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LysoStories™

A Publication from your Healthcare Advocates

Welcome

We hope that you enjoy reading this winter issue of LysoStories, a newsletter designed by Health Care Advocates for patients and families with lysosomal storage disease (LSDs). If you have a suggestion for an article or would like to tell your story please contact a member of the Publication Committee

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What's New

There are a few important websites that have been helpful recently during this unprecedented time of change in the lysosomal storage disorder community in particular with regard to Fabry and Gaucher:

Current listings of clinical trials *(search by disorder)*
www.clinicaltrials.gov

Updated National Gaucher Foundation website
(Pharmaceutical Company tab)
www.gaucherdisease.org

Supply updates from Genzyme
www.genzyme.com/supplyupdate

Fabry Support and Information Group
www.fabry.org

The National Fabry Disease Foundation
www.TheNFDF.org

Living with Pompe disease: A Family Story

By Michelle Hackenberry

Hello. My name is Michelle. My husband Mark and I have 3 children – Tyler, Mollie and Abby. Two of our three children are affected by Pompe Disease.

In February 2006, after approximately 6 long years of searching for answers, Tyler was diagnosed with Pompe Disease. A muscle biopsy confirmed that he was suffering from this rare genetic disease. Tyler was a fairly active young boy, but it wasn't hard to notice that he had difficulty keeping up with the other kids his age. As he got older, he was diagnosed and treated for migraine headaches. In addition to the migraines, he started experiencing increased irritable bowel symptoms. During routine labwork to monitor the effects of the medications being used to treat both his migraines and irritable bowel, it was discovered that his liver function panel was abnormal. This was the beginning of a lot of testing (EMGs, labwork, MRIs, muscle biopsies and even a liver biopsy was scheduled). Over a short period of time, Tyler started experiencing fatigue and increased muscle and joint pain that started in his legs and eventually affected his whole body. Simple daily functions such as climbing stairs, attending school, sitting and standing in any position for prolonged times and sleep patterns were becoming more difficult. We were devastated to hear that Tyler was found to also have a cardiomyopathy with a moderate dilation of his aorta. It was heartbreaking to know that the most important muscle in his body was affected by this disease.

Not long after Tyler's diagnosis, we had met with a geneticist and the metabolic medicine physicians at Children's Hospital of Philadelphia (CHOP) where we learned the inheritance pattern of the disease. Knowing the affects Pompe has on the muscles and the importance of early detection, we chose to have Mollie and Abby both undergo genetic testing immediately. In August 2006, again we were devastated to find out that our youngest daughter, Abby had Pompe disease. Mollie was not affected by Pompe and it was determined that she was not a carrier. As you can imagine, in 2006 our lives changed forever- becoming much more complicated than I could have ever imagined.

Abby was 10 years old when the diagnosis was made. Her age at the time of diagnosis is significant to our family because Tyler was around the same age when he started showing signs and symptoms of this debilitating disease. Abby has mild asthma- like symptoms, irritable bowel, and occasional muscle pains in her feet and legs. To date, she does not have the cardiac complications that affect her brother.

Tyler and Abby have been receiving care for their disease for 3 years now. Overall, they both remain stable and we are hopeful that this will continue to help Abby stave off the terrible effects of the disease and that progression of the disease process will continue to slow down for Tyler.

When we initially found out that we had a diagnosis, it was such a relief to know the condition had a name. That relief was short-lived when we realized how rare and progressive the disease was and that there was not a cure. The more we learned about the disease, the more alone and isolated we

Continued on page 2

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Patient volunteers can become valuable contributors to Fabry disease education efforts for the greater good of the community. Patients should contact us at info@TheNFDF.org for assistance and to let us know about their contribution to our overall NFDF education efforts. To date I have personally shown angiokeratoma to over 500 dermatologists in various physician forums around the country. The resulting diagnoses from these education physician opportunities can change the course of someone else's life.

We are beginning a similar program and call-for-action by people with Fabry disease to improve recognition by eye doctors. Please visit our website in the "Education" then "Fabry Disease Eye Symptoms" tabs for more details. These are not new concepts and many Fabry patients have already contributed. We are hoping to facilitate significantly more education opportunities.

The examples above in combination with our website and Fabry family meetings represent some of the NFDF's ongoing education efforts. More programs will be developed and administered as resources become available. Increased education is so important.

