Fabry Disease

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Fabry Disease Information Page.

Fabry disease (also called alpha-galactosidase-A deficiency) is caused by the lack of or faulty enzyme needed to metabolize lipids, fat-like substances that include oils, waxes, and fatty acids. The mutated gene allows lipids to build up to harmful levels in the autonomic nervous system (which controls involuntary functions such as breathing and digestion), cardiovascular system, eyes, and kidneys. Symptoms usually begin during childhood or adolescence and may include:

- burning sensations in the arms and legs that gets worse with exercise and hot weather,
- small, non-cancerous, raised reddish-purple blemishes on the skin,
- clouding in the corneas,
- impaired blood circulation and increased risk of heart attack or stroke,
- enlarged heart,
- kidneys may become progressively impaired, leading to renal failure, and
- decreased sweating, fever, and gastrointestinal difficulties.

Fabry disease is the only X-linked lipid storage disease (where the mother carries the affected gene on the X chromosome that determines the child's gender and passes it to her son). Boys have a 50 percent chance of inheriting the disorder and her daughters have a 50 percent chance of being a carrier. A milder form is common in females, and occasionally some affected females may have severe symptoms similar to males with the disorder.