February 2008

LysoStories™

A Publication from your Healthcare Advocates

What's New

The 10th International Mucopolysaccharide & Related Diseases Symposium is being held in Vancouver, BC June 26th – June 29th 2008

For more information please see the website www.mpssymposium2008.com

Center News

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Welcome

We hope that your enjoy reading this 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 Publications Committee.

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Patient Story

Living with MPS I

By Jennifer Prince

When you hear of MPS1, most people think about something technical or just give you a look of confusion, so I spare them the full pronunciation "Mucopolysaccharidosis." This means our body is missing an enzyme used to breakdown complex carbohydrates, so since our body doesn't break them down they get stored and cause problems. MPS 1 is such a rare disease, most people have never heard of it. It affects every organ in our body as well as our physical appearance. My sister and I both have it and we found this out when I was 8 and my sister was 5. It has been tough trying to keep up with our friends. There are many times when we have to ask for help doing the simplest things that most people take for granted, like tying your shoes or brushing your hair. We also spend many days in doctor's offices waiting to be called.

It all started when I failed the eye exam at school. My mom took us to Sears to get checked out and then from there we were sent from one doctor to another not knowing what was going on or what was causing my vision problem. One day, after waiting for the doctor to come back into the room after several minutes of wondering why he was in the hall talking to about four other Doctors, he says you have a very rare disease. I didn't really understand what that meant, but I knew it was bad. After that day, our lives were forever changed.

Because our disease was so rare, the doctor asked if we wouldn't mind if some of other doctors in the same field could come and look at our eyes. Of course, we didn't mind because what would we do without Science. Little did I know when we walked into the exam room there were about 30 doctors sitting and waiting to look at mine and my sister's eyes. It was pretty cool actually!

This disease doesn't have a cure from what I have been told and can affect how long I live, which is scary. My parents found out about a clinical trial being done in North Carolina at UNC, so we flew there to see if we could be included. We were approved and began to receive the enzyme our bodies were missing. Well, actually my sister got the real thing and I got saline for 8 months because no one could know who was getting it for the clinical trial, but we notice right away with my sister and she started to feel better. After the 8 months, we both got the real drug and Emory Genetics Department was able to work with the pharmaceutical company to complete the trial at home. I missed flying but it was nice to be back home. After several years, with our help the drug Aldurazyme was approved for all the children with MPS 1 to start receiving. This won't cure our disease but it will slow down the progression.

Having this genetic disease has allowed me to be able to do some awesome things, like going on a Make A Wish trip to Disney World and going on the Disney Cruise, and meeting some famous people and not so famous people but all wonderful just the same. I even got to meet the Atlanta Falcons, Atlanta Thrashers and ride the Zamboni… that was great! But, nothing was as exciting as my trip to Switzerland. This was my Graduation present from my parents. They were nervous, to let me go to another country without them but my Cousin lives there so they knew I would be OK. She is also a Nurse so that made it

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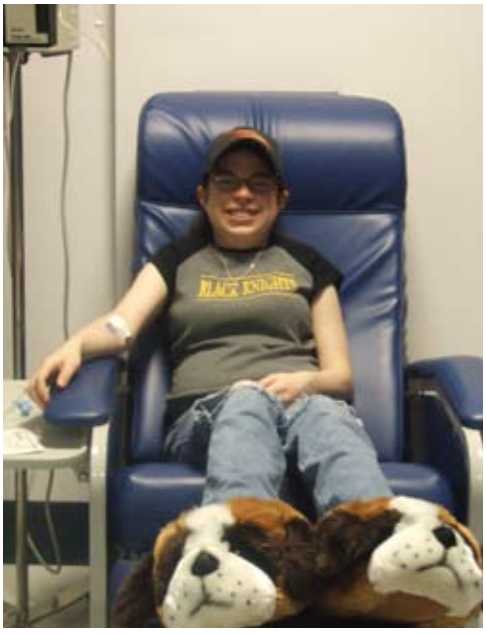

even better. My Cousin, Sherry took me to see some amazing things like "The Matahorn", Lake Geneva and Germany. I have never seen so much snow and art. I even walked around Zurich by myself and felt so grown up and safe. I couldn't believe I could do all that walking and not get tired. I couldn't do that before receiving the enzyme.

The best thing I think about having this disease is it has helped me to be a stronger person and not be afraid to share my experiences with others so that maybe I can help the others that can't help themselves. In early November 2006, I was the only "kid" to speak at the Children's Healthcare Rally in Atlanta. I was so scared, but I knew I had to do it. I stood there shivering in the cold and gave the best speech I could have ever imagined.

