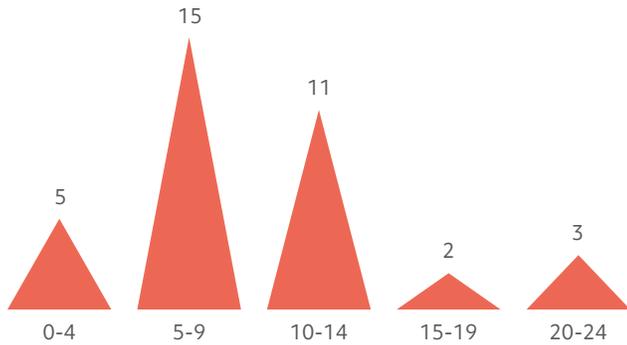


Simons Searchlight Registry Update SETBP1

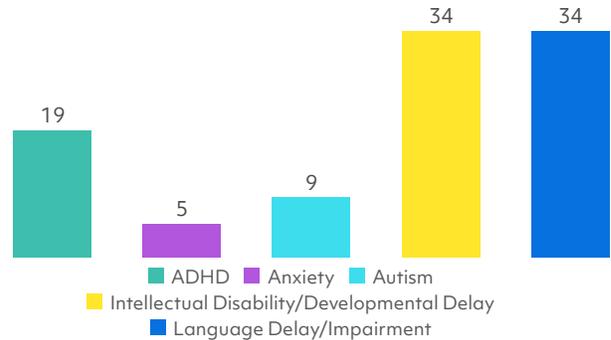
Data in these four graphs are from the medical history information collected in Simons Searchlight from 36 participants with SETBP1-related syndrome.

April 2025

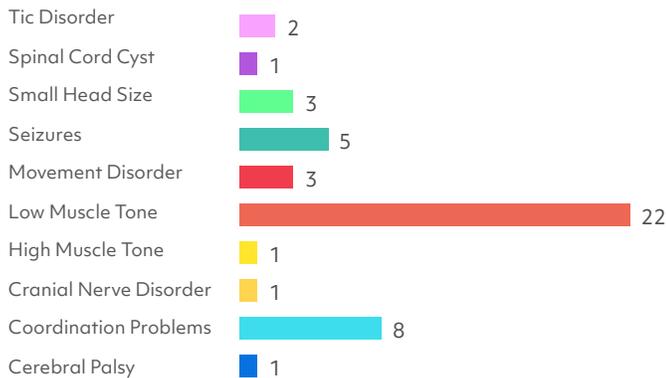
Ages in Years



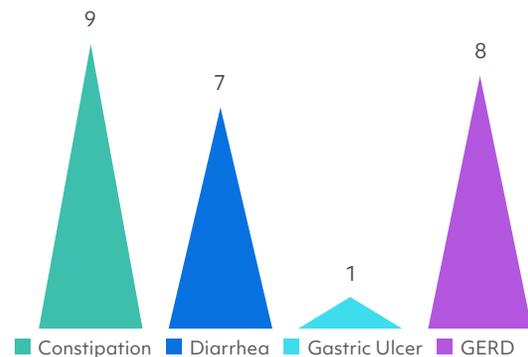
Developmental and Behavioral Conditions



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. Graphs include individuals with pathogenic or likely pathogenic variants, and without additional variants.

How to participate?

The information in this report is made possible by the active participation of the SETBP1 community! Progress for individuals in your community with SETBP1 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

113



STEP 2

Provide your genetic lab report

91



STEP 3

Share your important medical history

57



STEP 4

Fill out surveys

69



STEP 5

Provide a blood sample if you are interested

22



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.



**27 SETBP1 participants
contributed Vineland-3
data to this report**



Scan the QR code below or click the following link
(www.bit.ly/Searchlight_Vineland_Graph) for a video from Dr. LeeAnne Green Snyder, former Clinical Research Scientist for Simons Searchlight, on how to read the Vineland Adaptive Behavior Scales growth charts.



