# Krabbe Disease

Krabbe’s disease is a sphingolipidosis caused by a deficiency of the enzyme galactocerebrosidase. This enzyme is required for the catabolism of galactocerebroside to ceramide and galactose and a deficiency of the enzyme causes accumulation of unmetabolized galactocerebroside which affects the growth and development of the myelin sheath around nerves. Infants with Krabbe’s disease are normal at birth but begin to show symptoms between the ages of 3 and 6 months with irritability, fevers, feeding difficulties, and developmental delay. Other symptoms include optic atrophy and peripheral neuropathy. The disease can be diagnosed by finding globoid cells, which are engorged macrophages with multiple nuclei in the parenchyma and around blood vessels.




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## Pathophysiology

**Autosomal Recessive**
Recessive-chocolate
Krabbe Disease is inherited in an autosomal recessive modality. This means two copies of the abnormal gene must be present in order for the disease to develop.

**Galactocerebrosidase Deficiency**
Galaxy-cerebro-guy-on-side
This enzyme is required for the catabolism of galactocerebroside to ceramide and galactose and a deficiency of the enzyme causes accumulation of unmetabolized galactocerebroside which affects the growth and development of the myelin sheath around nerves.

**Increased Psychosine**
Up-arrow Psycho-sun
Galactocerebrosidase normally breaks down psychosine, and a lack of the former leads to an increased buildup of psychosine. Increased psychosine levels are believed to lead to widespread destruction of oligodendroglia in the CNS and to subsequent demyelination, as psychosine preferentially accumulates in white matter.

## Signs and Symptoms

**Developmental Delay**
Developmental-bus
Infants with Krabbe's disease are normal at birth but begin to show symptoms between the ages of 3 and 6 months with irritability, fevers, feeding difficulties, and developmental delay. The developmental delay is directly related to damage of the myelin sheaths caused by accumulation of galactocerebroside.

**Peripheral Neuropathy**
Purple-wavy Neuron-extremities
Peripheral neuropathy is a general term that describes damage to the nerves of the peripheral nervous system. Symptoms can include abnormal sensory function and motor symptoms. Peripheral neuropathy is a common finding in Krabbe's disease due to damage of the myelin sheaths in the peripheral nerves.

**Optic Atrophy**
Optic @-trophy glasses
Optic atrophy occurs when there is loss of some or most of the fibers in the optic nerve. Optic atrophy can lead to vision changes or vision loss and is a common finding in Krabbe's disease due to damage of myelin.

## Diagnosis

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**Globoid Cells**

Globe

Globoid cells are engorged macrophages with multiple nuclei found in the parenchyma and around blood vessels in individuals with Krabbe's disease. Finding globoid cells can be diagnostic of Krabbe's disease.

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