

# Krabbe Disease

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*~Service Learning Initiative in Biochemistry 5614 (Autumn 2018)~*

# History and Occurrence

## History

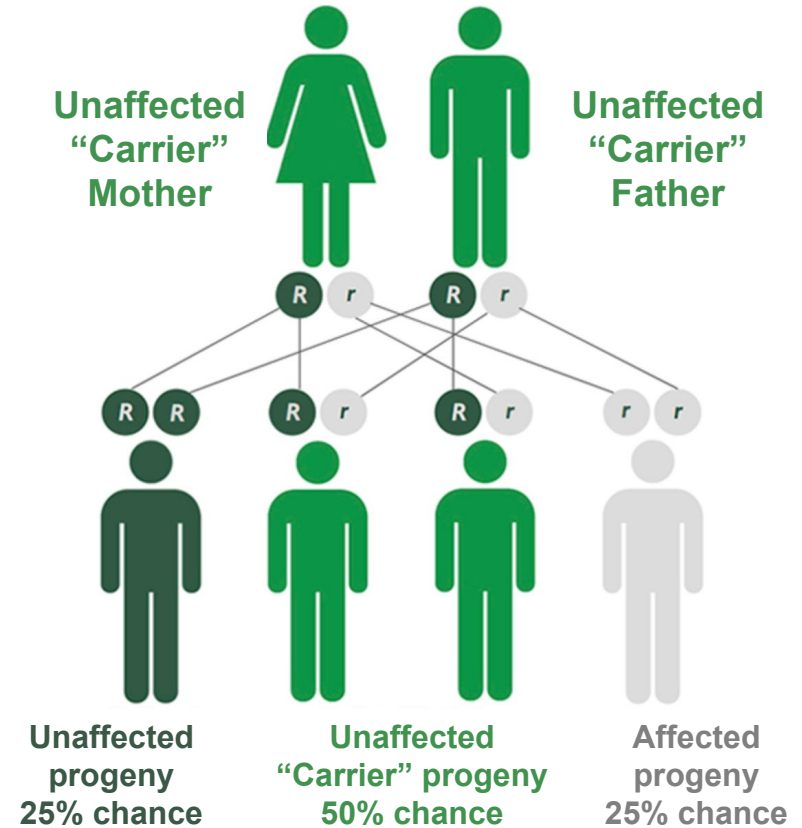
- First case published in 1916 by Knud Krabbe

## Occurrence

- 1/100,000 individuals in the US
- Several isolated communities in Israel have greater incidence: (1/160)
- Males and females at equal risk

