Supplementary File 2. Data from case reports of individuals with pathogenic variants in autism-associated genes

Autism gene	ID cases	Autism cases	Autism cases with comorbid ID	Autism cases without ID	Variant types in autism/ID cases	Ref.
ADNP	11/11	7/11	7/7	0/7	Unknown	[1]
	Mild - 9/73 Moderate - 26/73 Severe - 38/73 TOTAL - 73/73	64/73	64/64	0/64	Unknown	[2]
	1/1 - Severe	1/1	1/1	0/1	Nonsense	[3]
	36/36	29/36	29/29	0/29	Missense - 1 Nonsense - 14 Frameshift - 13 Deletion - 1	[4]
	13/13	13/13	13/13	0/13	Unknown	[5]
ANK2	1/1 - Moderate (IQ=40-60)	1/1	1/1	0/1	Frameshift	[6]
ANKRD11	Moderate - 6/7 Mild - 1/7 TOTAL - 7/7	7/7	7/7	0/7	Nonsense - 1 Frameshift - 3 Deletions - 3	[7]
	1/1	1/1	1/1	0/1	Frameshift	[8]
	2/2	1/2	1/1	0/1	Nonsense	[9]
ARID1B	127/143	77/143	77/77	0/77	LOF - 118 Deletions - 18 Splice site - 7	[10]
	1/1 - Severe	1/1	1/1	0/1	Frameshift	[11]
	8/8 - Moderate/Severe	1/8	1/1	0/1	Nonsense	[12]
	1/1	1/1	1/1	0/1	Frameshift	[13]

Autism gene	ID cases	Autism cases	Autism cases with comorbid ID	Autism cases without ID	Variant types in autism/ID cases	Ref.
ASH1L	6/13 - Mild 5/13 - Severe 2/13 - Yes TOTAL - 13/13	3/13	3/3	0/3	Nonsense - 2 Frameshift - 1	[14]
	1/1 - IQ<30	1/1	1/1	0/1	Missense	[15]
ASXL3	1/1	1/1	1/1	0/1	Nonsense	[16]
	3/12 - Moderate 1/12 - Moderate-Severe 6/12 - Severe 1/12 - Profound TOTAL - 11/12	9/12	8/9	1/9	Nonsense - 6 Frameshift - 2	[17]
	1/1 - Severe	1/1	1/1	0/1	Frameshift	[18]
	1/1 - Profound	1/1	1/1	0/1	Frameshift	[19]
	2/2 - Severe	2/2	2/2	0/2	Nonsense - 2	[20]
	1/1	1/1	1/1	0/1	Biallelic Missense	[21]
	1/1	1/1	1/1	0/1	Frameshift	[22]
BCL11A	1/9 - Mild-Moderate 1/9 - Moderate-Severe 6/9 - Moderate 1/9 - Severe TOTAL - 9/9	2/9	2/2	0/2	Missense - 1 Frameshift - 1	[23]
	2/7	2/7	1/2	1/2	Missense	[24]
CHD2	1/1	1/1	1/1	0/1	Frameshift	[25]
	2/2	2/2	2/2	0/2	Nonsense - 1 Splicing - 2	[26]

Autism gene	ID cases	Autism cases	Autism cases with comorbid ID	Autism cases without ID	Variant types in autism/ID cases	Ref.
CHD8	1/10 - Mild 3/10 - Moderate 1/10 - Moderate-Severe 2/10 - Severe TOTAL - 7/10	7/10	6/7	1/7	Nonsense - 4 Splicing - 1 Frameshift - 1	[27]
	7/10	9/10	6/9	3/9	Missense - 2 Nonsense - 4	[28]
	11/11	6/11	6/6	0/6	Unknown	[1]
	1/2 - Severe 1/2 - IQ=33 TOTAL - 2/2	2/2	2/2	0/2	Frameshift - 2	[15]
	11/17	14/17	9/14	5/14	Deletion - 1 Nonsense - 1 Splicing - 1 Frameshift - 3	[29]
	9/15	13/15	8/13	5/13	Nonsense - 4 Splicing - 1 Frameshift - 3	[30]
	1/10 - Below average IQ 1/10 - Mild 3/10 - Low IQ 4/10 - Extremely low IQ TOTAL - 9/10	10/10	9/10	1/10	Deletions - 1 Nonsense - 3 Splicing - 1 Frameshift - 4	[31]
CUL3	1/1 - Mild	1/1	1/1	0/1	Missense	[32]
DDX3X	69/69	27/69	27/27	0/27	Unknown	[33]
	28/28	6/28	6/6	0/6	Unknown	[34]

