Krabbe Disease

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Krabbe Disease Information Page.

Krabbe disease is a rare, inherited metabolic disorder in which harmful amounts of lipids (fatty materials such as oils and waxes) build up in various cells and tissues in the body and destroy brain cells. Krabbe disease, also called globoid cell leukodystrophy, is characterized by globoid cells (cells that have more than one nucleus) that break down the nerve's protective myelin coating. Krabbe disease is caused by a deficiency of *galactocerebrosidase*, an essential enzyme for myelin metabolism. The disease most often affects infants, with onset before age 6 months, but can occur in adolescence or adulthood. Symptoms include:

- severe deterioration of mental and motor skills,
- muscle weakness,
- hypertonia (inability of a muscle to stretch),
- myoclonic seizures (sudden, shock-like contractions of the limbs),
- spasticity (involuntary and awkward movement),
- unexplained fever,
- blindness,
- difficulty with swallowing,
- deafness.