SIMONS
SEARCHLIGHT
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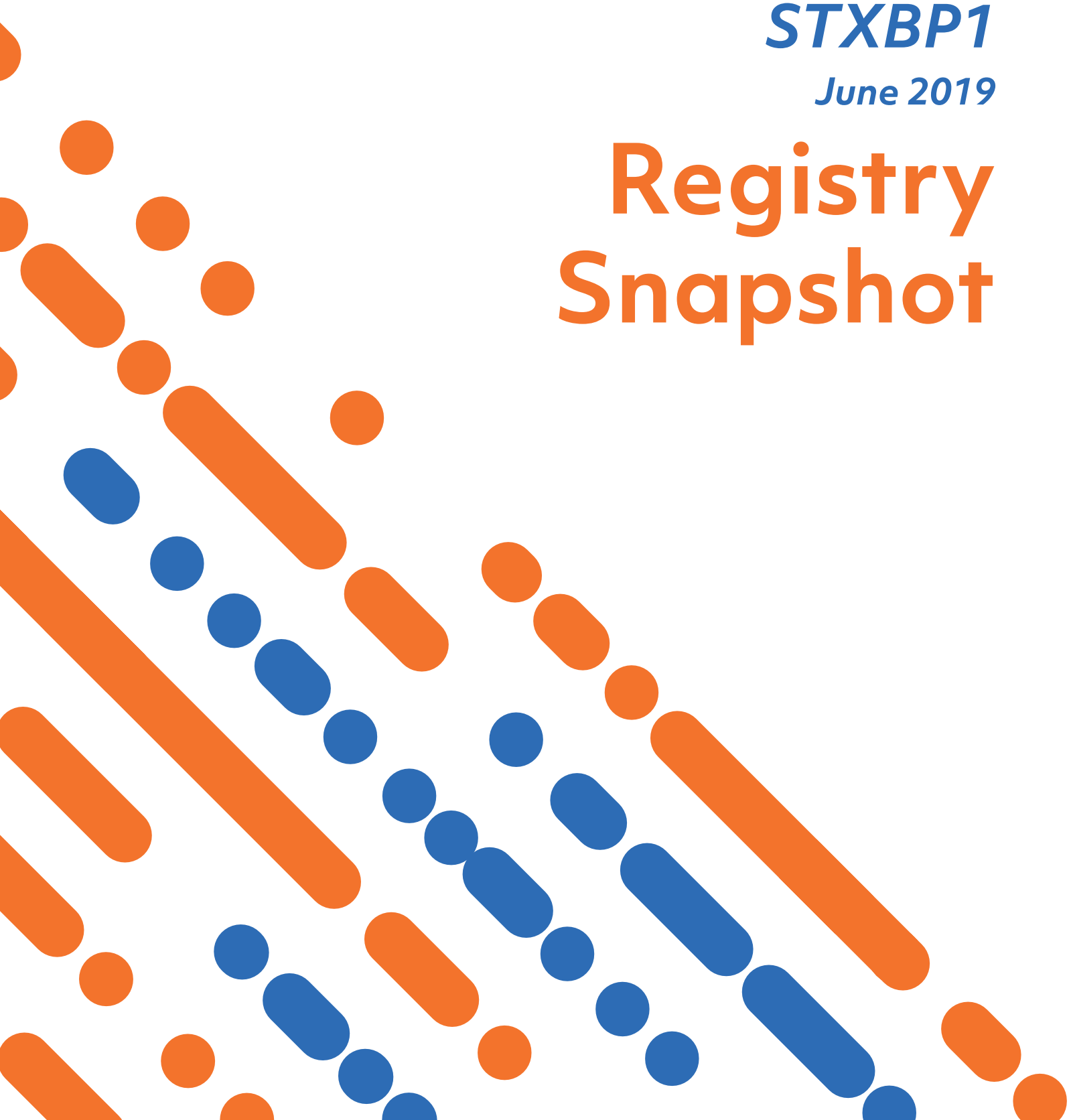
STXBP1

June 2019

Registry Snapshot



About this Report

Most of the information in this report comes from *STXBP1* 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 39 participants.

