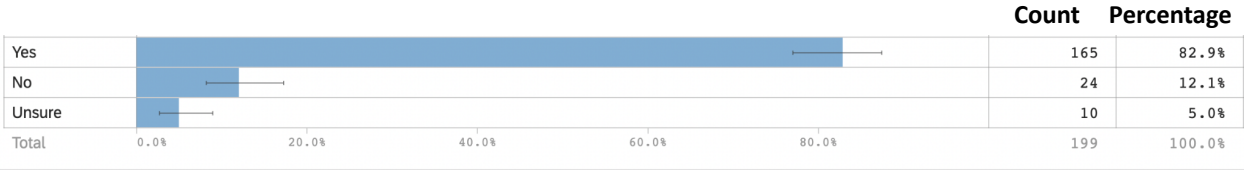
Collective Voices in Gut1 Deficiency Project Survey – Report

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 **seizures**.

Seizures:

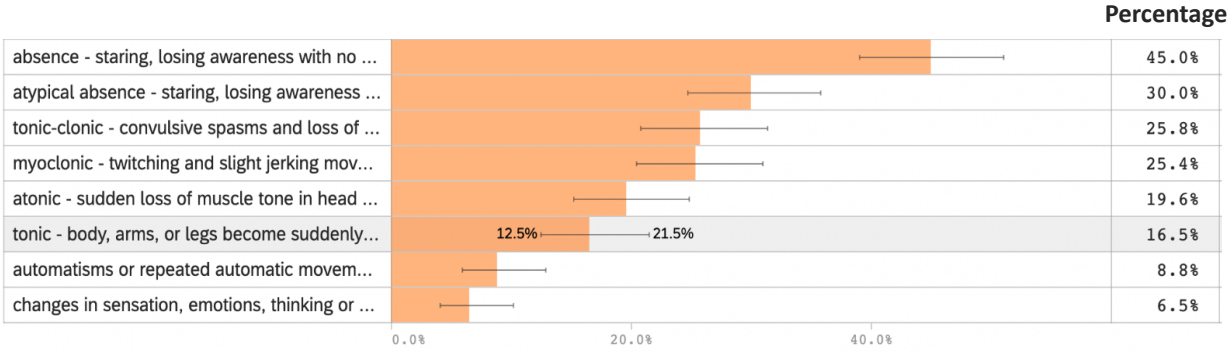
1. **Have you ever had any type of seizures?**



The majority (~83%) of patients surveyed reported experiencing seizures and 12% of surveyed patients report not experiencing seizures. Interestingly, 5% of patients surveyed report not being sure about experiencing seizures.

2. **Which of the following have been the most similar to your type or types of seizure?**

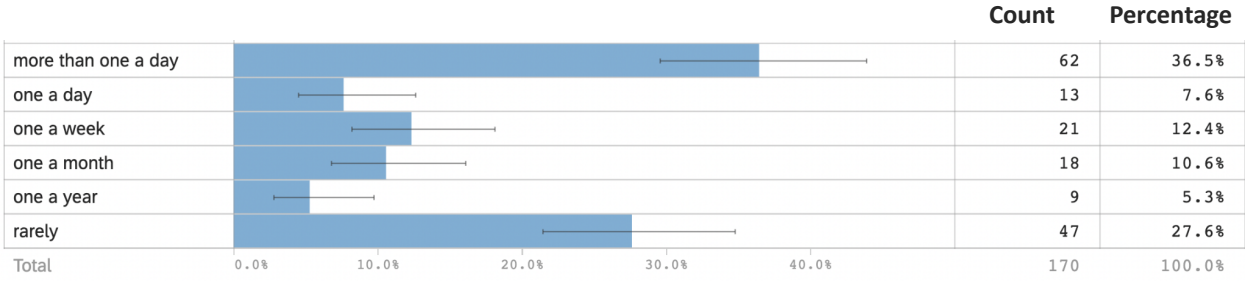
This question was a multiple choice question and surveyors could choose all the options that applied.



