Supplement to Notifiable genetic variants on commercially-available SNP arrays

Supplemental Table 1. Full information on genetic variants identified in this study from the GeneTest database. Included are information on disease, affected gene, variants tested in CLIA-labs, variants for which unique SNP identifiers could and could not be located, and presence of variants and proxies in HapMap and on commercial genotyping arrays.

Disease	Gene	Chr	Variants listed on Gene Tests	Variants for which SNPids could not be determined	Variants with SNPids	Total with SNPids	SNPs in HapMap: (Variant) rsID	SNP arrays on which variant is present: (rsID) array(s)	Proxy SNPs in HapMap: (SNP) Proxy SNP	SNP arrays on which Proxy SNP is present: (Proxy SNP) Array(s)
21-Hydroxylase Deficiency	*CYP21A2*	6p21.3	13	4 - P30L, P31L, I173N, I236N	9 - (V237E) rs12530380, (M239K) rs6476, (V281L) rs6471, (Q318X) rs7755898, (R356W) rs7769409, (I172N) rs6475, (P453S) rs6445, (R357T) rs7769409, (656A/C>G) rs6467	9	(V237E) rs12530380; (M239K) rs6476	(rs12530380) AG		
Achondroplasia	*FGFR3*	4p16.3	2	1 - G375C	1 - (G380R) rs28931614	1				
Alpha-1-Antitrypsin Deficiency	*SERPINA1*	14q32.1	2	0	2 - (Q342K) rs17850837, (Q264V) rs17580	2				
Alpha-Thalassemia	*HBA1*	16p13.33 -p13.11	0	0	0	0				
Beta-Thalassemia	*HBB*	11p15.5	10	3 - IVS1-5,Codon 15, HbE	7 - (IVS1-1) rs33943001 , (delAA>K8stop) rs35497102 , (insG,K8S9>stop) rs35699606 , (FS41/42delTTCT) rs34532766 , (-29A>Gtatabox) rs34598529 , (-88C>T) rs33944208 , (IVS2nt1) rs33945777	7				
Biotinidase Deficiency	*BTD*	3p25	8	5 - G98d7i3, R157H, F403V, Q456H, R538C	3 - (D444H) rs13078881, (A171T) rs13073139 , (D252G) rs28934601	3	(D444H) rs13078881; (A171T) rs13073139	(rs13078881) AG		
Bloom Syndrome	*BLM*	15q26.1	2	2 - 2281del6/ins 7 (blmASH), insT2407	0	0				
BRCA1 Hereditary Breast/Ovarian Cancer	*BRCA1*	17q21	4	4 - Tyr978X, 185delAG, 5382insC, 6174delT	0	0				
BRCA2 Hereditary Breast/Ovarian Cancer	*BRCA2*	13q12.3	1	1 - 6174delT	0	0				
Canavan Disease	*ASPA*	17p13	4	1 - 433-2 (intron 2 acceptor site)	3 - (Y231X) rs12948217, (E285A) rs28940279, (A305E) rs28940574	3				
Carnitine Palmitoyl-transferase IA (liver) Deficiency	*CPT1A*	11q13.1-q13.2	2	2 - P479L, G710E	0	0				
Carnitine Palmitoyl-transferase II	*CPT2*	1p32	14	12 - S38fs, S113L,	2 - (P50H) rs28936375, (Y628S) rs28936673	2				

Deficiency				R124x, L178F, M214T, P227L, Q413fs, R503C, G549D, Q550R, P604S, R631C,						
CFTR-Related Disorders	*CFTR*	7q31.2	16	12 - R347P, G85E, R117H, Q359K, G542X, S549R, G551D, R553X, W1089X, D1152H, W1282X, N1303K	4 - (F508) rs332, (I148T) rs35516286, (I507V) rs1801178, (M1101K) rs36210737	4				
Colon Cancer (APC I1307K related)	*APC*	5q21-q22	2	0	2 - (I1307K) rs1801165, (E1317Q) rs1801166	2				
Cystinosis	*CTNS*	17p13	5	5 - W138x, L158P, D205N, 1035insC, 537del21	0	0				
Diabetes and Hearing Loss	*MTTL1*	M	n/a							
Dentatorubral-Pallidoluysian Atrophy (DRPLA)	*ATN1*	12p13.31	0	0	0	0				
Early-Onset Primary Dystonia (DYT1)	*TOR1A*	9q34	1	1 - 302Edel	0	0				
Factor V Leiden Thrombophilia	*F5*	1q23	1	0	1 -(R506Q) rs6025	1	(R506Q) rs6025	(rs6025) IM,IMD,IBC	(rs6025): rs1894692, r^2=1	
Factor V R2 Mutation Thrombophilia	*F5*	1q23	1	0	1 - (H1299R) rs1800595	1				
Familial Adenomatous Polyposis	*MUTYH*	1p34.3-p32.1	3	1- A459D	2 - (Y165C) rs34612342, (G382D) rs36053993	2				
Familial Dysautonomia	*IKBKAP*	9q31	5	4 - R696P, IVS20(+6)T>C, 2507+6T>C, 2204 +6T>C	1 - (P914L) rs28939712	1				
Familial Mediterranean Fever	*MEFV*	16p13.3	14	8 - E167D/G167A, T267I,	6 - (E148Q) rs3743930, (P369S) rs11466023, (R408Q) rs11466024, (M680I) rs28940580, (M694I)	6	(P369S) rs11466023; (R408Q)	(rs11466023) AG,IBC; (rs11466024)	(rs11466023) : rs11466024; (rs11466024)	(rs11466024) AG,IBC; (rs11466023)

