About this Report

Most of the information in this report comes from DYRK1A families completing the Simons VIP 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 7 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.
Participation

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

1. Add children to account
2. Complete consents for all family members
3. Complete Simons VIP Connect Questionnaire
4. Upload or email your genetic lab report
5. Genetic Counselor phone interview
6. Complete online research surveys
7. Continue participating yearly

If you have any questions about enrollment, please contact a Simons VIP Connect Coordinator. By phone: 855.329.5638 or email: coordinator@simonsvipconnect.org

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

<table>
<thead>
<tr>
<th>Category</th>
<th>Number of Families</th>
</tr>
</thead>
<tbody>
<tr>
<td>Families Registered</td>
<td>19</td>
</tr>
<tr>
<td>Parent &amp; Individual Consented</td>
<td>15</td>
</tr>
<tr>
<td>Lab Reports Submitted</td>
<td>12</td>
</tr>
<tr>
<td>Medical History Phone Call</td>
<td>7</td>
</tr>
<tr>
<td>Data Released to Researchers</td>
<td>5</td>
</tr>
</tbody>
</table>

Number of Families
**Total Males and Females**

Among the 16 participants for whom we have age and gender information, there are 7 males and 9 females who have the DYRK1A genetic change.

**Age Range**

The average age is 11 years old. Ages range from 3 years to 24 years, and 14 of the 16 participants are under 18.

**Developmental & Behavioral Diagnoses**

Simons VIP completed diagnostic history interviews with 7 participants.

- 1 participant has been diagnosed with autism spectrum disorder
- 6 participants have reports of language disorder or delay
- 5 participants have reports of intellectual disability or developmental delay
- 4 participants have been diagnosed with attention-deficit/hyperactivity disorder
Most Commonly Reported Conditions

DYRK1A families reported a variety of associated medical conditions. The graphic below shows the most common medical conditions reported by the 7 participants for whom we have medical history data.

- **Hypertonia**: 6 reports of hypertension
- **Microcephaly**: 6 reports of microcephaly
- **Eczema**: 4 reports of eczema
- **Heart Conditions**: 3 reports of heart conditions*
- **Seizures**: 5 reports of seizures
- **Bone Conditions**: 2 reports of bone conditions (scoliosis & club foot)
- **Eye Conditions**: 4 reports of eye conditions, including strabismus (3) and optic nerve change (2)

* heart conditions include: atrial septal defect, pulmonic stenosis, & heart murmur
THANK YOU

Source:
SimonsVIPConnect.org
sfari.org/resources/sfari-base