GENETICS AND GENODERMATOSES

FABRY DISEASE: CASE REPORT

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Background: Fabry disease is a X-linked, hereditary, lysosomal storage disease caused by alpha-galactosidase A (α-Gal A) enzyme deficiency, which results in the accumulation of neutral glycosphingolipid globotriaosylceramide (Gb3) on the vascular endothelium and visceral tissues, such as skin, heart, kidneys and central nervous system.

Observation: 37-year-old male patient presents small erythematous lesions throughout the body that grow, rupture, bleed and reappear continuously since age of 13. Over the years he began to experience seizure episodes associated with fever and joint pain (knees and elbows). Family history revealed a sibiling with Fabry disease. Exams showed α-Gal A enzyme deficiency, confirming the diagnosis. Clinical examination revealed multiples abdomen, scrotal and penis angiokeratomas. Complementary investigation was normal, except for the audiometry that showed sensorineural hearing loss in a notch in the left ear and 510mg of proteinuria of 24 hours. Patient is currently waiting the start of enzyme resorption therapy.

Key message: The diagnosis was based on the dosage of enzymatic activity, subsequently confirmed by the mutation in the α-GAL gene. It is clinically characterized by chronic pain and acroparesthesia, gastrointestinal disturbances, angiokeratoma, progressive renal impairment, cardiomyopathy, and stroke. Renal involvement occurs due to Gb3 accumulation in renal cells and may present glomerular and tubular function disorders. Proteinuria is a commom feature, caused by the affected Podocytes. Angiokeratomas are present as initial manifestation. Otological problems should also be considered. Sensorineural hearing loss is common and patients should undergo careful evaluation. Additionally, neurological manifestations occur, such as Parkinson's, dementia and epilepsy, as they are caused by cerebrovascular disease and accumulation of glycolipids in neurons.