

SPARK Gene List

The SPARK gene list contains 167 single genes ([blue](#)), 43 copy number variants ([orange](#)) and 5 chromosomal differences ([green](#)) 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 del	16p12.2 del	ACTB	DMPK	MTOR*	SETD2
1q21.1 dup	16p13.11 del	ADNP	DNMT3A	MYT1L	SETD5
2p16.3 del	16p13.3 del	ADSL	DSCAM	NAA15	SHANK2
2q37.3 del	17p11.2 del	AFF2	DYNC1H1	NBEA	SHANK3
3q29 del	17p11.2 dup	AHDC1	DYRK1A	NCKAP1	SHOC2
3q29 dup	17p13.3 del	ALDH5A1	EBF3	NEXMIF	SIN3A
5p- del	17p13.3 dup	ANK2	EHMT1	NF1	SLC6A1
5q35 del	17q11.2 del	ANK3	EIF3F*	NIPBL	SLC9A6
5q35 dup	17q11.2 dup	ANKRD11	EP300	NLGN2	SMARCC2
6q16 del	17q12 del	ARHGEF9	FMR1	NLGN3	SON
7q11.23 del	17q12 dup	ARID1B	FOXP1	NLGN4X	SOS1
7q11.23 distal del	17q21.3 del	ARX	GIGYF1	NR4A2	SOS2
7q11.23 dup	17q21.3 dup	ASH1L	GIGYF2	NRAS	SOX5
8p23.1 dup	22q11.2 del	ASXL3	GRIN1*	NRXN1	SPAST
9q34 del	22q11.2 dup	ATRX	GRIN2A*	NRXN2	SRCAP
9q34 dup	22q11.2 central del	AUTS2	GRIN2B	NRXN3	STXBP1
15q11.2q13.1 del	22q11.2 central dup	BAZ2B	HIVEP2	NSD1	SYNGAP1
15q11.2q13.1 dup	22q13.3 del	BCKDK	HNRNPH2*	PACS1*	TANC2
15q13.3 del	Xq28 dup	BCL11A	HNRNPU	PCDH19	TAOK1
15q15 del		BRAF	HRAS	PHF21A	TBCK
15q24A_C del		BRSK2	IQSEC2	PHF3	TBR1
16p11.2 del		CACNA1C	IRF2BPL	PHIP	TCF4
16p11.2 distal del		CAPRIN1	KANSL1	POGZ	TCF20
16p11.2 dup		CASK	KCNB1	POMGNT1	TLK2
		CASZ1	KCNQ3*	PPP1CB	TRIO
		CBL	KDM3B	PPP2R5D*	TRIP12
		CDKL5	KDM6B	PSMD12	TSC1
		CHAMP1	KMT2A	PTCHD1	TSC2
		CHD2	KMT2C	PTEN	TSHZ3
		CHD3	KMT5B	PTPN11	UBE3A
		CHD7	KRAS	RAF1	UPF3B
		CHD8	LZTR1	RAI1	VPS13B
		CIC	MAGEL2	RALGAPB	WAC
		CNOT3	MAP2K1	RELN	WDFY3
		CREBBP	MAP2K2	RERE	YY1*
		CSDE1	MBD5	RFX3	ZBTB20
		CTCF	MBOAT7	RIMS1	ZNF292
		CTNNB1	MECP2	RIT1	ZNF462
		CUL3	MED13	RORB	
		DDX3X	MED13L	SCN1A	
		DEAF1*	MEF2C	SCN2A	
		DHCR7	MEIS2	SCN8A	
		DLG4		SETBP1	

Chromosomal Differences

Trisomy 21 (Down syndrome)

XO (Turner syndrome)

XXY (Klinefelter syndrome)

XXYY

XXXY

*Only certain variants in these genes are returned.