LysoStories™
A Publication from your Healthcare Advocates

May 2006

Welcome

We hope you enjoy reading this second edition of LysoStories, a newsletter designed by Healthcare Advocates for patients and families with lysosomal storage diseases (LSDs). Our first issue of LysoStories included a story about a man with Gaucher disease, an article about home infusion therapy, a summary of monitoring guidelines for patients with Type 1 Gaucher disease, information about the Gaucher Registry, and links to monitoring guidelines for other LSDs. In this issue you will find an article about women and Fabry disease, a story written by the daughter of a man with Fabry disease, and an article about effective ways people deal with longterm medical conditions.

Your feedback is important to us! Please take a minute to complete and return the enclosed business reply card to let us know what you like about LysoStories and your recommendations for additions or changes.

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Updates on recent meetings

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The Gaucher family meeting at the Georgia Aquarium on April 2, 2006 was our biggest and best Gaucher meeting ever, with a dynamic and informative presentation by Jeffrey Marko, MD, emergency room physician and Gaucher patient. Our thanks to the National Gaucher Foundation for their generous support of this meeting.

Upcoming meetings

Our next MPS family meeting is scheduled for June 11, 2006 from 1:30 to 3:00 pm at Maggie’s Little House in Buckhead. Our guest speaker will be Dierdre Monroe, MD, Call Sara Cooper at 404-778-8536 or 1-800-200-1524 for more information.

Please feel free to contact us at 1-800-200-1524 or 404-778-8565 or visit our website at www.genetics.emory.edu/LSDC.

Patient Story

My Unsung Hero
By Amanda Luchento

It’s funny how something tragic can actually bring people closer than they ever were. For example, Michael had had a ‘hard’ life, with a serious illness. He was also not really able to see his kids or wife. But, in 1995 his life became even harder when he was diagnosed with Fabry disease (which is a lack of a certain enzyme in the body). Then in 1999 at the age of 39 it was found that Michael’s kidneys were no longer working and he was in need of a transplant. He could no longer work and had to be on a dialysis machine. In 2000, he received a transplant and could be free from the machine he had to be connected to early in the morning and late at night. It was said that he would die at the age of 40 like most people who suffer from Fabry’s disease, but because of the excellent treatment, he is living strong at the age of 47.

If you haven’t guessed already, this man Michael is my dad. He means so much to me that I don’t know what I’d do without the strange jokes he uses to gross out my friends and I even his obsession over electronics and super heroes. The main reason he is my unsung hero is because of his will to live. There are a lot of symptoms of Fabry disease such as: damaging of the nerves in the body, severe depression, and/or short term memory loss, but he tries his best to live his life. He still doesn’t work and is Mr. Mom for the household and I think that men of any age should learn from him. He is a loving respecting husband, father, and friend to his loved ones and I know I would be nowhere without him.

The day my dad got called in for his kidney everything was in chaos. My mom was off the walls. I was yelling my eyes out, my next door neighbors were cheering and my dad was just smiling. It was something like in the bible where the battle of St. Michael and the devil would be. My dad was St. Michael and the devil was his disease. He likes to tell that story because of the tattoo on his leg. My dad’s favorite part of the story is when St. Michael wins. He is like St. Michael because he is strong but, not just physically. He is also strong mentally and emotionally.

There were days I would break down and cry because daddy had to go to the hospital again. I was so afraid he wouldn’t come

(continued...)
The psychological responses of people affected by Gaucher Disease are as varied as the disease itself. It is clear that the disease itself has been the subject of many of the effects of enzyme replacement therapy upon the quality of life of Gaucher patients. But there is another very important form of information, the psychological impact of Gaucher Disease. There is an existing body of knowledge, however, regarding the psychological factors involved in diagnosis and disability, including arthritis, chronic pain, substance abuse, and life-threatening disorders. Some of that information has relevance for the Gaucher community and, perhaps, for the entire lysosomal storage disease (LSD) community. Ineffective Denial Since the beginning of enzyme replacement therapy for Gaucher and other lysosomal diseases, some medical practitioners have noted certain clinical responses from a few of their patients. There have been patients who, despite medical test results, and, in some cases, their own diagnosis, continued to deny that they had the disease, define the impact of their disease as mild. Practitioners who Specialize in Fabry Disease would have the advantage of an effective enzyme replacement therapy, have been dismayed by the lack of enthusiasm from some of the people that would benefit if treatment were available. They have known that many people live with the medical consequences of the disorder. There have been individuals who cannot deal with their disease, multiple fractures or failed joint replacements, who use assistive devices, experience chronic pain, or have to give up their labor-intensive activities, who neither decline treatment. What does this mean? The phenomenon of the minimization of the impact of a disorder has been characterized by some writers as the psychological process of “denial” and has been described as “infectious denial,” because the patient seems to be ignoring a potentially crushing reality. And the use of that psychological defense has been viewed historically as less than desirable for mental health.

