Frequently asked questions about glucose transporter type 1 deficiency syndrome

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What is Glucose Transporter Deficiency Syndrome?

Glucose transporter type 1 deficiency syndrome (GLUT1DS) is a defect in carrying glucose into the brain. Glucose is the essential fuel for the brain and is provided from carbohydrates in the diet. The transport defect results in an "energy failure" in brain.

Is it contagious?

GLUT1 is not contagious. For most people, it is a novel genetic mutation in a particular gene known as SLC2A1. Some cases follow an autosomal-dominant transmission. This means that you only need to get the abnormal gene from one parent in order to inherit the condition. About 30% of individuals do not have a gene mutation and therefore the cause is unknown.

How is it diagnosed?

It can be diagnosed by a low concentration of glucose in the spinal fluid. It can also be diagnosed by testing of glucose transport in red blood cells. Genetic testing for the SLC2A1 gene confirms this syndrome in about 70% of cases.

What are the symptoms?

Most patients show symptoms of this syndrome in infancy and early childhood with epilepsy, global developmental delay, and a complex movement disorder.

Is this syndrome common?

Glut1 is considered a rare condition however it is likely that it is not being tested for and therefore it may be more common than thought.

What is the treatment?

According to a recent publication in 2008 by a group of medical experts, "The Ketogenic Diet is the treatment of choice for Glut 1". These experts also agree that in the vast majority of patients, seizure control by the ketogenic diet is sufficient and that anti-seizure medication can be withdrawn. The ketogenic diet is a high-fat, low-carbohydrate, moderate protein diet. More information about this diet can be found under the FAQ link of our web-site:charliefoundation.org.

What benefits can be expected if the ketogenic diet is started?

Increased alertness and ability to learn are usually experienced soon after starting the ketogenic diet. In addition, improvement in muscle strength and walking are often experienced.

Detecting this disorder early is the key to helping the brain receive the energy it needs to grow and to develop.

How long should the diet be followed?

The diet should be maintained strictly especially during childhood years. Some doctors agree that a more liberal diet may be used during adulthood such as lower ratios of 2:1 or 1:1, the Low Glycemic Index Treatment or the Modified Atkins Diet.