The report shows data collected in Simons Searchlight from SLC6A1 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.

**About this Report**

**Ages in Years**

Individual participants: 37

**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.

**SLC6A1 Progress**

Includes families registered with all variant types

- Registered Families: 104
- Person with Genetic Change Consented: 98
- Lab Report Approved: 74
- Medical History Phone Call: 58
- Data Released to Researchers*: 60

* Most recent data release - February 2021
**Developmental & Behavioral Diagnoses**
Individual participants: 37

- Intellectual Disability/Development Delay: 27
- Language Impairment: 12
- Autism: 16
- ADHD: 6
- OCD: 2
- Anxiety: 3

**Seizures**
Individual participants: 37

- Myoclonic: 19
- Simple Partial: 4
- Complex Partial: 1
- Atonic Drop Attack: 1
- Infantile Spasm: 1
- Petit Mal: 1
- Grand Mal: 4

**Seizure Medications Reported Most Effective**
Individual participants: 31

- Valproic Acid: 12

**Neurological Problems**
Individual participants: 37

- Clumsy: 9
- Low Muscle Tone: 2
- High Muscle Tone: 2
- Large Head Size: 2
- Small Head Size: 2
- Movement Disorder: 15
- Tic Disorder: 2