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Case report: Becker's muscular dystrophy

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Abstract
A case report of 27yrs old male patient presented with c/o difficulty in climbing stairs, lifting heavy weight, difficulty in standing from sitting position since last 12ys. Significant calf muscle hypertrophy was seen on examination. A muscular dystrophy was the probable diagnosis and to confirm this various investigations were carried out, including electromyography (EMG) and creatinine kinase (ck) levels. A cardiac workup followed to assess for dilated cardiomyopathy which is associated with Becker's Muscular Dystrophy. It mainly affects male and prevalence in females is extremely low, as BMD is a X linked disorder.

Keywords: becker's muscular dystrophy, electromyography, dystrophin

Introduction

Case Report

A 27yrs old male patient presented to us at Neuromedicine department of BMHRC (Bhopal) with complaints of difficulty in lifting weight, difficulty in climbing upstairs, difficulty in standing from sitting position since last 12 yrs. Patient had to take support while standing from sitting position (Gowers sign present). Patient also complaints of weakness of proximal upper limb and pain in calf muscles. No c/o breathlessness, chest pain, palpitation. Detailed clinical evaluation was done and it was noticed that there was wasting of muscles (deltoid) in upper limb and calf muscle hypertrophy. Higher functions were normal, no cranial nerves were involved. Power in upper limb was 5/5 and in lower limb was 4/5 and reflexes were normal. Sensation in both upper limb and lower limb were normal. No involuntary movement present. No fasciculations present. Waddling gait was present. CPK was done repeatedly which was on the higher side, EMG findings were small polyphasic MUPs with reduced IP observed in all the muscles studied s/o Beckers Muscular Dystrophy. ECG and Echocardiography was done which were normal suggesting no cardiac involvement. Patient's younger brother was also suffering from the same disorder. No positive history from the maternal side. Physiotherapy was advised to patient at the time of discharge.

