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 close to 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 153 single genes (blue) and 19 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	CREBBP	KIAA2022	PHF3	SMARCC2
2p16.3	ADNP	CTNNB1	KRAS	PHIP	SON
3q29	ADSL	CTCF	KMT2A	POGZ	SOS1
5q35	AFF2	CUL3	KMT2C	POMGNT1	SOS2
7q11.23	AHDC1	DDX3X	LZTR1	PPP1CB	SOX5
8p23.1	ALDH5A1	DHCR7	MAGEL2	PPP2R5D	SPAST
15q11.2 BP1-BP2 deletion	ANK2	DLG4	MAP2K1	PSMD12	SRCAP
	ANK3	DMPK	MAP2K2	PTCHD1	STXBP1
15q11.2-q13.1	ANKRD11	DNMT3A	MBD5	PTPN11	KMT5B (SUV420H1)
15q13.3	ARHGEF9	DSCAM	MBOAT7	PTEN	
15q15	ARID1B	DYRK1A	MECP2	RAF1	SYNGAP1
16p11.2	ARX	EBF3	MED13	RAI1	TAOK1
16p12.1	ASH1L	EHMT1	MED13L	RELN	TANC2
16p13.3	ASXL3	EP300	MEIS2	RERE	TBCK
17p11.2	ATRX	FMR1	MYT1L	RFX3	TBR1
17q11.2	AUTS2	FOXG1	NAA15	RIMS1	TCF20
17q12	BAZ2B	FOXP1	NBEA	RIT1	TCF4
17q21.3	BCKDK	GIGYF1	NCKAP1	RORB	TLK2
22q11.2	BCL11A	GIGYF2	NF1	SCN1A	TRIO
22q13.3	BRAF	GRIN2B	NIPBL	SCN2A	TRIP12
	BRSK2	HECTD4	NLGN2	SCN8A	TSC1
	CACNA1C	HIVEP2	NLGN3	SETBP1	TSC2
	CASK	HNRNPH2	NRAS	SETD2	TSHZ3
	CDKL5	HNRNPU	NR4A2	SETD5	UBE3A
	CHAMP1	HRAS	NRXN1	SHANK2	UPF3B
	CHD2	IRF2BPL	NRXN2	SHANK3	VPS13B
	CHD3	IQSEC2	NRXN3	SHOC2	WAC
	CHD7	KANSL1	NSD1	SIN3A	WDFY3
	CHD8	KCNB1	PACS1	SLC6A1	ZBTB20
	CIC	KDM3B	PCDH19	SLC9A6 (NHE6)	ZNF292
	CNOT3	KDM6B	PHF21A		ZNF462

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.

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