Whole exome sequencing in extended families with autism spectrum disorder implicates four candidate genes
Human Genetics
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Online Resource 3: Variants with association test p-value less than or equal to 0.05, or Mendelian inheritance pattern, by family.

Table OR3a: AU119 variants with a segregation pattern consistent with Mendelian inheritance or association p ≤ 0.05. Bold indicates variants with p≤0.05. Pattern denotes whether the variant was present in a pattern consistent with Mendelian inheritance, based on WES only.

							Mean dosage					Predicted function		
chr	**position**	**rsname**	**ref**	**alt**	**pattern**	**p-value**	**affected**	**unaffected**	**1KGP**	**dbSNP**	**GVS**	**DBSNP**	**Gene**
7	**121,944,239**	**.**	**T**	**C**	**yes**	**0.007**	**0.87**	**0.44**	**0**	**dbSNP_134**	**synonymous**	**missense**	***FEZF1***
22	**38,071,707**	**rs75882122**	**A**	**G**	**no**	**0.025**	**0.49**	**0.14**	**0.05**	**dbSNP_52**	**utr-5**	**utr--5**	***LGALS1***

Table OR3b: AU625 variants with a segregation pattern consistent with Mendelian inheritance or association p≤ 0.05. Bold indicates variants with p≤0.05. Pattern denotes whether the variant was present in a pattern consistent with Mendelian inheritance, based on WES only. * indicates genes known to have a high rate of false positives in exome sequencing as indicated by Fajardo et al 2011 (Detecting false positive signals in exome sequencing, Human Mutation 33(4):609-613).

							Mean dosage				Predicted function		
chr	**position**	**rsname**	**ref**	**alt**	**pattern**	**p-value**	**affected**	**unaffected**	**1KGP**	**dbSNP**	**GVS**	**DBSNP**	**Gene**
1	22,165,963	rs139500146	T	C	yes	0.12	1.00	0.35	0.02	dbSNP_134	missense	missense	*HSPG2**
1	22,181,360	rs116788687	G	C	yes	0.12	1.00	0.36	0.02	dbSNP_132	missense	missense	*HSPG2**
5	**102,490,411**	**rs35671301**	**T**	**G**	**yes**	**0.00**	**1.00**	**0.02**	**0.01**	**dbSNP_126**	**missense**	**missense**	***HISPPD1***

Table OR3c: AU366 variants with a segregation pattern consistent with Mendelian inheritance or association p≤ 0.05. Bold indicates variants with p≤0.05. Pattern denotes whether the variant was present in a pattern consistent with Mendelian inheritance, based on WES only.

							Mean dosage				Predicted function		
chr	**position**	**rsname**	**ref**	**alt**	**pattern**	**p-value**	**Affected**	**unaffected**	**1KGP freq**	**dbSNP**	**GVS**	**DBSNP**	**Gene**
7	23,306,141	rs75801644	G	A	yes	0.47	1.00	0.57	0.03	dbSNP_131	missense	missense	*GPNMB*
22	**26,688,831**	**rs137203**	**G**	**T**	**yes**	**0.00**	**1.00**	**0.01**	**0.01**	**dbSNP_78**	**missense**	**missense**	***SEZ6L***

Table OR3d: AU071 variants with a segregation pattern consistent with Mendelian inheritance or association p≤ 0.05 Bold indicates variants with p≤0.05. Pattern denotes whether the variant was present in a pattern consistent with Mendelian inheritance, based on WES only. .* indicates genes known to have a high rate of false positives in exome sequencing as indicated by Fajardo et al 2011 (Detecting false positive signals in exome sequencing, Human Mutation 33(4):609-613). ** this variant did not have a segregation pattern consistent with Mendelian inheritance due to missing genotypes in two affected individuals.

