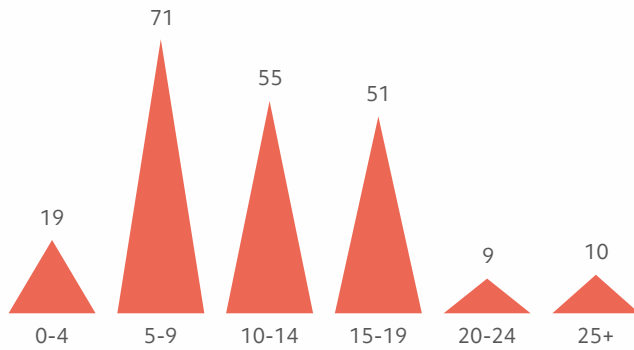# SIMONS SEARCHLIGHT

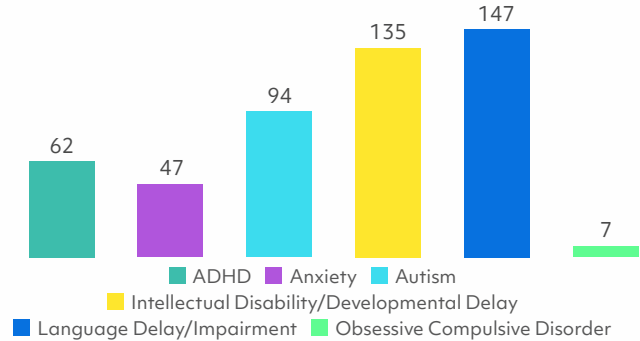# Simons Searchlight Registry Update **16p11.2 deletion**

Data in these four graphs are from the medical history phone interviews collected in Simons Searchlight from 215 participants with a 16p11.2 deletion.
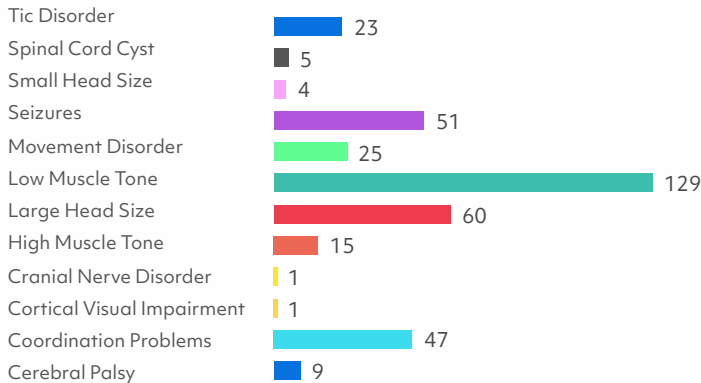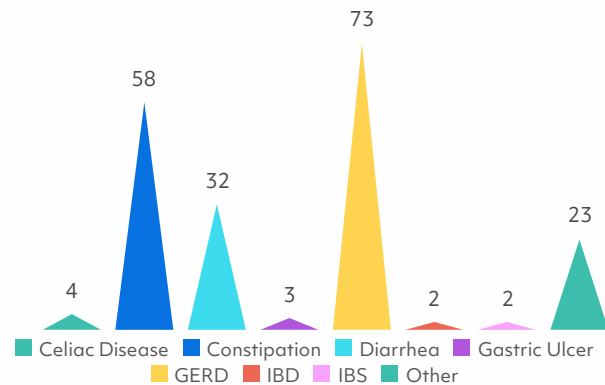
August 2023

Ages in Years

Age	Count
0-4	19
5-9	71
10-14	55
15-19	51
20-24	9
25+	10

Developmental and Behavioral Conditions

Condition	Count
ADHD	62
Anxiety	47
Autism	94
Intellectual Disability/Developmental Delay	135
Language Delay/Impairment	147
Obsessive Compulsive Disorder	7

