

What Information SPARK Collects, and Why

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Acknowledgements

SPARK Families

SPARK Team

Clinical Sites

Libby Brooks, M.S.



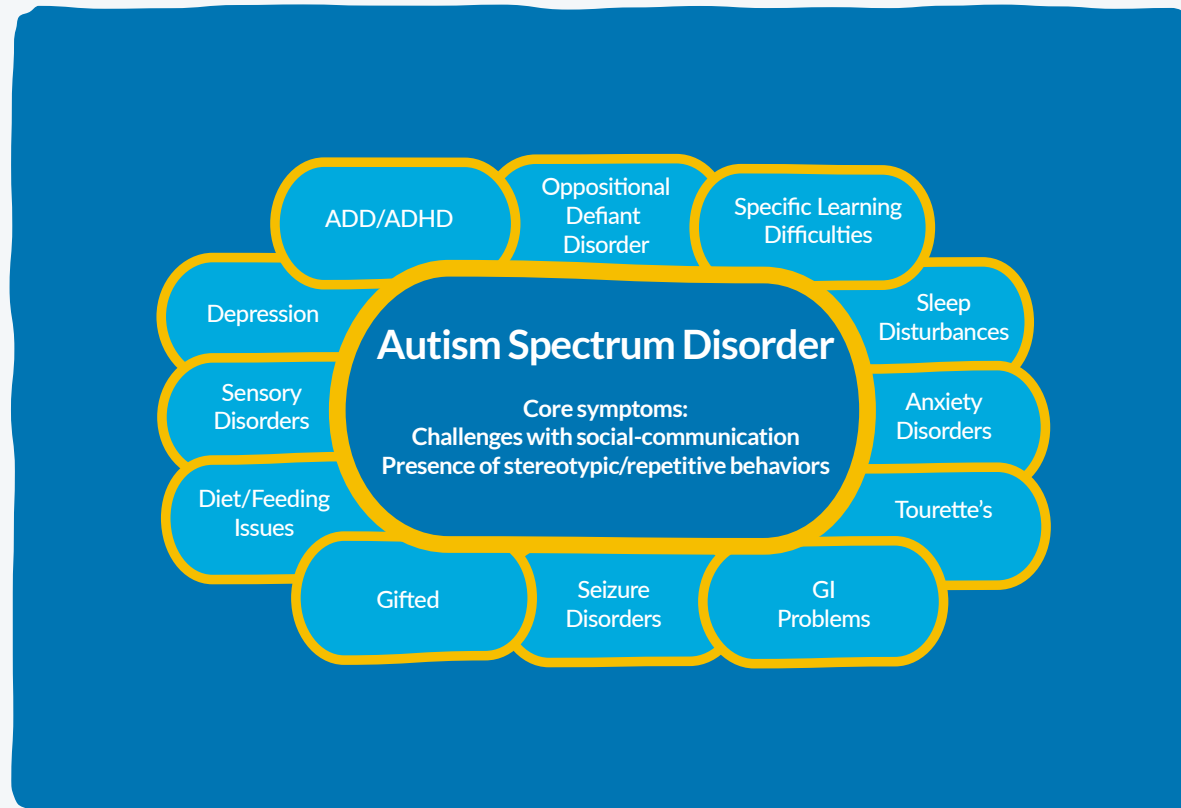
Agenda for today's webinar



- I. Define phenotyping
- II. Describe some of our research goals in SPARK
- III. Describe our measures and what they are about
- IV. Review some of our first findings in SPARK

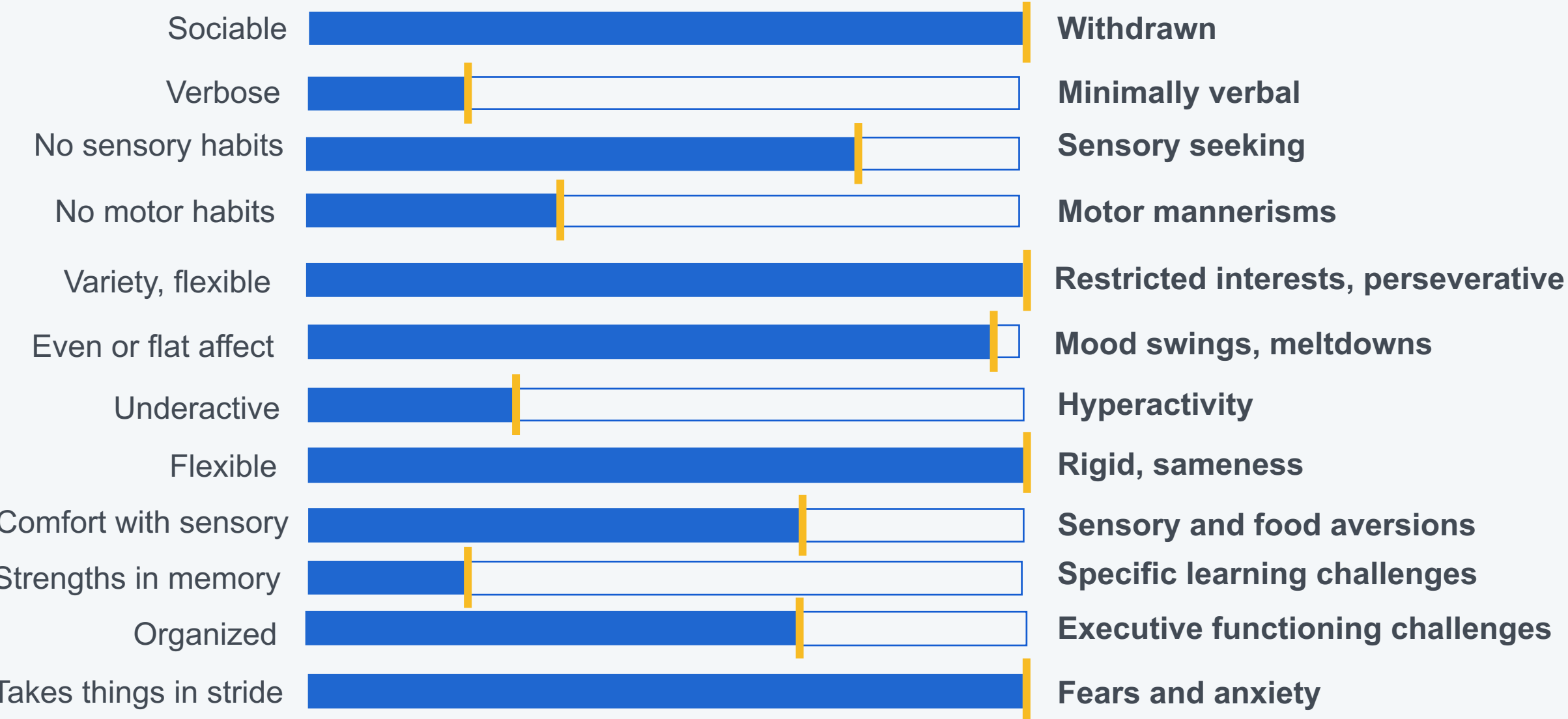
What is phenotyping?

- Phenotyping refers to measuring and describing the **traits** –behaviors, personality, abilities, medical conditions and appearance– that result from your genotype (our genes) and your environment.



