



**SPARK** 



# A 5-Year Progress Report

Transforming the way autism  
research is done.



 **5 YEARS OF  
SPARK**



[SPARKforAutism.org](http://SPARKforAutism.org)

**SPARK is a growing community of autistic individuals, their families, and researchers on a mission to advance the understanding of autism.**

**The goal of SPARK is to build the largest, long-term research cohort to better understand the causes of autism and to help improve lives.**



[SPARKforAutism.org](https://SPARKforAutism.org)

Letter from Dr. Wendy Chung

## Transforming Autism Research



**Thank you for your commitment to science and participation in SPARK!**

**As SPARK celebrates its 5th anniversary, I would like to take a moment to reflect on how far we have come and look forward to the future.**

SPARK launched in April 2016. At that time, we had a vision to power autism research so we could find more answers. Because of you, there has been real progress! Over the past five years, autistic adults and families have not only been participants in the study, but have also served on three SPARK advisory boards, on our staff, and as community partners. They guide how our research is done and what questions we should ask. We now have over 100,000 people with autism and 175,000 of their family members participating in SPARK and being matched with research studies that are performed by leading autism researchers. So, what have we learned about autism in these past five years?

Over 100 genes have been linked to autism. We hope that by understanding the genetic basis for autism, we will better understand what makes individuals with autism special and how to best support them.

### **Over 100 genes have been linked to autism.**

Some interventions may directly focus on a genetic treatment for some people in the future, whereas other treatments could be medications, behavioral or educational strategies, or technological support. Importantly, the genetic studies are just one aspect of what SPARK does.

We have also built the largest data resource in the world on individuals with autism.

Using SPARK data, researchers have new insights about autism, including the occurrence of regression with autism, challenges with developmental coordination disorder, and “camouflaging” in teens. We were inspired to find that some minimally verbal children learn to talk even after the age of four. We also found that many more autistic adults need support for anxiety and depression than previously thought, and this issue is most pressing for women. Insights such as these can power change and help to ensure that attention and resources are focused in these areas, especially when the research is trustworthy and based upon large numbers of participants. We invite you to read the sections in this report that detail some of the results from SPARK research.

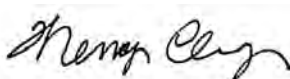
SPARK is committed to supporting autism research for decades to come. There are exciting things on the horizon. Here are just a few:

- As SPARK participants age, we will have a better understanding of how things change over their life course.
- Technology changes so fast – things that were impossible a few years ago are now commonplace. We will carry out a pilot study that collects data from wearable devices like Fitbits or Apple Watches to make it easier to sense how people are feeling based upon their heart rate.
- The Simons Foundation Autism Research Initiative (SFARI) is sponsoring several new studies of sleep and online assessments of behavior in babies and adults in SPARK.
- A small number of SPARK participants with specific genetic variants are eligible for a clinical trial that is

testing a new medication (arbaclofen) that may help with autism challenges with speech.

We are here for the long term to watch what happens over the life course for your family, to be able to listen to you as you go through those different phases with successes and challenges, and together, to share and help each other on this journey.

Here's to the next five years!



Wendy Chung, M.D., Ph.D.  
Principal Investigator, SPARK



There are a lot of resources that aren't available to many families, because of income. I've been that parent. So, I feel it would be very beneficial to get research out to those families so that they are able to take advantage of all the resources available to them, to help their children in the most efficient way possible.

Kanesha Burch, autism mom



**275,075**  
**PARTICIPANTS**  
**JOINED**



Since launching in 2016, **over a quarter of a million people**, including **over 100,000 people with autism**, have joined.

**OUR**  
**CLINICAL**  
**SITE**  
**PARTNERS**



The SPARK Clinical Site Network includes **31 top research centers** and **academic institutions** across the country.

# LARGEST RESEARCH STUDY

## About SPARK

**SPARK is now the largest study of autism ever, with over 275,000 participants. We are working to address the gaps in autism research and transforming the way that autism research is done.**

We collect medical, behavioral, and genetic information from families and autistic adults across the country. With your contribution, we are making strides in understanding autism.

SPARK strives to listen to and include all voices in the autism community.

Because SPARK has participants aged 2 to 92, we are able to study autism across the lifespan. There are complex challenges in the different life stages, and SPARK is committed to serving the autism community for decades to come.

