Family Responses
Family Session - Goal & Methods

- A series of questions aimed at understanding the voice of the patient through the caregiver were asked.
- Participants engaged in an interactive question session.
- Caregivers were split out among tables & tasked with answering 1 question as a group.
- A group leader was asked to summarize the discussion.
- Caregivers also shared individual perspectives using Menti.
Breakout Session Questions

1. If you could heal one thing about *STXBP1* for your child what would it be & why?
2. What keeps you up at night about your STXer & why?
3. What aspect of clinical trials make you the most uncomfortable & why?
   • What could help?
4. What aspects of clinical trials are the most exciting & why?
5. Would you consider gene therapy? Why or why not?
6. Are there other types of therapies/drugs that give you hope? Why?
7. What are some key features/aspects of *STXBP1* that you feel parents & clinicians should observe?
   • How might we measure them?
   • Have you seen differences in these features when your child is feeling better or worse?
8. What is the best thing you have done as a caregiver for self-care?

9. What are some deal-breakers to having your child participate in research & why?
   • What would you like researchers to know and why?

10. What research do you want to see happen on STXBP1 Disorders in next 1-5 years & why?

11. What is the most difficult aspect of having a family member with STXBP1 & why?

12. What are some pros/cons to the registry?
   • What could help you with those concerns?
What we would heal....

- Personal care / potty especially at puberty because of abuse risk
- Communication especially wants & needs
- Behavioral / safety for siblings, family & self
What keeps us up at night...

- Regression due to epilepsy (during puberty?)
- Communication – finding ways to talk to our child
- Physical safety
What excites us about clinical trials & why…

- Repurposing already existing medication –
  - already known safety profile,
  - faster availability to our community
- Looking at natural resources in remote locations
- Researchers are noticing our organization & parent participation!
What gives us discomfort about clinical trials…

- Unknown side-effects
- Invasive procedures or treatments
  - “STXers are happy kids who seem unaffected with their condition; we would hate put them through anything that could affect their quality of life”
- Having to repeat invasive interventions
- Drug / intervention may not help universally

What could help:

- More personalized treatments
- Non-life threatening / altering
We would consider gene therapy if…

- High likelihood of success
- Benefits outweigh risks
- Low risk of adverse reaction to treatment
These other treatments hope...

- Boost working genes
- Phenylbutyrate
- Natural remedies
- Other “sensory” issues to understand the world better (beyond STXBP1)
What should researchers observe?

- Child’s ability to communicate

- Sleep / lack of restful sleep and negative effects (eg, not being able to participate in activities as a result)

- Common behavioral traits:
  - Leg cross
  - Mouth grabbing
  - Non-verbal communications

- How to measure if it works:
  - Progression over time

- “We can’t tell if these have gotten better over time because we can’t communicate with them”. It is hard to know if they are just having a good or a bad day or if it is a medication.
Best things we do for self-care…

- Making a conscious decision to make time for self
- Sources for respite
- Letting others provide care
- Physical & mental health
- Recognize limitations

Example:

“We have a program through our church that let’s us have time to go out and they take siblings to”
The most difficult aspect of having family member with STXBP1

- Funding – getting the money for medical intervention & supplies. This is especially hard if we make too much to get aid.

- Lack of communication skills – not sure if child is cold, hurt. Example: What does ear tugging mean?

- Having to always be their advocate – they have no voice. Always pushing, going “the extra distance” for them, driving far to get right resources & help.
Deal-breakers to participation in research are…

- Data security and protection

- Feedback and results:
  - How do I know my child’s sample used for a research study?
  - What were the results?
  - It is really important that we have a feedback loop and share information

- Concern about pain / discomfort of sample collection
Pros & Cons to Data Registry

Pros:
- Research community wants it
- Everything in one spot
- Way to connect with new families
- Getting correct information to the researchers
- Sharing data

Cons:
- Ease of researchers obtaining data
- Unclear where I am in the process
- How to correct changes
- More Simons follow-up but more concise
- Privacy concerns
- Fear that if we don’t have enough people who have completed surveys and there won’t be enough representation