Study of clinical and genetic characteristics of limb-girdle muscular dystrophy in Iranian patients

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ABSTRACT

Limb–girdle muscular dystrophy (LGMDs) is a bothersome muscle disease associated with weakness of the shoulder and pelvic girdle.

Objectives

The study is aimed to determine the genetic diversity and relative frequency of 6 various forms of LGMD in Iranian children.

Materials and Methods

In this descriptive research, 60 children referred to the neurology or emergency department of the Pediatric Medical Center during April 2019 to April 2020 were studied.

Additional tests (muscle biopsy and genetic testing) were performed in order to confirm the diagnosis of LGMDs.

Quantitative evaluations such as disease level, motor, respiratory, and cardiac functions and molecular analysis were performed using statistical analysis.

Results

Out of 60 children with muscular weakness and suspected of having limb-girdle muscular dystrophy, a total of 41 patients with a mean age of 11.1 were studied. 22 patients were diagnosed with genetic tests and 19 patients with muscle biopsy. 26.8% had alpha sarcoglycanopathy, 24.4% had beta sarcoglycanopathy, 17.1% had gamma sarcoglycanopathy, 7.3% had calpainopathy, 7.3% had dysferlinopathy, 7.3% had dystroglycanopathy, 7.3% had titinopathy and one patient had laminopathy.

Among genetically proven individuals, 27.3% had SGCB mutation and 18.2% had SGCA mutation. Conclusions: The prevalence of alpha and beta sarcoglycanopathy phenotypes in the study population shows that the severity of clinical involvement may be predicted by SGCB gene mutation and sarcoglycan expression.

Conclusion

The prevalence of alpha and beta sarcoglycanopathy phenotypes in the study population shows that the severity of clinical involvement may be predicted by evaluating various mutations in the SGCB gene and the mentioned clinical symptoms.

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