An individual phenotypic profile

Common in ASD:



Finding Subtypes of ASD



→ Can we group individuals into “**types**” of ASD based on similar clusters of traits?

→ Do these types of ASD have different genetic bases, and different paths to well-being?

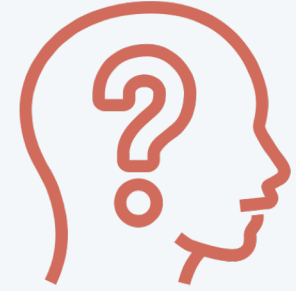
Summary: Goals of phenotyping

Autism is different for everyone, but many different types of people are lumped under the umbrella term of “ASD”



- How and why are some people with ASD different from others?- What is underlying these differences?
- Do some traits tend to **cluster** together, and do they link to particular genes?
- Are there other factors in the environment that interact with our genes to contribute to ASD?

A few of our questions in SPARK



- **Who** is in SPARK?
- How and why are **boys and girls** with autism different?
- What are **early signs** of autism, and how do **babies** with autism develop?
- What are **predictors** of improved language and independence?
- What other **medical** issues are common in autism?
- How do children with autism **change** as they grow into adulthood?
- What are the **needs of adults** with autism?

What information we collect through SPARK surveys

- Registration questions
 - Age when diagnosed
 - Who made the diagnosis
 - Current language level and cognitive level
 - Any services?
- Basic Questionnaire - Medical Screening
- Background History Form



What we collect through standardized measures

- What is a **standardized measure**?
 - A standardized measure is a questionnaire or test that aims to **combine or compare** many people on the same trait, and so it must use the **same** methods.
 - Same questions and same scoring for everyone
 - Better quality if information is measured or asked more than once, in different ways
- There are many published standardized tests out there!
- Current SPARK Standardized measures:
 - Social Communication Questionnaire – Lifetime (Rutter)
 - Repetitive Behavior Scale – Revised (Bodfish)
 - Coordination Questionnaire (Wilson)





Why do we collect these measures?

- Core **ASD** traits
- **Other** behavior diagnoses
- **Impact** of ASD behaviors
- **Markers** of rare genetic variants
- **Environmental factors**
- Demonstrate the strength of our diagnosis data
- Check if **siblings** have signs of ASD
- Other important factors

For example,

- Developmental level
- Socioeconomic status

SPARK Measures

- Diagnosis details
- Social Communication Questionnaire
- Repetitive Behavior Scale-Revised
- Social Responsiveness Scale (2020)
- Background History
- Basic Medical Screening Questionnaire
- Vineland Adaptive Behavior Scales (2019)
- Child Behavior Checklist (CBCL) - Adult (ABCL) (2019)

Where does your SPARK data go?

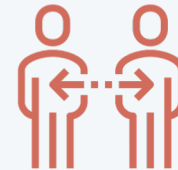
Clues to discovering **new autism risk genes**, to report to families and to the world



Data for the community:
Snapshots



Research Match



Approved researchers look for certain characteristics in individuals to join new studies

Share with **approved researchers** around the world, to make new discoveries

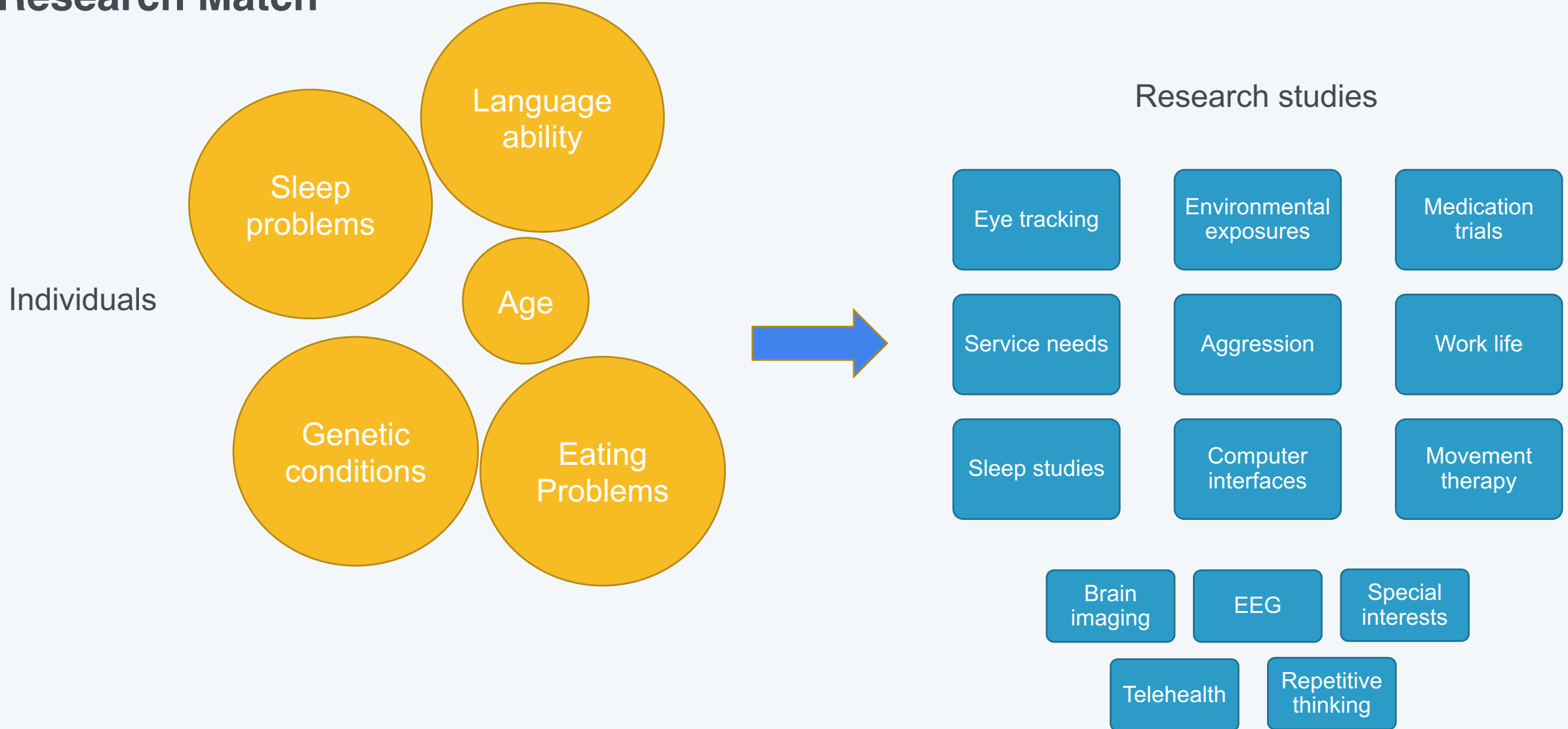


Ongoing SPARK phenotypic research



- Predictors of outcomes in adulthood
- Differences between males and females
- A method to rate the effect of genetic changes
- What factors affect parent stress

Research Match



SPARK Snapshots

Pregnancy, Birth History and Associated Conditions

Percent with Birth Complications

7%



SPARK families reported many concerns about pregnancy and birth for their children, but serious complications causing brain injury are no more likely than they are in the general population.

