



Family focus group summary report

Initial report

Prepared March 8, 2022

Participants:

Focus group 1 – March 4

- Bainbridge-Ropers Syndrome (age 24)
- Bohring-Opitz Syndrome (age 4)
- Bohring-Opitz Syndrome (age 10 months)
- Bainbridge-Ropers Syndrome (age 20)
- Bainbridge-Ropers Syndrome (age 9)
- Bainbridge-Ropers Syndrome (age 24)

Focus group 2 – March 7

- Bohring-Opitz Syndrome (age 33)
- Bainbridge-Ropers Syndrome (age 4)

Focus group 3 – March 8

- Bainbridge-Ropers Syndrome (age 13)

Summary feedback on key questions

What are your unanswered medical questions?

Broad themes	
Future life/services needed	4
How/why did mutation happen (preventable?)	2
What are limits of child’s abilities?	1
Specific themes	
Concerns about teeth/crowding	1
Concerns about feet/needing surgery	1
Seizure onset or return of seizures	2
Muscle tone (related to movement)	2
Gut/GI/constipation	1
Anxiety/behavioral	1

If you could wave a magic wand and fix/alleviate one symptom, what would it be?

Brain malformation	1
Communication	7
Mobility/independent movement	4
Anxiety	1

If you could decide where to spend research money, what would you spend it on?*

Does the location of the mutation correlate to severity of symptoms? (To predict child's future)	2
What interventions/therapies/equipment are needed?	2

**NOTE: We did not get to this question with the first focus group on Friday, March 4; if you were in this group and would like to provide an answer, please email your response to Amanda Johnson (amanda@arrefoundation.org)*

Other broad themes of interest:

- Strong connection through music
- Strong need for routine
- Cycles of progression and regression (have skills/abilities, then lose them)
- Being nonverbal does not mean non-communicative
- Not wanting a diagnosis or being wary of a diagnosis in case the diagnosis was degenerative or there was a known limited life span