SIMONS SEARCHLIGHT

Driven by science. United by hope.

The Simons Searchlight Gene List

The Simons Searchlight gene list contains 180 gene changes (orange) and 24 copy number variants (purple) that are known to be associated with autism and other neurodevelopmental disorders. Any result returned by SPARK, another SFARI initiative, are eligible in Simons Searchlight.

Genetic Changes We Study

1q21.1
2p16.3
2q34 duplication
2q37 deletion
3q29
5p deletion
5q35
6q16 deletion
7q11.23
8p23.1
9q34 duplication
15q11.2-q13.1
15q13.3 deletion
15q15 deletion
15q24 deletion
16p11.2*
16p12.2 deletion**
16p13.11
16p13.3 deletion
17p11.2
17q11.2
17q12
17q21.3
22q11.2
22q13.3 deletion

ACTB
ACTL6B
ADNP
ADSL
AFF2
AHDC1
ALDH5A1
ANK2
ANK3
ANKRD11
ARHGEF9
ARID1B
ARX
ASH1L
ASXL3
ATRX
AUTS2
BAZ2B
BCKDK
BCL11A
BRAF
BRSK2
CACNA1C
CAPRIN1
CASK
CASZ1
CDKL5
CHAMP1
CHD2
CHD3
CHD7
CHD8
CIC
CNOT3
CREBBP
CSDE1
CSNK2A1

CTBP1
CTCF
CTNNB1
CUL3
DDX3X
DEAF1
DHCR7
DLG4
DMPK
DNMT3A
DSCAM
DST
DYRK1A
EBF3
EHMT1
EIF3F
EP300
FMR1
FOXG1
FOXP1
GIGYF1
GIGYF2
GRIN1
GRIN2A
GRIN2B
GRIN2D
HIVEP2
HNRNPH2
HNRNPU
HRAS
IQSEC2
IRF2BPL
KANSL1
KAT6A
KATNAL2
KCNB1
KCNQ2

KCNQ3
KDM3B
KDM5B
KDM6B
KRAS
KMT2A
KMT2C
KMT2E
KMT5B*
LZTR1
MAGEL2
MAP2K1
MAP2K2
MBD5
MBOAT7
MECP2
MED13
MED13L
MEIS2
MYT1L
NAA15
NBEA
NCKAP1
NEXMIF**
NF1
NIPBL
NLGN2
NLGN3
NLGN4X
NRAS
NR3C2
NR4A2
NRXN1
NRXN2
NRXN3
NSD1
PACS1

PBRM1
PCDH19
PHF21A
PHF3
PHIP
POGZ
POMGNT1
PPP1CB
PPP2B
PPP2R1A
PPP2R5D
PSMD12
PTCHD1
PTPN11
PTEN
PURA
RAF1
RAI1
RELN
RERE
REST
RFX3
RIMS1
RIT1
RORB
SCN1A
SCN2A
SCN8A
SETBP1
SETD2
SETD5
SHANK2
SHANK3
SHOC2
SIN3A
SLC6A1
SLC9A6

SMARCA4
SMARCC1
SMARCC2
SON
SOS1
SOS2
SOX5
SPAST
SRCAP
STXBP1
SYNCRIP
SYNGAP1
TAOK1
TANC2
TBCK
TBR1
TCF20
TCF4
TLK2
TRIO
TRIP12
TSC1
TSC2
TSHZ3
UBE3A
UPF3B
USP9X
VPS13B
WAC
WDFY3
ZBTB20
ZNF462
ZNF292

* *Includes deletions and duplications that include at a minimum the BP4 - BP5 region (proximal) or BP2 - BP3 (distal) region*

** *Formerly known as 16p12.1 deletion*

* *Formerly known as SUV420H1*

** *Formerly known as KIAA2022*

This list was updated May 2020. **simonssearchlight.org**