



Research Roadmap

ARRE
ASXL RARE RESEARCH ENDOWMENT

Family Survey results

Prepared June 8, 2022

Demographics questions

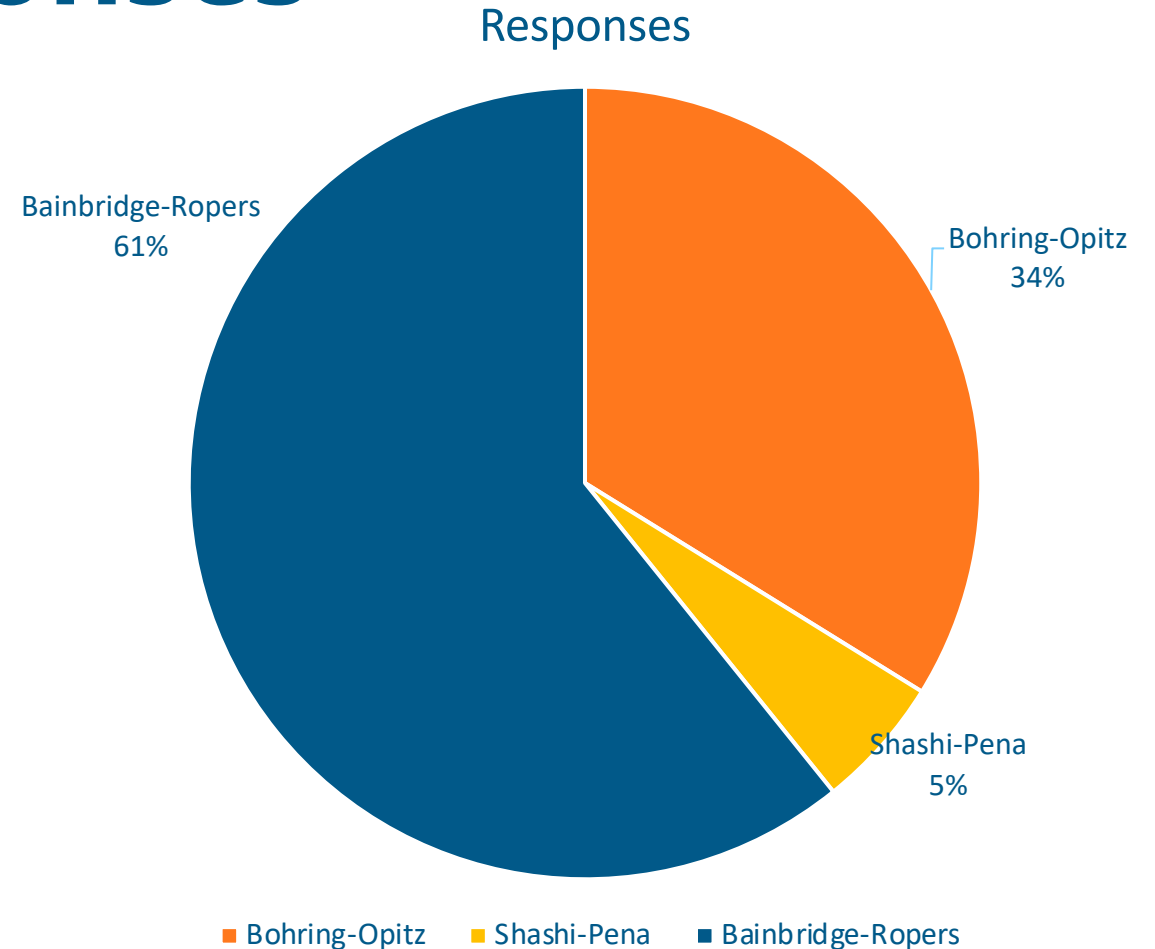
- Which syndrome the patient has
- Patient's birth year
- Patient's year of diagnosis
- Patient's type of diagnosis (clinical diagnosis, diagnosis through genetic testing, or both)

