man with genetically-confirmed CMT and myelodysplastic syndrome presented with heliotrope rash and generalized proximal weakness 4 years after allogeneic bone marrow transplant (BMT). The BMT was initially complicated by chemotherapy-induced worsening of his hereditary polyneuropathy and cGVHD with ocular and mucosal involvement, which stabilized with corticosteroids. Examination revealed severe generalized weakness with prominent axial component and head drop in addition to phenotypic CMT findings of a glove-stocking polyneuropathy with bilateral foot drop. Serological evaluation showed elevated aldolase of 13.8 (0-7.7 U/L) with normal creatinine kinase of 68 (39-308 U/L), negative antinuclear antibodies, and myositis antibody panel including anti-Jo1 antibodies. Needle EMG showed a generalized necrotizing myopathy. Muscle biopsy of the adductor magnus revealed perifascicular atrophy with perimysial inflammation composed primarily of CD4+ cells and macrophages suggestive of dermatomyositis. Despite 4 years of intermittent IV immunoglobulin, corticosteroids, rituximab, methotrexate, and tacrolimus, no significant improvement was seen.

SUMMARY/CONCLUSION: The immunopathogenesis of cGVHD and dermatomyositis is complex and not completely understood. cGVHD is thought to be primarily T-cell mediated although abnormal B-cell activity has been described. Similarly, dermatomyositis is mainly humorally-mediated, but T-cell infiltrates have been reported. Shared immunologic dysfunction may potentially give rise to the development of dermatomyositis in the setting of cGVHD. Treatment is with immune-modulating medications, but symptoms may remain refractory despite aggressive therapy.
NERVE CONDUCTION STUDY AND ELECTROMYOGRAPHY EXHIBITING RICHE–CANNIEU ANASTOMOSIS IN THE SETTING OF BILATERAL ULNAR NEURITIS: A CASE REPORT
Travis Coats (Syracuse, NY)

INTRODUCTION: Riche–Cannieu anastomosis (RCA) is an anatomic variant neural connection between the deep branches of the ulnar nerve and the recurrent branch of the median nerve at the thenar eminence. RCA clinical presentation varies by the extent of anomalous ulnar innervation of the hand.

OBJECTIVE: To emphasize the importance that practitioners be aware of anatomic variants that may alter Nerve Conduction Study (NCS) and needle Electromyography (EMG) findings.

CASE REPORT: The patient presented with right ring and little finger paresthesia and left global hand numbness. Physical examination (PE) revealed positive Tinel's sign at the bilateral cubital fossae. No atrophy, weakness, or sensory impairments were observed. NCSs exhibited relative slowing of conduction velocity (CV) of bilateral ulnar nerves at the elbows. Left and right median nerve stimulation to abductor pollicis brevis (APB) motor revealed reduced amplitudes. Left ulnar stimulation to adductor digiti minimi motor showed decreased CV. APB responses were elicited bilaterally from ulnar stimulation. Needle EMG revealed minimally decreased recruitment of the right first dorsal interosseous muscle. Other NCS and needle EMG findings were within normal limits.

SUMMARY/CONCLUSION: PE findings coupled with diminished CV at bilateral elbows with correction distally is suggestive of ulnar neuritis. RCA is suggested by PE findings coupled with bilateral APB response to ulnar nerve stimulation and diminished APB response to median nerve stimulation bilaterally. Practitioner knowledge of RCA prevented misdiagnosis of polyneuropathy or other conditions, allowing for proper treatment and the prevention of patient morbidity.

Travis Coats, MD
Resident and Fellow Member Award Recipient

INTRAOPERATIVE PERIPHERAL NERVE LESION LOCALIZATION USING “INCHING” DIRECT SSEP TECHNIQUE.
Watcharasarn Rattananan (Boston, MA), Mirela Simon (Boston, MA), Reiner Henson See (Boston, MA)

INTRODUCTION: Intraoperative localization of a peripheral nerve lesion can be challenging because the lesion may not be visually identified. We present a new application of short-latency somatosensory evoked potentials (SSEPs) obtained by direct nerve stimulation and used to demonstrate the integrity of ascending neuronal tracts via the “inching” technique. To our knowledge, this is the first report of using “inching direct SSEPs.” This newly adapted method can provide valuable information to surgeons in determining an appropriate site for nerve grafting.

OBJECTIVE: To describe the use of “inching” direct SSEPs in localizing a lesion of peripheral nerve.

CASE REPORT: A 23-year-old male presented with left knee dislocation, resulting in a complete left foot drop. Physical examination showed weakness in ankle dorsiflexion and eversion. MRI of the knee revealed multiple ligamentous injuries. An EDX study showed a left fibular axonal neuropathy localized distal to the takeoff to the short head of biceps femoris muscle. After 6 months, no clinical improvement was noted; hence, a surgical intervention with nerve graft was planned. During the surgical exploration, a lesion was identified very distally. There was no perceptible lesion at a more proximal site. Nerve-to-nerve conduction study recording proximal to this lesion revealed absent responses. At this point, the direct SSEP study with the “inching” technique was performed and was able to identify a very proximal conduction block lesion just distal to the sciatic nerve bifurcation.

SUMMARY/CONCLUSION: This case illustrates the valuable application of “inching” direct SSEP technique intraoperatively in challenging peripheral nerve lesions. This method helps to localize nerve lesions accurately in combination with other intraoperative monitoring modalities.
ROLE OF QUANTITATIVE SUDOMOTOR AXON REFLEX TESTING (Q-SWEAT) IN MONITORING TREATMENT OF AUTOIMMUNE SMALL FIBER NEUROPATHY
Elise Madar (Hershey, PA), Max Lowden (Hershey, PA)

OBJECTIVE: To evaluate quantitative sudomotor axon reflex testing (QSART) via Q-Sweat (WR Medical Electronics Co., Maplewood, Minnesota) as a tool for monitoring treatment response in a case of autoimmune small fiber neuropathy (SFN).

CASE PRESENTATION: A 19-year-old man presented with tingling, severe neuropathic pain in all extremities, and sensory ataxia several weeks post-viral pneumonitis. Two months after a 3-day course of IV immunoglobulin and initiation of daily mycophenolate therapy, the patient reported significant reduction in symptoms.

RESULTS: Laboratory studies showed elevated erythrocyte sedimentation rate at 33 mm/hr. Cerebrospinal fluid (CSF) showed elevated protein 107 mg/dL, albumin 60.3 mg/dL, and IgG 9.5 mg/dL, and CSF cell count was normal at 2/µL. Needle EMG/NCSs showed a sensory axonal polyneuropathy, and a skin biopsy demonstrated absent intraepidermal nerve fiber densities (IENFDs) at all 3 sites confirming a SFN. A repeat skin biopsy performed at 1 year demonstrated reinnervation, with IENFDs of 0.2/mm from the foot and calf, consistent with the improved clinical picture. At 2 years, the patient's symptoms continued to improve; however, a third skin biopsy demonstrated absent IENFDs at all 3 sites. Following the negative biopsy, Q-Sweat testing was performed to further evaluate for reinnervation, which identified sweat production at all locations.

SUMMARY/CONCLUSION: This case creates awareness of the utility of QSART as a tool to monitor treatment of SFN. The gold standard modality used to demonstrate reinnervation is IENFDs on skin biopsy. This method failed to display reinnervation at 2 years post-treatment despite marked clinical improvement. Q-Sweat testing was able to provide objective evidence of reinnervation through intact sweat production.

VELOPHARYNGEAL ELECTROMYOGRAPHY IN INFANTS WITH PIERRE ROBIN SEQUENCE

INTRODUCTION: Pierre Robin sequence (PRS)—retrognathia, glossoptosis, and cleft palate clinical triad—has a more severe clinical course and worse speech outcome than isolated cleft palate.

OBJECTIVE: To use velopharyngeal needle EMG to search for possible associations of respiratory, feeding, and speech outcomes.

METHODS: We designed a retrospective study of 92 children with isolated (72) or syndromic (20) PRS. Clinical grading systems were used to classify respiratory and feeding disorders. Needle EMG investigated the levator veli palatini muscle bilaterally. Outcome measures included: (1) the need for respiratory support, (2) the duration of enteral feeding, and (3) repeated phonological evaluations using the Borel-Maisonny classification of velopharyngeal insufficiency.

RESULTS: Velopharyngeal needle EMG patterns were normal (41; 44%), myogenic (44; 48%), or neurogenic (7; 8%). Neurogenic needle EMG signs were detected exclusively in patients with syndromic PRS. The frequencies of respiratory complications, the duration of enteral feeding, and the degree of velopharyngeal insufficiency were not statistically associated with abnormal EMG findings.

SUMMARY/CONCLUSION: Soft palate muscle denervation was a marker of syndromic PRS, but velopharyngeal needle EMG changes were not a predictor of phonological outcomes. Surgical techniques used for cleft palate repair remain the key prognostic factor for speech outcome in children with PRS.
ALCOHOLIC NEUROPATHY: CLINICAL CHARACTERISTICS BASED ON A CASE SERIES
Munazza Ahmed (Steubenville, OH), Igor Titoff (Pittsburgh, PA), Victoria Titoff (Pittsburgh, PA), Heather Maury (Pittsburgh, PA), Sandeep Rana (Pittsburgh, PA)

INTRODUCTION: Alcohol abuse is a well-known etiology of peripheral neuropathy; however, clinical characteristics of alcoholic neuropathy are not well delineated.

OBJECTIVE: To outline demographics (age, sex, and socioeconomic status), symptomatology, EDX, and laboratory findings, as well as response to abstinence.

METHODS: Retrospective chart review of 9 patients diagnosed with alcoholic neuropathy treated in 2014-2016.

RESULTS: The average age of patients was 50 years, with about equal incidence in males and females. All were socioeconomically well off; 8 out of 9 had stable careers. Only 1 had psychiatric illness. At least moderate exposure to alcohol spanned years in all except 1 who had only been drinking heavily for 6 months. Some of the patients were predominantly wine or beer drinkers. Others listed vodka and mixed drinks. All patients presented with numbness and pain in distal extremities and difficulty ambulating. Features of dysautonomia, namely high resting pulse, lightheadedness, and violaceous hue in distal extremities, was noted in some patients. Body mass index, B12, and albumin levels did not indicate malnourishment. NCSs revealed sensorimotor axonal peripheral neuropathy. On follow up, all patients reported symptomatic improvement or stabilization once they became abstinent.

SUMMARY/CONCLUSION: Alcoholic neuropathy should be suspected in middle-aged well-off patients presenting with painful neuropathy, particularly when no other etiology is forthcoming. Habitual wine drinking with dinner or beer consumption over a period of years can trigger neuropathy, which likely is secondary to toxic effects of alcohol rather than malnutrition.

PERIPHERAL NEUROPATHY IN PARKINSON’S DISEASE A CASE-CONTROL STUDY
Proel Perez Galdos (Lima, PE)

INTRODUCTION: Parkinson’s disease (PD) traditionally is seen as a motor disorder characterized by the triad of tremor, stiffness, and bradykinesia. Now, there is more interest in the concept that PD is a complex systemic disorder with many non-motor symptoms, including sensory disorders, and peripheral neuropathy being the most common characteristics in PD than had been previously thought.

OBJECTIVE: To determine the characteristics and prevalence of peripheral neuropathy in PD.

METHODS: A study of 23 case control PD patients in a peripheral neuropathy clinic was conducted (age of PD patients: 66.1±11.7 years; age of control subjects: 66.6±10.8 years; average disease duration: 7.7±7.5 years). Neurophysiologic tests were performed, and the Hoehn-Yahr scales (HY) and activities of daily living (ADL) of Schwab–England were applied.

RESULTS: Motor axonal or mixed neuropathy was present in 39.1% of PD patients and 17.4% of control subjects (OR: 3.05). Neuropathy was related to time of disease (>7 years) (p=0.042) and the stage 3-4 of HY (p=0.049).

SUMMARY/CONCLUSION: We observed a high prevalence of peripheral neuropathy in PD patients; its causality is not well established. Yet, the effects associated with the duration of disease affects the patient’s disability and hence their quality of life.

Proel Perez Galdos, MD
IFCN North American Chapter Fellowship Award Recipient
IDIOPATHIC OVERACTIVE BLADDER: IS IT OF NEUROPATHIC ORIGIN?
Rowaida Ali (New Cairo, EG), Naglaa A. Gadallah (Cairo, EG), Abeer Elzohiery (Sheraton, EG)

INTRODUCTION: Overactive bladder has devastating effects on quality of life. It can be associated with other medical comorbidities, such as urinary tract infections; thus, identifying its etiology can help with proper management.

OBJECTIVE: To investigate the possible association of subtle neurogenic affection in patients with idiopathic overactive bladder.

METHODS: A cross-sectional cutoff study was conducted on 30 females suffering from idiopathic overactive bladder (previously diagnosed by urodynamic study where other causes were excluded) as well as 10 healthy female volunteers. They were all subjected to thorough clinical assessment and electrophysiological studies in the form of: determination of pudendal nerve terminal motor latency, sacral reflexes latencies, pudendal somatosensory evoked potential P1 latency (PSSEP P1), and needle EMG of both the external anal and external urethral sphincters (patients only).

RESULTS: The patients showed a statistically significant prolongation of pudendal nerve terminal motor latency and sacral reflexes latencies than the control subjects (p<0.01), but they showed a non-significant difference regarding PSSEP P1 latency (p>0.05). Neuropathic needle EMG findings were positive in 22 patients (73.3%) in external anal sphincter needle EMG and in 23 patients (76.6%) in external urethral sphincter needle EMG.

SUMMARY/CONCLUSION: There is a possible attributing element of neuropathic affection in patients with idiopathic overactive bladder. We recommend integrated clinical, urodynamic, and electrophysiological studies of the pelvic floor to be included in evaluation of any overactive bladder patients.

Rowaida Ali, Master Degree PMR
IFCN North American Chapter Fellowship Award Recipient

MEDIAN NERVE AREA IN DIABETIC CARPAL TUNNEL SYNDROME PATIENTS: A RETROSPECTIVE REVIEW OF 1030 CASES
Ana Moreira (Campinas, Sao Paulo), Lisa Hobson-Webb (Durham, NC), Santoshi Billakota (New York, NY)

INTRODUCTION: Diabetes mellitus (DM) is a well-known risk factor for CTS, but the association of higher median nerve cross-sectional area (CSA) and DM is poorly defined.

OBJECTIVE: To determine if median nerve CSA is higher in diabetic CTS as compared to nondiabetic CTS.

METHODS: A total of 1030 consecutive patients presenting to the Duke EMG Laboratory during 2013-2014 with a final EDX diagnosis of CTS were examined. Median nerve CSA at the wrist (W-CSA) and forearm (F-CSA) were measured and a wrist-to-forearm ratio (WFR) was calculated. Statistical analysis was performed with ANCOVA test and Bonferroni correction using as covariates age, gender, pregnancy, handedness, and body mass index (BMI).

RESULTS: A total of 714 of these patients (mean age: 56.0±14.0 years; 36.8% male, 63.2% female; mean BMI: 32.2±8.0; 41.9% diabetic) underwent ultrasound. There was no difference between W-CSA of nondiabetic and diabetic patients (right W-CSA p=0.992, mean=12.8±4.7, left W-CSA p=0.253, mean=11.7±4.8). The same was observed with F-CSA (right F-CSA p=0.498, mean=6.5±2.2, left F-CSA p=0.181, mean=6.1±2.3) and WFR (right-WFR p=0.706, mean=2.1±0.9, left WFR p=0.444, mean=2.0±0.8). Bonferroni-correction p-values were also nonsignificant (value=1) for all the analysis. There were no differences in the severity of CTS between groups (right hand p=0.237, left hand p=0.458).

SUMMARY/CONCLUSION: There is no evidence that median nerve CSA is higher in diabetic patients with CTS as compared to nondiabetic CTS.
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MEDIAN NERVE AREA AND BODY MASS INDEX IN CARPAL TUNNEL SYNDROME PATIENTS: A RETROSPECTIVE REVIEW OF 1030 CASES
Ana Moreira (Campinas, SP), Lisa Hobson-Webb (Durham, NC), Santoshi Billakota (New York, NY)

INTRODUCTION: Obesity is a risk factor for CTS, but the association between median nerve cross-sectional area (CSA) and body mass index (BMI) is unclear.

OBJECTIVE: To determine if median nerve CSA is higher in overweight CTS patients.

METHODS: During 2013-2014, 1030 consecutive patients presenting to the Duke EMG Laboratory with a final EDX diagnosis of CTS were examined. Median nerve at the wrist (W-CSA) and forearm (F-CSA) were measured and wrist-to-forearm ratio (WFR) was calculated. Patients were classified into BMI groups: I=underweight/normal, II=overweight, III=moderately obese, IV=severely obese, V=morbidly obese. Statistical analysis was performed with ANCOVA test and Bonferroni correction using as covariates age, gender, pregnancy, and handedness.

RESULTS: A total of 714 patients (mean age: 56.0±14.0 years) underwent ultrasound and EDX testing. There was a significant difference between BMI groups for W-CSA (right W-CSA mean 12.8 mm², p=0.000; left W-CSA mean 11.7 mm², p=0.002), especially between underweight/normal and morbidly obese patients (right W-CSA mean 11.8 mm² versus 14.3 mm², p=0.001; left W-CSA mean 11.1 mm² versus 13.2 mm², p=0.004), and between overweight and morbidly obese patients (right W-CSA mean 12.2 mm² versus 14.3 mm², p=0.000; left W-CSA mean 11.3 mm² versus 13.2 mm², p=0.001), but not for F-CSA or WFR.

SUMMARY/CONCLUSION: In the current study, higher BMI is associated with increased W-CSA in patients with CTS.

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A RARE CASE OF IDIOPATHIC, PARTIAL ANTERIOR INTEROSSEOUS NERVE PALSY
Shahd Haidar (Hershey, PA), Aiesha Ahmed (Hershey, PA)

INTRODUCTION: Anterior interosseous nerve (AIN) palsy is a rare condition accounting for less than 1% of all nerve palsies in the upper limb. Multiple etiologies have been discussed in the literature and rarely have idiopathic cases been noted. Within the idiopathic category, no partial AIN palsy has been reported.

CASE REPORT: A healthy 63-year-old female presented with sudden-onset inability to flex her right thumb. She reported no trauma or illness but had previously developed a sudden onset of right shoulder pain which quickly resolved leaving her with right thumb flexion weakness. Cervical MRI was unremarkable. EDX testing revealed normal NCSs but the needle EMG, which included extensive sampling of muscles, showed active denervation in only the right flexor pollicis longus muscle with no axonal continuity.

DISCUSSION: In 1952, Kiloh and Nevin described the pathophysiology of AIN syndrome to be a neuritis. Others have reported cases due to trauma, venipuncture, or compression. There have been few cases of idiopathic AIN palsy, none of which have shown partial involvement of the flexor pollicis longus muscle. It is vital to recognize the etiology of AIN syndrome to prevent unnecessary surgical intervention. In non-traumatic cases of AIN palsy, spontaneous resolution of symptoms begins within the first 6 months.

CONCLUSION: Our patient’s symptoms improved significantly within the first 4 months of initial onset. Additional reports of idiopathic AIN syndrome may help to reduce the number of unnecessary surgical interventions.
CARPAL TUNNEL SYNDROME ELECTROPHYSIOLOGICAL PARAMETERS ACQUIRED USING THE E-NORMS TECHNIQUE
Joe Jabre (Los Angeles, CA), Joao Kouyoumdjian (Sao Jose do Rio Preto, SP), Vanessa Ferreira (Sao Jose do Rio Preto, SP)

INTRODUCTION: The e-norms technique, developed by Jabre, relies on identifying normal studies from a mixed normal and abnormal laboratory cohort.

OBJECTIVE: To use the e-norms technique to derive reference values for 3 median nerve absolute latencies.

METHODS: From January 2002 to March 2016, we selected 8688 consecutive patients from the EMG database with a reported conclusion of CTS or those deemed normal. Exclusions included any systemically-associated disease, nerve injuries, and peripheral neuropathy. The CTS group comprises 1882 patients, 84.1% female with mean age 49.2 years (19-95). The normal group comprises 1794 patients, 76.1% female with a mean age 41.4 years (14-84). CTS EDX assessment (using just right-hand values) was based on: (1) median sensory peak latency (MSP) to the index or middle fingers, 14 cm (≥3.70 ms); (2) median mixed palm peak latency (MPP), palm-to-wrist, 8 cm (≥2.30 ms); and (3) median motor distal latency (MMDL), wrist-abductor pollicis brevis, 8 cm (≥4.30 ms). If these 3 absolute latencies were normal, we studied comparative latencies, sensory median-to-ulnar ring finger, sensory median-to-radial thumb, and/or mixed palmar median-to-ulnar.

RESULTS: The e-norms derived practical limits were MSP ≥3.40 ms (3538 hands), MPP ≥2.20 ms (1561 hands), and MMDL ≥4.04 ms (3029 hands).

SUMMARY/CONCLUSION: The e-norms technique proved useful in identifying normal values for NCSs. The new reference values obtained were less for all 3 parameters studied: MSP (3.40 ms versus 3.70 ms), MPP (2.20 ms versus 2.30 ms), and MMDL (4.04 ms versus 4.30 ms).

Joao Kouyoumdjian, MD, PhD
IFCN North American Chapter Fellowship Award Recipient

MEDIAN AND ULNAR NERVE CONDUCTION REFERENCE VALUES BASED ON THE E-NORMS METHOD
Dongqing Zhu (Shanghai, PR), Yu Zhu (Syracuse, NY), Joe Jabre (Los Angeles, CA)

INTRODUCTION: NCSs are commonly used in the diagnosis of a variety of neuromuscular diseases. However, normal values for most nerves and age groups are not well documented for the Chinese population.

OBJECTIVE: To setup reference values of median and ulnar NCSs based on our laboratory (Huashan Hospital, Shanghai) population, equipment, and environment.

METHODS: One hundred needle EMG studies from January 2015 to December 2016 were reviewed, and latencies, amplitudes, and conduction velocity (CV) of median and ulnar nerves were recorded (motor and sensory nerves included, and patients’ age). E-norms were derived from this cohort (age range: 15-77 years) using the technique developed by Jabre. Data that lie in the plateau part of an inverted S curve derived from sorted data were used to calculate descriptive statistics.

RESULTS: The e-norms data were as follows: (1) for median motor NCSs: latency was 4.24±0.41 ms (3.42-5.07), amplitude was 11.62±1.59 mV (8.44-14.8), and CV was 56.73±3.77 m/s (49.2-64.26); (2) for median sensory NCSs: latency was 2.48±0.29 ms (1.9-3.06), amplitude was 19.59±3.46 µV (12.67-26.51), and CV was 47.45±7.1 m/s (33.26-61.64); (3) for ulnar motor NCSs: latency was 2.62±0.22 ms (2.17-3.06), amplitude was 12.96±1.21 mV (10.54-15.37), and CV was 54.17±3.73 m/s (46.71-61.63); (4) for ulnar sensory NCSs: latency was 2.02±0.11 ms (1.79-2.25), amplitude was 12.05±1.87 µV (8.32-15.78), and CV was 53.59±2.98 m/s (47.63-59.55). (Note: The distance for median and ulnar motor distal latency studies were both 7 cm.)

SUMMARY/CONCLUSION: The e-norms method seems to be useful in developing individual laboratory reference values from its own laboratory population. The variation in our test is low, 5.5-15.5%, and our data compared favorably with the published range.

Dongqing Zhu, MD
AANEM Foundation for Research and Education Award Recipient
IS IT POSSIBLE TO ELICIT LONG LATENCY SOMATOSENSORY EVOKED POTENTIALS WITH MECHANICAL STIMULATION?
Joel Gutierrez (La Habana, CU), Rachel Pérez-Lalana (La Habana, CU), Yodeisy Ferrer González (La Habana, CU), Norge Santiesteban (La Habana, CU), Lestayo Zurina (La Habana, CU), Benn Smith (Scottsdale, AZ)

INTRODUCTION: Scalp recorded long latency somatosensory evoked potentials (LLSEPs) have been previously elicited with different types of electrical stimulation. It is controversial whether LLSEPs can be also elicited with mechanical stimulation.

OBJECTIVE: To compare the electrophysiological features of LLSEPs elicited with electrical versus mechanical stimulation.

METHODS: LLSEPs were recorded in 20 healthy subjects (age: 31±8 years) from Cz-A1+A2 in response to 2 types of stimuli: electrical stimulation (ES) of the median nerve at the wrist (5 mA) and mechanical stimulation (MS) of the index finger fingernail with a reflex hammer synchronized with the EMG machine. Each type included 25 stimuli, given at variable inter-stimulus intervals (from 10 to 20 seconds). Peak latencies of the most prominent negative (N1) and positive (P1) deflections (between 80 and 300 ms), as well as the P1-N1 amplitude, were measured for both types of stimulation.

RESULTS: Very reproducible (CCR>90) LLSEP waveforms were elicited in all subjects in response to both types of stimuli. MS elicited responses with latencies significantly shorter than ES both for N1 (111±14 ms versus 130±11 ms; p<0.000) and P1 (189±40 ms versus 222±28 ms; p<0.000). N1–P1 amplitudes were very similar for both conditions (ES: 49±22 µV versus MS: 49±16 µV; p=0.84).

SUMMARY/CONCLUSION: These results demonstrate that it is possible indeed to elicit LLEPs with MS of the fingernails. The shorter latencies recorded with MS could be related to the activation of faster or more excitable fibers and/or receptors. LLEPs could be a very useful tool for the evaluation of peripheral and central somatosensory pathways.

Joel Gutierrez, MD, PhD
IFCN North American Chapter Fellowship Award Recipient

EFFECTS OF HYPERBARIC OXYGEN ON SENSORY CONDUCTION PARAMETERS IN A GROUP OF EGYPTIAN DIABETIC NEUROPATHY PATIENTS A LONGITUDINAL CROSS-SECTIONAL STUDY
Mohamed Sherif El Morsi (Alexandria, EG), Waj Meintjes (Cape Town, EG)

INTRODUCTION: Research work assessing the effect of hyperbaric oxygen therapy (HBOT) on diabetic neuropathy using EDX studies is scarce. HBOT has potential benefit in diabetic neuropathy based on its anti-ischemic and metabolic modulatory effects.

OBJECTIVE: To objectively assess this potential benefit using sensory NCSs.

SUBJECTS/METHODS: A group of Egyptian diabetic patients were selected, and sensory NCSs were performed on their peripheral nerves in the upper and lower limbs. The sensory NCSs were repeated after 10 sessions of HBOT, and results from both settings were compared and analyzed.

RESULTS: Statistical analysis showed significant differences between before and after HBOT in sensory amplitudes and conduction velocities. The most significant changes were found in the sensory amplitudes and conduction velocities of the median, ulnar, and sural nerves.

CONCLUSION: This result confirms the value of HBOT as adjunct treatment in diabetic neuropathy.

Mohamed Sherif El Morsi, MD
AANEM Foundation for Research and Education Award Recipient
FACTORS ASSOCIATED WITH INTUBATION IN HIV+ PATIENTS PRESENTING WITH CRITICAL ILLNESS MYOPATHY OR POLYNEUROPATHY, A NEW YORK STATEWIDE PLANNING AND RESEARCH COOPERATION SYSTEM DATABASE REPORT

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INTRODUCTION: Critical illness myopathy (CIM) and polyneuropathy are complications observed in human immunodeficiency virus (HIV)+ patients.

OBJECTIVE: To evaluate factors associated with intubation and mechanical ventilation (MV) in HIV+ patients diagnosed with chemotherapy induced polyneuropathy (CIP) or CIM.

METHODS: We sought factors associated with MV in HIV+ adult inpatients diagnosed with CIM or CIP, utilizing the New York Statewide Planning and Research Cooperation System database for the 1998-2014 period. Data were analyzed via χ², Mann-Whitney U, and t-test.

RESULTS: Comparing MV versus non-MV groups, 158 HIV+ CIM/CIP patients were analyzed: 41% (65) necessitated MV, 88% (57) of these requiring >96 hours. The MV group had significantly higher pneumonia rates (52% versus 16%, p<0.001). Mean age was similar (50±10.5 years versus 49±13.5 years) as well as rates of concomitant diabetes, hypertension, congestive heart failure, chronic obstructive pulmonary disease, obesity, or corticosteroid treatment. Median hospitalization length was significantly prolonged in the MV group (34 [range: 23-54] days versus 10 [range: 5-22] days, p<0.001). Mortality was significantly higher in the MV group (34% versus 5%, p<0.001); discharges home were higher in the non-MV group (12% versus 36%, p<0.001). Average hospitalization charge was significantly higher in the MV group. ($345,000 versus $71,200, p<0.001).

SUMMARY/CONCLUSION: MV is associated with significant increase in hospitalization charges and length of stay in HIV+ CIM/CIP patients. Corticosteroid administration was not significantly different between the studied groups. There is work in progress to further identify risks associated with MV in HIV+ patients with CIM/CIP.


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INTRODUCTION: Guillain–Barré syndrome (GBS) has been reported as a possible sequela of influenza vaccination.

OBJECTIVE: To investigate the association between GBS and influenza vaccination in children.

METHODS: The Vaccine Adverse Event Reporting System (VAERS) is a self-reporting public database for post-vaccine events. VAERS data for 1991-2015 were analyzed, and we identified patients less than 18 years of age reporting GBS after influenza vaccination (Codes FLU3, FLU4, FLUN3, FLUN4, and FLUR3). The initial 6 weeks after vaccination were defined as a risk period for possible cause-effect, the subsequent 6 weeks as a control period, and beyond 13 weeks as remote. Case-centered and self-controlled case analysis study designs were employed.

RESULTS: There were 92 cases of GBS (mean age 7.8±5.53 years; 48.91% male) reported after influenza vaccination for the defined period: 80 (86.95%) during the risk period, 11 (11.95%) during the control period, and 1 (1.08%) in the remote period. Cases reported during the risk period were via Brighton Criteria levels, in descending order of diagnostic certainty levels: I (9%), II (2%), III (12%), and IV (77.5%). Case-centered analysis yielded reciprocating results.

SUMMARY/CONCLUSION: Incidence of GBS after flu vaccination in children was 2.5/10 million vaccinations, below that seen the general population. However, analyses yielded an unbalanced distribution of GBS reports after vaccination, most cases reporting within the risk period. This may suggest that some cases could be triggered by vaccination. Further investigation and the implementation of active surveillance after influenza vaccination is warranted.
FACTORS ASSOCIATED WITH INTUBATION IN CRITICAL ILLNESS POLYNEUROPATHY (CIP), AN ANALYSIS OF NEW YORK STATEWIDE PLANNING AND RESEARCH COOPERATION SYSTEM (SPARCS) DATABASE REPORT FROM 1998 TO 2014.

Francisco Gomez (West Orange, NJ), Jesyree Veitia (West Orange, NJ), Abu Nasar (Newark, NJ), Jeffrey Kornitzer (Newark, NJ), Nizar Souayah (Newark, NJ)

INTRODUCTION: Critical illness polyneuropathy (CIP) can complicate management of severely ill patients, especially those requiring mechanical ventilation (MV).

OBJECTIVE: To evaluate factors associated with MV in patients with CIP.

METHODS: We analyzed factors associated with MV in adult inpatients with CIP utilizing the New York Statewide Planning and Research Cooperation System (SPARCS) database for the 1998-2014 period. Analysis was performed via chi-square, Mann-Whitney U, and t-test.

RESULTS: Included were 2945 CIP patients; 886 (30%) underwent MV, 810 (91%) of these necessitated >96 hours MV. Patients requiring MV were younger (62±15.7 years versus 64±16.5 years). Factors seen more frequently in the MV group included congestive heart failure (30.9% versus 19.8%, p<0.001) and pneumonia (51.8% versus 7.3%, p<0.001). Factors observed less frequently in intubated patients included diabetes (19% versus 28.6%, p<0.001), and hypertension (21.7% versus 37.3%, p<0.001). In the MV group, 1.2% of the population had received IV immunoglobulin versus 5% in the non-MV group (p<0.001).

Gender, chronic obstructive pulmonary disease, parkinsonism, multiple sclerosis, obesity, and corticosteroid or plasmapheresis treatments did not vary significantly between groups. Median length of stay was significantly longer in the MV group (37 [range: 23-56] days versus 14 [range: 8-21] days, p<0.001). Mortality and hospitalization charges were higher in the MV group (28.9% versus 3.2%, p<0.001; $399,800 versus $77,300).

SUMMARY/CONCLUSION: MV has been associated with increased mortality, hospitalization charges, and median length of hospitalization in CIP patients. There is work in progress to further determine risk factors associated with MV in CIP.

Francisco Gomez, MD
Resident and Fellow Member Award Recipient

FACTORS ASSOCIATED WITH INTUBATION AND MECHANICAL VENTILATION IN CRITICAL ILLNESS MYOPATHY (CIM), AN ANALYSIS OF NEW YORK STATEWIDE PLANNING AND RESEARCH COOPERATION SYSTEM (SPARCS) DATA 1998-2014.

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INTRODUCTION: Intubation may be required in the setting of critical illness myopathy (CIM).

OBJECTIVE: To evaluate factors associated with intubation in patients with CIM.

METHODS: We analyzed factors associated with intubation and mechanical ventilation (MV) in adult patients with CIM using the New York Statewide Planning and Research Cooperation System (SPARCS) database for the 1998-2014 period. Data were analyzed via chi-square, Mann-Whitney U, and t-test.

RESULTS: Of the 3922 CIM patients analyzed, 973 (25%) underwent intubation, 862 (89%) of which required >96 hours MV. Intubated patients tended to be younger (62±16.2 years versus 68±15.7 years). Pneumonia patients required MV more frequently (47.6% versus 8.3%, p<0.001), as well as those receiving plasmapheresis (0.7% versus 0.1%, p<0.001) or corticosteroids (5.9% versus 0.6%, p<0.001). Factors observed less frequently in the MV group included diabetes (22.7% versus 35.1%, p<0.001), hypertension (23.3% versus 44.6%, p<0.001), and obesity (8% versus 11.06%, p<0.001). Gender, concomitant congestive heart failure, chronic obstructive pulmonary disease, parkinsonism, multiple sclerosis, or lupus erythematosus did not vary significantly between groups. Length of stay for MV patients was significantly longer (34 [range: 22-54] days versus 14 [range: 9-18] days, p<0.001). Mortality and hospital charges were higher in the MV group (22.5% versus 2.5%, p<0.001; $363,000 versus $80,300, p<0.001). Discharges to home were higher in the non-MV group (20.3% versus 7.6%).

SUMMARY/CONCLUSION: Patients with CIM requiring MV had higher mortality. It remains to be seen whether early and aggressive treatment of some of the identified factors associated with MV, including pneumonia, could possibly reduce these instances.

Francisco Gomez, MD
Resident and Fellow Member Award Recipient
CLINICAL UTILITY, SAFETY AND TOLERABILITY OF PAIRED-PULSE TRANSCRANIAL MAGNETIC STIMULATION IN THE FIRST SEIZURE CLINIC
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INTRODUCTION: Paired-pulse transcranial magnetic stimulation (ppTMS) can be used to measure cortical excitability in the interictal state; long-interval intracortical inhibition (LICI) being reduced in patients with epilepsy when compared to healthy control subjects. In this study, we retrospectively reviewed the clinical utility of measuring LICI in patients presenting with a first unprovoked seizure. Data regarding safety and tolerability are also presented.

METHODS: We identified 132 patients who presented with a first unprovoked seizure referred for a sleep deprived electroencephalogram (sdEEG) and ppTMS. Clinical, electrographic, neuroimaging, and ppTMS data at presentation were collected, and longterm outcomes were obtained by reviewing hospital medical records and via telephone interviews. We assessed sensitivity, specificity, and predictive value of LICI for a diagnosis of epilepsy at greater than 3 years of followup, and group level differences in cortical excitability.

RESULTS: ppTMS was well tolerated and safe (1.5% per subject seizure risk). LICI (at 200 ms and 250 ms interstimulus intervals) showed high specificity (93.3% and 80%) but low sensitivity (21.1% and 28.8%) for a diagnosis of epilepsy. Positive predictive value of ppTMS for an eventual diagnosis of epilepsy was 85.7%. At a group level, LICI was significantly reduced in those diagnosed with epilepsy compared to those without further unprovoked seizures (p=0.019 at 200 ms interstimulus intervals [ISI] and p=0.044 at 250 ms ISI).

SUMMARY/CONCLUSION: We provide retrospective evidence that ppTMS can be useful in predicting longterm outcome after presentation with a first unprovoked seizure and can be used safely as part of an initial evaluation in the first seizure clinic.

RAPIDLY PROGRESSIVE DEMENTIA, PROFOND CORTICAL ATROPHY, AND AUTONOMIC NEUROPATHY IN A YOUNG MAN WITH LOW LEVELS OF GANGLION ACETYLCHOLINE RECEPTOR (G-ACHR) ANTIBODIES
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INTRODUCTION: Ganglionic acetylcholine receptor (G-Achr) antibodies are usually described with autonomic neuropathy and previously not observed in the setting of rapidly progressive dementia with cortical atrophy.

OBJECTIVE: To describe a case of rapidly progressive dementia, cortical atrophy, and autonomic symptoms in a young man with low titers of G-AChr antibodies.

METHODS: A 33-year-old man who presented initially with knee pain was found to have progressive memory impairment, dysphagia, 40-lb weight loss, unsteady gait, and urinary retention over the prior 6 months. On examination he appeared cachectic, with psychomotor slowing, poor attention, and recall, and had full strength, increased tone, diffuse hyperreflexia, transient upper extremity asymmetrical myoclonic jerks, and an ataxic gait. Workup revealed severe cortical atrophy and a mildly elevated serum titer for G-Achr antibodies (0.06). Treatment with IV steroids, IV immunoglobulin, and plasmapheresis revealed some delayed clinical improvement. Repeat antibody testing about 3.5 months later was negative. All other testing for paraneoplastic, autoimmune, infectious, gastrointestinal (GI), and heavy metal etiologies were negative. Cerebrospinal fluid was remarkable for elevated protein (70) and negative 14-3-3 protein. Positron emission tomography scan was without abnormalities. Autonomic function test was grossly normal, but skin biopsy revealed markedly reduced epidermal nerve fiber density consistent with small fiber neuropathy.

SUMMARY/CONCLUSION: It is reasonable to assume that G-Achr antibodies were related to this patient’s autonomic symptoms and small fiber neuropathy. They have been observed in GI disorders such as dysphagia. Given the otherwise negative workup, the presence of G-Achr antibodies needs clarification in future studies as to whether they are playing a role in the rapidly progressive dementia and diffuse cortical atrophy seen.
THE VALUE OF ELECTROPHYSIOLOGICAL Typing AND Conduction block FOR Prediction OF Functional outcome in Guillain-Barré Syndrome

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INTRODUCTION: Guillain–Barré syndrome (GBS) mainly consists of acute inflammatory demyelinating polyradiculoneuropathy (AIDP) and acute motor axonal neuropathy (AMAN). In AMAN, conduction block (CB) could be reversible or followed by axonal degeneration.

OBJECTIVE: To identify the correlation between CB and the functional outcome for patients with AIDP or AMAN.

METHODS: We prospectively recruited 52 GBS patients for serial electrophysiological tests and disability evaluation on admission and 1 month and 6 months after treatment with IV immunoglobulin. Patients were classified into AIDP, AMAN, equivocal, or normal. AMAN patients who had followup electrophysiological tests were further classified into 3 types.

RESULTS: Electrophysiological study showed 20 patients with AIDP, 24 with AMAN, 7 equivocal, and 1 normal. Probable or definite CB was observed in 11 AIDP and 16 AMAN patients. AMAN with CB had more reduction of Hughes grade at 1 month (1.71±0.83 versus 0.43±0.79, p=0.003) and a lower percentage of patients with slow recovery (unable to walk independently at 6 months) (7% versus 57%, p=0.025) than AMAN without CB. There were no significant differences in reduction of Hughes grade at 1 month between AIDP with and without CB. Among the 13 AMAN patients who were followed up, 4 had typical AMAN without CB (type 1), 7 had reversible CB (type 2), and 2 had CB and subsequent axon degeneration (type 3). Hughes grades at nadir were similar, while patients with reversible CB (type 2) had the greatest Hughes grade reduction at 1 month.

SUMMARY/CONCLUSION: EDX results of those with AMAN with CB, especially reversible CB, might be a marker of good recovery.
WHAT IS THE MOST SENSITIVE ELECTRODIAGNOSTIC CRITERIA FOR THE DIAGNOSIS OF GUILLAIN BARRÉ SYNDROME
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INTRODUCTION: Clinical electrophysiological studies are important in the evaluation of patients with Guillain–Barré syndrome (GBS) Several criteria have been proposed, both for acute inflammatory demyelinating polyneuropathy (AIDP) and for the axonal form acute motor axonal neuropathy (AMAN). They have different sensitivities for diagnosis in clinical practice.

OBJECTIVE: To assess the sensitivity of different EDX criteria for early detection and characterization of GBS in an Argentinian cohort.

METHODS: We retrospectively compared 7 published sets of electrophysiological criteria (designated by first author: Albers, Van Den Berg, Ho, Hadden, Italian Guillain-Barré Study Group, Cornblath, and Rajabally) in patients with a clinical diagnosis of GBS, who were consecutively included in our International Guillain-Barré Syndrome Outcome Study (IGOS) cohort between 2014 and 2017. Electrophysiological tests were performed twice: at diagnosis and within 4 weeks.

RESULTS: Fourteen patients were included (mean age: 45.9 years; 9 male). The criteria with the highest sensitivity for AIDP in the first study was from Albers (71.42%) followed by Van Den Berg and Hadden (64.3% and 57%, respectively). The second study increased the sensitivity to 82.46%, 80.51%, and 66.23% (Van Den Berg, Albers, and Hadden, respectively). AMAN was identified in 35.7%, increasing up to 44.79% with the second study, under the Rajabally criterion, followed by Ho (14.28%).

SUMMARY/CONCLUSION: AIDP was the most prevalent form of GBS. The highest sensitivity was with the Albers and Van Den Berg criteria. Sensitivity is further increased in equivocal cases when a second study is performed. Using the recent criteria by Rajabally, GBS can be characterized as axonal in over 40% of patients.

Maria Lucia Rattagan, MD
IFCN North American Chapter Fellowship Award Recipient

SELPERCEIVED ABILITY TO ESTIMATE VARYING WEIGHTS IN PERIPHERAL NEUROPATHY COMPARED TO NON-PERIPHERAL NEUROPATHY PATIENTS
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INTRODUCTION: Common symptoms of polyneuropathy (PN) are weakness and sensory impairment.

OBJECTIVE: To assess the perception of varying weights at the ankle in PN and non-PN groups.

METHODS: A total of 38 subjects with electrophysiologically-documented PN and normal subjects (aged 18 to 80; 16 PN, 22 control subjects) participated. An initial weight (N0) was applied to the subject's ankle for 1 minute to assess the weight while sitting. Then, the N0 was replaced with the next weight (N1); the subject had 1 minute to assess the N1 and provide a response: heavier, lighter, or the same as the previous weight (N0). This process was repeated 10 times in the same sequence for every subject. The weight differences were from 0, 0.25, 0.5, 1.0, and 2.0 lb. Both the participants and examiners were blinded to the correct responses until the end of the study.

RESULTS: The control group outperformed the PN group at weight differences of 0 (55% to 38% correct responses, respectively), 0.25 (63.6% to 53.1%), 1.0 (84.1% to 68.8%), and 2.0 lb (100% to 94%). At 0.5 lb difference, the PN group outperformed the control subjects (63% to 52%).

SUMMARY/CONCLUSION: The data suggest that PN subjects have a lower likelihood of correctly assessing the varying weights at the ankle compared to normal subjects. The changes in weights of 0 and 1.0 lb seem to provide a greater value in distinguishing PN from normal subjects. Future study with more subjects can help clarify if the perception of varying weights at the ankle has a diagnostic value in identifying PN subjects.

Maria Lucia Rattagan, MD
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JITTER PROBABILISTIC CHARACTERIZATION
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INTRODUCTION: Bayesian probabilistic characterization has already been used for quantitative EMG but not for jitter analysis. Instead of the classical statistical method, probabilistic characterization uses sequential cumulative probability.

OBJECTIVE: To evaluate the performance of jitter probabilistic characterization.

METHODS: We retrospectively analyzed 20 control subjects and 20 patients with serologically-confirmed myasthenia gravis (MG). All patients were submitted to orbicularis oculi concentric needle voluntary jitter measurements of 20 pairs of apparent single fiber action potentials (ASFAPs), adding up 797 mean consecutive difference (MCD) measurements. Jitter was characterized in 4 different levels: A: <25 µs, B: 25-45 µs, C: 45-65 µs, and D: >65 µs. We calculated the probability of an abnormal result for each jitter level. Then, we used the Bayesian sequential approach in each subject until the cumulative probability of a normal or abnormal result was >99%.