## SPARK Research Match

**The SPARK Research Match program pairs members of the SPARK community with other autism research studies.**

These studies may involve anything from an online survey about autism services to an in-person study at a hospital. SPARK has helped researchers learn about gender identity, genetics, sleep, food selectivity, and depression. Research Match gives you the chance to be represented in research and share your unique experience.

**Research Match accomplishments from its launch in July 2017 through August 2021:**

- Over **90,000 SPARK families** have been invited into new research studies.
- More than **40,000 families** have responded to at least one study invitation.

- Researchers have published **24 articles** from studies supported by Research Match.
- Over **160 research studies** have been approved and 100 launched, with the others in progress.

**275,075**

TOTAL  
PARTICIPANTS  
IN SPARK

**106,725**

INDIVIDUALS WITH  
AUTISM INCLUDING:

**16,636**

ADULTS  
WITH AUTISM

# WHO ARE OUR PARTICIPANTS?

**26,415**

FEMALES  
WITH AUTISM

**80,310**

MALES WITH  
AUTISM



## What SPARK Has Learned

In the five years since SPARK launched, there have been over 50 publications that use SPARK or SPARK Research Match data. In the next few pages, we summarize some of the main findings. For a full list of SPARK publications visit our publications page <https://bit.ly/SPARKpubs>

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**SCIENTISTS HAVE USED  
SPARK DATA TO FURTHER  
AUTISM RESEARCH.**



## About Genetics

**Scientists have found over 100 genes that are strongly linked to autism, largely due to de novo or new genetic variants that are present in the person with autism, but not present in either of the parents.<sup>1</sup> These genes cluster into three important biological categories:**

1. Controlling the activity of other genes
2. Communication between neurons
3. Internal structures around which cells are organized

In 2021, a new study showed that genetic variants inherited from parents (even if the parent does not have autism) can be important as well.<sup>2</sup> The genes affected by inherited variants that are passed down from a parent tend to be different from the set of autism genes affected by de novo variants. The study also showed that the inherited variants were more commonly found in families that had multiple people with autism than in families that had only one person with autism.

Going forward, SPARK will continue to work towards identifying all of the genes involved in autism. A comprehensive understanding of autism's genetic foundation will help us understand how the brain of a person with autism works differently. This knowledge will help develop better supports and treatments.

## About Behavior and Development

Researchers have been studying some of the most common features of autism in thousands of families in SPARK. They found that children have a lot of special interests, but most parents are not concerned that these interests interfere with daily life.<sup>3</sup>

There are certain kinds of interests, such as a TV show, a particular schedule, or a favorite object, that seem to cause more difficulties in transitioning through the day. People with autism also have many sensory issues, including difficulty

perceiving body signals, such as hunger or discomfort.<sup>4</sup>

Researchers have also learned that although there may be concerns about handwriting and physical coordination in older children, most children

with autism seem to be born with good basic motor control. Delays in early milestones like first steps may signal a more severe underlying genetic condition.<sup>5</sup>

## 70,000 REPORTS RETURNED

Just under 70,000 families have received reports from five different screening tools used in SPARK, reflecting their individual behavior and development.



SPARK will have very clear benefits for research, but as I understand it, this may take some time. [My son] may not be a direct beneficiary, but as a mom, I feel that I have an obligation to contribute to our understanding of what causes autism and how to provide better treatments. And I think that could be my small step to help many others like [my son].

Sunghee Park, autism mom

## About Co-occurring Conditions

Autistic individuals and their families face many challenges every day. Rates of depression, anxiety, and attention-deficit/hyperactivity disorder are all higher among people with autism and their family members in SPARK compared to the general population. Learning more about how these conditions interact and how to treat these conditions will provide a better quality of life.

- SPARK researchers looked specifically at rates of attention-deficit hyperactivity disorder (ADHD) and anxiety disorders in autistic children.<sup>6</sup> This study suggested that health care providers should pay special attention to females when considering an autism spectrum diagnosis. The researchers found that girls had different behavioral features than boys and that more specific diagnostic measures are needed with girls.
- Three other studies using SPARK data reinforced issues with motor skills in children with autism. One SPARK study found that autistic children in SPARK were more than 22 times as likely to have motor coordination issues as the general population.<sup>7</sup>

Another study found that motor problems were not related to intellectual disability in children in SPARK. Parent-reported motor difficulties in activities, such as sports and handwriting, were almost universal.<sup>8</sup> A third study found that although these kinds of motor difficulties continued into adolescence, less than one-third of teen participants received physical therapy.<sup>9</sup>

By increasing awareness of the severity and impact of motor impairments in autistic children, the researchers hope that doctors and insurance companies will begin to incorporate appropriate therapies to decrease these challenges.



