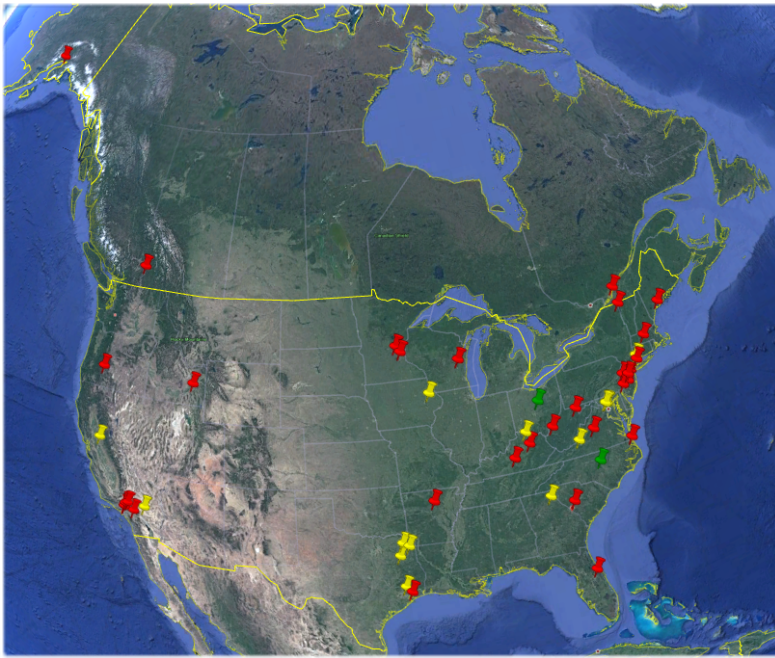
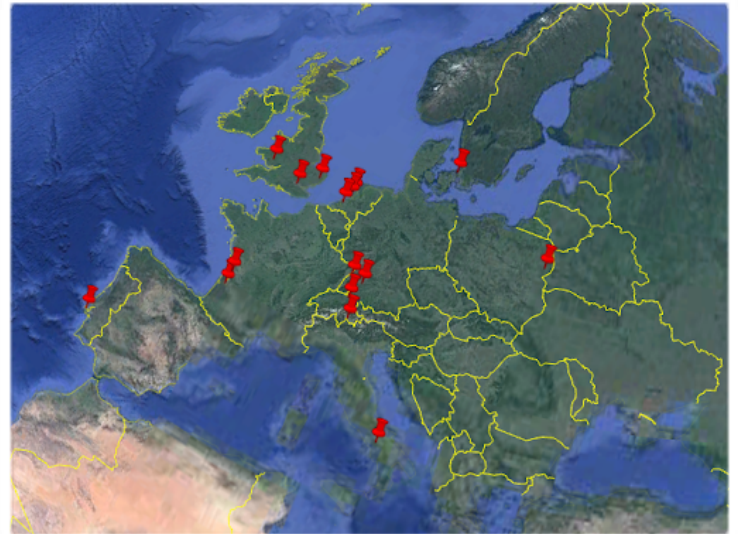


# ASXL REGISTRY NEWSLETTER

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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

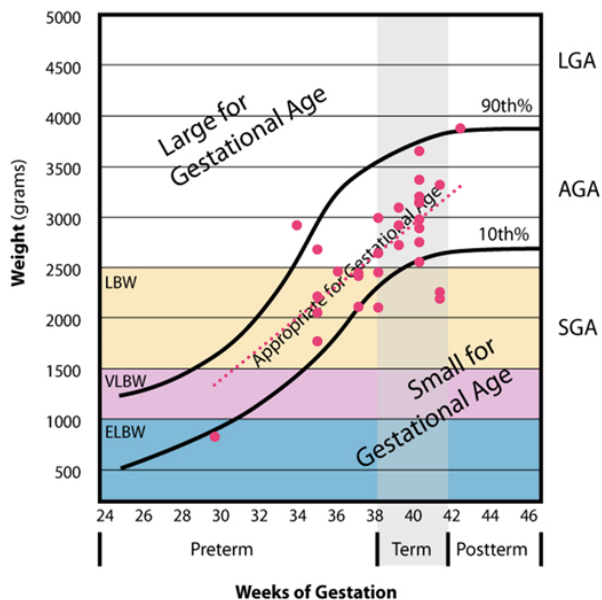
## AN UPDATE:

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.