Neurological Conditions

Condition	Count
Tic Disorder	23
Spinal Cord Cyst	5
Small Head Size	4
Seizures	51
Movement Disorder	25
Low Muscle Tone	129
Large Head Size	60
High Muscle Tone	15
Cranial Nerve Disorder	1
Cortical Visual Impairment	1
Coordination Problems	47
Cerebral Palsy	9

Gastrointestinal Conditions

Condition	Count
Celiac Disease	4
Constipation	58
Diarrhea	32
Gastric Ulcer	3
GERD	73
IBD	2
IBS	2
Other	23

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 16p11.2 deletion community! Progress for individuals in your community with 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
504

STEP 2
Provide your genetic lab report
293

STEP 3
Share your important medical history
225

STEP 4
Fill out surveys
243

STEP 5
Provide a blood sample if you are interested
107

STEP 6
Update us every year
Log in to see next steps

Information Spotlight:
Deeper Dive into Development

We use a standardized measure called the Vineland Adaptive Behavior Scales (Vineland-3) to measure each person's development in communication, self-care, social skills, and in younger children, motor skills.

We can learn how skills develop over time by looking at growth charts. The sample graph on the next page is an example growth chart, and the graphs afterwards show real Vineland-3 data from your community.

When we plot everyone together, we can see what people can do at younger and older ages. This is one snapshot in time. The more people fill out their Vineland over time, the more we can estimate what true longitudinal growth looks like for everyone in your community.

125 16p11.2 deletion participants contributed Vineland-3 data to this report

Scan the QR code below or use the following link (bit.ly/Searchlight_Vineland_Graph) for a video from Dr. LeeAnne Green Snyder, Clinical Research Scientist for Simons Searchlight, on how to read the Vineland Adaptive Behavior Scales growth charts.
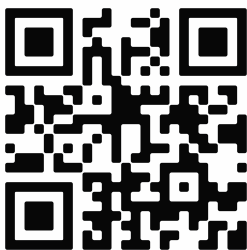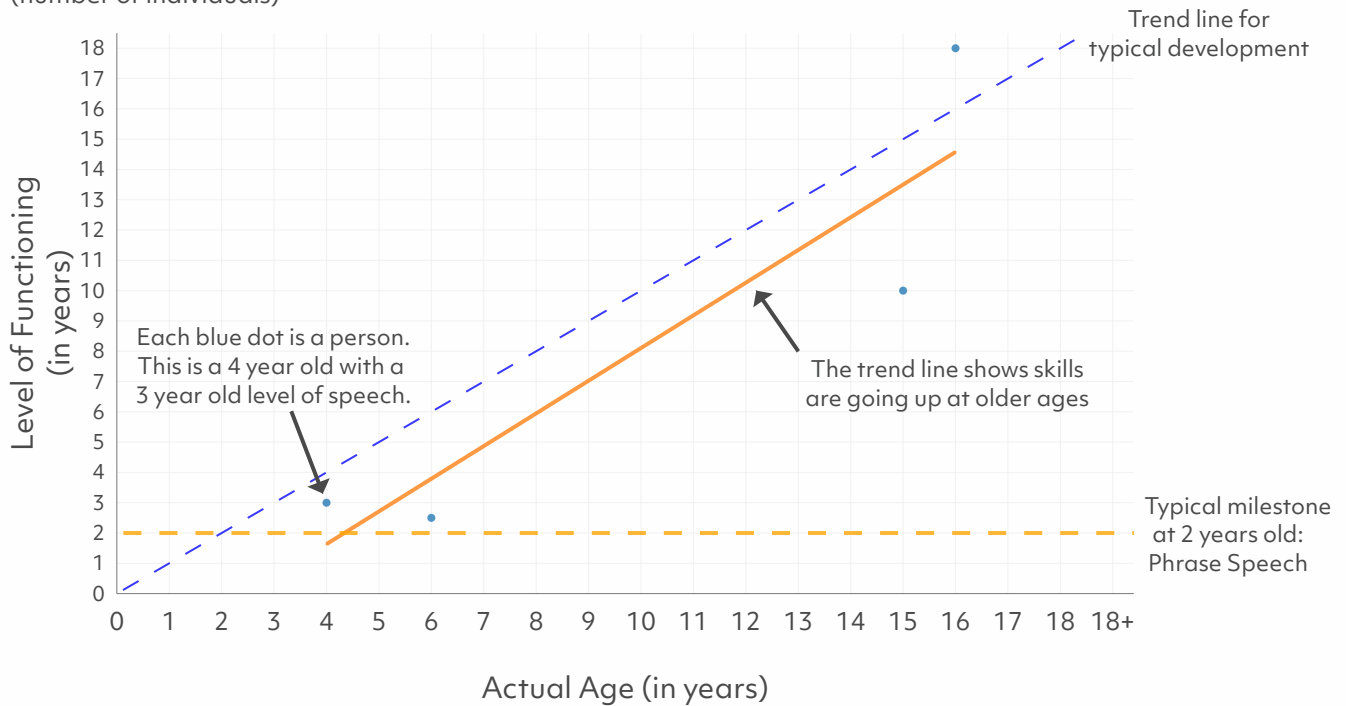
We will continue to collect Vineland data every year. Over time, we will have a better understanding of how individuals with 16p11.2 deletion change as they get older.

