Simons Searchlight Registry
Update SETBP1

April, 2022

Ages in Years

Developmental and Behavioral Conditions

Neurological Problems

Gastrointestinal Problems

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

How to Participate
The information in this report is made possible by the active participation of the SETBP1 community! Progress for individuals in your community with SETBP1 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

68
54
30
41
12
Information Spotlight: Quality of Life

The Quality of Life Inventory is a scientific survey where we asked caregivers to describe how their family member with SETBP1 responds to life experiences.

It measures quality of life by looking at the following areas: physical health, psychological well-being, social relationships, leisure, and independence.

These graphs reflect the percentage of people who reported "Often" or "Very Often" for each question.

### Health & Well-being

- **Has energy for daily activities**: 92%
- **Is in good health**: 100%
- **Is alert and aware**: 92%
- **Sleeps well**: 69%

### Feelings & Emotions

- **In a good mood**: 92%
- **Smiles**: 100%
- **Expresses happiness using body language**: 100%
- **Laughs or giggles**: 92%
- **Unsettled for no reason**: 8%
- **Displays aggression**: 8%
- **Feels upset or angry**: 15%
- **Withdrawn, low mood**: 0%
- **Intentionally self harms**: 0%
- **Dislikes changes in routine**: 23%
- **Displays anxiety or agitation**: 23%

The questionnaire was given to family members of people 2 years and older.

13 people with SETBP1 are shown.

We gratefully acknowledge Dr. Jenny Downs of the Telethon Kids Institute.

QI-Disability Parent Version © Telethon Kids Institute, 2017, 2021
Information Spotlight: Quality of Life

"Quality of life is a common survey of patient-centered outcomes in clinical trials and natural history studies."

- Alison Holbrook, PhD

**Daily Life**

- Expresses their needs: 77%
- Makes own choices for activities: 85%
- Likes using technology: 69%
- Helps with routines: 62%
- Enjoys making things: 62%

**Activities & the Outdoors**

- Enjoys moving their body: 69%
- Enjoys feeling steady during activities: 77%
- Enjoys physical activities: 77%
- Enjoys outings: 62%
- Enjoys time outdoors: 85%

**Family & Friends**

- Happy when they are understood: 92%
- Feels relaxed while making eye contact: 92%
- Initiates greetings with others: 85%
- Happy to be included: 92%
- Enjoys socializing at mealtimes: 92%
- Enjoys interacting with others: 100%
- Excited by upcoming activities: 85%

Thank you for contributing knowledge to Simons Searchlight!