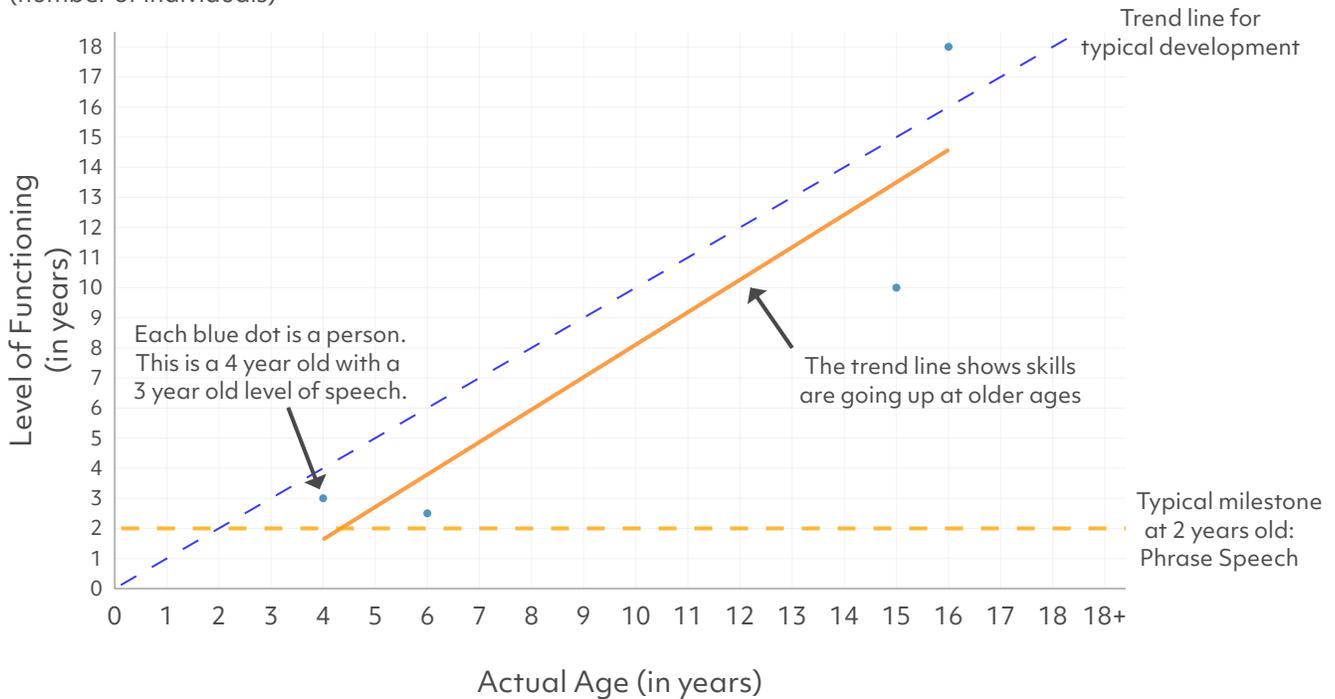
We will continue to collect Vineland data every year. Over time, we will have a better understanding of how individuals with SETBP1 change as they get older

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.

Sample graph:

Developmental Levels in Speech at Younger and Older Ages

Expressive Language Development (number of individuals)



What does this mean as my child gets older?

