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Original Research Article

Profile, types, duration and severity of muscular dystrophy: a clinical study at a tertiary care hospital

Viswajyothi P., Rambabu M. V.*

Department of General Medicine, Malla Reddy Institute of Medical Sciences, Suraram, Hyderabad, Telangana, India

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***Correspondence:**
Dr. Rambabu M. V.,
E-mail: drmvrambabu@gmail.com

ABSTRACT

Background: The cause of muscular dystrophies is genetic. It is a disorder of muscular system. The classification of the muscular dystrophies is based on the signs and symptoms. Present study was done to evaluate the profile, types, duration and severity of muscular dystrophy at a tertiary care hospital.
Methods: A hospital based cross sectional study was carried out at department of General Medicine, Malla Reddy Institute of Medical Sciences, Hyderabad from October 2015 to December 2017. During the study period, it was possible to study the 20 cases of muscular dystrophy.
Results: Muscular dystrophy was more common in males. Maximum cases were of Duchenne type of muscular dystrophy. Majority of the patients presented at 5-10 years of age. Muscular dystrophy was seen in early childhood. Out of 10 patients of Duchenne muscular dystrophy five patients were of grade I. There was no correlation between the duration of the disease and the severity of the disability. All patients had lower limb proximal weakness. Pathological Q wave (width > 30 ms) and Pathological Q wave (depth >more than 25% of the QRS amplitude) were present in 35% of the cases. All patients had rhythm NSR and QRS +60 to +75. Conduction abnormality was present in 5% of the cases. In half of the patients, serum creatinine kinase levels are moderately elevated.
Conclusions: Muscular dystrophies are a common disorder of the childhood. Detailed studies will help to focus more light on this condition to improve outcome in future patients.

Keywords: Becker, Clinical study, Duchenne, Muscular dystrophy

INTRODUCTION

Among all the types of muscular dystrophies, children are commonly affected by Duchenne muscular dystrophy. Disease progresses very fast and severely. There is loss of mobility. This can occur as early as the age of 10 years. Some can be affected by insufficiency of respiratory system. This can lead to requirement of support on ventilator to the affected patient. This can occur at around the age of 20 years.[1]

Next most common type of muscular dystrophy after Duchenne muscular dystrophy is the Becker muscular dystrophy. This is considered to be less severe as compared to the Duchenne muscular dystrophy. It is also variable as compared to the Duchenne muscular dystrophy.

After Duchenne muscular dystrophy and Becker muscular dystrophy the third common type of muscular dystrophy is limb girdle muscular dystrophy. It can mimic either Duchenne muscular dystrophy or Becker muscular dystrophy. It is common in children. There is reduced mobility of the affected patient. But fortunately it is more likely to occur at around the age of 40 years of life. Heart is not involved in this type.[2]

All these three types of muscular dystrophies are known to carry a long course. Over the period of time, there is decreased mobility. Due to decreased mobility the quality of life of the affected patients is deteriorated.[3]

The cause of muscular dystrophies is genetic. It is a disorder of muscular system. The classification of the muscular dystrophies is based on the signs and symptoms. Dystrophin glycoprotein complex is important to maintain the skeletal muscle fibers integrity. The problem in this dystrophin glycoprotein complex leads to muscular dystrophy.[4]

Dystrophin, 427 kDa protein product of the dystrophin gene, is one of the largest components of the DGC. [5] Its absence or severe deficiency is seen in Duchenne muscular dystrophy. Becker muscular dystrophy cases show its reduced levels. [6]

Duchenne muscular dystrophy is the most common type of muscular dystrophy and constitutes nearly 80% of the total cases. One in every 3500 live born males can suffer from Duchenne muscular dystrophy. [7]

Present study is carried out to throw light on the picture of the muscular dystrophies in South Indian set up.

