



2021 – 2023 Strategic Plan

Summary for External Partners

Approved by the Board of Directors on July 21, 2022

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ABOUT

ASXL syndromes are the result of random genetic changes in ASXL genes that occur around the time of conception. There are three ASXL syndromes: ASXL1/Bohring-Opitz Syndrome, ASXL2/Shashi-Pena Syndrome and ASXL3/Bainbridge-Ropers Syndrome. Many children have global developmental delays and complex medical issues that can include severe neurological and respiratory issues. Most individuals are completely dependent on caretakers for every aspect of their care, though there is significant variation in each individual's abilities and function.

The number of people with an ASXL syndrome is unknown. Only a few cases of each syndrome have been described in the medical literature. Based on the grassroots network of families that have formed on Facebook, we estimate the global number of diagnosed cases of Bohrning-Opitz Syndrome at 150-200, Shashi-Pena Syndrome at 12-15, Bainbridge-Ropers Syndrome at around 300. It is probable that there are a significant number of undiagnosed cases worldwide. Whole exome sequencing, the specialized genetic test required to conclusively diagnose these syndromes, has only become available in the last 10 years. As access to this diagnostic genetic test increases, the frequency of diagnosis is increasing. While much of the published research to date has focused on young children, we are learning of older individuals in their 20s who have lived undiagnosed until relatively recently.

Due to the limited research on ASXL syndromes, there are significant unknowns about how the ASXL genes work and what they do. Similarly, there are few evidence-based publications for medical professionals to reference in managing the care of someone with an ASXL syndrome. A significant challenge of the ASXL community is that families often see specialists who do not have the knowledge or experience to properly manage their care due to the rarity and unknowns of their syndrome. In many cases, parents and caregivers become the experts in their child's care and take on an outsized role in educating medical providers and advocating for their child's needs.

The ARRE Foundation is the only organization dedicated to all three ASXL syndromes. Through research and education, we aim to improve the quality of life for these individuals and their families. All members of the ARRE Foundation volunteer team have been personally impacted by this group of disorders and we are highly motivated to continue to elevate the level of care these families receive and improve the scientific understanding of these disorders.

Vision and mission

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.

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

Guiding principle

The guiding principle in the development of this plan is as follows:

The ARRE Foundation is the link between the patient community and the medical/research community.

We help families:

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

We help medical professionals/researchers:

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

Values

To accomplish our mission, the ARRE Foundation embodies the key values of integrity, respect and community. We act in the best interests of ASXL-affected individuals and their families.

- **Integrity:** We conduct operations in a manner that is honest, transparent, and ethical. We demonstrate good stewardship of the resources entrusted to the ARRE Foundation.
- **Respect:** We value and listen to all individuals and appreciate their diverse backgrounds, experiences, styles, approaches, and ideas.

- **Community:** We collaborate with ASXL families, friends, advisers, doctors, scientists, and other partner organizations to build relationships and maximize our accomplishments.

Key stakeholders

The ASXL community is small and we believe it will take the entire ASXL community to make meaningful progress toward our goals. The ARRE Foundation will engage all stakeholders in our work.

ASXL-affected families and caregivers

- Newly or recently diagnosed families
- Existing/known families
- Angel families, whose child has passed away

Professional community

- Clinicians and clinician-researchers
- Researchers
- Allied health professionals (therapists, nutritionists, etc.)

Donors and partners

- Current and active funders
- Prospective funders and partners
- Other ASXL syndrome-focused organizations and support groups

ENVIRONMENTAL ASSESSMENT

Accomplishments to date

The ARRE Foundation was founded by a parent in 2018, and launched with the support of other parents, family members and key members of the medical and scientific community. There has been significant progress planning and collaborating with key stakeholders to create a framework for success over the last three years.

Key accomplishments include:

- Hosted two ASXL medical/scientific conferences in 2018 and 2019
 - Clinic day to meet with medical specialists
 - Sample collection to enroll in registry

- Networking with other families
- ASXL Professional Symposium for exchange of ideas related to research
- Funded five initial research grants, each with a two-year funding commitment (2020-2021)
- Funded the ASXL Patient Registry research coordinator at UCLA
- Made significant strides in building the organization's operational infrastructure
 - Built website and social media accounts with a strong visual brand
 - Benchmarked with other rare syndrome foundations/organizations
 - Developed initial governance structure and organizational policies
 - Established relationships with necessary professional advisers, including legal support, investment management and accounting
 - Hired first paid staff person, a Chief Development Officer, in early 2021

Strengths, Weaknesses, Opportunities and Threats

The ARRE Foundation Board of Directors and leadership volunteers completed a strategic planning survey in March 2021. A summary of their assessment of the organization's key strengths, weaknesses, opportunities, and threats are summarized as follows:

Strengths:

- The ARRE Foundation bridges all three ASXL syndromes.
- The ASXL community is passionate and led by enthusiastic and highly motivated leaders.
- The ARRE Foundation has demonstrated success securing financial support for research.

