

## Case Report



## Duchenne Muscular Dystrophy in Male Child Diagnosed by Dystrophin Gene Deletion Test

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### ABSTRACT

Duchenne muscular dystrophy is an x-linked recessive disease, affecting 1 in 3600 to 6000 live male births, DMD is inherited musculoskeletal disease. DMD is named by a French neurologist Guillaume Benjamin Amand Duchenne in 1860. Dystrophies are caused by mutation in the dystrophin gene DMD (Xp21.2). Multiplex Ligation dependent Probe Amplification has been used as the initial diagnostic test of choice. MLPA can diagnose 70% of DMD patients, having deletions/duplications. Creatine kinase levels can also be used as a diagnostic marker for DMD. Genetic testing is mandatory irrespective of biopsy, results in the muscle biopsy is not required if the diagnosis is secured first by genetic testing. Current management of DMD involves physiotherapy and corticosteroid therapy, which delays loss of ambulation for 1-3 yrs.

**Keywords:** Duchenne muscular dystrophy, multiplex ligation dependent probe amplification.

### QUICK RESPONSE CODE →

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### INTRODUCTION

Duchenne Muscular Dystrophy is an debilitating early onset, severe, rapidly progressive musculoskeletal disorder. It is associated with a functional deficiency of dystrophin. It is belonging to a pathological group of diseases known as dystrophinopathies<sup>1</sup>.

DMD occur as a result of mutations such as deletions (60-65%), duplications (5-15%) in the dystrophin gene (DMD locus Xp21.2), leads to absence or defect in the dystrophin protein results in progressive muscle degeneration and loss of independent ambulation.

Multiplex Ligation dependent Probe Amplification is a widely used method and is initial diagnosis test of choice for DMD. MLPA diagnose patients with deletions/duplication of patient with point mutations, need direct sequencing of all coding regions. Muscle biopsy and creatine kinase levels are considered as a diagnostic markers<sup>2</sup>.

Infants are rarely symptomatic. Poor head holding in infancy may be the earlier sign of weakness. A Gowers sign and trendelenburg gait occurs at 5-6yrs age. Some are confined to wheelchair by 7yrs of age. Respiratory complications like weak and ineffective cough, frequent respiratory infections, decreasing respiratory reserve and pharyngeal weakness. Pseudo hypertrophy of calves and

wasting of thigh muscles are classic features of DMD. Cardiomyopathy is seen in 50-80% of patients. Mental retardation, epilepsy is slightly most common in DMD patients.

Use of glucocorticoids in DMD slows the decline in muscle strength and function in DMD. And treat cardiac and respiratory complications. Use dietary supplements like coenzyme. Q10, carnitine, amino acids (glutamine, arginine) and antioxidants (fish oil, vitamin E)<sup>3</sup>.

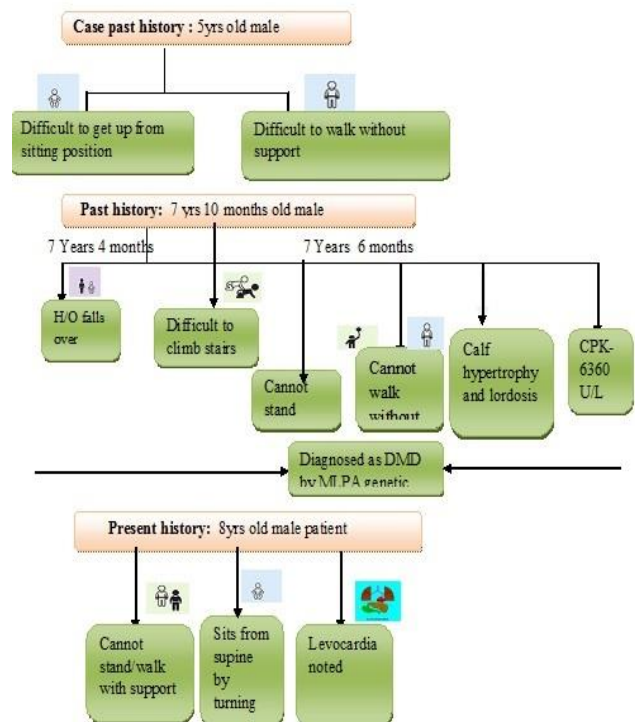


Figure 1: Past and present medical history



**CASE REPORT****Chief complaints**

A 7yrs 10 months old 2<sup>nd</sup> borne male child to NCM, brought with c/o difficulty to get up from sitting position noticed from 5yrs age, progressed to difficulty to walk without support and h/o falls down. Presently child cannot stand/walk and climb stairs without support since last 6 months. No h/o dyspnea while walking, change in speech, seizures.

