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Research Article

PREVALENCE OF DUCHENNE MUSCULAR DYSTROPHY: A CASE REPORT

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ABSTRACT

Muscular dystrophy MD comprises a group of diseases characterized by progressive muscle weakness that induces functional deterioration. It is caused by mutation in dystrophin gene located on x chromosomes. This study aimed to investigate that certain people are affected by muscular dystrophy in Tirupati [A.P] particularly patients affected by DMD and BMD and to figure out the patients affected by different types of MD. The characteristics of different types of MD are discussed .The patients diagnosed with DMD [classified severe progressive MD] BMD (classified slow progressive MD) Around 43 cases have been reported in the study area and 12 case were been reported and projected in case study. The result revealing depicts that 12 out of 7 are affected by DMD and 2 cases are affected by BMD remaining 3 refused muscle biopsy but having the symptoms of MD. Since most centers in developing countries have limited facilities for investigation of patients with MD and similar disorders. Laboratory techniques performed like ECG, EMG, 2DECHO, Serum creatine kinase and Muscle biopsy by using western blot method was performed. The main cause of concern is the ignorance and poverty line and below poverty line are the primary reasons. More over Muscular Dystrophy is prevailing in some part of socio economic zones as well. By giving proper hospitalization, awareness we can improve the life span of MD patients.

Keywords: Muscular dystrophy, Duchenne muscular dystrophy, Becker Muscular dystrophy, clinical findings.

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INTRODUCTION

The muscular dystrophies are a group of common and distressing neurological conditions with scarcely any effective treatment options available to date, despite the rapid strides in developing molecular diagnosis and the possibility of both pharmacological and genetic therapeutic strategies. The term ‘muscular dystrophy’ traditionally refers to a group of genetically determined, progressive, degenerative disorders of muscle. Many patients can be diagnosed clinically as muscular dystrophy. The usual investigations carried out for such patients are estimation of serum creatine kinase (CK), electrophysiological evaluation and histopathological examination. Diagnosis of muscular dystrophies and their classification for the purpose of prognostication requires evaluation with special immunohistochemical staining of histopathology specimens, which are available only at selected centers in developing countries like India. In developing countries, most centers have limited facilities for the investigation of such patients. As a result, investigations for patients with suspected muscular dystrophies are carried out according to the availability of investigative facilities, with no specific guidelines for their use. There are a number of myopathic disorders and some neurogenic disorders like congenital myopathies, mitochondrial disorders and spinal muscular atrophies, which may clinically mimic muscular dystrophies and may be difficult to differentiate

from the latter, unless sophisticated investigations like immunohisto chemical staining of muscle biopsy specimens or genetic analyses are carried out. [1-4]

MATERIALS AND METHODS

Detailed phenotype was noted in all the patients.Laboratory evaluation included complete heamogram,urinenalysis,serum biochemical analysis including muscle enzymes,ECG,2D ECHO, chest X-ray, nerve conduction(NC) and electromyography(EMG) and Muscular Biopsy western blot studies were performed. Some patients underwent a quadriceps muscle biopsy; serial sections were stained for light microscopy.

CASE REPORT

In Tirupati, 43 patients with muscular dystrophy were observed. In our study, we have collected case report of 12 patients suffering with different types of MD since three years. The information was collected from Department of Neurology in various hospitals of Tirupati (AndhraPradesh, INDIA).

Duchenne muscular dystrophy (DMD)

Out of 12,7 cases of DMD-one was female and remaining 6 patients were male.5 patients started walking normally, but had the onset of the disease before 5years of age.2 had onset of disease between 5 and 8 years. The mean age of onset was seen to be 6 years. The onset of disease was found in pelvic girdle musculature in 5 patients and with calfs hypertrophy.2 cases manifested onset with crural muscle weakness. The distribution of muscle weakness is central to this disorder. In 4 patients, weakness was global. These were advanced cases. Remaining 3 patients had pelvic girdle weakness, shoulder girdle weakness, distal muscle weakness, leg weakness and fore arm weakness. Facial weakness was seen in 2patients, hypertrophy was seen in the calves in 6 of the patients.2 patients had gluteal, 3 patients each had deltoid and triceps and 2 patients had quadriceps hypertrophy. Mental backwardness was psychometrically confirmed in 2 patients, serum total creatine phosphokinase was found elevated (1675IU) , electromyogram was myopathic in 6 patients and ECG,persistent tachycardia commonly noted.atrial and ventricular beats and more complex or sustained ventricular ectopy which increase with age and ventricular dysfunction.Chronic respiratory insufficiency was reported in all patients. Obstructive sleep apnea is predominant in 4 patients. Intellectual disability is seen in 1 patient. Orthopedic complications like scoliosis develops in almost all patients, osteoporosis and loss of bone mineral density were observed.