RESULTS: The probability of abnormality for each jitter level was determined as: A: 20%, B: 73%, C: 95%, and D: 99%. The cumulative probability achieved more than 99% up to the sixth potential in 77% of the subjects. However, 2 control subjects were labeled as abnormal studies, while 4 MG subjects were labeled as normal studies. Additionally, 2 control subjects could not be characterized up to the 20th recording. The method accuracy was 80%.

CONCLUSION: Probabilistic characterization requires fewer recordings than the statistical method. However, our study showed poor accuracy for jitter characterization. One possible reason is that MCD values in each patient are not randomly scattered during the entire examination, but rather clustered according to the sampled region.

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UTILITY OF CONVENTIONAL AUTONOMIC STUDIES IN INDIVIDUALS WITH PRE-DIABETES, DIABETES, AND NON-DIABETIC PATIENTS: A VETERANS AFFAIRS STUDY.

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INTRODUCTION: Previous studies demonstrated that individuals with prediabetes and diabetes can suffer several types of autonomic neuropathy. Abnormalities with heart rate response to deep breathing (HRDB) and quantitative sudomotor axon reflex testing (QSART) can be recognized early. Can conventional autonomic function (ANF) studies help to quantify and characterize these different entities?

OBJECTIVE: To evaluate autonomic study results in individuals with prediabetes, diabetes, and non-diabetes from Edward Hines Veterans Affairs (VA) hospital.

METHODS: We chose 171 patients from our autonomic laboratory over the last 2 years. Conventional studies (HRDB, Valsalva maneuver, QSART, and tilt table testing, or TTT) were performed in all. Their HbA1C was tested within 6 months of ANF.

RESULTS: In non-diabetics (n=21), abnormalities were found with: QSART=11 (52.3%), HRDB=6 (28.6%), Valsalva=5 (23.8%), and TTT=8 (38.1%). In prediabetics (n=24), abnormalities were found with: QSART=9 (37.5%), TTT=3 (12.5%), HRDB=4 (16.7%), and Valsalva=3 (12.5%). In the diabetic population (n=126), abnormalities were found with: QSART=71 (56.3%), HRDB=33 (26.2%), Valsalva=49 (38.9%), and TTT=37 (29.4%).

SUMMARY/CONCLUSION: At our institution, prediabetics demonstrated an abnormal QSART, but were less likely to have an abnormal HRDB or more than 2 autonomic abnormalities in comparison to diabetic patients. QSART and HRDB are unable to distinguish between groups; however, Valsalva phases and TTT seem to be more abnormal in diabetics compared to prediabetics. Combining QSART either alone or in any combination with HRDB, Valsalva, or TTT did not significantly differentiate prediabetic or diabetic versus nondiabetic groups.
ACUTE MOTOR AXONAL NEUROPATHY AND MYASTHENIA GRAVIS OVERLAP SYNDROME
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INTRODUCTION: Myasthenia gravis (MG) is a disorder of neuromuscular transmission caused by antibodies to the acetylcholine receptor (AChR). In this report, we describe simultaneous onset of acute motor axonal neuropathy (AMAN) and MG.

CASE REPORT: A 69-year-old man with insulin-dependent diabetes mellitus and cervical fusion presented with a 1-week history of rapidly progressive bilateral shoulder and neck extension weakness and orthopnea. MG was suspected. Neurologic examination was notable for minimal asymmetric fluctuating ptosis, mild facial diplegia, and less than antigravity strength of the neck extensors and shoulder abductors. Reflexes were diminished, and there was sensory loss in the distal feet. NCSs were technically limited by excessive sweating, tachycardia, and tachypnea. Needle EMG revealed widespread fibrillation potentials and reduced recruitment. AMAN was suspected; anti-ganglioside serologies were requested prior to initiating IV immunoglobulin and demonstrated asialo-GM1 and GD1a antibodies. Minimal clinical improvement occurred. Further evaluation revealed positive AChR binding antibodies and a 50% decrement on 3-Hz repetitive nerve stimulation of the trapezius, prompting initiation of plasma exchange and prednisone (60 mg). Repeat needle EMG revealed widespread spontaneous activity and unstable motor unit potentials with reduced recruitment consistent with the expected evolution of AMAN. Strength recovered over the next 5 weeks.

CONCLUSION: Guillain–Barré syndrome and MG overlaps are rare, with 1 AMAN–MG case previously reported. This case highlights the importance of routine needle EMG during the evaluation of suspected MG as acute denervation exaggerates the decrement and postactivation exhaustion observed during 3-Hz repetitive nerve stimulation.

PREDICTIVE VALUE OF INTRAOPERATIVE BULBOCAVERNOSUS REFLEX DURING UNTETHERING SURGERY FOR POST-OPERATIVE VOLUNTARY VOIDING
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INTRODUCTION: Neurogenic bladder is one of the major disabilities in tethered cord syndrome. Intraoperative monitoring of the bulbocavernosus reflex (BCR) is known to help predict and prevent bladder dysfunction after untethering surgery. However, its predictive value for postoperative voiding function has not been confirmed in children with spinal dysraphism.

OBJECTIVE: To evaluate clinical significance of intraoperative BCR during untethering surgery of tethered cord syndrome to predict postoperative voiding function.

METHODS: We performed a retrospective review of 64 pediatric patients who underwent untethering surgery and whose BCR at baseline was obtainable. They were classified based on whether BCR was preserved or lost during surgery. As a functional outcome, voluntary voiding without need of assistive technique (such as intermittent catheterization or Valsalva maneuver) was checked at admission, at discharge, 2 months, and 6-12 months after surgery.

RESULTS: Among the 64 patients, BCR was lost during surgery in 12 and preserved in 52. The positive predictive value of intraoperative BCR (failure to void/loss of BCR) was 58.3%, 50%, and 44.4% at discharge, 2 months, and 6-12 months after surgery, respectively. The negative predictive value (independent voiding/preservation of BCR) was 67.3%, 76.9%, and 91.7% at the same time points.

SUMMARY/CONCLUSION: Intraoperative BCR during untethering surgery in children with spinal dysraphism can predict longterm bladder function with high specificity (86.8%) and moderate sensitivity (57.1%). It indicates that when BCR is preserved, voluntary voiding function can be reliably expected after surgery.
CASE SERIES OF SUSPECTED TARSAL TUNNEL SYNDROME IN JAPAN
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INTRODUCTION: Tarsal tunnel syndrome (TTS) is usually diagnosed based on clinical symptoms and signs and EDX investigations, typically NCSs, although the standard diagnostic criteria are yet to be established. Furthermore, TTS may be rarer in Japan than in western countries, possibly due to the custom that the people commonly remove their shoes at home. There are few large case series of TTS in Japan, especially those with EDX studies.

OBJECTIVE: To present a case series of TTS in Japan and investigate the diagnostic role of electrophysiological measures.

METHODS: We searched our EMG database from 2008 to 2016 with the keyword of TTS, and retrospectively reviewed clinical and EDX of extracted patients. The entry criteria included: (1) clinical diagnosis of TTS by the referring doctor, (2) numbness of sole and a positive Tinel’s sign at the ankle, and (3) that both the tibial motor NCSs and plantar sensory NCSs were conducted.

RESULTS: Enrolled were 12 patients (age range: 20-82 years; 9 men, 3 women). TTS was confirmed by NCSs in 3 patients with abnormal plantar sensory NCSs on the affected side. In 2 of them, tibial motor NCSs showed a bilobed compound muscle action potential (CMAP) indicating the isolated delay of the medial plantar nerve. NCSs were normal or nondiagnostic for the other 9 patients, although 1 had an abnormal needle EMG only on the affected side.

CONCLUSION: TTS is in general difficult to confirm electrophysiologically. Bilobed CMAP may be a previously undescribed sign of TTS.
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IMPACT OF SLEEP-DISORDERED BREATHING ON MOTOR AND NON-MOTOR SYMPTOMS IN MULTIPLE SYSTEM ATROPHY
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INTRODUCTION: Although several studies suggest that sleep-disordered breathing (SDB) is a frequent symptom of multiple system atrophy (MSA), whether SDB has influence on the motor and non-motor symptoms of MSA is unknown.

OBJECTIVE: To elucidate whether SDB has an impact on other clinical features of MSA; and meanwhile, to explore the associations between SDB and other sleep symptoms.

METHODS: A total of 40 MSA patients and 40 healthy volunteers (HVs) underwent video polysomnography (PSG) in the current study. All the MSA individuals were assessed using the Epworth Sleepiness Scale (ESS), Unified Multiple-System Atrophy Rating Scale (UMSARS), Hamilton Depression Scale (HAMD), Hamilton Anxiety Scale, Frontal Assessment Battery (FAB), Parkinson’s Disease Questionnaire-39 (PDQ-39), and Montreal Cognitive Assessment (MoCA).

RESULTS: The apnea-hypopnea index (AHI) of the MSA patients recorded by PSG was 16.4±20.2. SDB was found in 65% of the MSA patients (26/40), which was significantly higher than for the HVs (8/40, 20%) (p=0.0001). Compared to the MSA patients without SDB, MSA individuals with SDB showed higher total UMSARS, UMSARS-Ⅱ, FAB, and HAMD scores, more frequent occurrence of excessive daytime sleepiness, hypopneas, longer mean times for hypopneas, and obstructive sleep apnea (OSA), as well as longer time for OSA. This study suggested that SDB is frequently seen in MSA patients.

SUMMARY/CONCLUSION: MSA individuals with SDB are prone to severe motor deficits, depression, frontal lobe dysfunction, and excessive daytime sleepiness.

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SEVERITY OF GLYCATED HEMOGLOBIN VERSUS DURATION OF DIABETES: ROLE OF CONVENTIONAL AUTONOMIC STUDIES IN A REVIEW FROM VETERANS AFFAIRS HOSPITAL.
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INTRODUCTION: Previous studies demonstrated that clinical autonomic failure varies with duration of diabetes and glycated hemoglobin (HbA1C), and cardiac adrenergic denervation develops with advanced diabetic neuropathy.

OBJECTIVE: To evaluate autonomic study results in individuals based upon the levels of HbA1C and duration of diabetes from Edward Hines Vetrans Affairs (VA) hospital.

METHODS: We chose 171 patients from our autonomic laboratory over the last 2 years. All patients completed 4 conventional autonomic function (ANF) studies, including heart rate response to deep breathing (HRDB), Valsalva maneuver, quantitative sudomotor axon reflex test (QSART), and tilt table testing; they also had their HbA1C tested within 6 months, irrespective of the reason for referral. Prediabetes is defined as HbA1C of ≥5.7 and diabetes (type 2) ≥6.4%.

RESULTS: About 80.0% of patients with HbA1C >9 had ≥3 abnormal ANF measures, compared to only 1 (4.2%) pre-diabetic (HbA1C <6.4) patient. Similarly, 32.6% of those with diabetes mellitus (DM) for >9 years had ≥3 abnormal ANF measures. In patients with DM for ≥4 years, abnormal Valsalva was seen in 40.2% and abnormal HRDB was demonstrated in 28.4%; whereas among prediabetics, only 12.5% had abnormal Valsalva and 16.7% demonstrated an abnormal HRDB. QSART was the most common ANF abnormality across all variables.

SUMMARY/CONCLUSION: At our institution, we found that patients with longer disease duration and higher HbA1Cs were more likely to have ≥3 abnormal ANF results, with HbA1C >9 having a stronger correlation. Abnormality with Valsalva phases and HRDB testing is seen with longer disease duration consistent with cardiac adrenergic and cardiovagal denervation. Early detection of autonomic dysfunction would be desirable for better individual risk stratification.
INSIGHTS INTO CLINICAL AND ELECTRODIAGNOSTIC FEATURES OF CHEMOTHERAPY INDUCED PERIPHERAL NEUROPATHY
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INTRODUCTION: Chemotherapy-induced peripheral neuropathy (CIPN) is a major cause of dose modification in chemotherapeutic regimens and creates significant morbidity and reduced quality of life. The natural history and diagnostic approach for early CIPN have not been well defined.

OBJECTIVE: To prospectively characterize the incidence and clinical and EDX features of CIPN.

METHODS: Patients with breast, ovarian, or colon cancer receiving neurotoxic chemotherapy were prospectively evaluated prior to treatment and at 3-6 week intervals. CIPN diagnosis was based on symptoms and signs including validated neuropathy impairment and examination scales: total neuropathy score (TNSc) and Utah Early Neuropathy Score (UENS). NCSs were performed in participants with CIPN. Distal leg skin biopsy for intra-epidermal nerve fiber density (IENFD) measurement was performed in those with normal NCS results.

RESULTS: Of the 80 patients reviewed, 34 (42%) developed clinical CIPN. The average TNSc score in this population was 5.5±2.3, with a UENS average of 8.2±4.9. The UENS pin-prick sub-score was abnormal in 27/34 (79%), and vibration sensation was reduced in 30/34 (88%). Of the 34 participants with CIPN, 19 (56%) had an abnormal NCS. In the 15 participants with a normal NCS, 7 consented to skin biopsies and 6/7 (88%) had normal IENFD.

CONCLUSION: Clinical CIPN occurred in 42% of patients. All patients reported sensory symptoms, and a minority experienced motor or autonomic involvement. Preferential small fiber involvement may account for the poor sensitivity of NCSs. The normal IENFD suggests a functional deficit rather than axonal degeneration, accounting for neuropathic symptoms in those with normal IENFD and NCSs.

THE SPECTRUM OF PERIPHERAL NERVE INJURIES FROM GUNSHOT WOUNDS: A CASE SERIES FROM A SINGLE EVENT
Ilya Zolotnik (Loma Linda, CA), Laura Nist (Loma Linda, CA), Jeffrey Rosenfeld (Loma Linda, CA), Bryan Tsao (Loma Linda, CA)

INTRODUCTION: The variety of injury following a mass casualty incident provides an opportunity to appreciate the diversity of peripheral nerve pathology caused by gunshot wounds (GSWs).

OBJECTIVE: To describe the mechanisms, pathophysiology, and prognosis of different GSW-related peripheral nerve injuries.

CASE STUDY: All patients from a single event were wounded by a .223 caliber assault rifle. Patient 1 suffered 7 GSWs with scapular fracture, adjacent soft tissue injury, and a partial axon loss suprascapular mononeuropathy. Patient 2 suffered 2 GSWs with laceration of the axillary artery. Thrombosis of this vessel led to ischemic monomelic neuropathy with a gradient of distal-to-proximal weakness and sensory loss. Patient 3 suffered 4 GSWs, 1 in the proximity of the left brachial plexus. This resulted in a pan-brachial plexopathy of mixed pathophysiology (axon loss and demyelinating conduction block).

CONCLUSION: Most GSWs result in a near miss to peripheral nerves but the effect can be as destructive as a direct hit. Their mechanism of injury, pathophysiology, and prognosis include: (1) contusion and stretch: with axon loss due to adjacent bone/soft tissue damage and variable prognosis; (2) proximal large vessel thrombosis: with axon loss from ischemic monomelic neuropathy in a distal-to-proximal gradient and poor prognosis; and (3) acute compression from projectile gases: with mixed axon loss and focal demyelination with good prognosis relative to the extent of demyelination and inversely proportional to the extent of axon loss.

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Resident and Fellow Member Award Recipient
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ULTRASOUND FOR THE INVESTIGATION OF ULNAR NEUROPATHY
Yao Feng Chong (Singapore, SG), Einar Wilder-Smith (Singapore, SG)

INTRODUCTION: Eliciting the localization and etiology of ulnar neuropathy can be difficult based on history, examination, and EDX investigations alone. Nerve ultrasound (US) can be a useful ancillary, point-of-care test.

OBJECTIVE: To determine the utility of ultrasound in the investigation of ulnar neuropathy.

METHODS: We retrospectively reviewed all patients with EDX-diagnosed ulnar neuropathy, from January 2013 to February 2014, who had also undergone US. The US images were reviewed to determine nerve cross-sectional area (CSA), echogenicity, severity of arthritis, and degree of perineural soft-tissue swelling at the elbow.

RESULTS: We recruited 28 patients. The site of neuropathy for 4 patients could be localized with US but not EDX testing; 1 could be localized with EDX testing but not US, 4 could not be localized with either, and the rest could be localized with both. US could elucidate the etiology in 24 patients: compression neuropathy at the sulcus (10), arthritis at the elbow (9), previous injury/surgery (3), contraceptive implant (1), and leprosy (1). As an evaluation of whether structure (US) could determine function (EDX), our multiple regression models found that CSA is the US parameter that most consistently predicts EDX parameters, e.g., every 0.1 cm² increase in CSA increases motor latency by 1.53 ms and decreases motor amplitude by 3.61 mV, when measured below the elbow with recording electrodes at the abductor digiti minimi. The severity of arthritis and the presence of nerve subluxation also predicts EDX parameters, but less consistently.

SUMMARY/CONCLUSION: Nerve US can be useful for investigating localization, etiology, and nerve function in ulnar neuropathy.

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Resident and Fellow Member Award Recipient

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RESTLESS LEG SYNDROME CAUSED BY SMALL FIBER NEUROPATHY TREATED WITH PENTOXIFYLLINE
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INTRODUCTION: Restless leg syndrome can be caused by a central mechanism or a peripheral neuropathy. In cases of peripheral neuropathy, treatment can be made with antiepileptics and/or antidepressants.

OBJECTIVE: To treat restless leg syndrome and small fiber neuropathy using pentoxifylline.

METHODS: Ten patients were screened for restless leg syndrome. To identify if the symptoms were central or peripheral, needle EMG and NCSs were performed. If the needle EMG was negative for large fiber neuropathy, epidermal nerve fiber density studies were obtained from skin biopsies. In the positive cases, the patients were treated with pentoxifylline, 400 mg 3 times a day.

RESULTS: In all 10 cases, the patients had mild-to-severe small fiber neuropathy from Protein Gene Product (PGP) 9.5 staining and counting per the method of Dr. Lauria and the European Federation of Neurological Societies. The patients were given 400 mg of pentoxifylline, 3 times a day. All 10 patients had 75-100% relief of their symptoms with only mild gastrointestinal side effects.

SUMMARY/CONCLUSION: Restless leg syndrome should be worked up for peripheral nerve disease. If positive for small fiber neuropathy by skin biopsy and PGP 9.5 staining, treatment with pentoxifylline may relieve the symptoms with much fewer side effects then other treatments. It is currently used for increasing circulation in peripheral vascular disease and improving wound healing in diabetic ulcers. By decreasing the size and improving the pliability of the red blood cells, more oxygen gets to the small nerve fibers which causes the fibers to regenerate and cause less pain.
A NOVEL DYSPHAGIA ASSESSMENT AND CLASSIFICATION MODEL TO EVALUATE MECHANISMS OF DYSPHAGIA IN NEUROMUSCULAR DISEASE
Lisa Williams (Santa Cruz, CA), Erik Henricson (Santa Cruz, CA), Rebecca Leonard (Santa Cruz, CA)

INTRODUCTION: Dysphagia in neuromuscular disease (NMD) is prevalent in up to 100% in some disorders and can result in failure to thrive, pneumonia, and death. Regular assessment and grading of severity to inform management in NMD is critical. However, the spectrum of swallowing abnormalities in NMD has not been fully characterized. Clinically relevant assessment tools have not been validated in this varied patient population.

OBJECTIVE: To develop a dysphagia classification system based on quantitative assessment of videofluoroscopic swallow studies (VFSSs).

METHODS: This study involved data abstraction from an NMD clinical database (1990-2013) undergoing VFSS; 19 NMD disease diagnoses were classified into 3 groups: upper motor neuron (UMN) disease, lower motor disease (LMN) disease, and combined UMN/LMN disease. Total pharyngeal transit time, pharyngeal constriction ratio (PCR), esophageal opening, hyoid displacement, and hyoid to larynx approximation data were collected.

RESULTS: The most common diagnoses were ALS, myotonic muscular dystrophy, and Duchenne muscular dystrophy (n=157). The mean (±SD) age of the cohort was 53.5 (21.8) years. Significant differences in PCR existed between groups. Using an ordinary least squares (OLS) regression model, PCR was elevated on average of 0.264 in the LMN relative to UMN when controlling for age, gender, and aspiration status (model r^2=0.38).

SUMMARY/CONCLUSION: LMN NMD is associated with a higher PCR than UMN NMD. A high PCR indicates poor pharyngeal constriction (>0.25) which is associated with dysphagia and aspiration, and may be a clinically-applicable outcome measurement tool used to determine management strategies and efficacy of treatments in NMD subtypes.

PHARYNGEAL-CERVICAL-BRACHIAL VARIANT OF GUILLAIN-BARRÉ SYNDROME WITH UNILATERAL WEAKNESS
Mohammad Abu Rub (Washington, DC), Perry Richardson (Washington, DC)

INTRODUCTION: The pharyngeal-cervical-brachial (PCB) variant of Guillain–Barré syndrome (GBS) is defined by rapid progression of oropharyngeal, neck, and shoulder weakness, with areflexia, sparing the legs. Needle EMG discloses axonal sensorimotor neuropathy affecting the arms. The reported key examination finding is symmetric and bilateral weakness. Patients whose presentations depart from consensus clinical criteria pose diagnostic challenge.

OBJECTIVE: To describe a challenging case of PCB presenting primarily with unilateral weakness.

CASE REPORT: A 53-year-old man with no significant past history, started complaining of left ptosis and diplopia shortly after a trip to East Africa, where he experienced symptoms of an upper respiratory tract infection and was treated for bronchitis. On initial evaluation, he also complained of dysphonia, dysarthria, and increased airway secretions, later progressing to respiratory distress and requiring intubation. Neurological examination was significant for bilateral lateral rectus weakness, right facial palsy, weakness of neck flexion and the proximal left upper extremity, with hyporeflexia of the upper extremities. Workup included a non-remarkable cerebrospinal fluid and a needle EMG 2 weeks after symptom onset showing a mild patchy axonal polyneuropathy affecting the left arm. He responded to plasmapheresis, with gradual improvement in weakness over more than 3 months. A repeat needle EMG at 3 months showed evidence of axonal injury with robust reinnervation of muscles of the proximal left arm.

CONCLUSION: Unilateral ophthalmoplegia and unilateral facial palsies are not uncommon in GBS. However, we found no reported cases of unilateral limb weakness in PCB. Neurologists should consider that the spectrum of variants of GBS may be broader than reported.
NERVE CONDUCTION PATTERNS IN GUILLAIN-BARRÉ SYNDROME ASSOCIATED WITH ZIKA VIRUS INFECTION

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INTRODUCTION: Zika virus (ZIKV) infection has been associated with Guillain-Barré syndrome (GBS), which diagnosis is clinical. However, NCSs provide information about subtypes, prognosis and outcomes

OBJECTIVE: To evaluate NCS patterns in GBS-associated ZIKV.

METHODS: This was a cross-sectional study in which 43 GBS patients (Brighton criteria) with ZIKV infection were included. NCS were evaluated according to new criteria set (AU). Motor conduction studies were evaluated in the median, ulnar, peroneal and tibial motor nerves whereas sensory nerve action potential and conduction velocity were measured in median, sural and ulnar nerves. F wave and H reflex were also registered. Statistical analysis was done in R 3.3.2.

RESULTS: Demyelinating form represented 74.4% of the cases. AMAN and AMSAN forms were present in 6.9% and 2.3% of cases, respectively. The most observed pattern in AIDP were distal motor latency and decreased conduction velocity at left median nerve (p=0.013, p=0.038, respectively). In AMAN patients a decrease of the motor amplitude at right ulnar nerve was the highest (p=0.007). There was a prevalent distal involvement in AIDP patients. A major compromise of median nerve was distinctive (left: 63.6%, right: 71.4%). AGA were present in 6(26%) cases, and were not associated with any specific pattern. Agreement between Hadden’s and Ho’s criteria and Uncini’s criteria was not found (Cohen’s kappa coefficient -0.0024, 95% CI -0.13 -0.13).

SUMMARY/CONCLUSION: AIDP is the most frequent pattern of GBS associated with ZIKV infection. Also, AGA were not associated with any specific pattern in our population, more acute studies are needed. Furthermore, there is a lack of agreement between Hadden’s criteria and new criteria set proposed by Uncini.

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AUTOIMMUNE HEPATITIS-RELATED SENSORY NEURONOPATHY

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INTRODUCTION: Sensory neuronopathies (SNs) are characterized by dorsal root ganglia damage. Clinically, SNs are characterized by non-length dependent sensory deficits and sensory ataxia. Several underlying conditions have been identified, and autoimmune diseases represent important etiologies to be considered. There are isolated reports of SNs associated with autoimmune hepatitis (AIH), but there are no systematic clinical and neurophysiological evaluations of this association in a large cohort.

OBJECTIVE: To evaluate the frequency of SNs among patients with AIH and to describe clinical and neurophysiological aspects of these patients.

METHODS: We evaluated transversally 67 AIH patients followed in a tertiary gastroenterology clinic. The patients were invited to neurological and neurophysiological examination. Descriptive statistics were employed to analyze the results.

RESULTS: Five patients (7%) fulfilled criteria for SN (AIH-SN). The female: male ratios were 47: 15 and 5: 0 for the non-SN and AIH-SN patients. Among the AIH-SN patients, 4/5 (80%) had ultrasonographic and histological diagnosis of cirrhosis and none of them were diabetic. Other neurophysiological diagnoses were radiculopathies (25%), CTS (5%), and polyneuropathy (4%). Sural and median sensory amplitudes for non-SN/AIH-SN patients were 19.5±9.6 μV/0.9±2.1 μV and 45.3±20.6 μV/4.2±4.6 μV with internal reference values of 20 μV and 20 μV, respectively.

SUMMARY/CONCLUSION: SN and AIH are related conditions. However, a mutual pathophysiological mechanism remains to be described and a shared epitope may be a common ground for both conditions.

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AANEM Foundation for Research and Education Award Recipient
63 RETROSPECTIVE REVIEW OF RESPONSE TO IVIG TREATMENT IN CIDP NAÏVE PATIENTS
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INTRODUCTION: Chronic inflammatory demyelinating polyneuropathy (CIDP) responds differently to first-line immunomodulatory therapies, such as IV immunoglobulin (IVIg), plasma exchange, or prednisone. Various clinical factors (CIDP phenotype, EDX results, and disease duration) help prognosticate responses to these.

OBJECTIVE: To investigate clinical factors associated with treatment response to IVIg in CIDP naïve patients.

METHODS: We retrospectively reviewed CIDP patients receiving IVIg from 2010-2016 at Keck Medical Center at USC. Patients met European Federation of Neurological Societies (EFNS)/peripheral nervous system (PNS) EDX criteria for CIDP, and we excluded patients with comorbid acquired neuropathies. We measured response to IVIg with the Inflammatory Neuropathy Cause and Treatment (INCAT) disability scale, and correlated responders and non-responders to various clinical factors. Response was defined as 1-point decrease on INCAT.

RESULTS: Of 18 patients reviewed, 72% were responders and 28% were non-responders. Non-responders showed a mean duration of symptoms to initiation of IVIg therapy of 65 months (33-120 months) versus 21 months (1-84 months) for responders. Of patients with predominant demyelinating pathophysiology, 85% (11/13) responded compared to 40% (2/5) of responders with predominant axonal loss. Typical phenotype and definite CIDP criteria on EDX examination correlated with 72% responders compared to 50% of responders with atypical phenotype and definite criteria. Responders with a greater than 1-point decrease on INCAT showed a mean duration of symptoms to treatment of 38 months versus 73 months in CIDP responders with decrease in INCAT by 1 point.

SUMMARY/CONCLUSION: Early diagnosis and treatment of CIDP impacts response in IVIg naïve patients and correlates with greater degree of functional recovery. Also, typical phenotypes, predominately demyelinating pathophysiology and definite criteria on EDX studies, favored response to IVIg.

64 ORTHODROMIC GREATER AURICULAR NERVE STIMULATION
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INTRODUCTION: The greater auricular nerve (GAN) is a superficial sensory branch of the C2/3 cervical plexus that innervates the external ear and posterior auricular region. Lesions of the GAN are most commonly seen iatrogenically in the setting of parotidectomy. The suggested mechanism of red ear syndrome involves peripheral C3 nerve root or branch nerve lesions. Lesions present clinically as either sensory loss in the region of the GAN or dysesthesia. These lesions have not yet been well demonstrated, in part due to the diminutive size of the nerves precluding imaging and the lack of reported NCS techniques. Antidromic NCSs have been demonstrated in 1 study. To our knowledge, an orthodromic technique has not been demonstrated previously.

OBJECTIVE: To demonstrate a technique for the orthodromic stimulation of the GAN.

METHODS: The location of the course of the GAN was identified in reference materials. Utilizing a Natus Viking® on a Nicolet® VikingQuest™ Machine (Middleton, Wisconsin), standard 3 mm recording and reference electrodes were placed at the posterior border of the sternocleidomastoid at C3. A stimulator was applied to the external helix of the ear with a cathode placed at the level of the concha. Stimulation at low levels (0.1 ms and 60 mA) resulted in recordable waveforms.

RESULTS: Successful stimulation of the GAN was achieved.

CONCLUSIONS: In normal control subjects, stimulation of the GAN using an orthodromic technique can be performed. Further work to replicate this stimulation within clinical populations, including patients with red ear syndrome, is required.
IMAGING MUSCLE CONTRACTION WITH ELECTRICAL IMPEDANCE
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INTRODUCTION: A technology that provides quantitative data on alterations in muscle properties during contraction, including stresses and deformation, currently does not exist. One approach for doing so is via the application of electrical impedance (EII) techniques.

OBJECTIVE: To utilize EII, in which an array of electrodes is placed over the muscle and impedance data acquired across multiple electrode combinations in rapid succession.

METHODS: A 32-channel tetrapolar-based EII system was used for impedance acquisition. Specifications include a 10 MHz bandwidth, about 100 dB signal-to-noise ratio, about 100/second frame rate, and continuously selectable frequencies. A printed circuit board-based array consisting of 20 electrodes was affixed to the skin overlaying the biceps with a Velcro band. EII was performed on 3 healthy individuals. Imaging was performed first at rest and then with progressively greater weights (5, 10, and 15 lb), keeping the arm in an identical posture throughout. Difference images were generated by subtracting at-rest values from contraction values.

RESULTS: Areas of increased and decreased conductivity were readily observed during the contraction. These areas intensified with increasing weight, consistent with greater deformation of the muscle fibers. The changes were consistent across individuals.

SUMMARY/CONCLUSION: Changes in the impedance of muscle are readily observable during a fixed isometric contraction. The mechanisms underlying these changes require further study. However, they are likely related to muscle fiber deformation and possibly depolarization and not simply due to increased blood flow since areas of both increased and decreased conductivity were observed. Further study of this innovative muscle imaging technique in healthy and diseased individuals is planned.

PREDICTIVE MODEL TO CALCULATE THE NORMATIVES VALUES IN NCSs OF THE UPPER ARM FOR ADULTS, IN CALI COLOMBIA
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INTRODUCTION: Normal values of NCSs are usually statistical measures obtained from the mean, SD, and percentiles. However, these approaches underestimate the influence of personal individual characteristics.

OBJECTIVE: To establish the limits of NCSs and their relationship with age, body mass index (BMI), sex, height, and temperature.

METHODS: Two hundred twenty-two healthy adults between 18 and 82 years of age were included in this study. All standard quality recommendations were incorporated in the design. Bootstrap technique was used to identify relevant associations between NCS parameters with age, sex, height, BMI, and temperature. The effect of individual characteristics on sensory and motor NCSs of median, ulnar, and radial nerve parameters was estimated with robust multivariate linear regression.

RESULTS: Reference values of sensory nerve action potentials for median, radial and ulnar nerves were adjusted according to relevant characteristics; our values for latency, amplitude, and conduction velocity for these nerves were 3.48 ms, 19.93 μV, 38.69 m/s; 2.86 ms, 11.88 μV, 41.27 m/s; and 3.47 ms, 19.58 μV, 39.67 m/s, respectively. Adjusted values of compound muscle action potentials for latency, amplitude, and conduction velocity of the median and ulnar nerves were 3.99 ms, 4.68 mV, 48.05 m/s and 3.35 ms, 4.26 mV and 50.41 m/s, respectively. Significant influence of age and temperature was found in most NCS parameters. Negative correlation was observed between height and velocity parameters.

SUMMARY/CONCLUSION: Adjusted reference values according to patient characteristics may represent an alternative to improve the method of diagnosis upper limb pathologies.
NORMATIVE VALUES OF NERVE CONDUCTION OF MEDIAN, ULNAR, RADIAL OF UPPER ARM IN A COLOMBIAN POPULATION

INTRODUCTION: Normative NCSs require quality.

OBJECTIVE: To generate reference values for sensory and motor NCS parameters of the upper arm among a sample of Colombian adults and identify factors that account for variations in the population.

METHODS: Occupational, clinical test, and NCSs were conducted among a sample of 222 healthy adults (18-82 years old), of whom 50% were older than 40. Age, sex, height, temperature, and body mass index variations in the estimates were included in the models. American Association of Neuromuscular and Electrodiagnostic Medicine quality recommendations were applied. Normative values for latency, amplitude, and velocity were calculated for sensory nerve action potentials (SNAPs) for the ulnar, median, and radial nerves and for compound motor action potentials (CMAPs) for the ulnar and median nerves. Estimates in the Colombian sample were compared to estimates in the United States.

RESULTS: The normative values for this population were determined as follows: median nerve SNAP latency (L): 3.50 ms, amplitude (A): 18.66 µV, and velocity (V): 40 m/s; median nerve CMAP L: 3.90 ms, A: 4.60 mV, and V: 49.00 m/s; ulnar nerve SNAP L: 3.50 ms, A: 10.37 µV, and V: 39.00 m/s; ulnar nerve CMAP L: 3.48 ms, A: 3.82 mV, and V: 50.82 m/s; and radial nerve SNAP L: 2.90 ms, A: 11.3 µV, and V: 50.8 m/s. Significant differences were found in the estimates across age groups.

SUMMARY/CONCLUSION: Compared to estimates from the United States, this Colombian sample showed lower normative values for sensory and motor nerve conduction in the ulnar and median nerves. These differences might be attributed to local temperature and height.

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BILATERAL IDIOPATHIC THENAR AND FIRST DORSAL INTEROSSEOUS MUSCLE HYPERTROPHY ASSOCIATED WITH CARPAL TUNNEL SYNDROME
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INTRODUCTION: Bilateral hypertrophy of thenar and first dorsal interosseous (FDI) muscles is a rare condition that has not been previously reported with CTS.

OBJECTIVE: To report a rare syndrome of bilateral hypertrophy of thenar and FDI muscles associated with CTS.

CASE REPORT: A 43-year-old male utility technician presented with a 2-year history of worsening bilateral thenar and FDI muscle hypertrophy associated with rapid fatigue and impaired grip. Physical examination showed symmetric bilateral thenar and FDI muscle hypertrophy and no other findings. Initial EDX studies did not reveal a definite abnormality. A hand MRI demonstrated a normal median nerve and enlarged but otherwise normal appearing thenar and FDI muscles. The creatine kinase (CK) level was normal and genetic testing for muscular dystrophies was negative. Over several months, he developed paresthesias in the median distribution bilaterally, and repeat EDX studies revealed bilateral median neuropathies at the wrist consistent with bilateral mild CTS. The patient underwent bilateral carpal tunnel release and a thenar muscle biopsy. Postoperatively, the hand paresthesias resolved. The muscle hypertrophy remained unchanged. The biopsy demonstrated mild denervation atrophy with associated reinnervation and no evidence of myopathy. Literature review revealed 3 patients with bilateral thenar and FDI muscle hypertrophy but without CTS. There was also 1 patient with unilateral thenar hypertrophy associated with CTS due to lipofibromatous hamartoma.

SUMMARY: This case is the first known patient with bilateral hypertrophy of the thenar and FDI muscles associated with CTS. The cause of the muscle hypertrophy remains undetermined.

SOMATOSENSORY EVOKED POTENTIALS IN VITAMIN B12 DEFICIENCY
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INTRODUCTION: It is usually believed that vitamin B12 deficiency mainly affects the dorsal column. In this study, we investigated which part of the sensory pathway is affected in vitamin B12 deficiency using NCSs and somatosensory evoked potentials (SEPs).

OBJECTIVE/METHODS: We retrospectively reviewed the EMG database of Teikyo University and Mitsui Memorial Hospital from 2008 to 2016, and enrolled patients (n=17) who presented with sensory symptoms and/or gait disturbance and whose serum B12 level was <170 ng/ml.

RESULTS: Spinal MRI demonstrated abnormalities in 4 out of 17 patients. NCS abnormalities were found in 3 out of 15 patients for the sural nerve, and in 5 out of 12 for the median nerve. When distal NCSs and peripheral segments of the SEPs were combined, conduction delay was detected in 12 out of 15 patients for the lower limbs and in 8 out of 13 for the upper limbs. Delay of central conduction time was rarer, found in 3 out of 15 patients in tibial SEPs. The N9 onset-to-P13/14 onset segment in median SEPs was abnormal in 4 out of 8 patients, although this is a mixture of peripheral and central conduction.

SUMMARY/CONCLUSION: Patients with vitamin B12 deficiency exhibited abnormalities of peripheral nerve conduction more frequently than those of central conduction. SEPs are useful in localizing the lesion along the sensory pathway, not only for central conduction time but for peripheral conduction time.
CLINICAL SPECTRUM OF NEUROPATHY AFTER TOTAL KNEE ARTHROPLASTY: A RETROSPECTIVE REVIEW OF 471 CASES
Scott Speelziek (Rochester, MN), Ruple Laughlin (Rochester, MN), Nathan Staff (Rochester, MN)

INTRODUCTION/OBJECTIVE: With an approximate 700,000 total knee arthroplasties (TKAs) performed annually in the United States, and a projected 3.48 million by 2030, there is a sizeable population who may be susceptible to a postoperative lower extremity neuropathy. The aim of this comprehensive review is to gain a better understanding of neuropathy after TKA including risk factors, etiologies, and clinical course.

METHODS: We identified patients who underwent TKA at Mayo Clinic Rochester between 1996 and September 2016 (14,450) and had a possible neuropathy ascertained via international classification of diseases (ICD) 9/10 diagnosis codes chosen to comprehensively capture the spectrum of lower extremity neuropathies (lumbosacral, femoral, sciatic, peroneal, tibial, etc.). A retrospective review of 471 identified cases was performed.

RESULTS: Of 350/471 cases reviewed to date, we have identified 43 new cases of neuropathy meeting inclusion/exclusion criteria: peroneal (25), sciatic (11), ulnar (1), tibial (1), sural (1), and lumbosacral plexopathy (1). Of particular interest is the 1 case of postsurgical inflammatory lumbosacral plexopathy identified. Needle EMG was used to aid diagnosis in 20/43 cases, with findings typically supporting an axonal process including reduced/absent compound muscle action potentials/sensory nerve action potential, fibrillations, and long duration motor unit potentials. Neurological outcome data are currently being investigated in this cohort.

SUMMARY/CONCLUSION: Neuropathy following TKA most commonly affects the peripheral nerves that are most at risk anatomically (peroneal, distal sciatic) during the surgical procedure; however, an inflammatory etiology should remain a consideration, which then may require a more aggressive treatment strategy.

Scott Speelziek, MD
Resident and Fellow Member Award Recipient

PREDICTION OF QUANTITATIVE SUDOMOTOR AXON REFLEX TESTING VALUES FOR DORSAL HAND
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INTRODUCTION: Quantitative sudomotor axon reflex testing (QSART) is useful in detection of various neuropathies related to diabetes and idiopathic small-fiber neuropathy. Our autonomic laboratory measures distal upper extremity sweat production on the dorsum of the hand, as most cases of small fiber neuropathy are worse distally. However, the density of sweat glands on the dorsum of the hand is almost 3 times more than other standard testing locations which makes the comparison challenging.

OBJECTIVE: To predict the amount of sweat production on the dorsum of the hand based on sweat production on the foot, proximal lateral leg, and forearm.

METHODS: This was a retrospective cross-sectional study of all adult patients referred to our autonomic laboratory of between 2015 and 2017 whose screening autonomic testing (QSART, heart rate variability to deep breathing, Valsalva maneuver, and tilt table test) was normal with a Composite Autonomic Severity Score of 10. We excluded subjects who use medications with a known effect on sweat production, and those with paresthesia, peripheral neuropathy, diabetes, and orthostatic hypotension. Multivariate linear regression modeling was used.

RESULTS: The amount of sweat production on dorsum of the hand is significantly higher than that of the other 3 standard testing locations. After gender, the strongest predictor of sweat production on hand is the amount of sweat production on the proximal lateral leg.

CONCLUSION: The amount of sweat production on the dorsum of the hand can be predicted with a multivariate linear regression model using gender and the sweat production on the 3 standard locations of the foot, proximal lateral leg, and forearm.
INTRODUCTION: Determining the underlying etiology of ataxia can be challenging, as hereditary ataxias are clinically and genetically heterogeneous. Whole exome sequencing (WES) does not identify repeat expansions but provides a broad approach for screening thousands of genes for pathogenic sequence variants.

OBJECTIVE: To establish the positive diagnostic rate (PDR) of WES in patients with ataxia and recognize factors contributing to the PDR.

METHODS: We reviewed the PDR of WES for specific clinical features and/or age in 1081 patients with ataxia referred to our laboratory.

RESULTS: WES yielded a 32.5% positive rate (351/1081 cases), defined as 1 or 2 pathogenic or likely pathogenic variants in a single gene, depending on mode of inheritance. Seventy-four percent of probands were younger than 20 years; PDR for less than 20 years was 35.6%. Conversely, probands older than 50 years had a lower PDR (14.2%). WES-trio (proband and parents sequenced concurrently) significantly improved outcome, increasing the PDR from 23.9% (WES-proband only) to 37.5% (p<0.01). PDR was influenced by specific accompanying clinical features: structural cerebellar abnormalities (40.3%), white matter disease (38.2%), seizures (34.3%), and extrapyramidal signs (32.1%). Pathogenic variants were reported in 179 distinct genes; 11/179 genes were newly associated with ataxia in recent years.

SUMMARY/CONCLUSION: WES has a diagnostic yield of >30% for patients with ataxia and should be included in the clinical workup. WES-trio testing is more effective than WES-proband only, yielding a higher PDR in children and adolescents. Individuals with complex presentations benefit most from WES. Our results and the growing number of newly described ataxia genes underscore the need for broad and flexible molecular diagnostic testing for ataxias.
**CONSTITUENT MUSCLE CONTRIBUTION TO THENAR COMPOUND MUSCLE ACTION POTENTIAL**

Darine Kassar (El Paso, TX), Mohtashim Qureshi (El Paso, TX), Mohammad Afzal (El Paso, TX), Anantha Vellipuram (El Paso, TX), Ihtesham Qureshi (El Paso, TX), Gustavo Rodriguez (El Paso, TX), Paisith Piriyawat (El Paso, TX), Salvador Cruz-Flores (El Paso, TX)

**BACKGROUND:** Mechanical ventilation (MV) in ALS allows prolongation of life and improves the quality of life.

**OBJECTIVE:** To study the impact of MV in ALS on in-hospital complications, life sustaining procedures, length of stay, and discharge disposition.

**METHODS:** ALS patients from the Nationwide Inpatient Sample database for 2011-2014 were identified using code 335.20 from the International Classification of Diseases, 9th edition. We compared ALS patients with and without MV by age, gender, race, medical comorbidities, All Patient Refined Diagnosis Related Groups (APR-DRG) severity, insurance status, median household income for patient's zip code, in-hospital complications and procedures, length of stay (LOS), total hospital charges, and mortality. Prolonged length of stay (PLOS) was defined as LOS ≥5 days. ALS patients without MV served as the reference in multivariate analysis.

**RESULTS:** Of 45,860 hospitalized patients with ALS, 7976 (17.4%) had MV. ALS with MV had a significantly higher rate of in-hospital complications (myocardial infarction, sepsis, pneumonia, deep vein thrombosis, pulmonary embolism, and urinary tract infection), in-hospital procedures (MV, gastrostomy, transfusions, and tracheostomy), LOS, total hospital charges, mortality. Mortality was no different between the groups.

**CONCLUSION:** ALS patients requiring MV have higher in-hospital complications, in-hospital procedures, LOS, and moderate-to-severe disability. Mortality was no different between the groups.

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**RACIAL AND ETHNIC DIFFERENCES IN HOSPITAL UTILIZATION, LIFE SAVING PROCEDURES AND PALLIATIVE CARE AMONG PATIENTS WITH ALS**

Darine Kassar (El Paso, TX), Mohammad Afzal (El Paso, TX), Mohtashim Qureshi (El Paso, TX), Anantha Vellipuram (El Paso, TX), Ihtesham Qureshi (El Paso, TX), Gustavo Rodriguez (El Paso, TX), Paisith Piriyawat (El Paso, TX), Salvador Cruz-Flores (El Paso, TX)

**BACKGROUND:** Racial/ethnic disparities exist in health care.

**OBJECTIVE:** To establish if race/ethnicity impacts hospital utilization, defined as length of stay (LOS), and lifesaving procedures, palliative care, and discharge disposition among patients with ALS in the United States.

