CASE 1995-1

Submitted by:

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Clinical History:

The patient is a 60-year-old female who presented with a 20-year history of gradually progressive proximal lower extremity weakness initially noted with rising from a squat position. During the previous ten years she had intermittent episodes of knee buckling with occasional falls. Over the past 2-3 years she noticed distal leg weakness with some foot dragging and some difficulty lifting her arms above the shoulders. Approximately one year ago, she fractured her left hip and now requires a walker for ambulation. She denied sensory symptoms, visual problems, speech problems, difficulty swallowing, and bowel and bladder dysfunction. The patient had no history of skin rashes, joint symptoms, weight loss, fevers, or cardiorespiratory and gastrointestinal symptoms. She also noted that episodes of severe weakness seemed to be precipitated in some instances by eating certain salads, spaghetti or pizza dinners. Her medications at the time of presentation included Diamox, micro-K, and calcium and vitamin supplements. The family history is remarkable for three sons, age 36-43 years, who have experienced episodic weakness and her father who also experienced similar episodic weakness and died at age 39 years of an aspiration pneumonia from suspected respiratory weakness. A paternal aunt and two paternal cousins, likewise, have problems with episodic weakness.

General physical examination was unremarkable except for temporal muscle atrophy and diminished muscle bulk especially in the hand intrinsics. There is no evidence of fasciculations, ptosis, facial weakness, and bulbar abnormalities. Reflexes were normal throughout as was the sensory examination. Laboratory findings were as follows Na = 146 mEq/l, K = 3.6 mEq/l, Cl = 109 mEq/l, glucose = 84 mg/dl, Ca = normal, Mg = normal, PO_4 = normal, B12 = normal, TSH = normal, ESR = normal, CRP = normal, CK = normal, aldolase = normal, ANA profile - negative. EMG studies showed a generalized myopathy predominantly involving the lower extremities with associated fibrillation potentials and occasional myotonic discharges.

Materials Submitted:

Kodachrome photomicrographs including an H&E stained section of skeletal muscle, acid phosphatase stained section of skeletal muscle and an electron micrograph.

Points of Discussion:

1) Diagnosis
2) Pathogenesis