When Baby has an abnormal test for Mucopolysaccharidosis type 1 (MPS I)

(MPS I 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 doctors determine if your baby has one of these disorders. Because an enzyme called Alpha-L-iduronidase (IDUA) was very low in your baby's first test, a secondary test was performed:

The secondary screening test indicated your baby has a pseudo-deficiency for MPS I.

What does it mean to have a pseudo-deficiency?

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

Babies with a pseudo-deficiency allele and an IDUA gene variant most likely do not have MPS I disease either. But baby may still need further evaluation with a metabolic geneticist to be certain.

What is MPS I?

Mucopolysaccharidosis type 1 (MPS I) is an inherited condition that affects many different parts of the body. It is considered a lysosomal storage disorder (LSD). People with MPS I 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 abnormal symptoms.

Where can I find more information?

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