

Supplemental Material for:

Interlocus gene conversion events introduce deleterious mutations into at least 1% of human genes associated with inherited disease

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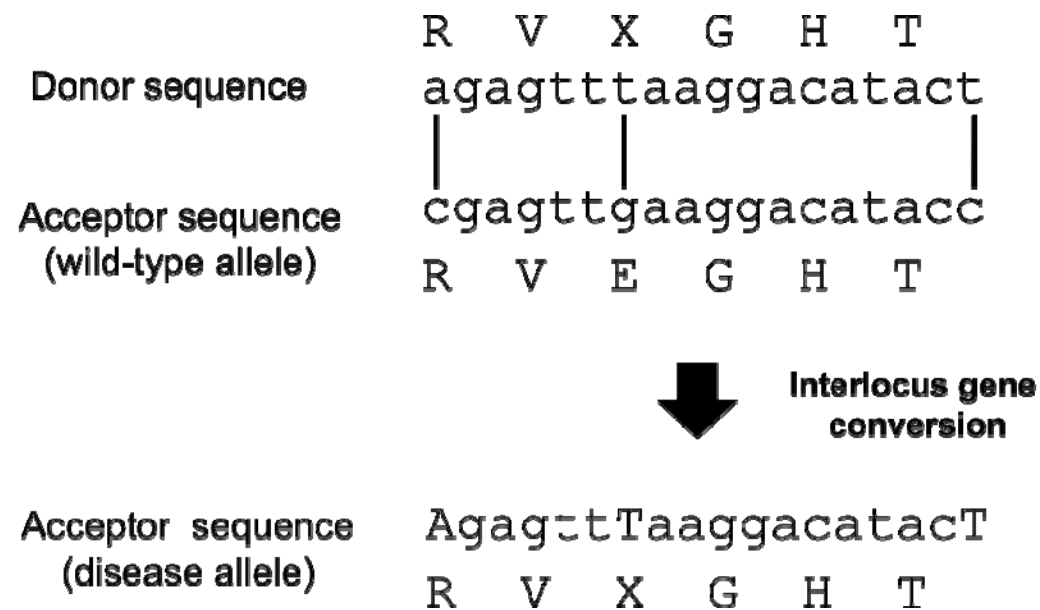
Methods

Validation of disease alleles originating from interlocus gene conversion

All candidate IGC-derived disease alleles identified by our BLAST searches were further examined to identify high-confidence IGC events. Such alleles required at least one disease-associated mutation plus another mutation to be shared by the IGC donor sequence. In addition, we excluded from our data set those cases where hypermutable CpG sites constituted all the diagnostic mutations. After careful examination of the available literature, we also disregarded several putative IGC events that were deemed likely to represent experimental artifacts. In particular, we identified several cases where primer pairs, which were originally considered to have amplified a single genomic locus or cDNA transcript, also appeared to be capable of amplifying sequences paralogous to the target genes. Such instances included disease alleles from the genes *CFC1* (Bamford et al. 2000), *GPD2* (Novials et al. 1997) and *HLA-DRB1* (Velickovic et al. 2004). In the first two of these cases, we noted that the primer pairs that had been originally designed to amplify the genes specifically, also perfectly matched the paralogous gene *CFC1B* and an unlinked processed pseudogene of *GPD2*, respectively. Given the publication years of the first two papers (1997 and 2000), it is likely that the presence of such sequences paralogous to the target genes would not have been appreciated at the time. In the latter case, both primers displayed insufficient specificity to permit unambiguous assignment of the reported mutations.

Disputed IGC events involving the von Willebrand factor (VWF) gene and its pseudogene

Recently, several cases of disease alleles putatively derived from IGC events involving the von Willebrand factor gene (*VWF*) and its pseudogene (*VWFP*) have been disputed, mostly on the grounds of inappropriate PCR conditions and/or polymorphisms in the pseudogene (Kasatkar et al. 2010). *VWFP* is a 2,907 bp long non-processed pseudogene (located on 22q11.1) that includes exons 23-34 of the *VWF* gene (located on 12p13.31). We however opted to retain these cases in our study for the following reasons. First, of the 65 single nucleotide differences between the *VWF* and *VWFP* coding regions, very few have been found to coincide with disease mutations that have originated via IGC involving this gene-pseudogene pair. In practice, the non-specific amplification of both *VWF* gene and pseudogene would have given rise to many more such mutations. This is also the case when only the 1,379 bp exon 28 of *VWF*, which includes all IGC-derived mutations hitherto described in *VWF*, is considered (the nucleotide differences between *VWF* and *VWFP* within exon 28 amount to 38/1,379 bp). Second, several authors succeeded in identifying IGC-derived mutations in DNA samples derived from patients but not from negative controls (Altschul et al. 1990). Third, cDNA was used in some studies as the only source of DNA to investigate mutations in the *VWF* gene (Eikenboom et al. 1994). We were unable to identify any ESTs corresponding to *VWFP* within either the NCBI human EST db or the EST trace archive (<http://www.ncbi.nlm.nih.gov/>), whereas dozens of expressed sequences corresponding to the *VWF* gene were retrievable (data not shown). Hence, the PCR amplification of *VWFP* from cDNA appears very unlikely given that *VWFP* is either not transcribed or else is transcribed at a much lower level than *VWF*. Finally, patients homozygous for IGC-derived mutations have been reported in some cases (Zhang et al. 1993; Gupta et al. 2005; Terai et al. 2010).



Supplemental Figure 1. Schematic representation of a hypothetical high-confidence IGC event. The paralogous donor and acceptor loci differ at multiple nucleotides, indicated by vertical lines between the sequences. High-confidence IGC events are required to include a pathological mutation and at least one secondary mutation found in the same disease allele (i.e. haplotype). In this example, the G8T nucleotide substitution in the acceptor gene introduces a stop codon (E3X, where X represents the stop codon) and is assumed to be responsible for an inherited disease. The disease allele also harbors two synonymous nucleotide changes, C1A and C18T, which support the inference that an IGC event was responsible for introducing the pathological mutation. Mutated nucleotides in the disease allele are given in uppercase.

Supplemental Table 1. HGMD mutations occurring within paralogous sequences with ≥ 200 bp and $\geq 92\%$ sequence identity

Gene	HGMD_ID	wildBASE	mutBASE	Acceptor Chr.	Mutation Position	% identity	Donor Chr.	Donor Start	Donor end	TYPE	Tag	CpG
<i>ABCC6</i>	CM012720	C	T	16	16203403	98.51	16	18511394	18511594	MISNON	DM	0
<i>ABCC6</i>	CM080016	G	A	16	16221013	99.50	16	18493775	18493975	MISNON	DM	1
<i>ABCD1</i>	CM000643	G	A	X	152659248	95.02	22	15248347	15248547	MISNON	DM	1
<i>ABCD1</i>	CM011743	G	A	X	152661906	97.01	22	15251034	15251234	MISNON	DM	1
<i>ABCD1</i>	CM066723	T	C	X	152659341	96.02	10	38934578	38934778	MISNON	DM	0
<i>ABCD1</i>	CM066723	T	C	X	152659341	96.52	22	15248440	15248640	MISNON	DM	0
<i>ABCD1</i>	CM940041	C	T	X	152661703	96.52	22	15250831	15251031	MISNON	DM	1
<i>ABCD1</i>	CM950056	G	A	X	152661679	95.52	22	15250807	15251007	MISNON	DM	1
<i>ABCD1</i>	CM950057	G	A	X	152661879	94.12	16	32394279	32394482	MISNON	DM	1
<i>ABCD1</i>	CM950057	G	A	X	152661879	97.01	22	15251007	15251207	MISNON	DM	1
<i>ABCD1</i>	CM960043	T	C	X	152661670	94.03	10	38936937	38937137	MISNON	DM	0
<i>ABCD1</i>	CM960043	T	C	X	152661670	96.02	16	32394491	32394691	MISNON	DM	0
<i>ABCD1</i>	CM960043	T	C	X	152661670	96.02	2	91394746	91394946	MISNON	DM	0
<i>ABCD1</i>	CM960043	T	C	X	152661670	95.52	22	15250798	15250998	MISNON	DM	0
<i>AKR7A3</i>	CM062419	G	A	1	19483828	92.04	1	19468315	19468515	MISNON	DP	0
<i>APOC3</i>	CM942309	T	C	11	116206745	99.50	22	20195	20395	MISNON	FP	0
<i>C4B</i>	CX033310	TCTCTCCAGTGATACAT	cctgtccagtgttagac	6	32071839	100.00	6	32104476	32104692	INDEL	DM	0
<i>CFC1</i>	CM002873	C	T	2	131072700	99.00	2	130996057	130996257	MISNON	DM	1
<i>CFH</i>	CM010322	T	C	1	194982960	99.00	1	195067547	195067747	MISNON	DM	0
<i>CFH</i>	CM062504	G	A	1	194949570	92.54	1	195120342	195120542	MISNON	DP	1
<i>CFH</i>	CM993344	C	T	1	194982942	99.00	1	195067529	195067729	MISNON	DM	1
<i>CFTR</i>	CP942282	ttttg	ctttta	7	116976121	97.57	20	25848022	25848227	INDEL	DM	0
<i>CFTR</i>	CP942282	ttttg	ctttta	7	116976121	94.66	20	28063022	28063223	INDEL	DM	0
<i>CYP11B1</i>	HM972177	C	T	8	143953695	99.00	8	143991169	143991369	MISNON	DM	1
<i>CYP11B2</i>	CM087150	G	T	8	143992748	93.53	8	143954626	143954826	MISNON	FP	0
<i>CYP11B2</i>	CM087151	G	A	8	143991821	94.03	8	143954149	143954349	MISNON	FP	1
<i>CYP11B2</i>	CM087152	A	G	8	143991135	95.02	8	143953463	143953663	MISNON	FP	0
<i>CYP11B2</i>	CM087153	G	C	8	143991106	94.53	8	143953434	143953634	MISNON	FP	0
<i>CYP11B2</i>	CM095226	C	G	8	143995613	92.04	8	143957487	143957687	MISNON	FP	0

Supplemental Table 1. Continued

Gene	HGMD_ID	wildBASE	mutBASE	Acceptor Chr.	Mutation Position	% identity	Donor Chr.	Donor Start	Donor end	TYPE	Tag	CpG
<i>CYP11B2</i>	CM920222	T	C	8	143991268	99.00	8	143953596	143953796	MISNON	DM	0
<i>CYP21A2</i>	CM880020	T	A	6	32115182	99.50	6	32082346	32082546	MISNON	DM	0
<i>CYP21A2</i>	CM880023	C	T	6	32116177	99.50	6	32083342	32083542	MISNON	DM	0
<i>CYP21A2</i>	CM900080	C	T	6	32116291	99.50	6	32083456	32083656	MISNON	DM	1
<i>CYP21A2</i>	CX065865	TCACATCGT	ccacaacga	6	32115558	98.56	6	32082721	32082929	INDEL	DM	0
<i>CYP2A6</i>	CM083626	G	C	19	46047578	95.02	19	46286720	46286920	MISNON	FP	0
<i>CYP2A6</i>	CM083626	G	C	19	46047578	94.53	19	46224763	46224963	MISNON	FP	0
<i>CYP2A6</i>	CM102863	C	A	19	46046011	98.51	19	46077777	46077977	MISNON	FP	0
<i>CYP2B6</i>	CM016107	C	T	19	46214555	96.52	19	46146863	46147063	MISNON	FP	1
<i>CYP2B6</i>	CM078372	C	T	19	46210210	93.03	19	46142144	46142344	MISNON	DM	1
<i>CYP2C19</i>	CM983294	A	G	10	96592613	94.03	10	96730860	96731060	MISNON	FP	0
<i>CYP2D6</i>	CM033368	G	A	22	40856707	96.52	22	40870280	40870480	MISNON	FP	1
<i>CYP2D6</i>	CM930187	C	T	22	40853887	93.56	22	40877079	40877280	MISNON	DP	1
<i>CYP2D6</i>	CM930187	C	T	22	40853887	93.60	22	40867488	40867690	MISNON	DP	1
<i>CYP2D6</i>	CM950334	G	T	22	40854979	93.03	22	40878183	40878383	MISNON	DM	0
<i>CYP2D6</i>	CM992872	G	A	22	40854979	93.03	22	40878183	40878383	MISNON	FP	1
<i>FCGR2B</i>	CM078107	C	T	1	159907841	96.02	1	159825912	159826112	MISNON	DP	0
<i>FCGR3A</i>	CM061766	G	C	1	159785046	98.51	1	159866304	159866504	MISNON	DP	0
<i>FCGR3A</i>	CM992625	G	T	1	159781166	98.01	1	159862511	159862711	MISNON	DFP	0
<i>GBA</i>	CM001166	T	C	1	153474643	97.01	1	153453267	153453467	MISNON	DM	0
<i>GBA</i>	CM082771	A	G	1	153473827	99.00	1	153452771	153452971	MISNON	DM	0
<i>GBA</i>	CM870010	T	C	1	153471667	98.51	1	153450941	153451141	MISNON	DM	0
<i>GBA</i>	CM900104	G	A	1	153472794	97.51	1	153452013	153452213	MISNON	DM	0
<i>GBA</i>	CM900107	G	C	1	153471632	98.51	1	153450906	153451106	MISNON	DM	0
<i>GBA</i>	CM910173	T	A	1	153474556	95.52	1	153453180	153453380	MISNON	DM	0
<i>GBA</i>	CM940807	G	A	1	153474589	96.02	1	153453213	153453413	MISNON	DM	1
<i>GBA</i>	CM960694	C	T	1	153475045	99.50	1	153453669	153453869	MISNON	DM	1
<i>GBA</i>	CM980832	T	G	1	153474629	97.01	1	153453253	153453453	MISNON	DM	0
<i>GBA</i>	CM980833	T	G	1	153474621	97.01	1	153453245	153453445	MISNON	DM	0
<i>GBA</i>	CM992893	T	C	1	153474607	97.01	1	153453231	153453431	MISNON	DM	0

Supplemental Table 1. Continued

Gene	HGMD_ID	wildBASE	mutBASE	Acceptor Chr.	Mutation Position	% identity	Donor Chr.	Donor Start	Donor end	TYPE	Tag	CpG
<i>GBA</i>	CX092868	GCTGTTGTGGTCGTG	cctgttggtgcgtc	1	153471618	99.53	1	153450891	153451105	INDEL	DM	0
<i>GH1</i>	CM092297	T	C	17	59348956	94.53	17	59311869	59312069	MISNON	DM	0
<i>GH1</i>	CM092298	G	C	17	59348520	96.02	17	59326134	59326334	MISNON	DM	0
<i>GH1</i>	CM092298	G	C	17	59348520	94.03	17	59340823	59341023	MISNON	DM	0
<i>GH1</i>	CM104401	T	A	17	59349457	93.53	17	59312371	59312571	MISNON	DM	0
<i>GH1</i>	CM970645	A	G	17	59349862	97.01	17	59304601	59304801	MISNON	DM	0
<i>GH1</i>	CM970645	A	G	17	59349862	94.53	17	59342184	59342383	MISNON	DM	0
<i>GH1</i>	CM970645	A	G	17	59349862	98.00	17	59312788	59312987	MISNON	DM	0
<i>GJA1</i>	CM014191	G	A	6	121810777	96.02	5	108414875	108415075	MISNON	DM	1
<i>GJA1</i>	CM014192	G	A	6	121810819	96.52	5	108414833	108415033	MISNON	DM	1
<i>GJA1</i>	CM030456	C	T	6	121809811	97.51	5	108415838	108416038	MISNON	DM	1
<i>GJA1</i>	CM040077	G	A	6	121810135	96.52	5	108415517	108415714	MISNON	DM	1
<i>GJA1</i>	CM055949	C	A	6	121810561	98.01	5	108415091	108415291	MISNON	DM	0
<i>GJA1</i>	CM055950	G	C	6	121810444	96.02	5	108415208	108415408	MISNON	DM	0
<i>GJA1</i>	CM055952	C	T	6	121810407	97.01	5	108415245	108415445	MISNON	DM	1
<i>GJA1</i>	CM055953	G	C	6	121810449	96.02	5	108415203	108415403	MISNON	DM	0
<i>GLUL</i>	CM057402	C	T	1	180620315	94.03	9	34908040	34908240	MISNON	DM	1
<i>GUSB</i>	CM092097	G	A	7	65077108	92.04	7	63212340	63212540	MISNON	DM	1
<i>GUSB</i>	CM092098	C	T	7	65077101	92.04	7	63212347	63212547	MISNON	DM	1
<i>GYP A</i>	CM0910231	T	G	4	145259346	95.52	4	145139944	145140144	MISNON	DM	0
<i>HBA2</i>	CR042844	G	A	16	163735	100.00	16	163635	163835	REG	DM	0
<i>HBG2</i>	CM960822	T	C	11	5232186	99.50	11	5227163	5227363	MISNON	DM	0
<i>HLA-A</i>	CM068505	G	T	6	30019243	93.53	6	29964343	29964543	MISNON	FP	0
<i>HLA-DRB1</i>	CM033913	A	T	6	32660007	92.00	6	32597704	32597903	MISNON	DP	0
<i>HSD3B2</i>	CM021276	A	C	1	119766766	93.03	1	119790136	119790336	MISNON	DM	0
<i>IFNA17</i>	CM041015	T	G	9	21217622	97.01	9	21176881	21177081	MISNON	DP	0
<i>IFNA17</i>	CM041015	T	G	9	21217622	97.01	9	21196447	21196647	MISNON	DP	0
<i>IFNA17</i>	CM041015	T	G	9	21217622	94.53	9	21229285	21229485	MISNON	DP	0
<i>KRT14</i>	CM950722	C	A	17	36993644	95.02	17	16663513	16663713	MISNON	DM	0
<i>KRT14</i>	CM950724	C	T	17	36993125	96.02	17	16662994	16663194	MISNON	DM	1

Supplemental Table 1. Continued

Gene	HGMD_ID	wildBASE	mutBASE	Acceptor Chr.	Mutation Position	% identity	Donor Chr.	Donor Start	Donor end	TYPE	Tag	CpG
<i>KRT18</i>	CM034541	A	G	12	51629531	92.04	5	40102716	40102916	MISNON	DM	0
<i>KRT18</i>	CM061831	C	G	12	51632278	93.03	6	29045022	29045222	MISNON	DM	0
<i>LHB</i>	CM022419	G	A	19	54211747	96.02	19	54227508	54227708	MISNON	FP	0
<i>LHB</i>	CM022419	G	A	19	54211747	96.02	19	54239581	54239781	MISNON	FP	0
<i>LHB</i>	CM022419	G	A	19	54211747	96.02	19	54218537	54218737	MISNON	FP	0
<i>LHB</i>	CM022419	G	A	19	54211747	96.02	19	54243306	54243506	MISNON	FP	0
<i>LHB</i>	CM022419	G	A	19	54211747	94.03	19	54231237	54231437	MISNON	FP	0
<i>LHB</i>	CM022419	G	A	19	54211747	93.53	19	54249942	54250142	MISNON	FP	0
<i>LHB</i>	CM993948	T	C	19	54211717	96.02	19	54227538	54227738	MISNON	DP	0
<i>LHB</i>	CM993948	T	C	19	54211717	96.02	19	54239611	54239811	MISNON	DP	0
<i>LHB</i>	CM993948	T	C	19	54211717	96.02	19	54218507	54218707	MISNON	DP	0
<i>LHB</i>	CM993948	T	C	19	54211717	96.02	19	54243276	54243476	MISNON	DP	0
<i>LHB</i>	CM993948	T	C	19	54211717	95.02	19	54231207	54231407	MISNON	DP	0
<i>LHB</i>	CM993948	T	C	19	54211717	95.02	19	54249912	54250112	MISNON	DP	0
<i>LHB</i>	CM993949	T	C	19	54211695	95.02	19	54231185	54231385	MISNON	DP	0
<i>LTF</i>	CM051945	G	C	3	46455962	99.50	3	734144	734344	MISNON	DP	0
<i>MRC1</i>	CM099897	G	A	10	18178636	98.01	10	17931612	17931812	MISNON	DP	1
<i>MYH7</i>	CM011799	C	T	14	22964847	98.01	14	22935747	22935947	MISNON	DM	1
<i>NF1</i>	CM000796	A	G	17	26583996	93.03	15	19399113	19399313	MISNON	DM	0
<i>NF1</i>	CM032012	C	T	17	26587083	93.03	15	19396329	19396529	MISNON	DM	0
<i>NF1</i>	CM087437	C	T	17	26551587	94.53	18	14147219	14147419	MISNON	DM	1
<i>NF1</i>	CM087525	A	G	17	26565668	96.52	2	131671876	131672076	MISNON	DM	0
<i>NF1</i>	CM087525	A	G	17	26565668	96.52	14	18565252	18565452	MISNON	DM	0
<i>NF1</i>	CM087525	A	G	17	26565668	96.52	14	19148007	19148207	MISNON	DM	0
<i>NF1</i>	CM087525	A	G	17	26565668	95.02	22	14725888	14726088	MISNON	DM	0
<i>NF1</i>	CM087526	C	T	17	26580376	96.02	15	19402801	19403001	MISNON	DM	1
<i>NF1</i>	CM087526	C	T	17	26580376	95.02	12	36886955	36887154	MISNON	DM	1
<i>NF1</i>	CM087527	A	G	17	26580422	95.02	15	19402755	19402955	MISNON	DM	0
<i>NF1</i>	CM096910	T	C	17	26551576	94.03	18	14147230	14147430	MISNON	DM	0
<i>NF1</i>	CM973234	C	G	17	26584259	93.03	15	19398854	19399050	MISNON	DM	0

Supplemental Table 1. Continued

Gene	HGMD_ID	wildBASE	mutBASE	Acceptor Chr.	Mutation Position	% identity	Donor Chr.	Donor Start	Donor end	TYPE	Tag	CpG
<i>NLRP7</i>	CM094534	A	G	19	60142467	92.04	19	60186414	60186614	MISNON	DM	0
<i>OPN1LW</i>	CM045801	T	C	X	153073353	97.51	X	153110384	153110584	MISNON	FP	0
<i>OPN1LW</i>	CM045801	T	C	X	153073353	97.51	X	153147502	153147702	MISNON	FP	0
<i>OPN1LW</i>	CM045802	G	T	X	153073361	97.51	X	153110392	153110592	MISNON	FP	0
<i>OPN1LW</i>	CM045802	G	T	X	153073361	97.51	X	153147510	153147710	MISNON	FP	0
<i>OR12D2</i>	CM035845	G	T	6	29472814	94.03	6	29493295	29493495	MISNON	FP	0
<i>PKD1</i>	CM003336	C	T	16	2096601	98.01	16	14927339	14927539	MISNON	DM	1
<i>PKD1</i>	CM003336	C	T	16	2096601	98.01	16	16325990	16326190	MISNON	DM	1
<i>PKD1</i>	CM003336	C	T	16	2096601	98.01	16	16370278	16370478	MISNON	DM	1
<i>PKD1</i>	CM003336	C	T	16	2096601	98.01	16	15135255	15135455	MISNON	DM	1
<i>PKD1</i>	CM003336	C	T	16	2096601	97.51	16	18345193	18345393	MISNON	DM	1
<i>PKD1</i>	CM003336	C	T	16	2096601	97.01	16	18384419	18384619	MISNON	DM	1
<i>PKD1</i>	CM010387	T	C	16	2093612	96.52	16	18341843	18342043	MISNON	DM	0
<i>PKD1</i>	CM010387	T	C	16	2093612	98.51	16	14930793	14930993	MISNON	DM	0
<i>PKD1</i>	CM010387	T	C	16	2093612	98.51	16	16329278	16329478	MISNON	DM	0
<i>PKD1</i>	CM010387	T	C	16	2093612	98.51	16	18381953	18382153	MISNON	DM	0
<i>PKD1</i>	CM010387	T	C	16	2093612	98.01	16	15132681	15132881	MISNON	DM	0
<i>PKD1</i>	CM010387	T	C	16	2093612	97.01	16	16372580	16372780	MISNON	DM	0
<i>PKD1</i>	CM014074	T	G	16	2094574	99.00	16	14929708	14929908	MISNON	DM	0
<i>PKD1</i>	CM014074	T	G	16	2094574	99.00	16	16328192	16328392	MISNON	DM	0
<i>PKD1</i>	CM014074	T	G	16	2094574	99.00	16	18342955	18343155	MISNON	DM	0
<i>PKD1</i>	CM034116	C	T	16	2098571	95.52	16	16368308	16368508	MISNON	DM	1
<i>PKD1</i>	CM034120	C	T	16	2087973	98.51	16	16378210	16378410	MISNON	DM	1
<i>PKD1</i>	CM034120	C	T	16	2087973	98.51	16	18336213	18336413	MISNON	DM	1
<i>PKD1</i>	CM074422	G	T	16	2093631	98.01	16	14930774	14930974	MISNON	DM	0
<i>PKD1</i>	CM074422	G	T	16	2093631	98.01	16	16329259	16329459	MISNON	DM	0
<i>PKD1</i>	CM074422	G	T	16	2093631	98.01	16	15132700	15132900	MISNON	DM	0
<i>PKD1</i>	CM074422	G	T	16	2093631	98.01	16	18381972	18382172	MISNON	DM	0
<i>PKD1</i>	CM074422	G	T	16	2093631	96.52	16	16372561	16372761	MISNON	DM	0
<i>PKD1</i>	CM074422	G	T	16	2093631	96.02	16	18341862	18342062	MISNON	DM	0

Supplemental Table 1. Continued

Gene	HGMD_ID	wildBASE	mutBASE	Acceptor Chr.	Mutation Position	% identity	Donor Chr.	Donor Start	Donor end	TYPE	Tag	CpG
<i>PKD1</i>	CM076395	T	C	16	2104845	99.00	16	14923340	14923540	MISNON	DM	0
<i>PKD1</i>	CM076395	T	C	16	2104845	99.00	16	16321991	16322191	MISNON	DM	0
<i>PKD1</i>	CM076395	T	C	16	2104845	99.00	16	16362020	16362220	MISNON	DM	0
<i>PKD1</i>	CM076395	T	C	16	2104845	99.00	16	18392677	18392877	MISNON	DM	0
<i>PKD1</i>	CM971179	T	C	16	2093780	96.52	16	14930625	14930825	MISNON	DP	0
<i>PKD1</i>	CM971179	T	C	16	2093780	96.02	16	16329110	16329310	MISNON	DP	0
<i>PKD1</i>	CM971179	T	C	16	2093780	96.02	16	18382121	18382321	MISNON	DP	0
<i>PKD1</i>	CM971180	G	C	16	2093777	96.52	16	14930628	14930828	MISNON	DP	0
<i>PKD1</i>	CM971180	G	C	16	2093777	96.02	16	16329113	16329313	MISNON	DP	0
<i>PKD1</i>	CM971180	G	C	16	2093777	96.02	16	18382118	18382318	MISNON	DP	0
<i>PKD1</i>	CM971182	T	C	16	2093768	98.01	16	15132837	15133037	MISNON	DP	0
<i>PKD1</i>	CM971182	T	C	16	2093768	97.01	16	14930637	14930837	MISNON	DP	0
<i>PKD1</i>	CM971182	T	C	16	2093768	96.52	16	16329122	16329322	MISNON	DP	0
<i>PKD1</i>	CM971182	T	C	16	2093768	96.52	16	16372424	16372624	MISNON	DP	0
<i>PKD1</i>	CM971182	T	C	16	2093768	96.52	16	18341999	18342199	MISNON	DP	0
<i>PKD1</i>	CM971182	T	C	16	2093768	96.52	16	18382109	18382309	MISNON	DP	0
<i>PKD1</i>	CM991043	T	C	16	2104491	98.50	16	16322346	16322545	MISNON	DM	0
<i>PKD1</i>	CM991043	T	C	16	2104491	98.50	16	16362375	16362574	MISNON	DM	0
<i>PKD1</i>	CM991043	T	C	16	2104491	98.50	16	18348838	18349037	MISNON	DM	0
<i>PKD1</i>	CM991043	T	C	16	2104491	98.00	16	14923695	14923894	MISNON	DM	0
<i>PKD1</i>	CM991043	T	C	16	2104491	98.00	16	18392323	18392522	MISNON	DM	0
<i>PMS2</i>	CX075740	GC	agt	7	5993190	95.57	7	6743808	6744008	INDEL	DM	0
<i>PRDM9</i>	CM098032	C	G	5	23563008	99.00	5	23562825	23563025	MISNON	DM	0
<i>PRDM9</i>	CM098032	C	G	5	23563008	97.51	5	23562741	23562941	MISNON	DM	0
<i>PRODH</i>	CM022054	T	A	22	17289902	98.01	22	18671158	18671358	MISNON	DM	0
<i>PRODH</i>	CM022055	T	C	22	17285934	96.02	22	18675278	18675478	MISNON	DM	0
<i>PRODH</i>	CM057358	C	T	22	17285859	97.01	22	18675353	18675553	MISNON	DP	1
<i>PRODH</i>	CM057359	C	T	22	17286998	98.51	22	18674213	18674413	MISNON	DP	1
<i>PRODH</i>	CM057360	C	T	22	17285899	96.02	22	18675313	18675513	MISNON	DP	1
<i>PRODH</i>	CM057555	G	A	22	17285964	97.01	22	18675248	18675448	MISNON	DP	1

