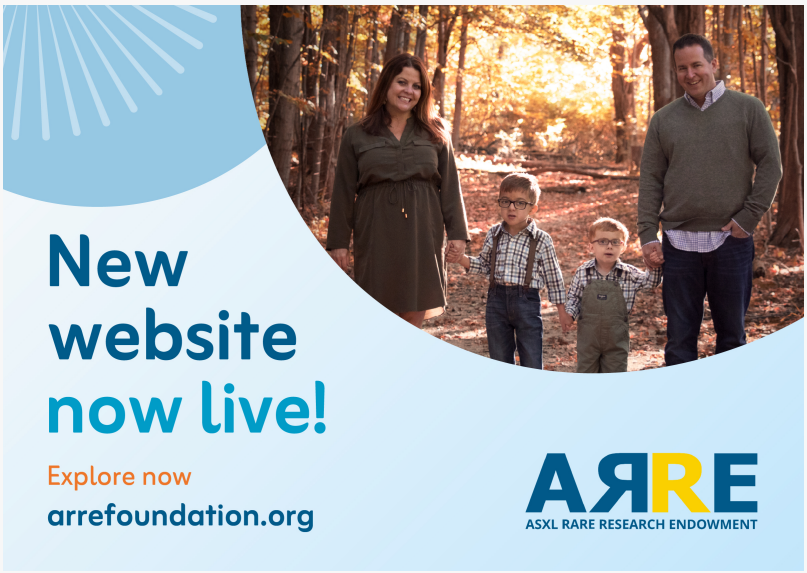
ARRE Foundation launches new website to help improve quality of life for ASXL community

GORHAM, ME, UNITED STATES, September 11, 2025 / EINPresswire.com/ -- The ASXL Rare Research Endowment (ARRE) Foundation has launched a new website designed to better serve families, caregivers, clinicians, and researchers. The site reflects the Foundation's mission to support research and improve care for individuals living with ASXL-related disorders, while also introducing major new features, including a dedicated section for professionals and a community resource database.



ASXL Family

ASXL-related disorders, including Bohring-Opitz Syndrome (ASXL1), Shashi-Pena Syndrome (ASXL2), and Bainbridge-Ropers Syndrome (ASXL3), are ultra-rare and complex genetic conditions. Bohring Opitz Syndrome was first described clinically in 1999 and was molecularly defined by pathogenic ASXL1 variants in 2011. Shashi Pena Syndrome was described in 2016 based on six children with ASXL2 variants. Bainbridge Ropers Syndrome was identified in 2013, when truncating ASXL3 mutations were linked to a distinctive neurodevelopmental disorder.

Because these syndromes have only recently been characterized, published literature is still scarce and families and their medical providers often face challenges finding reliable, consolidated information to guide the complex care of individuals with ASXL-related disorders. Likewise, many clinicians encounter these disorders very rarely.

"The new website is one of the best patient advocacy websites I have ever seen," said Bianca Russell, MD, clinical geneticist at the University of California, Los Angeles, and a member of the ARRE Foundation's Medical and Scientific Advisory Board. "My patients represent numerous rare genetic disorders and I refer families and their pediatricians to patient advocacy websites all the time. The accessibility of the content on the new ARRE Foundation website will make a real

difference for clinicians and their ability to give their patients the best possible care."

The website also introduces a database of materials for parents and caregivers, titled the ASXL Resource Library. This comprehensive collection of guides, tools, and materials supports parents and caregivers in managing daily life. Together, these new resources strengthen the bridge between families and the medical community, ensuring that education and care improve in parallel.

"This website is a reflection of how far we've come as a community since my son was one of the earliest individuals diagnosed with Bohring-Opitz Syndrome in 2014," said Laura Badmaev, Chair of the ARRE Foundation, which she founded in 2018. "It's about providing families with trusted tools and information they can use today while providing clinicians with resources that will raise the standard of care tomorrow. By bringing families and professionals together, we're laying the foundation for better care and, in the future, treatments and clinical trials."

The launch of the new website follows a period of significant growth for the ARRE Foundation, including the expansion of its Medical and Scientific Advisory Board and the appointment of Chief Scientific Officer, Dr. Karen Ho. With these developments, the ARRE Foundation continues to drive forward research and education to support the ASXL community worldwide.

Amanda Johnson
ASXL Rare Research Endowment (ARRE) Foundation
+1 607-382-5109
amanda@arrefoundation.org
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