							Mean dosage				Predicted function		
chr	**position**	**rsname**	**ref**	**alt**	**pattern**	**p-value**	**Affected**	**unaffected**	**1KGP**	**dbSNP**	**GVS**	**DBSNP**	**Gene**
1	**877,523**	**rs200195897**	**C**	**G**	**No****	**0.01**	**0.82**	**0.04**	**0**	**dbSNP_137**	**missense**	**missense**	***SAMD11***
1	3,697,663	rs11547615	C	T	yes	0.12	1.00	0.34	0.03	dbSNP_120	missense	missense	*LRRC47*
1	3,755,638	.	C	T	yes	0.12	1.00	0.34	0	none	missense	unknown	*CEP104*
4	663,916	rs61760239	C	A	yes	0.13	1.00	0.38	0.03	dbSNP_129	utr-3	utr-3-prime	*PDE6B*
4	166,388,900	rs144727363	C	T	yes	0.12	1.00	0.34	0	dbSNP_134	missense	missense	*CPE*
4	169,317,237	rs202066594	C	G	yes	0.12	1.00	0.34	0	dbSNP_137	missense	missense	*DDX60L**
4	173,730,541	rs144181630	A	G	yes	0.12	1.00	0.34	0	dbSNP_134	missense	missense	*GALNTL6*
10	27,497,191	rs7918793	G	A	yes	0.13	1.00	0.45	0.05	dbSNP_116	missense	missense	*ACBD5*
12	16,342,622	rs111967344	G	A	yes	0.12	1.00	0.34	0.01	dbSNP_132	missense	missense	*SLC15A5*
12	18,865,819	rs144902254	A	G	yes	0.12	1.00	0.34	0	dbSNP_134	missense	missense	*PLCZ1*
12	20,876,168	rs144285413	C	T	yes	0.12	1.00	0.34	0.01	dbSNP_134	missense	missense	*SLCO1C 1*
12	21,196,367	.	T	A	yes	0.12	1.00	0.34	0	none	missense	unknown	*SLCO1B 7*
12	21,196,466	.	C	A	yes	0.12	1.00	0.34	0	none	missense	unknown	*SLCO1B 7*
12	27,648,704	.	TAGATACGGAAG	T	yes	0.38	1.00	0.68	0	none	frameshift	unknown	*C12orf70*
12	29,630,166	rs189316643	C	T	yes	0.38	1.00	0.67	0.003	dbSNP_135	splice-3	splice-donor-variant	*OVCH1*
12	40,740,686	rs33995883	A	G	yes	0.39	1.00	0.69	0.02	dbSNP_126	missense	missense	*LRRK2*
12	49,168,798	rs3730071	C	A	yes	0.39	1.00	0.69	0.02	dbSNP_107	missense	missense	*ADCY6*
12	52,579,331	rs140694010	C	T	yes	0.38	1.00	0.69	0	dbSNP_134	missense	missense	*KRT80*
19	57,184,265	rs200338534	T	G	yes	0.12	1.00	0.37	0	dbSNP_137	near-gene-5	unknown	*none*

Table OR3e: AU754 variants with a segregation pattern consistent with Mendelian inheritance or association p≤ 0.05. Bold indicates variants with p≤0.05. Pattern denotes whether the variant was present in a pattern consistent with Mendelian inheritance, based on WES only. . * indicates genes known to have a high rate of false positives in exome sequencing as indicated by Fajardo et al 2011 (Detecting false positive signals in exome sequencing, Human Mutation 33(4):609-613) ** this variant did not have a Mendelian segregation pattern because the genotype was missing in one of the WES individuals.

							Mean dosage				Predicted function		
chr	**pos**	**rsname**	**ref**	**alt**	**pattern**	**p-value**	**affected**	**unaffected**	**1KGP**	**dbSNP**	**GVS**	**DBSNP**	**Gene**
1	**214,816,297**	**rs3795514**	**A**	**G**	**yes**	**0.03**	**1.00**	**0.35**	**0.04**	**dbSNP_107**	**missense**	**missense**	***CENPF****
1	**216,062,273**	**rs189748047**	**C**	**T**	**yes**	**0.03**	**1.00**	**0.34**	**0**	**dbSNP_135**	**missense**	**missense**	***USH2A****
7	**72,992,858**	**rs76029572**	**C**	**G**	**No****	**0.04**	**0.97**	**0.35**	**0.04**	**dbSNP_131**	**missense**	**missense**	***TBL2***
10	**98,031,160**	**rs7916154**	**T**	**C**	**yes**	**0.03**	**1.00**	**0.33**	**0.01**	**dbSNP_116**	**utr-5**	**utr-variant-5-prime**	***BLNK***
10	**98,155,678**	**rs41291628**	**G**	**A**	**yes**	**0.03**	**1.00**	**0.34**	**0.02**	**dbSNP_127**	**missense**	**missense**	***TLL2***
10	**98,742,043**	**rs112594620**	**A**	**G**	**yes**	**0.03**	**1.00**	**0.33**	**0.02**	**dbSNP_132**	**missense**	**missense**	***C10orf12***
10	**102,089,635**	**rs117403721**	**C**	**A**	**yes**	**0.03**	**1.00**	**0.33**	**0.01**	**dbSNP_132**	**missense**	**missense**	***PKD2L1***
11	18,740,269	rs201466035	G	A	yes	0.23	1.00	0.67	0	dbSNP_137	missense	missense	*IGSF22*
12	21,695,439	rs61733199	T	C	yes	0.23	1.00	0.67	0.02	dbSNP_129	missense	missense	*GYS2*
12	27,648,710	rs3751223	C	T	yes	0.25	1.00	0.68	0.05	dbSNP_107	missense	missense	*C12orf70*
12	29,449,986	.	G	A	yes	0.23	1.00	0.67	0	none	missense	unknown	*FAR2*
21	**28,210,457**	**rs71317487**	**T**	**C**	**yes**	**0.03**	**1.00**	**0.34**	**0.004**	**dbSNP_130**	**missense**	**missense**	***ADAMTS1***
21	**40,190,408**	**rs61735785**	**C**	**A**	**yes**	**0.03**	**1.00**	**0.34**	**0.01**	**dbSNP_129**	**missense**	**missense**	***ETS2***

