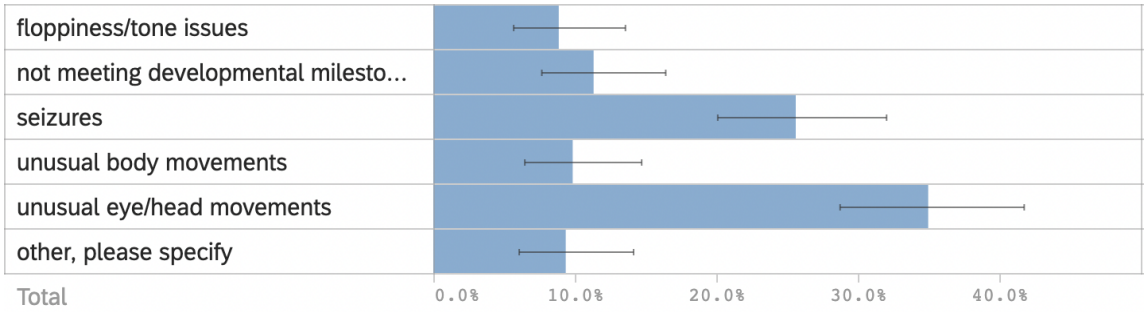
Collective Voices in Gut1 Deficiency Project Survey – Report

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 sections of **symptoms and movement episodes**.

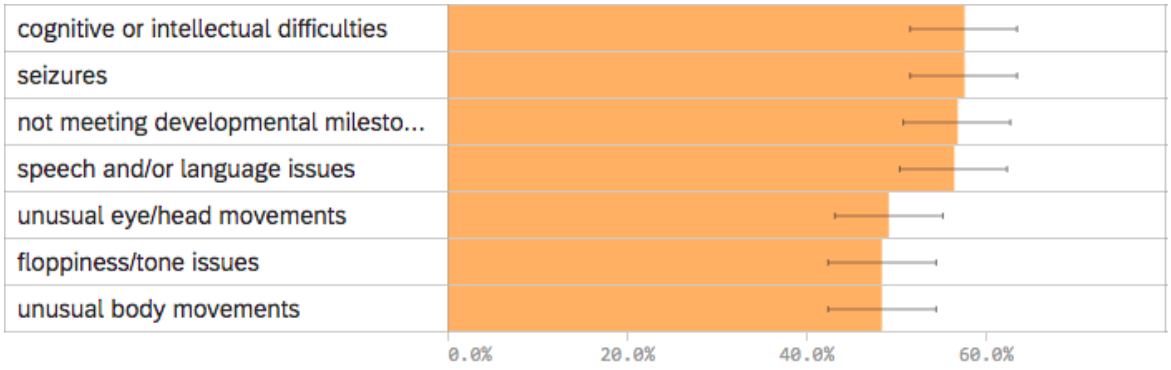
Symptoms:

1. **What was the first symptom noticed that raised concern?**



The first symptoms that raised concern were most commonly reported as unusual eye/head movements followed by seizures.
Less commonly reported were floppiness/tone issues, not meeting developmental milestones, and unusual body movements.

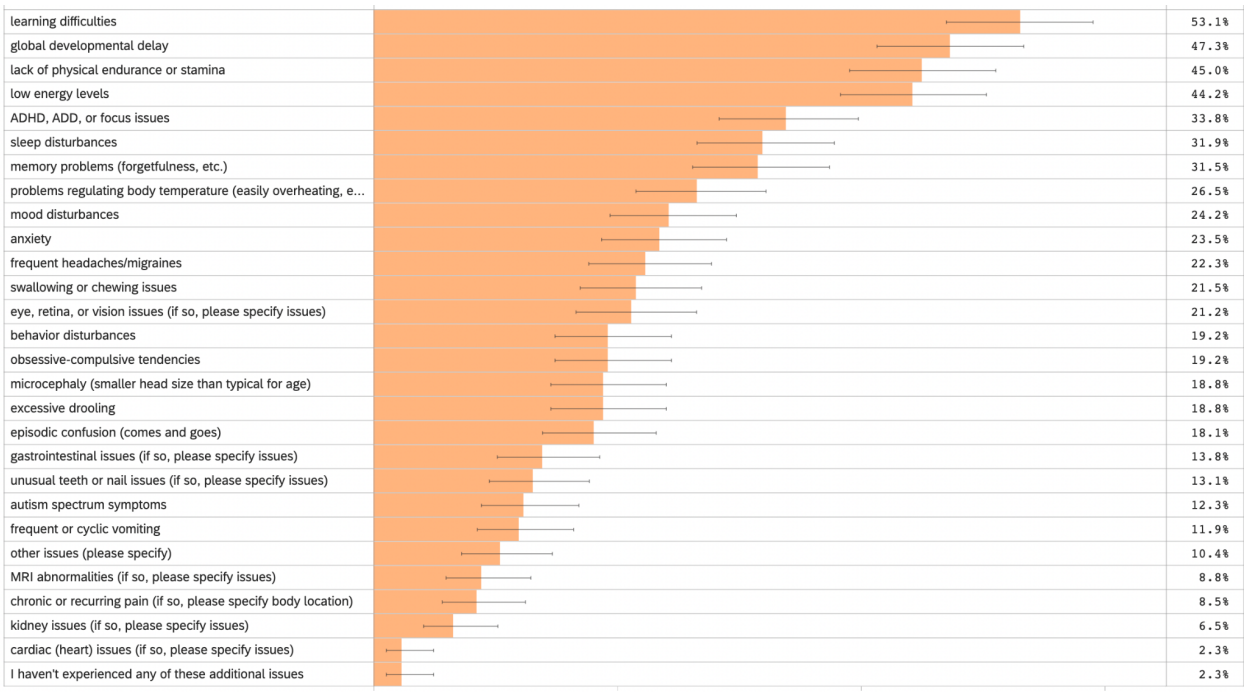
2. **Which of these more common Glut1 Deficiency symptoms have you experienced?**