Kindly note that these charts are from self-reported online research surveys and errors are possible. The skills shown are only estimates and may not apply to everyone. Remember that everyone develops at their own pace, and individual perspectives and responses may vary. Please let us know if you have any questions.

SIMONS
SEARCHLGHT

Sample graph:
Developmental Levels in Speech at Younger and Older Ages

Expressive Language Development
(number of individuals)

Trend line for typical development

Each blue dot is a person. This is a 4 year old with a 3 year old level of speech.

The trend line shows skills are going up at older ages

Typical milestone at 2 years old: Phrase Speech

Level of Functioning (in years)

18
17
16
15
14
13
12
11
10
9
8
7
6
5
4
3
2
1
0

0 1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 18+

Actual Age (in years)
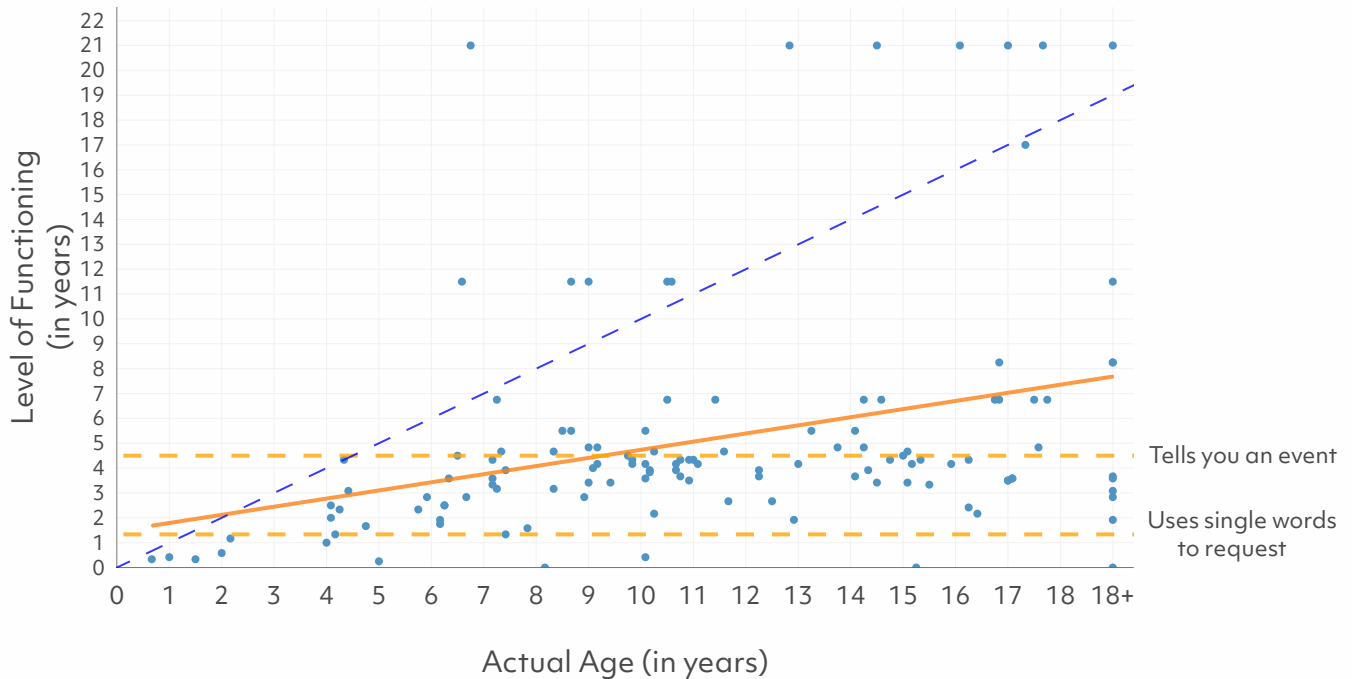
What does this mean as my child gets older?

