Neurodevelopment in Glut-1
Understanding the Impact on Brain Function and Development

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How does Glut-1 affect the brain and development?
GLUT1 deficiency syndrome (GLUT1DS, OMIM 606777) is a treatable epileptic encephalopathy resulting from impaired glucose transport into the brain. The essential biochemical finding is a low glucose concentration in the cerebrospinal fluid (CSF; hypoglycorrhachia; mean 1.7 [SD 0.3mmol/L]) in the setting of normal glucose in the blood (normoglycaemia)

What is the Role of Glut 1 and the brain
Glucose is the primary energy source for brain cells. It is crucial for neuronal growth, synapse formation, and neurotransmitter synthesis. Therefore, the consequences of Impaired Glucose Transport include energy deficit in brain cells. Lack of glucose at the propre time in development can lead to disruption in normal brain development and function.

What are the Symptoms of GLUT1 Deficiency?
The symptoms are universally neurological including seizures which can appear in infancy and be accompanied by abnormal eye movements. Many neurologists do not consider these typical of Glut-1 and do not send testing. For those that present later, there may be developmental delays including in the motor systems with ataxia or unsteady gait and dystonia, or abnormal movements. Other symptoms included cognitive impairments and speech difficulties.

Diagnosis of GLUT1 Deficiency
The diagnosis used to require a Lumbar puncture, or spinal tap to measure the glucose in the CSF and compare with that in the blood. However, now the gene is known and can be tested through blood or cheek swab. The gene is called SLC2A1.
It may be part of an epilepsy panel or found when doing a whole exome or whole genome test.

Treatment Approaches
So far, the mainstay of treatment depends on a Ketogenic Diet which is a High-fat, low-carbohydrate diet to provide alternative energy source (ketones) for the brain.
Other Therapeutic Strategies include the use of Antiseizure medications as well as supportive therapies (physical, occupational, speech therapy). Currently, scientists are looking into gene therapy and novel pharmacological approaches. We don't' know the long term outcome in adults, but data is accumulating. Because it is a treatable disorder with diet, there are efforts to include it on the newborn screening program.