

# The Simons Searchlight Gene List

The Simons Searchlight gene list contains 151 gene changes (orange) and 23 copy number variants (purple) that are known to be associated with autism and other neurodevelopmental disorders.

## Genetic Disorders We Study

|                          |         |         |         |         |
|--------------------------|---------|---------|---------|---------|
| 1q21.1                   | ACTB    | DDX3X   | MED13   | SCN8A   |
| 2p16.3 deletion          | ACTL6B  | DEAF1   | MED13L  | SETBP1  |
| 2q34 duplication         | ADNP    | DHCR7   | MEF2C   | SETD2   |
| 2q37 deletion            | ADSL    | DLG4    | MEIS2   | SETD5   |
| 2q37.3 deletion          | AFF2    | DNMT3A  | MYT1L   | SHANK2  |
| 5q35                     | ALDH5A1 | DSCAM   | NAA15   | SIN3A   |
| 6q16 deletion            | AHDC1   | DYNC1H1 | NBEA    | SLC6A1  |
| 7q11.23 duplication      | ANK2    | DYRK1A  | NCKAP1  | SLC9A6  |
| 9q34 duplication         | ANK3    | EBF3    | NEXMIF  | SMARCA4 |
| 15q11.2 BP1-BP2 deletion | ANKRD11 | EHMT1   | NIPBL   | SMARCC2 |
| 15q13.3 deletion         | ARHGEF9 | EIF3F   | NLGN2   | SON     |
| 15q15 deletion           | ARID1B  | EP300   | NLGN3   | SOX5    |
| 15q24 deletion           | ARX     | FOXP1   | NLGN4X  | SPAST   |
| 16p11.2*                 | ASH1L   | GIGYF1  | NR3C2   | SRCAP   |
| 16p12.2 deletion**       | ASXL3   | GNB1    | NR4A2   | STXBP1  |
| 16p13.11 deletion        | ATRX    | GRIN1   | NRXN1   | SYNCRIP |
| 16p13.3 deletion         | AUTS2   | GRIN2A  | NRXN2   | SYNGAP1 |
| 17q11.2 dup              | BCKDK   | GRIN2B  | NSD1    | TANC2   |
| 17p13.3                  | BCL11A  | GRIN2D  | PACS1   | TAOK1   |
| 17q12                    | BRSK2   | HECW2   | PACS2   | TBR1    |
| 17q21.3                  | CACNA1C | HIVEP2  | PHF21A  | TCF20   |
| Xp11.22 duplication      | CAPRIN1 | HNRNPH2 | PHF3    | TCF7L2  |
| Xq28 duplication         | CASK    | HNRNPU  | PHIP    | TLK2    |
|                          | CASZ1   | IQSEC2  | POMGNT1 | TRIO    |
|                          | CHAMP1  | IRF2BPL | PPP3CA  | TRIP12  |
|                          | CHD2    | KANSL1  | PPP2R1A | UPF3B   |
|                          | CHD3    | KATNAL2 | PPP2R5D | USP9X   |
|                          | CHD8    | KCNB1   | PSMD12  | VPS13B  |
|                          | CIC     | KDM3B   | PTCHD1  | WAC     |
|                          | CLCN4   | KDM5B   | RALGAPB | WDFY3   |
|                          | CNOT3   | KDM6B   | RELN    | YY1     |
|                          | CREBBP  | KMT2A   | RERE    | ZBTB20  |
|                          | CSDE1   | KMT2C   | REST    | ZNF292  |
|                          | CSNK2A1 | KMT2E   | RFX3    | ZNF462  |
|                          | CSNK2B  | KMT5B   | RIMS1   |         |
|                          | CTBP1   | MAOA    | RORB    |         |
|                          | CTCF    | MAOB    | SCN1A   |         |
|                          | CTNNB1  | MBD5    | SCN1B   |         |
|                          | CUL3    | MBOAT7  | SCN2A   |         |

\* 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