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Case Report

A case report on Duchene muscular dystrophy in an Indian family

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ABSTRACT

Duchenne muscular dystrophy (DMD) is commonest X-linked syndrome muscular dystrophy in the children, present in early childhood and characterized by the proximal muscle weakness and the calf hypertrophy in exaggerated boys. There is generally delay in the motor development and ultimately wheelchair confinement leading to premature death from cardiac or the respiratory complications. For treatment we use various modalities like corticosteroid therapy. We also used intermittent positive pressure ventilation that could help in developments in function, quality of life, ambulation and life expectancy though novel therapies still focus to provide the cure for this disease. Clinically, it is categorized by the progressive muscle wasting ultimately leading to the premature death. Here we presented a case of Duchene muscular dystrophy in a 13 year old boy with clinical presentation.

Keywords: Calf hypertrophy, Corticosteroid, Muscle weakness, Gower sign

INTRODUCTION

Duchenne muscular dystrophy (DMD), an atypical inherited musculo-skeletal disorder which shows the clinical characteristics of the progressive muscular weakness at early stage and the pathologic features of fibrosis, and fatty replacement, predominantly late in disease course. It is recessive X-linked disorder befalling 1 in 3500 male births, and named after the French neurologist Guillaume Benjamin Amand Duchenne in 1860.[1]

DMD has very high mutation-rate with distinctive, and persistent clinical presentation. Patients generally become wheelchair bound by age of 12 and expire in their late teens to early twenties.[2,3]

It is commonest and severe form of the muscular dystrophy, starts at 3 to 5 years of age and characterized by the proximal muscle weakness and the calf hypertrophy in the affected boys. DMD has very high mutation-rate with distinctive and persistent clinical presentation. Patients generally become wheelchair bound by age of 12 and expires in their late teens to the early twenties. According to PubMed literature, around 150 cases been reported till death.[4]

High-quality multidisciplinary care can slow disease progression, prolong functional independence and prolong life expectancy.[5] Early detection of the disease, better clinical practice guidelines and increased ventilator use along with early intervention has improved the life expectation of these patients. A study in France showed that the mean life expectancy increased from 25.77 years for those born between 1955 and 1969 to 40.95 years for those born between 1970 and 1994.[6]

CASE REPORT

A 13 year old male patient presented to our department with chief complain of walking difficulty and difficulty in standing from sitting position. His parents give history of falling repeatedly, fatigue muscle weakness and incapability to climb upstairs. His IQ level is claimed to be normal. There is no history of cranial nerve involvement and muscular pain. In family history

patients' younger brother age 5 years with symptoms of inability to walk also have similar symptoms but less prominent. There is also history of similar illness in one child from maternal side and death due to it.

On general physical investigation, the child is thin built and having difficulty in walking, standing and getting up from the sitting position, and climbing upstairs. Patient also have proximal muscle weakness, calf hypertrophy and positive Gower's sign. There is no muscle tone, twitching of muscles, and cranial nerve investigation is found to be with in normal limit.

The patient's creatine kinase was 329.9 IU/l (normal 50-150 IU/l) and the muscle biopsy from the left quadriceps showed the rounded small muscle fibres including evidence of degeneration, and absence of the dystrophin protein. He was identified as case of DMD. He was currently bed bound due to weakness and contractures of all the limbs deformities.

He was treated with physiotherapy regularly, limb strengthening exercises, 0.9 mg/kg/day deflazacort and management of the respiratory infections, time to time cardiac and the respiratory follow up, genetic counselling, and other therapies.

Figure 1: Patient age 13 years.

Figure 2: Younger brother (5 years) of patient.

Table 1: Biochemical reports.

