This newsletter is sponsored by Genzyme Corporation.

LysoStories™
A Publication from your Healthcare Advocates

(continued from page 3)

1. Parent Participation in Meetings:
To establish and maintain an effective team atmosphere be positive.

- Don't allow your emotions to interfere; be factual, prepared, and ask for clarification.
- Know the philosophy of your principal. Does he/she truly believe all children can learn?
- Educate the team on LSDs. This will give them more information to plan your child's educational program.
- Learn about tests and measurements.
- Dress professionally.
- Gather all your child's medical and educational records that are pertinent to the meeting. Review your child's records. What are their strengths and weaknesses, what do you want them to achieve this year, what are your long-term goals for your child, how does your child's disability affect his or her ability to learn?
- Write down questions and concerns.

2. During the Meeting:

- Sit next to the administrator or the person who will ultimately make the decisions.
- Ask questions, define and describe issues, problems, and offer solutions.
- Stay on task.
- Learn to be a negotiator. Prioritize important issues.
- Take notes. If it is not written down, it never happened!
- If at all possible both parents should attend the meeting and present a unified front. If not take a friend or advocate so you don't feel vulnerable and can process the meeting together afterwards.
- Do not sign the IEP at the meeting if unsure. Take it home and read it.

3. After the Meeting:

- Add documents from the conference to your files.
- Send a follow-up letter of your recollections about what happened and a brief thank you letter. Include your understanding of what the school agreed to, and issues that went unresolved.
- Keep written records, keep written records, oh, did I say keep written records.
- Take care of yourself or you will become exhausted and burn out. Pace yourself.

In closing I would like to extend encouragement to those families who have children with disabilities. Having a child with special health care needs means becoming his or her voice. Do not give up; there are many knowledgeable professional advocates that will assist you through this process and ultimately improve the quality of life for your child.

September 2007

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What's New

Take a look at the updated MPSI website at www.mpsldisease.com

Don't miss the National Gaucher Foundation Conference in Atlanta on October 14 and 15, 2007. This year's conference, "Science Evolving - Where It's Taking Us - A Look into the Future for Gaucher Disease" will be held at the Westin Peachtree Plaza, 210 Peachtree Street, Atlanta, Georgia. To register, visit the NGF website at www.gaucherdisease.org or email: rivers@gaucherdisease.org.

Center News

Lysosomal Storage Disease Center Emory University

Please join us for our upcoming Annual MPS Family Meeting on Sunday, September 23rd at Maggiano's Little Italy, 4400 Ashford Dunwoody Rd., Atlanta, GA 30346, phone 770-804-3313. (Note that this is the Perimeter Maggiano's, not the Buckhead location). The meeting will provide all local MPS patients and families an opportunity to visit with each other and share information. Please RSVP to Heather Clark at the Emory LSDC at 404-778-8536 or email hclark@genetics.emory.edu.

The LSDC group at Emory (left to right): Karen Grinzaid, Eleanor Botha, Dawn Laney, Dr. Paul Fernhoff, Heather Clark, Erin Rooney and infusion nurse Trudi Holbrook.

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.

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Patient Story

Invisible Disabilities
By Nathaniel Goldman
A 19 year old with Fabry disease

I wish I had a cast on my arm, so people would see it and ask, "How can I help?" My disabilities, however, are invisible. I have two challenges: a learning disability, which makes it difficult to read and write; and an enzyme deficiency, called Fabry, which makes it almost impossible to walk long distances, play sports, or be out on a hot day. These invisible disabilities have taught me to deal with many situations. I have learned about myself, about others, and about life in general.

These disabilities have taught me to take care of myself in several specific ways. First, I have to acknowledge my disabilities. When friends go on a hike, I must use good judgment and stay behind, even if I want to go. Admitting, especially to myself, that I cannot do what others do has been a hard lesson to learn. Second, I must be my own advocate. As Rabbi Hillel said, "If I am not for myself, who will be for me?" For example, I need to make arrangements for extended time to complete tests and assignments, and to plan ahead to obtain recorded tapes of assigned text books. Third, I need to communicate the effects of my disabilities to others. This is especially difficult because my limitations are not apparent to other people. When a coach, or teacher, or employer tells me to do some physical activity that I cannot do, I have to explain that I am unable to do it. If he or she says, "no excuses," I have to continue to explain and not be intimidated.

