

Whole exome sequencing in extended families with autism spectrum disorder implicates four candidate genes

Human Genetics

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Online Resource 1: selected phenotype data for affected individuals. ASD=Autism Spectrum Disorder, BAP=Broader Autism Phenotype. Age at time of IQ testing is shown only if it is different from the age at which ADOS was administered. IQ scores are for age-appropriate Wechsler tests unless otherwise noted (†=Mullen).

Family	ID	dx	Sex	Age yrs(mo)	Version	Module	ADOS scores			Severity Score	IQ scores		
							Social affect	Rest. and rep. behaviors	Total		Age yrs(mo)	Piq	viq
AU119	2043	ASD	M	11(1)	WPS	3	6	1	7	4		120	97
AU119	2039	ASD	M	9(7)	WPS	3	10	1	11	7		133	128
AU119	2037	ASD	F	3(1)	G	1	9	3	12	6	15(3)	89	94
AU119	2036	ASD	M	6(2)	G	3	7	0	7	4		66	134
AU119	2033	ASD	F	18(1)	WPS	4	7	0	7	4		97	87
AU119	2031	ASD	M	11(4)	G	3	10	3	13	8		117	100
AU119	2030	ASD	M	13(1)	G	4	10	4	14	8		97	108
AU119	2029	ASD	F	14(7)	G	4	19	3	22	10		128	106
AU119	2026	ASD	M	9(0)	G	3	12	3	15	9		114	128
AU119	2027	ASD	M	8(10)	G	3	18	3	21	10	8(8)	125	128
AU119	2048	ASD	M	4(1)	WPS	2	5	3	8	5		126	75
AU625	16028	ASD	M	9(2)	WPS	3	4	2	6	3		100	136
AU625	16030	ASD	M	3(11)	WPS	2	10	1	11	6		-	-
AU625	16023	ASD	M	5(2)	G	2	19	6	25	10	11(0)	78	53
AU366	27012	ASD	F	4(9)	WPS	3	5	2	7	4		114	72
AU366	27013	ASD	F	6(3)	G	3	1	1	2	1		106	75
AU366	27014	ASD	M	3(8)	G	2	4	1	5	2		67†	64†
AU071	1003	ASD	M	11(9)	G	3	8	2	10	6		67	97
AU071	1004	ASD	M	10(6)	G	3	10	2	12	7		78	103
AU071	1005	BAP	M	7(2)	G	3	1	1	2	1		122	103
AU071	1013	ASD	F	12(5)	WPS	3	6	1	7	4		89	106
AU754	24017*	BAP	F									64†	75†
AU754	24016	ASD	F	6(5)	WPS	1	17	3	20	8		80	50
AU754	24015	ASD	F	7(9)	WPS	3	10	1	11	7		103	70

AU754	24011	BAP	M	19(5)	WPS	4	1	3	4	2		145	135
AU754	24009	BAP	M	7(10)	WPS	3	3	0	3	2		97	94
AU599	6003	ASD	M	9(3)	WPS	3	7	2	9	6		103	114
AU599	6004	ASD	M	5(4)	WPS	2	6	1	7	3		103	69
AU599	6015	ASD	M	18(6)	WPS	4	11	1	12	7		89	92
AU599	6014	ASD	M	20(5)	WPS	4	9	1	10	6		111	95
AU113	26013	ASD	M	32(11)	WPS	4	13	3	16	9		80	76
AU113	26011	ASD	M	11(5)	G	2	14	4	18	8		92	50
AU113	26010	ASD	M	14(2)	G	4	11	1	12	7	14(7)	120	111
AU640	21013	ASD	F	16(8)	WPS	3	11	4	15	9		100	106
AU640	21015	ASD	M	12(7)	WPS	3	17	5	22	10		117	81
AU640	21014	ASD	M	15(8)	WPS	3	6	4	10	6		89	89
AU136	18006	ASD	M	11(2)	G	3	7	1	8	5		69	78
AU136	18007	ASD	F	6(8)	G	2	12	1	13	6		108	50
AU136	18008	ASD	F	4(1)	G	1	16	1	17	6		58†	27†
AU136	18003	ASD	M	40(10)	WPS	2	12	5	17	**		55	55
AU053	25011	ASD	M	16(4)	G	3	5	1	6	3		83	59
AU053	25013	ASD	M	11(9)	G	2	14	3	17	7		97	50
AU053	25014	ASD	M	39(5)	WPS	4	5	1	6	3		108	89

\*ADOS scores were unavailable on this individual. Diagnosis was based on ADI-R and professional judgment.

\*\* The severity score could not be computed for AU136-18003, because this subject is older than the maximum age for this score.