various social networking opportunities are providing new ways for the Fabry and make their lives easier. Activities like organizing the 2010 “Charles Kleinschmidt Fabry Family Camp” Fabry disease assistance and support: Increased education is so important. Meetings represent some of the NFDF’s ongoing education efforts. More We are hoping to facilitate significantly more education opportunities. In the “Education” then “Fabry Disease Eye Symptoms” tabs for more details. Fabry disease to improve recognition by eye doctors. Please visit our website around the country. The resulting diagnoses from these education physician contributions to our overall NFDF education efforts. To date I have personally education efforts for the greater good of the community. Patients should...
Life Transitions in Lysosomal Storage Diseases: A Focus on Teens

By Dawn Laney, MS, GCC

“Not in the stars to hold our destiny but in ourselves.” — William Shakespeare

“You’re not the boss of me, NO!”

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 more successful adults. In individuals suffering from a chronic disease, such as a lysosomal storage disease, the teenage 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 we will provide 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 taking part in planning and learning about medical care, they can become more comfortable with taking on the care they will need. I am sure that each of you understands the challenge of maneuvering throughout 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 know that you were an integral part of helping our kids get the care that is necessary to fight this disease that affects them. You can image what it’s like having such a large team of caregivers to help you. As you can see how different personalities and ideas on how best to meet the needs of both our children, 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 our payment details with billing offices. Billing professionals have told us that our account is very complicated and they have a difficult time trying to sort out the medical billing. If you have questions about your own family’s insurance, feel free to contact one of us.

2. Take a quick test. Before starting off on a transition journey, figure out what your teen knows about their medical care. Basic health care questions like the “Ready to Manage Your Own Health Care?” form created by the PACER Center, Inc. are a great start and direct health care providers to make the transition easier.

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 healthy as possible. It is important for teens to learn why they do all the things 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 ask if they think the plan is making sense.

4. Know your healthcare providers. It is important for teens to know exactly what happening with their doctor or health care providers and their medical specialists. A list with contact information for each care provider is very helpful for quick reference. This knowledge will help them know how and when to ask for the care they need.

5. Learn about insurance. It is important for teens to discuss this with their physicians, parents, and helpful billers. When talking to your child, be very clear about what their insurance is needed to know and how insurance availability changes over time. This information will help them understand critical that teens know what happens to their insurance in different scenarios.

6. Plan for adulthood. Teens should think about what they might do to the over the next years and immediately 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? Teens, parents, and healthcare providers need 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, medical genetics/genetic counselors are able to work well 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 easy for some, and others may know who they are and be immediately transferred to another doctor.

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 situation is that a primary care doctor does not know immediately about as opposed to requiring an over the counter medication.

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 okay to speak up when they feel uncomfortable with a healthcare provider or need a little more explanation about something.

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. When diagnosing and participation from everyone, this can be a smooth transition.

Resources

3. Several useful Transition Quiznaires and Resources such as can be found at http://www.nationalcancerpatients.org.uk in the special education section.
4. Disease and treatment summary sheets: several paper-based options at http://departments.adulthealth.medscape.com 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.doc.edu/auto/teens/parents/resources.asp

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 illness. The National Fabry Disease Foundation (NFDF) distributes and discusses them. We will do our best to expand this program to 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, and clinical staff. We will develop educational materials 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 major symptoms of Fabry disease and every physician group will be informed of our disease. Earlier diagnosis is critical to living better and longer lives. People are interested to see how others are faring with Fabry disease.

As the National Fabry Disease Foundation (NFDF) enjoys our fifth year in operation, we continue to strive for greater clarity about our purpose and our programs that the beginning years. We have always been clear that our overall purpose is to serve and support families with Fabry disease. We just didn’t have the means and understanding of how to get there. We should have 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 works best in our operations.

To begin with, we view the Fabry community as a congregation 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 including 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 LysoStories, we would like to update you on a couple of our main focus areas.

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