Having invisible disabilities also has affected how I relate to others. I know that I should not make judgments based only on what I see. I recently met the mother of a friend and she did not get off the couch to introduce herself. I might have thought she was being rude, but instead, I wondered if she had an invisible disability or other hidden reason that prevented her from getting up. Additionally, I have become very sensitive to people's feelings. I understand that everyone's pain is not visible, and that words and actions can cause wounds that are not seen.

I have also changed my philosophy about major concepts in life. I know from personal experience that something does not have to be seen to exist. Invisible does not mean nonexistent. This has helped me accept the existence of concepts that I believe are real, even though I cannot offer visible proof, like prejudice, love, hate, faith, or God.

My invisible disabilities have helped me look at life through more understanding eyes. I know who I am and what I must do for myself. I am careful not to prejudge others. I accept certain universal beliefs about life that can not be proven. Having disabilities that are not readily observed by others is not just a part of who I am, but has actually changed who I have become.

Identifying a Dragon

By Toby Director

"Why are you so invested in finding something wrong with your sons?" came the question from a close relative. "Something ***is*** wrong with them," I answered, "I'm just invested in trying to find out what it is." That determination finally paid off. After a long search, with several false detours, we finally did obtain the correct diagnosis. We could, then, begin to knowledgeably address the problem.

One of the difficulties with diagnosing Fabry Disease is that the symptoms send the patient to a variety of specialists who concentrate on one organ system and never see the whole picture; they are like the blind men who describe an animal by feeling only its trunk, its leg, its tusk, or its tail, and who never identify it as an elephant. In our case, however, we were dealing with a "dragon", the name patients have begun to use as a euphemism to refer to Fabry Disease.

We visited gastroenterologists to treat digestive problems, dermatologists to look at angiokeratomas, neurologists to analyze hand and foot numbness and burning, endocrinologists to do a work up for delayed growth, cardiologists to check out chest pain and heart murmurs, orthopedists to investigate foot and leg pains, geneticists to search for ethnic-related diseases, and a variety of doctors to help explain the lack of sweating, fatigue, depression, and anxiety. Two excellent physicians at Emory Children's Center, Dr. Lillian Meacham (endocrinology) and Dr. Bess Schoen (gastroenterology), stood at an important crossroad, and guided us in the right direction. They each took that significant step beyond treating the symptoms in their own specialty and insisted that we look further for the underlying cause.

The search finally led us to the Mayo Clinic. An admitting doctor looked at the angiokeratomas and said, "You need to see a dermatologist." Within several hours, the dermatologist examined my sons and said, "You need to see the pediatric-dermatologist." A short time later, the pediatric-dermatologist took another look and said, "You need to see the genetic-pediatric-dermatologist." When the genetic-pediatric-dermatologist finished her exam, she opened a book to a page with the title, Fabry Disease. On the page were listed about thirteen characteristics of Fabry Disease and about eleven of them described our personal family "dragon."

Of course, we followed up with skin biopsies and blood tests to confirm the diagnosis and eventually to identify our family's specific genetic mutation. But as soon as we read the description on that page, we knew the dragon was out of the bag (or book)!

A Simple Blood Test: Diagnosis of Fabry Disease

By Dawn Laney, MS, CGC

The situation described in Toby's story is not unusual. Many individuals affected by Fabry disease visit many specialists before a diagnosis of Fabry is considered. However, once the suspicion is raised, testing for Fabry disease is easy and only involves a blood test. When testing, a small sample of blood is drawn at a local physician's office or hospital and then sent to a qualified lab for testing. Initial testing in males usually requires measurement of the α-galactosidase enzyme which is deficient in Fabry disease. Testing in females usually requires two tests: the α-galactosidase enzyme test ***and*** the Fabry DNA test in which the Fabry gene (called GLA) is sequenced or studied in detail. This DNA sequencing test looks for "mutations" or changes in the GLA gene that cause Fabry disease. Results of testing are available 2-3 weeks from the time blood is received at the lab. Insurance usually covers the tests as long as the individual insurance precertification requirements have been met.

