

KENTUCKY NEWBORN SCREENING PROGRAM

PARENT TEACHING SHEET

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Krabbe Disease

CAUSE

Krabbe Disease is a disorder that prevents the nervous system and brain from functioning normally. This disease is due to a decrease of an enzyme, called galactocerebroside beta-galactosidase (GALC). This enzyme normally helps maintain a protective coating on nerves (myelin) that helps the nerves conduct signals better. When the enzyme doesn't function, toxins build up that damage brain cells and nerve cells throughout the body.

IF NOT TREATED

Signs of Krabbe disease can begin any time from infancy to childhood, but many cases have symptoms starting shortly after birth. This is called early onset Krabbe disease. The major symptoms of early onset Krabbe disease are:

- Loss or underdevelopment of motor skills
- Muscle tone becomes floppy
- Hearing loss or sensitivity to loud noises
- Irritability
- Vision loss

Lower weight or lower rate of weight gain than that of children similar in age, a condition known as "failure to thrive"

TREATMENT OPTIONS

Generally, treatment for the disorder is symptomatic and supportive but may not cure this disease. Physical therapy may help maintain or increase muscle tone and circulation. Medications may be used for treating specific symptoms. For some individuals with Krabbe, detecting it early and beginning treatment prior to the onset of symptoms may help to limit some of the severe health outcomes associated with the condition. The specialists involved in the diagnosis and treatment of Krabbe disease will discuss all options for further testing and treatment.

