



# OHIO DEPARTMENT OF HEALTH

246 North High Street  
Columbus, Ohio 43215

614/466-3543  
[www.odh.ohio.gov](http://www.odh.ohio.gov)

John R. Kasich/Governor

Richard Hodges/Director of Health

July 6, 2016

To: Primary Care Providers  
From: Ohio Department of Health Laboratory

RE: Krabbe disease added to the Ohio Newborn Screening Panel

The Ohio Newborn Screening Program is now screening for Krabbe disease. Krabbe disease is a progressive neurodegenerative disease with a variable age of onset and severity. In the most severe infantile form, symptoms begin in the first few months of life. Affected babies present with irritability, feeding problems, and muscle rigidity. Neurologic deterioration progresses rapidly and death usually occurs in the second year.

Krabbe disease is an autosomal recessive disease that is classified as both a lysosomal storage disease and a leukodystrophy. It is caused by a deficiency of the enzyme, galactocerebrosidase (GALC), which is needed for metabolism of galactolipids. Decreased enzyme activity results in demyelination of the brain and nerve cells throughout the body and accumulation of toxic amounts of psychosine in the lysosomes. There is no cure for Krabbe disease; however, hematopoietic stem cell transplantation may slow the progression of the disease if performed before symptoms present. Without newborn screening, a diagnosis of Krabbe disease cannot be made in time for parents to consider treatment options.

Urgent attention is needed when the newborn screening has identified a baby as at-risk for Krabbe disease. The primary medical provider needs to initiate the diagnostic work-up within 3 days of notification. It is recommended that the medical provider start by consulting with a Regional Genetic Center with expertise in diagnosing Krabbe disease (a list of genetic centers is attached). The baby should be scheduled for diagnostic testing within 48 hours of the consult. The testing may include mutation analysis and sequencing of the GALC gene, galactocerebrosidase enzyme activity, and psychosine levels. It is important that testing be done at a laboratory that has experience in these assays and can provide results within a week to ten days. If clinical evaluation is suggestive of Krabbe disease, the infant will be referred to a pediatric neurologist for further testing. Infants identified with severe infantile Krabbe disease will work with a multidisciplinary team to determine the most appropriate treatment options. Infants identified with later onset forms of Krabbe will be offered periodic neurologic evaluations to assess current clinical status.

The Ohio Department of Health is mandated by law (Ohio Administrative Code 3701-55) to perform Krabbe screening of specimens received by the laboratory on and after July 1, 2016. The law gives parents/guardians the right to refuse screening for Krabbe disease. The law does not allow parents to refuse screening for any other conditions included on the Ohio Newborn Screening Panel except for religious objection. Parents must be given information about their right to refuse this screening prior to the collection of the blood specimen. The ODH has developed an insert, "*Screening for Infantile Krabbe Disease*", for the "*Why Must My Baby Be Screened*" pamphlet to inform families about screening for Krabbe disease and their right to refuse this screening. (The pamphlet and Krabbe insert can be viewed and downloaded from the Ohio Newborn Screening web site: <http://www.odh.ohio.gov/odhPrograms/phl/newbrn/nbrn1.aspx>)

Please contact the Newborn Screening Program if you have any questions.

Regards,  
Bureau of Public Health Laboratory  
8995 East Main St, Bldg 22, Reynoldsburg, OH 43068  
Phone: 1-888-ODH-LABS; 614-466-2278  
Email: [odhlabs@odh.ohio.gov](mailto:odhlabs@odh.ohio.gov)

# Ohio Department of Health Newborn Screening Program Krabbe Disease Evaluation

## **Cincinnati**

- Cincinnati Children's Hospital Medical Center  
Biochemical Genetics  
Division of Human Genetics  
3333 Burnet Avenue (MLC: 4006)  
Cincinnati, OH 45229  
P (513) 636-4760 F (513) 636-7297  
After 5pm and on weekends,  
Physician Priority Link (513) 636-7997

- University Hospitals Case Medical Center  
Rainbow Babies and Children's Hospital  
Center for Human Genetics  
11000 Euclid Avenue, Lakeside 1500  
Cleveland, OH 44106  
P (216) 844-3936 F (216) 844-7497  
After 5pm and on weekends,  
On-call Line (216) 292-9597

## **Columbus**

- Nationwide Children's Hospital  
700 Children's Drive  
Columbus, OH 43205  
P (614) 722-3543 F (614) 722-3546  
After 5pm and on weekends,  
PCTC Line (614) 355-0221

## **Northern Ohio**

University Hospitals Case Medical Center  
Rainbow Babies and Children's Hospital  
(Cleveland) - see contact information above -  
will perform clinical evaluation for Krabbe  
disease for babies in the Northern Ohio. This  
includes babies who would otherwise be  
referred to:

- Akron Children's Hospital
- Cleveland Clinic Foundation
- MetroHealth Medical Center
- Toledo Children's Hospital
- Rocket Pediatrics (Toledo)

## **Dayton**

- Dayton Children's Hospital  
One Children's Plaza  
Dayton, OH 45404-1815  
P (937) 641-3800 F (937) 641-5325  
After hours & weekends,  
On-call Line (937) 641-3000  
Ask to page Metabolic doctor on-call

## **North Western Ohio**

University Hospitals Case Medical Center  
Rainbow Babies and Children's Hospital  
(Cleveland) and Nationwide Children's Hospital  
(Columbus) - see contact information above -  
will perform clinical evaluation for Krabbe  
disease for babies in North West Ohio. This  
includes babies who would otherwise be  
referred to:

- Toledo Children's Hospital
- Rocket Pediatrics (Toledo)

## **Cleveland**