

Collective Voices in Gut1 Deficiency Project Survey – Report

The Glut1 Deficiency Collective Voices Project was designed to have a better understanding of the patient and family experience across a broad range of areas. The goals for the survey were to better define the range of symptoms, identify gaps in treatment and patient care, identify the gaps in knowledge and understanding of this disease, better understand disease burdens, identify the most important components of a future natural history study, develop and prioritize a patient-led strategic research plan, and to develop better and more effective clinical trials for potential future treatments.

This time we are reporting the section about **Adulthood**.

1. Are you an adult (over 18 years of age)?

Of the respondents that completed the survey, about 20% reported being over 18 years of age.

Seizures	Count	Percentage
yes	38	19.9%
no	153	80.1%

2. Did your symptoms change over time once you reached adulthood or during adulthood?

The majority of adults that responded to the survey felt that their symptoms had changed once they reached adulthood or during adulthood.

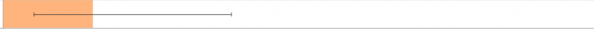
Seizures	Count	Percentage
yes	28	73.7%
no	9	23.7%
I'm not sure	1	2.6%

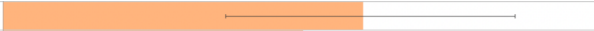
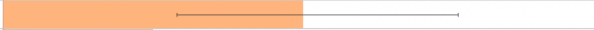
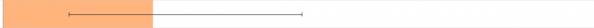
3. How have your symptoms changed in adulthood?

The majority of respondents felt that seizures improved with adulthood.

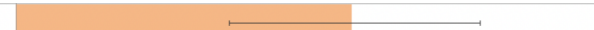



Seizures	%	Count
improved	60.7%	17
did not change	17.9%	5
worsened	10.7%	3
I do not have this symptom	10.7%	3





On the other hand, movement episodes and stamina/energy worsened.

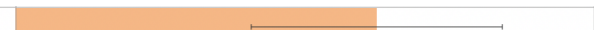

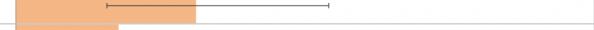

Movement Episodes		%	Count
worsened		53.6%	15
improved		35.7%	10
did not change		10.7%	3

Stamina/ Energy		%	Count
worsened		42.9%	12
did not change		35.7%	10
improved		17.9%	5

Cognitive or intellectual difficulties, speech/communication issues, and memory did not change for most.




Intellectual difficulties		%	Count
did not change		46.4%	13
improved		32.1%	9
worsened		17.9%	5
I do not have this symptom		3.6%	1

Speech/Communication		%	Count
did not change		39.3%	11
improved		32.1%	9
worsened		21.4%	6
I do not have this symptom		7.1%	2

Memory		%	Count
did not change		50.0%	14
worsened		25.0%	7
improved		14.3%	4
I do not have this symptom		10.7%	3

4. How would you compare your overall quality of life in adulthood in comparison to childhood?

Most felt that their quality of life had improved in adulthood.

		Count	Percentage
improved in adulthood		17	44.7%
stayed the same in adulthood		13	34.2%
worsened in adulthood		8	21.1%

5. Are you following a ketogenic diet now as an adult?

Of the adult respondents, roughly half of them report being on a ketogenic diet now.

	Count	Percentage
yes	21	55.3%
no	17	44.7%

6. Are you able to manage a ketogenic diet on your own without help?

Most do not feel that they are able to manage a ketogenic diet on their own without help.

	Count	Percentage
yes	5	19.2%
no	21	80.8%

7. Are you able to manage and take medications on your own without help (keep up with refills, measure or count out dose, and take the medication?)

Similarly, most feel unable to manage and take medication on their own without help, although there is a larger percentage who feel they *are* able to manage medications than those who feel they are able to manage a ketogenic diet.

	Count	Percentage
yes	17	44.7%
no	21	55.3%

8. Do you have children?

Only a small percentage of the adults surveyed have children.

	Count	Percentage
yes	4	10.5%
no	34	89.5%



9. Do any of your children also have Glut1 Deficiency?

Of the 4 adults with children, 3 have children with Glut1 Deficiency.

	Count	Percentage
yes	3	75.0%
no	1	25.0%



10. How do your children's Glut1 Deficiency symptoms compare to yours?

2 of the 3 adults whose children also have Glut1 reported their children's symptoms as comparable to their own. 1 reported their child's symptoms as more severe.

		Count	Percentage
about the same		2	66.7%
more severe		1	33.3%


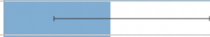

11. Are you able to care for your child or children independently without help?

Most reported being able to care for their child on their own, without help.

		Count	Percentage
yes		3	75.0%
no		1	25.0%

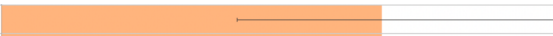

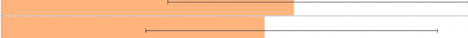
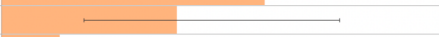


12. Did you complete high school?

Almost 70% of respondents had completed high school.

		Count	Percentage
yes		26	68.4%
no		6	15.8%
I'm still in high school		6	15.8%


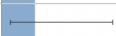
13. After you completed your regular schooling years, have you done any of the following?

After having completed regular schooling years, many have taken part in other activities such as attending a day program, volunteering, and even going to college.

		Count	Percentage
attend a day program		13	34.2%
volunteer		13	34.2%
go to college		10	26.3%
work part time		9	23.7%
I have not yet completed my regular schooling ...		6	15.8%
work full time		2	5.3%

14. What is your relationship status?

Most of the respondents are single, although ~10% report being in a relationship or have a spouse/partner.

		Count	Percentage
single		34	89.5%
dating		2	5.3%
have a spouse/partner		2	5.3%

15. Does your spouse/partner need to help provide caregiving for you?

Of the respondents with a spouse/partner, neither needed help providing caregiving.

	Count	Percentage
no	2	100.0%

A horizontal bar chart with a blue bar extending to the right, representing 100% of the data for the 'no' category. The bar is positioned above the 'no' label in the table below.