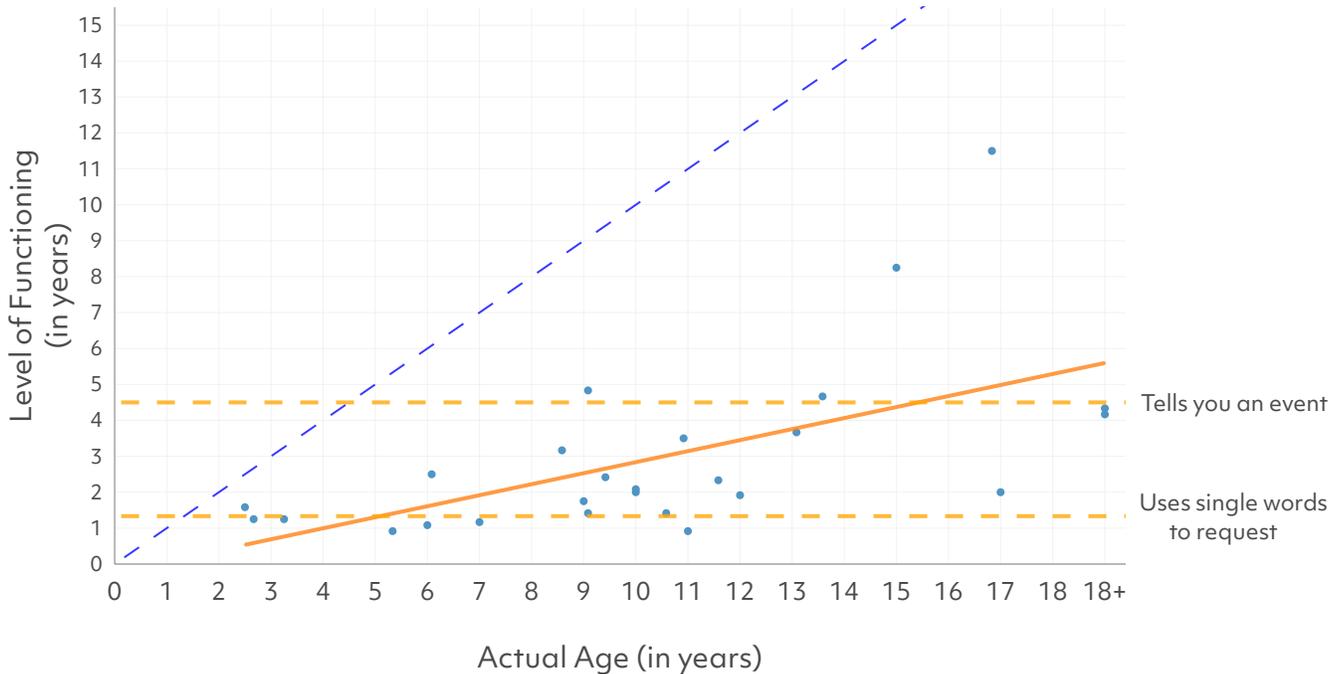
Along the bottom is the actual age of the person.

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 SETBP1

Expressive Language Development (26 individuals)



1 participant excluded as a potential outlier, defined here by level of functioning in years exceeding actual age in years.

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

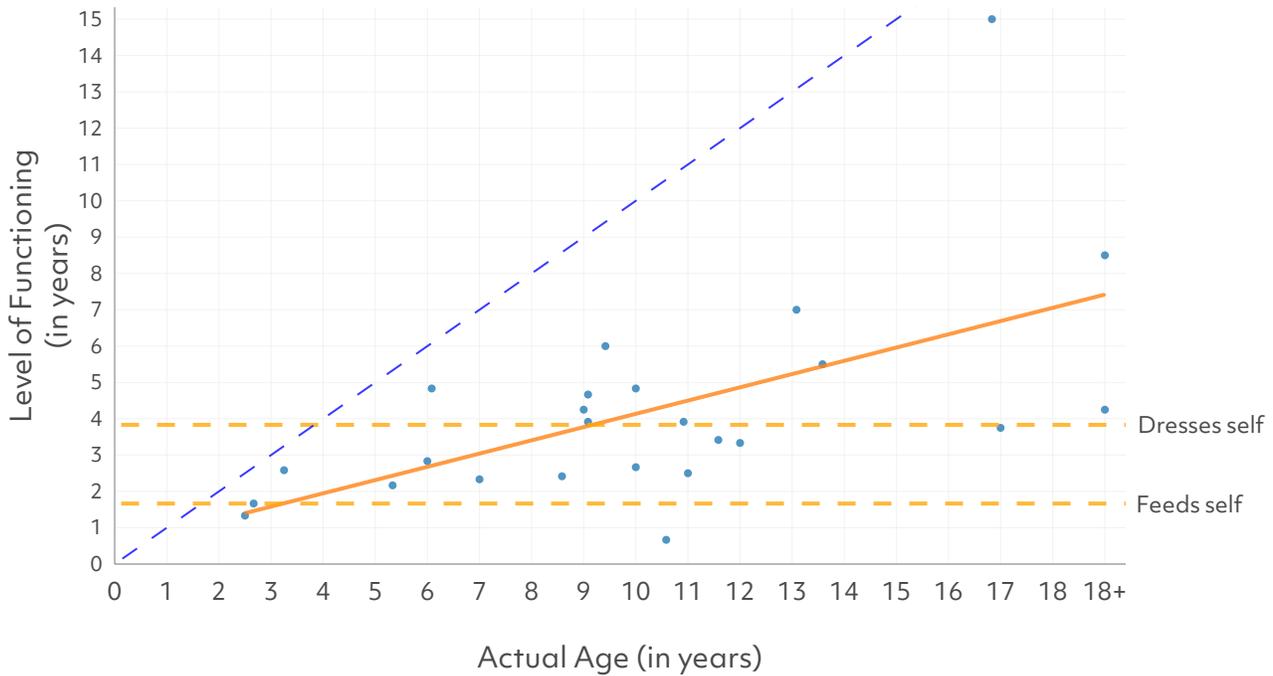
Along the
bottom is the
actual age of
the person.