**METHODS:** ALS patients from the Nationwide Inpatient Sample database for years 2011-2014 were identified using code 335.20 from the International Classification of Diseases, 9th edition. We compared 4 race categories: white, black, hispanic, and other (i.e., Asian or Pacific Islander, Native American, etc.) by age, gender, race, medical comorbidities, in-hospital complications and procedures, LOS, total hospital charges, median household income for patient's zip code, insurance status, in-hospital mortality, palliative care (PC), and do not resuscitate (DNR). The white race category served as the reference group.

**RESULTS:** Of 43,123 patients with ALS, 32,274 (74.8%), 4,668 (10.8%), 3,539 (8.3%), and 2,642 (6.1%) were White, Black, Hispanic, and Others, respectively. Either or all minorities had a significantly higher rate of in-hospital complications (sepsis, pneumonia, pulmonary embolism, and urinary tract infection), in-hospital procedures (mechanical ventilation and transfusions) compared to Whites. Mean LOS and hospital charges were also higher for minorities compared with Whites. Whites had a higher rate of in-hospital mortality, palliative care, and DNR as compared to minorities.

**CONCLUSION:** Minorities have a greater utilization of life sustaining procedures and longer LOS while Whites have greater utilization of palliative care/hospice and a higher in-hospital mortality. The reasons for these differences are unclear but may reflect cultural differences and not problems with access to care.
REDUCTION OF OPIOID DOSE WITH THE USE OF SUBCUTANEOUS BOTULINUM TOXIN TYPE A IN PATIENTS WITH PAINFUL DISTAL SYMMETRIC POLYNEUROPATHY-A PILOT STUDY  
Shalvinder Seehra (Columbia, MO), Raghav Govindarajan (Columbia, MO)

BACKGROUND: Chronic neuropathic pain is a disabling condition. It is common clinical practice to use opioids in managing severe breakthrough pain thus putting patients at risk of developing dependence/addiction. There is an urgent unmet need for finding alternatives given the current opioid epidemic.

OBJECTIVE: To assess the efficacy of 3 monthly subcutaneous botulinum toxin injections (administered in clearly defined areas of allodynia in the lower limb) in reducing the dose of opioids.

METHODS: This is a retrospective chart review of 5 patients. A visual analog pain scale (VAS) was used to measure pain score pre- and post-injections. Opioid dose changes were left to the discretion of the treating physician. All patients were on a stable dose of pain modulating drugs.

RESULTS: Included were 5 patients (average age: 62 years; range: 55-68 years; 3 men, 2 women; all Caucasian): 3 with idiopathic neuropathy and 2 with neuropathy associated with TS-HDS antibodies. Two patients were on 1200 mg three times a day of gabapentin and 3 were on 100 mg of amitriptyline at bedtime in addition to 75 mg of topiramate. The average VAS pre-injection was 8. Patients were on an average of 6 oral morphine dose equivalents (range: 4-12). After 2 injections (in 2 patients) and after 3 injections (in 3 patients), the VAS was reduced to 5 (range: 4-6, p<0.05) and the morphine equivalent dose was reduced to 3 (range: 4-6, p<0.05).

CONCLUSION: Three monthly botulinum toxin type A injections (after 2-3 sessions) can reduce the need of opioid dose by average 50-60% in patients with chronic neuropathic pain and well-defined allodynia.

Shalvinder Seehra, BS  
Resident and Fellow Member Award Recipient

DOES THE USE OF TOPICAL ANESTHETIC REDUCE THE PAIN OF EMG?  
Klaudia Kukulka (Chicago, IL), Raghav Govindarajan (Columbia, MO)

BACKGROUND: Pain and discomfort during needle EMG are common and sometimes can lead to premature termination of needle EMG thus resulting in an incomplete interpretation.

OBJECTIVE: To assess if use of topical lidocaine HCl 4% spray (1-4 ml) reduces the pain of needle EMG.

METHODS: This is a retrospective chart review of 40 patients (20 had received topical anesthetic and 20 had not). Pregnant patients were excluded from the study. All patients had needle EMG performed by a single board certified neuromuscular physician. Clinical and demographic data were extracted from the chart. All patients were given a post EMG visual analog pain scale (VAS) and the scores between the 2 groups were compared.

RESULTS: The average age of the patients was 45 years (range: 32-78 years); 35 Caucasians and 5 African Americans (25 men, 15 women) were included. Lumbosacral radiculopathy was the indication in 15, peripheral neuropathy in 10, carpal tunnel/cubital tunnel syndrome in 10, and cervical radiculopathy in 5. The VAS was 4.5 (range: 2-8) in patients with no anesthetic, and the VAS was 4 (range: 2-8) in patients with anesthetic (p=0.3). Patients with fibromyalgia had a median VAS of 6 (range: 6-8, p<0.05). Patients with allodynia had a trend towards reduced post-needle EMG pain with topical anesthetic (VAS 3.5, p=0.48). Age, sex, race, indication, prior experience with needle EMG, and muscles examined did not show any statistical difference.

CONCLUSION: In this pilot study, the use of topical spray did not alter post-needle EMG pain perception. Fibromyalgia was associated with higher pain, and there was a trend toward lower pain in patients with allodynia.

Klaudia Kukulka, BS  
Resident and Fellow Member Award Recipient
EFFICACY OF ULTRASOUND-GUIDED CORTICOSTEROID INJECTIONS IN THE TREATMENT OF ULNAR NEUROPATHY AT THE ELBOW
John Norbury (Greenville, NC), Carrie McShane (Greenville, NC), Jason Lee (Greenville, NC), Abigail DeFarrell (Greenville, NC)

INTRODUCTION: The efficacy of ultrasound-guided corticosteroid injections in the treatment of ulnar neuropathy at the elbow (UNE) has limited support in the literature.

OBJECTIVE: To describe the effect of a single ultrasound-guided corticosteroid injection on patients with clinical UNE and what factors might predict a good response.

METHODS: Patients with clinical and/or EDX evidence of UNE received a diagnostic neuromuscular ultrasound measuring the cross-sectional area (CSA) of the ulnar nerve at 3 locations about the elbow. They then received a single ultrasound-guided injection of 1 cc of 6 mg betamethasone and 1 cc of 1% lidocaine at the location of maximum nerve swelling. Followup phone interviews were conducted at 2 and 12 weeks.

RESULTS: Ten patients were enrolled in the study; the average age was 56.5 years. The completion rates for the 2 week and 12 week phone interviews were 10/10 and 9/10, respectively. Six out of 10 indicated the injection improved their symptoms. Only patients with at least 1 nerve CSA above 10 mm2 responded favorably to the injection.

SUMMARY/CONCLUSION: Corticosteroid injections around the ulnar nerve may be efficacious in a subset of patients with UNE. An increased nerve CSA may be positively correlated with a favorable outcome following the injection.

Shelby Herr, BS
Resident and Fellow Member Award Recipient
A CRITICAL NEED FOR PHYSICAL AND OCCUPATIONAL THERAPY IN PATIENTS WITH CHARCOT MARIE TOOTH PERIPHERAL NEUROPATHY: PRELIMINARY RESULTS OF AN ONLINE SURVEY.
James Nussbaum (New York, NY), Allison Moore (New York, NY)

INTRODUCTION: Charcot–Marie–Tooth (CMT) disease is characterized by distal motor and sensory dysfunction, often limiting independent mobility and quality of life (QOL). There is a paucity of literature on patient-reported symptoms and their impact on function and QOL.

OBJECTIVE: To summarize the preliminary results of an ongoing (through June 2017) survey by the Hereditary Neuropathy Foundation, and to identify and justify the critical need for physical (PT) and occupational therapy (OT) in CMT patients.

METHODS: A survey to demonstrate the impact and prevalence of CMT symptoms was implemented via an online, anonymous platform.

RESULTS: Of 1747 respondents, 88.9% report having CMT, the remainder being family members/caretakers. Almost every respondent reported many near falls per month, and on average, reported >2 falls to the ground/month. Respondents reported problems with walking long distances (88.1%), climbing up/down stairs (83.0%), maintaining balance (91.9%), getting up from a chair/ground (67.2%), standing for long periods (82.0%), and trips/falls (79.2%). Only 26.2% reported not using any orthosis or assistive device. Despite overwhelming reports of (hand 82.4%, ankle 87.1%) weakness, imbalance (91.9%), fatigue (78.8%) and falls, only 5.2% reported receiving OT, and less than 20% PT.

SUMMARY/CONCLUSION: The overwhelming majority of CMT patients report numerous symptoms which limit independent mobility and QOL. In most diseases, PT/OT therapists can objectively assess, document, and skillfully treat deficits in functional strength, mobility, and balance. Yet, CMT patients report not receiving routine PT/OT. It may be beneficial for CMT patients to be evaluated by PT/OT to establish baselines, and to participate in skilled PT/OT treatment to address impairments limiting function, mobility, and QOL.

SMALL FIBER NEUROPATHY IN PAIN CLINIC
Francis Lagattuta (Santa Maria, CA), Sheng Zhong (Santa Maria, CA)

INTRODUCTION/OBJECTIVE: This retrospective review involved patients in an outpatient pain clinic with positive screening for small fiber neuropathy.

RESULTS: Six hundred thirty-three patients were given a 5-question screening tool and were identified as at risk for having small fiber neuropathy. A total of 258 (40.7%) patients had abnormal epidermal nerve fiber density (ENFD) findings consistent with small fiber neuropathy. Of these, the average body mass index (BMI) was 32.2. There were 94 (36.4%) patients with a BMI under 25, and 164 (63.5%) patients with a BMI greater than 24.9. A total of 50 (19%) patients with an abnormal ENFD had been previously diagnosed with at least 1 chronic disease (e.g., diabetes, prediabetes, hypertension, hepatitis C, thyroid condition). Of these, 45 (90%) had diabetes and 16 (12%) had hypertension.

CONCLUSION: Nerves are very sensitive, and injury to the nerves is usually caused by microvascular insults. The nerve damage can be caused by anything that impairs the microcirculation. This includes diabetes, hypertension, lipid dysfunction, and many other factors that need to be explored. Early microvascular neuropathy (EMN) is the earliest nerve injury in diabetes and may be the cause of chronic pain in over 40% of patients with burning and diffuse pain. Screening for early microvascular neuropathy with ENFD is necessary, and treatment may prevent permanent large fiber neuropathy and other diseases of vascular injury such as retinopathy, renal disease, and autonomic neuropathy.
SUCCESSFUL COMMUNICATION USING BRAIN-EYE INTERFACE MESSENGER APPLICATION IN PATIENTS WITH ALS: A CASE REPORT
Won Ah Choi (Seoul, KP), Seong-Woong Kang (Seoul, KP), Mi Ri Suh (Seoul, KP)

INTRODUCTION: Patients with ALS usually face progressive bulbar dysfunction and undergo tracheostomy, leaving only eye-gaze function for communication.

OBJECTIVE: To describe the effect of a newly developed brain–eye interface messenger application as a new communication tool for patients with non-verbal ALS.

CASE REPORT: A newly developed brain–eye messenger application using the eye gaze was tested in February 2017 in 2 patients with ALS. Correct character per minute (CCPM) for 18- and 21-character sentences was compared with a former communication tool of each patient. ALS quality of life questionnaire (K-ALSQOL) was taken before and after using the application. A 49-year-old man was diagnosed with ALS in 2012. He underwent tracheostomy in 2016, but was nonverbal since 2015 due to severe bulbar dysfunction and has been using the letter board. For 18- and 21-character sentences, his CCPM was 10.2 and 9.3 using the letter board and 27.6 and 22.4 using the application. His K-ALSQOL improved from 258 to 329 points after using the new application.

A 60-year-old woman was diagnosed with ALS in 2012. She underwent tracheostomy in 2013, and she had difficulty in communication using her mouth shape. She failed the sentence test using her former communication tool. However, she scored 8.0 and 9.9 in CCPM using the application for 18- and 21-character sentences. Her K-ALSQOL improved from 228 to 236 points after using the new application.

SUMMARY/CONCLUSION: A new brain–eye interface application can promote communication efficiency in patients with ALS, and thus alleviate their quality of life.

IMPROVING EMG PROCEDURE ACCESS BY ANSWERING QUESTION VIA ELECTRONIC CONSULT (E-CONSULT): DANGEROUS OR COST EFFECTIVE?
Jasvinder Chawla (Darien, IL), Rishi Garg (Hines, IL), Susan Vaughan (Hines, IL)

INTRODUCTION: Timely access to care, including needle EMG, has been a priority for Veterans Health Administration (VHA). To improve access, VHA implemented electronic consults (e-consults) in 2011 as an alternative to in-person visits. Given the increased wait times for a needle EMG, our study was designed to evaluate whether the need for a needle EMG could be addressed via e-consult in lieu of the procedure.

METHODS: Of the needle EMG requests sent to Neurology from August to December 2016, 33/315 consults were answered via e-consults, which were based upon chart review by a neurologist. Three of these 33 consults were recommended for autonomic testing. Approximately 3-6 months following the e-consult recommendation, another chart review was performed to assess clinical status.

RESULTS: We found that 30/33 patients improved when offered conservative measures such as wrist splints for CTS, elbow pads for ulnar neuropathy, or heating pads for the neck or back pain. Two of 33 patients worsened, 1 with lumbar pain and 1 with pain in the little finger. Needle EMGs performed on these 2 patients did not reveal any significant findings. Two separate patients expressed their dissatisfaction; however, they improved with reassurance.

SUMMARY/CONCLUSION: As demonstrated by our study, e-consults may be a useful tool to improve veteran access to needle EMG and can both be safe and cost effective with appropriate screening. Unlike other studies, ours is the first in which needle EMG requests were converted to e-consults and addressed accordingly. Without compromising patient care, practicing cost-effective medicine is an achievable goal. We plan to do a cohort study to better assess outcomes.
2 CASES OF NEW PEDIATRIC SMA3 DIAGNOSES IN PATIENTS THAT HAD PREVIOUSLY BEEN DIAGNOSED WITH OTHER NEUROMUSCULAR DISORDERS
Irma Reyes (Los Angeles, CA), Leighmaria Ramosplatt (South Pasadena, CA)

INTRODUCTION: Chromosome 5 spinal muscular atrophy (SMA) is among the most common disorders seen in pediatric neuromuscular centers, generally types 1 and 2. There is a wide spectrum of SMA clinical presentations, especially types 3 and 4.

OBJECTIVE: To describe the clinical presentation of 2 patients eventually diagnosed with SMA type 3 who were previously diagnosed with other neuromuscular disorders.

CASE REPORT: Patient 1 is a 16-year-old boy who presented with a 4-year history of gradual weakness, more prominent proximally. He had a Gower sign, significant scapular winging, and elevated creatine kinase (800-1500 U/L). His deep tendon reflexes (DTRs) were preserved. Neuromuscular panels from 2 companies showed a heterozygous variant of uncertain significance (VUS) in DNAJB6, a gene associated with limb-girdle muscular dystrophy 1E/D. He was later found to have tongue fasciculations. Needle EMG demonstrated mildly reduced compound muscle action potential (CMAP) amplitude with neurogenic motor unit action potentials. He had 0 copies of SMN1 and 3 of SMN2.

Patient 2 is a 4-year-old girl who had been borderline to attain gross motor skills. On examination, she had proximal and distal weakness. Her DTRs were 1/4 in the lower and 2/4 in the upper extremities. Needle EMG/NCSs demonstrated borderline CMAP amplitude with neurogenic changes on needle examination. A Charcot–Marie–Tooth (CMT) gene panel demonstrated a heterozygous VUS in PRX. She was diagnosed with an axonal CMT. Tongue fasciculations were eventually noted. She had 0 copies of SMN1 and 3 of SMN2.

SUMMARY/CONCLUSION: Ambulatory pediatric patients with non-genetically-confirmed neuromuscular disorders should be evaluated for SMA type 3.
RESULTS OF NORTH STAR AMBULATORY ASSESSMENTS IN THE PHASE 3 ATALUREN CONFIRMATORY TRIAL IN PATIENTS WITH NONSENSE MUTATION DUCHENNE MUSCULAR DYSTROPHY
Niren Shah (South Plainfield, NJ), Francesco Muntoni (London, EN)

INTRODUCTION: Ataluren treats the underlying cause of nonsense mutation Duchenne muscular dystrophy (nmDMD) by promoting readthrough of premature stop codons to produce full-length functional dystrophin.

OBJECTIVE: Ataluren Confirmatory Trial (ACT) DMD was a phase 3, randomized, double-blind, placebo-controlled trial of ataluren that enrolled males aged 7-16 years with nmDMD and baseline 6-minute walk distance (6MWD) ≥150 m and ≤80% predicted. Eligible patients were randomized 1:1 to receive ataluren or placebo orally 3 times daily for 48 weeks.

METHODS: The North Star Ambulatory Assessment (NSAA) is a validated functional scale to measure disease progression specifically in ambulant boys with DMD. It consists of 17 activities ranging from standing from a chair to jumping. Each activity is scored as 0, 1, or 2; the sum of these scores forms the total score, which is linearized to a 0 (worst) to 100 (best) score.

RESULTS: The intent-to-treat population of ACT DMD consisted of 228 patients (ataluren, n=114; placebo, n=114). Overall, patients who received ataluren gained a 1.5-point advantage in the NSAA observed score compared with patients who received placebo (mean NSAA scores, ataluren: −7.0; placebo: −8.5; p=0.270). In addition, fewer patients who received ataluren shifted from a score of 1 or 2 (able to perform function) to 0 (unable to perform function) across all 17 activities compared with those who received placebo. Between-group differences across each of the activities consistently favored ataluren, ranging from 1% (lift head) to 11% (jump) to 12% (rise from chair).

SUMMARY/CONCLUSION: In conclusion, ataluren is the first drug to demonstrate a benefit compared with placebo as assessed by NSAA scores in nmDMD.

AMBULATORY OUTCOME MEASURES AFTER THE INITIAL 48-WEEK PERIOD OF A LONG-TERM, OPEN-LABEL TREATMENT WITH ATALUREN IN PATIENTS PREVIOUSLY ENROLLED IN ATALUREN CLINICAL TRIALS FOR NONSENSE MUTATION DUCHENNE
Niren Shah (South Plainfield, NJ)

INTRODUCTION: Ataluren treats the underlying cause of nonsense mutation Duchenne muscular dystrophy (nmDMD) by promoting readthrough of a premature stop codon to produce full-length functional dystrophin.

OBJECTIVE: In this ongoing, long-term open-label access trial in Europe, Israel, Australia, and Canada, physical function was assessed in patients with nmDMD receiving ataluren (NCT01557400).

METHODS: Males with nmDMD who had received ataluren in previous trials were enrolled; the gap between last prior exposure and entry to this trial ranged 801-1334 days. Patients are to receive ataluren for up to 240 weeks. Results from the first 48 weeks for ambulant patients (able to run/walk 10 minutes in ≤30 seconds upon study entry) are reported. Study assessments were performed at baseline and every 12 weeks during treatment. Change from baseline in 6-minute walk distance (6MWD), timed function tests, and North Star Ambulatory Assessment (NSAA) total score were summarized.

RESULTS: Overall, 94 patients were enrolled, 50 (53.2%) of whom were ambulant at study entry. At baseline, mean (SD) age was 12.1 (2.08) years, body mass index was 22.8 (4.63) kg/m², and 6MWD was 341.6 (108.11) m. Corticosteroids were used by 47 (94.0%) ambulant patients. Mean (SD) change from baseline in 6MWD, time to stand from supine, time to run/walk 10 m, and NSAA score are reported. One patient lost ambulation during these 48 weeks of treatment.

SUMMARY/CONCLUSION: The results suggest a slowing of disease progression in patients receiving ataluren for the first 48 weeks of treatment, compared to natural history, in older and severe patients.
IMPORTANCE OF WALKING-SPEED ASSESSMENT AS AN INDICATOR OF FUNCTIONAL IMPROVEMENT IN ADULTS WITH SPASTIC HEMIPARESIS AFTER REPEATED ADMINISTRATIONS OF ABOBOTULINUMTOXINA

Alberto Esquenazi (Elkins Park, PA), Allison Brashear (Winston Salem, NC), Gustavo Suarez (Basking Ridge, NJ), Claire Vilain (Les Ulis, FR), Philippe Picaut (Les Ulis, FR), Jean-Michel Gracies (Créteil, FR)

INTRODUCTION: Walking speed (WS) is an important predictor of walking capability along a continuum from limited household ambulation to unlimited community ambulation. Walking ability has important health implications in providing protective effects against secondary complications following stroke or traumatic brain injury and is associated with improvements in quality of life.

OBJECTIVE: To evaluate the 10-Meter Walk Test (10-MWT) in patients with spastic hemiparesis as an indicator of functional improvement following repeated abobotulinumtoxinA (aboBoNT-A, Dysport®) treatment.

METHODS: This is a phase 3, multicenter, prospective, double-blind, randomized, placebo-controlled, single-treatment-cycle study (NCT01249404) comparing aboBoNT-A with placebo in adults with chronic hemiparesis affecting the lower limb, followed by an open-label multiple-cycle extension study (≤4 additional treatment cycles; NCT01251367). Muscle tone was the primary endpoint; the secondary endpoint was unassisted Comfortable Barefoot WS (CBWS), assessed using the 10-MWT.

RESULTS: In the double-blind study, mean change from baseline in CBWS showed no significant difference between placebo and aboBoNT-A at Week 4 (W4) post-treatment. Clinically relevant improvements in CBWS were observed across open-label treatment cycles, with mean (SD) change from double-blind baseline reaching +0.07 (0.12), +0.08 (0.13), +0.08 (0.13) and +0.09 (0.14) m/s at W4 across cycles 1-4, respectively (+0.06 m/s is considered clinically meaningful). AboBoNT-A exhibited a safety profile consistent with previous clinical experience.

SUMMARY/CONCLUSION: The last patient completing the study is expected in March 2018.
INCOBOTULINUMTOXINA SUSTAINABLY IMPROVES UPPER LIMB SPASTICITY – POOLED ANALYSIS OF TWO PHASE 3 TRIALS

Michael Munin (Pittsburgh, PA), Petr Kanovsky (Olomouc, CZ), Michael Althaus (Frankfurt, GE), Angelika Hanschmann (Frankfurt, GE), Irena Pulte (Frankfurt, GE), Reinhard Hiersemnel (Frankfurt, GE)

INTRODUCTION: The efficacy and safety of incobotulinumtoxinA for upper limb post-stroke spasticity has been demonstrated in 2 phase 3 studies; both studies included a placebo-controlled main period (MP) and an open label extension (OLEX). The OLEX period, study 3001, included 3 additional injections of incobotulinumtoxinA; study 0410 included ≤5 additional injections.

OBJECTIVE: The objective of this post-hoc analysis was to assess key efficacy outcomes using pooled data from both studies.

METHODS: Subjects received either 400 U fixed doses of incobotulinumtoxinA at 12-week intervals (study 3001) or ≤400 U doses at flexible intervals (≥12 weeks; study 0410). Outcome measures included Ashworth Scale (AS) scores and Disability Assessment Scale (DAS) scores. An additional AS arm sum-score was calculated by combining AS scores for flexed wrist, clenched fist, flexed elbow, thumb-in-palm, and pronated forearm.

RESULTS: The pooled analysis included 465 subjects (incobotulinumtoxinA, n=283; placebo, n=182). The AS arm sum-score during the myotonic discharge (MP) improved from baseline by a mean (SD) of 3.23 (2.55) at 4 weeks postinjection (placebo, 1.49 [2.09]). Sum-score improvements during the OLEX were 4.38 (2.85), 4.87 (3.05), and 5.03 (3.02) in cycles 1-3, respectively. The DAS responder rate (principal target domain) increased from 47.4% for incobotulinumtoxinA-treated subjects during the MP to 66.6% during OLEX cycle 3. A dose dependence was observed for AS responder rates during the MP for individual joints, with a greater effect observed in the larger versus smaller muscle groups.

SUMMARY/CONCLUSION: Results are consistent with previous studies and demonstrate that repeated treatment with adequate doses of incobotulinumtoxinA enables sustained improvement of muscle tone and disability.

EFFICACY OF INCOBOTULINUMTOXINA IN THE TREATMENT OF SHOULDER SPASTICITY

Rashid Kazerooni (Raleigh, NC), Djamel Bensmail (Garches, FR), Astrid Scheschonka (Frankfurt, GE), Birgit Flatau-Baqué (Frankfurt, GE), Olivier Simon (Frankfurt, GE), Jörg Wissel (Berlin, GE)

INTRODUCTION: There are limited data on treatment of shoulder spasticity using botulinum toxin. Because of proximity to auxiliary breathing muscles, treatment of the shoulder using botulinum toxin is considered challenging; however, use of the shoulder is important for improved functionality and quality of life (QOL) in many patients.

OBJECTIVE: To evaluate the efficacy and safety of incobotulinumtoxinA to treat shoulder spasticity using a post-hoc analysis of the TOWER study (NCT01603459).

METHODS: One hundred fifty-five subjects received escalating fixed doses of incobotulinumtoxinA in 3 treatment cycles (400, 600, and 600-800 U, respectively). Assessments of muscle tone (Ashworth Scale, or AS) for all clinical patterns were conducted during the injection visit and a control visit 4 weeks postinjection. A post-hoc analysis compared subjects who received treatment in the shoulder with those who did not. An AS shoulder sum-score (AS-SSS) was determined by combining AS scores for shoulder adductors, extensors, and internal rotators. The EQ-5D instrument was used to assess QOL. Adverse events were monitored throughout.

RESULTS: Eighty-four subjects received incobotulinumtoxinA treatment in the shoulder during cycle 3 (mean shoulder dose: 118.4 U); 57 subjects did not. Among those treated, the mean (SD) AS-SSS improvement was −1.7 (1.8) during cycle 3, compared with −0.9 (1.4) for untreated subjects. Multiple regression analysis (adjusting for AS-SSS baseline) revealed a significant dose dependence for effect of incobotulinumtoxinA on AS-SSS (p=0.0081). All dimensions of the EQ-5D improved across injection cycles. Treatment-related adverse events indicating toxin spread to the lung (e.g., respiratory depression) were not observed.

SUMMARY/CONCLUSION: Results demonstrate the safety and efficacy of incobotulinumtoxinA for treating shoulder spasticity and supports further clinical evaluation.
EFFICACY OF INCOBOTULINUMTOXINA IN TREATMENT OF LOWER LIMB SPASTICITY, INCLUDING PES EQUINOVARUS IN ADULTS
Rashid Kazerooni (Raleigh, NC), Djamel Bensmail (Garches, FR), Astrid Scheschonka (Frankfurt, GE), Birgit Flatau-Baqué (Frankfurt, GE), Olivier Simon (Frankfurt, GE), Jörg Wissel (Berlin, GE)

INTRODUCTION: IncobotulinumtoxinA has been shown to be safe and effective for the treatment of upper limb (UL) spasticity.

OBJECTIVE: This post-hoc analysis assessed the effectiveness of incobotulinumtoxinA for treating lower limb (LL) spasticity, including pes equinovarvarus.

METHODS: The TOWER study (NCT01603459) included 3 cycles with escalating fixed total doses of incobotulinumtoxinA (400, 600, and 600-800 U, respectively). Adult subjects (n=155; 18-80 years) with UL and LL spasticity were enrolled. Assessments included the Ashworth Scale (AS), AS responder rates (i.e., subjects with ≥1 point improvement at 4-weeks), and Resistance to Passive Movement Scale (REPAS).

RESULTS: One hundred nine subjects received LL treatment with incobotulinumtoxinA in cycle 1; 100 subjects received pes equinovarvarus treatment. The mean±SD dose for pes equinovarvarus was 166.3±94.9 U during cycle 1. At 4 weeks, the mean improvement±SD in ankle joint AS score was 0.63±0.76 in subjects treated for pes equinovarvarus and 0.16±0.63 in subjects not treated for this pattern. The incobotulinumtoxinA dose used to treat pes equinovarvarus significantly affected AS improvement (p=0.0096 using multiple regression analysis adjusting for AS baseline). Fifty-five percent of treated subjects were considered AS responders, compared with 12.7% of untreated subjects (p<0.0001). At 4 weeks, the mean improvement±SD in REPAS LL scores from baseline among those who received LL treatment was 1.6±2.0, compared with 0.3±1.5 in subjects who did not receive treatment. Multiple regression analysis on LL REPAS baseline value and LL dose demonstrated a significant effect of LL dose administered (p=0.0022).

SUMMARY/CONCLUSION: Results demonstrate that incobotulinumtoxinA is a safe and effective treatment for LL spasticity.

VITALITY-ALS PHASE 3 TRIAL OF TIRASEMTIV, A FAST SKELETAL TROPONIN ACTIVATOR, AS A POTENTIAL TREATMENT FOR PATIENTS WITH ALS: STUDY DESIGN AND BASELINE CHARACTERISTICS
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INTRODUCTION: Tirasemtiv demonstrated statistically significant effects versus placebo on slow vital capacity (SVC) after 3 months of treatment in patients with ALS in a large phase 2b study (BENEFIT-ALS).

OBJECTIVE: Ventilatory Investigation of Tirasemtiv and Assessment of Longitudinal Indices after Treatment for a Year (VITALITY-ALS; NCT02496767) further assesses the effect of tirasemtiv versus placebo on respiratory function in ALS.

METHODS: VITALITY-ALS is a phase 3 multinational (North America and Europe), double-blind, randomized, placebo-controlled, stratified, parallel-group study. Eligible participants (diagnosis ≤24 months; minimum SVC ≥70% predicted) were randomized 3:2:2:2 to placebo or target total daily Tirasemtiv dose groups of 250, 375, or 500 mg after 2 weeks of open-label tirasemtiv (125 mg twice a day) to establish tolerability. Outcomes include change in SVC from baseline to 24 weeks and secondary outcomes related to respiratory function loss based on 1 year of treatment/followup.

RESULTS: Enrolled patients are, on average, 57.6 years, 7.7 months from diagnosis, and 20.6 months from first symptoms and have an average predicted SVC of 90.7%; 65% are male. As seen previously with tirasemtiv, the most common adverse events (AEs) in the open-label phase are dizziness, fatigue, and nausea. Approximately 24% patients withdrew from VITALITY-ALS during the 2-week, open-label phase, primarily due to AEs, similar to what was observed during the first 2 weeks of tirasemtiv treatment in BENEFIT-ALS.

SUMMARY/CONCLUSION: VITALITY-ALS evaluates the hypotheses that tirasemtiv significantly reduces SVC decline over 24 weeks versus placebo and meaningfully impacts other clinical outcomes. Study completion is expected in the fourth quarter of 2017.
DURATION OF TREATMENT EFFECT OF INCOBOTULINUMTOXINA IN UPPER LIMB SPASTICITY
Michael Munin (Pittsburgh, PA), Petr Kanovsky (Olomouc, CZ), Michael Althaus (Frankfurt, GE), Angelika Hanschmann (Frankfurt, GE), Irena Pulte (Frankfurt, GE)

INTRODUCTION: The efficacy and safety of incobotulinumtoxinA in upper-limb post-stroke spasticity (ULPSS) has been confirmed in 2 phase 3 studies. Study 0410 included a randomized placebo-controlled period and an open-label extension (OLEX) with flexible reinjection intervals (≥12 week intervals with doses ≤400 U). Study 3001 included fixed 12-week treatment intervals (TIs). In study 0410, investigators and subjects mutually determined the need for reinjection.

OBJECTIVE: In this post-hoc analysis, the duration of treatment effect for incobotulinumtoxinA TIs was assessed.

METHODS: All TIs between 2 consecutive incobotulinumtoxinA injections were included, except TIs prior to the end of study visit and those with doses <300 U. Outliers and factors other than a medical need for reinjection were accounted for using duration thresholds applied during the analysis; the number of subjects with ≥1 TI above a threshold was calculated.

RESULTS: A total of 347/437 incobotulinumtoxinA TIs met the inclusion criteria (range of intervals observed: 9-49 weeks). Over half (54.8%) of the re-injections were administered at week ≥14. The mean incobotulinumtoxinA TI was 15.46 weeks (SD 4.63 weeks). A majority of subjects (59.1%) had ≥1 TI with reinjection at week ≥16; 33.1% of subjects had 1, 22.1% had 2, and 3.9% had 3. Many subjects (42.5%) had ≥1 TI with reinjection at week ≥18.

SUMMARY/CONCLUSION: Results demonstrate variability in the duration of treatment effect, which supports the use of flexible and individualized dosing intervals for the treatment of ULPSS. The duration of treatment effect was ≥14 weeks after most treatments; however, many experienced effects lasting up to 20 weeks.

DURATION OF EFFECT OF ABOBOTULINUMTOXINA IN ADULT PATIENTS WITH LOWER LIMB SPASTICITY POST-STROKE OR TRAUMATIC BRAIN INJURY
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INTRODUCTION: Few studies have assessed treatment intervals during repeated botulinum toxin injections of any form. In a double-blind (DB) single-treatment, and long-term open-label (OL) extension study, abobotulinumtoxinA (aboBoNT-A, Dysport*) was efficacious in patients with lower limb spasticity, with no unexpected safety findings.

OBJECTIVE: To evaluate re-treatment intervals after repeated injections of aboBoNT-A in hemiparetic adults with lower limb spasticity.

METHODS: This is a phase 3, international, multicenter, DB, single-treatment study of aboBoNT-A in the hemiparetic lower limb (NCT01249404), followed by a long-term OL extension study (≤4 additional treatment cycles over ≤18 months; NCT01251367). Re-treatment, per investigator’s judgement, was possible at weeks 12, 16, 20, and 24.

RESULTS: Among patients who received aboBoNT-A in the DB study and were treated in OL cycle 1, 20% were re-injected at week 16 or later (week 16, 10%; week 20, 5%; week 24 or later, 5%). For those patients who received a second OL treatment cycle, 32% were re-injected at week 16 or later (week 16, 17%; week 20, 9%; week 24, 7%). For those patients who received a third OL treatment cycle, 15% were re-injected at week 16 or later.

SUMMARY/CONCLUSION: These data demonstrate the long time to re-treatment of aboBoNT-A in lower limb spasticity, with 15-32% of patients re-injected at week 16 or later across repeated cycles. A longer time to re-treatment may reduce the burden associated with frequency of injections for patients and their caregiver/families. These data also highlight the need for a tailored approach in lower limb spasticity treatment for hemiparetic adults.
CHARACTERISTICS AND TREATMENT PATTERNS OF COMMERCIALLY INSURED PEDIATRIC CEREBRAL PALSY PATIENTS IN THE UNITED STATES

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INTRODUCTION: Cerebral palsy (CP), the most common childhood motor disability, causes spasticity in the majority of diagnosed children. Spastic CP patients may utilize a variety of treatment options, including, but not limited to, botulinum toxin type A (BoNT-A), anti-spasticity medications, and baclofen.

OBJECTIVE: To describe real-world demographics, clinical characteristics, and utilization of BoNT-A to treat pediatric spastic CP in the United States.

METHODS: A retrospective analysis of claims from a U.S. commercial insurance database was conducted to identify pediatric patients with CP (ICD-9-CM 343.X) between 1/1/2010 and 12/31/2015. Variables included: patient demographics (pre-index date), comorbidities, and utilization of BoNT-A (Botox® [onabotulinumtoxinA] and Dysport® [abobotulinumtoxinA]) and other spasticity management therapies (on or after index date). For BoNT-A, mean dose administered was evaluated for all 343.X ICD-9 codes.

RESULTS: A total of 33,679 pediatric patients had a CP diagnosis; 16.6% hemiplegic, 20.3% diplegic, 25.5% quadriplegic, and 37.6% unspecified. CP was frequently diagnosed among children 6-12 years of age. Prevalent comorbidities included epilepsy (31.8%), respiratory (35.3%), and gastrointestinal (31.3%) problems. BoNT-A (onabotulinumtoxinA: 97.8%, abobotulinumtoxinA: 2.2%) was given to 12.4% of CP children. Mean dose (SD) administered per patient ages 2-17 years (mean age: 9.1 years) was 311.3 units (284.8) for onabotulinumtoxinA (n=5639) and 810.8 units (458.2) for abobotulinumtoxinA (n=119).

SUMMARY/CONCLUSION: Real-world use of BoNT-A to treat pediatric spastic CP remains low. Treatment is multifaceted and multidisciplinary, with further analyses needed to understand the unmet needs of this patient population.
FIRST-IN-HUMAN PRELIMINARY PK AND SAFETY DATA ON A NOVEL RECOMBINANT ACID A-GLUCOSIDASE, ATB200, CO-ADMINISTERED WITH THE PHARMACOLOGICAL CHAPERONE (PC) AT2221 IN PATIENTS WITH LATE-ONSET POMPE DISEASE

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INTRODUCTION: Pompe disease, caused by deficiency of lysosomal acid alpha-glucosidase (GAA), leads to progressive glycogen accumulation and muscular weakness. A next-generation approach using a combination of optimally glycosylated recombinant human GAA (rhGAA) and a pharmacological chaperone (PC) is in early clinical trials.

OBJECTIVE: To evaluate preliminary safety (n=13) and pharmacokinetic/pharmacodynamic (PK/PD) (n=10) data in enzyme replacement therapy (ERT)-experienced and -naive patients.

METHODS: In adults with Pompe disease, study ATB200-02 (NCT02675465) is designed to evaluate safety, tolerability, PK, and PD, including muscle enzymes/urinary Hex4, of the novel optimally glycosylated IV rhGAA enzyme replacement therapy, ATB200, co-administered with the oral PC AT2221.

RESULTS: Single 4-hour IV infusions of ATB200 at 5, 10, and 20 mg/kg demonstrated slightly greater than dose-proportional exposures. Oral administration with AT2221 prior to infusion increased total GAA protein terminal-phase partial AUCs (area under the curve) (tmax to 24 hours post-dose) by approximately 25% and plasma half-life by approximately 45% relative to ATB200 20 mg/kg alone. Plasma AT2221 demonstrated dose-proportional kinetics with peak concentrations 2 hours into infusion. Markers of muscle injury (aspartate aminotransferase, alanine aminotransferase, and creatine kinase) improved or stabilized. Urinary Hex4 levels decreased. Adverse events (AEs) have been mild; none led to treatment discontinuation. No serious AEs or infusion-associated reactions (IARs) were reported after 150+ total infusions in all patients.

SUMMARY/CONCLUSION: ATB200/AT2221 is safe and well tolerated, with no IARs reported to date. Muscle enzymes decreased or stabilized. Increased exposure and half-life of ATB200 co-administered with AT2221 confirms the intended mechanism of action of stabilization in circulation by AT2221.
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FINDING MARKERS IN ALS DIAGNOSIS
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INTRODUCTION: ALS is an uncommon illness caused by motor neuron degeneration, and upper, lower, and bulbar muscles are affected. Diagnosis is based on El Escorial criteria. Some research also reports degeneration in non-motor structures.

OBJECTIVE: To evaluate electrophysiological and image techniques such as markers in the diagnosis of ALS and to correlate these results.

METHODS: From January 2014 to January 2016, 20 patients with an ALS diagnosis and 20 healthy subjects were evaluated. All participants underwent sensory and motor NCSs, needle EMG, and somatosensory evoked potentials (SSEPs), as well as MRIs. Post-processing MRI techniques such voxel-based morphometric, diffusion techniques and corticospinal tract tractography were applied.

RESULTS: NCS results were positive in 90% of the patients, SSEPs were positive in 60%, and needle EMG abnormalities were observed in 100%. An anatomic MRI was positive in 60% of the patients, it showed non-specific abnormalities such as cortical atrophy and ventricle enlargement. Fractional anisotropy was reduced, mean diffusivity was increased, and fiber number was diminished in ALS patients in relation to healthy subjects. Also, grey and white matter were reduced in motor and somatosensory cortex, cerebellum and corpus callosum, respectively, in ALS patients in relation to healthy subjects. Clinical, EDX, and MRI parameters were in correlation.

SUMMARY/CONCLUSION: Electrophysiological studies confirmed the ALS diagnosis in 100% of cases. Electrophysiological studies and new image parameters could be markers in ALS diagnosis.

Bárbara Hernandez, MD, PhD
IFCN North American Chapter Fellowship Award Recipient

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THE PORTRAYAL OF ALS IN CINEMA AND TELEVISION
Jose David Avila (Danville, PA)

INTRODUCTION: ALS, like other neurologic disorders, has been portrayed in cinema and television. However, literature analyzing the nature and accuracy of its representation is limited.

OBJECTIVE: To describe the depiction of ALS in English language cinema and television.

METHODS: A list of films was developed from various sources, including a PubMed search, other publications, and online resources as well as the author's own experience. All films were reviewed by a neuromuscular specialist.

RESULTS: Forty-three films were identified, most of which were full-length documentaries. There were also theatrical and television films, short films, and miniseries. In most films the individual with ALS was the main character. Different aspects of the disease have been portrayed, including the onset and progression, the burden on spouses and caregivers, and end-of-life decisions. Some documentaries have shown the effect of the illness in individuals with different occupations: athletes, musicians, artists, and filmmakers themselves. The lives of 2 well-known figures suffering from ALS, the late Yankees player Lou Gehrig and the theoretical physicist Stephen Hawking, have been the subject of multiple films. Several actors have received critical acclaim and accolades for their roles. Documentaries display a more accurate depiction of the disease compared to other films.

SUMMARY/CONCLUSION: ALS has been portrayed in several films, addressing different aspects of the disease with variable levels of accuracy. It is relevant for neuromuscular physicians to be familiar with these depictions as they may influence the public's perception of the illness. Additionally, some films or portions of them may be used for educational purposes.
GUT INFLAMMATION AND DYSBIOSIS IN MOTOR NEURON DISEASE
Julie Rowin (Westchester, IL), Barbara Jung (Chicago, IL), Jun Sun (Chicago, IL)

INTRODUCTION: Emerging evidence supports a potential role for intestinal dysbiosis in the development of neurodegenerative diseases. We have demonstrated gut dysbiosis and increased intestinal permeability in ALS SOD1G93A mice, and have shown that improvement in gut integrity and microbial homeostasis significantly prolongs lifespan in this model. However, little is known about the intestinal microbiome in patients with motor neuron disease (MND).

OBJECTIVE: We describe stool microbiome analysis in 4 patients with MND: 2 with bulbar-onset ALS, 1 with lower motor neuron disease (brachial amyotrophic diplegia) with lymphoproliferative disease, and 1 with upper and lower motor neuron disease associated with celiac disease.

METHODS: Genova Diagnostics Gastrointestinal Effects® comprehensive stool profile was performed to evaluate for infection, inflammation, and bacterial imbalance, diversity, and abundance.

RESULTS: Three of 4 patients showed elevated inflammatory markers (fecal secretory IgA [sIgA], calprotectin, and/or eosinophilic protein X (EPX). Three of 4 patients showed dysbiosis indicated by decreased diversity of the microbiome and lower Firmicutes/Bacteroidetes (F/B) ratio. The 1 patient without inflammatory markers in the stool had biopsy-confirmed celiac disease and was gluten-free for 6 months. The 1 patient without dysbiosis was taking probiotic supplementation.

SUMMARY/CONCLUSION: This preliminary look into the gastrointestinal health of 4 patients with heterogenous MNDS revealed dysbiosis (decreased diversity and F/B ratio) and/or elevated inflammatory markers (sIgA, calprotectin, and/or EPX) in all cases. These studies, in conjunction with findings in ALS SOD-1 mice, suggest that further study of the interplay of gut health with the onset and progression of MND may reveal novel therapeutic targets for disease modulation.
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A RARE PRESENTATION OF ALS: PTOSIS AND SLURRED SPEECH
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INTRODUCTION: ALS is a progressive motor neuron disease usually presenting with atrophy and weakness of the extremities or bulbar muscles. Extraocular muscle involvement has been thought to be nonexistent, however several case reports have described patients with palsy of these muscles.

CASE REPORT: This is a 61-year-old man with bulbar-onset ALS with presenting symptoms of left eye ptosis and dysarthria. Initial examination was significant for spastic speech, left eye ptosis, and mild tongue atrophy as well as increased muscle stretch reflexes without sensory abnormalities. His workups, including anti-acetylcholine and anti-muscle-specific receptor tyrosine kinase (MuSK) antibodies, were normal. An EDX study showed active denervation associated with chronic reinnervation in the cranial, cervical, and lumbosacral regions. Within 1 year, followup examination was notable for worsening weakness of the extremities, bilateral ptosis, anarthria, and severe limitation of vertical gaze. The oculocephalic maneuver elicited a full range of motion.

SUMMARY/CONCLUSION: In this case, vertical eye movements were severely limited, however full movements were elicited by the oculocephalic maneuver which is suggestive of a supra-nuclear origin of this abnormality. The involvement of these muscles occurred within 1 year of diagnosis; providing further evidence that oculomotor involvement has the potential to occur very early in the ALS disease process. A possible explanation for the development of the supra-nuclear palsy could be neuronal loss or impairment of regions including the rostral interstitial nucleus of the medial longitudinal fasciculus and substantia nigra, which are important in the generation of vertical saccades.

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CLINICOSOCIAL FACTORS THAT INFLUENCE DIAGNOSTIC DELAY IN ALS
Drew Young (Columbia, MO), Raghav Govindarajan (Columbia, MO)

INTRODUCTION: ALS is a progressive neurodegenerative disease with an average survival time after diagnosis of 3 years. Thus, early diagnosis and supportive treatment of the disease is paramount to ensuring patients have the best possible quality of life.

