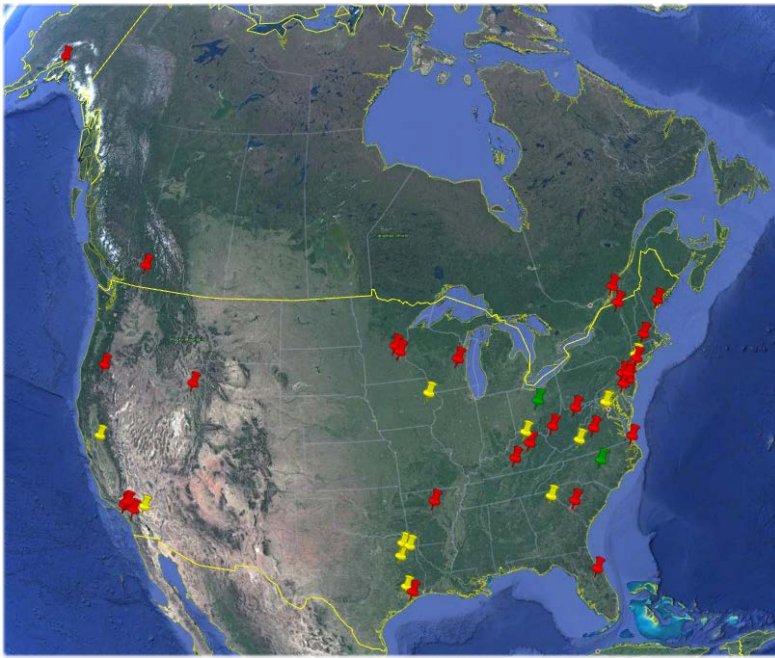
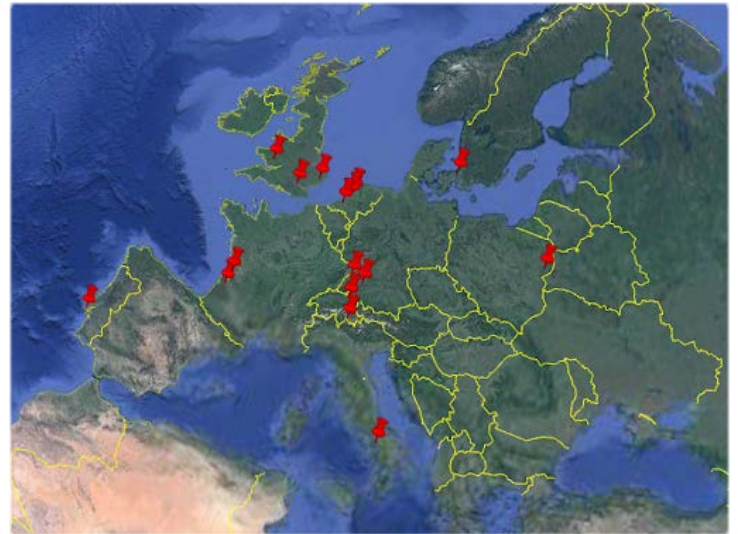


ASXL REGISTRY NEWSLETTER

Bianca Russell, MD
Wen-Hann Tan, BMBS
Loren Pena, MD



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



24 US states & 13 countries; not shown: Brazil & China

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

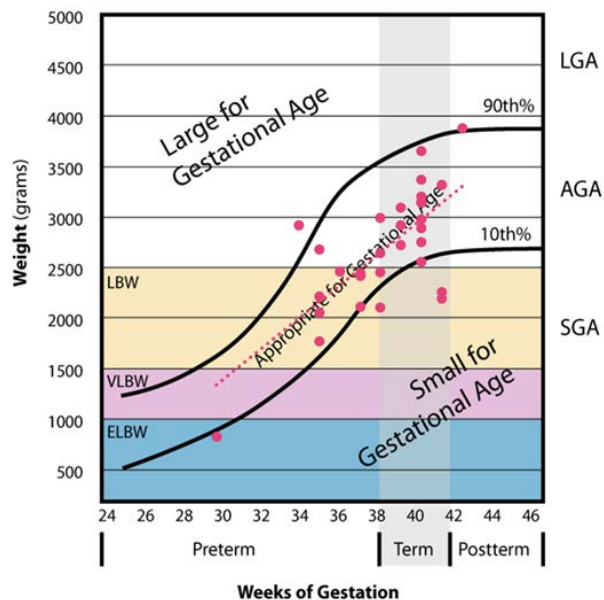
AN UPDATE:

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.

HIGHLIGHTED REGISTRY FINDINGS

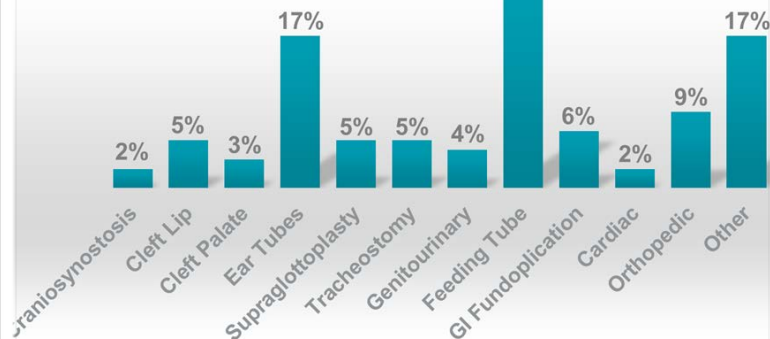
Birth Weights



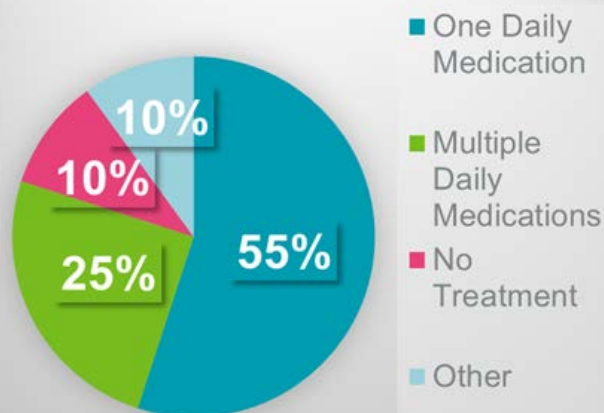
BOS MOBILITY



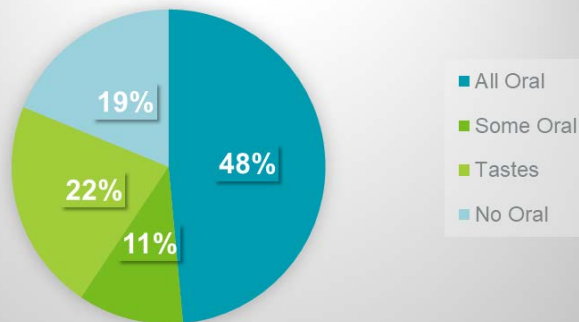
Surgeries



BOS Seizure Management



BOS Feeding Habits



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