

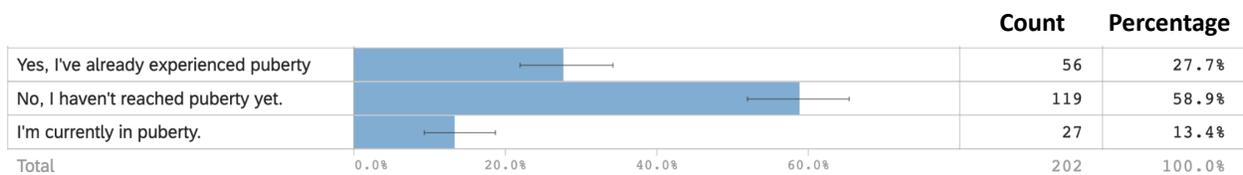
Collective Voices in Gut1 Deficiency Project Survey – Report

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 **Puberty**.

Puberty:

1. Have you experienced puberty yet?



Close to 30% of respondents reported having experienced puberty, while ~13% reported currently experiencing it and ~59% reported they have not reached puberty yet.

Note: the data showed from this point on was calculated using an n=260 (Total number of responses)

2. Did your symptoms change in puberty?



The majority (~56%) of patients surveyed reported experiencing changes in their symptoms when they reached puberty, while 17% report no changes in their symptoms and 26% report not being sure about experiencing changes.

3. How did your symptoms change in puberty?

Seizures:

		Percentage	Count
worsened		7.7%	20
I do not have this sympt...		3.1%	8
improved		2.7%	7
did not change		2.7%	7

The majority of the respondents (~8%) reported their seizures worsened, and the remaining of respondents, about equally, reported that their seizures either improved, did not change or they don't experience them.

Movement episodes

		Percentage	Count
worsened		11.5%	30
improved		2.3%	6
did not change		2.3%	6
I do not have this symptom		0.0%	0

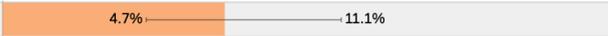
About 11% of the respondents report their movement episodes worsened during puberty, while about 2% report improvement or no change of this symptom.

Cognitive or intellectual difficulties

		Percentage	Count
did not change		10.4%	27
worsened		3.5%	9
improved		2.3%	6
I do not have this symptom		0.4%	1

The majority of respondents reported that their cognitive or intellectual difficulties did not change during puberty. ~3% reported this symptom worsened and ~2% reported improvement.

Anxiety

		Percentage	Count
worsened		7.3%	19
did not change		3.8%	10
I do not have this symptom		3.5%	9
improved		1.5%	4

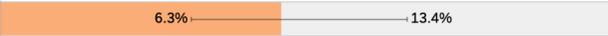
Most of the surveyors (~7%) reported that their anxiety worsened during puberty. At a similar percentage (3.5-3.8%), surveyors reported either not experiencing anxiety or not experiencing changes in this symptom.

Energy levels/Stamina

		Percentage	Count
worsened		9.6%	25
did not change		4.6%	12
improved		1.9%	5
I do not have this symptom		0.4%	1

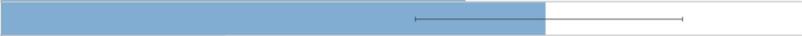
About 10% of respondents reported their levels of energy/stamina worsened during puberty, while in about 5% of them they reported not having changes.

Speech communication issues

		Percentage	Count
did not change		9.2%	24
worsened		5.4%	14
improved		1.5%	4
I do not have this symptom		0.0%	0

Most of the respondents (~9%) reported not experiencing changes in their speech and communication issues, while about 5% reported worsening of this symptom.

4. Did any new symptoms begin for the first time in puberty that did not exist before?

		Count	Percentage
yes		29	37.7%
no		34	44.2%
I'm not sure		14	18.2%

Most of the surveyors (44%) reported no new symptoms began in puberty, while ~38% reported new symptoms began during this stage and 18% reported not being sure.

5. What new symptoms started in puberty that did not exist before?

This question was a multiple choice question and surveyors could choose all the options that applied.

		Percentage	Count
new types of movement episodes I ne...		4.2%	11
movement episodes for the first time		3.8%	10
anxiety		3.1%	8
new types of seizures I never had before		2.7%	7
energy/stamina issues		1.9%	5
other		1.9%	5
cognitive issues		1.5%	4
speech/language issues		1.5%	4
seizures for the first time		0.0%	0

Most of the patients experiencing new symptoms in puberty report having new movement episodes (4%), similarly, others report experiencing movement episodes for the first time during puberty.

At a slightly lower percentage, patients report anxiety as a new symptom in puberty, while others report issues with energy and stamina.

6. Did treatment need to change during puberty?

		Count	Percentage
Yes		35	45.5%
No		42	54.5%

54% of the respondents report not changing their treatment during puberty, while 45 report changing their treatment.

7. How did treatment need to change during puberty?

This question was a multiple choice question and surveyors could choose all the options that applied.

		Percentage	Count
diet changes		8.5%	22
new medications		7.3%	19
other (please specify)		1.5%	4
new therapies		1.2%	3

Most of the changes in treatment were related to the diet, followed by the use of new medications. Other respondents reported trying therapies and making other changes in treatment but those changes were not specified.

It would be important to follow up and find out which were the changes made with regards to the diet, as well as which new medications or which new therapies were introduced and whether or not these changes were effective.