

# The Consumer Advocate's Voice



## Lori Wise: Fabry Disease Awareness

I am a 57 year-old female with a strong family history of Fabry disease. Since Fabry is an X-linked disorder, females with Fabry have a 50% chance of passing it during each pregnancy. Males with the Fabry gene transmit it to all of their daughters and none of their sons. Because of its rarity and its costly therapeutic drug therapy, having Fabry has required a lot of time and attention with regard to insurance companies. Fortunately for patients receiving the Enzyme Replacement Therapy (ERT), Genzyme provides a Case Manager whose assistance navigating through the insurance rules and policies has proven priceless. I have been receiving ERT with Fabrazyme since late 2004, shortly after it had been approved by the FDA (in the U.S.). In addition to an incredible Case Manager, I have an extensive network of physicians who, along with much diligence on my part, contribute greatly to our goal of keeping me feeling as well as possible.

So, what is Fabry disease?

Fabry disease is an inherited disorder caused by a defective gene. A person who inherits the gene is unable to produce enough of an essential enzyme called alpha-galactosidase A,

or alpha-GAL. Alpha-GAL breaks down a fatty substance called globotriaosylceramide, or GL-3, so that it can be removed from the body. Since a person with Fabry disease does not produce enough alpha-GAL, GL-3 is not removed from the body, but instead builds up in the cells. Over many years, as GL-3 slowly builds up in the walls of blood vessels and other tissues, it is associated with progressive damage. Major organ systems involving the heart, kidney and brain may eventually stop functioning properly causing potentially life-threatening problems.

Because Fabry disease is rare and causes a wide variety of symptoms, it can be mistaken for other diseases. Therefore, people may have the disease for a long time before it is accurately diagnosed. This is a concern because the longer a person has Fabry disease, the more damage is likely to occur in the body's organs and tissues and the more serious the person's condition may become. The earlier Fabry disease is diagnosed, the earlier doctors can start treatment to manage symptoms and try to prevent further health problems.

Note: This is the 4th consecutive year that the Governor of Colorado has signed a proclamation designating April: Fabry Disease Awareness Month.

I have been a Medical Laboratory Technician for over thirty years and worked at National Jewish Health for 17 of those years and at Quest Diagnostics for the last 14. With MSGRC, I have been a Consumer Advocate for 8 years and also serve on the Newborn Screening Panel. In addition, I am a Consumer and voting member with the Colorado Newborn Screening Advisory Committee. I am an extremely passionate individual with a strong desire to help enable those persons, who might otherwise, not have a voice. I have just accepted a new position with Coram Specialty Infusion. I look forward to being a "Patient Relations Specialist" and working with patients again.

