

[Open Peer Review on Qeios](#)

2-Hydroxyglutaric Aciduria

National Cancer Institute

Source

National Cancer Institute. *2-Hydroxyglutaric Aciduria*. NCI Thesaurus. Code C128187.

A group of genetic disorders characterized by elevated urinary concentrations of 2-hydroxyglutaric acid. Three different types have been identified based on the stereoisomeric composition of the elevated alpha-hydroxyglutaric acid metabolites. Additionally, the disease may be categorized by the genetic mutation that is causative. Genes associated with 2-hydroxyglutaric aciduria are L2HGDH, D2HGDH, IDH2, and/or SLC25A1. Generally, there is nervous system involvement, but the clinical manifestations are variable and are dependent on the specific type of defect present.