Simons Searchlight Registry Update for ASXL3 (Bainbridge-Ropers)

December 2021

Participant Age at Most Recent Interview

![Bar chart showing age distribution of participants.]

Developmental & Behavioral Diagnoses

- Intellectual Disability/Development Delay: 38
- Language Impairment: 20
- Autism: 32
- ADHD: 2
- Anxiety: 3
- OCD: 4

Notes:
- Graphs show counts of individuals in each category. Individual participants may appear in more than one category if they report multiple conditions.
- Graphs show data as of the last medical history interview completed on participants who have a pathogenic or likely pathogenic variant.

Neurological Problems

- Tic Disorder: 6
- Movement Disorder: 5
- Small Head Size: 13
- Large Head Size: 2
- Low Muscle Tone: 9
- High Muscle Tone: 6
- Clumsy: 4
- Cerebral Palsy: 1
- Cortical blindness: 11

Gastrointestinal Problems

- GERD: 28
- Diarrhea: 14
- Constipation: 3
- Celiac Disease: 1

Notes:
- Data in the four graphs are from the medical history phone interviews collected in Simons Searchlight on 44 participants with ASXL3.

How to Participate

The information in this report is made possible by the active participation of the ASXL3 community! Progress for individuals in your community with a ASXL3 variant is shown below -- log in to your simons-searchlight.org dashboard today to check for new surveys and tasks. Your data could hold the clues geneticists need to find answers.

**STEP 1**
Sign up online.

**STEP 2**
Provide your genetic lab report.

**STEP 3**
Share your important medical history.

**STEP 4**
Fill out surveys.

**STEP 5**
Provide a blood sample if you are interested.

**STEP 6**
Update us every year.

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Log in to see next steps.
Information Spotlight: **Deeper Dive into Sleep**

**The Simons Searchlight Sleep Supplement** is a custom survey. We worked with members of the Simons Searchlight community and sleep researchers to design these questions. It asks caregivers about children's sleep habits and the impact of health issues on sleep behaviors. The survey looks at children between ages 2 and 18 years of age. 17 children with ASXL3 are represented.

### Getting Enough Sleep

- **Usually (64.71%)**
- **Sometimes (5.88%)**
- **Rarely (17.65%)**
- **Never (11.76%)**

### Medical issues related to sleep problems in children with ASXL3*

- 1. Seizures
- 1. Mood disorder
- 1. Anxiety
- 2. Movement disorder
- 2. Itchy skin
- 3. Breathing problems
- 3. Sleep apnea/snoring
- 2. Pain
- 3. GI symptoms

Children with ASXL3 fall asleep in **69 minutes** on average.

### Best Ways to Help Sleep: Community Recommendations*

- 1. Withholding naps
- 1. Parent lying in their bed until they fall asleep
- 2. Bedtime routine or ritual
- 2. Sound or white noise machine
- 8. Medication

* Caregivers were able to select more than one answer

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**Thank you** for contributing knowledge to Simons Searchlight!