An average of 57% of patients experienced cognitive or intellectual difficulties, seizures, not meeting developmental milestones, and speech and/or language issues. Slightly less commonly reported at an average of 50% was unusual eye/head movements, floppiness/tone issues, unusual body movements.

Interestingly, although unusual eye/head movements were less commonly reported than other symptoms, those that did experience it, reported it as being one of the first symptoms to raise concern.

3. **<u>What other symptoms have you experienced?</u>**

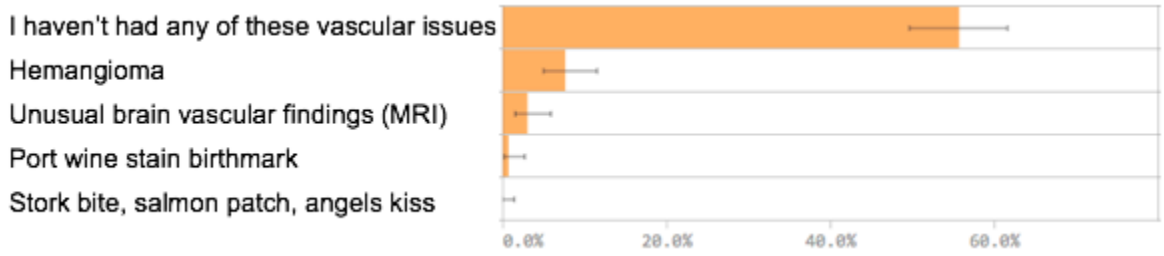


Other symptoms commonly experienced by Glut1 patients include learning difficulties, global developmental delay, lack of physical endurance or stamina, and low energy levels.

Correlation between number of symptoms and age of symptom onset:

Younger age of symptom onset is correlated with a higher number of symptoms. Respondents who showed symptoms before a year of age were more likely to report speech/language issues. Those who showed symptoms before 6 months were more likely to report unusual eye or head movements.
These results support eye/head movements as a characteristic symptom of Glut1 among infants. This is important to know as it is oftentimes the first step in achieving a diagnosis.

4. **<u>Have you had any of these vascular issues?</u>**

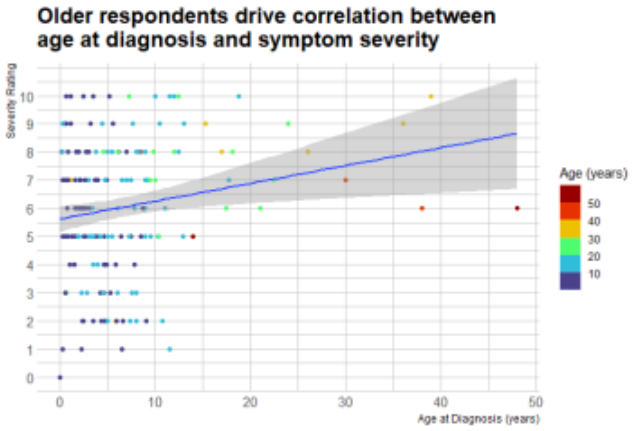


The majority of Glut1 patients did not report having experienced vascular issues (55.8%), However, of the ones that did, hemangioma (sometimes called a strawberry mark) was the most common.