Fig 1: Calf muscle hypertrophy

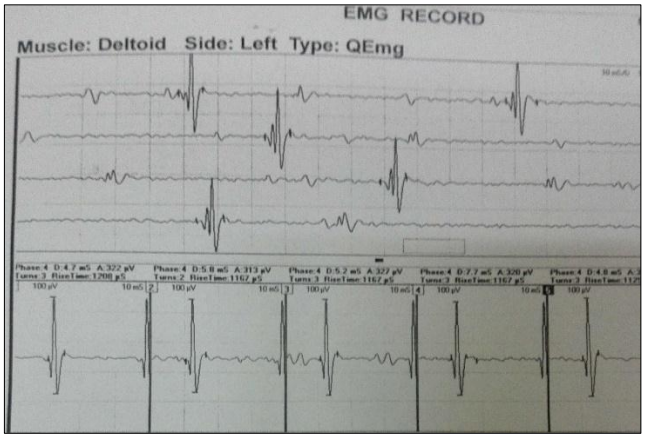

Fig 2

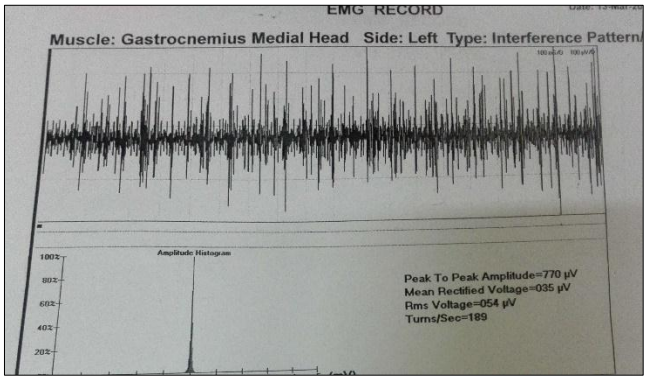

Fig 3

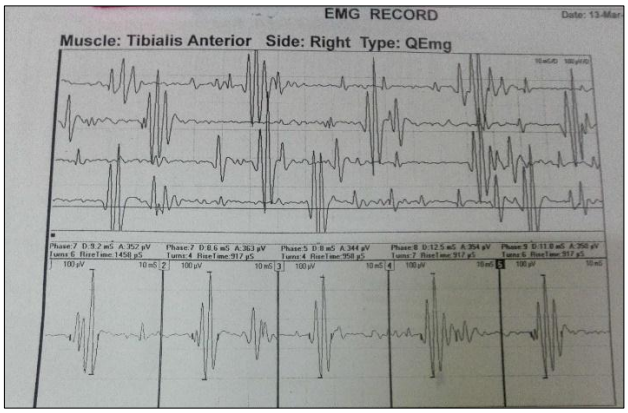

Fig 4

Discussion

The muscular dystrophies are a group of inherited disorders characterized by progressive muscle wasting and weakness. Becker's muscular dystrophy is similar to the more common muscular dystrophy Duchenne muscular dystrophy but the clinical course is milder. There is muscle wasting and weakness which is mainly proximal. The incidence of almost 1 in 30000 live births. BMD is caused by abnormalities of dystrophin gene which is responsible for the muscle protein dystrophin. Dystrophin is a large filamentous protein of 3685 residues and a molecular weight of 427 kDa[6]. In BMD abnormal but only partly functional dystrophin is produced. It is a X linked recessive inherited disorder and most cases involve exon deletions in the dystrophin gene Xp21.This mutation leads to the translation and production of a semi-functional dystrophin gene(unlike DMD where the resulting dystrophin gene is nonfunctional ; 30-80% normal dystrophin in BMD compared to only 5% in DMD). Nicholas *et al.* [2] suggested that different exon deletions resulted in different disease severity and hence different disease progression. Nicolas *et al.* [2] studied four prevalent in frame exon deletions (45-47,45-48,45-49,45-51) and showed differences in the rate of disability progression to the point of being wheelchair bound (reached earlier in deletions 45-47and 45-49 compared to deletion 45-48), as well as differences in the age of onset of dilated cardiomyopathy (onset delayed by 11yrs in deletion 45-48 and by 14yrs in deletions 45-49,in comparison to exon deletions 45-47) (Nicolas *et al.*, 2015) [2].

BMD presents within the ages of 5 and 15 with various signs and symptoms [1].The pattern of muscle loss in BMD usually begins with the hips and pelvic area, the thighs and the shoulders. To compensate for weakening muscles the person may walk with a waddling gait, walk on his toes or stick out of the abdomen. The rate of muscle degeneration varies a great deal from one person to another. Some men require wheelchairs by their 30s or later while some manage for many years with minor aids such as canes. On examinations a rather characteristic sign in BMD is pseudo hypertrophy of the calves, which may be quite obvious on inspection. However, it is known that the pseudo hypertrophy is realistically a combination of actual muscle hypertrophy together with fatty deposition (Mauro *et al.* 2014) [3]. A sign which helps differentiate DMD from BMD is the preservation of strength of the neck flexors. Because muscular dystrophy does not affect nerves directly, touch and other senses remain normal, as does control over the smooth or involuntary muscles of the bladder and bowel and sexual functions. Muscle deterioration in BMD usually isn't painful in itself.

Like muscles in the limbs, heart muscles also can be weakened by lack of dystrophin. People with BMD often develops cardiomyopathy-heart muscle weakness because of deficiency of dystrophins. Dilated cardiomyopathy in BMD is not necessarily related to the skeletal sign and symptoms (Mavrogeni *et al.*, 2015) [5]. Cardiac involvement is prominent feature of disease and heart failure is the common cause of early death in BMD patients [7-10].

Respiratory muscles often stay strong in BMD for many years, but eventually, they may become weaker over a period of time.

Complications and their treatment

Regarding treatment, there is no cure for BMD. Therefore the main aim of treatment is to control patients symptoms as they arise, as well as a supportive approach with a multidisciplinary team in order to improve the health related quality of life. The team should include physiotherapy, occupational therapy, as well as speech therapy. Physiotherapy focuses on strengthening muscles as well as helping the patients to be as physically functional as possible. Occupational therapy focuses on aiding the patient with activities of daily living, as well as education and job difficulties encountered due to the disease. Speech therapy is needed in BMD patients.

Musculoskeletal complications

Weakness can lead to joint contractures and scoliosis, which may require orthotic or orthopedic treatment.

Cardiac complications

It includes cardiomyopathy and congestive heart failure. Atrial and ventricular arrhythmias may be life threatening. Regular cardiac monitoring from diagnosis including clinical evaluation, ecg, echocardiography.

Respiratory deteriorations once started should involve noninvasive intermittent positive pressure ventilation

Diagnosis

It is based on neurological examination and including following tests [5].

CPK blood test- CPK levels rise in the blood due to muscle damage.

EMG-It is used to examine the response of the muscles to stimulation. Decreased response is seen in muscular dystrophy. It typically reveals increased insertional and spontaneous activity in the form of fibrillation potentials and PSWs (positive sharp waves), along with brief small, polyphasic MUAPs (motor unit action potential) with early recruitment.

Muscle biopsy- Changes in the structure of muscles cells and presence of fibrous tissue or other aberrant structure are characteristic of different forms of muscular dystrophy.

Genetic Testing- Dystrophin gene deletion analysis shows deletions in about 98% of specific cases.

Summary

BMD is a X linked recessive disorder affecting mainly males. Most patients of BMD fist experiences difficulty between ages 5 and 15yrs [1]. By definition, patients with BMD walk beyond age 15.Cardiac involvement occurs in BMD. Diagnosis required clinical evaluation, EMG, muscle biopsy. Uses of glucocorticoids has not been adequately studied in BMD.

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