METHODS

A hospital based cross sectional study was carried out at department of General Medicine, Malla Reddy Institute of Medical Sciences, Hyderabad from October 2015 to December 2017. During the study period, it was possible to study the 20 cases of muscular dystrophy.

Protocol of the study was submitted to the Institutional Ethics Committee. After its approval, the study began. Informed consent was taken from all patients included in the present study.

Patients willing to participate, able to give proper history and cooperative for clinical examination was included in the present study. Seriously ill patients, not willing to participate, not able to give proper history were excluded from the present study.

20 consecutive cases of muscular dystrophy admitted in the neurology ward during the study period and satisfying the inclusion and exclusion criteria of the present study were included in the present study.

Detailed history and thorough clinical examination was carried out and recorded in the pre designed, pre tested, and semi structured study questionnaire prepared for the present study.

Detailed history included gender of the patient, history of consanguineous marriage in the parents of the patient, family history of muscular dystrophy, age at presentation of muscular dystrophy in the patient, age at onset of the muscular dystrophy in the patient, the interval from the age at onset to the time the patient first presented was taken as duration of the disease was asked and recorded in the questionnaire of the present study.

Symptoms were inquired into like difficulty in getting up from squatting position, frequent falls, abnormality of the gait, difficulty in climbing stairs; weakness of both the upper and lower limbs was noted and recorded in the questionnaire of the present study.

Detailed clinical examination like tendon reflexes, examination for skeletal deformity, sensory system examination, physical examination of the cardiovascular system, type of muscular dystrophy was carefully done and recorded.

All patients were subjected to routine laboratory tests viz. complete blood count, hemoglobin, ESR, complete urine analysis, blood sugar, and blood urea, serum electrolytes, creatinine phosphokinase were carried out and recorded. All patients also underwent X ray chest. ECG was done in all cases. 2D echo was done in all patients.

Motor nerve conduction studies were done on medium ulnar, peroneal and tibial nerves on both sides. Electromyography was done with concentric needle.

A neurostar, Medelec MS 92B electromyography was used the room temperature was maintained between 25 to 300C. Muscle biopsy of the vastus lateralis in the majority of patients showed degeneration of muscle fibers with regeneration and interstitial fatty infiltration.

Depending upon the severity of disability at presentation the patients were graded as

- Grade I: Ambulant
- Grade II: Ambulant with support
- Grade III: Wheel chair bound
- Grade IV: Bed ridden

The data was recorded in the Microsoft Excel Worksheet and analyzed using proportions.

RESULTS

Table 1 shows sex wise distribution of study subjects. Muscular dystrophy was more common in males than females. Compared to 90% incidence of muscular dystrophy in males it was only 10% in females.

Table 1: Distribution of study subjects as per sex.

Sex	Number	Percentage
Male	18	90
Female	02	10
Total	20	100

Table 2: Distribution of study subjects as per the type of muscular dystrophy.

Type of muscular dystrophy	Number	Percentage
Duchenne	10	50
Becker	07	35
Limb girdle	02	10
Female Duchenne	01	05
Total	20	100

Maximum cases were of Duchenne type of muscular dystrophy in 50% followed by Becker type of muscular dystrophy in 35% of the cases. There was only one case of female Duchenne type of muscular dystrophy.

Table 3: Age at presentation of muscular dystrophy.

Age at presentation (years)	Number	Percentage
0-5	02	10
5-10	08	40
10-15	04	20
15-20	02	10
20-25	01	10
25-30	02	10
30-35	01	10
Total	20	100

Majority of the patients presented at 5-10 years of age i.e. 40% to the hospital followed by 10-15 years of age i.e. 20%. Only one case presented at 30-35 years of age.

Table 4: Age at onset of muscular dystrophy.

Age at onset (years)	Number	Percentage
0-5	10	50
5-10	04	20
10-15	02	10
15-20	0	0
20-25	02	10
25-30	02	10
30-35	0	0
Total	20	100

Half of the patients gave history that muscular dystrophy age of onset was between 0-5 years followed by 5-10 years at age group of 5-10 years. Only 10% of the patients had age of onset at 25-30 years of age. Thus, muscular dystrophy is seen in early childhood.