Most of the surveyors (45%) report absence seizures as the most common type of seizure experienced. Other types of seizures reported with less frequency include atypical absence (30%), tonic clonic (26%), myoclonic (25%), atonic (20%) and tonic (16%). In summary, most of

the patients report experiencing seizures that belong to the generalized seizures type described by the CDC.

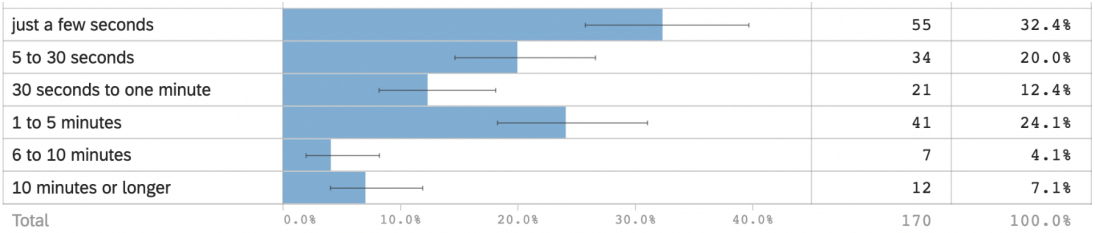
3. **What has been the typical frequency pattern of your seizures?**



	Count	Percentage
more than one a day	62	36.5%
one a day	13	7.6%
one a week	21	12.4%
one a month	18	10.6%
one a year	9	5.3%
rarely	47	27.6%
Total	170	100.0%

Most of the patients surveyed (36%) report experiencing more than one seizure a day. At lower percentages, survey participants report having seizures at higher frequencies. Interestingly, about 28% of respondents report rarely having seizures.

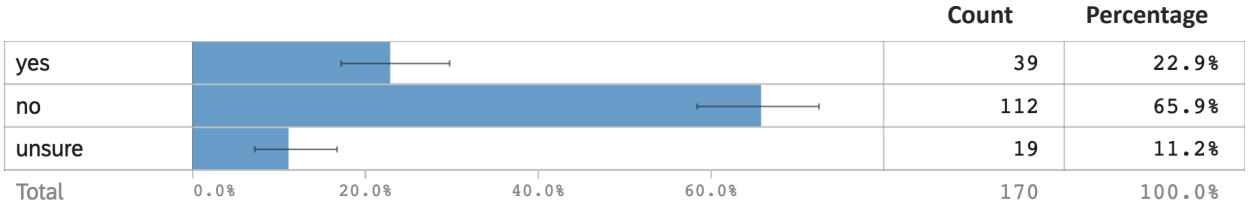
4. **How long have your seizures typically lasted?**



	Count	Percentage
just a few seconds	55	32.4%
5 to 30 seconds	34	20.0%
30 seconds to one minute	21	12.4%
1 to 5 minutes	41	24.1%
6 to 10 minutes	7	4.1%
10 minutes or longer	12	7.1%
Total	170	100.0%

Seizures experienced in 32 % of surveyed patients last just a few seconds. About 24% of patients experiencing seizures have seizures that last between 1 and 5 minutes and 7 % of patients experience seizures lasting 10 minutes or longer.

5. **Do you still have seizures currently?**

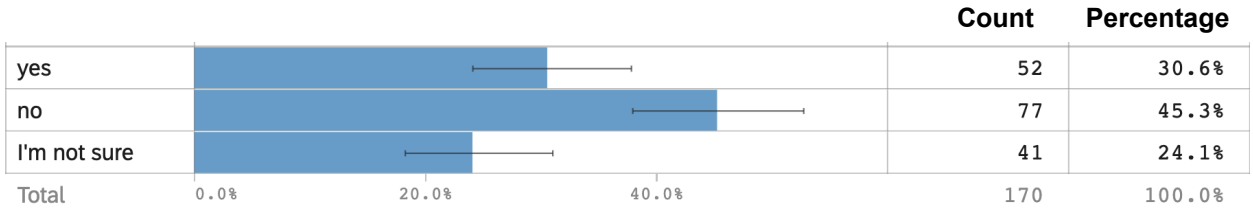


	Count	Percentage
yes	39	22.9%
no	112	65.9%
unsure	19	11.2%
Total	170	100.0%

A significant percentage of the population (66%) do not currently experience any seizures, while 23% of surveyed patients are still experiencing them. Interestingly 11% of surveyed patients are

