

Donations with Known Genetic Diagnoses (SNVs or CNVs) *#

Available cases with a genetic variant and number of cases confirmed or under review for ASD as of October 1, 2025

Genetic Finding	Number of Cases with a Genetic Variant**	Number of Cases Confirmed or Under Review for ASD
Angelman Syndrome	3	1
Trisomy 21	4	0
STXBP1	2	0
SETBP1	1	0
SCN2A	5	1
SCN1A	2	1
KMT2A	1	1
GRIN2B	1	0
GRIN1	1	0
FOXG1	2	0
FMR1	2	2
DDX3X	1	0
CDKL5	1	0
BRSK2	1	0
ANK2	1	1
22q13.3 Del	8	6
22q11.2 Del	2	0
1q21.1 Dup	1	0
16p11.2 Dup	1	1
XYY	1	1
UBA5	1	0
TUBA1A	1	0
Trisomy 9	1	1

Ring 14	1	0
PPP21RA	1	0
PCDH15	1	1
PARK2	1	0
Maternal Uniparental Disomy 15	2	1
MACROD2	1	0
KIF1A	4	0
HMBS	1	0
CLCN4	1	0
ATP7B	1	1
AGAP2	1	1
20q11.23	1	1
15q11.1q11.2 Del	1	0
14q12-q13 Del	1	0
7q36.6 Dup	1	1
15q11.2q13.1 Dup	22	20
9q34 Dup	1	0
22q11.2 Dup	1	0
15q11.2q13.1 Del	5	1

*Note that genomic analysis on all cases is being performed at the New York Genome Center, but not all cases have been analyzed yet. Some cases could have a genetic diagnosis still to be discovered. Genomic data is made available for distribution via SFARI Base.

** The total number of donations does not include donations to the legacy program, the Autism Tissue Program, but some tissue remains.

Autism BrainNet accepts donations with a genetic diagnosis, with or without a diagnosis of autism; the information is available in SFARI Base.