The information presented in this report is a summary of data contributed by *STXBP1* families with genetic changes classified as "pathogenic" or "likely pathogenic". To present an accurate picture of conditions related to *STXBP1*, 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

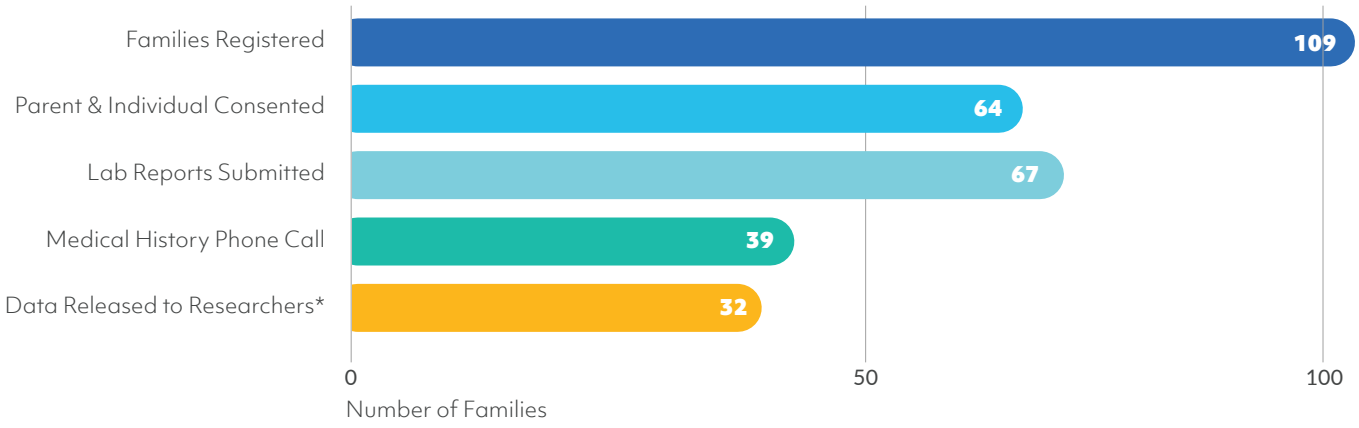
3. Discuss your medical history with a genetic counselor

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 *STXBP1* families in this process?

Finish any missing steps to contribute your family's data!



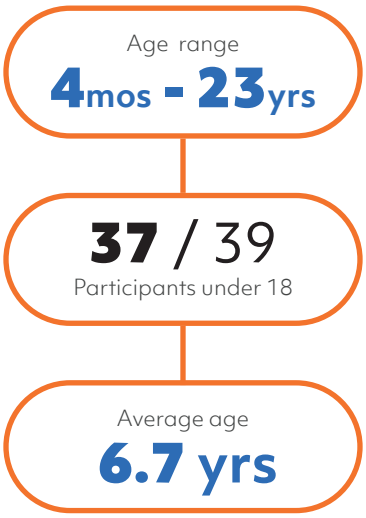
* Additional data to be release July 2019

Total Males and Females

Among the 39 participants included, there are 16 males and 23 females who have the *STXBP1* genetic change.

