Fabry Disease

National Cancer Institute

Source

National Cancer Institute. Fabry Disease. NCI Thesaurus. Code C84701.

A rare X-linked inherited lysosomal storage disorder characterized by deficiency of the enzyme alpha-galactosidase A. It results in the accumulation of glycolipids in the blood vessels and tissues. Signs and symptoms include hypertension, cardiomyopathy, angiokeratomas, neuropathy, hypohidrosis, keratopathy, proteinuria, and renal failure.