

ASXL REGISTRY NEWSLETTER

An Update:

The registry has fully transitioned to UCLA as the parent site with affiliations at Cincinnati Children's Hospital, Duke University, and Boston Children's Hospital. We are currently working on new surveys for participants. We also have a new contact email and we are currently recruiting new participants. ASXL-Chromatin-Registry@mednet.ucla.edu We are excited to introduce our research team and share next steps for our biobank and registry.

COVID-19 Update

The registry remains open as we continue to enroll families and develop new surveys. Sample collection for the biobank will resume when samples can be safely processed post-pandemic.

Meet the Team



Bianca E. Russell, M.D.

University of California - Los Angeles

Dr. Russell is a clinical geneticist at UCLA who sees patients with metabolic and genetic conditions. She has been following patients with ASXL-related disorder since 2013 and has made this the research focus of her career. She started the ASXL Registry as a resident at Cincinnati Children's and is excited about all the new growth and opportunities at UCLA.



Loren Pena, M.D., PhD

Cincinnati Children's Hospital

Dr. Pena is a geneticist at Cincinnati Children's Hospital. Along with Dr. Shashi, Dr. Pena described the first patients with ASXL2 in 2016 and has continued to stay involved with the ASXL community. She is glad to apply her knowledge and experience to partner with families in the diagnostic journey and to provide hope in the form of future treatments for patients affected with rare disorders.



Wen-Hann Tan, BMBS

Boston Children's Hospital

Dr. Tan is a clinical geneticist at Boston Children's Hospital who sees patients with a wide variety of genetic and metabolic conditions. He is also actively involved in the Bohring-Opitz syndrome community through his involvement in the ASXL Registry, ASXL Rare Research Endowment Foundation, and the Bohring-Opitz Syndrome Foundation.



Vandana Shashi, MBBS, M.D.

Duke Medical Center

As part of the efforts in ultra-rare and undiagnosed diseases, Dr. Shashi's team described Shashi-Pena syndrome due to variants in ASXL2. Further understanding of the medical and developmental findings in individuals with this condition would let medical professionals provide better management to the patients and in the long-term, work with the families towards specific treatments.

Why have the Registry and Biobank?

Rare diseases are just that; rare! It is hard for doctors to know how to best take care of patients when little is known about the condition including treatment and expected outcomes.

By joining together, families can share their experiences and knowledge through a registry. Your participation helps families get better care by collecting important information that the medical community needs. It also helps produce scientific literature and leads to new research findings.

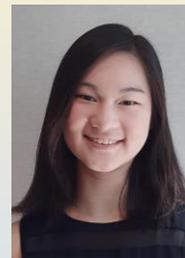
The biobank is a research tool where we collect blood and skin samples from participating families to be used for research. The biobank helps us recruit basic scientists to study these rare disorders using the deidentified data provided by you. Like the registry, it helps us work toward a better understanding of the ASXL-related conditions and possible treatments in the future.



Rebecca Kianmahd

Research Study Coordinator

Rebecca is a 4th year Psychology major & Disability Studies minor student on a pre-med track at UCLA. She is continuing as Dr. Russell's Research Coordinator for the ASXL Registry & Biobank. She looks forward to enrolling families, and answering questions. She is honored to have this opportunity.



Anna Yu

Research Study Coordinator

Anna is a 2nd year Human Biology and Society major and Applied Developmental Psychology minor student on a pre-med track at UCLA. She is a new study coordinator who will be assisting Dr. Russell with running the Registry and Biobank. She is excited to be a part of the team.

Our Locations:



For more information or to enroll in the Registry or Biobank, contact us at: ASXL-Chromatin-Registry@mednet.ucla.edu



We are grateful for the continued financial support of the BOS Foundation and ARRE. We continue to utilize the funds provided by the BOS Foundation in 2019 and to grow the program thanks to a significant gift from ARRE in early 2020.