Supplemental Table 1. Continued

Gene	HGMD_ID	wildBASE	mutBASE	Acceptor Chr.	Mutation Position	% identity	Donor Chr.	Donor Start	Donor end	TYPE	Tag	CpG
<i>PRODH</i>	CM057556	G	A	22	17285842	97.01	22	18675370	18675570	MISNON	DP	1
<i>PRODH</i>	CM057558	G	A	22	17285977	97.01	22	18675235	18675435	MISNON	DP	1
<i>PROS1</i>	CM095623	C	T	3	95094634	95.02	3	90353824	90354024	MISNON	DM	1
<i>PROS1</i>	CM951045	C	T	3	95128785	97.51	3	90389002	90389202	MISNON	DM	1
<i>PROS1</i>	CM951056	C	T	3	95086403	96.02	3	90345695	90345895	MISNON	DM	1
<i>PRSS1</i>	CM993429	C	T	7	142137986	95.02	7	142159392	142159592	MISNON	DM	0
<i>PRSS1</i>	CM993429	C	T	7	142137986	92.54	9	33786548	33786748	MISNON	DM	0
<i>PRSS1</i>	CP015809	GC	at	7	142139363	93.00	7	142160677	142160876	INDEL	DM	0
<i>RANBP2</i>	CM090148	C	T	2	108734881	98.51	2	106418911	106419111	MISNON	DM	1
<i>RANBP2</i>	CM090148	C	T	2	108734881	98.01	2	107842633	107842833	MISNON	DM	1
<i>RANBP2</i>	CM090148	C	T	2	108734881	97.51	2	87875651	87875851	MISNON	DM	1
<i>RANBP2</i>	CM090148	C	T	2	108734881	97.51	2	87055338	87055538	MISNON	DM	1
<i>RDX</i>	CM071969	G	A	11	109607824	92.54	11	4188074	4188274	MISNON	DM	1
<i>RHCE</i>	CM011810	G	C	1	25619817	99.50	1	25471572	25471772	MISNON	FP	0
<i>RHCE</i>	CM011811	G	T	1	25589961	97.51	1	25500529	25500729	MISNON	FP	0
<i>RHCE</i>	CM015257	C	G	1	25589931	97.01	1	25500559	25500759	MISNON	FP	0
<i>RHCE</i>	CM015258	A	G	1	25589916	96.52	1	25500574	25500774	MISNON	FP	0
<i>RHCE</i>	CM099530	T	A	1	25589828	97.51	1	25500662	25500862	MISNON	FP	0
<i>RHCE</i>	CM930645	T	C	1	25607789	99.50	1	25483708	25483908	MISNON	FP	0
<i>RHCE</i>	CM961250	C	G	1	25589895	96.52	1	25500595	25500795	MISNON	FP	0
<i>RHCE</i>	CM995294	G	C	1	25591104	96.02	1	25500038	25500238	MISNON	FP	0
<i>RHD</i>	CM004833	G	C	1	25471673	99.50	1	25619716	25619916	MISNON	FP	0
<i>RHD</i>	CM004838	T	C	1	25505759	92.54	1	25584736	25584936	MISNON	FP	0
<i>RHD</i>	CM024420	C	T	1	25516140	99.50	1	25574343	25574543	MISNON	FP	1
<i>RHD</i>	CM025462	A	G	1	25502530	97.01	1	25587960	25588160	MISNON	FP	0
<i>RHD</i>	CM074989	A	T	1	25500763	97.51	1	25589727	25589927	MISNON	FP	0
<i>RHD</i>	CM074990	G	A	1	25500750	97.01	1	25589740	25589940	MISNON	FP	0
<i>RHD</i>	CM074991	G	C	1	25500660	97.01	1	25589830	25590030	MISNON	FP	0
<i>RHD</i>	CM074992	G	A	1	25500675	96.52	1	25589815	25590015	MISNON	FP	1
<i>RHD</i>	CM074995	G	C	1	25500696	96.52	1	25589794	25589994	MISNON	FP	0

Supplemental Table 1. Continued

Gene	HGMD_ID	wildBASE	mutBASE	Acceptor Chr.	Mutation Position	% identity	Donor Chr.	Donor Start	Donor end	TYPE	Tag	CpG
<i>RHD</i>	CM076467	A	T	1	25521006	98.51	1	25569478	25569678	MISNON	FP	0
<i>RHD</i>	CM076483	T	C	1	25483809	99.50	1	25607688	25607888	MISNON	FP	0
<i>RHD</i>	CM994712	T	A	1	25500081	96.52	1	25591061	25591261	MISNON	FP	0
<i>RHD</i>	CM994713	A	T	1	25500131	96.02	1	25591011	25591211	MISNON	FP	0
<i>RHD</i>	CM994716	T	G	1	25500630	97.51	1	25589860	25590060	MISNON	FP	0
<i>ROCK1</i>	CM071970	C	T	18	16787610	99.50	18	108330	108530	MISNON	FTV	1
<i>SBDS</i>	CM075001	A	C	7	66095736	96.02	7	71940120	71940316	MISNON	DM	0
<i>SBDS</i>	CM102156	G	T	7	66097713	98.01	7	71938196	71938396	MISNON	DM	0
<i>SBDS</i>	CP035464	TA	ct	7	66096708	98.51	7	71939162	71939363	INDEL	DM	0
<i>SBDS</i>	CX030185	TAA	agttcaagtatc	7	66095805	96.62	7	71940132	71940338	INDEL	DM	0
<i>SFTPA1</i>	CM033024	C	G	10	81041735	99.00	10	80988997	80989197	MISNON	DP	0
<i>SFTPA1</i>	CM033024	C	G	10	81041735	99.00	10	81308942	81309142	MISNON	DP	0
<i>SMG1</i>	CM077638	C	A	16	18777852	98.01	16	68812033	68812233	MISNON	FTV	0
<i>SULT1A1</i>	CM090528	C	T	16	28525827	95.02	16	28512219	28512419	MISNON	DFP	0
<i>TARDBP</i>	CM085730	C	A	1	11005088	92.00	2	238806037	238806235	MISNON	DM	0
<i>TAS1R3</i>	CM098260	C	T	1	1259417	99.50	1	269679	269879	MISNON	DP	1
<i>TIMM8A</i>	CM014112	C	T	X	100488199	98.01	2	162642070	162642270	MISNON	DM	1
<i>TLR2</i>	CM015327	C	T	4	154845538	97.51	4	154820391	154820591	MISNON	DFP	1
<i>TLR2</i>	CM035918	C	A	4	154845401	92.04	4	154820254	154820454	MISNON	DP	0
<i>UBE2NL</i>	CM0910038	T	G	X	142795134	92.50	4	173537850	173538049	MISNON	FTV	0
<i>UGT1A3</i>	CM042130	T	C	2	234302651	96.52	2	234292246	234292446	MISNON	FP	0
<i>UGT1A3</i>	CM042130	T	C	2	234302651	94.03	2	234286417	234286617	MISNON	FP	0
<i>UGT1A3</i>	CM042130	T	C	2	234302651	93.03	2	234320639	234320838	MISNON	FP	0
<i>UGT2B15</i>	CM004865	G	T	4	69570689	99.50	4	69218580	69218780	MISNON	FP	0
<i>VWF</i>	CM074634	G	A	12	5998704	97.51	22	15558481	15558681	MISNON	FP	1
<i>VWF</i>	CM087824	T	G	12	5999159	95.52	22	15558936	15559136	MISNON	DM	0
<i>VWF</i>	CM095110	C	G	12	5998107	95.52	22	15557884	15558084	MISNON	DM	0
<i>VWF</i>	CM910404	T	A	12	5998740	97.51	22	15558517	15558717	MISNON	DM	0
<i>VWF</i>	CM920715	C	T	12	5997870	97.01	22	15557647	15557847	MISNON	DM	1
<i>VWF</i>	CM930727	C	T	12	5999048	98.01	22	15558825	15559025	MISNON	DM	1

Supplemental Table 1. Continued

Gene	HGMD_ID	wildBASE	mutBASE	Acceptor Chr.	Mutation Position	% identity	Donor Chr.	Donor Start	Donor end	TYPE	Tag	CpG
<i>VWF</i>	CM930735	G	A	12	5998020	95.02	22	15557797	15557997	MISNON	DM	1
<i>VWF</i>	HM070121	G	C	12	5998431	96.52	22	15558208	15558408	MISNON	FP	0
<i>ZNF600</i>	CM067728	T	C	19	57962196	95.02	19	57748712	57748912	MISNON	DP	0

wildBASE: base in the wild-type (non-disease) allele. mutBASE: base in the disease allele. TYPE: type of mutation. MISNON: missense or nonsense mutation. REG: regulatory mutation Tag: effect of mutation. DM: Disease-causing mutations. DP: Disease-associated polymorphisms. DFP: Disease-associated polymorphisms with supporting functional evidence. FP: Functional polymorphisms. FTV: Frameshift or Truncating Variant. In the CpG column, “1” and “0” denote that the mutation occurs or does not occur in a CpG site, respectively.

Supplemental Table 2. New high-confidence disease-related IGC events: Gene, disease, and mutation information

Disease/phenotype	Acceptor gene	Donor gene	Type of Mutation	Mutation(s) in disease alleles	HGMD ID	PMID
Pseudoxanthoma elasticum, autosomal recessive	<i>ABCC6</i>	<i>ABCC6P1</i>	Misnon	Q378X 1077A>G 1132C>T 1141T>C	CM012720	11692167
Poor metaboliser, association with	<i>CYP2D6</i>	<i>CYP2D7P1</i>	Misnon	L91M;H94R 271C>A 281A>G	CM994689 CM994690	10471072
Heart malformations	<i>GJA1</i>	<i>GJA1P1</i>	Misnon	R239W;S251T;A253P; P283L;T290N 715C>T;752G>C;757G>C; 848C>T;869C>A	CM055952;CM055950;CM055953; CM055951;CM055949	15978203
			Misnon	R362Q;R376Q 1085G>A;1127G>A	CM014191;CM014192	11470490
Neurofibromatosis type 1?	<i>NF1</i>	NF1P1 or NF1P2	Misnon	R873C; K874Q; S876S; 2828C>T; 2831A>C; 2839T>C	CM087526	19076627
Hyperprolinaemia, association with	<i>PRODH</i>	<i>LOC440792</i>	Misnon	V427M;R431H;L441P; 1279G>A;1292G>A;1322T>C;	CM057558;CM057555;CM022055;	11891283 15662599 15494707
			Misnon	R453C; T466M;A472T 1363C>T;1397C>T;1414G>A	CM025458;CM057358;CM057556	11891283 12217952
Complement C4b deficiency	<i>C4B</i>	<i>C4A</i>	Indel/Misnon	3359_3375 delTCTCTCCAGTGATACAT insCCTGTCCAGTGTAGAC L1120P;S1121C;I1124L;H1125D	CX033310	12626442
mRNA stability, assoc. with	<i>CYP2A6</i>	<i>CYP2A7</i>	Regulatory, 3'UTR	CYP2A6*1B. Described as 1695A>G CYP2A6*1B. Described as 1698C>A	CR060575;CR060576	16378601

Misnon=missense/nonsense. PMID: PubMed ID. Each row represent a disease allele. Two non-overlapping IGC-derived disease alleles have been found in *GJA1* and *PRODH*.

Supplemental Table 3. New high-confidence disease-related IGC events: Gene conversion information

Acceptor gene	Donor gene	Mutation(s) in disease alleles	Acceptor/Donor chromosome(s)	Donor location	Donor type	MaxCT	MinCT
<i>ABCC6</i>	<i>ABCC6P1</i>	Q378X	16p13.11/16p12.3	Linked	Non-processed pseudogene	505	56
<i>CYP2D6</i>	<i>CYP2D7P1</i>	L91M;H94R	22q13.1/22q11.2-qter	Linked	Non-processed pseudogene	673	11
<i>GJA1</i>	<i>GJA1P1</i>	R239W;S251T;A253P; P283L;T290N R362Q;R376Q 1085G>A;1127G>A	6q22-q23/5q21.3	Unlinked	Processed pseudogene	214 164	155 71
<i>NF1</i>	NF1P1 or NF1P2	R873C; K874Q; S876S;	17q11.2/15q11.2 or 22q11.1	Unlinked	Non-processed pseudogene	77	12
<i>PRODH</i>	<i>LOC440792</i>	V427M;R431H;L441P; 1279G>A;1292G>A;1322T>C R453C;T466M;A472T 1363G>T;1397C>T;1414G>A	22q11.2/22q11.21	Linked	Non-processed pseudogene	231 309	44 58
<i>C4B</i>	<i>C4A</i>	3359_3375 delTCTCTCCAGTGATACAT insCCTGTCCAGTGTTAGAC L1120P;S1121C;I1124L;H1125D	6p21.3/6p21.3	Linked	Functional paralog	669	17
<i>CYP2A6</i>	<i>CYP2A7</i>	CYP2A6*1B. Descr. as 1695A>G CYP2A6*1B. Descr. as 1698C>A	19q13.2/19q13.2	Linked	Functional paralog	184	4

MaxCT: maximal conversion tract. MinCT: minimal conversion tract. Linked is defined for the purposes of the current analysis as being located on the same chromosome. Unlinked: on different chromosomes. Each row represent a disease allele. Two non-overlapping IGC-derived disease alleles have been found in *GJA1* and *PRODH*.

Supplemental Table 4. HGMD mutations analyzed and high-confidence (HC) IGC-caused mutations detected

	HGMD data set	HC Mutations	HC Genes
Missense/nonsense	57,255	30	16
• DM	54,411	19	
• DP	1,117	8	
• FP	1,310	3	
• DFP	352	0	
• FTV	65	0	
Regulatory	1,770	2	2
• DM	643	0	
• DP	351	0	
• FP	356	2	
• DFP	420	0	
• FTV	0	0	
Indels	1,463	3	3
• DM	1,435	3	
• DP	4	0	
• FP	20	0	
• DFP	4	0	
• FTV	0	0	
Complex	1045	26	26
• DM	970	25	
• DP	14	0	
• FP	53	1	
• DFP	8	0	
• FTV	0	0	
TOTAL	60,488	61	47*
• DM	56,489	47	
• DP	1,472	8	
• FP	1,686	6	
• DFP	776	0	
• FTV	65	0	

The number of mutations refers to mutations detected during the course of this work and literature surveys (Chen et al. 2007; Chuzhanova et al. 2009). The total number of mutations is higher than the number of IGC events because several events involve different kinds of mutation, which are also reported in the “Complex mutations” category. The total number of genes with IGC-derived mutations is greater than the 30 discovered genes because some mutations are described in the Complex mutations category as well as in other categories. DM: Disease-causing Mutations. DP: Disease-associated Polymorphisms. FP: Functional Polymorphisms. DFP: Disease-associated Polymorphisms with supporting functional evidence. FTV: Frameshift or Truncating Variant.

Supplemental Table 5. Experimental evidence of functional effect in gene conversion events involving two paralogous genes

Acceptor gene	Donor gene	Functional evidence in disease allele	Experiments/assays	PMID
<i>C4B</i>	<i>C4A</i>	Undetectable total hemolytic complement of the patient's serum due to lack of C4B	Hemolytic assay	12626442
<i>CFH</i>	<i>CFHR1</i>	Lower affinity of the recombinant CFH protein to C3b/C3d Defective capacity to control complement activation	C3b/C3d binding assays Hemolytic assays	16470555
<i>CYP11B2</i>	<i>CYP11B1</i>	High levels of urinary 18-hydroxycortisol due to	gas chromatography/mass spectrometry	15062555
<i>CYP2A6</i>	<i>CYP2A7</i>	Low expression of the protein encoded by the <i>CYP2A6*5</i> allele	Protein expression and coumarin 7-hydroxylase activity assays in yeast	10544257
<i>CYP3A7</i>	<i>CYP3A4</i>	Low hepatic mRNA expression of the <i>CYP3A7*1C</i> allele	RT-PCR	11279519
<i>GH1</i>	<i>GH2</i>	Decreased expression level of mutated allele	Luciferase reporter gene assay	12655557
<i>GYPA</i>	<i>GYPE</i>	Protein from the GPSta(TF) allele expressed on erythrocytes	Western blot	10862083
<i>HBA1</i>	<i>HBA2</i>	$\alpha 121$ allele, IVSI,5 G→C β^+ -thalassemia mutation	Mean corpuscular volume assay PCR	16503550
<i>HBD</i>	<i>HBB</i>	Hemoglobin Parchman, $\delta\beta\delta$ allele, protein detected <i>in vivo</i>	Tryptic digestion of hemoglobin Restriction analysis of HBB gene	7123235
<i>HBG1</i>	<i>HBG2</i>	Elevated level of fetal hemoglobin (Hb F) due to high expression of the A γ -globin gene	HPLC measurement of Hb A ₂ and Hb F levels transient protein expression assays	9703422
<i>IGLL1</i>	<i>IGLL3</i>	Defective folding and secretion of the mutant protein	COS7 cells transfection with expression vectors for the mutant $\lambda 5/14.1$	9419212
<i>OPN1LW</i>	<i>OPN1MW</i>	RCP/GCP locus linked to the disorder The C203R mutation in the disease allele is the most common inactivating mutation in blue cone monochromacy	Linkage analysis PCR	8666378
<i>PRSS1</i>	<i>PRSS2</i>	Recombinant cationic trypsinogen double-mutant N29I-N54S showed increased susceptibility for spontaneous activation to trypsin	Trypsin activity <i>in vitro</i>	15776435

PMID: PubMed ID.

Supplemental Table 6. Summary of paralogous sequences with ≥ 200 bp and $\geq 92\%$ sequence identity

	IGC genes	HGMD genes	Non-HGMD genes
Total genes examined	30	3,196	16,181
Genes with paralogous sequences	30	441 (14%)	2,800 (17%)
Total paralogous sequences	55	1,171	9,458
<i>Features of paralogous sequences:</i>			
• functional genes	19 (35%)	454 (39%)	2,825 (31%)
• pseudogenes	36 (65%)	717 (61%)	6,563 (69%)
• linked functional genes	14 (74%)	254 (56%)	2,340 (81%)
• linked pseudogenes	25 (70%)	254 (35%)	2,529 (39%)
• total linked	39 (71%)	508 (43%)	4,869 (52%)

'Linked' is defined for the purposes of the current analysis as being located on the same chromosome. IGC genes: genes involved in IGC events.

Supplemental Table 7. Potential IGC-mediated deleterious mutations

	Amino acid replacements	Genes
HGMD genes – PolyPhen	11,920	1,035
HGMD genes - stop codons	1,122	332
Non-HGMD genes – PolyPhen	43,597	4,475
Non-HGMD genes – stop codons	6,965	2,184
All genes – PolyPhen	55,517	5,510
All genes - stop codons	8,087	2,516
All genes – all deleterious mutations	63,604	5,854

Polyphen: probably damaging mutations identified using PolyPhen-2.

Supplemental Table 8. Potential IGC donor sequences causing deleterious mutations

	HGMD genes	Non-HGMD genes
Total donor sequences	769	3179
Functional genes	141 (18%)	577 (18%)
Pseudogenes	628 (82%)	2602 (82%)
Linked functional genes	124 (88%)	466 (81%)
Linked pseudogenes	318 (51%)	1060 (41%)

Only results for donor sequences sharing $\geq 92\%$ sequence identity over ≥ 200 bp with the gene region harboring the replaced amino acid are shown. 'Linked' is defined for the purposes of the current analysis as being located on the same chromosome.

Supplemental Table 9. Potential IGC-mediated premature stop codons.

Gene Symbol	ProteinID	GeneID	Site	Gene type	Acceptor Chr.	Acceptor Start	Acceptor End	Donor Chr.	Donor Start	Donor End	Distance	Donor Type
ABCC6	NP_001162	NM_001171	Q378X	HGMD	chr16	16203300	16203500	chr16	18511348	18511623	2307848	Gene
ACTB	NP_001092	NM_001101	Y362X	HGMD	chr7	5533846	5534046	chr6	89042323	89042838	na	Gene
ADAM21	NP_003804	NM_003813	R133X	non-HGMD	chr14	69988626	69996375	chr14	69781982	69784380	204246	Gene
ADAMTS7	NP_055087	NM_014272	R995X	non-HGMD	chr15	76838599	76890828	chr15	76064720	76066050	772549	Gene
ADAMTS7	NP_055087	NM_014272	W1415X	non-HGMD	chr15	76838599	76890828	chr15_r	509198	510318	na	Gene
ADAMTS7	NP_055087	NM_014272	W1415X	non-HGMD	chr15	76838599	76890828	chr15_r	612569	613710	na	Gene
ADAMTS7	NP_055087	NM_014272	W1415X	non-HGMD	chr15	76838599	76890828	chr15	80406783	80407924	3515955	Gene
ADAMTS7	NP_055087	NM_014272	W1415X	non-HGMD	chr15	76838599	76890828	chr15	80714110	80715230	3823282	Gene
ADAMTS7	NP_055087	NM_014272	W757X	non-HGMD	chr15	76838599	76890828	chr15_r	515130	515878	na	Gene
ADAMTS7	NP_055087	NM_014272	W757X	non-HGMD	chr15	76838599	76890828	chr15_r	618479	619227	na	Gene
ADAMTS7	NP_055087	NM_014272	W757X	non-HGMD	chr15	76838599	76890828	chr15	80401224	80401972	3510396	Gene
ADAMTS7	NP_055087	NM_014272	W757X	non-HGMD	chr15	76838599	76890828	chr15	80708550	80709298	3817722	Gene
ADAMTS7	NP_055087	NM_014272	W757X	non-HGMD	chr15	76838599	76890828	chr15	83611682	83612430	6720854	Gene
ADAMTS7	NP_055087	NM_014272	W946X	non-HGMD	chr15	76838599	76890828	chr15_r	510928	511655	na	Gene
ADAMTS7	NP_055087	NM_014272	W946X	non-HGMD	chr15	76838599	76890828	chr15_r	614320	615047	na	Gene
ADAMTS7	NP_055087	NM_014272	W946X	non-HGMD	chr15	76838599	76890828	chr15	80405446	80406173	3514618	Gene
ADAMTS7	NP_055087	NM_014272	W946X	non-HGMD	chr15	76838599	76890828	chr15	80712773	80713500	3821945	Gene
ADAMTS7	NP_055087	NM_014272	W946X	non-HGMD	chr15	76838599	76890828	chr15	83607481	83608208	6716653	Gene
AKR7A3	NP_036199	NM_012067	R106X	HGMD	chr1	19485249	19485449	chr1	19469829	19470017	15232	Pseudogene
ALG1	NP_061982	NM_019109	E443X	HGMD	chr16	5074717	5074917	chr7	6936857	6936991	na	Gene
ALG1	NP_061982	NM_019109	E451X	HGMD	chr16	5074741	5074941	chr11	3386486	3386620	na	Gene
ALG1L	NP_001182152	NM_001195223	E158X	non-HGMD	chr3	127130807	127138577	chr7	6936607	6937325	na	Gene
ALG1L	NP_001182152	NM_001195223	E166X	non-HGMD	chr3	127130807	127138577	chr11	3386236	3386954	na	Gene
ALG1L	NP_001182152	NM_001195223	Q180X	non-HGMD	chr3	127130807	127138577	chr16	5074496	5075214	na	Pseudogene
ALG1L	NP_001182152	NM_001195223	Q180X	non-HGMD	chr3	127130807	127138577	chr11	3386236	3386954	na	Gene
ALG1L	NP_001182152	NM_001195223	Q180X	non-HGMD	chr3	127130807	127138577	chr11	67315998	67316716	na	Gene
ALG1L	NP_001182152	NM_001195223	Q180X	non-HGMD	chr3	127130807	127138577	chr7	6936607	6937325	na	Gene
AMZ2	NP_001028744	NM_001033572	G98X	non-HGMD	chr17	63755739	63764900	chr17	60399067	60399776	3355963	Gene
ANKRD20A1	NP_115626	NM_032250	L792X	non-HGMD	chr9	67516580	67559660	chr1	142219593	142220734	na	Gene
ANKRD20A4	NP_001092275	NM_001098805	C515X	non-HGMD	chr9	68671800	68714929	chr1	142051297	142052123	na	Gene

<i>ANKRD20A4</i>	NP_001092275	NM_001098805	L792X	non-HGMD	chr9	68671800	68714929	chr1	142219593	142220734	na	Gene
<i>ANKRD20A4</i>	NP_001092275	NM_001098805	L792X	non-HGMD	chr9	68671800	68714929	chr21_r	819786	820927	na	Gene
<i>ANKRD30B</i>	NP_001138501	NM_001145029	E367X	non-HGMD	chr18	14738238	14842737	chr10	44971981	44972885	na	Gene
<i>ANKRD36</i>	NP_001157787	NM_001164315	Q1835X	non-HGMD	chr2	97142959	97279633	chr2	88883092	88883750	8259209	Gene
<i>ANKRD36</i>	NP_001157787	NM_001164315	Q1836X	non-HGMD	chr2	97142959	97279633	chr13_r	4517	5175	na	Gene
<i>ANKRD36</i>	NP_001157787	NM_001164315	R1838X	non-HGMD	chr2	97142959	97279633	chr2	88883092	88883750	8259209	Gene
<i>ANKRD36B</i>	NP_079466	NM_025190	Q1247X	non-HGMD	chr2	97487692	97572860	chr2	88883092	88883750	8603942	Gene
<i>ANKRD36B</i>	NP_079466	NM_025190	Q1248X	non-HGMD	chr2	97487692	97572860	chr13_r	4517	5175	na	Gene
<i>ANKRD36B</i>	NP_079466	NM_025190	R1250X	non-HGMD	chr2	97487692	97572860	chr2	88883092	88883750	8603942	Gene
<i>ANKRD49</i>	NP_060174	NM_017704	W151X	non-HGMD	chr11	93866800	93872392	chr5	136220966	136221942	na	Gene
<i>ARF6</i>	NP_001654	NM_001663	Y31X	non-HGMD	chr14	49429485	49433522	chr7	99073856	99074574	na	Gene
<i>ARHGAP5</i>	NP_001025226	NM_001030055	E481X	non-HGMD	chr14	31616245	31698685	chr7	54969666	54970603	na	Gene
<i>ARHGAP5</i>	NP_001025226	NM_001030055	R480X	non-HGMD	chr14	31616245	31698685	chr7	54969666	54970603	na	Gene
<i>ARL17</i>	NP_001096624	NM_001103154	L100X	non-HGMD	chr17	41732689	41794876	chr17	34443458	34444071	7288618	Gene
<i>ARSE</i>	NP_000038	NM_000047	W581X	HGMD	chrX	2862799	2862999	chrY	12977873	12978229	na	Gene
<i>ASNS</i>	NP_001664	NM_001673	R63X	non-HGMD	chr7	97319364	97339790	chr2	94921721	94922433	na	Gene
<i>ASNS</i>	NP_001664	NM_001673	Y140X	non-HGMD	chr7	97319364	97339790	chr21_r	1606986	1607728	na	Gene
<i>ASNS</i>	NP_001664	NM_001673	Y140X	non-HGMD	chr7	97319364	97339790	chr21_r	93489	94231	na	Gene
<i>ASNS</i>	NP_001664	NM_001673	Y140X	non-HGMD	chr7	97319364	97339790	chr22_r	182196	182938	na	Gene
<i>ASNS</i>	NP_001664	NM_001673	Y140X	non-HGMD	chr7	97319364	97339790	chr2	94916079	94916821	na	Gene
<i>ASNS</i>	NP_001664	NM_001673	Y140X	non-HGMD	chr7	97319364	97339790	chr4_r	483824	484566	na	Gene
<i>ASNS</i>	NP_001664	NM_001673	Y140X	non-HGMD	chr7	97319364	97339790	chr4_r	710519	711261	na	Gene
<i>ASNS</i>	NP_001664	NM_001673	Y140X	non-HGMD	chr7	97319364	97339790	chrY	11759676	11760418	na	Gene
<i>ASNS</i>	NP_597680	NM_133436	R63X	non-HGMD	chr7	97319364	97339413	chr2	94921721	94922433	na	Gene
<i>ASNS</i>	NP_597680	NM_133436	Y140X	non-HGMD	chr7	97319364	97339413	chr21_r	1606986	1607728	na	Gene
<i>ASNS</i>	NP_597680	NM_133436	Y140X	non-HGMD	chr7	97319364	97339413	chr21_r	93489	94231	na	Gene
<i>ASNS</i>	NP_597680	NM_133436	Y140X	non-HGMD	chr7	97319364	97339413	chr22_r	182196	182938	na	Gene
<i>ASNS</i>	NP_597680	NM_133436	Y140X	non-HGMD	chr7	97319364	97339413	chr2	94916079	94916821	na	Gene
<i>ASNS</i>	NP_597680	NM_133436	Y140X	non-HGMD	chr7	97319364	97339413	chr4_r	483824	484566	na	Gene
<i>ASNS</i>	NP_597680	NM_133436	Y140X	non-HGMD	chr7	97319364	97339413	chr4_r	710519	711261	na	Gene
<i>ASNS</i>	NP_597680	NM_133436	Y140X	non-HGMD	chr7	97319364	97339413	chrY	11759676	11760418	na	Gene
<i>ATMIN</i>	NP_056066	NM_015251	K370X	non-HGMD	chr16	79626958	79638452	chr9	97095217	97096250	na	Gene
<i>ATMIN</i>	NP_056066	NM_015251	R491X	non-HGMD	chr16	79626958	79638452	chr9	97095217	97096250	na	Gene