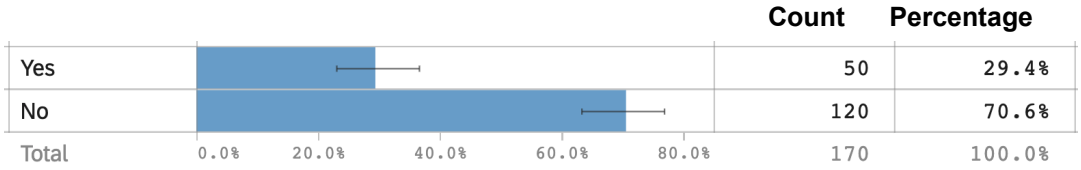
unsure of whether they are experiencing seizures or not. This information suggests that more education is necessary around which signs to look for when patients are experiencing seizures.

6. **Have you experienced seizure clusters (star and stop, but occur in groups one after another?)**



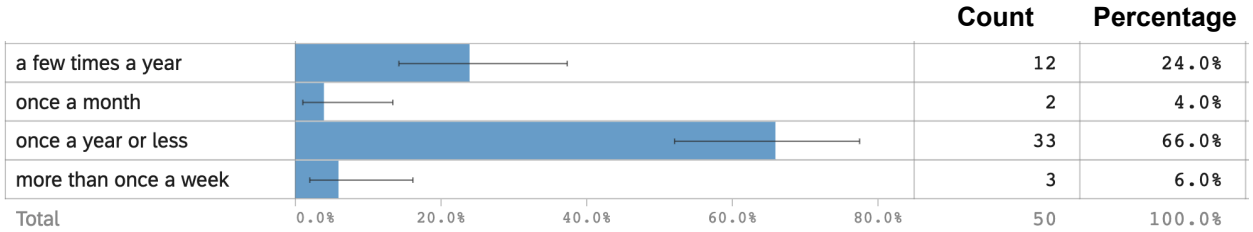
45% of patients who have experienced seizures report not having seizure clusters, while 31% report experiencing this clusters. 24 % of surveyed patients are not sure of their occurrence. Once more, this result shows the importance of educating patients about seizure types and signs to recognize them.

7. **Have you ever had to use rescue medications to try to stop prolonged seizures?**



The majority of our respondents (71%) report not having to use rescue medication to stop prolonged seizures. It is important to highlight that most of the surveyed patients do not experience prolonged seizures and therefore, the need for this type of medication is not as prevalent.

8. **How frequently have you had to use rescue medications for seizures?**



The majority (66%) of the patients who use rescue medications for prolonged seizures, use these medication once a year or less. While 24% of patients using this type of medication use it only a few times a year.

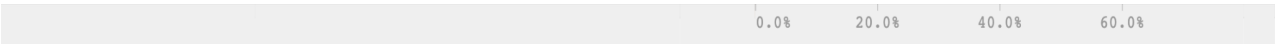
Is there a relationship between the types of seizures experienced and the length of seizures?

Length of seizure		Type of seizure		Percentage
just a few seconds	(55)	absence - staring, losing awarenes...	20 of 55	36.4%
5 to 30 seconds	(34)	absence - staring, losing awarenes...	19 of 34	55.9%
30 seconds to one mi...	(21)	absence - staring, losing awarenes...	12 of 21	57.1%
1 to 5 minutes	(41)	absence - staring, losing awarenes...	21 of 41	51.2%
6 to 10 minutes	(7)	absence - staring, losing awareness ...	3 of 7	42.9%
10 minutes or longer	(12)	absence - staring, losing awareness...	2 of 12	16.7%

Type of seizure: Absence - Staring, losing awareness with no movement.

Absence seizures in the majority of patients experiencing this type of seizure, last between 5 seconds to 10 minutes.

