A RARE CASE OF MANIFESTING FEMALE CARRIER IN PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY

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Introduction: Duchenne muscular dystrophy affects females in very rare instances. Approximately 8% of female DMD carriers have muscle weakness and are designated as manifesting DMD carriers. Dystrophin is a rod-shaped cytoplasmic protein, which is localized on the cytoplasmic surface of the plasma membrane of skeletal and cardiac muscles, which is absent in the case of DMD.

Discussion: A 34 year old female presented with a complain of proximal thigh muscle weakness for the last 20 years. She was born a floppy baby, with poor ventral suspension and generalized asymmetrical hypotonia. She started walking independently at the age of 4 years up till 10 years, here after requiring crutches assistance. She defaulted clinic appointment, and came back to us when she was 17 years old with a complain of left hip pain and bilateral lower limb asymmetrical proximal myopathy. She was ambulating using walking frame from 16yrs old till date. X-ray left hip showed avascular necrosis for which she underwent left total hip replacement at the age of 21 years old. Quadriceps muscle biopsy revealed skeletal muscle fibers of various sizes composed of atrophic and hypertrophic muscle with preservation of mosaic pattern of dystrophin positive and negative fibers. Impression was muscular dystrophy. Her serum CK level was within normal range. ECG and ECHO findings were unremarkable.

Conclusion: In Duchenne dystrophinopathic carriers, muscular weakness is predominantly asymmetrical(81.8%). Dystrophin immunohistochemistry is a useful tool to diagnose what type of myopathy is present and whether mosaicism is present in female myopathic patients. The surface membranes of muscles in DMD patients do not react with anti dystrophin anti-serum, while those from carriers show a mosaic pattern. Raised serum CK levels are seen in 45% to 70% of cases, which indicate progressive worsening of muscle involvement, which was not the case in our patient.