Simons Searchlight Registry Update for 16p11.2 duplication

December 2021

Participant Age at Most Recent Interview

Developmental & Behavioral Diagnoses

- Intellectual Disability/Development Delay
- Autism
- Language Impairment
- ADHD
- Anxiety
- OCD

Neurological Problems

- Tic Disorder
- Movement Disorder
- Small Head Size
- Large Head Size
- Low Muscle Tone
- High Muscle Tone
- Clumsy
- Seizures

Gastrointestinal Problems

- GERD
- Diarrhea
- IBS
- Constipation
- Celiac Disease

Notes:
- Graphs show counts of individuals in each category. Individual participants may appear in more than one category if they report multiple conditions.
- Graphs show data as of the last medical history interview completed on participants who have participated in the online portion of the study.

How to Participate

The information in this report is made possible by the active participation of the 16p11.2 duplication community! Progress for individuals in your community with a 16p11.2 duplication is shown below -- log in to your simonssearchlight.org dashboard today to check for new surveys and tasks. Your data could hold the clues geneticists need to find answers.

STEP 1
Sign up online.

STEP 2
Provide your genetic lab report.

STEP 3
Share your important medical history.

STEP 4
Fill out surveys.

STEP 5
Provide a blood sample if you are interested.

STEP 6
Update us every year.
Information Spotlight: **Deeper Dive into Sleep**

The Simons Searchlight Sleep Supplement is a custom survey. We worked with members of the Simons Searchlight community and sleep researchers to design these questions. It asks caregivers about children's sleep habits and the impact of health issues on sleep behaviors. The survey looks at children between ages 2 and 18 years of age. 37 children with a 16p11.2 duplication are represented.

**Getting Enough Sleep**

- Always (2.7%)
- Usually (45.95%)
- Sometimes (29.73%)
- Rarely (16.22%)
- Never (5.41%)

**Children with a 16p11.2 duplication fall asleep in 41 minutes on average.**

**Medical issues related to sleep problems in children with a 16p11.2 duplication***

- Seizures
- Breathing problems
- Itchy skin
- Restless legs
- Movement disorder
- Autonomic dysfunction
- Sleep apnea/snoring
- Medication side effects
- Runny nose
- Mood disorder
- Pain
- Anxiety
- GI symptoms

**Best Ways to Help Sleep: Community Recommendations***

- Discipline
- Sound or white noise machine
- Parent lying in their bed until they fall asleep
- Medication
- Bedtime routine or ritual

*Caregivers were able to select more than one answer.

Thank you for contributing knowledge to Simons Searchlight!