SUPPLEMENTARY MATERIALS

Rare genetic variants in *CX3CR1* and their contribution to the increased risk of schizophrenia and autism spectrum disorders

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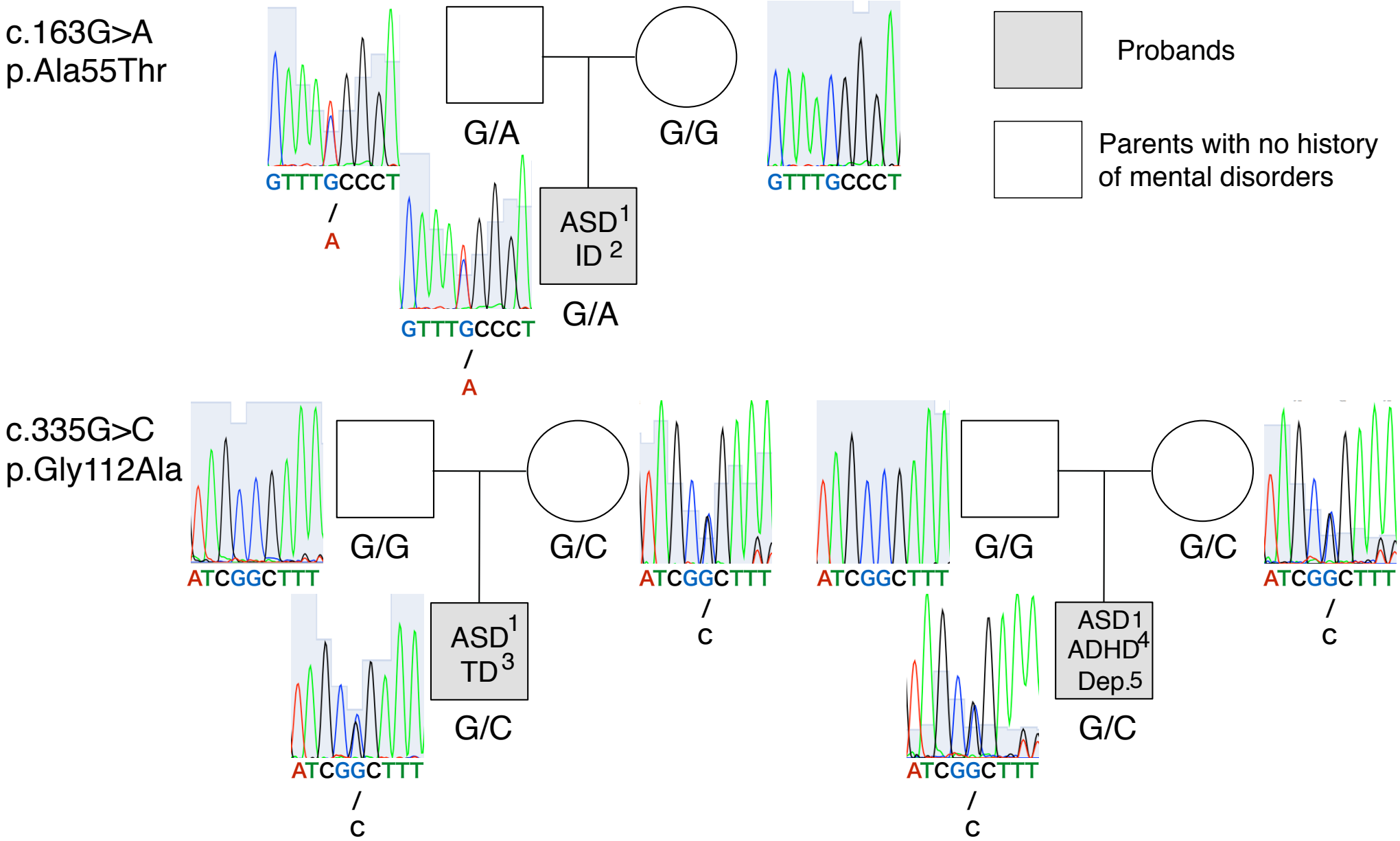

S1 Fig. Family-trio analysis with parental genotype data

Note: The genotypes of the tested individuals are indicated on the lower-side.

