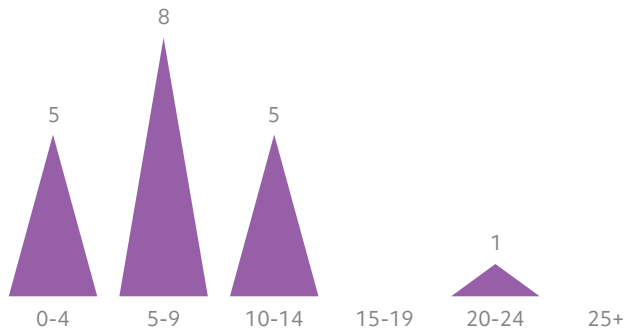


Simons Searchlight Registry Update SETBP1

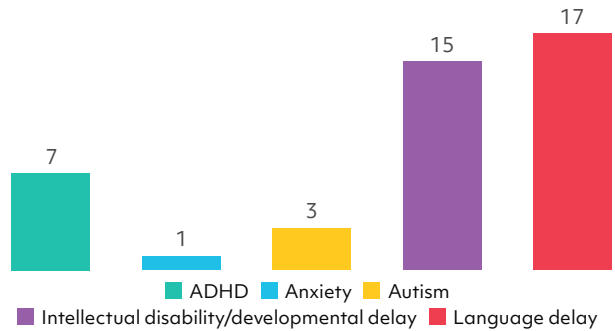
April, 2022

Data in these four graphs are from the medical history phone interviews collected in Simons Searchlight on 19 participants with SETBP1.

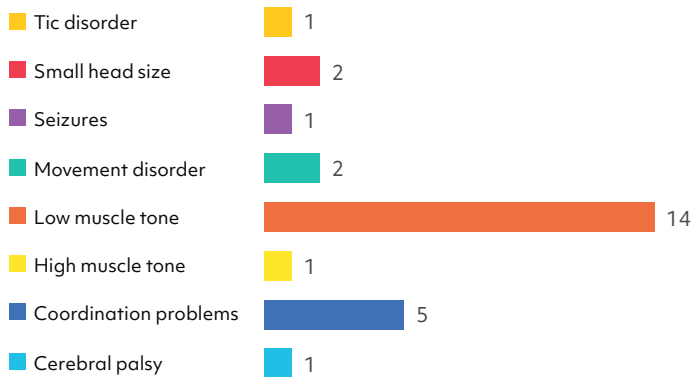
Ages in Years



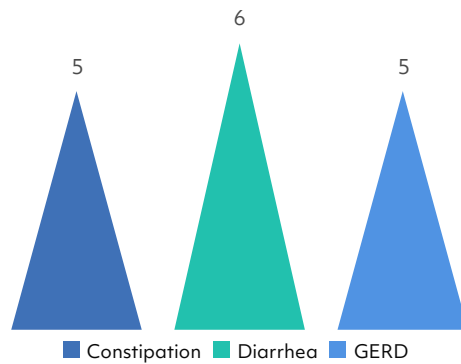
Developmental and Behavioral Conditions



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



68

STEP 2

Provide your genetic lab report



54

STEP 3

Share your important medical history



30

STEP 4

Fill out surveys



41

STEP 5

Provide a blood sample if you are interested



12

STEP 6

Update us every year



Log in to see next steps

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.

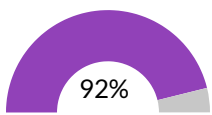
It measures quality of life by looking at the following areas: physical health, psychological well-being, social relationships, leisure, and independence.

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

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

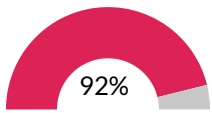
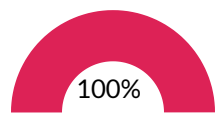
13 people with SETBP1 are shown.

Health & Well-being



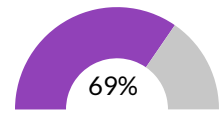
Has energy for daily activities

Is in good health



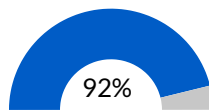
Is alert and aware

Sleeps well

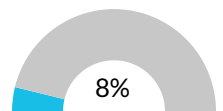


Feelings & Emotions

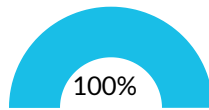
In a good mood



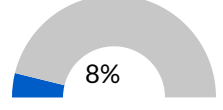
Unsettled for no reason



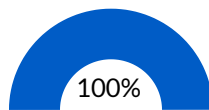
Smiles



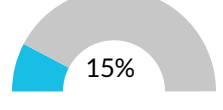
Displays aggression



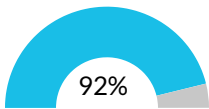
Expresses happiness using body language



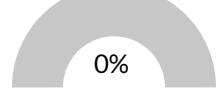
Feels upset or angry



Laughs or giggles



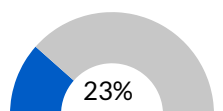
Withdrawn, low mood



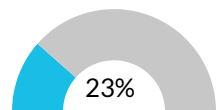
Intentionally self harms



Dislikes changes in routine



Displays anxiety or agitation



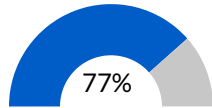
"Quality of life is a common survey of patient-centered outcomes in clinical trials and natural history studies."

- Alison Holbrook, PhD

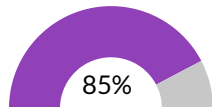


Daily Life

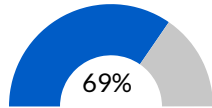
Expresses their needs



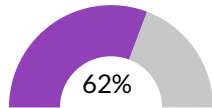
Makes own choices for activities



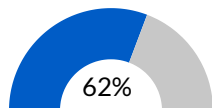
Likes using technology



Helps with routines

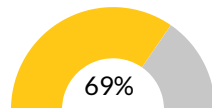


Enjoys making things

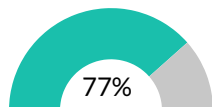


Activities & the Outdoors

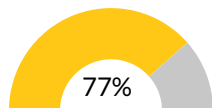
Enjoys moving their body



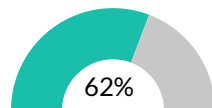
Enjoys feeling steady during activities



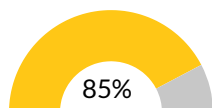
Enjoys physical activities



Enjoys outings

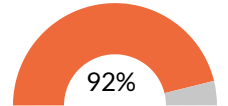


Enjoys time outdoors

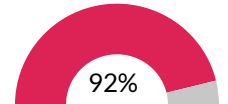


Family & Friends

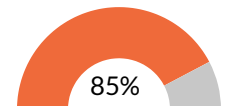
Happy when they are understood



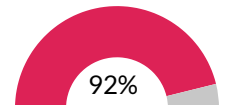
Feels relaxed while making eye contact



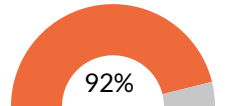
Initiates greetings with others



Happy to be included



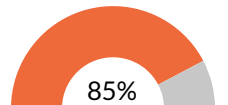
Enjoys socializing at mealtimes



Enjoys interacting with others



Excited by upcoming activities



Thank you

for contributing knowledge to Simons Searchlight!