Table 5: Duration of disease in muscular dystrophy.

Duration of disease (years)	Number	Percentage
0-2	09	45
2.1-4	05	25
4.1-6	03	15
6.1-10	01	05
10.1-15	02	10

The interval from the age of onset to the time the patient first presented was taken as duration of disease. In 9 patients (45%) the symptoms existed for over two years before seeking medical aid. 5 patients had the disease for less than 4 years.

Table 6: Degree of disability at presentation in muscular dystrophy.

Type of muscular dystrophy	No. of patients (%)	Degree of disability (%)			
		I	II	III	IV
Duchenne	10 (50)	5 (50)	2 (20)	1 (10)	2 (20)
Becker	07 (35)	7 (100)	0	0	0
Limb girdle	02 (10)	2 (100)	0	0	0
Female Duchenne	01 (05)	1 (100)	0	0	0

Out of 10 patients of Duchenne muscular dystrophy five patients were of grade I, two patients were in grade II, one patient was in grade III and two patients were in grade IV. For other types of muscular dystrophies, all patients belonging to those categories were having grade I disability.

Table 7: Correlation between duration of disease and severity.

Duration of disease (years)	No. of patients (%)	Degree of disability (%)			
		I	II	III	IV
0-2	09 (45)	4 (44.5)	2 (22.2)	1 (11.1)	2 (22.2)
21.-4	05 (25)	5 (100)	0	0	0
4.1-6	03 (15)	3 (100)	0	0	0
6.1-10	01 (05)	1 (100)	0	0	0
10.1-15	02 (10)	2 (100)	0	0	0

As the duration of disease increased the degree of disability did not increase. In fact, number of patients with increased duration of disease decreased. There was no correlation between the duration of the disease and the severity of the disability.

Table 8: Clinical features in the study subjects.

Initial symptoms	Number	Percentage
Difficulty in getting up from squatting	06	30
Frequent falls	03	15
Abnormality of gait	05	25
Difficulty in climbing stairs	04	20
Weakness of both upper and lower limbs	02	10
Total	20	100

Majority i.e. 30% of the patients had difficulty in getting up from squatting position followed by abnormality of

gait in 25% of the cases. This was followed by difficulty in climbing stairs in 20% of the cases.

Table 9: Nature of distribution of weakness in muscular dystrophy.

Distribution of weakness	Number	Percentage
Lower limb proximal	20	100
Lower limb distal	04	20
Axial muscles	02	10
Neck drop	02	10
Respiratory muscles	01	05
Total	20	100

All patients had lower limb proximal weakness. 20% had lower limb distal weakness. 10% of the patients had axial muscle weakness and neck drop each. 5% of the cases had respiratory muscles weakness.

Table 10: ECG abnormalities in the study subjects.

ECG abnormalities	Number	Percentage
Rhythm NSR	20	100
QRS +60 to +75	20	100
RAO/LAO	01	05
R/S VI in VI	14	70
Pathological Q wave (width >30 ms)	07	35
Pathological Q wave (depth > more than 25% of the QRS Amplitude)		
Conduction abnormality	01	05

Pathological Q wave (width >30 ms) and Pathological Q wave (depth > more than 25% of the QRS amplitude) were present in 35% of the cases. All patients had rhythm NSR and QRS +60 to +75. Conduction abnormality was present in 5% of the cases.

Table 11: Serum creatinine kinase levels in the study subjects.

Serum creatinine kinase	Number	Percentage
Markedly elevated > 1001	04	20
Moderately elevated 500-1000	10	50
Minimally elevated 100-500	06	30
Total	20	100

In half of the patients, serum creatinine kinase levels are moderately elevated. 30% of the patient's serum creatinine kinase levels were minimally elevated. Only 20% of the patients had markedly elevated serum creatinine kinase levels.

