



Kidney Patients Seek Information about Fabry Disease

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What is Fabry Disease?

Fabry disease is an inherited, rare disorder that is caused when a person's GLA gene has changes or mutations that keep it from working correctly. This non-working GLA gene is unable to produce enough of an important enzyme called alpha-galactosidase A whose job it is to break down a fatty substance called globotriaosylceramide or GL3. The GL3 builds up in cells all over the body, in particular in the walls of blood vessels which leads to burning nerve pain in the hands and feet, lack of sweating, fatigue, diarrhea, and eventually damaged organs such as the kidneys, heart, and the brain.

How is it inherited?

The gene that causes Fabry disease is on the X chromosome. Males have one X and one Y chromosome and females have two X chromosomes. When a father has Fabry disease, his only X chromosome has the non-working GLA gene, so all of his female offsprings will inherit the non-working GLA gene and will have Fabry disease. Since Fabry disease only involves the X chromosome and the only way to have a son is for the father to pass on his Y chromosome, male offsprings of a man with Fabry disease will not inherit the non-working GLA gene and will not have Fabry disease. When a mother has Fabry disease, she has a 50% chance with each pregnancy of passing on the non-working GLA gene to either a son or a daughter,







leading to Fabry disease in that child. In this case both girls and boys would be affected.

Since we know how Fabry runs in families, it is easy to figure out who in the family is at risk for Fabry disease and to have genetic testing performed. Through studies, Fabry experts have found that when one family member is diagnosed with Fabry disease, then an average of five of their relatives will also have the disease. Since there is a FDA approved treatment for Fabry disease and we know the earlier treatment starts, the better it works, it can be helpful to know as early as possible. Some people may not know that testing is available or they may feel guilty, angry or scared of the potential test results, but in this case, knowledge is power. It can be useful to talk to a genetic counselor about testing options and decision about testing for genetic disease. You can learn about genetic counselors and find one near you at: www. aboutgeneticcounselors.com

In order to make it easier for the family members of someone with Fabry disease to get tested, Emory University and the American Association of Kidney Patients (AAKP) have partnered to create the Fabry Disease Family Testing and Education project. Through this project, Emory University offers FREE testing to people who have Fabry in their family and know the change in the GLA which causes the condition. As part of the program, people can also learn more about Fabry disease and available resources. To participate in the testing program people who would like to test for Fabry disease should talk to their doctor and then, the at-risk person can contact directly Emory and order a saliva collection and testing kit by mail. A kit can be ordered by calling the Emory Fabry Center at 404-778-8518 or by emailing Robin Vinson at robin.vinson@ emory.edu or Dawn Laney at dawn.laney@ emory.edu. In addition to providing the free testing, research from the project has found that family members who have corneal whorls (easily found by any eye doctor), pain in hands/feet, decreased sweating, or a purplish-pink rash in

the "bathing trunk area" called angiokeratomas are more likely to have Fabry disease than other family members.

Are Females Affected by Fabry Disease?

Yes, females do have Fabry disease. At one time, it was thought that they were "just" carriers of the disease but did not have any of the symptoms. But now it is known that they can experience symptoms, from mild to severe. Females may not have any or as many symptoms as males because they have two X chromosomes and the one that is working can still produce alpha-galactosidase A. It is important for women to be followed regularly by their doctor even if they have few symptoms since the disease continues throughout life and additional problems may develop.

Do children have symptoms of Fabry Disease?

Yes, children may have symptoms and being a rare disease, it may be difficult to diagnose if no one else in the family has symptoms of Fabry disease. Sometimes children have been told it is "growing pains" when they complain of pain in their hands and feet, cannot tolerate heat, or have GI problems, such as pain after eating, nausea, or diarrhea.

Boys with Fabry disease typically first have symptoms when they are six years old compared to girls who have their first symptoms around eight years old.





However, symptoms have been reported as early as age three years in both genders. Diagnosis of Fabry disease often occurs years after the symptoms have begun.

Children with Fabry disease may have symptoms of kidney problems. According to the Fabry Registry, approximately 15% of boys and girls under the age of 18 years of age have proteinuria defined as a urinary protein: urinary creatinine ratio ≥ 0.3 or urinary protein levels e0.3g/day. It has been documented that, although rare, children with Fabry disease can experience renal failure as young as 16 years of age. Given the importance of early treatment of Fabry disease, the early detection of this disease has been added to the newborn screening blood test in Missouri and Illinois. Other states are in the process of adding Fabry to their testing as well and some babies in New York City are tested depending on their hospital of birth.

What are ways that the kidneys are affected by Fabry Disease?

According to the Fabry Registry (an international database sponsored by Genzyme that collects information on Fabry disease patients), a number of people with Fabry disease experience kidney problems. Nineteen percent of the females in the Registry had chronic kidney disease, Stage 3 or higher and 2% of them reached End-Stage Renal Disease (ESRD). For males, 34% had chronic kidney disease, Stage 3 or higher and 14% of them reached End-Stage Renal Disease (ESRD).

In addition, heart problems were reported most frequently as a serious problem for males and females. And both males and females in the Registry reported having a lower quality of life after they were in their mid-thirties.

Where can I find more information?

Fabry Support and Information Group (FSIG): www. fabry.org National Fabry Disease Foundation (NKDF): www. thenfdf.org ThinkGenetic: www.thinkgenetic.com FabryCommunity(Genzyme):www.fabrycommunity. com Genzyme Case Managers: 1-800-7454447 (option3) Fabry Family and Education Testing Project: http:// genetics.emory.edu/patient-care/lysosomal-storagedisease-center/lab-testing.html National Organization for Rare Disorders (NORD): www.rarediseases.org

References

A Guide for Women Living with Fabry Disease 2014. Genzyme Corporation.

Emory University School of Medicine, http://genetics. emory.edu/patient-care/lysosomal-storage-diseasecenter/lab-testing.html

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