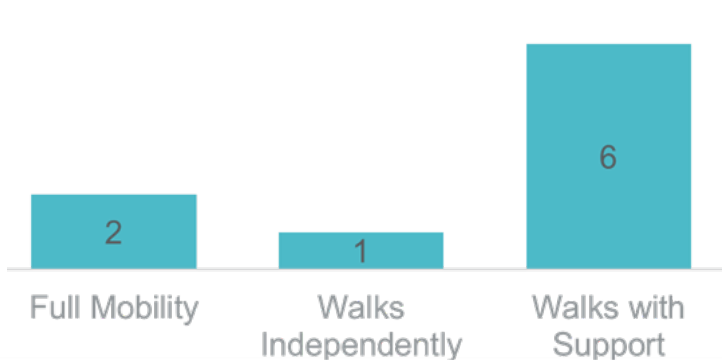
Contact email remains: [ASXLRegistry@cchmc.org](mailto:ASXLRegistry@cchmc.org) and we will inform you when it changes.

# HIGHLIGHTED REGISTRY FINDINGS

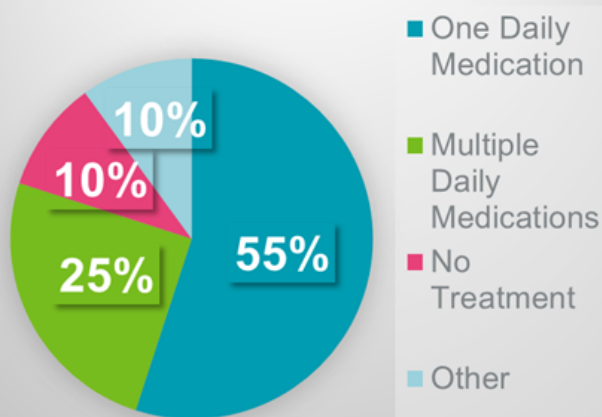
Birth Weights



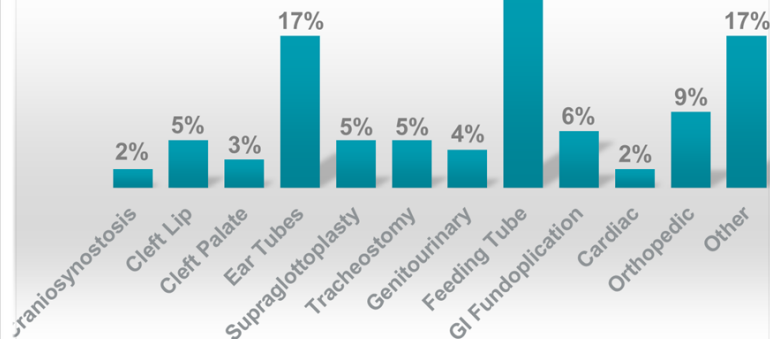
BOS MOBILITY



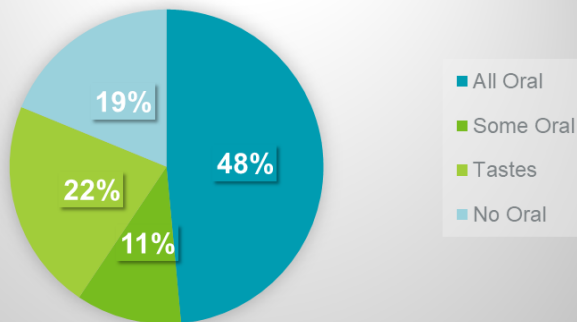
BOS Seizure Management



Surgeries



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