

Table S5. Descriptive and diagnostic data by patient

Subject	Age (y)	NVIQ estimate (standard score/DQ)	Vineland Adaptive Behavior Scales-II (standard score)					ADI-R ^a			ADOS-2 ^b				DSM-5	Consensus diagnosis	Peabody Picture Vocabulary Test-4 (standard score)	Expressive Vocabulary Test-2 (standard score)	
			ABC	Communication	Socialization	Daily Living	Motor	A: Social	B: Comm	C: RRSB	Module	Social Affect	Restricted & Repetitive Behavior	Overall Total					Severity Score
S1	12	10.4	33	36	38	33	56	23	13	6	1	20	7	27	10	ASD	ASD	ND	ND
S2	5	30.6	49	44	55	43	61	16	12	3	1	16	3	19	6	ASD	ASD	ND	ND
S3	7	18.8	49	45	51	52	40	17	10	7	1	2	3	5	2	No ASD	No ASD	ND	ND
S4	3	19.5	55	49	68	55	59	13	12	2	1	19	7	26	10	ASD	ASD	ND	ND
S6	5	35	53	54	49	53	64	25	21	6	1	15	5	20	8	ASD	ASD	ND	ND
S7	7	74	75	75	87	69	75	12	11	1	3	12	3	15	9	No ASD	No ASD	75	70
S8	9	14.6	53	53	51	54	59	28	14	6	1	20	8	28	10	ASD	ASD	ND	ND
S10	9	60	72	72	78	71	88	6	4	3	3	5	1	6	3	No ASD	No ASD	78	70
S11	6	17.3	42	36	53	34	51	28	14	7	1	18	2	20	7	ASD	ASD	ND	ND
S12	42	0.97	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
S13	15	15.5	36	37	43	35	59	29	13	4	1	14	5	19	6	ASD	ASD	ND	ND
S14	4	33	49	42	61	46	56	21	13	5	1	18	2	20	7	ASD	ASD	60	ND

^a ADI-R cutoff scores for autism are: Social = 10, Communication (nonverbal) = 7, Repetitive behaviors and restricted interests = 3.

^b ADOS-2 Module 1 (Pre-verbal/single words) cutoff scores for autism and ASD are 16 and 11 for *few to no words*, and 12 and 8 for *some words*. ADOS-2 Module 3 (Fluent speech) cutoff scores for autism and ASD are 9 and 7, respectively.

ABC Adaptive Behavior Composite, ADI-R Autism Diagnostic Interview-Revised, A: *Social* Qualitative abnormalities in reciprocal social interaction, B: *Comm* Communication, C: *RRSB* Restricted, repetitive, or stereotyped patterns of behavior, ADOS-2 Autism Diagnostic Observation Schedule, Second edition, DQ developmental quotient, DSM-5 Diagnostic and Statistical Manual of Mental Disorders, Fifth edition, IQ intellectual quotient, NVIQ nonverbal IQ, ND not done, y years

Table S6. ASD and intellectual ability classifications in individuals with *SHANK3* mutations

Classification	n	%
ASD classification (n=11)		
ASD classification on ADOS	9	82
ASD classification on ADI-R	8	73
Consensus diagnosis of ASD	8	73
Nonverbal IQ classification (n=12)		
Mild ID (IQ 55-69)	2	17
Severe ID (IQ 25-40)	3	25
Profound ID (IQ <25)	7	58

ASD autism spectrum disorder, *ADI-R* Autism Diagnostic Interview-Revised, *ADOS-2* Autism Diagnostic Observation Schedule, Second Edition, *ID* intellectual disability, *IQ* intellectual quotient

Table S7. Language and motor functioning in individuals with *SHANK3* mutations

Domain	Mean age equivalent \pm SD in months (range)
Language	
Receptive language	
Mullen	10.0 \pm 3.1 (5-14)
Vineland-II	12.6 \pm 5.3 (6-21)
Expressive language	
Mullen	7.9 \pm 6.0 (2-23)
Vineland-II	10.2 \pm 4.2 (7-21)
Motor	
Fine motor skills	
Mullen	16.7 \pm 5.3 (9-27)
Vineland-II	20.4 \pm 10.5 (6-36)
Gross motor skills	
Mullen	22.2 \pm 4.7 (16-28)
Vineland-II	26.8 \pm 6.1 (17-36)

Language and motor age equivalents in individuals assessed with both the Mullen Scales of Early Learning and the Vineland Adaptive Behavior Scales II (n=9; S1-S4, S6, S8, S11, S13, and S14).