ALS: Making the Diagnosis

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Disclosures

- No relevant financial relationships to disclose
- No conflicts of interest
- No discussion of off-label or investigational drug use

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Goals

By the end of this talk, you should be able to:

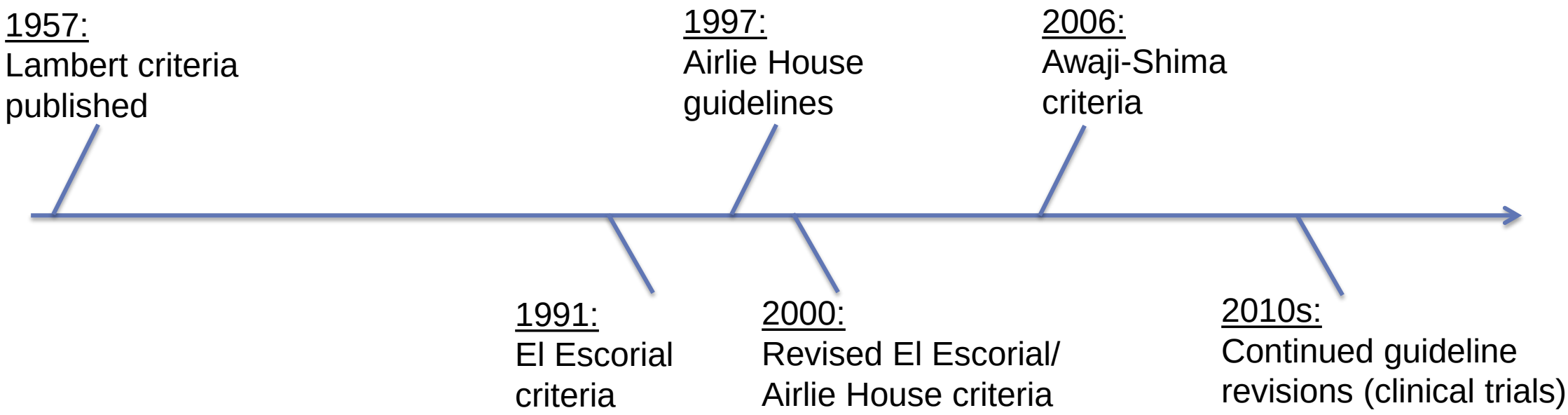
- Describe the approach to diagnosing ALS using existing clinical criteria
- Identify common electrodiagnostic (EDX) findings in ALS
- Recognize EDX findings that may prompt evaluation for mimickers

Diagnosing ALS

- Can be challenging
  - o Clinical diagnosis
  - o Phenotypic heterogeneity
  - o Early in disease course

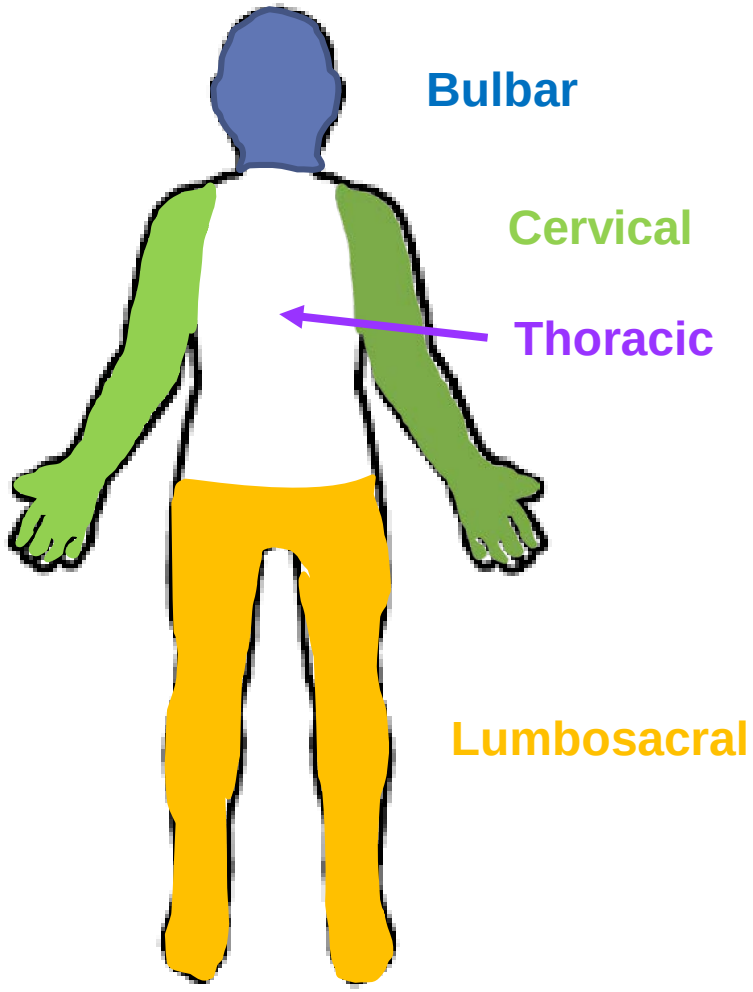
- Most do not meet criteria for a definite diagnosis early on
  - o Limitations to existing criteria

