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DUCHENNE MUSCULAR DYSTROPHY- A CASE REPORT.

Paediatric Medicine

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ABSTRACT

DMD begins in early childhood and runs a relatively rapid and progressive course. The incidence is about 1 in 3,500 live male birth1. It is due to the mutation on short arm of X- chromosome at XP21 which is one of the largest gene. Early diagnosis and multidisciplinary approach to care are essential to enhance the quality of life.

KEYWORDS

Dystrophin Gene, Gower Sign, Calf Hypertrophy, Muscle Weakness, Progressive Muscle Wasting, Muscular Dystrophy, X-linked Recessive.

1. INTRODUCTION:

Duchenne muscular dystrophy is the most common hereditary neuromuscular disease affecting all races and ethnic groups[2]. The disease is inherited as an X linked recessive trait and named after a French neurologist Guillaume Benjamin Amand Duchenne in 1860[1]. The abnormal gene is at Xp21 locus. It's clinical features are progressive muscle weakness, hypertrophy of calves , intellectual impairment, progressive fibrosis.

2. Case Presentation:

A 8 year old male presented to the OPD with difficulty in standing up from sitting position since 1 year and difficulty in walking for last 6 months, it was gradual in onset and progressive in nature. The mother also noticed gradual thinning of his upper limbs and increase thickening of lower limb. The child had difficulty while putting on trousers but he had no difficulty in putting on slippers. On examination there was bilateral calf muscle hypertrophy. The power of trunk muscle, hip flexors and extensors were 3/5. Provisional diagnosis of muscular dystrophy was made and investigations were done. The serum creatinine kinase level was 520 IU/L . The multiplex PCR of DMD gene showed deletion identified in 49,47 and 45 exons. Patient was diagnosed as Duchenne muscular dystrophy and tablet prednisolone was started along with physiotherapy.

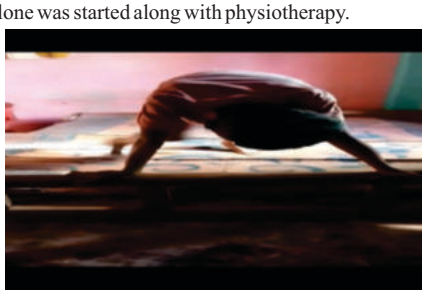

Figure 1: calf muscle hypertrophy.

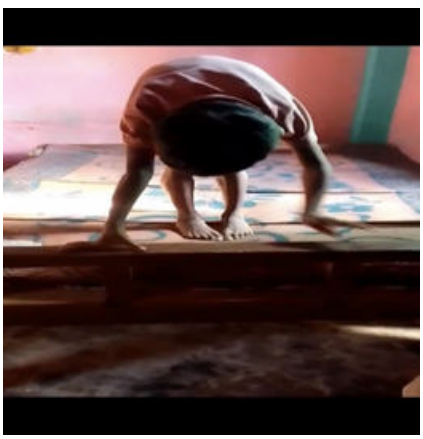

Figure 2: Gower sign.

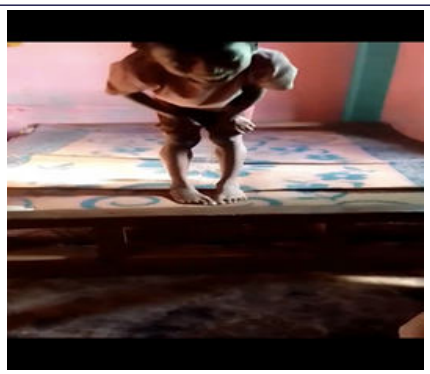

Figure 3: Gower sign

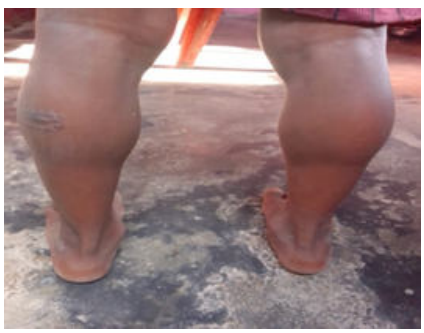

Figure 4: Gower sign.

Investigation:

Complete blood count: Hemoglobin: 10.8 g/dl, Total count: 6,200, platelet: one lakh fifty thousand
KFT: urea: 11.08 mg/dl, creatinine: 0.27mg/dl
Serum electrolytes: sodium: 115.5mmol/l, potassium: 4.76mmol/l, chloride 93.61mmol/l.
CRP: <0.5mg/dl.
Serum creatine kinase: 520 IU/L.
ECHO: normal
Electromyography: MUAPs low amplitude and area, polyphasic motor unit potentials.
Nerve conduction study: normal.
Gene analysis (multiplex PCR of DMD gene): deletion identified in 49,47 and 45 exons.

Treatment:

Patient was started with tablet prednisolone @0.75mg/kg/day[2]. Immunization with influenza virus was given. Physiotherapy was started and nutritional care was advised. Patient was counselled about the condition of the disease and advised for routine follow-up.

3. DISCUSSIONS:

Dystrophin is a cytoskeletal protein and is located subsarcolemally.

The DMD gene encodes the dystrophin protein, which plays a crucial role in maintaining the structural integrity of muscle fibres. Absence of dystrophin leads to delocolalization of dystrophin associated proteins from the membrane, disruption of the cytoskeleton with resultant membrane instability and increased susceptibility to mechanical stress[3]. The absence of nitric oxide synthase also leads to dystrophic features, resulting in progressive muscle weakening.

At birth, children usually do not have symptoms. At 2-3 years of age, children cannot jump with both feet off the ground. By 5-6 years, they have difficulty in rising from chair and climbing stairs. By 7 years the weakness becomes more evident. There is weakness of the proximal muscles of the lower limb as in which patient uses his hands and arms to walk up their own body from a squatting position due to lack of hip and thigh muscle strength suggestive of Gower sign[4]. They develop lordosis due to paraspinal muscle weakness and waddling gait due weakness of gluteal muscle. Between 9 and 12 years, children usually become wheel chair dependent. Eventually respiratory muscles get involved. Death in boys with DMD occurs in late teens to 20s[5]. the causes of death are respiratory failure, pneumonia, intractable heart failure or occasionally aspiration.

Elevated serum creatine kinase levels can be an early indicator. Confirmative diagnosis is by genetic testing. Current management of DMD involves only supportive and preventive care with corticosteroids and physiotherapy which can delay the loss of ambulation 1-3 years but not cure the disease. Cardiac care, nutritive care should also be given. Pulmonary infections should be promptly treated.

4. CONCLUSION:

DMD is a progressive disorder where the progression can be delayed by proper supportive and preventive care. Recent research focuses on developing more effective treatments including gene therapies and exon-skipping drugs. Early diagnosis and comprehensive care remain essential in managing this condition. Prenatal counselling and genetic tests such as multiplex ligation-dependant probe amplification are being used to offer hope in this progressive and eventually fatal muscle dystrophy to prolong and improve the quality of patient's life[6,7].

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