Pompe carriers and Pompe Pseudo-deficiency alleles

A small sample of your patient's blood was collected soon after birth and sent to the SC DPH Public Health Laboratory for testing. This blood test is called Newborn Screening. Across the US, newborns are screened for several metabolic and genetic disorders.

A secondary lab test indicated your patient is a carrier of Pompe Disease or has a Pompe pseudo-deficiency allele.

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

Carriers of Pompe disease are individuals who have a variant (mutation) in one of their two *GAA* genes. These individuals still have one *GAA* gene without a mutation. ***Note, carriers of Pompe disease do not have signs or symptoms of Pompe disease.***

However, there is a 1 in 4 (25%) chance a carrier may have a child with Pompe disease if their partner is also a carrier of Pompe. Both parents of a child with Pompe disease are nearly always carriers of the condition.

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

A pseudo-deficiency allele is a change in the *GAA* gene which results in lower GAA enzyme activity, but not low enough to cause Pompe disease. **Note, *babies with only pseudo-deficiency allele(s) do not have Pompe disease. This is a false positive/normal finding*.**

*Babies with a pseudo-deficiency allele **and** other GAA gene variants most likely do not have Pompe disease. However, they still need to undergo further evaluation with a pediatric metabolic geneticist to be certain*.

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 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). The forms differ regarding 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: https://www.acmg.net/PDFLibrary/Pompe.pdf