



How does SPARK analyze participants' DNA?



See the journey your DNA takes from saliva sample to genetic results.



It may take several years until you hear from us about your DNA submission. We outline each step and its timeline in this document.

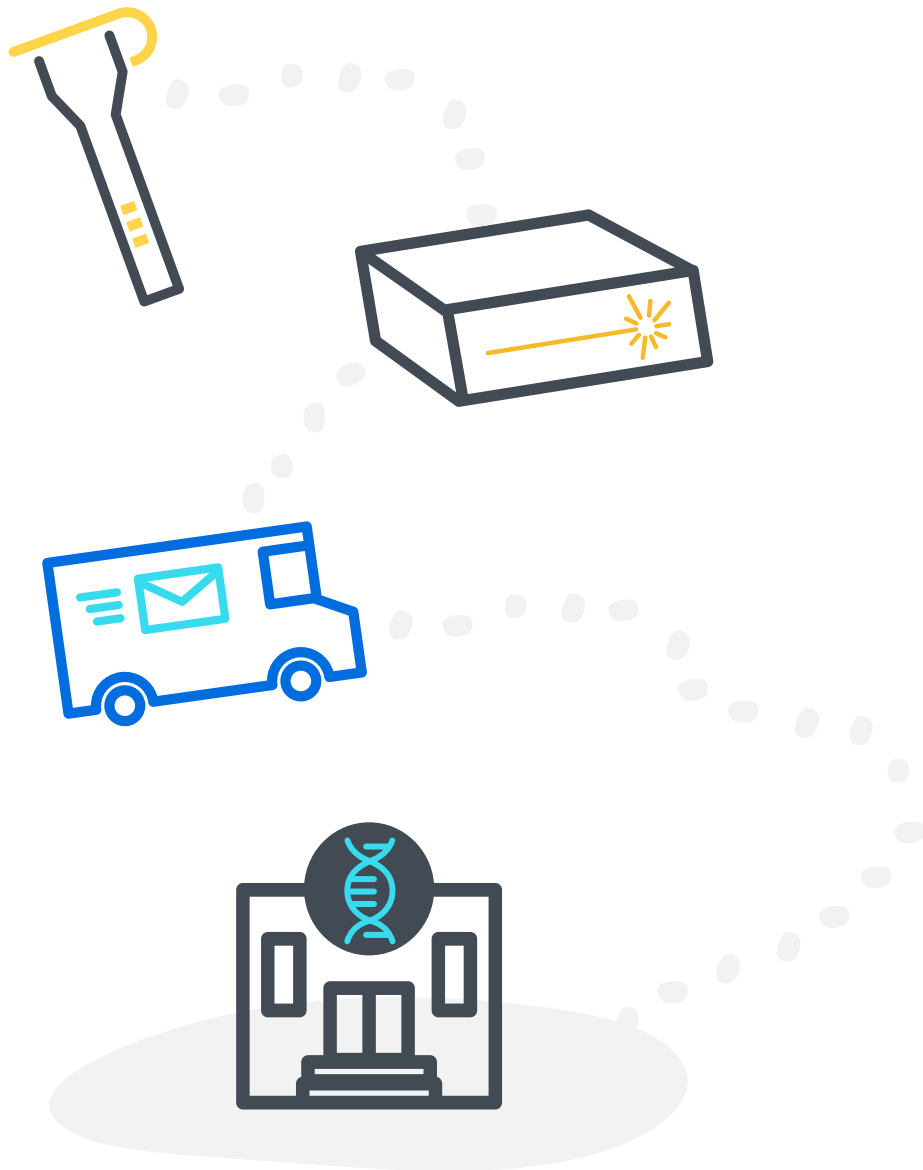
1.



Send saliva sample to the lab



Duration: 1-2 weeks



2.



Scientists isolate DNA from saliva



Duration: 2 weeks



Where: External Facility



Five to ten percent of samples can't be processed. This may be because the samples were not labeled properly. Or because there was not enough saliva to isolate DNA. In these cases, SPARK will send you another saliva kit.

3.



DNA samples are gathered for analysis



Duration: 2-4 months



Where: External Facility



This process used to take much longer, 3 to 9 months. This was because SPARK had to collect batches of 15,000 DNA samples before the DNA could be analyzed. This is no longer the case.

4.



