

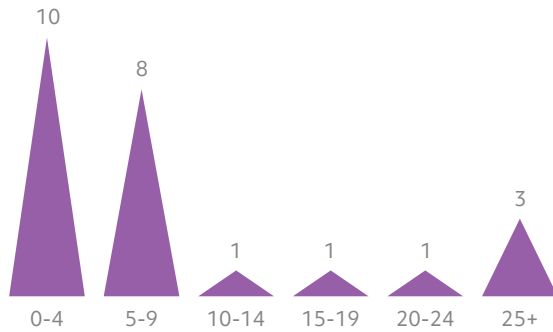
Simons Searchlight Registry Update MED13L

April, 2022

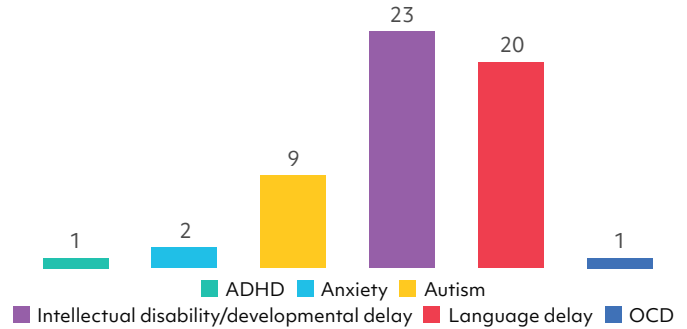
SIMONS
SEARCHLIGHT

Data in these four graphs are from the medical history phone interviews collected in Simons Searchlight on 24 participants with MED13L.

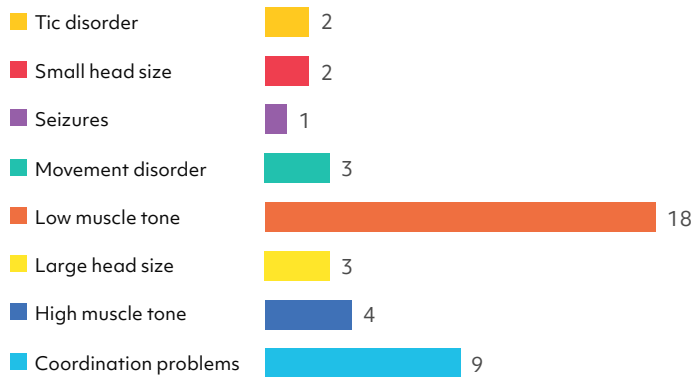
Ages in Years



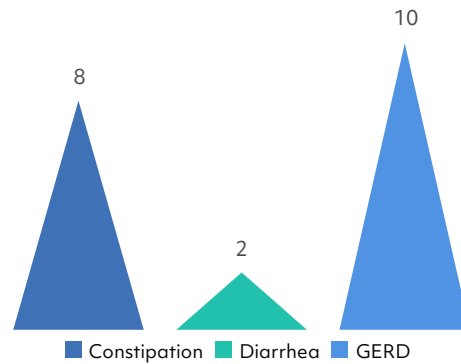
Developmental and Behavioral Conditions



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 MED13L community! Progress for individuals in your community with MED13L 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



91

STEP 2

Provide your genetic lab report



65

STEP 3

Share your important medical history



34

STEP 4

Fill out surveys



55

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 MED13L 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.