So, just because someone has a disease or some kind of disability does not mean they can't excel in many things. I have started college and plan to major in Art and Business. I would like to one day be a Photo Journalist. So, I like to say, "Children don't have disabilities, they have different abilities!"

Managing MPS I Disease: Monitoring Your Body from Head to Toe

By Heather Clark, MS, CGC
and Ellen DeVincentis, MS, RD, CDN

MPS I Background
Mucopolysaccharidosis Type I disease (MPS I) is one of many types of lysosomal storage diseases (LSDs). This autosomal recessive condition is caused by the accumulation of a particular substance known as glycosaminoglycans (GAGs) in the lysosomes of cells causing problems with bodily organs and tissues.

Within the diagnosis of MPS I there are three subtypes of the condition based on the severity of disease symptoms. Only a trained specialist in medical genetics can determine into which subtype the patient 'fits.' The three subtypes are identified as Hurler syndrome, Hurler-Scheie syndrome, and Scheie syndrome. All three of these syndromes are the result of an absence, or insufficient levels, of one particular enzyme called alpha-L-iduronidase. Hurler syndrome is the most severe of the MPS I subtypes. Children born with Hurler syndrome are usually recognized in the first few years of life and their cognitive development slows between ages 2 to 4 years. There are many physical abnormalities seen in individuals with MPS I including hernias, delayed growth or short stature, joint stiffness, hearing problems, distinct facial features, eye problems, heart problems, enlarged organs (liver, spleen, heart) and breathing problems. Hurler-Scheie syndrome is less severe than Hurler syndrome and may not be recognized until ages 3 to 8 years. Children with this subtype may have mild to moderate learning difficulties or mental retardation and many of the other physical characteristics of Hurler syndrome. Scheie syndrome is the mildest form of MPS I, with the diagnosis often being made after age 10 years. Patients with Scheie syndrome may have normal intelligence or mild learning disabilities. They may also have some of the other physical problems listed previously.

How can we monitor MPS I disease?
Once a person has been diagnosed with MPS I, their doctor may order a number of tests and evaluations. The purpose of these multiple tests and evaluations are to help determine the extent of the disease and the best possible treatments and therapies. Because MPS I affects multiple systems within the body, the initial assessments are varied and assess the body from "head to toe."

The MPS I Registry has developed a minimum schedule of assessments for monitoring patients with MPS I disease (www.MPSIRegistry.com). A physical examination with a review of medical history and vitals assessment (height, weight, head circumference, blood pressure) is recommended every six months. Additional assessments include:

- **BRAIN**: MRI every other year (to screen for excess fluid in the brain, known as hydrocephaly).
- **SPINE**: MRI every other year (to screen for narrowing of the spine , known as stenosis).
- **EYES**: Yearly ophthalmology exam (to check vision & screen for corneal clouding).
- **EARS, NOSE & THROAT**: Yearly audiology exam (to check hearing). Yearly ENT exam (to check for ear tubes, tonsils & adenoids, and evaluate airway).
- **MOUTH**: Yearly dental exam (to screen for dental disease).
- **HEART**: Echocardiogram and EKG every other year (to screen for heart & valve problems).
- **LUNGS**: Annual pulmonary function tests (to check for breathing problems). Sleep study every other year (to screen for sleep apnea).
- **ABDOMEN**: MRI every other year (to measure spleen & liver volumes).
- **URINARY**: Urinalysis and GAG screen every 6 months (to measure GAG levels).
- **BONES**: Skeletal survey X-ray every other year (to check for bone problems).
- **HANDS**: Nerve conduction velocity tests every other year (to check for carpal tunnel syndrome).
- **LEGS & FEET**: Yearly orthopedic exam (to check for joint problems). Physical therapy as needed
- **BLOOD**: For patients on enzyme therapy, antibody testing every 3 months to 1 year.

Why are all these tests important?
Your doctor will use some the results from these tests to determine how the MPS I disease is affecting your body. By performing baseline tests and comparing them to future assessments, the doctor is better able to assess the stage or degree of the disease and also track improvement or disease progression.

Treatment
Although there is no cure for MPS I, there is a treatment available consisting of an enzyme replacement therapy (ERT). This ERT is called Aldurazyme and is administered through an intravenous infusion once every week. The goal of this type of treatment is to stabilize, prevent, or delay the onset of MPS I symptoms and problems. In addition to ERT, many patients will continue to receive adjunctive treatments such as physical therapy, occupational therapy, and speech therapy.