Research priority questions

- If we knew more about _____, life would be better.
- Which symptom or challenge is the most important to you that we try to alleviate or improve?
- How should the ARRE Foundation balance the following:
 - Investing in research that **addresses the underlying cause** of ASXL syndromes
 - Investing in research that **addresses the symptoms** of ASXL syndromes
 - *NOTE: Families responded to this question by choosing a point on a sliding scale where 0 points was the underlying cause and 100 points was addressing symptoms*

Family Survey responses

- 205 complete responses
 - From 28 countries
 - In 6 languages
- By syndrome:
 - Bohring-Opitz: 69
 - Shashi-Pena: 11
 - Bainbridge-Ropers: 124



Survey results overview

Analysis process

- Coded all responses into standard categories
- Used counts of each coded response to count number of responses
 - Grouped like categories together
 - i.e. “GI – vomiting” and “GI – feeding” > broad “GI-related” category

Example coding:

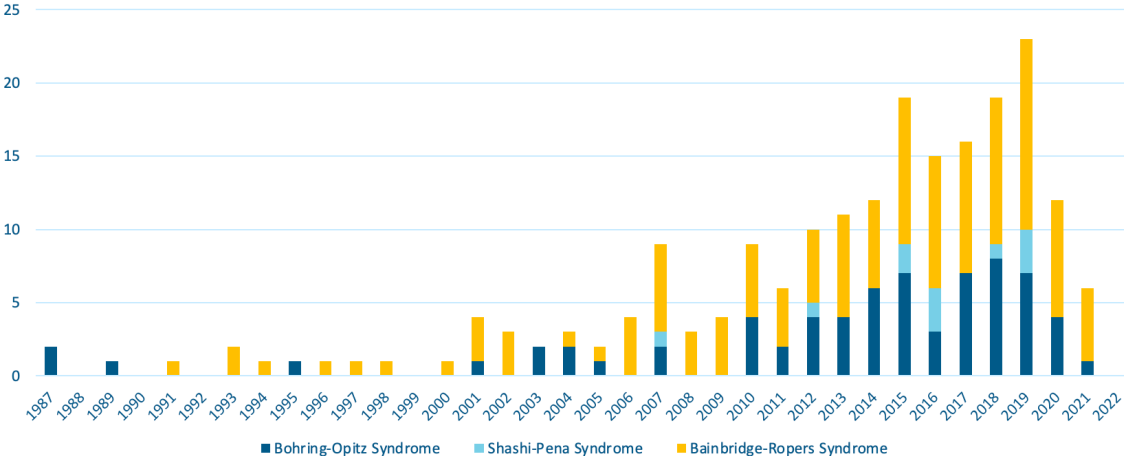
ASXL1/Bohring-Opitz	109
All	1
Appreciating danger	1
Behavior: autistic behaviors	1 6
Behavior: general	1
Behavior: self-harm	4
Breath holding	1
Cardiac concerns	2
Colic	1
Communication: apraxia	1 11
Communication: general	5
Communication: nonverbal	3
Communication: speech	2
Cure	1
Dental complications	1
Developmental delay	2
Family QOL	1
GI: feeding	5 21
GI: general	6
GI: motility	2
GI: reflux	2
GI: vomiting	6
Growth	1 3
Growth: weight gain	1
Growth: weight loss	1
Hyperventilation	1
Hypotonia	2
Immune response	1
Independence	1
Infections	1

Demographics

- People are getting diagnosed faster
- There are likely many people in their 20s and 30s (and likely older) living undiagnosed

Age group	# people	Avg. age of Dx
Infant/toddler (2021-2022)	6	.5 years
Preschool (2018-2020)	54	1.6 years
Child (2010-2017)	98	3.6 years
Teen (2004-2009)	25	10.1 years
Adult (2003 and earlier)	21	20.6 years

Patients by birth year (by syndrome)



Research priority themes

- Families want to know what they are dealing with and what's coming
 - The syndrome itself
 - Future – what's next, what their child is capable of
- There is commonality across syndromes about the most challenging symptoms to address
 - GI-related symptoms
 - Communication
 - Behavior
 - Intellectual disability

All responses

“If we knew more about _____, life would be better.”

