About this Report

The report shows data collected in Simons Searchlight from CSNK2A1 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.

CSNK2A1 Progress
Includes families registered with all variant types

<table>
<thead>
<tr>
<th>Category</th>
<th>Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>Registered Families</td>
<td>59</td>
</tr>
<tr>
<td>Person with Genetic Change Consented</td>
<td>52</td>
</tr>
<tr>
<td>Lab Reports Approved</td>
<td>39</td>
</tr>
<tr>
<td>Medical History Phone Call</td>
<td>24</td>
</tr>
<tr>
<td>Data Released to Researchers *</td>
<td>28</td>
</tr>
</tbody>
</table>

* Most recent data release - February 2021
Developmental & Behavioral Diagnoses
Individual participants: 24

- Intellectual Disability/Developmental Delay: 19
- Autism: 6
- Language Impairment: 14
- ADHD: 5
- Anxiety: 3

Neurological Problems
Individual participants: 24

- Clumsy: 6
- Low Muscle Tone: 19
- Large Head Size: 1
- Small Head Size: 6
- Movement Disorder: 6
- Tic Disorder: 1
- Cerebral Palsy: 2
- Cortical Blindness: 1

Gastrointestinal Problems
Individual participants: 24

- GERD: 8
- Diarrhea: 2
- Constipation: 13

Seizures
Individual participants: 24

- Myoclonic: 1
- Simple Partial: 1
- Complex Partial: 1
- Infantile Spasm: 1
- Petit Mal: 1
- Grand Mal: 5