

Supplemental Table 1. Markers used in Stages 1-3 of the analysis, with associated lod scores, for affecteds only (AO) and reduced penetrance models.

Marker	cM	AO	Reduced Penetrance		
		Stage 1	Stage 1	Stage 2	Stage 3
GATA198B05	3.7	-2.15	-2.70		
D22S420	5.0	-1.04	-1.52		
AGAT120	15.5	-0.46	-1.06		
D22S539	17.0	0.28	-0.60		
D22S345	22.9	-0.12	-0.78		
ATTT019M	23.3	-0.82	-1.72		
D22S315	26.4	0.56	-0.75		
D22S689	37.6	0.97	-0.14		
D22S280	41.5	0.58	-0.85	-0.93	-0.91
rs4420793	53.44			-0.85	-0.91
rs240080	54.09			-0.86	-0.91
D22S685	43.5	2.14	1.40	-0.87	-0.91
rs202916	54.88			-0.87	-0.91
rs134215	55.64			-0.96	-0.91
rs138794	56.28				-0.96
rs1076139	56.36			1.34	
rs5999860	56.89			1.58	
rs5750116	56.99				1.81
rs1573708	57.64			1.81	1.81
D22S683	46.5	2.58	1.81	1.81	1.81
rs1557540	58.36			1.81	1.81
D22S283	47.2	2.55	1.79	1.81	1.81
rs6000293	59.09			1.76	1.76
rs6000386	59.78			1.70	1.69
D22S692	48.8	2.27	1.49	1.65	1.64
rs5750310	60.53			1.61	1.61
rs4821567	61.16			1.47	1.52
rs5750370	61.75				1.52
rs5756489	61.78			1.42	
rs5995385	62.38			1.36	1.52
D22S1045	51.3	1.87	1.07	1.34	1.52
D22S445	51.4	1.82	1.01	1.28	1.44
rs6000645	62.95			1.20	1.30
rs4328731	63.40				-0.86
rs133710	63.5			0.84	
rs5750576	64.07			-1.29	-0.90
rs7292074	64.86				-1.14
rs713863	64.76			-1.67	
rs139412	65.23				-2.08
rs11704525	65.33			-1.74	
rs107140	66.06			-1.83	-2.07
D22S423	55.1	-0.01	-1.28	-1.92	-2.04
D22S274	62.7	-0.64	-1.29		
D22S532	65.3	-0.86	-1.15		
D22S1169	78.7	-1.74	-1.52		
TCTA014M	79.5	-2.71	-2.91		

Supplemental Table 2. The probability that the observed allele at each Stage 1 marker locus shares the same FGL as the D22S683 tagging allele, as estimated from sampled inheritance vectors.

Marker	Affected Subject							
	IV.4.a	IV.4.b	IV.3.a	IV.3.b	IV.3.c	IV.2.a	V.1.a	IV.2.b
GATA198B05	0.12	0.12	0.11	0.11	0.02	0.88	0	0.88
D22S420	0.16	0.16	0.15	0.14	0.02	0.78	0	0.78
AGAT120	0.44	0.44	0.44	0.44	0	0.21	0	0.21
D22S539	0.55	0.55	0.55	0.55	0	0.17	0.04	0.17
D22S345	0.97	0.97	0.97	0.97	0	0.01	0.01	0.01
ATTT019M	1	1	1	1	0	0	0	0
D22S315	1	1	1	1	0.13	0	0	0
D22S689	0.98	0.98	0.98	0.98	0.6	0.01	0.01	0.01
D22S280	1	1	1	1	0.78	0.01	0.01	0
D22S685	1	1	1	1	0.86	0.37	0.37	0.37
D22S683	1	1	1	1	1	1	1	1
D22S283	1	1	1	1	0.98	0.97	1	0.98
D22S692	0.65	0.65	0.65	0.65	0.61	0.86	0.98	0.87
D22S1045	0.4	0.4	0.4	0.4	0.33	0.75	0.97	0.71
D22S445	0.38	0.38	0.37	0.37	0.31	0.74	0.97	0.7
D22S423	0	0	0	0	0	0.55	0.95	0.43
D22S274	0	0	0	0	0	0.14	0.93	0.15
D22S532	0	0	0	0	0	0	0.93	0
D22S1169	0	0	0	0	0	0	0.92	0
TCTA014M	0	0	0	0	0	0	0.92	0

Supplementary Table 3: Exome variant counts in each of the five sequenced individuals, in the Stage 3 region of interest, for all 68 variants that appeared in at least one exome. The frequency of the variant allele was obtained from HapMap-CEU, 1000 Genomes – EUR(*), or the NHLBI exome sequencing project exome variant server(**). Variants with a frequency of 5% or less are highlighted in **bold**.