Percent Born Prematurely

12%



Premature birth occurs slightly more often in babies who are later found to have autism than it does in other babies. Birth complications such as prematurity are known to increase the risk for developmental disability, and some research says it increases the risk for autism.

Adults with autism also struggle with depression and anxiety. Independent adults with autism report depression and anxiety at over three times the rate of the general population.



Associated conditions reported by Dependent Adults with ASD

20% have a diagnosis of depression
33% anxiety disorder
40% ADHD

Associated conditions reported by Independent Adults with ASD

49% have a diagnosis of depression
39% anxiety disorder
40% ADHD

Pregnancy, Birth History and Associated Conditions

eating problems
cognitive impairment
sleep problems
anxiety
ADHD
depression

Associated Conditions in Children with Autism

5% have a diagnosis of depression
18% have anxiety
36% have ADHD
14% have cognitive impairment
59% have sleep problems
58% have eating problems

The rate of attention deficit (ADHD) in SPARK is over 3 times higher than it is in the general population. Diagnoses of conditions like ADHD and anxiety may rise now that the new diagnostic system used by doctors encourages all other conditions to be documented.

Over half of parents report that their child has problems with sleep and eating.

Seizures

The rate of seizure disorders in SPARK is higher than it is in the general population. But SPARK's reported rate is much lower than the rates reported by other autism studies. We need everyone in SPARK to finish their basic medical screening survey to give us a complete picture.



What are we finding in SPARK?



What have we collected in SPARK so far?

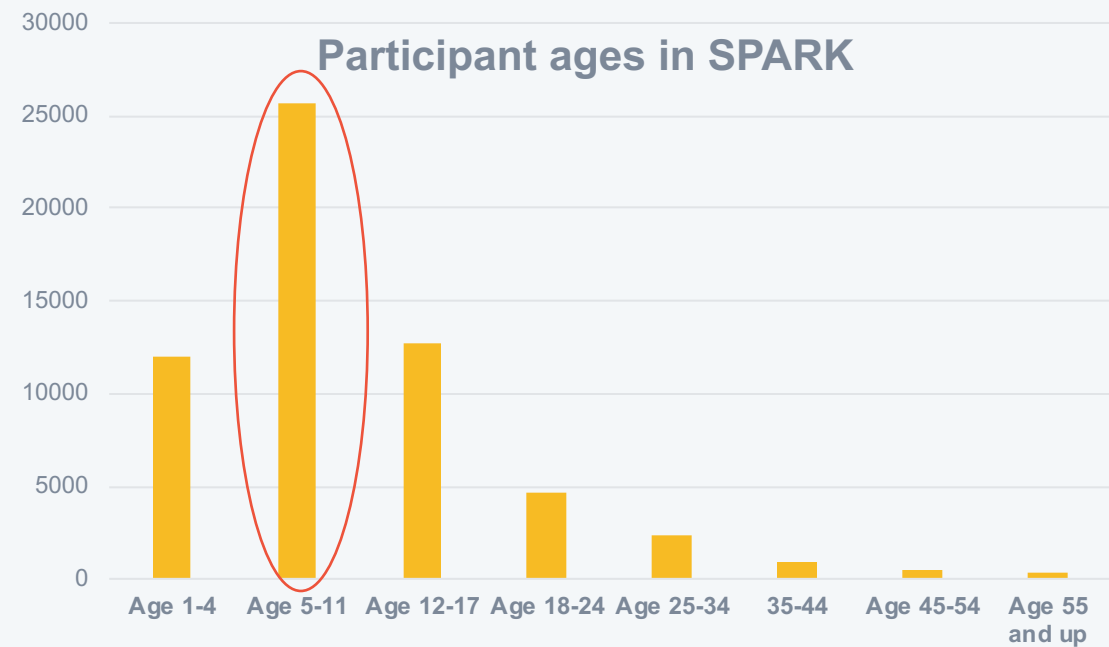
Measure	Purpose	Subject	How many families completed it	How many in our last Data Release
Round 1				
Social Communication Questionnaire-Lifetime	Core behavior ASD risk	ASD children & NASD sibs	65%	43,184
Basic Screening	Medical markers Complications	All family members	66%	90,549
Round 2				
Repetitive Behavior Scale-Revised	Core behavior RRBs	ASD children/dependents	51%	23,011
Coordination Questionnaire	Motor delays	ASD children	51%	16,705
Background History Questionnaires	Demographics Development Family background	ASD kids ASD adults NASD sibs	49% 54.5% 48.5%	23,488 1,660 10,302

Registration and Background History

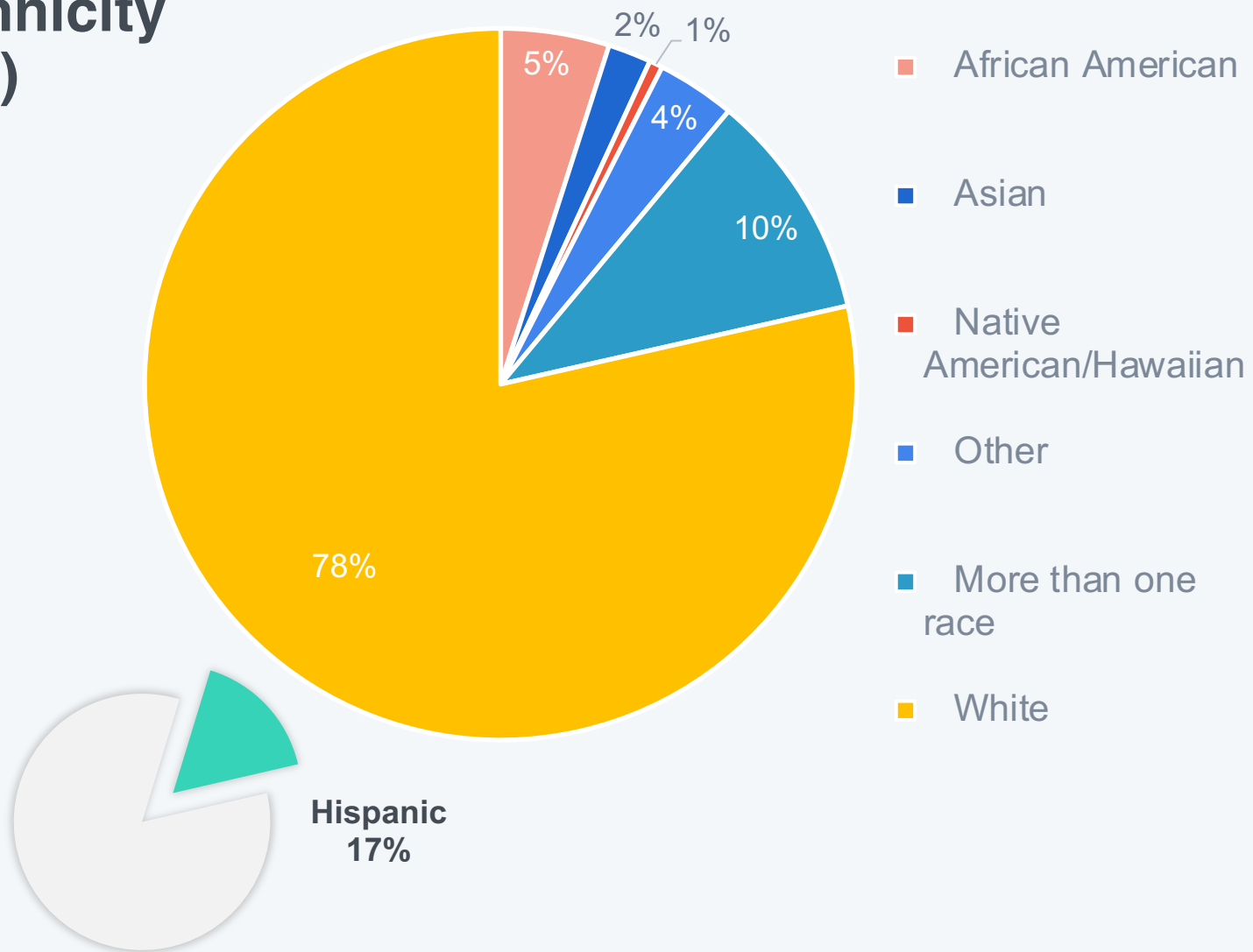


Who is in the SPARK community?

