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Fabry Disease: Linking Inflammation, Multiorgan Pathophysiology and Emerging Therapeutic Strategies

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ABSTRACT

Fabry Disease (FD) is a rare X-linked disease caused by mutations in the GLA gene that result in decreased α-galactosidase A activity, as well as progressively increasing concentrations of Globotriaosylceramide (Gb3) and Globotriaosylsphingosine (lyso-Gb3) in various cells of the body including blood vessels, heart muscles, and kidney cells. Normally, there are two types of FD: classical and late-onset, and some females may have other manifestations due to the X-inactivation of chromosome. Other than accumulation of the substrate, the propagation of FD is due to a permanent activation of the innate immunity as mediated by the TLR-4/NF-κB signaling pathway responsible for the secretion of pro-inflammatory cytokines and contributing to hypertrophy, myocardial, and renal fibrosis, regardless of the substrate. Instead of classical mechanisms of cell dysfunctions and tissue remodeling, the inflammatory component provides an explanation for multi-systemic pathology in FD. The diagnosis of FD necessitates assessing enzyme activity, confirming the genotype, and obtaining a professional opinion, which is much more challenging when it comes to women and those individuals with variant genotypes. Most important approaches to disease-modifying treatment of FD include Enzyme replacement therapy (ERT) and Oral Cheprone therapy (OCT). Nevertheless, the effectiveness of such methods is in dispute. Substrate reduction therapy (SRT) as well as gSRT (genetic substrate reduction via RNA interference) and gene replacement therapy utilizing viral vectors are very promising, yet exist only on paper so far. The goal of this review is to compile information regarding the mechanisms of pathophysiology of FD with a special focus on the underlying role of the inflammation in FD and provide the assessment of the recent developments in the therapeutic interventions from the use of enzymes to the usage of genes.

Keywords: Fabry disease(FD), Lysosomal storage disorder(LSD), Alpha-galactosidase A deficiency(alpha-Gal A), Globotriaosylceramide(Gb3), Enzyme replacement therapy(ERT), Gene therapy.

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