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Walking Against The Odds: The Clinical Story Of Limb-Girdle Muscular Dystrophy

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Abstract

We report the case of a 7-year-old female child who presented with non progressive bilateral lower limb weakness for four years, difficulty in getting up from a cross-legged sitting position for one month, and increased difficulty in walking and pain in the right leg for four days. Neurological examination revealed normal muscle tone, positive Gower's sign, and calf muscle hypertrophy. Deep tendon reflexes were present with bilateral extensor plantar responses. Investigations revealed elevated serum creatinine phosphokinase levels. Electromyography and nerve conduction studies were abnormal, suggestive of a primary muscle disease. This case emphasises the need for clinical awareness to enable early diagnosis and prevent misdiagnosis in children with underlying myopathies.

Keywords: NIL

Introduction

Limb-girdle muscular dystrophy (LGMD) represents a clinically and genetically heterogeneous group of inherited progressive myopathies characterized predominantly by weakness and wasting of the proximal muscles of the pelvic and shoulder girdles. To date,

more than 30 different subtypes of LGMDs have been identified, with an estimated prevalence for all forms of LGMD between 1 in 14,500 and 1 in 123,000 and incidence of 1–6 cases per 100,000 population. LGMDs demonstrate both autosomal dominant and autosomal recessive inheritance patterns. The age of onset may range from early childhood to adulthood, and disease progression varies from rapidly progressive forms leading to early disability to milder phenotypes with slow progression over decades. Histopathological evaluation often reveals dystrophic changes with muscle fibre size variation, necrosis, regeneration, and endomysial fibrosis. However, definitive diagnosis relies on molecular genetic testing, which is essential for accurate subtype classification, prognostication, and genetic counselling. Management remains largely supportive and rehabilitation to maintain functional independence and improve quality of life.

We present a case of limb-girdle muscular dystrophy to emphasize its clinical spectrum, diagnostic challenges, and the importance of early recognition and comprehensive management in improving patient outcomes.

Case Report:

A 7 year old female child presented with bilateral lower limb weakness for four years, which was insidious in onset and non progressive. She had difficulty in getting up from cross legged sitting posture since 1 month and increased difficulty in walking and pain in the right leg since 4 days. Birth history and developmental milestones were normal. There was no history of similar complaints in the family.

Neurological examination showed higher mental functions including speech, gait and cranial nerves were normal, muscle tone was normal with positive Gower's sign and hypertrophy of both calves. Power was 4/5 at both shoulders, 4/5 at both elbows, 5/5 at both wrists, 3/5 at both hip joints, 4/5 at both knees,

4/5 at both ankles. All deep tendon reflexes were normal with knee reflex of 1+ and ankle reflex of 2+, with bilateral extensor plantar responses. Sensory system was normal. There were no cerebellar signs, skull and spine was normal.

Investigations showed haemogram, serum electrolytes and metabolic workup and urine routine were normal. Serum Creatinine phosphokinase was 7091 IU/L (Reference: 24-190 IU/L). Cardiac screening showed normal 2D ECHO findings. Electromyography and nerve conduction studies were abnormal; suggestive of a primary muscle disease. Genetic testing was confirmatory and showed 2 variants; Limb girdle muscular dystrophy-6 and Limb girdle muscular dystrophy-10.

Case Discussion:

Limb-girdle muscular dystrophy (LGMD) comprises a genetically and clinically heterogeneous group of inherited muscular dystrophies characterized predominantly by progressive weakness involving the pelvic and shoulder girdle musculature. The estimated prevalence ranges from 1.6–4.7 per 100,000 population; however, the actual burden is likely underestimated because of phenotypic overlap and limited access to molecular diagnostic facilities, particularly in resource-limited settings [1]. Recent advances in next-generation sequencing have significantly improved the understanding and classification of LGMD, enabling identification of overlapping and atypical phenotypes.

The present case highlights a childhood-onset LGMD presenting with progressive proximal muscle weakness, difficulty in climbing stairs, waddling gait, positive Gower's sign, calf hypertrophy, and markedly elevated serum creatine phosphokinase levels. These findings are consistent with a primary dystrophic myopathy. The chronic progressive course, preserved sensory examination, absence of inflammatory manifestations, and myopathic pattern on electromyography further supported the diagnosis. Early onset of symptoms in pediatric patients is often associated with a relatively severe disease phenotype and greater functional impairment during adolescence [2].

Genetic analysis revealed variants associated with LGMD type 6 and type 10, suggesting either compound heterozygosity or overlapping genotypic expression. Such findings underscore the increasing complexity of genotype–phenotype correlations in LGMD. Titin-related muscular dystrophies, particularly LGMD type 10, exhibit considerable clinical variability ranging from mild proximal weakness to severe childhood-onset muscular dystrophy with respiratory or cardiac involvement [3]. The coexistence of variants involving different LGMD-associated genes in the present case may explain the early presentation and distinctive clinical phenotype.

