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A NOVEL MUTATION OF THE ALPHA GALACTOSIDASE GENE LINKED WITH FABRY DISEASE AND THERAPY-RESISTANT DEPRESSION

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Introduction: Fabry disease (FD) is an X- chromosome linked recessive glycolipid storage disease caused by deficient activity of the lysosomal enzyme Alpha Galactosidase A, resulting in progressive accumulation of aberrant catabolites like Globotriaosylsphingosin (lyso-Gb3) in vulnerable cells. The disorder can affect multiple organ systems including the central nervous system. A high proportion of patients with FD are at increased risk of developing neuropsychiatric symptoms.
Materials and methods: A 56 year old female inpatient suffering from treatment resistant recurring depression with psychotic symptoms for a period of over two decades was diagnosed with asymptomatic hypertrophic cardiomyopathy in a routine health checkup. The incidental finding lead to an elaborate diagnostic work-up including genetic testing for FD as a possible cause.
Result: A novel, hitherto undescribed mutation in the Alpha Galactosidase Gene (c.1025..1027delGAC) affecting enzyme activity was detected. In addition, lyso-GB3 concentration was increased above the normal level, in concordance with the diagnosis of FD.
Conclusion: In this patient, undiagnosed FD associated with a novel mutation of the Alpha Galctosidase Gene may have caused predominantly psychiatric symptoms and contributed to therapy resistance.

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