We joined SPARK to help with research. If [our daughter] can help others behind her, then why not.

Jodie Zeyer, autism mom

# PARTICIPATE AND DISCOVER



## About COVID-19's Impact

**The coronavirus disease 2019 (COVID-19) pandemic has had a wide-ranging impact on the autism community. SPARK surveyed participants in a series of questionnaires to learn more about:**

- How the COVID-19 pandemic has affected access to services and therapies for people with autism
- Overall emotional well-being of parents or caregivers, children or dependents, and autistic adults
- Availability and effectiveness of online and telehealth services
- Attitudes towards the COVID-19 vaccines

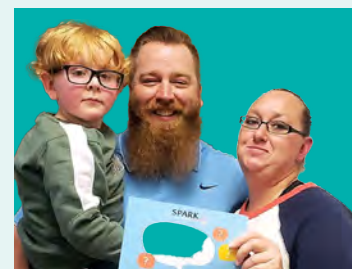
**Parents, caregivers, and autistic adults were surveyed starting in March 2020. Surveys were repeated every few months.**

Over 9,000 parents and caregivers and 636 autistic adults took part in the initial survey. Learning more about how COVID-19 is impacting the autism community will help us to better meet the needs of the community in times of national disruption; understand what does and does not work for online and telehealth services; advocate for supportive therapies, services, and policies; and document and share the unique experiences of the autism community during the pandemic.

- During the early months of the pandemic (March 2020 - April 2020), parents raising a child with autism reported higher levels of psychological distress when compared to parents in the U.S. as a whole. In addition to experiencing greater psychological distress, nearly 50 percent of the families of children with autism were at risk for a crisis, experiencing moderate to high levels of general distress.<sup>10</sup>
- In addition to impacts on mental health, children with autism experienced significant disruption. This includes a complete lack of access to their services and therapies for autism. Disruptions were reported most for high-intensity services, such as applied behavior analysis, special education, speech language therapy, and physical/occupational therapy. Most services were disrupted for a majority of autistic individuals across all age groups. Medical services were the least disrupted. While some services had switched to online or telehealth delivery, most individuals with autism were not receiving remote services early in the pandemic, and those who did generally did not find them very helpful.<sup>10</sup>

 **75,000**  
**PARTICIPANTS'**  
**DNA**  
**SEQUENCED**

DNA from 75,000 participants, including 38,000 people with autism has been sequenced and made available to qualified researchers.



These reports highlight the need to tailor assistance, both for mental health and autism-related services to these families during times of national crisis, as well as to make quality services accessible in online or telehealth formats. In addition, more research on online and telehealth services is needed to determine how to successfully conduct therapy this way, and whether different groups of individuals need different telehealth approaches.

**There is a need to improve online and telehealth support for autistic adults to ensure high-quality services can continue uninterrupted.**

- For autistic adults, the COVID-19 pandemic also presented challenges. Compared to their feelings before the pandemic, by May 2020, two-thirds of autistic adults reported feeling pandemic-related distress.<sup>11</sup> A related study found that autistic adults who were younger, female, and had a mental health diagnosis before the pandemic were more likely to report being negatively impacted by COVID-19 and have greater difficulty coping.<sup>12</sup>

Less hopefulness predicted greater distress over time, as well as feeling less benefit from online services. Researchers suggest that these findings provide areas to target in therapy, for example, positivity and coping skills. When developing supports for autistic adults during crises, researchers suggest that certain groups may need additional services and attention. In addition, similar to the experiences of parents and caregivers, there is a need to improve online and telehealth support for autistic adults to ensure high-quality services can continue uninterrupted.

## 830 AUTISTIC PEOPLE WITH GENETIC FINDINGS

SPARK has notified more than 500 families and autistic people about a genetic cause for autism.



**The attractiveness to us of SPARK was, first and foremost, the stature, the scale, and the scope of the scientific exploration of the root causes of autism.**

David Browne, autism dad



## About Adults

### Over 8,000 independent, autistic adults participate in SPARK and share information about their life experiences with autism.