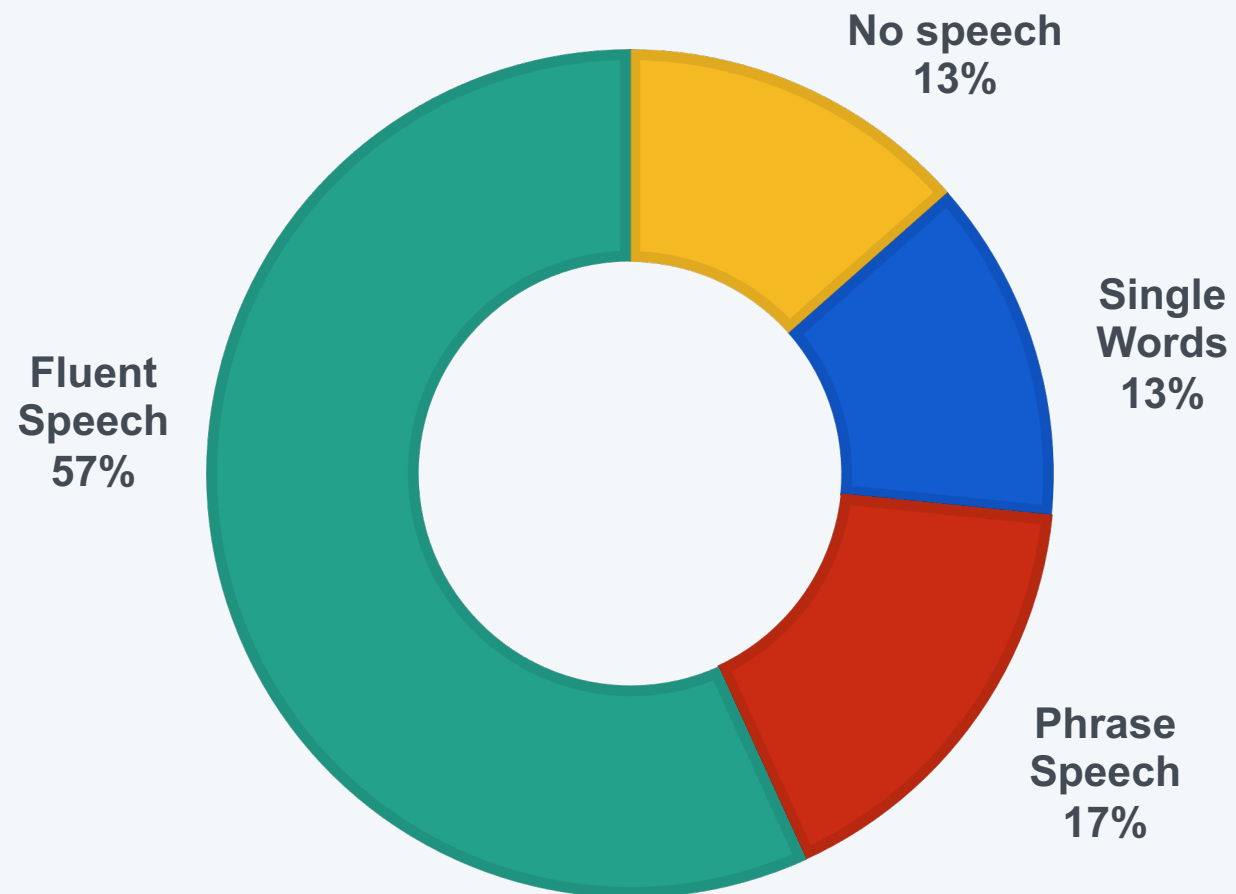
	n (%)
Everyone in SPARK	150,064
ASD (age 1-85 years)	59,218
Adults over 18	8,713
Adults with guardians	5,025
Adults without guardians	3,688
Children under 18	50,505
Male/Female probands (%)	80/20%
Individuals with intellectual disability	10,527 (19%)
Adults with guardians	2,241 (46%)
Children	8,286 (16%)
Minimally verbal children/dependents (single or no words)	14,432 (26.6%)
Multiplex families (2+ ASD members)	6,552
Siblings without ASD	24,076




Race and ethnicity (Total 25,281)





Language levels



Background History


Early development 


School and services 

Family history of ASD 

Adult supports 

Employment 

Relationships 

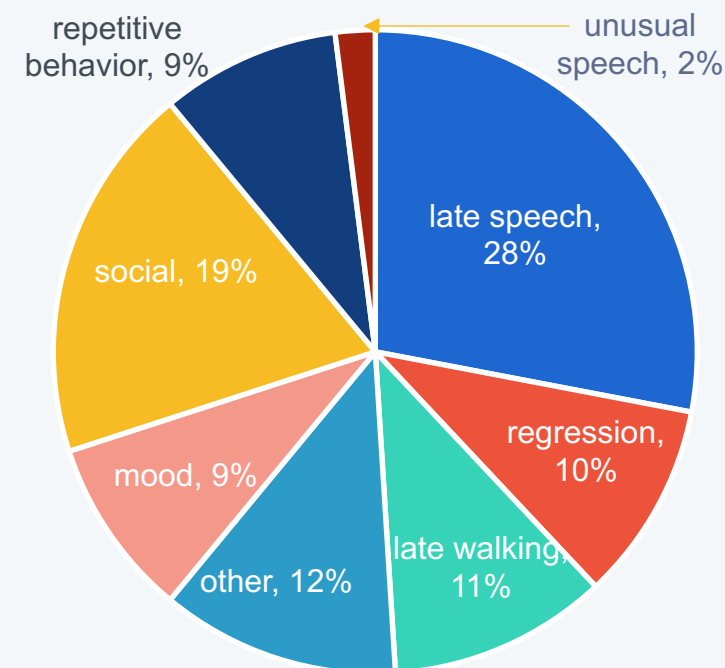
Interests and hobbies 

Early development in 21,253 Children with ASD

	Average age	On time?
Sitting up	6 months	✓
Walking	14 months	✓
Single words*	21 months	
Phrase speech (over age 2)	33 months	

