Late-onset Pompe Disease (LOPD)

Signs and Symptoms of LOPD

Late-onset Pompe disease (LOPD) can appear at any age. Its signs and rate of progression vary widely but are usually milder and slower to worsen than with infantile-onset Pompe disease (IOPD). This is because individuals with LOPD have more GAA enzyme in their bodies to break down the glycogen stored in the lysosomes of muscle cells.

There is a wide range in how much GAA enzyme people with LOPD have. There are also other factors that are not yet fully understood that affect LOPD progression. This means the effects of LOPD are unique to each person. Some children and adults with LOPD have milder symptoms than others. Some symptoms are:

- Muscle weakness (trouble walking, using stairs, getting off the floor, etc.)
- Trouble with breathing (may first show as morning headaches or nausea)
- Feeling tired
- Digestive problems
- Difficulty chewing and swallowing

These and other symptoms can look like different medical conditions. This makes LOPD hard to diagnose, which delays treatment.

Parents who live in a state that screens newborns for Pompe disease learn early that their baby may have Pompe disease. The diagnosis of LOPD is confirmed by lab tests that measure a person's GAA enzyme activity. Even if there are no signs or symptoms, parents should make sure a doctor who knows how to treat LOPD sees their child. They should make a plan to monitor their child's LOPD. The AMDA Patient Advocate, Marsha Zimmerman, RN, can help families find care. She can be reached at marsha.zimmerman@amda-pompe.org. You can also learn more at: IPA Pompe Connections-Finding a Treating Physician.

In states where newborn screening for Pompe disease is not offered or if a child was born after it was available, it can take a long time to find out the cause of medical problems. This is time that is lost in starting treatment. The AMDA and other organizations are working to ensure all states screen for Pompe disease. You can search which states screen for Pompe disease at https://www.babysfirsttest.org

Some Signs of LOPD at Different Ages

Babies do not have an enlarged heart within their first year, as happens in babies with IOPD. Some babies with LOPD may not learn to sit, crawl, or stand, or they may have developmental delays when it comes to motor milestones. Some babies and children may lose the gains they make. They may also have breathing and feeding problems.

LOPD weakens muscles in the back and pelvis. Children may have trouble walking, keeping their balance, or sitting up straight.
Their muscle weakness may make it hard to keep up with their peers when playing sports and being active. Hip and leg muscle weakness may cause them to sway their hips or waddle when walking. Children may have problems eating and gaining weight. They may also have frequent lung or other respiratory infections.

Adults may experience similar symptoms of muscle weakness like the swaying walk, trouble getting up from a chair, or falling often. They may get tired easily. They may have difficulty breathing when exercising or climbing stairs. They may have lower back pain, muscle aches, and other pain.

The good news is that there is effective treatment and supportive therapy for living a full life with LOPD. How LOPD affects a person is greatly improved with treatment and supportive therapy which can slow the disease and help with managing it. Please see the next page for more information. You can also learn more at:

- IPA/Pompe Connections
  https://worldpompe.org/resources/patient-focused-publications/
  - The Signs & Symptoms of Pompe
  - Adapting to Living with Pompe
- Baby's First Test-Pompe
  https://www.babysfirsttest.org/newborn-screening/conditions/pompe

Acid Maltase Deficiency Association
PO Box 700248
San Antonio, Tx 78270
www.amda-pompe.org

Enzyme Replacement Therapy (ERT)

ERT is the main treatment for LOPD and IOPD at this time. It has been studied and advanced since the mid-1990s and was first approved as a treatment for Pompe in 2006. ERT replaces the GAA enzyme that is missing in the body or is in just a tiny amount. This GAA enzyme is made in the laboratory. It is given through the person's vein into the bloodstream. ERT is done on a regular schedule for life.

Researchers are developing promising new treatments such as gene therapy, chaperone therapy, and substrate reduction therapy. Some of the new treatments are combined with ERT.

Starting Treatment for LOPD

Treatment for LOPD is started at various ages. It depends on the patient's symptoms and severity. A team of family, different kinds of specialist doctors, and therapists is needed to treat LOPD.

Children and adults who have slight but observable signs of LOPD should start ERT right away. People with symptoms at diagnosis should also start ERT right away. Those without symptoms should be observed by their health care provider to determine when treatment should be started. (Reference: Cupler, E.J. et al. Consensus Treatment Recommendations for Late-onset Pompe Disease. *Muscle Nerve* 45:319-333, 2012).

All people with LOPD should be followed closely by their health care providers.

Learn more at AMDA:
https://amda-pompe.org/treatment

Supportive Therapies

Therapies are adapted to the age and needs of each person with LOPD.

- Physical therapy helps babies gain or keep muscle strength as they grow. It can help infants, children, and teens develop motor skills meant for their ages like crawling, running, and playing ball. However, as muscle weakness increases, people with LOPD may need to use a cane, walker, or wheelchair at some point in their life.
- Respiratory therapy is needed for lung infections and breathing problems. At any age, people with LOPD may need to use a machine to help them cough normally and to breathe, especially at night. This is called non-invasive ventilation.
- Dietary therapy may be needed for those who have trouble eating. They may need special diets and some may need a feeding tube.
- Therapies for speech, hearing, learning, and other challenges may be needed over the years.

Learn more at IPA/Pompe Connections -
https://worldpompe.org/resources/patient-focused-publications/

- Exercise and Physical Therapy
- Breathing Problems in Pompe Disease
- Nutrition and Diet Therapy
- The Emotional Impact of Pompe Disease
- Common Health Concerns

AMDA Resources

A diagnosis of LOPD may be overwhelming. The AMDA's goal is that nobody will ever be alone when questions and needs arise. **There is hope for the future. We are here to help.** We have many resources and programs to support families and patients. Two important resources are:

Patient Advocate: The AMDA's Patient Advocate, Marsha Zimmerman, is a registered nurse. She has been working as a Pompe Patient Advocate since 2001. She has experience assisting families with everything from access to therapy to disease education and tips for communicating with medical professionals. She also provides emotional support. Whatever questions or issues you may have, Marsha Zimmerman can help. Her email address is: marsha.zimmerman@amda-pompe.org

Mentor Program: This program puts families and patients in touch with other people who have been through similar experiences. AMDA volunteer mentors from the Pompe community are available to help in dealing with a new diagnosis. They can share experiences, feelings, and resources related to Pompe disease. Reach out to Morgan.Burroughs@amda-pompe.org or through links on the website: www.amda-pompe.org

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