OBJECTIVE: To identify the clinical, demographic, and social predictors of delay in diagnosing ALS.

METHODS: This was a retrospective chart review including 50 patients that received complete care for ALS between 2010 and 2016. Demographics, social factors, site of onset, and diagnostic tests run on individual patients were gathered.

RESULTS: Of the 50 patients (12 upper limb-onset, 25 lower limb, 13 bulbar; 29 male, 21 female; 95% Caucasian) included in the study, the average age of diagnosis was 60 years, with bulbar-onset at 65 years, and with upper limb-onset at 56 years. Lower limb-onset ALS was diagnosed 27 months after symptom onset (p<0.01), upper limb at 12 months, and bulbar at 7 months. Patients with upper limb-onset saw a range of 1-2 physicians before a diagnosis was established, bulbar 1-3, and lower limb 2-5. Older male patients, those with lower limb-onset, those with prior documented history of alcohol abuse, those with frontotemporal dementia or behavior, and those receiving care at the VA had delay in diagnosis (p<0.05). Patients with history of frontotemporal behavior, lower limb-onset, and alcohol abuse (current) had the longest delay (30 months, p<0.01).

CONCLUSION: Older male patients with lower limb onset ALS, frontotemporal behavior, and current alcohol abuse caused significant delay in the diagnosis of ALS.

Drew Young, BS
Resident and Fellow Member Award Recipient
THE PENN UPPER MOTOR NEURON SCORE AS A PREDICTOR OF MORTALITY IN ALS
Christyn Edmundson (Cambridge, MA), Lauren Elman (Philadelphia, PA), Nabila Dahodwala (Philadelphia, PA)

INTRODUCTION: The Penn Upper Motor Neuron Score (PennUMNS) is a novel, standardized clinical score that quantifies upper motor neuron (UMN) findings in ALS patients. The relationship between the PennUMNS and mortality in ALS has not been established.

OBJECTIVE: To evaluate whether the PennUMNS at diagnosis is a predictor of mortality in ALS.

METHODS: This was a retrospective cohort study of patients in the PennALS Center database. Inclusion criteria were (1) diagnosis of an ALS-spectrum disease and (2) ≥2 clinic visits recorded. Patients whose first visit was >6 months after diagnosis were excluded. We collected the PennUMNS and additional demographic and clinical characteristics. Patients were divided into tertiles based on PennUMNS at diagnosis. Characteristics of patients in each tertile were compared using Pearson’s chi-squared test. Survival between tertiles was compared using the Cox proportional hazards model, adjusting for potential confounding variables. Primary endpoints were death or tracheostomy.

RESULTS: This study included 587 patients. There was no significant difference in age, gender, body mass index, or forced vital capacity at diagnosis between PennUMNS tertiles. There was no significant difference in time to endpoint between tertiles. Hazard ratios (with 95% CI) for patients in the middle and upper tertiles compared with the lowest tertile were 1.15 (0.091-1.48) and 1.19 (0.93-1.54), respectively. In contrast, the ALS Functional Rating Scale-Revised, forced vital capacity, and age at diagnosis were significantly correlated with time to endpoint.

SUMMARY/CONCLUSION: The PennUMNS at diagnosis did not correlate with time to death or tracheostomy in this sample. These results suggest that UMN burden of disease at diagnosis is not an independent predictor of survival in ALS.

Christyn Edmundson, MD
Resident and Fellow Member Award Recipient
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CORRELATION OF CREATINE KINASE LEVELS WITH CLINICAL AND ELECTROMYOGRAPHIC FEATURES AND SURVIVAL IN ALS
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INTRODUCTION: Serum creatine kinase (CK) could be mildly to moderately elevated in some ALS patients, although the mechanism and influencing factors of CK elevation and whether it’s a predictor of survival of ALS are still not well understood. OBJECTIVE: To evaluate CK levels of ALS patients and to explore the relationship between CK levels and the clinical characteristics and survival.

METHODS: We analyzed the CK levels of 185 ALS patients who underwent long-term follow-up. The relationship between CK levels and clinical features and spontaneous needle EMG activity were analyzed by univariate analysis and multiple linear regression. Kaplan–Meier and Cox proportional hazards models were used for survival analyses.

RESULTS: Baseline serum CK was raised in 43% of participants. The median CK level was 160 U/L (range: 20-2574 U/L), and 99% of patients had a CK level less than 1000 U/L. CK levels were significantly higher in male patients than in female patients (204 [169] U/L versus 117 [111] U/L, p<0.001) and in patients with limb-onset ALS than with bulbar-onset ALS (p<0.001). CK levels were also correlated with serum creatinine (p=0.011) and the spontaneous potential score of needle EMG (p=0.037) but not correlated with age, disease duration, or body mass index. Log CK was independently correlated with survival of ALS patients (hazard ratio=0.457, 95% CI 0.221-0.947, p=0.035) after adjusting for confounding factors.

SUMMARY/CONCLUSION: Serum CK levels of ALS patients were correlated with gender, site of onset, serum creatinine, and spontaneous activity in needle EMG. Serum CK could be an independent prognostic factor for survival of ALS patients.

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MULTIDISCIPLINARY AND COMPREHENSIVE CARE FOR PATIENTS WITH ALS
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INTRODUCTION: ALS is a progressive, ultimately fatal motor neuron disease. Management requires input from multiple providers, often delivered through multidisciplinary clinics, which have been shown to improve quality of life and prolong survival. Our institution has depended on separate clinics without multidisciplinary coordination.

OBJECTIVE: Our Process Improvement (PI) project aims to assess delays and identify opportunities to improve communication.

METHODS: With institutional PI approval, a database was created to track existing and new patient progress through key clinics: neurology, pulmonology, sleep medicine, and speech pathology (SLP). A monthly multidisciplinary meeting was established to facilitate cross-specialty discussion of patients.

RESULTS: To date, 15 patients (4 female, mean age: 56 years) have been entered into the database. Data collection is ongoing for 9 (2 deaths, 4 moved away). Mean days from referral to neurology evaluation, needle EMG/neuromuscular consult, and diagnosis is 16, 32, and 72. Mean days from referral to SLP evaluation, swallow study, and pulmonology consultation is 37, 45, and 79. Patients in the system prior to project initiation averaged 214 days to connect to all key clinics; patients entering the system since initiation take an average of 32 days.

SUMMARY/CONCLUSION: Patients enter into the ALS care system through several channels, resulting in variable times to key encounters. Time between needle EMG and diagnosis accounts for much of the delay from referral to diagnosis, perhaps due to workup of alternative diagnoses. Participation in monthly multidisciplinary meetings has subjectively improved communication and fastened access to care. Input from additional services may allow identification of further areas for improvement.

Kaye Sedarsky, MD
Resident and Fellow Member Award Recipient
113 SQUAMOUS CELL CARCINOMA OF THE BASE OF THE TONGUE MIMICKING BULBAR ONSET ALS
Miguel Chuquilin (Gainesville, FL)

INTRODUCTION: Bulbar-onset ALS presents with progressive dysphagia and dysarthria. Tongue squamous cell carcinoma can cause tongue paralysis secondary to hypoglossal nerve infiltration. In rare cases, it can mimic motor neuron disease.

OBJECTIVE: To describe a case of base of the tongue squamous cell carcinoma mimicking bulbar-onset ALS.

CASE REPORT: A 67-year-old woman presented with 3 years of slowly progressive dysarthria and dysphagia. She had associated weight loss due to decreased oral intake. Needle EMG performed elsewhere showed denervation in tongue muscles, and she was given a diagnosis of motor neuron disease. On examination, facial strength was normal, palate elevated at the midline, gag was present, cough was weak, tongue was curved to the right, and she was unable to move it out or to either side. There were no tongue fasciculations. The rest of neurological examination, including muscle strength, was normal. Needle EMG showed no active denervation in other body regions (atypical for motor neuron disease), a pharyngeal MRI was ordered, which showed infiltrating tumor at the base of the tongue including both lingual neurovascular bundles. Biopsy showed focal non-keratinizing invasive squamous cell carcinoma, related to human papillomavirus (HPV) infection. The patient was scheduled for chemoradiation therapy.

SUMMARY/CONCLUSION: Base of the tongue squamous cell carcinoma can cause progressive tongue paralysis and mimic motor neuron disease. It should be in the differential diagnosis of progressive dysphagia and dysarthria without other weakness or denervation in needle EMG to prevent delay in diagnosis and treatment.

112 GASTROINTESTINAL COMPLICATIONS WITH NOURISHMENT VIA PEG TUBE IN ALS PATIENTS: A RETROSPECTIVE REVIEW
Samuel Dang (Tampa, FL), Maryam Alshammeri (Tampa, FL), Brittnay Harvey (Tampa, FL), Natalie Tucker (Tampa, FL), Jerrica Farias (Tampa, FL), Catherine Jones (Tampa, FL), Adam Hart (Tampa, FL), Raul Alsina (Tampa, FL), Clifton Gooch (Tampa, FL), Tuan Vu (Tampa, FL)

INTRODUCTION: The American Academy of Neurology guideline regarding the care of ALS patients recommends patients with worsening dysphagia or significant weight loss to have enteral nutrition administered via percutaneous endoscopic gastrostomy (PEG) tube to stabilize weight. Adequate nutrition has been shown to prolong survival and improve quality of life (QOL).

OBJECTIVE: To determine the incidence of gastrointestinal (GI) complications associated with PEG tubes in our ALS population which may degrade QOL.

METHODS: We performed a chart review of patients in our ALS Association Certified Center of Excellence who had PEG tube placement between April 2011 and December 2016 to identify GI complications after PEG placement.

RESULTS: During the study period, 141 patients had PEG tube placement. Ten of the patients had no additional information after the PEG placement and were excluded from further analysis. In the remaining 131 patients, we found GI complications in 96 (73%). Immediate complications included PEG site pain, seeping fluid/discharge, PEG site rash/inflammation, bleeding, infection, malodor, and swelling. Longer term complications included constipation, diarrhea, abdominal pain, nausea/vomiting, bloating, bloody stools, and gastroesophageal reflux disease (GERD). The diarrhea and constipation were severe enough to require treatment. One patient had cholecystitis.

SUMMARY/CONCLUSION: In our experience, GI complications were common with PEG tube placement. Constipation was the most common symptom; tube feeding formulas often had to be adjusted along with other appropriate preventive measures. Significant postoperative pain was experienced by almost one-fourth of the patients, necessitating proactive pain control measures. By identifying these complications, preemptive measures can be taken to prevent degradation of patients’ QOL.
INTRODUCTION: ALS is a progressive and non-curable motor neuron disease with a median survival rate of 3-5 years. Prior studies have shown that advanced age at the time of symptom onset, bulbar type, the degree of functional impairment, respiratory complications, and dysphagia are the primary causes of morbidity and mortality.

OBJECTIVE: To consider the other factors that might potentially be responsible for increased morbidity and mortality of ALS patients including the use of riluzole, the presence of the percutaneous gastrostomy (PEG) tube, depression, the presence of limb pain and cramps, and family support.

METHODS: Using electronic medical records, we reviewed 51 deceased ALS cases retrospectively at Saint Louis University Hospital between 2010 to 2015.

RESULTS: In our patient population, the disease course ran from 10 months to 4 years in both limb- and bulbar-onset; 45% patients were on riluzole. Out of the 51 patients, 36 received PEG tube. During their clinic visits, 60.7% patients suffered depression. Only 15% of patients had severe limb pain and cramps during illness. Almost all patients had strong family support.

SUMMARY/CONCLUSION: Riluzole showed no significant benefit on the course of the disease in either limb- or bulbar-onset ALS. Patients with bulbar- and limb-onset ALS received PEG tube equally. However, patients with bulbar-onset ALS received PEG 1 year sooner than the limb-onset ALS. No clear association was noted between depression and increased mortality in ALS patients. Severe limb pain affected patients negatively while strong family support impacted them positively.

INTRODUCTION: ALS is a neurodegenerative disorder. It may be mimicked by disorders affecting different levels of the motor system from cortex to muscle.

OBJECTIVE: To assess the reliability of motor unit number estimation (MUNE) in differentiating between ALS, cervical spondylosis, and cervical spondylotic myelopathy (CSM).

METHODS: Forty subjects were included in this study, divided into 4 groups: 10 healthy volunteers, 10 patients with cervical spondylosis, 10 patients with CSM, and 10 patients with ALS. All patients were subjected to thorough clinical assessment and cervical MRI, and manual incremental method and multiple point stimulation techniques were performed for MUNE.

RESULTS: In both manual incremental and multiple point stimulation methods, the ALS group showed a statistically significant difference compared to control subjects and other studied groups (p=0.001). However, no statistically significant difference was found on comparing the cervical spondylosis and CSM groups to each other and to the control group (p=0.999).

SUMMARY/CONCLUSION: The MUNE technique can give valuable information, helping to differentiate ALS from cervical spondylosis and CSM. It may be considered as a complementary tests for ALS, for the detection of motor unit loss.
UTILITY OF REPETITIVE NERVE STIMULATION TEST FOR ALS DIAGNOSIS

INTRODUCTION: Patients with ALS frequently present with decremental responses in the repetitive nerve stimulation (RNS) test. However, its role in ALS diagnosis has received little investigation. We previously documented that spontaneous activities in needle EMG of the trapezius muscle were highly specific to ALS. RNS of the trapezius muscle is especially promising, together with its minimal discomfort.

OBJECTIVE: To investigate the diagnostic utility of RNS in differentiation between ALS and cervical spondylotic amyotrophy (CSA), an important ALS mimic, especially in Japan.

METHODS: Patients were prospectively enrolled, and the diagnosis was confirmed by followup. RNS was performed on the abductor pollicis brevis (APB), upper trapezius (trapezius), and deltoid muscles. A decremental response exceeding 5% was considered abnormal.

RESULTS: Enrolled were 53 ALS patients (35 men, 18 women; age: 65.8±13.8 years) and 37 CSA patients (31 men, 6 women, age: 62.1±10.8 years). Abnormal decremental responses were observed in 32%, 51%, and 75% of ALS patients and 3%, 0%, and 20% of CSA patients for the APB, trapezius, and deltoid muscles, respectively. The sensitivity for 23 ALS patients with upper limb-onset was 78% for the trapezius and 100% for the deltoid muscles.

CONCLUSION: An abnormal decremental response in the trapezius muscle was 100% specific to ALS in comparison with CSA. Its presence would exclude CSA and strongly suggest ALS. No decrement in the deltoid muscle might exclude ALS in patients having symptoms with upper limb-onset. RNS is useful in differentiation between ALS and CSA.

HEREDITARY SPASTIC PARAPLEGIA WITH SPG11 MUTATIONS CAUSE SPINAL MUSCULAR ATROPHY
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INTRODUCTION: Mutations in the SPG11 gene account for most of the autosomal recessive hereditary spastic paraplegias (HSPs). The disease has a wide phenotypic variability but peripheral nervous system (PNS) involvement is not well characterized.

OBJECTIVE: To elucidate the type and frequency of PNS involvement in HSP-SPG11.

METHODS: We performed NCSs and monopolar needle EMG testing (Neuropack MEB-9200 Electromyographer) in 25 patients with confirmed molecular diagnosis of HSP due to SPG11 mutations.

RESULTS: Sensory nerve action potentials were essentially normal while compound muscle action potentials (CMAPs) unfolded striking abnormalities in 19/25 cases. Myography disclosed remodeled potentials in 24/25 patients, all of whom displayed chronic neurogenic changes affecting at least upper and lower extremities. In 12/22 individuals tested, bulbar myotomes displayed chronic neurogenic changes as well. Four patients displayed signs of acute denervation. There was no clear distal–proximal gradient, and in 9 there were significant asymmetries. Jointly, NCS and needle EMG changes indicated a chronic motor neuron disorder, resembling type 3 spinal muscular atrophy, in 24/25 patients. There was significant inverse correlation between the NCS-based score (sum of CMAPs) and age (r=−0.44, p=0.045). Correlation analysis also indicated more significant atrophy in patients with longer disease duration (r=0.71, p<0.001) and older age (r=0.85, p<0.001).

SUMMARY/CONCLUSION: We were able to demonstrate that PNS involvement in HSP-SPG11 is frequently encountered and is mainly due to motor neuronopathy. This finding is in line with neuropathology of HSP-SPG11, which reveals neuronal aggregates affecting the spinal cord anterior horns.

Ingrid Faber, MD, PhD
IFCN North American Chapter Fellowship Award Recipient
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A CASE OF ANTI-KU POSITIVE MYOSITIS PRESENTING WITH CHIEF COMPLAINT OF FACIAL NUMBNESS, TRIGEMINAL-LIKE PAIN AND PROGRESSIVE DYSPHAGIA
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INTRODUCTION: Anti-Ku antibodies are myositis-associated antibodies seen in autoimmune conditions, particularly scleroderma, and the most frequently associated symptoms are arthralgias and Raynaud’s phenomenon.

OBJECTIVE: To highlight that neurologic symptoms, both sensory and motor, may be the presenting complaints.

CASE REPORT: A 52-year-old female with a history of vitiligo was referred with a 1-year history of left facial numbness and intermittently associated electric shock like pain and progressive dysphagia, mild distal hand numbness, and minimal complaint of weakness at initial consult. Testing prior to consultation revealed normal esophageal motility, esophagogastroduodenoscopy, MRI of the brain, and lumbar puncture. Examination revealed decreased sensation in the left face and mild weakness in both proximal and distal upper extremity muscles and in hip flexors. There was no skin thickening or joint swelling. Blink reflex was normal. Needle EMG showed positive sharp waves diffusely. Laboratory results revealed creatine kinase 248 U/L (26-169), antinuclear antibodies >1:640, positive SSA antibody, and negative Scl-70. Deltoid muscle biopsy showed moderate multifocal perivascular centered inflammation (T-lymphocytes and plasma cells) and mild segmental myofiber degeneration. Anti-Ku antibody was positive. After starting high-dose steroids, all of the patient’s symptoms notably improved.

SUMMARY/CONCLUSION: This case highlights that patients with anti-Ku associated myositis may have neurologic manifestations of an associated autoimmune disease (in this case Sjögren’s syndrome) yet lack the more common rheumatologic examination findings or symptoms. Other findings not previously described but seen in this case were the severe dysphagia and facial pain as a principal manifestation of the condition despite minimal proximal and distal upper limb weakness.

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NOVEL HETEROZYGOUS MUTATIONS IN PLEC GENE CAUSING EPIDERMOLYSIS BULLOS A SIMPLEX WITH MUSCULAR DYSTROPHY, CASE SERIES OF TWO AFFECTED SISTERS
Ikjae Lee (Birmingham, AL), Cuixia Tian (Cincinnati, OH), Anna Hurst (Birmingham, AL)

INTRODUCTION: Epidermolysis bullosa simplex with muscular dystrophy is an autosomal recessive disorder characterized by early childhood-onset of progressive muscular dystrophy and blistering skin changes. It is caused by homozygous or compound heterozygous mutations in the plectin (PLEC) gene. The precise phenotype–genotype correlations have yet to be defined.

OBJECTIVE: To describe 2 affected sisters with a detailed clinical presentation, examination findings, MRI results, and confirmed novel PLEC mutations that have not yet been reported.

CASE REPORTS: Case 1: A 5-year-old female born premature at 25 weeks was evaluated for motor delay. Epidermolysis bullosa (EB) was diagnosed at age 5 months with recurrent skin blisters. She sat alone at 18 months, walked at 2 years, and never could jump with both feet. Physical examination revealed small stature, dysmorphic findings of micrognathia, high-arched palate, poor dentition with erosions, hand and feet blisters, and nail deformation. Neurological examination showed generalized muscle weakness, decreased reflexes, and delayed time with sit-to-stand, 30-feet-run, and 4-steps-climbing. Creatine phosphokinase was elevated at 519 U/L. Muscle MRI showed asymmetric atrophy of the quadriceps, mild fatty infiltration in the quadriceps and gluteus maximus, and subtle increased T2 signal in the quadriceps bilaterally. Genetic testing confirmed heterozygous mutations in PLEC (c.11912delA and c.6276dupA) causing frame shift stacking leading to premature stop codons. Case 2: The 3-year-old sister was also diagnosed with EB and found to have same heterozygous PLEC mutations. She had delayed motor development and elevated creatine kinase. Muscle MRI was unremarkable.

CONCLUSION: We present clinical evidence that the novel compound heterozygous PLEC mutations are pathogenic for EB simplex with muscular dystrophy.
A RANDOMIZED PLACEBO-CONTROLLED CLINICAL TRIAL EVALUATING EFFICACY AND SAFETY OF IGIV-C IN CORTICOSTEROID DEPENDENT PATIENTS WITH GENERALIZED MYASTHENIA GRAVIS

Rhonda Griffin (Research Triangle Park, NC), Junliang Chen (Research Triangle Park, NC), Kim Hanna (Research Triangle Park, NC), Elsa Mondou (Research Triangle Park, NC)

INTRODUCTION: Myasthenia gravis (MG) is an autoimmune disease affecting the neuromuscular junction with clinical symptoms of generalized muscle fatigue and weakness. While symptoms can be improved with acetylcholinesterase inhibitors, therapy often requires mitigation of the autoimmune process. Treatment modalities include corticosteroids (CSs)/immunosuppressives, plasma exchange, or IV immunoglobulin (IVIg). In MG, IVIg has shown improvement for exacerbations but additional data are needed to confirm its effectiveness as maintenance treatment. CSs have been considered an appropriate treatment for immunomodulation of MG which has primarily been based on clinical experience rather than randomized controlled trials. Long term CS use is complicated by severe side effects, thus reducing the CS dose is of clinical interest. A study conducted by Grifols S.A. (Barcelona, Spain) is evaluating efficacy of IGIV-C in a maintenance setting to achieve CS dose reduction while maintaining a similar MG control level.

OBJECTIVE: To evaluate the efficacy of IGIV-C versus placebo in reducing the CS maintenance dosage as measured by the percentage of patients who achieve 50% or greater CS dose reduction at week 39.

METHODS: This is a randomized controlled study to assess IGIV-C efficacy and safety in CS-dependent patients with generalized MG. Patients meeting eligibility criteria are stratified by baseline CS dose and randomized to receive IGIV-C or placebo for 36 weeks. Patients are to maintain their background MG regimen while CS is tapered per the protocol-specified algorithm.

RESULTS: Results are not yet available since the clinical trial is ongoing.

SUMMARY/CONCLUSION: The IGIV-C effectiveness in the treatment of MG as maintenance therapy during and post CS reduction will be determined.
ANTI-HMGCR NECROTIZING MYOPATHY PRESENTING WITH VERY MILD PHENOTYPE AND MYALGIA IN A STATIN-NAÏVE PATIENT

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INTRODUCTION: Anti-hydroxy-3-methylglutaryl-coenzyme A-reductase (HMGCR) antibodies are associated with necrotizing autoimmune myopathies. Oftentimes, the phenotype is characterized by severe weakness, although not all patients are affected to this extent. Most commonly, these autoantibodies occur following statin exposure, although they are also well described in statin-naïve patients. We describe a patient who had neither developed frank muscle weakness nor had exposure to statins, highlighting the phenotypic variability of this disease.

OBJECTIVE: To illustrate a case of anti-HMGCR-mediated myopathy presenting with myalgias and elevated creatine kinase (CK) in a young woman who had never received statins.

CASE REPORT: A 40-year-old woman presented with approximately 1 year of progressive low back pain. She also reported progressive lower extremity muscle pain, worse with light exercise. Her neuromuscular examination was normal, with intact muscle strength throughout. Her CK was elevated to 2300. Needle EMG revealed irritative findings restricted to the paraspinal muscles. Muscle biopsy was notable for rare necrotic fibers and a paucity of inflammation. She required no immunomodulatory treatment and continues to have normal strength and elevated CK with prolonged observation.

SUMMARY/CONCLUSION: The spectrum of anti-HMGCR-mediated muscle diseases is broad. We report a patient who was not only statin-naïve, but also had only mild manifestations, requiring no immunosuppression to this point. The diagnosis of anti-HMGCR myopathy should thus be considered in any patient with necrotizing fibers on muscle biopsy, regardless of their extent of weakness or history of statin use.

MUSCLE PATHOLOGY QUANTITATIVE EVENTS IN 18 PATIENTS WITH MITOCHONDRIAL DISORDERS

Joao Kouyoumdjian (Sao Jose Do Rio Preto, SP), Carla Graca (Sao Jose do Rio Preto, SP), Vanessa Ferreira (Sao Jose do Rio Preto, SP)

INTRODUCTION: Mitochondrial disorders can lead to 4 main events on muscle pathology: (1) mitochondrial accumulations in muscle fibers, detected by modified trichrome Gomori (“ragged-red fibers,” RRFs); (2) mitochondrial accumulations in muscle fibers, detected by succinate-dehydrogenase staining (succinate dehydrogenase, or SHD+, or "ragged-blue fibers"); (3) cytochrome-c-oxidase negative fibers (COX−); and (4) COX+SDH combination (COMBO+), where unapparent COX-deficient fibers stained strong blue from SDH.

OBJECTIVE: To quantify RRFs, SHD+, COX−, and COMBO+ fibers in muscle biopsies with mitochondrial findings.

METHODS: We retrospectively selected 18 muscle biopsies (deltoid) cases (females: 83.3%; mean age: 38.6 years, range: 5 months-70 years) with mitochondrial abnormalities based on Walker criteria, depending on age, the percentage of RRFs/COX− fibers, and clinical picture, and/or Sleigh criteria, depending on the percentage of RRFs, SDH+, and COX− fibers.

RESULTS: The series comprised chronic progressive external ophthalmoplegia (66.7%), proximal myopathy (22.2%), idiopathic hyperCKemia (11.1%), Kearns–Sayre syndrome (5.6%), mitochondrial encephalomyopathy with ragged red fibers and stroke-like episodes (5.6%), and a dystrophic pattern (5.6%). The mean percentage, SD, and range, for the events were: RRFs (3.95±3.16%, 0.42-10.24); SDH+ (7.55±6.10%, 1-21); COX− (10.9±7.19%, 0-29); and COMBO+ (14.22±12.78%, 0-46). General findings showed slight muscle fiber diameter, the absence of necrosis or proliferation of connective tissue, few fibers with internal nuclei, and some cases with fiber type grouping.

SUMMARY/CONCLUSION: In ascending order, the quantitative findings frequency were RRFs, SDH+, COX−, and COMBO+ fibers. The importance of the COMBO+ technique should be stressed when non-detectable COX-deficient fibers can be found.

Joao Kouyoumdjian, MD, PhD
IFCN North American Chapter Fellowship Award Recipient
PD-1 INHIBITOR-ASSOCIATED MYOPATHY: AN EMERGING AUTOIMMUNE MYOPATHY
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INTRODUCTION: Programmed death-1 (PD-1) is an inhibitory molecule in the immune system, which plays an important role in promoting self-tolerance and preventing autoimmunity. Recently, PD-1 inhibitors have been increasingly used in cancer immunotherapy and their immune-related adverse events have been increasingly recognized. Myopathy in patients undergoing PD-1 inhibitor therapy has not been well-described.

OBJECTIVE: To report a series of patients with myopathy during PD-1 inhibitor therapy.

CASE REPORT: Among 653 cancer patients treated with PD-1 inhibitors (pembrolizumab=389; nivolumab=264), 6 patients developed myopathy (pembrolizumab=6). Myopathy occurred after a median of 2 cycles of PD-1 inhibitor therapy (range: 1-4). All patients had mild neck flexor and/or proximal limb weakness, and 4 also had either bulbar or extraocular weakness. Repetitive stimulation of proximal nerves in 4 patients was normal. All 6 had EDX evidence of myopathy. Five patients underwent muscle biopsy (necrotizing myopathy=2; nonspecific myopathic changes=3). Median creatine kinase level was 388.5 U/L (range: 23-7, 307). Both necrotizing myopathy patients had severe bulbar weakness and/or ophthalmoparesis with mild limb involvement, and negative anti-signal recognition particle and anti-hydroxy-3-methylglutaryl-coenzyme A-reductase (HMGCR) antibodies. PD-1 inhibitors were discontinued. Five patients received immunotherapy (steroid monotherapy=2; steroid and plasma exchange=3). Both patients with necrotizing myopathy died despite combined treatment. Non-necrotizing myopathy patients responded to immunotherapy.

SUMMARY/CONCLUSION: Myopathy occurs in 0.92% of patients receiving PD-1 inhibitors and is more common with pembrolizumab. Extraocular or bulbar weakness is frequent. Muscle biopsy is important to identify a subset of patients with necrotizing myopathy who require aggressive immunotherapy given their grave prognosis.

OPTIMAL PLACEMENT OF NEEDLE EMG FOR EXTENSOR INDICIS: A CADAVERIC STUDY
Dong Hwee Kim (Ansan-si, KP), Jin Young Im (Ansan-si, KP), Hong Bum Park (Ansan-si, KP), Seok Jun Lee (Ansan-si, KP)

INTRODUCTION: The extensor indicis (EI) muscle is the most distal muscle innervated by the radial nerve. It is used when evaluating a radial nerve lesion or cervical radiculopathy by needle EMG.

OBJECTIVE: To identify the mid-point of EI muscle as the optimal site for needle EMG.

METHODS: Eighteen upper limbs of 10 adult cadavers were dissected. The midpoint (MP) of EI was marked at the middle of the musculotendinous junction and proximal origin of EI. The forearm length (FL) was measured from the radial head to the ulnar styloid process (USP). The forearm width (FW) at the level of EI was measured. The distance from USP to MP (USP_MP) as parallel to the line between the radial head to USP and from the medial border of ulnar bone to MP (UMB_MP) were measured.

RESULTS: The median values of FL and FW were 23.0 cm and 47.6 mm, respectively. The median values of USP_MP and UMB_MP were 49.6 mm and 16.1 mm, respectively. The percentage of USP_MP to FL and UMB_MP to FW were 21.3% and 32.9%, respectively.

SUMMARY/CONCLUSION: The MP of EI is approximately 5 cm proximal to USP level, about distal 20% of forearm length and 16.1 mm lateral to medial border of ulnar bone, about 30% of forearm width.
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CHOLINERGIC URTICARIA WITH EXERTIONAL HEAT ILLNESS AS PRESENTATION OF A NOVEL RYR-1 MUTATION
Randall Brown (Columbia, MO), Raghav Govindarajan (Columbia, MO)

BACKGROUND: Mutations in the skeletal muscle ryanodine receptor (RYR1) gene are a common cause of neuromuscular disease. Recent studies have shown additional phenotypes including exertional myalgia and heat intolerance/illness.

OBJECTIVE: To report an index case of a novel RYR1 mutation with exertional heat illness and present the phenotypic-genotypic spectrum of the family.

CASE REPORT: A 45-year-old male presented with recurrent episodes of heat exertion followed by generalized hives on mild physical exertion since his 20s. He had noted weakness when he was getting up from the floor after playing with his children. Neurological examination showed symmetric (4/5) weakness with hip flexion and knee extension. Creatinine kinase level was 400 U/L (52-336 U/L, Mayo Clinic). Vastus lateralis biopsy with nicotinamide adenine dinucleotide (NADH) stain showed central cores with predominance of type 1 fibers. Whole exome sequencing showed a heterogenous missense mutation K1393R, p.Lys1393Arg (AAG>AGG): c.4178A>G in exon 29 of RYR1. He had 4 children, 2 daughters (aged 13, 9 years) and 2 sons (fraternal twins, aged 6 years). Further history had revealed the twins were hypotonic with poor sucking and feeding at birth. They needed gastric tube for feeding. They also had a delay in meeting their milestones, especially motor. The daughters had attained their milestones at appropriate age but started developing exertional heat intolerance. Whole exome sequencing of the children revealed the heterogeneous K1393R mutation in the exon 29 of the RYR1 gene.

CONCLUSION: RYR1 mutations present with varying severity of phenotypes with the same gene mutation within the family and across the families.

Randall Brown, BS
Resident and Fellow Member Award Recipient

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A CASE OF ANDERSEN-TAWIL SYNDROME: RARE HEREDITARY PERIODIC PARALYSIS WITH RARE CLINICAL MANIFESTATION
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INTRODUCTION: Andersen–Tawil syndrome (ATS) is a rare hereditary autosomal dominant channelopathy caused by mutations in the KCNJ2 gene that affects membrane excitability of cardiac and skeletal muscle.

OBJECTIVE: To report an infrequent hereditary channelopathy with an unusual presentation.

CASE REPORT: We report the case of a 75-year-old man who has a medical history of hypertension, ischemic cardiopathy, and diabetes mellitus type 2. His mother and sister died of cardiopathy in the middle adult age. At age 12, he started having episodes of paraparesis lasting minutes or hours, triggered by exercise. These occurred with variable frequency: daily, monthly, or even being asymptomatic for 12 months. He entered the hospital at age 70 with progressive leg weakness, leading him to be wheelchair dependent. Neurological examination showed paraparesis, lower limb areflexia, hypertelorism, micrognathia, low-set ears, and clinodactyly. Sensory and motor NCSs were normal. Electrophysiological results of Fournier protocol correlated with clinical pattern type V. Exon 10 and 30 of the CACNA1S gene were studied, but no mutations were found. Exome sequencing revealed a heterozygous variant in the KCNJ2 gene c.224C>T (p.Thr75Met). On followup, he developed extreme bradycardia requiring a cardiac pacemaker; afterwards, his condition worsened and the patient died.

SUMMARY/CONCLUSION: We report a rare genetic mutation associated with ATS. This syndrome should be considered in patients having periodic paralysis, family medical history, and characteristic facies associated with electrocardiographic arrhythmias, QT prolongation, and bone structural deformities.

Micke Enrique De arco Espinosa, MD
IFCN North American Chapter Fellow Award Recipient
A RARE VARIANT OF THOMSEN DISEASE PRESENTING IN PREGNANCY
Christyn Edmundson (Boston, MA), Amanda Guidon (Boston, MA)

INTRODUCTION: Myotonia congenita (MC) is a nondystrophic myotonia caused by autosomal dominant (Thomsen disease) and autosomal recessive (Becker disease) mutations of the CLCN1 gene. Phenotypic severity varies significantly both within and between affected families.

OBJECTIVE: To describe a case of Thomsen disease caused by a rarely reported mutation, with symptoms manifesting exclusively during pregnancy.

CASE REPORT: A 34-year-old woman presented for neuromuscular evaluation of new-onset muscle stiffness during her first trimester of pregnancy. She had no prior history of muscle stiffness and no family history of neuromuscular disorders. Physical examination revealed grip myotonia. A needle EMG performed 2 years prior to pregnancy for right calf weakness showed abnormal spontaneous activity in several muscles without frank myotonic discharges. A subsequent needle EMG performed during the second trimester of pregnancy showed diffuse myotonic discharges. Genetic testing revealed a heterozygous c.774+1G>A mutation of the CLCN1 gene, previously reported in 2 families with Thomsen disease. In one, symptoms were characterized by juvenile onset of fluctuating clinical myotonia. In the second, mutation carriers were asymptomatic or described with mild, nonspecific muscular symptoms. This patient’s muscle stiffness resolved without treatment several weeks after uncomplicated delivery of a healthy infant.

SUMMARY/CONCLUSION: This case expands our understanding of the phenotypic range of disease caused by the c.774+1G>A mutation of the CLCN1 gene. This case also illustrates the potential for initial presentation or exacerbation of MC in pregnancy, demonstrated by both clinical and EDX parameters in this patient. The etiology of this phenomenon is unclear, but may be due to progesterone effects on chloride channel conductance.

Christyn Edmundson, MD
Resident and Fellow Member Award Recipient

DEBUNKING THE MYTH: DENERVATED MUSCLE IS THE SOLITARY CAUSE OF MUSCLE SPONTANEOUS ELECTRICAL ACTIVITY
Roger Coletti (Lewes, DE)

INTRODUCTION: Treatment of chronic muscle spasm is confounded by the belief that spontaneous electrical activity (SEA) is only found in denervated muscle. SEA referred to herein is continuous chaotic electrical activity and without evidence of reciprocal inhibition, to be distinguished from the variety of presentations of transient increased insertional activity. Prior reports have shown that needle EMG evidence of SEA is present in acquired chronic muscle spasm which was successfully treated with needle EMG-guided chemodenervation utilizing phenoxybenzamine.

OBJECTIVE: To correct needle EMG misinterpretation of nerve function and muscle pathology.

METHODS: We present a survey of clinical outcomes of 93 patients with SEA treated with needle EMG-guided chemodenervation.

RESULTS: A steady state of pain relief was achieved within 1 week of the injection procedure; 76% of patients reported having had years of prior pain, 50% of patients reported complete relief of pain. Regarding the impact on overall health, wellbeing, or ability to function, 55% of patients reported a major impact and 71.4% of patients reported pain relief that lasted over 3 months.

SUMMARY/CONCLUSION: Rapid resolution of pain and disability with a high degree of sustained pain relief is inconsistent with interpretation of SEA in these patients as the result of denervated muscle. Incorrect interpretation of muscle denervation as solitary cause of SEA needs to be abandoned. The clinical impact of this finding will promote further research and treatment of the pathological state of acquired chronic muscle spasm and resultant chronic pain.
ROLE OF QUANTITATIVE EMG, MOLECULAR GENETICS IN DETECTING CARRIERS OF DUCHENNE MUSCLE DYSTROPHY
Hanan Soliman (Giza, EG), Hanan Soliman (Giza, EG), Mona Nada (Cairo, EG), Ayatallah Farouk (Cairo, EG), Hanan El-Gendy (Cairo, EG), Manal Gaber (Beni Suef, EG)

INTRODUCTION: Duchenne muscle dystrophy (DMD) is an X-linked severe progressive muscular dystrophy caused by deficiency of dystrophin gene. Quantitative electromyography (QEMG) is one of the most important electrophysiological tests used in diagnosis of DMD.

OBJECTIVE: To evaluate the role of QEMG in the detection of myopathic carriers in DMD and assess the increase of micronuclei among carriers of DMD.

METHODS: The study included 30 female carriers proved by positive genetic study for DMD and 30 matched control subjects. All subjects underwent clinical evaluation, including laboratory tests of creatine phosphokinase (CPK) level, a molecular genetics study, and electrophysiological studies (routine and QEMG studies) of both upper and lower limbs.

RESULTS: A statistically significant decrease in the amplitude and duration of motor unit action potentials, with a statistically significant increase in CPK level and a number of binucleated micronucleated lymphocytes, were seen in the female carriers in comparison to the normal subjects.

SUMMARY/CONCLUSION: QEMG and genetic study have an important role in the detection of female carriers of DMD.

Manal Gaber, MBBS, Master
IFCN North American Chapter Fellowship Award Recipient

OPTIMAL PLACEMENT OF NEEDLE EMG FOR SUPINATOR: A CADAVERIC STUDY TO SAFE NEEDLE EMG SITE FOR SUPINATOR: A CADAVERIC STUDY.
Jin Young Im (Ansan-si, KP), Dong Hwee Kim (Ansan-si, KP), Hong Bum Park (Ansan-si, KP), Seok Jun Lee (Ansan-si, KP), Ki Hoon Kim (Ansan-si, KP), Byung Kyu Park (Ansan-si, KP)

INTRODUCTION: Needle EMG evaluation of the supinator muscle is important when localizing the level of involvement in a posterior interosseous nerve (PIN) injury. OBJECTIVE: To identify the optimal and safe needle insertion site to the supinator muscle.

METHODS: Twenty upper limbs of 11 adult cadavers were dissected. Distances from the radial head (RH) to midpoint of the dorsal wrist were measured. We made the RH the 0 point in the virtual quadrant and took this line as the X-axis. The medial side of the RH at the fully pronated position was defined as the positive direction of the Y-axis. Distances from the RH to the PIN at different levels as the coordinates of the X- and Y-axes were measured.

RESULTS: The median length of the RH to the mid-point of the dorsal wrist was 233.5 mm. The median lengths between the RH to the proximal and distal points of the supinator muscle were 11.5 mm and 100.6 mm. The median X coordinate values from the RH to the PIN were 21.9 mm, 41.5 mm, and 66.0 mm and the median Y were 8.8 mm, 1.5 mm, and −5.9 mm at the level of inlet, mid-point, and outlet of supinator muscle, respectively.

SUMMARY/CONCLUSION: The ideal length between the RH to the site of needle insertion is about 10-40 mm on the line between the RH and mid-point of dorsal wrist.
SUCCESSFUL TREATMENT OF LONGSTANDING CHRONIC MUSCLE SPASM WITH EMG GUIDED CHEMODENERVATION
Roger Coletti (Lewes, DE)

INTRODUCTION: Chronic muscle spasm represents a significant cause for chronic pain. Treatment of chronic pain with opioid medications has led to opioid addiction, and overdose deaths are currently recognized as a national crisis. Treatment modalities are needed to treat truly chronic pain when associated with chronic muscle spasm. Novel treatment modalities utilizing needle EMG-guided chemodenervation have been previously described. Preliminary assessment of the success of such treatments for longstanding chronic muscle spasm can be provided by patient surveys of such treatment.

OBJECTIVE: To identify success rates of needle EMG-guided chemodenervation with phenoxybenzamine in patients with pain duration of greater than 1 year.

METHODS: Ninety-three sequential patients treated with this technique were surveyed by mail. Forty-two responded.

RESULTS: Of the respondents, 31 (74%) reported years of pain duration. Of those, 50% reported complete relief of pain (81% of which reported relief of pain for greater than 3 months) and 27.4% reported moderate relief of pain (44% of which reported pain relief for greater than 3 months). The average duration of pain when specified was 5 years and the longest was 15 years. A single treated patient, not in this survey, reported near complete pain relief and return of function after 35 years.

SUMMARY/CONCLUSION: Truly longstanding chronic muscle spasm and pain can be successfully treated in a significant portion of patients with stable outcomes utilizing the previously described technique of needle EMG-guided chemodenervation with phenoxybenzamine. In this unselected patient population with longstanding chronic pain, results support further clinical research to establish the utility of this treatment modality.

LENALIDOMIDE TREATMENT LEADS TO NEAR COMPLETE NEUROLOGICAL RECOVERY IN MONOCLONAL GAMMOPATHY ASSOCIATED NEMALINE MYOPATHY- CASE REPORT AND REVIEW OF LITERATURE
Garima Agrawal (Goodlettsville, TN), Rishi Agarwal (Bowling Green, KY), Abhimanyu Ghose (Chandler, AZ), Saulius Girnius (Cincinnati, OH)

INTRODUCTION: Sporadic late-onset nemaline myopathy (SLONM) is a rare debilitating disease. The association of monoclonal gammopathy with SLONM has been reported previously and portends unfavorable prognosis. Treatment options of monoclonal gammopathy with SLONM include stem cell transplant, immunomodulators, and IV immunoglobulin (IVIg). Other than transplant, no other treatment has shown impressive response. We report the first case of effective treatment of IVIg-refractory SLONM in a transplant naive patient with the immunomodulatory drug lenalidomide.

CASE REPORT: A 65-year-old white female with Parkinsonism presented with slowly progressive facial numbness along with neck drop, dysphagia, difficulty chewing, taste changes, anorexia, and hoarse voice. She was found to have IgG lambda monoclonal gammopathy 0.7 gm/dl. Bone marrow biopsy showed 5-10% plasma cells with complex cytogenetics. Needle EMG showed a complex bilateral chronic trigeminal neuropathy, left facial neuropathy, and myopathy of axial muscles. Cervical muscle biopsy was consistent with sporadic late-onset nemaline myopathy. Neurologically, she deteriorated quickly and was soon wheelchair bound and unable to extend her neck. She was treated with IVIg for 5 days followed by every 2 weeks without improvement. Due to treatment failure with IVIg and presence of multiple myeloma, she was started on lenalidomide and dexamethasone. Within 1 year of treatment initiation, she had a complete hematologic and excellent neurological response, remission that remains ongoing and persistent an additional 14 months later.

SUMMARY/CONCLUSION: Lenalidomide/dexamethasone has clinical activity in plasma cell dyscrasias associated SLONM. Transplant ineligible SLONM patients may be offered lenalidomide at the earliest to prevent morbidity and mortality. Further studies are warranted to provide more support for this approach.
AN HETEROZYGOUS MAN WITH PHENOTYPE OF ADULT-ONSET GLYCOGEN STORAGE DISEASE TYPE 2

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INTRODUCTION: Pompe disease is a rare genetic lysosomal storage disorder caused by a deficiency of the enzyme acid α-glucosidase (GAA). This enzyme is necessary for the degradation of glycogen within lysosomes. The decrease of its activity leads to progressive accumulation of glycogen in several tissues, mainly in the cardiac and skeletal muscle.

OBJECTIVE: Pompe disease is considered an autosomal recessive disorder. However, the clinical symptoms in heterozygosis patients has not been completely established. We report a patient within heterozygosis with signs of myopathy compatible with late-onset Pompe disease.

