Simons Searchlight Registry Update PPP2R1A

Data in these four graphs are from the medical history phone interviews collected in Simons Searchlight from 11 participants with PPP2R1A.

October 2023

Notations: Graphs show counts of individuals in each category. Individual participants may appear in more than one category if they report multiple conditions.

### Neurological Conditions
- Tic Disorder: 1
- Small Head Size: 2
- Seizures: 5
- Movement Disorder: 2
- Low Muscle Tone: 11
- Large Head Size: 3
- High Muscle Tone: 4
- Cortical Visual Impairment: 1
- Coordination Problems: 2
- Cerebral Palsy: 3

### Gastrointestinal Conditions
- Constipation: 3
- Diarrhea: 1
- GERD: 5
- IBS: 1

### Developmental and Behavioral Conditions
- ADHD: 1
- Anxiety: 1
- Autism: 3
- Intellectual Disability/Developmental Delay: 9
- Language Delay/Impairment: 10
- Obsessive Compulsive Disorder: 1

### How to participate?

The information in this report is made possible by the active participation of the PPP2R1A community! Progress for individuals in your community with PPP2R1A 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

Log in to see next steps.
Information Spotlight:
Child Behavior Checklist for Ages 1.5–5

The Child Behavior Checklist (CBCL) is a survey used for caregivers to share their concerns about their children’s behaviors and emotions.

Caregivers completed the CBCL for Ages 1.5-5 (CBCL/1.5-5) for 7 children in the Simons Searchlight PPP2R1A community.

Behavioral and Emotional Concerns
Ages 1.5-5 Years (7 individuals)

- Aggression
- Sadness
- Anxiety
- Attention/Hyperactivity
- Oppositional Behavior

Tip: Longer bars mean more concerns reported by more people.

Top Behavioral and Emotional Concerns
Ages 1.5-5 Years (7 individuals)

- Chews on non-food items
- Doesn’t respond when spoken to
- Frequently wakes during the night
- Trouble sleeping alone

Tip: Bubbles show top 4 concerns reported in the group.

Kindly note that these summaries are from self-reported online research surveys and errors are possible. The behaviors and emotions shown may not apply to everyone, and individual caregiver perspectives and levels of concern may vary. Please contact us with any questions.