Blood parameters	Values
Sodium (mmol/l)	139
Potassium (meq/l)	3.9
Calcium (mg/dl)	10
CPK-MB (U/l)	329.9

DISCUSSION

DMD is commonest muscle dystrophy in our India in addition to the world, occurs by mutations in the dystrophin gene as the result of which body is incapable to synthesize protein dystrophin necessary for muscle contraction. Every time muscle contracts, the damage occurs which is fixed but with a deficient protein leading in repaired muscle that is also the damaged one. The continuous succession of the damage, and repair, and ultimately replacement of the muscle with fibro-fatty tissue is accountable for clinical signs of the progressive muscle wasting and degeneration that was generally evident by 3 to 4 years.[4]

DMD is caused by the mutations in gene which encodes 427-kDa cytoskeletal protein dystrophin. An increased knowledge of function of dystrophin and its part in muscle led to the greater understanding of pathogenesis of DMD. This, together with developments in genetic toolkit of molecular biologist were leading to several different treatment approaches. Gene therapy could be achieved by viruses or plasmids, mutations could be corrected by chimaera plasts and the short DNA fragments, mutations exon skipping can be induced by oligonucleotides and read through of nonsense mutations can be achieved by aminoglycoside antibiotics. Blocking proteasome degradation path can steady any reduced dystrophin protein and up regulation of various other proteins can also avert dystrophic process. Muscles can be repopulated by myoblasts, or stem cells. All, or a combination, of these approaches hold great promise for the treatment of this devastating disease.[2]

DMD gene contains seventy-nine exons but accounts only for 0.6% of gene; rest made of the large introns. Large size of DMD gene make it vulnerable to mutations, causes loss of function of dystrophin, leading in prematurely reduced, and unbalanced dystrophin protein. Majority of the mutations are intra-genic deletions, which account for 65-72.0% of all patients of DMD. The exact mechanism of dystrophin deficiency causing degeneration of muscle fibres is unclear. Absence of dystrophin at plasma membrane cause delocalization of dystrophin related proteins from membrane, disruption of cytoskeleton with subsequent membrane instability and increased vulnerability to mechanical stress. Furthermore, altered membrane permeability, and an abnormal calcium homeostasis thought to play the role, with an increased concentration of cytosolic calcium leading to an activation of proteases like calpains.[7] Singh et al reported a deletion rate of 73.0% in North Indian patients

including both DMD and the Beckers muscular dystrophy.[8] Among a southern Indian DMD population the reported deletion rate was 62.1% and 78% of which was located in the distal hotspot region.[9]

Affected boys present with running difficulty or getting-up from ground, toe-walking or frequent falls. Patients have waddling gait, lumbar lordosis and calf enlargement that disappear on sitting. There was a weakness of proximal muscles of lower limb which the patient uses his hands, and arms to walk up themselves from squatting position because of lack of strength in hip and thigh muscle suggesting of Gower's sign.[10] In this case, affected child presented clinically with signs of the delayed motor development, walking difficulty and stairs climbing, muscle weakness and positive Gower's sign. Oral manifestations include wide dental arches, open bite, delayed eruption, large tongue and retro gnathic facial morphology. The development of the malocclusion in such patients was linked to involvement of orofacial muscles by disease that was apparent in present case.[5,10]

Current DMD management involved physiotherapy and corticosteroid therapy that delayed but didn't cure the disease. Prenatal counselling and various other genetic modalities were being tested to offer hope in this fatal and progressive muscle dystrophy to prolong and improve quality of life in such patients. DMD can be a devastating condition to the family, however, improvements in research and high-quality multidisciplinary care have prolonged the life expectancy and can maintain participation in activities that the patient feels are important for a longer period of time.

CONCLUSION

DMD is a condition affect many boys and families, remaining dystrophy like limb girdle, congenital, myotonic diseases not able to diagnose due to lack of hospitals and diagnostic facilities. Recent advance in symptomatic management with the careful use of corticosteroids, respiratory support and physiotherapy improve muscle strength. Stem cell therapy remaining search for a cure remains elusive, although may promising and novel therapies are in progress, some of which have entered the stage of human trials. The main cause of concern is the malnutrition, environmental pollution, gene mutation, ignorance and the poverty. Population below poverty line is the primary reasons for higher incidence of MD prevailing in some parts of world.

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