				F579L, I692del, M694V, K695R, A744S, R761H	rs28940578, (V726A) rs28940579		rs11466024	AG,IBC	: rs11466023	AG,IBC
FANCC-Related Fanconi Anemia	*FANCC*	9q22.3	5	5 - Q13X, IVS4+4, 322delG, Arg548X, Leu554Pro	0	0				
FGFR1-Related Craniosynostosis Syndromes	*FGFR1*	8p11.2-p11.1	1	1 - P252R	0	0				
FGFR2-Related Craniosynostosis Syndromes	*FGFR2*	10q26	2	2 - S252W, P253R	0	0				
FGFR3-Related Craniosynostosis Syndromes	*FGFR3*	4p16.3	2	0	2 - (P250R) rs4647924, (A391E) rs28931615	2				
FRAXE Syndrome	*FMR1*	Xq27.3	0	0	0	0				
Friedreich Ataxia	*FXN*	9q13-q21.1	0	0	0	0				
Galactosemia	*GALT*	9p13	12	10 - S135L, T138M, Q169K, Q188R, L195P, Y209C, K285N, X380R, IVS2-2, 5'UTR -119del	2 - (L218L) rs2070075, (N314D) rs2070074	2	(N314D) rs2070074; (L218L) rs2070075	(rs2070074) A6,I3,I5,I6,I6Q,IM, IMD,IC,ICQ	(rs2070075): rs12553321, r^2=1	(rs12553321) IM,IMD
Gaucher Disease	*GBA*	1q21	8	6 - 84Gins, IVS2+1G>A, N370S, V394L, R463C/H, R496H	2 - (D409H) rs1064651, (L444P) rs35095275	2				
GJB2-Related DFNA 3 Nonsyndromic Hearing Loss and Deafness	*GJB2*	13q11-q12	0	1- 35delG	0	0				
GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness	*GJB2*	13q11-q12	11	10 - W24X, M34T, W77X/R, 167delT, 235delC, 101delAG, R184P, V37I, 313del14, delE120	1 - (35delG) rs1801002	1				

GJB6-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness	*GJB6*	13q12	1	1- D13S1830 (del)	0	0				
Glucose-6-Phosphate Dehydrogenase Deficiency	*G6PD*	Xq28	5	1 - 1376G>T (R459L)	4 - (V68M) rs1050828, (N128D) rs1050829, (S188F) rs5030868, (A225T) rs5030869	4	(G202A) rs1050828	(rs1050828) A6,I1,IM,IMD,IBC		
Glycogen Storage Disease Type 1a	*G6PC*	17q21	5	4 - Q27delC (fs), 130x, delF327 (459insTA), Q347X	1 - (R83C) rs1801175	1				
Glycogen Storage Disease Type V	*PYGM*	11q13	3	3 - R49X, G204S, K542T	0	0				
Hemoglobin Constant Spring	*HBA2*	16p13.3	0	0	1- (X143E) rs41464951	1				
Hemoglobin E	*HBB*	11p15.5	0	0	2- (E121Q) rs3394627, (E26K) rs33950507	2				
Hemoglobin S Beta-Thalassemia	*HBB*	11p15.5	1	1- c.20A>T	0	0				
Hemoglobin SC	*HBB*	11p15.5	0	0	1- (E6K) rs33030165	1				
Hemoglobin SS	*HBB*	11p15.5	0	0	1- (E6V) rs334	1	(Glu6Val) rs334			
Hemophilia A	*F8*	Xq28	0	0	0	0				
Hereditary Fructose Intolerance	*ALDOB*	22q13.1	4	3 - A174D, Y203X, N334X	1 - (A149P) rs1800546	1				
Hereditary Non-Polyposis Colon Cancer	*MLH1*	3p21.3	2	2- 454-1G>A, 320T>G	0	0				
Hereditary Pancreatitis	*PRSS1*	7q34	3	3- A16V, R122H, N291I	0	0				
	SPINK1	5q32	2	0	2 - (M1T) rs28935768, (N34S) rs17107315	2	(N34S) rs17107315	(rs17107315) AG,IM,IMD		
Hexosaminidase A Deficiency	*HEXA*	15q23-q24	9	8 - R247W, R249W, G269S, 1278insTATC, IVS7+1G>A, IVS9+1G>A, IVS12(+1)G>C, G805A	1 - (R178L) rs28941770	1				
HFE- Associated Hereditary Hemochromatosis	*HFE*	6p21.3	8	2- E168Q/X, W169X	6 - (H63D) rs1799945, (S65C) rs1800730, (V53M) rs28934889, (V59M) rs28934890, (Q127H) rs28934595, (C282Y) rs1800562	6	(H63D) rs1799945; (C282Y) rs1800562	(rs1799945) IM,IMD,IBC; (rs1800562) AS,A5,A6,I3,I5,I6, I6Q,IM,IMD,IC,ICQ, IBC	(rs1799945): rs198846, r^2=1; rs129128, r^2=1; rs198833, r^2=1	(rs198846): I6,IM,IMD; (rs129128): AN,A5,A6,I3,I5, I6,I6Q,IM,IMD, IC,ICQ
Homocystinuria	*CBS*	21q22.3	2	1- G307S	1 - (I278T) rs5742905	1				