- Some children who were late to talk were still able to learn after age 7

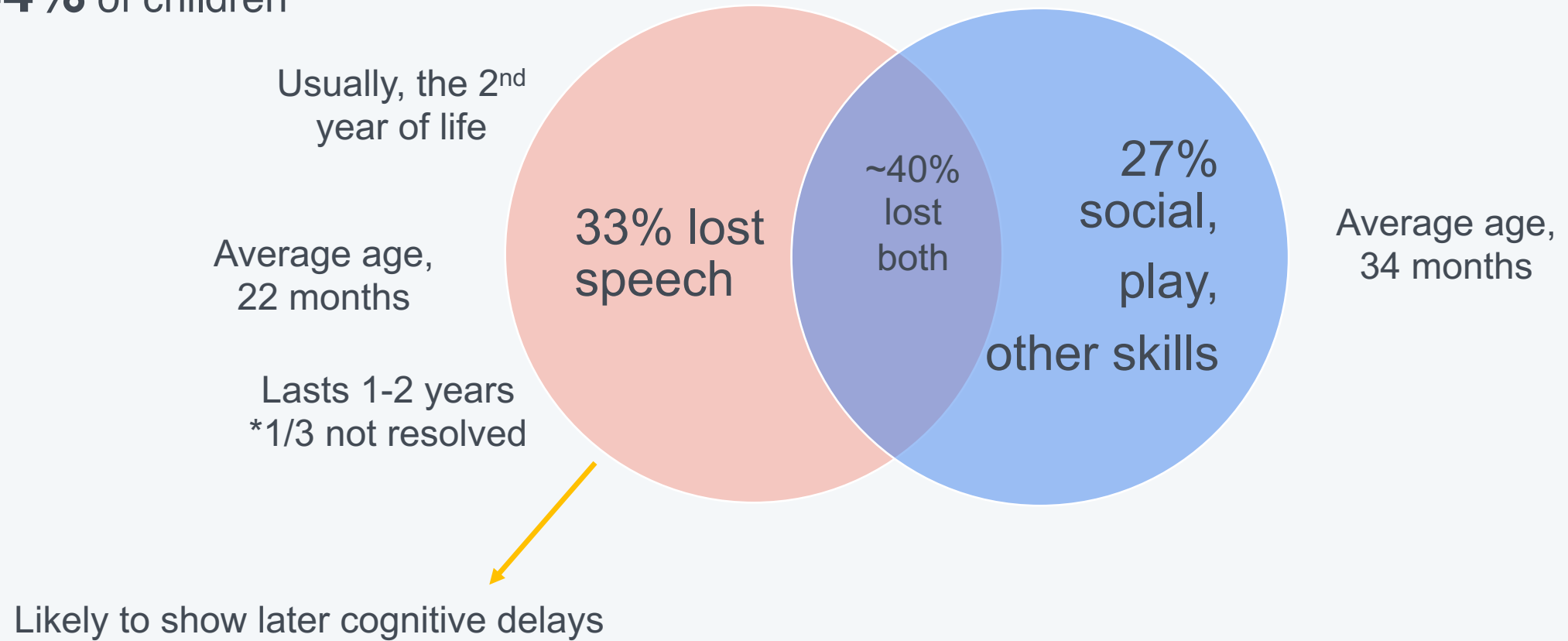
First concern



- Typically in the 1st or 2nd year of life
- Average age of first concern was **22 months**

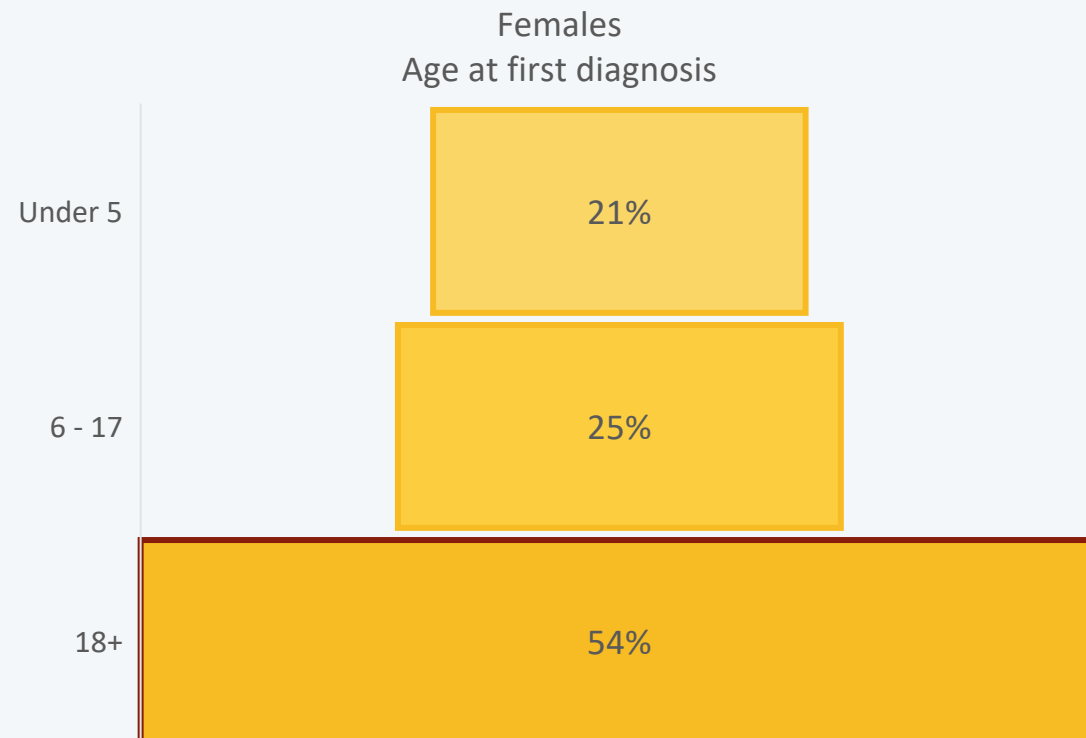
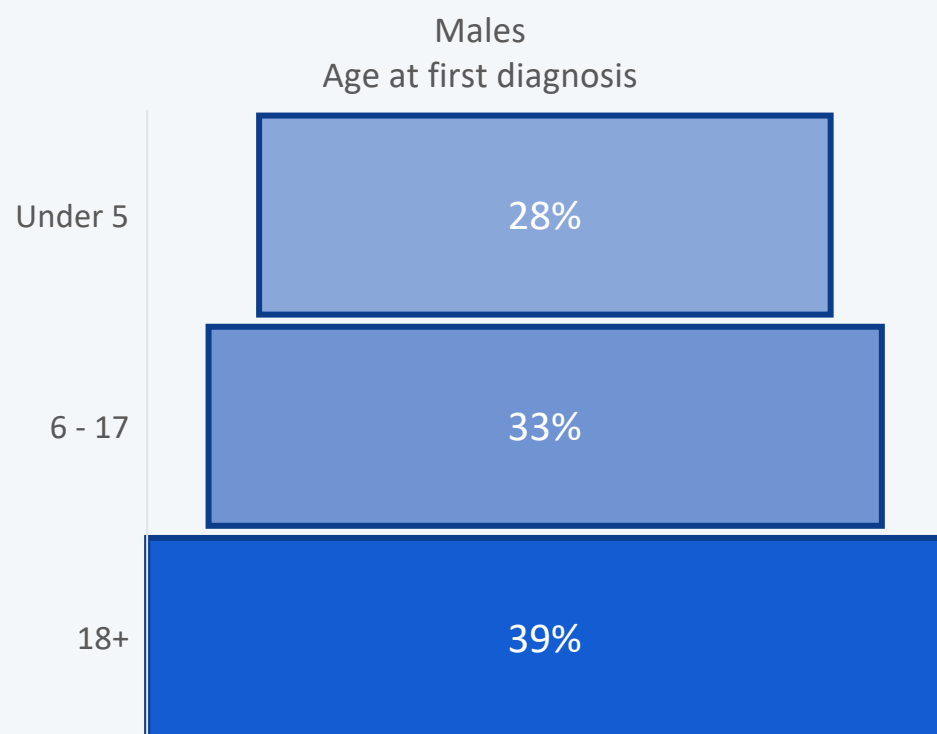
Regression

