



ARRE

ASXL RARE RESEARCH ENDOWMENT
Donor Summary 2019



FOREWORD

Dear Friends

Thank you for your donation to the AARE Foundation! We greatly appreciate your continued support.

AARE key tenets include good stewardship of our resources and transparency. Our donor summary shows how your support benefits ASXL syndrome patients and highlights our 2019 accomplishments.

AARE Foundation maximizes funds toward research that will increase our understanding of the ASXL genes and improves the treatment of individuals with congenital ASXL mutations.

Please contact us to learn about more ways to support AARE through planned giving, volunteer efforts, and conference sponsorships.

Thank you for making a difference in the lives of these patients & their families!



Laura Badmaev

laura@arrefoundation.org





PRIORITIES

ASXL Registry & Biobank

These are foundational tools based on patient data & samples, which researchers will utilize to study and increase our knowledge of these syndromes.

Research Grants

Effective research will bring an understanding of the underlying mechanisms and potential treatments for ASXL syndromes.



Conferences

These meetings bring together professionals and families to share information and exchange ideas that will move our mission forward.

ASXL Centers of Excellence

Future ASXL Centers of Excellence are hub hospitals where ASXL patients will see medical specialists who have clinical expertise and can improve their care plans.



“ I have struggled immensely to just have an answer as to why my little girl was so sick. Now I struggle to keep up with her changing needs and unpredictable development. I am so grateful for the work AARE is doing. I can't thank them enough, truly. They are the piece of hope that I so desperately need.
– Amy

London, Bohring-Opitz Syndrome, ASXL1





ASXL REGISTRY & BIOBANK

A registry focused on the natural history, management and treatment of patients with Bohring-Opitz Syndrome (ASXL1), Shashi-Pena Syndrome (ASXL2) and Bainbridge-Ropers Syndrome (ASXL3).

Sponsors & Collaborators:

- University of California, Los Angeles
- Boston Children's Hospital
- Children's Hospital Medical Center, Cincinnati
- Duke University

The HIPPA compliant & IRB-approved, patient-driven Registry incentivizes the study of ASXL syndromes.

Researchers from institutions around the world have access to this freely available patient data and samples, which they use for clinical and scientific studies.

ARRR has provided funding to UCLA to:

- Scale the ASXL registry & ability to process more samples
- Hire an ASXL Registry Coordinator, who will help to increase enrollments, strengthen protocols, and prioritize 3 publications



“ Our son was diagnosed with Bohring Opitz Syndrome, a very rare genetic diagnosis, in 2014 at age 11. There are only about 100 children in the world with this syndrome so it is very difficult to find information, connect with other families, and have scientists take enough notice to study our syndrome, both for those currently affected and for future diagnoses. The ASXL Rare Research Endowment Foundation (ARRE) has been an incredible resource for our son and our entire family. Being able to attend conferences, connect with other parents, and learn from some of the experts from around the world has impacted our ability to care for our son and his sisters. I can't wait to see what they bring to us next!! – *Lori*

Tristan, Bohring-Opitz Syndrome, ASXL1





RESEARCH GRANTS

The ASXL Rare Research Endowment Foundation invites application that propose novel studies on the molecular and genetic mechanisms underlying the function of the ASXL genes.

ARRE will develop a diverse research portfolio that will build on successful studies as well as support innovative investigations with high potential rewards. Key focus areas include:

- Gain or loss of function
- Transcripts expressed post nataly
- Protein expression
- Pathogenic variants

ARRE has approved 5 strong and diverse scientific and clinical research proposals from qualified researchers that will be funded for 2020 -2022.

Each proposal is focused on a different niche and allows for a broad covering of this field:

- Dr. Rosanna Weksberg, Sick Kids Toronto
- Dr. Feng-Chun Yang, University of Texas Science Health Center at San Antonio
- Dr. Valerie Arboleda, UCLA
- Dr. Dale Frank, Israel Institute of Technology
- Dr. Micha Drukker, Leiden University



“ We are thankful for non-profits like ASXL Rare Research Endowment, we hope that with continued research we will know more about how we can help our little ones! Rare diseases, like Shashi-Pena Syndrome-ASXL2 need LOTS of TLC! And of course "ALL" rare disease need TLC!
– Michelle



Noah, Shashi-Pena Syndrome, ASXL2



CONFERENCES

Hosted a successful conference at the University of Michigan with >20+ scientific and clinical speakers & over 30 families, which ignited collaborations, professional engagement, and 5 research proposal submissions

Enrolled families in the Simons VIP, NIH FaceBase, ASXL Registry, and collected patient samples for its ASXL biobank, which has resulted in collaborations with researchers who are developing mouse models, organoids from induced pluripotent stem cells of ASXL individuals, “omics” studies, and methylation signature patterns that will help with diagnosis and identify potential drug targets