<i>ATP5H</i>	NP_006347	NM_006356	W153X	non-HGMD	chr17	70546549	70554669	chr9	2890073	2890821	na	Gene
<i>ATP5H</i>	NP_006347	NM_006356	Y152X	non-HGMD	chr17	70546549	70554669	chr9	2890073	2890821	na	Gene
<i>AZGP1</i>	NP_001176	NM_001185	W154X	non-HGMD	chr7	99402285	99411671	chr7	99421078	99421883	9407	Gene
<i>BCAP31</i>	NP_005736	NM_005745	E228X	non-HGMD	chrX	152619140	152643032	chr16	32790519	32791129	na	Gene
<i>BCAP31</i>	NP_005736	NM_005745	E228X	non-HGMD	chrX	152619140	152643032	chr16	33699195	33699805	na	Gene
<i>BCAP31</i>	NP_001132929	NM_001139457	E295X	non-HGMD	chrX	152619140	152642743	chr16	32790519	32791129	na	Gene
<i>BCAP31</i>	NP_001132929	NM_001139457	E295X	non-HGMD	chrX	152619140	152642743	chr16	33699195	33699805	na	Gene
<i>BCLAF1</i>	NP_001070909	NM_001077441	R69X	non-HGMD	chr6	136619693	136652682	chr5	110313122	110314482	na	Gene
<i>BMS1</i>	NP_055568	NM_014753	G1012X	non-HGMD	chr10	42597959	42650391	chr22	20992812	20993434	na	Gene
<i>BMS1</i>	NP_055568	NM_014753	Q100X	non-HGMD	chr10	42597959	42650391	chr10	46179651	46180267	3529260	Gene
<i>BMS1</i>	NP_055568	NM_014753	Q100X	non-HGMD	chr10	42597959	42650391	chr10	48569418	48570034	5919027	Gene
<i>BMS1</i>	NP_055568	NM_014753	Q322X	non-HGMD	chr10	42597959	42650391	chr10	46172022	46172725	3521631	Gene
<i>BMS1</i>	NP_055568	NM_014753	Q322X	non-HGMD	chr10	42597959	42650391	chr10	48561790	48562493	5911399	Gene
<i>BMS1</i>	NP_055568	NM_014753	Q899X	non-HGMD	chr10	42597959	42650391	chr22	20990936	20991900	na	Gene
<i>BMS1</i>	NP_055568	NM_014753	R1076X	non-HGMD	chr10	42597959	42650391	chr15	18625179	18625825	na	Gene
<i>BMS1</i>	NP_055568	NM_014753	R1076X	non-HGMD	chr10	42597959	42650391	chr15	19636431	19637077	na	Gene
<i>BMS1</i>	NP_055568	NM_014753	R1076X	non-HGMD	chr10	42597959	42650391	chr22	20993842	20994497	na	Gene
<i>BMS1</i>	NP_055568	NM_014753	R1076X	non-HGMD	chr10	42597959	42650391	chr9	42801346	42802001	na	Gene
<i>BMS1</i>	NP_055568	NM_014753	R1076X	non-HGMD	chr10	42597959	42650391	chr9	66703085	66703740	na	Gene
<i>BMS1</i>	NP_055568	NM_014753	R1076X	non-HGMD	chr10	42597959	42650391	chr9	67017864	67018519	na	Gene
<i>BMS1</i>	NP_055568	NM_014753	R1076X	non-HGMD	chr10	42597959	42650391	chr9	69008241	69008896	na	Gene
<i>BMS1</i>	NP_055568	NM_014753	R1076X	non-HGMD	chr10	42597959	42650391	chr9	69373690	69374345	na	Gene
<i>BMS1</i>	NP_055568	NM_014753	R1076X	non-HGMD	chr10	42597959	42650391	chr9	69574438	69575093	na	Gene
<i>C11orf2</i>	NP_037397	NM_013265	Y41X	non-HGMD	chr11	64620258	64635756	chr8	6927699	6928507	na	Gene
<i>C14orf109</i>	NP_001092091	NM_001098621	C143X	non-HGMD	chr14	92721127	92723178	chr5	90362313	90363130	na	Gene
<i>C14orf109</i>	NP_056491	NM_015676	C105X	non-HGMD	chr14	92721048	92723184	chr5	90362313	90363130	na	Gene
<i>C15orf43</i>	NP_689661	NM_152448	E137X	non-HGMD	chr15	43036194	43058713	chr15	43003896	43004494	31700	Gene
<i>C18orf25</i>	NP_659492	NM_145055	E12X	non-HGMD	chr18	42007985	42100953	chr2	188799911	188800617	na	Gene
<i>C21orf63</i>	NP_478067	NM_058187	Q61X	non-HGMD	chr21	32706622	32809568	chr4	3976475	3977178	na	Gene
<i>C21orf63</i>	NP_478067	NM_058187	Q76X	non-HGMD	chr21	32706622	32809568	chr4	9343842	9344545	na	Gene
<i>C6orf66</i>	NP_054884	NM_014165	W133X	HGMD	chr6	97445729	97445929	chr15	47235854	47236336	na	Gene
<i>C9orf144B</i>	NP_001135389	NM_001141917	Q827X	non-HGMD	chr9	34713049	34719535	chr9	34822359	34825834	102824	Pseudogene
<i>C9orf86</i>	NP_078994	NM_024718	W520X	non-HGMD	chr9	138822194	138855460	chr18	14993785	14994548	na	Gene

CA5A	NP_001730	NM_001739	R32X	non-HGMD	chr16	86479125	86527613	chr16	30254972	30255612	56223513	Gene
CBWD1	NP_001138828	NM_001145356	R337X	non-HGMD	chr9	111037	169075	chr9	69471571	69472211	69302496	Gene
CBWD2	NP_742000	NM_172003	R356X	non-HGMD	chr2	113911737	113970251	chr9	69471571	69472211	na	Gene
CBWD5	NP_001020087	NM_001024916	R308X	non-HGMD	chr9	69671823	69729991	chr9	69471571	69472211	199612	Gene
CBWD6	NP_001078926	NM_001085457	R356X	non-HGMD	chr9	68494357	68552413	chr9	69471571	69472211	919158	Gene
CD33	NP_001763	NM_001772	R89X	non-HGMD	chr19	56420146	56435086	chr19	56406010	56406905	13241	Gene
CDRT15	NP_001007531	NM_001007530	R24X	non-HGMD	chr17	14079897	14080875	chr17	13868306	13869357	210540	Gene
CDRT15L2	NP_001177719	NM_001190790	E63X	non-HGMD	chr17	20423628	20424816	chr17	15608492	15609255	4814373	Gene
CEACAM1	NP_001171744	NM_001184815	W178X	non-HGMD	chr19	47703297	47724501	chr19	47002741	47003513	699784	Gene
CENPJ	NP_060921	NM_018451	E212X	HGMD	chr13	24382056	24382256	chr13	23989878	23990210	391846	Gene
CEP170	NP_001035863	NM_001042404	S694X	non-HGMD	chr1	241354352	241485331	chr4	119653889	119656218	na	Gene
CFHR4	NP_001188480	NM_001201551	W444X	non-HGMD	chr1	195123766	195154466	chr1	195025138	195025763	98003	Gene
CNTNAP3	NP_387504	NM_033655	C676X	non-HGMD	chr9	39062763	39278300	chr9	65342158	65342876	26063858	Gene
CNTNAP3	NP_387504	NM_033655	R399X	non-HGMD	chr9	39062763	39278300	chr9_r	140006	140769	na	Gene
CNTNAP3	NP_387504	NM_033655	R399X	non-HGMD	chr9	39062763	39278300	chr9	40486812	40487575	1208512	Gene
CNTNAP3B	NP_001188309	NM_001201380	C676X	non-HGMD	chr9	43624880	43862469	chr9	65342158	65342876	21479689	Gene
CNTNAP3B	NP_001188309	NM_001201380	R399X	non-HGMD	chr9	43624880	43862469	chr9_r	140006	140769	na	Gene
CNTNAP3B	NP_001188309	NM_001201380	R399X	non-HGMD	chr9	43624880	43862469	chr9	40486812	40487575	3137305	Gene
COIL	NP_004636	NM_004645	W404X	non-HGMD	chr17	52370559	52393410	chr14	38924106	38925409	na	Gene
COL12A1	NP_004361	NM_004370	R1757X	HGMD	chr6	75903895	75904095	chr2	14056584	14056736	na	Gene
CPD	NP_001295	NM_001304	R872X	non-HGMD	chr17	25730067	25820801	chr1	16187014	16187798	na	Gene
CROCC	NP_055490	NM_014675	R240X	non-HGMD	chr1	17121031	17172061	chr1	16855336	16856003	265028	Gene
CROCC	NP_055490	NM_014675	R240X	non-HGMD	chr1	17121031	17172061	chr1	16949629	16950296	170735	Gene
CROCC	NP_055490	NM_014675	R809X	non-HGMD	chr1	17121031	17172061	chr1	16674636	16675390	445641	Gene
CROCC	NP_055490	NM_014675	R859X	non-HGMD	chr1	17121031	17172061	chr1	16823150	16823805	297226	Gene
CROCC	NP_055490	NM_014675	R997X	non-HGMD	chr1	17121031	17172061	chr1	16820778	16821448	299583	Gene
CSPG4	NP_001888	NM_001897	R491X	non-HGMD	chr15	73753717	73792244	chr15_r	446282	448431	na	Gene
CSPG4	NP_001888	NM_001897	R491X	non-HGMD	chr15	73753717	73792244	chr15_r	672006	674155	na	Gene
CSPG4	NP_001888	NM_001897	R491X	non-HGMD	chr15	73753717	73792244	chr15	80541283	80543432	6749039	Gene
CSPG4	NP_001888	NM_001897	R491X	non-HGMD	chr15	73753717	73792244	chr15	80780553	80782702	6988309	Gene
CSPG4	NP_001888	NM_001897	R491X	non-HGMD	chr15	73753717	73792244	chr15	80927869	80930018	7135625	Gene
CSPG4	NP_001888	NM_001897	R491X	non-HGMD	chr15	73753717	73792244	chr15	83538286	83540723	9746042	Gene
CSPG4	NP_001888	NM_001897	R606X	non-HGMD	chr15	73753717	73792244	chrY	24742667	24744573	na	Pseudogene

CSPG4	NP_001888	NM_001897	R606X	non-HGMD	chr15	73753717	73792244	chrY	26036636	26038542	na	Pseudogene
CYCS	NP_061820	NM_018947	Y98X	HGMD	chr7	25129768	25129968	chr3	10074660	10074971	na	Gene
CYP21A2	NP_000491	NM_000500	Q319X	HGMD	chr6	32116079	32116279	chr6	32083387	32083602	32477	Gene
CYP4Z1	NP_835235	NM_178134	R341X	non-HGMD	chr1	47305746	47356579	chr1	47105955	47106646	199100	Gene
CYP51A1	NP_000777	NM_000786	S18X	non-HGMD	chr7	91579398	91601776	chr3	82938105	82940128	na	Gene
DHFRL1	NP_001182572	NM_001195643	E102X	non-HGMD	chr3	95259455	95264757	chr6	31441896	31442803	na	Gene
DNAJB3	NP_001001394	NM_001001394	K60X	non-HGMD	chr2	234316134	234317400	chr2	234328000	234328904	10600	Gene
DNAJB3	NP_001001394	NM_001001394	Y89X	non-HGMD	chr2	234316134	234317400	chr2	234328000	234328904	10600	Gene
DRD5	NP_000789	NM_000798	W188X	HGMD	chr4	9393215	9393415	chr1	147169370	147170044	na	Gene
DRD5	NP_000789	NM_000798	W188X	HGMD	chr4	9393215	9393415	chr2	91236558	91237232	na	Gene
DRD5	NP_000789	NM_000798	W455X	HGMD	chr4	9394016	9394216	chr1	147170038	147170406	na	Gene
DRD5	NP_000789	NM_000798	W455X	HGMD	chr4	9394016	9394216	chr2	91236196	91236564	na	Gene
DSE	NP_001074445	NM_001080976	Q609X	non-HGMD	chr6	116707975	116866135	chr9	70414789	70416092	na	Gene
DSE	NP_001074445	NM_001080976	R654X	non-HGMD	chr6	116707975	116866135	chr9	70414789	70416092	na	Gene
DSE	NP_001074445	NM_001080976	W98X	non-HGMD	chr6	116707975	116866135	chr9	70413621	70414954	na	Gene
DUB3	NP_958804	NM_201402	C207X	non-HGMD	chr8	12032085	12033678	chr8	12280305	12282394	246627	Gene
DUB3	NP_958804	NM_201402	E249X	non-HGMD	chr8	12032085	12033678	chr8	12276145	12277676	242467	Gene
DUB3	NP_958804	NM_201402	G313X	non-HGMD	chr8	12032085	12033678	chr4	8968957	8971046	na	Gene
DUB3	NP_958804	NM_201402	L237X	non-HGMD	chr8	12032085	12033678	chr8	7862032	7863470	4168615	Gene
DUB3	NP_958804	NM_201402	L267X	non-HGMD	chr8	12032085	12033678	chr8	12280305	12282394	246627	Gene
DUB3	NP_958804	NM_201402	W201X	non-HGMD	chr8	12032085	12033678	chr8	7862032	7863470	4168615	Gene
EFTUD1	NP_078856	NM_024580	R132X	non-HGMD	chr15	80209615	80342159	chr15	82562381	82563021	2220222	Gene
ELL2	NP_036213	NM_012081	K194X	non-HGMD	chr5	95246557	95323531	chr1	156412014	156414394	na	Gene
EMB	NP_940851	NM_198449	R145X	non-HGMD	chr5	49727787	49772991	chr1	120999997	121000589	na	Gene
ENPEP	NP_001968	NM_001977	Y102X	non-HGMD	chr4	111616677	111703942	chr11	50085690	50086687	na	Gene
EPN2	NP_055779	NM_014964	R139X	non-HGMD	chr17	19081282	19180621	chr18	27800665	27801689	na	Gene
ESCO1	NP_443143	NM_052911	G836X	non-HGMD	chr18	17363259	17434691	chr11	49048630	49049378	na	Gene
ESCO1	NP_443143	NM_052911	G836X	non-HGMD	chr18	17363259	17434691	chr11	89137450	89138198	na	Gene
ESCO1	NP_443143	NM_052911	G836X	non-HGMD	chr18	17363259	17434691	chr11	89447411	89448045	na	Gene
EXTL2	NP_001430	NM_001439	W284X	non-HGMD	chr1	101110528	101133006	chr2	176415616	176416523	na	Gene
FAM120A	NP_055427	NM_014612	L1094X	non-HGMD	chr9	95253993	95368218	chr3	43053368	43054194	na	Gene
FAM22A	NP_001092808	NM_001099338	Q795X	non-HGMD	chr10	88975184	88984713	chr10	52114335	52115626	36859558	Gene
FAM22A	NP_001092808	NM_001099338	S83X	non-HGMD	chr10	88975184	88984713	chr10	52106259	52107148	36868036	Gene

FAM58B	NP_001098987	NM_001105517	I67X	non-HGMD	chr1	198449278	198450266	chrY	27181866	27183043	na	Gene
FAM75A3	NP_001076593	NM_001083124	C1337X	non-HGMD	chr9	40690290	40696537	chr9	89725341	89728489	49028804	Pseudogene
FAM75A3	NP_001076593	NM_001083124	C1337X	non-HGMD	chr9	40690290	40696537	chr9	89933943	89937091	49237406	Gene
FAM75A3	NP_001076593	NM_001083124	Q1289X	non-HGMD	chr9	40690290	40696537	chr9	89933943	89937091	49237406	Gene
FAM75A3	NP_001076593	NM_001083124	Q158X	non-HGMD	chr9	40690290	40696537	chr9	94686733	94688630	53990196	Gene
FAM75A6	NP_001138668	NM_001145196	Q158X	non-HGMD	chr9	43564497	43570726	chr9	94686733	94688630	51116007	Gene
FAM75C1	NP_001138596	NM_001145124	R1156X	non-HGMD	chr9	89722696	89728392	chr9	83862168	83863168	5859528	Gene
FAM75C1	NP_001138596	NM_001145124	R170X	non-HGMD	chr9	89722696	89728392	chr9	94686733	94687841	4958341	Gene
FAM75C1	NP_001138596	NM_001145124	R752X	non-HGMD	chr9	89722696	89728392	chr9	83862634	83864474	5858222	Gene
FAM75C2	NP_001159609	NM_001166137	R164X	non-HGMD	chr9	89934039	89939720	chr9	94686733	94687841	4747013	Gene
FAM75C2	NP_001159609	NM_001166137	R746X	non-HGMD	chr9	89934039	89939720	chr9	83862634	83864471	6069568	Gene
FAM75D3	NP_997299	NM_207416	W623X	non-HGMD	chr9	83748234	83754829	chr9	83721392	83722845	25389	Gene
FAM75D4	NP_001138669	NM_001145197	W623X	non-HGMD	chr9	83733162	83739733	chr9	83721392	83722845	10317	Gene
FAM86B1	NP_001077006	NM_001083537	Y283X	non-HGMD	chr8	12077021	12089033	chr4	9313337	9313965	na	Gene
FAM86B2	NP_001131082	NM_001137610	E4X	non-HGMD	chr8	12327494	12338223	chr4	4007723	4008318	na	Gene
FAM86B2	NP_001131082	NM_001137610	Y317X	non-HGMD	chr8	12327494	12338223	chr4	9313337	9313965	na	Gene
FAM86C	NP_689776	NM_152563	G70X	non-HGMD	chr11	71176204	71189928	chr3	127126249	127126871	na	Gene
FAM90A10	NP_001157919	NM_001164447	K103X	non-HGMD	chr8	7663908	7666920	chr8	7117441	7118147	545761	Gene
FAM90A20	NP_001157925	NM_001164453	K103X	non-HGMD	chr8	7139945	7142956	chr8	7117441	7118147	21798	Gene
FAM90A5	NP_001157927	NM_001164455	K103X	non-HGMD	chr8	7132323	7135334	chr8	7117441	7118147	14176	Gene
FAM90A8	NP_001157922	NM_001164450	K103X	non-HGMD	chr8	7633318	7636328	chr8	7117441	7118147	515171	Gene
FAM90A9	NP_001157920	NM_001164448	K103X	non-HGMD	chr8	7656261	7659272	chr8	7117441	7118147	538114	Gene
FANCD2	NP_149075	NM_033084	Q448X	HGMD	chr3	10064566	10064766	chr3	10010836	10010988	53578	Gene
FAT1	NP_005236	NM_005245	Y2529X	non-HGMD	chr4	187745930	187881981	chr20	10816983	10817989	na	Gene
FCGR1A	NP_000557	NM_000566	Q224X	HGMD	chr1	148028246	148028446	chr1	120729852	120730139	27298107	Gene
FCGR1A	NP_000557	NM_000566	Q246X	HGMD	chr1	148028312	148028512	chr1	147643338	147643583	384729	Gene
FCGR2B	NP_003992	NM_004001	Q57X	HGMD	chr1	159907743	159907943	chr1	159825975	159826241	81502	Pseudogene
FCGR3A	NP_000560	NM_000569	R270X	HGMD	chr1	159779388	159779588	chr1	159860872	159861054	81284	Pseudogene
FEM1A	NP_061178	NM_018708	E37X	HGMD	chr19	4742877	4743077	chr13	18137377	18139365	na	Gene
FEM1A	NP_061178	NM_018708	E37X	HGMD	chr19	4742877	4743077	chr18	14383642	14383905	na	Gene
FEM1A	NP_061178	NM_018708	Q343X	HGMD	chr19	4743795	4743995	chr18	14382307	14383164	na	Gene
FEN1	NP_004102	NM_004111	Q333X	HGMD	chr11	61320308	61320508	chr1	91567176	91567661	na	Gene
FGD5	NP_689749	NM_152536	E816X	non-HGMD	chr3	14835472	14951076	chr3	13953591	13954249	881223	Gene

<i>FKBP6</i>	NP_003593	NM_003602	Y61X	HGMD	chr7	72381206	72381406	chr7	72079190	72079291	301915	Gene
<i>FKBP9</i>	NP_009201	NM_007270	Y193X	non-HGMD	chr7	32963529	33013068	chr7	55746253	55746947	22733185	Gene
<i>FLJ46321</i>	NP_001001670	NM_001001670	W626X	non-HGMD	chr9	83793506	83799991	chr9	83721392	83724858	68648	Gene
<i>FOLR3</i>	NP_000795	NM_000804	W60X	non-HGMD	chr11	71524418	71528582	chr11	71565206	71565900	36624	Gene
<i>FOSL1</i>	NP_005429	NM_005438	R169X	non-HGMD	chr11	65416267	65424573	chr4	129295744	129297056	na	Gene
<i>FRG1</i>	NP_004468	NM_004477	W48X	non-HGMD	chr4	191098967	191121353	chr9	67928129	67928769	na	Gene
<i>FSCN1</i>	NP_003079	NM_003088	E54X	non-HGMD	chr7	5598979	5612812	chr15	32573663	32575362	na	Gene
<i>FTL</i>	NP_000137	NM_000146	Y165X	HGMD	chr19	54161671	54161871	chrX	30558336	30558860	na	Gene
<i>GAPDH</i>	NP_002037	NM_002046	Y276X	non-HGMD	chr12	6513917	6517797	chrX	46183838	46185162	na	Gene
<i>GBP1</i>	NP_002044	NM_002053	Y47X	non-HGMD	chr1	89290574	89303631	chr1	89648216	89648907	344585	Gene
<i>GGT1</i>	NP_038347	NM_013430	W25X	non-HGMD	chr22	23309717	23354972	chr2	91327447	91328111	na	Gene
<i>GH2</i>	NP_072051	NM_022557	W214X	non-HGMD	chr17	59311303	59312955	chr17	59348290	59349233	35335	Pseudogene
<i>GH2</i>	NP_072052	NM_022558	G159X	non-HGMD	chr17	59311305	59312955	chr17	59340537	59341249	27582	Pseudogene
<i>GH2</i>	NP_072052	NM_022558	W210X	non-HGMD	chr17	59311305	59312955	chr17	59302880	59303664	7641	Pseudogene
<i>GPAA1</i>	NP_003792	NM_003801	W611X	non-HGMD	chr8	145209511	145213107	chr2	110099346	110100103	na	Gene
<i>GPAA1</i>	NP_003792	NM_003801	W611X	non-HGMD	chr8	145209511	145213107	chr2	110578378	110579135	na	Gene
<i>GPX1</i>	NP_000572	NM_000581	U49X	HGMD	chr3	49370468	49370668	chrX	13306781	13307347	na	Gene
<i>GPX1</i>	NP_000572	NM_000581	W162X	HGMD	chr3	49369850	49370050	chrX	13306781	13307347	na	Gene
<i>GTF2I</i>	NP_001157108	NM_001163636	Q369X	non-HGMD	chr7	73709965	73812958	chr7	72230812	72231497	1478468	Gene
<i>GTF2I</i>	NP_001157108	NM_001163636	Q369X	non-HGMD	chr7	73709965	73812958	chr7	74266810	74267495	453852	Gene
<i>HADHA</i>	NP_000173	NM_000182	Q748X	HGMD	chr2	26267570	26267770	chr12	8680730	8681473	na	Gene
<i>HBZ</i>	NP_005323	NM_005332	E7X	non-HGMD	chr16	142853	144504	chr16	152871	153466	8367	Gene
<i>HDHD1A</i>	NP_036212	NM_012080	K53X	non-HGMD	chrX	6976960	7076231	chr1	150363454	150364463	na	Gene
<i>HERC2</i>	NP_004658	NM_004667	E57X	non-HGMD	chr15	26029782	26240890	chr15	18948421	18949037	7080745	Gene
<i>HERC2</i>	NP_004658	NM_004667	E57X	non-HGMD	chr15	26029782	26240890	chr15	20907298	20907914	5121868	Gene
<i>HERC2</i>	NP_004658	NM_004667	E57X	non-HGMD	chr15	26029782	26240890	chr15	26655993	26656609	415103	Gene
<i>HERC2</i>	NP_004658	NM_004667	K1656X	non-HGMD	chr15	26029782	26240890	chr16	32085595	32086271	na	Gene
<i>HERC2</i>	NP_004658	NM_004667	K1656X	non-HGMD	chr15	26029782	26240890	chr16	32678997	32679673	na	Gene
<i>HERC2</i>	NP_004658	NM_004667	K1656X	non-HGMD	chr15	26029782	26240890	chr16	33028379	33029055	na	Gene
<i>HERC2</i>	NP_004658	NM_004667	W3936X	non-HGMD	chr15	26029782	26240890	chr15	28916855	28917438	2675965	Gene
<i>HIST2H2BF</i>	NP_001154806	NM_001161334	Y43X	non-HGMD	chr1	148020868	148050552	chr1	120707316	120708193	27312675	Gene
<i>HIST2H2BF</i>	NP_001154806	NM_001161334	Y43X	non-HGMD	chr1	148020868	148050552	chr1	147665176	147666053	354815	Gene
<i>HNRNPA1</i>	NP_112420	NM_031157	Q127X	non-HGMD	chr12	52960754	52965297	chr2	33714823	33715925	na	Gene

<i>HNRNPA1</i>	NP_112420	NM_031157	Q127X	non-HGMD	chr12	52960754	52965297	chr4	79128161	79128939	na	Gene
<i>HNRNPA1</i>	NP_112420	NM_031157	Q127X	non-HGMD	chr12	52960754	52965297	chr6	32401091	32401764	na	Gene
<i>HNRNPA1</i>	NP_112420	NM_031157	Q127X	non-HGMD	chr12	52960754	52965297	chrX	73609669	73610663	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	K105X	non-HGMD	chr13	52089605	52115920	chr2	194760098	194760972	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	L21X	non-HGMD	chr13	52089605	52115920	chr3	42905607	42906433	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	P182X	non-HGMD	chr13	52089605	52115920	chr2	206992354	206993345	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	Q127X	non-HGMD	chr13	52089605	52115920	chr6	32401091	32401764	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	Q127X	non-HGMD	chr13	52089605	52115920	chrX	73609669	73610663	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	Q300X	non-HGMD	chr13	52089605	52115920	chr1	58286188	58286870	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	Q300X	non-HGMD	chr13	52089605	52115920	chr6	32401495	32402180	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	Q300X	non-HGMD	chr13	52089605	52115920	chr6	5554212	5554897	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	Q302X	non-HGMD	chr13	52089605	52115920	chr2	201635807	201637266	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	Q36X	non-HGMD	chr13	52089605	52115920	chrX	99997344	99998275	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	R140X	non-HGMD	chr13	52089605	52115920	chr4	75023312	75024144	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	R196X	non-HGMD	chr13	52089605	52115920	chr1	189381526	189382964	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	Y305X	non-HGMD	chr13	52089605	52115920	chr6	5554212	5554897	na	Gene
<i>HNRNPA1L2</i>	NP_001011724	NM_001011724	Y314X	non-HGMD	chr13	52089605	52115920	chr6	5554212	5554897	na	Gene
<i>HSD3B1</i>	NP_000853	NM_000862	W356X	HGMD	chr1	119858637	119858837	chr1	119789423	119790232	68405	Gene
<i>ID2</i>	NP_002157	NM_002166	Y37X	non-HGMD	chr2	8739563	8742034	chr3	62084671	62085572	na	Gene
<i>IGF2BP2</i>	NP_006539	NM_006548	Q188X	non-HGMD	chr3	186844220	187025521	chr1	144989283	144990238	na	Gene
<i>IGSF3</i>	NP_001533	NM_001542	E806X	non-HGMD	chr1	116918553	117011837	chr13_r	107823	108733	na	Gene
<i>IGSF3</i>	NP_001533	NM_001542	E806X	non-HGMD	chr1	116918553	117011837	chr2	91318531	91319441	na	Gene
<i>INTS4</i>	NP_291025	NM_033547	Q254X	non-HGMD	chr11	77267413	77383365	chr7	64247813	64248417	na	Gene
<i>INTS4</i>	NP_291025	NM_033547	Q254X	non-HGMD	chr11	77267413	77383365	chr7	64647716	64648320	na	Gene
<i>INTS4</i>	NP_291025	NM_033547	Q254X	non-HGMD	chr11	77267413	77383365	chr7	64759031	64759635	na	Gene
<i>INTS4</i>	NP_291025	NM_033547	R742X	non-HGMD	chr11	77267413	77383365	chr7	64810564	64811243	na	Gene
<i>INTS4</i>	NP_291025	NM_033547	Y711X	non-HGMD	chr11	77267413	77383365	chr7	64300070	64300749	na	Gene
<i>INTS4</i>	NP_291025	NM_033547	Y711X	non-HGMD	chr11	77267413	77383365	chr7	64810564	64811243	na	Gene
<i>IPMK</i>	NP_689416	NM_152230	S309X	non-HGMD	chr10	59621283	59697700	chr13	22309177	22310144	na	Gene
<i>KIR2DL3</i>	NP_056952	NM_015868	Q305X	non-HGMD	chr19	59941785	59956316	chr19_r	254875	255524	na	Pseudogene
<i>KIR3DL2</i>	NP_006728	NM_006737	R437X	non-HGMD	chr19	60053709	60070474	chr19	59955378	59956087	97622	Pseudogene
<i>KIR3DL2</i>	NP_006728	NM_006737	R445X	non-HGMD	chr19	60053709	60070474	chr19_r	178198	178907	na	Pseudogene
<i>KIR3DL2</i>	NP_006728	NM_006737	R445X	non-HGMD	chr19	60053709	60070474	chr19	59955378	59956087	97622	Pseudogene

