



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.

Living with Pompe disease is about more than just dealing with symptoms, understanding genetic issues, or juggling doctor appointments. It is also about living in the face of new physical and emotional challenges. This section offers resources and insights to help patients and caregivers deal with the effects of the disease on everyday life.

Patients should know that they are not alone. Although Pompe disease is rare, there are active patient groups and caring health professionals throughout the world who can help you to manage the challenges of living with this disease. While each experience will always be unique, reaching out to others can bring patients and caregivers both support and new perspectives on their situation.

Although not everything suggested in this brochure may apply to each patient's situation now, some may become helpful as time goes by. Remember that Pompe disease is progressive, so patient's needs will likely evolve over time. A positive attitude, creative problem-solving skills, and the support of others will help with meeting new challenges head on.

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

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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. (for more information please refer to the *Where to Learn More* in this brochure).

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 stressors mount, try these tips to help keep yourself going:

- **Focus on the things you can control.** Pompe disease affects everyone differently, so it can be difficult to predict exactly how it will affect your life. 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 reading and learning as much as you can about Pompe disease. Contact the groups listed in *Where to Learn More* in this brochure, 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 ways for dealing with Pompe disease in day to day life:** The symptoms and challenges of Pompe disease will inevitably require changes in your daily routine and lifestyle. But these changes don't always mean you have to give up your independence or the activities you enjoy. Instead, you just need to learn new strategies and approaches to adapt to your situation. These can be as simple as adjusting expectations about what you can do in a single day. Or, they may be hands-on changes such as modifying your home or workstation to better fit your needs.
- **Keep up with regular routines:** Living with Pompe disease can place a burden on regular daily activities, such as attending school, going to work, and participating in hobbies. You may need to take extra time off from school or work in order to go to doctor appointments and for your biweekly ERT. Keeping up with regular routines can also help you to feel better, both physically and mentally. You just need to brainstorm new ways of doing things with your family and friends.

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- **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. Reaching out for emotional support is also important. Patient organizations can connect families for sharing experiences and practical advice. Other ways to connect with other patients and families living with Pompe disease is by telephone, e-mail, or through an online support group such as the GSDNet. 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. For more information about the GSDNet refer to the *Where to Learn More* section in this brochure).
- **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.
- **Accepting Outside Help:** If you or a loved one needs round-the-clock care, you need to be realistic about how much you and your family can handle. Some people may need specialized medical care that family members cannot provide. Accepting the help of nurses, home health aides and other professional caregivers may be difficult, but it can be a sound option in many cases.

Q: Are there any tips for caregivers?

A: The challenges of Pompe disease affect not just patients, but their family members as well. As the disease progresses and patients face greater physical challenges, they must turn more and more to parents, spouses, and others for support with daily activities. If you are taking care of a loved one with Pompe disease, you should remember to take care of yourself too. Caregiving can be draining - both physically and mentally. Reaching out to other family members and friends can help you to find time each day 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 for you too.

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.

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

A: Caring for a child with Pompe disease requires both accommodating their medical and physical needs as well as providing emotional, mental, and social support. 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. The following is a list of some strategies and tips that may help:

- Talk to your child's doctors and care providers about the best ways to explain Pompe disease and its effects to children.
- Make young children understand that Pompe disease is nobody's fault.
- Discuss whether older children want to talk to friends and classmates about their condition or not.
- Helping children remain as active and independent as possible by giving them tasks, chores, and activities tailored to their age and abilities.
- Encouraging involvement in sports, art, and music programs that meet a child's needs for social interaction and physical activity.
- Adapt a child's interests and hobbies to their changing condition, and explore new activities to replace past ones.
- Become an advocate for your child's special needs.

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 federation of Pompe disease patient groups around the world. To find the contact for your country, visit the IPA Web site at www.worldpompe.org.
- **Acid Maltase Deficiency Association (AMDA):** The AMDA was formed to assist in funding research and to promote public awareness of Acid Maltase Deficiency, also known as Pompe's Disease. Visit the website www.amda-pompe.org
- **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

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- **American College of Medical Genetics (ACMG) Practical Guideline:** Pompe Disease Diagnosis and Management Guideline 2006. Vol. 8. No. 5. The *ACMG guidelines were designed as an educational resource for physicians and other health care providers.*
- The Genzyme Corporation's **Pompe Community website** www.pompe.com: offers the Pompe community comprehensive information on the disease, as well as resources and support to help manage the challenges it may bring.
- **GSDNet** (listserv@listserv.icors.org): Join the GSDNet Listserv to exchange emails with people around the world living with Pompe disease. **To subscribe to GSDNet by email:**

"To" Line: Type the following onto the "To" Line: listserv@listserv.icors.org

"CC" Line: Leave the CC line blank

"Subject" Line: Leave the subject line blank.

Email Message: Type the following message into the body of the email:
Subscribe GSDNet <add your name>.



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