When Baby has an abnormal test for Pompe Disease
Decreased or absent Acid Alpha-Glucosidase (GAA)

A small sample of your baby's blood was collected soon after birth and sent to the DHEC Public Health Laboratory for testing. This testing is called Newborn Screening. In SC, newborns are screened for several metabolic and genetic disorders. Sometimes, a secondary test is needed to help your doctor determine if your baby has one of these disorders. In most cases, the secondary test will be normal. However, if your baby does have a disorder, prompt treatment will give him or her the best chance to grow up healthy.

Because an enzyme called Acid Alpha-Glucosidase (GAA) was very low or absent in your baby's first test and secondary test results were abnormal, he or she could possibly have Pompe disease (aka Glycogen Storage Disease - GSD Type II).

Please be aware that a low Acid Alpha-Glucosidase (GAA) can also be found in babies who do not have Pompe. However, your baby needs to be evaluated very quickly, as a precaution.

What is Pompe Disease?

Pompe is an inherited condition that affects many different parts of the body. It is considered a lysosomal storage disorder because people with Pompe have lysosomes (the recycling center of each cell) that cannot break down certain types of complex sugars. This causes undigested sugar molecules and other harmful substances to build up in cells throughout the body, resulting in a variety of symptoms.

There are three forms of Pompe (classic infantile onset, non-classic infantile onset, and late onset), which differ in regard to disease severity and age of onset. The symptoms and long-term outcome of each form also vary widely. For the best possible outcome, it is important to detect Pompe early and begin proper treatment immediately.

Signs of infantile-onset Pompe disease (IOPD) begin before or shortly after birth and include:

- Muscle weakness (myopathy)
- Poor muscle tone (hypotonia)
- Failure to gain weight and grow at expected rate (failure to thrive)
- Difficulty breathing
- Trouble feeding
- Respiratory infections
- Hearing problems

Signs of non-classic infantile onset Pompe usually occur by age one and include:

- Delayed motor skills
- Progressive muscle weakness (myopathy)
- Difficulty breathing

Late-onset Pompe disease (LOPD) may develop in childhood, adolescence, or even adulthood. It is also associated with progressive muscular weakness and difficulty breathing. However, the symptoms are usually milder and progress more slowly than the other forms of Pompe.

If your baby shows any of these signs, be sure to contact your baby's doctor immediately.

How will I know if my baby really has Pompe?

If your baby's newborn screening result showed decreased or absent Acid Alpha-Glucosidase (GAA) enzyme activity, the newborn screening test will be repeated, and confirmatory tests will be done to help the doctors determine if your baby has Pompe. Usually, the results of these tests take a few weeks to come back. You may also be referred to a doctor who specializes in this kind of metabolic disorder.

What do I need to do until I know the final results?
Follow your doctor's instructions very carefully.

How is Pompe treated?

Your baby's health care provider may recommend your baby go on enzyme replacement therapy (ERT) to improve the signs and symptoms of Pompe. This treatment aims to supplement the enzymes that are present at very low levels in your baby's lysosomes. ERT can stabilize your baby's organ functions, improve motor skills, and reduce heart enlargement.

Physical Therapy
Physical therapy will be necessary to help infants, children, and adolescents develop motor skills, maintain range of motion, and strengthen muscles and joints.

Respiratory Therapy
Because lung infections, breathing difficulties and heart enlargement are common in children with Pompe, your baby may need to see a pulmonologist (lung doctor), respiratory therapist, or cardiologist (heart doctor).

Dietary Treatments
Many babies with Pompe have trouble feeding and gaining weight. A nutritionist or dietitian can help you monitor your baby's calorie intake and plan a diet that will provide the nutrients he or she needs for proper growth and development. Some children with Pompe are managed on a soft diet, while others may require a feeding tube.

What else should I do to keep my baby as healthy as possible?

Treating illnesses before they become serious and following your baby's treatment plan carefully are the best things you can do to help your baby grow and develop.

Internet References: http://www.babysfirsttest.org/