When one member of a family is diagnosed with Fabry disease, it is crucial to test other at-risk relatives. A recent multi-center study found that when an individual is diagnosed with Fabry disease, on average there are five more affected family members. The large number of affected relatives can be explained by the way in which the condition is inherited. Fabry disease is inherited in an X-linked pattern. This means that the non-working GLA gene that causes Fabry disease is located on the X-chromosome. Females have two X chromosomes while males have one X- and one Y-chromosome. If a female has Fabry Disease, each of her sons and daughters is at 50% risk for the disease. If a male has Fabry disease, all of his daughters and none of his sons are at risk to develop symptoms of Fabry disease.

A genetic counselor or medical geneticist can help identify at-risk family members by meeting with a family and creating and analyzing a detailed family history. At-risk family members may then be tested by taking a sample of their blood and looking for the mutation known to be present in their affected family members. This type of testing is called "targeted" DNA analysis in that it only looks for the mutation in the GLA gene known to cause Fabry disease in that particular family.

Once a family member is diagnosed with Fabry disease, testing other family members is now easier thanks to the ***Fabry Family Member Testing Project***. This joint project between the American Association of Kidney Patients (AAKP) and Emory Genetics Laboratory provides free Fabry disease testing to at-risk individuals whose family mutation is known. For more information about the Fabry Family Member Testing Project, please call the Emory Lysosomal Storage Disease Center at 404-778-8565 or 800-200-1524, or visit the project's website at http://www.genetics.emory.edu/LSDC/fabryproj.php

Advocating for your Child's Educational Needs

By Kendra J. Bjoraker, Ph.D., L.P.

"Succeeding in school is one of the most therapeutic things that can happen to a child!."

It is that time of year again; the excitement of getting your children ready for the new school year…buying new shoes, clothes and supplies. And although this holds true for families with lysosomal storage diseases (LSDs), feelings of anxiety, fear, and uncertainty also arise. The complex and continuous nature of their conditions and the high level of skill their care demands differentiates children who are medically fragile and often developmentally disabled from the general population of children with chronic conditions. Families identify, obtain, coordinate, and monitor a wide range of services for their child, one of which is educational services.

Advocating for educational services is overwhelming and often frustrating for many parents with children requiring more than the "regular" curriculum in order to learn. The following suggestions are offered to help assist your child as they enter school this fall.

The basic foundation for being effective in collaborating with regular and special educators is knowledge of the laws, regulations, and rules governing special education. Read the law, learn the law, and do not rely on anyone else to tell you what the special education laws mean for

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your child. This is the key that will unlock the door for your child. I encourage all parents to obtain a copy of their states rules and regulation through the Department of Education.

Individual with Disabilities Education Act (IDEA)

The IDEA has six principles. The first four principles reflect the actual processes that schools follow in order to provide each student with a disability, the benefits of a free appropriate education in the least restrictive environment. The last two are the procedures that parents and students can use to hold the schools accountable for carrying out the first four principles.

Children with LSDs typically are classified under "Other Health Impaired." The term includes a chronic or acute health problem resulting in limited strength, limited vitality or alertness, where special education and related services are needed because the disease adversely affects his/her educational performance.

During the Individualized Education Program (IEP) meeting, parents need to make sure their child's disease is clearly defined and a description of the disease is added to the IEP. It is important that the team, including the teacher, knows what the disease is and how it is manifested in the educational setting. For example, if you have a child with a diagnosis of Hurler syndrome (MPSI), you would define it and the symptoms (orthopedic, language, inattention, corneal clouding). It is important to realize that if a child has a disability but does not need special education services, the child will not be eligible under IDEA but may be eligible under Section 504 of the Rehabilitation Act. Section 504 does not receive federal money for providing 504 accommodations, so there is less incentive for serving children under these laws, nor do the children have an IEP under a 504, so parents beware.

Children with LSDs also begin receiving services early. Children (Ages 3- 9) may qualify in one or more of the following areas: physical development, cognitive, development, communication development, social or emotional development, or adaptive development. Infants and toddlers (Ages 0 –3) who experience the trauma of life-threatening illness and treatments are entitled by federal law to early-intervention programs that try to head off developmental delays. These services do not depend on a family's ability to pay. According to a child's and family's needs, the child may receive physical and occupational therapy, speech therapy, and special instruction. The family may be entitled to family services, such as training, counseling, or case management to help coordinate services.

