

GLUT1 DEFICIENCY FOUNDATION

Reaching for a Bright Future



www.G1DFoundation.org

Shedding Light

As parents, we know that our children depend on us for everything. It is our purpose and pleasure to help them grow, learn and thrive. But what happens when they don't progress as expected, or if they have alarming symptoms that don't respond well to treatment? We look for answers, but sometimes those answers are difficult to find. This booklet aims to help shed light on the rare genetic disorder known as Glut1 Deficiency.

What is Glut1 Deficiency?

Glucose Transporter Type 1 Deficiency Syndrome (Glut1 Deficiency, Glut1 DS, G1D or De Vivo Disease) is a genetic disorder that impairs brain metabolism. Glut1 (a protein) is responsible for transporting glucose (a sugar) across the blood-brain tissue barrier. The Glut1 protein is made by the SLC2A1 gene, located on chromosome 1. If this gene is damaged by a mutation, the protein is not made, and glu-

cose cannot be transported into the brain cells. Glucose is the primary source of fuel for the brain. A metabolic fuel source is necessary for cells to make energy. Patients with Glut1 Deficiency have insufficient cellular energy to permit normal brain growth and function.

Think of it this way: the Glut1 Deficiency brain is always being starved and cannot perform its brain functions adequately. What are these brain functions? They include our ability to think, learn, socialize and move. Disturbances in these functions represent the symptoms and signs of Glut1 Deficiency.



Signs and Symptoms

Glut1 Deficiency is manifested by an array of signs and symptoms, and these neurological findings may vary considerably from one patient to another. Some symptoms may be present all the time (like walking difficulties), while other signs may come and go (like seizures or poor balance). These findings can be clustered under three major domains: **cognition, behavior** and **movement**.

Cognitive symptoms affect intelligence and can range from subtle learning difficulties to severe intellectual disabilities. Examples include:

- Speech and language development;
- Learning colors, letters and numbers; and
- Learning to count and read

Behavioral symptoms affect relations with other people.

Attention span, tractability, mental and motor activity, and social relatedness influence our success in group settings like school. ADHD can be a bothersome set of behavioral symptoms.

Movement symptoms may affect the quality or quantity of motor activity.

Walking may be difficult because legs are stiff (spasticity), balance is poor (ataxia) or posture is twisted (dystonia). These abnormalities may be constant or intermittent.

Other intermittent symptoms may include headaches, confusion, loss of energy or different types of seizures.



Diagnosis

In order to make a proper and early diagnosis, it is important to know the various symptoms that Glut1 Deficiency causes. However, other diseases may also share some of these signs and symptoms; sorting out diagnostic possibilities is the art of medicine. When Glut1 Deficiency is suspected, a lumbar puncture (spinal tap) should be performed. The spinal fluid concentrations of glucose and lactate, if lower than normal, support the clinical suspicion and justify further laboratory tests. Glucose transport can be analyzed in the red blood cells, as the protein is identical to that found in the brain. Gene-sequencing analysis can also be performed to look for a genetic mutation in the SLC2A1 gene.



Treatments

Although there is currently no cure for Glut1 Deficiency, there are effective treatment methods which nourish the growing brain and prevent and/or control the signs and symptoms. The Ketogenic Diet is currently the core of this treatment. Anti-seizure medications are generally not effective, since they do not provide nourishment to the starved brain.

Ketogenic Diet

The Ketogenic Diet was created in 1921 to control seizures. It is a high-fat, low-protein, low-carbohydrate diet that favors the burning of fat for energy in lieu of glucose. Ketone bodies are formed when fat is metabolized, passing into the brain and replacing glucose as a source of energy. In 1967, it was proven that the brain could use these ketones as an alternate fuel source for energy. Even today, we know that the brain only has two choices for metabolic fuel: glucose is the preferred fuel

and ketones are the alternative fuel. When glucose cannot be used effectively, the brain needs an equivalent amount of ketones to nourish it properly. Early diagnosis and treatment is the key to protecting brain growth and function during development. The Ketogenic Diet mitigates many of the symptoms associated with Glut1 Deficiency. The bigger challenge, however, is feeding the brain adequately to facilitate normal growth and development. The Ketogenic Diet must be carefully crafted and tailored to meet the needs of each patient and should only be used under the care of medical professionals.



Therapy Benefits

Rehabilitative services are beneficial since most Glut1 Deficiency patients experience movement disturbances as well as speech and language disorders. Occupational, physical, and speech/language therapies are very beneficial. Therapists are often able to co-treat patients with integrated interventions.



Learning and School Performance

Although the ability levels among Glut1 Deficiency patients vary greatly, studies have proven that clinical features are common in the majority of cases. It is very important to emphasize the strengths as you work to remediate the weaknesses.

Weak areas include:

- Lowered IQ and adaptive-behavior scores;
- Expressive-language deficits;
- Weaknesses in fine-motor skills;
- Limited visual attention to details; and
- Difficulties in seeing the “big picture”

Strong areas include:

- Receptive language or understanding;
- Social skills;
- Fun-loving and empathetic personalities;
- Playful sense of humor; and
- Varied interests

Resources

Glut1 Deficiency Foundation www.g1dfoundation.org

Mission: The Glut1 Deficiency Foundation is a volunteer, nonprofit family organization dedicated to:

- Educating others about Glut1 Deficiency by creating a forum for sharing support, experiences, resources, and information between patients, families, and healthcare professionals.
- Increasing awareness of and advocacy for Glut1 Deficiency.
- Supporting and funding researchers as they work for a cure.

Additional Resources

www.charliefoundation.org
www.childbrainfoundation.org
www.colleengiblinfound.org
www.glut1.de
www.matthewsfriends.org
www.milestonesforchildren.org

Glut1 Deficiency Research Center Locations

North America



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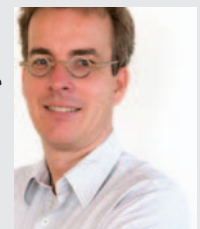


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