History of ALS Diagnostic Criteria



Regions/segments

- UMN signs
  - Weakness without atrophy
  - Hyperreflexia (preserved reflexes in atrophic muscles)
  - Primitive reflexes (Babinski sign)
  - Spasticity
- LMN signs
  - Weakness with atrophy
  - Low or normal tone
  - Reflex loss
  - Fasciculations



Revised El Escorial Criteria

Degree of diagnostic certainty	Definition
Definite ALS	UMN + LMN signs in **3 regions**
Probable ALS	UMN + LMN signs in **2 regions** (some UMN signs rostral to LMN)
Laboratory-supported probable ALS	UMN+ LMN signs in **1 region** OR UMN in **≥ 1 region(s)** **+** EMG evidence of active denervation in **≥ 2 segments** (≥ 2 muscles of different root and nerve origin)
Possible ALS	UMN + LMN signs in **1 region** UMN signs only **≥ 2 regions** LMN signs rostral to UMN signs

Fibrillations or positive sharp waves

Brooks et al. Revised El Escorial Criteria, 2000.

Awaji Criteria

Degree of diagnostic certainty	Definition
Definite ALS	UMN + LMN signs in **3 regions**
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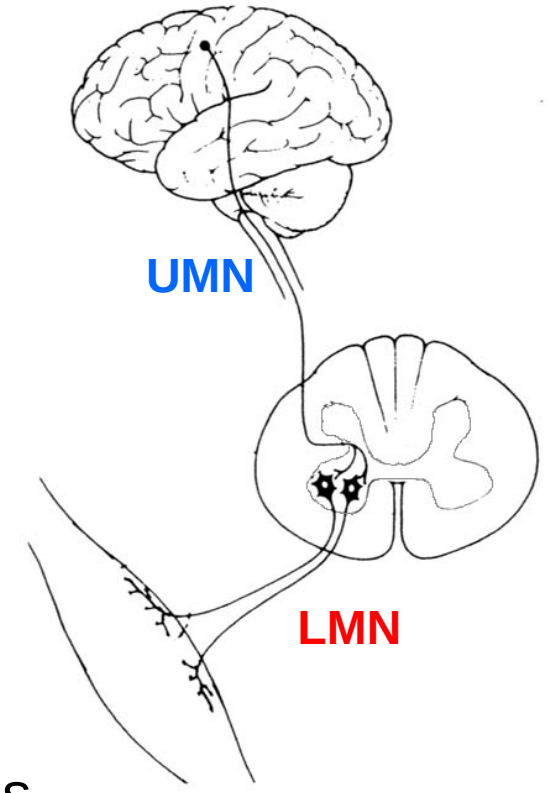


- Includes both clinical and EMG findings
- Active denervation
  - Fibrillations or positive sharp waves
  - Fasciculations with chronic neurogenic changes
- Literature suggests helpful in bulbar-onset ALS for earlier diagnosis