Becker muscular dystrophy (BMD)

Out of 12 patients, 2 patients were affected by this type. Both were male. They had born of consanguineous parentage. They had onset of symptoms in the hip girdle, calf hypertrophy and with toe walking at onset. They were not mentally backward.Face, hip muscles, shoulder girdle was affected.Mean serum CK was elevated (4000IU), EMG was myogenic; muscle biopsy was diagnostic of dystrophy in both the patients.

The remaining patient investigations revealed elevated serum CK values,EMG was myogenic in all patients,they were not mentally backward.Face,hip muscles,shoulder girdle was affected.In muscle biopsy,there was no dystrophy abnormalities.So,we can assume that they were autosomal recessive muscular dystrophy or limb girdle muscular dystrophy.But,due to lack of diagnostic specificity like dystrophin immuno staining and polymerase chain reaction(PCR) based dystrophin gene analysis for various muscle proteins and there is no detailed family history, we cannot asses the particular type of MD of which these patients were affected.

RESULTS AND DISCUSSION

Out of the 12 patients studied with laboratory evaluation, serum biochemical analysis including muscle enzymes, ECG, EMG, 2D Echo, Chest X-ray, studies were performed. The mean CK value was 1675 IU for DMD boys. Muscle biopsies of boys with DMD can be demonstrated by western blot method analysis using antibodies directed against different epitopes of dystrophy. The amount of dystrophin protein as well as size of the protein in DMD boys has less than 4% of the normal quantity of dystrophin is present when carboxyl terminal antibodies are used. Family history was positive in 2 patients and four patients were born of consanguineous marriage and one of the patients was born on non- consanguineous marriage. The mean age at presentation was 14-21 years. Two patients were affected by Becker muscular dystrophy born of consanguineous marriage. Mean CK value was 4000IU. They underwent muscle biopsy by western blot method. The Immunoreactivity to carboxyl terminal antibodies is absent in DMD and is therefore useful in differentiating DMD from BMD. Due

to non-availability of gel-electrophoresis to study the deletion patterns of BMD has not performed. Remaining patients refused biopsy but they are having symptoms of muscular dystrophy like progressive muscular wasting, poor balance respiratory difficulty was noted. Those patients mean age was 20-40 years affecting by different types of MD. An ECG performed on 12 patients by no abnormality of rhythm was detected in any patient, 2D-Echo cardiograph was performed in12 patients and was abnormal in 9 patients those who are affecting by DMD and BMD muscular diseases, suggesting reduced contractility of the myocardium and segmental wall motion abnormality.

Table 1: Summary of 2 Patients with Becker Muscular Dystrophy

FEATURES	Patient – 1	Patient – 2
Gender	Male	Male
Age at Presentation (Years)	14	15
Age at onset (years)	13	14
First symptom	Hip girdle	Calf hypertrophy
Family History And Inheritance	AD	Nill
Contracture	Neck extensors	Elbow flexors
Weakness	Proximal UL	Proximal UL/PL
Wasting	Biceps, Triceps	Arm, Anterior tibial
Muscular stretch reflex	Diminished	Diminished(UL)
Cardiac involvement	Myogenic	Myogenic
Micellaneous	Nill	Hyptonia
CK	Elevated (7 times)	Elevated
NC Study	Normal	Normal
EMG	Myogenic	Myogenic

Table 2: Summary of 7 Patients with Duchenne Muscular Dystrophy

Patient	1	2	3	4	5	6	7
Gender(Female-F, Male-M)	M	M	M	M	M	M	M
Age at presentation (Years)	2	7	18	17	18	6	5
Age at onset (years)	1	3	1	4	6	3	1
First Symptom	DMF	MB	DMF	DMF	DMF	DMF	DMF
Family history and inheritance	AD	AD	AD	AD	AD	AD	AD
Contracture	Ankles Biceps	Biceps Lliopsoas	Biceps Ankles	Biceps Ankles	Biceps Ankles	Biceps Ankles	Biceps Lliopsoas
Weakness	P L\L	P U\L	muscle	P U\L	P U\L	P L\L	P L\L
Wasting	Neck	extensors	flexors	Neck	extensors	Neck	flexors
Muscular stretch reflex	D	D	D	D	D	D	D
Micellaneous	Hyptonia	Hyptonia	NII	NII	NII	NII	NII
CK level (1U/L	E	E	E	E	E	E	E
NC study	N	N	N	N	N	N	N
EMG	M	M	M	M	M	M	M

NOTE:-

DMF - DELAYED MOTOR FUNCTION, D - DIMINISHED, E - ELIVATED, N - NORMAL, M - MYOGENIC.

CONCLUSION

Duchenne and Becker muscular dystrophy 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 and respiratory support increase, 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 linear below poverty line is the primary reasons for MD prevailing in some part social economic zone. The government has to curb the

situations or circumstances of these ailments which is quite imperative.

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