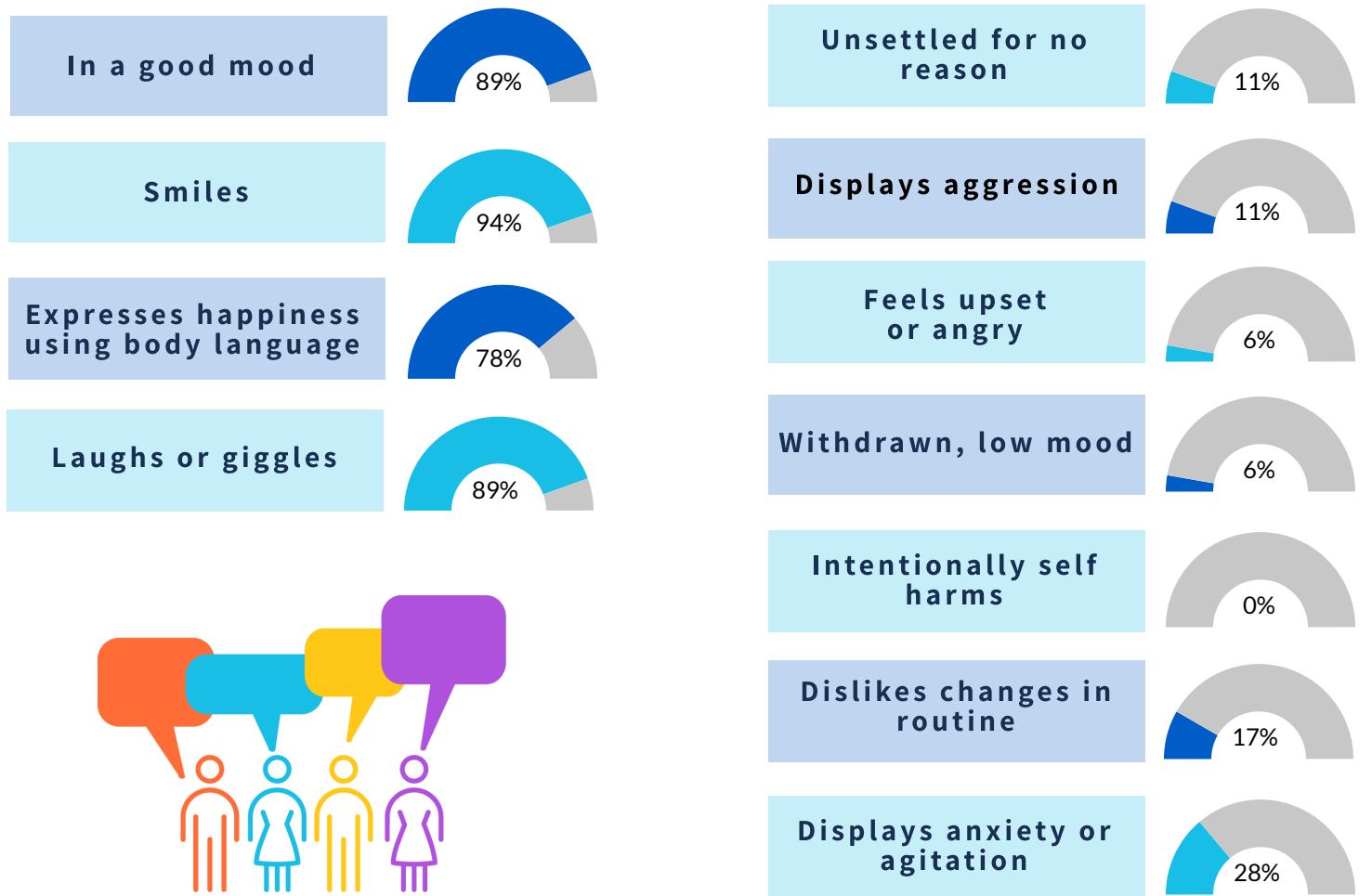
The questionnaire was given to family members of people 2 years and older.

18 people with **MED13L** are shown.

Health & Well-being



Feelings & Emotions

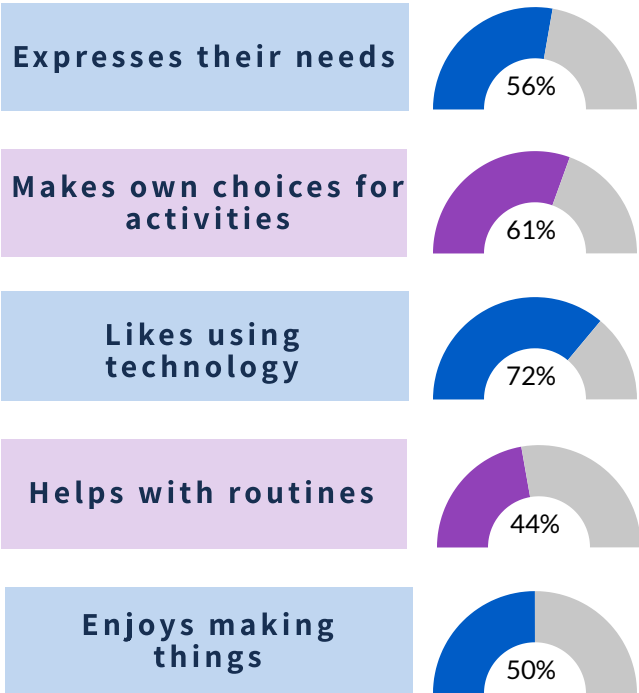


"Quality of life is a common survey of patient-centered outcomes in clinical trials and natural history studies."

