ORIGINAL ARTICLE

ORO-FACIAL DYSFUNCTION IN PATIENTS WITH MUSCULAR DYSTROPHIES

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ABSTRACT

Muscular dystrophies are a group of genetic neuromuscular disorders characterized by skeletal muscle weakness and wasting. Our aim was to observe the facial morphology and certain features of oro-facial function in patients with different forms of muscle dystrophy. This study was conducted in the Division of Morbid Anatomy and Histopathology at University of Health Sciences after ethical review committee approval. A total of 100 patients with clinical suspicion of muscular dystrophy reporting at the Children Hospital and Institute of Child Health and Pakistan Society of Rehabilitation of Disabled (PSRD) Lahore from January 2010-2015were included.Among 100 patients, 77 were males while 23 were females. Mean age for onset of muscle weakness was 6.40± 4.226 years. A total of 6 patients presented with dysmorphic faces and jaw muscle weakness along with other characteristics suggesting other forms of muscular dystrophies. Muscular dystrophies involve facial morphology and alter the functioning of jaw muscles that carries both diagnostic and therapeutic implications.

Key Words: *Muscle dystrophy, Dysmorphic face, Jaw muscle weakness.*

INTRODUCTION

Muscular dystrophies are inherited disorders caused by genetic mutations. These mutations result in either a dysfunction or lack of proteins that are needed for maintaining muscle cell stability, thus may lead to progressive destruction and weakness in the muscle.[1] Muscular dystrophies are clinically divided into six types as Duchenne (DMD), Becker (BMD), Limb-girdle (LGMD), Congenital (CMD), Facio-Scapulo-Humeral Muscular dystrophy (FSHMD), distal myopathies and myotonic dystrophy (MD).[2]

Duchenne muscular dystrophy (DMD) is the most common X-linked recessive disease.[3] It involves most serious myopathies affecting approximately 1: 3500 live born males.[4] It usually appears between 2 and 5 years of age along with clinical warning sign of muscle weakness in the limbs and pseudo hypertrophic muscles of calf.[5] Patients are wheelchair bound at the age of 10-12 years because of the loss of ambulation.[6]

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The disease is caused by mutations in the dystrophin gene 3 that encodes the sarcolemmal protein product, a part of the dystrophin glycoprotein complex and also contacts the motor protein actin to the cytoskeleton.[7] It gives mechanical firmness to striated muscles.[8] Deficiency of dystrophin, collapses dystrophin glycoprotein complex thus leading to instability of muscle cell membrane, abandoned calcium influx sterile inflammation, and advanced muscle degeneration.[9,10] The masticatory muscles are involved later along with time span while the oro-facial muscles are affected approximately after 2 years resulting in muscle imbalance and severe craniofacial and dental abnormalities.[11]

Duchenne muscular dystrophy interrupts the oro-facial function and brings the difficulties in the pre-oral phase of swallowing.[12] The bite force and mouth opening distance in these patients are considerably lower than maximum.[13] Macroglossia, tongue propelling, mouth breathing and lip incompetency commonly seen in DMD patients.[14] Mandibular elevator muscles are rotated downward and backward because of progressive weakness thus leading to turning of the mandible away from the maxilla resulting in long face deformity with open bite.[11]

The development and morphology of the craniofacial structures and muscles is effected in patients with DMD. As a result of weakness of the jaw muscles, along with changes in the craniofacial morphology, may affect mandibular function, involving mandibular range of motion (ROM).[12,13]

Facio-scapulo-humeral muscular dystrophy (FSHD) is an autosomal dominant disease with an increase occurrence of de novo cases.[14] There is markedly variable clinical involvement with a wide range of severity in FSHD even within the same family.[15] It is the third most common muscular dystrophy involving the facial muscles resulting in a characteristic clinical appearance that may contribute to reduced facial wrinkles.[16]

Congenital muscle dystrophy related with Laminin alpha-2 deficiency (MDC1A) is characterized by late or halted motor milestones, feeding problems, marked muscle weakness, atrophy, and facial dismorphism.[17,18] Limb girdle muscular dystrophy type B1 an autosomal dominant type of limb girdle muscle dystrophy associated with mutations in LMNA.[1] Facial muscle weakness is sometimes distinctive feature in patients with autosomal dominant limb girdle muscle dystrophy.[19] This study was therefore designed to determine the frequency and pattern of oro-facial changes in patients presenting with different forms of muscular dystrophies. Thus it will be helpful in diagnostic and therapeutic purpose.

