

Accessing Care and Resources for Pompe Disease

Getting a Diagnosis

Pompe disease is a rare condition. Initial symptoms can look like many other diseases, which can make it hard to diagnose Pompe. Getting a diagnosis may involve a long diagnostic journey. Patients identified through state newborn screening programs have the benefit of early diagnosis, but not all states screen for Pompe disease.

Diagnostic tests and procedures for Pompe include: a medical history and physical exam, procedures like an EKG that records heart activity, lab tests such as tests for GAA enzyme activity or genetic testing for mutation analysis, and imaging like a CT or MRI scan. Read about the diagnostic journeys of people with Pompe disease who have kindly shared their stories at AMDA's Meet the Patients - <https://amda-pompe.org/patients/>

Because Pompe disease is rare, many health care providers have not treated it. If you need help locating medical care and/or supportive therapy, please contact AMDA's Patient Advocate, Marsha Zimmerman, RN, at marsha.zimmerman@amda-pompe.org.

It Takes a Team

Each person with Pompe disease has different medical care and supportive therapy needs. It takes a team of the patient and/or family, primary care doctor, medical specialists, and therapists to treat Pompe. The primary care doctor, like an internal medicine doctor, or for children, a pediatrician, makes a referral to a medical specialist like a neurologist or geneticist. In most cases, this specialist becomes the team leader for Pompe disease care once a diagnosis is made. For other medical care needs, patients should see their primary care doctor.

Preparing for Your Doctor Visit

Be prepared to share your medical history and that of your family. Doctors will ask for this information, usually during your first visit. It is important to write down:

- When symptoms began
- If and how symptoms have changed over time
- Previous doctor visits and tests
- Any treatments that have been tried

Preparing ahead of time for all your medical appointments allows you to think about what you hope to get out of the visit. You will have time to consider what to say about your worries, symptoms, and other information the doctors may need. It may also help you to write down the questions you have for your doctor to take with you to the appointment. When hearing a lot of new information at once, it can be difficult to remember your questions, so a written list may be helpful.

Learn more at:

NIH Genetic and Rare Diseases Information Center – Getting a Diagnosis for Pompe Disease

<https://rarediseases.info.nih.gov/diseases/5714/glycogen-storage-disease-due-to-acid-maltase-deficiency/diagnosis>

Society to Improve Diagnosis in Medicine- Patient Toolkit to Prepare for a Medical Appointment

<https://www.improvediagnosis.org/patients-toolkit/>



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Working with Your Team

A wide range of expertise is needed for Pompe treatment. Most people with Pompe are under the care of multiple specialists. For example, a patient might be referred to a pulmonologist for lung and respiratory problems, a cardiologist for heart problems, a gastroenterologist for digestion problems, and a physical therapist to help with mobility issues. As the medical care and supportive therapy needs of you or your child change over time, the team members may change.

Multidisciplinary team members should communicate with each other on an ongoing basis throughout treatment. You will also need to work closely with your team to make sure you get the services you or your child need. Be sure each team member knows the care you are receiving from other team members. Keep a list of your team members and their contact information that you can share with the other team members. Coordinated, multidisciplinary care is essential to help ensure the best outcomes.

Learn more at IPA/Pompe Connection:

- <https://worldpompe.org/resources/patient-focused-publications/>
 - Getting the Right Care for Pompe Disease

Talking with Your Doctor

Before your medical appointments, think about questions to ask the doctors. For example, are they treating other Pompe patients? Will they help you with referrals to other medical specialists? How is insurance handled? For more examples of questions, refer to IPA/Pompe Connection-Finding a Treating Physician.

Other helpful tips are:

- Keep track of symptoms and reactions to treatment. This information can help your doctors provide better care.
- Take notes during appointments.
- Make sure you understand your diagnosis and treatment. If you don't, ask questions.
- Repeat back to the doctor what you have been told to be sure you are clear. Ask your doctor for written instructions if you need them.
- Find out how to review your medical records so you can track your care.
- Ask your doctor about how best to communicate and where to find more information.

Learn more at:

- Talking with Your Doctor or Health Care Provider <https://www.nih.gov/institutes-nih/nih-office-director/office-communications-public-liaison/clear-communication/talking-your-doctor>
- National Library of Medicine – Talking with Your Doctor <https://medlineplus.gov/talkingwithyourdoctor.html>

For Parents

You must take an active role in managing your child's care. Work with your child's doctor on next steps after diagnosis. Ask questions, take notes, and keep records. It will help ensure good care for your child, who may see multiple medical specialists and therapists over the years. Because Pompe disease is an inherited condition, you may want to talk with a genetics specialist, especially if you have other children. Ask your child's doctor about a referral for genetic services.

Some children with Pompe have developmental delays. The Milestone Tracker App (<https://www.cdc.gov/ncbddd/actearly/milestones-app.html>) tracks a child's milestones from age 2 months to 5 years. This Center for Disease Control and Prevention app has illustrated checklists. It offers tips for encouraging your child's development and what to do if there is concern. If you think that your child is not meeting milestones, ask your child's doctor about arranging for a developmental evaluation and care.

Learn more at:

- Baby's First Test – Accessing Care <https://www.babysfirsttest.org/newborn-screening/conditions/pompe>
- For Parents: Getting the Most from a Doctor's Visit Nemours Children's Hospital <https://kidshealth.org/en/parents/talk-doctor.html>

Accessing and Paying for Care

Medical care and supportive therapy for Pompe disease have different costs such as medical co-pays, medicines, lab tests, and other expenses. AMDA's webinar, Insurance 101, guides viewers through a discussion of the main types of health insurance coverage, types of health insurance plans, things to consider when choosing a plan, open enrollment, qualifying events.

Many companies with FDA-approved treatments have assistance programs as well. Specifically, they often have programs to assist patients with any insurance-related questions.

Some non-commercial resources to help people with Pompe disease with expenses are:

- Accessia Health Foundation - <https://accessiahealth.org/medications/>
- Patient Access Network Foundation - <https://www.panfoundation.org/disease-funds/pompe-disease/>
- Patient Advocate Foundation Co-pay Relief - <https://copays.org/funds/pompe-disease/>
- The Assistance Fund/Pompe Disease Financial Assistance Program—<https://tafcares.org>
- The National Organization for Rare Diseases (NORD) offers:
 - CareGiver Aid - <https://rarediseases.org/patient-assistance-programs/caregiver-respite/>
Grants of up to \$250 annually for those who qualify to give caregivers a break to attend a conference, event or have an afternoon or evening away from caregiving.
 - Emergency Relief - <https://rarediseases.org/patient-assistance-programs/medicalert-assistance-program/>
Provides eligible individuals with a MedicAlert product and a 3-year membership.

For other guidance about assistance with accessing care, please contact AMDA's Patient Advocate, Marsha Zimmerman, at marsha.zimmerman@amda-pompe.org.

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