Name	Build 37 position	Variant Frequency	Exomed individuals				
			II.1.b	IV.3.a	IV.4.a	V.1.a	IV.5.a
rs743810	35,742,925	0.32	1	1	1	1	1
rs133427*	35,815,880	0.15	0	0	0	0	1
rs7292	36,007,045	0.55	1	1	0	1	0
rs7293	36,007,075	0.53	1	1	0	1	0
rs34014315*	36,055,406	0.02	0	1	0	0	0
rs17723764*	36,122,517	0.23	0	1	0	0	0
rs2076671	36,122,930	0.36	2	1	2	2	2
rs2076672	36,123,083	0.18	0	1	0	0	0
rs2076673	36,124,860	0.35	2	1	2	2	2
rs132618	36,537,500	0.36	2	1	2	1	0
rs61731692*	36,537,725	0.08	0	1	0	0	0
rs3827346*	36,537,893	0.29	0	0	0	1	0
rs132642	36,545,137	0.94	2	2	2	2	0
rs132653	36,556,823	0.80	2	2	2	2	0
rs6000172	36,587,202	0.62	1	2	1	1	0
rs6000174	36,587,279	0.62	1	2	1	1	0
rs2227167	36,587,486	0.62	1	2	1	1	0
rs2227168	36,587,511	0.63	1	2	1	1	0
rs132700	36,587,704	0.29	1	0	1	1	1
rs2227169	36,587,952	0.61	1	2	1	1	0
rs2007468	36,591,380	0.67	1	2	1	1	0
rs9610445	36,597,744	0.13	0	1	0	0	0
rs80587	36,598,049	0.60	2	2	1	1	1
rs132736	36,598,058	0.60	2	2	1	1	1
rs6000181	36,598,081	0.09	0	1	0	0	0
rs132760	36,623,731	1.00	2	2	2	2	2
rs2239785	36,661,330	0.70	1	2	1	2	2
rs136174	36,661,536	0.84	1	2	1	2	2
rs136175	36,661,566	0.84	1	2	1	2	2
rs136176	36,661,646	0.84	1	2	1	2	2
rs136177	36,661,842	0.83	1	2	1	2	2
rs2269529	36,684,354	0.28	0	0	1	0	0
rs2269530	36,684,358	0.28	0	0	1	0	0
.***	36,684,412	0.0005	0	0	1	0	0
.***	36,689,462	0.0002	1	1	1	1	0
rs710181	36,691,607	0.98	2	2	2	2	2
rs2277841	36,900,271	0.67	1	2	1	2	2
rs760718	36,900,806	0.51	1	2	1	2	2
rs1048012	37,163,329	0.80	2	2	2	2	2
rs2072708	37,267,767	0.64	1	2	2	2	1
rs2072712	37,271,802	0.11	0	0	1	0	0
rs2075939	37,271,882	0.82	1	2	2	2	2
rs11552115*	37,273,742	0.11	0	0	1	0	0
rs1801117*	37,329,999	0.16	1	1	1	0	0
rs2235321	37,462,926	0.37	1	0	0	1	0

rs855791	37,462,936	0.60	1	0	1	1	1
rs4820268	37,469,591	0.54	1	0	1	1	1
rs2111833	37,480,797	0.33	1	0	0	1	0
rs2235324	37,485,724	0.39	1	0	0	1	0
rs11704654	37,499,386	0.16	0	0	1	0	1
rs228953	37,531,436	0.42	1	0	0	0	0
rs17812681*	37,578,388	0.014	0	0	1	0	0
rs229519	37,578,579	0.45	0	0	1	1	1
rs17812699	37,578,652	0.11	0	0	1	0	0
rs7290488	37,581,383	0.19	2	1	1	1	0
rs229526	37,581,422	0.18	0	1	0	0	0
rs229527	37,581,485	0.46	0	1	0	1	1
rs2071710	37,603,021	0.34	0	0	0	1	1
rs229569	37,603,051	0.40	0	0	0	1	1
rs86582	37,603,390	0.40	0	0	0	1	1
rs86583*	37,603,744	0.42	0	0	0	1	1
rs1064498*	37,622,815	0.25	0	0	0	1	2
rs2239774	37,637,653	0.17	0	0	0	1	1
rs2239822	37,699,377	0.70	1	2	1	1	0
rs8142615	37,770,066	0.42	0	1	0	1	0
rs7292751	37,770,357	0.46	0	1	0	1	0
rs2071856	37,770,630	0.29	2	1	1	1	1
rs2071857	37,771,158	0.45	0	1	0	1	1