# Krabbe: An autosomal recessive disorder

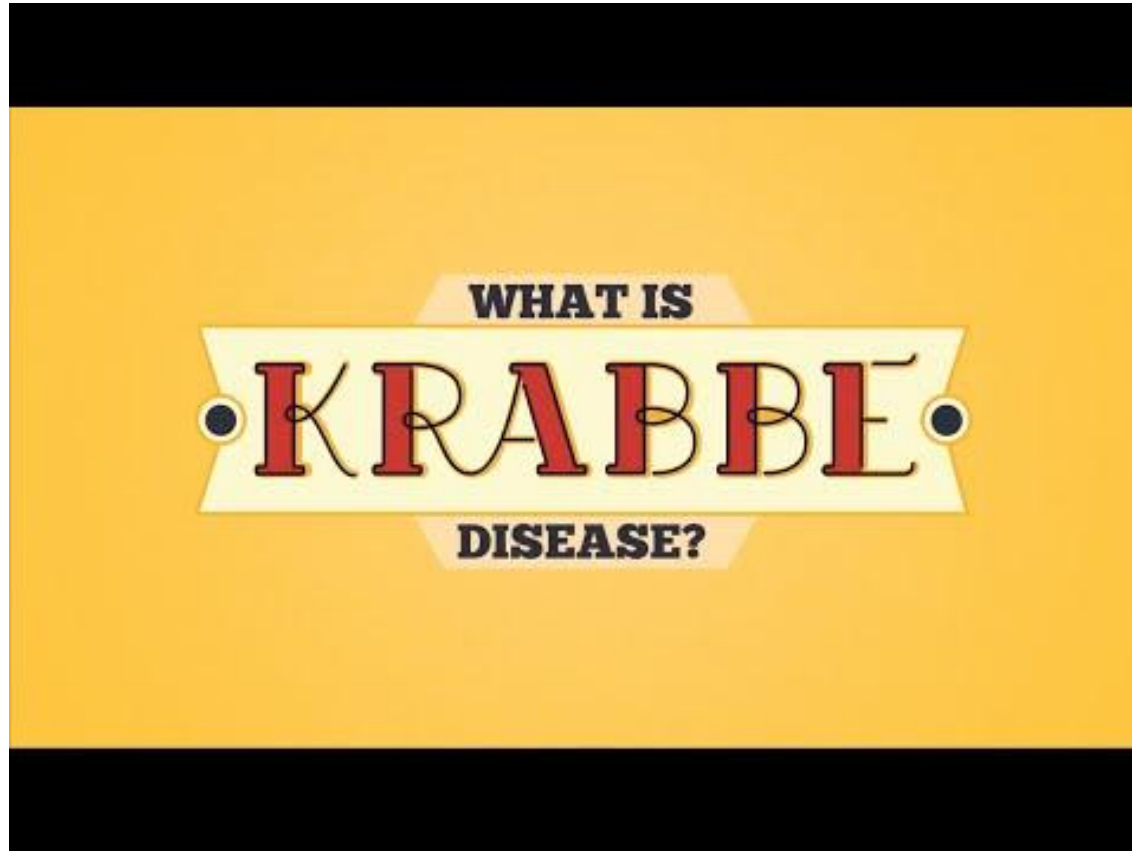
Video on Mendelian Inheritance



HNEkidshealth. "Autosomal Recessive Inheritance - Genetics." YouTube, YouTube, 30 Mar. 2015, [www.youtube.com/watch?v=Nv6qUsKYodA](http://www.youtube.com/watch?v=Nv6qUsKYodA).

How do you get Krabbe Disease? Digital image retrieved October 30, 2018 from <https://www.huntershope.org/family-care/leukodystrophies/krabbe-disease/>

# Krabbe Disease: An Overview

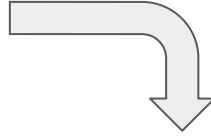


Judsons Legacy. What is Krabbe Disease? Video File retrieved 30 October 2018 from

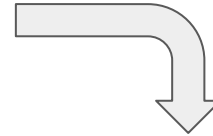
<https://www.youtube.com/watch?v=-73qE2mHYWY&feature=youtu.be>

# Biochemical Features

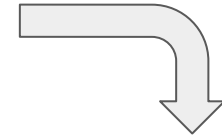
Galactosylceramidase  
(GALC) deficiency



Buildup of galactosyl-  
ceramide and psychosine



Buildup of  
galactolipids



Impairs myelin formation  
(affects neuronal function)

