

## Genetic Analysis of 457 SPARK Families Reveals Genes Linked to Autism

Results from SPARK's 'pilot' phase — the first group of families enrolled in the study — show that about 1 in 10 families get a genetic result.

*Summary Reports describe results from newly published research using data from SPARK participants.*

### ■ Study Title

Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes

### ■ What was the study about?

The study presents the first genetic findings from SPARK. Scientists calculated how many SPARK participants get a genetic diagnosis and looked for new genes that are linked to autism.

### ■ How was the study done?

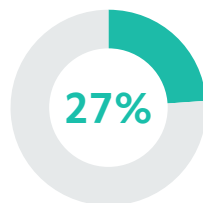
SPARK scientists analyzed genetic data from almost 500 families who enrolled in SPARK between December 2018 and December 2019. This included:

- 418 families with one child who has autism.
- 39 families with two or more children who have autism.

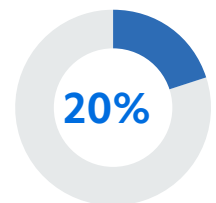
### ■ What did the researchers learn?

- One in ten families that enroll in SPARK get a genetic result.

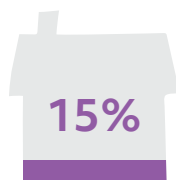
- Participants who are most likely to get a genetic result are those who have autism and seizures



- Participants who are most likely to get a genetic result are those who have intellectual disability



- In families with two or more children who have autism, about 15 percent got a genetic result.



- In families with one child who has autism, about 10 percent got a genetic result.



- Families in the SPARK pilot group had changes in 26 different genes that are linked to autism. Most of these genes had already been strongly linked to autism. But the SPARK analysis provided new support for several other genes. One of these genes, called BRSK2, meets the criteria for an autism risk gene.

## ■ What was new and innovative about the study?

The study shows that it's possible to:

- enroll a large number of participants in a research study online.
- get good DNA samples from mail order spit kits.
- find new autism risk genes through online research communities.

## ■ What do the findings mean?

The findings predict how many SPARK participants can expect to get a genetic diagnosis. The study also found new genes that are linked to autism. This means that the SPARK study will eventually increase the number of people who get a genetic diagnosis. The findings highlight the power of researchers and families working together.

## ■ What are scientists and participants saying?

**Scientist (Jacob Michaelson, Ph.D., SPARK Investigator at the University of Iowa):** "These are results from active, engaged families and individuals, many of whom are having genetic findings returned to them by a genetic counselor as a result of this research," says Jacob Michaelson, Ph.D., a SPARK investigator from the University of Iowa. They are people who continue to advance autism research by participating in follow up studies (through SPARK's research matching program) that look at specific aspects of autism that they are passionate about and that they have a personal connection to. So the genes are important, but it's the thriving community behind those genes that makes this study significant to me. From that perspective, this paper marks the beginning of a new era of genetic research in autism."

**Participant (Lynn Vigo, whose daughter got a genetic diagnosis through SPARK's research):** "Like most moms, I have always wondered if I did or didn't do something to cause or contribute to her autism, such as eating or not eating something," Vigo says. "Knowing it was a spontaneous genetic event at the time of conception made it concrete for me that I had no control over that."

## ■ What's next?

SPARK scientists are now analyzing genetic data from a much larger group of participants. The goal is to uncover more autism risk genes. As scientists find more genes that are linked to autism, the percentage of families that receive a genetic diagnosis will likely grow.

## ■ Reference

Feliciano P. et al. NPJ Genom. Med. 4, (2019) PubMed