5. **<u>How would you describe your severity of Glut1 Deficiency on a scale of 0 to 10?</u>**

When describing the severity of Glut1 Deficiency, respondents averaged a 6 on a scale from 0 - 10. There are many factors that can impact this response including age at diagnosis and length of time from first symptom to diagnosis:

Age at diagnosis:
Our data shows that the older the age of diagnosis, the higher severity was ranked. This is driven, however, by older participants who tended to be both diagnosed when older and report more severe symptoms:

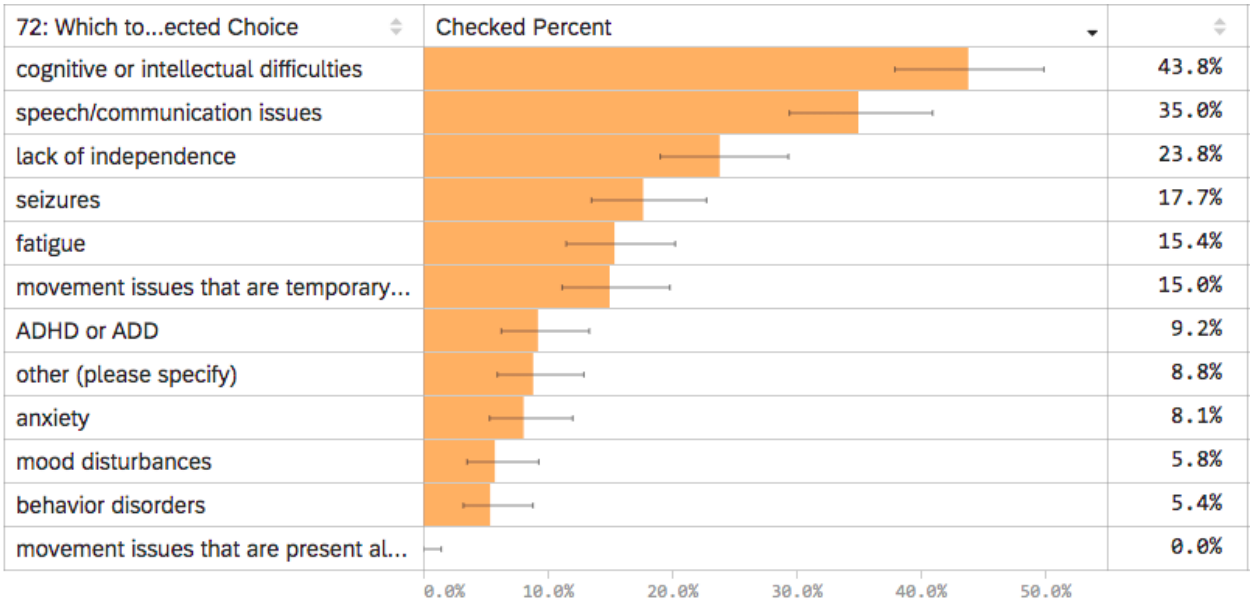


When we look at respondents that are 10 and younger only, the correlation between age of diagnosis and severity is no longer present. One possibility for this is that patients with less

severe symptoms are getting diagnosed in the younger cohorts and in the past were either not diagnosed or misdiagnosed for older groups.

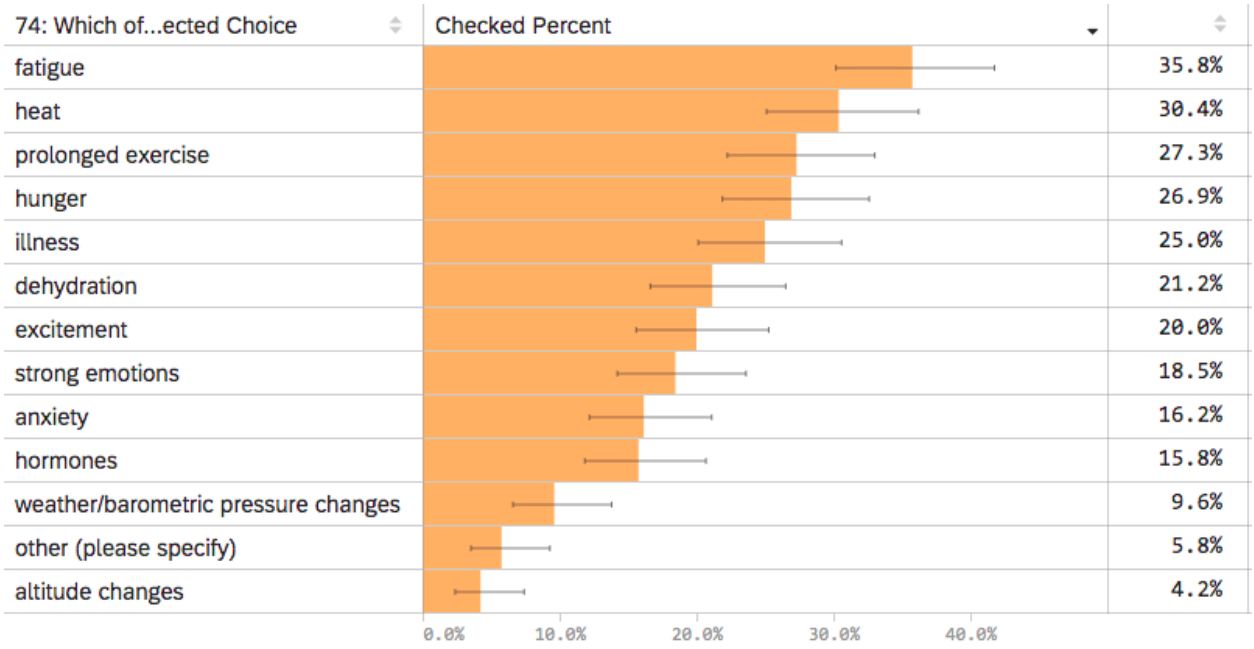
A similar relationship is seen when we look at the length of time from first symptom to diagnosis and severity. It seems likely that as awareness of Glut1 Deficiency has increased and diagnosis/testing methods have improved, patients are diagnosed more quickly and less severe cases are detected more frequently.

