AN UPDATE:

The registry is currently in an inactive state while we transition to UCLA as the host with affiliations at Cincinnati, Duke, and Boston Children’s Hospital. Please stay tuned- we will inform you when the registry is active again and are looking forward to all that is in store for us at UCLA.

Contact email remains: ASXLRegistry@cchmc.org and we will inform you when it changes.

NEXT STEPS:
- Data collection for ASXL2 and ASXL3
- Focus on understanding neurological features
- Further collaborations to create and build a biorepository (collection of biological material)

BOS REGISTRY DATA:
- 40 completed BOS survey responses
- Age range: 1-23 years with a mean age of 7 years
- 57% male and 43% female
- 4/40 (10%) of patients are deceased due to respiratory, cardiac, or GI failure
  - Ages: 2, 2, 6, and 18

Enrollment: ASXL1 (52), ASXL2 (2), ASXL3 (15)

24 US states & 13 countries; not shown: Brazil & China
**Communication:**
- 81% understand more than they speak
- Most communicate with non-verbal expressions
- 4 participants use mostly assistive devices
- 4 participants speak a few basic words
- 1 participant reports normal language

**Summary findings:**
- Late diagnosis primarily by whole exome sequencing
- No identifiable relationship between severity and location of mutation
- Normal birth weight
- More oral feeding and mobility than previously reported