Weaknesses:

- The current research landscape is very small.
- There is broad lack of awareness of the disease area, including among families and the medical/scientific community.
- The ARRE Foundation currently relies heavily on a single funder.

Opportunities:

- There is opportunity to broaden the organization's fundraising sources.
- The networks within the community (families, medical professionals and

researchers) can be more deliberately expanded.

- There is a wealth of knowledge in family learnings and experience that could be used to develop resources for care management.

Threats:

- Limited fundraising potential and/or loss of the organization's primary funder could threaten the organization's financial viability.
- Families could lose engagement with the ARRE Foundation because the research-related work is too abstract and not directly connected to their immediate needs.
- With such a small community, a lack of collaboration between all relevant parties (families, professionals and other groups/organizations) could limit the community's progress.

STRATEGIC PLAN

This strategic plan is intended for the years 2021 and 2022, with some activities extending into 2023. The plan is intentionally narrow in scope and focus to build a solid base for future growth.

Strategic priorities

The central strategic priorities of the ARRE Foundation are defined as pillars of our strategic plan and are supported by goals. The pillars and goals of the strategic plan are as follows:

Pillar 1: Invest in and drive ASXL research

Goals:

- Engage and build the ASXL research community
- Consistently fund promising research grants
- Fund a focused portfolio of basic research and clinical research grants
- Facilitate the sharing of data, resources and ideas among the ASXL research community
- Encourage family participation in the ASXL Patient Registry and Biobank

Pillar 2: Improve the clinical care and management of those with ASXL syndrome

Goals:

- Build the global network of clinicians and other allied health professionals who have experience treating ASXL syndromes

- Develop resources with must-know information for newly diagnosed families and their care team
- Develop topic-specific resources for common challenges experienced by those with an ASXL syndrome

Pillar 3: Educate, empower and connect the ASXL community of patients, families, medical professionals and researchers

Goals:

- Build a diverse, supportive and inclusive community of families, medical professionals and researchers
- Host regularly scheduled, topic-specific virtual education sessions for families
- Provide meaningful engagement opportunities for those who wish to contribute to advancing the organization’s work

Pillar 4: Strengthen organization’s governance structure

Goals:

- Build a sustainable organizational infrastructure for long-term success
- Develop and implement a scalable fundraising strategy to broaden and diversify the organization’s financial support
- Build and deploy a strategic communications plan to support key priorities
- Build trust by stewarding resources wisely and communicating transparently with all stakeholders
- Leverage relationships within the rare disease community to accelerate our work

Scope

The ARRE Foundation’s scope has been redefined since launching in 2018 to bring laser focus on efforts that provide families with information and resources that will help improve clinical management and outcomes related to ASXL syndromes.

The following chart summarizes what activities and initiatives are in scope, out of scope, and somewhere in the middle where might collaborate with other groups or organizations:

In scope <i>Own these; core mission</i>	Gray area <i>Potential collaboration</i>	Out of scope <i>No time/money investment</i>
<p>Raising money for and funding research</p> <p>Building a research network & driving research agenda based on family priorities</p> <p>Developing family-friendly resources to improve clinical management and outcomes</p> <p>Facilitating connections between clinicians and other health professionals to grow the number of experiences/specialized professionals</p>	<p>Connecting new families to knowledgeable doctors and other health professionals</p> <p>Coordinating and centralizing the collective knowledge of families</p> <p>Connecting families to each other</p>	<p>Directly supporting individual families (money or advice)</p> <p>General awareness-raising activities (t-shirts, wristbands)</p> <p>Hosting events with no medical/research information</p> <p>Providing resources related to indirect topics, such as adaptive equipment, home modifications, legal considerations (conservatorship), disability/special needs services by location</p> <p>Things not specific to ASXL community</p>

These scope changes from the ARRE Foundation’s previous work include:

Refocusing the ASXL Family Conference

The ASXL Family Conference serves an important informational purpose for families, as well as bringing the family community together face-to-face. It also serves as an opportunity for the medical and research community to meet the people their work and research benefits. With the revised scope of the strategic plan, the ARRE Foundation will continue to host ASXL Family Conferences but will hone the program to focus narrowly on topics that aid in medical management or are research oriented. We will no longer include topics that are not specific to the ASXL community (see below). The ASXL Family Conferences will continue to serve as a critical opportunity for enrollment in the ASXL Patient Registry, collection of biobank samples, and other research-oriented activities that can take advantage of having many patients and families together in one place.

