SPARK’s Past, Present, and Future:
Moving Autism Research Forward

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SPARK, Past to Present:

What has SPARK accomplished since its inception?
What is SPARK’s mission?

1. To better understand the causes of autism and to help improve lives.

2. To unite the entire autism research community.
What have we achieved?

Built an unprecedented research community:

• 30 clinical sites
• 333,184 participants, including individuals with autism and first-degree family members
• 139,275 research participants with autism
• 113,267 children 25,842 adults

Accelerated research:

• Since launching Research Match in 2017, SPARK has recruited over 58,000 SPARK participants and families for nearly 300 research studies
• More than 300 scientists used SPARK data
• Over 130 scientific papers published using SPARK data or recruiting SPARK participants, covering a range of topic from motor impairments, to mental health, to rate genetic variants
• Helped researchers discover >100 autism genes
• Advancing understanding of aggression, autism across the lifespan, camouflaging, LGBTQIA+ issues, and more…
What has SPARK given back to the community?

- Transformed the community’s experiences into scientific knowledge
- 220,000+ families have received reports from six different screening tools
- Notified 8.6% of families and autistic people about a genetic cause
- Invited participants with a genetic diagnosis to join Simons Searchlight as the next step
- 85 webinars with researchers and clinicians
- 187 stories about our participants, treatments and therapies, and other topics important to the community
SPARK found a genetic result in 8.6% of participants with autism

- Analysis of 21,532 people with autism
- Overall, 8.6% of participants have a genetic finding
- Females are 1.5 times more likely to have a genetic finding compared to males
Forty percent of results are de novo

- 40% of all genetic findings have “de novo” inheritance, which means the genetic change is new in the person with autism
- In these cases, we are able to confidently test both parents and say the variant is not present in either parent
- If both parents submit DNA samples, there is a higher chance of receiving a genetic finding
Update on Return of Genetic Results

Secondary findings returned by SPARK include:

- Hereditary breast and ovarian cancer (HBOC)
- Familial hypercholesterolemia
- Lynch syndrome
SPARK Research:

How has SPARK data been used to advance our understanding of autism?
Parents’ perspectives on receiving a genetic diagnosis of autism

- Parents report both benefits and limitations to receiving a genetic diagnosis
- **Benefits** – an end to the diagnostic journey, feelings that knowledge = power, more patience with their child, better able to prepare for the future, can identify more specific advocacy groups for support, etc.
- **Limitations** – feelings of sadness, decreased hope, lack of treatments, new uncertainties, increased worry about the future, etc.
- Parents resolve these mixed feelings in different ways, including acceptance, ambivalence, weighing the tradeoffs, etc.
- **Providers should be aware of these potential pros and cons**, and help parents anticipate the mixed reactions and feelings

Experiences of Black families in the autism community

- Over 100 Black caregivers shared their experiences seeking a diagnosis and treatment services for their child
- Caregivers reported personal, community, and systemic barriers
  - 75% encountered providers who lacked humility, compassion, patience & understanding
  - 65% were hindered by their initial feelings of fear and denial
  - 60% experienced racial disparities
- Barriers were associated with increased parent stress
- Becoming a strong advocate (90%), moving from denial to acceptance (75%), and having positive treatment experiences (70%) were the most common facilitators in overcoming challenges
- Recommendations for professionals:
  - Greater humility, compassion & understanding
  - More training about autism
  - Increased support after diagnosis
  - More Black support groups

Behavior Parent Training (BPT) Engagement Among Young Children with Autism

While around 23% of families surveyed were deemed appropriate for BPT, and 93% were open to participating, approximately 69% had not been recommended a BPT program.

<table>
<thead>
<tr>
<th></th>
<th>Percent in Sample (n = 110)</th>
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<tbody>
<tr>
<td></td>
<td>Very Dissatisfied</td>
</tr>
<tr>
<td>Learning parenting techniques</td>
<td>2%</td>
</tr>
<tr>
<td>Child behavior improvement</td>
<td>3%</td>
</tr>
<tr>
<td>Parent-child relationship improvement</td>
<td>&lt;1%</td>
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<tr>
<td>Overall satisfaction</td>
<td>4%</td>
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Telehealth interventions are effective in improving fine motor skills and social communication.

