BOS &ASXL Related Disorders Registry

Cincinnati Children's Hospital and Boston Children's Hospital

How it works

Patients and families (including angels) with BOS or mutations in ASXL genes are enrolled in our study which is approved by the hospital ethics committees. Parents complete simple questionnaires online about their child's medical history and may submit photographs as well as test results and notes from their physicians.

The information is kept on a secure server. Identified information can only be accessed by the researchers in charge of the study. De-identified content may be available to other researchers, members of the registry advisory board, and the study participants.

Please contact us with questions and to enroll in this study.

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Our Registry

We have created short questionnaires that a busy family can answer in 15 minutes. Angels will only be surveyed once but we will continue to provide updates about the Registry. Our questionnaires are organized into categories and ask questions that are important to you and researchers. The Registry Advisory Board is composed of family members, researchers and BOS foundation members who help to make sure that the registry best suits the needs of the community. With appropriate enrollment, we hope to have 3-4 questionnaires in year one. Participants will be updated about their data via a regular newsletter.

Why it matters

Rare diseases are just that; rare! It is hard for physicians to know how to best take care of patients when little is known about the disease including treatment and expected clinical outcomes. But by joining together, families can share their experiences and knowledge by being involved in clinical registries. Your participation helps patients with BOS and ASXL mutations get better care by collecting important information that the medical community needs. A well run registry also helps produce scientific literature and leads to new research findings.

Dr. K. Nicole Weaver is the Principle Investigator