Simons Searchlight Registry Update

**CTNNB1**
March 2021

About this Report

The report shows data collected in Simons Searchlight from CTNNB1 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: 31

**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.

CTNNB1 Progress
Includes families registered with all variant types

* Most recent data release - February 2021
Developmental & Behavioral Diagnoses
Individual participants: 31

- Intellectual Disability/Development Delay: 22
- Autism: 4
- Language Impairment: 13
- ADHD: 3
- OCD: 1
- Anxiety: 1

Neurological Problems
Individual participants: 31

- Clumsy: 13
- Low Muscle Tone: 28
- High Muscle Tone: 18
- Small Head Size: 24
- Movement Disorder: 11
- Tic Disorder: 1
- Cerebral Palsy: 10

Gastrointestinal Problems
Individual participants: 31

- GERD: 7
- Diarrhea: 2
- Constipation: 5