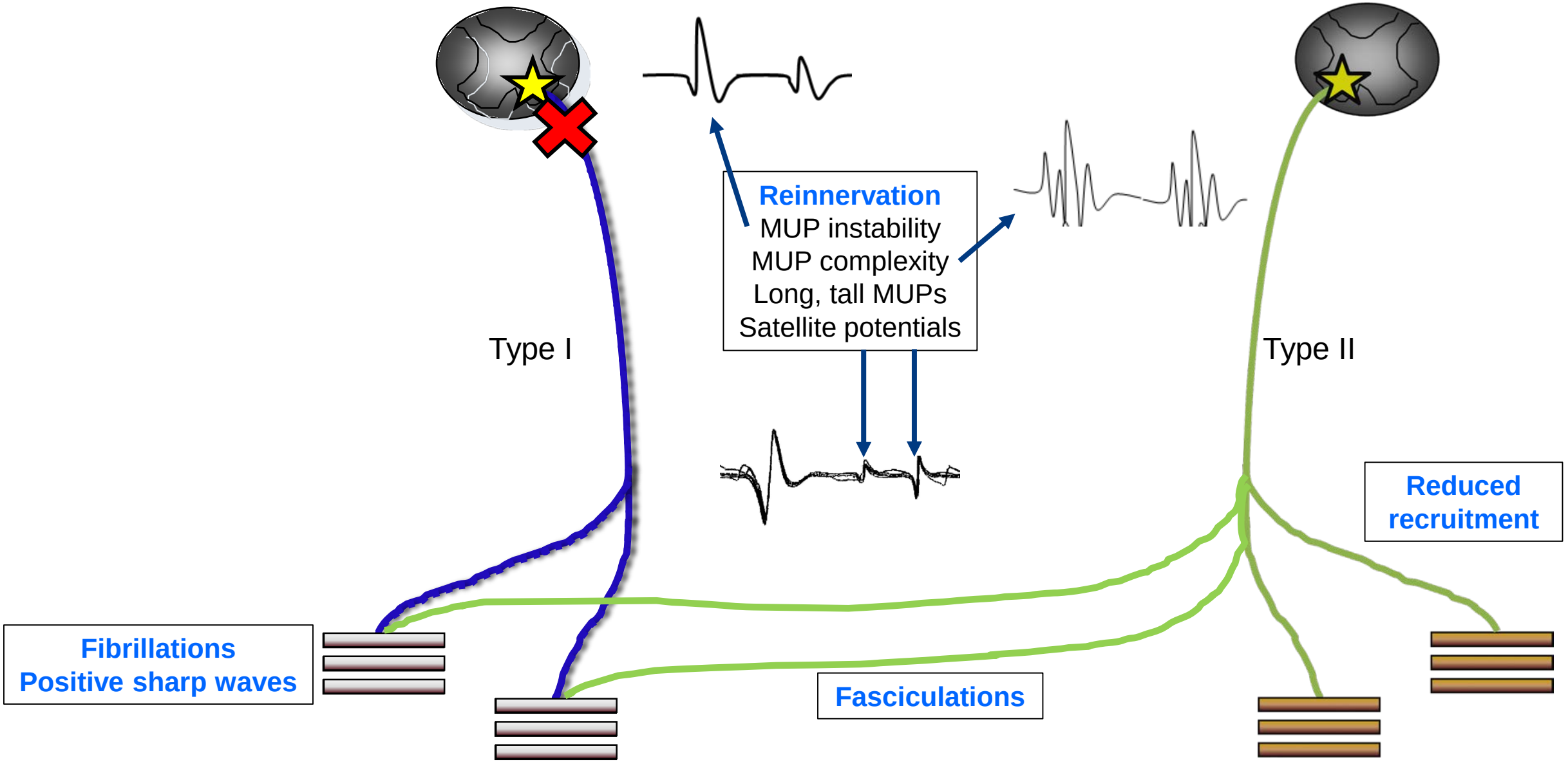
de Carvalho et al. Electrodiagnostic criteria for diagnosis of ALS, 2008.
Geevasinga et al. Awaji criteria improves diagnostic sensitivity in ALS, 2016.

Why perform EMG/NCS?

- Confirm extent of lower motor neuron involvement

- Exclude ALS mimickers
  - Motor-predominant neuropathies (CIDP, MMN)
  - Inclusion body myositis
  - BFS or CFS
  - Other motor neuron diseases (need clinical context)
    - SMA, SBMA, PLS, PBP

Common EMG/NCS Changes


Reinnervation
MUP instability
MUP complexity
Long, tall MUPs
Satellite potentials
Type I
Type II
Reduced recruitment
Fibrillations
Positive sharp waves
Fasciculations