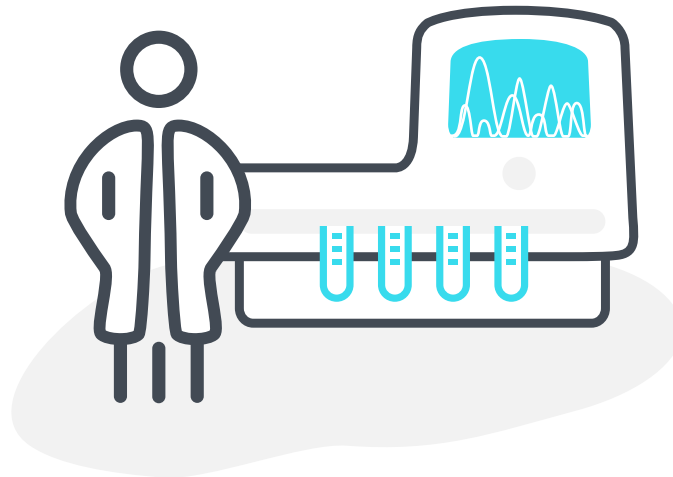
DNA is read letter by letter, a process known as sequencing



Duration: 9 months



Where: External Facility



- About two to three percent of samples cannot be sequenced. This may be because the samples were not labeled properly or contaminated. In these cases, SPARK will send you another saliva kit.
- The type of sequencing is called exome sequencing. This means that scientists read the parts of DNA that produce proteins.

5.



SPARK receives sequence data from an external lab and analyzes the data, searching for changes in the DNA sequence



Duration: 4-6 months



Where: SPARK



- Changes in the DNA can include new or inherited changes. These changes may involve a single letter of DNA or span many letters, known as copy number variations.
- Computer programs analyze DNA for different types of changes. The process involves huge amounts of data – a petabyte, or a million gigabytes. This can be time-consuming. For comparison, it would take 2,000 years to listen to a petabyte of music on MP3 files.
- SPARK staff and scientific experts who collaborate with SPARK search for different types of changes. SPARK then compiles the results.
- Automated tools can find about half of the DNA changes. Scientists analyze more difficult cases by hand, a process that can require many hours.

6.



Scientists narrow down the list of DNA changes to genes or regions that are on SPARK's gene list. Genes on this list have been linked to autism in multiple unrelated people.



Duration: 3-6 months



Where: SPARK



Learn more



SPARK gene list

bit.ly/SPARKGeneList

"What Makes an Autism Gene?"

bit.ly/WhatMakesAnAutismGene

7a.



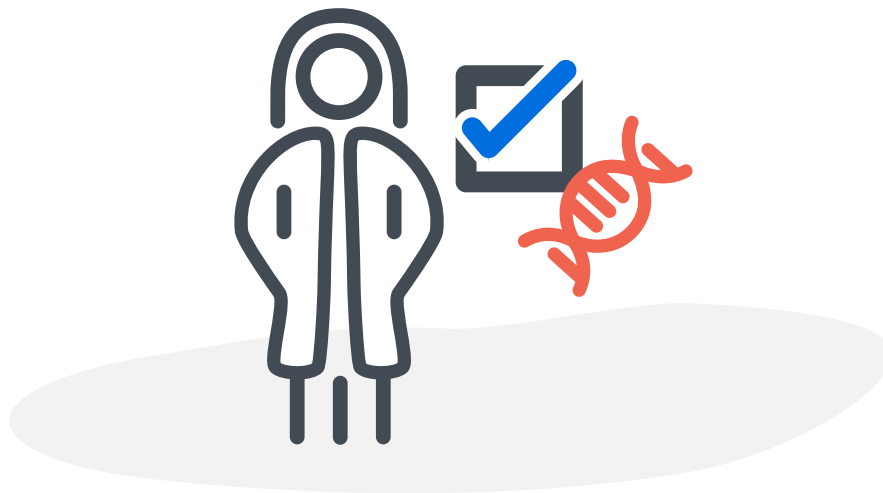
If a gene change is found that is linked to autism, a clinical genetics lab reanalyzes your DNA and confirms the result.



Duration: 3 months



Where: External Facility



- Some gene changes result in a non-working copy of the gene. Other gene changes alter how well the gene works. Some changes affect many genes.
- Early results suggest that about 10 percent of participants will get a genetic result. That number will grow as more genes that are linked to autism are discovered.

