Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

Fabry Disease

Fabry Disease belongs to a group of disorders known as lysosomal storage disorders. Lysosomes are the 'recycling centres' of the cells. In this disorder there is a deficiency of the enzyme alpha-galactosidase A which is needed to break down a fatty substance called globotriaosylceramide. The enzyme deficiency results in the build-up of this substance in the cells causing damage to various organs, blood vessels and the nervous system. Although there is a wide spectrum of symptoms associated with Fabry Disease, each individual may be affected differently and may, or may not have all of the symptoms. The most commonly seen symptoms include episodic pain, most notable in the hands and feet, groups of small, dark red spots on the skin, clouding of the cornea, hearing problems, the inability to perspire properly, and gastrointestinal signs. There is a risk of complications such as progressive kidney damage, heart attack, and stroke. A milder form appears later in life.

Synonyms

Alternative names for this condition are:

- Alpha-Galactosidase A Deficiency
- Anderson-Fabry Disease
- Angiokeratoma Corporis Diffusum
- Angiokeratoma Diffuse
- Ceramide Trihexosidase Deficiency
- GLA Deficiency

Further information about this condition is available from Climb.

Disclaimer

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