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## **Pompe Disease**

### **Information for Physicians and Other Health Care Professionals**

#### **Definition**

Pompe disease is an inherited metabolic disorder in which harmful amounts of glycogen accumulate within lysosomes of cells. Individuals with Pompe disease do not produce enough of one of the enzymes ( $\alpha$ -glucosidase or GAA) needed to metabolize glycogen. Over time, this excessive storage of glycogen in the lysosomes can cause permanent cellular and tissue damage, particularly in the heart and skeletal muscles.

#### **Clinical Symptoms**

Infantile Pompe disease is the result of a severe deficiency of GAA. Symptoms begin in the first months of life, with feeding problems, poor weight gain, muscle weakness, and hypotonia. Hypertrophic cardiomyopathy is the hallmark of the disorder and, in the untreated patient, typically results in death by 1 year of age.

Juvenile or adult Pompe disease is the result of a less severe deficiency of GAA. Symptoms can appear as early as the first year or as late as the sixth decade of life. The disorder resembles limb girdle muscular dystrophy with symptoms including muscle weakness progressing to respiratory weakness. The heart is rarely involved and not to the extent observed in infantile Pompe disease.

#### **Newborn Screening and Definitive Diagnosis**

In Illinois, newborn screening for Pompe disease is performed by determination of GAA activity. **If newborn screening results indicate an abnormal GAA activity, referral should be made immediately to a metabolic disease specialist.**

#### **Treatment**

If a diagnosis of infantile Pompe disease is established, enzyme replacement therapy should be started as soon as possible. For patients with a diagnosis of later onset Pompe disease, clinical follow-up will determine when enzyme replacement therapy should be initiated. In addition to enzyme replacement therapy, supportive treatment such as physical therapy has an important role. Evaluation by additional medical specialists such as neurologists, pulmonologists and cardiologists may be appropriate.

#### **Incidence**

The incidence of Pompe disease is estimated to be approximately one in 40,000 births.

#### **Inheritance Patterns**

Pompe disease is inherited in an autosomal recessive pattern. Parents of a child with Pompe disease are unaffected, healthy carriers of the condition, and have one normal gene and one abnormal gene. With each pregnancy, carrier parents have a 25 percent chance of having a child with Pompe disease (inheriting two copies of the abnormal gene). Carrier parents have a 50 percent chance of having a child who is an unaffected carrier, and a 25 percent chance of having an unaffected, non-carrier child. These risks would hold true for each pregnancy. **Genetic counseling is recommended for families planning future pregnancies.**

## **Pathophysiology**

In Pompe disease, a lysosomal enzyme deficiency leads to an accumulation of glycogen within in the lysosomes, primarily in cardiac and skeletal muscle. The build-up of glycogen in the lysosomes causes clinical findings of the disease. Because the cytoplasmic pathway for glycogen synthesis and degradation is intact, none of the metabolic abnormalities (such as hypoglycemia or metabolic acidosis) common in patients with other forms of glycogen storage disease are observed.

## **Key Points for Parents**

Reassure parents that not all infants identified as having low GAA activity through newborn screening will have Pompe disease. If the infant should turn out to have Pompe disease, treatment is available which may ameliorate the clinical symptoms of the disorder. If the child needs additional testing or diagnostic evaluation, make certain the parents understand the importance of following the pediatrician's and/or specialist's recommendations for additional testing and referrals.

## **Following Confirmation of Diagnosis**

These guidelines should be followed after a diagnosis of Pompe disease has been confirmed:

- 1) Follow up with the child's metabolic disease specialist.
- 2) Use a multidisciplinary approach for long-term management including specialists from pediatrics, genetics, cardiology, pulmonology and other disciplines as needed.
- 3) Ensure that parents understand that treatment for Pompe disease is not curative and that morbidity cannot always be prevented.
- 4) Recommend genetic counseling services to help the parents understand the complexity surrounding the carrier state and inheritance of this disease.
- 5) Provide parents information on support services, such as the [Acid Maltase Deficiency Association](#), [United Pompe Foundation](#), early intervention service providers, the local health department, and the University of Illinois at Chicago Division of Specialized Care for Children (DSCC).
- 6) Additional information about newborn screening can be found at:
  - Baby's First Test: <http://www.babysfirsttest.org/>  
Health Resource and Service Administration (HRSA), Grant no. U36MC16509, Quality Assessment of the Newborn Screening System.
  - National Center for Biotechnology Information: <http://www.ncbi.nlm.nih.gov/gtr/>  
National Center for Biotechnology Information, U.S. National Library of Medicine, 8600 Rockville Pike, Bethesda MD, 20894 USA.