Hirayama Disease

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Last month’s case presented a 21-year-old shrimp peeler who developed weakness of his fingers five years earlier that progressed to a point where he was unable to perform his job. CPK and 530 U/L and EMG revealed chronic diffuse denervation of the arms and muscles with normal sensory and motor responses. Watch the exam at PracticalNeurology.com.

Chronic unilateral or bilateral pure motor weakness of the hand and muscles in a young patient is not common.

DIFFERENTIAL DIAGNOSIS

• Cervical cord pathology such as Syringomyelia: dissociated sensory loss is typically present.
• Brachial plexus pathology: sensory findings are usually present.
• Motor neurons disease: ALS, spinal muscular atrophy (SMA).
  – Cervical Spines are usually investigated before neuromuscular referrals are made.
  – The lack of pain, radicular or sensory symptoms and normal sensory SNAPs and cervical MRIs ruled out most of the mentioned possibilities except:
    • distal myopathy (usually not unilateral) and spinal muscular atrophy.
• EMG/NCS demonstration of chronic distal denervation with normal sensory responses and no demyelinating features limited the diagnosis to MND.
• Segmental denervation pattern further narrows the diagnosis to Hirayama disease (HD).

HIRAYAMA DISEASE

• HD is a sporadic and focal form of SMA that affects predominantly males between the ages of 15 and 25 years.
• Weakness and atrophy usually starts unilaterally in C8-T1 muscles of the hands and forearm (typically in the dominant hand).
  – In roughly one-third of cases, the other hand is affected and weakness may spread to the proximal muscles.
  – DTRs are normal or brisk, unlike most SMA cases where the reflexes are decreased or absent.
  – After progressive course of six years or less, the progression plateaus.
  – Extreme exacerbation of weakness in the cold and focal hyperhidrosis are reported.
  – The disease is more common in India.
• Hypothesis: Radiological forward displacement of the cervical dural sac and compressive flattening of the cervical cord during flexion suggest that this is a form of cervical myelopathy. The resulting ischemia leads to preferential damage of the motor neurons. Decompressive surgery is unlikely to be effective due to the chronic nature of the neurological insult.