Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

GLUT1 Deficiency Syndrome

GLUT1 Deficiency Syndrome is a rare disorder characterized by infantile seizures, developmental delay and learning disabilities. Glucose Transporters are part of glycoproteins, which are involved in transporting glucose to most cells in the body. This disorder is caused by defects on the Glucose Transporter Type 1 Deficiency gene (SLC2A1) which encodes the Glut-1 protein.

Symptoms of GLUT1 Deficiency Syndrome are not present at birth and do not usually show until infancy with frequent seizures. The first sign may be rapid eye movements. Symptoms include low glucose levels in the fluid that surrounds the brain and spinal cord, a small head and neurological symptoms. A non-epileptic form of this disorder accounts for approximately 10% of cases of GLUT1 Deficiency Syndrome

Synonyms

Alternative names for this condition are:

- De Vivo Disease
- Glucose Transporter Protein Syndrome
- Glucose Transporter Type 1 Deficiency Syndrome
- GLUT1 Deficiency
- GLUT1-DS

Further information about this condition is available from Climb.

Disclaimer

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