When Baby has a decreased level of Galactocerebrosidase *(GALC)*
and **an elevated level of Psychosine (PSY)**

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. In most cases, the secondary test will be normal. However, if your baby does have an abnormal test, prompt treatment will give him or her the best chance to grow up healthy.

Because an enzyme called Galactocerebrosidase (GALC) was low and a chemical called Psychosine (PSY) was elevated in your baby's blood, he or she could possibly have a newborn screening condition.

Please be aware that positive newborn screening results can also be found in healthy babies who do not have a condition. However, your baby needs to be evaluated very quickly, as a precaution.

What is Krabbe Disease?

Krabbe disease is a rare, inherited metabolic disorder in which harmful amounts of lipids (fats) build up in various cells and tissues in the body. Those affected by Krabbe disease typically appear healthy until the onset of the disease.

Signs of Infantile Krabbe can begin any time from birth to 3 years of age. Typically, the infantile signs do not become apparent until the baby is between 2 to 6 months of age. Until then, a baby with Krabbe may appear to develop normally. There are two ways in which Krabbe can develop: **Infantile onset** or **Late onset**.

Infantile onset of Krabbe leads to neurological deterioration and without treatment, survival beyond 2 years of age is uncommon. The main symptoms of infantile onset Krabbe are:

- Loss or underdevelopment of motor skills
- Muscle tone becomes floppy
- Hearing loss or sensitivity to loud noises
- Irritability and Vision loss
- Low weight or lower rate of weight gain than children similar in age, a condition known as "failure to thrive"

For the best possible outcome, it is very important to detect Infantile Krabbe early and begin proper treatment immediately.

The **later onset** types of Krabbe begin in childhood or early adulthood. These symptoms have a slower progression than the infantile form. Symptoms may include:

- Vision problems and hearing loss
- Difficulty conducting basic movements
- Muscle rigidity (stiffness) may also be present

The later onset forms have variable conditions, meaning there is no definitive set of stages like the early onset form. If your baby shows any of these signs, be sure to contact your baby's health care provider immediately.

How will I know if my baby really has Krabbe disease?

If your baby's newborn screening result showed abnormal activity, the newborn screening test was repeated, and secondary testing was performed to help doctors determine if your baby may have a condition. Usually, the results of these tests take a few days to come back. You may also be referred to a doctor who specializes in this kind of disorder, called a metabolic specialist/clinical geneticist, or hematologist/oncologist.

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

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.

Where can I get more information?

Internet Reference: https://www.huntershope.org/newborn-screening/nbs-family-guide/