What is Krabbe?

Globoid Cell Leukodystrophy, more commonly known as Krabbe (crab á) disease, is an inherited disorder that affects the central and peripheral nervous systems. Children who inherit the disorder lack an important enzyme (GALC) that is needed for the production of normal myelin (white matter). Myelin is the protective covering of the nerve cells. It is essential to normal bodily function.

For more information about Krabbe disease, please visit www.huntershope.org/krabbeNBS

Hunter James Kelly

Hunter’s Hope was established in 1997 by Jim Kelly, Hall of Fame Quarterback, and his wife Jill, after their infant son, Hunter (2/14/97—8/5/05) was diagnosed with Krabbe Leukodystrophy. The mission of the Foundation includes education, awareness, research, and family support.

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What is Newborn Screening?

A few drops of blood were taken from your baby’s heel and placed on a card that was sent to your state’s newborn screening lab. The results were then sent to your baby’s pediatrician and the hospital where your baby was born.

Newborn screening looks for serious diseases that would not otherwise be detected until the baby got sick. For these diseases, early detection is essential so that treatment can be given before irreversible consequences occur. Newborn screening does not diagnose diseases, but identifies which babies need additional testing to confirm or rule out these diseases. These diseases are very rare, and they are treatable if caught early.

To learn more about Krabbe Newborn Screening, please visit www.huntershope.org/KrabbeNBS

What does a positive screen mean?

If your baby has a positive screen for Krabbe disease, your baby’s doctor or someone from the hospital will contact you. You will be asked to bring your child in for additional testing as soon as possible.

Just because your baby received a positive screen for Krabbe disease does not mean that he/she has the disease. A baby’s immature system may not yet be making the right amount of certain substances. Or, your child may be a carrier of Krabbe. Carriers never show any of the symptoms of the disease, but could potentially pass the gene on to their future children.

The majority of infants with a positive screen for Krabbe do not have the disease.

There are varying forms of the disease and it is important that a diagnosis is made as soon as possible to ensure that appropriate treatment is administered. For infants that are affected by the most severe form of the disease, there is HOPE – through early diagnosis, children who receive treatment before the disease is too far progressed have the potential for a healthy life.

If you have been contacted about your baby’s newborn screening results, your baby needs to be seen by a medical expert as soon as possible.