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

(MPS I Carrier)

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. 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 is a carrier for MPS I.

What does it mean to be a carrier of MPS I?

Carriers of MPS I are individuals who have a variation (change) in one of their two IDUA genes. These individuals still have one IDUA gene without a change. **Carriers of MPS I do not have signs or symptoms of MPS I disease.**

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

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) because 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 molecules 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 References: http://www.babysfirsttest.org/