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ISSN No: 2319-5886

International Journal of Medical Research & Health Sciences, 2016, 5, 10:182-186

Early Therapeutic intervention for Limb Girdle Muscular Dystrophy in Late Adolescence – A Case Report

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ABSTRACT

This paper reports the benefits of strength & endurance training based programme in the early rehabilitation of Limb girdle muscular dystrophy (LGMD) patient. LGMD is an autosomal X-linked disorder, mainly involve shoulder & pelvic muscles with variable rate of progression occur in first & second decade of life. An intervention of 30 days (5 days per week for 6 weeks) was given to the patient & prognosis was observed on various outcome variables like Muscular dystrophy Functional Rating Scale (MDFRS), Berg Balance Sclae (BBS) & Brroke & Vignos Scale before & after the intervention. Considerable improvement was seen by applying both theory of strength & endurance training in patient with LGMD.

Keywords- LGMD, Muscular Dystrophy, Endurance training, Physiotherapy, Strength

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INTRODUCTION

Limb-girdle muscular dystrophy (LGMD) is a purely descriptive term, generally reserved for childhood- or adult-onset muscular dystrophies that are distinct from the much more common X- linked dystrophinopathies, which include Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) (Pegoraro, 2007). Walton and Nattrass first proposed limb-girdle muscular dystrophy (LGMD) as a nosological entity in 1954. Their definition included the following characteristics:

- Expression in either male or female sex
- Onset usually in the late first or second decade of life (but also middle age)
- Usually autosomal recessive and less frequently autosomal dominant
- Involvement of shoulder or pelvic-girdle muscles with variable rates of progression
- Severe disability within 20-30 years
- Muscular pseudohypertrophy and/or contractures uncommon

LGMDs are typically nonsyndromic & like other muscular dystrophies, is primarily a disorder of voluntary/ skeletal muscles. These are the muscles you use to move the limbs, neck, trunk and other parts of the body that are under voluntary control. Individuals with LGMD generally show weakness and wasting restricted to the limb musculature, proximal greater than distal. Proximal weakness refers to weakness of the muscles closer to the centre of the body (including the shoulder, pelvic girdle, upper thighs, and upper arms). Distal weakness refers to weakness in muscles farther from the centre of the body (including lower legs and feet, lower arms and hands) (Pegoraro, 2007).

Often, people with LGMD first notice a problem when they begin to walk with a “waddling” gait because of weakness of the hip and leg muscles. They may have trouble getting out of chairs, rising from a toilet seat or climbing stairs. Onset, progression, and distribution of the weakness and wasting may vary considerably among individuals and genetic subtypes the involuntary muscles, except for the heart (which is a special type of involuntary muscle), aren’t affected in LGMD. Digestion, bowel, bladder and sexual function remain normal. The brain, intellect and senses also are unaffected in LGMD. Cardiopulmonary complications sometimes occur in later stages of the disease (MDA). Over time, muscle weakness and atrophy can lead to limited mobility and disability
Establishing the Diagnosis (Pegoraro, 2007)

- The clinical course of the limb-girdle muscular dystrophies is typically progressive, though some individuals may show mild symptoms and/or the disease may stabilize.
- Serum creatine kinase (CK) concentration is usually elevated.
- Muscle biopsy typically shows degeneration/regeneration of muscle fibers ("dystrophic changes").
- In some LGMDs (i.e., sarcoglycanopathy, calpainopathy, dysferlinopathy, and glycosylation defects, or dystroglycanopathy) the diagnosis can be established based on "biochemical testing," i.e., immunostaining/immunoblotting of a muscle biopsy to determine if specific proteins are present or absent.
- In some cases, molecular genetic testing can be used to identify the specific disease-causing mutations.
- Inflammatory myopathy should be excluded during the diagnostic process.