Caused by Cystathionine Beta-Synthase Deficiency										
Huntington Disease	*HD*	4p16.3	0	0	0	0				
Hyper-lipoproteinemia Type III/ Familial Dysbeta-lipoproteinemia	*ApoE*	19q13.2	2	0	2 - (C112R) rs429358, (C158R) rs7412	2	(Cys112Arg) rs429358			
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	*SLC25A15*	13q14	1	1 - F188del	0	0				
Hypochondroplasia	*FGFR3*	4p16.3	9	6 - Asn328Ile, Ile538Val, N540T, N540S, K650M, K650Q	3 - (G380R) rs28931614, (N540K) rs28933068, (K650N) rs28928868	3				
Infantile Myopathy and Lactic Acidosis (Fatal and Non-Fatal Forms)	*MTTT*	M	n/a							
Isovaleric Acidemia	*IVD*	15q14-q15	1	0	1 - (A282V) rs28940889	1				
Krabbe Disease	*GALC*	14q31	0	0	0	0				
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	*HADHA*	2p23	3	3 - E474Q, E10Q, Q342X	0	0				
Medium Chain Acyl-Coenzyme A Dehydrogenase Deficiency	*ACADM*	1p31	9	9 - Y42H, K304E, K329E, R206C (250C>T), G267R (799G>A), 503A>G, 583 G>A, 616C>T, 617G>A	0	0				
MELAS Syndrome	*MTTL1*	M	n/a							
MERRF (Myoclonic Epilepsy associated with Ragged Red Fibers)	*MTTH*	M	n/a							
Metachromatic Leukodystrophy	*ARSA*	22q13.31	2	1-459+1G>A (SDEx2+1)	1- (P426L) rs28940893	1				
Mitochondrial DNA-Associated Leigh Syndrome and	*MTATP6, MTTL1, MTTK,*	M	n/a							

NARP	*MTND1, MTND3, MTND4, MTND5, MTND6, MTCO3, MTTW, MTTV*									
MTHFR Deficiency and Thermolabile Variant	*MTHFR*	1p36.3	2	0	2 - (A222V) rs1801133, (E429A) rs1801131	2	(A222V) rs1801133; (E429A) rs1801131	(rs1801133) AH,I1,I3,I5,I6,I6Q, IM,IMD,IC,ICQ,IBC (rs1801131) A6,I6Q,IM,IMD,IBC	(rs1801131): rs4846049, r^2=1	(rs4846049) IBC
MTRNR1-Related Hearing Loss and Deafness	*MTRNR1*	M	n/a							
MTTS1-Related Hearing Loss and Deafness	*MTTS1*	M	n/a							
Mucolipidosis IV	*MCOLN1*	19p13.3-p13.2	6	6 - IVS3(-2)A>G, 5534A>G, 406-2A>G, 511_6943del, T232P, D362Y	0	0				
Multiple Endocrine Neoplasia Type 2	*RET*	10q11.2	4	4 - C618R (or others), E768D, A883F, M918T	0	0				
Myotonic Dystrophy Type 1	*DMPK*	19q13.2-q13.3	0	0	0	0				
Niemann-Pick Disease Due to Sphingomyelinase Deficiency	*SMPD1*	11p15.4-p15.1	7	7 - L302P, P330fs, N370S, L444P, 84insG, R496L, R608del	0	0				
Phenylalanine Hydroxylase Deficiency	*PAH*	12q23.2	11	2 - I65T, G272x	9 - (L48S) rs5030841, (R158Q) rs5030843, (R261X) rs5030850, (R261Q) rs5030849, (R408W) rs5030858, (R408Q) rs5030859, (Y414C) rs5030860, (IVS10int-546) rs5030855, (IVS12+1G>A) rs5030861	9	(R261Q) rs5030849			
Plasminogen Activator Inhibitor I	*SERPINE1*	7q21.3-q22	1	0	1 - 4G/5G (rs1799762)	1				
Prader-Willi Syndrome	*PWCR*	15q11-q13	0	0	0	0				
Prothrombin G20210A Thrombophilia	*F2*	11p11	1	0	1 - 3('UTR SNP) rs1799963	1				
Rett Syndrome	*MECP2*	Xq28	14	9 - S134C, P152R,	5 - (R106W) rs28934907, (R133C) rs28934904, (A140V) rs28934908,	5				

