

Table S1: Variant call statistics.

sample	mean target coverage	# of passing exonic indels	# of passing exonic snps	# of synonymous snps	# of non synonymous snps	# of missense snps	# of nonsense snps	#number of silent snps	# of indel codon change	# of indel frameshifts	# of indel causing stop/start mutations
II-1	129.8	471	20821	10832	9989	9608	70	10832	272	196	3
II-2	135.8	450	20754	10892	9862	9484	67	10892	251	196	3
II-4	146.2	472	20849	10939	9910	9526	67	10939	272	199	1
II-6	137.5	488	20894	10886	10008	9630	76	10886	279	205	4
II-8	130.1	470	20867	10915	9952	9582	67	10915	259	207	4
II-9	130.2	470	20989	10960	10029	9654	68	10960	280	187	3
II-11	117.4	448	20806	10911	9895	9514	72	10911	247	198	3
II-19	119.9	449	20731	10881	9850	9477	71	10881	254	194	1
II-23	127.8	480	20869	10882	9987	9611	62	10882	268	208	4
III-2	109.2	426	20793	10910	9883	9529	69	10910	248	176	2
III-12	138.0	447	20789	10916	9873	9511	67	10916	259	185	3

Table S2. Population frequency of *SEC16A* and *MAMDC4* wild-type and deletion alleles using a large replication cohort.

Gene	Cohort	Allele	Number of Alleles	Percentage (%)
<i>SEC16A</i>	Controls	Wild-type	2239	99.16
		Deletion	19	0.84
	AxSpA	Wild-type	1826	99.13
		Deletion	16	0.87
<i>MAMDC4</i>	Controls	Wild-type	2236	98.59
		Deletion	32	1.41
	AxSpA	Wild-type	1862	98.62
		Deletion	26	1.38

Table S3. Linkage disequilibrium (LD) analysis in the nuclear family for the genetic variants detected in chromosome 9.

CHR	POS1	POS2	SNP1	SNP2	r ²	D	D'
chr9	139266496	139277994	rs4077515	rs147271628	1	0.222222	1
chr9	139266496	139368953	rs4077515	rs3812594	1	0.222222	1
chr9	139277994	139368953	rs147271628	rs3812594	1	0.222222	1
chr9	139266496	139370954	rs4077515	indelSEC16A	1	0.222222	1
chr9	139368953	139370954	rs3812594	indelSEC16A	1	0.222222	1
chr9	139277994	139370954	rs147271628	indelSEC16A	1	0.222222	1
chr9	139266496	139378914	rs4077515	rs34376913	1	0.222222	1
chr9	139277994	139378914	rs147271628	rs34376913	1	0.222222	1
chr9	139370954	139378914	indelSEC16A	rs34376913	1	0.222222	1
chr9	139368953	139378914	rs3812594	rs34376913	1	0.222222	1
chr9	139368953	139564387	rs3812594	rs146989870	1	0.222222	1
chr9	139277994	139564387	rs147271628	rs146989870	1	0.222222	1
chr9	139266496	139564387	rs4077515	rs146989870	1	0.222222	1
chr9	139378914	139564387	rs34376913	rs146989870	1	0.222222	1
chr9	139370954	139564387	indelSEC16A	rs146989870	1	0.222222	1
chr9	139368953	139748276	rs3812594	indelMAMDC4	1	0.222222	1
chr9	139378914	139748276	rs34376913	indelMAMDC4	1	0.222222	1
chr9	139266496	139748276	rs4077515	indelMAMDC4	1	0.222222	1
chr9	139277994	139748276	rs147271628	indelMAMDC4	1	0.222222	1
chr9	139564387	139748276	rs146989870	indelMAMDC4	1	0.222222	1
chr9	139370954	139748276	indelSEC16A	indelMAMDC4	1	0.222222	1
chr9	139368953	140064315	rs3812594	rs7019671	0.25	0.111111	1
chr9	139266496	140064315	rs4077515	rs7019671	0.25	0.111111	1
chr9	139378914	140064315	rs34376913	rs7019671	0.25	0.111111	1
chr9	139564387	140064315	rs146989870	rs7019671	0.25	0.111111	1
chr9	139370954	140064315	indelSEC16A	rs7019671	0.25	0.111111	1
chr9	139277994	140064315	rs147271628	rs7019671	0.25	0.111111	1
chr9	139748276	140064315	indelMAMDC4	rs7019671	0.25	0.111111	1
chr9	140064315	140100317	rs7019671	rs113809617	1	0.222222	1
chr9	139378914	140100317	rs34376913	rs113809617	0.25	0.111111	1
chr9	139266496	140100317	rs4077515	rs113809617	0.25	0.111111	1
chr9	139564387	140100317	rs146989870	rs113809617	0.25	0.111111	1
chr9	139370954	140100317	indelSEC16A	rs113809617	0.25	0.111111	1
chr9	139277994	140100317	rs147271628	rs113809617	0.25	0.111111	1
chr9	139748276	140100317	indelMAMDC4	rs113809617	0.25	0.111111	1
chr9	139368953	140100317	rs3812594	rs113809617	0.25	0.111111	1
chr9	139266496	140128107	rs4077515	rs142918575	1	0.222222	1
chr9	139378914	140128107	rs34376913	rs142918575	1	0.222222	1
chr9	139277994	140128107	rs147271628	rs142918575	1	0.222222	1
chr9	139564387	140128107	rs146989870	rs142918575	1	0.222222	1
chr9	139368953	140128107	rs3812594	rs142918575	1	0.222222	1
chr9	139748276	140128107	indelMAMDC4	rs142918575	1	0.222222	1
chr9	139370954	140128107	indelSEC16A	rs142918575	1	0.222222	1
chr9	140064315	140128107	rs7019671	rs142918575	0.25	0.111111	1
chr9	140100317	140128107	rs113809617	rs142918575	0.25	0.111111	1

Table S4. GLIDERS (Genome-wide Linkage DisEquilibrium Repository and Search engine) contents for rs3812594 (sec16A snp upstream of novel indel). GLIDERS contain pairwise associations with $r^2 \geq 0.3$ across the human genome for any SNP genotyped within HapMap phase 2 and 3, regardless of distance between the markers.