All comorbidities were diagnosed by experienced psychiatrists according to *Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition* (DSM-5) criteria.

c, coding DNA; p, protein

[1]Autism spectrum disorder; [2]Intellectual disability; [3]Tic disorder; [4]Attention deficit hyperactivity disorder; [5]Depression

S1 Table. Association results for each phenotype

					ASD				SCZ				Control	
Exon	Ref	Val	Position	Variant	Allele Count[a]	MAF	Odds Ratio	*P* value[b]	Allele Count[a]	MAF	Odds Ratio	*P* value[b]	Allele Count[a]	MAF
2	G	A	3:39266347	p.Ala55Thr	2/1148	0.0017	13.3 [1.04-Inf]	0.047	5/5306	0.00099	7.2 [1.04-Inf]	0.045	1/7652	0.00013

Note: Genomic position based on NCBI builds GRCh38 (Ensembl Transcript ID ENST000000399220).
Amino acid position is based on NCBI reference sequence NP_001328.1.
Ref, reference; Val, variant; MAF, minor allele frequency; ASD, autism spectrum disorders;
SCZ, schizophrenia; p, protein
[a]minor allele / major allele; [b]P values were calculated by one-tailed Fisher's exact test

S2 Table. Clinical characteristics in individuals with CX3CR1-Ala55Thr

Diagnosis	SCZ	SCZ	SCZ	SCZ	SCZ	ASD	ASD	HC
Age of eveluation (years)	45	52	62	65	67	19	31	34
Gender	M	M	M	M	M	M	F	F
Congenital and developmental phenotypes								
Developmental delay	-	NA	-	-	NA	+	-	NA
Autistic-like symptoms	-	NA	-	-	NA	+	+	NA
Seizures	-	NA	-	-	NA	-	-	NA
Educational years	16	NA	8	9	NA	12 (Special needs)	14	NA
Occupation	Office worker	NA	Office worker	-	Office worker	-	Temporary worker (employee with a disability)	NA
Marriage	-	-	-	-	+	-	-	NA
Age at psychosis onset (years)	35	19	19	14	30	-	14	-
Core symptoms						-	-	-
Persecutory and grandiose delusions	+	+	-	-	+			
Disorganized speech	+	+	+	+	+			
Grossly disorganized behavior	+	+	-	+	+			
Catatonic behavior with mutism and stupor	-	-	+	-	-			
Cognitive decline	+	NA	+	+	+			
Other neuropsychiatric features	Risperidone-induced dyskinesia	-	-	-	-	Firesetting behavior		-
Hospitalizations	Three times 1.6 years	Two time 13 years	Two times 12 years	Two times 49 years	Three times 30 years	-	-	-
Treatment	Perospirone 40mg/d	NA	Electroconvulsive therapy Insulin shock therapy	Chlorpromazine 150mg/d	NA	-	-	-
Brain imaging	No significant findings	NA	NA	NA	NA	NA	NA	NA
Other physiological features	DM, HT, HC	NA	-	-	NA	-	Irritable bowel syndrome Premenstrual syndrome	-
Psychiatric disorders in first-degree relatives	Depression (Mother)	SCZ (Brother)	-	-	-	NA	-	-

Note: SCZ, schizophrenia; ASD, autism spectrum disorders; HC, healthy control; M, male; F, female; +, feature present; −, feature absent; NA, not assessed

S3 Table. Primer sequences for validating each variant

Variant	Forward primer (5'-3')	Reverse primer (3'-5')
p.Ala55Thr p.Gly112Ala	AATGCTGATGACGGTGATGAAGAA	TCTACTCCGTCATCTTTGCCATTG
p.Met138Ile	GGGTAGTCACCAAGGCATTCATT	GCCTTCTTCTTCATCGGCTTTT

S4 Table. Probe sequences for TaqMan SNP assays

Variant	Forward primer (5'-3')	Reverse primer (3'-5')	Reporter 1 (5'-3')	Reporter 2 (5'-3')
p.Ala55Thr	CATTGGCCTGGTGGGAAATTTG	CTCTTGGGCTTCTTGCTGTTG	VIC-TTGGTAGTGTTTGCCCTCAC-NFQ	FAM-TTGGTAGTGTTTACCCTCAC-NFQ

Note: A TaqMan probe consists with a FAM or VIC dye label on the 5' end, and nonfluorescent quencher (NFQ) on the 3' end