				R168x, P225R, R255x, G269fs, R270x, R294x, R306x	(T158M) rs28934906, (R306C) rs28935468					
Smith-Lemli-Opitz Syndrome	*DHCR7*	11q12-q13	11	11 - T93M, L109P, W151X, L157P, V326L, R352W, R352Q, C380Y, R404C, R446Q, IVS8-1G>C	0	0				
Spinal and Bulbar Muscular Atrophy	*AR*	Xq11-q12	0	0	0	0				
Spinal Muscular Atrophy	*SMN1*	5q12.2-q13.3	0	0	0	0				
Spinocerebellar Ataxia Type 1	*ATXN1*	6p23	0	0	0	0				
Spinocerebellar Ataxia Type 2	*ATXN2*	12q24	0	0	0	0				
Spinocerebellar Ataxia Type 3	*ATXN3*	14q24.3-q31	0	0	0	0				
Spinocerebellar Ataxia Type 6	*CACNA1A*	19p13	0	0	0	0				
Spinocerebellar Ataxia Type 7	*ATXN7*	3p21.1-p12	0	0	0	0				
Spinocerebellar Ataxia Type 8	*ATXN8*	13q21	0	0	0	0				
Spinocerebellar Ataxia Type10	*ATXN10*	22q13	0	0	0	0				
Thanatophoric Dysplasia	*FGFR3*	4p16.3	13	13 - R248C, S249C, G370C, S371C, Y373C, K650M, K650E, X807G, X807R, X807L, X807S, X807C, X807W	0	0				
Tyrosinemia Type I	*FAH*	15q23-q25	5	5- Q64H, P261L, W262X, G337S, E357X	0	0				
Von Willebrand	*VWF*	12p13.3	0	0	0	0				

Disease										
XX Male Syndrome	*SRY*	Yp11.3	0	0	0	0				
XY Gonadal Dysgenesis	*SRY*	Yp11.4	0	0	0	0				

Supplemental Table 2. Information on NHGRI GWAS catalog of variants associated with common diseases. This catalog includes only SNPs with p-values for association < $9.5x10^{-6}$ from GWAS assaying ≥ 100,000 variants.

Disease	Gene	SNPid	SNP Genotyping Arrays	AA	Ab	bb	FHS MAF	Risk allele (if given)	CEU	CHB	JPT	YRI
Bone and Musculoskeletal Disorders												
Knee osteoarthritis	PTGS2,PLA2G4A	rs4140564	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.059	-	0.076	0.000	0.000	0.076
Rheumatoid arthritis	TNFAIP3, OLIG3	rs10499194	AX				0.292	C	0.175	0.011	0.091	0.100
	TRAF1-C5	rs3761847	I1,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.583	G	0.483	0.511	0.545	0.350
	Intergenic	rs11761231	AN,A5,A6,IM,IMD,IBC	3616	3740	1012	0.344	C	0.400	0.767	0.711	0.183
		rs743777	AS,A6,IBC	3796	3688	837	0.322	G	0.275	0.133	0.102	0.575
		rs2837960	AN,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	5167	2607	434	0.212	G	0.102	0.216	0.125	0.250
		rs3816587	AX,AS,A5,A6,I1,IM,IMD,IBC	2801	4131	1450	0.419	C	0.442	0.656	0.722	0.542
Restless legs syndrome	BTBD9	rs3923809	AG,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	3863	3524	809	0.314	A	0.259	0.651	0.640	0.342
	MEIS1	rs2300478	AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	4664	3126	554	0.254	G	0.242	0.222	0.273	0.125
	BTBD9	rs9296249	AX,AN,A5,A6,I2,I5,I6,I6Q,IM,IMD,IBC	5051	2921	412	0.223	T	0.200	0.544	0.500	0.458
	MAP2K5, LBXCOR1	rs12593813	AS,A5,A6,I2,I5,I6,I6Q,IM,IMD	3966	3538	880	0.316	G	0.317	0.614	0.678	0.842
Cardiovascular Diseases												
Coronary spasm in women		rs10498345					0.000	T	0.000	0.189	0.221	0.138
Atrial fibrillation	Intergenic	rs958546	AH,A6				0.293	-	0.381	0.311	0.364	0.342
	KIAA1598	rs4776472	AH,A6				0.241	-	0.242	0.474	0.569	0.043
	CNTN5	rs10501920	AH,AS,A5,A6	6534	1728	109	0.116	-	0.092	0.167	0.189	0.225
Heart failure	KIAA1598	rs740363	AH,AN,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	2319	4166	1882	0.474	-	0.458	0.089	0.091	0.333
Major CVD	Intergenic	rs499818	AH,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.279	-	0.208	0.122	0.091	0.058
Coronary disease	CDKN2A,CDKN2B	rs1333049	AN,A5,A6,IBC	2137	4229	1924	0.487	C	0.492	0.478	0.511	0.175
		rs17672135	AN,A5,A6,IBC	6542	1677	104	0.113	C	0.150	0.089	0.144	0.092
		rs688034	AN,A5,A6,IBC	3712	3663	892	0.329	T	0.291	0.000	0.000	0.058
		rs8055236	AN,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	5555	2499	278	0.183	G	0.208	0.111	0.167	0.608
	MTHFD1L	rs6922269	AX,AN,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	4535	3253	588	0.264	A	0.300	0.022	0.011	0.725
	Pseudogene	rs2943634	AN,A5,A6,IBC	3632	3733	990	0.342	C	0.358	0.133	0.089	0.608
Atrial fibrillation/atrial flutter	PITX2,ENPEP	rs2200733	AX,AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	6283	1865	170	0.133	T	0.117	0.422	0.467	0.300
		rs10033464	I1,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC				0.091	T	0.117	0.233	0.209	0.250
Hypertension	RYR2,CHRM3,ZP4	rs2820037	AS,A5,A6,IBC	5754	2385	245	0.171	T	0.158	0.100	0.033	0.433
		rs2398162	AS,A5,A6,IM,IMD,IBC	4932	2853	431	0.226	A	0.217	0.633	0.625	0.100
Stroke	IMPA2	rs7506045	AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	6843	1299	78	0.089	-	0.075	0.333	0.167	0.167
	Intergenic	rs10486776	AX,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.074	-	0.058	0.178	0.070	0.000
	AIM1	rs783396	AG,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	7086	1206	78	0.081	-	0.067	0.033	0.080	0.100
Myocardial infarction	CDKN2A,CDKN2B	rs10757278	IBC				0.509	G	0.500	0.474	0.528	0.051
Respiratory Disorders												
Childhood	Intergenic	rs7216389	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.512	T	0.483	0.611	0.711	0.875