Fabry disease assistance and support:

The NFDF's programs to assist and support the Fabry community range from small actions such as providing information by phone or email to major activities like organizing the 2010 "Charles Kleinschmidt Fabry Family Camp" at Victory Junction, NC. In between these extreme ends of the spectrum, we are trying to keep the Fabry community informed and make their lives easier.

We have put together an extensive listing on our website of physicians and staff members who treat Fabry disease. We are providing free access to medical journal articles about Fabry disease for our members. Our efforts to provide various social networking opportunities are providing new ways for the Fabry disease community to interact and to receive news, information and support. NFDF social networking options include Inspire, Face Book, Twitter, a Wordpress Blog, and LinkedIn. We hope there is something for everyone.

We are very excited about our Fabry family camp opportunity which is going to be great fun! Please see our website and our recently published e-newsletter for more information about all of these and other activities.

The NFDF welcomes suggestions about other ways we can support the Fabry community. We are trying to build a strong community with a community voice that carries our needs to the doorsteps of legislators, government agencies, corporate and organization sponsors, and others who can help to improve the lives of people living with our disease.

The NFDF feels strongly that the Fabry community needs all the help we can get and that alone, none of us can do enough to support our community. Please enjoy and participate in the efforts of our sister organization, the Fabry Support and Information Group (FSIG) at www.fabry.org, and the various clinical centers around the country. Many major treatment center websites are included in our "Find a Doctor" database.

We have enjoyed sharing a little about our organization with you. Lastly, if you believe in what we are doing and you are able to provide support, please swing by the "donate" link on the home page at www.TheNFDF.org. Thank you from our hearts!

Sincerely, Jerry

Jerry Walter

Founder and President (and patient)

started to feel. There was not a lot of information out there, but we were directed to the Genzyme website that at least gave us good information that was not as scary and intimidating as some of the other information we were finding on the internet. I have found comfort in reaching out to others who have Pompe Disease or who have a family member affected by the same disease as Tyler and Abby. It has been extremely helpful for us to actually meet others with Pompe disease and to form friendships that are like no other. We share stories, solutions and encouragement to one another. Many of us – no matter what the disease process is- need to pull strength from others from time to time as we face similar realities of what life has dealt us.

Having two children with Pompe disease certainly has raised its share of challenges. We both have full-time careers and one of the biggest challenges we face is planning and coordinating schedules, care, doctor appointments, testing, school activities/conferences and all the other normal activities that we do each day. At times, we have Tyler and Abby receiving their care on the same day. This is very complicated process because Tyler receives his care at CHOP and Abby receives her's at home with a homecare nurse. Many times multiple appointments are scheduled on any given day – appointment days usually turn into all day events. My husband Mark and I must function as a tag team to get the jobs done. We attempt to alternate the appointment schedules, but at times this is impossible. Financial challenges are another major factor that at times have consumed our lives. With the increasing costs of medical care, insurance premiums on the rise and out of pocket expenses that would make most people shudder, we try to make the best of the situation. Mark handles most of the financial information and spends hours on the phone trying to straighten out the medical benefits and working out payment details with billing offices. Billing professionals have told us that our account is very complicated and they have a difficult time trying to help sort out the medical billing info – so imagine the frustration we feel when trying to sort through the explanation of benefits, claim forms and piles of invoices we must pay to make sure our children can continue to receive the care they need. I am sure that each of you understands the challenge of maneuvering thru the medical system. We have many medical professionals and other support individuals that are part of the team that takes care of Tyler and Abby. We are very grateful to each of them for helping our kids get the care that is necessary to fight this disease that affects them. You can imagine with having such a large medical team also comes many different personalities and ideas on how to best meet the needs of both our children. This at times creates conflict, miscommunication and frustration with those involved in determining the care Tyler and Abby so desperately need. As parents, we take care of things that are normally left up to medical staff to handle – making sure our kids' medications and testing are properly preauthorized and following up on the completion of medical forms for hospital clearances – making sure this is done as to not disrupt the scheduling of an important surgical procedure or test. At times, our frustration stems from the lack of understanding of the important and urgent nature of the requests we make to our physicians. To physicians, we may come across as "pushy" or "demanding". In our eyes, we are advocating for our children –attempting to obtain the best medical care available to Tyler and Abby in hopes for the best outcomes possible.