METHODOLOGY

A total of 100 patients belonging to all age groups, both male and female with strong clinical suspicion of muscular dystrophy, irrespective of positive family history, were included by convenient sampling technique. These patients reported at the Children Hospital and Institute of Child Health and Pakistan Society of Rehabilitation of Disabled (PSRD), Lahore from January 2010-2015. This study was conducted in the Department of Morbid Anatomy and Histopathology at University of Health Sciences after the approval by Ethical Review Committee of University. Informed written consents were taken from guardians of pediatric patients while adult patients gave consent by themselves.

RESULTS

Among 100 patients, 77 were males while 23 were females only. Age range for all patients was 01- 31 years while the mean age for the onset of muscle weakness was 6.40±4.226 years.

A total of 6 patients presented with dysmorphic faces including coarse facial structures, broadened epicanthic folds, prominent upper jaw and high arched palate along with other characteristics suggesting different forms of muscular dystrophies. Among 6 patients with dysmorphic facial features 3 patients were suggestive of DMD as fulfilling the criteria described by Pereirasuch as predominantly proximal muscle weakness with symmetrically involvement, exercise intolerance, positive Gower's sign, muscle hyporeflexia / hypotonia,contractures and dysmorphic faces.[20] While other 3 patients presented with clinical features suggestive of musculardystrophies other than the typical DMD (Table 1).

The first male patient (age: 15 years) presented with prominent maxilla, high arched palate and jaw muscle weakness along with lower limbs involvement as a primary weakness subsequently symmetrical and severe involvement of the upper limb girdle and impaired hearing were indicative of FSHMD or LGMD. The second male patient (age: 05 years) presented with dysmorphic face along with limb and upper limb girdle involvement was indicative of MDC 1A or LGMD.

The third male patient (age: 03 years) presented with dysmorphic face with broad nasal bridge and jaw muscle weakness along with symmetrical limb involvement and spared girdles; thus suggestive of the probability of LGMD (Fig 1).

TABLE: 1- CLINICAL FEATURES IN PATIENTS SUSPECTED OF MUSCULAR DYSTROPHIES OTHER THAN DMD

Clinical Feature	Case 1	Case2	Case 3
Age present (years)	15 years	05 years	03 years
Age of onset (years)	11 years	Prenatal	Within first 6 months
Gender	Male	Male	Male
Ambulation	Partial	Nil	Nil
Dysmorphic face	Present	Present	Present
Jaw muscle weakness	Present	Absent	Present
Limb Involvement	Symmetrical	Symmetrical	Symmetrical
Upper Girdle involvement	Present	Present	Absent
Impaired hearing	Present	Absent	Absent
	FSHMD/LGMD	MDC1A/LGMD	LGMD 1B

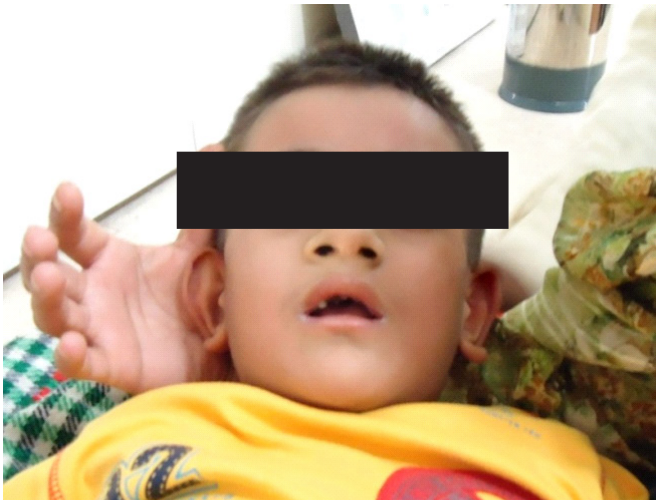

Fig 1: Dysmorphic face with broad nasal bridge and jaw muscle weakness in 03years male patient with clinical features suggestive of LGMD

