



2022 Annual Report

ARRR

ASXL RARE RESEARCH ENDOWMENT

What we do

We are the link between the family community and the medical and research community.

We help families:

- Understand best practices for care management
- Continually learn more about ASXL-related disorders, their symptoms, and complications
- Drive the research agenda in the direction that is most meaningful to them

We help medical professionals and researchers:

- Understand the impact of their work on families
- Get the samples and data they need for research; we also directly fund some research
- Learn from each other based on their own experiences with ASXL patients

Our mission

Our mission is to support research and education that lead to improved quality of life for individuals with ASXL syndromes.

Our vision

We envision a world where all individuals with an ASXL syndrome have access to evidence-based treatments and engaged healthcare professionals who have knowledge and experience treating ASXL syndromes.

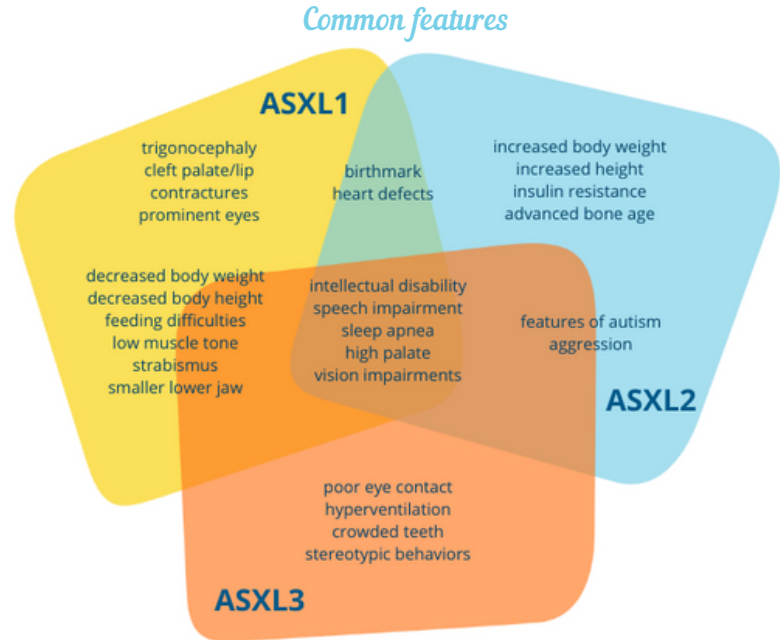
About ASXL-related disorders

ASXL syndromes are caused by a variant (mutation) in one of the three ASXL genes:

- **ASXL1/Bohring-Opitz Syndrome (BOS)**
- **ASXL2/Shashi-Pena Syndrome (SPS)**
- **ASXL3/Bainbridge-Ropers Syndrome (BRS)**

Each of these rare syndromes has a large spectrum of potential symptoms that vary in severity.

With so little understanding of what the ASXL genes do and the progression of these disorders, families are the world's best experts and advocates in their children's care.



arrefoundation.org/asxl-syndromes

Dear friends,

Effie Parks, a rare disease mom and host of the Once Upon a Gene podcast, said recently, “There is no wizard behind the curtain.” There is no magic ASXL button we can push to find the many answers we need to help our kids live their healthiest, most fulfilling lives. It is up to us as families to fund the research, connect the right people to do the right studies, and bring more research attention to ASXL-related disorders.

And that’s exactly what we’re doing together as a community.

This report shares the highlights of the progress we have made in the last year. I invite you to join me in marveling that 40% of the ARRE Foundation’s support comes from individual donors with a connection to an ASXL family (page 17). **It is families who are driving ASXL research forward.**

The ARRE Foundation has an incredible team of volunteers, a rapidly growing network of researchers, and a strong infrastructure to support the work we need to do. We have spent the last year+ building our Research Roadmap (page 11-12) to define what those next steps are and ensuring our priorities are centered on the needs of families.

I am so proud of the progress we are making, and I am so grateful to you for supporting this work. I can’t wait to see you in Baltimore in July 2024!

Laura Badmaev

Founder and chair, ARRE Foundation

Mom to Alex, Bohring-Opitz Syndrome (ASXL1)



2022 Leadership

Board of Directors

Laura Badmaev, *Chair*
Daniel Ordower, *Treasurer*
Julie Lopez
Melissa Richards
Mike Salad

Volunteers

Lauren Adams
Sheri Bermejo
Lee Esposito
Sankar Madhavan
Lauren O'Neil
Jamie Ordower
Sarah Scott



Medical and Scientific Advisors

Stephanie Bielas, PhD
Natasha Ludwig, PhD
Bianca Russell, MD
Wen-Hann Tan, BMBS

2022 ASXL Research Symposium and Family Conference

The 2022 ASXL Research Symposium and Family Conference at UCLA was the highlight of the year. We welcomed over 100 members of the ASXL community including families with Bohring-Opitz, Shashi-Pena, and Bainbridge-Ropers Syndromes, researchers, doctors, and new friends from other rare disorder organizations.

Travel grant recipients

We awarded our first travel grants, which are travel stipends so young investigators can attend research conferences to learn and present their work. Four young investigators received travel grants and presented their research at the trainee poster session during the ASXL Research Symposium.



