The Emory Lysosomal Storage Disease Center (LSDC) Clinic is now held on the first Friday of every month. This specialized clinic was created to serve the individual needs of patients and families with lysosomal storage diseases. Services provided in or through the clinic include: review of medical history, physical examination, laboratory studies, imaging studies, discussion of treatment options, assessment of ongoing treatment plans, genetic counseling, and other services as needed. Having a dedicated clinic for LSD's allows patients to meet other individuals with similar diagnoses who are dealing with some of the same health-related issues and concerns.

To schedule an appointment at the Emory LSDC Clinic, call (404) 727-3930 or 1-800-200-1524.

WHAT ARE LYSOSOMAL DISEASES?

The Lysosomal Storage Diseases are a group of conditions in which certain substances (substrates) build up in compartments of the body's cells called lysosomes. These conditions are caused by missing or poorly functioning enzymes important in the breakdown of these substances. Over time, excessive amounts of the substrates accumulate and cause damage to involved systems and organs in the body.

NEW EMORY LSDC BROCHURE

Our Lysosomal Storage Disease Center Brochure is now ready for mailing. All of our families and referring physicians will receive copies of the brochure in the near future. Please contact us if you need additional copies or if you know of others who would be interested in learning more about our center.

OUR TREATMENT TEAM

Paul M. Fernhoff, MD, FAAP, FACMG
Karen A. Grinzaid, MS, CGC
Eleanor Geller Botha, MS, CGC
Dawn J. Laney, MS, CGC

CONTACT US:
Email: LSDC@genetics.emory.edu
Phone: 404-712-8438 or 1-800-200-1524
Fabry disease is a progressive, destructive, and life-threatening lysosomal storage disorder caused by the partial or complete deficiency of the enzyme a-galactosidase A.

Fabry disease is a rare inherited X-linked disorder which predominantly affects males, but carrier females can also be affected to a mild or severe degree because of X-chromosomal inactivation.

Deficiency of a-galactosidase A leads to accumulation of glycosphingolipids (particularly globotriaosylceramide (GL-3)) in visceral tissues and the vascular endothelium throughout the body, leading to episodic crisis of visceral tissues and the vascular system. Without treatment for uremia, the average lifespan for hemizygotes with Fabry disease is 41 years.

With the advent of renal dialysis or transplantation, the median survival is about 50 years.

Although Fabry disease usually presents in childhood or adolescence, it is often not diagnosed until adulthood.

Early diagnosis and intervention are critical, since organ system damage is progressive and can be irreversible or even life-threatening. Varied presentations and diffuse symptoms often hamper an accurate and timely diagnosis.

Diagnosis of Fabry disease is confirmed by low or absent a-galactosidase A activity in plasma or serum, leukocytes, tears, biopsied tissues, or cultured skin fibroblasts. Since female heterozygotes can have a wide range of enzymatic activity, mutation or DNA analysis may be necessary to establish carrier status.

For additional information on Fabry disease call Genzyme Medical Information at 1 (800) 745-4447 to locate a Lysosomal Storage Disease Center nearest you.
We are the parents of 2 children with mucopolysaccharidosis (MPS I), more commonly referred to as Hurler, Hurler-Scheie, or Scheie syndrome. MPS I disease is a rare, inherited, lysosomal storage disorder caused by the deficiency of the lysosomal enzyme alpha-L-iduronidase. Deficiency of this enzyme results in the progressive accumulation of non-degraded material (called glycosaminoglycans, or GAG) in cells throughout the body. Here is our story....

In 1984, my wife Wanda and I were blessed with a baby boy, Michael, and with a baby girl named Ashley in 1987. For 5 years everything was normal in our family. One day in 1989, a daycare provider asked a question that would change our lives forever, "Mrs. Frix, did you know your son can't raise his arms above his head?" It was also brought to our attention that Michael couldn't do jumping jacks. The daycare provider thought he was just goofing around when in fact there was a problem far more serious then we could even imagine. Immediately, our first response was of course he can. We checked Michael that evening and his elbows would only come up to just under his eyes. There was something blocking his arms from going above his head.

We returned to the United States where orthopedics diagnosed her with Hurler-Scheie Syndrome by Dr. Curtis Rogers.

For the most part, as Michael’s progress, we noticed problems similar to Michael's, it never occurred to us that she could be affected with the same syndrome as Michael. Several doctors diagnosed her with "growing pains".

After learning about Michael and Ashley’s syndrome, we started noticing more things that they were unable to do. We had been questioning why, why, why, after seeing so many doctors. Now all the puzzle pieces were beginning to fit together.

Michael was born with hernias which were identified at 2 weeks of age. He had surgery to repair the hernias when he was one month old. He also had 4 sets of tubes inserted in his ears as well as another hernia repair in his stomach area. We thought these were just normal problems, but Ashley had the same surgeries as well. Michael's joints progressively got stiffer.

Mrs. Frix, did you know your son can't raise his arms above his head?

Ashley’s disease had progressed, for the most part, as Michael’s progressed. However, Ashley had mucopolysaccharide storage in areas different from Michael. In October 2001, Ashley was seen by her eye doctor and referred to a neurologist because of fluid collecting in her head. She had a VP Shunt placed to correct that problem. In March 2002, when she went to have braces put on her teeth, we were told her jaws had not developed normally. Her jaws were the size of a 5 year old. Ashley was 15 years old. Soon after, Ashley started losing her balance and falling for no apparent reason. After extensive testing by her neurosurgeon, she was diagnosed as having storage build up on her spinal cord; which was clamping down and causing her to lose her balance. In 2003, she had spine surgery to relieve this build up on her spine. Ashley had always had breathing problems due to mucopolysaccharide storage in her lung tissue. She was low on energy and felt bad much of the time.