Helpful Hints and Recommendations
MPS I is a complex disease that requires individual assessments and management plans. The timing of these MPS I Registry assessment recommendations are only generalized suggestions. Your physician may decide to order these tests and evaluations more often or less frequently, depending on a person's individual symptoms. It may be helpful for the patient with MPS I to identify one particular physician who has experience with this condition to coordinate all the testing and evaluations. If the MPS patient makes the commitment to a treatment plan, he or she must be an active participant in the care plan. Communication and compliance are necessary for a good outcome in the management of MPS I.

Resources

- National MPS Society www.mpssociety.org
- MPS I Registry www.MPSIRegistry.com
- Genzyme MPS I info www.MPSIDisease.com

About the authors
Heather Clark is a genetic counselor and clinical coordinator at the Lysosomal Storage Disease Center at Emory University in Atlanta, GA. She works with patients and families with MPS I and related disorders. Heather can be reached via email at hclark@genetics.emory.edu or by phone at 404-778-8565.
Ellen DeVincentis is a metabolic nutritionist and coordinator for the Metabolism Program at The Women & Children's Hospital in Buffalo, NY. She works with patients and families with MPSI, Fabry disease, Pompe disease and Gaucher disease. Ellen can be reached via email at edevincentis@upa.chob.edu or by phone at 716-878-7496.

The Road to Treatment: Navigating the complexities of the insurance system

By CeeCee Fairley and Cindy Ochs

For most people with a lysosomal storage disorder, the road to diagnosis and treatment is long and confusing. With the first symptoms begins a long journey through the medical system; it often requires visits to many doctors before a diagnosis is reached. However, once this occurs and enzyme replacement therapy is recommended, a new journey begins—this time, patients and families face the maze of the insurance system.

This system is complex. There are many types of insurance coverage and plans, including: private, group or employer, state, and national medical plans. This is confusing enough. Adding to the challenge is the fact that current recommendations suggest lifelong enzyme replacement therapy—and medication is expensive. Understandably, this can cause significant concern for those with a lysosomal storage disorder and their families. This article will provide information that will help you navigate the complexities of the insurance system.

There are some facts of which you may be unaware. An estimated 79% of patients currently treated with Cerezyme for Gaucher disease have no lifetime maximum in their insurance policies. A "lifetime maximum" is the maximum amount of money an insurance company will pay in benefits, or medical expenses, to a person covered by the plan during his or her lifetime. Neither Medicaid nor Medicare include lifetime maximums.

Patient support groups may be able to provide financial assistance to those who qualify. For Gaucher disease, the National Gaucher Foundation should be contacted. For Fabry, MPS I, and Pompe disease, Patient Support Incorporated should be contacted. The information for disease specific patient support groups and financial assistance is located at the end of the article.

Genzyme Treatment Support (GTS) is a service that is available to patients with Fabry, Gaucher, MPS I, and Pompe disease. GTS is a free service provided by the Genzyme Corporation; it provides confidential one-on-one insurance counseling to patients and their families who are insured by a United States health insurance policy. Those interested in using this service can call 1-800-745-4447 to speak to a case management specialist. GTS case managers are nurses, social workers, or other health care professionals who have expertise in reimbursement, insurance, and the health care delivery system. A GTS case manager can review your current insurance coverage and, if necessary, try to obtain and maintain the insurance coverage you may need for long-term enzyme replacement therapy. This service is voluntary and any information shared with your case manager will remain confidential.

A case management specialist may assist in many ways, including:

- Educating third-party payers about your lysosomal storage disease and the benefits of enzyme replacement therapy.
- Providing individuals with information about available health insurance and helping them to review their current policies.
- Coordinating the exchange of information between physicians, third-party payers, and patients to obtain coverage approval for enzyme replacement therapy.
- Serving as a resource to those who are seeking alternative funding options.

Following is an example of how one GTS case manager helped a family whose child was reaching his lifetime maximum.

A 12-year-old child was approaching his one-million-dollar lifetime maximum. The father worked for a small company that didn't offer health insurance and the mother was unemployed. However, the child did not qualify for state-funded programs such as Medicaid because the family income was over the eligibility limit. The family's own private insurance company offered a one-time increase of the lifetime maximum up to $100,000 per year for 10 years. Was this their best or only option? Anxious, and hoping for guidance, the parents called GTS for help.

