The report shows data collected in Simons Searchlight from 1q21.1 duplication 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.

Participants

Graphs include data and information strictly from the online phase of our study.

About this Report

Simons Searchlight Registry Update
1q21.1 duplication
March 2021

Ages in Years
Individual participants: 48

ծ0-4 13 10 15
5-9 7
10-14
15-19 3
20+

1q21.1 duplication Progress
Includes families registered with all variant types

- Registered Families: 81
- Person with Genetic Change Consented: 91
- Lab Reports Approved: 66
- Medical History Phone Call: 50

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.
### Developmental & Behavioral Diagnoses

- **Individual participants:** 48

- Intellectual Disability/Developmental Delay: 15
- Autism: 13
- Language Impairment: 11
- ADHD: 9
- OCD: 1
- Anxiety: 1

### Neurological Problems

- **Individual participants:** 48

- Clumsy: 9
- Low Muscle Tone: 16
- High Muscle Tone: 1
- Large Head Size: 19
- Small Head Size: 1
- Cyst in the Spinal Cord: 1
- Movement Disorder: 3
- Tic Disorder: 4

### Gastrointestinal Problems

- **Individual participants:** 48

- GERD: 13
- Diarrhea: 12
- Constipation: 2
- Inflammatory Bowel Disease: 1
- Gastric Ulcer: 3
- Other: 2

### Seizures

- **Individual participants:** 48

- Simple Partial: 2
- Complex Partial: 2
- Petit Mal: 1
- Grand Mal: 2