Ashley’s progress, as a result of this treatment, is remarkable to say the least. Her breathing and lung capacity has increased. She is more limber, she doesn’t hurt as much, and she feels all-around better than she has in so many years.

We thank all the folks that had a part in the development of Aldurazyme. While it isn’t a cure for MPS I, it is a great help to the quality of life for Ashley. She can now brush the back of her hair for the first time in her life, and she can go to the mall and shop from end to end.

We hope that after reading Michael and Ashley’s story you are more aware of MPS I disease. For additional information regarding MPS I disease signs and symptoms, please call the Lysosomal Storage Disease Center at Emory University at 800-200-1524 or Genzyme Medical Information at 1-800-745-4447.

**OUR STORY**

Ashley had the same surgeries as well. Michael's disease was most noticeable in his knees; he had a "space-walk" type of walk. Ashley's disease was most noticeable in her hands and fingers. She also had stiffness in her joints; her arms were as limited as Michael's and could not go above her head.

We noticed other limitations: our children loved to play ball however, they could not throw a ball up in the air. Their shoulder and arm rotation was such that they couldn't get any arc on anything they threw overhand. They loved playing in the band; however, their stiff joints and inability to get enough air in their lungs, led to them having to drop out of the band. Breathing, hearing, and vision were all affected. Michael wore hearing aids for some of his early years and his vision was affected with corneal clouding.

As Michael grew older, we needed to watch for other complications of Hurler-Scheie syndrome such as heart disease due to mucopolysaccharide storage in the heart valves. Michael started having seizures in 1998, and on August 22, 1998 he had his final seizure at the Atlanta Braves baseball game. Michael's heart got off rhythm. He died on August 22, 1998 of cardiac arrhythmias.

Ashley’s arms, legs and fingers would no longer straighten. It seemed the older the children got, the stiffer their joints became. Michael’s disease was most noticeable in his knees; he had a "space-walk" type of walk. Ashley's disease was most noticeable in her hands and fingers. She also had stiffness in her joints; her arms were as limited as Michael's and could not go above her head.

Her jaws were the size of a 5 year old, she was 15 years old. Her arms and fingers would no longer straighten. It seemed the older the children got, the stiffer their joints became. Michael’s disease was most noticeable in his knees; he had a "space-walk" type of walk. Ashley's disease was most noticeable in her hands and fingers. She also had stiffness in her joints; her arms were as limited as Michael's and could not go above her head.

We noticed other limitations: our children loved to play ball however, they could not throw a ball up in the air. Their shoulder and arm rotation was such that they couldn't get any arc on anything they threw overhand. They loved playing in the band; however, their stiff joints and inability to get enough air in their lungs, led to them having to drop out of the band. Breathing, hearing, and vision were all affected. Michael wore hearing aids for some of his early years and his vision was affected with corneal clouding.

As Michael grew older, we needed to watch for other complications of Hurler-Scheie syndrome such as heart disease due to mucopolysaccharide storage in the heart valves. Michael started having seizures in 1998, and on August 22, 1998 he had his final seizure at the Atlanta Braves baseball game. Michael's heart got off rhythm. He died on August 22, 1998 of cardiac arrhythmias.

He gave us contacts with Genzyme and through much work on both our parts Ashley started taking Aldurazyme, enzyme replacement therapy, in October 2003 at Emory University.

Ashley’s progress, as a result of this treatment, is remarkable to say the least. Her breathing and lung capacity has increased. She is more limber, she doesn’t hurt as much, and she feels all-around better than she has in so many years.

We thank all the folks that had a part in the development of Aldurazyme. While it isn’t a cure for MPS I, it is a great help to the quality of life for Ashley. She can now brush the back of her hair for the first time in her life, and she can go to the mall and shop from end to end.

We hope that after reading Michael and Ashley’s story you are more aware of MPS I disease. For additional information regarding MPS I disease signs and symptoms, please call the Lysosomal Storage Disease Center at Emory University at 800-200-1524 or Genzyme Medical Information at 1-800-745-4447.

**MPS I**

**MAJOR SIGNS & SYMPTOMS**

- Joint stiffness
- Skeletal deformities
- Coarse facial features
- Enlarged liver and spleen
- Corneal clouding
- Obstructive airway disease
- Recurrent ear and nose infections
- Valvular heart disease
- Inguinal/umbilical hernias

**Resources**

www.mpsl.com
www.mpslregistry.com
FACTS ABOUT THE REGISTRY

LYSOSOMAL STORAGE DISEASE REGISTRIES

A COLLECTIVE RESOURCE TO OPTIMIZE OUTCOMES

The Registry provides physicians access to a resource that can help better manage patients.

• Monitor disease progression through patient specific reports
• Exchange clinical data among participating physicians to facilitate clinical decision-making
• Access information on current treatment guidelines and practice patterns
• Potentially improve patient outcomes through information and collaboration

THE REGISTRY BENEFITS EVERYONE - DO YOUR PART!

COMING SOON

WHAT’S IN STORE in Future Issues...

► You and the Registry
► Pompe Disease
► Clinical Trial Update
► Your Case Manager and You

Special thanks to the National Gaucher Foundation for making this newsletter possible.

HAVE YOU SEEN THESE?

Joint deformity and organomegaly: symptoms that can be found in MPS I disease.
(courtesy of Nat. MPS Soc., Inc.)

EMORY UNIVERSITY SCHOOL OF MEDICINE

Have you seen these lately?