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Case Report
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Physiotherapy in Calpainopathy (LGMD2A): A Case Report

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Abstract

Limb-Girdle Muscular Dystrophy-Type 2A (LGMD-2A) is an autosomal recessive disorder triggered by a mutation in the Calpain-3 gene (CAPN3) contributing to partial or complete protein deficiency. LGMD-2A is the most prevalent form of the disease in India, accounting for 47% of cases in the heterogeneous group. Here, we record a 22-year-old female with trouble walking due to proximal muscle weakness since one year and an elevated Creatine Phosphokinase (CPK) with abnormal muscle biopsy finding. The patient was granted an 8-week intervention and a prognosis was established.

Keywords: LGMD-2A; Muscular dystrophy; Physiotherapy

Introduction

The most prevalent autosomal recessive limb-girdle muscular dystrophy (LGMD) is Calpainopathy. The frequency of the LGMDs is difficult to determine because of their heterogeneity. LGMDs are graded into dominant (LGMD1) and recessive (LGMD2) types according to the pattern of inheritance. Form 2A (LGMD2A) limb-girdle muscular dystrophy is known as Calpainopathy which is a result of mutations in the CAPN3 gene [1-6]. The disease begins in childhood, usually between the ages of 8 and 15, with recorded onset ages ranging from 2 to 40 years.

Case Presentation

A girl, 22, was suspected of having a muscle disease. She was a first child born to a non-consanguine family, delivered without any complication. Her younger brother did not have any similar signs. The patient is an undergraduate student who has no history, allergies or prescription and can attend classes. In April 2018, she started having problems walking and ascending stairs. After walking up 2 to 3 stairs, she was able to walk with assistance and was exhausted.

Normal cranial functions with speech were demonstrated by neurological examination. Both reflexes of the superficial and deep tendons were acceptable. Test of the motor nerve of both lower limbs showed a score of 3/5 in hips, 4/5 in knees and 3/5 in ankles. Dorsiflexion and flexion in the plantar had been high. However, both upper limbs reported scoring of 3/5 in the shoulder, 4/5 in the elbow and 3/5 in the wrist. Compared to normal levels of 22 to 198 U/L, her serum CPK level was elevated to 3591 U/L.

Patient's muscle biopsy examination from right quadriceps was done in January 2019, which revealed the presence of lobulated fibers and incomplete fascicles. There was no sign of inflammation or vasculitis. Right Calf hypertrophy was present (Table 1). She exhibits slight curvature of the spine. Based on muscle involvement in disease, elevated CPK and muscle biopsy findings, the patient was suspected of having Duchenne muscular dystrophy. After reviewing her reports in august 2019 doctor diagnosed her with LGMD 2A (Calpinopathy) [2].

On observation, built was a mesomorphic and patient stand with slight lateral curvature towards left. Anterior pelvic tilt was seen. Ear, eye, head and facial expressions on observation were symmetrical. The pattern of respiration was symmetrical & pattern of breathing was thoracoabdominal. Functional foot flat has been observed.

On examination, a Mini-Mental Status Examination (MMSE) and 30/30 patient scoring were used to determine higher mental functions. Both deep and superficial sensations (checked at a dermatomic level) are intact.

Waddling was seen on the gait examination. The Functional Independence Measurement (FIM)

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Table 1: Muscle girth measurements.

Area	Right (cm)	Left (cm)
Arm	27.5	27.5
Forearm	18	18
Thigh	47	47
Calf	31	28

for functional evaluation was used, with a score of 121/126. Transfer skills and escalation were the main areas of problem in the FIM.

Full pneumological (spirometry and polysomnography) and cardio logical tests (echography, 24-h ECG Holter testing, cardiac MRI, thorough cardio logical physical evaluation with a focused medical history and cardiovascular reflex analysis) showed no significant anomaly.

After evaluating the critical problem area found is sit-in difficulties for tasks & transitions, tightness in the calf muscle, trouble ascending stairs & continuous walking causes fatigue quickly. The main goals of rehabilitation with physiotherapy are:

- To maintain muscle power and gain endurance.
- To prevent secondary complications (deformities and contractures).
- To correct and maintain posture.
- Maintenance of respiratory function.
- Gait training.
- To promote ADLs.

The treatment protocol followed according to the problem list is as described below physiotherapy management

To maintain ROM and muscle strength

- Active ROM exercises for both Upper limbs& Lower Limbs.
- Active ROM of scapular retraction & protraction
- Wall push-ups
- Abdominal muscle strengthening (tabletop, crunches, side crunches)
- SLR- supine, side-lying
- Pelvic bridging

To prevent secondary complications

- Stretching of
- Calf muscle
- Hamstrings
- Adductors of hip
- Medial arch support

Maintenance of respiratory function

- Diaphragmatic breathing exercise
- Deep breathing exercise
- Thoracic expansion exercises

Gait training & Posture re-education

- Maintenance of correct posture
- Gait training on the parallel bar

Endurance training

- Advised to do cycling, walk at home
- Swimming for 1 hour (without fatigue)

After delivering the physiotherapy care referred to above.

The home exercise regimen was also included within the procedure, including pulley exercise of the enarthrosis, AROM exercise of the bilateral upper limb and lower limb and calf and hamstring muscle self-stretching of the patient. After the 8-week recovery regimen, substantial improvements in the evaluation outcome were reported. The patient was now able to get up from the seat to stand with less support/hands, climb 18-19 stairs in one go, feel less exhaustion during exercises than before and even after a long walk.

After giving the physiotherapy as mentioned above, treatment for 8-weeks, the patient was reassessed. The patient was now able to rise from sitting to stand with less use of support/hands, climb up 18-19 stairs in one go, felt less fatigue during exercises than before & also after prolonged walking, and feeling less stiffness in lower limbs.

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