Myelin sheath  
functions like  
insulation on a wire

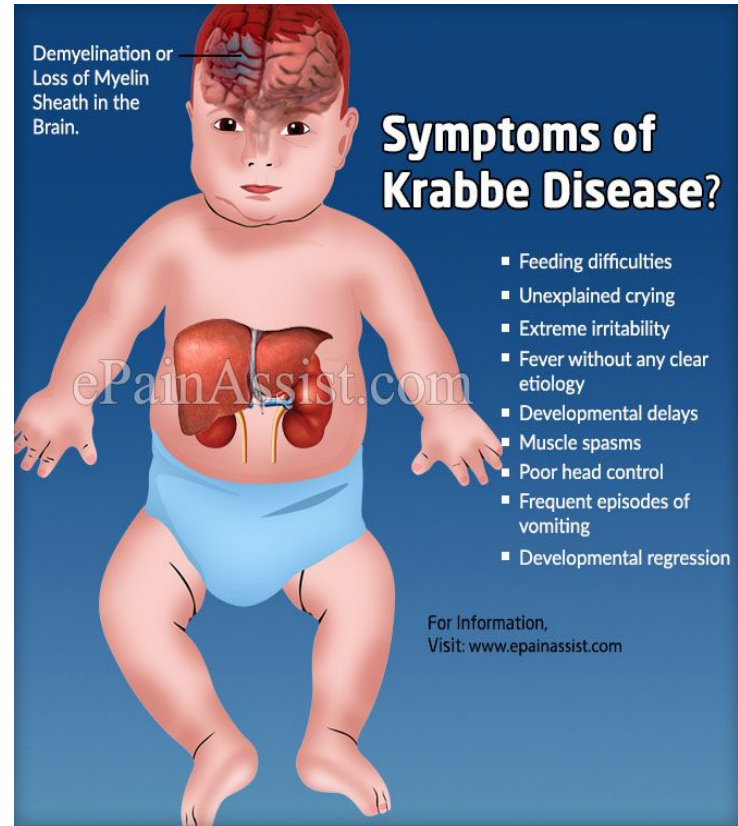


Digital image retrieved 30 November 2018 from

<https://hackaday.io/project/11951/instructions?page=2>

# Symptoms (in infants)

- Difficulty in feeding
- Fever without infection/etiology
- Decreased alertness
- Delayed intellectual and physical development
- Hearing and vision loss



Kerker, P.. What are the Symptoms of Krabbe Disease?

Digital image Retrieved 30 October 2018 from

<https://www.epainassist.com/genetic-disorders/what-is-krabbe-disease>

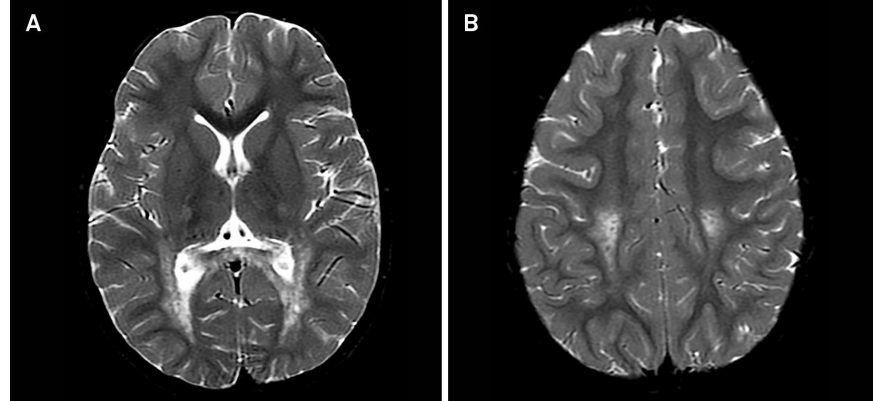
# Neonatal diagnosis

## Newborn screening test:

- Blood test assessing GALC enzyme activity in leukocytes
  - Low activity indicates Krabbe disease

