



Getting the right care for Pompe disease

It takes a team of healthcare providers to meet all the needs that may arise as Pompe disease progresses. Each member of your healthcare team has unique skills to offer. One doctor may serve as team leader and coordinate the care given by all healthcare providers. But because Pompe disease is so rare, you may see providers who have never treated anyone with this neuromuscular disorder. It will help to work closely with the medical team to get the treatment and services you or your child really need. Learning all you can about Pompe disease can help you take charge of your care. This handout will get you started.



Q I know it's important to learn about Pompe disease, but where do I begin?

A You can find good sources of information on the Internet. Much of it comes from groups that were started by patients or parents of children with Pompe disease. You can also learn a lot from groups that support research into other neuromuscular disorders, as well as from government agencies and major medical centers around the world. (The list of resources in *Where to learn more* on the last page can help you get started.)

Q How is Pompe disease treated?

A Researchers are making good progress toward finding effective ways to treat Pompe disease or slow the progression of symptoms. For now, however, treatment focuses on

supportive therapies to manage symptoms and make it easier to live with the disease. The goal of supportive therapies is to help people with Pompe disease keep up their strength and prevent health problems caused by muscle weakness. Therapies are tailored to the specific needs of the patient. Respiratory therapy can provide the support needed for breathing. Physical therapy can help increase movement, lessen stiffness, and ease pain. Dietary therapy helps to maintain calories and prevent weight loss. Everyone with Pompe disease faces new challenges as the disease slowly progresses. Getting the right therapy from a knowledgeable medical team combined with the support and care of those around you can help you live each day to its fullest.

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 Who are the key people on my healthcare team?

A It takes a wide range of skills to meet the needs of someone with Pompe disease. In addition to your family doctor or primary care provider, the chart below lists possible members of your medical team.

The team will need you to keep them informed if health changes or new symptoms occur. Be sure each team member knows the care you are getting from other team members. If you have a child with Pompe disease, you will need to keep track of your child's symptoms and reactions to treatment and assist with supportive therapies.

Caring for Pompe Disease: Possible Members of the Medical Team

Team Member	Role
Pediatrician	Diagnoses and treats infants and children with all kinds of illnesses, including metabolic diseases. May be your child's primary care provider
Neurologist or pediatric neurologist	Diagnoses and treats neuromuscular diseases
Cardiologist	Treats the heart problems that may occur with Pompe disease
Pulmonologist or respiratory specialist	Treats the breathing problems caused by muscle weakness
Gastroenterologist	Treats problems related to digestion and feeding difficulties
Orthopedist	Treats scoliosis (curvature of the spine), contractures (muscle tightness), and other joint or bone problems related to muscle weakness
Respiratory therapist	Helps patients learn exercises to strengthen the muscles used for breathing
Dietitian	Creates eating plans to manage weight loss and swallowing problems
Genetics counselor	Discusses issues related to family risk and family planning and arranges for prenatal diagnosis
Social worker and psychologist	Help people cope with their fears and worries and deal with practical concerns
Occupational therapist and physical therapist	Help people do exercises to strengthen weak muscles and learn new ways to do everyday tasks

Q **How do I find a medical center that has experience in dealing with Pompe disease?**

A There are only a handful of medical centers around the world that specialize in treating Pompe disease. But clinics and rehabilitation centers that treat similar neuromuscular disorders or rare genetic diseases may also provide expert care for people with Pompe disease. In the United States, the Muscular Dystrophy Association (MDA) has more than 200 hospital-affiliated clinics that offer comprehensive care for children and adults with neuromuscular disorders. You can find a complete listing of MDA clinics at www.mdausa.org/clinics. In Europe and other continents, there are also many specialized centers for people with neuromuscular disorders. To locate medical centers in other countries, see *Where to learn more* on the next page.

Q **Beyond medical advice, where can I turn for help with all the practical challenges I will face?**

A There are many agencies, individuals, and patient groups that can assist you in getting the support you will need. They can help you review your finances, apply for government benefits and financial aid, find medical and social services, set up an education plan for your child, arrange for home care, learn your legal rights, and deal with employment issues and insurance problems. To find sources of support in your community, your country, or on the Internet, see *Where to learn more* on the next page.

Q **I feel like I know more about Pompe disease than some of the healthcare providers who are treating my child. What can I do about this?**

A Many caregivers and patients with rare disorders like Pompe disease feel that the only way to be sure of getting the right treatment is to learn all they can about the disease. Knowledge can be a real advantage when you are dealing with a large number of healthcare providers. You should keep track of what everyone is doing. This may help prevent mistakes or errors from occurring. There may be times when it is necessary to educate a pediatrician, or family physician, or emergency room doctor who has not had much experience with Pompe disease. Think of your knowledge as a skill you have to share with the healthcare team. And when you have concerns, do not hesitate to offer your opinion or question why something is being done. Remember that no one has a greater stake in your care or your child's care than you do.

Where to learn more

Information, advice, and support

- The **International Pompe Association (IPA)** can direct you to Pompe disease patient groups around the world. To find the contact for your country, visit the IPA Web site at www.worldpompe.org
- The **Acid Maltase Deficiency Association (AMDA)** offers information on special needs trust funds. Go to www.amda-pompe.org, and click on *AMDA's Pompe's Disease Communication Program*
- The **Pompe Community** Web site (www.pompe.com), sponsored by Genzyme, has a comprehensive list of patient groups and other services for people with Pompe disease

Medical centers specializing in neuromuscular disorders

To locate medical centers that specialize in treating neuromuscular disorders, contact the neuromuscular disorders (NMD) association for your country

- In the **United States**, contact the **Muscular Dystrophy Association (MDA)** at www.mdaua.org/clinics
- In **Europe**, contact the **European Alliance of Neuromuscular Disorders Association (EAMDA)** Web site (www.eamda.net)
- In **other continents**, visit the **World Alliance of Neuromuscular Disorder Associations (WANDA)** Web site (www.wandaweb.org) and click on *Your Country*

NOTES:

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