6. **<u>Which top 3 symptoms do you feel most negatively impact your quality of life?</u>**



Our data shows that cognitive or intellectual difficulties, speech/communication issues, and lack of independence negatively impact the Glut1 community the most- even more so than seizures, and movement episodes. These results highlight the importance of services and therapies to support development.

7. **Do you feel there are triggers that bring on, worsen, or aggravate your symptoms?**



60% of respondents feel that there are triggers for their symptoms, with fatigue being the most common. Heat, prolonged exercise, hunger, and illness were also commonly reported.

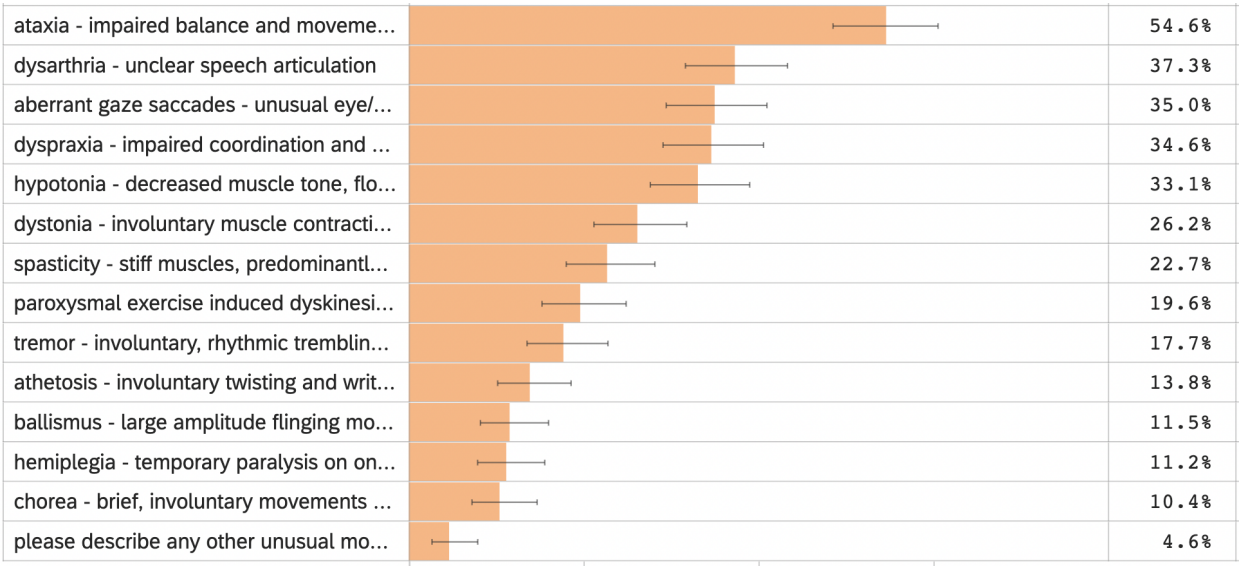
Movement episodes

8. **Have you experienced muscle issues or movement episodes?**

Most of the patients surveyed (about 77%), report having experienced movement episodes.

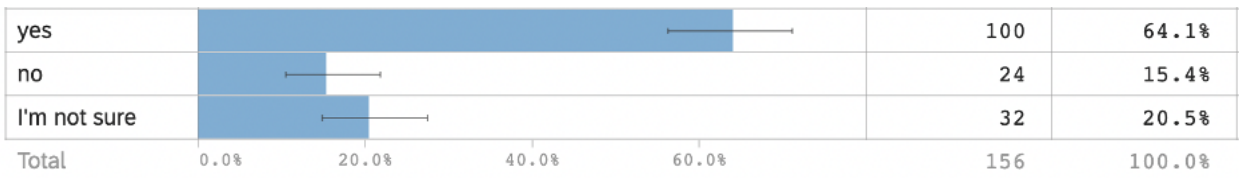
9. Movement episodes or issues in Glut1 Deficiency are often complex and unique. Have you experienced any that are similar to the following?

