

What is Glut1 Deficiency?

Glut1 Deficiency is a rare genetic condition that affects brain metabolism. It is caused by a mutation in the SLC2A1 gene, which regulates the glucose transporter protein type 1 (Glut1). Glut1 is the principal transporter of glucose, the primary source of energy, across the blood-brain barrier. More than 100 different types of mutations and deletions of this gene have been found to date in Glut1 Deficiency patients.

Impaired glucose transport associated with Glut1 Deficiency creates an energy crisis in the brain and often results in seizures, movement disorders, and developmental delays. The standard of care treatment is a ketogenic diet. This low-carbohydrate, high-fat diet causes the body to produce ketones, which are used as an energy source by the brain and other tissues when glucose is limited.

For more information:

GeneReviews®

www.ncbi.nlm.nih.gov/books/NBK1430/

Genetics Home Reference

ghr.nlm.nih.gov/condition/glut1-deficiency-syndrome



www.G1DFoundation.org

Diagnosing Glut1 Deficiency

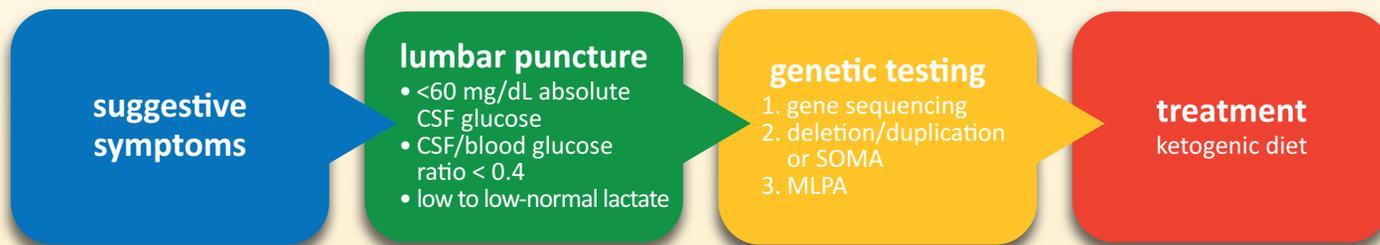
Glut1 Deficiency can be diagnosed through low glucose levels in the cerebral spinal fluid, a low blood to CSF glucose ratio, genetic testing, and a specialized red blood cell uptake assay.

Recommended diagnostic protocol is to screen suspected patients with CSF/blood analysis and follow up with genetic testing to confirm.

Referrals for genetic testing can be facilitated and expedited by involving neurology, metabolic, and genetic specialists from larger institutions experienced in working with insurance and

genetic testing companies to secure coverage. Gene testing companies should be selected for a comprehensive approach including screening, more sensitive testing and physician/geneticist collaboration.

Approximately 90% of cases are caused by de novo mutations, although Glut1 Deficiency can be inherited through an autosomal dominant pattern with rare cases of autosomal recessive inheritance. Genetic analysis of additional family members may be warranted.



Visit www.G1DFoundation.org to learn more about diagnosing and treating G1D.