Could it be Fabry Disease?

Clusters of signs and symptoms could help distinguish Fabry disease—a progressive, potentially life-threatening disorder—from more common conditions.

Questions to Ask Your Patient

	YES	NO
1. Do you have chronic tingling or burning in your hands or feet?	☐	☐
2. Have you ever experienced episodes of extreme pain in your hands and/or feet of unknown cause, possibly accompanied by fever?	☐	☐
3. Do you have trouble sweating or exercising?	☐	☐
4. Do you find heat or cold hard to tolerate?	☐	☐
5. Do you frequently have gastrointestinal problems such as pain and bloating after eating, or nausea, cramps, or diarrhea?	☐	☐
6. Do you have small raised reddish-purple spots on your skin, especially in the "bathing trunk" area?	☐	☐
7. Do you have a family history of early cardiac or valvular disease, renal failure, or stroke?	☐	☐

Progressive Signs and Symptoms

Fabry disease is progressive and affects multiple organ systems. This chart indicates signs and symptoms that may appear at various stages of life.

Most males with the defective gene are subject to significant morbidity and mortality.[1] While females with the defective gene demonstrate a wide range of disease severity, most develop symptoms.[2,3]

Symptoms	Childhood	Adolescence	Adulthood
Episodic pain crises	●	●	●
Acroparesthesia	●	●	●
Hypohidrosis/anhidrosis	●	●	●
Corneal and lenticular opacities	●	●	●
Recurrent fever	●	●	●
Heat and cold intolerance	●	●	●
Psychosocial manifestations	●	●	●
Proteinuria		●	●
Gastrointestinal distress		●	●
Angiokeratomas		●	●
Fatigue		●	●
Renal insufficiency			●
Neurological complications			●
Cerebrovascular disease			●
Cardiac dysfunction			●
Hearing loss and tinnitus			●

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Take Action

What To Do If You Suspect Fabry Disease

If you suspect that a patient has Fabry disease, a geneticist can help establish a definitive diagnosis and provide information on intervention. For information on locating a geneticist near you, please contact **Genzyme Medical Information** at **617-768-9000** (option 2) or **800-745-4447** (option 2).

Diagnosing males:

- Alpha galactosidase enzyme assay is diagnostic.
- Males typically have <1% normal alpha-galactosidase in plasma and leukocytes.[1]

Diagnosing females:

- Enzyme assay alone is frequently insufficient for diagnosis.
- DNA-based diagnosis is required in females with normal to low-normal enzyme activity levels, and is advisable in all suspected patients.

Ocular assessment:

- Corneal whorling, visible through slit lamp ophthalmoscopy, is present in >90% of Fabry disease patients.[4]
- A slit lamp exam by an eye care professional may help establish the need for further testing.

Resources

www.FabryCommunity.com
www.FabryRegistry.com
www.GeneTests.org

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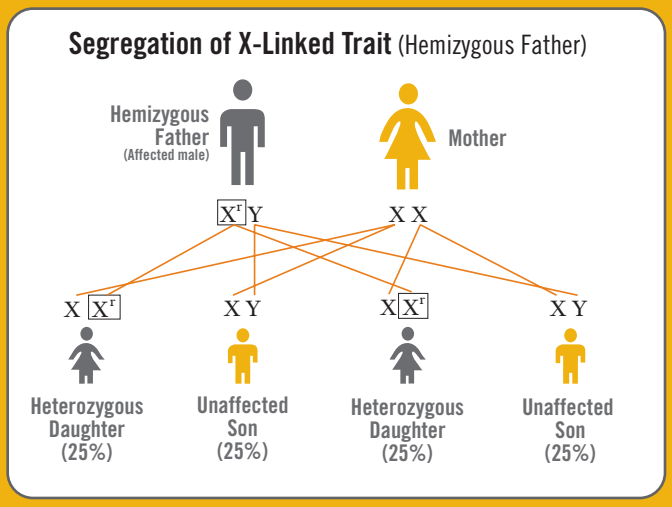
Genzyme Corporation
500 Kendall Street
Cambridge, MA 02142 U.S.A.
800-745-4447 or 617-768-9000
Fax 800-737-3642 or 800-591-7178
Monday - Friday 8:00 am - 6:00 pm EST

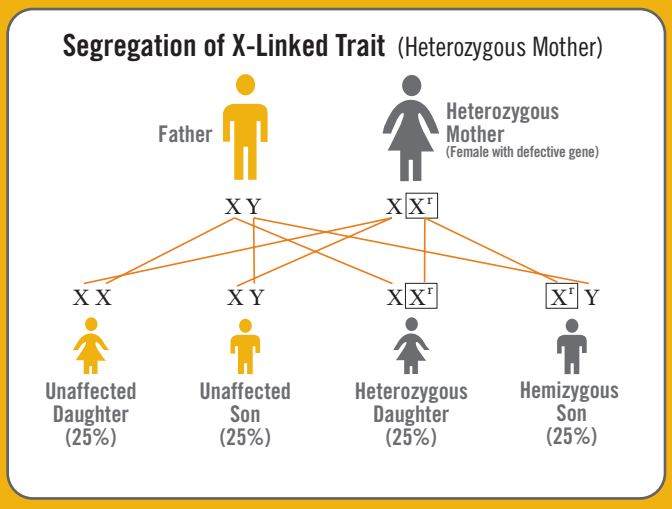
Fabry@genzyme.com

How Fabry Disease is Inherited

Fabry disease is an X-linked genetic disorder. Males with the defective gene pass it on to all of their daughters and none of their sons. Females with the defective gene have a 50% chance with each pregnancy of passing the gene to each of their offspring.

Because females have two X chromosomes in every somatic cell, symptom manifestation is variable, ranging from mild to severe, and potentially affecting some organ systems while sparing others.





1. Desnick RJ, Ioannou YA, Eng CM. α-Galactosidase A deficiency: Fabry disease. In: The Metabolic and Molecular Bases of Inherited Disease. New York: McGraw Hill, 2001;3733-74.
2. Wang RY, Lelis A, Mirocha J, Wilcox WR. Heterozygous Fabry women are not just carriers, but have a significant burden of disease and impaired quality of life. Genet Med 2007;9:34-45.
3. Wilcox WR, Oliveira JP, Hopkin RJ, et al. Females with Fabry disease frequently have major organ involvement: Lessons from the Fabry Registry. Mol Genet Metab 2007; doi:10.1016/j.ymgme. 2007.09.013.
4. Franceschetti A. Fabry disease: ocular manifestations. In: Bergsma D, Bron AJ, Cotlier E (eds). The Eye and Inborn Errors in Metabolism. Vol. 12, No. 3. New York: AR Liss Co., 1976;195-208.