asthma, ORMDL3 expression												
Cancer												
Breast cancer	ECHDC1,RNF146	rs2180341	AN,A5,A6	4864	3002	515	0.241	G	0.275	0.267	0.211	0.342
	COLIA1	rs2075555	AX,AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	6200	2024	150	0.139	-	0.183	0.411	0.367	0.233
	Intergenic	rs6556756	AH,AN,A5,A6,IM,IMD	6365	1742	113	0.120	-	0.100	0.400	0.500	0.025
	Intergenic	rs1154865	AH				0.223	-	0.153	0.211	0.122	0.275
	Intergenic	rs1978503	AH,AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	5480	2542	322	0.191	-	0.167	0.189	0.136	0.192
	ABCC4	rs1926657	AX,A6,I6Q,IM,IMD,IBC				0.172	-	0.158	0.422	0.386	0.417
	FGFR2	rs2981582	A6,IM,IMD				0.416	G	0.417	0.300	0.200	0.517
	TNCR9,LOC6,43714	rs3803662	AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	4347	3331	669	0.280	C	0.300	0.711	0.556	0.533
	MAP3K1	rs889312	I6Q,IMD,ICQ				0.279	A	0.308	0.500	0.545	0.328
	Intergenic	rs13281615	I6Q,IMD,ICQ				0.418	T	0.458	0.568	0.611	0.433
	LSP1	rs3817198	AS,A5,A6,I2,I5,I6,I6Q,IM,IMD,IBC	3901	3628	832	0.316	T	0.342	0.100	0.144	0.125
	FGFR2	rs1219648	A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.418	G	0.417	0.367	0.295	0.467
	TNRC9	rs3803662	AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	4347	3331	669	0.280	C	0.300	0.711	0.556	0.533
	Intergenic	rs13387042	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.481	A	0.633	0.133	0.111	0.775
Colorectal cancer	DQ515897	rs7014346	AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	3433	3853	1090	0.360	A	0.267	0.356	0.216	0.467
	Intergenic	rs3802842	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.293	C	0.242	0.375	0.300	0.342
	SMAD7	rs4939827	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.468	T	0.492	0.744	0.795	0.767
	Intergenic	rs10795668	I2,I5,I6,I6Q,IM,IMD				0.327	C	0.362	0.352	0.405	0.008
	EIF3H	rs16892766	AS,A6,I2,I5,I6,I6Q,IM,IMD	7132	636	249	0.071	A	0.117	0.000	0.000	0.229
	SMAD7	rs4939827	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.468	T	0.492	0.744	0.795	0.767
	Intergenic	rs6983267	A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.508	G	0.458	0.389	0.341	0.983
	ORF DQ515897	rs10505477	AH,AN,A5,A6,I6Q,IMD,ICQ	2117	4213	2054	0.496	A	0.442	0.378	0.341	0.900
Lung cancer	CHRNA3	rs8034191	A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.361	G	0.433	0.044	0.011	0.142
	CRP	rs2808630	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.305	G	0.283	0.244	0.114	0.183
	IL1RAP	rs7626795	AG,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	6209	1910	154	0.134	G	0.133	0.200	0.205	0.450
	CHRNA3, CHRNA5, CHRNB4	rs8034191	A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.361	G	0.433	0.044	0.011	0.142
Melanoma	CDC91L1	rs910873	A6,IM,IMD				0.098	T	0.092	0.000	0.000	0.000
Neuroblastoma	FLJ22536, FLJ44180	rs6939340	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.469	G	0.425	0.644	0.682	0.883
Prostate cancer	MSMB	rs10993994	I1,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.391	T	0.339	0.411	0.477	0.800
	KLK3	rs2735839	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.129	G	0.092	0.356	0.400	0.400
	Intergenic	rs7931342	I2,I5,I6,I6Q,IM,IMD				0.455	G	0.508	0.227	0.302	0.883
	SLC22A3	rs9364554	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.271	T	0.233	0.364	0.318	0.008
	LMTK2	rs6465657	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.434	C	0.525	0.089	0.057	0.000
	NUDT10, NUDT11, LOC340602, GSPT2, MAGED1	rs5945572	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				-	A	0.389	0.059	0.106	0.333
	EHBP1	rs721048	IM,IMD				0.177	A	0.117	0.022	0.045	0.000
	Intergenic	rs1529276	AH				0.187	-	0.183	0.000	0.034	0.000
	CTDSPL	rs9311171	AH,A6,I6Q,IM,IMD,IC,ICQ				0.159	-	0.175	0.089	0.011	0.567
	HAPLN1	rs4466137	AX,AN,A5,A6	4757	3079	504	0.245	-	0.225	0.167	0.159	0.175
	PKHD1	rs10498792	AX,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.111	-	0.108	0.211	0.111	0.267
	Intergenic	rs345013	AX				0.128	-	0.117	0.156	0.205	0.058
	TCF2	rs4430796	I2,I5,I6,I6Q,IM,IMD,IBC				0.487	A	0.467	0.722	0.667	0.390
	Intergenic	rs1859962	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.531	G	0.492	0.511	0.800	0.792
	Intergenic	rs16901979	AS,A5,A6	7947	429	6	0.026		0.025	0.289	0.156	0.542
	Intergenic	rs6983267	A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.508	G	0.458	0.389	0.341	0.983
Prostate cancer (aggressive)	MSMB	rs10993994	I1,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.391	T	0.339	0.411	0.477	0.800
	Intergenic	rs10896449	I2,I5,I6,I6Q,IM,IMD				0.461	G	0.508	0.089	0.033	0.708
	CTBP2	rs4962416	I2,I5,I6,I6Q,IM,IMD				0.271	C	0.229	0.011	0.011	0.233
	JAZF1	rs10486567	AH,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.231	G	0.267	0.844	0.867	0.317

