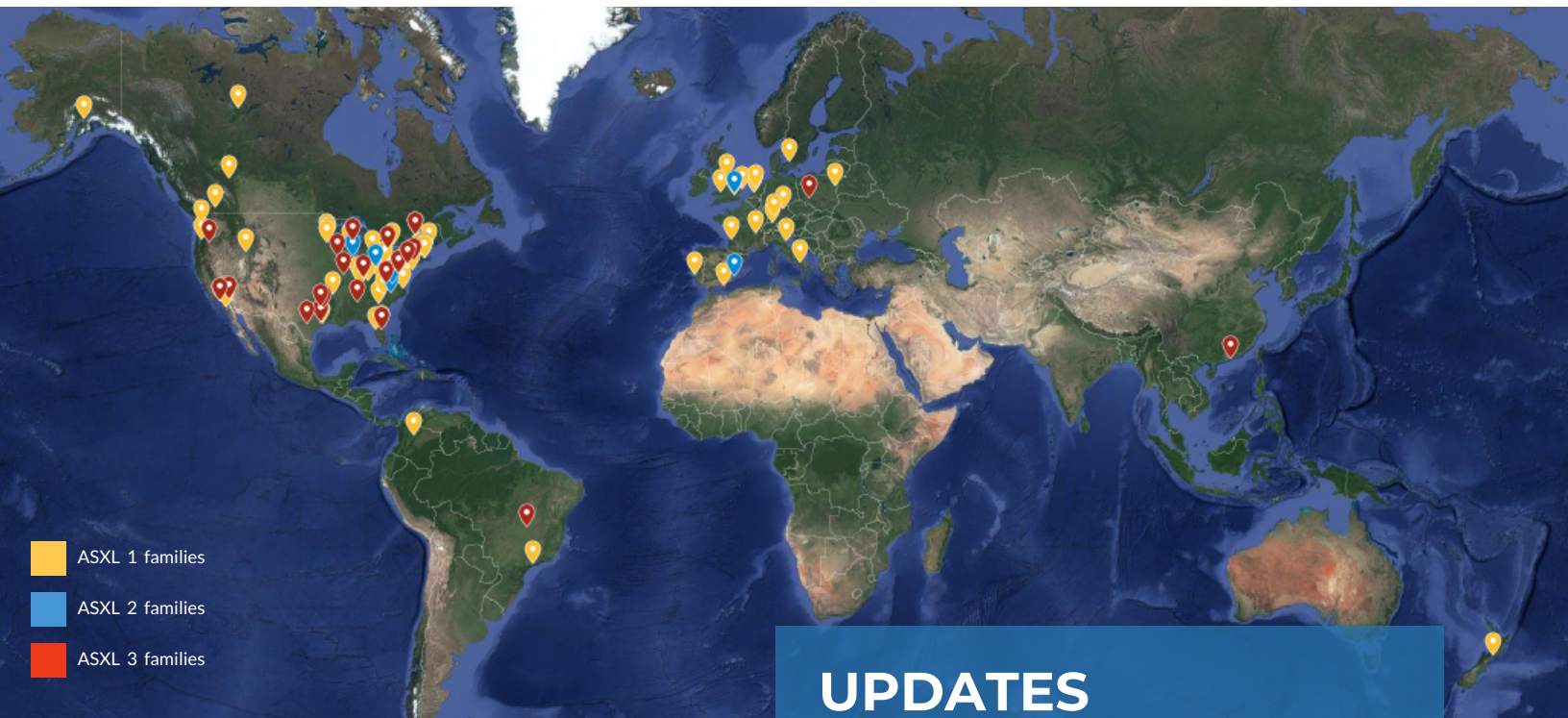


ASXL REGISTRY NEWSLETTER

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Current Focus of the Registry

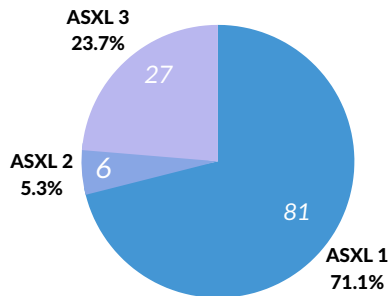
The Clinical Registry for ASXL-Related Disorders and Disorders of Chromatin Remodeling is currently focusing on updating participant data. As part of this initiative, all participants that were originally enrolled in the registry through Cincinnati Children's, are being **contacted for re-enrollment** in the current UCLA protocol through DocuSign. **Genetic testing results** are crucial to our work and essential for complete enrollment. If you do not have a copy of your child's genetic testing and would like our staff to contact your medical team to attain these results, please email us.

UPDATES

- The registry remains open for enrollment and we are actively recruiting new participants.
- ASXL biobank is now collecting samples.
- All fully enrolled ASXL1 families should have received online surveys. If you think you should have received a survey or have questions, please contact us.
- All participants will now be enrolled using DocuSign. Re-enrollment reminders have been sent to registered emails.
- We now have consents translated into **Spanish, French, Italian, and German**. We are working hard to accommodate families from all over the world and are thrilled that we are hearing from families with diverse backgrounds.

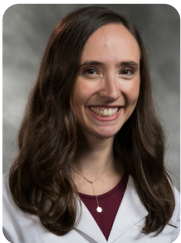
Current Registry Enrollment Numbers

As of February 2, 2022, there are 114 families from all across the globe enrolled or in the process of enrolling in our registry.



Next Steps for ASXL 3

We have formed an ASXL3 Registry Advisory Board to assist in developing surveys to meet the needs of the ASXL3 community. This survey will allow us to examine the issues that are most important to you. Following the families' interest, we will be looking into developmental assessments. If you are interested in getting involved, please email us.



Welcome Dr. Jennifer Cohen

Dr. Cohen works at Duke University and sees patients with ASXL-Related Disorders. We are excited to welcome her to the registry and clinical team.



We hope to see you at the UCLA Meyer and Renee Luskin Conference Center July 21-23, 2022 for our next ASXL scientific and family meeting.

Thursday, July 21: Scientific Programming
Friday, July 22: Family Programming
Saturday, July 23: Family Programming

We will be there to answer your questions, provide updates, and enroll new families.

More information at www.arrefoundation.org/conference.
This page will be frequently updated.

GRANT FUNDING

We have received a NIH grant to support a scientific and family meeting in the summer of 2022. This funding supports making childcare available to all families during the meeting and encourages involvement of junior investigators. Additionally, we have recently received another grant to look at the neurologic features of BOS. Please stay tuned for details!

We are grateful for the continued financial support of the BOS Foundation and ARRE.

Thank you!