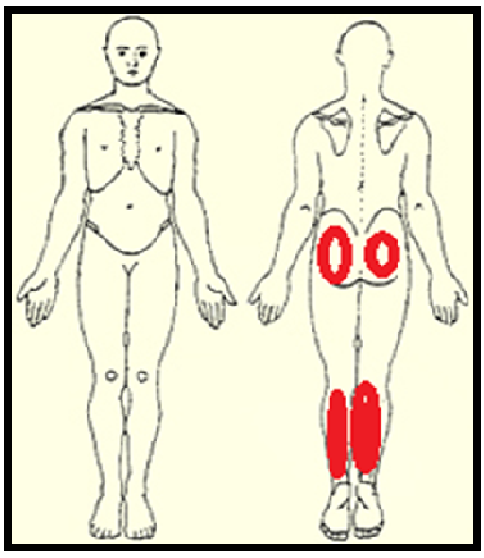
When a person is diagnosed as having a muscle disease, questions arise about the prognosis, possible interventions and genetics. However, people with muscle disease are usually also concerned about everyday issues such as participation in sports, work and hobbies. Weakness and impaired cardiorespiratory function are common in people with muscle disease. In healthy persons the best intervention to improve strength and cardiorespiratory function is training. Strength training or aerobic exercise programmes in people with muscle disease might maximise muscle and cardiorespiratory function and prevent additional disuse atrophy (Voet *et al*., 2011).

Strength training, which is performed to improve muscle strength and muscle endurance, or aerobic exercise programmes, which involve training at moderate levels of intensity for extended periods of time (for example, distance cycling) might optimise physical fitness and prevent additional muscle wasting in people with muscle disease(Voet *et al*., 2011).

Because many of the LGMDs are caused by defects in structural proteins in the DGC, a similar approach as in DMD/BMD is on theoretical grounds probably justified, in which high-resistance and eccentric training should be avoided, whereas low-resistance training may be justified. On the other hand, in those forms of LGMD where non-structural proteins are involved, high-resistance training may very well be proven to be beneficial. However, in the few existing reports on the effects of muscle training programmes in LGMD, the underlying molecular defect has not been known (Ansved, 2001).

CASE REPORT

A 19 year old male presented with complaint of feeling of heaviness & tightness in the bilateral lower leg region since few month. He also felt difficulty in doing sit to stand activities. He noticed these from last 1 month because he experienced more difficulty than before, in rising from sitting to standing. Then, patient consults a local doctor in nearby hospital, where echocardiography & serological test was done. In serology, the Creatinine phosphokinase levels were markedly increased. The doctor advised him to take physiotherapy treatment & referred him to another hospital for further evaluation & treatment. There various diagnostic tests like Electromyography (EMG), muscle biopsy was done. EMG of right deltoid & vastus lateralis muscles was done which shows myopathic process. Muscle biopsy of left biceps shows regenerating muscle fibers & infiltration by adipose tissue & patient was diagnosed as a case of Limb Girdle Muscular Dystrophy. (The source of history was patient himself & medical history was recorded from the investigatory reports carried by him).



Patient was not taking any medications except some home remedies (Ayurvedic medicine) & supplements (Vitamins, Calcium). No any genetic & family history wass found. Family members are cooperative & supported in the treatment of patient. Patient was able to perform ADL's independently. He was a student & able to attend classes & able to drive. Yet, no restriction of any activity was seen. Signed consent for the publication of this case report was obtained from the patient concerned.

On Observation, built was mesomorphic & on lateral view, patient stands with lordotic posture, anterior pelvic tilt & winging of scapula was seen (Posture of a patient was assessed by using postural analysis scale which is enclosed at the end of the case report). Ear, eye, head & facial expressions on observation were symmetrical. The pattern of respiration was symmetrical & type of respiration was thoraco-abdominal. The wasting of biceps & brachioradialis muscle was seen bilaterally. Hypertrophy of calf muscle was present bilaterally.

On Examination, the higher mental functions was assessed by Mini Mental Status Examination (MMSE) & the patient score 30/30. All the superficial, deep & cortical sensations were found to be intact (checked at dermatomal level). On Motor Examination, muscle tone as per Modified Ashworth Scale (MAS) was grade 0 i.e tone was normal in bilateral upper limb & lower limb. The range of motion (ROM) was assessed by goniometer & it was within normal limits.