Facilitated family informational workshops including special needs trusts & conservatorship, augmentative communication, Make-A-Wish, social worker support, and ADA home accommodations

Organized & provided opportunities at no cost to families for meals, adaptive bowling, therapy dogs, accessible playground activities, and transportation, as well as travel scholarships and hotel subsidies

July 19-21, 2019

ASXL Conference at the University of Michigan

- Professional Research Symposium
- Family Informational Workshops
- Clinical Presentations & Registry Enrollments
- Adapted Activities & Family Fun Day

Fall 2020 (*Cancelled due to COVID*)

Professional Research Symposium at Boston Children's Hospital

Summer 2021

ASXL Family Conference (Virtual)



“ ASXL Rare Research Endowment Foundation has been great the last two years in organizing the second ASXL conference. We were able to attend this year thanks the travel scholarship we received. The event was extremely helpful for us helping us leaning more about this rare condition and also for having the opportunity to discuss with the nation experts in the ASXL associated conditions. The opportunity we had of meeting other families affected by this rare condition has been instrumental in our learning of their management of several co-morbidities associated to Bainbridge Ropers. We are extremely grateful for all the resources provided to us in addition to the travel scholarship, like meals, hotel discounts, etc.

Looking forward to next year conference!!

– *Maria*

Gabriel, Bainbridge-Ropers Syndrome, ASXL3





CENTERS OF EXCELLENCE

Vision

- Elected hub hospitals and specialists around the world that demonstrate exemplary care for ASXL patients
- Travel and financial support to enable families and providers to establish and maintain these practices at the ASXL CoE (Centers of Excellence)
- Find a Doctor database, which will provide contact information for specialists who are ASXL experts

Professional Awareness & Open Sourcing

ARRE will support the broad dissemination of ASXL related papers by providing travel scholarships to researchers presenting ASXL related works at conferences outside of their home country. This will increase awareness of ASXL internationally and help our researchers continue to network and build relationships across borders.

ARRE will also offer grants to authors of ASXL relevant papers that cover the cost of publishing open source, facilitating synergy, and fostering collaboration.

Current Focus

- Build the professional network of researchers and clinicians
- Collect patient data and publish results
- Create awareness and identify potential specialist candidates in the professional community for future medical clinics
- Deepen relationships with hospital & university sponsors of the ASXL Registry



“ This foundation is wonderful. They helped with getting us to a convention for my son who has BRS. We got so much knowledge from the speakers. It was amazing for our whole family. It was also really cool to be a part of the calendar. Thanks for all that you do.

– *Andrea*

Pierce, Bainbridge-Ropers Syndrome, ASXL3



ADVOCACY & COMMUNITY

117 ASXL Symposium
Attendees

495 Social Media
Followers (+143%)

260 Newsletter
Subscribers (+250%)



“ ARRE brings research and families together! My family and I were so proud to be apart of ARRE's first annual conference this past July. It was an event that involved learning from top medical geneticists about current research, bio-banking, and meeting other families that share our experiences. This is a one of kind nonprofit that focuses on improving the lives of our chronically ill children. Thank you for all you do, ARRE.
– Sheri

Brandon, Bohring-Opitz Syndrome, ASXL1



ADVOCACY & COMMUNITY

26 Conference Live
Stream Videos

2 Campaign
Fundraisers

6 Family Stories

10 Education/
Health/Tips



“ AARE has united a group of families who have a child with an ASXL gene mutation. This nonprofit has also given us a voice as well as hope for our children and their futures. AARE advocates for research and educates families regarding best practices for medical interventions as well as therapeutic interventions. This summer we were able to attend the AARE Family Meeting and it was an incredible experience to be able to meet other families just like ours!

– *Caitlin*

Issac, Shashi-Pena Syndrome, ASXL2





RECOGNITION



The **GreatNonprofits Top-Rated Awards** is the one and only people's choice award where volunteers, donors, and people served by nonprofits are asked to share stories of inspiration, express their appreciation, and potentially help nonprofits earn a spot on the prestigious GreatNonprofits [Top-Rated Nonprofits List](#)



NORD, a 501(c)(3) organization, is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them. NORD, along with its more than 300 patient organization members, is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and patient services.



The Highest Level of Recognition Offered by **GuideStar**: GuideStar Platinum encourages nonprofit organizations to share their progress and results in important new ways—moving way beyond simplistic financial ratios—to reflect the changes organizations are making in the world.



“ AARE does so much to help people affected by the ultra-rare ASXL syndromes! They help families connect, help caregivers be better advocates and help bring much-needed awareness to medical professionals.





