When Baby has a GAA Pseudo-deficiency

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. Because an enzyme called Acid Alpha-Glucosidase (GAA) was low in your baby's first test, a secondary test was performed:

The secondary screening test indicated your baby has a GAA pseudo-deficiency.

What does it mean to have a *pseudo-deficiency*?

A pseudo-deficiency is a variation in the baby's body which results in lower GAA enzyme activity, but not low enough to cause disease. ***Babies with only a GAA pseudo-deficiency do not have Pompe disease.***

What is Pompe Disease?

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

There are three forms of Pompe Disease (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.

Where can I find more information?

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