If you have a Fabry patient, you likely have a Fabry family

Help identify family members at risk of Fabry disease

ABOUT FABRY DISEASE

Fabry disease is an often life-threatening, panethnic, and heterogeneous inherited disorder caused by a lysosomal enzyme (alpha-galactosidase A) deficiency. The resulting progressive accumulation of globotriaosylceramide (GL-3) in the vasculature and other cell types or tissues eventually leads to major organ system damage including renal insufficiency, cardiac disease, and premature stroke.

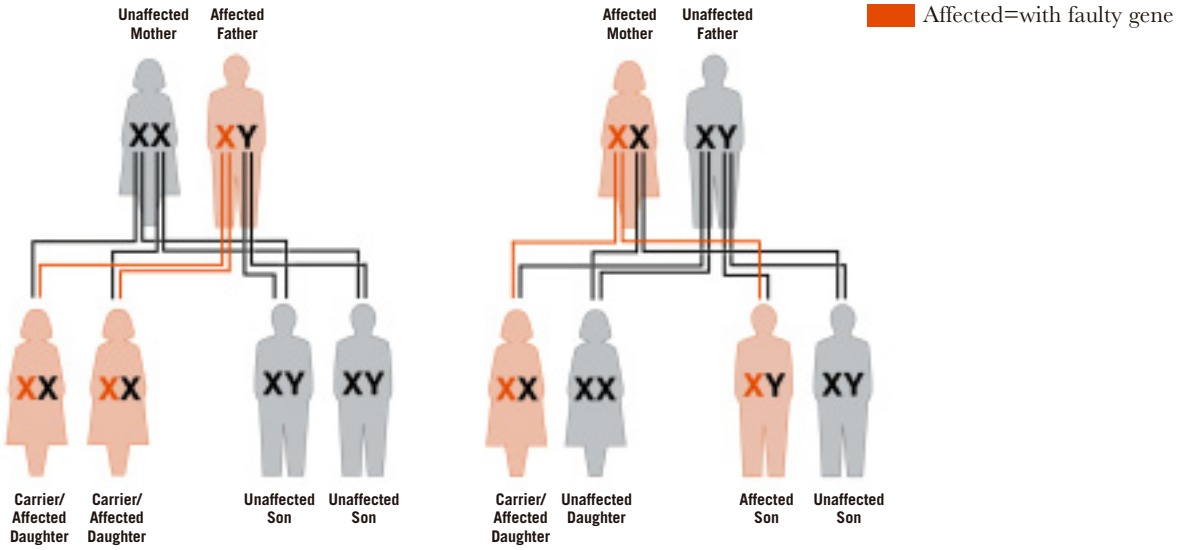
While Fabry disease is rare, it may be prevalent within a Fabry family. Early diagnosis and intervention may lead to improved outcomes.

THE FAMILY CONNECTION

Understanding the X-linked mode of inheritance of Fabry disease may help you identify a Fabry patient's family members who are at risk.

- Males with the disease pass the faulty gene on to all of their daughters but none of their sons.
- Females have a 50% chance with each pregnancy of passing the faulty gene to each of their sons and daughters.

Unlike many other X-linked disorders, females with the faulty gene can have varying degrees of disease manifestations.



Males with the faulty gene transmit it to all of their daughters and none of their sons.

Females with the faulty gene have a 50% chance of passing it during each pregnancy.

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Make the next Fabry diagnosis – your patient's family members may also be at risk

THE URGENCY IS REAL

Fabry disease is chronic and progressive, and presents at different stages and with different symptoms. Family members at risk may or may not be experiencing symptoms currently.

- Even in the absence of overt clinical signs, the disease may be progressing.
- Left undiagnosed and untreated, Fabry disease can cause significant organ damage.
- Early identification and intervention may lead to improved outcomes.

SPEAK WITH YOUR FABRY PATIENT ABOUT THE RISK

- Find out about your patient's family (i.e. children, siblings, parents, aunts, uncles and cousins).
- Understand that if your patient is male, all of his female children will have the faulty gene. His mother will usually also have the faulty gene.
- Recognize that if your patient is female, each of her children have a 50/50 chance of having Fabry disease. She inherited the disease from either her father or her mother.

CONFIRM THE DIAGNOSIS

Encourage your Fabry patient to speak with those family members at risk. You can play an important role in helping patients get the resources they need:

- Family members at risk should be tested, regardless of whether they are currently experiencing symptoms.
- Diagnosis can be confirmed with a blood test.
- Genetic counselors can provide education to patients and their families about Fabry disease, testing, and the risk to other family members.

EARLY IDENTIFICATION IS KEY

Early diagnosis is important, and can result in earlier intervention. When you have a Fabry patient, you are in the unique position to help identify the family members who may also be at risk of having this potentially life-threatening disease. Please don't delay.

ACT NOW

Information on testing: Call Genzyme at 800-745-4447 or 617-768-9000

Locate a genetic counselor: Contact National Society of Genetic Counselors at www.nsgc.org or 610-872-7608

Information on Fabry disease: Visit www.fabrycommunity.com

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