<i>KIR3DL2</i>	NP_006728	NM_006737	R445X	non-HGMD	chr19	60053709	60070474	chr19	59986648	59987357	66352	Pseudogene
<i>KIR3DL2</i>	NP_006728	NM_006737	R445X	non-HGMD	chr19	60053709	60070474	chr19	60033113	60033822	19887	Pseudogene
<i>KIR3DL2</i>	NP_006728	NM_006737	R445X	non-HGMD	chr19	60053709	60070474	chr19	59970206	59970915	82794	Gene
<i>KIR3DL3</i>	NP_703144	NM_153443	Q383X	non-HGMD	chr19	59927795	59939815	chr19_r	254875	255500	na	Pseudogene
<i>KIR3DL3</i>	NP_703144	NM_153443	Q383X	non-HGMD	chr19	59927795	59939815	chr19_r	271335	271960	na	Pseudogene
<i>KIR3DL3</i>	NP_703144	NM_153443	Q383X	non-HGMD	chr19	59927795	59939815	chr19_r	287515	288140	na	Pseudogene
<i>KIR3DL3</i>	NP_703144	NM_153443	Q383X	non-HGMD	chr19	59927795	59939815	chr19	60050916	60051541	111101	Pseudogene
<i>KRT18</i>	NP_000215	NM_000224	Q357X	HGMD	chr12	51632192	51632392	chr16	71320730	71321299	na	Gene
<i>KRT18</i>	NP_000215	NM_000224	Q357X	HGMD	chr12	51632192	51632392	chr6	29044932	29045471	na	Gene
<i>KRT8</i>	NP_002264	NM_002273	R40X	HGMD	chr12	51584812	51585012	chr8	62653333	62654781	na	Gene
<i>KRT8</i>	NP_002264	NM_002273	Y267X	HGMD	chr12	51579905	51580105	chr3	171695924	171697114	na	Gene
<i>KRTAP9-2</i>	NP_114167	NM_031961	C86X	non-HGMD	chr17	36636425	36637430	chr17	36680839	36681860	43409	Gene
<i>LOC100130932</i>	NP_001140165	NM_001146693	Q54X	non-HGMD	chr19	14460757	14461690	chr18	50187011	50187732	na	Gene
<i>LOC100130932</i>	NP_001140165	NM_001146693	R63X	non-HGMD	chr19	14460757	14461690	chr1	203586748	203587469	na	Gene
<i>LOC100132396</i>	NP_001180559	NM_001193630	R244X	non-HGMD	chr8	7821268	7847345	chr4	8994594	8995327	na	Gene
<i>LOC646508</i>	NP_001182116	NM_001195187	S187X	non-HGMD	chr19	58476622	58480012	chr19	58493893	58495325	13881	Gene
<i>LOC650293</i>	NP_001035160	NM_001040071	K137X	non-HGMD	chr4	9094611	9095260	chr11	67498248	67499392	na	Gene
<i>LOC650293</i>	NP_001035160	NM_001040071	L131X	non-HGMD	chr4	9094611	9095260	chr10	15089834	15090981	na	Gene
<i>LOC650293</i>	NP_001035160	NM_001040071	Q132X	non-HGMD	chr4	9094611	9095260	chr8	11928501	11929648	na	Gene
<i>LOC650293</i>	NP_001035160	NM_001040071	S42X	non-HGMD	chr4	9094611	9095260	chr4	9069964	9071102	23509	Gene
<i>LOC650293</i>	NP_001035160	NM_001040071	W128X	non-HGMD	chr4	9094611	9095260	chr11	3577416	3578563	na	Gene
<i>LOC650293</i>	NP_001035160	NM_001040071	W128X	non-HGMD	chr4	9094611	9095260	chr13	67374238	67375385	na	Gene
<i>LOC650293</i>	NP_001035160	NM_001040071	W128X	non-HGMD	chr4	9094611	9095260	chr4	9123528	9124573	28268	Gene
<i>LOC650293</i>	NP_001035160	NM_001040071	W128X	non-HGMD	chr4	9094611	9095260	chr7	97414197	97415251	na	Gene
<i>LOC650293</i>	NP_001035160	NM_001040071	W128X	non-HGMD	chr4	9094611	9095260	chr9	92034106	92035253	na	Gene
<i>LOC650293</i>	NP_001035160	NM_001040071	Y27X	non-HGMD	chr4	9094611	9095260	chr8	11928501	11929648	na	Gene
<i>LOC650293</i>	NP_001035160	NM_001040071	Y87X	non-HGMD	chr4	9094611	9095260	chr9	92034106	92035253	na	Gene
<i>LPGAT1</i>	NP_055688	NM_014873	R192X	non-HGMD	chr1	209983421	210070737	chr8	30960638	30961626	na	Gene
<i>LYST</i>	NP_000072	NM_000081	Q2694X	HGMD	chr1	233973870	233974070	chr13	31571759	31571950	na	Gene
<i>MAPK6</i>	NP_002739	NM_002748	Q512X	non-HGMD	chr15	50098702	50145754	chr8	48003525	48004387	na	Gene
<i>MAPK6</i>	NP_002739	NM_002748	Q572X	non-HGMD	chr15	50098702	50145754	chr8	110551292	110552577	na	Gene
<i>MAPK6</i>	NP_002739	NM_002748	R399X	non-HGMD	chr15	50098702	50145754	chr10	50649640	50650520	na	Gene
<i>MATR3</i>	NP_954659	NM_199189	L55X	HGMD	chr5	138671068	138671268	chr1	106236008	106236436	na	Gene

<i>MATR3</i>	NP_954659	NM_199189	R167X	HGMD	chr5	138671404	138671604	chr1	106236008	106236436	na	Gene
<i>MCART1</i>	NP_219480	NM_033412	P87X	non-HGMD	chr9	37877593	37894350	chr18	7124780	7126065	na	Gene
<i>MCART1</i>	NP_219480	NM_033412	R174X	non-HGMD	chr9	37877593	37894350	chr8	81669351	81670741	na	Gene
<i>MCART1</i>	NP_219480	NM_033412	R194X	non-HGMD	chr9	37877593	37894350	chr6	66554781	66556099	na	Gene
<i>MCART1</i>	NP_219480	NM_033412	R57X	non-HGMD	chr9	37877593	37894350	chr8	81669351	81670741	na	Gene
<i>MCART1</i>	NP_219480	NM_033412	W283X	non-HGMD	chr9	37877593	37894350	chr6	66555463	66556148	na	Gene
<i>MCART2</i>	NP_001029344	NM_001034172	P87X	non-HGMD	chr18	27593656	27594841	chr18	7124780	7126062	20467594	Gene
<i>MCART2</i>	NP_001029344	NM_001034172	R174X	non-HGMD	chr18	27593656	27594841	chr8	81669351	81670741	na	Gene
<i>MCART2</i>	NP_001029344	NM_001034172	R194X	non-HGMD	chr18	27593656	27594841	chr6	66554781	66556099	na	Gene
<i>MCART2</i>	NP_001029344	NM_001034172	R57X	non-HGMD	chr18	27593656	27594841	chr8	81669351	81670741	na	Gene
<i>MEP1A</i>	NP_005579	NM_005588	R650X	non-HGMD	chr6	46869052	46915478	chr9	42326227	42327032	na	Gene
<i>MEP1A</i>	NP_005579	NM_005588	R650X	non-HGMD	chr6	46869052	46915478	chr9	43154796	43155601	na	Gene
<i>MEP1A</i>	NP_005579	NM_005588	R650X	non-HGMD	chr6	46869052	46915478	chr9	44089807	44090612	na	Gene
<i>MEP1A</i>	NP_005579	NM_005588	R650X	non-HGMD	chr6	46869052	46915478	chr9	69971258	69972063	na	Gene
<i>MEP1A</i>	NP_005579	NM_005588	Y403X	non-HGMD	chr6	46869052	46915478	chr9	42328217	42329184	na	Gene
<i>MEP1A</i>	NP_005579	NM_005588	Y403X	non-HGMD	chr6	46869052	46915478	chr9	69973248	69974215	na	Gene
<i>MKRN1</i>	NP_038474	NM_013446	E296X	non-HGMD	chr7	139799308	139825838	chrX	73297389	73299136	na	Gene
<i>MKRN1</i>	NP_038474	NM_013446	W467X	non-HGMD	chr7	139799308	139825838	chr2	153415164	153415834	na	Gene
<i>MLL3</i>	NP_733751	NM_170606	R1092X	non-HGMD	chr7	151462942	151764023	chr1	147154436	147155103	na	Gene
<i>MLL3</i>	NP_733751	NM_170606	R1092X	non-HGMD	chr7	151462942	151764023	chr13_r	173435	174102	na	Gene
<i>MLL3</i>	NP_733751	NM_170606	R1092X	non-HGMD	chr7	151462942	151764023	chr21	10036555	10037222	na	Gene
<i>MLL3</i>	NP_733751	NM_170606	R1092X	non-HGMD	chr7	151462942	151764023	chr2	91251443	91252110	na	Gene
<i>MR1</i>	NP_001181929	NM_001195000	W151X	non-HGMD	chr1	179269183	179297697	chr1	179201147	179201955	67228	Gene
<i>MRPL32</i>	NP_114109	NM_031903	R181X	non-HGMD	chr7	42938463	42943978	chrX	53851456	53852093	na	Gene
<i>MTMR12</i>	NP_001035536	NM_001040446	W725X	non-HGMD	chr5	32262867	32348871	chr11	100584605	100585638	na	Gene
<i>MXRA5</i>	NP_056234	NM_015419	E2055X	non-HGMD	chrX	3236607	3274684	chrY	12609361	12610610	na	Gene
<i>MYO5B</i>	NP_001073936	NM_001080467	E1655X	HGMD	chr18	45617957	45618157	chr9	67847098	67848351	na	Gene
<i>NANOG</i>	NP_079141	NM_024865	Q214X	non-HGMD	chr12	7833261	7839922	chr7	29218739	29219520	na	Gene
<i>NAT8</i>	NP_003951	NM_003960	Q168X	non-HGMD	chr2	73721357	73723045	chr2	73781010	73782190	57965	Pseudogene
<i>NAT8</i>	NP_003951	NM_003960	W16X	non-HGMD	chr2	73721357	73723045	chr2	73781010	73782190	57965	Pseudogene
<i>NBEA</i>	NP_056493	NM_015678	R145X	non-HGMD	chr13	34414423	35144873	chr14	18788076	18788812	na	Gene
<i>NBEA</i>	NP_056493	NM_015678	R145X	non-HGMD	chr13	34414423	35144873	chr14	18926584	18927320	na	Gene
<i>NBEA</i>	NP_056493	NM_015678	R145X	non-HGMD	chr13	34414423	35144873	chr2	132196210	132196946	na	Gene

<i>NBEA</i>	NP_056493	NM_015678	R145X	non-HGMD	chr13	34414423	35144873	chr22	14504487	14505223	na	Gene
<i>NBPF1</i>	NP_060410	NM_017940	Q140X	non-HGMD	chr1	16762998	16812569	chr1	147369325	147370043	130556756	Gene
<i>NBPF1</i>	NP_060410	NM_017940	W791X	non-HGMD	chr1	16762998	16812569	chr1	147367544	147368250	130554975	Gene
<i>NBPF1</i>	NP_060410	NM_017940	W8X	non-HGMD	chr1	16762998	16812569	chr1	147372302	147372945	130559733	Gene
<i>NBPF10</i>	NP_001034792	NM_001039703	L279X	non-HGMD	chr1	144004727	144080041	chr1	147372302	147372984	3292261	Gene
<i>NBPF10</i>	NP_001034792	NM_001039703	W8X	non-HGMD	chr1	144004727	144080041	chr1	147372302	147372945	3292261	Gene
<i>NBPF14</i>	NP_056198	NM_015383	Q71X	non-HGMD	chr1	146470265	146492472	chr1	147369262	147370043	876790	Gene
<i>NBPF14</i>	NP_056198	NM_015383	W180X	non-HGMD	chr1	146470265	146492472	chr1	147367544	147368250	875072	Gene
<i>NBPF15</i>	NP_001164226	NM_001170755	L8X	non-HGMD	chr1	146824811	146862891	chr1	147372302	147372945	509411	Gene
<i>NBPF15</i>	NP_001164226	NM_001170755	W249X	non-HGMD	chr1	146824811	146862891	chr1	147367544	147368250	504653	Gene
<i>NBPF20</i>	NP_001032764	NM_001037675	Q140X	non-HGMD	chr1	143326315	143540167	chr1	147369325	147370043	3829158	Gene
<i>NBPF3</i>	NP_115640	NM_032264	Q196X	non-HGMD	chr1	21639217	21683980	chr1	147369268	147370043	125685288	Gene
<i>NBPF3</i>	NP_115640	NM_032264	R36X	non-HGMD	chr1	21639217	21683980	chr1	16807341	16807972	4831245	Gene
<i>NBPF3</i>	NP_115640	NM_032264	W346X	non-HGMD	chr1	21639217	21683980	chr1	145053271	145053935	123369291	Gene
<i>NBPF3</i>	NP_115640	NM_032264	W64X	non-HGMD	chr1	21639217	21683980	chr1	147372299	147372981	125688319	Gene
<i>NBPF4</i>	NP_001137461	NM_001143989	E622X	non-HGMD	chr1	108567485	108588226	chr5	85624437	85625161	na	Pseudogene
<i>NBPF6</i>	NP_001137460	NM_001143988	E622X	non-HGMD	chr1	108794426	108814783	chr5	85624437	85625161	na	Pseudogene
<i>NCOR1</i>	NP_006302	NM_006311	Q17X	non-HGMD	chr17	15874132	16059599	chr7	57663192	57663799	na	Gene
<i>NDUFAF1</i>	NP_057097	NM_016013	W125X	HGMD	chr15	39476074	39476274	chr19	61545809	61546600	na	Gene
<i>NDUFAF1</i>	NP_057097	NM_016013	W136X	HGMD	chr15	39476041	39476241	chr19	61545809	61546600	na	Gene
<i>NF1</i>	NP_000258	NM_000267	K613X	HGMD	chr17	26574605	26574805	chr2	131671395	131671538	na	Gene
<i>NF1</i>	NP_000258	NM_000267	Q589X	HGMD	chr17	26574533	26574733	chr14	18564801	18564914	na	Gene
<i>NF1</i>	NP_000258	NM_000267	Q589X	HGMD	chr17	26574533	26574733	chr14	19148545	19148658	na	Gene
<i>NF1</i>	NP_000258	NM_000267	Q589X	HGMD	chr17	26574533	26574733	chr22	14726426	14726539	na	Gene
<i>NF1</i>	NP_000258	NM_000267	R304X	HGMD	chr17	26551489	26551689	chr18	14147168	14147341	na	Gene
<i>NF1</i>	NP_000258	NM_000267	S1546X	HGMD	chr17	26612878	26613078	chr15	19390394	19390504	na	Gene
<i>NGRN</i>	NP_001028260	NM_001033088	H152X	non-HGMD	chr15	88609898	88616447	chr6	72317425	72318344	na	Gene
<i>NHEDC1</i>	NP_001094344	NM_001100874	R305X	non-HGMD	chr4	104025641	104160345	chr10	38984114	38984724	na	Gene
<i>NOX4</i>	NP_001137309	NM_001143837	S416X	non-HGMD	chr11	88697169	88962427	chr11	49341932	49342554	39354615	Gene
<i>NOX4</i>	NP_058627	NM_016931	S440X	non-HGMD	chr11	88697169	88864301	chr11	49341932	49342554	39354615	Gene
<i>NRBF2</i>	NP_110386	NM_030759	Q171X	non-HGMD	chr10	64563012	64584792	chr3	46039542	46040899	na	Gene
<i>NRBF2</i>	NP_110386	NM_030759	Q87X	non-HGMD	chr10	64563012	64584792	chr3	46039542	46040899	na	Gene
<i>OR10G2</i>	NP_001005466	NM_001005466	R85X	non-HGMD	chr14	21171905	21172838	chr14	21139637	21141066	30839	Gene

OR1D5	NP_055381	NM_014566	R172X	non-HGMD	chr17	2912712	2913651	chr17	3090470	3091905	176819	Gene
OR2M3	NP_001004689	NM_001004689	S36X	non-HGMD	chr1	246432992	246433931	chr1	246351607	246353042	79950	Gene
OR2M5	NP_001004690	NM_001004690	S36X	non-HGMD	chr1	246375072	246376011	chr1	246351607	246353042	22030	Gene
OR2M7	NP_001004691	NM_001004691	S36X	non-HGMD	chr1	246553554	246554493	chr1	246351607	246353042	200512	Gene
OR4A47	NP_001005512	NM_001005512	Q232X	non-HGMD	chr11	48466920	48467850	chr11	49898289	49899058	1430439	Gene
OR4A5	NP_001005272	NM_001005272	S289X	non-HGMD	chr11	51267953	51269024	chr11	48588611	48589353	2678600	Gene
OR4C13	NP_001001955	NM_001001955	Y58X	non-HGMD	chr11	49930518	49931547	chr11	51339693	51341077	1408146	Gene
OR4D10	NP_001004705	NM_001004705	E231X	non-HGMD	chr11	59001478	59002414	chr11	59015451	59016877	13037	Gene
OR4N2	NP_001004723	NM_001004723	Q24X	non-HGMD	chr14	19365447	19366371	chr15	19914576	19915990	na	Gene
OR5AK2	NP_001005323	NM_001005323	Q20X	non-HGMD	chr11	56512964	56513894	chr11	56541892	56543171	27998	Gene
OR5H6	NP_001005479	NM_001005479	Y51X	HGMD	chr3	99465871	99466071	chr3	99439838	99440812	25059	Gene
OR5M8	NP_001005282	NM_001005282	C126X	non-HGMD	chr11	56014486	56015422	chr11	56024221	56025572	8799	Gene
OR5M8	NP_001005282	NM_001005282	R121X	non-HGMD	chr11	56014486	56015422	chr11	56024221	56025572	8799	Gene
OR7D4	NP_001005191	NM_001005191	L104X	non-HGMD	chr19	9185525	9186547	chr19	9206570	9208002	20023	Gene
OR7D4	NP_001005191	NM_001005191	Y60X	non-HGMD	chr19	9185525	9186547	chr19	9206570	9208002	20023	Gene
OTUD4	NP_001096123	NM_001102653	E693X	non-HGMD	chr4	146274251	146320282	chr12	3818674	3822191	na	Gene
PAICS	NP_006443	NM_006452	R403X	non-HGMD	chr4	56996671	57022291	chr8	4633100	4633764	na	Gene
PARG	NP_003622	NM_003631	R645X	non-HGMD	chr10	50696330	51041337	chr10	45725752	45726413	4969917	Gene
PARG	NP_003622	NM_003631	R645X	non-HGMD	chr10	50696330	51041337	chr10	51354873	51355534	313536	Gene
PARP4	NP_006428	NM_006437	Q1059X	non-HGMD	chr13	23893068	23984948	chrY	27183556	27184232	na	Gene
PDCD6IP	NP_001155901	NM_001162429	R451X	non-HGMD	chr3	33815069	33886199	chr15	20693652	20694328	na	Gene
PDCD6IP	NP_001155901	NM_001162429	R451X	non-HGMD	chr3	33815069	33886199	chr15	26848666	26849342	na	Gene
PDE4DIP	NP_001185761	NM_001198832	R1311X	non-HGMD	chr1	143562780	143751349	chr1	16749532	16750562	126812218	Gene
PDE4DIP	NP_001185761	NM_001198832	R1311X	non-HGMD	chr1	143562780	143751349	chr1	16880598	16881577	126681203	Gene
PDE4DIP	NP_001185761	NM_001198832	R1311X	non-HGMD	chr1	143562780	143751349	chr1	17055543	17056573	126506207	Gene
PDE4DIP	NP_001185761	NM_001198832	R1806X	non-HGMD	chr1	143562780	143751349	chr1	147314167	147314804	3562818	Gene
PDE4DIP	NP_001185761	NM_001198832	R1946X	non-HGMD	chr1	143562780	143751349	chr1	144727883	144728544	976534	Gene
PDE4DIP	NP_001185761	NM_001198832	R1946X	non-HGMD	chr1	143562780	143751349	chr1	146024930	146025591	2273581	Gene
PDE4DIP	NP_001185761	NM_001198832	R1963X	non-HGMD	chr1	143562780	143751349	chr1	147326330	147327075	3574981	Gene
PDE4DIP	NP_001185761	NM_001198832	S1277X	non-HGMD	chr1	143562780	143751349	chr1	16749532	16750562	126812218	Gene
PDE4DIP	NP_001185761	NM_001198832	S1277X	non-HGMD	chr1	143562780	143751349	chr1	16880598	16881577	126681203	Gene
PDE4DIP	NP_001185761	NM_001198832	S1277X	non-HGMD	chr1	143562780	143751349	chr1	17055543	17056573	126506207	Gene
PDPR	NP_060460	NM_017990	Q376X	non-HGMD	chr16	68705029	68752685	chr16	72935927	72936621	4183242	Gene

<i>PEX12</i>	NP_000277	NM_000286	R154X	HGMD	chr17	30928287	30928487	chr13	89442487	89443059	na	Gene
<i>PKD1</i>	NP_000287	NM_000296	E2810X	HGMD	chr16	2093528	2093728	chr16	15132443	15133066	13038715	Pseudogene
<i>PKD1</i>	NP_000287	NM_000296	E2810X	HGMD	chr16	2093528	2093728	chr16	14930608	14931084	12836880	Gene
<i>PKD1</i>	NP_000287	NM_000296	Q1483X	HGMD	chr16	2100619	2100819	chr16	16365109	16366608	14264290	Gene
<i>PKD1</i>	NP_000287	NM_000296	Q554X	HGMD	chr16	2106490	2106690	chr16	14921602	14921760	12814912	Gene
<i>PKD1</i>	NP_000287	NM_000296	Q554X	HGMD	chr16	2106490	2106690	chr16	16320253	16320411	14213563	Gene
<i>PKD1</i>	NP_000287	NM_000296	Q554X	HGMD	chr16	2106490	2106690	chr16	18350972	18351130	16244282	Gene
<i>PKD1</i>	NP_000287	NM_000296	R2430X	HGMD	chr16	2096498	2096698	chr16	15135151	15135432	13038453	Gene
<i>PKP4</i>	NP_001005476	NM_001005476	R1104X	non-HGMD	chr2	159021721	159246186	chr2	209753574	209754181	50507388	Gene
<i>PLA2G10</i>	NP_003552	NM_003561	C90X	HGMD	chr16	14689507	14689707	chr16	21319902	21320024	6630195	Gene
<i>PLA2G10</i>	NP_003552	NM_003561	C90X	HGMD	chr16	14689507	14689707	chr16	21752330	21752452	7062623	Gene
<i>PLA2G10</i>	NP_003552	NM_003561	C90X	HGMD	chr16	14689507	14689707	chr16	22456292	22456414	7766585	Gene
<i>PLA2G10</i>	NP_003552	NM_003561	R57X	HGMD	chr16	14691829	14692029	chr16	15360336	15360506	668307	Gene
<i>PLA2G10</i>	NP_003552	NM_003561	R57X	HGMD	chr16	14691829	14692029	chr16	16399783	16399953	1707754	Gene
<i>PLA2G10</i>	NP_003552	NM_003561	R57X	HGMD	chr16	14691829	14692029	chr16	18314655	18314825	3622626	Gene
<i>PLA2G10</i>	NP_003552	NM_003561	R57X	HGMD	chr16	14691829	14692029	chr16	29397701	29397871	14705672	Gene
<i>PLA2G10</i>	NP_003552	NM_003561	R57X	HGMD	chr16	14691829	14692029	chr16	30137042	30137212	15445013	Gene
<i>PLA2G12A</i>	NP_110448	NM_030821	Q27X	non-HGMD	chr4	110850593	110870691	chr1	52606731	52607752	na	Gene
<i>PLAGL2</i>	NP_002648	NM_002657	G465X	non-HGMD	chr20	30243967	30259207	chr4	85384690	85385915	na	Gene
<i>PLEKHM1</i>	NP_055613	NM_014798	S338X	HGMD	chr17	40901551	40901751	chr17	60241443	60241793	19339692	Gene
<i>PPFIBP1</i>	NP_003613	NM_003622	R47X	non-HGMD	chr12	27568311	27739764	chr12	27167125	27167837	400474	Gene
<i>PPIAL4A</i>	NP_001137355	NM_001143883	K151X	non-HGMD	chr1	143074818	143075603	chr18	4992504	4993294	na	Gene
<i>PPIAL4G</i>	NP_001116540	NM_001123068	K151X	non-HGMD	chr1	142558666	142559404	chr18	4992504	4993294	na	Gene
<i>PPP1R12B</i>	NP_002472	NM_002481	R805X	non-HGMD	chr1	200584452	200824320	chrY	23946727	23947340	na	Gene
<i>PPP1R12B</i>	NP_002472	NM_002481	R805X	non-HGMD	chr1	200584452	200824320	chrY	26834350	26834963	na	Gene
<i>PPP1R12B</i>	NP_002472	NM_002481	S694X	non-HGMD	chr1	200584452	200824320	chrY	23944270	23945003	na	Gene
<i>PPP1R12B</i>	NP_002472	NM_002481	S694X	non-HGMD	chr1	200584452	200824320	chrY	26836687	26837420	na	Gene
<i>PPP2R5C</i>	NP_001155198	NM_001161726	W120X	non-HGMD	chr14	101297887	101464081	chr3	52382213	52383285	na	Gene
<i>PRAMEF10</i>	NP_001034450	NM_001039361	F168X	non-HGMD	chr1	12875313	12880681	chr1	12822442	12823379	51934	Gene
<i>PRAMEF10</i>	NP_001034450	NM_001039361	F168X	non-HGMD	chr1	12875313	12880681	chr1	13094360	13095297	213679	Gene
<i>PRAMEF10</i>	NP_001034450	NM_001039361	Q315X	non-HGMD	chr1	12875313	12880681	chr5	65064428	65065485	na	Gene
<i>PRAMEF10</i>	NP_001034450	NM_001039361	W170X	non-HGMD	chr1	12875313	12880681	chr1	12936833	12937920	56152	Gene
<i>PRAMEF10</i>	NP_001034450	NM_001039361	W170X	non-HGMD	chr1	12875313	12880681	chr1	13047164	13048251	166483	Gene

