Presentation: Family perspectives
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The Glut1 Deficiency Foundation launched a new survey to continue to learn and to have a better understanding of the patients and families perspectives about clinical trial readiness. The goal is to help capture the needs and priorities of patients and families so that we can help ensure research is patient-centered and focused on things that matter most to our community.

The results presented during this talk, are a combination of results collected in our collective voices survey which was launched in 2020, as well as answers collected at our regional research ready series meetings. This survey is an ongoing project and we want to hear from all the people in our community. If you are a patient or family member interested in participating, below you can find a QR code available that you can scan and it will take you to the survey. The survey is available in English and Spanish.

At the time of the presentation, we had 44 participants; most of the patients represented were female (57%) and most of the participants were within the 7-12 years old age range (34%).

The top three research priorities for participants, are to have new and better treatments (64%), followed by having a better understanding of the diet (40%), its side effects, long term effects, and micronutrients, among others. Participants also expressed that basic research to better understand the disease and its mechanisms (35%) is one of their top 3 priorities.

Participants expressed that the to three most beneficial outcomes for a new treatment would be to eat a normal diet (46%), followed by improved cognition (40%) and better speech/communication (37%). Having less seizures and less movement issues were not seen as a priority for participants.

Regarding new treatments, most of the participants will be willing to try a pill or tablet (89%), followed by an oil or liquid form of therapy (81%) and a dietary therapy (78%).

We asked participants about their willingness to take part in clinical trials. The majority of them are willing to participate in clinical trials with a 66% and maybe at a 30%. 85% of the participants reported that having telemedicine or remote virtual appointments available would make it more likely for them to participate. The majority of participants indicated that the most common outcome measures they would be willing to use in a clinical trial are, first, bloodwork and cognitive assessments, both at at a 89%, second, EEGs and Speech and language assessments (85%) and third walking and/or gait assessments and endurance tests (80%).

Participants feel that the top three symptoms that impact their quality of life the most are: cognitive or intellectual difficulties in first place (48%), followed by speech and communication issues (46%) and challenges around the diet (34%).

Looking back at the results from our collective voices survey, to have a better understanding of the most impactful symptoms on quality of life, most participants reported that math and writing/composition are the hardest subjects at school. In addition, most of the participants reported having deficits in attention/focus, visual attention, planning and organization, and memory among others.

The most common challenges around speech and language, are issues with fluency and minor articulation. In addition, participants reported that on average they are able to speak well enough to communicate at around 3.5 years of age.

Regarding challenges around the ketogenic diet, most participants reported the diet makes them feel different and that the diet impacts their familie's celebrations and holidays. Despite the challenges, most participants (83%) indicated that the benefits of the diet outweigh the challenges.

Finally, we asked adult participants about their quality of life and most of them (42%) indicated that they feel their quality of life compared to childhood has improved and that the impact or severity of GLUT1 Deficiency is 6 in a 1 to 10 scale.

In summary, the results presented here are a preliminary report of the families and patients perspectives. We want to hear from all the people in our community, so if you are a patient of family member, please consider taking this survey. Thank you!