CASE REPORT: We have identified a 59-year-old man with suggestive symptoms of adult-onset Pompe disease, specifically slowly progressive lower limb muscle weakness. He had sustained elevated creatine kinase (>500 UI/L, normal range: 75-185). Muscle biopsy was pathologic. The diagnosis of Pompe disease was based on reduced GAA enzymatic activity in dried blood spot (DBS) and purified lymphocytes (0.64 μmol/L/h, normal >75). He was found to have a novel heterozygous mutation in the GAA gene (c.1249A>C, exon 7, p.Asn417His). DNA was extracted from peripheral blood lymphocytes. The study did include laboratory examination of the blood, needle EMG, electrocardiography, pulmonary functional test, MRI, and muscle biopsy.

SUMMARY/CONCLUSION: Heterozygous individuals are not always asymptomatic, and probably the residual GAA enzyme activity correlates with a later age of onset and slower disease progression. Since enzyme replacement therapy is effective in Pompe disease, further studies are required to identify subjects in this group of individuals who may benefit from enzyme replacement therapy.

UPPER AND LOWER EXTREMITY MUSCLE STRENGTH DECLINES OVER TIME IN A PROSPECTIVE, OBSERVATIONAL GNE-MYOPATHY (HIBM) NATURAL HISTORY STUDY

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INTRODUCTION: GNE myopathy (GNEM) (also known as hereditary inclusion body myopathy, or HIBM) is a rare, severely debilitating adult-onset myopathy with progressive muscle weakness caused by a defect in sialic acid biosynthesis.

OBJECTIVE: To better understand the clinical presentation and progression of GNEM.

METHODS: This is an international, prospective, observational study enrolling up to 100 subjects with a genetically-confirmed diagnosis of GNEM. Muscle strength is measured by hand-held dynamometry (HHD) at baseline, 6, and 12 months. Individual muscle group strength using HHD measured in the upper extremities (UEs)—grip, shoulder abductors, and elbow flexors and extensors—and lower extremities (LEs)—hip flexors, extensors, abductors, and adductors and knee flexors—are combined to generate composite scores (UEC and LEC). Strength is compared with age- and sex-matched normative values.

RESULTS: Seventy two subjects (mean age: 40 years; 54% men; 92% white) completed 12 months. Thirty subjects (42%) could walk ≥200 m in the 6-minute walk test at baseline. Mean baseline muscle strength for all subjects was 35% and 19% of predicted normal values for the UEC and LEC, respectively. In the ≥200 m subgroup, predicted strength was 64% for UEC and 38% for LEC. Muscle strength (LS mean, 95% CI) declined by month 12, by −1.48 kg (-3.25, 0.30) for the UEC and −2.57 kg (4.32, −0.81) for the LEC. In the ≥200 m subgroup, the decline for UEC was similar (−1.51 kg [−5.28, 2.26]), and for LEC was greater (−3.84 kg [−7.52, −0.16]). Followup data for 24 months will be presented.

SUMMARY/CONCLUSION: These findings demonstrate the progressive decline of muscle strength in GNEM subjects in as little as 1 year.
FAILURE OF HYPOGLOSSAL NERVE STIMULATION FOR OBSTRUCTIVE SLEEP APNEA
Kaye Sedarsky (Bethesda, MD), Brian Robertson (Bethesda, MD), Jacob Collen (Bethesda, MD), Jonathan Smith (Bethesda, MD)

INTRODUCTION: Hypoglossal nerve stimulation (HNS) is an alternative treatment for select patients with obstructive sleep apnea (OSA) and difficulty tolerating continuous positive airway pressure (CPAP).

OBJECTIVE: To present a patient with severe OSA and excessive daytime sleepiness (EDS) who failed treatment with HNS and was later found to have myotonic muscular dystrophy type 1 (DM1).

CASE PRESENTATION: A 39-year-old man with EDS was diagnosed with severe OSA after polysomnography at an outside center demonstrated an apnea–hypopnea index (AHI) of 44 events/hour. He reported a paternal history of OSA but denied other notable medical history. After several weeks of CPAP therapy, he was unwilling to continue and elected to undergo HNS placement with postoperative resolution of snoring and improved sleep quality. However, followup HNS titration revealed an AHI of 43 events/hour with predominantly central sleep apnea. At followup, the patient's wife reported that he had a family history of DM1, and the patient endorsed difficulty releasing hand tools, hypophonia, and aspiration. Neurologic examination demonstrated flaccid dysarthria, temporalis atrophy, bifacial weakness, tongue atrophy without fasciculation, and grip and percussion myotonia. EDX testing revealed myotonic discharges. Genetic testing confirmed DM1.

SUMMARY/CONCLUSION: This case highlights the importance of screening for occult neuromuscular disease prior to consideration of HNS for OSA. Because neuromuscular diseases are proportionately rare, such conditions are often missed when evaluating patients for a prevalent disease such as OSA. Managing EDS and sleep disordered breathing in DM1 is challenging, often requiring multiple modalities and coordination between specialty providers.

Kaye Sedarsky, MD
Resident and Fellow Member Award Recipient

MYASTHENIA GRAVIS COEXISTING WITH FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY - A CASE REPORT
Nino Khizanishvili (Tbilisi, GE), Nana Kvirkvelia (Tbilisi, GE), Roman Shakarishvili (Tbilisi, GE), Maia Beridze (Tbilisi, GE)

OBJECTIVE: To investigate a facioscapulohumeral muscular dystrophy (FSHMD) patient with coexisting myasthenia gravis (MG).

CASE REPORT: A male patient, aged 67, was clinically investigated appealing to neurological consultation due to difficulties with breathing, swallowing, and speech. These symptoms began 1 year ago; they fluctuate, increasing after physical activity and vice versa. He had difficulty swallowing solid food and fluids as well. After physical activity, expression of fatigue, dysphagia and dysarthria was increased. The weakness was found in respiration muscles. After some rest, speech and swallowing improved and difficulty breathing went away, but weakness in the limb and trunk muscles did not change. A neostigmine test was positive for bulbar and respiration muscles. The patient also had weakness of facial, shoulder, and upper limb muscles beginning from age 12. The patient's mother and sister were diagnosed as FSHMD type 1. Genetic and serological laboratory testing, needle EMG, and mediastinum CT was performed.

RESULTS: FSHMD was diagnosed by molecular genetic diagnostics. Needle EMG revealed characteristic changes to a muscular dystrophy: decreased duration and amplitude of motor unit potentials during an interference curve of muscle's maximal contraction. Stimulation EMG revealed a postsynaptic type of neuromuscular transmission damage. A high level of antibodies against acetylcholine receptors and titin were revealed in blood serum. Thymus hyperplasia was seen on mediastinal CT. After 2-3 months of treatment with anticholinergic drugs, the patient's dyspnea, dysarthria, and hypophonia resolved. Therapy was continued by average maintaining doses of methylprednisolone.

SUMMARY/CONCLUSION: The present case is the rare example of MG coexisting with FSHMD.

Nino Khizanishvili, MD
AANEM Foundation for Research and Education Award Recipient
DIFFERENTIATION BETWEEN AUTOIMMUNE MYOPATHY AND MUSCULAR DYSTROPHIES
Olivia Yambem (Memphis, TN), Tulio Bertorini (Memphis, TN), Mariallan Shadle (Memphis, TN)

INTRODUCTION: Polymyositis is not a common autoimmune myopathy, except when associated with connective tissue disorder; this condition sometimes is difficult to differentiate from muscular dystrophies with prominent inflammation in the muscle biopsies.

OBJECTIVE: To analyze clinical and histological characteristics of patients with polymyositis, and to differentiate from patients with proven muscular dystrophies with prominent inflammation in muscle biopsies.

METHODS: This study entailed clinical evaluation, muscle biopsy studies with routine stains, and immunohistochemistry including staining for major histocompatibility complex-I (MHC-I) antigen in 6 patients with autoimmune myopathy and 6 with muscular dystrophies.

RESULTS: More chronic myopathic features were seen in biopsies of dystrophic patients, and MHC-I upregulation was patchy. Whereas, in autoimmune myopathies this was diffuse, but not always.

SUMMARY/CONCLUSION: Differentiation of the 2 conditions is difficult. Thus, in cases where the diagnosis is not clear, other studies such as western blot or DNA testing (e.g., for calpainopathy and dyserlinopathy) are helpful, even if there is no positive family history. However, clinical correlation and analysis of phenotype are of utmost importance.

Olivia Yambem, MD
Resident and Fellow Member Award Recipient

SAFETY OF PHENOXYBENZAMINE CHEMODENERVATION WITH REPEATED INJECTIONS
Roger Coletti (Lewes, DE)

INTRODUCTION: Chemodenervation with botulinum toxin A is known to cause weakness and atrophy with repeated injections. Alternative medications with a similar duration of action without this side effect would clearly be preferable. Phenoxybenzamine has been shown to be a chemodenervation agent useful in the treatment of chronic muscle spasm. The issue of its safety with repeated use has not been reported. Several hundred patients have been treated without evidence of weakness or atrophy when 1 or 2 injections were delivered to a given muscular region. However, no reports of the outcome of multiple injections have been published.

OBJECTIVE: To document the outcome regarding weakness and atrophy of a single known case of multiple injections to establish parameters of safety for this medication.

METHODS: A single patient following a two-level lumbar laminectomy developed severe post-laminectomy syndrome. An MRI at 3 months following surgery was interpreted as worse than pre-surgery. Needle EMG-guided chemodenervation was performed at approximately 2-week intervals with a total of greater than 10 injections for post-laminectomy syndrome to resolve.

RESULTS: A final MRI at 7 months post-surgery showed resolution of neural impingement and resolution of radiculopathy symptoms. No evidence of weakness or atrophy was observed on musculoskeletal physical examination.

SUMMARY/CONCLUSION: Initial results of multiple injections with phenoxybenzamine during needle EMG-guided chemodenervation suggest it may be safely used for repeated interventions. The known mechanism of action indicates this drug should not be toxic to muscle or nerve. Further investigation is needed to determine if the specific dose utilized played a significant role in this outcome.
THE DIAGNOSTIC ODYSSEY OF A FEMALE MANIFESTING CARRIER OF DUCHENNE MUSCULAR DYSTROPHY
Gaden Osborne (Albany, NY), Derreece Reid (Albany, NY), Valerie Arias (Albany, NY)

INTRODUCTION: Duchenne muscular dystrophy (DMD) is an X-linked recessive disorder characterized by progressive skeletal muscle weakness and atrophy secondary to dystrophin gene mutation. Muscle biopsy shows a myopathic pattern and abnormal dystrophin staining. DMD is typically diagnosed in young males. Most heterozygous female carriers are asymptomatic; 2.5-7.9% of these females are manifesting carriers (MCs) developing symptoms secondary to skewed X inactivation. We present a case of an MC, initially diagnosed with myositis after incomplete workup, leading to a delay in care.

CASE REPORT: On examination, a 37-year-old woman with 6 years of bilateral lower extremity pain and progressive weakness, elevated creatinine kinase, and transaminitis showed diffuse hyporeflexia and proximal weakness. She was diagnosed with inflammatory myopathy in 2009 due to: (1) MRI with focal edema several muscles, (2) needle EMG showing fibrillations and decreased amplitude at the biceps femoris and tibialis anterior, and (3) nonspecific muscle biopsy. Symptoms progressed despite steroid trial. A second muscle biopsy showed a mild noninflammatory myopathy and abnormal dystrophin stain. Recent genetic testing showed exons 46 through 55 deletion, a pathologic variant of DMD, diagnosing her as an MC. Later investigations revealed a positive family history for DMD.

SUMMARY/CONCLUSION: Similarities in muscle pathology may lead to misdiagnosis of DMD as myositis, delaying accurate diagnosis particularly in female MCs. Diagnosis may be further obscured if patients respond to steroids, which treats DMD and certain inflammatory myopathies. Our case emphasizes the importance of obtaining a family history, thorough physical examination, and laboratory workup including dystrophin staining and genetic testing, in this population.

LATE-ONSET MULTIPLE ACYL-COENZYME A DEHYDROGENASE DEFICIENCY NOT CAUSED BY ETFDH, ETFA OR ETFB GENE MUTATIONS
Diana Mnatsakanova (Pittsburgh, PA), Jose David Avila (Danville, PA), David Lacomis (Pittsburgh, PA)

INTRODUCTION: Multiple acyl-coenzyme A dehydrogenase deficiency (MADD) or glutaric aciduria type II is a rare, autosomal recessive disorder of fatty acid, amino acid, and choline metabolism. It is caused by mutations in the electron transfer flavoprotein dehydrogenase (ETFDH) gene or, less often, the alpha (ETFA) and beta (ETFB) subunits of the electron transfer protein. The late-onset form of the disease typically manifests with chronic weakness, exercise intolerance, and/or myalgia.

OBJECTIVE: To provide a clinical, biochemical, and pathological overview of a patient with diagnosis of MADD.

CASE REPORT: A 61-year-old woman presented with chronic multifocal pain involving the low back, hips, thighs, and knees. She also complained of progressive leg weakness, requiring a wheelchair. She denied arm weakness and bulbar deficits. There was no family history of neuromuscular disorders. Examination demonstrated mild bilateral hip flexor weakness. Serum creatine kinase was elevated (2240 IU/L [26-292]). EDX examination showed short duration motor unit action potentials in most leg and some proximal arm and thoracic paraspinal muscles, with no spontaneous activity. Muscle biopsy revealed changes consistent with a lipid storage myopathy. Urine organic acids demonstrated elevated ethylmalonate, methylsuccinate, and 2-hydroxyglutarate. Acylcarnitine analysis showed multiple increased species, from C4 to C18. Sequencing of ETFDH, ETFA, and ETFB exons and adjacent introns failed to demonstrate a causative mutation. She was treated with riboflavin without improvement.

SUMMARY/CONCLUSION: The clinical, biochemical, and pathological findings in this patient support the diagnosis of MADD. The genetic defect may be present in promoter or deep intronic regions of ETFDH, ETFA, and ETFB; riboflavin transporter; or other unidentified genes.

Diana Mnatsakanova, MD
Resident and Fellow Member Award Recipient
GENETIC TESTING IN LIMB-GIRDLE MUSCULAR DYSTROPHY: A SINGLE CENTER EXPERIENCE
Alexandru Olaru (Danville, PA), Jose David Avila (Danville, PA)

INTRODUCTION: Limb-girdle muscular dystrophy (LGMD) is a heterogeneous group of inherited myopathies characterized by progressive shoulder- and/or hip-girdle muscle weakness. Obtaining a genetic diagnosis is critical as it allows planning and surveillance of disease-specific complications, implementation of anticipatory care, cost-effective use of medical resources, participation in research, and accurate genetic counseling.

OBJECTIVE: To analyze the use of genetic testing in LGMD patients followed across a large rural health care system.

METHODS: This retrospective study included patients seen within Geisinger Health System from 2000 until 2016. Electronic medical records were searched for patients bearing a diagnosis of muscular dystrophy based on international classification of diseases (ICD) codes. Clinical, EDX, pathological, and genetic testing records were reviewed.

RESULTS: Forty-five patients were identified. There were 27 (60%) men and 18 (40%) women. Twenty-five patients (56%) had an elevated creatine kinase (CK), with a median value of 2304 IU/L. Six cases (13%) had a normal CK, and there were no data in the remaining 14. Twenty-six patients (58%) underwent EDX testing and muscle biopsy. Genetic testing was performed in 17 patients (38%) and was diagnostic only in 3 (7%). Five cases had variants of unknown significance, and results of other 3 are pending. Genetic testing was considered or discussed in additional 10 cases (22%) but was ultimately not performed.

SUMMARY/CONCLUSION: A large proportion of patients in our cohort did not have a genetic diagnosis. Genetic testing is likely underutilized. Other contributing factors include those lost to followup and cost of testing. Using an algorithmic approach to genetic testing may improve diagnostic outcomes.

Alexandru Olaru, MD
Resident and Fellow Member Award Recipient

JAPANESE-AMERICAN MAN PRESENTING WITH MDA5-POSITIVE DERMATOMYOSITIS WITH SCLERODERMA OVERLAP AND GLOMERULONEPHRITIS
Cina Sasannejad (Redondo Beach, CA), Marcus Cimino (Torrance, CA), Margaret Adler (Redondo Beach, CA), Luis Chui (Rancho Palos Verdes, CA)

INTRODUCTION: Anti-melanoma differentiation associated gene 5 (MDA5) antibodies are found in a minority of dermatomyositis (DM) patients with a phenotype that includes interstitial lung disease (ILD) and cutaneous ulcerations. Positive anti-MDA5 antibodies have been reported in patients with rheumatologic disease on only 2 prior occasions.

CASE REPORT: A 32-year-old Japanese-American man with history of cocaine use presented with a 1.5-year history of progressive weakness associated with 40-lb weight loss, arthralgias, dysphagia, discolored painful fingers, and Raynaud’s phenomenon. He was admitted for cardiogenic shock, ejection fraction 10-20%. Examination revealed painful distal finger pressure ulcers, erythematos palmar macules with sclerodactyly and gangrene; diffuse alopecia; and proximal muscle weakness. Laboratory results showed elevated aldolase, normal creatine kinase, and cryoglobulinemia. Anti-Scl-70, anti-ribonucleic acid-polymerase-III, anti-centromere, and anti-polymyositis/Scl antibodies were negative. Hepatitis C antibody was positive 1/3 times, but given an undetectable viral load, was deemed false-positive. Needle EMG/NCSs found diffuse, irritable myopathy with length-dependent axonal sensory neuropathy. Muscle biopsy found scattered perivascular T- and B-cells, with autophagic vacuoles on electron microscopy. Renal biopsy revealed glomerulonephritis with full-house immune complex mesangial deposition. Additional tests found upper esophageal sphincter dysfunction and calcified pelvic soft tissues. No evidence of ILD. Anti-MDA5 antibodies were detected. Finger ulcers, strength, and repeat needle EMG improved after 2 IV immunoglobulin courses.

SUMMARY/CONCLUSION: Anti-MDA5/scleroderma overlap with glomerulonephritis is a novel presentation of MDA5-DM. This case also contributes to pathologic findings in MDA5-DM as autophagic vacuoles are not well described. Lastly, the concurrent, new onset cardiomyopathy raises the possibility of its relationship with MDA5-DM.
MANAGEMENT OF WEIGHT LOSS AND ABDOMINAL FULLNESS WITH MEXILETINE IN MYOTONIC DYSTROPHY TYPE 1
Amit Sachdev (East Lansing, MI), Noha Aljehani (East Lansing, MI), Kemar Green (East Lansing, MI)

INTRODUCTION: Gastrointestinal (GI) symptoms are underappreciated in patients with myotonic dystrophy type 1 (DM1). Myotonia is an early, prominent symptom in DM1 affecting skeletal muscle and contributing to decreased dexterity, gait instability, oropharyngeal swallowing dysfunction, and muscle pain. Less well understood is the postulated smooth muscle dysfunction and decreased gastric motility in DM1. Mexiletine is commonly used to treat skeletal muscle myotonia in DM1. However, treatment with sodium-channel-blocking agents is accompanied by a high incidence of GI side effects due to induced smooth muscle dysfunction. Mexiletine inhibits nonadrenergic, noncholinergic mediated relaxation of the lower esophageal sphincter and gastric slow wave activity. To our knowledge, we report the first case of mexiletine-associated improvement of GI smooth muscle associated symptoms.

OBJECTIVE: To report the utility of mexiletine in the management of postprandial GI distress in DM1.

CASE REPORT: A 29-year-old male with drug metabolism and pharmacokinetics (DMPK) gene mediated DM1 reported minimal skeletal muscle myotonia in DM1. However, treatment with sodium-channel-blocking agents is accompanied by a high incidence of GI side effects due to induced smooth muscle dysfunction. Mexiletine inhibits nonadrenergic, noncholinergic mediated relaxation of the lower esophageal sphincter and gastric slow wave activity. To our knowledge, we report the first case of mexiletine-associated improvement of GI smooth muscle associated symptoms.

DYSPHAGIA IN A PATIENT WITH DERMATOMYOSITIS: IT'S NOT ALWAYS DUE TO THE UNDERLYING MUSCLE DISEASE
Jatinder Patti (Worcester, MA), Kate Daniello (Worcester, MA), Bella Isabelita (Worcester, WA), Lan Qin (Westborough, MA)

INTRODUCTION: Patients with dermatomyositis have increased incidence of cancer, most commonly adenocarcinoma of lung, breast, ovaries, stomach, pancreas, and bladder. Dermatomyositis as a paraneoplastic manifestation of tonsillar carcinoma is a rare occurrence.

OBJECTIVE: To discuss a rare case of dermatomyositis associated with tonsillar neoplasm in an African American woman.

METHODS: A 52-year-old African American woman presented with a 3-month history of progressive bilateral proximal arm and leg pain and weakness associated with an erythematous facial rash. Evaluation revealed elevation in creatine phosphokinase (CPK), negative anti-Jo-1 antibody, myopathic changes on needle EMG, and muscle biopsy showing characteristic features of dermatomyositis. Six months after treatment with prednisone and methotrexate, the patient reported new swallowing problems and intermittent hoarseness despite improved muscle strength and normalization of CPK. CT of the chest, abdomen, and pelvis along with pan-endoscopy were negative for malignancy. Barium swallow showed moderate esophageal motility dysfunction. Her dysphagia did not respond to prednisone and methotrexate. Approximately 1.5 years after presentation, she developed a right-sided neck mass; biopsy showed metastatic squamous cell carcinoma. Repeat pan-endoscopy revealed a right tonsillar carcinoma.

RESULTS: The patient underwent radical neck dissection and tonsillectomy followed by chemotherapy and radiation with improvement in swallowing difficulties.

SUMMARY/CONCLUSION: Tonsillar cancer is rarely associated with dermatomyositis. High suspicion of nasopharyngeal cancer should be maintained especially in myositis patients with dysphagia in whom routine evaluation for malignancy is unremarkable. Considering the rarity of this association, reporting of each new case is valuable in highlighting this association and prompting earlier detection.
PERIPHERAL NEUROPATHY AS THE PRESENTING FEATURE OF JACOB CREUTZFELDT DISEASE
Sandeep Rana (Pittsburgh, PA), Abhishek Purohit (Houston, TX), Laxmi Shah (Monroeville, PA), Kamaldeep Nandra (Cole Bay, SX), Malis Arwani (Pittsburgh, PA)

POOP INTRODUCTION: Creutzfeldt–Jakob disease (CJD) is a rare human prion disease that typically presents with rapidly progressive dementia, myoclonus, and ataxia. Electroencephalograms often have periodic sharp wave complexes, and diffusion-weighted MRI has been described as having T2 hyperintensities in the basal ganglia along with a cortical ribboning pattern.

OBJECTIVE: To present a unique case of CJD which initially presented with peripheral neuropathy followed thereafter by ophthalmoparesis and rapidly progressive dementia.

CASE REPORT: A 60-year-old Caucasian male on immunomodulatory therapy for rheumatoid arthritis presented with a 3-month history of burning dysesthesias in his feet. He did not have a history of diabetes mellitus or alcohol abuse. On examination he had sensory impairment in the stocking distribution and diminished reflexes consistent with diagnosis of peripheral neuropathy. Initial workup, including screening tests for common treatable causes for peripheral neuropathy (i.e., B12, thiamine level, serum protein electrophoresis, and thyroid function tests), was negative. NCSs and needle EMG revealed sensorimotor polyneuropathy with axonal and demyelinating features. One month later, he presented with diplopia and ataxia. Over the subsequent few months he developed rapidly progressive dementia which then led to his demise. He had “cortical ribboning” on MRI of the head, and cerebrospinal fluid analysis revealed positive 14-3-3 protein suggestive of CJD. His family reported that the patient’s mother had died of CJD, which had been confirmed on autopsy.

SUMMARY/CONCLUSION: Painful peripheral neuropathy can be a presenting feature of prion disease (i.e., CJD) and should be in the differential diagnosis when no cause for neuropathy is forthcoming.

BRACHIAL PLEXUS INJURY ASSOCIATED WITH EXTRACORPOREAL MEMBRANE OXYGENATION
Prachi Kale (White Plains, New York), Charlotte Zisman (Valhalla, NY), Jin Li (Scarsdale, NY), Brij Singh Ahluwalia (Valhalla, NY), Anila Thomas (Valhalla, NY)

BACKGROUND: Extracorporeal membrane oxygenation (ECMO), a cardiopulmonary support system, is commonly used to treat critically ill cardiac patients. Neurologic complications with ECMO include ischemic stroke and intracerebral hemorrhage. Brachial plexus injury is uncommon.

OBJECTIVE: To present 2 cases of brachial plexopathy after initiation of ECMO.

CASE REPORTS: Case 1: A 72-year-old man with cardiac arrest underwent placement of an Impella® (Abiomed, Danvers, Massachusetts) device in the groin. The Impella device was switched to ECMO via right axillary artery cannulation after which the patient had right upper extremity weakness with complete sensory loss. Case 2: A 54-year-old man with aortic dissection underwent placement of ECMO through right axillary artery cannulation after which the patient developed right upper extremity weakness with complete sensory loss. Needle EMG on Case 1 was consistent with right brachial plexopathy. The patient did not have MRI of the brachial plexus due to recent cardiac surgery. CT of the chest revealed no hematoma in the area of the plexus. This patient was lost to followup. Needle EMG on Case 2 was consistent with right brachial plexopathy. CT of the chest revealed a hematoma in the area of the brachial plexus, which was drained. Nine months after the injury, this patient continued to improve clinically and on needle EMG.

SUMMARY/CONCLUSION: We present 2 patients with brachial plexopathy after the initiation of ECMO via axillary artery cannulation. The mechanism of injury can include direct trauma to the plexus during axillary artery cannulation or compression by hematoma. One patient developed plexopathy from hematoma. Early recognition is important for prompt treatment of structural lesions to avoid permanent injury.
RARE ELECTRODIAGNOSTIC FINDINGS IN A PATIENT WITH WHIPLASH
Shannon Schultz (Hershey, PA), William Jens (Hershey, PA), Aiesha Ahmed (Hershey, PA)

INTRODUCTION: Parsonage–Turner syndrome is a rare acute immediate complication of autologous stem cell transplantation (ASCT) in multiple myeloma attributed to graft-versus-host disease, chemotherapy, and immunosuppression.

INTRODUCTION: Neuronal traction injuries are uncommon yet known complications of trauma. In whiplash, spinal accessory and long thoracic nerve injuries have been reported individually but rarely at the same time.

CASE DESCRIPTION: A 57-year-old male suffered a non-displaced C2 fracture, left T5-6 rib fractures, and a fracture of the left ulnar styloid after a motor vehicle accident. Weeks later, he developed pain and progressive weakness of his right upper extremity. MRI showed a very mild upper subscapular tear and right trapezius atrophy. Plexus imaging showed a normal plexus with subacute denervation in the right trapezius muscle. NCSs noted CTS unrelated to the presenting complaint. On needle EMG, chronic denervation was observed in the right serratus anterior muscle with active and chronic denervation in the trapezius muscle.

DISCUSSION: Whiplash is a soft tissue injury associated with hyperextension injuries; 300/100,000 whiplash victims will present to the ER with whiplash associated disorders (WADs), with up to 44% having symptoms for over 3 years. The presence of spinal accessory and long thoracic nerve injury is an incredibly rare complication of trauma, with only 1 case ever reported.

SUMMARY/CONCLUSION: The emergence of our patient’s weakness had increased his WAD grading to a 3; however, with rapid evaluation and good physical therapy, he is improving. Long thoracic and spinal accessory nerve injuries as well as brachial plexus injuries should remain within the differential following traction type damage to the cervical spine. Early diagnosis and treatment of these injuries has been shown to result in better functional outcomes.

GUILLAIN-BARRÉ SYNDROME RELATED TO PLASMODIUM FALCIPARUM MALARIA
Shorog Althubait (Montreal, QC), Rami Massie (Montreal, CA), Erin K. O’Ferrall (Montreal, CA)

INTRODUCTION: Guillain–Barré syndrome (GBS) is rarely preceded by parasitic infections. We present a patient with GBS triggered by Plasmodium falciparum malaria (PFM) without central nervous system involvement.

OBJECTIVE: GBS related to PFM is rare condition which needs to be seriously considered in travelers from malaria’s endemic regions.

CASE REPORT: A 26-year-old male returned from a trip to Burkina Faso. Six days after his trip, he developed fever, chills, and malaise with generalized fatigue shortly followed by severe weakness in both legs that progressed over 2 days. He had difficulty ambulating and reported numbness around the ankles. He was diagnosed with severe PFM by blood smear and antigen capture test with a 3.94% parasitemia level. That was complicated by disseminated intravascular coagulation. His examination showed areflexia in the arms and legs; proximal and distal weakness of the legs; and reduced vibration sensation in the toes. NCSs of the legs demonstrated prolonged distal motor latencies, mildly reduced motor amplitudes, and mildly reduced sensory conduction velocities. F waves were absent in the arms and legs. Cerebrospinal fluid analysis, MRI of the brain, and complete spine were unremarkable. Other infectious diseases were ruled out. At followup 1 month after his presentation, his reflexes had returned and his strength had partially improved. Needle EMG showed normalization of the NCSs with evidence of acute denervation and reinnervation. Needle EMG was consistent with a radiculoplexopathy.

SUMMARY/CONCLUSION: GBS related to PFM is rare condition which needs to be considered in patients who present with subacute weakness after returning from regions where malaria is endemic.
NERVE ELASTOGRAPHY: LITERATURE REVIEW AND PERSONAL EXPERIENCE
Mohamed A Bedewi (Alkharj, KSI), Daniele Coraci (Rome, IT), Luca Padua (Rome, IT)

INTRODUCTION: Nerve ultrasound (US) is a very important tool in a neurophysiological laboratory, allowing morphological characterization of a nerve lesion. In the last few years elastography (E-US) has been used for peripheral nervous system evaluation. Two types of E-US exist: strain and shear-wave. The first one is based on rhythmic vertical compression on the evaluated site, and the obtained image shows grading based on color difference of blue, green, and red. Shear-wave E-US quantifies the propagation of the waves in the examined tissues. These methods allow the evaluation of tissue hardness degree, especially by using the ratio between the pixel values of peripheral nerves.

OBJECTIVE: To assess if there is a difference in median nerve stiffness between the wrist and forearm by means of sonoelastography.

METHODS: We evaluated the median nerves of 10 asymptomatic subjects, on both sides, by means of strain and shear-wave E-US. We developed a post-processing imaging analysis to quantify the amount of blue, green, and red colors of median nerve picture.

RESULTS: We found significant lower levels of red and green colors in the wrist, in comparison with the forearm. These findings suggested higher stiffness of the median nerve at wrist. In 10 asymptomatic subjects an evaluation of nerve stiffness between the wrist and forearm was performed with shear-wave E-US. The data showed different elasticity in between the median nerve at the wrist and the median nerve at the forearm with both techniques.

CONCLUSION: Our study shows that E-US may add additional information about nerve condition. Further studies are needed to obtain an effective stiffness index and understand the real usefulness of E-US in peripheral neuropathies. AUTHORS: Should the highlighted text be changed to: The data showed similar elasticity in the 2 nerve segments.

POTENTIAL NEURAL PLASTICITY AND NERVE REMODELLING OF THE PERIPHERAL NERVOUS SYSTEM IN CHRONIC GIANT HEMOPHILIC PSEUDOTUMORS
Hunaid Hasan (Galveston, TX), Ahmad Yusuf Solaiman (Galveston, TX), Elena Shanina (Galveston, TX)

INTRODUCTION: Small acute hematomas in anatomical spaces produce dramatic peripheral nerve dysfunction secondary to compression/traction injury. Acute nerve elongation >6% affects nerve function and >11% leads to irreversible vascular nerve changes. Hemophilic pseudotumors of the upper extremities are extremely rare. Our patient presented with minimal neurological dysfunction despite giant chronic hemophilic pseudotumors.

OBJECTIVE: To understand the neurobiology and biomechanics of peripheral nerve remodeling post-mechanical stress.

CASE REPORT: A 29-year-old male with Hemophilia A presented with bilateral forearm masses since birth having expanded enormously over last 2 years, producing a 57-cm forearm circumference. Examination demonstrated normal left upper extremity motor/sensory function, mild right finger abductor weakness, and little finger hypoesthesia. EDX testing demonstrated right ulnar axonal neuropathy and active/chronic denervation changes in the abductor digiti minimi/first dorsal interosseous. Forearm MRI/MR neurography showed extensive bilateral mildly-enhancing masses (left 25x12x11 cm, right 19x16x14 cm) with osteolytic ulna and radius destruction and neurovascular displacement. Calculated median/ulnar nerve elongation on the right was 13/16% and the left was 8/10%, respectively. Biopsy showed chronic hemophilic hemorrhage.

DISCUSSION: Peripheral nerve injury and the subsequent remodeling process have distinct pathophysiology in acute and chronic settings. Our patient’s calculated nerve elongation/displacement by masses was expected to produce significant neurological dysfunction. He presented with minimal neurological symptoms suggesting different mechanisms in chronic versus acute injury. Both events cause nerve traction/compression; however, the adaptive and remodeling processes differentiate functional outcome in slow injury, with greater potential for neural plasticity and preservation of peripheral nerve function. Better understanding may help identify areas for intervention, promoting neurorehabilitation in the peripheral nervous system.
PROGNOSTIC FACTOR FOR AXONAL GUILLAIN BARRÉ SYNDROME
Seung Hwa Rhie (Seoul, KP), Dae Yul Kim (Seoul, KP), Eun Jung Sung (Seoul, KP)

INTRODUCTION: Acute motor axonal neuropathy (AMAN) is a pure axonal subtype of Guillain–Barré syndrome (GBS). Two patterns of recovery are seen in patients with AMAN. Some patients with AMAN recover within days, whereas others have slow and poor recovery.

OBJECTIVE: To find different factors between axonal GBS patients who can walk independently in a month and who cannot.

METHODS: We reviewed the records of 205 patients who were admitted with a diagnosis of GBS. Neurophysiological criteria for GBS according to Van den Bergh was used for sorting axonal type of GBS.

RESULTS: In this study, 28 patients (13.7%) were categorized with an axonal type of GBS according to criteria. Mean onset age was 51.4±13 years. Most patients had an infectious antecedent prior to the onset of weakness, the most frequent being gastrointestinal infection (44.4%). At 1 month after admission, 8 patients (30%) were able to walk for 10 meters without help, 11 (41%) needed assistance to walk, 4 (15%) were bedridden, and 4 (15%) needed respiratory support. Some factors including preceding infection, interval from symptom to IV immunoglobulin, and mechanical ventilation were analyzed as possible predictors of a poor outcome, but only GBS disability score at admission was related with independent gait at 1 month, and the Medical Research Council sum score tended to be associated with independent gait at 1 month.

SUMMARY/CONCLUSION: In our study we demonstrate that, even in the first day of admission, lower scores on the GBS disability score are associated with worse outcome.

LIMITATIONS ON THE USE OF ULTRASOUND AS A SCREENING ALGORITHM FOR CTS, A MINI-CASE SERIES
Matthew Drakeley (Pittsburgh, PA), John Frampton (Pittsburgh, PA), Michael Munin (Pittsburgh, PA)

INTRODUCTION: Recent literature has described an algorithm using ultrasound as a screening test for focal median mononeuropathy of the wrist (FMMW). Some argue that peripheral nerve ultrasound imaging (PN-USI) may replace EDX studies.

OBJECTIVE: To determine the utility of ultrasound as a screening modality for FMMW.

METHODS: Here we present 3 case reports of patients presenting with possible CTS who were evaluated with EDX studies and PN-USI. The median nerve diameter was measured at the carpal tunnel inlet and in the proximal forearm. All of these patients were evaluated in the same EMG laboratory in a large academic institution over a 2-month period.

RESULTS: All 3 cases had normal PN-USI but grossly abnormal EDX studies.

SUMMARY/CONCLUSION: To our knowledge, there are few similar case reports published in the literature and this clinical scenario may be more common than initially thought. These cases demonstrate that although PN-USI may be a useful as part of a multivariate analysis for CTS, it does not replace EDX testing as the gold standard.
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A NEW VARIANT OF CIDP WITH RESPIRATORY INSUFFICIENCY AND NOVEL ANTIBODIES WITHOUT POEMS SYNDROME: DO WE NEED TO LOOK BEYOND THE FENCE?
Harmanpreet Tiwana (Hershey, PA), Divpreet Kaur (Hershey, PA), Aiesha Ahmed (Hershey, PA), Sankar Bandopadhyay (Hershey, PA)

INTRODUCTION: Respiratory insufficiency due to diaphragm or intercostal muscles is rare in chronic inflammatory demyelinating polyneuropathy (CIDP), and is seen in association with POEMS syndrome (polyneuropathy, organomegaly, endocrinopathy, monoclonal gammopathy, and skin changes).

OBJECTIVE: To study an unusual case of CIDP with respiratory depression without associated POEMS syndrome.

CASE REPORT: A 71-year-old male without previous diagnosis of CIDP presented to the ICU in a central Pennsylvania tertiary care referral center with respiratory insufficiency and neck drop. He had 2 prior admissions for extremity weakness over the preceding 3 months, and received IV immunoglobulin (IVIg) infusions. A CIDP diagnosis was made based on clinical presentation, needle EMG, sural nerve biopsy, and cerebrospinal fluid examination. Increasing dyspnea mandated intubation and spontaneous mode ventilation. The patient also had autonomic dysfunction. As respiratory depression is rare in CIDP patients, other causes such as POEMS syndrome were explored. Whole body scanning showed no evidence of organomegaly. No evidence of endocrinopathy, monoclonal gammopathy, or skin changes were seen to support POEMS syndrome. He was started on steroids and plasma exchange, and improvement in arm and neck muscle strength followed. Neuronal potassium channel, striational antibodies, and paranodal antibodies including anti-contactin were positive.

SUMMARY/CONCLUSION: CIDP with severe respiratory embarrassment, autonomic dysfunction unrelated to POEMS syndrome, and the presence of anti-nodal and K-channel antibodies raise the possibility of new faces of this disease with protean manifestations and associations. Antibodies against contactin-1 and neurofascin-155 define specific disease subtypes distinct from patients with typical CIDP. Importantly, these patients have aggressive presentation and poor response to IVIg.

Harmanpreet Tiwana, MD
Resident and Fellow Member Award Recipient

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ELECTRODIAGNOSTIC STUDIES IN PATIENTS WITH ADULT ONSET GM2 GANGLIOSIDOSES
Tanya Lehky (Bethesda, MD), Katharine Alter (Bethesda, MD), Camilo Toro (Bethesda, MD), Cynthia Tifft (Bethesda, MD)

INTRODUCTION: Late-onset Tay–Sachs (LOTS) disease and Sandhoff disease (SD) are recessive lysosomal storage disorders due to mutations in hexosaminidase A (HEXA) (LOTS) or hexosaminidase B (HEXB) (SD) affecting different subunits of the enzyme β-hexosaminidase A. Adult variants often manifest as a lower motor neuronopathy (LMN) and sensory neuropathy (SN). SD can have an early-onset Fabry disease-like painful SN distinct from LOTS presentation.

OBJECTIVE: To undertake an electrophysiological and ultrasound characterization of the peripheral nervous system (PNS) and muscles of subjects with GM2 gangliosidosis (LOTS).

METHODS: Thirteen subjects with the diagnosis of adult-onset GM2 gangliosidosis were evaluated under protocol 02-HG-0107 (NCT00029965). Standard NCSs, needle EMG, and B-mode ultrasound of major muscle groups were performed.

RESULTS: Eight subjects (mean age: 39.8±9.8 years, range: 27-49 years) with LOTS and 5 with SD (57-41, mean 50.8±7.0) were evaluated. All had evidence of LMN, with the triceps (12/13 or 92%) and quadriceps (12/13 or 92%) the most affected. SN occurred in 80% of SD patients and 50% of LOTS. Sensory impairment was pronounced in those with SD and mild or asymptomatic in those with LOTS. Muscle ultrasound showed selective hyperechogenicity and atrophy of the triceps and deltoid and quadriceps and the tibialis anterior with relative sparing of the biceps, hamstrings, and gastrocnemius. Ultrasound findings paralleled clinical weakness.

SUMMARY/CONCLUSION: LOTS and SD have similar electrophysiological presentations of LMN and SN. SN in SD may be more complex with small fiber or posterior column involvement. Ultrasound further characterizes the unique distribution pattern of involvement in adult GM2 gangliosidosis preferentially targeting the triceps and quadriceps.
EARLY ELECTROPHYSIOLOGICAL FINDINGS IN ACUTE INFLAMMATORY DEMYELINATING POLYRADICULONEUROPATHY (AIDP) VARIANT OF GUILLAIN-BARRE SYNDROME IN COMPARISON WITH THE GLOBAL DATA
Ahmad Wali (Karachi, PK), Sara Khan (Karachi, LA), Duresshwar Kanwar (Karachi, LA), Safoora Ahmed (Karachi, LA)

INTRODUCTION: Acute inflammatory demyelinating polyradiculoneuropathy (AIDP) is the most common subtype of Guillain–Barré syndrome (GBS) in Pakistan. Electrophysiological findings in the first 2 weeks of presentation and their comparison to global data has not been studied.

OBJECTIVE: To validate and evaluate the variability of early neurophysiologic findings in patients clinically-suspected of AIDP.

METHODS: Charts of 87 patients, diagnosed with AIDP clinically and via NCSs, January 2010-July 2015, were studied. Excluded were patients with concomitant diseases that can cause neuropathies (i.e., diabetes, hypothyroidism, and B12 deficiency) and exposure to chemotherapeutic drugs.

RESULTS: The chart review showed (worst affected nerves in parentheses): (1) decreased compound muscle action potentials (CMAPs): 85% (posterior tibial), (2) slow conduction velocities: 75% (posterior tibial), and prolonged motor distal latency: 71% (median), (3) absent H reflexes: 65%, (4) temporal dispersion: 64% (ulnar), and conduction block: 52% (posterior tibial), (5) prolonged F waves: 39% (posterior tibial), (6) prolonged sensory latencies: 48% (median), (7) sural sparing pattern: 73% (p=0.016), (8) conduction block: 73%, and temporal dispersion: 64% in nerves with very low distal CMAPs, (9) abnormal blink reflex: 37%, and (10) (during needle EMG) spontaneous activity: 90% (p=0.039) and rapid firing rate motor units: 59% in those with very low CMAPs.

SUMMARY/CONCLUSION: The AIDP chart study revealed an increased frequency of abnormalities in CMAPs and sensory nerve action potentials, prolonged distal latencies, and temporal dispersion/conduction blocks. There was an increased frequency of a sural sparing pattern with no significant differences in conduction velocities and abnormalities of H reflexes/F responses. The blink reflex can help in the diagnosis of AIDP. A needle EMG and a followup study is recommended.

Ahmad Wali, MD
IFCN North American Chapter Fellowship Award Recipient

THE TALE OF TWO SOLDIERS REMEMBERED 101 YEARS LATER
Jose David Avila (Danville, PA)

INTRODUCTION: In October 1916 Georges Guillain, Jean Alexandre Barré, and André Strohl published their seminal paper describing 2 soldiers of the French Sixth Army with a disease now known as Guillain–Barré syndrome (GBS). Jean-Baptiste Landry reported a similar illness in 1859 and termed it “ascending paralysis.”

OBJECTIVE: To revise the 2 cases described by Guillain, Barré, and Strohl.

METHODS: Review of the original report from 1916 and other related historical articles.

RESULTS: Case 1 was a 25-year-old man with progressive paresthesia and limb weakness. Examination revealed distal predominant weakness. Case 2 was a 35-year-old man with pain, progressive limb weakness, and difficulty walking. Examination demonstrated weakness involving the face, legs, and distal arms. Both had areflexia and no objective sensory loss. Case 2 also had hypertonia, myoclonus, and a maculopapular erythematous rash. Ventilatory and autonomic dysfunction were not observed. Cerebrospinal fluid (CSF) analysis showed elevated protein without pleocytosis in both cases. Case 1 was treated with bed rest, muscle frictions, and several chemicals. Both patients improved over the following weeks. The authors highlighted the CSF findings and good prognosis of the disease. They suspected an infectious or toxic cause and postulated a simultaneous pathology of the spinal roots, peripheral nerves, and muscles.

SUMMARY/CONCLUSION: These cases illustrate the classic clinical presentation and CSF profile of GBS. The atypical features in case 2 may suggest a GBS mimic. The syndrome was considered separate from Landry’s ascending paralysis owing to its good prognosis. There is no uniform explanation as to why Strohl’s name was omitted in the eponym.
OSTEOCHONDROMA AS A CAUSE OF DEEP PERONEAL NERVE COMPRESSION: A CASE REPORT
Julia Reilly (Charlestown, MA), Jennifer Baima (Worcester, MA), Mathew Most (Worcester, MA)

OBJECTIVE: To demonstrate a case of a proximal lesion with limited effect of some distal muscles and more profound effect on the extensor hallucis longus (EHL).