Autism gene	ID cases	Autism cases	Autism cases with comorbid ID	Autism cases without ID	Variant types in autism/ID cases	Ref.
DYRK1A	6/6	6/6	6/6	0/6	Nonsense - 2 Frameshift - 4	[35]
	1/20	3/20	1/3	2/3	Nonsense	[36]
KMT2A	5/6	5/6	4/5	1/5	Missense - 1 Nonsense - 1 Frameshift - 2	[37]
	0/2	1/2	0/1	1/1	Splicing - 1	[38]
	18/33 - Mild 11/33 - Moderate 4/33 - Severe TOTAL - 33/33	2/33	2/2	0/2	Missense - 2	[39]
	38/58	4/58	4/4	0/4	Unknown	[40]
MECP2	1/1	1/1	1/1	0/1	Nonsense	[41]
MYT1L	1/1 - IQ=60	1/1	1/1	0/1	Nonsense	[15]
NRXN1	11/11 - Severe	1/11	1/1	0/1	Bi-allelic/deletion	[42]
	8/21	12/21	2/12	10/12	Deletions - 2	[43]
	25/27	20/27	19/20	1/20	Deletions - 19	[44]
	1/1	1/1	1/1	0/1	Deletion	[45]
POGZ	9/10	5/10	5/5	0/5	Missense - 1 Nonsense - 3 Duplication - 1	[46]
	1/1	1/1	1/1	0/1	Missense	[47]
	1/1	1/1	1/1	0/1	LOF	[48]
	1/1 - Severe	1/1	1/1	0/1	Frameshift	[49]
	24/25	17/23	17/17	0/17	Intronic - 2 Nonsense - 7 Frameshift - 8	[50]

Autism gene	ID cases	Autism cases	Autism cases with comorbid ID	Autism cases without ID	Variant types in autism/ID cases	Ref.
POGZ	5/5	2/5	2/2	0/2	Nonsense - 1 Frameshift - 1	[51]
	6/6	1/7	1/1	0/1	Nonsense	[52]
	1/1 - Severe	1/1	1/1	0/1	Missense	[53]
SCN2A	1/1	1/1	1/1	0/1	Deletion	[54]
	1/1	1/1	1/1	0/1	Missense	[55]
	1/1 - Severe	1/1	1/1	0/1	Missense	[56]
	1/1	1/1	1/1	0/1	Splicing	[57]
	15/33	5/33	3/5	2/5	Missense - 2 Nonsense - 1	[58]
SETD5	8/9	2/9	2/2	0/2	Intronic - 1 Nonsense - 1	[59]
	7/7	5/7	5/5	0/5	Unknown	[60]
SHANK3	29/30	22/30	21/22	1/22	Missense - 1 Splicing - 2 Nonsense - 4 Frameshift - 14	[61]
	0/1	1/1	0/1	1/1	Frameshift	[62]
	1/1	1/1	1/1	0/1	Frameshift	[63]
	5/5	2/5	2/2	0/2	Unknown	[64]
SYNGAP1	1/1 - Moderate	1/1	1/1	0/1	Frameshift	[65]
	1/8 - Moderate 6/8 - Moderate/Severe 1/8 - Severe TOTAL - 8/8	3/8	3/3	0/3	Missense - 2 Frameshift - 1	[66]
	1/1	1/1	1/1	0/1	Splicing	[67]
	1/1 - Mild	1/1	1/1	0/1	Missense	[68]
	1/1 - Severe	1/1	1/1	0/1	Splicing	[69]

Autism gene	ID cases	Autism cases	Autism cases with comorbid ID	Autism cases without ID	Variant types in autism/ID cases	Ref.
SYNGAP1	1/11 - Mild 6/11 - Moderate 4/11 - Moderate/Severe TOTAL - 11/11	4/11	4/4	0/4	Missense - 2 Splicing - 1 Frameshift - 1	[70]
TCF20	16/16	9/16	9/9	0/9	Missense - 1 Nonsense - 1 Deletion - 3 Frameshift - 4	[71]
	26/26	18/26	18/18	0/18	Unknown	[72]
	1/5 - borderline (IQ=79) 2/5 - Mild 1/5 - Mild-Moderate 1/5 - Moderate TOTAL - 5/6	5/6	4/5	1/5	Rearrangement - 2 Frameshift - 2	[73]

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