This was a multiple-choice question and surveyors could check all the answers that applied.



The data shows that ataxia is the most common type of movement episode for our patients. Other movement or muscle related issues that are also common for our patients are dysarthria, aberrant gaze saccades and dyspraxia. Some patients reported that another unusual movement they have experienced is oromandibular dystonia. Oromandibular dystonia, is characterized by forceful muscle contractions of the face, jaw, and/or tongue. The muscle contractions cause involuntary movements that interfere with opening and closing the mouth and may affect chewing and speech (Definition taken from the Dystonia MedicalResearch Foundation's website).

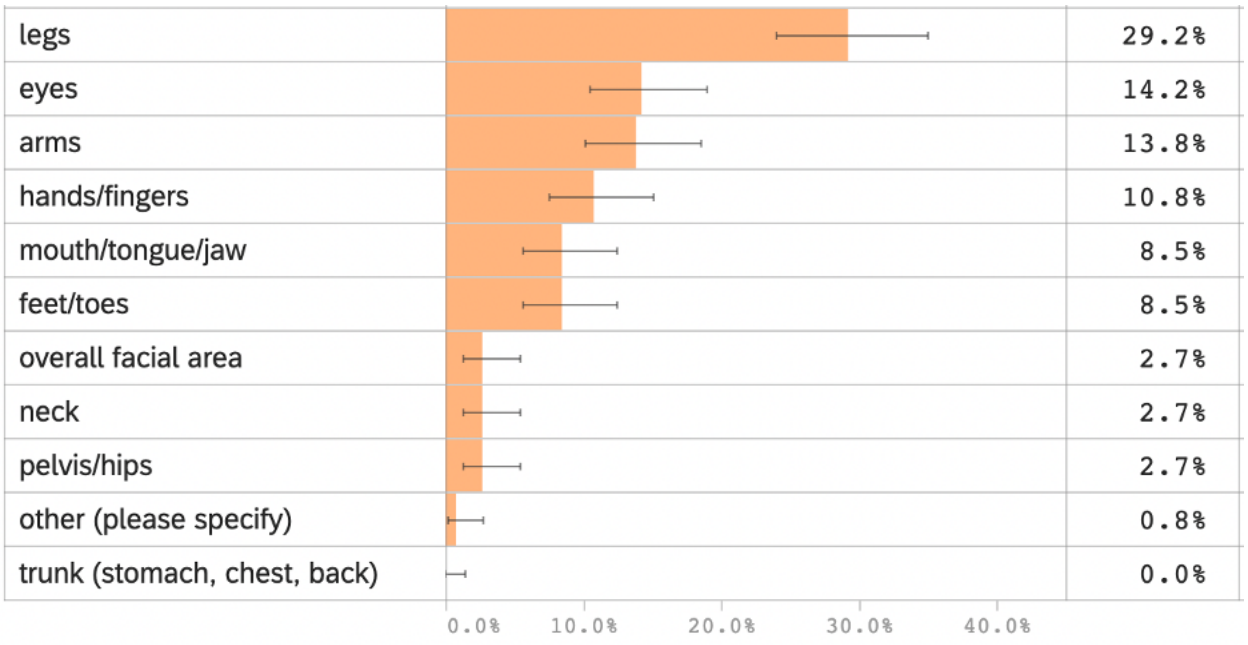
10. Have movement episodes occurred more frequently in certain body parts?



64% of the patients surveyed report that movement episodes occur more frequently in certain body parts.

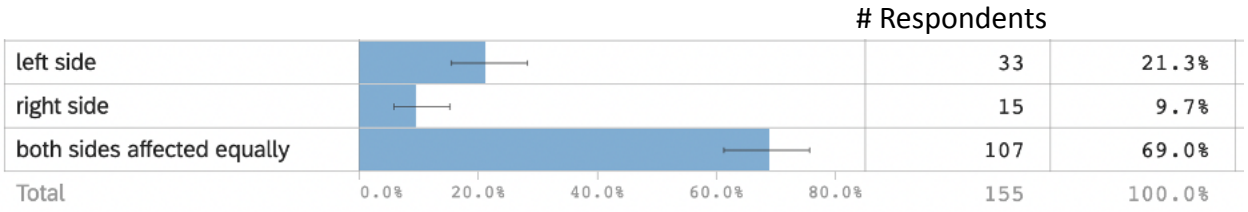
11. **Which body parts have been most frequently affected during movement episodes?**

This was a multiple-choice question and surveyors could check all the answers that applied.