- **Affects 44%** of children



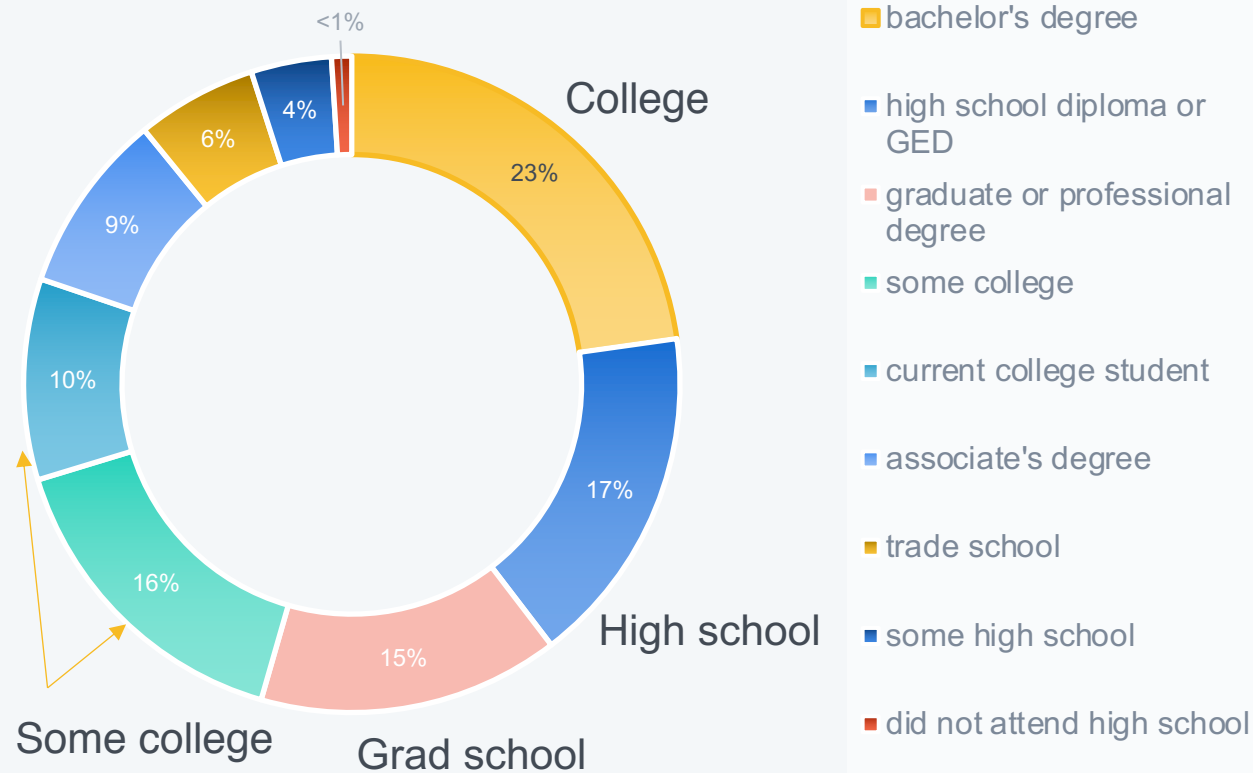
Adults in SPARK

Age at first diagnosis

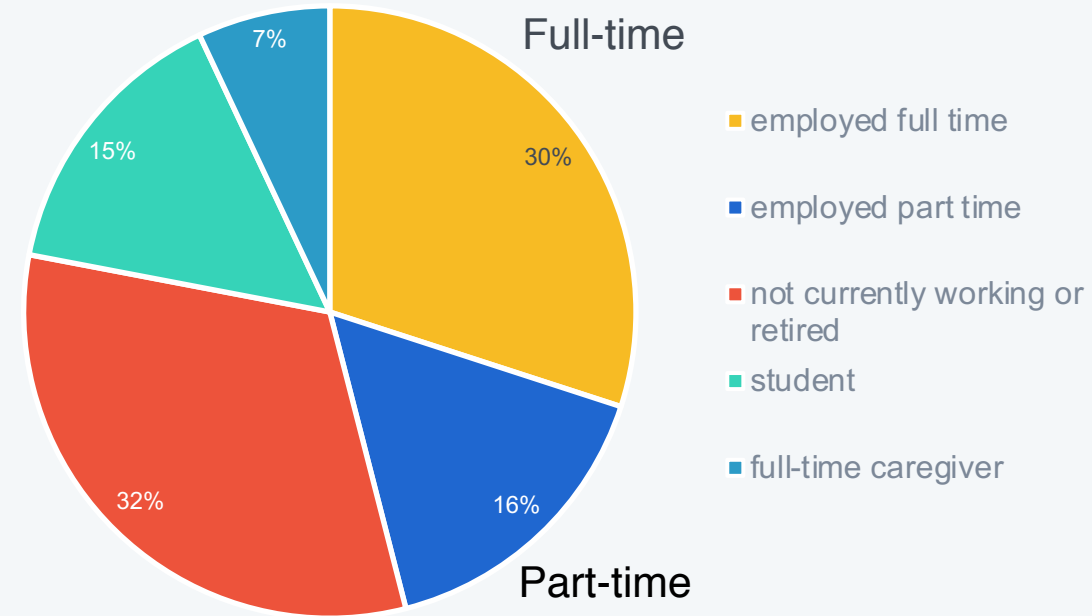


Adults in SPARK Education & Employment

Education Level



Employment



1,660 adults

Basic Questionnaire: Medical Screening



Basic Questionnaire

Medical Screening

I. Medical and environmental complications

- **Pregnancy & birth** complications: prematurity, insufficient oxygen, brain hemorrhage, alcohol or drug exposure
- Severe **vision or hearing** impairments
- **Neurological problems:** traumatic brain injury, brain infection (such as meningitis), lead poisoning



II. Major medical issues that commonly occur in **genetic syndromes**

- Seizures
- Growth conditions – diagnosed short stature, underweight, obesity, microcephaly and macrocephaly
- Birth defects



