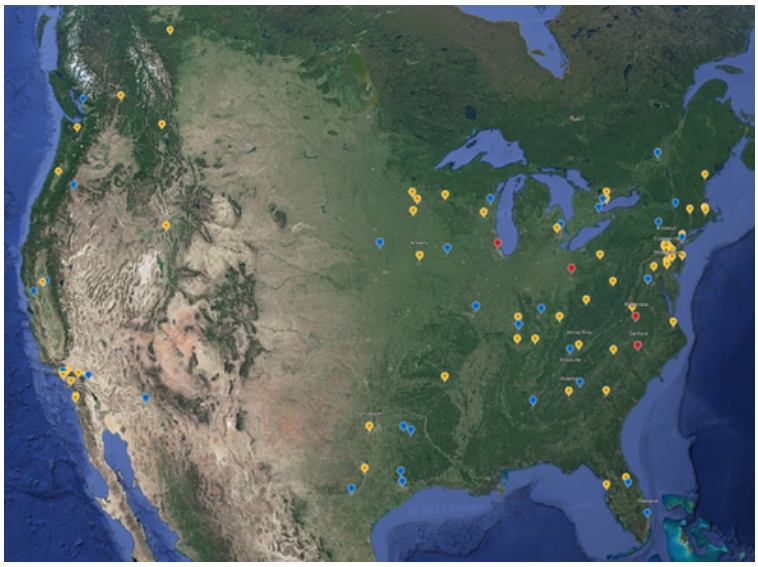
AUGUST 2023

ASXL NEWSLETTER

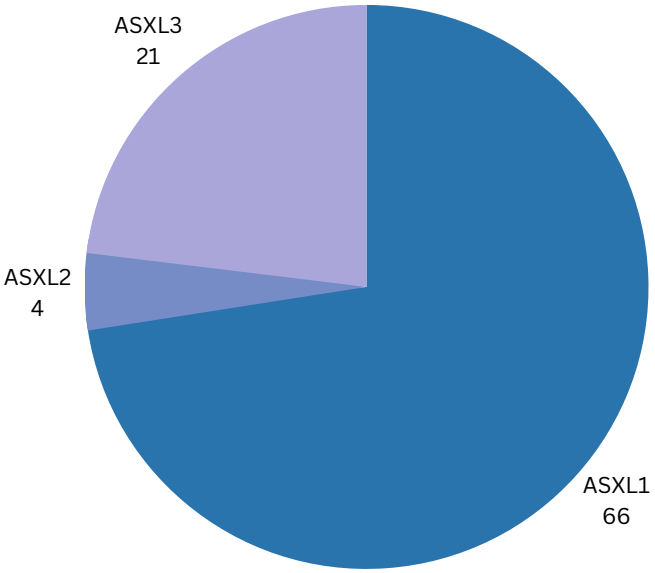
BIANCA RUSSELL, MD
LOREN PENA, MD PHD
JENNIFER COHEN MD
WEN HANN-TAN, MD
VANDANA SHASHI,MBBS MD



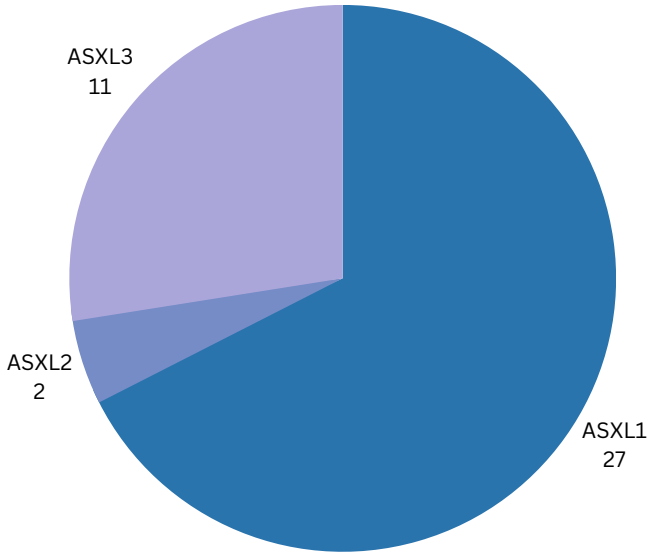
Registry Around the Globe

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

ASXL Registry Enrollment (completed)



