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A Review on Fabry Disease

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Abstract: *Fabry disease is the most common of the lysosomal storage disorders and results from deficient activity of the enzyme alpha-galactosidase A (α-Gal A), leading to progressive lysosomal deposition of globotriaosylceramide and its derivatives in cells throughout the body. The classic form, occurring in males with less than 1% α-Gal A enzyme activity, usually has its onset in childhood or adolescence with periodic crises of severe pain in the extremities (acroparesthesia), the appearance of vascular cutaneous lesions hyperhidrosis), characteristic corneal and lenticular opacities, and proteinuria. Gradual deterioration of renal function to end-stage kidney disease (ESKD) usually occurs in men in the third to fifth decade. In middle age, most males successfully treated for ESKD develop cardiac and/or cerebrovascular disease, a major cause of morbidity and mortality. Heterozygous females typically have milder symptoms at a later age of onset than males. Rarely, females may be relatively asymptomatic throughout a normal life span or may have symptoms as severe as those observed in males with the classic phenotype. In contrast, late-onset forms occur in males with greater than 1% . Alpha Gal A activity. Clinical manifestations include cardiac disease, which usually presents in the sixth to eighth decade with left ventricular hypertrophy, cardiomyopathy, arrhythmia, and proteinuria; kidney failure, associated with ESKD but without the skin lesions or pain; or cerebrovascular disease presenting as stroke or transient ischemic attack.*

Keywords: Fabry disease.

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