Our data shows that the body parts that are most commonly affected by movement episodes are the legs. Other body parts affected but in a lower percentage include eyes and arms.

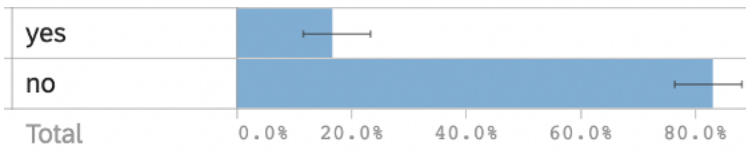
12. **Which side of the body is most affected by unusual movement episodes?**



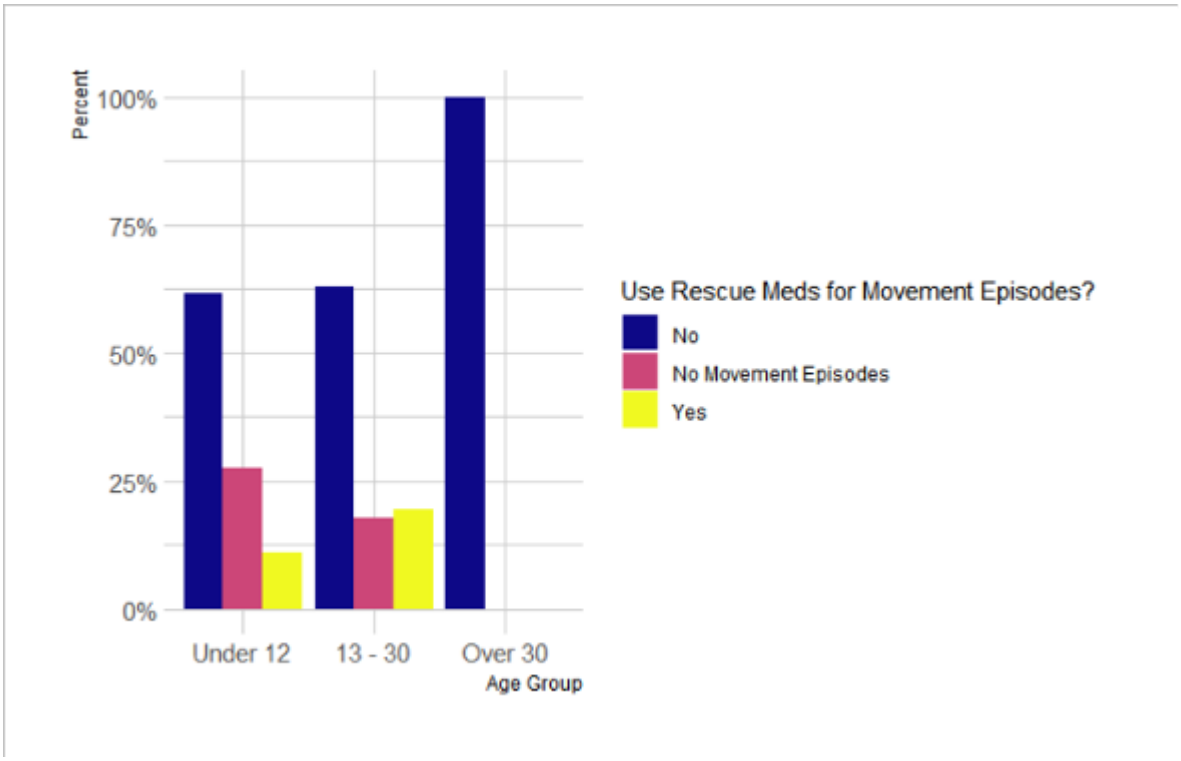
69% of surveyed patients report both sides of the body to be equally affected.

13. **Have you ever had to use rescue medications to try to stop prolonged movement episodes?**

	# Respondents	
yes	26	16.8%
no	129	83.2%
Total	155	100.0%



The great majority of surveyed patients (83%) report not having to use rescue medications for prolonged movement episodes.