ASXL Biobank Enrollment (completed)



Email ASXL-CHROMATIN-REGISTRY@mednet.ucla.edu to check your registry enrollment status or if you are interested in participating in the Biobank

Puberty Survey

We recently completed a puberty survey through the Registry looking at puberty in individuals with ASXL syndromes.

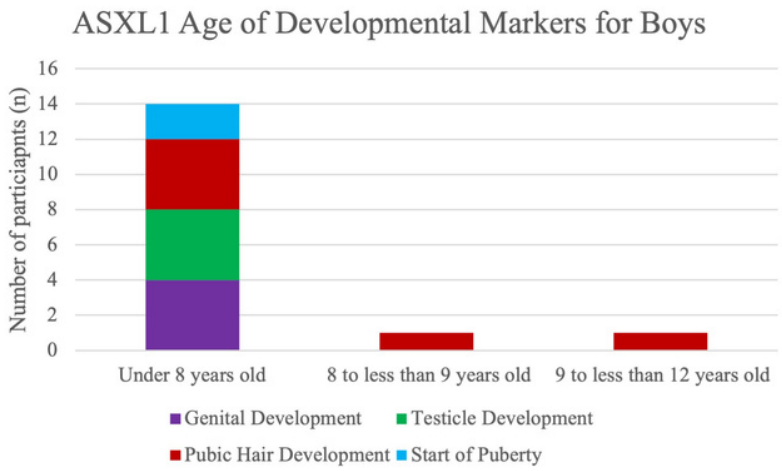
Amanda Piring
UCLA Undergraduate student

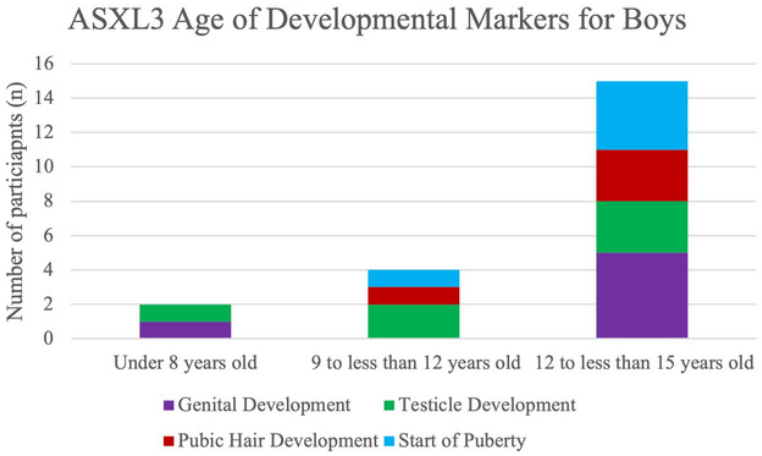
Dr. Rebecca Hicks
UCLA Pediatric Endocrinologist

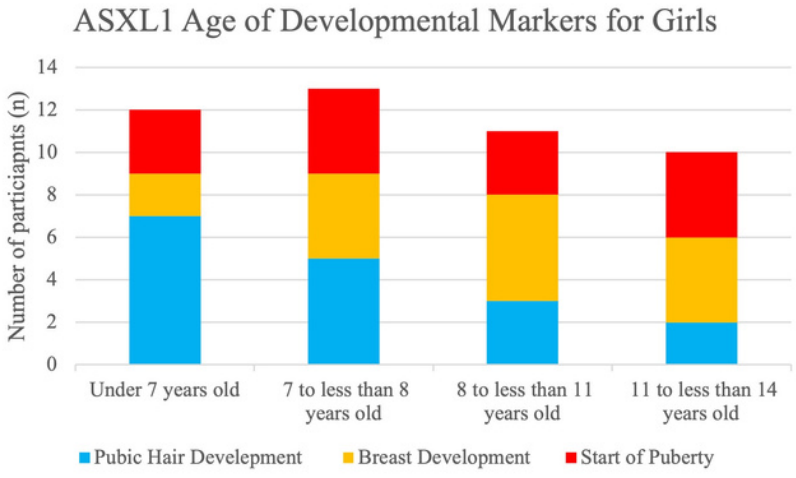
Dr. Bianca Russell
UCLA Pediatric Geneticist