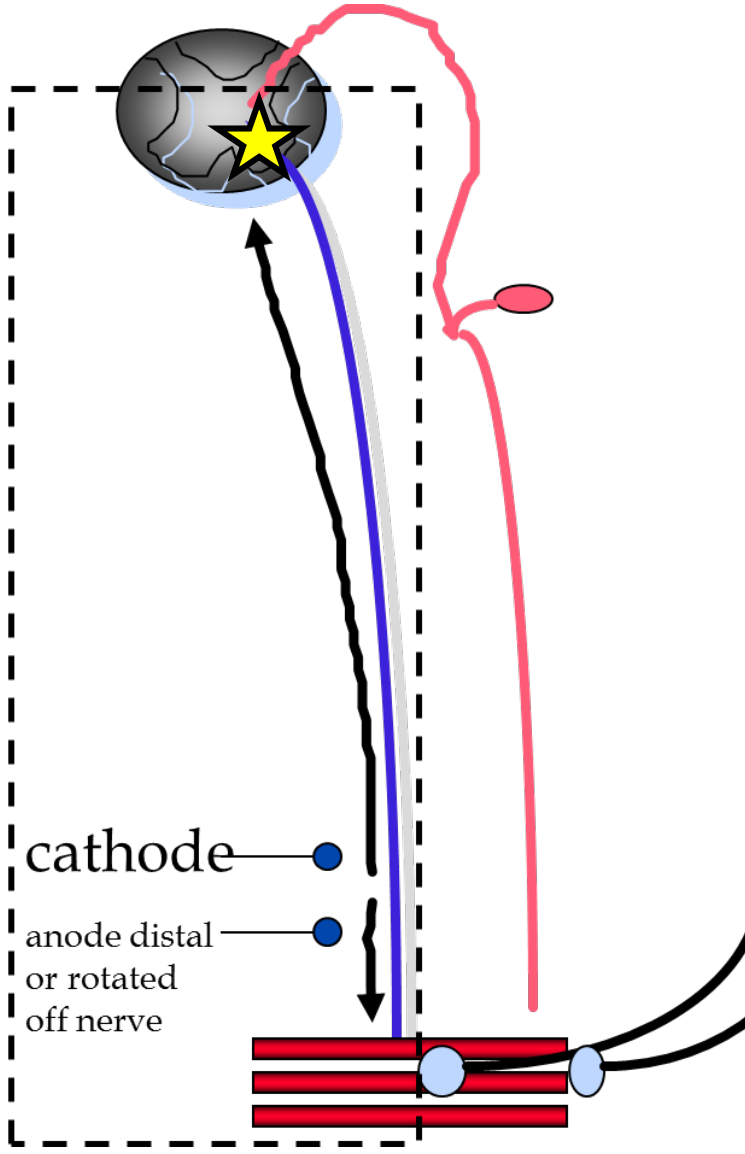
NCS in ALS

- **Sensory NCS** usually normal

- **Motor NCS** normal early on
  - Slow, progressive motor axon loss + compensatory reinnervation
  - CMAP amplitude drops once # of functioning motor units are ~50% normal (later finding)
  - Distal latencies and conduction velocities normal until amplitudes are very low

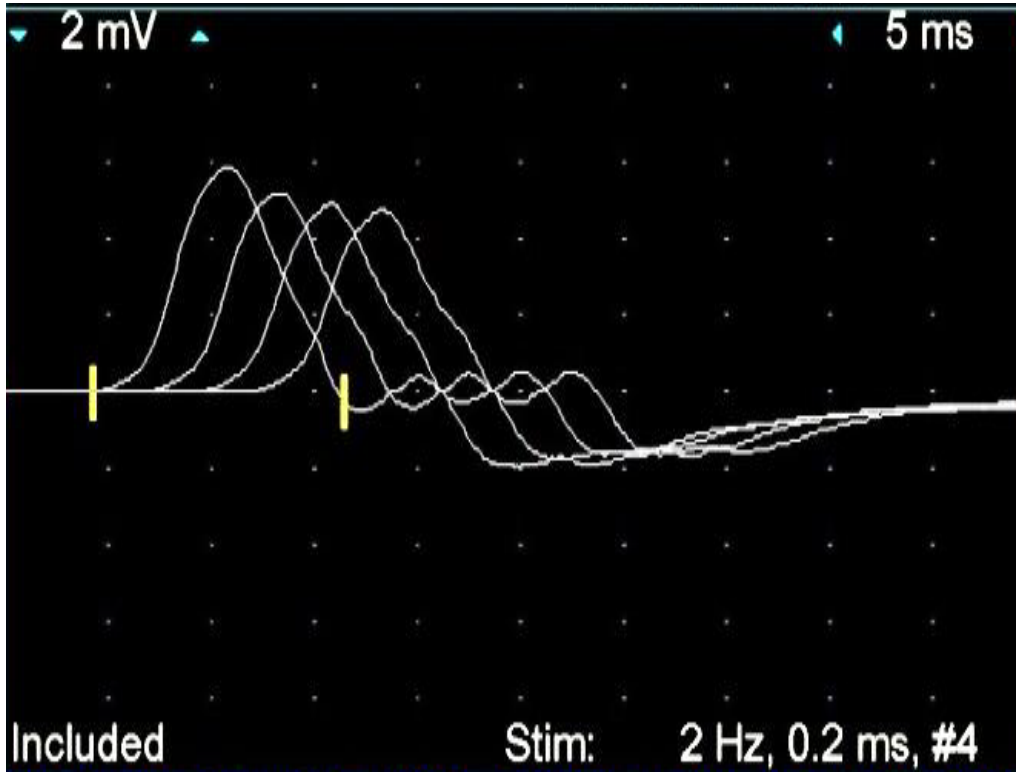
Motor NCS in ALS (con't)

- **F-waves** usually normal
  - o May be absent if CMAP amplitude very low
  - o F-wave persistence or decreased variability
  - o F-wave latency should not be prolonged relative to F-estimate (think of proximal slowing)
- May need to exclude proximal conduction block
  - o Beware of pseudo-conduction block



