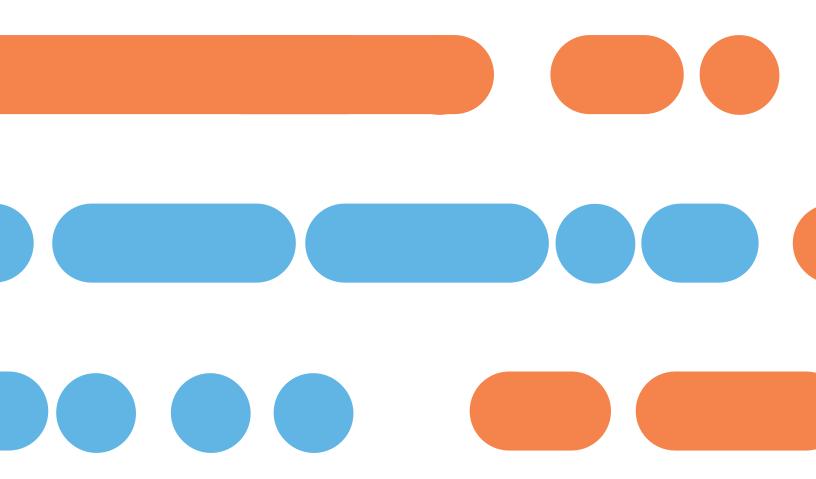
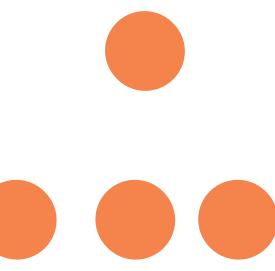


# DYRK1A Registry Snapshot





### **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**

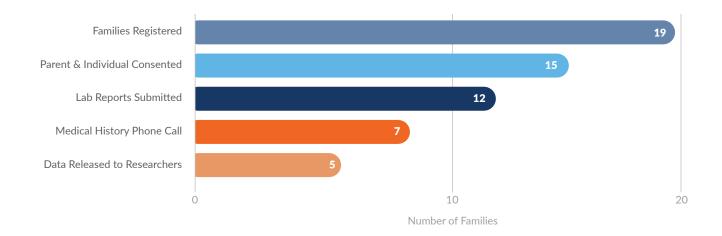
After registering for a research account, what does it take for a participant's data to be released to researchers?



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!



### **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**

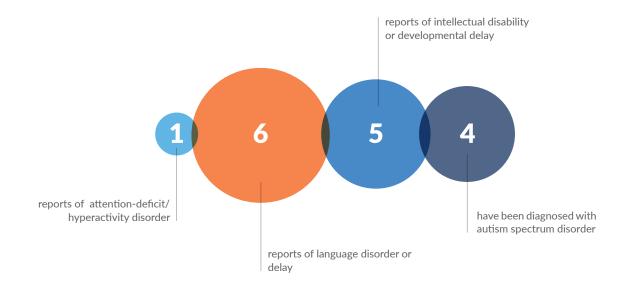
The average age is 11 years old. Ages range from 3 years to 24 years, and 14 of the 16 participants are under 18.





### **Developmental & Behavioral Diagnoses**

Simons VIP completed diagnostic history interviews with 7 participants.



# **Reported Conditions**

### **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.

Seizures

5 reports of seizures

Hypertonia

6 reports of hypertonia

Eczema
4 reports of eczema

Heart
Conditions
3 reports of heart

### **6** reports of microcephaly

**Microcephaly** 

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

Bone Conditions
2 reports of bone



# **THANK YOU**







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