After discussing the situation, the GTS case manager did some research and identified a possible alternative plan with a different insurance company. The child's parents requested information from the company, and, upon its arrival they reviewed it with the case manager. As a follow-up, the case manager called the insurer to receive specific information about how the child's treatment would be covered if he was enrolled in this plan. The plan, an HMO, covered home infusions with no co-pay as long as the home care agency obtained preauthorization. In addition, the plan had no lifetime maximum. This came as a relief to the parents, who ultimately chose to enroll their child in the plan.

This is just one example of how a GTS case manager might help a family to better navigate the insurance system. Because of the complexities of the system, each family's situation requires individual assessment. Outcomes, including obtaining insurance, are not guaranteed. However, GTS can research individual options and alternatives in an effort to find the best possible choices. To discuss coverage with a GTS case manager, call 800-745-4447.

Patient Support Groups

Gaucher Disease
National Gaucher Foundation
www.gaucherdisease.org

Financial assistance may be available through the Care and Care+Plus program. For those who qualify, these programs may be able to provide financial aid to people with Gaucher disease. Information about these programs and how to apply is available by calling Barbara Lichtenstein at 866-346-8176 or 1-301-963-4489. It is also available at www.gaucherdisease.org (click the Gaucher disease tab and then Support programs). Information on this topic can also be found at www.gauchercare.com.

Fabry Disease
Fabry Support and Information Group (FSIG)
www.fabry.org

Financial assistance may be available through Patient Services Incorporated: contact 804-744-3813 or www.uneedpsi.org

MPS I Disease
National MPS Society
www.mpssociety.org
1-919-806-0101

Financial assistance may be available through Patient Services Incorporated: contact 804-744-3813 or www.uneedpsi.org

Pompe Disease
United Pompe Foundation
www.unitedpompe.com
1-559-227-1898

Financial assistance may be available through Patient Services Incorporated: contact 804-744-3813 or www.uneedpsi.org

Editors' Note
Insurance case management programs are available at other pharmaceutical companies and are listed below.

BioMarin Pharmaceuticals (Naglazyme for MPS VI)
BioMarin Patient and Physician Support (BPPS)
866-906-6100, Option 1 for Naglazyme
e-mail: bpps@bmrn.com

Shire (Elaprase for MPS II)
OnePath℠ Support Services
1-866-888-0660. Case managers are available Monday through Friday, 8:30 a.m. to 8:00 p.m. Eastern Time.
http://www.elaprase.com/patients_families/support/services/

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Join the NGF in a Live Online Gaucher Meeting

By Sharon Adams, Director of Meetings and Support Services

The National Gaucher Foundation (NGF) has made it possible for anyone with a computer to be able to attend Gaucher meetings – online! Beginning in 2006, the NGF began hosting live online Gaucher meetings through a service called WebEx. Through the miracle of technology, WebEx allows anyone to view, listen and participate in a meeting, without ever leaving the comfort of their home or office. All you need is a computer to view the presentations and a telephone line to listen to the audio portion of the presentations. You can even submit your questions online to the speaker for him or her to address during the meeting. If you don't have access to a computer, you can call in and listen to the audio portion if you'd like.

The NGF emails invitations out with easy instructions on how to join in the meeting. Instructions are also posted on the NGF's website listed below. You may think that you have to be computer savvy to participate but, in fact, it's very simple to do! Approximately 10 -15 minutes prior to the start of a meeting, you will click on the web address provided in the invitation. You will see instructions on your screen that will easily walk you through the set-up process. You will need to have the Meeting ID number and passcode which are listed on the invitation.

Once you are logged into the meeting on your computer, you will need to call a toll free number (also provided) to access the audio portion of the meeting. It's that easy!

The NGF, through WebEx meetings, provides people the opportunity to learn the latest developments in research and treatment options for Gaucher disease, without ever having to leave their homes. The NGF encourages you to visit its website regularly to stay up to date on upcoming meetings and events. Visit us at www.gaucherdisease.org today!

Here is a list of archived patient meetings on Gaucher disease. They can be seen on-line via WebEx by copying and pasting the web address into your browser.

Risks to health, current management options and future perspectives
Web Meeting
Speaker:
Gregory M. Pastores, M.D.
http://www.gaucherdisease.org/video15.php

So You Think You Know Everything About Gaucher Disease
Web Meeting
Emory University
Speakers:
Paul M. Fernhoff, MD, FAAP, FACMG
Karen Grinzaid, MS, CGC
http://www.gaucherdisease.org/video12.php

Moving from Childhood To Adulthood With Gaucher Disease
Web Meeting
Presentation by:
Nadene Henderson, MS, CGC, University of Pittsburgh
David Finegold, MD
http://www.gaucherdisease.org/video14.php