CASE REPORT: A 15-year-old boy presented to clinic for evaluation of a right knee mass. He endorsed a history of prior patellar dislocations, most recently causing him to fall onto his lateral knee, prompting him to first notice the mass. He noted foot numbness and lower extremity weakness after this injury. He denied fevers, chills, night sweats, or unintentional weight loss. On examination, he had 0/5 strength of his right EHL and 4/5 strength of ankle dorsiflexion. Sensation was intact to light touch. Imaging revealed a mass along the lateral aspect of the fibular neck, thought to be a benign osteochondroma. A needle EMG was ordered prior to surgical intervention to assess the etiology of his weakness. Needle EMG findings were consistent with a deep peroneal axonal neuropathy, primarily involving the fascicles to the EHL. The patient underwent excision of the osteochondroma, and after 6 weeks his EHL was 4/5 on manual muscle testing.

SUMMARY/CONCLUSION: Nerve fibers of the peroneal nerve are at risk for injury in several locations along the lower extremity. A common location for injury is the fibular head, as the nerve may be tethered at the origin of the peroneus longus. This patient's osteochondroma was located at the fibular neck, and his primary symptom was EHL weakness. Needle EMG revealed the superficial peroneal nerve was spared and the lesion was distal to the bifurcation but affected that muscle preferentially.

BRACHIAL PLEXOPATHY AFTER CONTRALATERAL RADIATION FOR HEAD AND NECK CANCER: A CASE REPORT
Julia Reilly (Charlestown, MA), Jennifer Baima (Worcester, MA)

OBJECTIVE: To discuss potential causes of brachial plexopathy after cancer treatment.

CASE REPORT: A 58-year-old man with history of right-sided squamous cell carcinoma of the head and neck treated with radio chemotherapy presented to clinic with acute left shoulder pain and weakness. He was last treated with radiation 3 years prior to presentation, in 3 phases with maximum of 7560 cGy. His symptoms began with acute severe left shoulder pain, which progressed to weakness after 3 days. He denied any preceding trauma or illness. His examination was notable for atrophy of the deltoid, infraspinatus, and supraspinatus; decreased sensation over the lateral arm; and weakness with shoulder abduction and external rotation. A needle EMG was ordered, with findings consistent with a severe upper trunk lesion of the brachial plexus as no motor units could be identified in the deltoid and infraspinatus. No myokymic potentials were noted. Repeat needle EMG demonstrated resolution of abnormal spontaneous activity seen in the deltoid and infraspinatus and evidence of reinnervation. Brachial plexus MRI revealed bilateral increased signal in the roots and trunks. Fifteen months after presentation, his strength improved to near full.

SUMMARY/CONCLUSION: Idiopathic brachial neuritis occurs after trauma, stress, surgery, and other causes. Often, the etiology remains unknown. Radiation-induced plexopathy occurs several years after treatment, commonly involving the upper trunk; however, it usually does not present with pain, and myokymic discharges are typically noted on needle EMG. Although the workup was consistent with idiopathic brachial plexopathy, given bilateral findings on imaging, it is possible that the history of radiation contributed to his presentation.
THREE CASES OF FASCICULAR TORSION OF THE PROXIMAL NERVE IN THE UPPER EXTREMITY
Jae Lim Kim (Gangnam-gu, KP), Duk Hyun Sung (Gangnam-gu, KP)

INTRODUCTION: Anterior interosseous nerve (AIN) and posterior interosseous nerve (PIN) syndromes have been regarded as mechanical compressive entrapment neuropathies; however, fascicular torsion of proximal nerves without a space-occupying lesion has been reported.

OBJECTIVE: To report 2 cases of AIN syndrome and a case of PIN syndrome showing fascicular torsion of proximal nerves without compressive lesion.

CASE REPORTS: (1) A 32-year-old woman presented with left finger extensor weakness. Needle EMG demonstrated PIN syndrome. MRI showed 4 hourglass-like constrictions with a thickened and enhanced left radial nerve at the level between the lateral intermuscular septum and radial nerve bifurcation. Within the surgical field, some constrictions like torsion were found. (2) A 47-year-old man presented with left finger flexor weakness. Needle EMG demonstrated AIN syndrome. MRI showed enlargement and enhancement of a posterolateral fascicle of the median nerve above elbow level. Interfascicular neurolysis showed a discolored segment along the median nerve and 2 constriction sites (proximal pronator teres branch and distal AIN branch). (3) A 49-year-old woman presented with right finger flexor weakness. Needle EMG demonstrated AIN syndrome. On MRI, the right median nerve from 6.7 cm above the radiohumeral joint to the issuing point of the AIN was enlarged and enhanced like string beads. Her sister and aunt had prior decompression surgery for AIN syndrome.

SUMMARY/CONCLUSION: Nerve fascicular torsion within the upper arm level without a space-occupying lesion does not suggest compressive neuropathy. It is noteworthy that family history is observed in 1 patient, and only motor fascicles of proximal nerve were affected. It is proposed that a new etiology such as disimmunity causes AIN and PIN syndrome.

A CASE OF POSTPARTUM NEURALGIC AMYOTROPHY IN A PATIENT WITH A STRONG FAMILY HISTORY OF THE CONDITION
Jacob Halvorsen (Madison, WI), Larry Kim (Madison, WI), Bonnie Weigert (Madison, WI)

INTRODUCTION: Neuralgic amyotrophy (NA) is a rarely diagnosed condition thought to be an immune-mediated inflammation typically affecting the nerves of the brachial plexus. It often presents as a sudden-onset shoulder/arm pain followed by weakness and preceded by stressors such as a viral illness, trauma, or surgery.

OBJECTIVE: To describe the history and EDX findings of a postpartum NA patient with a strong family history and symptoms preceded by an abdominal illness.

RESULTS: A 23-year-old female with a 3-week history of a cesarean section requiring general anesthesia presented to the ER for 2 weeks of bilateral arm pain and weakness. At 11 days post-cesarean section, her course was complicated by an abdominal illness with vomiting and diarrhea lasting several days. Deep vein thrombosis was ruled out with bilateral upper extremity ultrasound, and the patient was sent home. For the next 6 weeks, she continued to have pain in her arms and weaknesses of her right thumb and left arm. A cervical MRI was negative for stenosis or radiculopathy. However, a needle EMG revealed severe denervation changes with high spontaneous activity and minimal motor unit potentials in her left triceps and right flexor pollicis longus muscles. Sampling of other left radial nerve and right anterior interosseous nerve-innervated muscles were normal. The patient endorsed a brother and a grandmother who were diagnosed with NA.

SUMMARY/CONCLUSION: The patient’s likely hereditary form of NA may have been triggered by multiple potential inciting factors, including a cesarean section requiring general anesthesia and a severe abdominal illness preceding the onset of her symptoms.
ULTRASOUND IN CONJUNCTION WITH ELECTRODIAGNOSIS IN THE DIAGNOSIS OF LATERAL FEMORAL CUTANEOUS NEUROPATHY (MERALGIA PARESTHETICA): A CASE SERIES

John Norbury (Greenville, NC), Eric Morrison (Greenville, NC), Hardeep Kainth (Greenville, NC)

INTRODUCTION: Lateral femoral cutaneous nerve (LFCN) neuropathy can be a challenging diagnosis in the EDX Laboratory due to technical challenges related to NCSs of the LFCN.

OBJECTIVE: To describe the utility of neuromuscular ultrasound (NMUS) in evaluating LFCN neuropathy.

METHODS: EDX studies and NMUS were performed on 2 patients with LFCN neuropathy.

RESULTS: Case 1 is a 68-year-old man status post right total hip arthroplasty resulting in complete loss of sensation in the lateral thigh. NCSs revealed an absent right LFCN response. NMUS revealed a right LFCN cross-sectional area (CSA) of 8.20 mm² and a left CSA of 3.10 mm². The nerve coursed much closer to the anterior superior iliac spine (ASIS) than on the left, and there was no evidence of nerve transection. Case 2 is a 41-year-old man who presented with left thigh numbness. NMUS was used to localize the nerve for reliable NCSs, which showed an absent left LFCN response. NMUS revealed a CSA of 3.14 mm² on the right and 11.16 mm² on the left, with the left LFCN also traveling in close proximity to the ASIS as in Case 1.

SUMMARY/CONCLUSION: NMUS is a useful adjuvant for both guiding NCs and determining the location of entrapment in a LFCN neuropathy. NMUS can improve the reliability of EDX data, localize the site of entrapment, and rule out nerve transection. Further research is needed to determine normative data for the LFCN and whether proximity to the ASIS may be associated with a LFCN neuropathy.

ACUTE AUTONOMIC AND SENSORY NEUROPATHY. A CASE SERIES

Joel Gutierrez (La Habana, CU), Jose-Alberto Palma (New York, NY), Carlos Mendoza (New York, NY), Horacio Kaufmann (New York, NY)

INTRODUCTION: Acute autonomic and sensory neuropathy (AASN) is considered a very infrequent variant of immune-mediated acute peripheral neuropathy/ganglionopathy. The number of cases documented in the literature is very scarce.

OBJECTIVE: To present a series of patients with AASN.

METHODS: NCSs, cardiovascular autonomic reflexes, spinal cord MRI, and standardized neurological examinations were evaluated in 3 previously healthy subjects (11-year-old male, 11-year-old female, and 37-year-old female), all of Asian ancestry, who presented with acute and severe sensory and autonomic deficits shortly after a minor infectious disease.

RESULTS: Autonomic disturbances included vomiting, diarrhea, anhidrosis, abdominal cramps, neurogenic pain, dry mouth and eyes, and dizziness upon standing and syncope. Sensory disorders included decreased perception for all sensory modalities with widespread patchy distribution and significant sensory ataxia. Impaired motor control with dysphagia was interpreted as reduced muscle power and prompted an early diagnosis of Guillain–Barré syndrome. Symptoms progressed for a few days and later stabilized. Neurological examination revealed very depressed or absent deep tendon, corneal, and gag reflexes with preserved muscle power. NCSs showed extremely reduced or absent sensory nerve action potentials with normal motor nerve conduction. Cardiovascular autonomic evaluation showed decreased heart rate variability, orthostatic hypotension without compensatory tachycardia, and very low or absent plasma norepinephrine levels. Spinal cord MRI showed extensive T2 hyperintensities of the posterior cords. One year after onset, the recovery was very poor.

SUMMARY/CONCLUSION: AASN is a very disabling disease which is frequently misdiagnosed. This is the second largest case series ever reported of patients with AASN.

Joel Gutierrez, MD, PhD
IFCN North American Chapter Fellowship Award Recipient
PHENOTYPES OF LATE-ONSET TRANSTHYRETIN AMYLOID NEUROPATHY
Diana Mnatsakanova (Pittsburgh, PA), David Lacomis (Pittsburgh, PA), Sasha Zivkovic (Pittsburgh, PA)

INTRODUCTION: Transthyretin amyloidosis is a rare cause of severe neuropathy, typically manifesting with progressive sensorimotor and autonomic neuropathy.

OBJECTIVE: To describe clinical phenotypes of late-onset transthyretin amyloid neuropathy.

CASE REPORT: Four patients (mean age: 69 years; range: 65-76 years) presented with progressive sensorimotor neuropathy without dysautonomia. EDX testing showed demyelinating polyneuropathy (2), asymmetric axonal polyneuropathy (1), and mononeuritis multiplex (1). Cerebrospinal fluid (CSF) analysis showed elevated CSF protein in 2 patients (range: 117-169 mg/dl). Nerve biopsy showed perivascular inflammation and multifocal axon loss in 1 patient and diffuse axon loss in another patient with negative amyloid stains. Muscle biopsy showed amyloid in 1 of 2 patients. Two patients were initially diagnosed with chronic inflammatory demyelinating polyneuropathy (CIDP) and 1 with probable vasculitic neuropathy, but they did not respond to treatment with IV immunoglobulin (2), plasma exchange (1), corticosteroids (2), and steroid-sparing agents (1). Genetic testing showed mutations in transthyretin (TTR) gene, including V30M (2), T60A (1), and P64L (1). Subsequently, histopathologic confirmation was obtained with deeper cuts on the nerve biopsy in 1 patient, and amyloid cardiomyopathy was demonstrated in 3 patients. One patient passed away at the age of 80, 6 years after the onset of neuropathy.

SUMMARY/CONCLUSION: Late-onset hereditary transthyretin amyloid neuropathy may mimic other acquired polyneuropathies including CIDP and vasculitic neuropathy and should be considered if there is no response to treatment. Dysautonomia may present late and may not be among the presenting features of neuropathy. High level of suspicion is required as nerve biopsy may be false-negative. Genetic testing may be needed to establish the diagnosis.

ACUTE UNILATERAL HYPOGLOSSAL PALSY AS A RESULT OF COMPRESSION BY THE POSTERIOR INFERIOR CEREBELLAR ARTERY
Hani Kushlaf (Cincinnati, OH)

INTRODUCTION: Acute hypoglossal palsy is rare. A vascular etiology such as internal carotid or vertebral artery dissection can be considered. A hypoglossal vertebral entrapment syndrome has been described; however, there are no prior reports of compression of the hypoglossal nerve by the posterior inferior cerebellar artery (PICA).

OBJECTIVE: To report the clinical presentation and results of diagnostic testing of a patient with acute unilateral hypoglossal palsy as a result of compression by PICA.

METHODS: A 33-year-old woman presented with sudden onset of slurring of speech. Over a few days, she noted that the right side of her tongue is smaller and twitching. She had no preceding febrile or flu like illness or a diarrheal illness. Neurologic examination revealed an atrophic right tongue with fasciculations, and right tongue deviation with tongue protrusion outside the mouth. She had only mild weakness with pushing into left cheek and no facial, bulbar, or limb weakness apart from the tongue weakness. Reflexes and sensory examination were normal. EDX testing revealed normal NCSs of the right arm and leg and normal needle EMG of the right arm, right leg, and paraspinal muscles. Decreased recruitment with fasciculations were observed in the right genioglossus muscle. MRI and MR angiography (MRA) of the brain with and without contrast showed compression of the right cisternal segment of the hypoglossal nerve by PICA and a decrease in its caliber at the site of compression.

SUMMARY/CONCLUSION: Acute unilateral hypoglossal palsy is rare; compression by PICA adds to its list of causes.
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POSTERIOR INTEROSSEOUS NEUROPATHY DUE TO SUPINATOR HYPERTROPHY IN A PATIENT WITH STEREOTYPICAL HAND MOVEMENTS AND AUTISTIC SPECTRUM DISORDER-CLINICAL, ELECTROPHYSIOLOGICAL AND MAGNETIC RESONANCE NEURO
Lauren Tucker (Columbia, MO), Raghav Govindarajan (Columbia, MO)

BACKGROUND: The localization and etiology of posterior interosseous nerve/neuropathy (PIN) remain highly diverse and at times controversial. Recent studies have emphasized the role of imaging to supplement electrodiagnosis. We report a case of PIN, correlated by EDX testing and imaging studies, caused by selective hypertrophy of the supinator due to repetitive stereotypical hand movement in a patient with autistic spectrum disorder.

CASE REPORT: A 20-year-old lady with autistic spectrum disorder was brought in for assessment after the mother noticed her daughter was no longer doing her stereotypical hand movements (rhythmic supination and elbow flexion) over the last month. Physical examination showed dorsoradial deviation of the wrist and 0/5 strength in finger extensors. Radial sensory testing with pin prick was normal. An NCS showed a normal radial sensory result, but the radial motor amplitude from the extensor digitorum communis was reduced (60% reduction as compared to the left). Radial sensory testing with pin prick was normal. An NCS showed a normal radial sensory result, but the radial motor amplitude from the extensor digitorum communis was reduced (60% reduction as compared to the left). Needle EMG showed +2 fibrillation and positive waves in the extensor carpi ulnaris, extensor digitorum communis, and extensor indicis proprius, with normal studies in the triceps, brachioradialis, supinator, and extensor carpi radialis longus localizing the lesion to the PIN. MR neurographic imaging of the radial nerve showed hypertrophy of the supinator with increased T2-weighted signal within the PIN just distal to its piercing of the supinator, confirming the EDX localization and providing a clue to its etiology.

CONCLUSION: Stereotypical hand movements can result in selective supinator hypertrophy and cause PIN, and they should be considered in the differential diagnosis especially in autistic spectrum disorders. MR neurography adds to the localization of the lesion and provides a clue to underlying etiology.

Lauren Tucker, BS
Resident and Fellow Member Award Recipient

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AN UNCOMMON CASE OF MULTIPLE SCLEROSIS AND GUILLAIN-BARRE SYNDROME OVERLAP: CLINICAL, ELECTRODIAGNOSIS AND NEUROIMAGING CASE STUDY
Chakrapani Pathikonda (Columbia, MO), Raghav Govindarajan (Columbia, MO)

BACKGROUND: There are very few cases reported of an overlap between Guillain–Barré syndrome (GBS) and multiple sclerosis (MS). We report a case of a patient who presented with rapidly ascending numbness and weakness and was diagnosed with acute inflammatory demyelinating polyneuropathy (AIDP). Three months later he returned with recurrent episodes of hemibody numbness and was diagnosed with MS.

CASE STUDY: A 32-year-old male presented with a weeklong history of ascending numbness and tingling. Examination showed 1+ ankle reflex bilaterally and muscles strength of 3/5 in the lower limbs and 4/5 in the upper limbs. Vibration sense was impaired in the feet. Needle EMG/NCSs showed absent H reflexes and prolonged F-wave latency. Spinal tap showed 0 white blood count or cells (WBC) and protein 70 mg/dl. He was treated with IV immunoglobulin for AIDP. He recovered completely in 6 weeks when he developed right hemibody numbness. He came back to the ER and was thought to have recurrence or a relapse of GBS symptoms (although different from the initial presentation) and underwent spinal tap which showed normal protein and WBC count. Needle EMG/NCSs were normal. MRI of the brain with or without contrast showed multiple open ring enhancing lesions in the frontal and parietal cortices along with T2-weighted hyperintensities (non-enhancing) perpendicular to the lateral ventricles fulfilling the neuroimaging criteria for MS. MRI of the rest of the neuraxis was normal.

CONCLUSION: Overlap of GBS with MS, although rare, should be considered as a differential in those patients who present with recurrent or relapsing symptoms of GBS or in those whose presentation is atypical for GBS (such as hemibody numbness).

Chakrapani Pathikonda, BS
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CLINICAL DIFFERENCES OF DIABETIC POLYNEUROPATHY OR CARPAL TUNNEL SYNDROME IN PATIENTS WITH DIABETES
Seung Hwa Rhie (Seoul, KP), Dae Yul Kim (Seoul, KP), Seoyon Yang (Seoul, KP)

INTRODUCTION: Patients with diabetes often suffer from diabetic polyneuropathy (DPN) and CTS. NCSs are a useful EDX method for detecting damage to large nerve fibers. NCSs help to make the accurate diagnosis of DPN or CTS. Though some studies have tried to determine possible risk factors which cause patients to develop DPN or CTS, it has not yet been investigated.

OBJECTIVE: To investigate whether there are clinical differences in patients with DPN or CTS.

METHODS: We reviewed 587 patients with diabetes mellitus who had an EDX study to investigate whether they had DPN or CTS. The patients were divided into DPN, CTS, and DPN+CTS groups. General characteristics, diabetes-related factors, and associated complications were compared between these groups.

RESULTS: Of the 587 diabetic patients, 111 patients had DPN, CTS, or both. Among those, 47 had DPN (42.3%), 29 had CTS (26.1%), and 35 had both (31.5%). The results showed that the duration of diabetes was associated with DPN. Patients who had both DPN and CTS had higher levels of hemoglobin A1c (HbA1c), PP2, and albumin/creatinine ratio as well as a longer duration of diabetes than the patients who had only DPN or CTS.

SUMMARY/CONCLUSION: These results suggest that diabetic patients with both DPN and CTS showed a higher level of HbA1c and PP2 and a longer duration of diabetes or renal complications, more so than seen in those who have only DPN or CTS. Therefore, NCSs are recommended for diabetic patients who have these risk factors in order to be screened for both DPN and CTS.

CASE REPORT OF INTERDIGITAL NEUROPATHY AFTER CARPAL TUNNEL RELEASE
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INTRODUCTION: CTS is the most common reason for EDX testing. EDX studies often follow carpal tunnel release. Complications after carpal tunnel release may include interdigital neuropathy.

OBJECTIVE: To describe a case of interdigital neuropathy after carpal tunnel release diagnosed via EDX studies.

CASE REPORT: A 38-year-old hairdresser with a past medical history of mixed connective tissue disease presented for EDX evaluation of left middle and ring finger numbness. Previously, the patient had been diagnosed with severe median mononeuropathy at the wrist via EDX studies 2 years prior. Subsequent to that, she underwent a carpal tunnel release. Postoperatively, her CTS symptoms improved significantly; however, she reported persistent middle and ring finger numbness. A repeat EDX study with standard NCSs was performed. Findings showed normal median and ulnar sensory and motor nerve latencies, amplitudes, and conduction velocities at the wrist. Due to difficulty examining for interdigital neuropathy in small branches via standard ring electrodes, special interdigital electrodes were obtained.

RESULTS: NCSs revealed axonal neuropathy of interdigital nerves of the middle and ring fingers, with normal findings in the index and middle fingers and the ring and little fingers. Contralateral interdigital NCSs of the middle and ring fingers showed normal amplitude.

SUMMARY/CONCLUSION: Interdigital neuropathy is challenging to diagnose with standard ring or disc electrodes. Special interdigital electrodes can assist in the evaluation and diagnosis of interdigital neuropathy.
**CASE OF BILATERAL ISCHEMIC LUMBOSACRAL PLEXOPATHY AFTER THE USE OF EXTRACORPOREAL MEMBRANE OXYGENATION (ECMO)**

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**INTRODUCTION:** Ischemic lumbosacral plexopathy (LSP) is a rare but known complication following aortoiliac vascular interventions. However, bilateral ischemic LSP due to circulatory compromise has not been well described.

**CASE REPORT:** A 52-year-old woman with history of systemic lupus erythematosus presented with respiratory failure and cardiogenic shock. Venoarterial extracorporeal membrane oxygenation (ECMO) was initiated due to persistent hypoxemia. After extubation, the patient complained of severe lower extremity pain and weakness. There were no bulbar or upper extremity deficits. Her neurological examination and EDX evaluation were consistent with bilateral LSP. Imaging of the spine, lumbosacral plexus, and pelvis was non-revealing. Cerebrospinal fluid analysis was unremarkable.

**RESULTS:** The lumbar plexus receives blood supply directly from the lumbar arteries as well as from the blood supply to the psoas major muscle in which the lumbar plexus is imbedded. Four cases of unilateral ischemic LSP were reported following kidney transplantation. A single case of bilateral LSP was reported after hypovolemic shock and cardiac arrest. There have been no reported cases in association with prolonged hypoxemia or the use of ECMO. We believe that our patient developed bilateral ischemic LSP secondary to circulatory and tissue oxygenation compromise along with preexisting factors such as small vessel atherosclerosis and possible hypercoagulable state.

**SUMMARY/CONCLUSION:** Ischemic LSP should be in the differential diagnosis in patients who develop weakness after circulatory collapse. The diagnosis can be supported by the use of EDX studies along with imaging to rule out other causes of LSP.

**IMPROVEMENT IN ANATOMICAL MANIFESTATIONS IN CARPAL TUNNEL SYNDROME PATIENTS AFTER TREATMENT WITH ONABOTULINUMTOXINA**

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**INTRODUCTION:** CTS is caused by mechanical compression of the median nerve within the carpal tunnel. Neuromuscular ultrasound (NMUS) has identified median nerve edema and thenar muscle compression of the nerve in CTS. Previous studies demonstrate that Botox®/onabotulinumtoxinA (Onabot) reduces thenar muscle size and contractile activity, resulting in decreased nerve compression.

**OBJECTIVE:** To determine if CTS patients with thenar muscle compression of the median nerve will show a significant decrease in compression and decreased nerve edema after Onabot injections.

**METHODS:** NMUS was performed on 10 patients (5 Onabot, 5 placebo) with mild-to-moderate bilateral CTS after non-dominant hands were injected with 40 units of Onabot or 0.4 cc normal saline into the abductor pollicis brevis and opponens pollicis muscles. Median nerve compression was evaluated by NMUS at baseline and 6, 12, and 18 weeks.

**RESULTS:** At 18 weeks, the median nerve had a decreased mean cross-sectional area of −0.6 mm² in the placebo group compared to −2.2 mm² in the Onabot group (p=0.040). Clinical significance was observed during stress testing at 18 weeks; there was a decreased mean compression of the median nerve of −4.7% compression in the placebo group compared to −12.6% compression in the Onabot treatment group (p=0.19). Compression for the Onabot group demonstrated a −20.5% median change, while the placebo group demonstrated no median change.

**SUMMARY/CONCLUSION:** Improvement in anatomical manifestations was statistically and clinically significant among the Onabot group. Additional studies with a higher subject number are required to confirm the efficacy of Onabot injections for CTS as a viable nonsurgical treatment.
ACQUIRED NEUROMYOTONIA, A PARANEOPLASTIC PHENOMENON LEADING TO THE DIAGNOSIS OF HODGKIN'S LYMPHOMA
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INTRODUCTION: Neuromyotonia is a rare disorder of peripheral nerve hyperexcitability characterized by continuous muscle fiber activity that translates into impaired muscle relaxation and clinical muscle twitches, cramps, and spasms. This disorder is thought to be caused by dysfunctional neuronal potassium channels and may be seen in association with autoimmune or paraneoplastic conditions.

OBJECTIVE: To present a case of a patient with neuromyotonia associated with Hodgkin’s lymphoma and to emphasize the importance of exhaustive workup for occult malignancy.

CASE REPORT: A 67-year-old woman complained of frequent muscle spasms and cramps involving her face, trunk, and extremities occurring spontaneously or with movement. She had no family history of similar symptoms. Neurological examination was notable for normal strength and involuntary flexion of the fingers spontaneously and after manual muscle testing. Needle EMG demonstrated spontaneous bursts of rapid firing (200-300 Hz) single motor unit potentials consistent with neuromyotonic discharges in several upper limb muscles. Extensive laboratory evaluation, including a paraneoplastic panel, and initial cancer surveillance were negative. Repeat CT scan 6 months later revealed a mass in the upper abdomen with pathology consistent with Hodgkin’s lymphoma. She was successfully treated with chemotherapy and has remained in complete remission now for over 2 years. Despite this, she continues to have neuromyotonia which is disabling without the use of carbamazepine.

CONCLUSION: Oncological evaluation is imperative in patients presenting with acquired neuromyotonia, and ongoing cancer surveillance should be pursued even if initial malignancy workup is unrevealing. Neuromyotonia may persist despite treatment of the underlying cancer.

Rocio Vazquez do Campo, MD
Resident and Fellow Member Award Recipient

PHRENIC NEUROPATHY IN HERPES ZOSTER INFECTION
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INTRODUCTION: Herpes zoster is a common infection caused by varicella zoster virus reactivation from latently infected ganglions along the entire neuraxis. Motor complications resulting in motor weakness is a rare complication of herpes zoster.

CASE REPORT: We describe 2 patients who developed ipsilateral phrenic nerve paralysis following herpes zoster in cervical dermatomes. The first was a 73-year-old man who developed a painful vesicular rash over his right arm and lateral forearm. He had no weakness or numbness. Five days after onset of the rash, he noticed dyspnea and orthopnea. On examination 3 weeks after onset, he had a vesicular rash along the right C5 and C6 dermatomes. His motor strength was normal. His right biceps and brachioradialis reflexes were depressed. The second patient was a 61-year-old man who developed a rash over the right shoulder and scapular area, with shortness of breath noted a week later. Dyspnea and orthopnea persisted. On examination 6 months after onset, he had residual scarring over the right scapula. Motor examination, sensory testing, and reflexes were normal. Both had an elevated hemidiaphragm on chest X-rays and no movement of hemidiaphragm on fluoroscopy. On NCSs, both had absent phrenic motor responses on the symptomatic side. Pulmonary function testing showed forced vital capacities of 2.77 liters and 3.67 liters (58% and 68% of predicted, respectively).

SUMMARY/CONCLUSION: Phrenic nerve palsy should be added to the list of causes of unilateral phrenic paralysis and considered in patients with dyspnea/orthopnea occurring shortly after herpes zoster infection of the upper limb.
INFLAMMATORY POLYRADICULOPATHY, A RARE VARIANT OF CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY

Thananan Thammongkolchai (Cleveland, OH), Bashar Katirji (Cleveland, OH)

INTRODUCTION: Chronic Inflammatory Demyelinating Polyneuropathy (CIDP) is a rare disease with several variants.

OBJECTIVE: To report cases with a focal variant of CIDP presenting with a sensorimotor polyradiculopathy variant who responded to IV immunoglobulin (IVIg).

CASE REPORT: Three woman, aged 69, 63, and 37 years, presented with progressive predominantly proximal weakness and numbness in both legs for 3 months to 3 years. Two had burning leg pain, up to the calf in 1 and to the torso in the other. Two had lumbosacral radicular pain. All had absent deep tendon reflexes in the legs. On EDX studies, all patients had normal sensory NCSs in the lower extremities. Needle EMG showed active denervation and reinnervation in several muscles in the lower extremities and cervical and lumbar paraspinal muscles. MRI of the spine with contrast demonstrated diffuse enhancement of cauda equina in 2 patients, which disappeared after treatment in 1. Cerebrospinal fluid examination revealed elevated protein (71-166 mg/dl) with no pleocytosis. There was no evidence of infection nor malignancy. All patients had dramatic response to pulse IVIg (60-100 gm every 3 weeks) after 2-3 courses. One patient received prednisone and azathioprine when IVIg was interrupted for 1 year.

SUMMARY/CONCLUSION: It is challenging to diagnose focal polyradiculopathy variants of CIDP due to the lack of demyelinating features on NCSs. The main clue would be preserved sensory nerve action potentials, needle EMG evidence of polyradiculopathy, enhancing roots on MRI, and no evidence of inflammatory, infectious, or neoplastic etiology. Relying on EDX criteria only one would not be able to diagnose these patients.

Thananan Thammongkolchai, MD
Resident and Fellow Member Award Recipient

FOLLOW-UP STUDY OF NERVE ULTRASOUND IN A PATIENT WITH PRIMARY NEUROLYMPHOMATOSIS

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INTRODUCTION: The followup study of nerve ultrasonography in patients with neurolymphomatosis has not been reported.

OBJECTIVE: To highlight the potential usefulness of nerve ultrasonography to identify and monitor peripheral nerve infiltration in patients with lymphoma.

CASE REPORT: We performed peripheral nerve ultrasonography in a patient with primary neurolymphomatosis before and after chemotherapy. The patient was a 54-year-old female with a 5-month history of asymmetric limb pain, paresthesia, and weakness. EDX studies and spinal cord MRI showed an axonal neuropathy involving the bilateral cervical and lumbosacral roots, brachial plexus, and left median nerve. Detection of malignant B lymphocytes by cytology and flow cytometry of cerebral spinal fluid confirmed the diagnosis of B-cell non-Hodgkin lymphoma. Nerve ultrasound showed dramatic enlargement of the upper, middle, and lower trunks of the left brachial plexus (cross sectional area [CSA] was 21 mm2, 30 mm2, and 26 mm2, respectively), middle trunk of the right brachial plexus (CSA 15 mm2), and proximal part of the left median nerve (CSA 15-18 mm2). Five months later, after 5 rounds of chemotherapy with rituximab and high-dose methotrexate, as well as intrathecal injection of cytosine arabinoside and dexamethasone, the patient had clinical improvement. Nerve ultrasound also showed alleviation of nerve enlargement. The CSAs of the upper, middle, and lower trunks of the left brachial plexus were 5 mm2, 14 mm2, and 14 mm2, respectively; the CSA of the middle trunk of the right brachial plexus was 11 mm2; the CSA of the proximal part of the left median nerve was 12-13 mm2.

SUMMARY/CONCLUSION: Peripheral nerve ultrasound could help locate the distribution of nerve involvement and reveal disease progression.
INTRAVENOUS IMMUNOGLOBULIN (IVIG) TREATMENT-RELATED FLUCTUATIONS IN CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY PATIENTS USING DAILY GRIP STRENGTH MEASUREMENTS: STUDY DESIGN AND JEFFREY ALLEN (MINNEAPOLIS, MN), MAMATHA PASNOOR (KANSAS CITY, KS), TED BURNS (CHARLOTTESVILLE, VA), SENDA AJOUD-DRISS (CHICAGO, IL), JOHN NEY (NEWTON, MA), ALBERT COOK (ATLANTA, GA), THOMAS BRANNAGAN (NEW YORK, NY), VICTORIA LAWSON (LEBANON, NH), JOHN KISSEL (COLUMBUS, OH), KENNETH GORSON (WELLESLEY, MA), RICHARD LEWIS (LOS ANGELES, CA), STACEY JENSEN (LENEXA, KS), TIMOTHY WALTON (LENEXA, KS)

INTRODUCTION: The optimal treatment approach for chronic inflammatory demyelinating polyneuropathy (CIDP) patients on chronic therapy is unknown.

OBJECTIVE: To describe an investigator-initiated, multicenter study that explores IV immunoglobulin (IVIg) treatment-related fluctuations in CIDP.

METHODS: The primary outcome measure is JAMAR (Lafayette Instruments, Lafayette, Indiana) grip strength (GS), performed daily for 6 months. Home nursing visits also capture results of the Rasch-built Overall Disability Scale (R-ODS), Timed Up and Go Test (TUG), Overall Neuropathy Limitations Scale (ONLS), Modified Fatigue Severity Scale (mFSS), and a visual analog pain severity scale (VAS) weekly for 6 months. Serum IgG levels are collected at 3 time-points surrounding IVIg infusions (peak, trough, and mid-cycle). Total recruitment of 30 subjects is anticipated. Upon study completion, “wear-off” frequency will be analyzed by assessing the proportion of subjects with GS and R-ODS intracycle fluctuation and the proportion of cycles in which GS and R-ODS fluctuation occurs. To determine the extent of “wear-off” maximum and minimum GS, R-ODS, TUGs, ONLS, and VAS scores will be analyzed.

RESULTS: Currently, 22 subjects from 4 sites have been enrolled (7 sites eligible for enrollment). This interim study report will provide preliminary representative data, demonstrating IVIg “wear-off” effects on GS and other outcome measures.

SUMMARY/CONCLUSION: By better understanding IVIg treatment-related fluctuations we expect that these results will facilitate development of CIDP treatment optimization strategies. We also expect that this information will be important in forming hypotheses to be tested in future studies (e.g., comparing different dosage intervals, optimal IVIg taper guidelines, or assessing the longterm outcome of short-term cycle to cycle clinical fluctuations).

UNUSUAL EMG/NCS AND ULTRASOUND FEATURES IN A PATIENT WITH NEUROPATHY ASSOCIATED WITH A SPTLC1 VARIANT
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INTRODUCTION: Hereditary sensory neuropathy type 1A (HSN1A) is an autosomal dominant disorder caused by mutations in serine palmitoyltransferase, long-chain base subunit 1 (SPTLC1). While severe sensory symptoms are characteristic of this neuropathy, patients can also have significant distal weakness. EDX studies typically show a length-dependent sensorimotor neuropathy with normal or intermediate velocities.

OBJECTIVE: To present EDX and ultrasonographic findings demonstrating proximal demyelination/dysmyelination and distal axonal loss in a patient with a SPTLC1 variant.

CASE REPORT: The patient is an 18-year-old woman with a family history of neuropathy who presented with “pins and needles” in her extremities starting around age 4 and progressive weakness of lower greater than upper extremities. Neurological examination showed distal-predominant weakness, tapered extremities (“licked candy-stick” appearance) without mutilation, distal sensory loss, and areflexia. Needle EMG/NCSs showed a symmetric, length-dependent, sensory-predominant sensorimotor, primarily demyelinating polyneuropathy. Interestingly, the demyelination was more prominent proximally compared to distally. On neuromuscular ultrasound, more proximal nerve segments had increased cross-sectional areas with enlarged fascicles, whereas distally the nerves appeared normal. Genetic testing revealed a SPTLC1 variant resulting in a predicted Ala339Ser.

SUMMARY/CONCLUSION: Here we describe an unusual finding of proximal greater than distal demyelination in a patient with neuropathy associated with a SPTLC1 variant. To our knowledge, this unique pattern has never been described. Testing of family members is in progress to definitively demonstrate linkage and conformity of phenotype.
TWO UNUSUAL CASES OF NEUROPATHIES WITH SERUM IGM BINDING TO TS-HDS AND RESPONSE TO IVIG
Samantha LoRusso (Columbus, OH), Stanley Iyadurai (Columbus, OH)

INTRODUCTION: Serum IgM binding to trisulfated heparin disaccharide (TS-HDS) is associated with a slowly progressive, painful, predominantly sensory polyneuropathy seen mostly in older adults. Response to immunotherapy has not yet been extensively studied in this population.

OBJECTIVE: To present 2 cases of TS-HDS neuropathy associated with acute-onset ataxia.

CASE REPORT: Patient 1 is a 70-year-old woman who presented to an outside facility with weakness, numbness, and ataxia that developed over weeks. She was suspected to have Guillain–Barré syndrome and was given treatment with IV immunoglobulin (IVIg). One and half years later, when seen in clinic, her neurological examination showed normal strength, reduced sensation to all modalities with choreoathetosis of her fingers, and areflexia. Needle EMG/NCSs showed a sensory neuronopathy. The patient was placed on monthly IVIg and the choreoathetosis has stopped. Patient 2 is a 3-year-old boy who presented with acute-onset ataxia and weakness. He showed significant clinical improvement after a course of IVIg.

SUMMARY/CONCLUSION: Here we present 2 cases of TS-HDS associated peripheral neuropathy. Both cases presented with ataxia, which was not reported previously, and showed a positive response to IVIg. These cases help to broaden the clinical phenotype of this potentially treatable entity.

BRACHIAL PLEXOPATHY POST TRANSCATHETER ARTERIAL CHEMOEMBOLIZATION
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INTRODUCTION: Brachial plexus injuries immediately after procedures account for approximately 7-10% of all brachial plexopathy. The most common presentation is painless weakness in the distribution of the upper brachial plexus, sometimes accompanied by paresthesia, occasionally bilateral. These injuries are predominantly demyelinating lesions caused by prolonged traction or compression, and most patients have rapid and complete recovery unless there is significant axonal loss. Transcatheter arterial chemoembolization (TACE) has become a popular procedure to treat refractory cancer patients. There have been no reports of brachial plexopathy post TACE.

OBJECTIVE: To present 2 cases of brachial plexopathy following a TACE procedure.

CASE REPORTS: Case 1: A 66-year-old woman developed left arm numbness and weakness immediately after TACE for hepatocellular carcinoma. MRI of the brachial plexus revealed subtle asymmetric T2/short-tau inversion recovery (STIR) hyperintensity of left C5 through C8 nerve roots and trunks of the brachial plexus, consistent with plexitis/neuritis. There was gradual improvement in arm weakness, more distally 1 week after the procedure. Case 2: A 69-year-old man woke up from general anesthesia after TACE for hepatocellular carcinoma with left arm weakness and numbness. MRI brachial plexus revealed no evidence of compressive lesion to the left brachial plexus. Needle EMG was consistent with brachial plexopathy. Strength gradually improved in distal followed by proximal muscles 3 months post operation.

SUMMARY/CONCLUSION: This is the first report of brachial plexopathy following TACE. Our cases indicate that brachial plexopathy post procedure may result from stretch injury and/or inflammatory nerve changes.
A NEW SYT2 MUTATION CAUSING PRESYNAPTIC NEUROMUSCULAR JUNCTION DYSFUNCTION AND DISTAL MOTOR NEUROPATHY (LEMS-CMT)
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INTRODUCTION: Autosomal dominant mutations in synaptotagmin-2 (SYT2), a synaptic calcium sensor, have been previously linked to presynaptic neuromuscular junction (NMJ) dysfunction and motor neuropathy in 2 families. Both pathogenic mutations affected the C2B domain of SYT2, which is essential for neurotransmitter release at the NMJs.

OBJECTIVE: To describe a new missense mutation in SYT2 causing a motor neuropathy with an associated presynaptic NMJ disorder.

CASE REPORT: A 50-year-old woman with normal developmental milestones presented with high arched feet, hammertoes, and occasional falls around the age of 8. She developed progressive bilateral leg and hand weakness, cramping, and mild paresthesias on distal extremities. Similar symptoms were reported by her maternal grandfather, 2 maternal uncles, her mother, and a younger sister. Her neurological examination revealed inability to walk on heels or toes, significant distal lower extremity weakness, and absent ankle deep tendon reflexes. Needle EMG/NCSs revealed normal sensory responses but globally reduced motor amplitudes with a >200% increment after brief isometric contraction. Slow (3-Hz) repetitive nerve stimulation of the ulnar nerve revealed a 40% decremental response in amplitude and a >200% increase in amplitude immediately after 1 minute of sustained muscle contraction. Voltage-gated calcium channel antibodies and a chest CT were normal. Sanger sequencing revealed an Ile371Lys mutation in the C2B domain of SYT2.

SUMMARY/CONCLUSION: SYT2-related neuropathy is a rare disease, but should be suspected in patients presenting with a combination of presynaptic NMJ dysfunction (resembling Lambert–Eaton myasthenic syndrome) and a predominantly motor neuropathy, especially in the context of a positive family history.

Nataly Montes-Chinea, MD
Resident and Fellow Member Award Recipient

POSTURAL ORTHOSTATIC TACHYCARDIA SYNDROME: EXPERIENCE AT A SINGLE CENTER
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INTRODUCTION: Postural orthostatic tachycardia syndrome (POTS) is chronic orthostatic intolerance with diverse and nonspecific clinical presentation.

OBJECTIVE: To evaluate demographics, clinical features, laboratory abnormalities, and disease course in patient population with POTS.

METHODS: This was a retrospective review of POTS patients at the University of Nebraska Medical Center, 2004-2016. Diagnosis was based on orthostatic tachycardia of at least 30 bpm from baseline/upright heart rate (HR) of 120 bpm without orthostatic hypotension and cardiac issues; endocrine causes were excluded. The 55 patients (age: 32±9 years; range: 19-54 years) were classified into 4 groups: neuropathic (abnormal quantitative sudomotor axon reflex testing, or QSART), hyperadrenergic (supine norepinephrine >600 pg/ml), mixed, and unclassified.

RESULTS: Clinical features included lightheadedness (96%), palpitations (85%), tremulousness (24%), breathlessness (47%), generalized weakness (22%), nausea/vomiting (69%), abdominal pain (24%), constipation (44%), and diarrhea (25%). Autonomic reflex screening results showed QSART, cardiovagal, and cardioadrenergic as abnormal in 25%, 11%, and 4%, respectively. For symptom control, 96% required at least 1 and 84% required more than 1 medication. Followup was 30±28 months (range: 1-144), and at last followup 15% were medication free, 53% unemployed and 40% able to work full- or part-time, and no mortality was observed. Group diagnoses were as follows: neuropathic POTS 16%, hyperadrenergic 24%, mixed 9%, and unclassified 51%. Clinical features and number of medications needed for treatment were not different between groups.

SUMMARY/CONCLUSION: POTS is a chronic disabling disease as evidenced by a number of patients being out of work and requiring longterm medications. A small number of patients had remission of symptoms. Neuropathic and hyperadrenergic POTS were similar in clinical presentation and treatment needed.
CLINICAL AND ELECTROPHYSIOLOGICAL CORRELATION IN CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY PATIENTS - A RETROSPECTIVE STUDY
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INTRODUCTION: Chronic Inflammatory Demyelinating Polyradiculoneuropathy (CIDP) has a wide clinical and electrophysiological spectrum.

OBJECTIVE: To identify the clinical, laboratory, and electrophysiological profile of CIDP patients.

METHODS: Data of patients diagnosed with CIDP from our centre over the past 22 years were reviewed and correlations between various parameters were analyzed.

RESULTS: Among 124 CIDP patients (81 [65.32%] male; mean age: 45.02±16.15 years), 70.2% had sensorimotor, 22.6% predominant motor, and 7.3% predominant sensory syndromes; 101 (81.45%) had typical CIDP and 23 (18.55%) atypical CIDP that included multifocal acquired demyelinating sensory and motor (MADSAM) neuropathy (11), multifocal motor neuropathy with conduction block (8), and the distal acquired demyelinating symmetric neuropathy (4). The electrophysiological evaluation revealed prolonged distal latencies in ≥2 nerves in 93 (75%), reduced conduction velocities in ≥2 nerves in 73 (66.13%), absent/prolonged F-wave latencies in ≥2 nerves in 120 (96.77%), partial conduction block in ≥1 nerve in 64 (51.61%), and temporal dispersion in ≥1 nerve in 88 patients (70.97%). Albuminocytological dissociation was present in 92.1% of typical CIDP versus 45.5% of MADSAM (p<0.001). Treatment varied; 62 patients (50%) were treated with steroids, 18 (14.5%) with Ig, 22 (17.7%) with therapeutic plasma exchange, and 22 (17.7%) with azathioprine/mycophenolate mofetil. Thirteen (10.5%) had a progressive course, 40 (32.3%) had a relapsing course, and 71 (57.3%) had a stable course. Hugh's grade ≥3 did not show significant correlation with severity of electrophysiological involvement or cerebrospinal fluid (CSF) protein level.

SUMMARY/CONCLUSION: Albuminocytological dissociation in CSF is highly specific for typical CIDP. Absence of F waves/prolongation of F-wave latencies are the most consistent electrophysiological finding. Clinical severity does not show significant correlation with extent of electrophysiological involvement.

