

ASXL NEWSLETTER

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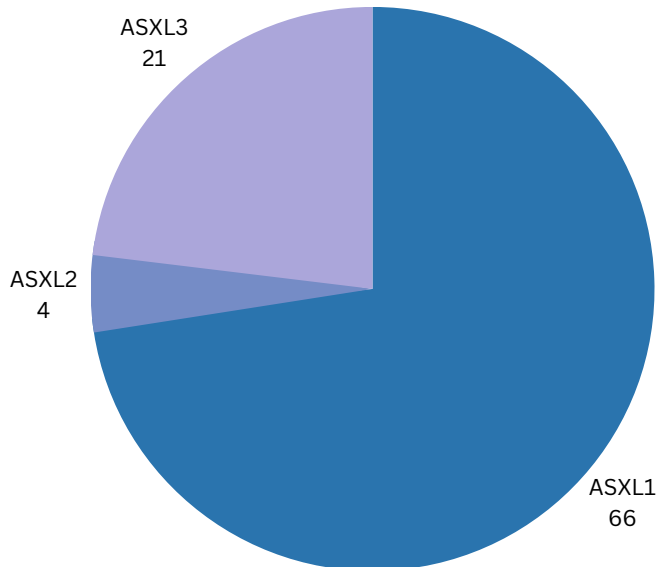
JENNIFER COHEN MD



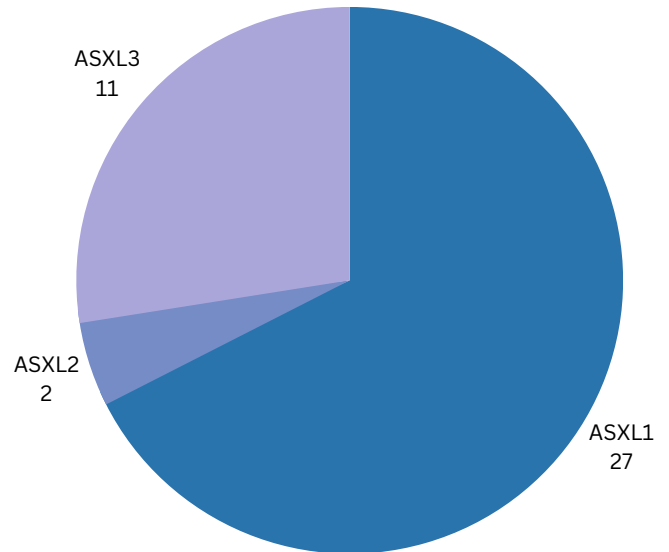
Registry Around the Globe

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.

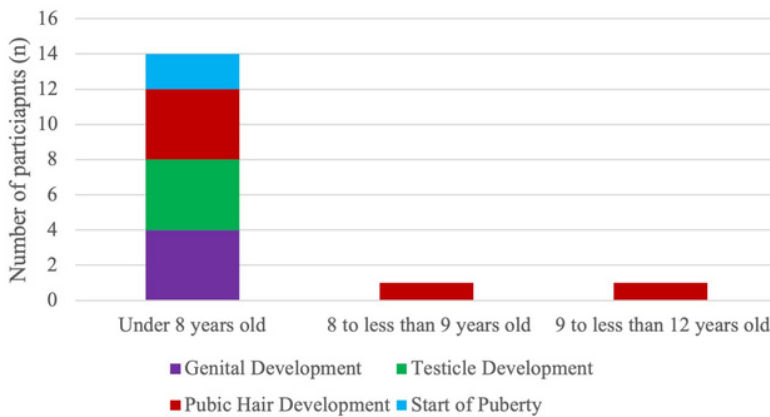
ASXL1: 37

ASXL2: 2

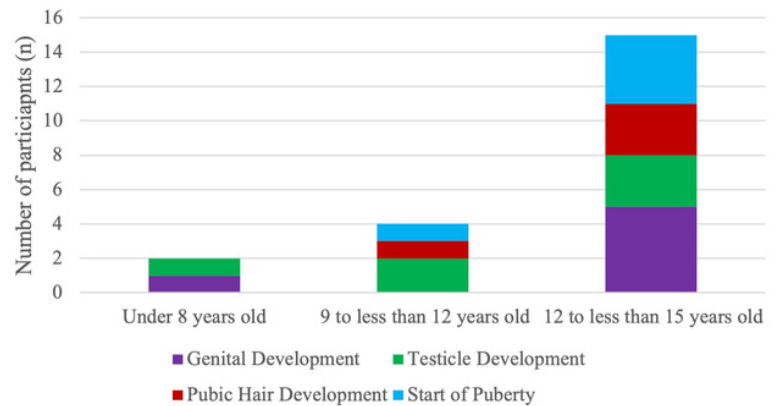
ASXL3: 16

*The results below are preliminary and unpublished.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.

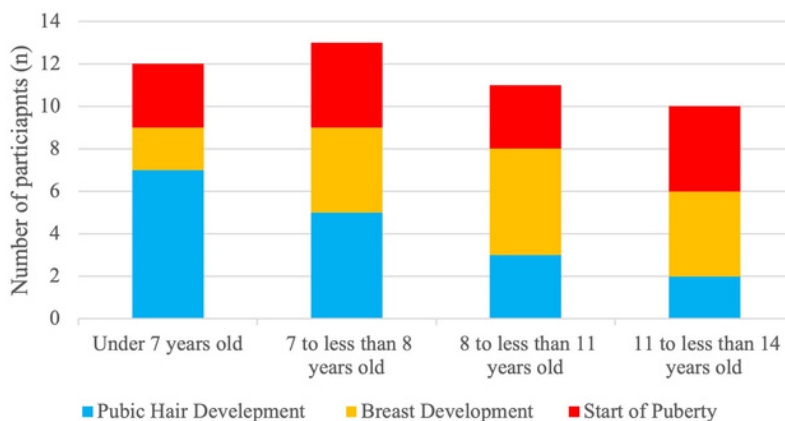
ASXL1 Age of Developmental Markers for Boys



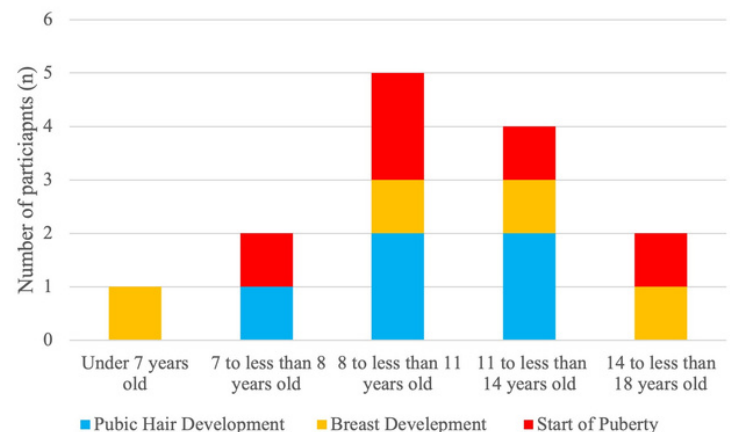
ASXL3 Age of Developmental Markers for Boys



ASXL1 Age of Developmental Markers for Girls



ASXL3 Age of Developmental Markers for Girls



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/>.

Nickname: JJ
First Name: Jon
Lastname: Smith
Sex: Male
Birth Date: 2012-06-14 YMD



A1B2 C3D4



name@email.com
name@anotheremail.com

EXAMPLE
© The CRID 2021

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

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



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