PRAMEF10	NP_001034450	NM_001039361	W170X	non-HGMD	chr1	12875313	12880681	chr1	13224491	13225578	343810	Gene
PRAMEF10	NP_001034450	NM_001039361	W253X	non-HGMD	chr1	12875313	12880681	chr1	13284560	13285650	403879	Gene
PRAMEF10	NP_001034450	NM_001039361	W253X	non-HGMD	chr1	12875313	12880681	chr1	13505357	13506447	624676	Gene
PRAMEF10	NP_001034450	NM_001039361	Y316X	non-HGMD	chr1	12875313	12880681	chr5	65064428	65065485	na	Gene
PRAMEF16	NP_001038945	NM_001045480	C168X	non-HGMD	chr1	13367840	13370846	chr1	12822442	12823370	544470	Gene
PRAMEF16	NP_001038945	NM_001045480	C168X	non-HGMD	chr1	13367840	13370846	chr1	13094360	13095288	272552	Gene
PRAMEF16	NP_001038945	NM_001045480	Q208X	non-HGMD	chr1	13367840	13370846	chr1	12936833	12937911	429929	Gene
PRAMEF16	NP_001038945	NM_001045480	Q208X	non-HGMD	chr1	13367840	13370846	chr1	13047164	13048242	319598	Gene
PRAMEF16	NP_001038945	NM_001045480	Q208X	non-HGMD	chr1	13367840	13370846	chr1	13224500	13225578	142262	Gene
PRAMEF16	NP_001038945	NM_001045480	Q254X	non-HGMD	chr1	13367840	13370846	chr1	12936833	12937911	429929	Gene
PRAMEF16	NP_001038945	NM_001045480	Q254X	non-HGMD	chr1	13367840	13370846	chr1	13047164	13048242	319598	Gene
PRAMEF16	NP_001038945	NM_001045480	Q254X	non-HGMD	chr1	13367840	13370846	chr1	13224500	13225578	142262	Gene
PRAMEF16	NP_001038945	NM_001045480	Q315X	non-HGMD	chr1	13367840	13370846	chr5	65064428	65065485	na	Gene
PRAMEF16	NP_001038945	NM_001045480	Y316X	non-HGMD	chr1	13367840	13370846	chr5	65064428	65065485	na	Gene
PRAMEF17	NP_001093321	NM_001099851	C168X	non-HGMD	chr1	13588674	13591651	chr1	12822442	12823370	765304	Gene
PRAMEF17	NP_001093321	NM_001099851	C168X	non-HGMD	chr1	13588674	13591651	chr1	13094360	13095288	493386	Gene
PRAMEF17	NP_001093321	NM_001099851	Q208X	non-HGMD	chr1	13588674	13591651	chr1	12936833	12937911	650763	Gene
PRAMEF17	NP_001093321	NM_001099851	Q208X	non-HGMD	chr1	13588674	13591651	chr1	13047164	13048242	540432	Gene
PRAMEF17	NP_001093321	NM_001099851	Q208X	non-HGMD	chr1	13588674	13591651	chr1	13224500	13225578	363096	Gene
PRAMEF17	NP_001093321	NM_001099851	Q254X	non-HGMD	chr1	13588674	13591651	chr1	12936833	12937911	650763	Gene
PRAMEF17	NP_001093321	NM_001099851	Q254X	non-HGMD	chr1	13588674	13591651	chr1	13047164	13048242	540432	Gene
PRAMEF17	NP_001093321	NM_001099851	Q254X	non-HGMD	chr1	13588674	13591651	chr1	13224500	13225578	363096	Gene
PRAMEF17	NP_001093321	NM_001099851	Q315X	non-HGMD	chr1	13588674	13591651	chr5	65064428	65065485	na	Gene
PRAMEF17	NP_001093321	NM_001099851	Y316X	non-HGMD	chr1	13588674	13591651	chr5	65064428	65065485	na	Gene
PRDM7	NP_001091643	NM_001098173	W437X	non-HGMD	chr16	88650474	88669839	chr5	23561846	23562591	na	Pseudogene
PRELID1	NP_037369	NM_013237	W65X	non-HGMD	chr5	176663440	176666556	chr6	127006229	127007355	na	Gene
PRKRIR	NP_004696	NM_004705	E249X	non-HGMD	chr11	75738651	75769528	chr10	53841355	53842583	na	Gene
PRKRIR	NP_004696	NM_004705	R701X	non-HGMD	chr11	75738651	75769528	chr8	79834685	79835403	na	Gene
PRKRIR	NP_004696	NM_004705	S620X	non-HGMD	chr11	75738651	75769528	chr10	53842089	53843107	na	Gene
PRKRIR	NP_004696	NM_004705	W535X	non-HGMD	chr11	75738651	75769528	chr1	238999094	239000511	na	Gene
PRODH	NP_057419	NM_016335	Q59X	HGMD	chr22	17303523	17303723	chr22	18667283	18667567	1363560	Gene
PROS1	NP_000304	NM_000313	Q563X	HGMD	chr3	95078580	95078780	chr3	90337849	90338082	4740498	Gene
PROS1	NP_000304	NM_000313	Y340X	HGMD	chr3	95094501	95094701	chr3	90353759	90353989	4740512	Gene

<i>PRPF4B</i>	NP_003904	NM_003913	R991X	non-HGMD	chr6	3966567	4010216	chr9	116650161	116650939	na	Gene
<i>PRSS3</i>	NP_001184027	NM_001197098	S38X	non-HGMD	chr9	33740818	33789229	chr9	33603596	33604257	136561	Gene
<i>PRSS3</i>	NP_031369	NM_007343	S102X	non-HGMD	chr9	33740463	33789229	chr9	33603596	33604257	136206	Gene
<i>PSG1</i>	NP_001171755	NM_001184826	R288X	non-HGMD	chr19	48062452	48075711	chr19	48040759	48041534	20918	Gene
<i>PSG11</i>	NP_002776	NM_002785	R98X	non-HGMD	chr19	48203648	48222471	chr19	48131141	48132024	71624	Pseudogene
<i>PSG11</i>	NP_002776	NM_002785	Y68X	non-HGMD	chr19	48203648	48222471	chr19	48049692	48050569	153079	Gene
<i>PSG2</i>	NP_112536	NM_031246	R98X	non-HGMD	chr19	48260201	48278733	chr19	48131141	48132024	128177	Pseudogene
<i>PSG3</i>	NP_066296	NM_021016	E363X	non-HGMD	chr19	47917633	47936508	chr19	48040099	48040853	103591	Gene
<i>PSG3</i>	NP_066296	NM_021016	R288X	non-HGMD	chr19	47917633	47936508	chr19	48040759	48041534	104251	Gene
<i>PSG3</i>	NP_066296	NM_021016	R98X	non-HGMD	chr19	47917633	47936508	chr19	48131141	48132024	194633	Pseudogene
<i>PSG3</i>	NP_066296	NM_021016	Y68X	non-HGMD	chr19	47917633	47936508	chr19	48049692	48050569	113184	Gene
<i>PSG4</i>	NP_002771	NM_002780	R288X	non-HGMD	chr19	48388693	48401630	chr19	48040759	48041534	347159	Gene
<i>PSG4</i>	NP_002771	NM_002780	Y68X	non-HGMD	chr19	48388693	48401630	chr19	48049692	48050563	338130	Gene
<i>PSG5</i>	NP_002772	NM_002781	Y68X	non-HGMD	chr19	48363734	48382528	chr19	48049686	48050569	313165	Gene
<i>PSG6</i>	NP_001027020	NM_001031850	R287X	non-HGMD	chr19	48098079	48113883	chr19	48040759	48041534	56545	Gene
<i>PSG7</i>	NP_002774	NM_002783	R288X	non-HGMD	chr19	48120123	48133170	chr19	48040759	48041534	78589	Gene
<i>PSG7</i>	NP_002774	NM_002783	Y68X	non-HGMD	chr19	48120123	48133170	chr19	48049692	48050569	69554	Gene
<i>PSG8</i>	NP_001123640	NM_001130168	R166X	non-HGMD	chr19	47948678	47961671	chr19	48040759	48041534	79088	Gene
<i>PSG9</i>	NP_002775	NM_002784	R288X	non-HGMD	chr19	48449274	48465522	chr19	48040759	48041534	407740	Gene
<i>PSMD12</i>	NP_002807	NM_002816	L425X	non-HGMD	chr17	62767080	62793183	chr3	73008327	73009693	na	Gene
<i>PVRIG</i>	NP_076975	NM_024070	W45X	non-HGMD	chr7	99654806	99657047	chr7	99787860	99788722	130813	Gene
<i>RABGEF1</i>	NP_055319	NM_014504	R8X	non-HGMD	chr7	65843077	65913883	chr7	65555615	65556291	286786	Gene
<i>RABGEF1</i>	NP_055319	NM_014504	R8X	non-HGMD	chr7	65843077	65913883	chr7	75598991	75599670	9685108	Gene
<i>RALBP1</i>	NP_006779	NM_006788	R272X	non-HGMD	chr18	9465529	9528106	chr2	100407994	100409252	na	Gene
<i>RALBP1</i>	NP_006779	NM_006788	R272X	non-HGMD	chr18	9465529	9528106	chr3	181524013	181525793	na	Gene
<i>RANBP6</i>	NP_036548	NM_012416	Q519X	non-HGMD	chr9	6001018	6005618	chr12	9550569	9552553	na	Gene
<i>RANBP6</i>	NP_036548	NM_012416	Q583X	non-HGMD	chr9	6001018	6005618	chr12	9550569	9552553	na	Gene
<i>RANBP6</i>	NP_036548	NM_012416	R819X	non-HGMD	chr9	6001018	6005618	chr12	9550569	9552553	na	Gene
<i>RANBP6</i>	NP_036548	NM_012416	Y630X	non-HGMD	chr9	6001018	6005618	chr12	9550569	9552553	na	Gene
<i>RBMX2</i>	NP_057108	NM_016024	S210X	non-HGMD	chrX	129363623	129374998	chr1	119428897	119430350	na	Gene
<i>RBMXL1</i>	NP_001156008	NM_001162536	S257X	non-HGMD	chr1	89217726	89231231	chr9	30678853	30680456	na	Gene
<i>RDX</i>	NP_002897	NM_002906	K107X	HGMD	chr11	109639940	109640140	chrX	27445989	27446327	na	Gene
<i>RETSAT</i>	NP_060220	NM_017750	R482X	non-HGMD	chr2	85422588	85435332	chr2	85396011	85396684	25904	Gene

<i>RHBDF1</i>	NP_071895	NM_022450	L787X	non-HGMD	chr16	48057	62629	chr3	14589113	14591550	na	Gene
<i>RHPN2</i>	NP_149094	NM_033103	R184X	non-HGMD	chr19	38161337	38247664	chr15	18726888	18727516	na	Gene
<i>RHPN2</i>	NP_149094	NM_033103	W511X	non-HGMD	chr19	38161337	38247664	chr16	33320008	33320660	na	Gene
<i>RIMKLB</i>	NP_065785	NM_020734	G369X	non-HGMD	chr12	8741784	8821054	chr21	36344135	36345792	na	Gene
<i>RMND5A</i>	NP_073617	NM_022780	S58X	non-HGMD	chr2	86800924	86858675	chr2	87136042	87136685	277367	Gene
<i>ROCK1</i>	NP_005397	NM_005406	R1330X	HGMD	chr18	16787507	16787707	chr18	108296	108502	16679005	Gene
<i>RPL10</i>	NP_006004	NM_006013	R116X	HGMD	chrX	153281917	153282117	chr19	9791817	9792131	na	Gene
<i>RPL15</i>	NP_002939	NM_002948	Q149X	non-HGMD	chr3	23933642	23937336	chr13	99600489	99601600	na	Gene
<i>RPL15</i>	NP_002939	NM_002948	R169X	non-HGMD	chr3	23933642	23937336	chr11	115325874	115326979	na	Gene
<i>RPL15</i>	NP_002939	NM_002948	R169X	non-HGMD	chr3	23933642	23937336	chr12	8504231	8505342	na	Gene
<i>RPL15</i>	NP_002939	NM_002948	R169X	non-HGMD	chr3	23933642	23937336	chr15	73055382	73056214	na	Gene
<i>RPL15</i>	NP_002939	NM_002948	R169X	non-HGMD	chr3	23933642	23937336	chr2	138752635	138753500	na	Gene
<i>RPL15</i>	NP_002939	NM_002948	R169X	non-HGMD	chr3	23933642	23937336	chr22	25612621	25613600	na	Gene
<i>RPL15</i>	NP_002939	NM_002948	W150X	non-HGMD	chr3	23933642	23937336	chr11	115325874	115326979	na	Gene
<i>RPL36AL</i>	NP_000992	NM_001001	K53X	non-HGMD	chr14	49155156	49157099	chr15	73265798	73266612	na	Gene
<i>RPL7</i>	NP_000962	NM_000971	R217X	non-HGMD	chr8	74365427	74368423	chr13	76453072	76453877	na	Gene
<i>RPL7</i>	NP_000962	NM_000971	R217X	non-HGMD	chr8	74365427	74368423	chr2	47219879	47220591	na	Gene
<i>RPL7</i>	NP_000962	NM_000971	R217X	non-HGMD	chr8	74365427	74368423	chrX	26615357	26616240	na	Gene
<i>RPS6KB1</i>	NP_003152	NM_003161	W276X	HGMD	chr17	55366525	55366725	chr17	57670930	57671019	2304205	Gene
<i>RPSA</i>	NP_002286	NM_002295	R120X	non-HGMD	chr3	39423207	39429036	chr4	81301093	81302171	na	Gene
<i>SALL1</i>	NP_002959	NM_002968	G298X	HGMD	chr16	49732639	49732839	chrX	49319392	49320234	na	Gene
<i>SBDS</i>	NP_057122	NM_016038	K62X	HGMD	chr7	66096605	66096805	chr7	71939161	71939337	5842356	Gene
<i>SDHA</i>	NP_004159	NM_004168	S509X	HGMD	chr5	293467	293667	chr5	1642362	1642481	1348695	Gene
<i>SFRS6</i>	NP_006266	NM_006275	S310X	non-HGMD	chr20	41519917	41525658	chr17	16460703	16461886	na	Gene
<i>SLC16A6</i>	NP_004685	NM_004694	G307X	non-HGMD	chr17	63774761	63799000	chr17	60380013	60381343	3393418	Gene
<i>SLC25A15</i>	NP_055067	NM_014252	G244X	HGMD	chr13	40280583	40280783	chr21	9920983	9921117	na	Gene
<i>SMG1</i>	NP_055907	NM_015092	L1278X	HGMD	chr16	18778397	18778597	chr16	21367268	21367435	2588671	Gene
<i>SMG1</i>	NP_055907	NM_015092	L1278X	HGMD	chr16	18778397	18778597	chr16	21799528	21799695	3020931	Gene
<i>SMG1</i>	NP_055907	NM_015092	L1278X	HGMD	chr16	18778397	18778597	chr16	22409113	22409280	3630516	Gene
<i>SMG1</i>	NP_055907	NM_015092	L1278X	HGMD	chr16	18778397	18778597	chr16	29447757	29447924	10669160	Gene
<i>SMG1</i>	NP_055907	NM_015092	L1278X	HGMD	chr16	18778397	18778597	chr16	30187742	30187909	11409145	Gene
<i>SMG1</i>	NP_055907	NM_015092	R164X	HGMD	chr16	18810997	18811197	chr16	29480334	29480474	10669137	Gene
<i>SMG1</i>	NP_055907	NM_015092	R164X	HGMD	chr16	18810997	18811197	chr16	30220041	30220181	11408844	Gene

<i>SOCS5</i>	NP_054730	NM_014011	Q133X	non-HGMD	chr2	46779602	46843431	chr3	44725573	44727632	na	Gene
<i>SPDYE3</i>	NP_001004351	NM_001004351	R543X	non-HGMD	chr7	99743260	99757755	chr7	5037518	5038077	94705183	Gene
<i>SPIC</i>	NP_689536	NM_152323	E195X	non-HGMD	chr12	100394787	100404906	chr17	30541843	30542990	na	Gene
<i>SPIC</i>	NP_689536	NM_152323	Y191X	non-HGMD	chr12	100394787	100404906	chr11	33764926	33765875	na	Gene
<i>SPRYD5</i>	NP_116070	NM_032681	R444X	non-HGMD	chr11	55407348	55415860	chr11	48929504	48930270	6477078	Gene
<i>SPRYD5</i>	NP_116070	NM_032681	W353X	non-HGMD	chr11	55407348	55415860	chr11	48929275	48930017	6477331	Gene
<i>ST13</i>	NP_003923	NM_003932	S362X	non-HGMD	chr22	39550546	39582633	chr2	188398185	188399668	na	Gene
<i>STAG3</i>	NP_036579	NM_012447	Y434X	non-HGMD	chr7	99613473	99649946	chr7	72107263	72107873	27505600	Pseudogene
<i>STAG3</i>	NP_036579	NM_012447	Y434X	non-HGMD	chr7	99613473	99649946	chr7	73937519	73938129	25675344	Pseudogene
<i>STAG3</i>	NP_036579	NM_012447	Y434X	non-HGMD	chr7	99613473	99649946	chr7	74832905	74833515	24779958	Gene
<i>STRC</i>	NP_714544	NM_153700	Q1353X	HGMD	chr15	41684107	41684307	chr15	41783619	41783816	99312	Gene
<i>SUZ12</i>	NP_056170	NM_015355	R286X	non-HGMD	chr17	27288156	27352170	chr17	26120759	26121360	1166796	Gene
<i>TAS2R48</i>	NP_795369	NM_176888	R253X	non-HGMD	chr12	11065537	11066437	chr12	11120965	11122358	54528	Gene
<i>TCP10L2</i>	NP_001138593	NM_001145121	E10X	non-HGMD	chr6	167504070	167516386	chr6	167715952	167716682	199566	Pseudogene
<i>TEKT4</i>	NP_653306	NM_144705	Y346X	non-HGMD	chr2	94900958	94906295	chr4_r	494459	495117	na	Pseudogene
<i>THOC4</i>	NP_005773	NM_005782	A169X	non-HGMD	chr17	77439006	77442758	chr17_r	2406319	2406974	na	Gene
<i>THSD1</i>	NP_061146	NM_018676	Q774X	non-HGMD	chr13	51849303	51878630	chr13	51639843	51640708	208595	Gene
<i>TIMM8A</i>	NP_004076	NM_004085	R80X	HGMD	chrX	100488096	100488296	chr2	162642116	162642430	na	Gene
<i>TLK2</i>	NP_006843	NM_006852	Q166X	non-HGMD	chr17	57910117	58046573	chr7	128018893	128019557	na	Gene
<i>TMEM161B</i>	NP_699185	NM_153354	R456X	non-HGMD	chr5	87526778	87600421	chr10	37667430	37668055	na	Gene
<i>TMEM161B</i>	NP_699185	NM_153354	W465X	non-HGMD	chr5	87526778	87600421	chr10	37667430	37668055	na	Gene
<i>TOX4</i>	NP_055643	NM_014828	R481X	non-HGMD	chr14	21015174	21037159	chr4	113596279	113598641	na	Gene
<i>TRIM48</i>	NP_077019	NM_024114	R85X	non-HGMD	chr11	54786233	54795171	chr2	95604120	95605033	na	Gene
<i>TSPY1</i>	NP_003299	NM_003308	E13X	non-HGMD	chrY	9914563	9917358	chrY	10355016	10355998	437658	Gene
<i>TSPY2</i>	NP_072095	NM_022573	E13X	non-HGMD	chrY	6174263	6177053	chrY	10355016	10355998	4177963	Gene
<i>TTC28</i>	NP_001138890	NM_001145418	R21X	non-HGMD	chr22	26704001	27405853	chr10	39063283	39063887	na	Gene
<i>TTC28</i>	NP_001138890	NM_001145418	R21X	non-HGMD	chr22	26704001	27405853	chr2	91526339	91526943	na	Gene
<i>TUBB</i>	NP_821133	NM_178014	S230X	non-HGMD	chr6	30796135	30801174	chr8	30328816	30330647	na	Gene
<i>TUBB8</i>	NP_817124	NM_177987	Q131X	non-HGMD	chr10	82827	85178	chr12	34209494	34210272	na	Gene
<i>TYW1</i>	NP_060734	NM_018264	W437X	non-HGMD	chr7	66099251	66341933	chr7	71831471	71832087	5489538	Pseudogene
<i>UBB</i>	NP_061828	NM_018955	S65X	non-HGMD	chr17	16225091	16226779	chr2	136803362	136804203	na	Gene
<i>UBE2NL</i>	NP_001013007	NM_001012989	L89X	HGMD	chrX	142795035	142795235	chr4	173537824	173538213	na	Gene
<i>UBE3A</i>	NP_570853	NM_130838	R401X	HGMD	chr15	23167050	23167250	chr21	31356243	31356572	na	Gene

UGT1A7	NP_061950	NM_019077	R254X	HGMD	chr2	234255984	234256184	chr2	234158837	234159676	96308	Gene
UGT1A8	NP_061949	NM_019076	R254X	HGMD	chr2	234191754	234191954	chr2	234177598	234177786	13968	Gene
UGT2A3	NP_079019	NM_024743	C23X	HGMD	chr4	69851898	69852098	chr4	70246917	70247264	394819	Gene
UGT2B11	NP_001064	NM_001073	W117X	non-HGMD	chr4	70100635	70115038	chr4	70308894	70310119	193856	Gene
UGT2B28	NP_444267	NM_053039	W117X	non-HGMD	chr4	70180805	70195357	chr4	70308894	70310119	113537	Gene
ULBP1	NP_079494	NM_025218	W158X	non-HGMD	chr6	150326835	150336539	chr6	150363403	150364208	26864	Gene
ULK4	NP_060356	NM_017886	R483X	non-HGMD	chr3	41263093	41978664	chr15	28194633	28195342	na	Gene
ULK4	NP_060356	NM_017886	R483X	non-HGMD	chr3	41263093	41978664	chr15	28460939	28461654	na	Gene
ULK4	NP_060356	NM_017886	R483X	non-HGMD	chr3	41263093	41978664	chr15	28663619	28664328	na	Gene
ULK4	NP_060356	NM_017886	R483X	non-HGMD	chr3	41263093	41978664	chr15	30502263	30502972	na	Gene
USP32	NP_115971	NM_032582	R1447X	non-HGMD	chr17	55609472	55824368	chr17	18359114	18360060	37249412	Gene
USP6	NP_004496	NM_004505	E1152X	non-HGMD	chr17	4972410	5019048	chr17	16642031	16642821	11622983	Gene
USP6	NP_004496	NM_004505	R1082X	non-HGMD	chr17	4972410	5019048	chr17	16642031	16642821	11622983	Gene
USP6	NP_004496	NM_004505	R1082X	non-HGMD	chr17	4972410	5019048	chr17	20270446	20271236	15251398	Gene
USP6	NP_004496	NM_004505	R1245X	non-HGMD	chr17	4972410	5019048	chr17	18359114	18360060	13340066	Gene
USP8	NP_005145	NM_005154	R1112X	non-HGMD	chr15	48503870	48580569	chr6	31353871	31354757	na	Gene
VMA21	NP_001017980	NM_001017980	W95X	HGMD	chrX	150324067	150324267	chr7	43773547	43773720	na	Gene
WASH1	NP_878908	NM_182905	R26X	non-HGMD	chr9	4510	19739	chr2	114062352	114063001	na	Gene
WBP11	NP_057396	NM_016312	Q389X	non-HGMD	chr12	14830678	14847668	chrX	79703022	79703758	na	Gene
XPO6	NP_055986	NM_015171	R279X	non-HGMD	chr16	28016816	28130691	chr14	20259600	20261377	na	Gene
YES1	NP_005424	NM_005433	Q536X	non-HGMD	chr18	711591	802327	chr22	24373873	24375449	na	Gene
YWHAZ	NP_003397	NM_003406	E35X	non-HGMD	chr8	101999979	102034799	chr9	34911934	34913168	na	Gene
ZNF181	NP_001025168	NM_001029997	Q436X	non-HGMD	chr19	39917319	39925614	chr19	39866740	39868637	48682	Pseudogene
ZNF43	NP_003414	NM_003423	E682X	non-HGMD	chr19	21779591	21810810	chr19	21267633	21269455	510136	Pseudogene
ZNF479	NP_150376	NM_033273	E413X	non-HGMD	chr7	57191267	57211513	chr7	63363458	63365094	6151945	Pseudogene
ZNF479	NP_150376	NM_033273	E413X	non-HGMD	chr7	57191267	57211513	chr7	63316876	63318413	6105363	Gene
ZNF492	NP_065906	NM_020855	E453X	non-HGMD	chr19	22608965	22642312	chr9	35137133	35138451	na	Gene
ZNF519	NP_660330	NM_145287	E485X	non-HGMD	chr18	14089941	14122489	chr15	19370321	19371285	na	Gene
ZNF519	NP_660330	NM_145287	R415X	non-HGMD	chr18	14089941	14122489	chr15	19370321	19371285	na	Gene
ZNF519	NP_660330	NM_145287	R443X	non-HGMD	chr18	14089941	14122489	chr15	19370321	19371285	na	Gene
ZNF614	NP_079316	NM_025040	E403X	non-HGMD	chr19	57208388	57223492	chr11	97941120	97942006	na	Gene
ZNF614	NP_079316	NM_025040	R335X	non-HGMD	chr19	57208388	57223492	chr11	97941487	97942373	na	Gene
ZNF614	NP_079316	NM_025040	R419X	non-HGMD	chr19	57208388	57223492	chr11	97941120	97942006	na	Gene

<i>ZNF654</i>	NP_060763	NM_018293	S404X	non-HGMD	chr3	88270951	88276504	chr8	13643896	13644737	na	Gene
<i>ZNF681</i>	NP_612143	NM_138286	E510X	non-HGMD	chr19	23713836	23733533	chr19	20598372	20599267	3114569	Pseudogene
<i>ZNF705A</i>	NP_001004328	NM_001004328	R244X	non-HGMD	chr12	8216416	8223909	chr4	8994594	8995327	na	Gene
<i>ZNF705D</i>	NP_001034704	NM_001039615	R244X	non-HGMD	chr8	11984255	12010434	chr4	8994594	8995327	na	Gene
<i>ZNF705G</i>	NP_001157929	NM_001164457	Q244X	non-HGMD	chr8	7202907	7207900	chr4	8994594	8995327	na	Gene
<i>ZNF735</i>	NP_001152996	NM_001159524	E385X	non-HGMD	chr7	63305015	63318103	chr7	62389064	62390100	914915	Gene
<i>ZNF736</i>	NP_001164376	NM_001170905	W296X	non-HGMD	chr7	63410620	63447452	chr5	105906422	105907674	na	Gene
<i>ZNF850</i>	NP_001180481	NM_001193552	Y32X	non-HGMD	chr19	41926219	41955556	chr20	25765090	25765730	na	Gene
<i>ZNF850</i>	NP_001180481	NM_001193552	Y32X	non-HGMD	chr19	41926219	41955556	chr20	25947450	25948090	na	Gene
<i>ZNF91</i>	NP_003421	NM_003430	Q76X	non-HGMD	chr19	23332337	23370109	chr19	23783110	23783708	413001	Gene

ProteinID and GeneID are from the RefSeq database. Site: wild-type amino acid followed by the codon position and the deleterious amino acid (X: stop codon). Gene type: the acceptor gene can either be in the HGMD or non-HGMD data set. Chr: chromosome. The “r” the end of the chromosome number indicates an unmapped scaffold on that chromosome. When available, the distance between donor and acceptor sequences is in base-pairs.

Supplemental Table 10. Potential IGC-mediated deleterious mutations in HGMD genes with known IGC-mediated mutations.