Muscle girth was assessed by lacote method

Left	From olecranon process	Right
	Above (arm)	
24.1 cm	6 cm(biceps)	24.9cm
25.2 cm	9cm(long head of biceps)	26cm
26.7 cm	12cm (triceps)	27.1cm
	Below (forearm)	
24.2cm	3cm	25.1cm
24.9cm	6cm	25.6cm
24.5cm	9cm	25.3cm
	From upper margin of Patella	
	Above (thigh)	
42.6cm	12cm	41.8cm
44.2cm	15cm	43.3cm
46.8cm	20cm	46cm
	Below (lower leg)	
35.6cm	15cm	35.5cm
34cm	20cm	33.9cm

Muscle strength were grades according to oxford method. In upper limb, the bilateral shoulder elevators, scapular retractors and elbow flexors muscles were graded 3+ and rest of the upper musculature were secure more than 4 grade. In lower limb, the abductors of hip, elevators of pelvis and calf musculature were graded as 3- and rest of the muscular were more than more than 4 grade on manual muscle chart..

For balance assessment, Berg Balance Scale were used & patient score 50/56. The main component effected in BBS were Sit to stand with support, Standing on one leg for more than 10 seconds, Tandem stance, Transfer from one chair to another. On gait assessment, patient was ambulates independently with normal base of support. For functional assessment Functional Independence Measure (FIM) was used & score obtained was 121/ 126. Main problem areas in FIM were transfer skills & stair climbing.

On the basis of history, investigation reports & examination a clinical diagnosis of Limb girdle muscular dystrophy was made. The BROOKE & VIGNOS scale was used to rate the grade of disease severity & patient obtained grade 2 for UL & grade 2 for LL. In disease specific MDFRS (Muscular dystrophy functional rating scale) patient score 79/96.

After assessing the main problem area identified were difficulties in sit to stand activities & transfers, tightness in calf muscle, difficulty in climbing stairs & prolonged walking causes fatigue easily. The main goals of the physiotherapy treatment is -

1. To maintain muscle power and gain endurance.
2. To prevent secondary complications (deformities and contractures).
3. To correct and maintain posture.
4. Maintenance of respiratory function.
5. Gait training.
6. To promote ADLs.

The treatment protocol followed according to problem list & goals were described in following table-

1. To maintain ROM & muscle strength	• Active ROM exercises for both UL & LL- 3 sets of 10 reps TD. • AROM of scapular elevation, protraction & retraction- 3 sets of 10 reps TD. • PNF B/L D2 flexion pattern of UL synchronize with breathing- 3 sets of 10 reps TD. • Wall squats- 3 sets of 10 reps with 5 second hold TD. • Static abdominal muscle strengthening - 3 sets of 10 reps with 5 second hold TD. • SLR(Supine, side, prone) without weight 10 reps with 10 sec hold BD. • Superman exercises in quadruped position. • Pelvic bridging- 10reps with 8 sec hold TD. (Note- all exercises should be done without fatigue so proper rest or relaxation time should be taken b/w exercises). For muscle strengthening a PRE regime is followed (Delorme method).
2. To prevent secondary complications (deformities and contractures).	• Stretching of calf muscle, hamstring, adductors & flexors of hip bilaterally- 20sec h0ld, 5 reps. • AFO night splint to prevent contractures of calf muscles. • Medial arch support shoes.
3. Maintenance of respiratory function.	• Deep breathing exercises- diaphragmatic breathing exercises. • Segmental breathing Exercises.
4. Balance training-	• Balance board- maintenance of balance on BB for 5 min. • Single leg stance • Tandem stance
5. To promote ADL's	• Sit to stand activities is done on various heights of stools & chairs. • A transfer from one chair to another chair.
6. Gait training & posture re-education.	• Maintenance of correct posture is taught to the patient & family members. • Side walking & heel walking • Agility training
7. Endurance training	• Patient is advice to do cycling (15 min BD) , treadmill(15 min BD) & swimming at home without fatigue.
8. Psychological counselling	• Patient & his Family members were educated about diseases & its outcomes so that they prepare themselves to cope up with the condition in the future.

