Fabry disease and new advances in treatment

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Abstract
Fabry disease is an X-Linked lysosomal storage disease, caused by deficient activity of lysosomal enzyme α-galactosidase A. The accumulation of globotriaosylceramide (GL-3) in the lysosomes causes lysosomal and cellular dysfunction and this in turn triggers the cascade of cells and tissue ischemia and fibrosis. The classic phenotype of Fabry disease is seen in most males with no enzyme activity and it affects multiple organ-systems. The early clinical manifestation of the disease occur in childhood with episodes of severe pain in the extremities (acroparesthesia), hypohidrosis, corneal and lenticular changes, and skin lesions (angiokeratoma). The renal failure, cardiovascular disease and stroke are the major causes of morbidity and mortality occurring later in life. Due to random chromosome X inactivation (Lyonization), the carrier females of Fabry disease may experience Fabry disease-related symptoms including acroparesthesia, gastrointestinal complains, renal and cardiac disease and/or strokes. In this article, after brief review of clinical presentations and diagnostic tests for the disease, we review the present therapeutic approaches and future directions in management of patients with Fabry disease.

Key words: Fabry disease; Lysosomal storage disease, Treatment, Enzyme replacement therapy

Introduction
Fabry disease (FD) is an X-Linked lysosomal storage disease, caused by deficient activity of lysosomal enzyme α-galactosidase A. As the result of α-galactosidase A deficiency glycosphingolipids, predominantly globotriaosylceramide (GL-3) and galabiosylceramide, accumulate in the lysosomes of various cells, such as in the vascular endothelium of multiple organs (1). The accumulation of GL-3 in the lysosomes causes lysosomal and cellular dysfunction and this in turn, triggers the cascade of cellular and tissue ischemia and fibrosis. The estimated prevalence of Fabry disease is about one in every 117000 live born males.

The classic phenotype of Fabry disease is seen