

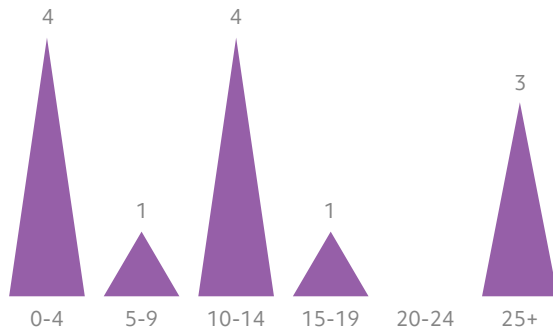
Simons Searchlight Registry Update DISTAL 16p11.2 deletion

SIMONS
SEARCHLIGHT

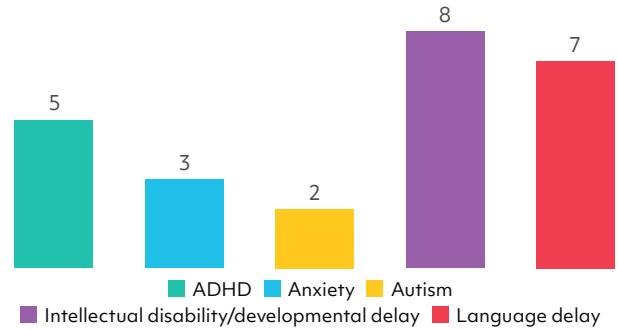
April, 2022

Data in these four graphs are from the medical history phone interviews collected in Simons Searchlight on 13 participants with DISTAL 16p11.2 deletion.

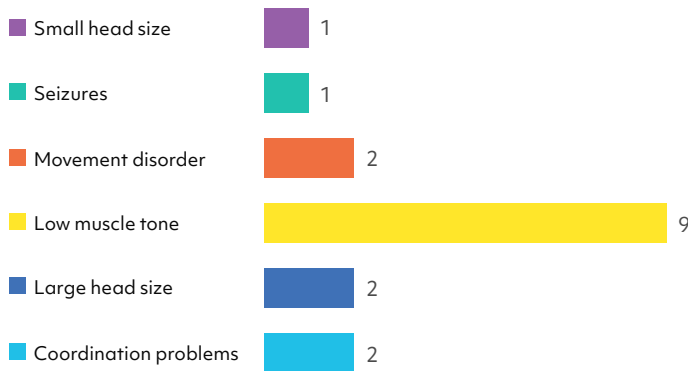
Ages in Years



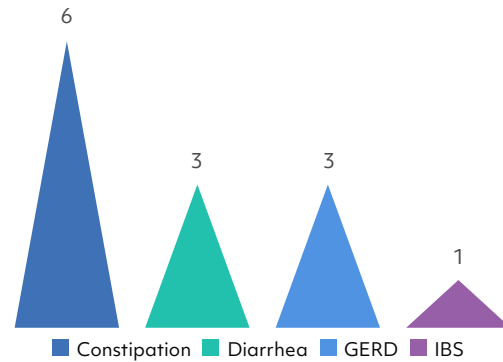
Developmental and Behavioral Conditions



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 DISTAL 16p11.2 deletion community! Progress for individuals in your community with DISTAL 16p11.2 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

Sign up online



59

STEP 2

Provide your genetic lab report



55

STEP 3

Share your important medical history



15

STEP 4

Fill out surveys



35

STEP 5

Provide a blood sample if you are interested



5

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 distal 16p11.2 deletion responds to life experiences.

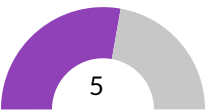
It measures quality of life by looking at the following areas: physical health, psychological well-being, social relationships, leisure, and independence.

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

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

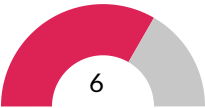
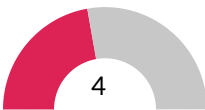
9 people with **distal 16p11.2 deletion** are shown.