DISCUSSION

The present study has shown that various functions of the oro-facial complex got affected in Duchene muscular dystrophy. Lip and posterior bite force exhibited lower values. As the patients gets older oro-facial complex is affected more. Bones continue to remodel during post-natal growth to keep a form that is suitable for their biomechanical function. Muscle function influences the skeletal and facial skeleton growth.[21] Duchenne muscular dystrophy interrupts the oro-facial function and brings the difficulties in the pre-oral phase of swallowing.[22] The bite force and mouth opening distance in these patients are considerably lower than maximum.[23] Macroglossia, tongue propelling, mouth breathing and lip incompetency commonly seen in DMD patients.[24]

Mandibular elevator muscles are rotated downward and backward because of progressive weakness thus leading to turning of the mandible away from the maxilla resulting in long face deformity with open bite.[11]

DMD affects muscles at different stages of the advancement of the disease like the weakening of the masseters that occurs earlier than labial muscles.[11] The discrepancy between lateral and anterior forces combined with the increased tongue volume, produces are more significant effects on dento- alveolar relations.[24] The posterior bite force is decreased that may be related with poor internal masseter muscle structure quality that gets wane with age, perhaps because of disease progression.[12] The progressive deterioration of function in the orofacial complex of DMD patients possibly influences the dentofacial growth thus giving rise to increasing malocclusions with age.[13]

The large inclined maxilla is present in these patients that may be because of palatal remodeling as a result of the increased tongue volume prompting mostly the posterior part of the maxilla. Mandibular elevator muscles cause the downward and backward rotation of the mandible away from the maxilla with a consequential long face deformity and open bite as a result of progressive weakness.[11] Factors affecting the masticatory system may lead to a decrease in the mandibular range of motion as well. Currently, functional association between the cranio-cervical and the masticatory system has been suggested.[25] A limited mouth opening may hinder feeding, maintenance of oral hygiene and dental care.[26]

Facial muscular involvement characterized by progressive hypotrophy and hyposthenia is associated with muscle decontraction, difficulties and muscle weakness, thus leading to an expression-less face.[27] Facial weakness is often the earliest obvious clinical feature in FSHD that shows the facial muscle involvement thus leading to reduced facial expression.[28] Asymmetric face followed by lower extremity muscle weakness is classically seen FSHD.[29] The similar feature also reported in our case.

Mercuri reported generalized hypotonia, severe limb muscle wasting and weakness in a patient with limb girdle muscle dystrophy (1B) at the age of 7 years. He also found dysmorphic face characterized by midface hypoplasia with a broad nasal bridge at age of 30 years showed.30In our study similar findings were observed in patient with limb girdle muscle dystrophy at the age of 3 years.

Klein and colleagues detailed severe facial weakness in certain congenital myopathies especially in subtype of laminin alpha-2 deficiency.[31] Similarly, in our study myopathic/ dysmorphic face alongwith symmetrical muscle involvement was seen during perinatal period thus presuming MDC 1A. Sparks also reported typical myopathic/dysmorphic facies in patients suffering with congenital muscle dystrophy.[32]

CONCLUSION

Muscular dystrophies not only affect the facial morphology but also alter the oro facial functioning. The imbalance between muscles and bones development leads to various dento-alveolar disorders. Thus consideration of orofacial function in patients with muscular dystrophies has both diagnostic as well as therapeutic implications as their life expectancy can be made better through proper management.

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