Psychosocial and clinical profile of pediatric and adult patients with LGMD

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Introduction

Limb-girdle muscular dystrophies (LGMDs) are a heterogeneous group of genetically inherited disorders characterized by progressive proximal muscle weakness. The clinical spectrum of LGMDs is broad, ranging from severe infantile forms that lead to loss of ambulation early in life to milder adult forms with little disability (Georganopoulou et al., 2021; Rathore & Khang, 2023). Beyond its physical impairment, these diseases also generate consequences for emotional and social health, which, in turn, directly affects the health-related quality of life (HRQoL) experienced by these individuals (Angelini & Rodríguez, 2024; Kovalchick et al., 2022). However, to date there is a notable lack of studies analyzing the psychosocial and emotional profile of both pediatric and adult patients with LGMD.

Objective: To compare scores between a group of LGMD patients with a homogeneous control group in HRQoL, and in behavioral and emotional symptomatology.

Methods

Participants

Sociodemographic and clinical data

	LGMD group	Control group	Total
Participants	20	20	40
Adults	9	9	18
Children	11	11	22
Sex (Adults)	6 women / 3 men	6 women / 3 men	12 women / 6 men
Sex (Children)	5 girls / 6 boys	5 girls / 6 boys	10 girls / 12 boys
Mean Age (Adults)	39.10 years	38.30 years	-
Mean Age (Children)	12 years	12 years	
LGMD Subtypes (Adults)	Alpha: 6 / Beta: 1 / Gamma: 3	-	
LGMD Subtypes (Children)	Alpha: 6 / Gamma: 4 / GMPPB: 1	-	

Instruments

Behavioral symptomatology

SDQ (children)

Emotional symptomatology

HADS (adults)

Quality of life

SF-12 (adults)
Pedsql (children)

Procedure

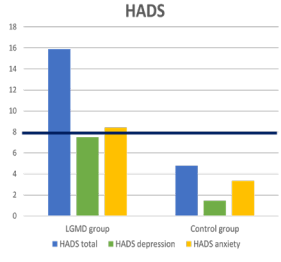
Period: November 2024 – April 2025

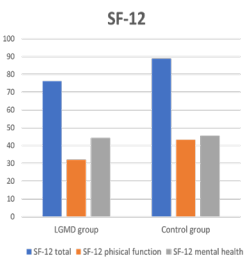
Via online (tele-neuropsychology):

- Test administration: 1 hour
- Questionnaire: 10 minutes

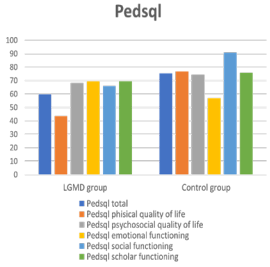
Results

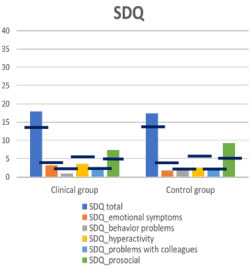
Adults





Children





Discussion

According to the results of pediatric patients with LGMD, their HRQoL, both in its physical and psychosocial dimension, was significantly decreased compared to healthy pairs. Specifically, impaired social functioning is consistent with greater social needs and more difficulties in prosocial behavior in children and adolescents with LGMD. These findings, while novel for this particular condition, reinforce the literature on social skills difficulties exhibited in other pediatric neuromuscular diseases (Darke et al., 2006; Gosar et al., 2021; García et al., 2024).

For adult patients with LGMD, their HRQoL was found to be significantly lower compared to the general population. This pattern was observed in the total HRQoL score, as well as in its physical and mental dimensions. These results are supported by previous research (Angelini & Rodríguez, 2024). In addition, the results regarding psychological symptomatology are consistent, as LGMD patients reported a higher number of depressive and anxiety symptoms compared to their healthy controls (O'Dowd et al., 2021; Peric et al., 2018).

The results in both cohort groups highlight that attention to psychological needs is necessary for LGMD patients, especially since it would provide valuable information to the natural study of these diseases (Georganopoulou et al., 2021).

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