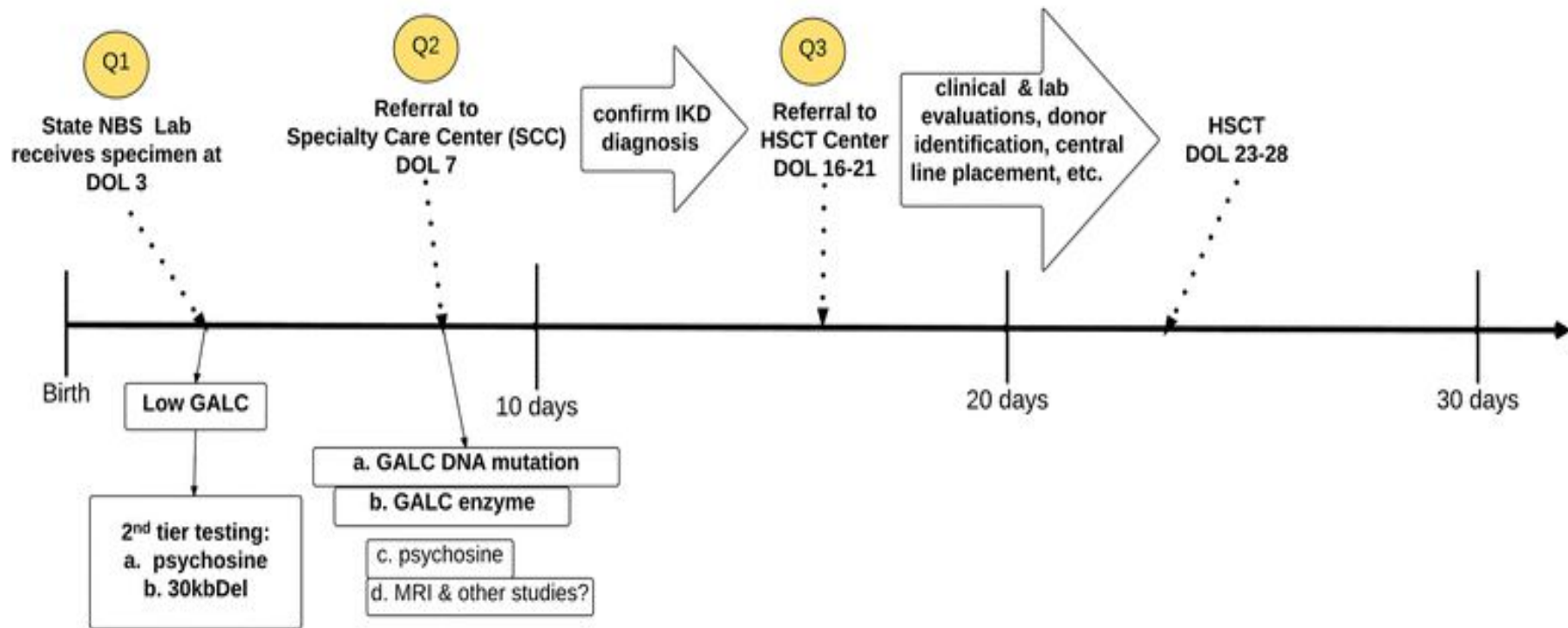
## Supporting tests

- MRI or other brain scans
- Nerve conduction studies
- Eye exam
- Genetic molecular testing



<https://healthjade.com/krabbe-disease/>

# Krabbe Disease Newborn Screening Timeline





# Prognosis

- Infantile Krabbe Disease: Onset before the age of one
  - Usually fatal before second birthday
  - Bone marrow transplant or umbilical cord blood stem cells can prolong life



By donating your  
**baby's cord blood,**  
you give  
**patients hope.**

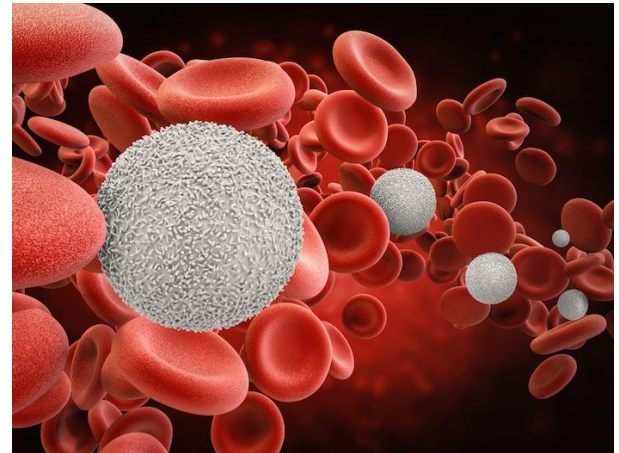
BE  THE MATCH.

“Cord Blood and Transplants.” Be the Match,  
National Marrow Donor Program,

[bethematch.org/transplant-basics/cord-blood-and-transplants/](http://bethematch.org/transplant-basics/cord-blood-and-transplants/)

# Therapy

- Hematopoietic Stem Cell Transplantation (HSCT)
  - Replaces affected cells with cells that have normal GALC function
  - Umbilical cord blood stem cell or bone marrow transplantation
  - Has to be repeated; lasts 2-4 years after treatment
- Anticonvulsants to manage seizures
- Drugs to improve muscle movement
- Physical Therapy (Supportive Treatment)



# Support groups

- The Legacy of Angels Foundation (TLOAF)
- Hunter's Hope Foundation
- United Leukodystrophy Foundation
- The Peace, Love & Trevor Foundation
- The Jackson Project



The Legacy of Angels  
FOUNDATION



United  
Leukodystrophy  
Foundation



THE  
**TREVOR**  
PROJECT



# Krabbe Disease: Project Team

<b>Name</b>	<b>Project Role</b>
Niki Grotewold (Text)	Symptoms
Erika Lindner (Illustrations)	Media/videos
Dan Branch	Prognosis
Sammy Gilbert	History
Miranda Somers	Occurrence, provenance, and genetics
Kaitlin Conner	Diagnosis
Jackie Fontana	Media/videos
Allison Webb	Biochemical Features
Jack Brooks	Therapy