[CAREGIVER EDITION]

JOHNS HOPKINS
MEDICINE

Adult Glut1 Deficiency Syndrome Survey

Participation in this survey is voluntary. Please complete this survey if you are a caregiver of an adult (18 years of age or older) with Glut1 Deficiency Syndrome (G1DS). One survey per person with G1DS, please. "My child" refers to the adult with G1DS in the survey. 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. I am a/an:
   - ❑ Mother of an adult child with G1DS
   - ❑ Father of an adult child with G1DS
   - ❑ Other care provider (please write relationship): ____________________
2. My child's age: _______
3. My child's gender: ❑ Male ❑ Female
4. My child's age when Glut1 Deficiency Syndrome was diagnosed: _______
5. My child's 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. Does your child 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 they have unusual eye/head movements as an infant? ❑ YES ❑ NO

3. Have you noticed any triggers for their 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 child's 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 they been given any diagnosis other than Glut1 Deficiency? ❑ YES ❑ NO
   If yes, please list: ______________________________
   ____
   ____
   ____
   ____
   ____

Has your child transitioned to an adult neurologist? ❑ YES ❑ NO
If yes, was this a smooth transition? ❑ YES ❑ NO

DIET INFORMATION:

1. What diet are they receiving currently **OR** have they 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 they 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 your child switched diets, at what age? ___________
From what diet to what diet? ________________________________
5. Are they followed at a ketogenic diet center? ❑ YES ❑ NO
6. Do they have an adult dietitian/nutritionist? ❑ YES ❑ NO
7. Has their 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 their 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. Does your child take ketogenic diet supplements?
❑ Multivitamin ❑ Calcium ❑ Polycitra / Cytra K / Potassium Citrate
❑ Vitamin D ❑ Carnitine ❑ Triheptanoin ❑ Other: ___________
14. Have they had high cholesterol levels while on the diet? (*check all that apply*)
❑ CHILD ❑ ADOLESCENT/TEEN ❑ ADULT
15. Do they take medications to treat diet side effects? (*check all that apply*)
❑ Kidney stones ❑ High cholesterol ❑ Gastroesophageal reflux
❑ Constipation ❑ Other: ___________
16. Do they currently have or have they ever had a feeding tube? ❑ YES ❑ NO
17. How effective is the diet in adulthood? ❑ LESS ❑ MORE ❑ SAME

Medications & Other Treatments:

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

Therapy:

1. Does your child receive any of the following therapy services? (*check all that apply*)
   ❑ Speech ❑ Occupational ❑ Physical ❑ Behavioral ❑ Other: __________
2. Did they 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 your child uses? ______________________

Social/Emotional Support:

1. Has your child participated in secondary school options? ❑ YES ❑ NO
2. Have they participated in vocational rehabilitation training? ❑ YES ❑ NO
3. Did they 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 them? (*check all that apply*)
   ❑ Volunteer work ❑ Part-time work ❑ Full-time work
   ❑ Day program ❑ Vocational program ❑ Other: ______________
5. How do they handle transportation? ______________________________
6. Is your child involved in: (*check all that apply*)
   ❑ Activities with a lot of friends ❑ Activities with 1-2 friends
   ❑ Support group activities ❑ Other: ______________________
7. Do they live: (*check all that apply*) ❑ Alone ❑ With family ❑ With friends
   ❑ In a group home ❑ Attend a day program
8. Are they: (*check all that apply*) ❑ Single ❑ Dating ❑ Married ❑ Divorced ❑ Separated
9. Are they able to provide their own daily living skills? (*check all that apply*)
   ❑ Dressing ❑ Cooking ❑ Household chores ❑ Managing finances ❑ Driving
10. How often do they feel happy? (*check all that apply*)
    ❑ Always ❑ Almost always ❑ Frequently ❑ Sometimes ❑ Rarely ❑ Never
11. Do they 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 them? ❑ YES ❑ NO
13. Do they have children? ❑ YES ❑ NO Do they have G1DS? ❑ YES ❑ NO
14. Do they have other family members who have G1DS? ❑ YES ❑ NO