Dermatologic Disorders												
Psoriasis	COG6	rs7993214	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.321	-	0.442	0.156	0.193	0.300
	SPATA2	rs495337	I1				0.395	-	0.430	0.330	0.398	0.076
Autoimmune Diseases												
Celiac disease	RGS1	rs2816316	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.187	C	0.217	0.178	0.284	0.267
	IL12A, SCHIP1	rs17810546	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.119	G	0.100	0.000	0.000	0.000
	IL1RL1,IL18R1,IL18RAP, SLC9A4	rs13015714	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.219	C	0.242	0.467	0.318	0.125
	LPP	rs1464510	AH,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.449	A	0.425	0.511	0.511	0.200
	TAGAP	rs1738074	I1,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.448	A	0.508	0.364	0.295	0.288
Systemic lupus erythematosus	ITGAM, ITGAX	rs11574637	AN,A6,I2,I5,I6,I6Q,IM,IMD	5303	2609	297	0.195	C	0.142	0.011	0.000	0.292
	C8orf13, BLK	rs13277113	I2,I5,I6,I6Q,IM,IMD				0.238	A	0.254	0.688	0.605	0.098
	BANK1	rs10516487	AH,AN,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	4134	3502	740	0.297	G	0.250	0.189	0.136	0.308
Systemic lupus erythematosus in women	ITGAM	rs9888739	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.136	T	0.108	0.011	0.000	0.708
	KIAA1542	rs4963128	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.346	-	0.350	0.033	0.022	0.550
	PXK	rs6445975	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.294	C	0.208	0.167	0.178	0.658
	Intergenic	rs10798269	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.346	-	0.375	0.244	0.256	0.533
Celiac disease	KIAA1109,TENR,IL2	rs6822844	I3,I5,I6,IM,IMD,IC,IBC				0.186	G	0.203	0.000	0.000	0.000
Crohn's disease	BSN, MST1	rs9858542	AS,A5,A6,I1,IM,IMD,IBC	4206	3454	718	0.292	A	0.242	0.044	0.078	0.200
	NKX2-3	rs10883365	AN,A5,A6,IBC	2095	4192	2087	0.500	G	0.500	0.456	0.500	0.508
	PTPN2	rs2542151	AN,A5,A6,IM,IMD,IBC	5855	2259	236	0.164	G	0.192	0.189	0.078	0.200
	IRGM	rs1000113	AS,A5,A6,IM,IMD,IBC	6837	1411	71	0.093	T	0.033	0.378	0.398	0.284
		rs9469220	AN,A5,A6,IM,IMD,IBC	2425	4076	1882	0.468	A	0.400	0.651	0.633	0.612
	IRGM	rs13361189	AN,A5,A6,IBC	6822	1433	74	0.095	-	0.025	0.422	0.389	0.533
	NKX2-3	rs10883365	AN,A5,A6,IBC	2095	4192	2087	0.500	G	0.500	0.456	0.500	0.508
	PTPN2	rs2542151	AN,A5,A6,IM,IMD,IBC	5855	2259	236	0.164	-	0.192	0.189	0.078	0.200
	Intergenic	rs10801047	AS,A5,A6,IBC	7114	1203	63	0.079	-	0.092	0.100	0.133	0.417
	ATG16L1	rs2241880	AG,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	2341	4117	1918	0.475	G	0.542	0.389	0.170	0.275
	Intergenic	rs224136	AS,A5,A6,I1,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	5862	2285	206	0.161	-	0.133	0.211	0.307	0.842
	Intergenic	rs1373692	I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC				0.402	-	0.400	0.833	0.922	0.333
Gastrointestinal Disorders												
Gallstones	ABCG8	rs11887534	No arrays, in HapMap				0.074	C	0.085	0.022	0.011	0.042
Inflammatory bowel disease	IL23R	rs7517847	I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC				0.423	C	0.500	0.422	0.364	0.117
	IL23R	rs11209026	AN,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	7156	1145	40	0.073	A	0.067	0.000	0.000	0.017
Metabolic Disorders												
Type 1 diabetes	RAB5B, SUOX, IKZF4, ERBB3, CDK2	rs1701704	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.317	C	0.292	0.278	0.261	0.095
	KIAA0350	rs2903692	I2,I5,I6,I6Q,IM,IMD				0.368	G	0.284	0.267	0.159	0.267
	SH2B3,LNK,TRAFD1,PTPN1	rs17696736	AN,A5,A6,I2,I5,I6,I6Q,IM,IMD,IBC	2490	4079	1795	0.458	G	0.350	0.000	0.000	0.000
	ERBB3	rs11171739	AS,A5,A6,IM,IMD,IBC	2874	4110	1387	0.411	C	0.375	0.278	0.267	0.842
	KIAA0350	rs12708716	AS,A6,IBC	3293	3275	1253	0.370	A	0.288	0.233	0.156	0.466
		rs11052552	AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	2124	4185	2054	0.496	G	0.483	0.330	0.398	0.858
		rs17388568	AS,A5,A6,IM,IMD,IBC	4807	3067	509	0.244	A	0.300	0.111	0.089	0.008
	ERBB3	rs2292239	AS,A5,A6,IM,IMD,IBC	3839	3659	882	0.324	A	0.300	0.256	0.256	0.417
	KIAA0350	rs12708716	AS,A6,IBC	3293	3275	1253	0.370	A	0.288	0.233	0.156	0.466
	PTPN2	rs2542151	AN,A5,A6,IM,IMD,IBC	5855	2259	236	0.164	-	0.192	0.189	0.078	0.200
	CD226	rs763361	AG,I1,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	2241	4140	1992	0.485	A	0.475	0.322	0.477	0.725
Type 2 diabetes	JAZF1	rs864745	AN,A5,A6,IBC	2111	4157	2112	0.500	T	0.482	0.189	0.216	0.237
	CDC123,CAMK1D	rs12779790	IBC				0.178	G	0.229	0.133	0.122	0.051
	TSPAN8,LGR5	rs7961581	AS,A5,A6,IBC	4155	3468	760	0.298	C	0.233	0.200	0.198	0.178
	THADA	rs7578597	AG,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	6628	1487	80	0.100	T	0.083	0.000	0.000	0.292
	ADAMTS9	rs4607103	AS,A5,A6,IBC	4525	3224	580	0.263	C	0.192	0.422	0.411	0.275
	FTO	rs9939609	AS,A5,A6,IBC	2935	4083	1346	0.405	A	0.450	0.122	0.167	0.517