Table OR3f: AU599 variants with a segregation pattern consistent with Mendelian inheritance or association p≤ 0.05. Bold indicates variants with p≤0.05. Pattern denotes whether the variant was present in a pattern consistent with Mendelian inheritance, based on WES only. . * indicates genes known to have a high rate of false positives in exome sequencing as indicated by Fajardo et al 2011 (Detecting false positive signals in exome sequencing, Human Mutation 33(4):609-613).

							Mean dosage				Predicted function		
chr	**position**	**rsname**	**ref**	**alt**	**pattern**	**p-value**	**affected**	**unaffected**	**1KGP**	**dbSNP**	**GVS**	**DBSNP**	**Gene**
1	228,033,258	rs61741607	T	A	Yes	0.08	1.01	0.41	0.03	dbSNP_129	missense	missense	*PRSS38*
6	**84,904,604**	**rs17790493**	**G**	**C**	**No**	**0.01**	**1.51**	**0.22**	**0.04**	**dbSNP_123**	**missense**	**missense**	***KIAA1009***
6	**90,024,862**	**.**	**A**	**G**	**Yes**	**0.01**	**1.00**	**0.20**	**0**	**dbSNP_134**	**missense**	**missense**	***GABRR2***
7	**142,836,646**	**rs75076193**	**C**	**T**	**Yes**	**0.00**	**1.00**	**0.00**	**0.01**	**dbSNP_131**	**missense**	**missense**	***PIP***
7	**149,522,165**	**.**	**G**	**A**	**Yes**	**0.01**	**1.00**	**0.20**	**0.01**	**dbSNP_132**	**coding**	**missense**	***SSPO****
7	**150,389,751**	**.**	**C**	**A**	**Yes**	**0.01**	**1.00**	**0.20**	**0**	**dbSNP_134**	**synonymous**	**missense**	***GIMAP2***
7	**151,945,007**	**rs2479172**	**C**	**T**	**Yes**	**0.01**	**1.00**	**0.20**	**0**	**dbSNP_134**	**synonymous**	**missense**	***MLL3****
7	**151,949,735**	**rs77652527**	**T**	**C**	**Yes**	**0.01**	**1.01**	**0.21**	**0.05**	**dbSNP_131**	**missense**	**missense**	***MLL3****
7	**151,970,856**	**rs10454320**	**T**	**A**	**Yes**	**0.01**	**1.00**	**0.20**	**0**	**dbSNP_119**	**synonymous**	**missense**	***MLL3****
7	**151,970,931**	**rs56850341**	**G**	**A**	**Yes**	**0.01**	**1.00**	**0.20**	**0**	**dbSNP_129**	**synonymous**	**missense**	***MLL3****
14	23,451,345	rs45543740	G	A	Yes	0.07	1.00	0.40	0.01	dbSNP_127	missense	missense	*AJUBA*
14	24,780,751	.	A	G	yes	0.07	1.00	0.40	0	dbSNP_137	utr-5	utr-5-prime	*LTB4R,LTB4R2*

Table OR3g: AU113 variants with a segregation pattern consistent with Mendelian inheritance or association p≤ 0.05. Bold indicates variants with p≤0.05. Pattern denotes whether the variant was present in a pattern consistent with Mendelian inheritance, based on WES only. ** this variant did not have a segregation pattern consistent with Mendelian inheritance due to a missing genotype in one affected individual.

