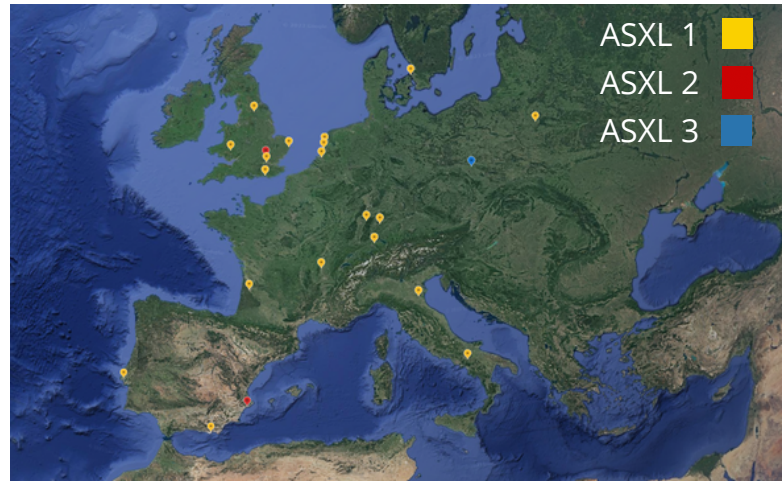
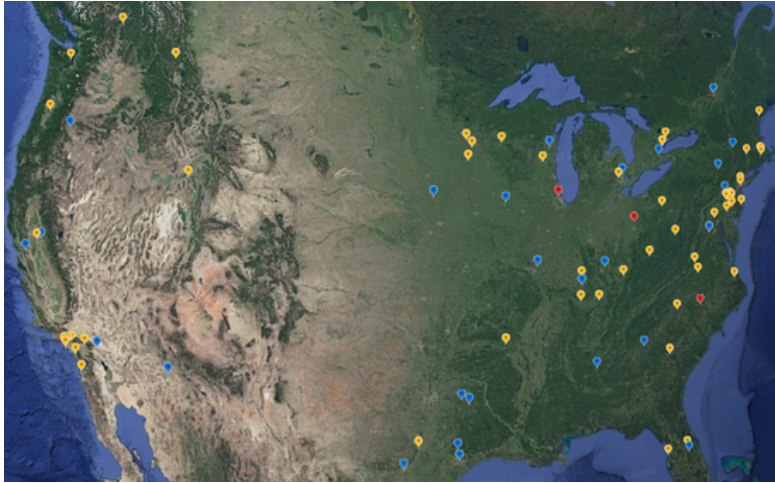