	CDKAL1	rs9465871	AS,A5,A6,IBC	5308	2705	371	0.206	C	0.150	0.478	0.456	0.617
		rs358806	AN,A5,A6,I2,I5,I6,I6Q,IM,IMD,IBC	5312	2606	316	0.197	-	0.212	0.227	0.291	0.105
		rs12304921	AS,A6,IBC	5933	2213	204	0.157	G	0.175	0.534	0.375	0.133
		rs1495377	AS,A5,A6,IBC	2134	4205	1966	0.490	G	0.467	0.256	0.307	0.155
	IGF2BP2	rs4402960	AS,A5,A6,I2,I5,I6,I6Q,IM,IMD,IBC	3847	3341	954	0.322	T	0.292	0.222	0.311	0.550
	CDKN2A,CDKN2B	rs10811661	AS,A5,A6,IBC	5657	2392	273	0.177	T	0.208	0.405	0.433	0.000
	CDKAL1	rs7754840	AS,A5,A6,I6Q,IMD,IBC	3872	3636	874	0.321	C	0.308	0.411	0.378	0.667
	IGF2BP2	rs4402960	AS,A5,A6,I2,I5,I6,I6Q,IM,IMD,IBC	3847	3341	954	0.322	T	0.292	0.222	0.311	0.550
	CDKN2A, CDKN2B	rs10811661	AS,A5,A6,IBC	5657	2392	273	0.177	T	0.208	0.405	0.433	0.000
	CDKAL1	rs7754840	AS,A5,A6,I6Q,IMD,IBC	3872	3636	874	0.321	C	0.308	0.411	0.378	0.667
	Intergenic	rs9300039	AX,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.094	C	0.108	0.300	0.278	0.169
	CDKAL1	rs7756992	AG,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	4340	3351	686	0.282	G	0.250	0.467	0.466	0.633
	CDKN2B	rs10811661	AS,A5,A6,IBC	5657	2392	273	0.177	T	0.208	0.405	0.433	0.000
	FTO	rs8050136	AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	2928	4042	1351	0.405	A	0.450	0.122	0.167	0.467
	CDKAL1	rs10946398	AS,A5,A6,IBC	3857	3635	875	0.322	C	0.308	0.411	0.378	0.667
	HHEX	rs5015480	AS,A5,A6,I2,I5,I6,I6Q,IM,IMD,IBC	3051	3997	1313	0.396	C	0.552	0.211	0.189	0.568
	SLC30A8	rs13266634	I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC				0.258	C	0.250	0.478	0.444	0.058
	HHEX	rs1111875	A6,I1,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC				0.395	G	0.442	0.678	0.591	0.142
Incident diabetes	TMEFF2	rs10497721	AX				0.094	-	0.042	0.367	0.250	0.025
Diabetic nephropathy	ELMO1	rs741301	A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.333	-	0.333	0.267	0.409	0.675
End-stage renal disease	PVT1	rs2648875	I6,IM,IMD				0.236	A	0.317	0.600	0.705	0.475
Neurodegenerative Disorders												
Amyotrophic lateral sclerosis	DPP6	rs10260404	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.381	C	0.392	0.200	0.140	0.350
	ITPR2	rs2306677	AG,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	6752	1532	90	0.102	-	0.083	0.200	0.205	0.254
	Intergenic	rs4363506	A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.425	-	0.325	0.544	0.489	0.283
		rs16984239	AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	5599	2486	299	0.184	-	0.183	0.322	0.211	0.025
		rs12680546	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.273	-	0.325	0.122	0.070	0.043
	ZFP64	rs6013382	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.466	-	0.466	0.267	0.367	0.525
	SUSD1	rs2782931	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.323	-	0.325	0.311	0.205	0.133
Parkinson's disease	Intergenic	rs1480597	AH,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.180	-	0.133	0.011	0.000	0.033
	BRDG1	rs2242330	I1,IM,IMD				0.225	-	0.190	0.233	0.136	0.093
	DLG2	rs10501570	AX,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.181	-	0.217	0.022	0.011	0.000
	SEMA5A	rs7702187	No arrays, not in HapMap				0.151	-	0.175	0.322	0.295	0.670
Multiple sclerosis	IL2RA	rs12722489	AN,A6,IMD	6159	2058	163	0.142	C	0.158	0.144	0.167	0.000
	IL7RA	rs6897932	AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	4656	3153	560	0.255	C	0.242	0.200	0.193	0.050
	KIAA0350	rs6498169	AN,A5,A6	3728	3689	955	0.334	G	0.392	0.578	0.533	0.200
	RPL5	rs6604026	AN,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ	4301	3367	715	0.286	C	0.258	0.011	0.011	0.250
	DBC1	rs10984447	AN,A5,A6	4958	2981	319	0.219	A	0.373	0.111	0.178	0.034
APOE*e4 carriers with late onset Alzheimer's disease	GAB2	rs2373115	AN,A5,A6,I1,IBC	5727	2396	249	0.173	G	0.108	0.422	0.533	0.350
Exfoliation glaucoma	LOXL1	rs3825942	I1,IM,IMD				0.161	G	0.172	0.111	0.125	0.415
Wet age-related macular degeneration	HTRA1	rs11200638	No arrays, not in HapMap				-	A	-	-	-	-
Age-related macular degeneration	CFH	rs380390	AX				0.400	C	0.450	0.078	0.068	0.233