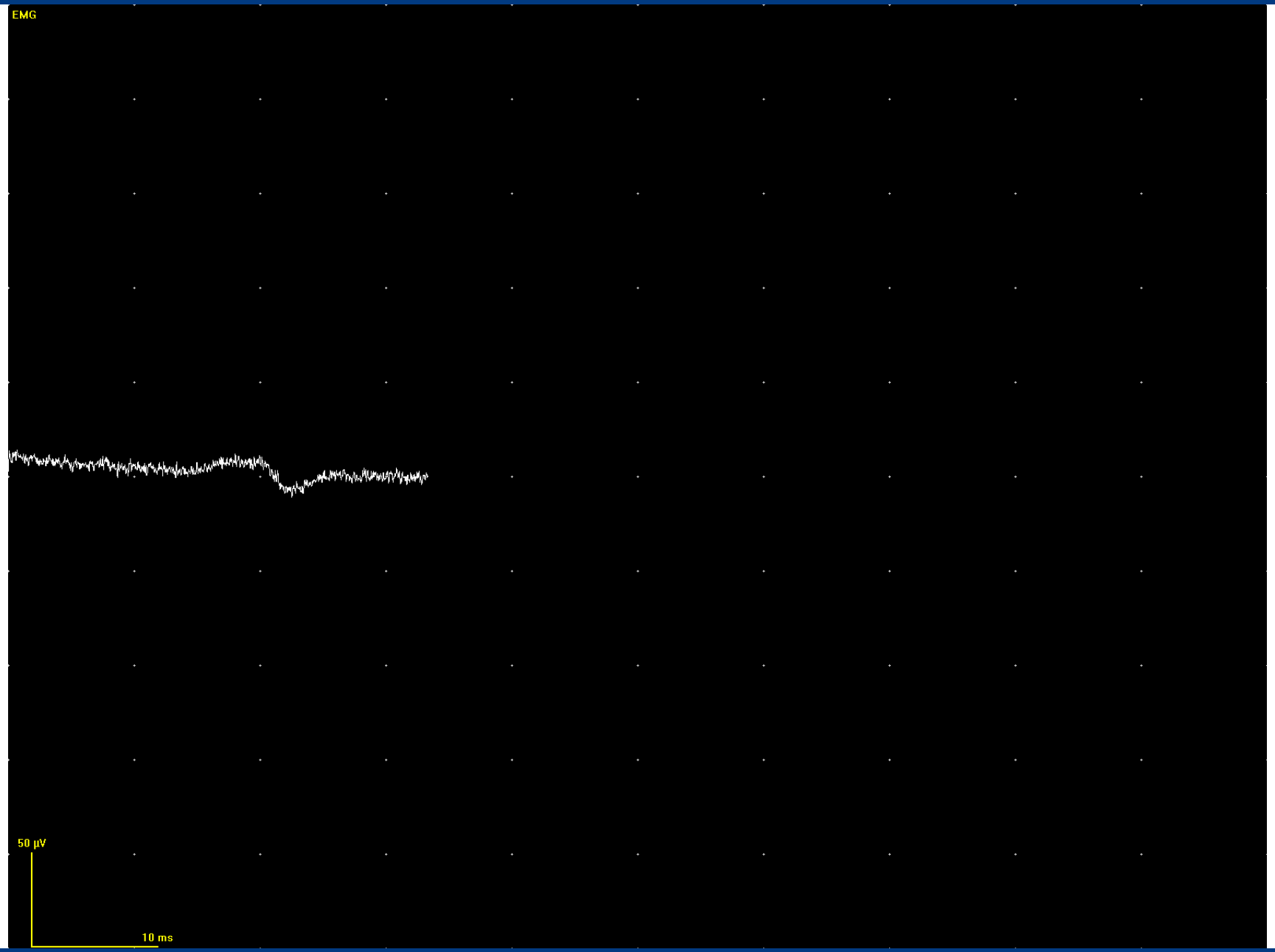
Repetitive stimulation (NCS)

- Decrement has been reported in up to 50% of patients with ALS
- Typically **< 10%** with baseline 2-Hz repetitive stimulation
- Can partially repair with brief exercise and worsen several minutes after exercise

