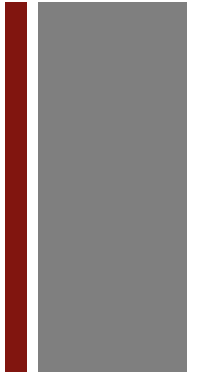




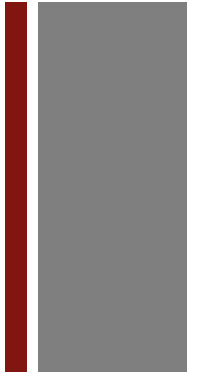
Family Responses

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



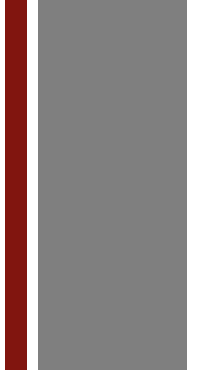
1. If you could heal one thing about *STXBP1* for your child what would it be & why?
2. What keeps you up at night about your STXer & why?
3. What aspect of clinical trials make you the most uncomfortable & why?
 - What could help?
4. What aspects of clinical trials are the most exciting & why?
5. Would 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?

Breakout Session Questions



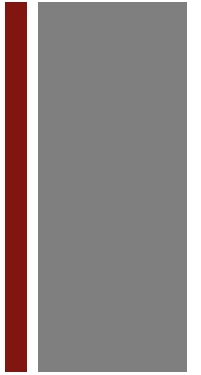
8. What is the best thing you have done as a caregiver for self-care?
9. What are some deal-breakers to having your child participate in research & why?
 - What would you like researchers to know and why?
10. What research do you want to see happen on STXBP1 Disorders in next 1-5 years & why?
11. What 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....



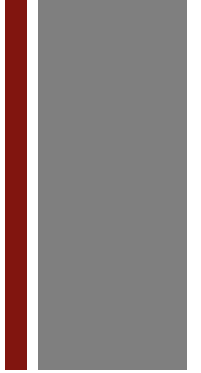
- Personal care / potty especially at puberty because of abuse risk
- Communication especially wants & needs
- Behavioral / safety for siblings, family & self

What keeps us up at night...



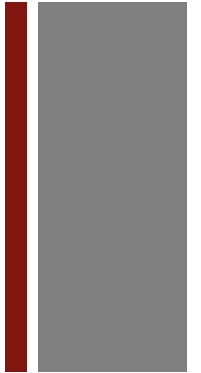
- Regression due to epilepsy (during puberty?)
- Communication – finding ways to talk to our child
- Physical safety

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...



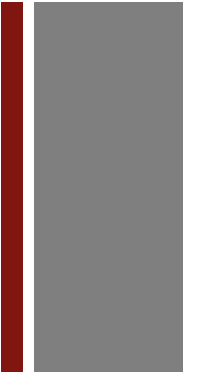
- Unknown side-effects
- Invasive procedures or treatments
 - “STXers are happy kids who seem unaffected with their condition; we would hate put them through anything that could affect their quality of life”
- Having to repeat invasive interventions
- Drug / intervention may not help universally

What could help:

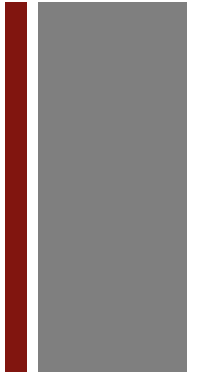
- More personalized treatments
- Non-life threatening / altering

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?

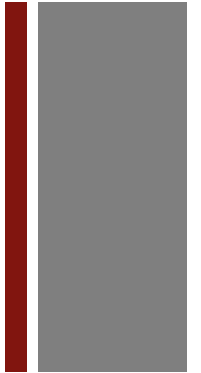
- Child's ability to communicate
- Sleep / lack of restful sleep and negative effects (eg, not being able to participate in activities as a result)
- Common behavioral traits:
 - Leg cross
 - mouth grabbing
 - non-verbal communications
- How to measure if it works:
 - Progression over time
- “We can't 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...

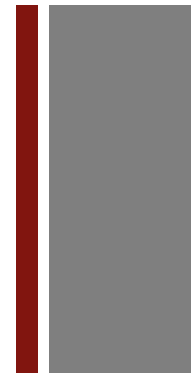
- Making a conscious decision to make time for self
- Sources for respite
- Letting others provide care
- Physical & mental health
- Recognize limitations

Example:

“We have a program through our church that let’s us have time to go out and they take siblings to”

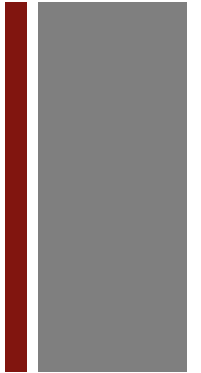


The most difficult aspect of having family member with STXBP1



- 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