Most of the information in this report comes from *DYRK1A* families completing the Simons Searchlight medical history phone call. Families also complete follow-up phone calls every year so we can track progress over time. We are reporting on a total of 17 participants.

The information presented in this report is a summary of data contributed by *DYRK1A* families with genetic changes classified as “pathogenic” or “likely pathogenic”. To present an accurate picture of conditions related to *DYRK1A*, genetic changes classified as "variants of uncertain/unknown significance" were not included here.

The data included in this report represents current information in the registry, prior to official data cleaning and release. It is not intended for publication.
Participation

After registering for a research account, what does it take for a participant’s data to be released to researchers?

1. Sign up online. SimonsSearchlight.org*
2. Provide your genetic lab report
3. Discuss your medical history with a genetic counselor
4. Unite with researchers and other families

* If you had previously signed up for Simons VIP, click the link in your email to link your old account with your new Simons Searchlight account. No need to re-register!

Where are DYRK1A families in this process?
Finish any missing steps to contribute your family’s data!

<table>
<thead>
<tr>
<th>Step</th>
<th>Number of Families</th>
</tr>
</thead>
<tbody>
<tr>
<td>Families Registered</td>
<td>32</td>
</tr>
<tr>
<td>Parent &amp; Individual Consented</td>
<td>25</td>
</tr>
<tr>
<td>Lab Reports Submitted</td>
<td>24</td>
</tr>
<tr>
<td>Medical History Phone Call</td>
<td>17</td>
</tr>
<tr>
<td>Data Released to Researchers</td>
<td>12</td>
</tr>
</tbody>
</table>
**Total Males and Females**

Among the 17 participants included, there are 8 males and 9 females who have the **DYRK1A** genetic change.

**Age Range**

The average age is 8 years old. Ages range from 2 years to 22 years, and 16 of the 17 participants are under 18.

**Developmental & Behavioral Diagnoses**

Simons Searchlight completed diagnostic history interviews with 17 participants.

- Reports of language disorder (without intellectual disability): 15
- Reports of anxiety: 8
- Reports of developmental delay or intellectual disability: 6
- Have been diagnosed with attention-deficit hyperactive disorder: 2
- Have been diagnosed with autism spectrum disorder: 2
DYRK1A REGISTRY SNAPSHOT

**Most Commonly Reported Conditions**

DYRK1A families reported a variety of associated medical conditions. The most common medical conditions were reported in the brain, eyes, and digestive system. The specific medical conditions that were reported are displayed in the graphs on this page.

**Neurological Conditions**

Note: Individual participants may have reported multiple conditions

![Graph showing neurological conditions]

- Large Head Size: 1
- Tic Disorder: 1
- Clumsy: 2
- Problems with Movement*: 4
- High Muscle Tone: 7
- Low Muscle Tone: 5
- Small Head Size: 15

*Including ataxia

**Gastrointestinal Conditions**

Note: Individual participants may have reported multiple conditions

![Graph showing gastrointestinal conditions]

- Constipation: 6
- Diarrhea: 5
- Reflux: 2
- Other (not specified): 2

**Vision Problems**

Note: Individual participants may have reported multiple conditions

- Crossed Eyes: 6
- Nearsighted: 5
- Optic Nerve Change: 5
- Repetitive Eye Movement: 2
- Farsighted: 2
- Astigmatism: 2
- Lazy Eye: 1
- Other: 1

Note: Individual participants may have reported multiple conditions
**Other Conditions Reported**

Some other conditions reported for this group of 17 participants are shown in the graphic below as total number of conditions reported. Individual participants may have reported multiple conditions across multiple systems.

*Heart issues include: congenital heart disease, atrial septal defect, murmur, palpitation.*

**Types of Seizures**

Note: Individual participants may have reported multiple conditions.

- **Tonic**: 1
- **Drop**: 1
- **Simple Partial**: 2
- **Petit Mal**: 4
- **Complex Partial**: 4
- **Grand Mal**: 5
- **Febrile**: 12
Medication Use
Simons Searchlight collected medication use from 16 participants. Below we list the number of participants taking each medication type. Participants may be taking multiple medication types.

Medication Reported to Work Best for Seizures*

*7 individuals reported improvement from medication, and 7 others no longer take medication.