Gene Symbol	ProteinID	GeneID	Site	Acceptor Chr.	Acceptor Start	Acceptor End	Donor Chr.	Donor Start	Donor End	Distance	Donor Type	In HGMD
GJA1	NP_000156	NM_000165	A40V	chr6	121809712	121809912	chr5	108414665	108416307	na	Pseudogene	yes
GJA1	NP_000156	NM_000165	R33Q	chr6	121809691	121809891	chr5	108414665	108416307	na	Pseudogene	no
GJA1	NP_000156	NM_000165	T204M	chr6	121810204	121810404	chr5	108414665	108416307	na	Pseudogene	no
GJA1	NP_000156	NM_000165	T290N	chr6	121810462	121810662	chr5	108414665	108416307	na	Pseudogene	yes
GJA1	NP_000156	NM_000165	Y185H	chr6	121810147	121810347	chr5	108414665	108416307	na	Pseudogene	no
NF1	NP_000258	NM_000267	E318Q	chr17	26551531	26551731	chr21	14294869	14295539	na	Pseudogene	no
NF1	NP_000258	NM_000267	E836A	chr17	26580167	26580367	chr22	14731813	14732744	na	Pseudogene	no
NF1	NP_000258	NM_000267	E836A	chr17	26580167	26580367	chr14	18558604	18559517	na	Pseudogene	no
NF1	NP_000258	NM_000267	E836A	chr17	26580167	26580367	chr14	19153937	19154850	na	Pseudogene	no
NF1	NP_000258	NM_000267	E836A	chr17	26580167	26580367	chr2	131665152	131666065	na	Pseudogene	no
NF1	NP_000258	NM_000267	F364V	chr17	26552110	26552310	chr18	14146351	14146961	na	Pseudogene	no
NF1	NP_000258	NM_000267	F894C	chr17	26580341	26580541	chr12	36886570	36887300	na	Pseudogene	no
NF1	NP_000258	NM_000267	F894Y	chr17	26580341	26580541	chr14	18558604	18559517	na	Pseudogene	no
NF1	NP_000258	NM_000267	F894Y	chr17	26580341	26580541	chr14	19153937	19154850	na	Pseudogene	no
NF1	NP_000258	NM_000267	F894Y	chr17	26580341	26580541	chr2	131665152	131666065	na	Pseudogene	no
NF1	NP_000258	NM_000267	F894Y	chr17	26580341	26580541	chr22	14731813	14732744	na	Pseudogene	no
NF1	NP_000258	NM_000267	G842S	chr17	26580185	26580385	chr22	14731813	14732744	na	Pseudogene	no
NF1	NP_000258	NM_000267	I719T	chr17	26577634	26577834	chr14	18561466	18562196	na	Pseudogene	no
NF1	NP_000258	NM_000267	I719T	chr17	26577634	26577834	chr14	19151263	19151993	na	Pseudogene	no
NF1	NP_000258	NM_000267	I719T	chr17	26577634	26577834	chr2	131667915	131668615	na	Pseudogene	no
NF1	NP_000258	NM_000267	I719T	chr17	26577634	26577834	chr22	14729129	14729835	na	Pseudogene	no
NF1	NP_000258	NM_000267	I913T	chr17	26580398	26580598	chr12	36886570	36887300	na	Pseudogene	no
NF1	NP_000258	NM_000267	K613X	chr17	26574605	26574805	chr2	131671395	131671538	na	Pseudogene	no
NF1	NP_000258	NM_000267	K798Q	chr17	26578635	26578835	chr14	18560435	18561021	na	Pseudogene	no
NF1	NP_000258	NM_000267	K798Q	chr17	26578635	26578835	chr14	19152436	19153022	na	Pseudogene	no
NF1	NP_000258	NM_000267	K798Q	chr17	26578635	26578835	chr2	131666988	131667577	na	Pseudogene	no
NF1	NP_000258	NM_000267	K798Q	chr17	26578635	26578835	chr22	14730303	14730889	na	Pseudogene	no
NF1	NP_000258	NM_000267	L578Y	chr17	26574500	26574700	chr2	131671145	131671788	na	Pseudogene	no
NF1	NP_000258	NM_000267	L578Y	chr17	26574500	26574700	chr14	18564551	18565164	na	Pseudogene	no
NF1	NP_000258	NM_000267	L578Y	chr17	26574500	26574700	chr14	19148295	19148908	na	Pseudogene	no

NF1	NP_000258	NM_000267	Q1033R	chr17	26581412	26581612	chr14	18557653	18558290	na	Pseudogene	no
NF1	NP_000258	NM_000267	Q589X	chr17	26574533	26574733	chr14	18564801	18564914	na	Pseudogene	no
NF1	NP_000258	NM_000267	Q589X	chr17	26574533	26574733	chr14	19148545	19148658	na	Pseudogene	no
NF1	NP_000258	NM_000267	Q589X	chr17	26574533	26574733	chr22	14726426	14726539	na	Pseudogene	no
NF1	NP_000258	NM_000267	R1204G	chr17	26584161	26584361	chr15	19398607	19399226	na	Pseudogene	yes
NF1	NP_000258	NM_000267	R304W	chr17	26551489	26551689	chr21	14294869	14295539	na	Pseudogene	no
NF1	NP_000258	NM_000267	R304X	chr17	26551489	26551689	chr18	14147168	14147341	na	Pseudogene	yes
NF1	NP_000258	NM_000267	R601Q	chr17	26574569	26574769	chr2	131671145	131671788	na	Pseudogene	no
NF1	NP_000258	NM_000267	S1546X	chr17	26612878	26613078	chr15	19390394	19390504	na	Pseudogene	no
PKD1	NP_000287	NM_000296	A3053T	chr16	2092324	2092524	chr16	14931606	14932363	12839082	Pseudogene	no
PKD1	NP_000287	NM_000296	A3053T	chr16	2092324	2092524	chr16	16330091	16330848	14237567	Pseudogene	no
PKD1	NP_000287	NM_000296	A3053T	chr16	2092324	2092524	chr16	18380583	18381340	16288059	Pseudogene	no
PKD1	NP_000287	NM_000296	E2810X	chr16	2093528	2093728	chr16	15132443	15133066	13038715	Gene	yes
PKD1	NP_000287	NM_000296	E2810X	chr16	2093528	2093728	chr16	14930608	14931084	12836880	Pseudogene	yes
PKD1	NP_000287	NM_000296	F2806L	chr16	2093540	2093740	chr16	16372145	16373268	14278405	Pseudogene	no
PKD1	NP_000287	NM_000296	F2806L	chr16	2093540	2093740	chr16	18341355	18342478	16247615	Pseudogene	no
PKD1	NP_000287	NM_000296	G2473E	chr16	2096369	2096569	chr16	18383516	18384846	16286947	Pseudogene	no
PKD1	NP_000287	NM_000296	G2530S	chr16	2096105	2096305	chr16	16370051	16371381	14273746	Pseudogene	no
PKD1	NP_000287	NM_000296	G2530S	chr16	2096105	2096305	chr16	18383516	18384846	16287211	Pseudogene	no
PKD1	NP_000287	NM_000296	G381V	chr16	2107750	2107950	chr16	16358234	16359417	14250284	Pseudogene	no
PKD1	NP_000287	NM_000296	G475R	chr16	2106915	2107115	chr16	15137083	15137741	13029968	Pseudogene	no
PKD1	NP_000287	NM_000296	H1369R	chr16	2100961	2101161	chr16	16364859	16366858	14263698	Pseudogene	no
PKD1	NP_000287	NM_000296	H1369R	chr16	2100961	2101161	chr16	18388039	18390038	16286878	Pseudogene	no
PKD1	NP_000287	NM_000296	I2872L	chr16	2093342	2093542	chr16	14930783	14931480	12837241	Pseudogene	no
PKD1	NP_000287	NM_000296	I2872L	chr16	2093342	2093542	chr16	16329268	16329965	14235726	Pseudogene	no
PKD1	NP_000287	NM_000296	I2872L	chr16	2093342	2093542	chr16	18381466	18382163	16287924	Pseudogene	no
PKD1	NP_000287	NM_000296	L2816P	chr16	2093510	2093710	chr16	14930358	14931334	12836648	Pseudogene	yes
PKD1	NP_000287	NM_000296	L2816P	chr16	2093510	2093710	chr16	15132193	15133316	13038483	Gene	yes
PKD1	NP_000287	NM_000296	L2816P	chr16	2093510	2093710	chr16	16328843	16329819	14235133	Pseudogene	yes
PKD1	NP_000287	NM_000296	L2816P	chr16	2093510	2093710	chr16	16372145	16373268	14278435	Pseudogene	yes
PKD1	NP_000287	NM_000296	L2816P	chr16	2093510	2093710	chr16	18341355	18342478	16247645	Pseudogene	yes
PKD1	NP_000287	NM_000296	L2816P	chr16	2093510	2093710	chr16	18381612	18382588	16287902	Pseudogene	yes
PKD1	NP_000287	NM_000296	L727P	chr16	2104743	2104943	chr16	14923094	14924364	12818151	Pseudogene	yes

PKD1	NP_000287	NM_000296	L727P	chr16	2104743	2104943	chr16	16321745	16323015	14216802	Pseudogene	yes
PKD1	NP_000287	NM_000296	L727P	chr16	2104743	2104943	chr16	16361774	16363044	14256831	Pseudogene	yes
PKD1	NP_000287	NM_000296	L727P	chr16	2104743	2104943	chr16	18391853	18393123	16286910	Pseudogene	yes
PKD1	NP_000287	NM_000296	L845S	chr16	2104389	2104589	chr16	14923094	14924364	12818505	Pseudogene	yes
PKD1	NP_000287	NM_000296	L845S	chr16	2104389	2104589	chr16	16321745	16323015	14217156	Pseudogene	yes
PKD1	NP_000287	NM_000296	L845S	chr16	2104389	2104589	chr16	16361774	16363044	14257185	Pseudogene	yes
PKD1	NP_000287	NM_000296	L845S	chr16	2104389	2104589	chr16	18348368	18349638	16243779	Pseudogene	yes
PKD1	NP_000287	NM_000296	L845S	chr16	2104389	2104589	chr16	18391853	18393123	16287264	Pseudogene	yes
PKD1	NP_000287	NM_000296	P2736R	chr16	2093750	2093950	chr16	14930358	14931334	12836408	Pseudogene	no
PKD1	NP_000287	NM_000296	P2736R	chr16	2093750	2093950	chr16	15132193	15133316	13038243	Gene	no
PKD1	NP_000287	NM_000296	P2736R	chr16	2093750	2093950	chr16	16328843	16329819	14234893	Pseudogene	no
PKD1	NP_000287	NM_000296	P2736R	chr16	2093750	2093950	chr16	16372145	16373268	14278195	Pseudogene	no
PKD1	NP_000287	NM_000296	P2736R	chr16	2093750	2093950	chr16	18341355	18342478	16247405	Pseudogene	no
PKD1	NP_000287	NM_000296	P2736R	chr16	2093750	2093950	chr16	18381612	18382588	16287662	Pseudogene	no
PKD1	NP_000287	NM_000296	P2835L	chr16	2093453	2093653	chr16	16372145	16373268	14278492	Pseudogene	no
PKD1	NP_000287	NM_000296	P2835L	chr16	2093453	2093653	chr16	18341355	18342478	16247702	Pseudogene	no
PKD1	NP_000287	NM_000296	Q1483X	chr16	2100619	2100819	chr16	16365109	16366608	14264290	Pseudogene	no
PKD1	NP_000287	NM_000296	Q554X	chr16	2106490	2106690	chr16	14921602	14921760	12814912	Pseudogene	no
PKD1	NP_000287	NM_000296	Q554X	chr16	2106490	2106690	chr16	16320253	16320411	14213563	Pseudogene	no
PKD1	NP_000287	NM_000296	Q554X	chr16	2106490	2106690	chr16	18350972	18351130	16244282	Pseudogene	no
PKD1	NP_000287	NM_000296	R2220W	chr16	2098408	2098608	chr16	18385549	18387245	16286941	Pseudogene	yes
PKD1	NP_000287	NM_000296	R2347H	chr16	2097808	2098008	chr16	16367652	16369348	14269644	Pseudogene	no
PKD1	NP_000287	NM_000296	R2430X	chr16	2096498	2096698	chr16	15135151	15135432	13038453	Pseudogene	yes
PKD1	NP_000287	NM_000296	R2477C	chr16	2096357	2096557	chr16	16325763	16327093	14229206	Pseudogene	no
PKD1	NP_000287	NM_000296	R2477C	chr16	2096357	2096557	chr16	16370051	16371381	14273494	Pseudogene	no
PKD1	NP_000287	NM_000296	R2518C	chr16	2096141	2096341	chr16	16370051	16371381	14273710	Pseudogene	no
PKD1	NP_000287	NM_000296	R2518C	chr16	2096141	2096341	chr16	18383516	18384846	16287175	Pseudogene	no
PKD1	NP_000287	NM_000296	R966W	chr16	2103149	2103349	chr16	14924712	14925397	12821363	Pseudogene	no
PKD1	NP_000287	NM_000296	S2745L	chr16	2093723	2093923	chr16	16372145	16373268	14278222	Pseudogene	no
PKD1	NP_000287	NM_000296	S2745L	chr16	2093723	2093923	chr16	18341355	18342478	16247432	Pseudogene	no
PKD1	NP_000287	NM_000296	T1115M	chr16	2101723	2101923	chr16	18388039	18390038	16286116	Pseudogene	no
PKD1	NP_000287	NM_000296	T1649M	chr16	2100121	2100321	chr16	18386745	18388537	16286424	Pseudogene	no
PKD1	NP_000287	NM_000296	T334M	chr16	2107891	2108091	chr16	18395480	18396663	16287389	Pseudogene	no

<i>PKD1</i>	NP_000287	NM_000296	V1129E	chr16	2101681	2101881	chr16	16364859	16366858	14262978	Pseudogene	no
<i>PKD1</i>	NP_000287	NM_000296	V1129E	chr16	2101681	2101881	chr16	18388039	18390038	16286158	Pseudogene	no
<i>PKD1</i>	NP_000287	NM_000296	V2019F	chr16	2099011	2099211	chr16	16366360	16368152	14267149	Pseudogene	no
<i>PKD1</i>	NP_000287	NM_000296	V2019F	chr16	2099011	2099211	chr16	18386745	18388537	16287534	Pseudogene	no
<i>PKD1</i>	NP_000287	NM_000296	V3057M	chr16	2092312	2092512	chr16	16330091	16330848	14237579	Pseudogene	no
<i>PKD1</i>	NP_000287	NM_000296	V3057M	chr16	2092312	2092512	chr16	16373390	16374147	14280878	Pseudogene	no
<i>PKD1</i>	NP_000287	NM_000296	V3057M	chr16	2092312	2092512	chr16	18340476	18341233	16247964	Pseudogene	no
<i>PKD1</i>	NP_000287	NM_000296	V3202I	chr16	2090173	2090373	chr16	15129086	15129744	13038713	Gene	no
<i>PKD1</i>	NP_000287	NM_000296	W3001L	chr16	2092480	2092680	chr16	15131310	15132067	13038630	Gene	no
<i>PKD1</i>	NP_000287	NM_000296	Y2168C	chr16	2098564	2098764	chr16	16367652	16369348	14268888	Pseudogene	no
<i>PKD1</i>	NP_000287	NM_000296	Y2168C	chr16	2098564	2098764	chr16	18385549	18387245	16286785	Pseudogene	no
<i>CYP21A2</i>	NP_000491	NM_000500	Q319X	chr6	32116079	32116279	chr6	32083387	32083602	32477	Pseudogene	yes
<i>CYP21A2</i>	NP_000491	NM_000500	R357W	chr6	32116193	32116393	chr6	32083137	32083852	32341	Pseudogene	yes
<i>CYP21A2</i>	NP_000491	NM_000500	V238E	chr6	32115467	32115667	chr6	32082515	32083125	32342	Pseudogene	no
<i>VWF</i>	NP_000543	NM_000552	C1031S	chr12	6005247	6005447	chr22	15564950	15565617	na	Pseudogene	no
<i>VWF</i>	NP_000543	NM_000552	G1209R	chr12	6001273	6001473	chr22	15560956	15561590	na	Pseudogene	no
<i>VWF</i>	NP_000543	NM_000552	N1005S	chr12	6005325	6005525	chr22	15564950	15565617	na	Pseudogene	no
<i>VWF</i>	NP_000543	NM_000552	V1730G	chr12	5995963	5996163	chr22	15555639	15556288	na	Pseudogene	no
<i>CYP2A6</i>	NP_000753	NM_000762	R64C	chr19	46047613	46047813	chr19	46079082	46079755	31269	Gene	no
<i>ABCC6</i>	NP_001162	NM_001171	Q378X	chr16	16203300	16203500	chr16	18511348	18511623	2307848	Pseudogene	yes
<i>SBDS</i>	NP_057122	NM_016038	G24R	chr7	66097667	66097867	chr7	71937921	71938549	5840054	Pseudogene	no
<i>SBDS</i>	NP_057122	NM_016038	K62X	chr7	66096605	66096805	chr7	71939161	71939337	5842356	Pseudogene	no
<i>SBDS</i>	NP_057122	NM_016038	N121T	chr7	66095634	66095834	chr7	71939906	71940564	5844072	Pseudogene	yes
<i>PRODH</i>	NP_057419	NM_016335	Q59X	chr22	17303523	17303723	chr22	18667283	18667567	1363560	Pseudogene	no

ProteinID and GeneID are from the RefSeq database. Site: wild-type amino acid followed by the codon position and the deleterious amino acid (X: stop codon). Chr: chromosome. When available, the distance between donor and acceptor sequences is given in base-pairs. In HGMD: amino acid replacement reported in the HGMD missense/nonsense mutations data set.

Supplemental Table 11. Potential IGC-mediated deleterious mutations in HGMD genes without known IGC-mediated mutations.

Gene Symbol	ProteinID	GeneID	Site	Acceptor Chr.	Acceptor Start	Acceptor End	Donor Chr.	Donor Start	Donor End	Distance	Donor Type	In HGMD
ABCA3	NP_001080	NM_001089	G1606S	chr16	2,267,871	2,268,071	chr16	2,414,363	2,415,021	146,292	Pseudogene	no
ABCD1	NP_000024	NM_000033	R617C	chrX	152,661,605	152,661,805	chr22	15,250,611	15,251,197	na	Pseudogene	yes
ABCD1	NP_000024	NM_000033	A626T	chrX	152,661,781	152,661,981	chr16	32,394,014	32,394,645	na	Pseudogene	yes
ABCD1	NP_000024	NM_000033	A626T	chrX	152,661,781	152,661,981	chr22	15,250,844	15,251,472	na	Pseudogene	yes
ABCD1	NP_000024	NM_000033	C631G	chrX	152,661,796	152,661,996	chr10	38,936,980	38,937,611	na	Pseudogene	no
ABCD1	NP_000024	NM_000033	H659L	chrX	152,661,880	152,662,080	chr10	38,936,980	38,937,611	na	Pseudogene	no
ABCD1	NP_000024	NM_000033	R660Q	chrX	152,661,883	152,662,083	chr10	38,936,980	38,937,611	na	Pseudogene	no
ABCD1	NP_000024	NM_000033	R689H	chrX	152,662,112	152,662,312	chr10	38,937,257	38,937,999	na	Pseudogene	no
ABCD1	NP_000024	NM_000033	R689S	chrX	152,662,112	152,662,312	chr22	15,251,118	15,251,860	na	Pseudogene	no
ACSM2B	NP_872423	NM_182617	D463N	chr16	20,461,877	20,462,077	chr16	20,399,131	20,399,774	62,103	Gene	no
ACTB	NP_001092	NM_001101	G158E	chr7	5,534,665	5,534,865	chr1	222,118,184	222,119,124	na	Pseudogene	no
ACTG1	NP_001605	NM_001614	A230P	chr17	77,092,820	77,093,020	chrY	18,378,028	18,379,649	na	Pseudogene	no
ACTG1	NP_001605	NM_001614	A230P	chr17	77,092,820	77,093,020	chrY	18,768,908	18,770,529	na	Pseudogene	no
ADH5	NP_000662	NM_000671	Y149C	chr4	100,216,894	100,217,094	chr1	238,399,843	238,401,464	na	Pseudogene	no
AKR1C3	NP_003730	NM_003739	G72D	chr10	5,128,633	5,128,833	chr10	5,164,541	5,165,211	35,708	Pseudogene	no
AKR1C4	NP_001809	NM_001818	G72D	chr10	5,232,175	5,232,375	chr10	5,164,535	5,165,211	66,964	Pseudogene	no
AKR7A3	NP_036199	NM_012067	R106X	chr1	19,485,249	19,485,449	chr1	19,469,829	19,470,017	15,232	Gene	no
ALDOA	NP_000025	NM_000034	G75K	chr16	29,986,284	29,986,484	chr3	52,201,976	52,202,982	na	Pseudogene	no
ALG1	NP_061982	NM_019109	R276Q	chr16	5,068,746	5,068,946	chr4	9,320,298	9,320,929	na	Pseudogene	no
ALG1	NP_061982	NM_019109	G323R	chr16	5,070,855	5,071,055	chr4	9,318,094	9,318,731	na	Pseudogene	no
ALG1	NP_061982	NM_019109	L325P	chr16	5,070,861	5,071,061	chr11	3,382,409	3,383,010	na	Pseudogene	no
ALG1	NP_061982	NM_019109	S369F	chr16	5,072,495	5,072,695	chr7	6,934,419	6,935,038	na	Pseudogene	no
ALG1	NP_061982	NM_019109	S370C	chr16	5,072,498	5,072,698	chr11	3,384,054	3,384,667	na	Pseudogene	no
ALG1	NP_061982	NM_019109	G384R	chr16	5,072,540	5,072,740	chr11	71,191,304	71,191,923	na	Pseudogene	no
ALG1	NP_061982	NM_019109	G384R	chr16	5,072,540	5,072,740	chr3	75,551,358	75,552,028	na	Pseudogene	no
ALG1	NP_061982	NM_019109	G384R	chr16	5,072,540	5,072,740	chr3	131,297,325	131,297,944	na	Pseudogene	no
ALG1	NP_061982	NM_019109	G384R	chr16	5,072,540	5,072,740	chr4	3,992,595	3,993,205	na	Pseudogene	no
ALG1	NP_061982	NM_019109	F427S	chr16	5,074,669	5,074,869	chr12	8,275,245	8,275,879	na	Pseudogene	no
ALG1	NP_061982	NM_019109	E443X	chr16	5,074,717	5,074,917	chr7	6,936,857	6,936,991	na	Pseudogene	no
ALG1	NP_061982	NM_019109	E451X	chr16	5,074,741	5,074,941	chr11	3,386,486	3,386,620	na	Pseudogene	no

ALG1	NP_061982	NM_019109	V457M	chr16	5,074,759	5,074,959	chr12	8,275,245	8,275,879	na	Pseudogene	no
ALOX15	NP_001131	NM_001140	H355R	chr17	4,485,798	4,485,998	chr17	6,505,012	6,505,709	2,019,014	Pseudogene	no
ALOX15	NP_001131	NM_001140	K344Q	chr17	4,485,831	4,486,031	chr17	6,505,012	6,505,709	2,018,981	Pseudogene	no
ALOX15	NP_001131	NM_001140	I289S	chr17	4,487,142	4,487,342	chr17	6,503,715	6,504,313	2,016,373	Pseudogene	no
APBA2	NP_005494	NM_005503	S249I	chr15	27,134,026	27,134,226	chr15	32,488,891	32,490,314	5,354,665	Pseudogene	no
APBA2	NP_005494	NM_005503	S249I	chr15	27,134,026	27,134,226	chr15	32,635,113	32,636,536	5,500,887	Pseudogene	no
AQP7	NP_001161	NM_001170	T160N	chr9	33,376,019	33,376,219	chr9	68,924,620	68,925,236	35,548,401	Pseudogene	no
ARSE	NP_000038	NM_000047	W581X	chrX	2,862,799	2,862,999	chrY	12,977,873	12,978,229	na	Pseudogene	yes
ARSE	NP_000038	NM_000047	G443V	chrX	2,864,764	2,864,964	chrY	12,971,646	12,972,223	na	Pseudogene	no
ARSE	NP_000038	NM_000047	G434S	chrX	2,864,791	2,864,991	chrY	12,971,646	12,972,223	na	Pseudogene	no
AURKA	NP_940835	NM_198433	H380Y	chr20	54,378,592	54,378,792	chr1	218,506,422	218,507,116	na	Pseudogene	no
AURKA	NP_940835	NM_198433	R46W	chr20	54,394,800	54,395,000	chr1	218,507,257	218,508,128	na	Pseudogene	no
BCR	NP_004318	NM_004327	R1090H	chr22	21,983,871	21,984,071	chr22	22,989,384	22,989,982	1,005,313	Pseudogene	no
BMPRI1A	NP_004320	NM_004329	L334S	chr10	88,668,942	88,669,142	chr11	120,737,522	120,739,032	na	Pseudogene	no
BMPRI1A	NP_004320	NM_004329	I349T	chr10	88,668,987	88,669,187	chr6	129,520,114	129,521,837	na	Pseudogene	no
C6orf66	NP_054884	NM_014165	W133X	chr6	97,445,729	97,445,929	chr15	47,235,854	47,236,336	na	Pseudogene	no
CD1A	NP_001754	NM_001763	R249L	chr1	156,493,242	156,493,442	chr1	156,565,535	156,566,313	72,093	Gene	no
CD1A	NP_001754	NM_001763	R249P	chr1	156,493,242	156,493,442	chr1	156,419,041	156,419,819	73,423	Gene	no
CD1A	NP_001754	NM_001763	D257N	chr1	156,493,266	156,493,466	chr1	156,565,535	156,566,313	72,069	Gene	no
CDH12	NP_004052	NM_004061	L161W	chr5	22,010,899	22,011,099	chr5	69,316,533	69,317,332	47,305,434	Pseudogene	no
CDH12	NP_004052	NM_004061	L161W	chr5	22,010,899	22,011,099	chr5	69,464,009	69,464,808	47,452,910	Pseudogene	no
CDH12	NP_004052	NM_004061	L161W	chr5	22,010,899	22,011,099	chr5	70,191,616	70,192,415	48,180,517	Pseudogene	no
CDH12	NP_004052	NM_004061	L161W	chr5	22,010,899	22,011,099	chr5	70,464,323	70,465,122	48,453,224	Pseudogene	no
CDH12	NP_004052	NM_004061	L161W	chr5	22,010,899	22,011,099	chr8	933,859	934,658	na	Pseudogene	no
CENPJ	NP_060921	NM_018451	E212X	chr13	24,382,056	24,382,256	chr13	23,989,878	23,990,210	391,846	Pseudogene	no
CES1	NP_001020365	NM_001025194	F480S	chr16	54,397,652	54,397,852	chr16	54,337,920	54,338,563	59,089	Pseudogene	no
CHEK2	NP_009125	NM_007194	R521W	chr22	27,413,853	27,414,053	chr15	18,756,358	18,756,947	na	Pseudogene	no
CHEK2	NP_009125	NM_007194	Y390C	chr22	27,421,686	27,421,886	chr15	18,748,513	18,749,174	na	Pseudogene	no
COL12A1	NP_004361	NM_004370	R1779H	chr6	75,903,829	75,904,029	chr2	14,056,334	14,056,986	na	Pseudogene	no
COL12A1	NP_004361	NM_004370	R1757X	chr6	75,903,895	75,904,095	chr2	14,056,584	14,056,736	na	Pseudogene	no
CR1	NP_000564	NM_000573	I224V	chr1	205,763,663	205,763,863	chr1	205,950,102	205,950,937	186,239	Pseudogene	no
CR1	NP_000564	NM_000573	G673V	chr1	205,800,788	205,800,988	chr1	205,950,102	205,950,937	149,114	Pseudogene	no
CR1	NP_000564	NM_000573	T1056M	chr1	205,815,529	205,815,729	chr1	205,947,897	205,948,483	132,168	Gene	no

CR1	NP_000564	NM_000573	A1073T	chr1	205,817,704	205,817,904	chr1	205,950,102	205,950,937	132,198	Pseudogene	no
CR1	NP_000564	NM_000573	R1094H	chr1	205,817,767	205,817,967	chr1	205,934,073	205,934,968	116,106	Gene	no
CR1	NP_000564	NM_000573	R1094H	chr1	205,817,767	205,817,967	chr1	205,950,102	205,950,937	132,135	Pseudogene	no
CR1	NP_000564	NM_000573	G1123V	chr1	205,817,854	205,818,054	chr1	205,950,102	205,950,937	132,048	Pseudogene	no
CR1	NP_000564	NM_000573	A1129V	chr1	205,817,872	205,818,072	chr1	205,950,102	205,950,937	132,030	Pseudogene	no
CUBN	NP_001072	NM_001081	D3224H	chr10	16,922,943	16,923,143	chr10	45,062,404	45,063,068	28,139,261	Pseudogene	no
CUBN	NP_001072	NM_001081	T2432I	chr10	16,996,991	16,997,191	chr10	42,518,421	42,519,064	25,521,230	Pseudogene	no
CUBN	NP_001072	NM_001081	T2432I	chr10	16,996,991	16,997,191	chr10	44,955,504	44,956,147	27,958,313	Pseudogene	no
CUBN	NP_001072	NM_001081	G2412A	chr10	16,997,051	16,997,251	chr10	42,518,421	42,519,064	25,521,170	Pseudogene	no
CUBN	NP_001072	NM_001081	G2412A	chr10	16,997,051	16,997,251	chr10	44,955,504	44,956,147	27,958,253	Pseudogene	no
CUBN	NP_001072	NM_001081	I2397T	chr10	16,997,744	16,997,944	chr10	42,519,208	42,519,920	25,521,264	Pseudogene	no
CUBN	NP_001072	NM_001081	G2372R	chr10	16,997,819	16,998,019	chr10	42,519,208	42,519,920	25,521,189	Pseudogene	no
CUBN	NP_001072	NM_001081	G2372R	chr10	16,997,819	16,998,019	chr10	44,954,655	44,955,358	27,956,636	Pseudogene	no
CUBN	NP_001072	NM_001081	G2173V	chr10	17,007,272	17,007,472	chr10	42,525,999	42,526,702	25,518,527	Pseudogene	no
CUBN	NP_001072	NM_001081	S2105F	chr10	17,007,635	17,007,835	chr10	42,526,336	42,527,003	25,518,501	Pseudogene	no
CYCS	NP_061820	NM_018947	Y98C	chr7	25,129,768	25,129,968	chr11	31,258,551	31,259,350	na	Pseudogene	no
CYCS	NP_061820	NM_018947	Y98X	chr7	25,129,768	25,129,968	chr3	10,074,660	10,074,971	na	Pseudogene	no
CYP2A13	NP_000757	NM_000766	M349T	chr19	46,291,963	46,292,163	chr19	46,107,370	46,108,073	183,890	Pseudogene	no
CYP2C8	NP_000761	NM_000770	T301M	chr10	96,795,514	96,795,714	chr10	96,759,170	96,759,828	35,686	Pseudogene	no
CYP2F1	NP_000765	NM_000774	V175R	chr19	46,319,143	46,319,343	chr19	46,024,085	46,024,767	294,376	Pseudogene	no
CYP2F1	NP_000765	NM_000774	A351M	chr19	46,322,452	46,322,652	chr19	46,019,707	46,020,395	302,057	Pseudogene	no
CYP3A4	NP_059488	NM_017460	P45L	chr7	99,215,480	99,215,680	chr7	99,131,055	99,131,647	83,833	Pseudogene	no
CYP4A11	NP_000769	NM_000778	R231C	chr1	47,173,255	47,173,455	chr1	47,381,769	47,382,433	208,314	Gene	no
DDX11	NP_085911	NM_030653	G354R	chr12	31,134,168	31,134,368	chr12	9,342,908	9,343,593	21,790,575	Gene	no
DRD5	NP_000789	NM_000798	P96S	chr4	9,392,939	9,393,139	chr1	147,168,716	147,169,614	na	Pseudogene	no
DRD5	NP_000789	NM_000798	P96S	chr4	9,392,939	9,393,139	chr2	91,236,988	91,237,886	na	Pseudogene	no
DRD5	NP_000789	NM_000798	G110E	chr4	9,392,981	9,393,181	chr1	147,168,716	147,169,614	na	Pseudogene	no
DRD5	NP_000789	NM_000798	G110E	chr4	9,392,981	9,393,181	chr2	91,236,988	91,237,886	na	Pseudogene	no
DRD5	NP_000789	NM_000798	W188X	chr4	9,393,215	9,393,415	chr1	147,169,370	147,170,044	na	Pseudogene	no
DRD5	NP_000789	NM_000798	W188X	chr4	9,393,215	9,393,415	chr2	91,236,558	91,237,232	na	Pseudogene	no
DRD5	NP_000789	NM_000798	S228P	chr4	9,393,335	9,393,535	chr2	91,236,308	91,237,482	na	Pseudogene	no
DRD5	NP_000789	NM_000798	G348C	chr4	9,393,695	9,393,895	chr1	147,169,120	147,170,294	na	Pseudogene	no
DRD5	NP_000789	NM_000798	G348C	chr4	9,393,695	9,393,895	chr2	91,236,308	91,237,482	na	Pseudogene	no