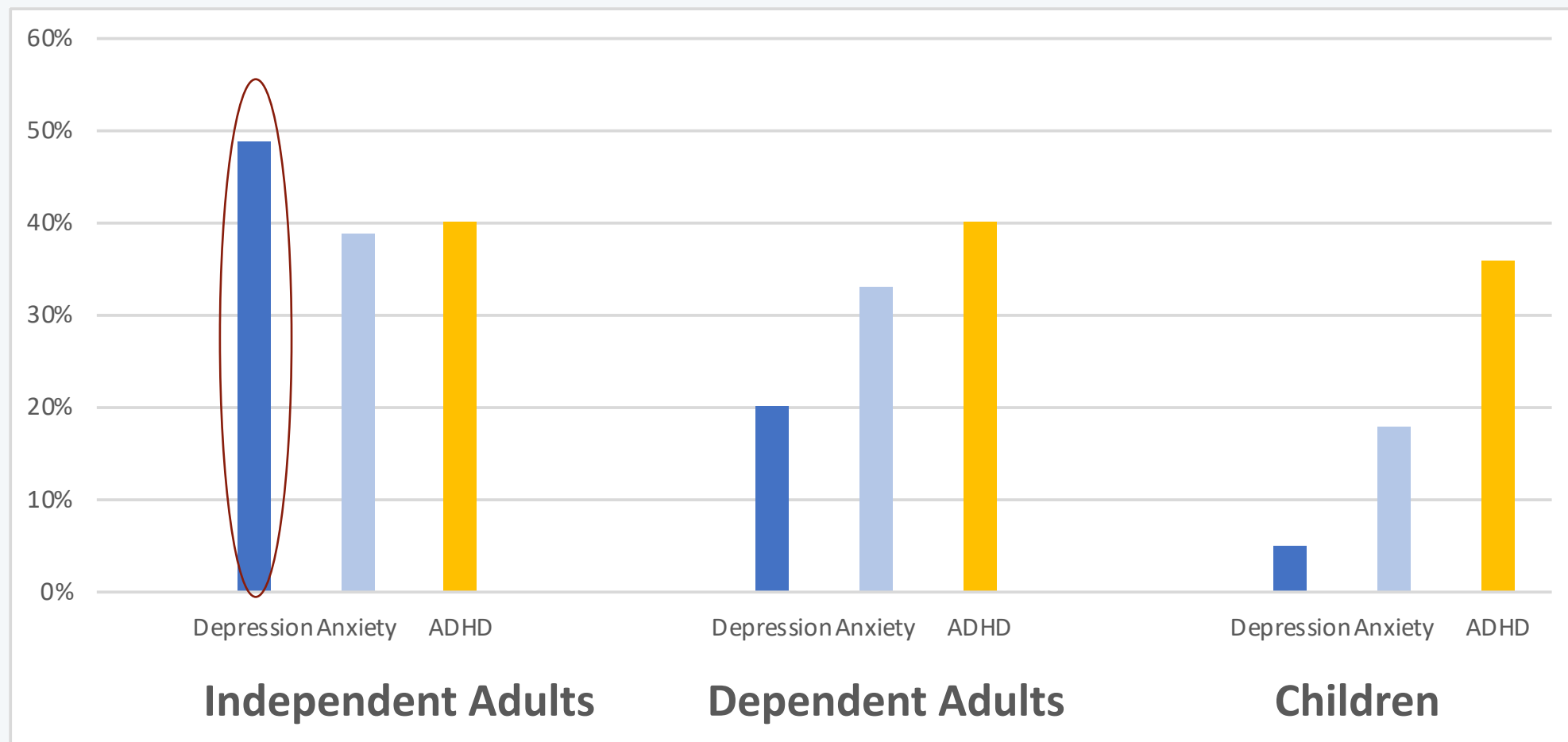
III. Other diagnoses

- For example, development, language, ADHD

IV. Previous **genetic diagnosis** or genetic study



What we've learned about other diagnoses



Current Standardized Measures:

Social-communication development

Repetitive behavior

Motor coordination



Social-Communication Questionnaire (SCQ)



What is it?

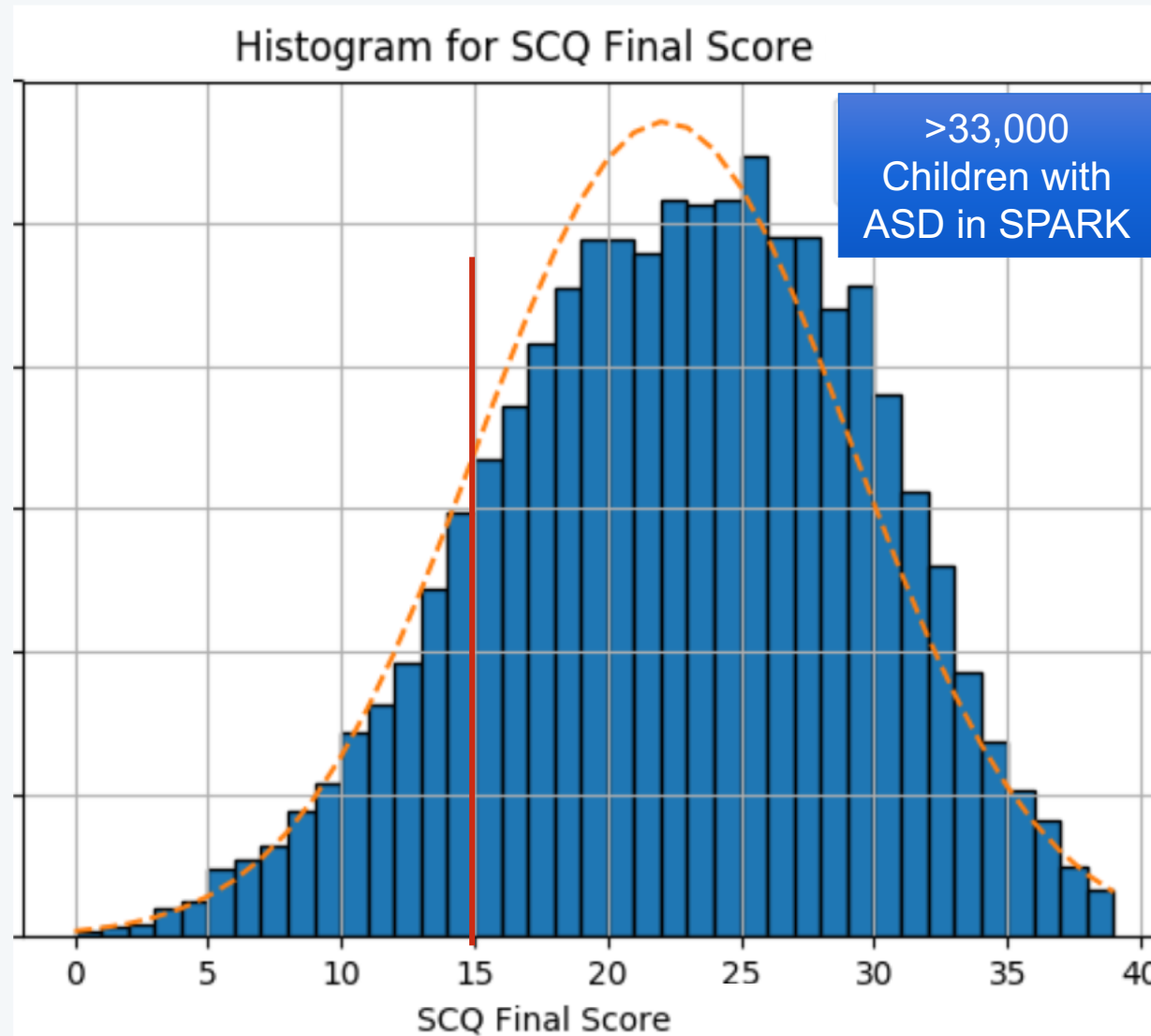
- Parent report of social, communication, and play behaviors both currently and in a past critical period (age 4)

Why collect the SCQ in research?