– Colleen

Alex, Bohring-Opitz Syndrome, ASXL1



DONATIONS



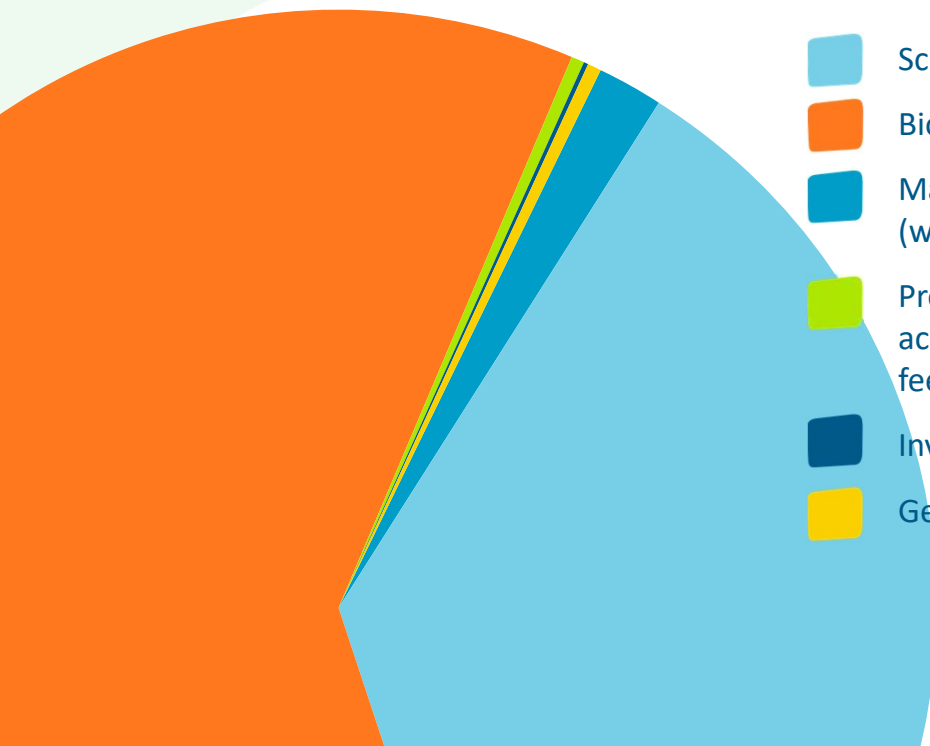
	Grants:	\$878,500.00
	Fundraisers:	\$10,972.00
	Direct donations:	\$2,751,66
	Investment income:	\$4,593,78







\$ 896,817.44



EXPENSES

\$ 122,036.55



	Scientific Conference (UCLA, July 2018)*	\$43,836.85
	Biobanking	\$75,000.00
	Marketing & Fundraising Expenses (website and credit card donation fees)	\$2,172.15
	Professional Services (lawyer, accountant, incorporation & license fees)**	\$425.00
	Investment expenses & fees	\$150.00
	General Expenses	\$452.55



“ This is an excellent organization that is providing much needed resources for families ASXL. We attended conference and as a newly diagnosed family it gave us so much valuable information and support, we needed.

- *Heather*

Emily, Bainbridge-Ropers Syndrome, ASXL3



THANK YOU!

#ASpire2XL *Achieving more together*

With your support, together our accomplishments set a strong foundation for our future. The expansion of the ASXL Registry, successful professional and family conference, & research proposals continue to drive our mission forward.

Your continued financial support and efforts to spread awareness and volunteer expertise are critical to our success. ARRE will strategically invest in our core priorities to bring our vision to fruition.



We look forward to a future success and invite you to join us on this incredible journey!

Laura Badmaev

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ARRE
Donor Summary 2019



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Thank you!



LEAVE YOUR LEGACY

Planned giving is an opportunity to make a significant difference in the lives of these children and their families.

To learn more and discuss planning your gift to AARE Foundation, please contact info@arrefoundation.org.

- Securities
- Real Estate
- Gifts by Will
- Life Insurance Policy
- Charitable Gift Annuity
- Charitable Remainder and Lead Trusts
- IRA Distributions



VOLUNTEER

Call for Volunteers!

Grandparents, Aunts, Uncles,
Family & Friends

ARE has grown exponentially
and needs your support!

- Marketing
- Website/Social Media Updates
- Corporate Sponsorships & Fundraising
- Conference Planning & Logistics
- Grant Writing
- Board Governance
- Accounting & Law Advisory Assistance



ARRE needs assistance from family, friends, caregivers, and neighbors to make our vision possible. We are a volunteer-based organization and your support is critical.

VOLUNTEER

Sign up to receive news and updates about ASXL Rare Research Endowment activities.

SUBSCRIBE

You can help the ASXL Rare Research Endowment remain at the forefront ASXL research: arrefoundation.org/donate

DONATE



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