
BACKGROUND: Fabry disease is an X-linked genetic disorder involving sphingolipid catabolism, which is caused by lysosomal alpha-galactosidase A deficiency. Ophthalmological findings such as corneal and lens opacities and conjunctival and retinal vessel abnormalities can be the only and/or the first recognizable symptoms, especially in heterozygous females.

METHODS: We report on a 34-year-old German woman with cornea verticillata. The alpha-galactosidase A activity was determined in leukocytes using a fluorescence substrate, and the sequence analysis of the alpha galactosidase A gene was performed with genomic DNA.

RESULTS: The alpha-galactosidase A activity in leukocytes was significantly low (0.24 nmol/min/mg protein; normal range, 0.4-1.2), which is compatible with a heterozygote for Fabry disease. The following sequence analysis revealed a heterozygous transition in position IVS5 + 2 T> C. Transition of thymine (T) to cytosine (C) affects the donor splice motive of exon 5 and most probably leads to an aberrant splicing procedure of the alpha-galactosidase A gene.

CONCLUSION: Our case emphasizes the importance of ophthalmological findings in Fabry disease. The subsequent biochemical and molecular analysis provides a secure diagnosis of female carriers of Fabry disease.