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Tables

Table 1: Cohort demographics

EU, Europe; US, United States.

	Cample	S	ex	Age	9	Autism		
Cohort	Sample size	n (male)	% (male)	mean	SD	n	%	
Genetic first cohorts								
16p11.2 deletion	82	43	52%	9.6	3.7	35	43%	
EU	12	7	58%	10.3	4.4	7	58%	
US	70	36	51%	9.5	3.6	28	40%	
16p11.2 duplication	50	35	70%	10.8	6.7	29	58%	
EU	17	16	94%	12.6	8.6	13	76%	
US	33	19	58%	9.9	5.4	16	48%	
22q11.2 deletion	370	182	49%	13.4	3.4	85	23%	
EU	215	102	47%	13.7	2.4	50	23%	
US	155	80	52%	12.9	4.4	35	23%	
22q11.2 duplication	45	35	78%	10.1	4.3	20	44%	
EU	11	9	82%	10.3	2.8	6	55%	
US	34	26	76%	10.0	4.7	14	41%	
Heterogeneous								
autism	2027	1753	86%	9.1	4.9	2027	100%	
EU	848	731	86%	8.6	4.8	848	100%	
US	1179	1022	87%	9.5	5.0	1179	100%	

Table 2: Comparison of CNV groups on IQ and autism measures

FSIQ, Full Scale Intelligence Quotient; VIQ, Verbal Intelligence Quotient; PIQ, Performance Intelligence Quotient; ADI-R, Autism Diagnostic Interview; RRB, Restricted, Repetitive, and Stereotyped Behaviours.

Separate MANCOVA analyses were ran for domain and subdomain scores to avoid including mathematically related scores in the same analysis. Total ADI-R score was analysed separately using an ANCOVA model. Age, gender and site were included as covariates. Post hoc contrasts are given in Supplementary Table 7.

Bold p-values indicate that the p-value is significant after BH-FDR 0.05 correction

*significant after correcting for IQ (Full results in Supplementary Table 3)

	16p11.2 deletion		16p11.2 duplication		22q11.2 deletion			22q11.2 duplication		Between group variation		Within group variation			
Score	N	mean	sd	N	mean	sd	N	score	sd	N	mean	sd	p-value	eta-squared %	eta-squared %
Cognitive scores															
FSIQ	80	81.3	15.4	50	70.9	22.4	337	70.3	13.5	32	88.1	21.1	<0.001	11.7	82.2
VIQ	80	78.2	18.6	49	73.6	24.4	329	72.8	13.9	32	84.2	18.9	<0.001	3.7	91.7
PIQ	81	85.7	15.2	50	71.9	22.4	333	72.3	14.3	32	87.1	21.7	<0.001	11.8	84.8
ADI-R scores															
Total score	82	22.0	11.8	50	27.7	14.0	370	16.7	11.9	45	21.2	15.4	<0.001*	7.4	88.7
Social domain	82	12.9	7.5	50	15.8	8.0	370	10.1	7.2	45	12.4	9.5	<0.001*	5.4	91.3
subdomains:															
Social interaction	82	2.4	1.9	50	2.9	2.0	370	2.2	2.0	45	2.5	2.2	0.120	1.0	96.6
Peer relationships	82	4.2	2.3	50	4.9	2.6	370	3.5	2.4	45	3.8	2.8	<0.001*	3.1	94.2
Shared enjoyment	82	2.5	2.2	50	3.2	2.0	370	1.7	1.8	45	2.5	2.4	<0.001*	5.7	90.1
Socioemotional reciprocity	82	3.9	2.5	50	4.8	2.5	370	2.7	2.2	45	3.7	2.9	<0.001*	7.9	89.8
Communication domain	82	5.8	3.7	50	6.5	4.9	370	4.5	3.8	45	5.4	4.3	<0.001*	3.1	93.6
subdomains:															
Gestures	82	2.4	2.5	50	3.1	2.9	370	1.7	2.3	45	2.7	2.6	<0.001*	3.6	94.7
Imagination & Imitation	82	3.5	1.8	50	3.4	2.3	370	2.8	2.0	45	2.7	2.1	0.009*	2.0	92.2
RRB domain	82	3.3	2.2	50	5.3	2.6	370	2.1	2.3	45	3.3	2.9	<0.001*	14.7	81.7
subdomains															
Unusual interests	82	0.8	1.0	50	1.6	1.1	370	8.0	1.0	45	0.9	1.0	<0.001*	4.2	92.8
Routines & rituals	82	0.5	0.9	50	1.1	1.3	370	0.5	1.0	45	8.0	1.1	<0.001*	3.2	96.2
Motor mannerisms	82	0.9	0.9	50	1.2	0.9	370	0.2	0.5	45	8.0	0.9	<0.001*	20.9	77.8
Sensorimotor interests	82	1.2	0.8	50	1.5	0.7	370	0.5	0.7	45	8.0	0.9	<0.001*	18.8	74.0