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

83% of individuals
are likely to be
using language at
age 4 and older.

Developmental Levels in Self Care at Younger and Older Ages in SETBP1

Self Care Skills Development
(25 individuals)



2 participants excluded as potential outliers, defined here by level of functioning in years exceeding actual age in years.

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

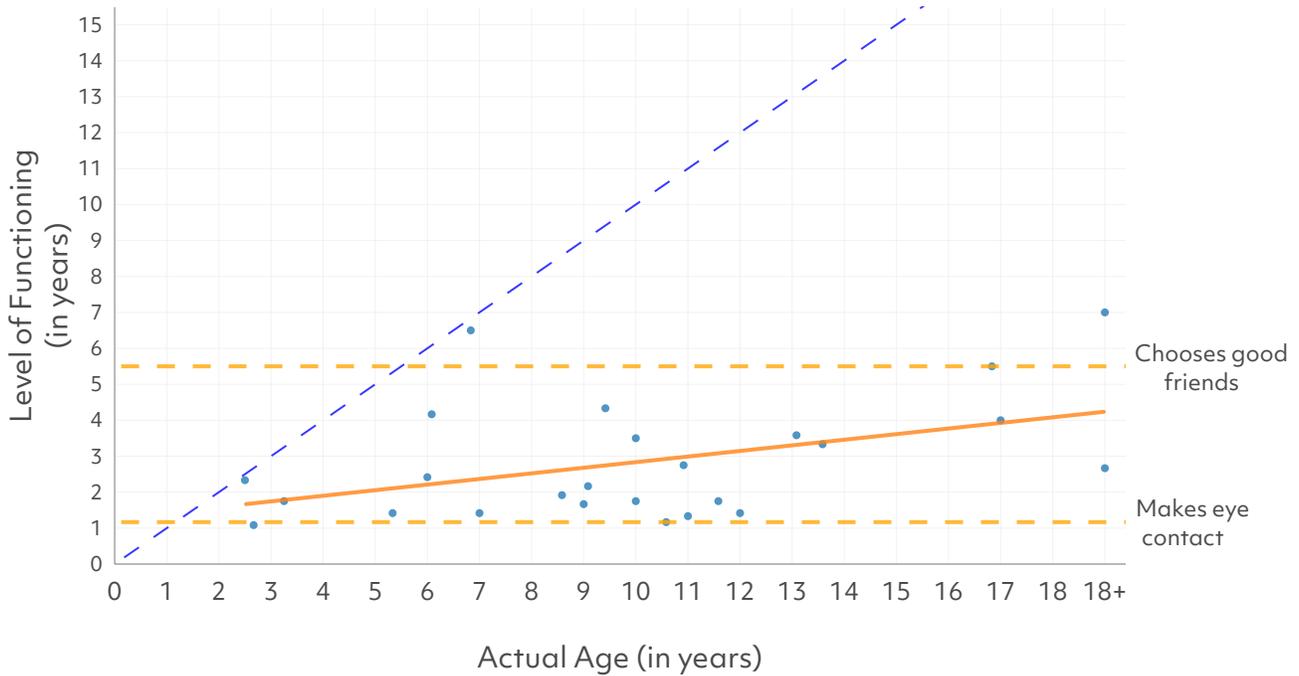
Along the
bottom is the
actual age of
the person.

