# Enzyme Replacement Therapy in Fabry Disease

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Fabry disease is an X-linked rare metabolic disease, caused by a deficient activity of the hydrolase α-Galactosidase A, and characterized by a progressive and systematic deposition of glycosphingolipids in many organs.

The disease is most severe in affected males. In the classic form (where the enzyme activity is absent) the clinical findings are represented by pain and paresthesias in the extremities, vessel ectasia (called angiokeratoma) in skin and mucous membranes, and hypohidrosis (a reduced sweating) during childhood or adolescence. Corneal and lenticular opacities may be present. Proteinuria, renal impairment,cardiac and neurological lesions develop with time, together with hypertension. When end stage renal disease occurs, dialysis or renal transplantation may be necessary. In heterozygous females a residual enzymatic activity may be demonstrated and they usually have asymptomatic or later onset disease manifestations, although rarely they could develop a disease as severe as in males.

A cardiac and a renal variant, where the heart and kidney are the only organs involved by the disease have been described too.

The recombinant human α-galactosidase A is now available for patients. Infusions of the enzyme replacement treatment have been demonstrated to be safe and effective. This study wants to evaluate the long term efficacy of enzyme replacement therapy in patients with Fabry disease and renal involvement.

Clinical period evaluations together with a genetic counselling will be offered to each patient.