



FABRY DISEASE, FAMILY CASE REPORT

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BACKGROUND

Fabry disease (FD) is a rare hereditary metabolic disorder linked to the X chromosome, caused by the partial or total deficiency of alpha-galactosidase A, related to mutations in the GLA gene (Xq21.3-q22). Its deficient activity may lead to the progressive accumulation of globotriaosylceramide and other glycosphingolipids in the lysosomes of various cell types throughout the body, causing severe complications of potentially fatal target organs.

Clinical manifestations of childhood or adolescence in the classical form include acroparasthesia, hypohidrosis, corneal opacities, stroke, cardiac anomalies, and renal disorders. In the variant form, the symptoms are usually mild and limited to heart or kidney. For this reason, patients often seek medical attention after the disease progresses. The diagnosis can be confirmed by the dosage of the alpha-galactosidase enzyme and genetic analysis. Enzyme replacement therapy is the basis of treatment.

CASE REPORT

Familial cases of Fabry's disease, involving of 4 relatives. The first patient is a male, 24 years old, has started during childhood episodes of recurrent abdominal pain, pain in the hands and feet (acroparesthesia) and reduced sweating during exercise. Physical examination showed angiokeratomas in upper body and presence of cornea verticillata. Laboratory tests: normal renal function and presence of microalbuminuria. Alpha-galactosidase enzyme dosage 0.26 mmol / mg protein / hour (normal > 1.4) and molecular analysis with homozygous mutation c.823delC (exon 6 in the alpha-galactosidase A gene), corroborating the diagnosis of FD at 14 years. After genetic and enzymatic research, her mother and sisters (from different parents) also received DF diagnosis. Mother had progression to chronic renal dialysis and cerebrovascular accident, while the sisters presented a clinic similar to the patient, abdominal pain, headache and acroparesthesia. All family members are receiving biweekly enzyme replacement, with control of the clinical condition

CONCLUSION

The triad composed of renal changes, angiokeratomas and limb pain in children or young males should always raise the suspicion of Fabry's disease, as in the cited patient. Because it's an X-linked pathology, most women are only carriers of the gene. In rare cases, as in the family case presented, female patients may be symptomatic. Often, there is a delay in diagnosis. Early enzyme replacement therapy may limit the severity of the manifestations, with improvement of clinical findings.