CASE 2001-02

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CASE HISTORY

This is a case of 27 year old male with life long difficulty coping with physical exercise, tiredness, and tendency to fall. His condition deteriorated during the last 5-6 years, and now he experiences shortness of breath and difficulty in climbing the stairs.

Neurological examination revealed thin, cachectic male weighing only 29 kg. Upper limbs showed moderate, global distal as well as proximal wasting, with very little corresponding weakness (Grade 4/5) in deltoids and triceps, and mild scapular winging. He was able to stand up from a sitting position without assistance. He was able to walk on his toes but not on his heels. Tendon reflexes were weak and he displayed mild hypotonia. His external ocular movement, as well as facial and neck muscles were normal. He had high palate, and no other abnormalities.

Laboratory investigation revealed mild elevation of CPK (2-5 times normal). He is one of thirteen children, seven of whom suffer from a similar disease. His mother and maternal grandmother also showed similar distribution of muscle atrophy and weakness. His father is healthy.

MUSCLE BX: Trichrome stain

DIAGNOSIS: