



Center News

Emory Lysosomal Storage Disease Center

Infusion 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. Our thanks to everyone who has donated to our Lysofriends fund! We are excited about the recent installation of our newly donated TVs.

Updates on recent meetings

The annual Fabry meeting on January 29, 2006 was a big success. Everyone enjoyed the presentation by the compassionate authors of "Sick and Tired of Feeling Sick and Tired."

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 Manko, 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 schedule for June 11, 2006 from 1:00 to 3:00 pm at Maggiano's Little Italy in Buckhead. Our guest speaker will be Joe Muenzer, 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



A happy patient at the new Emory Genetic Infusion Center.

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.

If you have a suggestion for an article or would like to tell *your* story, please contact a member of the Publications Committee:

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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. But, in 1995 his life became even harder when he was diagnosed with Fabry's 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 or 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 each day. 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 no where without him.

The day my dad got called in for his kidney everything was in chaos. My mom was off the walls. I was balling my eyes out, my next door neighbors were cheering and my dad was just smiling. It was like something in the bible where the battle of St. Michael and the devil would be. My dad was St. Michael and the devil dragon was the 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

(cont.)

back that I would think to the future and see me sad without him there beside me. Most of all I remember crying because I thought about how daddy and I couldn't dance at my wedding. It was a bad way to think but that's what people do when they are worried, they think bad thoughts. When he went to an infusion, just for 2-3 hours, I would sit home and look back on the afternoons we would go get sushi from our favorite restaurant or go out driving and never know where we'd end up. Its days like those people take for granted but

I could never do that. He is only one man, and he can only live once. He is my daddy, my father, my unsung hero.

This article was written by Amanda who was 13 at the time. She is now 15 years old and has been evaluated for Fabry disease. She remains asymptomatic at present and her health has been very good. Amanda lives an active life at school and at home. She is a good student; she plays recreational Basketball. Amanda has recently been accepted to

attend the Fine Performing Arts School; there she is planning to study Drama and singing. Her goal is to perform on Broadway. Amanda also has interests in science especially genetics.

Michael has been receiving Enzyme Replacement Therapy at St. Peter's University Hospital every two weeks. His Fabry disease is stable. Michael takes care of his family; he is a stay at home dad.

Fabry Disease: Important Facts for Women

Department of Human Genetics
Division of Medical Genetics
Emory University School of
Medicine

What is Fabry disease? Fabry disease is a genetic lysosomal storage disorder that affects about 1 in 40,000 men and an unknown number 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 trunk area, frequent diarrhea and constipation, abdominal pain, kidney failure, heart problems, hearing loss, strokes and early death.

The condition is caused by lack of an enzyme called α -galactosidase which breaks down a fatty substance in the body called globotriaosylceramide, or GL3. In families affected by Fabry disease, men who inherit the Fabry gene always develop related health problems, while women with the gene have more variable symptoms. The difference in expression between men and women is related to the way in which the condition is inherited. Fabry follows an X-linked pattern of inheritance, meaning that the gene for Fabry disease is located on the X-chromosome. Men, who have only one X chromosome, are unable to produce the enzyme if they inherit the Fabry gene and therefore always have symptoms. Women, on the other hand, have two X chromosomes. If they inherit one Fabry gene, they have a second gene on their other X chromosome which may be able to produce enzyme. Women with one Fabry gene and one normal copy of the gene are called "heterozygotes." Their α -galactosidase levels may vary from normal to very low. In the past, it was believed that women who were heterozygotes were carriers and would not have Fabry related health problems. We now know that women can and do have symptoms of Fabry disease. In some cases, their health problems are as severe as those of their male counterparts. Since women who carry one copy of a Fabry gene may have symptoms of

the disease, it is important that they discuss Fabry disease with their doctor and obtain appropriate referrals to monitor their health.

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 genetics appointment and evaluation, the second part is a simple blood test that measures the levels of the α -galactosidase enzyme in your 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 is run on the same blood drawn for the enzyme test.

As a woman, can I have health problems related to Fabry disease? Yes, women who inherit the Fabry gene may experience any of the above listed symptoms of the disease.

Are there certain tests suggested to check my health? Yes. If you have copy of the Fabry gene, it is important that you have heart and kidney studies to monitor these organs every year. Other studies, such as brain MRIs and hearing tests are also recommended, but on a less frequent basis. A Lysosomal Storage Disease Center near you that specializes in Fabry disease can monitor you and other family members and make recommendations to your primary care physician. Information about Fabry-specific monitoring recommendations is available at www.fabryzyme.com.

What is the treatment for women with Fabry disease? The best treatment for a woman affected by Fabry disease depends on her individual health problems and needs. Enzyme replacement therapy is available to help reduce the amount of fatty GL3 build-up in the body's tissues and prevent irreversible damage to organs. Dilantin or Tegretol is often prescribed for Fabry related pains. Reglan is often used to help alleviate the stomach and intestinal issues associated with Fabry disease. At different points in a woman's life, she may choose different treatments.

Will it hurt my baby if I become pregnant during treatment for Fabry disease? The answer to this question depends on the treatment a patient is undergoing. BEFORE becoming pregnant any woman wishing to consider pregnancy should discuss their medications with their doctor. No human studies have been done to establish the safety and efficacy of enzyme replacement therapy

for Fabry disease during pregnancy. This drug should be used during pregnancy only if clearly needed. If a woman who is taking Dilantin or Tegretol to control her hand and foot pain wishes to become pregnant, she should discuss her medications with her doctor before becoming pregnant. Taking Dilantin and Tegretol during pregnancy increases the risk for birth defects and developmental delays in the baby. If you become pregnant unexpectedly, call your doctor as soon as you learn of your pregnancy to discuss medication use.

Who else in my family could be affected by Fabry disease? Fabry disease is an X-linked inherited disorder. Men with Fabry disease pass the gene on to all of their daughters and to none of their sons. Heterozygote 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 discuss Fabry disease with any family member who could be affected by the disease. If you would like a copy of a letter explaining Fabry disease to your family, and/or testing information, contact one of the Lysosomal Storage Disease Centers listed below or contact the Fabry Support and Information Group (FSIG) at www.fabry.org or 1-660-463-1355.

When should I test my children to see if they have Fabry disease? Since the FDA's approval of the enzyme replacement therapy treatment, many families are now choosing to test their children at an earlier age. Early diagnosis means early intervention. A team member of your local Lysosomal Storage Disease Center can discuss the decision to test your children with you further. Please refer to the listing below of Lysosomal Storage Disease Centers in the U.S.

Where can I call to learn more about Fabry disease? A Lysosomal Storage Disease Center near you (see listing on the back page) can be a great resource.

Genzyme Medical Information (www.Fabrycommunity.com) can provide accurate information on enzyme replacement therapy for Fabry disease.

The Fabry Support and Information Group or FSIG (www.fabry.org) is another valuable source of information.

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. There has been some study of the effect of enzyme replacement therapy upon the quality of life of Gaucher patients. But there is otherwise very little scientific information specific to the psychological impact of Gaucher Disease. There is an existing body of knowledge, however, regarding the psychological factors in illness 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 enigmatic responses from a few of their patients. There have been patients who, despite medical test results and, in some cases, their own disability and acute pain, define the impact of their disease as mild. Practitioners who themselves are excited by the availability of an effective enzyme replacement therapy, have been dismayed by the lack of enthusiasm from some of the people they would like to treat; a lack of interest in treatment from the very people who live with the medical consequences of the disorder. There have been individuals who have endured multiple fractures or failed joint replacements, who use assistive devices, experience chronic pain, or have altered their schedules and activities, who nevertheless 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 “ineffective denial,” because the patient seems to be ignoring a potentially crushing reality. And the use of psychological defenses 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! The “denial” of their illness did not mean that they were consistently blocking the disease from their awareness. Nor were they refusing medical treatment. Rather, they were making conscious decisions to maintain normalcy in their

lives, for as long as possible. What appeared to be the psychological defense of “denial” seemed really to be patients’ conscious efforts to avoid defining themselves as “sick.”

Other patient populations have been confronting some of the issues now faced by the lysosomal storage disease community: Individuals at risk for Huntington’s Disease must decide whether to be tested for a disorder that, if present, may take decades to appear. Carriers of mutations that predispose individuals to breast cancer or many other disorders, must decide whether to be tested and possibly treated, and then decide how – or whether – to communicate the findings to other family members. The personal and family implications are enormous. Would a decision to deny the issue necessarily be evidence of poor coping? Or would a decision to defer the knowledge reflect awareness that the issue is complicated? A test result itself could have life-changing consequences. Depending upon the unique family and personal circumstances, deciding to defer acquisition of certain types of genetic information may have compelling justification.

Is denial of illness good or bad? The answer, I believe, lies in its effect upon the individual’s ability to maintain a predictable, pleasurable lifestyle which is not consumed by the disorder or its treatment. The lessons from breast cancer treatment are important: Retaining normalcy is helpful, but it does not replace the medical treatment. The quality of a person’s life will be adversely affected by a disorder allowed to progress into irreversible consequences. The best healthcare services in the world cannot always turn back time to recall a metastasized cancer, to give the smoker back his lungs, the Fabry patient back his kidneys, or to give the Gaucher patient parts and functions adversely affected by disease. But the wish to maintain normalcy during the course of treatment is desirable and realistically attainable for the patient with a treatable lysosomal storage disease. Receiving infusions at home or at work may be one strategy for incorporating treatment into a normal routine. Even infusions in a medical setting can present an opportunity for relaxation, for correspondence, reading, completing paperwork, or for socialization. Infusions need to be only a relatively small part of everyday life, leaving the rest of the week for activities which have nothing to do with lysosomal disease. There are many healthful behaviors which are personal investments in present and future wellness. Eating a proper diet, limiting alcohol consumption, avoiding smoking, engaging in regular exercise, developing hobbies and social interests, managing stress, and having fun can all be components of a healthy lifestyle. The active treatment of known health risks including, for some of us, a lysosomal storage disease, is part of our investment in future health.

Maybe Denial *is* Just a River in Egypt!

What may appear to be denial may, in fact, be a practical adaptation to illness. A focus upon the constraints imposed by a disorder is limiting indeed. A focus upon life’s possibilities *despite* the disorder creates limitless possibilities for productivity and joy.

An adaptive, positive approach to the illness can take many forms. The reports of “denial” among patient populations may actually represent adaptive efforts to keep the disorder in its own “compartment,” where it cannot contaminate everyday life.

What Else Works?

The tools for practicing a healthful, optimistic approach are everywhere. Indeed, groups of individuals with similar concerns – people who are vulnerable to abuse of drugs and alcohol are some of the most notable examples – have formed groups for mutual support. Adopting the language of “recovery,” such groups empower individuals to regain control of their lives and to fully integrate with community and family life. These groups connect peers who understand the needs of each other, cheer each other’s successes, and prod members to move beyond helplessness and self-pity. The action of providing a coping role-model for people who may be struggling, and advocating for their recovery, is itself an uplifting experience. Advocacy for others requires the advocate to adopt an optimistic mindset.

Connecting with other similarly affected people can take the form of attending patient and professional meetings and sharing one’s experiences in person. Others have found writing or speaking about their experiences to be helpful. The internet has provided a readily available means for finding and connecting with others. At least one discussion group already exists for the Gaucher Community, but no special skill or permission is required to form other groups. All that is required is the desire to connect with others. Similar to the other recovery groups which have existed for many years, members benefit from exposure to successful role-models, and receive the types of support, encouragement, and even prodding, that can come only from people with first-hand experience.

Maintaining a healthful response to a lysosomal storage disease can mean allotting time to tests and treatments. And a healthy life can mean *denying*, or maybe just ignoring for a while, a disorder. In the context of taking care of mind and body in many other ways, the limited time devoted to each treatment is an investment in a healthy future containing everything else that life offers.

Fabry Centers in the U.S.

Center name and address	Contact person	Phone #	Email
St. Peter's University Hospital Institute for Genetic Medicine MOB Room 2190 254 Easton Avenue New Brunswick, NJ 08901	Nita Patel, RNC, BSN Rashmi Chikarmane, MSc, CCGC	(732) 745-6659	npatel@saintpetersuh.com rchikarmane@saintpetersuh.com
Cincinnati STAR Center for LSDs Cincinnati Children's Hospital 3333 Burnet Avenue ML-4006 Cincinnati, OH 45229	Laurie Bailey, MS	(800) 647-4805 (513) 636-4507	Laurie.Bailey@cchmc.org
Massachusetts General Hospital Lysosomal Storage Disorders Program	Virginia Clarke, RN	(617) 726-8830 (617) 726-5732 (appointments)	vclarke1@partners.org
University of Pittsburgh Lysosomal Disorders Center Division of Medical Genetics 3705 Fifth Avenue Pittsburgh, PA 15213	Nadene Henderson, MS, CGC Michele Graham, RN	(800) 334-7980 (412) 692-3476	nadene.henderson@hgen.pitt.edu
Children's Hospital of Philadelphia Lysosomal Storage Disease Center Section of Metabolic Diseases 9th floor, South Tower, Suite 20 34th Street and Civic Center Blvd Philadelphia, PA 19104	Joan Hart, program coordinator	(215) 590-3692	hartj@email.chop.edu
Children's National Medical Center Division of Medical Genetics & Metabolism	Sandra Yang, MS, CGC	(202) 884-4168	syang@cnmc.org
Emory University School of Medicine Lysosomal Storage Disease Center Department of Human Genetics 2165 N. Decatur Road Decatur, GA 30033	Dawn Laney, MS, CGC	(800) 200-1524 (404) 778-8518	dlaney@genetics.emory.edu
NYU School of Medicine Neurogenetics	Carol Fisher, RN	(212) 263-8344	fisher01@med.nyu.edu
Children's Memorial Hospital	Shanna Randolph, RN, MS, APN	(773) 880-4560	srandolph@childrensmemorial.org
University of California San Francisco Stanford Lysosomal Storage Disease Center	Cecilia Fairley, MS	(800) 901-9997	fairleyc@peds.ucsf.edu

Upcoming Patient Meetings

MPS1

- July 27-29, 2006
National MPS society 21st Annual MPS society Family Conference
Covington, Kentucky
- June 29 - July 2, 2006
The 9th International Symposium on Mucopolysaccharide and Related Diseases
Venice-Lido Italy

Pompe

- November 2006
International Pompe Association Acid Maltase Deficiency Association Patient Conference
San Antonio, TX
- September 15-16, 2006
Association for Glycogen Storage Disease
Hershey, PA

Gaucher Disease

- July 18-22, 2006
7th Meeting of the European Working Group on Gaucher Disease
Cambridge, United Kingdom
- June 24-25, 2006
2nd Lysosomal Disease and the Brain Conference
NIH and Children's Gaucher Research Fund
Sacramento, California
- Expression of Hope
Sponsored by the NGF
A global program featuring works of art by the community touched by lysosomal disease. For more info go to the NGF website at www.gaucherdisease.org