SNP	Ch r	Pos. (bp)	MAF	Distance	r2	D'	Chi Sq	P	P(BFC)
rs4487900	9	138423122	0.433	65kb	0.34	1	0.35 6	2.68E-12	1
rs3812584	9	138432540	0.455	56kb	0.37	1	0.34 5	5.30E-13	0.372
rs10870164	9	138435503	0.442	53kb	0.35	1	0.34 6	1.91E-12	1
rs10870165	9	138436422	0.451	52kb	0.37	1	0.33 8	1.10E-12	0.771
rs10781538	9	138440750	0.478	48kb	0.41	1	0.36 5	2.43E-14	0.017
rs8413	9	138443132	0.491	45kb	0.46	1	0.38 0.38	7.88E-17	5.53E-5
rs10781542	9	138447260	0.496	41kb	0.44	1	0.37 6	1.18E-15	0.00083
rs1127152	9	138455420	0.491	33kb	0.46	1	0.40 2	7.68E-17	5.39E-5
rs3812591	9	138461433	0.311	27kb	0.98	1	0.97 7	2.77E-49	1.94E-37
rs11145756	9	138484406	0.5	4kb	0.45	1	0.38 4	4.89E-16	0.00034
rs3812595	9	138488883	0.308	109bp	1	1	1 1	1.21E-50	8.49E-39
rs4379550	9	138496247	0.491	7kb	0.46	1	0.40 2	7.68E-17	5.39E-5
rs7045859	9	138497759	0.491	8kb	0.44	1	0.37 3	1.99E-15	0.0014
rs9314867	9	138507667	0.478	18kb	0.41	1	0.43 4	2.29E-15	0.0016
rs6563	9	138509005	0.495	20kb	0.46	1	0.46 8	2.69E-17	1.89E-5
rs11146021	9	138447679	0.32	41kb	0.92	98	0.90 6	3.72E-44	2.61E-32
rs10448340	9	138439890	0.362	48kb	0.71	95	0.70 5	1.35E-28	9.47E-17
rs11145930	9	138423837	0.362	64kb	0.68	93	0.67 8	8.15E-27	5.72E-15
rs10870166	9	138436737	0.348	52kb	0.72	93	0.71 1	1.65E-29	1.16E-17
rs1130635	9	138437690	0.342	51kb	0.73	93	0.72 0.72	1.85E-30	1.30E-18
rs10747044	9	138438950	0.348	49kb	0.72	93	0.71 1	1.65E-29	1.16E-17
rs4400499	9	138424862	0.302	63kb	0.79	89	0.77 4	1.83E-34	1.28E-22
rs11145974	9	138436565	0.304	52kb	0.78	89	0.76 4	7.26E-35	5.09E-23
rs10870194	9	138446855	0.491	41kb	0.36	89	0.30 1	1.14E-11	1

rs11145917	9	138422021	0.406	66kb	0.45	83	1	9.08E-16	0.00064
						0.	0.59		3.07E-
rs11145910	9	138420315	0.302	68kb	0.64	82	3	4.38E-24	12
						0.	0.22		
rs10781499	9	138386226	0.491	102kb	0.3	81	4	1.95E-10	1
						0.	0.23		
rs4077515	9	138386317	0.487	102kb	0.31	81	2	7.95E-11	1
						0.	0.24		
rs3812558	9	138388956	0.491	99kb	0.3	81	4	1.71E-10	1
						0.	0.24		
rs10781500	9	138389159	0.491	99kb	0.3	81	4	1.71E-10	1
						0.	0.23		
rs11794847	9	138391568	0.495	97kb	0.3	81	8	3.17E-10	1
						0.	0.23		
rs10781507	9	138391879	0.482	96kb	0.31	81	9	1.48E-10	1
						0.	0.25		
rs3812570	9	138395025	0.496	93kb	0.3	81	9	1.37E-10	1
						0.	0.25		
rs10781510	9	138398994	0.486	89kb	0.31	81	6	7.99E-11	1
						0.	0.24		
rs4266763	9	138409646	0.491	79kb	0.3	81	4	1.71E-10	1
						0.	0.57		8.77E-
rs10781511	9	138400587	0.304	88kb	0.62	8	7	1.25E-23	12
						0.	0.57		8.77E-
rs563	9	138416306	0.304	72kb	0.62	8	7	1.25E-23	12
						0.	0.42		
rs3812555	9	138381754	0.373	107kb	0.48	79	9	9.13E-17	6.40E-5
						0.	0.32		
rs3812547	9	138372316	0.397	116kb	0.4	77	2	8.17E-13	0.573

NOTE: There is no SNP in the region of the indel in MAMDC4 (position 139,748,276) and rs3812594.