Underage alcohol use in autistic youth age 16-20 yrs

UNDERAGE DRINKING BY AUTISTIC AND NON-AUTISTIC YOUTH

- **16%** Drank in last month
- **30 days** Number of drinking days per year
- **36 days** Number of drinking days per year
- **5%** Blacked out from drinking
- **37%**
- **10%**

(From SPARK Summary Report)

Unmet need for social engagement associated with increased depressive symptoms in autistic teens and young adults

Understanding aggression in autism across childhood

Verbal aggression peaks at age 6-12 years, then decreases. Similar pattern for non-autistic youth.

In autistic youth, physical aggression is highest <6 years of age, then decreases.

Disruptive behaviors fairly constant through age 12 years, then decrease. Similar pattern for non-autistic youth.

Sexual identity, mental health, and quality of life in autistic adults

Mean item scores for abbreviated World Health Organization Quality of Life domains and Autism Spectrum Quality of Life questionnaire for heterosexual and minority groups

Autistic adults who identified as a sexual minority reported more mental health symptoms (e.g. anxiety, depression) and lower quality of life

Sleep and antipsychotic medications as risk factors for cardiovascular disease (CVD)

Around 87% of autistic adults surveyed self-reported at least one CVD risk factor.

Antipsychotic medication usage (15% of sample) associated with having diabetes.

(Image from SPARK Summary Report)

Anticholinergic medications associated with memory and cognitive decline in autistic adults

Longitudinal study with middle-aged and older autistic adults (40-83 years)

Use of anticholinergic medications associated with self-reported cognitive decline

RESEARCH MATCH STUDY

Medications with high anticholinergic effects:

- Antihistamines including diphenhydramine & hydroxyzine
- Antidepressants such as amitriptyline, doxepin, & imipramine
- Antipsychotics such as chlorpromazine, clozapine, & olanzapine
- And many others

McQuaid GA. et al. Increased anticholinergic medication use in middle-aged and older autistic adults and its associations with self-reported memory difficulties and cognitive decline. Autism Research. 2023 Dec. [Figure from G. Wallace presentation Feb.’24]
Self-reported cognitive decline among middle and older age autistic adults

Percentage endorsements of each item on the Ascertain Dementia 8 (AD8) measure

High rates (30%) of individuals screened positive, suggesting potential cognitive decline

The Future of SPARK:

What has SPARK accomplished since its inception?
SPARK Research Match connects members of the SPARK community to autism research studies. Researchers worldwide use Research Match to connect to autistic individuals and families who are willing to volunteer for online or in person studies.

SPARK participants are eligible for Research Match! To increase your eligibility:

• Complete your dashboard surveys
• Send in your saliva kits, if you consented to the genetic portion of the study
• Participation is always voluntary!
• You may receive information about the study from the study team itself. In addition, SPARK will often prepare a Research Match Summary Report and an article about the study.
New research study

Monitoring sleep and activity at home

**Smartwatch**
Heart rate, skin temperature, movement, and skin conductance (anxiety).

**Headband**
Measures brain activity and movement during sleep.

**Sensor**
Small mat that is placed under the mattress. Records movements, heart rate, breathing, and time in bed.
Design and goals

Core principles:

- Objective
- Direct
- Longitudinal
- Scalable
- Remote
- Standardized
- Fast data sharing

Why do autistic individuals have 3 times more sleep problems than controls?

- Collect data from 100 SPARK families with one autistic child and one non-autistic child (10-17 y/o) who live in the same household
- Record each child’s sleep for 2-3 weeks
- Connect sleep data with genetic data
- 40% completed within 4 months

Mihca Hacohen
Ilan Dinstein
SPARK’s commitment to understanding autism

SPARK is making important progress possible. We are committed to supporting autism research for decades to come. There are exciting things on the horizon.

To understand the complexity of autism — what makes us the same and what makes us different — we need large numbers of people to participate.

We are committed to understanding:

• Diversity, Equity, and Inclusion in the autism community
• Quality of life in autistic adults
• Genetic causes of autism
• Co-occurring mental health conditions
• Differences in males and females with autism
• …and so much more!
What does the future hold?

• *Continue recruiting!*
• Increase diversity through targeted initiatives
• Learn about the life course of people with autism and their families
• Retain participants after they turn 18
• Short and quick research surveys
• Continue accelerating all types of research through Research Match
• Discover more about autism genetics
• Notify participants if we find genetic variants related to autism and related to certain serious medical conditions