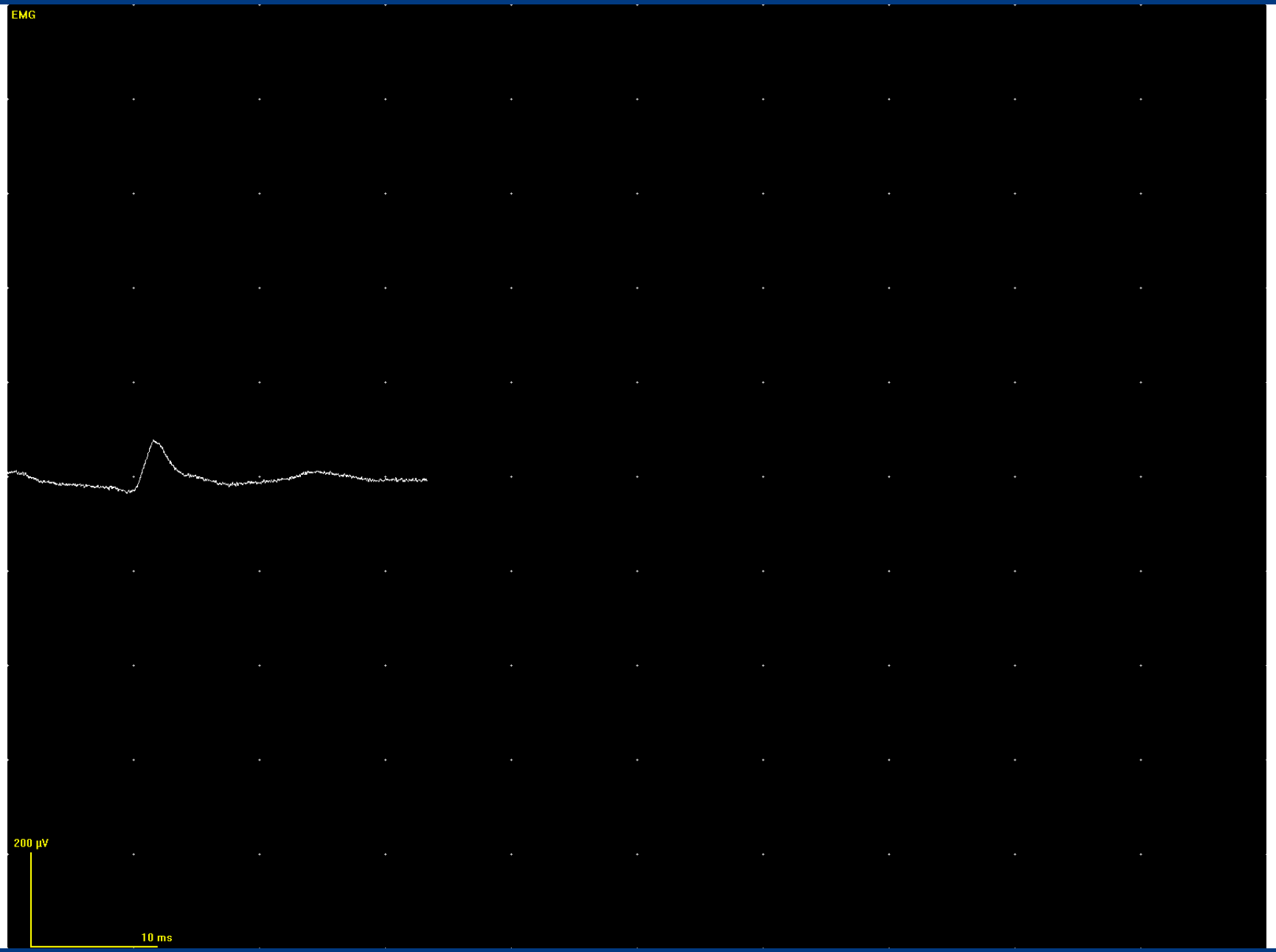


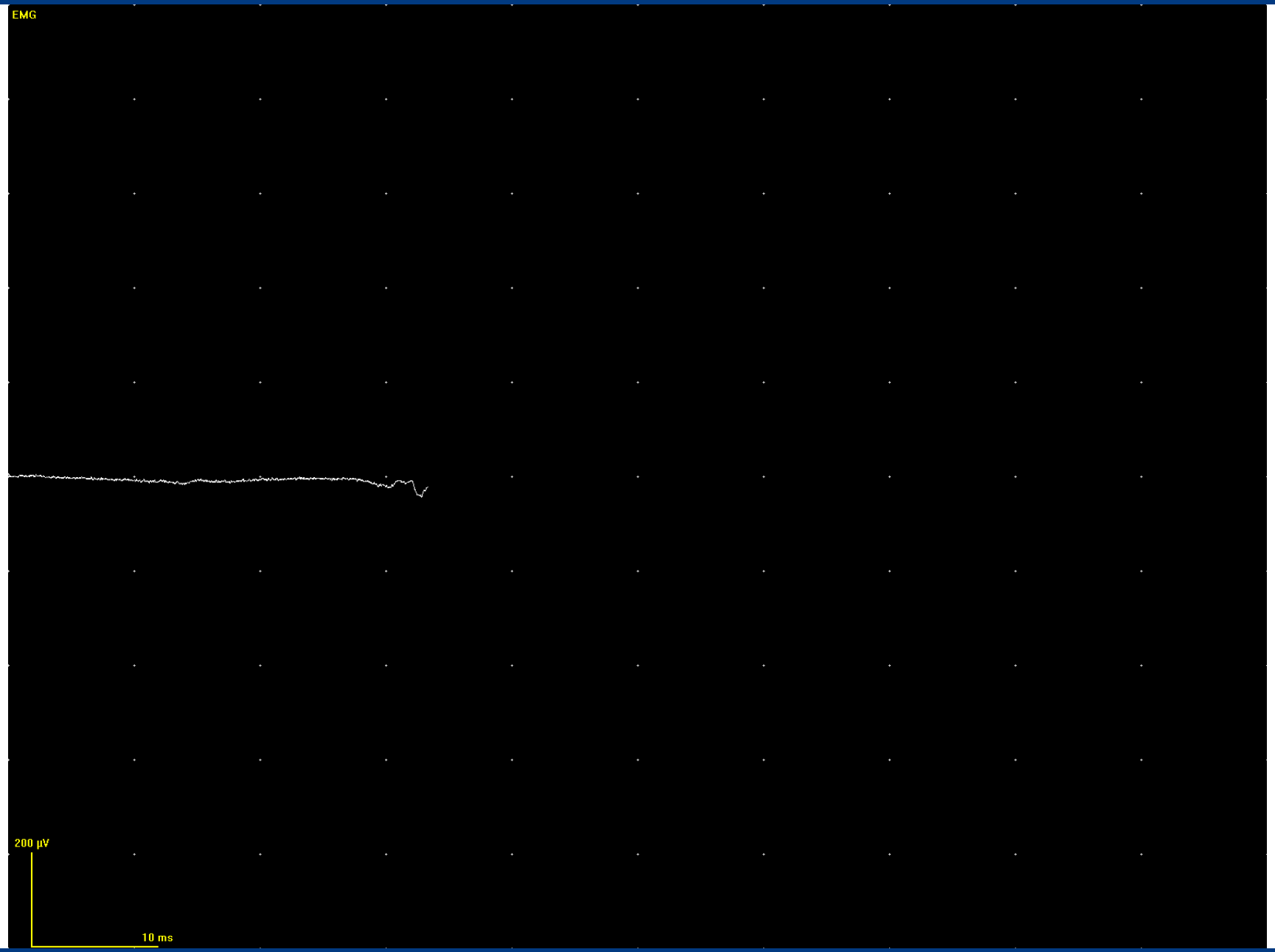
Sounds of EMG in ALS

Spontaneous activity



Voluntary activation

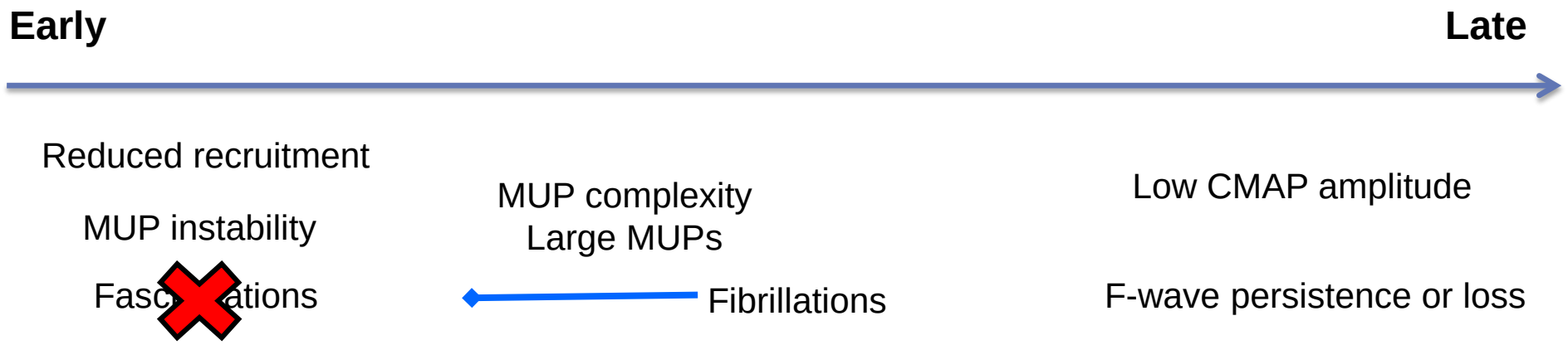

EMG
200 µV
10 ms


EMG
200 µV
10 ms


EMG
300 µV
10 ms

Temporal EMG/NCS Changes

Within single muscle:



Approach to EMG/NCS

NCS

If weakness isolated to lower limb:

- Peroneal (EDB) + F-wave
- Tibial (AH) + F-wave
- Sural sensory

If weakness isolated to upper limb:

- Ulnar (ADM) + F-wave
  - Consider 4-point or proximal stimulation if LMN findings only on exam
- Median (APB) + F-wave
- Ulnar and median antidromic sensory