Psychiatric Disorders												
Bipolar disorder	PALB2,NDUFAB1,DCTN5	rs420259	AS,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	4212	3401	654	0.285	A	0.200	0.378	0.333	0.425
		rs6458307	AN,A5,A6,I3,I5,I6,IM,IMD,IC,IBC	4004	3613	642	0.296	-	0.336	0.222	0.273	0.567
		rs683395	AN,A5,A6,I3,I5,I6,I6Q,IM,IMD,IC,ICQ,IBC	6846	1463	72	0.096	G	0.100	0.067	0.100	0.208
		rs3761218	AS,A5,A6,IM,IMD,IBC	3029	3964	1376	0.401	C	0.408	0.289	0.267	0.212
		rs2953145	AS,A5,A6,IBC	5136	2775	395	0.215	C	0.208	0.067	0.011	0.200
	DGKH	rs1012053	I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.162	A	0.150	0.533	0.522	0.075
Neuroticism	PDE3D	rs702543	AH,I3,I5,I6,I6Q,IM,IMD,IC,ICQ				0.565	A	0.492	0.690	0.727	0.042
Schizophrenia	CCDC60	rs11064768	AG,A6,I2,I5,I6,I6Q,IM,IMD	6992	1287	65	0.085	A	0.095	0.000	0.000	0.000
	CDF2RA	rs4129148	AS,A5,A6	3670	3685	988	0.339	C	0.342	0.515	0.379	0.669