Is Denial Always Bad? I attended a research symposium several years ago with several dozen psychologists, and I vividly recall the moment we heard the jaw-dropping results of a study of female breast cancer patients. Those women who evidenced “denial” of their illness were living longer than their most stoic peers. Women who experienced a “loss of control” of their body and their disease were living consistently shorter lives. Gaucher patients who do not intend to live the constraints imposed by a disorder is limiting healthier strategies. An adaptive, positive approach to the illness can be components of a healthy lifestyle. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers. The active treatment of health problems can mean that life offers containing everything else that life offers.
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sad without him there beside me. Most of all I
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when they are worried, they think bad things. When he went to an infusion, just for 2-3 hours, I
would sit home and look back on the afternoons we would go out dancing and never know where to
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The disease is important because it is important to discuss Fabry disease with any family
members benefit from exposure to successful role-
models, which can take the form of attending patient and profes-
sionals, members benefit from exposure to successful role-
models. These groups provide the patient and family members an opportunity to learn about Fabry disease, to make sense of its impact, and to share their experiences. An informed understanding can empower patients and families to take control of their lives and to live as independently as possible.

Fabry Disease: Important Facts for Women

What is Fabry disease?

Fabry disease is a genetic lysosomal storage disorder that affects about 1 in 40,000 people, predominantly males, of their female family members.

Signs of Fabry disease include severe pain in the hands and feet, the inability to sweat, fatigue, a pinkish-purple skin rash typically in the bathing treatment, frequent infections, abdominal pain, kidney failure, heart problems, hearing loss, strokes and early death.

The condition is caused by a lack of an enzyme called a-galactosidase which breaks down a fatty substance in the body called globotriaosylceramide, GL3. In affected females, the enzyme is not produced or is produced in low levels. It is inherited in an X-linked pattern of inheritance, meaning that the gene for Fabry disease is located on the X-chromosome. The risk is that women who are heterozygotes will be carriers and would not have Fabry related symptoms. If a woman is a carrier, she will pass the Fabry gene on to one of her sons or her daughters with 50% probability.

What is the treatment for women with Fabry disease?

The treatment for Fabry disease depends on whether the condition is inherited in an X-linked pattern of inheritance. If the condition is inherited in an X-linked pattern of inheritance, the gene for Fabry disease is located on the X-chromosome. The risk is that women who are heterozygotes will be carriers and would not have Fabry related symptoms. If a woman is a carrier, she will pass the Fabry gene on to one of her sons or her daughters with 50% probability.

How do I determine if I have Fabry disease?

A diagnosis of Fabry disease in a woman has three parts. The first part is a genetic appointment and evaluation, the second part is a simple blood test that measures the level of a specific protein in the patient's blood, and the third part is a genetic test that determines if you carry the Fabry gene on one of your X-chromosomes. This genetic test can also be done for men.

When should I test my children to see if they have Fabry disease?

If you have a family history of Fabry disease, it is important that you have a heart and kidney study if you have symptoms. In addition to these organs, you may also be tested for other Fabry-specific studies, such as brain MRIs and hearing tests. Fabry-specific studies are also recommended, but on a less frequent basis. A Lysosomal Storage Disease Center can discuss the decision to test one's children to see if they have Fabry disease.

If you had a genetic test performed on you at age 13 and were told you have Fabry disease, do you have Fabry disease?

Yes. If you have copy of the Fabry gene, the Fabry gene may experience any of the above listed symptoms of the disease.

Is there a genetic or family history of Fabry disease?

Yes, women who inherit the Fabry gene may experience any of the above listed symptoms of the disease.