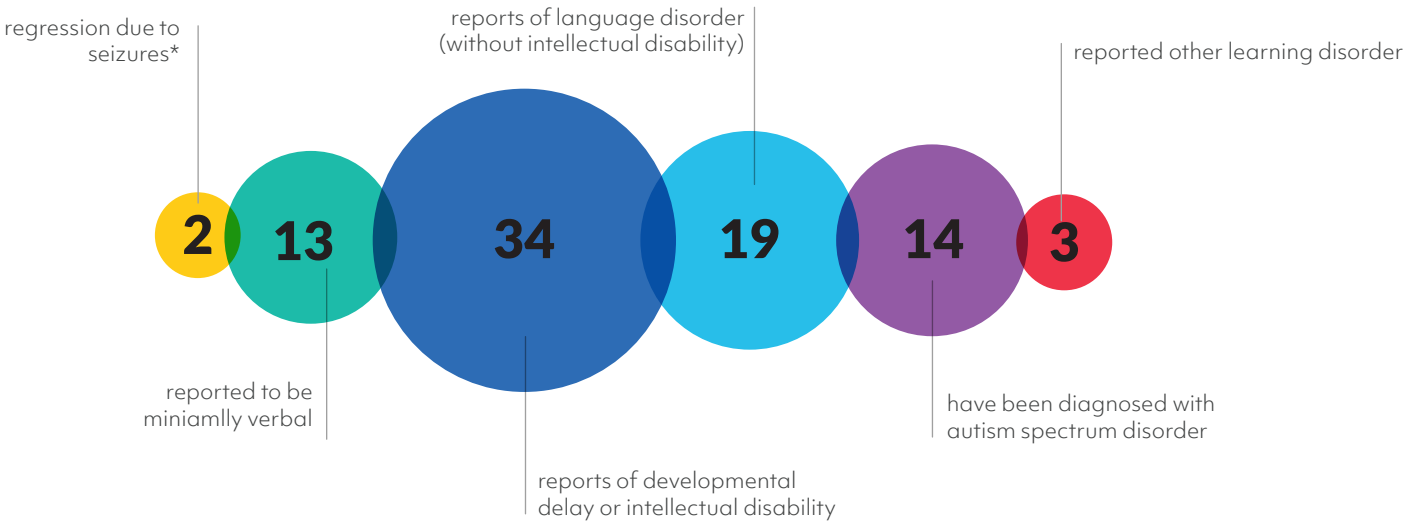
Age Range

The average age is 6.7 years old. Ages range from 4 months to 23 years, and 37 of the 39 participants are under 18.



Developmental & Behavioral Diagnoses

Simons Searchlight completed diagnostic history interviews with 38 individuals.



* Regression and sensory processing may be underestimates; more families may report these issues over time

** Other conditions reported once: ADHD, sensory processing disorder without autism, pica

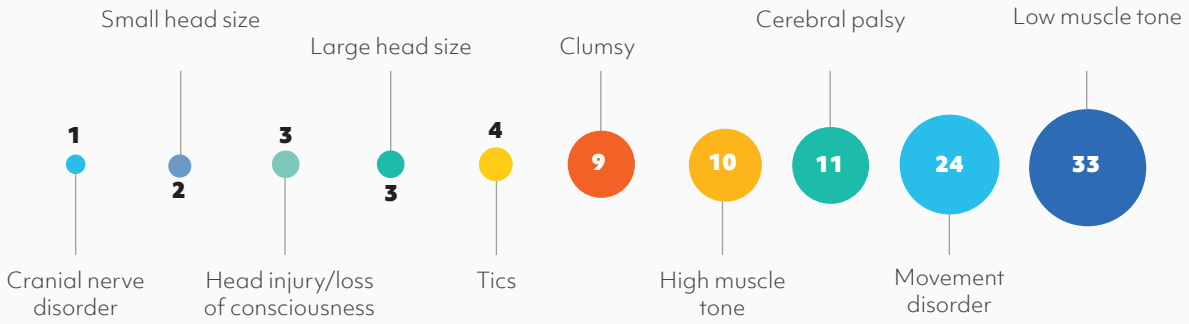
Reported Conditions

Most Commonly Reported Conditions

STXBP1 families reported a variety of associated medical conditions. Some 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

