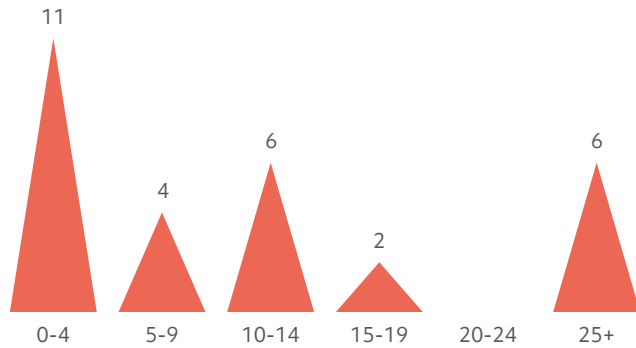


Simons Searchlight Registry Update **Distal 16p11.2 deletion**

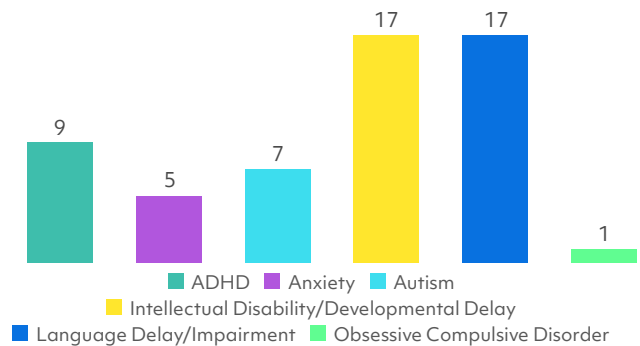
Data in these four graphs are from the medical history information collected in Simons Searchlight from 29 participants with Distal 16p11.2 deletion syndrome.

January 2025

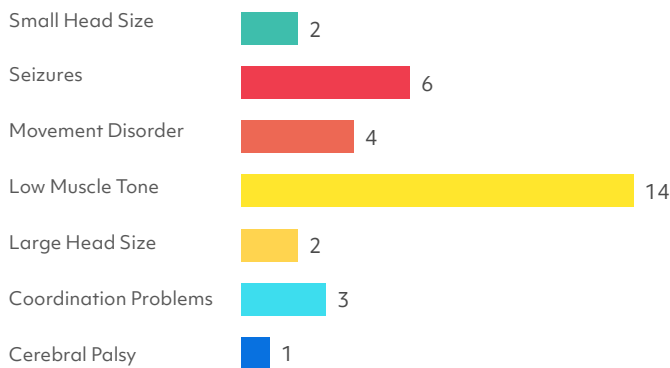
Ages in Years



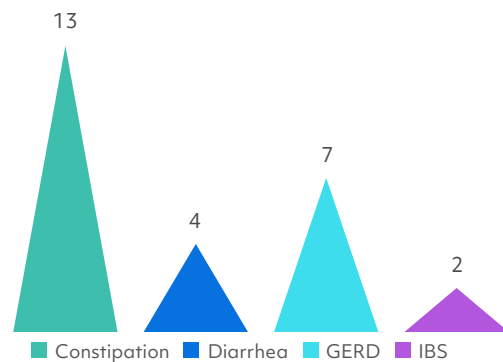
Developmental and Behavioral Conditions



Neurological Conditions



Gastrointestinal Conditions



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

94



STEP 2

Provide your genetic lab report

83



STEP 3

Share your important medical history

37



STEP 4

Fill out surveys

58



STEP 5

Provide a blood sample if you are interested

6



STEP 6

Update us every year

Log in to see next steps

Information Spotlight | 2024 Surveys in Review

Thank you for being a crucial part of Simons Searchlight and sharing your story through our online surveys. Together, we're making strides in rare neurogenetic conditions, thanks to your unwavering support.

2024 survey highlights include:

- The **Annual Medical History Survey**, to give researchers a key tool to understand your genetic story, identify unique medical details, and track changes over time.
- The **Vineland Adaptive Behavior Scales – Third Edition (VABS-3)**, the most common assessment tool for functioning level and intervention needs for individuals with developmental disabilities, in research, school and clinic.
- The **Seizure History Survey and Annual Seizure Update**, for researchers to learn about seizures onset, frequency, treatment, and impact over time.

In 2024, the Distal 16p11.2 deletion community in Simons Searchlight completed*:

6 new medical histories, totaling **45** medical history updates to date,
12 new Vineland entries, totaling **39** Vinelands to date**,
19 new seizure history updates, totaling **69** seizure surveys to date,

and many surveys on sleep, quality of life, behavior, and more.



In 2024, genetic communities across ALL of Simons Searchlight completed*:

994 new medical histories, totaling **6082** medical history updates to date,
1344 new Vineland entries, totaling **5071** Vinelands to date**,
1908 new seizure history updates, totaling **6582** seizure surveys to date,

and many surveys on sleep, quality of life, behavior, and more.



Make sure you are counted in research in 2025 by visiting your dashboard at www.bit.ly/Simons_Searchlight_Dashboard and staying up to date on your surveys.

*Counts reflect number of surveys, not participants. Participants can fill out multiple surveys over time, so that researchers learn more about these genetic conditions across the lifespan.

**New Vineland entries are from the VABS-3. Total entries include Vineland-II and VABS-3.