Home and Hospital Services

Because the treatment for some lysosomal storage diseases can involve bone marrow transplantation, many children qualify for home and hospital educational services. Home and hospital teaching is designed to provide instruction to public school students who are unable to attend a regular school program due to a physical or emotional condition which is verified by a physician. Instructional services are available to all qualified students during convalescence or treatment time in a medical institution or therapeutic treatment center, or at the student's place of residence. The length of instruction for students in a full-day program is six hours per week; however, parents should know this time is not fixed and can be increased through a team decision.

Some children with LSDs also receive enzyme replacement therapy (ERT); therefore, missing school up to one day a week. In the state of Minnesota, there is a section in our rules and regulations entitled "Care and Treatment." Children receiving ERT qualify to receive direct instruction at the facility in which they are receiving ERT or another agreed upon place. Parents need to check with their state regulations to receive services for their child's excessive absences due to medical treatment.

Individualized Educational Program (IEP)

The IEP is not a "one-size-fits-all" program. Individualized Educational Programs are unique when a child has a neurodegenerative disease. For children with degenerative conditions, the IEP may include related services such as physical and occupational therapy or other services to address the child's needs in the areas of self-help, mobility, and communication. This is to help maintain the child's present level of functioning for as long as possible so that the child benefits from special education. The IEP team is required to review a child's existing health data which may include evaluations and information from parents, medical professionals or neuropsychologists who know the child and the child's specific medical conditions. The IEP team can then include appropriate services designed to extend current skills throughout the child's enrollment in school, especially if the disease results in negative progression and cannot be fully corrected or stabilized.

Remember, because your child's medical status often changes, an IEP team meeting can be requested more than once a year. The contents of an IEP should include: the present levels of academic achievement and functional performance, annual goals, educational progress, and special education with related services. Also included should be statements of the special education and related services, supplementary aids and services (based on peer-reviewed research), program modifications or supports for school personnel, and finally, accommodations and alternate assessments.

Accommodations, modifications, and alternative assessments may be necessary for a special needs child to succeed at school. The terms accommodations and modification are frequently used interchangeably, but they are not identical in their effect on teaching and learning and have important differences. Both terms are included in IDEA and are described below.

Accommodations are defined as alternative ways for students to acquire information or share what they have learned with you. Accommodations do not lower the difficulty level nor expectations for the student's achievement, although there may be changes in teaching materials used, testing materials, or even in the instructional environment. Once these changes are made, the standards of achievement remain the same. For example, a child may be unable to read an assigned textbook; therefore the accommodation would permit the student to listen to a taped version. Once the student has heard the story, however, he or she must take part in all required testing and assigned work. Accommodations are individualized to suit the student learning style and developmental level. Other examples are highlighting, rehearsal, color coding, memory joggers, visual cues, number lines, alphabet strips, flip charts, organization/transition cards.

Modifications are more intensive changes to the difficulty level and /or the quantity of material to be learned. Modifications also may, in fact, change the way material is presented and the nature of testing. Modifications create a different standard for children whose disabilities require more intense adjustments. An example of the modification may be seen in a spelling test that reduces the number of words to be studied. Basic modification decisions become long-range goals for the child's educational program, and they serve as a guide for educational decision-making on the daily and long-term basis.

For students with more complex special needs, an alternate assessment may be most appropriate. Alternative assessments measure a different area of skill or concepts than normally used for a given grade level or subject.

Assistive technology includes "any item, regardless of its origin, that is used to increase, maintain, or improve functional capacities of a child with a disability," but excludes "a medical device that is surgically implanted, or the replacement of such a device." Assistive technology is considered in children with LSDs with significant orthopedic, visual, hearing, or language impairments.

Because some children with LSDs are both medically and cognitively involved, many should be eligible for Extended School Year (ESY). This is not summer school. ESY services are provided to maintain the skills or behaviors the child has developed as identified on the IEP. There are six factors that the IEP team should consider in deciding if the child is eligible for ESY one of which is regression and recoupment. Regression refers to a decline in knowledge and skills that can result from an interruption in education. Recoupment is the amount of time it takes to regain the prior level of functioning. The issue is whether the benefits derived by the child during the regular school year will be significantly jeopardized if they are not provided an educational program during the summer months (or breaks).

Parent Participation

The following are suggestions that I have gathered over the years from parents and professionals, from being a former special education teacher myself, as well as a parent of children with disabilities.

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