BOS		SPS		BRS	
Future-related	13	Future-related	5	Syndrome itself	22
Syndrome itself	10			Future-related	21
Behavior	6			Communication	18
GI-related	5			GI-related	14
Pain	5			Behavior	11

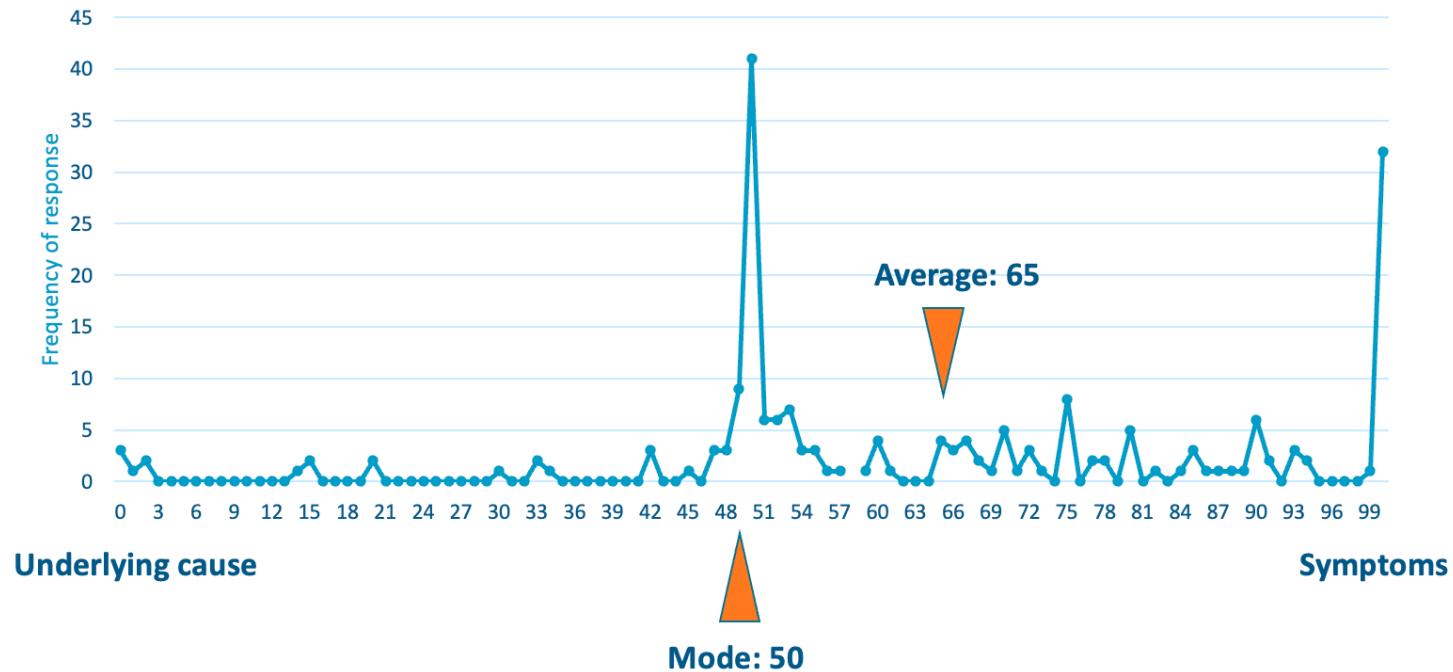
All responses

Most important symptom to address

BOS		SPS		BRS	
GI-related	21	Hypotonia	4	Communication	55
Communication	11	Behavior	3	Behavior	39
Respiratory issues	11	Intellectual disability	3	GI-related	27
Mobility	9	Seizures	2	Intellectual disability	13
Intellectual disability	7			Sleep	13

Research priority themes

- There is broad agreement across syndromes to pursue research that leans slightly more heavily toward symptom management



Response by syndrome	Average rating
Bohring-Opitz	63
Shashi-Pena	67
Bainbridge-Ropers	65