At times, our family rides the emotional roller coaster that most certainly affects each and every family that has been diagnosed with a debilitating disease or condition. I myself must admit that I have bouts of frustration and anxiety that includes ranting and raving, tearful outbursts and sleepless nights that result from the challenges and disappointments we face on a regular basis. For my own sanity, I allow myself to have my "pity party" and then move on to what is important – helping my kids to stay as healthy and happy as possible, achieve relief from the muscle pain they experience, encourage them to remain as active as possible and to focus on the fact that specialized medical care is available to them. Pompe disease does not define who we are, but has played a major role in who we have become. It too has had some positive impact on our lives. It has certainly made us stronger as a family. Our family bond is second to none. We have openness with discussing the good, the bad and the ugly. We do not hold anything back from our kids. Honesty when answering their questions can be very stressful and difficult at times, but important to us that they are getting the information they seek. We encourage them to ask questions, participate in decisions in regards to their care and most importantly learn to become advocates for themselves. This is very important to me that they take on this role on their own behalf. As a parent, I will always advocate for my children, but my hopes are that they will become more comfortable in participating and staying directly involved in the care they need.

Everyday we hope and pray for a cure. We are forever grateful for the existing medical care that allows Tyler and Abby to live as normal a life as possible. We are excited and encouraged about ongoing research that will hopefully improve treatments available to our children and others affected by Pompe. Life has unfortunately dealt our family – most importantly, Tyler and Abby a terrible blow. Because of the many caring people who have dedicated their lives and careers to studying, understanding and developing treatments for Pompe disease my kids have a chance at beating the odds.

Thank you for allowing me to share our story.

Life Transitions in Lysosomal Storage Diseases: A Focus on Teens

By Dawn Laney, MS, CGC

It is not in the stars to hold our
destiny but in ourselves.
-- William Shakespeare

"You're not the boss of me, NO!"
--The song from Malcolm in the Middle, by
They Might Be Giants

The teenage years are the life period during which teens begin separating their thoughts, goals, and self-image from that of their parents in order to become successful adults. In individuals affected by a chronic disease, such as a lysosomal storage disease, the teen years should also include a transfer from parent directed to parent-directed medical care. As you might expect, avoiding a fast, confusing crash of medical needs onto a teen in favor of a gradual planned and more peaceful transition of medical control takes planning and small steps. Here are my top 10 tips for preparing for the transition from parent to patient directed medical care:

1. **Start early!** The early teen years are a great time to communicate and plan. Schedule a meeting including parents, the patient, and core medical team members to put together a transition plan. This meeting should include a discussion about the importance of self directed medical care as teens enter adulthood. If teens can start by talking with parents and health care providers, it can make the transition easier AND give them the chance to learn why to take healthcare seriously.
2. **Take a quick test.** Before starting off on a transition journey, figure out what the teen knows about life, their disease, and their medical care. Basic health care questionnaires like the "Ready to Manage Your Own Health Care?" form created by the PACER Center, Inc. are a great start and direct you to areas that need attention. (See resource #2)
3. **Know your disease and treatment plan.** Teens should work closely with their key health care providers to understand their condition and what needs to be done to keep them as healthy as possible. It is important for teens to learn why they do all that they do medically (treatments, pills, doctor's appointments, and tests). If they understand how and why they do things, it provides a framework in which to rank them in importance in day to day life. This process can be facilitated further using a disease and treatment summary sheet. (See resource #3)

4. **Know your healthcare providers.** It is important for teens to know each of their doctors/health care providers and their medical specialties. A list with contact information for each healthcare provider is very helpful for quick reference. This knowledge will help them know how and when to contact the correct health care provider during a medical issue.
5. **Learn about insurance.** It is important for teens to discuss insurance with their physician, parents, and helpful billing folks (like Genzyme Care Coordination). Important points to review would include why insurance is needed and how insurance availability changes over time. It is also critical that teens know what happens to their insurance in different life scenarios.
6. **Plan for adulthood.** Ask teens to think about what they may like to do over the next years and discuss it with their parents and healthcare providers. Do they want to continue in school after high school? Do they want to work right away? What would be their realistic dream job? Teen, parents, and healthcare providers can work together to help teens understand how to make their goals a reality AND discuss practical issues, like insurance, that go with them.
7. **Meet the new doctors.** Some doctors, often medical geneticist/genetic counselors are able to work with patients for their entire life. Other doctors, like pediatricians, are primary care physicians only until age 18-21 years. A change from "kid-focused doctors" to "adult doctors" can be easier if the teen and parents know who they are and meet them before it is officially time to move care.
8. **Learn when a health issue requires treatment.** Some health issues are more serious than others. Providers and parents can help teens learn when a stomach ache is something that a primary care doctor needs to know about immediately as opposed to requiring an over the counter medication and waiting for it to go away.
9. **Advocate for yourself.** Teens know themselves best, so they must become involved in their healthcare life. They need to teach people about their disease and needs. Teens need to know that it is ok to speak up if they are uncomfortable with a healthcare provider or need a little more explanation about something.
10. **DON'T PANIC!** It may seem like a lot of things to do and know, but millions of teens move from being a kid to an adult every year. With planning and participation from everyone, this can be a smooth transition.