Family education and connections

The ASXL Family Conference provided a full day of education sessions with medical topics presented by healthcare professionals in the morning and families sharing their experiences in the afternoon. In between sessions, families enjoyed connecting with each other.





Scientific updates and discussions

The ASXL Research Symposium included scientific updates from the core group of researchers who are working in ASXL-related disorders. The meeting generated new ideas and research collaborations in this growing network of professionals who are studying how the ASXL genes work and how best to treat their related disorders.



7 studies collect data

Research teams from seven studies enrolled participants and collected data from children and families at the conference. Having so many families in one place is an invaluable research opportunity to collect blood samples and data that is advancing our understanding of ASXL-related disorders.



Highlights from 2022

Networking nationally to build relationships

Our presentation about our Research Roadmap family survey earned a top score at the National Organization for Rare Disorders annual summit providing an opportunity to share ASXL family research priorities on a national stage. We were also invited to attend the Rare Entrepreneur Bootcamp, hosted by Ultragenyx and Global Genes. Amanda Johnson attended the meeting to learn about the drug development process for ultra-rare genetic disorders and build connections with industry partners.

Research network grows to 34 professionals in 2022

We grew the ASXL research network by 14 people in 2022. These are scientists, doctors, and other healthcare professionals who have an interest in ASXL-related disorders and the ASXL genes.

ASXL-related disorders recognized in legislation

In December, President Biden signed a \$1.7 trillion government spending bill with a provision that recognizes the three ASXL-related disorders. The language in the bill encourages the National Institutes of Health to "expand funding for basic, clinical, and translational research" into these syndromes.





ASXL1/Bohring-Opitz Syndrome

Rory

He loves Tiny Desk and Daniel Tiger.

His biggest challenge is keeping a consistent weight.

He is most proud of playing his toy piano.



Research publications

We celebrate every time researchers publish new information about ASXL genes or ASXL-related disorders. These articles of significance were published in 2022:

DNA methylation signature associated with Bohring-Opitz syndrome: a new tool for functional classification of variants in ASXL genes*

Z. Awamleh, et al. (2022)

European Journal of Human Genetics

Familial Bainbridge-Ropers syndrome: Report of familial ASXL3 inheritance and a milder phenotype

S. Schirwani and E. Woods, et al. (2022)

American Journal of Medical Genetics

PR-DUB preserves Polycomb repression by preventing excessive accumulation of H2Aub1, an antagonist of chromatin compaction

J. Bonnet, et al. (2022)

Genes & Development

arrefoundation.org/published-research



**Research supported by the ARRE Foundation*



ASXL3/Bainbridge-Ropers Syndrome

Luke

He loves his big brother Jake.

His biggest challenge is sleep.

He is most proud of his sense of humor.

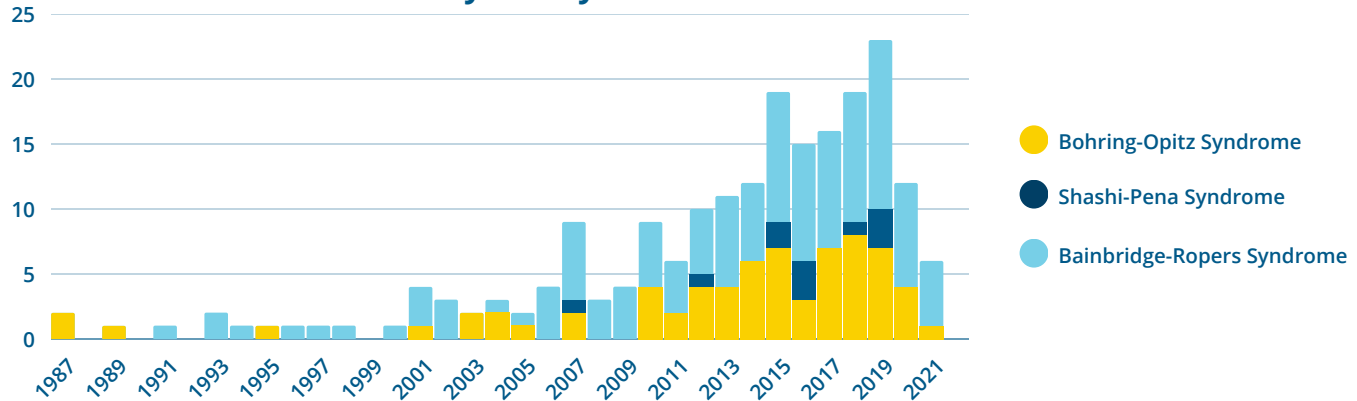


Research Roadmap

A strategic plan to lead us toward life-improving treatments as quickly and efficiently as possible

One of our biggest initiatives of the year was launching the development of our Research Roadmap, our strategic plan for future research investments. The plan is based on priorities identified by 205 families in a survey conducted in May. We then hosted a strategic planning meeting with stakeholders from all parts of our community to identify the top research needs. Insights from the survey and meeting are now being developed into a written strategic plan that includes a timeline, level of effort, cost, and resources. See page 13 for some of the initiatives already underway.

