

Head Drop as an Atypical Presentation of Suspected Limb-Girdle Muscular Dystrophy: A Case Report

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INTRODUCTION

Limb-Girdle Muscular Dystrophy (LGMD) is a **highly heterogeneous group** of inherited disorders. Weakness of the proximal limbs, including shoulder or pelvic girdle musculature, is a well-known feature. However, clinical manifestations can vary depending on the subtype. We report a patient with suspected autosomal dominant LGMD type 3, who presented mainly with head drop, which has not been reported in this subtype to our knowledge.

CASE DESCRIPTION

18-year-old female

Chief complaints

: **progressive head drop**

- aggravated about 10 months prior to presentation

Past history

- intellectual disability
- both renal cysts
- operation history (+)

both bicanalicular lacrimal duct stent insertion due to both nasolacrimal duct obstruction (2008.05.23.)

Family history : none

Physical examination

- **cervical hyperflexion** : aggravated during ambulation
- **thoracic hyperkyphosis** and **lumbar hyperlordosis**
- **pes planus** on both sides.
- manual muscle test : not testable due to poor cooperation
 - no apparent motor weakness
- increased muscle tone in both hands and ankles
- self ambulation
 - : possible, but with **impaired forward gaze**
- muscle atrophy or hypertrophy : (-)
- myotonia : (-)

Whole genome sequencing (2025.11.18.)

: **variant of undetermined significance**

in **HNRNPDL** gene

: suspicious of

autosomal dominant Limb-Girdle Muscular Dystrophy type 3 (LGMD3)

Table 1. The results of whole genome sequencing

Gene	Variant	Classification
HNRNPDL	Genomic Position : 4-82428403-C-T (GRCh38)	VUS
	cDNA: NM_031372.4:C.487G>A	
	Protein: NP_112740.1:p.Asp163Asn	
	Zygosity: Heterozygous	
	Inheritance: Unknown	

Further evaluation

- **laboratory tests** (2026.03.19.) : non-specific
- **additional imaging studies** (2026.03.19.)
 - : fixed pes planus on both sides



Fig. 1. Foot X-ray showing pes planus on both sides. Meary's angle (right/left) was measured at 9.5/15.6 degrees, and calcaneal pitch angle (right/left) was measured at 12.1/14.6 degrees.

Symptomatic management

- **head drop**
 - Philadelphia orthosis
 - proper posture and strengthening exercises
- **pes planus**
 - University of California Biomechanics Laboratory orthosis



Fig. 2. Before: On presentation, cervical hyperflexion with thoracic hyperkyphosis and lumbar lordosis is prominent. **After:** About 6 months later, following Philadelphia orthosis apply and education on proper posture and strengthening exercises, the caregiver said the **forward gaze during ambulation got better than before.**

Known clinical features of LGMD3

- proximal musculature weakness
- limitations in fingers and toes flexion
- scapular winging
- **head drop : has not been well described in this subtype**

CONCLUSION

This report suggests that **careful clinical assessment combined with genetic evaluation** help identify **peculiar presentations** within the rarely reported spectrum of LGMD. Further research is needed about **clinical manifestations and course of LGMD3**. **Guidelines regarding management and rehabilitation strategies** need to be established in the future. Also, with this patient, serial follow-up and additional evaluation like electrodiagnostic studies and family studies should be considered for a more confirmative diagnosis.