

CASE 1992-4

Submitted by: Dr. Jans Muller, (Indiana University School of Medicine, Division of Neuropathology, Indianapolis, IN) and Dr. Hans H. Goebel (Johannes Gutenberg Universitaet, Abt. Neuropathologie, Mainz, Germany)

These are cryostat frozen sections stained with the modified one-step Trichrome stain of Comori for frozen sections of two brothers, ages 39 and 50, and a cousin once removed, age 29. A pedigree through four generations reveals that all the involved family members eventually hail from one great-grandmother; inheritance is simple autosomal dominant. All biopsies are from the vastus lateralis.

Sections from the first case are those of a 39 year old male who at age 22 years noted intermittent numbness and pain in his calves radiating proximally and aggravated by physical activity. He did not do much about it. In the 4 to 5 years prior to his biopsy, some gross tremor of both arms developed. There was also an increase in fatigue. He also developed some cramping of the leg muscles on exertion, this resulted in limitation of function. Muscular strength seemed to improve on resting. There was somewhat of a nasal speech. There was also slight difficulty in swallowing. Examination revealed proximal muscular atrophy which had developed over the ten years prior to the biopsy. There was a mildly broad based gait, strength was only slightly impaired in the quadriceps, hamstrings, and the anterior tibialis as well as the shoulder girdle muscles. Ankle reflexes were absent on both sides. The remainder of the neurologic examination was normal. Electromyography revealed a neurogenic pattern. Creatine Phospho-Kinase (CPK) at the time was 32 units (normal 0-20 units). Two previous muscle biopsies were obtained elsewhere, from the gastrocnemius muscle in 1969 and from the deltoid muscle in 1972.

The other members of the family generally show slight progressive weakness with sometimes gait disturbances and often some pain on exercise. Weakness never becomes severe and not a single member of the family has died from the disease. CPK's are only slightly elevated, up to 2 or 3 times baseline maximally; electromyography is sometimes interpreted as neurogenic, sometimes as myopathic. Nerve conduction times, when performed, were normal. Some see a physician as early as the third decade; most come in older, sometimes as late as the sixth decade.