Could it be **FABRY DISEASE?**

QUESTIONS TO ASK YOUR PATIENT

	YES	NO
1. Do you have frequent 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?	☐	☐

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

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.[1]

Most males with the disease-causing pathogenic variant are subject to significant morbidity and mortality.[2] While females with the pathogenic variant demonstrate a wide range of disease severity, most develop symptoms.[3,4]

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

TAKE ACTION

DIAGNOSING MALES:

- Alpha galactosidase enzyme assay is diagnostic.
- Males with classic disease typically have <1% normal alpha-galactosidase activity in plasma and leukocytes.[2]

DIAGNOSING FEMALES:

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

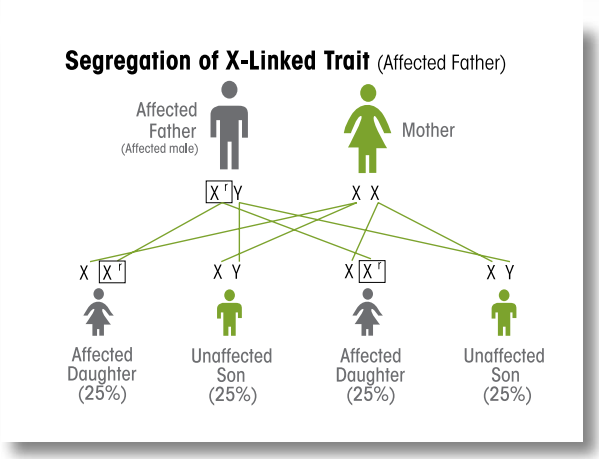
OCULAR ASSESSMENT:

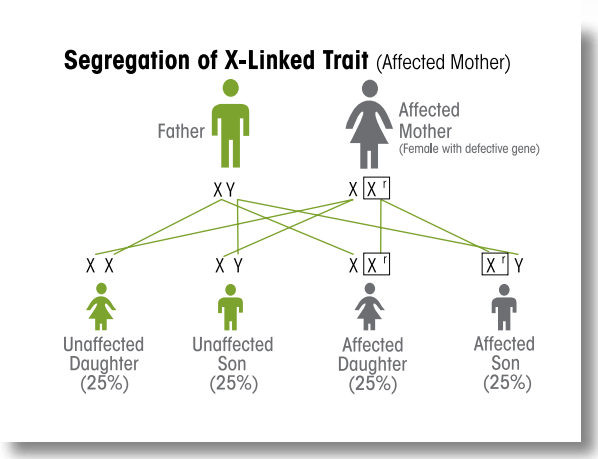
- Corneal whorling, visible through slit lamp ophthalmoscopy, is present in >90% of classic Fabry disease patients.[5]
- A slit lamp exam by an eye care professional may help establish the need for further testing.

HOW FABRY DISEASE IS INHERITED

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

Because females have two X chromosomes in every somatic cell, Fabry disease symptoms are more variable in females than they are in males. However, potentially life-threatening complications can develop, even in females whose initial presentation may suggest a more moderate disease course.





WHAT TO DO IF YOU SUSPECT FABRY DISEASE

If you suspect that a patient has Fabry disease, refer to a geneticist. A geneticist can help establish a definitive diagnosis and provide information on disease management.

SANOFI GENZYME RESOURCES

For Providers:
Sanofi Genzyme
Medical Information
1-800-745-4447, option 2

For Patients:
Connect with a Sanofi Genzyme Case Manager online at www.careconnectpss.com or call 1-800-745-4447, Option 3, Monday through Friday, 8:00 AM to 6:00 PM ET

www.discoverfabry.com

Notice: This document is intended to describe common clinical considerations, but may not be appropriate for every patient or case. Decisions surrounding patient care depend on the physician's professional judgment in consideration of all available information for the individual case.

1. Germain DP. Fabry disease. Orphanet journal of rare diseases. 2010 Dec;5(1):30. **2.** Desnick RJ, Ioannou YA, Eng CM. -Galactosidase A Deficiency: Fabry Disease. In: Valle D, Beaudet AL, Vogelstein B, Kinzler KW, Antonarakis SE, Ballabio A, Gibson K, Mitchell G. eds. . New York, NY: McGraw-Hill; 2014. http://ommbid.mhmedical.com/content.aspx?bookid=971&Sectionid=62644837. **3.** 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. **4.** 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. **5.** 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.

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