Along the bottom is the actual age of the person.

The dots are placed at the level at which they are expressing themselves.

A percentage of individuals are likely to be using language at age 4 and older.

Developmental Levels in Speech at Younger and Older Ages in 16p11.2 deletion

Expressive Language Development
(125 individuals)

Level of Functioning (in years)

Actual Age (in years)

Tells you an event

Uses single words to request

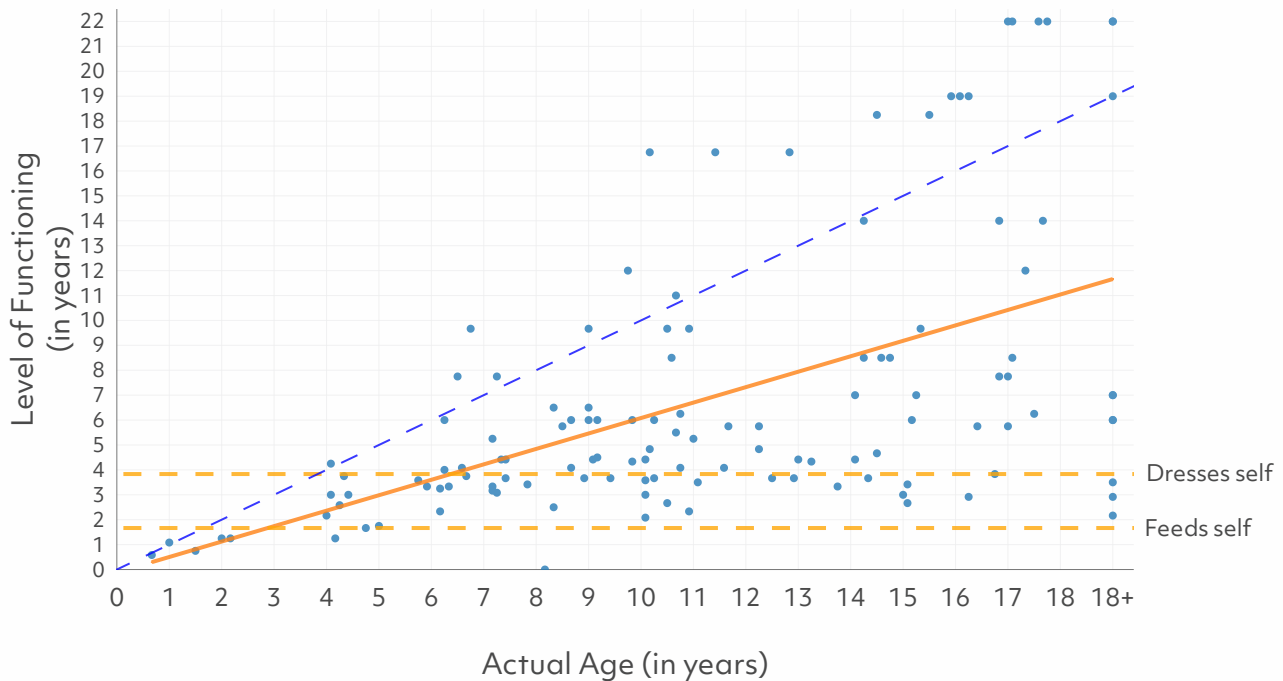**What does this mean as my child gets older?**