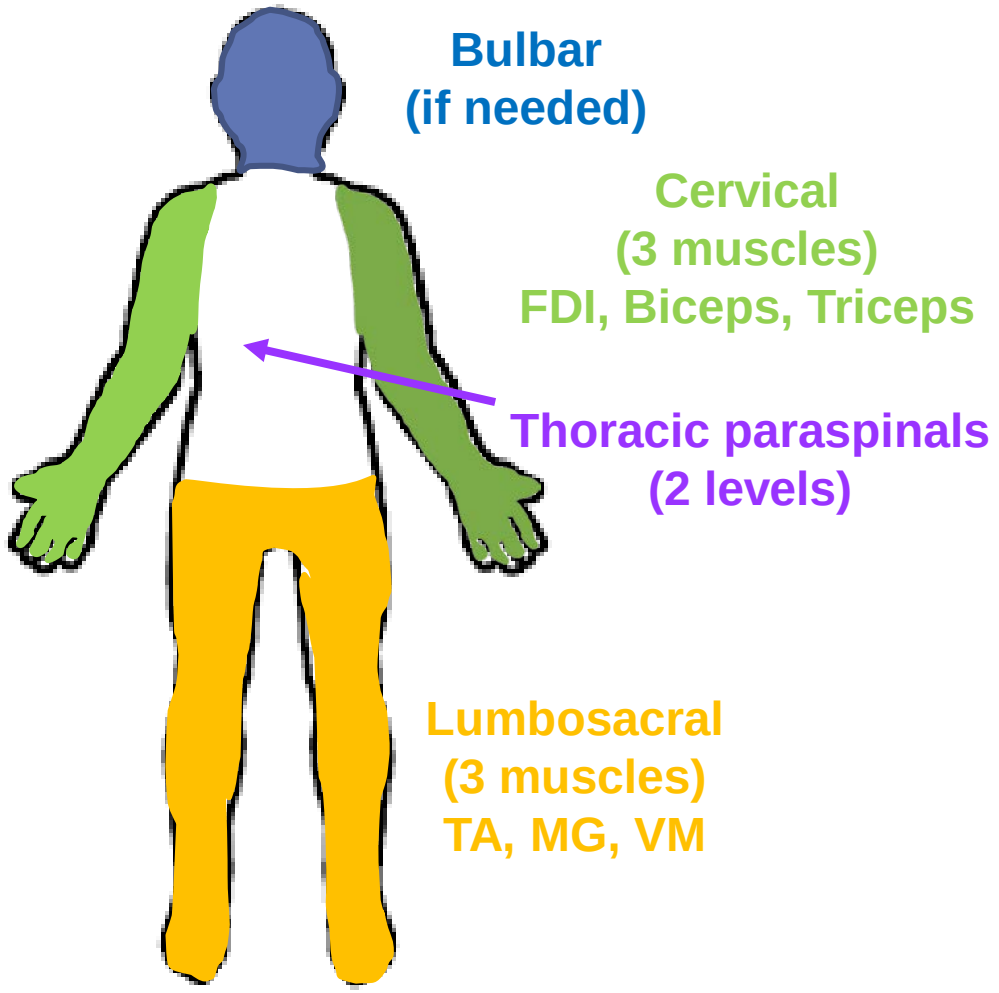
If weakness generalized:

- Peroneal (EDB) + F-wave
- Sural sensory

- Ulnar (ADM) + F-wave
- Median antidromic sensory

Needle EMG

- In obvious cases, may only need to do enough to show the process is diffuse
- Sample several muscles in each segment
  - Different root and nerve contribution
  - Relaxed muscle to assess for fasciculations
  - Distal/weak muscles higher yield
- If bulbar-onset may need to do cranial nerve-innervated muscles
  - Masseter, genioglossus, orbicularis oris



When to think about mimickers…

NCS

- True proximal conduction block on motor NCS
- Absent or prolonged F-waves with near normal motor amplitudes
- Prominent distal latency and conduction velocity abnormalities on motor NCS
- Sensory NCS abnormalities without pre-existing peripheral neuropathy or mononeuropathies

Needle EMG

- Absence of fasciculation potentials
- Large MUP of simple morphology
- Normal MUP (UMN syndrome, BFS/CFS)

Motor-predominant/demyelinating neuropathies
MMN (conduction block)

Kennedy's disease (SBMA)

Take-home points

- Making the diagnosis of ALS can be challenging
  - Requires combination of clinical and EDX findings
  - Revised El Escorial criteria used clinically
- EMG/NCS can:
  - Confirm extent of lower motor neuron involvement
  - Exclude ALS mimickers
- Standard NCS are typically normal with low CMAP amplitudes later in the disease
- Needle EMG shows
  - **Fasciculations**, fibrillations
  - Long duration, high amplitude, **unstable**, **complex MUP** with reduced recruitment

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