Fabry disease is a multisystemic genetic disorder that ultimately results in irreversible, potentially life-threatening disease of the kidney, heart, and brain.

Substrate Accumulation and Resulting Organ Dysfunction

Fabry disease is characterized by the progressive and unrelenting cellular accumulation of a lipid substrate called globotriaosylceramide (or GL-3).

- Caused by deficiency of the lysosomal enzyme alpha-galactosidase A (or α-GAL), which usually metabolizes GL-3 and keeps it from accumulating.
- Without enough of this essential enzyme, GL-3 accumulates in the lysosomes of most cell types.

Pervasive accumulation of GL-3 eventually causes tissue ischemia and fibrosis.

Fabry is Progressive: Early Diagnosis and Intervention are Critical

Diagnosis is straightforward and can be accomplished by enzyme assay in a blood sample. A number of laboratories across the country offer this assay.

Know Fabry Disease

Affects Men, Women, and Children

- Fabry disease affects both males and females of all ethnicities and ages.
- Women, in particular, can experience significant organ damage in the absence of overt symptomatology.

Fabry is an inherited disorder marked by the progressive cellular accumulation of globotriaosylceramide (GL-3). GL-3 build-up leads to devastating consequences that can be irreversible.

X-linked Inheritance Means One Diagnosis Can Lead to Many

- Fathers with Fabry disease will pass it to all daughters but no sons.
- Mothers with Fabry disease have a 50/50 chance with each pregnancy of passing the gene to sons and daughters.
- Easy to determine who is at risk within a Fabry family, enabling earlier diagnosis of family members.

Fabry is Progressive: Early Diagnosis and Intervention are Critical

Diagnosis is straightforward and can be accomplished by enzyme assay in a blood sample. A number of laboratories across the country offer this assay.

Contact Genzyme Medical Information at 800-745-4447 for more information on diagnostic testing or for additional information on Fabry disease.

Although many organs can be affected, diagnosis of Fabry disease is of the utmost importance because early intervention can save lives.
Pathology at a Glance

GL-3 accumulates in tissues throughout the body, triggering a cascade of manifestations that begin with pain, gastrointestinal problems and quality of life issues, and lead to life-threatening complications involving the kidney, heart, and brain.