NEWBORN SCREENING DISORDER FACT SHEET

Glycogen storage disorder type II (Pompe)

NEWBORN SCREENING FINDINGS

A baby with low **acid alpha glucosidase** (**GAA) activity** on their newborn screening specimen(s) may have **Glycogen storage disorder type II** (**Pompe disease)**. Diagnostic tests are needed to rule out or confirm Pompe disease.

ABOUT THE CONDITION

Glycogen storage disorder type II, Pompe Disease, is a treatable disorder that occurs in approximately one in every 28,000 births. The disorder is caused by changes in the acid alpha glucosidase (GAA) gene. This change results in decreased activity of the GAA enzyme, which is responsible for breaking down glycogen, a complex sugar, in the body's cells. If glycogen is not broken down, this can lead to an abnormal buildup within the body, which causes muscle damage.

Parents who have a child with Pompe disease typically each carry one altered copy of the gene involved in this process. That means, with each pregnancy, each baby will have a one in four chance of having Pompe disease.

SYMPTOMS

There are different types of Pompe disease that range from mild to serious. Symptoms of Pompe disease may first appear shortly after birth, or not until adulthood. These symptoms may include heart problems, muscle weakness, or difficulty breathing. If left untreated, Pompe disease can hinder a person's ability to move, eat, or breathe on their own, or even cause death.

TREATMENT

Pompe disease is treated by replacing the missing GAA enzyme through enzyme replacement therapy (ERT). Although ERT does not cure the disease, it can improve heart and muscle function, growth, development, and increase survival.

NEXT STEPS

The next step is to get a blood test to check the activity of the GAA enzyme to rule out or confirm Pompe disease. If the blood test is abnormal, a visit to a specialist may be needed. They may recommend additional tests or treatment.

RESOURCES

For more information about Pompe disease, please see the Disorders section of our website: www.doh.wa.gov/nbs. Information can also be found at https://www.babysfirsttest.org/.

Washington State Department of Health Newborn Screening Program
1610 NE 150th Street, P.O. Box 55729, Shoreline, WA 98155
Phone: 206-418-5410 | Fax: 206-363-1610 | www.doh.wa.gov/nbs |Email: NBS.Prog@doh.wa.gov

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