CASE 2001 - 03

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CLINICAL HISTORY:

Within three years of each other, two sisters (the first one aged 28 / the second one aged 25) developed in their 20's a neuromuscular disorder characterized by weakness in the proximal limb muscles, steadily and relently progressing over a period of about a year, culminating in respiratory failure. The sisters were born and developed· normally, and were healthy and active in sports up to the time when muscle weakness started. Family history indicates that the patients father died at the age of 40 years because of muscle disease and a paternal grandfather, paternal uncle and a paternal female cousin also suffer from myopathy. Another sister, aged 20 years, is normal. The first sister was investigated; transcripts show that she had calf hypertrophy and that despite full ventilatory support she was conscious and oriented; a muscle biopsy displayed similar changes to the ones present in the material submitted herein.

Physical examination of the second patient records that she is alert and follows commands though is fully intubated. She replies to enquiries by writing and when she is disconnected from ventilator, her breathing becomes shallow. She has bilateral calf hypertrophy. There is decreased strength: ankle dorsiflexion 2/5, plantar flexion acceptable, knee extension 0/5, knee flexion 4/5, hip flexion 0, hip extension 3/5, upper extremity extension 4/5, except for elbow flexor which are 5/5 and the neck extensors are also 5/5. Cranial nerve innervated muscles are spared. Reflexes are symmetrical in the upper extremities; the knees are 0 and the toes are down going. CPK 393 to 512. Muscle biopsy was performed (31/08/2000).

MATERIAL SUBMITTED:

1) Either semithin section - toluidine blue or cryosection -H&E
2) Lantern slide of electron photomicrograph

POINT FOR DISCUSSION: Diagnosis