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 WebEx meetings through a company 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 meeting. If you don't have access to a computer, you can call in and listen to the audio portion of the meeting. You will need to have the Meeting ID number and passcode which are in the invitation. You will see instructions on your screen that will easily walk you through the set-up process. You may think that you can even submit your questions online to the speaker for him or her to address during the meeting. If you have questions about attending WebEx meetings, please contact the Meetings Department at NGF.

By Sharon Adams, M.S., CGC

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 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 noticed right away with my sister and she started to feel better. After the 8 months, we both got the real drug and notice right away with my sister and she started to feel better. 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. I have 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 doctors' offices waiting to be called.

For more information please see the website www.mpsysymposium2008.com

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

- Karen Ginzrau, MS, CGC
  Emory University School of Medicine
  (404) 778-8516
  kginzrau@genetics.emory.edu

- Nadene Henderson, MS, CGC
  University of Pittsburgh
  (412) 734-1554
  etn.henderson@pitt.edu

- Erin O’Rearte, MS, CGC
  Genzyme
  (412) 734-1554
  ginerva.mellum@genzyme.com

- Rita Paud, BNC, BSN
  Saint Peter’s University Hospital
  (732) 745-6611
  rypad@steadmanruth.com

- Lisa Sondheimer King, M.S., CGC
  University of Washington
  (206) 987-1406
  ksking@uw.edu

(continued on page 2)
The MPS I Registry has developed a minimum schedule of assessments for monitoring the extent of the disease and the best possible treatments and therapies. Because MPS I and evaluations. The purpose of these multiple tests and evaluations are to help determine to moderate learning difficulties or mental retardation and many of the other physical and breathing problems. Hurler-Scheie syndrome is less severe than Hurler syndrome and are usually recognized in the first few years of life and their cognitive development slows. All three of these syndromes are the result of an disease symptoms. Only a trained specialist in medical genetics can determine into which diseases (LSDs). This autosomal recessive condition is caused by the accumulation of a Mucopolysaccharidosis Type I disease (MPS I) is one of many types of lysosomal storage MPS I Background

Mucopolysaccharidosis Type I disease (MPS I) is one of many types of lysosomal storage disorders. This autosomal recessive condition is caused by an enzyme deficiency that results in the accumulation of a particular substance known as glycosaminoglycans (GAGs) in the lysosomes of cells caused problems with healthy 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. These 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 does not progress beyond age 2 to 4 years.

There are many physical abnormalities seen in individuals with MPS I including bulging eyes, deformed hands or fingers, sleep apnea, deviated nasal septum, distinctive facial features, eye problems, heart problems, enlarged liver (giant 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 normal intelligence 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 difficulties. They may also have some of the other physical problems listed previously.

How can we monitor MPS I disease? Once a patient 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.MPSRegistry.com). A physical examination with a review of medical history and vital assessments (height, weight, blood pressure, red blood count) is recommended every six months. Additional assessments include:

- **BRAIN:** MRI every other year (to screen for excess fluid in the brain, known as hydrocephalus).
- **SPINE:** MRI every other year (to screen for narrowing of the spine, known as kyphosis).
- **EYES:** Ophthalmologic exam (to check vision and for corneal edema).
- **EARS, NOSE & THROAT:** Ear, nose, and throat exam (to check hearing).
- **MOUTH:** Yearly dental exam (to screen for dental disease).
- **HEART:** Echocardiogram and ECG every other year (to screen for heart valve problems).
- **LUNGS:** Annual pulmonary function tests (to screen for breathing problems).
- **STOMACH:** Upper GI exam (to screen for ulcers).
- **ADRENALS:** MRI every other year (to screen for adrenal tumors).
- **URETAN:** Urology and GAG screen every 4 months (to measure GAG levels).

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 signs of a lysosomal storage disorder, it is often frightening to read so many different disorders in one day. After learning that this occurs once 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 Cerazyme for Gaucher disease have no lifetime maximum in their insurance plans. 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 this article.

About the authors

CeeCee 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. Her contact can be reached via email at mccvee@emory.edu or by phone at 404-778-8565.