Vineetha Venugopal, MD, DNB, DM
IFCN North American Chapter Fellowship Award Recipient

COMBINED NEUROGENIC AND ARTERIAL THORACIC OUTLET SYNDROME IN A PATIENT WITH EMBOLI OF RADIAL AND ULNAR ARTERIES: A CASE REPORT
Erika Moody (San Antonio, TX), Faye Tan (Houston, TX)

INTRODUCTION: A 56-year-old female with history of hypothyroidism and latent tuberculosis on anti-tubercular agents presented with right forearm pain, numbness, and cold hand for years without history of inciting trauma.

OBJECTIVE: To review diagnosis techniques for documenting coexistence of neurogenic and vascular thoracic outlet syndrome (TOS).

METHODS: The patient was diagnosed with arterial TOS and embolic disease to the right radial and ulnar arteries prior to presentation. She subsequently underwent thrombolysis of the arterial occlusions followed by decompression with anterior scalenectomy and cervical and first rib resection. She presented to the needle EMG clinic 3 months postoperatively with complaints of residual right medial forearm numbness, tingling, and pain with associated weakness. Physical examination revealed mild atrophy of the right abductor pollicis brevis (APB). Manual muscle testing was 5/5 throughout bilateral upper extremities except 4+/5 on right hand grip and 4/5 on right little finger abduction. Postoperative CT angiogram of the right upper extremity demonstrated right subclavian artery narrowing of 25% with abduction of the arm.

RESULTS: Right ulnar sensory and motor NCSs showed decreased amplitudes. Needle EMG demonstrated abnormalities in the right APB, extensor indicis proprius, and first dorsal interosseous with increased insertional activity, fibrillation potentials, positive sharp waves, decreased recruitment, and many polyphasic potentials. Data were most consistent with a right lower trunk brachial plexopathy.

SUMMARY/CONCLUSION: This is a case report of a rare diagnosis of arterial TOS (<1% of all cases) coexisting with neurogenic TOS. NCSs and needle EMG 3 months postoperatively demonstrated a right lower trunk brachial plexopathy (C8/T1).
RAPIDLY PROGRESSIVE POEMS SYNDROME WITH ABSENT M PROTEIN IN SERUM OR URINE, NORMAL BONE SURVEY, AND NORMAL NON-TARGETED BONE MARROW BIOPSY
Payam Soltanzadeh (Cleveland, OH), Jason Valent (Cleveland, OH)

INTRODUCTION: POEMS syndrome (polyneuropathy, organomegaly, endocrinopathy, monoclonal gammopathy, and skin changes) is a paraneoplastic mixed axonal and demyelinating polyneuropathy due to an underlying plasma cell neoplasm. Diagnosis of POEMS syndrome is often delayed as this syndrome is rare and can be mistaken for other neuromuscular diseases.

OBJECTIVE: To present a patient with rapidly progressive disabling polyneuropathy due to POEMS syndrome who did not show typical non-neurologic features of POEMS.

CASE REPORT: A 48-year-old man presented with a several month history of progressive sensorimotor polyneuropathy associated with demyelinating features on EDX studies. His cerebrospinal fluid profile showed albumin protein dissociation, and MRI of the lumbar spine revealed cauda equina enhancement. Treatment with steroids, plasma exchange, and IV immunoglobulin did not help with the presumed diagnosis of chronic inflammatory demyelinating polyneuropathy (CIDP), and he became quadriparetic. Serum and urine immunofixation were negative for M protein. Vascular endothelial growth factor (VEGF) level was significantly high (454 pg/mL). A skeletal bone survey was unrevealing and bone marrow biopsy identified 1% non-clonal plasma cells. Whole-body fluorodeoxyglucose (FDG)-positron emission tomography (PET)/CT 1 month after the bone marrow biopsy showed several sclerotic and lytic lesions. A CT-guided bone biopsy revealed a lambda restricted plasmacytoma confirming the diagnosis of POEMS. Over a 5-month period, treatment with lenalidomide, cyclophosphamide, and dexamethasone led to significant improvement of his neuromuscular deficits.

CONCLUSION: Patients with POEMS syndrome might not show all the diagnostic criteria, and this syndrome should be considered in every patient with “atypical/refractory CIDP.” Measurement of VEGF level and whole body FDG-PET/CT can be very useful.

ISOLATED FINGER FLEXION NEUROMYOTONIA IN ELDERLY WOMEN WITH CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY; AN INTRIGUING TREND
Fatmah Al-Zahmi (Chicago, IL), Senda Ajroud-Driss (Chicago, IL)

INTRODUCTION: Acquired neuromyotonia is defined as generalized peripheral nerve hyperexcitability manifesting as spontaneous continuous muscle activity. Generalized muscle stiffness and pain are the main complaints. Focal neuromyotonia is a rare occurrence.

OBJECTIVE: To present a case of focal neuromyotonia and discuss similar cases in literature.

METHODS: A 64-year-old woman with history of severe chronic obstructive pulmonary disease (COPD) on oxygen therapy was evaluated for bilateral involuntary flexion of the ring and middle fingers for 1 year. She was previously seen by an orthopedic surgeon and had ultrasound of the hand which didn't show any evidence of fibromatosis. She has no involuntary movements elsewhere, no muscle spasms or cramps. A NCS was unremarkable. Needle EMG showed neuromyotonic discharges in the flexor digitorum superficialis, biceps, flexor carpi radialis, flexor carpi ulnaris, and brachioradialis. No abnormal discharges were seen in other muscles of the upper or lower extremities.

RESULTS: Review of the literature revealed 4 cases with isolated finger flexion and neuromyotonic discharges on needle EMG. All of them were elderly women (ages 70-78 years), with history of severe COPD. One case tested positive for anti-voltage gated potassium channel antibody. The mechanism by which these clinically-similar patients develop this almost identical phenotype is not clear. Previous authors have discussed the possibility of peripheral nerve hyperexcitability caused by chronic hypoxia and the use of sympathomimetic medications as a potential etiology.

SUMMARY/CONCLUSION: Isolated finger flexion represents a form of focal neuromyotonia affecting elderly women with COPD. Cases maybe under reported as they are referred to hand surgeons and may not be evaluated by neurologist.
DEMYELINATING NEUROPATHIES IN THE SUPER-ELDERLY: CASE REPORT, LITERATURE REVIEW, AND SPECIAL CONSIDERATIONS
Lauren Ottenhoff (Maywood, IL), Ryan Jacobson (Maywood, IL)

INTRODUCTION: Demyelinating neuropathies present unique diagnostic and management challenges to neuromuscular specialists. “Super elderly” patients are here defined as those greater than 85 years of age. In this complex patient population, demyelinating neuropathies are only rarely identified.

OBJECTIVE: To describe a case of an acquired demyelinating neuropathy presenting in a super elderly patient, and to review such presentations and their management in the literature.

CASE REPORT: A 93-year-old man with a history of chronic kidney disease and dementia presented to the EMG laboratory for assessment of progressive weakness and sensory change. The patient and his caregivers reported approximately 4 months of progressive symptoms. He had been walking independently 2 months prior. He then developed bilateral lower extremity followed by upper extremity weakness, and neuropathic-sounding pain of the feet and hands. His neurological examination was notable for proximal and distal weakness of the legs, bilateral hand weakness, and areflexia. NCSs revealed numerous features of demyelination, including slowed conduction velocities, very prolonged distal latencies, and conduction block in several motor nerves. The patient’s presentation was thought consistent with chronic inflammatory demyelinating polyradiculoneuropathy. He was started on therapy with oral corticosteroids.

SUMMARY/CONCLUSION: Acquired demyelinating neuropathies are seldom identified in the super elderly. In this population, attention towards possible monoclonal gammopathy is especially important. Treatment-wise, medical comorbidities including coronary artery disease, other risk factors for ischemia, and diabetes must be taken into account. Finally, the patient’s mobility and ease of presenting for infusions of corticosteroids or Ig must factor into clinical decisionmaking.

EXAMINING OUTCOMES OF MULTIFOCAL MOTOR NEUROPATHY CASES USING THE NATIONWIDE INPATIENT SAMPLE, FOR 2003-2012
Tamara Opila (Sharon Hill, PA), Nicole Benjamin (Sharon Hill, PA), Nizar Souayah (Newark, NJ)

INTRODUCTION: Multifocal motor neuropathy (MMN) is classified as a neuromuscular disorder occurring in single nerves without corresponding sensory loss. Males are affected at nearly 3 times the rate of females, and symptoms typically appear between 20-50 years of age.

OBJECTIVE: To assess inpatient outcomes for MMN cases treated with IV immunoglobulin (IVIg) and alternative therapies, when stratifying by gender and socioeconomic factors.

METHODS: A retrospective analysis (2003-2012) using the Nationwide Inpatient Sample (NIS) was conducted for patients with a primary diagnosis of MMN. Patient demographics were assessed; length of stay (LOS), charges, and disposition status were calculated.

RESULTS: The average age of patients was 56.2±21.1 years. Despite a greater prevalence documented among males, a majority of female cases were observed (53.5%). Total charges and LOS averaged $47,276 and 6.8 days, respectively. A linear trend was exhibited for daily hospitalization costs, while LOS remained steady during this period. Charges ranged $4727-10,418/day (p<0.001); this trend persisted even after accounting for inflation. The treatment plan included IVIg for 13.8% of patients; plasmapheresis and blood transfusions were also represented among common hospital procedures. Non IVIg patients demonstrated a similar rate for routine discharges compared to IVIg patients (54%; p=0.70); the latter had a significantly shorter LOS of 5.0 days (p=0.03). A greater proportion of patients receiving IVIg had private insurance compared to those receiving alternative therapies (35%; p=0.06).

SUMMARY/CONCLUSION: Daily hospitalization charges increased while LOS remained steady. These observations may be attributed to patient acuity; early intervention and maintenance therapy are critical to prevent hospitalization.
FULMINANT ISOLATED VASCULITIS OF THE PERIPHERAL NERVOUS SYSTEM IN A PATIENT WITH HEPATITIS C VIRUS-ASSOCIATED CRYOGLOBULINEMIA
Kourosh Rezania (Chicago, IL), Peter Pytel (Chicago, IL), Reeti Greenwald (Chicago, IL), Lena Derani (Chicago, IL), Raymond Roos (Chicago, IL)

INTRODUCTION: Hepatitis C virus (HCV) is the most common cause of cryoglobulinemic vasculitis (CV). Although neuropathy can be the initial presentation, other systemic manifestations (purpuric skin rash, arthralgia, and nephropathy) also usually occur during the disease course.

OBJECTIVE: To present a patient with HCV related CV fulminant mononeuritis multiplex without other systemic manifestations.

CASE REPORT: A 52-year-old diabetic male presented with burning pain in the left foot and leg, followed by progressive weakness and numbness and pain in the right leg and bilateral hands over the next 2 months. Examination showed a right facial paresis, muscle strength of 1/5 in bilateral hand grasp and interossei, 0/5 in bilateral ankle dorsiflexion, and 3/5 in bilateral ankle plantar flexion. There was stocking/glove sensory loss and allodynia, absent ankle jerks, and ataxic steppage gait. Workup showed positive HCV antibody and viral load of 13 million IU/ml; significantly high serum C-reactive protein, cryoglobulin, and rheumatoid factor; an IgM kappa monoclonal; and very low serum concentrations of C3/C4. Needle EMG demonstrated a severe multifocal axonal polyneuropathy. Small-to-medium vessel vasculitis with fibrinoid necrosis of the vessel walls with extensive loss of large myelinated axons was present in sural nerve biopsy. Plasmapheresis was administered followed by rituximab infusions and a course of oral sofosbuvir and ribavirin. During about 3 years of followup, there has been persistent clearance of HCV viremia and cryoglobulinemia, and marked improvement of his neuropathy.

SUMMARY/CONCLUSION: HCV related CV may manifest with isolated fulminant vasculitis of the peripheral nerves, which responds favorably to antiviral and B cell depletion treatment.

NON-TRAUMATIC SCIATIC NEUROPATHIES: CLINICAL AND ELECTRODIAGNOSTIC FEATURES IN 112 PATIENTS
Rejo Cherian (Cleveland, OH), Yuebing Li (Cleveland, OH)

INTRODUCTION/OBJECTIVE: Sciatic neuropathy is common but analyses of large series is lacking. We undertook a retrospective review of clinical and EDX features of patients diagnosed with sciatic neuropathy, unrelated to penetrating trauma.

METHODS: We reviewed 112 patients with EDX-confirmed non-traumatic sciatic neuropathy at our center from 2002 to 2012.

RESULTS: The study included 51 males and 61 females (mean age: 52.3 years). Onset was acute (<1 week) in 62.5% of patients. The most common etiologies were hip or knee surgeries (49.1%), acute external compression (14.3%), and perioperative positioning (9.8%). Other etiologies (26.8%) included vascular procedure, radiation, infection, and vasculitis. Combined sensory and motor deficits in the distal leg was the most common presentation, and foot drop was seen in 77.7% of patients. EDX studies revealed a preferential involvement of the peroneal (fibrillar) division in 39.3% and tibial in 5.4% of patients. On NCSs, peroneal motor and superficial peroneal sensory studies were mostly abnormal (92.9% respectively), followed by sural sensory (83.9%) studies. Needle examination of the tibialis anterior and tibialis posterior provided the highest EDX yield (92.0% and 85.6%, respectively). Axon loss findings were predominant, and demyelinating features were seen in 4.5% of patients. Among the 100 patients with a followup (mean period of 2.6 years), 35% showed significant improvement.

SUMMARY/CONCLUSION: Non-traumatic sciatic neuropathy encompasses a variety of etiologies but typically involves surgery or external compression. EDX data reveal a preferential involvement of the peroneal division and mostly are axonal. Significant subsequent improvement is only seen in one-third of patients.

Rejo Cherian, MD
Resident and Fellow Member Award Recipient
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NEURALGIC AMYOTROPHY MIMICKING ANTERIOR INTEROSSEOUS SYNDROME: CONSTRICTION OF MEDIAN NERVE WITH ABERRANT ANASTOMOSIS

Dong Hwee Kim (Ansan-si, KR), Jong Woong Park (Ansan-si, KR)

INTRODUCTION: We report a case with clinical manifestations of neuralgic amyotrophy (NA) probably related to severe constriction of the proximal median nerve (MN) and aberrant anastomosis from the musculocutaneous nerve.

CASE REPORT: A 42-year-old woman who had experienced left shoulder pain developed severe weakness in the left thumb and index finger flexors following electrical pain in the left elbow and middle and ring fingers. Muscle strength of the flexor digitorum profundus and flexor pollicis longus were both grade 1; the other MN-innervated muscles were grade 3. There was hypesthesia on the ulnar side of the left middle finger and radial side of the ring finger. The left "okay" sign was positive. Tinel sign of the left MN was positive below the axillae. Serial median motor NCSs demonstrated progressive decline of amplitude from 5.5 to 0.1 mV for 3 weeks. Median sensory NCSs showed significant axonal loss of the left middle and ring fingers (76.4% and 75.8%, respectively). Initial ultrasonography of left MN demonstrated no swelling but suspected constriction. After steroid pulse therapy, motor and sensory symptoms were not improved. Followup ultrasonography showed diffuse swelling and hourglass-like constriction of the MN and aberrant anastomosis with the musculocutaneous nerve distal to severe constriction of left MN. Surgical exploration demonstrated compatible findings with ultrasonographic examination, and direct end-to-end neurorrhaphy was performed after segmental resection of the constricted region. After surgery, motor and sensory symptoms were much improved.

CONCLUSION: In patients with suspected NA, ultrasonographic examination is recommended for evaluating the morphologic change of the nerve.

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ELECTROPHYSIOLOGIC FEATURES OF PERIPHERAL NEUROPATHY IN COPPER DEFICIENCY AND COPPERIEDUS


INTRODUCTION: Copper is both essential and toxic to humans. Copper deficiency (CD) and toxicity (CT) may cause multisystem dysfunctions including the central/peripheral nervous systems. CD causes myeloneuropathy with electrophysiologic features resembling vitamin B12 deficiency. However, the electrophysiologic features of peripheral neuropathy in CT have not been reported in the literature.

OBJECTIVE: To evaluate the electrophysiologic features in CD and CT.

METHODS: We retrospectively reviewed our neuromuscular clinic/EMG laboratory database, January 1, 2014 to December 31, 2016 to identify CD or CT patients. Subjects with zinc abnormality or those with no EDX study were excluded. Data were obtained from NCSs and needle EMG performed on 1 arm and 1 leg. Distal latency (DL), amplitude, and conduction velocity (CV) of NCSs and needle EMG waveforms were collected and analyzed.

RESULTS: Four CD patients (M/F=3/1; age: 54.5±19.8 years; copper=55.3±9.7 mcg/dL [ref=72-166]; zinc=73±17.5 mcg/dL [ref=56-134]) and 3 CT patients (M/F=1/2; age: 57±8.2 years; copper=215±10.8 mcg/dL; zinc=72.3±14.6 mcg/dL) were included. Statistically significant differences (p<0.05) were observed between CD and CT in the arms only, such as motor NCSs (median: amplitude 5.9±4.3 mV and CV 49.3±23.7 m/s versus 7.0±2.3 mV and 63.7±3.5 m/s; ulnar: 7.9±5.5 mV and 46.5±16 m/s versus 9.0±2.9 mV and 7.9±5.5 m/s) and sensory NCSs (radial: CV 51.0±4.2 m/s versus 53.3±1.5 m/s; ulnar: 44.0±12.5 m/s versus 52.0±2.0 m/s) in CD versus CT, respectively. CTS was seen in sensory NCSs of median nerve in both CD (CV 41.5±17.7 m/s) and CT (44.3±10.4 m/s). Needle EMG did not show significant differences between the groups.

SUMMARY/CONCLUSION: Different patterns of electrophysiologic features between CD and CT suggests different pathophysiology.
LENALIDOMIDE-RESPONSIVE ANTI-MAG NEUROPATHY
Amro Stino (Columbus, OH), Yvonne Efébera (Columbus, OH)

INTRODUCTION: Anti-myelin-associated glycoprotein (MAG) neuropathy is a disabling condition that has limited therapeutic options, largely due to a lack of consensus regarding proper outcome measures to track treatment responsiveness. Rituximab is the most studied therapy with possible treatment effect, although its efficacy has been questioned by 2 negative randomized controlled clinical trials.

OBJECTIVE: To examine the efficacy of lenalidomide, an immunosuppressive agent with proven efficacy in monoclonal gammopathies and POEMS syndrome (polyneuropathy, organomegaly, endocrinopathy, monoclonal gammopathy, and skin changes), in anti-MAG neuropathy.

CASE REPORT: A 76-year-old woman was seen in our clinic for anti-MAG neuropathy. She had immunoglobulin M (IgM) monoclonal gammopathy, anti-MAG titers >1:102400, distal demyelination on NCS (with secondary axonal loss), notable immediate sway on eye closure, and gait unsteadiness. She showed no improvement in quality of life or function 9 months following rituximab infusion. She was placed on lenalidomide 5 mg days 1-21 (in a 28 day cycle). Seven months following initiation of lenalidomide therapy, IgM levels dropped 71%, the inflammatory Rasch-built overall disability scale (I-RODS) score improved from 32 to 39, she was able to ambulate with good balance, and Romberg testing showed no sway.

SUMMARY/CONCLUSION: Lenalidomide may prove to be therapeutically efficacious in patients with anti-MAG neuropathy, especially those refractory to rituximab and/or with advanced axonal loss. However, our findings must be cautioned by the lack of consensus regarding the best outcome measures to track treatment responsiveness. The planned IMAGiNe study, aimed at identifying the best outcome measures in anti-MAG neuropathy, should pave the way for improved evaluation of drug efficacy.

DIAGNOSTIC UTILITY OF FIBULAR MOTOR AMPLITUDE IN THE DIAGNOSIS OF L5 RADICULOPATHY IN PATIENTS REFERRED FOR RADIATING LOWER LIMB PAIN
Brendan McNeish (Southbury, CT), Anita Craig (Ann Arbor, MI), Sandra Hearn (Ann Arbor, MI), Ann Laidlaw (Ann Arbor, MI), Mark Ziadeh (Ann Arbor, MI), James Richardson (Ann Arbor, MI)

INTRODUCTION: Electrodiagnosis of radiculopathy traditionally depends on needle EMG. However, pain associated with needle EMG limits applicability in some clinical populations. We explored the utility of NCSs for stratifying patients according to likelihood of needle EMG-confirmed L5 radiculopathy.

OBJECTIVE: To determine the diagnostic accuracy of fibular motor amplitude (FMAmp; recording L5-innervated extensor digitorum brevis) in identifying needle EMG-confirmed L5 radiculopathy.

METHODS: EDX reports coded for “L5 Radiculopathy” and “No Abnormality” in patients referred for radiating lower limb pain were identified and reviewed by American Board of Electrodiagnostic Medicine certified EDX physicians to determine absence of confounding conditions and presence of appropriate NCSs and needle EMG (including at least 2 L5-innervated muscles).

RESULTS: Radiculopathy subjects (n=104) had diminished FMAmp as compared to control subjects (n=113) (3.4±2.3 mV versus 6.2±2.3 mV, respectively; p<0.001). Receiver Operator Characteristics Curve demonstrated an Area Under Curve=0.819 (95% CI, 0.763, 0.876; p<0.001). Specific cut-point analyses showed that only 4.8% of radiculopathy patients had FMAmp >7.5 mV, while 24.8% of control subjects had FMAmp exceeding this value; 50% of radiculopathy patients had FMAmp <2.9 mV. Optimal combined sensitivity/specificity (60%/92%, respectively) occurred at 3.6 mV.

SUMMARY/CONCLUSION: FMAmp demonstrates potential for predicting likelihood that patients referred with radiating lower limb pain will have needle EMG-confirmed L5 radiculopathy. In particular, determination of a FMAmp cut-off above which a needle EMG-detectable radiculopathy would be atypical (7.5 mV based on our preliminary data) may be useful for patients who do not tolerate the needle EMG examination. Similar research at additional root levels should be pursued.
RECURRENT CRANIAL NERVE VII NEUROPATHY DUE TO IMMUNOGLOBULIN G4 RELATED DISEASE: A CASE REPORT
Alison Walsh (Philadelphia, PA), Anthony Allen (Philadelphia, PA)

INTRODUCTION: Immunoglobulin G4-related disease (IgG4-RD) is an immune mediated disease comprised of previously viewed unrelated disorders with shared histopathologic features of storiform fibrosis, obliterative phlebitis, and lymphoplasmacytic infiltration. Clinically, IgG4-RD has been associated with many fibroinflammatory conditions (autoimmune pancreatitis, sclerosing cholangitis, retroperitoneal fibrosis, and idiopathic hypertrophic pachymeningitis).

OBJECTIVE: To describe a case of a woman with recurrent cranial nerve palsies due to IgG4-RD.

CASE REPORT: A 33-year-old woman presented with new right facial numbness and weakness. She had a history of bilateral Bell’s palsy affecting her right facial nerve 3 years prior and affecting her left facial nerve 1 year prior. On examination she had facial diplegia, with abnormal sensation affecting right V2 and V3 distributions. Ophthalmoscopic examination demonstrated papilledema bilaterally. MRI of the brain showed pachymeningeal thickening and enhancement affecting primarily the right cerebral hemisphere and brainstem. Lumbar puncture demonstrated elevated opening pressure, mildly elevated lymphocyte count, and elevated protein. CT of the chest/abdomen/pelvis was unremarkable. Infectious, neoplastic, and other autoimmune conditions were excluded. IgG4 levels were significantly elevated. She was treated with high dose corticosteroids with improvement.

SUMMARY/CONCLUSION: A relatively newly described entity, IgG4-RD, can have primary neurologic appearances, in addition to the more often reported gastrointestinal and other systemic conditions. We describe a patient with recurrent bilateral cranial nerve neuropathies due to IgG4-RD. This case adds to our knowledge of the growing spectrum of IgG4-RD manifestations. Practitioners must be aware of this neurologic complication so as to accurately diagnose this disorder, impart early interventions, and uncover subclinical associated conditions.

NEURALGIC AMYOTROPHY IN A 15-YEAR-OLD WITH NEURAPRAXIC BLOCK
Nick Kinback (Elkins Park, PA), Ziva Petrin (Philadelphia, PA), Channarayapatna Sridhara (Elkins Park, PA)

INTRODUCTION: A 15-year-old healthy swimmer with acute-onset of pain in the right shoulder woke with severe pain 4 months ago with no numbness, tingling, inciting events, or neck pain. Pain completely resolved in 1 week. After pain resolved, she noted weakness of the right shoulder with prominence of the scapula. The right scapula was elevated, protracted with a depressed glenoid. There was minimal atrophy of the serratus anterior with winging of the medial scapula with shoulder flexion. Scapular retraction was normal. Other shoulder muscles were normal.

OBJECTIVE: To evaluate EDX abnormality in the shoulder girdle muscles.

CASE REPORT: The right long thoracic latency was insignificantly prolonged compared to the left, with symmetric compound muscle action potentials. Right median sensory/motor NCSs and H latency of the right median nerve was normal. No abnormalities were noted in insertional or spontaneous activity. There was increased duration and polyphasicity of motor unit action potential in the serratus anterior, deltoid, and rhomboid with reduced recruitment in the rhomboid. This is typical of neuralgic amyotrophy (NA) with involvement of some of C5-innervated muscles with old axon loss. Typical findings in NA are patchy demyelination and axonal degeneration. Conduction block is rare. The patient's young age is atypical for idiopathic NA. She lacks the typical dysmorphic features of hereditary NA, but genetic mutation of the SEPT9 gene should be considered. Patchy C5 root pathology cannot be excluded but lacks an inciting event. EDX examination of scapular muscles with no acute axon loss suggests neurapraxic block proximal to Erb's point.

SUMMARY/CONCLUSION: This is an atypical case of NA with unusual presentation of conduction block with possible genetic mutation.
MISDIAGNOSIS IN SENSORY NEURONOPATHIES
Alberto Martinez (Campinas, BR), Ingrid Faber (Brasília, BR), Raphael Casseb (Campinas, BR), Maximiliano Carneiro (Campinas, BR), Anamarli Nucci (Campinas, BR), Marcondes França Jr (Campinas, BR)

INTRODUCTION: Sensory neuronopathies (SNs) represent a distinct group of peripheral nerve system (PNS) disorders characterized by sensory ataxia and non-length sensory deficits. Since the first descriptions made by Derek Denny-Brown in 1948 great improvements have been made toward a simpler diagnostic workflow, such as the diagnostic criteria proposed by Antoine and colleagues. Earlier diagnoses of SNs enable recognition of potentially serious underlying conditions, such as Sjögren’s syndrome and pulmonary small cell lung neoplasm. Despite that, PNS impairment in general may resemble a challenging differential diagnosis which might lead to SN misdiagnosis.

OBJECTIVE: To evaluate the time delay and the different previous diagnoses of SN patients.

METHODS: We retrospectively analyzed 20 consecutive patients’ data with SN who were regularly followed in a tertiary neuromuscular clinic.

RESULTS: Nine (45%) were men, and 11 (55%) were women. Mean age at onset and disease duration were 41 years (range: 22-63 years) and 10.8±7.36 years, respectively. None of these patients was first evaluated in our center when their SN symptoms first appeared. Mean time of diagnosis delay was 6.6±6.5 years (range: 1-21 years). Every patient was evaluated by a mean of 4.4±7.36 specialists (including neurologists, general practitioners, rheumatologists, orthopedists, etc.) and received 3.1±1.0 wrong diagnoses with 12 different treatments.

SUMMARY/CONCLUSION: Despite the fact that SNs are considered rare conditions, their misdiagnosis may represent a significant feature. A high suspicion is necessary, not only by neurologists but also by other medical specialties, to improve this scenario and thereafter the general care of these patients.

Alberto Martinez, MD
AANEM Foundation for Research and Education Award Recipient

HORNER'S SYNDROME IN A PARANEOPLASTIC MULTIFOCAL ACQUIRED Demyelinating Sensory and Motor Neuropathy: A Case Report
Erin Manning (New York, NY)

INTRODUCTION: There is 1 reported previous case of a Horner’s syndrome associated with chronic inflammatory demyelinating polyneuropathy (CIDP).

CASE REPORT: A 63-year-old woman presented with a chief complaint of progressive weakness and numbness and pain in the left arm. She had a history of left breast cancer treated 14 years ago with lumpectomy and full axillary node dissection followed by radiation therapy and chemotherapy. She developed progressive weakness, pain, and numbness 5 months prior to presentation. Her examination showed distal left arm weakness and atrophy, hyperesthesia in the ring and little fingers and half of the middle finger, and edema in left forearm and hand. Her needle EMG/NCSs of the arms showed evidence of demyelination in the median and ulnar nerves of the left arm. MRI of the left brachial plexus showed fascicular enlargement of the C8-T1 nerve roots and the medial cord. She was diagnosed with CIDP of the multifocal acquired demyelinating sensory and motor neuropathy (MADSAM) variant. She started IV immunoglobulin with stabilization of her weakness and subsequent worsening of her weakness and sensory symptoms. IV methylprednisolone was added with initial stabilization and then worsening of symptoms. She developed mild right ptosis and pupil dilatation. During the workup for the cause of the Horner’s syndrome she was found to have pulmonary nodules. She was diagnosed with metastatic breast cancer. There has been some improvement in sensory symptoms with cancer treatment.

SUMMARY/CONCLUSION: This is the first case report of a patient with paraneoplastic MADSAM and a Horner’s syndrome.
IMMUNE RESPONSIVE CIDP RESOLVING WITH ANTERIOR MEDIASTINAL MASS RESECTION
Omer Suhaib (Oklahoma City, OK), Joon-Shik Moon (Oklahoma City, OK), Tyler Webb (Oklahoma City, OK), Eduardo De Sousa (Oklahoma City, OK)

INTRODUCTION: POEMS syndrome (polyneuropathy, organomegaly, endocrinopathy, monoclonal gammopathy, and skin changes) is a monoclonal plasma cell disorder associated with osteosclerotic myeloma, Castleman's disease (CD), and increased vascular endothelial growth factor (VEGF). CD is a lymphoproliferative disease with unicentric and multicentric variants, occasionally causing peripheral neuropathy.

OBJECTIVE: To report a patient with chronic inflammatory demyelinating polyneuropathy (CIDP)-like immunoresponsive polyneuropathy and anterior mediastinal mass.

CASE REPORT: A 57-year-old man presented with prednisone-responsive foot drop and proximal upper/lower extremity weakness over 8 weeks subsiding 3 times over last 3 years, with chronic constipation, gynecomastia, erectile dysfunction, and hyperhidrosis without organomegaly or orthostatic intolerance. CIDP diagnosis compatible with European Federation of Neurological Societies (EFNS)/ peripheral nervous system EDX criteria. Cerebrospinal fluid: protein 77, 9 white blood cells, 24 red blood cells. Serum immunofixation: atypical immunoglobulin G (IgG) without monoclonality, mildly elevated free kappa light chains, later IgG kappa chain monoclonal gammopathy. Antinuclear antibodies negative in 2012, 1:640 in 2013, 1:360 in 2016, unremarkable reflex panel. VEGF ranged 158-824 (normal <115). Positron emission tomography (PET)/CT: metabolically-active anterior mediastinal mass. Glycated hemoglobin, B12, thyroid-stimulating hormone, human immunodeficiency virus, interleukin 6, human herpesvirus 8, bone-marrow biopsy; urine porphyria/heavy metals, skeletal survey, celiac and paraneoplastic panels unremarkable. Last recurrence subsided after prednisone and IVIg; VEGF normalized to 67 (5-12-16) before thymectomy (5-27-16). Thymic pathology: unicentric CD with features of hyaline vascular and plasma cell variants versus consistent with POEMS. Bence–Jones kappa protein recently positive. He remains asymptomatic with variable VEGF levels. Recent PET/CT revealed complete metabolic resolution.

SUMMARY/CONCLUSION: The patient had clinical presentation and response to treatment mimicking CIDP, with complete resolution following resection of a metabolically-active anterior mediastinal mass.

MULTIFOCAL ACQUIRED DEMYELINATING SENSORY AND MOTOR NEUROPATHY PRESENTING INITIALLY AS ANTERIOR INTEROSSEOUS NERVE SYNDROME: ELECTRODIAGNOSTIC AND MRI CORRELATION
Brion Reichler (New York, NY), Darryl Sneag (New York, NY)

INTRODUCTION: Multifocal acquired demyelinating sensory and motor neuropathy (MADSAM) is characterized by a multifocal pattern of motor and sensory nerve involvement, evolving over months to years, with demyelination on NCSs. Reports of imaging are rare in the literature.

OBJECTIVE: To present the MRI findings of a case of early MADSAM presenting initially as anterior interosseous nerve syndrome (AINS).

CASE REPORT: A man presented with right pincer grip weakness, with subsequent stepwise development of pronation weakness, lateral antebrachial cutaneous (LAC) sensory loss, and finally left AINS, over months. On examination, there was weakness of right proximal median and bilateral anterior interosseous muscles, but sparing of the abductor pollicis brevis. On NCS, distal median function was normal. No demyelinating features could be demonstrated in any of the testable nerves. Right LAC sensory response was absent. Needle EMG showed neurogenic changes in muscles innervated by the proximal median nerve and anterior interosseous nerve, only. MRI of the brachial plexus was normal. MRI of the elbow region of both arms showed signal hyperintensity and enlargement of fascicles of the proximal median nerve.

SUMMARY/CONCLUSION: The diagnosis of MADSAM could not be definitively established in this patient based on demyelinating features on NCSs, given the proximal pattern of involvement. However, the stepwise clinical course over months with multifocal involvement suggests this as the only reasonable diagnosis. While Parsonage–Turner syndrome is the most common cause of AINS in clinical practice, a stepwise course over months would be highly atypical.
POEMS SYNDROME: AN IMPORTANT MIMIC OF CHRONIC INFLAMMATORY Demyelinating POLYNEUROPATHY
Matthew Imperioli (Farmington, CT)

INTRODUCTION: POEMS syndrome, which stands for polyradiculoneuropathy, organomegaly, endocrinopathy, monoclonal plasma cell disorder, and skin changes, is a rare paraneoplastic syndrome due to an underlying plasma cell neoplasm. The EDX characteristics of the neuropathy include a primary demyelinating or mixed axonal and demyelinating sensorimotor neuropathy that can mimic chronic inflammatory demyelinating polyneuropathy (CIDP).

CASE REPORT: A 57-year-old man presented with 6 months of progressive symmetrical sensory loss and weakness with associated areflexia. Physical examination findings included leukonychia, subtle gynecomastia, and hyperpigmentation of the extremities. A NCS revealed a demyelinating sensorimotor polyneuropathy without evidence of conduction block with an acute chronic polyradiculopathy seen on needle EMG of the lower extremities. Laboratory testing was notable for a biclonal immunoglobulin A lambda gammopathy, thrombocytosis of 667 10^3/µL, and a vascular endothelial growth factor (VEGF) level over 4 times the upper limit of normal at 357 pg/mL. There were multiple osseous lesions with diffuse nerve root enhancement on MRI of the lumbar spine. Cerebrospinal fluid (CSF) analysis was only remarkable for a protein level of 320. A bone marrow biopsy of the L3 vertebral body was consistent with a plasma cell neoplasm.

SUMMARY/CONCLUSION: POEMS syndrome is a rare disorder that can mimic CIDP both in its clinical presentation and EDX features. Not all patients with POEMS develop the full complement of symptoms classically described. A high index of suspicion and close physical examination are needed as features of this condition can be subtle. The vertebral lesions, lack of conduction block on NCSs, and thrombocytosis are other clues to suggest POEMS syndrome.

NEUROPATHIC DISTAL FOOT PAIN AS AN ATYPICAL PRESENTATION OF A TREATABLE PLANTAR PLATE TEAR
Amit Sachdev (East Lansing, MI), Meredith Herman (East Lansing, MI), Karl Dunn (East Lansing, MI)

INTRODUCTION: Distal dysesthesias in older aged individuals are often assumed to represent sensory peripheral polyneuropathy. Peripheral sensory polyneuropathies can present asymmetrically. Prognosis for recovery of normal sensation is dismal. Remarkable prolonged asymmetry should prompt additional evaluation for mononeuropathy. We report an unusual presentation of a plantar plate tear diagnosed in the evaluation of persisting unilateral sensory loss. Recent understanding of plantar plate pathology suggested a non-surgical treatment approach involving lesional steroid injections is successful in achieving resolution of injury. To our knowledge this represents the first report of digital nerve injury being utilized to identify an underlying plantar plate lesion.

OBJECTIVE: To describe a case of treatable orthopedic injury detected by methodical localization of unilateral dysesthesias.

CASE REPORT: A 58-year-old male presented in referral for shooting foot pain for years. Sensory examination revealed left sided numbness of the plantar aspect of the foot just proximal to the thumb and index fingers. No other foot deformities were obvious. The right side was spared. Bilateral lower extremity needle EMG revealed absent left medial plantar response. Motor responses were normal. Needle EMG was normal. MRI of the left foot without contrast revealed a tear of the lateral collateral ligament and lateral to mid fibers of the plantar plate at the second metatarsophalangeal joint. There was adjacent perineural fibrosis surrounding the second plantar digital nerve.

CONCLUSIONS: Careful history and examination are essential in asymmetric, long-standing, or progressive but focal foot pain and should prompt consideration of peripheral mononeuropathy secondary to orthopedic injury. Needle EMG and MRI maybe useful in the further evaluation of these patients.
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CASES OF LATE ONSET MYASTHENIA GRAVIS PRESENTING WITH ISOLATED BULBAR WEAKNESS WITHOUT OCULAR INVOLVEMENT
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INTRODUCTION: Foot drop is commonly encountered in clinical practice. Common acquired causes include compressive peroneal neuropathy, sciatic neuropathy, lumbar plexopathy, lumbar radiculopathy, motor neuron disease, stroke, and multiple sclerosis.

OBJECTIVE: To ensure awareness of an unusual but easily correctable cause of foot drop.

CASE REPORT: We describe a rare case of foot drop in a 46-year-old female with cerebral palsy, hypothyroidism, and C4-5 cervical myelopathy status post surgical correction who presented with left foot drop for 1 week. Her family reported no bowel movement for 2 weeks. Neurological examination showed normal cranial nerves and strength of 3/5 in upper extremities and right lower extremities. Ankle dorsiflexion on the left was 1/5, eversion was 0/5, and plantar flexion was 1/5. Sensory examination was limited due to cerebral palsy. Gait was deferred as she was wheelchair bound. MRI of the brain was unremarkable. MRI of the cervical spine showed C4-5 fusion with some myelomalacia of the cord at the same level. MRI of the lumbar spine did not show any acute abnormality. MRI of the pelvis showed large amount of stool in the rectum with mass effect on the left sciatic nerve. She underwent manual disimpaction. In the next few days she was able to move her left foot minimally.

SUMMARY/CONCLUSION: Chronic constipation should be considered in the differential diagnosis of extra spinal causes of foot drop. Timely treatment may result in favorable prognosis.

Rocio Garcia Santibanez, MD
Resident and Fellow Member Award Recipient
**IMPACT OF MEDICATIONS ON MYASTHENIA GRAVIS**

Rohit Gummi (Columbia, MO), Raghav Govindarajan (Columbia, MO)

OBJECTIVE: To assess the impact of certain medications that are contraindicated in patients with myasthenia gravis (MG) (those listed by Myasthenia Gravis Foundation of America on myasthenic exacerbation).

METHODS: A retrospective chart review of MG patients at the University of Missouri Hospital was performed. This included 127 patients (average age: 61.9 years, average disease duration: 8.8 years) seen from 2011 to 2016. Patient demographics and causes of exacerbations were recorded. The total number of exacerbations for every patient that was prescribed a contraindicated medication after diagnosis with MG was compared to the total exacerbations for each MG patient that was not prescribed any of the contraindicated medications. A two sided t-test was performed.

RESULTS: The patients had experienced 212 total exacerbations, with 106 requiring visits to the ER, and 141 requiring admission with an average duration of 6.2 days. Of the exacerbations, contraindicated medications played a part in the plurality at 19%. Patients that were prescribed contraindicated medications also had a significantly higher number of exacerbations, with an average of 2.1, than patients not prescribed contraindicated medications, who had an average of 0.79 (p<0.01). Beta-blockers, specifically, were found to have a significant association with a higher number of exacerbations (p<0.01).

CONCLUSION: Certain medications (some more than others) are common triggers of MG exacerbations. It is important to increase awareness among patients and physicians about these medications and subsequent risk for patients.

Rohit Gummi, BS  
Resident and Fellow Member Award Recipient

**CLINICODEMOGRAPHIC PREDICTORS OF THE IMPACT OF INFECTIONS ON MYASTHENIA GRAVIS**

Natalie Kukulka (Columbia, MO), Raghav Govindarajan (Columbia, MO)

BACKGROUND: Myasthenia gravis (MG) is an autoimmune disease which necessitates use of longterm immunosuppressive treatment, and thus makes those with MG vulnerable to infections.

OBJECTIVE: To assess the impact of infections on MG and its exacerbations.

METHODS: A retrospective chart review was performed on 127 MG patients (average age: 61.9 years, average disease duration: 8.8 years, 95%). All acquired infections (vaccine preventable infections, or VPIs, included pneumonia and seasonal influenza; vaccine non-preventable infections, or VNPIs, included opportunistic infections) were noted for each patient, compared to the immunization records, and analyzed for their significance in MG exacerbation.

RESULTS: A total of 212 flare-ups requiring 106 ER visits, 141 hospitalizations, and an average admission for 6 days were noted. Infections were responsible for 34% of all MG exacerbations, 44% of ER visits, and 40% of hospital admissions as well as the second longest average duration of a hospital admission (approximately 7 days at total cost of $11,000-14,000). VPIs were the most common reason for MG exacerbation needing an ER visit and hospitalization (60%), whereas only 20% of those with VNPIs needed an ER visit and admission. Common VPIs included pneumonia (16.5%) and influenza (11%). Two patients had developed infection despite vaccine (both influenza), whereas the rest were not immunized. The most common VNPI was an upper respiratory infection (20%). Older patients (both at the diagnosis and current age) were at an increased risk factor for a VPI (p<0.05) but not for a VNPI.

CONCLUSION: Infections are one of the most common triggers for MG exacerbations and contribute to prolonged admissions and hospital costs.

Natalie Kukulka, BS  
Resident and Fellow Member Award Recipient
CASES OF LATE ONSET MYASTHENIA GRAVIS PRESENTING WITH ISOLATED BULBAR WEAKNESS WITHOUT OCULAR INVOLVEMENT
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INTRODUCTION: Propensity for early ocular muscle involvement in myasthenia gravis (MG) is common and can lead to an early diagnosis. It has been theorized that fewer acetylcholine receptors may be present in ocular muscles, or even the response of ocular muscles to an autoimmune phenomena is more pronounced. Isolated bulbar weakness in late-onset MG (LOMG) is rare, and only 14 documented cases have been reported in literature.

OBJECTIVE: To present 2 cases of seropositive of LOMG with bulbar weakness as initial presentation without ocular symptoms.

CASE REPORTS: Case 1: A 62-year-old man presented with intermittent dysphagia accompanied by dysarthria and dysphonia over 2 weeks, without associated visual, sensory, motor, or gait instability. A few days later the patient developed left facial and neck extensor weakness. Only after negative workup of MRI of the brain, CT angiogram (head/neck), and endoscopy was LOMG was considered. Case 2: A 79-year-old male presented with dysphagia, dysarthria, and right facial weakness. Initially, acute inflammatory demyelinating neuropathy (AIDP) was considered. LOMG was considered after negative workup for AIDP. Both cases had elevated acetylcholine receptor antibodies. Repetitive stimulation showed significant decrement to confirm MG. Both patients responded to pyridostigmine treatment.

SUMMARY/CONCLUSION: LOMG with focal bulbar weakness is uncommon and presents clinical challenges. MG should be considered in the differential diagnosis of focal bulbar weakness, along with stroke and AIDP. Initial misdiagnosis is not uncommon, and careful attention to history should be placed on fluctuation of symptoms. Early diagnosis and treatment can improve clinical outcome and avoid unnecessary invasive procedures such as contrast studies and endoscopy.
ECULIZUMAB IN REFRACTORY MYASTHENIC CRISIS  
Crystal Yeo (Houston, TX), Milvia Pleitez (Houston, TX)

INTRODUCTION: No evidence based studies for treatment options in refractory myasthenia crisis exist. Rituximab improved respiratory status in two case studies. Eculizumab improved clinical outcomes in acetylcholine receptor antibody-positive refractory myasthenia gravis (MG) in REGAIN Phase-3. It has not been reported in refractory myasthenic crisis.

OBJECTIVE: To report promising results with eculizumab in refractory myasthenic crisis.