| Along the bottom is the actual age of the person. | The dots are placed at the level at which they express themselves. | 95% of individuals are likely to be using language at age 4 and older. |

Developmental Levels in Self Care at Younger and Older Ages in 16p11.2 deletion

Self Care Skills Development
(125 individuals)

Level of Functioning (in years)

Actual Age (in years)

- - - Dresses self
- - - Feeds self

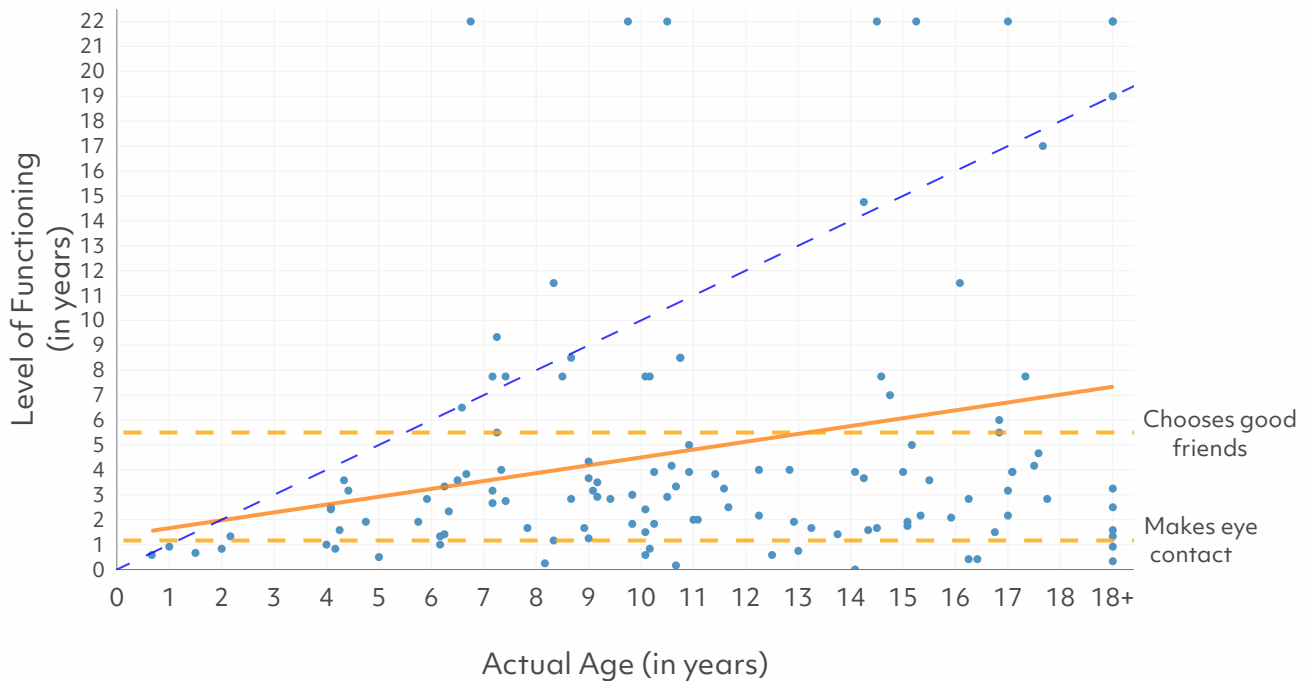| **What does this mean as my child gets older?** | Along the bottom is the actual age of the person. | The dots are placed at the level at which they can do things for themselves. | 98% of individuals are likely to be feeding themselves at age 4 and older. |