Cindy Ochs is a medical writer based in Miami, Florida. Prior to becoming a writer, she was employed by the medical writing division of a large pharmaceutical company where she specialized in preparing patient information on a variety of conditions. Cindy holds a bachelor’s degree in Molecular Biology from the University of Miami and a master’s degree in Science Communications from the University of Florida. She can be reached via email at cindy@cochs.com.
The MPS I Registry has developed a minimum schedule of assessments for monitoring a disease that affects multiple systems within the body, the initial assessments are varied and assess the extent of the disease and the best possible treatments and therapies. Because MPS I, which is caused by the absence or insufficient levels of one particular enzyme called alpha-L-iduronidase, is usually recognized in the first few years of life and their cognitive development does not progress as normal. Therefore, the MPS I Registry assessment recommendations are only generalized guidelines. Your doctor will use some of these results to determine how the MPS I disease is affecting your child. 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.

Why are all these tests important?
Your doctor will use some of these results to determine how the MPS I disease is affecting your child. 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.

Treatments
Although there is no cure for MPS I, there is a treatment available consisting of an enzyme replacement therapy (ERT). The ERT is called Alkazylase 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 general guidelines. Your physician may decide to order these tests and evaluations more often or less often, depending on a patient’s individual symptoms. It may be helpful for the parent of an MPS I patient to identify one particular physician who has experience with this condition to coordinate the testing and evaluations. If the patient’s physician makes a treatment commitment to a patient, 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.

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.

Heather Clark
Genzyme MPS I info
www.MPSIDisease.com

userManager@genzyme
www.mpssociety.org

BioMarin Patient and Physician Support (BPPS)
www.BioMarin.com
1-866-506-8680 or www.uneedpsi.org

Shire (Elaprase for MPS II)
www.shire.com
1-888-888-0660

MediMutual
www.medi-mutual.com

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

Burlington Pharmaceuticals (Naglazyme for MPS VI)
www.burlington.com
1-800-744-3813 or www.uneedpsi.org

CPhA
www.cpha.com

Shire
www.shire.com
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 presentations on WebEx. 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 emailed invitations out with easy instructions on how to join in the meeting. Instructions are also posted on the NGF’s website. You can visit the website at www.gafound.org/services to watch presentations and see an archive of previous meetings. You can also call the NGF at 1-800-334-7980.

By Sharon Adams, Director of Meetings and Support Services

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. For more information please see the website www.mps2meeting2008.com.

What’s New
The 10th International Mucopolysaccharidosis & Related Diseases Symposium is being held in Vancouver, BC June 26th – June 29th 2008.

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

Welcome
We hope that you 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.

• Karen Ginzaid, MS, CGC
Emory University School of Medicine
(404) 778-8516
kgrinzaid@genetics.emory.edu

• Nadene Henderson, MS, GOG
University of Pittsburgh
(800) 556-7900
nadene.henderson@hcn.pitt.edu

• Erin O’Reilly, MS, CGC
Genzyme
(412) 734-1554
erin.o'reilly@genzyme.com

• Nita Paul, BNC, BSN
Saint Peter’s University Hospital
(732) 745-6661
npaul@saintpetersuh.com

• Nita Patel, RNC, BSN
Saint Peter’s University Hospital
(732) 745-6661
npatel@saintpetersuh.com

• Lisa Sniderman King, M.S., CGC
University of Washington
(206) 987-1406
lsking@washington.edu

Living with MPS I
By Jennifer Prince

When you hear MPS I, 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 I 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 thing 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 20 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 noticed right away with my sister and the drastic change 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 a home. I missed flying but it was nice to be back home. After several years, with our help the drug Alkeran was approved for all the children with MPS I 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… those are the best! It 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 made it easier. In 2006, the NGF began hosting presentations on WebEx. 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 emailed invitations out with easy instructions on how to join in the meeting. Instructions are also posted on the NGF’s website. You can visit the website at www.gafound.org/services to watch presentations and see an archive of previous meetings. You can also call the NGF at 1-800-334-7980.

by Sharon Adams, Director of Meetings and Support Services

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 noticed right away with my sister and the drastic change 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 a home. I missed flying but it was nice to be back home. After several years, with our help the drug Alkeran was approved for all the children with MPS I 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… those are the best! It 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 made it easier.

In 2006, the NGF began hosting presentations on WebEx. 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 emailed invitations out with easy instructions on how to join in the meeting. Instructions are also posted on the NGF’s website. You can visit the website at www.gafound.org/services to watch presentations and see an archive of previous meetings. You can also call the NGF at 1-800-334-7980.