CASE 2001 - 03

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CASE REFERENCE NUMBERS: B - 4098 / 00-SC-12711

CLINICAL HISTORY:

Within three years of each other, two East Indian sisters (ZM aged 28 living in the United Arab Emirates / SS aged 25 living in Toronto) developed in their 20's a neuromuscular disorder characterized by weakness in the proximal limb muscles, steadily and relentlessly 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 sister ZM was investigated in UAE; 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 patient SS in Toronto 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