NEEDLE EMG IN THE DIAGNOSIS OF AMYOTROPHIC LATERAL SCLEROSIS

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Amyotrophic lateral sclerosis (ALS) is a rare disease but not an uncommon diagnosis to consider in the EMG laboratory. It is characterized by loss of both upper and lower motor neurons. The former is difficult to study objectively, but the latter can be studied by needle EMG. The distinguishing feature of lower motor neuron loss in ALS is that it starts with focal weakness (which brings the patient to medical attention) but progresses within the initial region and to other regions. Before a muscle becomes clinically weak, about 50% of axons must have degenerated, the percentage beyond where collateral reinnervation of surviving motor neurons cannot compensate. Thus, the role of needle EMG is to document a wide distribution of subclinical denervation, and thus exclude radiculopathies and neuropathies which would not be wide spread. Of note, nerve conduction studies have a limited but important role in documenting "motor neuron disease" by a normal sensory response in a nerve innervating a weak muscle.

The World Federation of Neurology put forth a set of diagnostic criteria to ensure enrollment of a uniform group of subjects in ALS clinical trials. The criteria were developed at a meeting in El Escorial, Spain, and there have been subsequent meetings with revisions at Airley House, VA and Awaji, Japan, and the criteria and revisions are referred to by the meeting site names. The diagnostic determinations are based on the distribution of upper and lower motor neuron findings, and are "definite" and "probable" ALS, and the differences reflect the distribution of involvement and not doubt about the diagnosis. There is "possible" ALS, which is consistent with lower motor neuron findings only (progressive muscular atrophy).

Since needle EMG can document subclinical denervation, EMG abnormalities can be used in place of clinical findings of muscle atrophy and weakness when making the diagnosis. The EMG findings of importance are to support "active" and "chronic" denervation. Active denervation in a muscle is reflected by the presence positive waves and fibrillations, and with the Awaji criteria fasciculations alone are equivalent to positive waves and fibrillations. Chronic denervation is reflected in reduced motor unit recruitment, increased amplitude and complexity.

The diagnosis of ALS is clinical, with no biomarker. The diagnostic strategy is based on documenting clinical upper motor neuron loss by pathologically brisk tendon reflexes, and lower motor neuron loss by muscle atrophy and weakness and supported by visible fasciculations. Since extensive subclinical denervation is a key distinguishing feature of ALS, it is appropriate to assess with EMG clinically involved muscle, and then look within the same region or limb to document that denervation is not within a nerve or root distribution. Other limbs should be studied to show widespread pathology. When involved, denervation in the tongue is highly supportive of ALS, but can be difficult to study. There are specific muscles that tend to be involved early in ALS ("index muscles") that are informative when looking for subclinical involvement. In the hand, the first dorsal interosseous, and in the leg, the anterior tibialis, are such muscles. Thoracic paraspinal muscles tend not to be involved with bony spine disease, and frequently show denervation early in the course of ALS. ALS is progressive, and if sufficient distribution of denervation cannot be demonstrated but the disease remains a consideration, a repeat study in several months may be diagnostic.

The El Escorial criteria and their revisions do not include features of frontotemporal lobe dementia or pseudobulbar affect, both of which occur to varying degrees in ~50% of ALS patients. Thus, their presence when there is clinical suspicion of ALS is strongly supportive of the diagnosis.
One form of motor neuron disease that could be confused with ALS is Kennedy disease (bulbospinal muscular atrophy). This is an x-linked disease which affects lower motor neurons and also sensory nerves, but not upper motor neurons. Early on there is prominent bulbar involvement. Weakness and EMG findings of denervation tend to be more symmetric than with ALS. The sensory nerve involvement results in reduced or absent tendon reflexes are markedly reduced or absent sensory nerve responses.