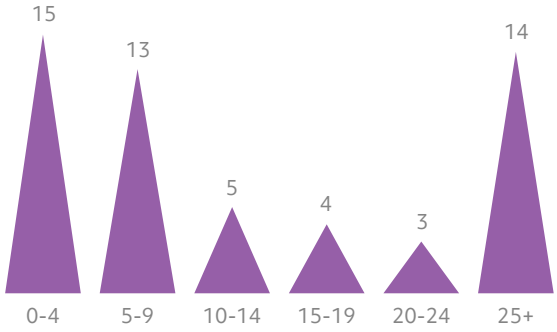
Simons Searchlight Registry Update 1q21.1 deletion

April, 2022

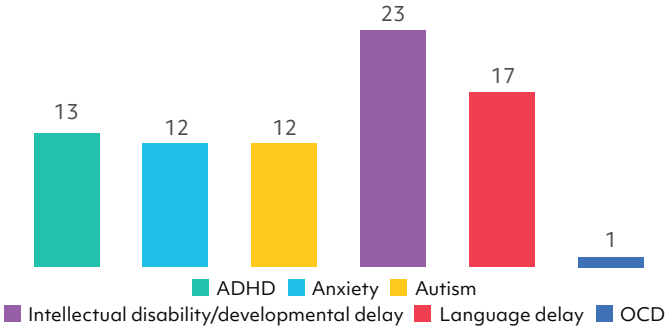
SIMONS SEARCHLIGHT

Data in these four graphs are from the medical history phone interviews collected in Simons Searchlight on 54 participants with 1q21.1 deletion.

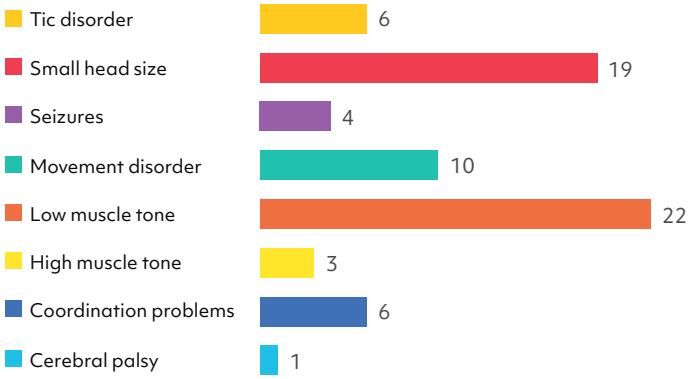
Ages in Years



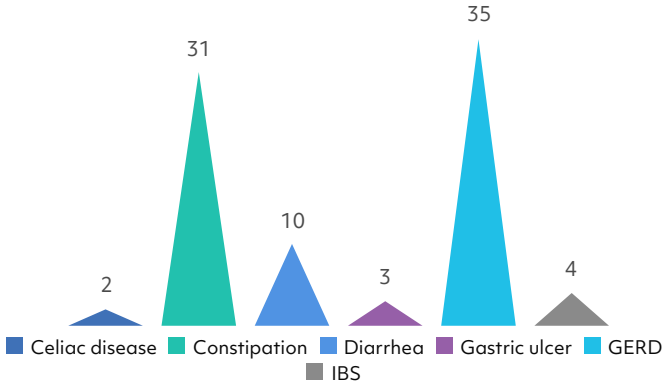
Developmental and Behavioral Conditions



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 1q21.1 deletion community! Progress for individuals in your community with 1q21.1 deletion 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	STEP 2	STEP 3	STEP 4	STEP 5	STEP 6
Sign up online	Provide your genetic lab report	Share your important medical history	Fill out surveys	Provide a blood sample if you are interested	Update us every year
131	70	56	57	19	Log in to see next steps

Information Spotlight: **Quality of Life**

SIMONS SEARCHLIGHT

The **Quality of Life Inventory** is a scientific survey where we asked caregivers to describe how their family member with 1q21.1 deletion responds to life experiences.

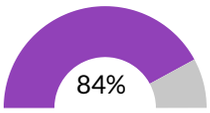
It measures quality of life by looking at the following areas: physical health, psychological well-being, social relationships, leisure, and independence.

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

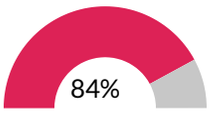
The questionnaire was given to family members of people 2 years and older.

19 people with **1q21.1 deletion** are shown.

Health & Well-being

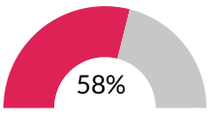


Has energy for daily activities

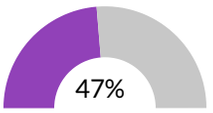


Is alert and aware

Is in good health

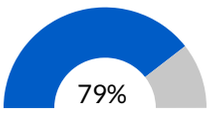


Sleeps well

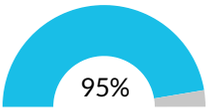


Feelings & Emotions

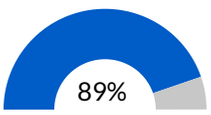
In a good mood



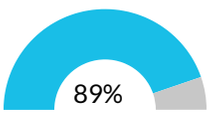
Smiles



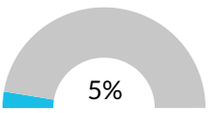
Expresses happiness using body language



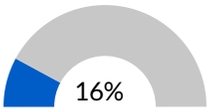
Laughs or giggles



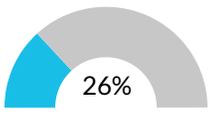
Unsettled for no reason



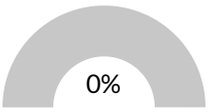
Displays aggression



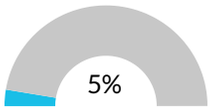
Feels upset or angry



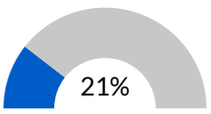
Withdrawn, low mood



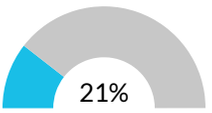
Intentionally self harms



Dislikes changes in routine



Displays anxiety or agitation



We gratefully acknowledge Dr. Jenny Downs of the Telethon Kids Institute.
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Information Spotlight: **Quality of Life**

SIMONS SEARCHLIGHT

"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	89%
Makes own choices for activities	84%
Likes using technology	95%
Helps with routines	84%
Enjoys making things	84%

Activities & the Outdoors

Enjoys moving their body	89%
Enjoys feeling steady during activities	84%
Enjoys physical activities	89%
Enjoys outings	79%
Enjoys time outdoors	89%

Family & Friends

Happy when they are understood	89%
Feels relaxed while making eye contact	74%
Initiates greetings with others	74%
Happy to be included	89%
Enjoys socializing at mealtimes	89%
Enjoys interacting with others	95%
Excited by upcoming activities	89%

Thank you

for contributing knowledge to Simons Searchlight!