Fabry's disease with minimal manifestations in girls

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Objective. Fabry disease (Anderson-Fabry disease) is a rare inherited lysosomal multisystem disease with X-linked inheritance due to the deficiency of lysosomal enzyme α-galactosidase A and leads to accumulation of sphingolipids (globoside, globotriaosylceramide) in the walls of blood vessels in all organs.

Methods. Clinical cases of low-symptomatic Fabry's disease in 2 girls (9 and 10 y.o.) with positive family history of the underlying disease are described.

Results. Girl 9 y.o. is granddaughter and girl 10 y.o. is daughter of a male patient with Fabry's disease. Girls had complaints of reduced sweating and febrile acroparastesia only.

Physical and sexual development according to age, the skin was clean.

Echocardiography, abdominal USE, pulmonary function test, cornea and fundus, ear, nose and throat examination were normal. Urine examination showed 1+ proteinuria both girls have. Glomerular filtration rates were normal. The lyso-Gb3 and galactosidase A levels analyzed in dried blood spots (DBS) by tandem mass spectrometry and molecular genetic analysis was performed. An increase lyso-Gb3 concentration and a change in the nucleotide sequence c.983G > C, leading to the replacement of p.G328A, described in the international database on *HGMD* mutations (CM 930337) in the hemizygous state has been identified.

Conclusion. These cases confirm that it is very important that the pediatricians become aware of the importance of genealogical anamnesis findings and clinical features of Fabry's disease, so they can participate in the identification of unrecognized patients.