The dots are
placed at the level
at which they can
do things for
themselves.

95% of individuals
are likely to be
feeding
themselves at age
4 and older.

Developmental Levels in Social Skills at Younger and Older Ages in SETBP1

Social Skills Development
(25 individuals)



2 participants excluded as potential outliers, defined here by level of functioning in years exceeding actual age in years.

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

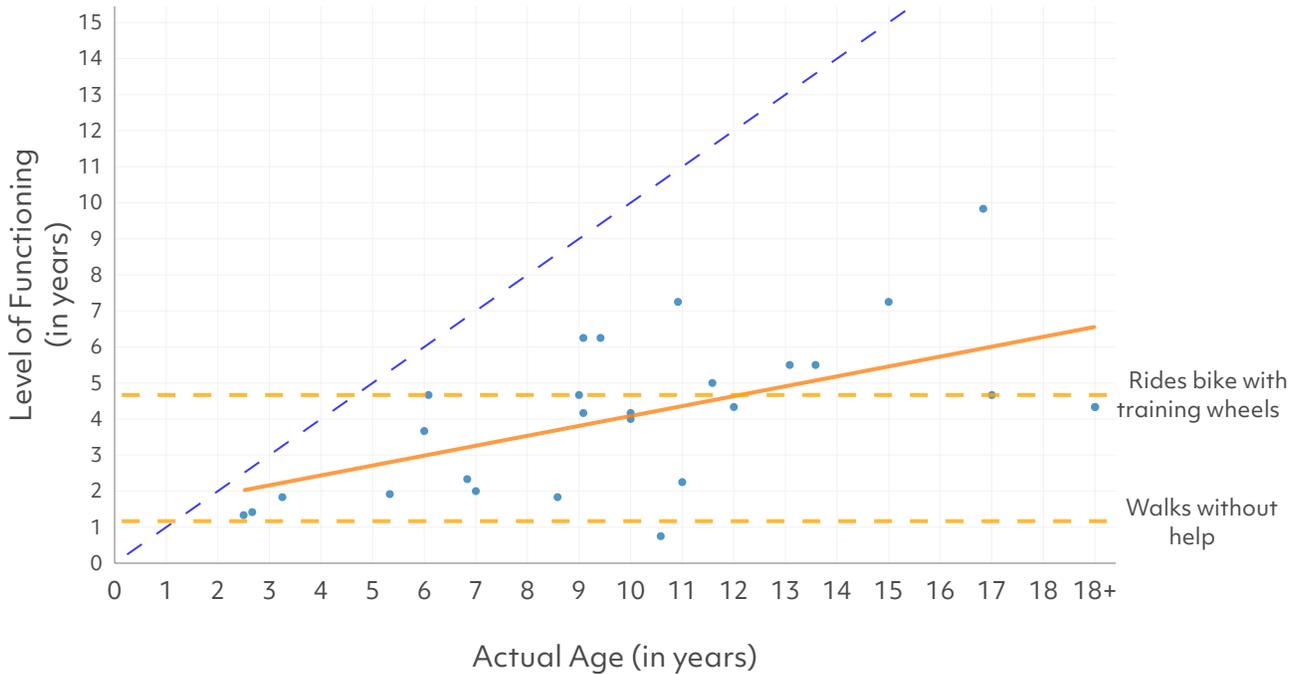
Along the
bottom is the
actual age of
the person.

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

100% 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 SETBP1

Motor Skills Development
(27 individuals)



**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.

96% of individuals
are likely to be
walking at age 4
and older.