What SPARK Gives Back to Its Participants

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Today’s talk

- What is SPARK?
- The process for return of genetic results
- SPARK’s current genetic research findings
- SPARK’s research matching program
- SPARK’s data reports and web content
My journey

- Diagnosis Day: July 27, 2007
- Diving into ASD research: September 2013
- SPARK Launch! April 2016
What is SPARK?

- We are a growing research community of individuals, families, and researchers on a mission to speed up the understanding of autism. Our aims are to:
  - Collect DNA and medical/behavioral information from 50,000 people with autism and both of their parents
  - Share that data freely with qualified researchers and connect our participants with other autism research studies
  - Discover breakthroughs that were never possible before
  - Return our findings to you
We are not looking for a “cure” for ASD.

We are only able to return genetic results to 10-20% of participants; this will increase over time as we learn more.

We are not developing genetic tests for autism. However, data from SPARK could be used to inform such tests.
Who is eligible to participate?

Individuals and Families with ASD are eligible if they:

1. Live in the United States
2. Are English speakers
3. Are an independent adult with ASD, or the parent of a child or dependent adult with ASD. Biological siblings are welcome too.
4. Have a professional diagnosis of autism
Your data is encrypted at all times in a secure database.

Your data is stripped of personally identifying information when shared with researchers.

Researchers can only access your data if they agree to protect your data and only use it for scientifically valid purposes.

SPARK holds a Certificate of Confidentiality from the NIH that further prohibits access to your data (by non-researchers) without your consent.

SPARK will inform you of other research studies, but you control what studies you want to participate in.
What Participation in SPARK Looks Like

- Easy to enroll online
- Choose how to participate
- Be contacted about new research opportunities
- Stay connected
Participation in SPARK

53,238 individuals with ASD
80,564 saliva specimens collected
209,447 questionnaires completed
How will SPARK accelerate better research?

Recruit 50,000 families with ASD, collect phenotypic & genetic info

Return genetic results so participants can be grouped by genetic similarities or phenotypic traits

Match participants to research studies
ASD is both genetic and environmental

Identical Twins 77%
Fraternal Twins 31%
Siblings 20%
How much of autism is genetic?

- **Genetic factors** play a large role in ASD (50% – 80%)
- **Large population study (2017)**
  - 3.5 million children in Sweden
  - Genetic contribution to ASD = 80%

- **Environmental Factors**
- **Combined interactions**

*Sandin et al. 2017*
What have we learned??

1970s Twin studies show autism is highly heritable

1991 Gene for Fragile X Syndrome discovered (FMRP)

1999 Gene for Rett syndrome discovered

2012 Total known genes explain ~5% of ASD

2018 Gene therapy clinical trial for Rett syndrome soon, new genetic therapy in Fragile X mouse leads to improvements

2018 Total known autism genes explain ~10-20% of ASD
Some genetic changes are NEW (de novo)
Some genetic changes are “X-linked”
Some genetic changes are inherited

Genetic change can come from mildly affected or unaffected parent

Inherited genetic changes

Child with genetic change in autism gene
How do we identify genetic results?

Variants found repeatedly in individuals with ASD but almost NEVER in people without ASD likely cause ASD (e.g. de novo)

Genetic variants in individuals with ASD

Genetic variants in people without autism (Thousands in each person)

Variants found repeatedly in individuals with ASD but rarely in people without ASD also can cause ASD (e.g. inherited)
**SPARK Genetics: DNA Extraction → Sequencing → Analysis**

- **Completed analysis: 457 families (all “trios”)**
  - 10.4% of families with a returnable result, 3.4% with a potential results

- **7,556 families in analysis**
  - 2,489 “trios” (mother, father, offspring with ASD)
  - 2,793 “quads” (mother, father, 2 offspring)
  - 2,274 parent-child duos (parent, offspring with ASD)
  - 13 families >4

- **6,528 families in progress**
  - 3,447 “trios”
  - 861 “quads”
  - 637 parent-child duos
  - 1583 individuals with ASD

- 48 families have received/will receive results (2018)
- ~750 families will receive results (2019)
- ~650 families will receive results (2019-20)
Analysis of 460 families

- We found results in 48 families (10.4%) in known ASD genes.
- We found genetic variants that are POSSIBLE results in an additional 18 families (4%).
- When we are more confident of these POSSIBLE results we will return them, but we need more data.
- We expect to keep identifying genetic results in ~10% of families.
- This will increase as we get more data.
- Discovering new autism genes is most effective if we have DNA from parents and siblings.

Genetic results in 10.4% of completed families
Participant Story: Return of Genetic Results

https://www.youtube.com/watch?v=OaihZ-5ouAE
My story

- Diagnosis Day: March 29, 1996
- Diving into ASD research: Launched the Interactive Autism Network (IAN) in 2006
- Moving forward: IAN is merging with SPARK to work toward a common goal of advancing autism research

Kiely Law and Isaac Law
Reporting results back to you

- **Snapshot 1:** Who is participating in SPARK?

- **Snapshot 2:** Independent adults with ASD in SPARK
Snapshot 1: Who is participating in SPARK?