Resources:

1. *A detailed booklet on "Adolescent transition care: A Guidance for nursing staff" http://www.rcn.org.uk/__data/assets/pdf_file/0011/78617/002313.pdf*
2. *Health Knowledge Questionnaire "Ready to Manage Your Own Health Care?": http://www.minnesotaschoolnurses.org/Ready_to_Manage_Questionnaire.pdf*
3. *Several useful Transition Questionnaires and Resources such as can be found at http://www.minnesotaschoolnurses.org/ in the special education section.*
4. *Disease and treatment summary sheets: several paper-based options at http://depts.washington.edu/healthtr/ and digital options such as the iPHONE application called My Med ID*
5. *Additional parent and teen focused transition worksheets and resources in English and Spanish. http://internet.dscc.uic.edu/dsccroot/parents/transition.asp*

LysoSupport

As the National Fabry Disease Foundation (NFDF) enjoys our fifth year in operation, we have much greater clarity about our purpose and our programs than in the beginning. We have always been clear that our overall purpose is to serve and support families with Fabry disease. We just didn't have as clear an understanding of how we should and how we could accomplish our mission. Over time our ideals have remained constant, but our many ideas have evolved over time. We have learned a lot. We are enjoying our successes, learning from our challenges, and continuing to enjoy the process of finding out what programs work well on behalf of our community.

To begin with, we view the Fabry community as a conglomeration of not just families with Fabry disease, but many others. Our overall community consists of people with Fabry disease and our families; our physicians, researchers and clinical staff; and our supporters and friends. We all have a significant role in helping to improve and prolong the lives of people with Fabry disease.

The NFDF's support to the Fabry community takes many forms involving the following areas: Fabry disease education, Fabry disease identification, individual and family assistance and support; facilitation and support of Fabry disease research; and advocacy for Fabry disease initiatives. In this issue of LysoStories, we would like to update you on a couple of our main focus areas.

Fabry disease education:

While we are fortunate to have several hundred physicians who have taken an interest in Fabry disease and some who have become experts and key opinion leaders, there is a great need to educate physicians everywhere to recognize and diagnose Fabry disease. Thousands of people are believed to be living with the life-altering and life-threatening symptoms of Fabry disease who still don't know what is causing their illnesses. The NFDF has made the welfare of these yet unknown Fabry patients a top priority. For those of us who already know we have Fabry disease, we are fortunate enough to know why we are ill. Everyone deserves to know and to have a chance at a better life.

With the help of some of our Fabry physician experts, we are in the process of developing targeted physician education materials for nephrologists, neurologists, cardiologists and pediatricians. We will develop educational handouts for each specialty and participate in 2010 physician conferences to distribute and discuss them. We will do our best to expand this program to include all the many major symptoms of Fabry disease and every physician group who has an opportunity to diagnose our disease. Earlier diagnosis is critical to living better and longer lives.

Also, to increase our education efforts, members of our community with Fabry disease can volunteer to provide real-life examples of Fabry disease symptoms to physicians. As an example, people with Fabry disease can join the NFDF's "Connecting the Dots Campaign" by visiting or writing their local hospital or independent dermatology clinics. Patients with a good display of angiokeratoma (the tell-tale skin finding of Fabry disease) may print the "NFDF letter to dermatologists" located on our website at www.TheNFDF.org. It is located in the "Education tab" (top tabs), then the "Fabry Disease Dermatology Symptoms" sub-tab on the right side. Patients can fill-out the patient information sheet and take it or send it with the letter and other materials we provide to their nearby clinics.

Continued on page 4