ASXL NEWSLETTER

BIANCA RUSSELL, MD
WEN HANN-TAN, MD

LOREN PENA, MD PHD
VANDANA SHASHI, MBBS MD

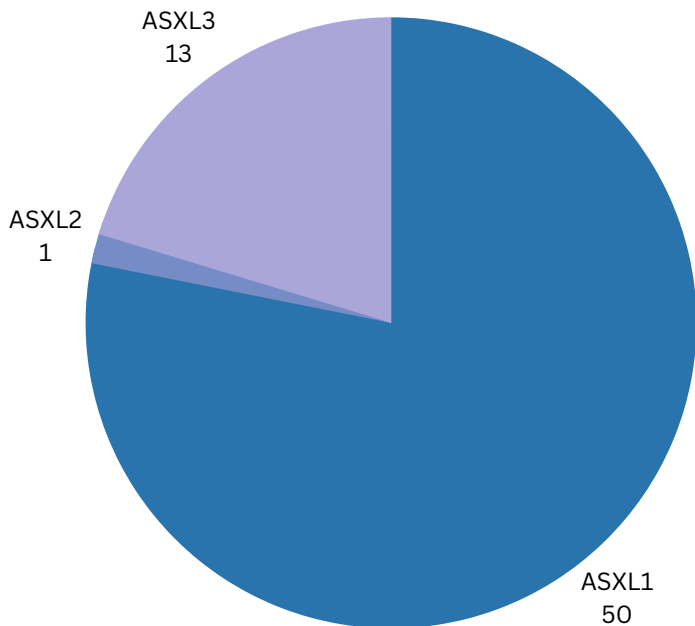
JENNIFER COHEN MD



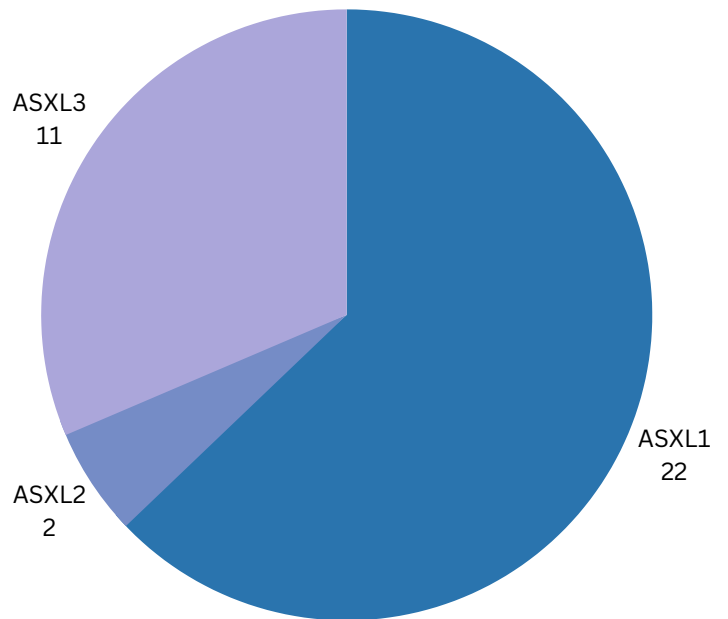
Registry Around the Globe

One hundred and thirty four families are enrolled in our Registry or are in the process of enrolling. Not pictured countries include: Australia, New Zealand, Brazil, Colombia, Chile, and China.

ASXL Registry Enrollment
(completed)



ASXL Biobank Enrollment
(completed)





July 2022 Conference

At the ARRE Foundation's ASXL Research Symposium and Family Conference at UCLA in July, we were able to make exciting enrollment progress.

We consented or began the enrollment process for **10 new participants in the ASXL Registry**, obtained **16 blood samples** for the Biobank, and consented **3 participants for our EEG study**.

We look forward to the next ASXL Family Conference in 2024!

RECENT PUBLICATIONS USING REGISTRY DATA

Awamleh et al. **DNA methylation signature associated with Bohring-Opitz syndrome: a new tool for functional classification of variants in ASXL genes.** European Journal of Human Genetics. April 2022.

Russell et al. **Clinical Findings in 39 Individuals with Bohring-Opitz Syndrome from a Global Patient Driven Registry with Implications for Tumor Surveillance and Recurrence Risk.** American Journal of Medical Genetics. February 2023.



Meet the Coordinator

Julia Sloan is the new Clinical Research Coordinator for the lead site of the ASXL Registry and Biobank at UCLA. She recently graduated from UCLA with a degree in Human Biology and Society and is hoping to become a genetic counselor. She is looking forward to working with the families and making lots of progress this year!

LOOKING TO GET MORE INVOLVED IN RESEARCH?

Research study now recruiting



Do individuals with Bohring-Opitz Syndrome have distinct brain waves?

For eligibility criteria and enrollment:
asxl-chromatin-registry@mednet.ucla.edu

We are excited to announce a study looking at the development and brain waves in children (<18 years old) with Bohring-Opitz Syndrome. We will attain video EEGs in your home to look at your loved one's brainwaves and will also conduct an interview about your child's development. Your loved one must be enrolled in the ASXL Registry to participate.

Additionally, coming soon is a new survey regarding puberty in individuals with ASXL conditions. If you would like to participate, please enroll in the Registry and provide your genetic testing so you can receive the survey! If you are already enrolled in the ASXL Registry, you will automatically receive the puberty survey when it is available.

Research study now recruiting

ASXL Puberty Survey
How does puberty impact individuals with ASXL syndromes?

Open to all ages

For eligibility criteria and enrollment:
asxl-chromatin-registry@mednet.ucla.edu



Thank you to the ARRE Foundation and the BOS Foundation for their continued financial support!