<i>DRD5</i>	NP_000789	NM_000798	W455X	chr4	9,394,016	9,394,216	chr1	147,170,038	147,170,406	na	Pseudogene	no
<i>DRD5</i>	NP_000789	NM_000798	W455X	chr4	9,394,016	9,394,216	chr2	91,236,196	91,236,564	na	Pseudogene	no
<i>FANCD2</i>	NP_149075	NM_033084	C342R	chr3	10,060,104	10,060,304	chr3	10,004,804	10,005,411	54,693	Pseudogene	no
<i>FANCD2</i>	NP_149075	NM_033084	L436M	chr3	10,064,530	10,064,730	chr3	10,010,586	10,011,238	53,292	Pseudogene	no
<i>FANCD2</i>	NP_149075	NM_033084	Q448X	chr3	10,064,566	10,064,766	chr3	10,010,836	10,010,988	53,578	Pseudogene	no
<i>FANCD2</i>	NP_149075	NM_033084	A586V	chr3	10,076,979	10,077,179	chr3	11,897,095	11,897,711	1,819,916	Pseudogene	no
<i>FCGR1A</i>	NP_000557	NM_000566	Q224X	chr1	148,028,246	148,028,446	chr1	120,729,852	120,730,139	27,298,107	Pseudogene	no
<i>FCGR1A</i>	NP_000557	NM_000566	Q246X	chr1	148,028,312	148,028,512	chr1	147,643,338	147,643,583	384,729	Pseudogene	no
<i>FCGR2B</i>	NP_003992	NM_004001	Q57X	chr1	159,907,743	159,907,943	chr1	159,825,975	159,826,241	81,502	Gene	no
<i>FCGR3A</i>	NP_000560	NM_000569	R270X	chr1	159,779,388	159,779,588	chr1	159,860,872	159,861,054	81,284	Gene	no
<i>FEM1A</i>	NP_061178	NM_018708	E37X	chr19	4,742,877	4,743,077	chr13	18,137,377	18,139,365	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	E37X	chr19	4,742,877	4,743,077	chr18	14,383,642	14,383,905	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	R50C	chr19	4,742,916	4,743,116	chr18	14,383,392	14,384,155	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	H128L	chr19	4,743,150	4,743,350	chr18	14,382,823	14,383,904	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	H128L	chr19	4,743,150	4,743,350	chr21	14,056,280	14,058,771	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	T152M	chr19	4,743,222	4,743,422	chr21	14,056,280	14,058,771	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	R178W	chr19	4,743,300	4,743,500	chr18	14,382,823	14,383,904	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	R178W	chr19	4,743,300	4,743,500	chr21	14,056,280	14,058,771	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	G194S	chr19	4,743,348	4,743,548	chr18	14,382,823	14,383,904	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	G194S	chr19	4,743,348	4,743,548	chr21	14,056,280	14,058,771	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	G213S	chr19	4,743,405	4,743,605	chr18	14,382,823	14,383,904	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	S223R	chr19	4,743,435	4,743,635	chr13	18,137,127	18,139,615	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	R294W	chr19	4,743,648	4,743,848	chr18	14,382,057	14,383,414	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	E302K	chr19	4,743,672	4,743,872	chr18	14,382,057	14,383,414	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	P339L	chr19	4,743,783	4,743,983	chr13	18,137,127	18,139,615	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	P339L	chr19	4,743,783	4,743,983	chr18	14,382,057	14,383,414	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	Q343X	chr19	4,743,795	4,743,995	chr18	14,382,307	14,383,164	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	P367A	chr19	4,743,867	4,744,067	chr21	14,056,280	14,058,771	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	T389S	chr19	4,743,933	4,744,133	chr13	18,137,127	18,139,615	na	Pseudogene	no
<i>FEM1A</i>	NP_061178	NM_018708	L464F	chr19	4,744,158	4,744,358	chr13	18,137,127	18,139,615	na	Pseudogene	no
<i>FEN1</i>	NP_004102	NM_004111	V82M	chr11	61,319,555	61,319,755	chr1	91,566,276	91,567,090	na	Pseudogene	no
<i>FEN1</i>	NP_004102	NM_004111	R100C	chr11	61,319,609	61,319,809	chr1	91,566,276	91,567,090	na	Pseudogene	no
<i>FEN1</i>	NP_004102	NM_004111	R129W	chr11	61,319,696	61,319,896	chr1	91,566,643	91,567,424	na	Pseudogene	no

<i>FEN1</i>	NP_004102	NM_004111	A174P	chr11	61,319,831	61,320,031	chr1	91,566,643	91,567,424	na	Pseudogene	no
<i>FEN1</i>	NP_004102	NM_004111	L183F	chr11	61,319,858	61,320,058	chr1	91,566,643	91,567,424	na	Pseudogene	no
<i>FEN1</i>	NP_004102	NM_004111	R192Q	chr11	61,319,885	61,320,085	chr1	91,566,643	91,567,424	na	Pseudogene	no
<i>FEN1</i>	NP_004102	NM_004111	G240S	chr11	61,320,029	61,320,229	chr1	91,566,926	91,567,911	na	Pseudogene	no
<i>FEN1</i>	NP_004102	NM_004111	Q333X	chr11	61,320,308	61,320,508	chr1	91,567,176	91,567,661	na	Pseudogene	no
<i>FH</i>	NP_000134	NM_000143	K337T	chr1	239,733,961	239,734,161	chr13	41,727,734	41,728,917	na	Pseudogene	no
<i>FKBP6</i>	NP_003593	NM_003602	Y61X	chr7	72,381,206	72,381,406	chr7	72,079,190	72,079,291	301,915	Pseudogene	yes
<i>FKBP6</i>	NP_003593	NM_003602	S62Y	chr7	72,381,209	72,381,409	chr7	72,078,940	72,079,541	301,668	Pseudogene	no
<i>FKBP6</i>	NP_003593	NM_003602	S62Y	chr7	72,381,209	72,381,409	chr7	74,861,228	74,861,829	2,479,819	Gene	no
<i>FLNB</i>	NP_001448	NM_001457	T140M	chr3	58,037,840	58,038,040	chr12	50,580,484	50,581,487	na	Pseudogene	no
<i>FLNB</i>	NP_001448	NM_001457	P141R	chr3	58,037,843	58,038,043	chr12	50,580,484	50,581,487	na	Pseudogene	no
<i>FLNC</i>	NP_001449	NM_001458	S2546T	chr7	128,284,384	128,284,584	chr7	128,340,863	128,341,584	56,279	Pseudogene	no
<i>FLNC</i>	NP_001449	NM_001458	Y2563D	chr7	128,284,435	128,284,635	chr7	128,340,863	128,341,584	56,228	Pseudogene	no
<i>FOLH1</i>	NP_004467	NM_004476	Y176H	chr11	49,164,782	49,164,982	chr11	89,031,179	89,031,816	39,866,197	Pseudogene	no
<i>FOXD4</i>	NP_997188	NM_207305	D285A	chr9	107,164	107,364	chr9	68,490,097	68,491,682	68,382,733	Gene	no
<i>FOXD4</i>	NP_997188	NM_207305	D285A	chr9	107,164	107,364	chr9	70,107,438	70,109,023	70,000,074	Gene	no
<i>FOXD4</i>	NP_997188	NM_207305	P272R	chr9	107,203	107,403	chr9	42,707,812	42,709,397	42,600,409	Gene	no
<i>FOXD4</i>	NP_997188	NM_207305	P272R	chr9	107,203	107,403	chr9	69,667,384	69,668,969	69,559,981	Gene	no
<i>FREM2</i>	NP_997244	NM_207361	R2278H	chr13	38,323,814	38,324,014	chr13	20,765,305	20,765,930	17,557,884	Pseudogene	no
<i>FTL</i>	NP_000137	NM_000146	Y165X	chr19	54,161,671	54,161,871	chrX	30,558,336	30,558,860	na	Pseudogene	no
<i>GLDC</i>	NP_000161	NM_000170	S1010L	chr9	6,522,949	6,523,149	chr4	57,156,221	57,156,891	na	Pseudogene	no
<i>GLUL</i>	NP_002056	NM_002065	R340C	chr1	180,620,164	180,620,364	chr5	151,210,018	151,210,724	na	Pseudogene	no
<i>GLUL</i>	NP_002056	NM_002065	S322N	chr1	180,620,218	180,620,418	chr5	151,210,018	151,210,724	na	Pseudogene	no
<i>GPX1</i>	NP_000572	NM_000581	R177C	chr3	49,369,805	49,370,005	chr21	27,437,287	27,438,059	na	Pseudogene	no
<i>GPX1</i>	NP_000572	NM_000581	W162X	chr3	49,369,850	49,370,050	chrX	13,306,781	13,307,347	na	Pseudogene	no
<i>GPX1</i>	NP_000572	NM_000581	F126S	chr3	49,369,958	49,370,158	chr21	27,437,502	27,438,196	na	Pseudogene	no
<i>GPX1</i>	NP_000572	NM_000581	P124T	chr3	49,369,964	49,370,164	chrX	13,306,531	13,307,597	na	Pseudogene	no
<i>GPX1</i>	NP_000572	NM_000581	U49X	chr3	49,370,468	49,370,668	chrX	13,306,781	13,307,347	na	Pseudogene	no
<i>GSTA1</i>	NP_665683	NM_145740	R20W	chr6	52,771,864	52,772,064	chr6	52,842,750	52,843,336	70,686	Pseudogene	no
<i>GSTA2</i>	NP_000837	NM_000846	G83R	chr6	52,727,622	52,727,822	chr6	52,830,775	52,831,430	102,953	Pseudogene	no
<i>GSTA2</i>	NP_000837	NM_000846	G83E	chr6	52,727,622	52,727,822	chr6	52,851,260	52,851,915	123,438	Pseudogene	no
<i>GSTA2</i>	NP_000837	NM_000846	R20W	chr6	52,730,544	52,730,744	chr6	52,842,750	52,843,336	112,006	Pseudogene	no
<i>GUSB</i>	NP_000172	NM_000181	F589S	chr7	65,066,666	65,066,866	chr7	63,217,949	63,218,610	1,848,056	Pseudogene	no

<i>GUSB</i>	NP_000172	NM_000181	W587G	chr7	65,066,672	65,066,872	chr7	63,217,949	63,218,610	1,848,062	Pseudogene	no
<i>GUSB</i>	NP_000172	NM_000181	R577L	chr7	65,066,702	65,066,902	chr22	22,331,800	22,332,440	na	Pseudogene	yes
<i>GUSB</i>	NP_000172	NM_000181	L566P	chr7	65,066,735	65,066,935	chr7	63,217,949	63,218,610	1,848,125	Pseudogene	no
<i>GUSB</i>	NP_000172	NM_000181	Y561D	chr7	65,066,750	65,066,950	chr5	69,252,062	69,252,702	na	Pseudogene	no
<i>GUSB</i>	NP_000172	NM_000181	Y561D	chr7	65,066,750	65,066,950	chr5	69,528,586	69,529,226	na	Pseudogene	no
<i>GUSB</i>	NP_000172	NM_000181	Y561D	chr7	65,066,750	65,066,950	chr5	70,127,111	70,127,751	na	Pseudogene	no
<i>GUSB</i>	NP_000172	NM_000181	Y561D	chr7	65,066,750	65,066,950	chr5	70,528,977	70,529,617	na	Pseudogene	no
<i>GUSB</i>	NP_000172	NM_000181	Y561D	chr7	65,066,750	65,066,950	chr8	869,354	869,994	na	Pseudogene	no
<i>GUSB</i>	NP_000172	NM_000181	E560D	chr7	65,066,753	65,066,953	chr22	22,331,800	22,332,440	na	Pseudogene	no
<i>GUSB</i>	NP_000172	NM_000181	N339S	chr7	65,077,288	65,077,488	chr5	98,897,184	98,897,803	na	Pseudogene	yes
<i>HADHA</i>	NP_000173	NM_000182	A754T	chr2	26,267,552	26,267,752	chr4	166,544,420	166,547,007	na	Pseudogene	no
<i>HADHA</i>	NP_000173	NM_000182	Q748X	chr2	26,267,570	26,267,770	chr12	8,680,730	8,681,473	na	Pseudogene	no
<i>HCN2</i>	NP_001185	NM_001194	P173L	chr19	541,364	541,564	chr15	28,302,302	28,305,306	na	Pseudogene	no
<i>HCN2</i>	NP_001185	NM_001194	P173L	chr19	541,364	541,564	chr15	30,392,371	30,395,045	na	Pseudogene	no
<i>HCN2</i>	NP_001185	NM_001194	P279Q	chr19	554,648	554,848	chr15	28,302,302	28,305,306	na	Pseudogene	no
<i>HCN2</i>	NP_001185	NM_001194	P279Q	chr19	554,648	554,848	chr15	30,392,371	30,395,045	na	Pseudogene	no
<i>HK2</i>	NP_000180	NM_000189	R381H	chr2	74,959,334	74,959,534	chrX	79,714,482	79,717,012	na	Pseudogene	no
<i>HK2</i>	NP_000180	NM_000189	A908F	chr2	74,971,446	74,971,646	chrX	79,713,779	79,714,980	na	Pseudogene	no
<i>HLA-B</i>	NP_005505	NM_005514	T162M	chr6	31,431,955	31,432,155	chr6	29,963,972	29,964,747	1,467,208	Gene	no
<i>HLA-B</i>	NP_005505	NM_005514	V36M	chr6	31,432,578	31,432,778	chr6	29,963,472	29,964,232	1,468,346	Gene	no
<i>HLA-C</i>	NP_002108	NM_002117	V100E	chr6	31,347,297	31,347,497	chr6	31,432,192	31,432,952	84,695	Gene	no
<i>HLA-C</i>	NP_002108	NM_002117	G69E	chr6	31,347,390	31,347,590	chr6	31,432,192	31,432,952	84,602	Gene	no
<i>HLA-DPB1</i>	NP_002112	NM_002121	D196N	chr6	33,160,828	33,161,028	chr6	33,203,345	33,204,165	42,317	Pseudogene	no
<i>HLA-DRB1</i>	NP_002115	NM_002124	T174I	chr6	32,657,341	32,657,541	chr6	32,630,196	32,630,977	26,364	Pseudogene	no
<i>HLA-DRB1</i>	NP_002115	NM_002124	G170A	chr6	32,657,353	32,657,553	chr6	32,630,196	32,630,977	26,376	Pseudogene	no
<i>HLA-DRB5</i>	NP_002116	NM_002125	T174I	chr6	32,595,154	32,595,354	chr6	32,630,187	32,630,977	34,833	Pseudogene	no
<i>HLA-DRB5</i>	NP_002116	NM_002125	G170A	chr6	32,595,166	32,595,366	chr6	32,630,187	32,630,977	34,821	Pseudogene	no
<i>HLA-E</i>	NP_005507	NM_005516	T235I	chr6	30,566,887	30,567,087	chr6	29,904,911	29,905,698	661,189	Gene	no
<i>HLA-E</i>	NP_005507	NM_005516	A266V	chr6	30,566,980	30,567,180	chr6	30,336,697	30,337,484	229,496	Gene	no
<i>HSD3B1</i>	NP_000853	NM_000862	D87Y	chr1	119,855,664	119,855,864	chr1	119,911,783	119,912,423	55,919	Pseudogene	no
<i>HSD3B1</i>	NP_000853	NM_000862	W356X	chr1	119,858,637	119,858,837	chr1	119,789,423	119,790,232	68,405	Pseudogene	no
<i>HSP90AA1</i>	NP_005339	NM_005348	N655Y	chr14	101,618,224	101,618,424	chr1	156,761,415	156,762,499	na	Pseudogene	no
<i>HSP90AA1</i>	NP_005339	NM_005348	A618V	chr14	101,618,335	101,618,535	chr1	156,761,415	156,762,499	na	Pseudogene	no

HSPA5	NP_005338	NM_005347	I602T	chr9	127,038,750	127,038,950	chr1	38,948,314	38,949,398	na	Pseudogene	no
HSPA5	NP_005338	NM_005347	R532H	chr9	127,038,960	127,039,160	chr1	38,948,314	38,949,398	na	Pseudogene	no
HSPA5	NP_005338	NM_005347	G519R	chr9	127,038,999	127,039,199	chr1	38,948,314	38,949,398	na	Pseudogene	no
HSPA5	NP_005338	NM_005347	E514K	chr9	127,039,014	127,039,214	chr1	38,948,314	38,949,398	na	Pseudogene	no
HSPA5	NP_005338	NM_005347	G507D	chr9	127,039,035	127,039,235	chr1	38,948,314	38,949,398	na	Pseudogene	no
HSPA5	NP_005338	NM_005347	I503M	chr9	127,039,047	127,039,247	chr1	38,948,314	38,949,398	na	Pseudogene	no
HSPA5	NP_005338	NM_005347	K296R	chr9	127,041,048	127,041,248	chr4	165,329,374	165,330,578	na	Pseudogene	no
HSPA5	NP_005338	NM_005347	E291K	chr9	127,041,063	127,041,263	chr1	38,946,996	38,948,461	na	Pseudogene	no
HSPA5	NP_005338	NM_005347	R289W	chr9	127,041,069	127,041,269	chr1	38,946,996	38,948,461	na	Pseudogene	no
HSPA5	NP_005338	NM_005347	H252Y	chr9	127,041,180	127,041,380	chr1	38,946,996	38,948,461	na	Pseudogene	no
HSPA5	NP_005338	NM_005347	E243K	chr9	127,041,207	127,041,407	chr1	38,946,996	38,948,461	na	Pseudogene	no
HSPA5	NP_005338	NM_005347	R74C	chr9	127,042,807	127,043,007	chr1	38,946,996	38,948,461	na	Pseudogene	no
HSPA8	NP_006588	NM_006597	G407D	chr11	122,434,978	122,435,178	chr3	139,082,608	139,083,458	na	Pseudogene	no
HSPA8	NP_006588	NM_006597	G407S	chr11	122,434,978	122,435,178	chr7	10,458,332	10,459,650	na	Pseudogene	no
HSPA8	NP_006588	NM_006597	V337F	chr11	122,435,399	122,435,599	chr1	245,460,326	245,461,443	na	Pseudogene	no
HSPA8	NP_006588	NM_006597	G312S	chr11	122,435,474	122,435,674	chrX	120,164,695	120,166,121	na	Pseudogene	no
HSPA8	NP_006588	NM_006597	D292N	chr11	122,435,534	122,435,734	chr12	4,077,601	4,078,484	na	Pseudogene	no
HSPA8	NP_006588	NM_006597	I282T	chr11	122,435,564	122,435,764	chrX	120,164,695	120,166,121	na	Pseudogene	no
HSPA8	NP_006588	NM_006597	L274V	chr11	122,435,588	122,435,788	chrX	120,164,695	120,166,121	na	Pseudogene	no
HSPA8	NP_006588	NM_006597	F245Y	chr11	122,435,675	122,435,875	chrX	120,164,695	120,166,121	na	Pseudogene	no
HSPA8	NP_006588	NM_006597	G224R	chr11	122,435,738	122,435,938	chr7	10,457,215	10,458,836	na	Pseudogene	no
HSPA8	NP_006588	NM_006597	G224R	chr11	122,435,738	122,435,938	chrX	120,165,618	120,166,345	na	Pseudogene	no
HSPD1	NP_002147	NM_002156	G110V	chr2	198,070,105	198,070,305	chr4	145,986,174	145,987,660	na	Pseudogene	no
HSPD1	NP_002147	NM_002156	A109S	chr2	198,070,108	198,070,308	chr5	21,918,336	21,920,374	na	Pseudogene	no
HSPD1	NP_002147	NM_002156	G94E	chr2	198,070,153	198,070,353	chr3	36,784,139	36,785,544	na	Pseudogene	no
IFNA17	NP_067091	NM_021268	K157R	chr9	21,217,601	21,217,801	chr9	21,196,280	21,197,346	20,255	Gene	no
IMPDH1	NP_000874	NM_000883	P228L	chr7	127,827,624	127,827,824	chr16	10,112,487	10,113,604	na	Pseudogene	no
IMPDH1	NP_000874	NM_000883	G226C	chr7	127,827,630	127,827,830	chr16	10,112,487	10,113,604	na	Pseudogene	no
KAL1	NP_000207	NM_000216	K329N	chrX	8,498,514	8,498,714	chrY	14,508,281	14,508,987	na	Pseudogene	no
KAL1	NP_000207	NM_000216	C172Y	chrX	8,524,999	8,525,199	chrY	14,479,526	14,480,247	na	Pseudogene	no
KAL1	NP_000207	NM_000216	H170P	chrX	8,525,005	8,525,205	chrY	14,479,526	14,480,247	na	Pseudogene	no
KAL1	NP_000207	NM_000216	S165L	chrX	8,525,020	8,525,220	chrY	14,479,526	14,480,247	na	Pseudogene	no
KAL1	NP_000207	NM_000216	L117F	chrX	8,525,164	8,525,364	chrY	14,479,526	14,480,247	na	Pseudogene	no

<i>KIR2DL1</i>	NP_055033	NM_014218	F202L	chr19	59,978,564	59,978,764	chr19	231,900	232,714	na	Gene	no
<i>KIR2DL1</i>	NP_055033	NM_014218	V344A	chr19	59,986,962	59,987,162	chr19	254,875	255,551	na	Gene	no
<i>KIR3DL1</i>	NP_037421	NM_013289	V440A	chr19	60,033,427	60,033,627	chr19	254,875	255,551	na	Gene	no
<i>KRT14</i>	NP_000517	NM_000526	R388C	chr17	36,993,022	36,993,222	chr17	16,662,730	16,663,457	20,329,565	Pseudogene	yes
<i>KRT14</i>	NP_000517	NM_000526	E312K	chr17	36,993,345	36,993,545	chr17	16,663,047	16,663,681	20,329,664	Pseudogene	no
<i>KRT14</i>	NP_000517	NM_000526	W305R	chr17	36,993,449	36,993,649	chr17	16,663,253	16,663,920	20,329,529	Pseudogene	no
<i>KRT14</i>	NP_000517	NM_000526	R282H	chr17	36,993,518	36,993,718	chr17	16,663,253	16,663,920	20,329,598	Pseudogene	no
<i>KRT14</i>	NP_000517	NM_000526	R153W	chr17	36,996,053	36,996,253	chr17	20,337,286	20,338,310	16,657,743	Pseudogene	no
<i>KRT14</i>	NP_000517	NM_000526	R153W	chr17	36,996,053	36,996,253	chr17	18,294,033	18,295,057	18,700,996	Pseudogene	no
<i>KRT14</i>	NP_000517	NM_000526	R153W	chr17	36,996,053	36,996,253	chr17	16,665,757	16,666,781	20,329,272	Pseudogene	no
<i>KRT14</i>	NP_000517	NM_000526	V133A	chr17	36,996,113	36,996,313	chr17	18,294,033	18,295,057	18,701,056	Pseudogene	yes
<i>KRT16</i>	NP_005548	NM_005557	R419C	chr17	37,020,031	37,020,231	chr17	20,345,722	20,346,440	16,673,591	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	R419H	chr17	37,020,031	37,020,231	chr17	18,285,912	18,286,630	18,733,401	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	R390C	chr17	37,020,118	37,020,318	chr17	18,285,912	18,286,630	18,733,488	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	R390C	chr17	37,020,118	37,020,318	chr17	16,674,120	16,674,904	20,345,214	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	L372P	chr17	37,020,172	37,020,372	chr17	16,674,120	16,674,904	20,345,268	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	Y368H	chr17	37,020,184	37,020,384	chr17	20,345,722	20,346,440	16,673,744	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	Y368H	chr17	37,020,184	37,020,384	chr17	18,285,912	18,286,630	18,733,554	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	R367C	chr17	37,020,187	37,020,387	chr17	20,345,722	20,346,440	16,673,747	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	R367C	chr17	37,020,187	37,020,387	chr17	18,285,912	18,286,630	18,733,557	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	R367C	chr17	37,020,187	37,020,387	chr17	16,674,120	16,674,904	20,345,283	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	E335L	chr17	37,020,585	37,020,785	chr17	16,674,707	16,675,344	20,345,241	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	E318K	chr17	37,020,636	37,020,836	chr17	20,346,243	20,346,880	16,673,756	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	E318K	chr17	37,020,636	37,020,836	chr17	18,285,472	18,286,109	18,734,527	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	E313K	chr17	37,020,651	37,020,851	chr17	20,346,243	20,346,880	16,673,771	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	E313K	chr17	37,020,651	37,020,851	chr17	18,285,472	18,286,109	18,734,542	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	E313K	chr17	37,020,651	37,020,851	chr17	16,674,707	16,675,344	20,345,307	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	K311Q	chr17	37,020,746	37,020,946	chr17	20,346,243	20,346,880	16,673,866	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	K311Q	chr17	37,020,746	37,020,946	chr17	18,285,472	18,286,109	18,734,637	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	K311Q	chr17	37,020,746	37,020,946	chr17	16,674,707	16,675,344	20,345,402	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	Y164C	chr17	37,021,874	37,022,074	chr17	16,676,007	16,676,707	20,345,167	Pseudogene	no
<i>KRT16</i>	NP_005548	NM_005557	V135A	chr17	37,021,961	37,022,161	chr17	18,294,033	18,295,087	18,726,874	Pseudogene	no
<i>KRT18</i>	NP_000215	NM_000224	G76D	chr12	51,629,352	51,629,552	chr2	189,884,687	189,885,726	na	Pseudogene	no

