SPARK Gene List

SPARKforAutism.org

SPARK
Simons Powering Autism Research

Autism Gene Discovery

Genetic changes are one of many causes of autism. Scientists have identified over 150 genes and segments of chromosomes, known as copy number variants (CNVs), that are related to autism. But there is still so much to learn.

Scientists predict that several hundred more genes related to autism have yet to be found. One of SPARK's main goals is to find them.

The more people who participate in SPARK, the faster we can find these genetic changes. If you or your family member has autism, your DNA may contain important clues that can help us to better understand autism genetics.

What are genes, chromosomes and genetic differences?

Genes

- Genes are made up of DNA. They provide instructions for making the proteins that our cells and body need in order to function.
- We all have the same genes. For example, we all have the gene called, CHD8.

Chromosomes

- Genes are located on chromosomes.
- Everybody has one set of chromosomes from their mother and one set from their father.

Genetic differences

- Except for identical twins, no two people have the same genetic make-up. Everyone has genetic differences that make them unique.
- Genetic differences can vary in their size, but a small change can still have a big impact:
  - Some people, such as those with Down syndrome, have extra copies of entire chromosomes.
  - Some people have chromosomes with regions that have been deleted, duplicated or rearranged. These changes, which scientists call copy number variants, often include multiple genes.
  - Some people have smaller genetic differences within single genes.

The SPARK Gene List

The SPARK gene list contains 157 single genes (blue) and 28 copy number variants (orange) that are known to be associated with autism. More information about each autism-linked gene or CNV, along with its associated symptoms, is available here.

Copy Number Variants	Single Genes				
1q21.1	ACTB	CNOT3	KDM3B	PCDH19	SMARCC2
2p16.3	ADNP	CREBBP	KDM6B	PHF21A	SON
2q11.2	ADSL	CSDE1	KIAA2022	PHF3	SOS1
2q37	AFF2	CTCF	KMT2A	PHIP	SOS2
3q29	AHDC1	CTNNB1	KMT2C	POGZ	SOX5
5p-	ALDH5A1	CUL3	KMT5B	POMGNT1	SPAST
5q35	ANK2	DDX3X	KRAS	PPP1CB	SRCAP
6q16	ANK3	DEAF1	LZTR1	PPP2R5D	STXBP1
7q11.23 distal	ANKRD11	DHCR7	MAGEL2	PSMD12	SYNGAP1
7q11.23	ARHGEF9	DLG4	MAP2K1	PTCHD1	TANC2
8p23.1	ARID1B	DMPK	MAP2K2	PTEN	TAOK1
9q34	ARX	DNMT3A	MBD5	PTPN11	TBCK
15q24A_C	ASH1L	DSCAM	MBOAT7	RAF1	TBR1
15q11.2-q13.1	ASXL3	DYRK1A	MECP2	RAI1	TCF20
15q13.3	ATRX	EBF3	MED13	RELN	TCF4
15q15	AUTS2	EHMT1	MED13L	RERE	TLK2
16p11.2	BAZ2B	EIF3F (F232V)	MEIS2	RFX3	TRIO
16p11.2 distal	BCKDK	EP300	MYT1L	RIMS1	TRIP12
16p12.2	BCL11A	FMR1	NAA15	RIT1	TSC1
16p13.11	BRAF	FOXG1	NBEA	RORB	TSC2
16p13.3	BRSK2	FOXP1	NCKAP1	SCN1A	TSHZ3
17p11.2	CACNA1C	GIGYF1	NF1	SCN2A	UBE3A
17q11.2	CAPRIN1	GIGYF2	NIPBL	SCN8A	UPF3B
17q12	CASK	GRIN2B	NLGN2	SETBP1	VPS13B
17q21.3	CASZ1	HIVEP2	NLGN3	SETD2	WAC
22q11.2 central	CDKL5	HNRNPH2	NR4A2	SETD5	WDFY3
22q11.2	CHAMP1	HNRNPU	NRAS	SHANK2	ZBTB20
22q13.3	CHD2	HRAS	NRXN1	SHANK3	ZNF292
	CHD3	IQSEC2	NRXN2	SHOC2	ZNF462
	CHD7	IRF2BPL	NRXN3	SIN3A	
	CHD8	KANSL1	NSD1	SLC6A1	
	CIC	KCNB1	PACS1	SLC9A6 (NHE6)	

How does a gene or CNV make it onto the list?

- We include genes and CNVs that have strong and consistent evidence that they are associated with autism.
- We update the list four times a year. New genes and CNVs are approved by the SPARK medical genetics committee.

What is the SPARK medical genetics committee?

- The members of the committee are experts in the study of autism genetics.
- The committee meets four times a year with SPARK's principal investigator, Dr. Wendy Chung, and SPARK's scientific director, Dr. Pamela Feliciano, to discuss the genetic findings from the SPARK study as well as updates to the SPARK gene list.
- You can learn more about the members of the committee in this video.

How does SPARK inform participants of genetic results?

- When we find that a participant with autism has a genetic change in one of the genes or CNVs on our list, we share those results with you as soon as we can, free of charge.
- Participants may decide if they want to hear about their results through their own medical provider or a SPARK-provided genetic counselor.
- It may take months or years for a participant to get a result. That's because SPARK's analyses are complex, and they take into account the latest advances in autism genetics.
- At this time, we are finding genetic changes related to autism in about 10 percent of families enrolled in SPARK.

To check the status of your sample(s), log in to your SPARK dashboard.

Contact us at info@SPARKforAutism.org if you have any questions.