Comparison with previously reported pediatric cases demonstrates both similarities and important differences. Alghamdi et al. described a 6-year-old boy with LGMD type 10 due to a homozygous TTN mutation who presented with progressive lower-

limb weakness, scapular winging, calf muscle atrophy, and distal involvement [4]. In contrast, our patient demonstrated predominant proximal muscle weakness with calf hypertrophy and without distal weakness or scapular winging. Similarly, childhood titinopathy cases reported by Baban et al. showed associated cardiac abnormalities and joint contractures in addition to proximal weakness [5]. Notably, our patient did not exhibit contractures, respiratory compromise, or cardiac involvement at presentation, suggesting a comparatively milder systemic phenotype despite early onset.

The differential diagnosis included Duchenne muscular dystrophy (DMD), Becker muscular dystrophy, polymyositis, and spinal muscular atrophy. DMD and Becker muscular dystrophy were considered less likely because of the female sex, absence of an X-linked inheritance pattern, and lack of dystrophin gene mutations. Polymyositis was excluded due to the absence of inflammatory signs, systemic manifestations, and the chronic insidious progression of symptoms. Spinal muscular atrophy was ruled out because of preserved deep tendon reflexes, absence of fasciculations, and normal sensory examination. Similar diagnostic challenges have been described in pediatric LGMD cohorts, where phenotypic overlap frequently delays definitive diagnosis until molecular testing is performed [6].

Currently, management of LGMD remains primarily supportive and multidisciplinary, focusing on physiotherapy, prevention of contractures, optimization of mobility, nutritional support, and

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periodic cardiac and respiratory surveillance. Early rehabilitation measures are essential to preserve functional status and improve

quality of life. Genetic counseling is equally important for family screening, prognostication, and future reproductive planning.

This case emphasizes the importance of considering LGMD in children presenting with progressive proximal muscle weakness and elevated creatine phosphokinase levels. It also highlights the utility of comprehensive genetic evaluation in identifying rare and overlapping LGMD subtypes, thereby facilitating accurate diagnosis, prognostication, and long-term multidisciplinary care.

Conclusion:

Limb-girdle muscular dystrophy should be considered in children presenting with progressive proximal muscle weakness, positive Gower's sign, calf hypertrophy, and elevated serum creatine phosphokinase levels. Early recognition remains challenging because of significant phenotypic overlap with other neuromuscular disorders. This case highlights the importance of detailed clinical evaluation combined with electrophysiological and molecular genetic testing in establishing an accurate diagnosis. Identification of overlapping LGMD-associated genetic variants further emphasizes the expanding complexity of genotype–phenotype correlations in paediatric muscular dystrophies. Early diagnosis facilitates timely rehabilitation, multidisciplinary surveillance, genetic counselling, and improved long-term functional outcomes.

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RESULTS

LIKELY PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED

SNV(s)/INDELS

Gene# (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification$
***SGCD* (+)** (ENST00000337851.9) NM_000337.6	Intron 6	c.502+1G>A **(5' Splice site)**	Homozygous	Limb-girdle muscular dystrophy 6 (OMIM#601287)	Autosomal recessive	Likely Pathogenic **(PM2_M, PVS1_VS)**
***TTN* (-)** (ENST00000589042.5) NM_001267550.2	Exon 47	c.11301G>A **(p.Met3767Ile)**	Likely compound Heterozygous	Limb-girdle muscular dystrophy 10 (OMIM#608807)	Autosomal recessive	Uncertain Significance **(PM2_Sup)**
	Exon 83	c.24095A>G **(p.Gln8032Arg)**				Uncertain Significance **(PM2_Sup)**

Abbreviation : **_VS** - Very Strong, **_S** - Strong, **_M** - Moderate, **_Sup** - Supporting





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INTERPRETATION:

Motor nerve conduction:

Right peroneal - EDB: Normal CMAP amplitude & velocity
Left peroneal - EDB: Normal CMAP amplitude & velocity
Right Tibial -AH : Normal CMAP amplitude & velocity
Left Tibial -AH : Normal CMAP amplitude & velocity

Sensory nerve conduction:

Right Sural : Normal SNAP amplitude & velocity
Left Sural : Normal SNAP amplitude & velocity
Right sup.peroneal : Normal SNAP amplitude & velocity
Left sup. peroneal : Normal SNAP amplitude & velocity

F wave study:

Right Tibial F wave: Normal
Left Tibial F wave: Normal

H reflex:

Right Tibial H reflex: No response
Left Tibial H reflex: No response

EMG: Electromyography study

Muscle	Spontaneous activity	MUP morphology	Recruitment pattern
Tibialis Anterior - Right	Nil	Small short polyphasic	Early & Full
Vastus Lateralis - Right	Nil	Small short polyphasic	Early & Full
Biceps Brachii - Right	Nil	Small short polyphasic	Early & Full

COMMENT: Nerve conduction study & Electromyography is abnormal suggestive of primary muscle disease. Kindly correlate clinically.

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