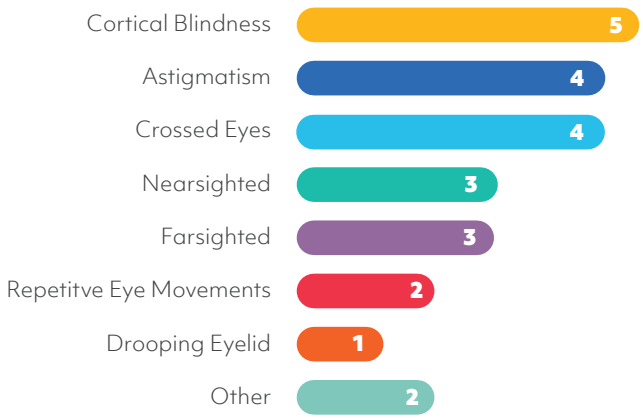


Gastrointestinal Conditions

Note: Individual participants may have reported multiple conditions

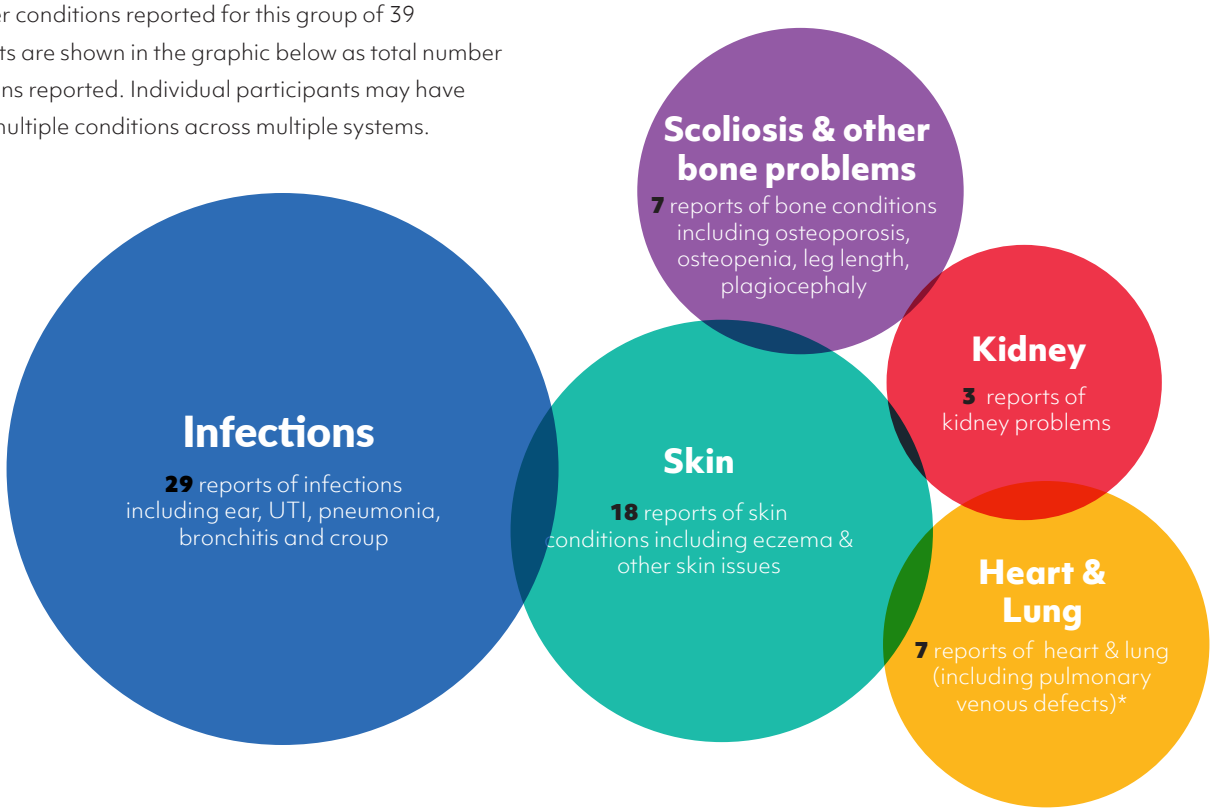
Vision Problems

Note: Individual participants may have reported multiple conditions



Other Conditions Reported

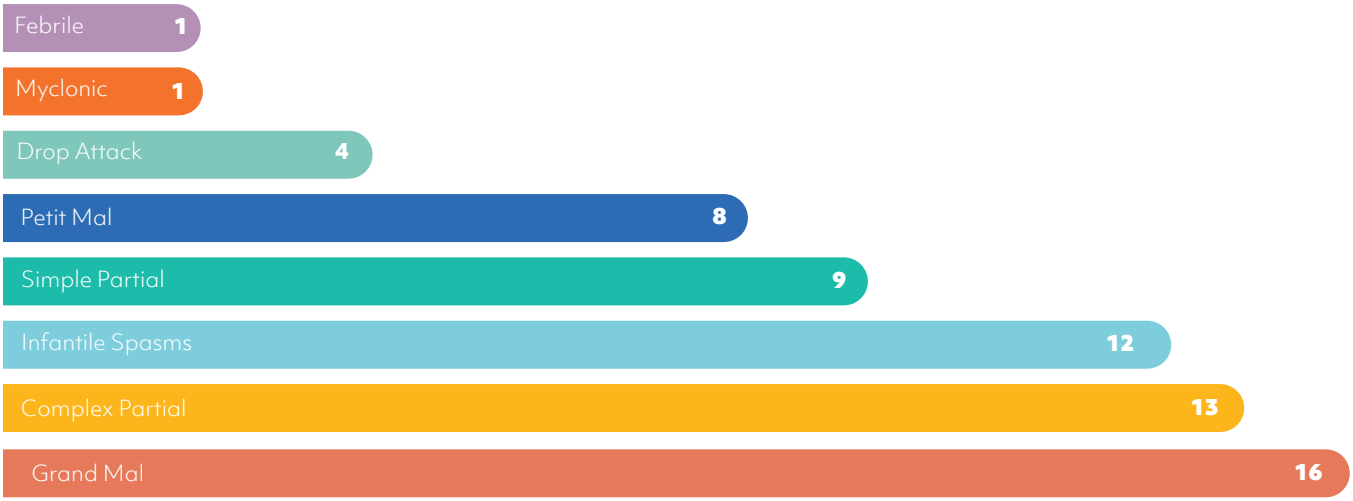
Some other conditions reported for this group of 39 participants are shown in the graphic below as total number of conditions reported. Individual participants may have reported multiple conditions across multiple systems.