CASE REPORT: A 79-year-old woman with hemolytic uremic syndrome, in remission after eculizumab and rituximab, presented with acute respiratory failure and weakness. She reported intermittent ptosis 1 year prior to presentation. Acetylcholine receptor, muscle specific kinase, LRP4, voltage-gated calcium channel, and striatal antibodies were negative. CT of the chest showed no thymoma. Repetitive nerve stimulation showed decrement consistent with MG. She received 5 plasma exchanges, improved, and was discharged on home oxygen. Days later, she was readmitted and intubated for severe respiratory distress. Despite Mestinon (pyridostigmine), corticosteroids, and IV immunoglobulin (IVIg), she remained ventilator dependent necessitating tracheostomy at 23 days ventilation. CD20/CD19 counts were negligible. At 25 days ventilation, she received eculizumab per the REGAIN phase 3 protocol. One week after starting, she tolerated pressure support weaning. Four days later, continuous positive airway pressure was tolerated. At 3 weeks, she tolerated tracheostomy collar, and at 4 weeks, tolerated it for 12-15 hours daily, walking around without ventilator support. At 6 weeks, she tolerated 48 hours tracheostomy collar. Muscle weakness improved significantly.

SUMMARY/CONCLUSION: Myasthenic crisis is usually reversible, however refractory cases to IVIg, plasma exchange, and rituximab exist. Our experience suggests eculizumab may be effective in refractory myasthenic crisis, and benefit seronegative MG patients.

Crystal Yeo, MD PhD  
Resident and Fellow Member Award Recipient
EFFICACY OF ECULIZUMAB ON MYASTHENIA GRAVIS - ACTIVITIES OF DAILY LIVING AND ITS RESPIRATORY, BULBAR, LIMB AND OCULAR DOMAINS IN PATIENTS WITH ACHR+ REFRACTORY GENERALIZED MYASTHENIA GRAVIS

James Howard (Chapel Hill, NC), Jing Jing Wang (New Haven, CT), Fanny O’Brien (Lexington, MA), Renato Mantegazza (Milan, IT)

INTRODUCTION: The myasthenia gravis activities of daily living (MG-ADL) is a patient-reported, validated 8-item questionnaire that emphasizes the functional impact of muscle weakness on activities of daily living and was used for the primary outcome of the 26-week, double-blind, placebo-controlled REGAIN study of anti-acetylcholine receptor refractory generalized myasthenia gravis (gMG) patients.

OBJECTIVE: To evaluate the effect of eculizumab on MG-ADL total score and the sub-domains of respiratory, bulbar, limb, and ocular in patients with refractory gMG in an open-label extension of REGAIN.

METHODS: Patients who completed REGAIN were allowed to continue into Study ECU-MG-302 to receive open-label eculizumab (maintenance dose: 1200 mg every 2 weeks). MG-ADL (primary efficacy endpoint) and safety were assessed over time.

RESULTS: The MG-ADL total score of patients treated with eculizumab/eculizumab (n=56) was unchanged from ECU-MG-302 baseline at each assessment through week 52. The REGAIN placebo patients (n=60) demonstrated change in MG-ADL total score from ECU-MG-302 baseline as early as week 1 (1.6; p<0.0001). The majority of the treatment effect occurred by week 8 (−2.6; p<0.0001), and was sustained through week 52 (−2.7; p<0.0001). A similar pattern of response was seen on the respiratory, bulbar, limb, and ocular domains of the MG-ADL. The most common adverse events were headache (26.5%), nasopharyngitis (23.9%), and diarrhea (14.5%).

SUMMARY/CONCLUSION: The treatment effect of eculizumab to improve overall ADL was sustained through ≥52 weeks of followup and was further evident across the respiratory, bulbar, limb, and ocular domains.

EFFICACY OF ECULIZUMAB IS MAINTAINED BEYOND 26 WEEKS IN PATIENTS WITH ACHR+ REFRACTORY GENERALIZED MYASTHENIA GRAVIS

James Howard (Chapel Hill, NC), Jing Jing Wang (New Haven, CT), Fanny O’Brien (Lexington, MA), Renato Mantegazza (Milan, IT)

INTRODUCTION: Results of the 26-week, double-blind, placebo-controlled REGAIN study suggest that eculizumab is an efficacious and well-tolerated treatment for patients with anti-acetylcholine receptor refractory generalized myasthenia gravis (gMG), as demonstrated by multiple patient- and/or physician-reported assessments—MG activities of daily living (MG-ADL), Quantitative MG (QMG), MG Composite (MGC), and MG quality of life (MG-QoL15).

OBJECTIVE: To evaluate eculizumab efficacy beyond 26 weeks based on MG-specific assessments.

METHODS: Patients who completed REGAIN were allowed to continue into Study ECU-MG-302 within 2 weeks. Each patient underwent an initial 4-week blind induction, before continuing on open-label eculizumab maintenance phase (1200 mg every 2 weeks). MG-ADL, QMG, MGC, and MG-QoL15 were assessed. Safety was also evaluated.

RESULTS: The MG-ADL total score of eculizumab/eculizumab patients (n=56) was unchanged from ECU-MG-302 baseline through week 52. The placebo/eculizumab patients (n=60) demonstrated improvement in MG-ADL total score from ECU-MG-302 baseline rapidly after starting eculizumab (1.6 at week 1; −2.4 at Week 8; both p<0.0001) that was sustained through week 52 (−2.7; p<0.0001). QMG, MGC, and MG-QoL15 total scores followed a similar pattern to MG-ADL (−4.6, p<0.0001; −5.1, p<0.0001, and −5.7, p=0.005, respectively, at week 52). The safety profile of eculizumab remained unchanged through longer exposure in ECU-MG-302 and was consistent with the known profile.

SUMMARY/CONCLUSION: The treatment effect of eculizumab was sustained through ≥52 weeks of followup and across a range of disease measures that included functional ability, muscle strength, and quality of life.
MYASTHENIA GRAVIS WITH GANGLIONIC ACETYLCHOLINE RECEPTOR AUTOANTIBODIES WITHOUT DYSAUTONOMIA
Omer Suhaib (Oklahoma City, OK), Joon-Shik Moon (Oklahoma City, OK), Tyler Webb (Oklahoma City, OK), Eduardo De Sousa (Oklahoma City, OK)

INTRODUCTION: Myasthenia gravis (MG) is an autoimmune neuromuscular junction disorder with fatigable weakness associated with pathogenic acetylcholine receptor antibodies (AChR-Abs) or muscle specific kinase (MuSK-Abs); 6-12% of cases are double seronegative with similar clinical and EDX characteristics to seropositive MG patients. Ganglionic acetylcholine receptor antibodies (GAR-Abs), typically part of a paraneoplastic panel, are present in autoimmune autonomic ganglionopathy patients with dysautonomia including orthostatic intolerance and gastrointestinal (GI) dysmotility. GAR-Abs were also described in seropositive MG patients who later developed dysautonomia, but not in MG without dysautonomia.

OBJECTIVE: To report a case of a seronegative MG patient with GAR-Abs but without dysautonomia.

CASE REPORT: An 80-year-old woman with diabetes, diabetic neuropathy, Graves’ disease, and thyroidectomy presented with fatigable bilateral blepharoptosis, diplopia, dysphagia, dysarthria, dysphonia, head drop, and proximal bilateral upper and lower extremity weakness. AChR-Ab, MuSK-Ab, thyroid-stimulating hormone, and creatine phosphokinase (CPK) were unremarkable. Chest/abdomen/pelvis CTs were unrevealing. Paraneoplastic panel was negative except for elevated GAR-Abs of 0.13 (normal <0.02). Symptoms and signs improved with pyridostigmine and recurred after ciprofloxacin and oxybutynin use. She declined corticosteroids due to prior side effects and current diabetes and partially responded to IV immunoglobulin, not clearly benefiting from longterm azathioprine therapy. She had significant improvement after rituximab. There were no symptoms or signs of dysautonomia (orthostatic intolerance, GI dysmotility, sicca syndrome, palpitations, or resting tachycardia).

SUMMARY/CONCLUSION: Her clinical course and response to treatment was similar to double seronegative MG without dysautonomia. This may be the first case of double seronegative MG with elevated GAR-Abs without dysautonomia. This patient responded to rituximab.

CLINICODEMOGRAPHIC PREDICTORS OF THE IMPACT OF INFECTIONS ON MYASTHENIA GRAVIS
N Kukulka (Columbia, MO), R Govindarajan (Columbia, MO)

INTRODUCTION: Myasthenia Gravis (MG) is an autoimmune neuromuscular disease which utilizes a long term immunosuppressive treatment.

OBJECTIVE: Evaluate the impact of acquired infections on Myasthenia Gravis exacerbations.

METHODS: A retrospective chart review was performed on 127 MG patients treated at the University of Missouri Hospital between 2011 and 2016. All acquired infections were noted for each patient, compared to the immunization records, and analyzed for their significance in MG exacerbation including: ED visits, hospitalizations, admission duration and mortality. Vaccine preventable infections (VPI) included pneumonia and seasonal influenza; vaccine non-preventable infections (VNPI) encompassed the rest.

RESULTS: The studied MG population revealed an average age of 61.9 years, average disease duration of 8.8 years, and a total of 212 flare-ups requiring: 106 ED visits, 141 hospitalizations, and an average admission for 6.2 days. Infections were responsible for: 34% of all MG exacerbations, 44% of ED visits, 40% of hospital admissions and second longest average duration of a hospital admission. Out of the nine flare-up categories contributing to ED visits, VPIs were the primary offender. In the studied population, 69% acquired a VNPI and 29% a VPI. Common VPIs included Pneumonia at 16.5% and Influenza at 11%. The most common VNPI was an upper respiratory infection at 20%. Older patients (both at the diagnosis and current age) were at an increased risk factor for VPIs (p<0.05).

SUMMARY/CONCLUSION: Infections are the highest trigger for MG exacerbations and contribute to ED visits, hospitalizations and prolonged admissions. A prompt re-evaluation of the vaccination guidelines for MG patients is of the upmost importance.
THE BENEFITS AND PITFALLS OF NEXT GENERATION SEQUENCING: 3 ILLUSTRATIVE CASES OF MYASTHENIA
K Scherer (Tucson, AZ)

INTRODUCTION: Next generation sequencing is an accessible, affordable and efficient method for the diagnosis of hereditary neuromuscular disorders. 3 cases demonstrate the clinical utility and potential pitfalls of NGS.

OBJECTIVE: Demonstrate utility and pitfalls of NGS.

METHODS: Case reports.

RESULTS:
Case 1
44-year-old man with several years of arm weakness had normal routine EMG, AchR and MuSK abs, and unremarkable muscle biopsy. Slow RNS showed a 27% decrement. NGS confirmed his diagnosis as CMSTA1. His treatment response will be reported.

Case 2
A 12-year-old boy followed since birth with hypotonia, proximal limb and axial weakness, dropped head, severe ptosis and double vision, and respiratory and feeding difficulties. Muscle biopsy was nonspecific. Pyridostigmine did not help. NGS confirmed a novel mutation in CHRA1 suspected to cause slow channel syndrome, a contraindication to pyridostigmine. He is now on fluoxetine and improving.

Case 3
A 44-year-old man had a 20 year history of mild-moderate limb weakness with no history or findings of any ocular, facial, bulbar, or respiratory dysfunction. Slow RNS showed a 50% decrement. NGS found a heterozygous novel variant in PLEC, suspected to alter mRNA splicing, raising the question of CMS. AchR binding abs subsequently came back elevated at 20.5 nmol/L, and chest CT found a thymoma. His treatment response will be reported.

SUMMARY/CONCLUSION: NGS is affordable and easy, and leads to appropriate diagnosis and treatment in properly selected cases. The clinician must complete laboratory, clinical and electrodiagnostic evaluation before ordering genetic testing to reduce diagnostic confusion due to high sensitivity of NGS for variants.

PEDiatric NEUromuscular Junction Disorders
S Bhatia (Atlanta, GA), S Verma (Atlanta, GA)

INTRODUCTION: Pediatric neuromuscular junction (NMJ) disorders include ocular, generalized myasthenia gravis (MG), congenital myasthenic syndromes (CMS) and infant botulism.

OBJECTIVE: To study the clinical and electrophysiological profile of pediatric NMJ disorders.

METHODS: Retrospective review of clinical presentation, laboratory (antibody, genetic testing), electrophysiology, functional measures and treatment of children followed in the Pediatric Myasthenia Clinic, Atlanta, Georgia from 2013 to 2017.

RESULTS: Forty six children, age 9.8±6.4 years (33 females; M: F 2.5:1), generalized MG 46% (21), ocular MG 22% (10), CMS 20% (9), infant botulism 7% (3) and MUSK 5% (2). Ptosis 91% (42), proximal weakness 67% (31), bulbar weakness 30% (14) and respiratory failure 17% (8) were common presentations. AChR antibody positive 86% (18) generalized and 20% (2) ocular MG. CMS mutations CHRNE (5), CHAT (3), DOK7 (1). Stimulated jitter analysis of orbicularis oculi muscle performed in 85% (39) showed jitter 54±38µs (normal < 26µs) and 26±30 % blocking. Quantitative Myasthenia Gravis and Motor Function Measure performed in generalized MG and CMS. Ocular MG treated with pyridostigmine 100% (10), steroids 60% (6); generalized MG received pyridostigmine 100% (21), steroids 100% (21), IVIG 95% (20), azathioprine 38% (8), thymectomy 38% (8), rituximab 10% (2) and PLEX 10% (2). CMS received pyridostigmine 100% (9), Firdapse™ (3, 4 DAP) 67% (6), albuterol 56% (5) and ephedrine 11% (1). BabyBIG® given to infant botulism.

SUMMARY/CONCLUSION: Generalized MG commonest subset followed by ocular MG and CMS. IVIG and 3, 4 DAP frequently used to treat generalized MG and CMS respectively. Stimulated jitter analysis was sensitive in detecting NMJ defects.
A CASE REPORT OF RECURRENT TAKOTSUBO CARDIOMYOPATHY IN A PATIENT DURING MYASTHENIA CRISIS

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INTRODUCTION: Patients with myasthenia crisis are known to have stress-induced cardiomyopathy secondary to proposed emotional or physical stress leading to high level of circulating catecholamines. We report a patient who developed recurrent Takotsubo cardiomyopathy during myasthenia crisis.

OBJECTIVE: To report a case of a patient with myasthenia gravis (MG) who developed recurrent stress-induced cardiomyopathy during MG crisis.

METHODS: Case report: A 63-year old female with seropositive myasthenia gravis on monthly infusion of intravenous immunoglobulin (IVIG), high dose daily oral steroids and pyridostigmine presented with shortness of breath and sudden onset chest pain. EKG showed ST elevation in anterolateral leads with troponemia. Coronary angiogram was unremarkable for occlusive coronary disease with left ventriculogram showing reduced wall motion with apical and midventricular segment hypokinesis suggestive of Takotsubo variant stress-induced cardiomyopathy. Her symptoms were attributed to MG crisis and echocardiographic findings along with her symptoms resolved completely after five cycles of plasmapheresis (PLEX). She had another similar episode one year later during myasthenia crisis with subsequent resolution in 5 days after PLEX.

SUMMARY/CONCLUSION: Takastubo cardiomyopathy can be one of the manifestations of Myasthenia crisis and needs meticulous cardiac monitoring in addition to respiratory parameters while administering rescue treatments like IVIG and PLEX.

WEST NILE VIRUS INDUCES A PROLONGED PROINFLAMMATORY STATE THAT MAY PROMOTE MYASTHENIA GRAVIS

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INTRODUCTION: New cases of serologically confirmed myasthenia gravis (MG) have been reported to occur several months after West Nile virus (WNV) infection. We offer a perspective on pathogenic mechanisms involved in WNV that may also contribute to the development of MG.

OBJECTIVE: To show that WNV increases expression of immunoregulatory proinflammatory proteins S100B, interleukin 17-A (IL-17A), and osteopontin (OPN) in human blood and tissues. These proteins are implicated in the development of human and experimental autoimmune MG.

METHODS: Human in vitro experiments and clinical studies.

RESULTS: Following WNV infection, we found that astroglial S100B is pathologically elevated in CSF, serum, and autopsy tissue. WNV also induced expression of IL-17A and OPN in human blood and tissues. Importantly, both IL-17A and OPN remained pathologically elevated in a subgroup of patients that recovered from WNV neuroinvasive disease in prior years.

SUMMARY/CONCLUSION: Proinflammatory proteins may remain elevated long after recovery from acute WNV infection. These proteins play an important role in autoimmune diseases: S100B activates the receptor for advanced glycation end products (RAGE), which amplifies autoimmunity and has been shown to promote human and experimental autoimmune MG (EAMG). IL-17A production by CD4+ T cells also plays a causal role in many autoimmune diseases and has been reported to contribute to loss of B-cell tolerance in EAMG. OPN, a multifunctional proinflammatory cytokine, promotes autoimmunity and tumorogenesis, and has recently been shown to be elevated in MG patients.

Collectively, these findings suggest WNV may represent an additional risk factor for MG.
SEQUENTIAL INTRAVENOUS IMMUNOGLOBULIN AND PLASMAPHERESIS THERAPY IN A SERONEGATIVE MYASTHENIA GRAVIS PATIENT WITH COMMON VARIABLE IMMUNODEFICIENCY DISEASE AND REFRACTORY WEAKNESS

G Small (Pittsburgh, PA)

INTRODUCTION: Both myasthenia gravis (MG), prevalence 1/5,000, and CVID, prevalence 1/30,000, are uncommon, immunopathogenic diseases. The individual we report with both diseases is an exceedingly rare finding, and this author only notes an individual on social media who may have both diseases, but not reported in the medical literature. Our reported patient is seronegative for binding, blocking, modulating and muscle specific kinase (MuSK) antibodies. Her repetitive stimulation testing negative, a frontalis muscle single fiber electromyogram positive, no other cause for her somatic and respiratory muscle weakness was identified. Fluctuating ptosis of her right levator palpebrae is chronically evident. Her clinical condition has only minimally improved, but not stabilized with IVIG, corticosteroids, thymectomy, pyrodistigmine and mycophenolate mofetil administration. Repeated hospitalizations requiring assisted ventilation are currently avoided with monthly IVIG therapy (2 grams/kilogram), and monthly plasmapheresis sessions.

OBJECTIVE: To highlight an unusual, but necessary therapeutic regimen for a critically ill seronegative MG patient

METHODS: Case report

RESULTS: Although therapeutic apheresis occurring close to IVIG administration is anti-intuitive for treating CVID, a disorder requiring exogenous immune globulin repletion, the patient's immunologist agreed the severity of her myasthenia symptoms, particularly ongoing respiratory failure, without other means of therapy besides apheresis, was reasonable. CVID patients frequently suffer other autoimmune disorders, among them pernicious anemia and connective tissue diseases, with negative biomarkers for these disorders. The patient ambulates freely and does not require assisted ventilation with this current therapeutic course.

SUMMARY/CONCLUSION: Severely ill MG patients may require novel approaches to treatment. This case may provide further insight into the seronegative MG state.

B-CELL DEPLETION IN LATE-ONSET MYASTHENIA GRAVIS IS SAFE AND EFFECTIVE; A CASE SERIES

S Sahai (Los Angeles, CA), R Lewis (Los Angeles, CA), A Maghzi (Los Angeles, CA)

INTRODUCTION: Late-onset myasthenia gravis (MG), defined as onset over 50 years of age, has therapeutic challenges different than earlier onset. Although treatment approaches for both groups are considered to be similar, the risks associated with immunosuppression are different given the advanced age.

OBJECTIVE: We aimed to report the safety and efficacy of B-cell depletion in late-onset MG.

METHODS: We retrospectively report five patients with late-onset MG who were treated with Rituximab. Results: The mean age of onset was 65 (range 53-74), four were female, and all were anti-AChR-positive (titers ranging 4.88-46.97). Two of five patients had a thymoma and underwent thymectomy. All patients presented with a variation of bulbar symptoms; two patients went into crisis requiring intubation prior to Rituximab. Three patients were refractory to prednisone therapy and three developed side effects including cushingoid features, shingles, and osteoporotic hip fracture. All patients were given two doses of Rituximab one gram IV infusions, two weeks apart. These patients were followed for 12-24 months and showed dramatic improvement in symptoms. All patients were able to discontinue or markedly reduce prednisone, plasmapheresis, and IVIG. Two patients did not require repeat Rituximab infusions for over one year.

SUMMARY/CONCLUSION: Based on this case series, B-cell depletion in late-onset MG appears to be effective for inducing remission in older patients with generalized anti-AChR-positive MG. The efficacy and side effect profiles of Rituximab compare favorably with prednisone, cyclosporine, azathioprine, and mycophenolate. It is appropriate to consider B-cell depletion as a first-line immunomodulatory treatment.
**PROLONGED POST TETANIC POTENTIATION**  
*L Gutmann (Iowa City, IA), M Shy (Iowa City, IA)*

**INTRODUCTION:** Post tetanic potentiation (PTP), seen with disorders of the neuromuscular junction (NMJ), reflects influx of Ca++ into terminal axons during tetanus, resulting in increased number of acetylcholine (Ach) vesicles released by each axonal action potential. In the pre-synaptic Lambert-Eaton syndrome there is increased amplitude of initially small muscle action potentials post-tetanically that persists 1-2 minutes. Prolongation of post tetanic potentiation (PTP) has now been reported in 2 disorders, one a genetically determined neuropathy/myasthenic disorder and the other a toxin.

**OBJECTIVE:** Discuss prolonged PTP and its relationship to synaptotagmin II (SYT2).

**METHODS:** Review literature on prolonged PTP

**RESULTS:** Prolonged PTP, up to 60 minutes, was reported in 2 families with a motor neuropathy and congenital myasthenic syndrome (having leg weakness and foot deformities) caused by heterozygous mutations of the synaptotamin II (SYT2) gene. The same phenomenon has been noted in botululism with PTP continuing up to 21 minutes. SYT2 is the synaptic vesicle calcium sensor in the terminal axon, allowing for fusion of Ach containing vesicles with the presynaptic membrane and synchronous release of Ach. The fusion requires a complex assembly process involving SNARE proteins. Both the SYT2 gene mutation and botulimum toxin affect normal SYT2 function, the mutation by altering amino acids in the calcium-binding domain and the toxin by binding to SYT2 as well as gangliosides GD1a and GT1b on the neural membrane. The mechanism for the PTP prolongation remains unknown.

**SUMMARY/CONCLUSION:** Prolonged PTP is a unique physiological abnormality that may represent a marker for a presynaptic NMJ defect involving altered SYT2.

**FOOD-BORNE BOTULISM: DIAGNOSTIC CHALLENGES**

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**INTRODUCTION:** The State of California has one of the highest incidences of botulism type A in the United States. The month of April shows a peak incidence of food-borne botulism.

**OBJECTIVE:** To describe two illustrative cases of food-borne botulism in Northern California in April 2017 and to analyze factors delaying antitoxin administration.

**METHODS:** Clinical evaluation, electrodiagnosis, mouse neutralization bioassay and C.Botulinum cultures.

**RESULTS:** An 85 year old woman developed bilateral ptosis, proximal limb and bulbar weakness. She was initially treated with IVIG for possible myasthenia gravis in another institution. However, an electrodiagnostic evaluation in our hospital showing small compound muscle action potential (CMAP) amplitudes alerted the possibility of botulism and she received the antitoxin one week after the onset of symptoms. The patient developed pneumonia, sepsis and eventually died. The diagnosis of botulism was strongly suspected based on positive cultures for C.Botulinum from feces. A second case of a 16 year old boy developed ophthalmoparesis, generalized weakness and respiratory failure. Based on the finding of small CMAP amplitudes a tentative diagnosis of botulism was made. The patient received the antitoxin within 24 hours of admission and had a favorable course. The diagnosis of botulism type A was made based on positive bioassay and positive cultures from gastric, fecal and food material.

**SUMMARY/CONCLUSION:** The diagnosis of botulism should always be suspected facing an acute descending paralysis. Small CMAP amplitudes in affected muscles are the most constant and reliable electrodiagnostic finding. Early antitoxin administration is fundamental for achieving a favorable outcome.
PRESCRIBING PATTERNS AND IMPLICATIONS OF MEDICATIONS KNOWN TO EXACERBATE MYASTHENIA GRAVIS
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INTRODUCTION: Multiple medications are known to exacerbate myasthenia gravis (MG). In some cases, prescription of these medications results in symptomatic worsening or even MG crisis. We hypothesize that these offending medications are prescribed to MG patients regularly and contribute to increased morbidity and cost of care. Currently there are no universally accepted tools to systematically reduce the prescription of these medications for myasthenics.

OBJECTIVE: Characterize the frequency and sequelae of ordering and administering medications that have the potential to exacerbate MG. This data will inform the development of a "drug-disease" interaction alert in the electronic medical record. We hypothesize that an alert will help reduce prescription of offending medications and improve overall health outcomes for MG patients.

METHODS: Single-center retrospective analysis of all MG patients for whom an order for a medication known or suspected to exacerbate MG symptoms was placed in the inpatient setting from 01/01/2010 to 12/31/2016. Prescription incidence will be quantified. Hospital length of stay, MGFA-PIS score, orders for intravenous immunoglobulin or plasma exchange, ICU admission rate, intubation rate, and mortality will be used to determine sequelae of medication prescription.

RESULTS: The University of Vermont Jeffords Quality Institute identified approximately 300 MG patients for analysis. Data review is ongoing.

SUMMARY/CONCLUSION: This study will provide data about the incidence and sequelae of offending medication prescriptions in the inpatient setting. Data will guide development of a "drug-disease" interaction alert to help reduce the incidence of offending medication prescriptions in myasthenics. Final study results will be presented.

CURATIVE STRATEGIES IN MYASTHENIA GRAVIS
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INTRODUCTION: The International Myasthenia Gravis Thymectomy trial (MGTX) has proven the role of thymectomy in myasthenia. Improvements in clinical status, including symptoms, adverse events and dose of steroids were all documented. However, rapid time to improvement and complete symptomatic remission (CSR) off all medication remains elusive. In part, this may reflect the inability of surgery to eradicate all responsible immunologic tissue and cellular memory. Clinical trials utilizing targeted immunologic induction followed by thymectomy may offer a more durable and rapid cure for myasthenia.

OBJECTIVE: Develop curative clinical treatment strategies and trials to enable early and complete remission of myasthenia gravis. Immuno-therapy targeted induction protocols, B and or T cell based, as directed by the particular type of myasthenia, followed by minimally invasive extended thymectomy. Figure 1.

METHODS: Multi-institutional trials of induction therapy for myasthenia gravis followed by thymectomy. Induction protocols directed by the antibody status of the patient, and known responses to certain immunologic therapy such as Rituximab. Limited phase II trials would enable rapid insight into the effectiveness of this strategy given the overall low early CSR rates of present therapy.

SUMMARY/CONCLUSION: Extended Thymectomy has been proven to improve the clinical course of myasthenia gravis. Directed immunologic therapy is quickly developing a role in the treatment of myasthenia. True cure, and rapid time to complete symptomatic remission are still lacking. Neurologist and surgeons should work together to develop and trial curative multidisciplinary approaches to myasthenia gravis. Combined Immunologic Induction and thymectomy should be investigated in multi-institutional trials.
TELE-REHABILITATION IMPROVE FUNCTION AND BALANCE IN MYASTHENIA GRAVIS
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INTRODUCTION: Weakness and fatigue in myasthenia gravis (MG) causes weakness and fatigue, which produces fear of falling and limitations with sustained activities. There is a need for rehabilitation programs that promote muscular endurance and better balance for adults with MG.

OBJECTIVE: Determine whether a program, introduced at the point-of-care in neurology and infusion clinics, is safe, well accepted, and enhances functional performance.

METHODS: Our progressive exercise program combines aerobic conditioning, muscular endurance, balance training, and pulmonary rehabilitation to improve gait and balance. Personalized exercises match each client's capacity and progress according to a standardized protocol based on client's achievement of milestones. For broader outreach, we embed this program on an interactive video exercise tele-rehabilitation (IVET) platform. A video avatar guides home exercise, while video captures client's performance to assure adherence, increase safety, and enhance visual feedback for unprecedented gains in proper body mechanics.

RESULTS: Twelve clients with a broad age range (52.8±17.4; range 25-73 years) and MG severity (IIa to IVb) showed a high level of acceptance for home exercise using this technology platform using a nominal ten-point scale (high rating for enjoyment, interest, confidence, safety, and perceived value). Trainers rate 75% of clients as safe and 50% as competent on exercises after a single training session. Training is ongoing to determine gains in gait and balance. Further studies are needed to determine the treatment fidelity, safety, and efficacy of IVET as a virtual therapist to improve function and health for individuals with MG.

ECULIZUMAB IN REFRACTORY MYASTHENIC CRISIS
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INTRODUCTION: No evidence based studies for treatment options in refractory myasthenia crisis exist. Rituximab improved respiratory status in two case studies. Eculizumab improved clinical outcomes in acetylcholine receptor antibody-positive refractory myasthenia gravis in REGAIN Phase-3. It has not been reported in refractory myasthenic crisis.

OBJECTIVE: To report promising results with eculizumab in refractory myasthenic crisis.

METHODS: A 79-year-old woman with hemolytic uremic syndrome, in remission after eculizumab and rituximab, presented with acute respiratory failure and weakness. She reported intermittent ptosis 1 year prior to presentation. Acetylcholine receptor, muscle specific kinase, LRP4, voltage-gated calcium channel, and striatal antibodies were negative. CT showed no thymoma. Repetitive nerve stimulation showed decrement consistent with myasthenia gravis. She received 5 plasma exchanges, improved, and was discharged on home oxygen. Days later, she was readmitted and intubated for severe respiratory distress. Despite pyridostigmine, corticosteroids, and IV immunoglobulin (IVIg), she remained ventilator dependent necessitating tracheostomy at 23 days ventilation. CD20/CD19 counts were negligible. At 25 days ventilation, she received eculizumab per the REGAIN phase 3 protocol. One week after starting, she tolerated pressure support weaning. Four days later, continuous positive airway pressure was tolerated. At 3 weeks, she tolerated tracheostomy collar, and at 4 weeks, tolerated it for 12-15 hours daily, walking around without ventilator support. At 6 weeks, she tolerated 48 hours tracheostomy collar. Muscle weakness improved significantly.

SUMMARY/CONCLUSION: Myasthenic crisis is usually reversible, however refractory cases to IVIg, plasma exchange, and rituximab exist. Our experience suggests eculizumab may be effective in refractory myasthenic crisis, and benefit seronegative myasthenia gravis patients.
PROGNOSTIC FACTORS OF THYMECTOMIZED PATIENTS WITH ACETYLCHOLINE RECEPTOR ANTIBODY POSITIVE MYASTHENIA GRAVIS


INTRODUCTION: Thymectomy remains the standard therapy for myasthenia gravis (MG). However, factors that influence the response to thymectomy still remain controversial.

OBJECTIVE: The aim of the study was to elucidate clinical variables that were associated with remission of thymectomized patients with acetylcholine receptor antibody (AchR Ab) positive MG.

METHODS: The records of 195 patients who underwent thymectomy for MG were analyzed retrospectively. All patients were positive with AchR Ab. We tried to identify the prognostic factors for clinical remission.

RESULTS: Of 195 patients who underwent thymectomy, 117 patients had thymomatous MG, 67 had thymic hyperplasia, and 17 displayed atrophy. The mean follow-up time was 8.2±5.6 years. At the time of the last clinical review, 53% of our patients (103/195) had achieved remission, including 11% of the patients (21/195) with complete stable remission (CSR). Non-thymoma pathology was a good prognostic factor for remission (HR: 1.55, 95% CI: 1.05-2.29, p = 0.026). In subgroup analysis of patients with thymoma, the AchR Ab titer was a significant prognostic factor (HR: 0.89, 95% CI: 0.82-0.97, p = 0.011).

SUMMARY/CONCLUSION: Non-thymoma pathology was a favorable factor for remission. The AchR Ab titer was a prognostic factor in thymomatous MG patients, but not in non-thymomatous MG patients. These findings may implicate the pathogenic role of thymus in AchR Ab positive MG.

Efficacy of Eculizumab Is Sustained Over 52 Weeks in Patients with ACHR+ Refractory Generalized Myasthenia Gravis: Interim Results from the Open-Label Extension of REGAIN

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INTRODUCTION: Eculizumab was effective and well-tolerated in the 26-week, randomized, double-blind, placebo-controlled REGAIN study of patients with AChR+ refractory generalized myasthenia gravis (gMG), demonstrated through multiple MG-specific assessments (MG-ADL, QMG, MGC and MG-QOL15).

OBJECTIVE: An open-label extension of REGAIN (ECU-MG-302) was conducted to evaluate the long-term safety and efficacy of eculizumab in patients with refractory gMG beyond 26 weeks.

METHODS: Of 118 patients who completed REGAIN (NCT01997229), 117 enrolled in ECU-MG-302 (NCT02301624) to receive open-label eculizumab (4-week blind induction then 1200 mg every 2 weeks). MG-ADL, QMG, MGC, MG-QOL15 and safety were assessed for two groups: eculizumab in REGAIN/eculizumab in ECU-MG-302; placebo in REGAIN/eculizumab in ECU-MG-302.

RESULTS: The effect of treatment on MG-ADL total score and MG-ADL respiratory, bulbar, limb and ocular domains, and QMG, MGC and MG-QOL15 total scores was maintained through week 52 in the eculizumab/eculizumab group (n=56). In the placebo/eculizumab group (n=60), improvement in MG ADL total score from open-label baseline was rapid (~2.4 at week 4; p<0.0001) and sustained through week 52 (~2.7; p<0.0001). Similar rapid, sustained improvements were seen in MG-ADL respiratory, bulbar, limb and ocular domains, QMG, MGC and MG-QOL15 total scores. The safety profile of eculizumab was consistent with the known profile.

SUMMARY/CONCLUSION: The treatment effect of eculizumab was sustained through ≥52 weeks and across measures of functional ability, muscle strength, quality of life and activities of daily living, including the respiratory, bulbar, limb and ocular domains. The response to eculizumab in the placebo/eculizumab group was consistent with that of eculizumab-treated patients in REGAIN.

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 IMMUNE PROFILING OF CIRCULATING FOLLICULAR HELPER T CELLS IN MUSK MYASTHENIA GRAVIS PATIENTS

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INTRODUCTION: Myasthenia gravis (MG) is an autoantibody-dependent, CD4 T-cell mediated autoimmune disease of the neuromuscular junction characterized by muscle fluctuating weakness and fatigability. A subset of CD4 T-cells critical to the development of autoantibody producing memory B-cells and plasmablasts are follicular helper T (Tfh) cells. Tfh cells promote B-cell responses through CD40L co-stimulation and interleukin-21. Although interaction between B-cells and Tfh cells occurs in lymphoid organ germinal centers, circulating Tfh cells have been identified and described as memory Tfh cells. In acetylcholine receptor antibody MG, circulating Tfh cell frequencies are elevated and they are superior at inducing antibody production compared with healthy controls. Data on Tfh cells in MuSK antibody MG is nonexistent.

OBJECTIVE: The objective of this study is to perform in-depth immune profiling of circulating Tfh cells in MuSK-MG patients compared with healthy controls.

METHODS: We will use polychromatic flow cytometry to identify circulating CD4+CD45RA-CXCR5+ Tfh cells from 20 MuSK-MG patients. Additional markers, including PD-1, ICOS, CXCR3, CCR6, IFN-γ, IL-2, IL-4, IL-17A and FOXP3, will define Tfh cell subsets, including Tfh1, Tfh2, Tfh17, and regulatory follicular T cells. We will measure Tfh cell functional capacity in in-vitro assays that measure total IgG production after culturing sorted Tfh cells with sorted naïve B cells.

RESULTS: This remains an ongoing study and we will share the results at the MGFA symposium.

Summary/Conclusion: Understanding Tfh cells in MuSK MG will lead to a better understanding of disease immunopathogenesis and may be a useful clinical biomarker or therapeutic target.

 AUTOIMMUNE LIMB GIRDLE MYASTHENIA

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INTRODUCTION: Limb girdle myasthenia (LGM) presents with prominent weakness of proximal limb muscles relatively sparing ocular and bulbar muscles. Etiology is more often inherited.

OBJECTIVE: To describe the presentation and clinical course of two patients diagnosed with LGM

METHODS: Retrospective chart review of patients

Results: AL, a 12 year old girl presented with complaints of lower extremity weakness after long distance running. On examination, she had mild weakness of the pelvic girdle muscles. Routine nerve conduction studies were normal, but repetitive nerve stimulation (RNS) showed decremental response in proximal muscles. Creatine kinase (CK) was normal and acetylcholine receptor antibody was positive. Pyridostigmine partially improved her weakness. Patient and parents refused steroid therapy. Eventually, she achieved remission after thymectomy. MB, a 16 year old girl, with celiac disease and Hashimoto’s thyroiditis, presented with complaints of delayed weakness in lower extremities after cheerleading and jazz dancing. Routine nerve conduction studies were normal, but RNS showed decremental response. CK was normal with positive acetylcholine receptor binding antibodies. Treatment with Pyridostigmine and low dose Prednisone improved exercise endurance.

SUMMARY/CONCLUSION: Autoimmune LGM is a rare clinical presentation and should be considered in patients presenting with fluctuating limb girdle muscle weakness.
A REDCAP DATABASE FOR THE MYASTHENIA GRAVIS CLINIC AND MULTICENTER COLLABORATIONS
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Since 1980, data on all patients seen in the Duke Myasthenia Gravis (MG) Clinic have been stored in a database, which was initially developed to track clinical information on our patients. A summary report of clinically-pertinent patient information is printed at each clinic visit and provides physician incentive to keep the database information current and accurate. The database now contains information on >17,000 clinic visits made by >1,800 patients with neuromuscular junction diseases. We have used data in the database in more than 60 publications and scientific presentations on the clinical features, electrodiagnostic and immunologic findings, treatments and patient outcomes.

We recently converted the database to REDCap (Research Electronic Data Capture) in order to have a readily-sharable web-based format that is compatible with current operating systems, and that contains multiple quality assurance and high-level HIPAA-compliant security features. REDCap is specifically designed to support data capture for collaborative research studies and is widely used in the academic research community, with more than 2000 institutional partners in over 100 countries.

Data in the database comply with the NIH-developed MG Common Data Element definitions to assure consistency among collaborating institutions. The REDCap database contains forms for capturing many scores and measures used for quantitating severity and outcomes, including the MGFA Clinical Class and Post-Intervention Status, MG-MMT, MG-ADL, MG-Composite and MG-QOL15R scores.

We will make our REDCap database available for use by MG Clinics that wish to maintain their data in a standardized format that will facilitate collaborations, especially for Comparative Effectiveness Research studies.

AN INSTITUTIONAL RETROSPECTIVE STUDY OF RITUXIMAB EFFECTIVENESS IN MYASTHENIA GRAVIS
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INTRODUCTION: Rituximab is a chimeric monoclonal antibody that binds CD20 and causes the depletion of B-Cells. Although initially designed to treat lymphoma, it has found wide use in the management of autoimmune conditions. Over the past several years, many case-series have shown its efficacy in the management of myasthenia gravis (MG), an auto-immune disorder of the neuromuscular junction.

OBJECTIVE: To determine the effectiveness of rituximab in treating various subtypes of myasthenia gravis.

Methods: This is retrospective chart review study of patients with myasthenia gravis seen in follow-up at the Johns Hopkins Hospital between January 1, 2015 and December 31, 2016.

RESULTS: We present results from 21 patients at our institution treated with rituximab: 10 patients with MuSK antibodies, 7 with AChR antibodies and 4 double sero-negatives. Age range was 25-79, with an average of 45 (+/- 17) years old. All patients responded positively to treatment. Patients received between 1 and 6 cycles of rituximab treatment. Steroid sparing agents were successfully stopped in 93% (15/16) of patients, usually at the time of their first infusion. Of those taking prednisone, 55% (10/18) were able to lower their daily dose, while 28% (5/18) were able to completely wean off steroids. Antibody titers decreased dramatically over time in 100% (10/10) MuSK patients.

SUMMARY/CONCLUSION: Our data suggests rituximab is highly effective in the management of all sub-types of myasthenia gravis.
REPORTING PATIENT REPORTED OUTCOME MEASURES IN MYASTHENIA GRAVIS PATIENTS PRESCRIBED INTRAVENOUS IMMUNOGLOBULIN IN THE HOME SETTING USING CAREEXCHANGE®
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INTRODUCTION: Intravenous immunoglobulin (IVIg) is an accepted treatment for myasthenia gravis (MG) crisis and is increasingly being used as maintenance therapy. MG patient reported outcome measures (PROMs) have been developed to report objective response to therapy. Specialty pharmacy gives a unique ability to provide real-time PROMs data collection in the home setting by a trained home infusion nurse. CareExchange is a PROMs system that captures and integrates pharmacy data, dosing, infusion nursing assessment data, physician progress notes, patient measured outcomes data, and patient response to therapy.

OBJECTIVE: To retrospectively review and analyze patient records and PROMs when IVIg therapy was prescribed by a treating physician and infused by BriovaRx Infusion Services (formerly AxelaCare) in the home setting.

Methods: PROMs and patient records were pulled from CareExchange and a Clinical Patient Records (CPR+) database, including demographics and dosing patterns (e.g. IVIg dose received per infusion (g/kg) and frequency).

RESULTS: A review of records was performed on over 300 MG patients prescribed IVIg by more than 170 physicians. Patient demographics, dosing regimens, interval of treatments, and a summary of PROMs will be presented. Such PROMs reporting will include respiratory assessments (NIF, PEF, FEV1), disability assessments (MGC, MG-ADL, mFSS, VAS), and QOL (MG-QOL15, SF-36, CHQ-PF28).

SUMMARY/CONCLUSION: CareExchange allows the collection of information not readily available to treating physicians between routine office visits and IVIg infusions. By using CareExchange, physicians may be able to optimize therapy and clinical outcomes in myasthenia gravis patients and provide future research opportunities.

COST COMPARISON BETWEEN RITUXIMAB, PLASMAPHERESIS AND INTRAVENOUS IMMUNOGLOBULIN FOR REFRACTORY MUSK ANTIBODY POSITIVE MYASTHENIA GRAVIS
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INTRODUCTION: Management of refractory Myasthenia Gravis (MG) often relies on frequent dosing of intravenous immunoglobulin (IVIg) or plasmapheresis (PLEX). Mounting evidence supports an excellent response to Rituximab, especially when MuSK positive, but insurance denials remain frequent. All three treatments are expensive but exact cost disparities are not widely known.

OBJECTIVE: Define costs associated Rituximab, PLEX, and IVIg in refractory MG.

METHODS: We constructed a Markov model to capture discounted lifetime costs associated with Rituximab, PLEX, and IVIg for a 25-year old female with refractory MuSK positive MG. Drug costs derive from Medicare average sales pricing, April 2017 (Rituximab, Gamunex-C, Albumin 5%) and infusion costs from up-to-date cms.gov physician fee schedule CPT codes. Dose assumptions included: IVIg 70gm every 3 weeks; PLEX every 10 days; Rituximab 375mg/m2 x 4, twice yearly for life. Efficacy was not accounted for; all treatments assumed as near equivalent for the purposes of this analysis. TreeAge Pro was used for all cost-analyses and discounting was 3% per year.

RESULTS: Rituximab, PLEX, and IVIg were associated with lifetime costs of $655,800, $1,323,300, and $2,210,380 respectively.

SUMMARY/CONCLUSION: Lifelong Rituximab costs approximately half that of PLEX and one-third that of IVIg, given our above assumptions. Such information should aid in supporting insurance authorizations for Rituximab use in refractory MG. To account for remission potential, variation in effectiveness and alternate dosing regimens, a cost-utility analysis would be further informative.
FEATURES OF LAMBERT-EATON MYASTHENIC SYNDROME WITH REPETITIVE NERVE STIMULATION IN A PATIENT WITH KNOWN MYASThenIA GRAVIS

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INTRODUCTION: Slow repetitive nerve stimulation at 2-3 Hz followed by 10 seconds of maximal voluntary exercise is frequently used to evaluate for disorders of the neuromuscular junction. We report a patient with known myasthenia gravis with features of Lambert-Eaton myasthenic syndrome (LEMS) on EDX testing.

OBJECTIVE: To describe a case of myasthenia gravis with some EDX features of LEMS.

METHODS: CASE REPORT: A 43-year-old male with a history of generalized weakness was evaluated as a second opinion on a prior diagnosis of myasthenia gravis. Fibular and median nerve proximal compound muscle action potential (CMAP) amplitudes were decreased at 0.3 mV and 3.5 mV, respectively. With 2 Hz repetitive nerve stimulation of the fibular nerve at the ankle and median nerve at the wrist, decrements of 29% and 60% were demonstrated, respectively. Post-exercise incremental responses were 12% and 77%, respectively, and there was repair of the fibular nerve decremental response. Needle examination showed unstable motor unit potentials in all tested muscles. A repeat study 15 months later showed mildly decreased CMAP amplitude only in the fibular nerve (3.4 mv) and did not show an incremental response post-exercise. VGCC antibodies were negative, and AChR binding antibodies were present at 11.4 nmol/L.

SUMMARY/CONCLUSION: This case illustrates that some patients may have features of both post- and presynaptic neuromuscular junction disorders with repetitive nerve stimulation.