Summary data from our first 18,000 participants with ASD
SPARK Participants - enrollment by sex (at birth)

Children with ASD

4 to 1
Ratio of Males to Females in Children Under 18

Adults with ASD

2 to 1
Ratio of Males to Females in Adults Over 18
SPARK participants - age of ASD diagnosis

Children with ASD

- Average Age When Boys Were Diagnosed: 4.1
- Average Age When Girls Were Diagnosed: 4.4

Adults with ASD

- Average Age When Adult Males Were Diagnosed: 14
- Average Age When Adult Females Were Diagnosed: 15.8
SPARK participants - language development

Children with ASD in SPARK

- 58% use longer complex sentences
- 17% use phrases
- 13% do not speak
- 12% are able to use single words to communicate
SPARK participants - other medical conditions

**Children with ASD**
- 5% have a diagnosis of depression
- 16.5% have anxiety
- 32% have ADHD
- 16% have cognitive impairment
- 19% have sleep problems
- 41% have eating problems

**Adults with ASD**
- 29% have a diagnosis of depression
- 35% have anxiety disorder
- 35% have ADHD
Snapshot 1: Additional topics

Percent Born Prematurely
12%

How Many People Have Had Genetic Testing Related to Autism in the Past

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.

1,917 - or - 16.6%

Most Common Genetic Findings Reported by Families in SPARK

SPARK
Snapshot 2: Independent Adults with ASD

SPARK SNAPSHOT
Independent Adults with Autism

Data from December 2015 to April 2018

1,941
Number of independent adults with autism enrolled in SPARK
Independent adults – enrollment by sex (at birth)

Sex (at birth) of independent adults with autism enrolled in SPARK

1,030 (53%) Female
911 (47%) Male
Independent adults - age

Ages of independent adults with autism enrolled in SPARK

- 502 people, age 18-24
- 650 people, age 25-34
- 390 people, age 35-44
- 251 people, age 45-54
- 148 people, age 55+
Independent adults - education

Education Independent adults with autism enrolled in SPARK

- Did not attend high school: <1%
- High school diploma or GED: 17%
- Associate’s degree: 6%
- Some college: 9%
- Bachelor’s degree: 10%
- Some college: 16%
- Graduate or professional degree: 23%
- Bachelor’s degree: 15%
Independent adults - sexuality

**Sexual orientation**

Independent adults with autism enrolled in SPARK

- **60%** Heterosexual
- **6%** Homosexual
- **8%** Bisexual
- **4%** Pansexual
- **10%** Asexual
- **6%** No response
- **1%** Queer
- **1%** Other
- **4%** Don’t know
Snapshot 2: Additional topics

**Lifestyle**

**Household income**

Employment income (e.g., salaries, wages, etc.) reported by independent adults with autism enrolled in SPARK is lower than the national average for employed people. Other autism studies have found this as well.

- 39% Under $20,999
- 17% $21,000-$35,999
- 11% $36,000-$50,999
- 20% $51,000-$100,999
- 13% Over $101,000

**Marital status**

Just over half the independent adults with autism enrolled in SPARK are single. Many are married or in a relationship. Only 6% are divorced.

- 51% Single
- 27% Married
- 6% Divorced
- 16% In a relationship / domestic partnership

**Employment**

Nearly half of the independent adults with autism enrolled in SPARK are employed either full time or part time. The unemployment rate among independent adults with autism enrolled in SPARK — that is, the proportion who are seeking but have not found paid work — is 22%. More female than male participants report being full-time caretakers.

**Housing**

- 22% Living alone
SPARK Research Match – Sharing new study opportunities

1. We let you know about other research studies
2. You decide if you want to participate
3. Your participation leads to new research findings
4. We send you updates about study results
Many different studies are looking for participants

Types of studies
- Online and in-clinic
- Local and national

Many different topics
- Treatments and therapies
- Brain imaging
- Early diagnosis
- Twins
- Baby siblings
- Environment
- Medications
- Special interests
- Sleep and Diet
Study Title:
Genes and Environment Autism Research Study (GEARS)

Study goal
To better understand the complex interactions of genes and environment (before, during, and just after pregnancy) that may cause autism.
Over 1,200 families participated in the online study which will also use SPARK genetic data.
Preliminary Report – environmental factors

Child’s birthplace

- Over 1,200 mothers of children enrolled in SPARK, ages 2 to 12 from across the U.S., completed the online GEARS survey. This was a tremendous response – more than 60% of those invited.

- Prior research has shown that some types of environmental exposures and other factors just before and after birth are linked to autism.

- GEARS participants reported their child’s birthplace as part of the survey. The GEARS team will use this information, along with those participants’ genetic and environmental data, to look for potential causes of autism.
Preliminary report – prenatal Factors

Medical problems and complications during pregnancy

- Everything that happens during pregnancy can affect a baby’s growth and development.

- Prior research has shown that medical complications during pregnancy may increase the risk of autism.

- GEARs participants reported many details about their pregnancy. The GEARs team will use this information to look for clues on how these earliest changes in a child’s environment (the womb) may interact with genes and potentially lead to the development of autism.

Fever during pregnancy

25%

One or more medical problems or pregnancy complications (examples: gestational diabetes, premature labor, high blood pressure)

44%
Your feedback is important

- How important is this research topic to you?
- How was your experience participating in this study?
- Any additional comments?
Advancing research together

- 37 studies are partnering with SPARK and YOU to accelerate research.
- To date, over 35,000 SPARK participants have been invited to join at least one outside study.
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Thank you!