

Krabbe's Leukodystrophy is a rare inherited lipid storage disorder caused by a deficiency of the enzyme galactocerebrosidase (GALC), which is necessary for the breakdown (metabolism) of the sphingolipids galactosylceramide and psychosine. Failure to break down these sphingolipids results in degeneration of the myelin sheath surrounding nerves in the brain (demyelination). Characteristic globoid cells appear in affected areas of the brain. This metabolic disorder is characterized by progressive neurological dysfunction such as mental retardation, paralysis, blindness, deafness and paralysis of certain facial muscles (pseudobulbar palsy). Krabbe's Leukodystrophy is inherited as an autosomal recessive trait. About 1 in 40,000 newborn babies in the United States is affected with Krabbe's Leukodystrophy. Males are affected as often as females. Krabbe's Leukodystrophy can be diagnosed by testing the activity of the enzyme galactocerebrosidase (galactosylceramidase) in fibroblast cells obtained from an infant or from a fetus by amniocentesis.