Who else in my family could be affected by Fabry disease?

Fabry disease is an X-linked inherited disorder. Women can only pass the gene for Fabry disease on to all of their daughters and to none of their sons. Heterozygous females, whether symptomatic or not, have a 50% chance of passing the Fabry gene to either their sons or their daughters. It is very important to note that Fabry disease is not a genetic disease, but a genetic disorder. If your Fabry gene comes from your father, you will have Fabry disease.

When I was 13 and was told I had Fabry disease, I was referred to a genetics specialist for a genetic appointment and evaluation. During the genetic appointment and evaluation, the genetics specialist will discuss the results of the genetic test to determine if you have Fabry disease. You will be referred to a Lysosomal Storage Disease Center for treatment and management of Fabry disease.

Where can I learn more about Fabry disease?

A Lysosomal Storage Disease Center near you (see listing on the back page) can be a great resource.

For more information on Fabry disease, please visit the website of the Fabry Foundation (www.fabry.org) or the website of the Fabry Support and Information Group (FSIG) (www.fabrys.com).

Staying in the Game, Playing Defense

By Wayne D. Rosenfield, Ph.D.

The psychological responses of people affected by Gaucher Disease are as varied as the disease itself. Some might have taken the full treatment of enzyme replacement therapy upon the quality of life of Gaucher patients. But there is another very different group of patients, most likely affected by the psychological impact of Gaucher Disease. There is an existing body of knowledge, however, regarding the psychological factors involved in quality of life, including arthritis, chronic pain, substance abuse, and life-threatening disorders. Some of that information has relevance for the Gaucher community and, for the entire lysosomal storage disease (LSD) community.

Ineffective Denial

Since the beginning of the enzyme replacement therapy for Gaucher and other lysosomal diseases, some medical practitioners have noted certain emotional responses from a few of their patients. There have been patients who, despite medical test results and, in some cases, life-threatening disorders, persist in their denial. There is evidence that effective denial, treatment have been dismayed by the lack of enthusiasm from some of the people that have been treated for Gaucher disease. The ability to maintain a positive attitude, to give the smoker lungs, to give the smoker back that I would think to the future and see me
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Upcoming Patient Meetings

MPHS
• July 27-29, 2006
National MPS society 21st Annual MPS society Family Conference
Covington, Kentucky
• June 29 - July 2, 2006
9th International Symposium on Macropoly saccharide and Related Disease
Venice-Lido Italy

Pompe
• November 2006
International Pompe Association acid Malron Deficiency Association Patient Conference
San Antonio, TX
• September 15-16, 2006
Association for Glycogen Storage Disease
Honolulu, HI

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Emory Lysosomal Storage Disease Center

Infection Center Updates
The Emory Center for Genetic Infusions, located in our clinic building at 2165 N. Decatur Road, continues to be a meeting place and medical home for our patients. We thank everyone who has donated to our Lysofriends fund. We are excited about the recent installation of our newly donated TVs.

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Patient Story
My Unsung Hero
By Amanda Luchento

It’s funny how something tragic can actually bring people closer than they ever were. For example, Michael had had a “hard” life, with a tiresome job. He was also not really able to see his kids or wife.

Michael was diagnosed with Fabry’s disease (which is a lack of enzyme activity) in 1995. My dad and mom were in shock when they were told the news. It was so afraid he wouldn’t come home for his wedding. I was so afraid he wouldn’t come back home for his wedding.

The day my dad got called in for his kidney every thing was in chaos. My mom was off the walls. I was bailing my eyes out, my next door neighbors were cheering and my dad was just smiling. It was like something in the bible where the harlot of St. Michael and the devil were. My dad was St. Michael and the devil was Michael. He is like St. Michael because he is strong but, not just physically. He is also strong mentally and emotionally.

There were days I would break down and cry because daddy had to go to the hospital again. I was so afraid he wouldn’t come back home for his wedding.

My dad’s favorite part of the story is when St. Michael and the devil would be. My dad wins. He is like St. Michael because he is strong but, not just physically. He is also strong mentally and emotionally.

I know I would be no where without him. My dad’s favorite part of the story is when St. Michael and the devil would be. My dad wins. He is like St. Michael because he is strong but, not just physically. He is also strong mentally and emotionally.

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