

HOW TO TREAT A PATIENT WITH FABRY DISEASE?

NEUROLOGICAL IMPLICATIONS

Professor Vida Demarin, FAAN, FAHA, FESO

Fellow of the Croatian Academy of Sciences and Arts, Head of the University Department of Neurology, Reference Centre for Headache, Reference Centre for Neurovascular Disorders Croatian Ministry of Health and Social Welfare, University Hospital Centre Sestre milosrdnice, Vinogradska 29, Zagreb

Fabry disease is an X-linked lysosomal disorder that leads to excessive deposition of neutral glycosphingolipids in the vascular endothelium of several organs and in epithelial and smooth muscle cells. Disease is characterized by the accumulation of the glycosphingolipid substrate, ceramide trihexoside and ceramide dihexoside in tissues. Progressive endothelial accumulation of glycosphingolipids accounts for the associated clinical abnormalities of skin, eye, kidney, heart, brain, and peripheral nervous system.

When young patients present with signs and symptoms of a stroke, along with a history of skin lesions, renal insufficiency or failure, and heart attacks, Fabry disease is a consideration. Clinical manifestations of Fabry disease comprise chronic pain, kidney impairment, skin lesions, ocular opacities, vascular deterioration, stroke and cardiac deficiencies leading to premature mortality.

Fabry disease is uncommon, although research suggests that Fabry mutations may be more frequent in cryptogenic stroke patients. Aggressive efforts to diagnose the etiology of stroke are necessary to plan secondary prevention strategies. Traditional secondary stroke prevention strategies are still necessary.

Treatment strategies involve combined efforts from multiple specialties. The diagnosis and care of these patients usually is best handled at tertiary care centers. Enzyme replacement therapy has recently become accessible. Agalsidase is recombinant form of the human enzyme α -Gal A, which is deficient in patients with Fabry disease. Data from clinical trials show a decrease in globotriaosylceramide levels following enzyme replacement, reversal in lipid tissue storage, stabilized or improved renal and cardiac function, and reduction or relief of neuropathic pain.