DISCUSSION

Muscular dystrophy was more common in males than females. Compared to 90% incidence of muscular dystrophy in males it was only 10% in females. Maximum cases were of Duchenne type of muscular dystrophy in 50% followed by Becker type of muscular dystrophy in 35% of the cases. There was only one case of female Duchenne type of muscular dystrophy. Majority of the patients presented at 5-10 years of age i.e. 40% to the hospital followed by 10-15 years of age i.e. 20%. Only one case presented at 30-35 years of age. Half of the patients gave history that muscular dystrophy age of onset was between 0-5 years followed by 5-10 years at age group of 5-10 years. Only 10% of the patients had age of onset at 25-30 years of age. Thus, muscular dystrophy is seen in early childhood. The interval from the age of onset to the time the patient first presented was taken as duration of disease. In 9 patients (45%) the symptoms existed for over two years before seeking medical aid. 5 patients had the disease for less than 4 years. Out of 10 patients of Duchenne muscular dystrophy five patients were of grade I, two patients were in grade II, one patient was in grade III and two patients were in grade IV. For other types of muscular dystrophies, all patients belonging to those categories were having grade I disability. As the duration of disease increased the degree of disability did not increase. In fact, number of patients with increased duration of disease decreased. There was no correlation between the duration of the disease and the severity of the disability. Majority i.e. 30% of the patients had difficulty in getting up from squatting position followed by abnormality of gait in 25% of the cases. This was followed by difficulty in climbing stairs in 20% of the cases. All patients had lower limb proximal weakness. 20% had lower limb distal weakness. 10% of the patients had axial muscle weakness and neck drop each. 5% of the cases had respiratory muscles weakness. Pathological Q wave (width >30 ms) and Pathological Q wave (depth > more than 25% of the QRS amplitude) were present in 35% of the cases. All patients had rhythm NSR and QRS +60 to +75. Conduction abnormality was present in 5% of the cases. In half of the patients, serum creatinine kinase levels are moderately elevated. 30% of the patient's serum creatinine kinase levels were minimally elevated. Only 20% of the patients had markedly elevated serum creatinine kinase levels.

Dey S et al studied 81 cases and found that the average age at onset was 3.9 years.[8] Calf hypertrophy and Valley sign was present in all cases. Facial weakness was seen in half of the cases. Deletion in distal exons was seen in 72.6% of the cases. Deletion in both proximal and distal exons was found in 16.4% of the cases. Proximal deletion was present in only 10.9% of the cases. The author stated that the clinical features and genetic pattern were not correlated.

Manjunath M et al found that age at onset mean was 45.3 years.[9] But we found that the age at onset was childhood age. The author stated that the disease mean duration was 53.3 months. We found that the majority of our cases had 0-2 years of disease duration. The author found that the mean value of creatine kinase was 12136. But in the

present study we found that majority of our cases had lower levels of creatine kinase.

Swaminathan B et al observed that the mean age at onset of the muscular dystrophy was 3.1 years.[10] Authors observed in the present study the similar findings i.e. majority of the children affected in the present study. The author reported that the mean age at presentation of the cases to the hospital was eight years. This finding is in accordance with the finding of the present study where we also noted that majority of the patients presented to our hospital at similar age groups. But the author reported a very high level of 11822.64 in their study which is contrary to the finding of the present study. We observed that majority of the patients had low levels of creatine kinase.

Rao MV et al noted that the majority of the patients in their study had markedly elevated creatine kinase.[11] This finding from this study is contrary to the finding from the present study. In the present study we observed that majority of the patients had low levels of creatine kinase. The author stressed that still more studies are required to throw a more light on the muscular dystrophies.

CONCLUSION

Muscular dystrophies are a common disorder of the childhood. Detailed studies will help to focus more light on this condition to improve outcome in future patients.

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