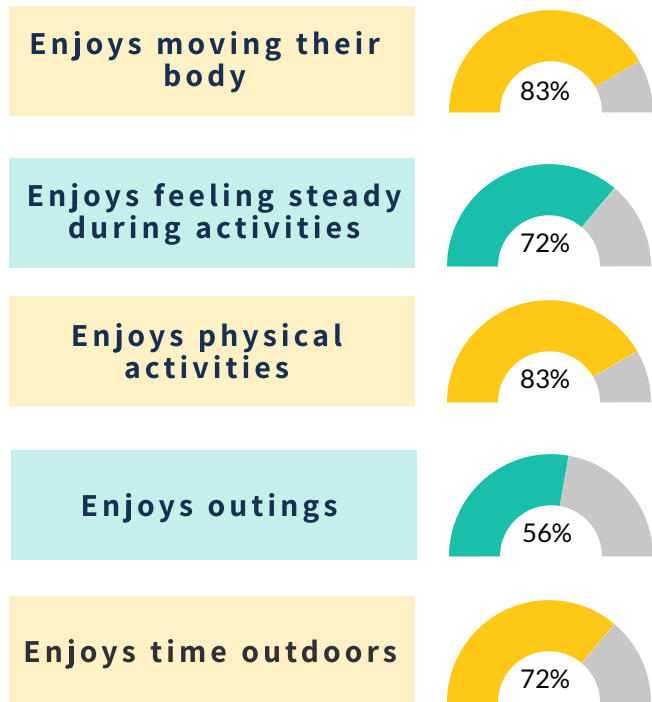
- Alison Holbrook, PhD



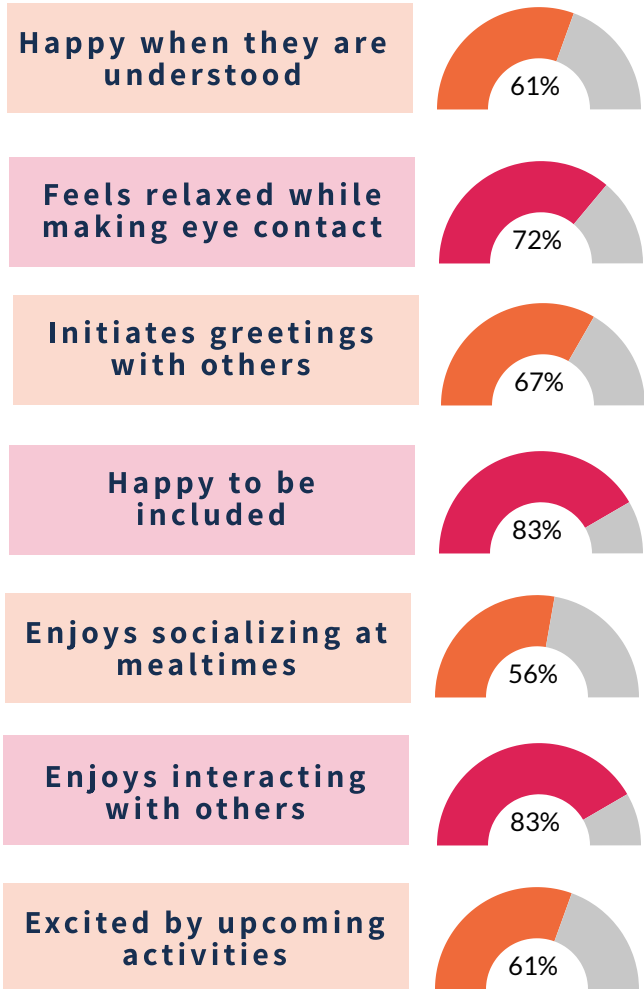
Daily Life



Activities & the Outdoors



Family & Friends



Thank you

for contributing knowledge to Simons Searchlight!