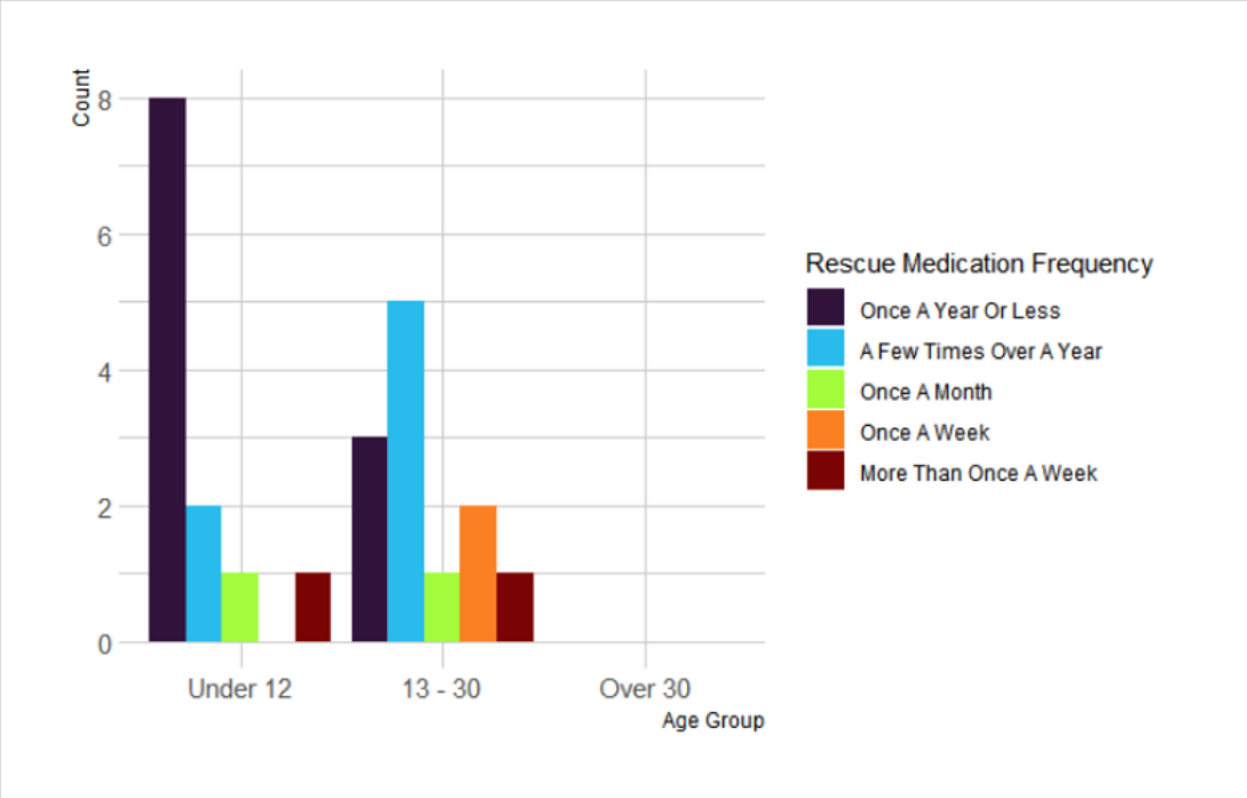
The plot above shows the use of rescue medications by age group, as well as the absence of movement episodes. According to this graph, no respondent over the age of 30 reports using rescue medication for movement episodes and only below 19% of respondents in the age group between 13-30 report using these medications. Less than 12% of respondents 12 and under report the use of these medications.

14. <u>How frequently have you had to use rescue medications for movement episodes?</u>

	# Respondents	
once a year or less	12	48.0%
a few times over a year	7	28.0%
once a month	2	8.0%
once a week	2	8.0%
more than once a week	2	8.0%
Total	25	100.0%