**Maternal history:** Birth ANC normal, Pregnancy Induced Hypertension at 7months GA, FT-LSCS, birth wt-2.7kgs, CIAB, no NICU stay/NNH/seizures. Discharged on day7.

**Development:** Neck control-4M,roll over -8M, sit with support -10M, stand with support-18M,walk with support-1y 8M, walk without support-2yr,cannot run, climb stairs with support-4yr, cannot climb stairs without support, fine UDG -8M ,scribbling-1Y 2M, drink from cup-1yr,eat on own-3yr,undress/wear slipper- 4-5yr,cannot dress/wear button ,Right handedness+.

**Language:** Monosyllable-8m, bisyllable-10m, 1<sup>st</sup> word with meaning -11m,phrases-2yr,body parts-4yrs,tell name/gender/parents name/ tell 1-20, A-Z, cannot tell rhymes/address/phone number.

**Social:** Social smile -5m, stranger anxiety- 1yr, wave bye bye-18m, bowel and bladder control-2yr, dry by night-1,2/2yr, indicate hunger+, knows colors, R-L discrimination. No money concept or arithmetic skills.

**Family:** NCM, elder sibling female -11yr, normal, younger sibling -7yr male –normal. No f/h/o similar complaints.

**Present Medical History****General Examination**

**Weight:** 22kgs, head circumferences: 48.5cm, others: No, dysmorphism/neurocutaneous marker. Valley sign +, B/L calf hypertrophy+, lordosis+.

**Systemic examination:** DTR: hyporeflexia

Others: plantar-flexor, Gait-cannot walk without support.

Tone: hypotonia.

Power: <3/5(proximal>distal weakness) cannot walk without support, cannot get up from sitting to standing position (previously lower sign+).

2D Echocardiography: It shows Levocardia

**Investigation:** CPK - 6366U/L, TFT-T3 162ng/dl, T4-7.64mcg/dl, TSH -2.69mIU/ml.

**Geniting test:** MLPA – Hemizygous deletion in exon 49-50 of DMD gene.

**Diagnosis:** Duchenne Muscular Dystrophy.

**Treatment:** T.omnacortil 10mg OD in October 2020 and given for a month and stopped, T.shelcal -500mg and physiotherapy.

**DISCUSSION**

DMD is the most common muscle dystrophy in India as well as worldwide caused by mutations in dystrophin gene. It shows clinical characteristics of progressive muscular weakness at an early stage and pathological features of fibrosis and fatty replacement particularly in the late disease. Fibrofatty tissue is responsible for the clinical signs of progressive muscle wasting and degeneration that is usually at 3-4yrs.

DMD gene contains 79 exons but account for only 0.6% of the gene. The large size of the DMD gene makes it susceptible to mutations, leading to loss of function of dystrophin.

Affected boys clinically presented with signs of delayed motor development. Oral manifestations include wide dental arches, large tongue, delayed eruption, open bite, and retrognathic facial morphology<sup>4</sup>. Immunostaining of a muscle biopsy with anti-dystrophic serum proved to be valuable in the diagnosis of DMD in a symptomatic female carrier<sup>5</sup>.

Early and accurate genetic diagnosis is crucial for optimal patient care, including early initiation of disease management<sup>6</sup>. Differential diagnosis of DMD include polymyositis, kennedydisean, facioscapulo humeraldystrophy, emery-dreifus muscular dystrophy, metabolic myopathies, spinal muscular atropy, physical medicine and rehibition for limb-girdle muscular dystrophy. DMD is confirmed based on bone mineral density test, echocardiogram, genetic analysis and clinical examination.

Common complications with DMD include contractures, scoliosis, breathing difficulties due to the weakness of diaphragm and chest muscle, facial and throat muscle. Heart is also affected by DMD. These complications can be treated by giving corticosteroids and physiotherapy<sup>7</sup>.

**CONCLUSION**

Males are most commonly affected with DMD. In females it can occur rarely. Early and accurate genetic diagnosis is crucial for optimal patient care, including early initiation of disease management. Multiplex Ligation dependent Probe Amplification has been used as the initial diagnostic test of choice. Lack of disease awareness and subsequent failure to recognize signs and symptoms of DMD, lead to delayed diagnosis. Open muscle biopsy is necessary if the differential diagnosis includes DMD among other types of muscular dystrophy.

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