

Assay

Test

Lysosomal storage disorder

A disease resulting from the storage or accumulation of material in the lysosomes. Fabry disease is a lysosomal storage disorder.

Protein

The building blocks of cells involved in all essential life functions (for example: cell growth, energy production, metabolism).

Proteinuria

Excessive protein in the urine.

Stroke

A sudden loss of brain function caused by a blockage or rupture of a blood vessel to the brain.

Whorling

A star burst pattern commonly found on the cornea of people with Fabry disease.

X chromosome

The sex chromosome associated with female characteristics. All females typically have two X chromosomes while males typically have only one X chromosome.

X-inactivation

A phenomenon in which one of two X chromosomes in every cell of a female's body is inactivated. The process of X-inactivation is random, which helps account for variation in the symptoms of females who carry the Fabry disease gene on only one X chromosome.

X-linked disease

A genetic disease in which the altered gene is carried on the X chromosome. Fabry disease is an X-linked disease