Length of seizure		Type of seizure		Percentage
just a few seconds	(55)	atonic - sudden loss of muscle tone...	8 of 55	14.5%
5 to 30 seconds	(34)	atonic - sudden loss of muscle tone...	2 of 34	5.9%
30 seconds to one mi...	(21)	atonic - sudden loss of muscle tone...	2 of 21	9.5%
1 to 5 minutes	(41)	atonic - sudden loss of muscle tone...	7 of 41	17.1%
6 to 10 minutes	(7)	atonic - sudden loss of muscle tone i...	0 of 7	0.0%
10 minutes or longer	(12)	atonic - sudden loss of muscle tone...	2 of 12	16.7%



Type of seizure: Atonic - sudden loss of muscle tone in head or body (also known as drop).

We analyzed the the relationship between type of seizure and the length of the seizure.
In the group of patients experiencing atonic seizures we don't see a clear pattern of length of the seizures. The length of time seems to vary from just a few seconds to 10 minutes or longer.

Length of seizure		Type of seizure		Percentage
just a few seconds	(55)	atypical absence - staring, losing a...	14 of 55	25.5%
5 to 30 seconds	(34)	atypical absence - staring, losing a...	10 of 34	29.4%
30 seconds to one mi...	(21)	atypical absence - staring, losing a...	3 of 21	14.3%
1 to 5 minutes	(41)	atypical absence - staring, losing a...	10 of 41	24.4%
6 to 10 minutes	(7)	atypical absence - staring, losing aw...	4 of 7	57.1%
10 minutes or longer	(12)	atypical absence - staring, losing a...	5 of 12	41.7%

Type of seizure: Atypical - Staring or losing awareness with twitches or slight movement in parts of the body (eyes, limbs).

Atypical seizures in most of the patients experiencing this type of seizure can last more than 6 minutes.

Length of seizure		Type of seizure		Percentage
just a few seconds	(55)	myoclonic - twitching and slight jer...	19 of 55	34.5%
5 to 30 seconds	(34)	myoclonic - twitching and slight jerk...	5 of 34	14.7%
30 seconds to one mi...	(21)	myoclonic - twitching and slight jerk...	3 of 21	14.3%
1 to 5 minutes	(41)	myoclonic - twitching and slight jerk...	6 of 41	14.6%
6 to 10 minutes	(7)	myoclonic - twitching and slight jerki...	0 of 7	0.0%
10 minutes or longer	(12)	myoclonic - twitching and slight jerk...	3 of 12	25.0%

Type of seizure: Myoclonic - Twitching and slight jerking movements but no loss of awareness.

Most of the patient experiencing myoclonic seizures report either experiencing them for just a few seconds or for 10 minutes or longer.

just a few seconds	(55)	tonic-clonic - convulsive spasms an...	1 of 55	1.8%
5 to 30 seconds	(34)	tonic-clonic - convulsive spasms an...	3 of 34	8.8%
30 seconds to one mi...	(21)	tonic-clonic - convulsive spasms an...	4 of 21	19.0%
1 to 5 minutes	(41)	tonic-clonic - convulsive spasms a...	12 of 41	29.3%
6 to 10 minutes	(7)	tonic-clonic - convulsive spasms and ...	2 of 7	28.6%
10 minutes or longer	(12)	tonic-clonic - convulsive spasms an...	4 of 12	33.3%

Type of seizure: tonic-clonic - Convulsive spasms and loss of awareness.

The majority of surveyed patients experiencing this type of seizure report having this episodes for 1 minute or longer.

Overall, the results show that there is not a clear pattern of length of time compared to the type of seizure experienced by patients, at least for seizures displayed in this report.

9. **<u>Which medications have been the most effective to rescue seizures?</u>**

Surveyed patients experiencing prolonged seizures reported different types of medications as being effective to control them. Four out of 46 patients report Diastat and Midazolam as the most effective medication to control prolonged seizures. Among some of the other medications reported are: Diazepam, Lorazepam, Prednisone and Stesolid.