

## A Review of the Pathogenesis, Clinical Manifestations and Management of Lysosomal Storage Disorder

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### ABSTRACT

**Background:** KD, or Globoid cell leukodystrophy, is a rare autosomal recessive neurodegenerative disease that results from a mutation of chromosome 14q31. Deficiency of galactocerebrosidase (GALC) enzyme, which breaks down galactosyl-ceramide leads to its accumulation and the generation of psychosine from an alternate catabolic pathway. Cytotoxicity of psychosine in the nervous system leads to severe neuronal damage. Diagnosis is best confirmed with fibroblast qualitative and quantitative analysis of galactocerebrosidase. The disease has no definitive cure and its prognosis is poor.

**Objective:** To elucidate a rare lysosomal storage disorder.

**Methodology:** Seventy-six publications were selected and 45 fits into the inclusion criteria [Medline (OvidSp), PubMed (USNLM), and Google Scholar.

**Discussion:** A lysosomal storage disorder with known variants from different molecular mechanisms, which include mutations of N279TGALC, HEK293T, Y551S, I546T, G270D, and WT GALC genes, Abnormalities occur in enzyme synthesis, endoplasmic reticulum trapping, vesicular transport, and/or post-translational modifications of co-factors/active sites binding.

Early Infantile KD: Children <6 months, characterized by neurological manifestations.

Late Infantile KD: 6 months to 3 years with symptoms similar to early Infantile KD.

Adolescent KD: >3 years of age. Presents with regression of motor skills and slower progression than the infantile-onset subtype.

The adult-onset KD: Visual problems followed by muscle stiffness, ataxia, and pain misdiagnosed as multiple sclerosis.

Investigated by enzyme activity, psychosine concentration, molecular testing, nerve conduction tests, brain imaging, and lumbar puncture. Treatment options include stem-cell transplantation and gene therapy. Symptomatic relief for irritability, seizure, and pain with muscles relaxant and feeding assistance may be helpful.

**Conclusion:** KD is an autosomal recessive neurodegenerative disorder with known variants. KD is primarily caused by the deletion of the GALC gene found on chromosome 14 (14q31). The mutation results in an enzyme deficiency, and the accumulation of psychosine, a toxic substance in neural cells. KD has no definitive cure.

**Keywords:** GALC, Globoid cell leukodystrophy, Autosomal

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