Patients by birth year



Age of diagnosis by age group

Age group (birth year)	Number of people	Average age of diagnosis
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



What did we learn from the research priority survey?

- “Future-related” questions are a top priority
- Symptoms related to gastrointestinal issues, communication, behavior, and cognition are the most important to address
- Families support balancing research investments between addressing the underlying cause of ASXL-related disorders and managing symptoms

Research Roadmap
ARRE

arrefoundation.org/research-roadmap

So far in 2023

While our research strategic plan is still being finalized, we got a jumpstart on implementing some of the key action items that will be in the strategic plan. So far in 2023, we have:

Launched the Research Roundtable series

These quarterly virtual meetings for the clinical and basic science communities provide a regular opportunity to continue discussions and build collaborations.

Built the ASXL Care Directory

This online directory features healthcare providers from around the world who have been recommended by ASXL families.

Developed a program to support newly diagnosed families

Our outreach program connects newly diagnosed families to life-improving resources faster through a welcome call and email series.

Hired a scientific advisor

We hired part-time scientific advising support to complete our research strategic plan and help to grow our research network.





ASXL2/Shashi-Pena Syndrome

Eva

She loves dancing, school, and family time.

Her biggest challenge is conquering physical challenges.

She is most proud of learning new things.



Getting involved

Participate in research

There are multiple research studies that are currently enrolling participants. Each study is important to our progress in finding treatments for our loved ones. We encourage all families to enroll in as many studies for which they are eligible and have capacity to join.

arrefoundation.org/research

Raise money for research

Your fundraising efforts support our family education programs, growth of the scientific community, and direct investments in research. Facebook fundraisers are the easiest way to raise funds, but your creative idea for a special event could be our next big fundraiser!

arrefoundation.org/fundraise

Build capacity as a volunteer

We run on the power of volunteers! No matter your skill set or the amount of time you have to contribute, we can use your talents to advance our mission.

arrefoundation.org/volunteer



Funded by families

Our work is largely funded by families who are part of our ASXL community, including direct contributions and lots and lots of fundraising efforts!

Friends and family members in our community hosted 66 Facebook fundraisers in 2022. By reaching out to their own networks for awareness days, birthday fundraisers, and GivingTuesday, families raised over \$27,800 through 441 donations in online fundraising.

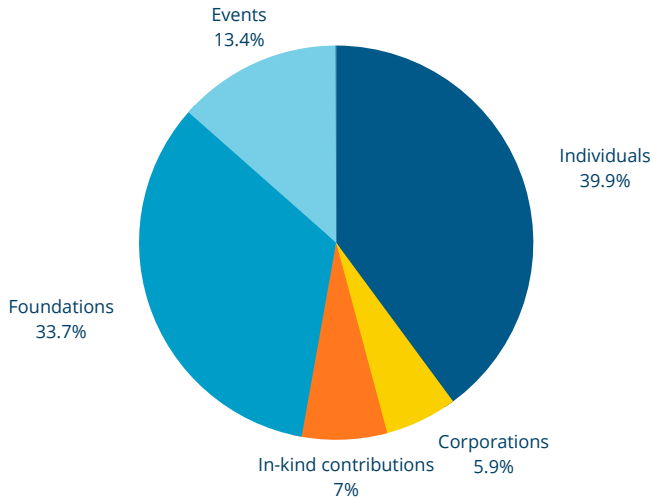
GIVING
TUESDAY



Shashi-Pena Syndrome Awareness Day
Scott Family

2022 financial report

Revenue by source



Revenue

Individuals	\$91,125
Corporations	\$13,547
In-kind contributions	\$15,946
Foundations	\$77,000
Events	\$30,723
Investment gains	\$86

Total revenue: \$228,427

Expense

Research grants & awards	\$64,762
Conference expenses	\$34,135
Research network & family education	\$110,328
G&A	\$14,324

Total expense: \$223,549

Looking ahead

Hosting the 2024 ASXL Research Symposium and Family Conference

Planning is underway for the 2024 ASXL Research Symposium and Family Conference at Kennedy Krieger Institute in Baltimore, Maryland. This meeting catalyzes the growth of our network of families and researchers, provides rich opportunities to collect data from families, and allows families and researchers to connect with and learn from each other.

Investing \$100,000 in ASXL research by the end of 2023

We're allocating \$100,000 to research-related investments by the end of the year guided by the priorities identified in our Research Roadmap. Our funding strategy will include investments in expanding animal and cell models in which to study our disorders, and funds for pilot studies in areas of clinical management that are most important to families.

Expanding capacity to collect samples critical for research

Samples of blood, skin, and other biological specimens from individuals with ASXL-related disorders are critical tools for advancing research. We've partnered with COMBINEDBrain to collect multiple types of biological samples in central biorepository that can be accessed by researchers around the world.



Save the Dates!

February 5: Bainbridge-Ropers Syndrome Awareness Day

February 29: Rare Disease Day

April 6: Bohring-Opitz Syndrome Awareness Day

July 18-21: 2024 ASXL Research Symposium and Family Conference

October 6: Shashi-Pena Syndrome Awareness Day

ARRE

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