Adult Glut1 Deficiency Syndrome Survey

Participation in this survey is voluntary. Please complete this survey if you are 18 years of age or older with Glut1 Deficiency Syndrome (G1DS). One survey per person with G1DS, please. Names are not recorded. Identifying individual information is not shared. Your completion of the survey or questionnaire will serve as your consent to be in this research study.

General Information:

1. My age:_______
2. My gender: ❑ Male ❑ Female
3. My age when Glut1 Deficiency Syndrome was diagnosed:_______
4. My Glut1 Deficiency Syndrome was diagnosed by:
   - ❑ Lumbar puncture
   - ❑ Genetic Testing – SLC2A1 autosomal dominant new mutation
   - ❑ Genetic Testing – SLC2A1 autosomal dominant inherited mutation
   - ❑ Genetic Testing – SLC2A1 autosomal recessive mutation
   - ❑ Based on symptoms
   - ❑ Other: ____

Thank you for your participation in this survey.

Please continue and complete the rest of the survey.
We will share the results at the next Glut1 Deficiency Foundation meeting.

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To preserve anonymity, please return survey via email to info@G1Dfoundation.org OR mail to:
Glut1 Deficiency Foundation • PO Box 737 • Owingsville, KY 40360

Glut1 Deficiency Syndrome Symptoms:

1. Do you have any of the following symptoms? (*check all that apply*)
   - ❑ Seizures
   - ❑ Dystonia
   - ❑ Parkinsonian symptoms
   - ❑ Migraine
   - ❑ Mobility issues
   - ❑ Memory difficulties
   - ❑ Depression
   - ❑ Anxiety
   - ❑ Paroxysmal exertional dyskinesias (PED)
   - ❑ Alternating Hemiplegia
   - ❑ Ataxia
   - ❑ Writer's Cramp
   - ❑ Cognitive difficulties
   - ❑ Speech difficulties
   - ❑ Obsessive-compulsive tendencies
   - ❑ Sleep difficulties
   - ❑ Other (please write in): ______________________________

2. Did you have unusual eye/head movements as an infant? ❑ YES ❑ NO

3. Have you noticed any triggers for your symptoms? ❑ YES ❑ NO
   If yes, what are the triggers? ______________________________
   ____
   ____

4. Did symptoms change at puberty? ❑ YES ❑ NO
   If yes, how did symptoms change? ______________________________
   ____
   ____

5. If female, do symptoms relate to your menstrual cycle? ❑ YES ❑ NO
   If yes, how are they related? ______________________________
   ____
   ____

6. Compared to childhood, if applicable, how are seizures now?
   - ❑ 100% gone (*seizure-free*)
   - ❑ 90-99% gone (*once in a while*)
   - ❑ 50-90% better (*improved, but still frequent*)
   - ❑ 0-50% improved (*really not better*)
   - ❑ Worse
   - ❑ Seizures began as an adult
   - ❑ Never had seizures

7. Compared to childhood, if applicable, how are abnormal movements now?
   - ❑ 100% gone (*free of abnormal movements*)
   - ❑ 90-99% gone (*once in a while*)
   - ❑ 50-90% better (*improved, but still frequent*)
   - ❑ 0-50% improved (*really not better*)
   - ❑ Worse
   - ❑ Movement began as an adulthood
   - ❑ Never had abnormal movements

8. Have you been given any diagnosis other than Glut1 Deficiency? ❑ YES ❑ NO
   If yes, please list: ______________________________
   ____
   ____
   ____
   ____
   ____

Have you transitioned to an adult neurologist? ❑ YES ❑ NO
If yes, was this a smooth transition? ❑ YES ❑ NO

DIET INFORMATION:

1. What diet are you receiving currently **OR** have you tried in the past?

CURRENT	PAST	
❑	❑	Ketogenic Diet (*circle ratio*): 3:1 4:1 Other ________
❑	❑	Modified Ketogenic Diet (*circle ratio*): 1:1 2:1 Other ________
❑	❑	Modified Atkins Diet – Net Carbohydrates: _____g/day
❑	❑	Low Glycemic Index Treatment
❑	❑	Medium Chain Triglyceride (MCT) Diet
❑	❑	Other (*please specify*): ______________________________
❑	❑	None/Regular

2. If you are not receiving any of these diets, why was it stopped or not started?
   ____
   ____

If you have answered NONE to 1 and 2, skip to MEDICATIONS.