In addition to completing surveys and providing genetic data, autistic adults participate in many other studies through our Research Match program. In the last two years, ten studies published new findings about autism in adulthood. By working together, autistic adults in SPARK and researchers increase our understanding of autism in the following areas:

- Mental Health** Two new studies focused on depression in autistic adults. Researchers found that most autistic adults with depression symptoms are getting diagnosed. But, only one-half of those who were currently depressed received treatment. Barriers to treatment included financial and insurance issues, lack of available services, and few providers familiar with autism and depression. Studies such as these highlight the need for more mental health services and providers.<sup>13, 14</sup>
- Autism and Aging** Few studies focus on older autistic adults. One recent study recruited 200 adults over age 50 from SPARK. Researchers found that older adults with autism were more likely to report tremors, muscle stiffness, muscle rigidity, and other movement issues. This study points to the need for more research on autism and aging.<sup>15</sup>
- Participant-Centered Research** Capturing real-life experiences from autistic people is important. Many of the surveys used in autism research were developed for non-autistic adults. Four new studies looked at how well some of these surveys capture data about depression, repetitive thinking, quality of life, and sensory challenges from those with autism. These findings will help researchers design better surveys and studies about autism.<sup>4, 16-18</sup>



I want scientists to figure out why I'm creative and imaginative. What is going on in my brain that makes me so unique?

Isaac Law, autistic adult

## 200 GENES

SPARK data has helped researchers discover new genes related to autism. There are now more than 200 genes known to play a role in autism and other neurodevelopmental conditions. Dozens of these genes have been discovered in the past 5 years.



## About Services

**Recent SPARK publications have shed new light on what might motivate families or individuals to seek care, as well as the types of care and services they receive.**

- One of SPARK's first Research Match studies looked at treatment patterns in children aged 3 to 17 with autism.<sup>19</sup> The researchers focused on understanding what non-drug treatments and services children were receiving, and they found that the most common were speech

and language therapy or occupational therapy, provided in school. Children living in rural areas were less likely to receive behavioral or speech and language therapy services, but there were no differences in service use between children with private insurance and children with Medicaid.

Overall, the study found that females were likely to be diagnosed 14 months later than their male counterparts. This was largely due to their mild or "atypical" presentations. Findings from this study underscore the need to better characterize and understand the female presentation of autism.

- Another recent study looked at factors that influence the age at which girls with autism are diagnosed.<sup>20</sup>

## 160 RESEARCH MATCH STUDIES

40,000 families have been part of over 160 studies through SPARK Research Match, ranging from the study of the neurobiology of atypical language development, to a clinical trial for a specific genetic cause of autism, to repetitive thinking patterns in autistic adults.



Of course, there's much more to parenting a child with autism than managing therapies and keeping the child safe. For the emotional part, I do my own intervention, like giving kisses, hugs, and a lot of love. This is one of the keys—the love. It's not only the research, it's not only the theories, the answers, the knowledge. It's also the love.

Jorge Rivera, autism dad



## Looking to the Future

As the SPARK community grows, so too does our understanding of autism.

- SPARK is working with researchers to better understand whether the younger siblings of children with autism may also be affected. The sibling of a person with autism has an increased chance of having autism.
- By following people through their life span we can learn about how autism changes as we age.
- There are over 160 Research Match studies that will publish their findings in the coming years. The research topics include parent training over telehealth, early social behaviors in infants, changes in brain anatomy in people with autism, Black families' experiences with obtaining a diagnosis of autism to create a culturally informed screening measure, social-communication abilities in both verbal and minimally verbal children, and mental and physical health outcomes for older autistic adults.
- SPARK will soon return genetic findings that are not related to autism but have serious or life-threatening consequences, such as the risks of cancer and heart disease. The number of people with these genetic findings will be small, but we believe it is our obligation to give participants this information.
- We will continue to return genetic results that are associated with autism. Most of the genetic results we have returned have been to people with autism and intellectual disabilities. As we learn more about the genetics of autism, we hope to get a better picture of the genetics of those with autism who do not have intellectual disabilities. We will return these genetic results as well.
- We are considering collecting data from wearable devices. Do you wear a Fitbit, Galaxy, Apple Watch, or other device? It might be the wave of the future.

