POMPE Connections

The emotional impact of Pompe disease

Even if you had a feeling that something was not quite right, nothing can really prepare you for the diagnosis of Pompe disease. It can be a shock to learn that you or someone close to you has a rare inherited muscle disease that is going to get worse over time. Maybe that is why many people feel numb when they hear the news. After a while, the shock and numbness may give way to a storm of emotions. Finding a way to cope with your feelings will help you deal with the challenges that you and your family may face. This handout describes the emotional changes you may go through and explains how you can help yourself come to terms with the diagnosis and adjust to living with Pompe disease.

Q **I have just received a diagnosis of Pompe disease. I feel so angry that this is happening to me. Is this a normal reaction?**

A Anger is a normal reaction to such painful and disturbing news. But it is not the only strong emotion that may surface when you receive a diagnosis of Pompe disease. It is also normal to feel scared, anxious, or upset when you cannot control what is happening to you. As symptoms of muscle weakness appear, you may struggle with feelings of depression. You may try to bargain your way out of the situation by making deals with yourself or with God. These are normal ways to mourn the losses and changes in your life. Parents and partners of people with Pompe disease often struggle with these feelings as well. In time, most people reach a point where they are ready to accept the diagnosis and learn how to live with the disease. Some find that acceptance becomes more of a challenge as Pompe disease progresses and muscle weakness makes it harder to get around. If you become overwhelmed by feelings of anger, panic, despair, or hopelessness, it is important to seek help. A professional counselor or spiritual advisor can help you work through your feelings and restore a sense of hope.

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Other names for Pompe disease

Acid alpha-glucosidase deficiency, acid maltase deficiency (AMD), glycogen storage disorder (GSD) type II, glycogenosis II, and lysosomal alpha-glucosidase deficiency. In different parts of the world, Pompe may be pronounced *"pom-PAY," "POM-puh,"* or *"pom-PEE."*

Q **How can I help myself deal with the diagnosis?**

A Everyone copes with challenges in a different way and you will have to figure out what works for you. You may be helped by strategies that have worked for other people who have lived with Pompe disease. When fear, frustration, anger, or stress mount, try these tips to help keep yourself going:

- **Focus on the things you *can* control.** You may not be able to control the course of the disease or how severe your symptoms will be, but you can do a lot to make life better for yourself. Start by learning as much as you can about Pompe disease. Contact the groups listed in *Where to learn more* on page 4, read the other handouts in this series, search for information on the Internet, and know what your insurance plan covers. Take charge of your healthcare by keeping track of your symptoms and choosing a team of experts to meet your needs

- **Develop a strong support network.** Let family, friends, and neighbors ease your burden by helping with errands, household chores, child care, and other daily tasks. Hire a responsible college student or licensed home health aide to help manage medical needs, assist with physical care, and provide companionship. Connect with other patients and families living with Pompe disease by telephone, e-mail, or through an online support group to get advice and information from people who truly understand what you are going through. You might consider going to a national or international Pompe disease meeting where you can meet other patients or parents and hear from experts about the latest research advances

- **Take time for yourself.** Dealing with all the challenges of Pompe disease can take up every minute of the day unless you set some limits. Learn to say no to tasks that someone else can do. If you are a full-time caregiver, get outside help if needed so that you can find time each day (or as often as you can) to exercise, keep up with hobbies and interests, or visit with friends. Staying active and making time to do things you enjoy will help make life as normal as possible

Q **What should I tell my family and friends about Pompe disease?**

A Sharing the news can be difficult, both for you and for those close to you. Some people may not know what to say. Some may have trouble accepting the diagnosis. They may pull away when you are counting on them to come through for you. Since most people have never heard of Pompe disease, they are likely to have a lot of questions about it. The more you understand about the diagnosis and what it means, the easier it may be to talk with others. It may be helpful to share some of the handouts in this series with your friends and family members.

Q **My child has just received a diagnosis of Pompe disease. What is the best way to talk with her about it?**

A It is important to be honest and open and give information in a way that your child can understand. That will depend on your child's age and maturity level. Young children, for example, need to be told that Pompe disease is no one's fault. Older children may be concerned about what to tell friends or classmates. You may want to ask your healthcare provider, nurse, or social worker for advice on what to say. Some families prefer to have the healthcare provider share the news, so be sure to let your child's healthcare provider know how you want to handle it. Watch for your child's reaction and ask how she is feeling. Answer any questions as well as you can and assure your child that she can talk to you whenever she has questions or concerns.

Q **What are some of the emotional changes a person with Pompe disease may go through as muscle weakness gets worse?**

A Muscle weakness that restricts movement or leads to serious breathing problems can make you or your child with Pompe disease more dependent on others for care. Even though you may know that using a scooter or wheelchair can help you be more active and independent, you may mourn the loss of mobility. Many people may advise you to "live in the present" rather than dwell on what you have lost. Others may urge you to plan ahead for future changes. The challenge is to find a way to do both. Living in the present will help you appreciate the joys in your life. Planning ahead for the future — for example, by adapting your home to meet the needs of someone in a wheelchair or moving to a home that is more accessible — can help you maintain as normal a life as possible.

While life with Pompe disease will always have its challenges, seeking prompt medical treatment and getting supportive therapy to aid breathing and preserve muscle function can improve your attitude and your quality of life by helping you feel stronger and more comfortable. And though it is important to be realistic, advances in treatment can bring hope for the future.

Where to learn more

These groups can help you find the support and information you need to cope with the emotional impact of Pompe disease:

- The **International Pompe Association (IPA)** is a global federation of groups of patients with Pompe disease. The IPA helps patients, family members, and healthcare providers from around the world share their experiences and knowledge across continents and cultures. To find the contact for your country, visit the IPA Web site at www.worldpompe.org

- You may also be able to connect with patients and families living with Pompe disease through an online support community, such as GSDnet. This electronic mailing list can be accessed through the **Association for Glycogen Storage Disease (AGSD)** in the United States or through the **Association for Glycogen Storage Disease (AGSD) of the UK.** To subscribe, visit www.agsdus.org or www.pompe.org.uk/agsdnet.html

- **Children Living with Inherited Metabolic Diseases (CLIMB)** works directly with families in the United Kingdom but also sends information to people in other countries. CLIMB promotes contact among patients, parents, and professionals through support groups, telephone counseling, a pen pal service that matches children with similar interests, a national conference, and other services. [The CLIMB Web site refers to Pompe disease as Glycogen Storage Disease Type II.] For more information, visit www.climb.org.uk