3. Age at start of a ketogenic diet: __________
4. If you have switched diets, at what age? __________
   From what diet to what diet? ______________________________
5. Are you followed at a ketogenic diet center? ❑ YES ❑ NO
6. Do you have an adult dietitian/nutritionist? ❑ YES ❑ NO
7. Has your ketogenic diet center discussed coming off the diet ever? ❑ YES ❑ NO
   If no, have they specifically said it's a lifelong treatment? ❑ YES ❑ NO
   Do you plan to stop the diet ever? ❑ YES ❑ NO ❑ NOT SURE
8. Has your ketogenic diet center discussed transitioning to the MAD? ❑ YES ❑ NO
9. Do you check ketones through: ❑ URINE ❑ BLOOD ❑ NEITHER
   If yes, what is an effective level for optimal symptom control? __________
10. Do you see a relationship between seizures and ketones?
    ❑ YES ❑ NO ❑ NOT SURE ❑ NO SEIZURES
11. Do you see a relationship between learning/behavior/mood/energy/movement problems and ketones? ❑ YES ❑ NO ❑ NOT SURE ❑ NO PROBLEMS
12. Did you see a change in ketones at puberty? ❑ YES ❑ NO
    If female, do you see a change in ketones during menstrual cycle? ❑ YES ❑ NO
    If yes, how?____
13. Do you take ketogenic diet supplements?
    ❑ Multivitamin ❑ Calcium ❑ Polycitra / Cytra K / Potassium Citrate
    ❑ Vitamin D ❑ Carnitine ❑ Triheptanoin ❑ Other: __________
14. Have you had high cholesterol levels while on the diet? (*check all that apply*)
    ❑ CHILD ❑ ADOLESCENT/TEEN ❑ ADULT
15. Do you take medications to treat diet side effects? (*check all that apply*)
    ❑ Kidney stones ❑ High cholesterol ❑ Gastroesophageal reflux
    ❑ Constipation ❑ Other: __________
16. Do you currently have or have you ever had a feeding tube? ❑ YES ❑ NO
17. How effective is the diet in adulthood? ❑ LESS ❑ MORE ❑ SAME

Medications & Other Treatments:

1. Are you on antiseizure medications? ❑ YES ❑ NO
   If yes, which one(s)? ______________________________
2. Have you been on antiseizure medications in the past? ❑ YES ❑ NO
   If yes, which one(s)? ______________________________
3. If yes to #1 or #2, were they helpful? ❑ YES ❑ NO
4. Do you take medications for any other symptoms of G1DS? ❑ YES ❑ NO
   If yes, please list the medications and what they treat.
   ____
5. Do you take any supplements (*besides those used with a ketogenic diet*) that are helpful? If so, what supplement and how does it help?
   ____
6. Do you have a Vagus Nerve Stimulator? ❑ YES ❑ NO
   If yes, is it helpful?
   ____

Therapy:

1. Do you receive any of the following therapy services? (*check all that apply*)
   ❑ Speech ❑ Occupational ❑ Physical ❑ Behavioral ❑ Other: __________
2. Did you receive any of the following in the past but no longer? (*check all that apply*)
   ❑ Speech ❑ Occupational ❑ Physical ❑ Behavioral ❑ Other: __________
3. What physical activities do you find helpful? ______________________
4. List any assistive devices you use? _________________________

Social/Emotional Support:

1. Have you participated in secondary school options? ❑ YES ❑ NO
2. Have you participated in vocational rehabilitation training? ❑ YES ❑ NO
3. Did you have help with transitioning from school services to more independence?
   ❑ YES ❑ NO If yes, what kind of assistance? ______________________
4. What job opportunities have been available to you? (*check all that apply*)
   ❑ Volunteer work ❑ Part-time work ❑ Full-time work
   ❑ Day program ❑ Vocational program ❑ Other: _______________
5. How do you handle transportation? ____________________________
6. Are you involved in: (*check all that apply*)
   ❑ Activities with a lot of friends ❑ Activities with 1-2 friends
   ❑ Support group activities ❑ Other: ____________________
7. Do you live: (*check all that apply*) ❑ Alone ❑ With family ❑ With friends
   ❑ In a group home ❑ Attend a day program
8. Are you: (*check all that apply*) ❑ Single ❑ Dating ❑ Married ❑ Divorced ❑ Separated
9. Are you able to provide your own daily living skills? (*check all that apply*)
   ❑ Dressing ❑ Cooking ❑ Household chores ❑ Managing finances ❑ Driving
10. How often do you feel happy? (*check all that apply*)
    ❑ Always ❑ Almost always ❑ Frequently ❑ Sometimes ❑ Rarely ❑ Never
11. Do you receive any type of financial support? (*check all that apply*)
    ❑ SSI disability ❑ Special needs trust ❑ Waiver programs
    ❑ None ❑ Other: ____________________
12. Does someone have guardianship care over you? ❑ YES ❑ NO
13. Do you have children? ❑ YES ❑ NO Do they have G1DS? ❑ YES ❑ NO
14. Do you have other family members who have G1DS? ❑ YES ❑ NO