**SPARK is committed to autism research for decades to come. We are forward-thinking with you in mind. We can make a difference together.**



**ADVANCING  
RESEARCH  
TOGETHER**

## Our Clinical Site Partners

**The nation's leading medical schools and autism research centers have joined with SPARK to help recruit individuals and families affected by autism.**



- |  |   |   |
|--|---|---|
| <ul style="list-style-type: none"> <li>• Autism Center at Texas Children's Hospital/Baylor College of Medicine</li> </ul>            | <ul style="list-style-type: none"> <li>• Medical University of South Carolina</li> </ul>                      | <ul style="list-style-type: none"> <li>• University of Iowa</li> </ul>  |
| <ul style="list-style-type: none"> <li>• Boston Children's Hospital</li> </ul>   | <ul style="list-style-type: none"> <li>• Nationwide Children's Hospital (Child Development Center)</li> </ul> | <ul style="list-style-type: none"> <li>• University of Miami's Center for Autism and Related Disabilities</li> </ul>                    |
| <ul style="list-style-type: none"> <li>• Center for Autism Research/The Children's Hospital of Philadelphia</li> </ul>               | <ul style="list-style-type: none"> <li>• OHSU (Oregon Health &amp; Science University)</li> </ul>             | <ul style="list-style-type: none"> <li>• University of Minnesota, Twin Cities</li> </ul>  |
| <ul style="list-style-type: none"> <li>• Children's Specialized Hospital</li> </ul>  | <ul style="list-style-type: none"> <li>• Rush University Medical Center</li> </ul>                            | <ul style="list-style-type: none"> <li>• University of Missouri, Thompson Center for Autism and Neurodevelopmental Disorders</li> </ul> |
| <ul style="list-style-type: none"> <li>• Cincinnati Children's Hospital Medical Center</li> </ul>                                    | <ul style="list-style-type: none"> <li>• Seattle Children's Hospital</li> </ul>                               |   |
| <ul style="list-style-type: none"> <li>• Emory University and Marcus Autism Center</li> </ul>  | <ul style="list-style-type: none"> <li>• Stanford University</li> </ul>                                       | <ul style="list-style-type: none"> <li>• University of North Carolina – Chapel Hill</li> </ul>  |
| <ul style="list-style-type: none"> <li>• Geisinger Autism Center &amp; Developmental Medicine Institute/The Emerge Center</li> </ul> | <ul style="list-style-type: none"> <li>• University of California, Davis MIND Institute</li> </ul>            | <ul style="list-style-type: none"> <li>• University of Utah</li> </ul>  |
| <ul style="list-style-type: none"> <li>• Kennedy Krieger Institute</li> </ul>  | <ul style="list-style-type: none"> <li>• University of California, Los Angeles</li> </ul>                     | <ul style="list-style-type: none"> <li>• Vanderbilt University Medical Center</li> </ul>  |
|  | <ul style="list-style-type: none"> <li>• University of California, San Diego and SARRC Phoenix</li> </ul>     | <ul style="list-style-type: none"> <li>• Yale University</li> </ul>   |



We were excited to share with [our son] the genetic information from SPARK, how he gets to be on the cutting edge of research, and how important it is that he can help other people.

Shannon McKee, autism mom

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# Pass It On!

**After reading this report, please consider passing it to someone with autism, their families, or those in their community.**

## Why Join?

You can play a role in powering autism research. Some of the benefits to you include:

- The possibility of finding a genetic cause of autism in your family.
- Getting personalized results from behavioral and developmental surveys.
- The chance to be invited to other autism research studies through SPARK Research Match.
- The opportunity to earn gift cards as part of your participation.
- Staying informed on the latest autism research through articles on our website, monthly webinars, newsletters, and the latest updates on SPARK's social media platforms.

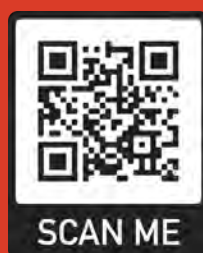
## How to Join

**Visit [SPARKforAutism.org](https://SPARKforAutism.org)** and you will be guided through a simple, easy process to become a member of our growing community. Enrollment only takes about 20 minutes. Your data will be protected and it's free!

**Together, we will advance autism research for years to come.**



[SPARKforAutism.org](https://SPARKforAutism.org)



SPARK: Simons Foundation Powering Autism Research  
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SPARK Principal Investigator: Wendy Chung, MD, PhD  
Western Institutional Review Board