							Mean dosage				Predicted function		
chr	**position**	**rsname**	**ref**	**alt**	**pattern**	**p-val**	**affected**	**unafffected**	**1KGP**	**dbSNP**	**GVS**	**DBSNP**	**Gene**
4	**520,973**	**rs201269761**	**G**	**A**	**yes**	**0.00**	**1.00**	**0.02**	**0**	**dbSNP_137**	**missense**	**missense**	***PIGG***
4	**1,656,767**	**rs146433325**	**G**	**A**	**No****	**0.00**	**1.00**	**0.01**	**0**	**dbSNP_134**	**missense**	**missense**	***FAM53A***
4	**4,198,948**	**rs112623841**	**GCGTTGC**	**G**	**yes**	**0.00**	**1.00**	**0.04**	**0.04**	**dbSNP_132**	**coding**	**cds-indel**	***OTOP1***
4	110,773,048	rs61745483	C	A	yes	0.12	1.00	0.36	0.04	dbSNP_129	missense	missense	*LRIT3*
4	119,673,889	rs141180741	G	A	yes	0.13	1.00	0.46	0.01	dbSNP_134	missense	missense	*SEC24D*
6	**161,508,880**	**rs35533223**	**A**	**C**	**yes**	**0.00**	**1.00**	**0.03**	**0.03**	**dbSNP_126**	**missense**	**missense**	***MAP3K4***
17	**11,543,588**	**rs17600516**	**G**	**A**	**yes**	**0.01**	**1.00**	**0.50**	**0.03**	**dbSNP_123**	**near-splice**	**synonymous-codon**	***DNAH9***
17	14,110,294	rs111541535	G	T	yes	0.10	1.00	0.33	0.01	dbSNP_132	missense	missense	*COX10*
17	27,893,565	.	CA	C	yes	0.10	1.00	0.35	0.03	none	frameshift	unknown	*ABHD15*

17	30,348,206	rs79573699	C	T	yes	0.65	1.00	0.85	0.03	dbSNP_131	missense	missense	*LRRC37B*
17	33,592,323	rs113644060	C	G	yes	0.11	1.00	0.34	0.02	dbSNP_132	missense	missense	*SLFN5*
17	33,880,305	rs79007502	T	C	yes	0.10	1.00	0.34	0.03	dbSNP_132	missense	missense	*SLFN14*
17	36,872,024	rs146278240	G	A	yes	0.11	1.00	0.34	0.01	dbSNP_134	missense	missense	*MLLT6*
17	36,872,922	rs145966494	G	T	yes	0.11	1.00	0.34	0.01	dbSNP_134	missense	missense	*MLLT6*
17	39,135,143	rs201414134	T	C	yes	0.12	1.00	0.34	0.004	dbSNP_137	missense	missense	*KRT40*
17	40,996,783	rs147588427	G	A	yes	0.11	1.00	0.34	0	dbSNP_134	stop-gained	stop-gained	*AOC2*
17	61,615,545	rs147985248	A	G	yes	0.12	1.00	0.34	0.02	dbSNP_134	missense	missense	*KCNH6*
17	73,900,941	.	A	G	yes	0.37	1.00	0.67	0	none	missense	unknown	*MRPL38*
17	79,502,218	rs186367879	G	A	yes	0.37	1.00	0.68	0.05	dbSNP_135	missense	missense	*FSCN2*
17	79,614,951	rs79350765	C	T	yes	0.37	1.00	0.68	0.05	dbSNP_131	missense	missense	*TSPAN10*
22	50,216,975	.	T	G	yes	0.11	1.00	0.34	0	none	missense	unknown	*BRD1*
22	50,313,452	rs149112831	C	T	yes	0.11	1.00	0.34	0.01	dbSNP_134	missense	missense	*CRELD2*
22	50,315,363	rs8139422	C	A	yes	0.10	1.00	0.34	0.03	dbSNP_116	missense	missense	*CRELD2*
22	50,316,906	rs145684970	C	T	yes	0.11	1.00	0.34	0.01	dbSNP_134	missense	missense	*CRELD2*