Table S3. Phenotypic information of individuals from the UK10K cohort with rare SATB1 missense variants. Wechsler Intelligence Scale for Children (WISC) test scores for individuals from the UK10K cohort, carrying rare SATB1 missense variants. Standard deviation scores (std score) were calculated by comparing individual scores of carriers to the mean test scores from UK10K non-carriers. Test scores that were lower compared to mean non-carrier scores are shaded in red, while test score that were higher compared to mean non-carrier scores are shaded in green. All carrier test scores were within 2.5 standard deviations compared to the mean non-carrier scores, and thus within normal range.

	UK10K non-carriers (n=1732, ±Std)	UK10K carriers (n=9, ±Std)	UK10K carriers								
Variant	-	-	rs148337599	rs148337599	rs148337599	rs148337599	rs148337599	rs760272331	rs760272331	rs185604711	rs185604711
Residue change (SATB1 NM_001131010.4)	-	-	p.S366L	p.S366L	p.S366L	p.S366L	p.S366L	p.V519L	p.V519L	p.A573T	p.A573T
gnomAD v2.1.1 frequency			6.61e-4 (allele 282848)					8.67e-6 (allele 230660)		1.17e-4 (allele 282890)	
WISC - Verbal IQ: F@8	111.92 (±16.57)	116.89 (±16.07)	103	133	139	103	111	133	128	99	103
VIQ std score	-	0.30 (±0.97)	-0.54	1.27	1.63	-0.54	-0.06	1.27	0.97	-0.78	-0.54
WISC - Performance IQ: F@8	103.49 (±16.79)	109.00 (±15.33)	104	125	115	119	109	115	90	80	124
PIQ std score	-	0.33 (±0.91)	0.03	1.28	0.69	0.92	0.33	0.69	-0.80	-1.40	1.22
WISC - Total IQ: F@8	109.12 (±16.00)	114.89 (±14.73)	104	133	132	111	111	130	111	88	114
IQ std score	-	0.36 (±0.92)	-0.32	1.49	1.43	0.12	0.12	1.30	0.12	-1.32	0.31
WISC - Verbal Comprehension Index: F@8	48.47 (±11.10)	51.79 (±12.51)	38	63	72	44	47	58	63	37	44
VCI std score	-	0.30 (±1.13)	-0.94	1.31	2.12	-0.40	-0.13	0.86	1.31	-1.03	-0.40
WISC - Perceptual Organisation Index: F@8	41.51 (±10.50)	43.80 (±8.37)	41	49	50	53	44	50	30	31	47
POI std score	-	0.23 (±0.80)	-0.05	0.71	0.81	1.09	0.24	0.81	-1.10	-1.00	0.52
WISC - Freedom from Distractability Index: F@8	22.17 (±5.96)	24.11 (±5.42)	27	26	22	22	28	35	19	20	18
FDI std score	-	0.33 (±0.91)	0.81	0.64	-0.03	-0.03	0.98	2.15	-0.53	-0.36	-0.70

≤-1.0 Std score compared to UK10K non-carriers
≤-0.0 Std score compared to UK10K non-carriers
≥1.0 Std score compared to UK10K non-carriers
≥0.0 Std score compared to UK10K non-carriers