Raw Mentimeter Results & Key Themes
How do you feel coming into this meeting?

- hopeful
- excited
- not alone and hopeful
- informed
- optimistic
- intrigued
- grateful
- very good
- helpful
What are the most difficult symptoms of STXBP1?
What else?

communication

potty unclear future
communication long term care
they seem locked inside behavioral issues
her future

self harm society doesn't understand
lack of safety awareness communication overall
long term outlook bathroom use
future

caregiving

motivated

choking
Would you invest in research even if it may not benefit your child?

- Yes: 24
- No: 5
What research do you want to see long-term?

cure
gene therapy
increase protein expression
cornell is covering a lot
specific treatment options
a cure
cures
treatments
cure or reduce challenges
issues with global development
How will you know if a therapy is working?

- decreased symptoms
- hitting milestones
- increased cognition
- observing progression
- faster inchstones
- reduced in seizures
- improved cognition
- long term study
- successful research
- improved abilities
- ability to communicate
- improved or less symptoms
- improve motor skills
- monitor symptoms
- progress
- communication
Would you participate in a natural history study to understand disease progression?

- Sign me up: 21
- It depends: 6
Would you participate in a drug study?

- Willing to try Phase 1 trial (first in humans): 7
- Only if a drug is on the market: 2
- Only if it has been used in Phase 2 trials: 13
- Only if it has been used in Phase 3 trials: 5
What was the most interesting thing we learned…

<table>
<thead>
<tr>
<th>Phenylbutyrate and Curcuman Oil</th>
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</thead>
<tbody>
<tr>
<td>Drug therapy options</td>
</tr>
<tr>
<td>The amazing research that is being done already</td>
</tr>
<tr>
<td>Where we are for research</td>
</tr>
<tr>
<td>Science works in network</td>
</tr>
<tr>
<td>Gene therapy</td>
</tr>
<tr>
<td>Very hard to narrow down . . . Detailed mechanism of the syntax binding disorder</td>
</tr>
<tr>
<td>The increased amount of research and awareness that surrounds STXBP1</td>
</tr>
<tr>
<td>Both meeting other families for the first time and possible treatments</td>
</tr>
<tr>
<td>Progress with curcurma essential oils</td>
</tr>
<tr>
<td>Pharma / compound studies</td>
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<tr>
<td>Model systems and drug information</td>
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</tbody>
</table>
What was the most interesting thing we learned…

| Research underway |
| Botanical and plant drugs |
| Even though there is a broad spectrum of disorders, there are still groupings of types of the STX disorder. |
| About phenylbutyral increasing brain activity |
| All of the different ways in which people are studying STXBP1 |
| Promising Research |
| Let the research is paying off and successful in the labs |
| Botanical and natural drugs |
| That the scientific community is actively researching STXBP1! |
| Progression in animal studies being done |
| Drug therapy options |
What was the most interesting thing we learned…

<table>
<thead>
<tr>
<th>Worm study</th>
</tr>
</thead>
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<tr>
<td>How the protein works and what is being done to understand what is normal and what is not...and hopefully that will lead to a list of direct symptoms proven by lack of this protein.</td>
</tr>
</tbody>
</table>

<table>
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<th>The various forms of research around the world</th>
</tr>
</thead>
<tbody>
<tr>
<td>The attention that our rare disease is receiving in the research field - especially the Cornell study that seems very promising.</td>
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</tbody>
</table>

| Mortality rate is low for STXBP1 |
What are your impressions of today?

Words: hopeful, community, excited, optimistic, inspiring, educated, valuable, well organized, sense of family, amazing, cure, dedication, family, detailed, change maker, researchers want to help, collaborative, collaboration, optimistic, positive families, great content, impressive, family excitement, exciting, motivating, organized, ecstatic, love, uncertainty, informed, epilepsy focused, lots of progress, thought provoking, change, unified, insightful, interesting, informative, uniting, very informative, inspiration, epilepsy, focused, lots of progress, thought provoking, change, unified, insightful, uniting, very informative, inspiration, epilepsy, focused, lots of progress, thought provoking, change, unified, insightful, uniting, very informative, inspiration, epilepsy, focused, lots of progress, thought provoking, change, unified, insightful, uniting, very informative, inspiration, epilepsy, focused, lots of progress, thought provoking, change, unified, insightful, uniting, very 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What are hurdles to joining the Simons Registry??

- Unwilling to share - No time
- Maybe - I need more information
- None!

21
4
1
What research do you want in the next 1-5 yrs

- Start to design new drugs
- Repurpose existing drugs
- Study disease progression
- Make cell lines to screen drugs
- Make animal models (e.g., fish, mice, worms)
- Gene therapy
Would you consider gene therapy?

- Yes: 12
- No: 1
- Maybe: 13
How often would you be willing to travel?

- Never: 0
- Once: 1
- 1-3 times if local: 2
- 3+ if local: 4
- 1-3 times flying nationally: 1
- 1-3 times flying internationally: 1
- 3+ flying nationally: 2
- 3+ flying internationally: 0
- Whatever it takes: 15
Is hospitalization OK during a study?

- No thank you: 1
- OK for 1 day: 11
- 1-3 consecutive days is OK: 8
- 3+ is OK: 6
Would you give a drug that is being tested?

- Never
- Some trade-off OK if main symptoms improve
- If nothing gets worse
- If works on some symptoms
- Only if works on all symptoms
What risks are acceptable in a drug study?

- Low - small # minor side-effects: 17
- Medium - moderate # of minor side-effects: 4
- High - larger # of minor side-effects, Small # Serious side-effects if reversible: 1
- None - No level of risk is acceptable: 2

Total: 24