* Heart conditions include: high BP, atrial septal defect, sinus tachycardia, pulmonary AVM (hereditary hemorrhagic telangiectasia), pulmonary fistula

Types of Seizures

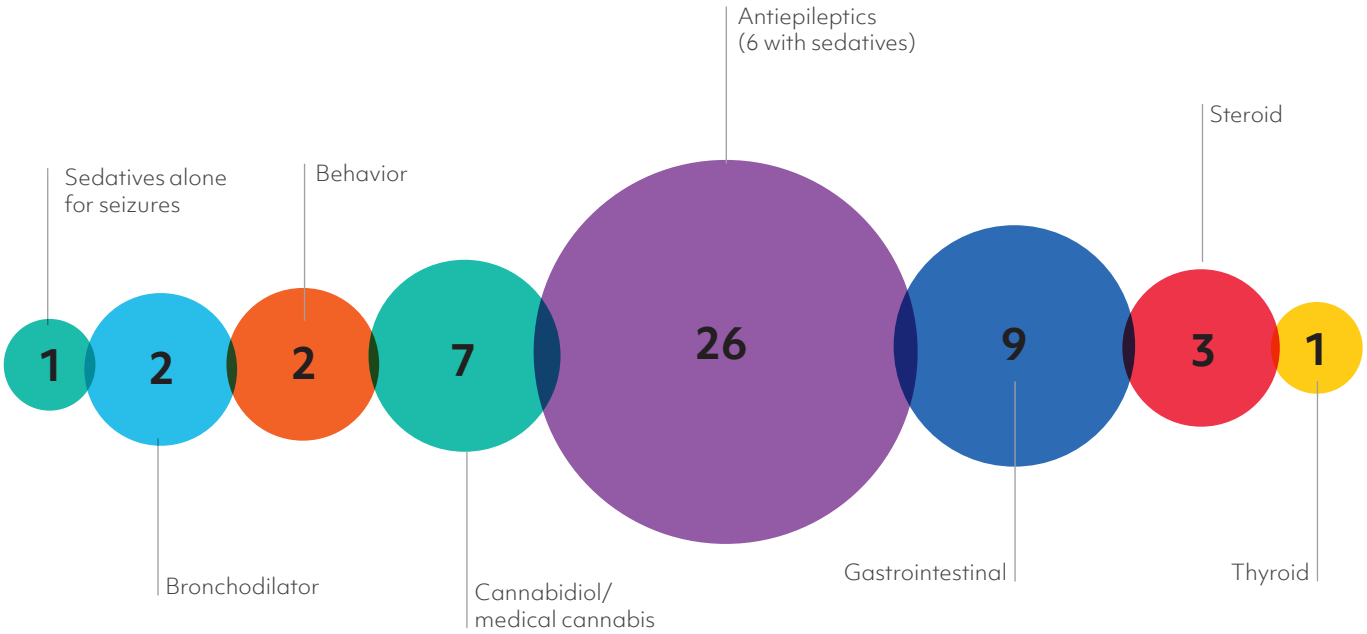
Note: Individual participants may have reported multiple conditions



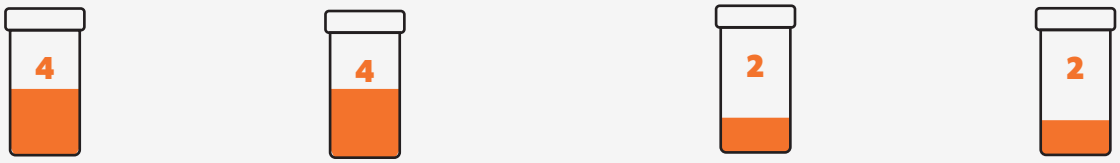
Medications

Medication Use

Simons Searchlight collected medication use from 38 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*



* Other meds less frequently reported by families to have good or complete seizure control: clonazepam (1) and phenobarb (1) (meds were weaned); Depakote/valproic acid (1); Tegretol/carbamazepine (1; med is still currently taken)

THANK YOU

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