COVER STORY

American Association of Kidney Patients is continuing the Free **Fabry Family Testing and Education Project** with its partner Emory University!

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"Isn't that wonderful?!" You might be thinking, "But what is Fabry disease and why is the AAKP helping to look for it in families?"

Fabry disease is one of many causes of kidney disease that runs in families. In addition to kidney disease, people with Fabry disease can also have burning pain in their hands and feet (particularly during fevers), decreased ability to sweat, a reddish-purplish rash around the "bathing suit area", eye findings called corneal whorls that do not usually affect vision, extreme fatigue, belly pain, alternating diarrhea and constipation, heart problems, and increased risk for strokes. The health problems seen in Fabry disease can start in childhood or may start later in life. Fabry disease is seen in people from all racial and ethnic groups around the world.

In an effort to help this patient population, the

AAKP developed a research and education program in 2007 in collaboration with Emory University which provided free testing for individuals at-risk and families affected by Fabry disease. The goals of the study are and continue to be to:

- Help identify individuals early in the disease state so treatment can begin immediately;
- Help affected individuals and their family members understand how this condition can impact their health and how it may affect any unborn family members;
- Develop resources such as informational brochures and webinars to increase the education available for individuals with this condition;
- Increase overall awareness of Fabry disease and;
- Collect data to hopefully lead to advancements in diagnosis, treatment and management of this condition.

Valuable research information was collected throughout the first phase of the study and AAKP is pleased to have received a five-year contribution from Sanofi Genzyme to continue this important work and increase the education and support available for individuals with Fabry disease.

How does someone get Fabry Disease?

Fabry disease happens when a person's body does not make enough of an enzyme called alpha-galactosidase A (alpha-Gal) because of changes or mutations in the GLA gene. When alpha-gal is not working, a fatty substance called globotriaosylceramide (GL-3 or Gb3) builds up in the body's lysosomes (the "recycling centers" of the cell). GL3 storage leads to narrowed blood vessels that cause health problems all over the body, particularly in the skin, kidneys, heart, brain, intestines, and nerves.

One of the unique things about Fabry disease, is that it runs in families in an "X-linked" pattern. That means that the GLA gene that causes Fabry disease is located on the X-chromosome. Women have two copies of the X-chromosome (XX) and men have one copy of the X-chromosome and one copy of a Y-chromosome (XY). If a man inherits an X chromosome with a nonworking GLA gene, they have the symptoms and health problems of Fabry disease. If a woman inherits an X chromosome with a nonworking GLA gene, they have a second, backup X-chromosome with a working GLA gene that can produce some alpha-Gal enzyme. In the past, it was believed that women who were "carriers" would not have Fabry related health problems because they had a normal second copy of the gene in the other X-chromosome. However, research has found that women are not just carriers; they can and do have Fabry related health problems. In some cases, woman can have health problems as severe as their male relatives.

Since Fabry disease is X-linked, it means that all of the daughters and none of the sons of a man with Fabry disease will have the condition. A woman with Fabry disease has a 50% chance for her sons or daughters to have the condition. We have found that when ONE person is diagnosed in a family, it means that on average FIVE of their relatives have it too. That is why the Fabry Family Testing and Education Project is so important, so people at risk to have Fabry disease can get tested easily and for free.



Treatment of Fabry Disease

Another important thing about Fabry disease is that there is a FDA approved treatment for this condition in the United States called Fabrazyme® (agalsidase beta). The hope is that if treatment is started early enough, it will stabilize kidney function and avoid end stage renal disease. If we can find people with Fabry disease through the Fabry Family Testing and Education Project, watch them closely for Fabry related health issues, and get them started on treatment early, they can live longer, healthier lives.

Research Findings

In addition to testing family members, Emory investigators are learning which family members are more likely to be affected by Fabry disease and what health problems they are having. Health issue information is reported by every person who gets testing through this project. AAKP and Emory's first joint study will be submitted for publication in the fall. So far we have learned some interesting information that can help the medical community diagnosis this condition:

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- That men and women who have a family history of Fabry disease and have the eye findings (corneal whorls) are more likely to have Fabry disease.
- Men who have a parent, sibling, or child with Fabry disease AND have pain in their hands and feet or problems tolerating hot or cold weather are more likely to have Fabry disease.

The Faces of Fabry Disease

The Fabry Disease Family Testing and Education project is an important step in helping to diagnosis, treat and manage this condition. As with any chronic condition, an overriding goal is to help those affected live their best life possible. The AAKP and Emory University have a unique partnership, where a patient advocacy organization meets academia. The result is a wonderful collaboration where not only is it important, life-changing research discovered by Emory, but those findings and additional education and support can be communicated easily to patients and families through the AAKP. Both organizations look forward to continuing this project and further helping this patient population.



Cheryl Barcomb *Living with Fabry Disease* Cheryl is a wife, mother of two and grandmother to three. She works full-time and is active in her church. Cheryl's daughter and grandson have also been diganosed with Fabry.



Michael Johnson Living with Fabry Disease Michael is a talented graphic artist and illustrator of books designed to empower and help kids affected by Fabry and other genetic conditions.



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