Simons Searchlight Registry Update PACS1

Ages in Years

- 0-4: 6
- 5-9: 10
- 10+: 5

Developmental & Behavioral Diagnoses

- Intellectual Disability/Development Delay: 21
- Language Impairment: 10
- ADHD: 12
- Autism: 1

Neurological Problems

- Low Muscle Tone: 9
- Clumsy: 4
- High Muscle Tone: 2
- Large Head Size: 1
- Small Head Size: 5
- Movement Disorder: 1
- Tic Disorder: 1
- Seizures: 10

Gastrointestinal Problems

- GERD: 9
- Diarrhea: 12
- Constipation: 1
- Irritable Bowel Syndrome: 2

Notes:
- Graphs show counts of individuals in each category. Individual participants may appear in more than one category if they report multiple conditions.
- Information on reported seizure types is included on the 2021 quarter 1 registry report available on simonssearchlight.org/PACS1.

Participation & Registration

This ever-growing database is made possible by the active participation of the PACS1 community! Progress for individuals in your community with a PACS1 variant is shown below – log in to your simonssearchlight.org dashboard today to check for new surveys and other priority tasks. Your unique data could hold the clues scientists need to make progress and find answers.

**STEP 1**
Sign up online.

**STEP 2**
Provide your clinical lab report.

**STEP 3**
Schedule medical history phone call with a genetic counselor.

**STEP 4**
Fill out surveys.

**STEP 5**
Provide a blood sample if you are interested.

**STEP 6**
We follow up with you over time.

Log in to see next steps

**Counts:**
- Low Muscle Tone: 41
- Clumsy: 26
- High Muscle Tone: 21
- Large Head Size: 21
- Small Head Size: 21
- Movements Disorder: 9
Information Spotlight: Child Behavior Checklist

The Child Behavior Checklist (CBCL) is a survey used to understand behavioral and emotional concerns in children.

The bars in the graphs show the overall level of concern reported by Simons Searchlight caregivers who completed the CBCL survey about their children with PACS1. Individual caregivers may have indicated a lower or higher level of concern than indicated by the overall group.

Behavioral Concerns for ages 1.5-5yrs

Behavioral Concerns for ages 6-18 yrs

Top Behavioral Concerns
Ages 1.5-5 yrs

Scared to attempt new things

Desires frequent attention

Frequently requests help

Specific fears (e.g., animals, places, situations)

Top Behavioral Concerns
Ages 6-18 yrs

Frequently distracted

Repeats certain behaviors

Demands frequent attention

Hyperactive