- ✓ Commonly used by researchers to confirm the presence of **signs** of autism in participants
- ✓ Shows 2 major types of **traits** in individuals: social-communication, and repetitive behavior
- ✓ We can look at how these traits, and how the overall number of signs, link to genetics and heritability (for example, in twins; Frazier et al., 2014)
- ✓ We can look at how these traits can predict other abilities

Social Communication Questionnaire

- 86% of children with ASD in SPARK are at or above 15



Repetitive Behavior Scale-Revised (RBS-R)

What is it?

- Measures the **amount** and **impact** of repetitive behaviors and special interests seen in children and adults with autism
- Includes body movements, special routines, and fascination with certain subjects



Why study the RBS-R?

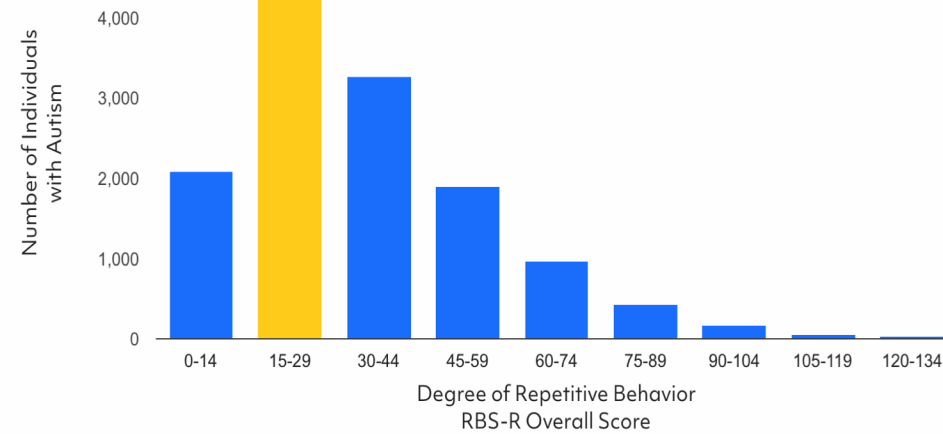


- ✓ One reliable subtype - **insistence on sameness** (IS) - is considered unique for genetic research: it may occur regardless of your other autism traits, language level, or IQ (Hus, Pickles, Cook, Risi, & Lord, 2007) (Bishop, Richler, & Lord, 2006; Cannon et al., 2010; Cuccaro, et al., 2003; Richler, Huerta, Bishop, & Lord, 2010; Szatmari et al., 2006)
- ✓ Insistence on sameness has been linked to specific **genetic** findings and runs in **families** (Buxbaum et al., 2001; Cannon, et al., 2010; McCauley et al., 2004; Shao et al., 2003; Silverman et al., 2008; Sutcliffe et al., 2005; Abramson et al., 2005; Silverman et al., 2002; Szatmari, et al., 2006)

Repetitive Behavior Scale

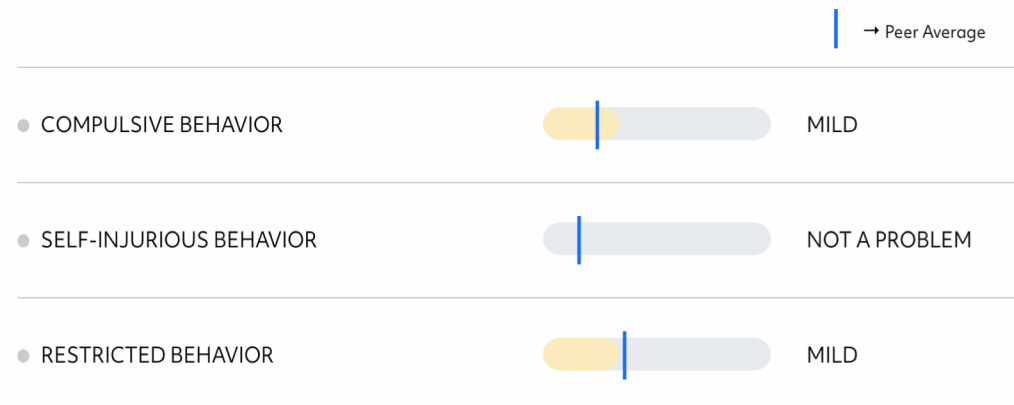
Individual Results (Sample)

- SPARK children have a high number of significant repetitive behaviors



TIGER'S RESULTS ?

You were asked to rate each behavior on the RBS-R as a mild, moderate or severe problem. The behaviors are then grouped into six major categories. The results below show how you rated Tiger's behaviors, and what other parents said in SPARK. When behaviors are more severe, parents may find it helpful to talk to their doctor for a referral to a behavior specialist.

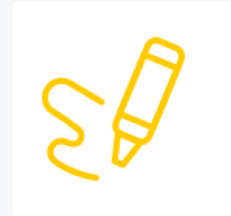


Coordination Questionnaire



What is it?

- Parent report of specific **fine motor** and **gross motor** skills (such as writing and running)
- Clinicians compare a child's score to what is expected for typically developing children of the same age, to screen for motor delays



Why study motor function in autism?

- ✓ Basic motor skills allow children to explore the world and interact with others
- ✓ Very early fine motor skill delays in infancy predict the extent of autism traits later
- ✓ Motor delays in infancy often go along with speech delay (Landa)
- ✓ In one of the first large genetic studies in autism -the Simons Simplex Collection- delays on the Coordination Questionnaire correlated most strongly with genetic changes (Buja)

We've only just begun to understand the importance of motor function in autism.

Coordination Questionnaire: Children with ASD in SPARK who have motor delays

How many children were reported to have delays in each age group?



How your data is helping research: Beyond SPARK



Researchers around the world are using SPARK data to study....

- Single genes that link to specific ASD behaviors
- Role of the oral microbiome
- Genetic syndromes
- Other issues, like ADHD
- Predicting risk factors for ASD
- Predicting risk for genetic variants using phenotype/traits
- Sex differences
- Motor skills
- Social-emotional skills
- Repetitive behaviors
- Neurological conditions



What's Next?



SPARK Measures coming to a dashboard near you

Social Responsiveness Scale

(John Constantino)

Core ASD social and repetitive behaviors

Vineland Adaptive Behavior Scales

(Celine Saulnier & Sara Sparrow)

Development and everyday skills

Children's Sleep Habits Questionnaire

(Judith Owens)

Modified-Checklist for Autism in Toddlers

(Debby Fein & Dianna Robbins) Early signs

Sensory Profile (Dunn)

SPARK Annual Update

Child Behavior Checklist Adult Behavior Checklist

(Achenbach)

Other behavior issues, attention and mood

Quality of Life

(World Health Organization, ASD-UK Newcastle)

Autism Impact Measure

(Steve Kanne & Micah Mazurek)

Core ASD social and repetitive behaviors

Aberrant Behavior Checklist

(Aman)

Challenging behaviors

Communication & Symbolic Behaviors Scale

(Amy Wetherby)

Early signs

Thank You

