Simons Searchlight Registry Update PPP2R1A

Data in these four graphs are from the medical history information collected in Simons Searchlight from 10 participants with PPP2R1A-related syndrome.

April 2024

**Ages in Years**

- 0-4: 6
- 5-9: 2
- 10-14: 1
- 15-19: 1
- 20-24: 1

**Developmental and Behavioral Conditions**

- Anxiety: 1
- Autism: 3
- Intellectual Disability/Developmental Delay: 8
- Language Delay/Impairment: 9
- Obsessive Compulsive Disorder: 1

**Neurological Conditions**

- Small Head Size: 2
- Seizures: 5
- Movement Disorder: 2
- Low Muscle Tone: 10
- Large Head Size: 3
- High Muscle Tone: 4
- Cortical Visual Impairment: 1
- Coordination Problems: 1
- Cerebral Palsy: 3

**Gastrointestinal Conditions**

- Constipation: 2
- Diarrhea: 1
- GERD: 1
- IBS: 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.
Simons Searchlight families live in 75 countries globally, including 23 PPP2R1A participants in 6 countries.

Most Simons Searchlight families live in the United States. See where 16 PPP2R1A participants live across the 50 states.

Thank you for participating in Simons Searchlight and helping us reach families worldwide!