We will seek the support of our partner organizations in fulfilling the social aspects of the ASXL Family Conference, including family activities and meals.

We will continue to host the ASXL Research Symposium for medical professionals and researchers which fills an otherwise unmet need of bringing the research community together for the sharing of information.

Focusing only on topics specific to the ASXL community

Many peripheral topics touch the ASXL patient community, but are not specific to our community, such as accessing disability services or building an adaptive home. We will focus our resources solely on topics that are directly related to the ASXL community. Many organizations already focus on these topics, and we may link these existing resources, but will not recreate them.

Research priorities informed by families

With limited financial resources to dedicate to research, we must choose what research we fund wisely. The needs of families should be considered in choosing what we fund as part of the balance between funding basic research that may yield fruit decades from now and funding clinical research that has the potential for higher impact sooner. The medical and scientific community will continue to guide the grant recipient selection process, but with a greater emphasis on understanding and meeting the nearer-term needs of families.

Key programs and services

The ARRE Foundation's central programs and services include the following:

Clinical and basic scientific research grants

We will continue to support clinical and basic scientific research that can further characterize the molecular causes and pathogenicity of the ASXL gene family, improve medical care plans, and meet the needs for families who face extraordinary challenges and uncertainty. The call for applications will be paused in 2021 while we seek additional funding to restart the program with stronger, more sustainable financial support in 2022 and beyond with the goal of providing at least one research grant per year to help the program build momentum.

Biennial ASXL Family Conference

We will host a biennial ASXL Family Conference (held on even-numbered years) with a hopeful return to an in-person meeting starting in 2022 in Los Angeles. Conferences will be streamed for live virtual participation and recorded for inclusion in online resource library of on-demand resources for families. Topics will be focused on medical management and research. The program will include opportunities for families to participate in research activities to advance the collection of patient information and samples. We will seek partnership opportunities with related organizations to support the social aspects of the ASXL Family Conference.

Care management tools for families

We will build a toolkit of web-based and printable resources to aid families in the clinical management of their children's care, including as advocates with medical professionals who are unfamiliar with treating ASXL syndromes. These resources will be developed using the collective wisdom of families and the medical community. We will also launch a series of regularly scheduled virtual education sessions (i.e. webinars) that connect the families with specialists on specific topics.

Annual ASXL Research Symposium

We will bring together the major medical and scientific leaders who are studying the ASXL genes and provide opportunities for physicians and researchers to present and share their work through collaborative methods. There will be an emphasis on the inclusion of trainees and junior faculty/residents in the meeting. The symposium will be held virtually in 2021, and will resume an in-person or hybrid virtual/in-person format in 2022 or when deemed safe to do so.

ASXL Professional Network

The ARRE Foundation's ASXL Professional Network seeks to grow and better engage the network for researchers, doctors and other allied health professionals who study and treat ASXL syndromes. We will encourage members to join the network, and will communicate regularly with this group by email and, eventually, a listserv to engage and inform this community who supports ASXL affected individuals.

ASXL Patient Registry and biobanking

The ARRE Foundation will continue to encourage families to enroll in the ASXL Patient Registry and provide biologic samples, which are critical tools and foundational for successful research. The ARRE Foundation will advocate for and disseminate regular updates from the patient registry to families to encourage long-term participation.

Meeting and education schedule

The ARRE Foundation's anticipated schedule of in-person and virtual educational opportunities for our key stakeholders are as follows:

Year	ASXL Research Symposium	ASXL Family Conference	Family-focused webinars
2021	Virtual		4-6 webinars (~ bi-monthly)
2022	In person – Los Angeles	In person – Los Angeles <i>Virtual option available</i>	4-6 webinars (~ bi-monthly)
2023	In person		4-6 webinars (~ bi-monthly)
2024	In person	In person <i>Virtual option available</i>	4-6 webinars (~ bi-monthly)
2025	In person		4-6 webinars (~ bi-monthly)
2026	In person	In person <i>Virtual option available</i>	4-6 webinars (~ bi-monthly)

- *In-person meetings would also have a component for virtual participation*
- *All family education (conference and webinars) to be recorded and added to resource library*