THE KIDNEY CITIZEN

DPC EDUCATION CENTER

An Introduction to Fabry Disease

By **Jack Johnson**, Executive Director Fabry Support & Information Group

Fabry disease is a rare lysosomal storage disease that is inherited, caused by a mutation on the X chromosome in a section of genetic code that produces the alpha-galactosidase A enzyme or A-gal for short. Lysosomes are the recycling centers of the cell and this is where A-gal goes to do its work of breaking down certain fatty glycolipid materials, specifically one with a big long name referred to as GL3 or sometimes called GB3. If you do not have sufficient functioning A-gal, GL3 builds up filling the lysosome and causing cellular damage.

Fabry is a disease that runs in my family. I was seeing Robert Desnick M.D., Ph.D., one of the leading researchers on Fabry disease and he encouraged me to start a support group for people with Fabry. With very little direction on how to accomplish this, my family and I set out to do just that. Starting in 1996, as one of those true kitchen table groups, we formed the Fabry Support & Information Group (FSIG). We thought we could help by spreading information about the disease, and so we started by producing newsletters.

We began with fewer than 20 names so that first newsletter did not have very wide distribution. But after creating a website, that quickly changed. Today, we have members all across the United States and more than 40 foreign countries around the world.

As the organization grew, it went from something we could manage on weekends and evenings to a full-time commitment. As a result, I found myself transitioning from my previous work life to a new one for FSIG. As the membership and needs of the Fabry patient community grew, the programs and services that were needed grew as well. Unlike so many rare diseases, treatments for Fabry were being developed for clinical trials needed participants. With keen interest from the medical

and pharmaceutical communities, FSIG received donations and grants to meet the needs of our growing organization.

In the beginning, we naively thought the need for a Fabry organization would diminish after a treatment became available. We were told the opposite would be true, and, after an Enzyme Replacement Therapy (ERT) received FDA approval in 2003, we found that to be the case.

The Fabry patient community is comprised of a very diverse group of people. Fabry is proving to be a complicated disease that is equally diverse in its impact on people. Fortunately, interest from the medical community in wanting to understand the disease more fully did not lessen once a treatment became available. Instead, interest increased, and efforts to develop improved next-generation treatments have intensified in recent years. Whether a cure can someday be developed is not known, but hope is still alive.

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When to Consider Fabry Disease as a Diagnosis*

Anyone who has a family member with Fabry disease and/or corneal whorls on eye exam should be tested for Fabry disease. In addition to those cases, anyone with at least two things below should consider testing for Fabry disease.

1. Family history of Fabry Disease
2. Decreased or no sweating in the heat or with exercise (anhidrosis or hypohidrosis)
3. Reddish-purple skin rash in the bathing trunk area (angiokeratomas)
4. Personal and/or family history of kidney failure
5. Personal or family history of “burning” or “hot” pain in the hands and feet, particularly during fevers (acroparesthesias)
6. Personal or family history of problems with overheating when exercising or having burning pain in the cold.
7. Corneal verticillata (“whorls”) that usually do not effect vision, found by eye doctors on eye exam with a slit lamp

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