Pompe disease is a rare inherited condition caused by the buildup of glycogen in the body’s cells. Glycogen is usually broken down by an enzyme called alpha-glucosidase (GAA). In individuals with Pompe disease, this GAA enzyme is missing or works poorly. This results in the buildup of glycogen, and this extra glycogen causes serious health problems.

What is Pompe disease?

Pompe disease is one of more than 40 different disorders which are included in newborn screening programs in every state. It is a rare inherited condition caused by the buildup of glycogen, and this extra glycogen causes serious health problems.

What is newborn screening?

Every state has a newborn screening program to detect these conditions at birth. Early diagnosis and treatment of these disorders often prevents lifelong disability or death. Most newborn screening programs test for Pompe disease when a baby's blood is drawn during routine newborn exams.

What does it mean to be a carrier of Pompe disease?

Carriers of Pompe disease are individuals who have one mutation in one out of their two GAA genes. These individuals still have one GAA gene without a mutation in it. Carriers of Pompe disease are more likely to have a child with Pompe disease if their partner is also a carrier. Both parents of a child with Pompe disease are nearly always carriers of the condition.

How do I know if my baby has Pompe disease?

Newborn screening results are not usually available until 3 to 6 weeks after birth. Parents are usually notified if their baby’s blood test is abnormal. Your baby will then undergo additional testing to determine if he or she definitely has Pompe disease, or that he or she may be a carrier of Pompe disease. Your baby may also need special evaluations of his or her heart, and so may be referred to a Pediatric Cardiologist as well.

Additional blood tests will be ordered by the geneticist to find out if your baby definitely has Pompe disease, or that he or she may be a carrier of Pompe disease. Additional blood tests will be ordered by the geneticist to find out if your baby definitely has Pompe disease, or that he or she may be a carrier of Pompe disease.

How does New York State screen for Pompe Disease?

Newborn Screening for Pompe Disease Information for Parents

How does New York State screen for Pompe Disease?

Newborn Screening Program staff can be contacted at 518-473-7552, Monday through Friday, 8:00 am until 4:45 pm or at nbsinfo@health.ny.gov. The NYS Newborn Screening Program can be reached between 8:30 am and 4:45 pm Monday through Friday.

Who can I call if I have additional questions about newborn screening for Pompe Disease?

Your baby’s doctor or the NYS Newborn Screening Program can provide additional information about newborn screening for Pompe disease.

What is newborn screening?

Newborn screening is a medical test performed on newborns to help detect certain health problems that may not be obvious at birth. This test is performed during routine newborn exams. Your baby’s doctor or the NYS Newborn Screening Program can provide additional information about newborn screening for Pompe disease.
Pompe disease:
The earlier symptoms appear and the more severe closely related to how much functioning GAA individual to individual, partly based on the age The symptoms of Pompe disease differ from of Pompe disease? What are the symptoms function abnormally. buildup of glycogen, and this extra glycogen causes with Pompe disease, this GAA enzyme is missing prevent the buildup of glycogen. In individuals glucosidase (GAA); when GAA is working it helps broken down by an enzyme called alpha-glycogen in the body's cells. Glycogen is usually disorders which are included in newborn screening Pompe disease is one of more than 40 different serious health problems. identify infants with rare disorders, which would Every state has a newborn screening program to What is newborn screening? Every state has a newborn screening program to identify infants with rare disorders, which would not usually be detected at birth. Early diagnosis and treatment of these disorders often prevents serious health problems. What is Pompe disease? Pompe disease is one of more than 40 different disorders which are included in newborn screening in New York State. It is a rare inherited condition caused by the buildup of a complex sugar called glycogen in the body's cells. Glycogen is usually broken down by an enzyme called alpha-glucosidase (GAA); when GAA is working it helps prevent the buildup of glycogen. In individuals with Pompe disease, this GAA enzyme is missing entirely or working poorly. This results in the buildup of glycogen, and this extra glycogen causes certain organs and tissues, such as muscle, to function abnormally. What are the symptoms of Pompe disease? The symptoms of Pompe disease differ from individual to individual, partly based on the age of the person at which his or her symptoms first appear. The age of onset of symptoms is usually closely related to how much functioning GAA enzyme is present (ie. the less active the enzyme is, the earlier symptoms appear and the more severe they are). As a result, there are two main forms of Pompe disease: Early onset. The GAA enzyme is completely or very nearly missing. In these babies, symptoms appear soon after birth and include poor muscle tone, an enlarged and weakened heart, poor feeding and growth, and difficulty breathing. Late onset. The GAA enzyme is only partially missing. In these individuals, the age of onset of symptoms is variable; symptoms may appear as early as the first few months of life, or as late as late adulthood. The primary symptom is muscle weakness that gets worse over time, and young children and babies may experience delayed developmental milestones. The heart is less likely to be involved in this form of Pompe disease. How does New York State screen for Pompe disease? NYS screens for Pompe disease by measuring the amount of GAA enzyme activity in a sample of the baby's blood. If there is a low amount of GAA enzyme activity, then genetic testing is done to look for mutations in the GAA gene. Mutations in this gene result in lower GAA enzyme activity. Every person has two copies of this gene. Pompe disease is an autosomal recessive genetic condition, meaning that individuals who have the condition have a gene mutation in both copies of their GAA gene. What does it mean to be a carrier of Pompe disease? Carriers of Pompe disease are individuals who have one mutation in one out of their two GAA genes. These individuals still have one GAA gene without a mutation in it. Carriers of Pompe disease do not have any signs or symptoms of the condition, but have a 1 in 4 (25%) chance to have a child with Pompe disease if their partner is also a carrier. Both parents of a child with Pompe disease are nearly always carriers of the condition. My baby had a positive newborn screen for Pompe disease. Does my baby definitely have the condition? If your baby was found to have two mutations in the GAA gene, then it is very likely that he or she has Pompe disease. Your baby will undergo further evaluation and testing with a specialist to determine if he or she has Pompe disease. Additional blood tests will be ordered by the geneticist to find out if your baby has Pompe disease. Your baby may also need special evaluations of his or her heart, and so may be referred to a Pediatric Cardiologist as well. These tests and evaluations are very important. If diagnosis and treatment of Pompe disease are delayed, it can lead to serious health complications for your child. In some cases, these complications are life threatening. What is the treatment for Pompe disease? Individuals with Pompe disease are treated with enzyme replacement therapy (ERT). This entails replacing the non-working GAA enzyme with a man-made form of it called alglucosidase alfa. This is done by giving the individual IV infusions of the enzyme twice a month. Once a baby or