A home exercise programme was also included in the treatment which comprises of pulley exercises of shoulder joint, AROM exercises of bilateral upper limb & lower limb and self stretching of calf & hamstring muscle was also taught to the patient. After a rehabilitation protocol of 30 days significant changes were observed in the assessment outcome. The patient was now able to rise from sit to stand without use of support/hands, climb up stairs without support, felt less fatigue during exercises than before & also after prolonged walking.

PROGNOSIS

After giving above mentioned physiotherapy treatment for 30 days (5 days per week for 6 weeks), patient was reassessed & following prognosis is observed in patient-

Patient was now able to stand from sitting position without use of hands, able to climb stairs without support & does not get fatigue early & feeling less stiffness & heaviness in the lower leg.

Table.- Endurance of Patient at Baseline & after 30 days

Exercise	Initial evaluation Fatigue after	30 days Evaluation Fatigue after
Supine SLR	8 reps	15 reps
Side SLR	7 reps	15 reps
Prone SLR	4 reps	10 reps
Wall Squat	5 reps	10 reps
Cycling	5 min	15 min
Single leg stance	Able to hold <5 sec	Able to hold >10 sec
Sit to stand activities	3 reps	7 reps

Table- Outcome measurements at baseline & after 30 days

Test	Baseline evaluation	After 30 days
Berg balance test	50/56	54/56
MDFRS	79/ 96	89/96
Brooke & Vignos Scale	Grade 2 for UL Grade 2 for LL	Grade 2 for UL Grade 1 for LL

DISCUSSION

The implication of both strength & endurance training was an effective & safe method in the early rehabilitation of LGMD. When studying patients with a progressive disease like LGMD, treatment design takes on an added

importance, especially if the disease produces rapid deterioration in health status. Individuals with LGMD were characterized by a progressive weakening and loss of functional skeletal muscles. The disability associated with LGMD depends on the pathogenesis, extent of clinical involvement, and rate of progression (Abresch *et al.,* 2009). The loss of functional muscle fibre and atrophy of disuse lead to weakness, fatigue, poor endurance, and associated functional impairments in these patients. Although several therapeutic treatments had been proposed for LGMD, there was currently no effective pharmacologic management. So, the patients have to rely on symptomatic treatment in which continuous physiotherapy plays a vital role. The primary goal for physiotherapy treatment was to maintain strength, function, independence, and quality of life. Exercise is an essential tool to maintain and improve strength, increase endurance, improve function, and enhance quality of life in individuals with LGMD.

The exercise prescription used in the case report mainly emphasize on the empirical benefits of strength & endurance training & significant differences were observed in the various outcome variables between baseline evaluation & after giving 30 days of physiotherapy treatment. The strength training (progressive resistance exercise) increases lean body mass, muscle protein mass, contractile force, power, and improves physical function. Exercise induces muscle hypertrophy by increasing the DNA content in the myofibrils, which in turn, increases the amount of muscle proteins, especially actin and myosin. Endurance training induces physiological adaptations that differ from strength training and reducing fatigue as shown by the ability to perform submaximal work with less effort for longer duration (Abresch *et al.,* 2009). The lack of exercise exacerbates the reduction in muscle mass and increased obesity, which contributes to the high prevalence of metabolic syndrome observed in patients with NMDs (Abresch *et al.,* 2009).

CONCLUSION

Considerable progress has been made in developing both theory of endurance and resistance exercise for maintenance of health status in individual with LGMD. The improvement in patient depends upon the intensity, frequency & nature of the exercise programme, but the level of training and kind of training depends on the type, stage, and severity of the disease. In addition to these, outcome variables which are indicators of patient satisfaction, functional ability, quality of life & emotional well being also determine the benefits obtained from particular type of training used in rehabilitation.

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