Health & Well-being



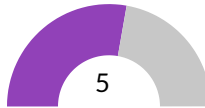
Has energy for daily activities

Is in good health



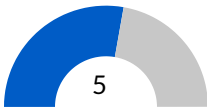
Is alert and aware

Sleeps well

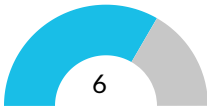


Feelings & Emotions

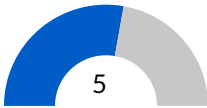
In a good mood



Smiles



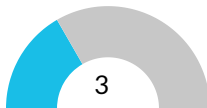
Expresses happiness using body language



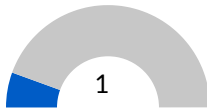
Laughs or giggles



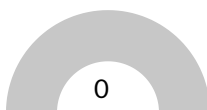
Unsettled for no reason



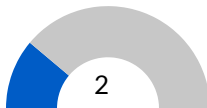
Displays aggression



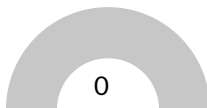
Feels upset or angry



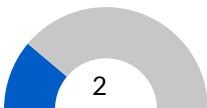
Withdrawn, low mood



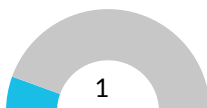
Intentionally self harms



Dislikes changes in routine



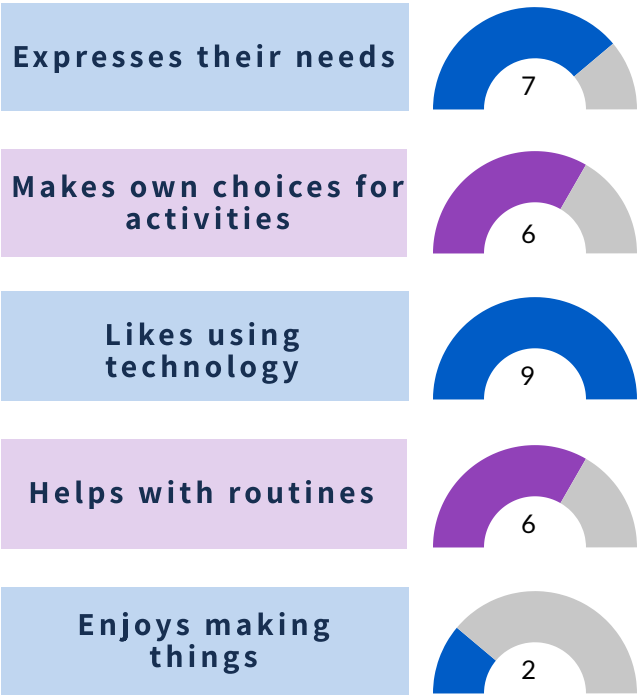
Displays anxiety or agitation



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

- Alison Holbrook, PhD

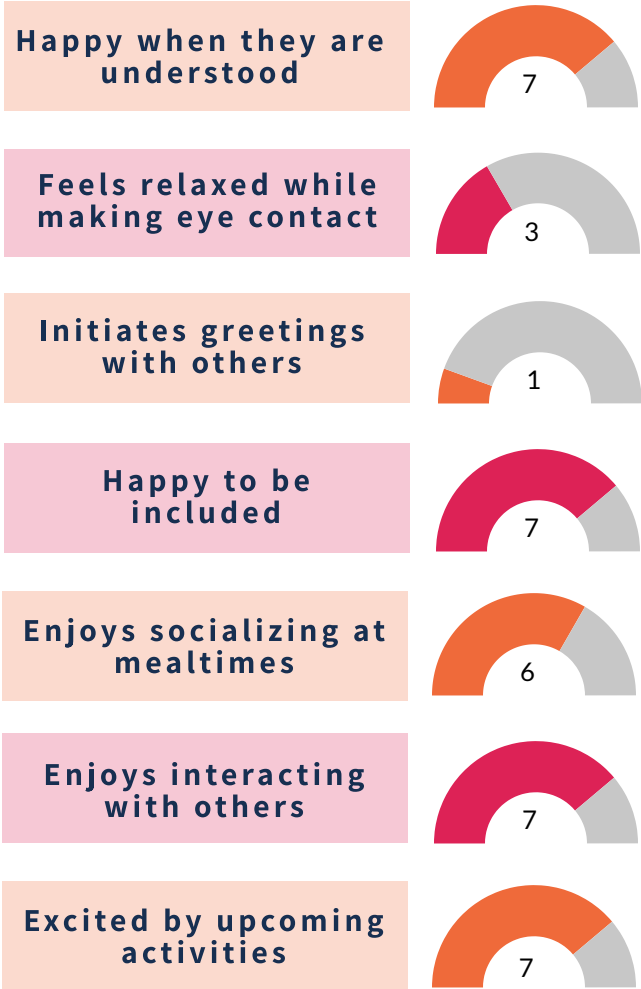
Daily Life



Activities & the Outdoors



Family & Friends



Thank you

for contributing knowledge
to Simons Searchlight!