WHAT IS FABRY DISEASE?

Approximately 1 out of every 40,000 men have classic Fabry disease.

Fabry disease is a rare genetic condition in which the body doesn't produce enough of an enzyme called alpha-galactosidase, resulting in a buildup of fat in the cells of the blood vessels and tissues of the kidneys, heart, skin and brain. Over time, this can lead to life-threatening problems, including kidney failure, heart attack or stroke. This disease is a type of lysosomal storage disorder.

01 | Causes and Types

Fabry disease is caused by a mutation in the GLA gene, located on the X chromosome. Due to the way the disease is inherited, men tend to develop more severe symptoms while women inherit a milder form.

The types of Fabry disease reflect the age at which symptoms first appear:

- **Classic type**: symptoms appear during childhood or the teenage years and may be noticeable as early as age 2. Symptoms will get progressively worse over time.
- **Late-onset/atypical type**: symptoms are usually milder and do not appear until a person is in their 30s or older.

02 | Symptoms

Symptoms of Fabry disease can vary based on age, severity of the condition, or which areas of the body are affected. General symptoms include:

- Episodes of pain in the hands or feet
- Cluster of rashes on the skin
- Reduced ability to sweat
- Cloudiness in the front part of the eye
- Ringing in the ears
- Hearing loss
- Problems with the gastrointestinal system
- Swelling in the legs, ankles or feet
- Kidney problems
- Heart problems

03 | Treatment

There is no cure for Fabry disease, although disease progression can be stopped if treated early enough. Enzyme replacement therapy is generally the first form of treatment. This will help slow down the buildup of fatty substances to prevent heart problems, kidney disease and other potential complications.

For more information on Fabry disease and supportive resources, please visit my.clevelandclinic.org.

Did You Know?

Late-onset or atypical Fabry disease is the more common form, affecting about 1 in every 1,500 to 4,000 men.

References

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