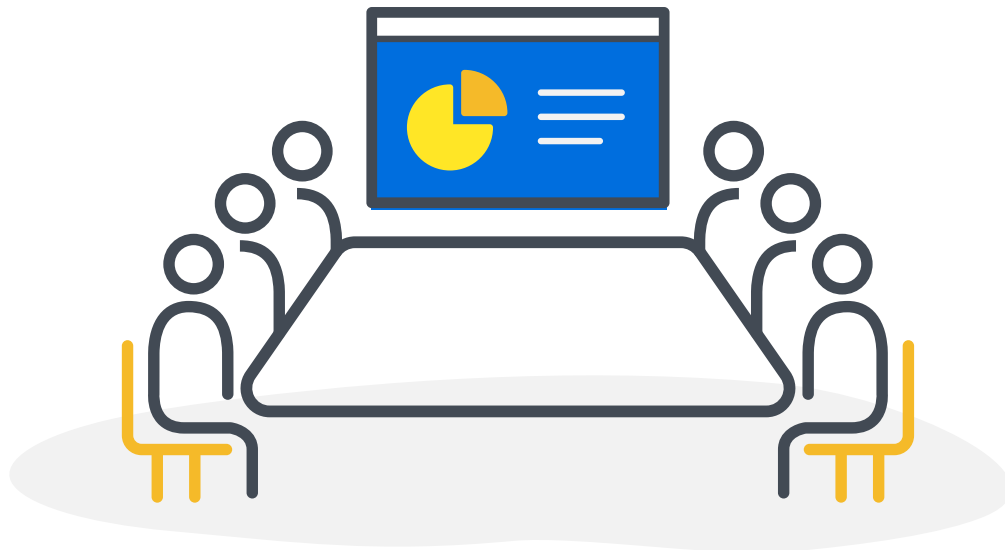
..... > **7b.** or >

In some cases, a participant has a gene change that may be linked to autism but hasn't yet met SPARK's criteria.

Additional experts review the results. If there is enough data to link the gene to autism, a clinical genetics lab reanalyzes your DNA and confirms the result.



Duration: The SPARK gene list is evaluated every 3 months



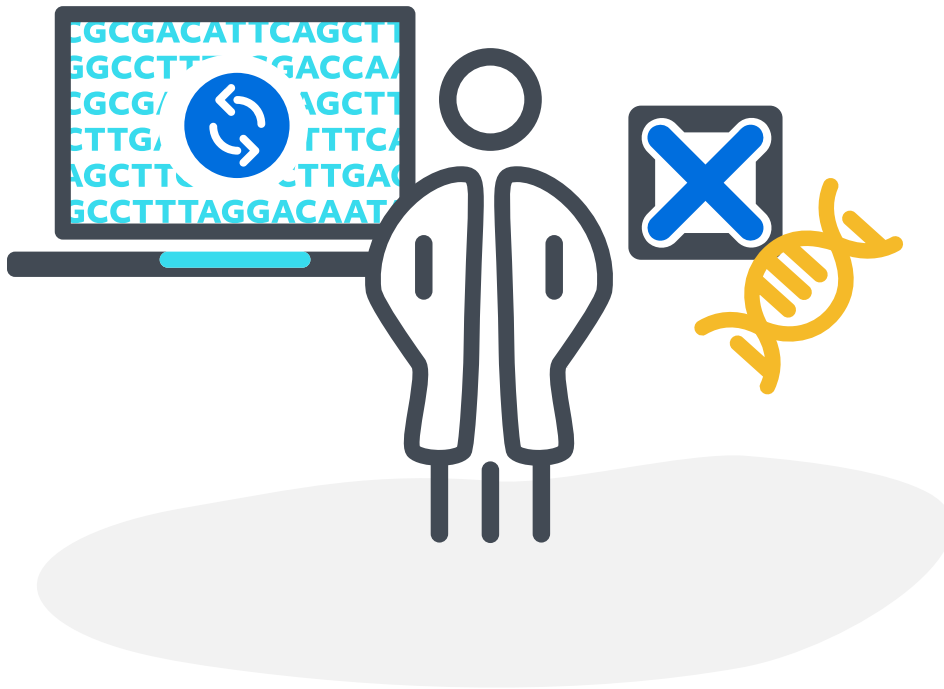
In these cases, scientists consult the medical genetics committee, which regularly reviews new research and adds to SPARK's list of autism-related genes.



7c.



In some cases, no autism-related gene changes are found.





If we have analyzed your DNA, we will let you know what we find. If we find a genetic change linked to autism, we will contact you. If we don't find a genetic change linked to autism, we will also let you know. This entire process may take several years.



Families are contacted



If an autism-linked gene change is found, a clinical genetics lab confirms the result.



Participants' DNA samples are re-analyzed as new genes are added to the list.



Duration:
Ongoing

SPARK

