

Supplementary Table 1. *SHANK3* variations identified in families with ASD and controls.

	Variations		Sex of proband	Transmission and ethnicity			Additional family information	Occurrence	
	Exon	Change ^a		Parents	Siblings	Ethnicity		ASD (n=227)	Controls ^b (n=190-333)
ASD									
E1	R12C	Male	Mother	Present in one unaffected brother and absent in one unaffected brother	European	The mother has social phobia traits; the father and the two brothers are healthy	1	0	
E5	A198G	Female	Mother	Absent in the affected brother	European	No history of psychiatric disorders in either parent	1	0	
E8	R300C	Male	Mother	Absent in the unaffected sister	Asian	Language delay in the father; the mother is healthy	1	0 ^c	
E21a	G1011V	Female	Mother	(3 brothers, DNA not available)	European	No history of psychiatric disorders in any family member	2	0	
E21a	G1011V	Male	Father	Present in the affected monozygotic twin	European	No history of psychiatric disorders in either parent			
E21a	R1066L	Male	Mother	Present in the unaffected sister	North African	The father has a depressive syndrome; the mother had depression during pregnancy	1	0	
E21a	ins(G)3680 at amino acid 1227	Male	<i>De novo</i> (germinal mosaicism in the mother)	Present in the two affected brothers and absent in the unaffected brother	European	No history of psychiatric disorders in either parent	1	0	
E21a	R1231H	Female	Mother	Absent in the affected brother	European	No history of psychiatric disorders in either parent	1	0	
E22b	S1566G	Male	Father	(2 unaffected siblings, DNA not available)	European	No history of psychiatric disorders in any family member	1	0	
ASD and Controls									
E21a	A963G	Female	Mother	(1 healthy brother, DNA not available)	European	No history of psychiatric disorders in any family member	2	1	
E21a	A963G	Male	Father	(1 brother, DNA not available)	European	The father has broad autism phenotype (very shy, some autistic traits); the younger brother has mental retardation and autistic traits (not autism)			
E22c	P1654T	Male	Father	Absent in the affected brother	European	The father talked late and has stereotyped body movements; the mother also was late to talk.	2	1	
E22c	P1654T	Male	Nd	(1 sister, DNA not available)	European	No history of psychiatric disorders in either parent or the sister			
Controls									
E6	A224T	Female	—	—	European		0	1	
E21a	V1333G	Male	—	—	European		0	2	

^a *SHANK3* cDNA sequence: NM_033517.1. ^b Exon 8 was sequenced in 333 controls; exons 1, 4-7, 10, 12, 18-20, 21a, and 22abc were sequenced in 270 controls; and exons 2, 3, 9, 11, and 13-17 were sequenced in 190 controls. Nd = not determined.