The report shows data collected in Simons Searchlight from CHAMP1 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.

Ages in Years

Individual participants: 22

CHAMP1 Progress

Includes families registered with all variant types

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

- Intellectual Disability/Development Delay: 14
- Autism: 5
- Language Impairment: 9
- ADHD: 1
- Anxiety: 1

Gastrointestinal Problems

- GERD: 14
- Diarrhea: 2
- Constipation: 12

Neurological Problems

- Clumsy: 2
- Low Muscle Tone: 21
- High Muscle Tone: 1
- Small Head Size: 8
- Movement Disorder: 2
- Tic Disorder: 1
- Cerebral Palsy: 1

Seizures

- Simple Partial: 1
- Atonic Drop Attack: 1
- Infantile Spasm: 1
- Petit Mal: 2
- Grand Mal: 2

Individual participants: 22