Gamma-sarcoglycanopathy (LGMDR5): clinical and genetic study of a pediatric Tunisian cohort

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ICNC 2022
17th INTERNATIONAL CHILD NEUROLOGY CONGRESS
ANTALYA, TURKEY | OCTOBER 3-7, 2022

INTRODUCTION

•Limb-girdle muscular dystrophy type R5 (**LGMDR5**) is an autosomal-recessive disorder caused by mutations in **gamma-sarcoglycan** encoding gene (1,2)

•It is characterized by **childhood** onset of progressive muscular dystrophy (1)

•It is **frequent in North African** populations (1,2)

OBJECTIVES

•To determine the clinical and genetic characteristics of a Tunisian series of LGMDR5

MATERIALS AND METHODS

•Retrospective study from 2004 to 2022 collecting demographic, clinical, biological, electrophysiological, and genetic data of patients with genetically confirmed LGMDR5

RESULTS

•**24 patients**

•**Demographic and clinical features** (Table 1)

•**Explorations** (Table 2)

•**Treatment**: Steroids at the dose of 0.75mg/kg/d : 10d ON/10d OFF (83% of patients)

•Mean **follow-up** period: 3 years

•**Evolution** (figure 1)

Table 1: Demographic and clinical features

Demographic and clinical features		N° of patients/families
No. of patients		24
No. of families		18
Sex, male/female		11/13
Consanguinity		17 families
Familial history of similar case		8 families
Mean age of onset (years)		3.7
Mean age at last evaluation (years)		9,9
Inaugural situation	Difficulties to run and to climb stairs	18
	Gait disturbance	4
	High CK level	2
Examination	Proximal lower limb weakness	24
	Calf hypertrophy	20
	Cognitive impairment	3

Table 2: Result of explorations

Exploration	Result (% patients)
Serum creatine kinase levels	Levels ranged from 2280 to 18461 U/L. (NV<150) (100%)
Transaminase	High level of AST and ALT (100%)
Electromyography	Myogenic pattern (100%)
Muscle biopsy	Dystrophic changes (17%)
Genetic study	Homozygous c.521delT mutation in the *SGCG* gene (100%)

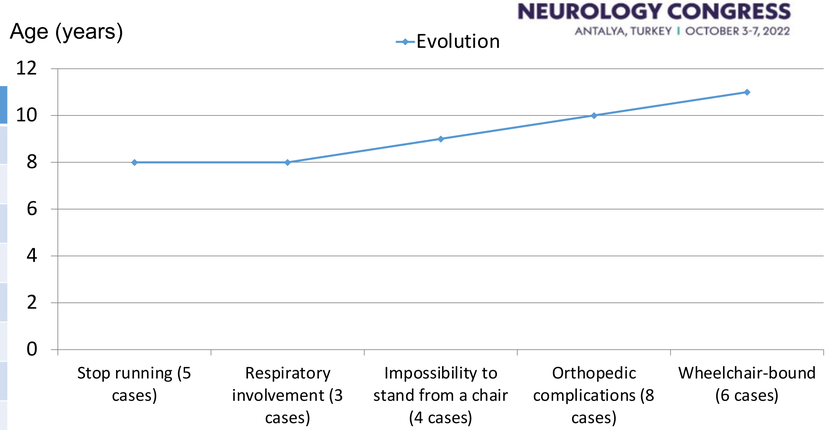

Figure 1: Evolution of our patients

DISCUSSION AND CONCLUSIONS

•LGMDR5 is a **Duchenne-like** autosomal recessive muscular dystrophy caused by mutations in the *SGCG* gene which encodes gamma-sarcoglycan (1,2)

•The mean age of onset is between **5 and 6 years** (1)

•**Calf hypertrophy** is common (1), seen in 83% of our patients

•Our study highlights the **recurrent founder mutation** in the Tunisian population

•Targeted mutational research in the LGMDR5 will enable early diagnosis and facilitate **genetic counseling**

REFERENCES

1-El Kerch F, Ratbi I, Sbiti A, et al. Carrier frequency of the c.525delT mutation in the SGCG gene and estimated prevalence of limb girdle muscular dystrophy type 2C among the Moroccan population. Genet Test Mol Biomarkers. 2014 Apr;18(4):253-6

2- Kefi M, Amouri R, Driss A, et al. Phenotype and sarcoglycan expression in Tunisian LGMD 2C patients sharing the same del521-T mutation. Neuromuscul Disord. 2003 Dec;13(10):779-87

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