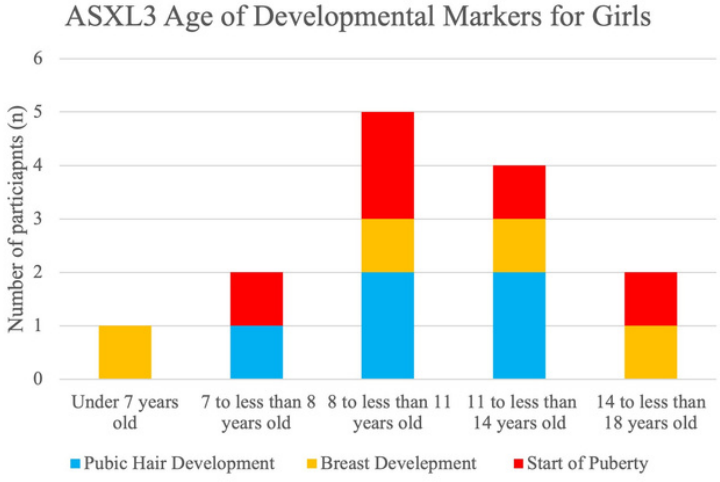
There was a total of 55 participants in the ASXL Registry that completed the survey.
ASXL1: 37
ASXL2: 2
ASXL3: 16

*The results below are preliminary and unpublished. Due to the small sample size of ASXL2 the results were not able to be quantified in a graph.









Significantly early ages of reported pubic hair development in males and females show possible precocious puberty in ASXL1. Preliminary findings indicate differences in ASXL1 and ASXL3 findings.

Clinical Research ID (CRID)

A Clinical Research ID (CRID) is a patient generated unique identifier that can be shared with researchers. This facilitates de-identified data sharing between researchers. We sent out a survey to all active participants to collect CRIDs in an effort for more collaborative research. Learn more at: https://thecrid.org/.

BOS EEG Study Updates

The BOS EEG study aims to identify an EEG signature for children with Bohring Opitz Syndrome. So far we have 27 participants, We are still looking for 3 more participants. Our preliminary data looks promising and we are excited about publishing the results of this study.

Upcoming Registry Surveys

Our next surveys will collect basic demographic information on ASXL2 and ASXL3 participants. Additionally, we plan to focus on sending out validated neurodevelopmental surveys to ASXL1/2/3 participants. These are standardized assessments that will allow for comparison with other syndromes.

RECENT PUBLICATIONS USING REGISTRY AND BIOBANK DATA

Lin, I., Wei, A., Awamleh, Z., Ning, A., Herrera, A., Singh, M., Weksberg, R., Russell, BE., Arboleda, VA. Truncating ASXL1 Mutations in Bohring-Opitz Syndrome Dysregulate Canonical and Non-Canonical Wnt-Signaling

Lin, I., Awamleh, Z., Wei, A., Russell, B., Weksberg, R., Arboleda, VA. ASXL1 mutations that cause Bohring Opitz Syndrome (BOS) or acute myeloid leukemia share epigenomic and transcriptomic signatures

ARRE
ASXL RARE RESEARCH ENDOWMENT

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

"The Best Life for All Families Living with BOS"
Bohring-Opitz Syndrome Foundation, Inc.
www.bos-foundation.org