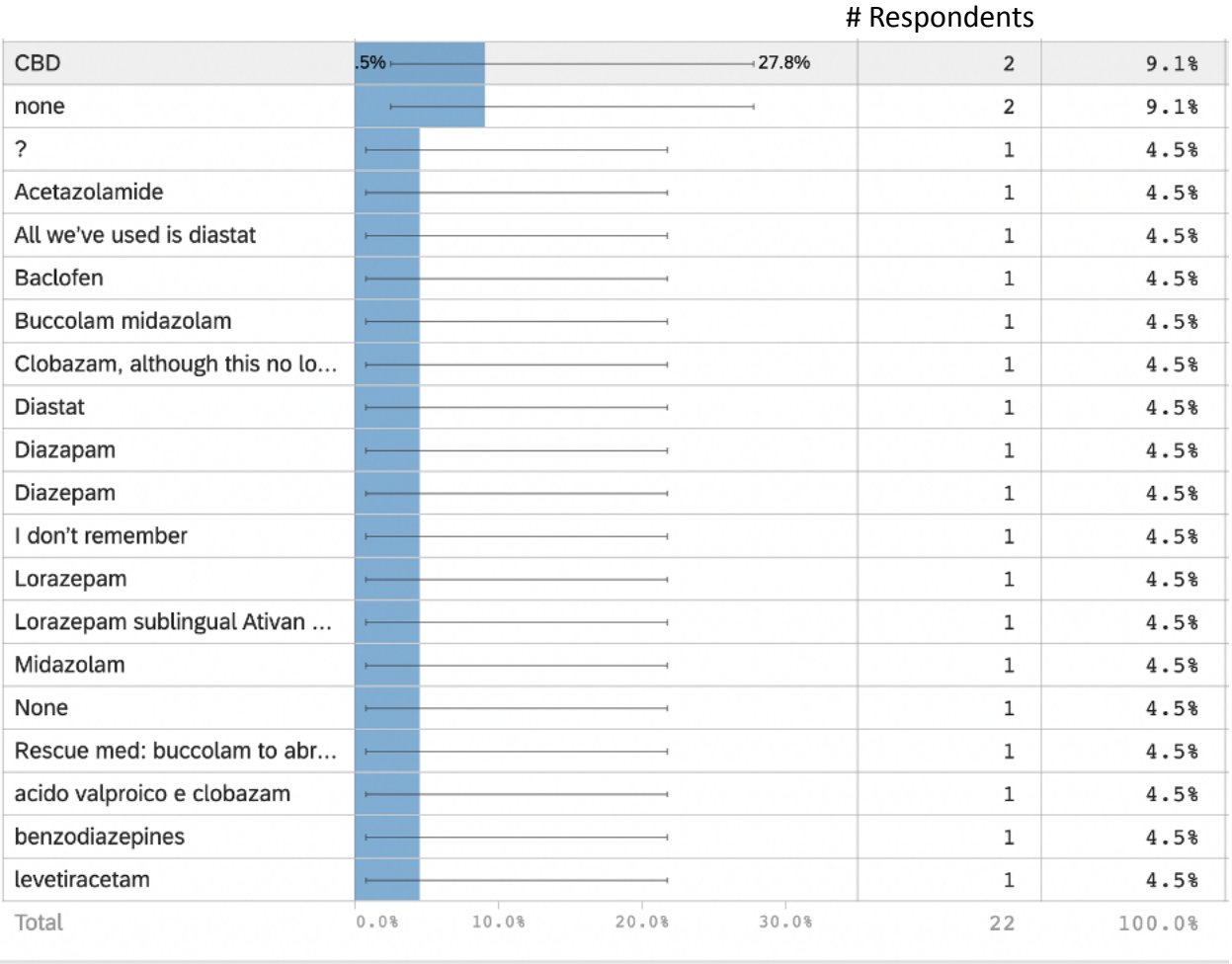
0.0% 20.0% 40.0% 60.0%

The majority of patients using rescue medications (48%) report having to use rescue medications for movement episodes once a year or less. 28% of respondents report using rescue medications a few times a year; and 8% report using this type of medications once a month, once a week or more than once a week.



The plot above depicts the responses regarding the use of rescue medications separated by age group. The plot suggests that the middle age group may also use rescue medications more frequently, but the sample sizes are quite small. 8/12 respondents under age 12 report using rescue medication once a year or less, compared to 3/12 respondents ages 13-30.

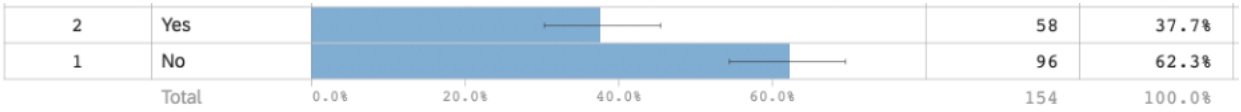
15. <u>Which medications have been most effective to rescue movement episodes?</u>



	# Respondents	
CBD	2	9.1%
none	2	9.1%
?	1	4.5%
Acetazolamide	1	4.5%
All we've used is diastat	1	4.5%
Baclofen	1	4.5%
Buccolam midazolam	1	4.5%
Clobazam, although this no lo...	1	4.5%
Diastat	1	4.5%
Diazapam	1	4.5%
Diazepam	1	4.5%
I don't remember	1	4.5%
Lorazepam	1	4.5%
Lorazepam sublingual Ativan ...	1	4.5%
Midazolam	1	4.5%
None	1	4.5%
Rescue med: buccolam to abr...	1	4.5%
acido valproico e clobazam	1	4.5%
benzodiazepines	1	4.5%
levetiracetam	1	4.5%
Total	22	100.0%

Our report shows that there is not a definite answer to this question. Surveyed patients report having used different medications to rescue movement episodes.

16. <u>Do you sometimes have difficulty distinguishing the difference between a seizure and a movement episode?</u>



		# Respondents	
2	Yes	58	37.7%
1	No	96	62.3%
	Total	154	100.0%

The majority of surveyed patients (62%) report not having difficulties differentiating between seizures and movement episodes.

Future directions:

Our next survey report will focus on the section regarding **seizures**.
If you have any questions regarding the information reported here, please do not hesitate to contact us:

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