Developmental Levels in Social Skills at Younger and Older Ages in 16p11.2 deletion

Social Skills Development
(125 individuals)

Level of Functioning (in years) vs Actual Age (in years)

Chooses good friends

Makes eye contact
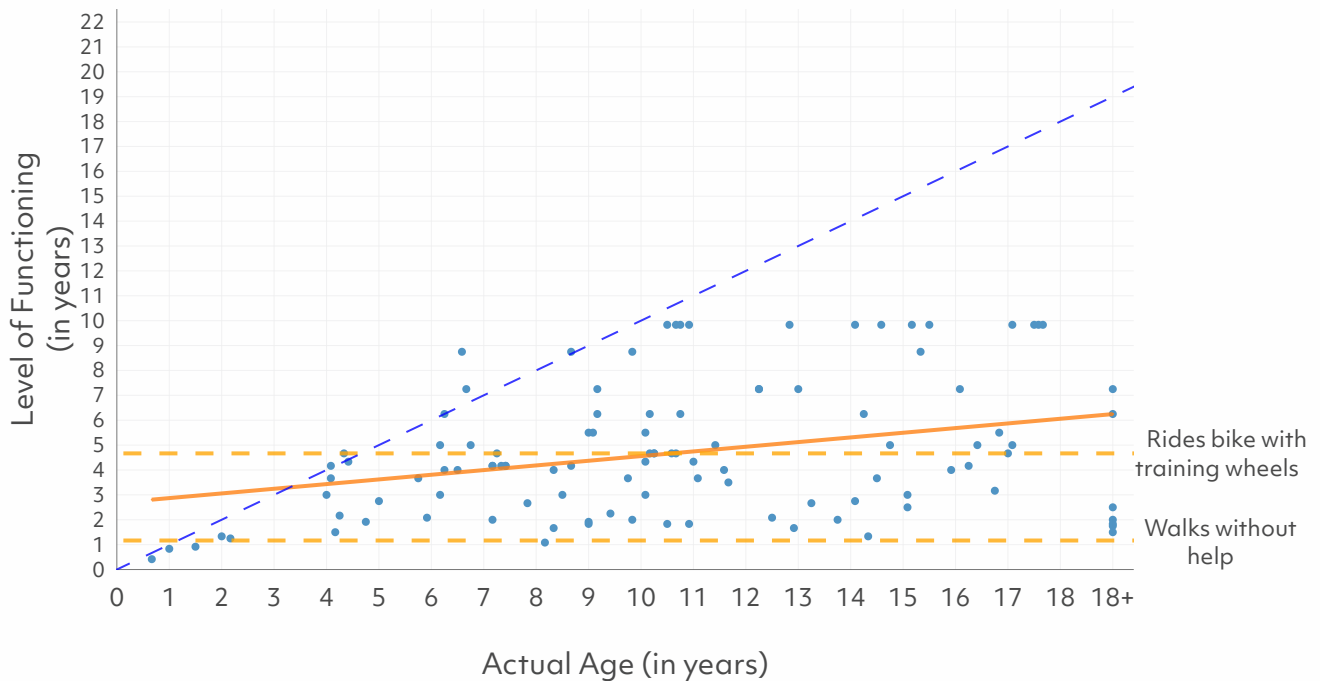
What does this mean as my child gets older?

Along the bottom is the actual age of the person.

The dots are placed at the level of their social behavior.

88% of individuals are likely to be making eye contact at age 4 and older.

Developmental Levels in Motor Skills at Younger and Older Ages in 16p11.2 deletion

Motor Skills Development
(108 individuals)

Level of Functioning (in years) vs Actual Age (in years)

Rides bike with training wheels

Walks without help

What does this mean as my child gets older?

| Along the bottom is the actual age of the person. | The dots are placed at the level of their motor skills. | 99% of individuals are likely to be walking at age 4 and older. |