The report shows data collected in Simons Searchlight from ASXL3 participants. Data from other published research are not included.

Data presented in this report come primarily from the medical history phone call. The data are current and may reflect information not yet available in an official researcher data release.

Graphs show counts of individuals in each category. Individual participants may appear in more than one category if they report multiple conditions.

This report may look different from previous reports because we have streamlined our process and updated the variants.

Variants

Graphs include individuals with variants classified as pathogenic or likely pathogenic unless otherwise noted. Variants of uncertain significance (VUS) and less typical variants are not included.

We reassess variants classified as VUS annually.

Participation & Registration

Learn more at SimonsSearchlight.org

STEP 1
Sign up online.

STEP 2
Provide your clinical lab report.

STEP 3
Schedule medical history phone call with a genetic counselor.

STEP 4
Fill out surveys.

STEP 5
Provide a blood sample if you are interested.

STEP 6
We follow up with you over time.

ASXL3 Progress
Includes families registered with all variant types

* Most recent data release - February 2021
**Developmental & Behavioral Diagnoses**

- **Intellectual Disability/Developmental Delay**: 34
- **Autism**: 18
- **Language Impairment**: 15
- **ADHD**: 2
- **OCD**: 3
- **Anxiety**: 3

**Individual participants:** 41

**Gastrointestinal Problems**

- **GERD**: 26
- **Diarrhea**: 3
- **Celiac Disease**: 13
- **Constipation**: 1

**Individual participants:** 41

**Neurological Problems**

- **Clumsy**: 6
- **Low Muscle Tone**: 9
- **High Muscle Tone**: 2
- **Large Head Size**: 12
- **Small Head Size**: 1
- **Cyst in the Spinal Cord**: 5
- **Movement Disorder**: 6
- **Tic Disorder**: 3
- **Cerebral Palsy**: 1
- **Cortical Blindness**: 37

**Individual participants:** 41

**Seizures**

- **Atonic Drop Attack**: 1
- **Petit Mal**: 5
- **Grand Mal**: 4

**Individual participants:** 41