KRT18	NP_000215	NM_000224	E82D	chr12	51,629,370	51,629,570	chr2	189,884,687	189,885,726	na	Pseudogene	no
KRT18	NP_000215	NM_000224	Q85K	chr12	51,629,379	51,629,579	chr9	80,841,396	80,842,684	na	Pseudogene	no
KRT18	NP_000215	NM_000224	N88K	chr12	51,629,388	51,629,588	chr5	122,999,453	123,001,248	na	Pseudogene	no
KRT18	NP_000215	NM_000224	V98A	chr12	51,629,418	51,629,618	chr22	19,167,777	19,168,642	na	Pseudogene	no
KRT18	NP_000215	NM_000224	V98E	chr12	51,629,418	51,629,618	chr4	117,061,062	117,062,830	na	Pseudogene	no
KRT18	NP_000215	NM_000224	V98E	chr12	51,629,418	51,629,618	chrY	5,501,082	5,502,514	na	Pseudogene	no
KRT18	NP_000215	NM_000224	L101Q	chr12	51,629,427	51,629,627	chr5	169,500,389	169,501,770	na	Pseudogene	no
KRT18	NP_000215	NM_000224	L101M	chr12	51,629,427	51,629,627	chr6	29,045,504	29,046,420	na	Pseudogene	no
KRT18	NP_000215	NM_000224	Q357X	chr12	51,632,192	51,632,392	chr16	71,320,730	71,321,299	na	Pseudogene	no
KRT18	NP_000215	NM_000224	Q357X	chr12	51,632,192	51,632,392	chr6	29,044,932	29,045,471	na	Pseudogene	no
KRT8	NP_002264	NM_002273	V462A	chr12	51,577,444	51,577,644	chr7	38,662,315	38,662,985	na	Pseudogene	no
KRT8	NP_002264	NM_002273	A363V	chr12	51,578,742	51,578,942	chr10	8,595,708	8,597,350	na	Pseudogene	no
KRT8	NP_002264	NM_002273	L360Q	chr12	51,578,751	51,578,951	chr11	118,978,535	118,979,721	na	Pseudogene	no
KRT8	NP_002264	NM_002273	Y286D	chr12	51,579,848	51,580,048	chr5	122,764,624	122,765,597	na	Pseudogene	no
KRT8	NP_002264	NM_002273	S274R	chr12	51,579,884	51,580,084	chr8	62,653,083	62,655,031	na	Pseudogene	no
KRT8	NP_002264	NM_002273	Y267X	chr12	51,579,905	51,580,105	chr3	171,695,924	171,697,114	na	Pseudogene	no
KRT8	NP_002264	NM_002273	Q266P	chr12	51,579,908	51,580,108	chr4	103,869,098	103,870,035	na	Pseudogene	no
KRT8	NP_002264	NM_002273	E141Q	chr12	51,581,924	51,582,124	chr15	68,937,856	68,939,741	na	Pseudogene	no
KRT8	NP_002264	NM_002273	E141Q	chr12	51,581,924	51,582,124	chr4	103,869,529	103,870,622	na	Pseudogene	no
KRT8	NP_002264	NM_002273	E141K	chr12	51,581,924	51,582,124	chr5	122,765,087	122,766,171	na	Pseudogene	no
KRT8	NP_002264	NM_002273	E141K	chr12	51,581,924	51,582,124	chr6	162,909,263	162,910,452	na	Pseudogene	no
KRT8	NP_002264	NM_002273	R40X	chr12	51,584,812	51,585,012	chr8	62,653,333	62,654,781	na	Pseudogene	no
KRT81	NP_002272	NM_002281	E160A	chr12	50,970,126	50,970,326	chr12	51,024,019	51,024,731	53,693	Pseudogene	no
KRT81	NP_002272	NM_002281	T79A	chr12	50,971,179	50,971,379	chr12	50,937,953	50,938,818	32,361	Pseudogene	no
LDHB	NP_002291	NM_002300	L324P	chr12	21,679,675	21,679,875	chrX	75,471,415	75,472,898	na	Pseudogene	no
LDHB	NP_002291	NM_002300	L317F	chr12	21,679,696	21,679,896	chrX	75,471,415	75,472,898	na	Pseudogene	no
LEFTY2	NP_003231	NM_003240	P301L	chr1	224,191,861	224,192,061	chr1	224,057,223	224,058,025	133,836	Pseudogene	no
LEFTY2	NP_003231	NM_003240	E292K	chr1	224,191,888	224,192,088	chr1	224,057,223	224,058,025	133,863	Pseudogene	no
LEFTY2	NP_003231	NM_003240	P286L	chr1	224,191,906	224,192,106	chr1	224,057,223	224,058,025	133,881	Pseudogene	no
LEFTY2	NP_003231	NM_003240	R265H	chr1	224,191,969	224,192,169	chr1	224,057,223	224,058,025	133,944	Pseudogene	no
LRP5	NP_002326	NM_002335	R494L	chr11	67,913,894	67,914,094	chr22	24,082,969	24,083,678	na	Gene	no
LYST	NP_000072	NM_000081	Q2694X	chr1	233,973,870	233,974,070	chr13	31,571,759	31,571,950	na	Pseudogene	no
MAD1L1	NP_003541	NM_003550	R714L	chr7	1,822,146	1,822,346	chr22	22,132,260	22,132,945	na	Pseudogene	no

<i>MAD2L1</i>	NP_002349	NM_002358	R184C	chr4	121,200,686	121,200,886	chr14	60,316,199	60,317,313	na	Pseudogene	no
<i>MAD2L1</i>	NP_002349	NM_002358	R182C	chr4	121,200,692	121,200,892	chr14	60,316,199	60,317,313	na	Pseudogene	no
<i>MATR3</i>	NP_954659	NM_199189	L55X	chr5	138,671,068	138,671,268	chr1	106,236,008	106,236,436	na	Pseudogene	no
<i>MATR3</i>	NP_954659	NM_199189	G162D	chr5	138,671,389	138,671,589	chr1	106,235,758	106,236,686	na	Pseudogene	no
<i>MATR3</i>	NP_954659	NM_199189	R167X	chr5	138,671,404	138,671,604	chr1	106,236,008	106,236,436	na	Pseudogene	no
<i>MICB</i>	NP_005922	NM_005931	K67G	chr6	31,581,403	31,581,603	chr6	29,820,977	29,821,728	1,759,675	Pseudogene	no
<i>MST1</i>	NP_066278	NM_020998	R108H	chr3	49,699,923	49,700,123	chr1	16,959,594	16,960,435	na	Pseudogene	no
<i>MST1</i>	NP_066278	NM_020998	L37S	chr3	49,700,217	49,700,417	chr1	16,959,594	16,960,435	na	Pseudogene	no
<i>MYO5B</i>	NP_001073936	NM_001080467	E1655X	chr18	45,617,957	45,618,157	chr9	67,847,098	67,848,351	na	Pseudogene	no
<i>MYO5B</i>	NP_001092	NM_001101	Y362X	chr7	5,533,846	5,534,046	chr6	89,042,323	89,042,838	na	Pseudogene	no
<i>NDUFAF2</i>	NP_777549	NM_174889	H132R	chr5	60,484,325	60,484,525	chr2	20,592,461	20,593,467	na	Pseudogene	no
<i>NEFM</i>	NP_005373	NM_005382	K902N	chr8	24,831,879	24,832,079	chr10	56,030,044	56,030,696	na	Pseudogene	no
<i>NOS2</i>	NP_000616	NM_000625	G1083W	chr17	23,110,038	23,110,238	chr17	23,004,453	23,005,150	104,888	Pseudogene	no
<i>NOS2</i>	NP_000616	NM_000625	Q899L	chr17	23,113,953	23,114,153	chr17	20,284,679	20,285,400	2,828,553	Pseudogene	no
<i>NOS2</i>	NP_000616	NM_000625	Q899L	chr17	23,113,953	23,114,153	chr17	18,346,853	18,347,574	4,766,379	Pseudogene	no
<i>NOS2</i>	NP_000616	NM_000625	Q899L	chr17	23,113,953	23,114,153	chr17	16,656,230	16,656,951	6,457,002	Pseudogene	no
<i>NOS2</i>	NP_000616	NM_000625	A680V	chr17	23,118,884	23,119,084	chr17	31,600,876	31,601,540	8,481,792	Pseudogene	no
<i>NOS2</i>	NP_000616	NM_000625	A680V	chr17	23,118,884	23,119,084	chr17	31,815,910	31,816,574	8,696,826	Pseudogene	no
<i>NOS2</i>	NP_000616	NM_000625	A680V	chr17	23,118,884	23,119,084	chr17	31,866,962	31,867,626	8,747,878	Pseudogene	no
<i>NOS2</i>	NP_000616	NM_000625	A680V	chr17	23,118,884	23,119,084	chr17	33,587,195	33,587,859	10,468,111	Pseudogene	no
<i>NOS2</i>	NP_000616	NM_000625	A680V	chr17	23,118,884	23,119,084	chr17	57,692,527	57,693,191	34,573,443	Pseudogene	no
<i>NOS2</i>	NP_000616	NM_000625	A680V	chr17	23,118,884	23,119,084	chr17	989,626	990,290	na	Pseudogene	no
<i>NR3C1</i>	NP_001018087	NM_001018077	F306S	chr5	142,759,579	142,759,779	chr16	86,057,993	86,059,011	na	Pseudogene	no
<i>NR3C1</i>	NP_001018087	NM_001018077	E285Q	chr5	142,759,642	142,759,842	chr16	86,057,993	86,059,011	na	Pseudogene	no
<i>NR3C1</i>	NP_001018087	NM_001018077	V276A	chr5	142,759,669	142,759,869	chr16	86,057,993	86,059,011	na	Pseudogene	no
<i>OR5H6</i>	NP_001005479	NM_001005479	Y51X	chr3	99,465,871	99,466,071	chr3	99,439,838	99,440,812	25,059	Pseudogene	no
<i>PDSS1</i>	NP_055132	NM_014317	Q162H	chr10	27,049,071	27,049,271	chr2	44,247,175	44,247,836	na	Pseudogene	no
<i>PEX12</i>	NP_000277	NM_000286	R154X	chr17	30,928,287	30,928,487	chr13	89,442,487	89,443,059	na	Pseudogene	no
<i>PGAM1</i>	NP_002620	NM_002629	R90Q	chr10	99,180,156	99,180,356	chr2	23,948,645	23,949,537	na	Pseudogene	no
<i>PGAM1</i>	NP_002620	NM_002629	R90Q	chr10	99,180,156	99,180,356	chr6	73,821,291	73,822,468	na	Pseudogene	no
<i>PGAM1</i>	NP_002620	NM_002629	T96I	chr10	99,180,174	99,180,374	chr3	9,364,877	9,366,123	na	Pseudogene	no
<i>PGAM1</i>	NP_002620	NM_002629	G97D	chr10	99,180,177	99,180,377	chr2	23,948,645	23,949,537	na	Pseudogene	no
<i>PGAM1</i>	NP_002620	NM_002629	N99S	chr10	99,180,183	99,180,383	chr2	23,948,645	23,949,537	na	Pseudogene	no

PGAM1	NP_002620	NM_002629	T238M	chr10	99,182,120	99,182,320	chr11	64,698,835	64,699,784	na	Pseudogene	no
PKP2	NP_004563	NM_004572	R212H	chr12	32,922,344	32,922,544	chr12	6,457,008	6,458,035	26,464,309	Pseudogene	no
PKP2	NP_004563	NM_004572	A195D	chr12	32,922,395	32,922,595	chr12	6,457,008	6,458,035	26,464,360	Pseudogene	no
PLA2G10	NP_003552	NM_003561	C100S	chr16	14,689,477	14,689,677	chr18	11,607,948	11,608,570	na	Pseudogene	no
PLA2G10	NP_003552	NM_003561	C90X	chr16	14,689,507	14,689,707	chr16	21,319,902	21,320,024	6,630,195	Pseudogene	no
PLA2G10	NP_003552	NM_003561	C90X	chr16	14,689,507	14,689,707	chr16	21,752,330	21,752,452	7,062,623	Pseudogene	no
PLA2G10	NP_003552	NM_003561	C90X	chr16	14,689,507	14,689,707	chr16	22,456,292	22,456,414	7,766,585	Pseudogene	no
PLA2G10	NP_003552	NM_003561	C67S	chr16	14,691,799	14,691,999	chr16	29,397,451	29,398,121	14,705,452	Pseudogene	no
PLA2G10	NP_003552	NM_003561	C67S	chr16	14,691,799	14,691,999	chr16	30,136,792	30,137,462	15,444,793	Pseudogene	no
PLA2G10	NP_003552	NM_003561	R57X	chr16	14,691,829	14,692,029	chr16	15,360,336	15,360,506	668,307	Pseudogene	no
PLA2G10	NP_003552	NM_003561	R57X	chr16	14,691,829	14,692,029	chr16	16,399,783	16,399,953	1,707,754	Pseudogene	no
PLA2G10	NP_003552	NM_003561	R57X	chr16	14,691,829	14,692,029	chr16	18,314,655	18,314,825	3,622,626	Pseudogene	no
PLA2G10	NP_003552	NM_003561	R57X	chr16	14,691,829	14,692,029	chr16	29,397,701	29,397,871	14,705,672	Pseudogene	no
PLA2G10	NP_003552	NM_003561	R57X	chr16	14,691,829	14,692,029	chr16	30,137,042	30,137,212	15,445,013	Pseudogene	no
PLA2G10	NP_003552	NM_003561	G55S	chr16	14,691,835	14,692,035	chr16	29,397,451	29,398,121	14,705,416	Pseudogene	no
PLA2G10	NP_003552	NM_003561	G55S	chr16	14,691,835	14,692,035	chr16	30,136,792	30,137,462	15,444,757	Pseudogene	no
PLA2G10	NP_003552	NM_003561	T50I	chr16	14,691,850	14,692,050	chr16	15,360,086	15,360,756	668,036	Pseudogene	no
PLA2G10	NP_003552	NM_003561	T50I	chr16	14,691,850	14,692,050	chr16	16,399,533	16,400,203	1,707,483	Pseudogene	no
PLA2G10	NP_003552	NM_003561	T50I	chr16	14,691,850	14,692,050	chr16	29,397,451	29,398,121	14,705,401	Pseudogene	no
PLA2G10	NP_003552	NM_003561	T50I	chr16	14,691,850	14,692,050	chr16	30,136,792	30,137,462	15,444,742	Pseudogene	no
PLEKHM1	NP_055613	NM_014798	F1013S	chr17	40,872,545	40,872,745	chr17	60,212,890	60,213,557	19,340,145	Gene	no
PLEKHM1	NP_055613	NM_014798	G998S	chr17	40,872,590	40,872,790	chr17	60,212,890	60,213,557	19,340,100	Gene	no
PLEKHM1	NP_055613	NM_014798	G971E	chr17	40,872,671	40,872,871	chr17	60,212,890	60,213,557	19,340,019	Gene	no
PLEKHM1	NP_055613	NM_014798	S706P	chr17	40,886,782	40,886,982	chr17	60,226,328	60,227,748	19,339,346	Gene	no
PLEKHM1	NP_055613	NM_014798	R544Q	chr17	40,887,268	40,887,468	chr17	60,226,328	60,227,748	19,338,860	Gene	no
PLEKHM1	NP_055613	NM_014798	R533Q	chr17	40,887,301	40,887,501	chr17	60,226,328	60,227,748	19,338,827	Gene	no
PLEKHM1	NP_055613	NM_014798	S338X	chr17	40,901,551	40,901,751	chr17	60,241,443	60,241,793	19,339,692	Pseudogene	no
PLEKHM1	NP_055613	NM_014798	A64T	chr17	40,911,052	40,911,252	chr17	60,250,954	60,251,702	19,339,702	Pseudogene	no
PLG	NP_000292	NM_000301	T28A	chr6	161,047,363	161,047,563	chr2	106,368,917	106,369,560	na	Pseudogene	no
PLG	NP_000292	NM_000301	A31P	chr6	161,047,372	161,047,572	chr2	87,097,930	87,098,573	na	Gene	no
PLG	NP_000292	NM_000301	A31P	chr6	161,047,372	161,047,572	chr2	87,832,630	87,833,273	na	Gene	no
PLG	NP_000292	NM_000301	H133R	chr6	161,052,105	161,052,305	chr2	87,093,370	87,094,004	na	Pseudogene	no
PLG	NP_000292	NM_000301	H133R	chr6	161,052,105	161,052,305	chr2	87,837,203	87,837,837	na	Pseudogene	no

PLG	NP_000292	NM_000301	H133R	chr6	161,052,105	161,052,305	chr2	106,373,543	106,374,159	na	Pseudogene	no
PLG	NP_000292	NM_000301	G703D	chr6	161,082,323	161,082,523	chr6	161,017,168	161,017,790	64,533	Pseudogene	no
PMS2	NP_000526	NM_000535	N775S	chr7	5,983,764	5,983,964	chr7	6,752,941	6,753,614	768,977	Pseudogene	no
PMS2	NP_000526	NM_000535	Q160K	chr7	6,008,566	6,008,766	chr7	66,397,790	66,398,475	60,389,024	Pseudogene	no
PMS2	NP_000526	NM_000535	Q160K	chr7	6,008,566	6,008,766	chr7	74,977,939	74,978,624	68,969,173	Gene	no
PMS2	NP_000526	NM_000535	Q160K	chr7	6,008,566	6,008,766	chr7	75,991,518	75,992,203	69,982,752	Pseudogene	no
PMS2	NP_000526	NM_000535	Q160K	chr7	6,008,566	6,008,766	chr7	101,765,844	101,766,529	95,757,078	Pseudogene	no
PMS2	NP_000526	NM_000535	T155M	chr7	6,008,581	6,008,781	chr7	99,764,145	99,764,863	93,755,364	Pseudogene	no
PMS2	NP_000526	NM_000535	T155M	chr7	6,008,581	6,008,781	chr7	101,765,844	101,766,529	95,757,063	Pseudogene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	66,397,790	66,398,475	60,388,997	Pseudogene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	72,121,361	72,121,992	66,112,568	Pseudogene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	72,149,337	72,150,055	66,140,544	Gene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	73,951,419	73,952,137	67,942,626	Gene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	74,348,439	74,349,157	68,339,646	Pseudogene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	74,762,704	74,763,422	68,753,911	Gene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	74,790,726	74,791,444	68,781,933	Pseudogene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	74,818,743	74,819,461	68,809,950	Gene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	74,977,939	74,978,624	68,969,146	Gene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	75,991,518	75,992,203	69,982,725	Pseudogene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	76,482,640	76,483,358	70,473,847	Pseudogene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	76,510,563	76,511,281	70,501,770	Pseudogene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	99,764,145	99,764,863	93,755,352	Pseudogene	no
PMS2	NP_000526	NM_000535	R151H	chr7	6,008,593	6,008,793	chr7	101,765,844	101,766,529	95,757,051	Pseudogene	no
PMS2	NP_000526	NM_000535	S90C	chr7	6,009,829	6,010,029	chr7	101,764,463	101,765,061	95,754,434	Pseudogene	no
PMS2	NP_000526	NM_000535	D70G	chr7	6,010,068	6,010,268	chr7	72,119,672	72,120,264	66,109,404	Pseudogene	no
PMS2	NP_000526	NM_000535	D70G	chr7	6,010,068	6,010,268	chr7	72,147,648	72,148,240	66,137,380	Gene	no
PMS2	NP_000526	NM_000535	D70G	chr7	6,010,068	6,010,268	chr7	73,949,945	73,950,537	67,939,677	Gene	no
PMS2	NP_000526	NM_000535	D70G	chr7	6,010,068	6,010,268	chr7	74,350,215	74,350,807	68,339,947	Pseudogene	no
PMS2	NP_000526	NM_000535	D70G	chr7	6,010,068	6,010,268	chr7	74,764,495	74,765,087	68,754,227	Gene	no
PMS2	NP_000526	NM_000535	D70G	chr7	6,010,068	6,010,268	chr7	74,792,520	74,793,112	68,782,252	Pseudogene	no
PMS2	NP_000526	NM_000535	D70G	chr7	6,010,068	6,010,268	chr7	74,820,500	74,821,092	68,810,232	Gene	no
PMS2	NP_000526	NM_000535	D70G	chr7	6,010,068	6,010,268	chr7	76,480,911	76,481,503	70,470,643	Pseudogene	no
PMS2	NP_000526	NM_000535	D70G	chr7	6,010,068	6,010,268	chr7	76,508,976	76,509,568	70,498,708	Pseudogene	no

<i>PMS2</i>	NP_000526	NM_000535	D70G	chr7	6,010,068	6,010,268	chr7	99,765,770	99,766,362	93,755,502	Pseudogene	no
<i>PMS2</i>	NP_000526	NM_000535	L42I	chr7	6,011,985	6,012,185	chr7	72,117,649	72,118,289	66,105,464	Pseudogene	no
<i>PMS2</i>	NP_000526	NM_000535	L42I	chr7	6,011,985	6,012,185	chr7	72,145,625	72,146,265	66,133,440	Pseudogene	no
<i>PMS2</i>	NP_000526	NM_000535	L42M	chr7	6,011,985	6,012,185	chr7	73,947,917	73,948,557	67,935,732	Gene	no
<i>PMS2</i>	NP_000526	NM_000535	L42I	chr7	6,011,985	6,012,185	chr7	74,352,195	74,352,835	68,340,010	Pseudogene	no
<i>PMS2</i>	NP_000526	NM_000535	L42I	chr7	6,011,985	6,012,185	chr7	74,766,469	74,767,109	68,754,284	Pseudogene	no
<i>PMS2</i>	NP_000526	NM_000535	L42I	chr7	6,011,985	6,012,185	chr7	74,794,494	74,795,134	68,782,309	Pseudogene	no
<i>PMS2</i>	NP_000526	NM_000535	L42I	chr7	6,011,985	6,012,185	chr7	74,822,477	74,823,117	68,810,292	Pseudogene	no
<i>PMS2</i>	NP_000526	NM_000535	E41K	chr7	6,011,988	6,012,188	chr7	73,947,917	73,948,557	67,935,729	Gene	no
<i>PPIA</i>	NP_066953	NM_021130	C161Y	chr7	44,807,431	44,807,631	chr2	129,970,090	129,971,078	na	Pseudogene	no
<i>PROS1</i>	NP_000304	NM_000313	V669F	chr3	95,075,702	95,075,902	chr3	90,333,909	90,334,567	4,741,135	Pseudogene	no
<i>PROS1</i>	NP_000304	NM_000313	Q563X	chr3	95,078,580	95,078,780	chr3	90,337,849	90,338,082	4,740,498	Pseudogene	yes
<i>PROS1</i>	NP_000304	NM_000313	S396N	chr3	95,087,904	95,088,104	chr3	90,347,183	90,347,853	4,740,051	Pseudogene	no
<i>PROS1</i>	NP_000304	NM_000313	Y340X	chr3	95,094,501	95,094,701	chr3	90,353,759	90,353,989	4,740,512	Pseudogene	no
<i>PROS1</i>	NP_000304	NM_000313	A303T	chr3	95,098,065	95,098,265	chr3	90,357,123	90,357,742	4,740,323	Pseudogene	no
<i>PRSS2</i>	NP_002761	NM_002770	C171Y	chr7	142,181,680	142,181,880	chr7	142,171,198	142,171,835	9,845	Pseudogene	no
<i>PSPH</i>	NP_004568	NM_004577	S109P	chr7	56,052,414	56,052,614	chr7	62,157,495	62,158,147	6,104,881	Pseudogene	no
<i>RANBP2</i>	NP_006258	NM_006267	P1966L	chr2	108,749,225	108,749,425	chr2	109,950,390	109,953,163	1,200,965	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	P1966L	chr2	108,749,225	108,749,425	chr2	110,725,324	110,728,097	1,975,899	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	P1966L	chr2	108,749,225	108,749,425	chr2	111,008,299	111,009,623	2,258,874	Pseudogene	no
<i>RANBP2</i>	NP_006258	NM_006267	P1966L	chr2	108,749,225	108,749,425	chr2	106,405,676	106,408,449	2,340,776	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	P1966L	chr2	108,749,225	108,749,425	chr2	112,861,817	112,864,590	4,112,392	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	Y2003C	chr2	108,749,336	108,749,536	chr2	87,862,503	87,865,276	20,884,060	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	Y2003C	chr2	108,749,336	108,749,536	chr2	87,065,915	87,068,688	21,680,648	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	T2027I	chr2	108,749,408	108,749,608	chr2	107,853,298	107,856,071	893,337	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	D2031G	chr2	108,749,420	108,749,620	chr2	107,853,298	107,856,071	893,349	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	D2031G	chr2	108,749,420	108,749,620	chr2	109,950,390	109,953,163	1,200,770	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	D2031G	chr2	108,749,420	108,749,620	chr2	110,725,324	110,728,097	1,975,704	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	D2031G	chr2	108,749,420	108,749,620	chr2	111,008,299	111,009,623	2,258,679	Pseudogene	no
<i>RANBP2</i>	NP_006258	NM_006267	D2031G	chr2	108,749,420	108,749,620	chr2	106,405,676	106,408,449	2,340,971	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	D2031G	chr2	108,749,420	108,749,620	chr2	112,861,817	112,864,590	4,112,197	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	D2031G	chr2	108,749,420	108,749,620	chr2	87,862,503	87,865,276	20,884,144	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	D2031G	chr2	108,749,420	108,749,620	chr2	87,065,915	87,068,688	21,680,732	Gene	no

<i>RANBP2</i>	NP_006258	NM_006267	R2039G	chr2	108,749,444	108,749,644	chr2	107,853,298	107,856,071	893,373	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	R2039G	chr2	108,749,444	108,749,644	chr2	109,950,390	109,953,163	1,200,746	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	R2039G	chr2	108,749,444	108,749,644	chr2	110,725,324	110,728,097	1,975,680	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	R2039G	chr2	108,749,444	108,749,644	chr2	111,008,299	111,009,623	2,258,655	Pseudogene	no
<i>RANBP2</i>	NP_006258	NM_006267	R2039G	chr2	108,749,444	108,749,644	chr2	106,405,676	106,408,449	2,340,995	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	R2039G	chr2	108,749,444	108,749,644	chr2	112,861,817	112,864,590	4,112,173	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	R2039G	chr2	108,749,444	108,749,644	chr2	87,862,503	87,865,276	20,884,168	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	R2039G	chr2	108,749,444	108,749,644	chr2	87,065,915	87,068,688	21,680,756	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	S2050R	chr2	108,749,477	108,749,677	chr2	107,853,298	107,856,071	893,406	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	S2050R	chr2	108,749,477	108,749,677	chr2	109,950,390	109,953,163	1,200,713	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	S2050R	chr2	108,749,477	108,749,677	chr2	110,725,324	110,728,097	1,975,647	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	S2050R	chr2	108,749,477	108,749,677	chr2	111,008,299	111,009,623	2,258,622	Pseudogene	no
<i>RANBP2</i>	NP_006258	NM_006267	S2050R	chr2	108,749,477	108,749,677	chr2	112,861,817	112,864,590	4,112,140	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	L2071P	chr2	108,749,540	108,749,740	chr2	107,853,298	107,856,071	893,469	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	L2071P	chr2	108,749,540	108,749,740	chr2	87,862,503	87,865,276	20,884,264	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	L2071P	chr2	108,749,540	108,749,740	chr2	87,065,915	87,068,688	21,680,852	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	R2076Q	chr2	108,749,555	108,749,755	chr2	106,405,676	106,408,449	2,341,106	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	L2108S	chr2	108,749,651	108,749,851	chr2	107,853,298	107,856,071	893,580	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	L2108S	chr2	108,749,651	108,749,851	chr2	109,950,390	109,953,163	1,200,539	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	L2108S	chr2	108,749,651	108,749,851	chr2	110,725,324	110,728,097	1,975,473	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	L2108S	chr2	108,749,651	108,749,851	chr2	112,861,817	112,864,590	4,111,966	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	Y2227C	chr2	108,750,008	108,750,208	chr2	107,853,298	107,856,071	893,937	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	A2245D	chr2	108,750,062	108,750,262	chr2	87,862,503	87,865,276	20,884,786	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	A2245D	chr2	108,750,062	108,750,262	chr2	87,065,915	87,068,688	21,681,374	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	R2259H	chr2	108,750,104	108,750,304	chr2	106,405,676	106,408,449	2,341,655	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	G2261D	chr2	108,750,110	108,750,310	chr2	109,950,390	109,953,163	1,200,080	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	G2261D	chr2	108,750,110	108,750,310	chr2	110,725,324	110,728,097	1,975,014	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	G2261D	chr2	108,750,110	108,750,310	chr2	106,405,676	106,408,449	2,341,661	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	G2261D	chr2	108,750,110	108,750,310	chr2	112,861,817	112,864,590	4,111,507	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	G2261D	chr2	108,750,110	108,750,310	chr2	87,862,503	87,865,276	20,884,834	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	G2261D	chr2	108,750,110	108,750,310	chr2	87,065,915	87,068,688	21,681,422	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	F2267S	chr2	108,750,128	108,750,328	chr2	107,853,298	107,856,071	894,057	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	F2267S	chr2	108,750,128	108,750,328	chr2	109,950,390	109,953,163	1,200,062	Gene	no

<i>RANBP2</i>	NP_006258	NM_006267	F2267S	chr2	108,750,128	108,750,328	chr2	110,725,324	110,728,097	1,974,996	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	F2267S	chr2	108,750,128	108,750,328	chr2	106,405,676	106,408,449	2,341,679	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	F2267S	chr2	108,750,128	108,750,328	chr2	112,861,817	112,864,590	4,111,489	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	P2277L	chr2	108,750,158	108,750,358	chr2	109,950,390	109,953,163	1,200,032	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	P2277L	chr2	108,750,158	108,750,358	chr2	110,725,324	110,728,097	1,974,966	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	P2277L	chr2	108,750,158	108,750,358	chr2	106,405,676	106,408,449	2,341,709	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	P2277L	chr2	108,750,158	108,750,358	chr2	112,861,817	112,864,590	4,111,459	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	D2298V	chr2	108,750,221	108,750,421	chr2	109,950,390	109,953,163	1,199,969	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	D2298V	chr2	108,750,221	108,750,421	chr2	110,725,324	110,728,097	1,974,903	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	D2298V	chr2	108,750,221	108,750,421	chr2	112,861,817	112,864,590	4,111,396	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	R2336M	chr2	108,750,335	108,750,535	chr2	87,862,503	87,865,276	20,885,059	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	K2338E	chr2	108,750,341	108,750,541	chr2	107,853,298	107,856,071	894,270	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	K2338E	chr2	108,750,341	108,750,541	chr2	109,950,390	109,953,163	1,199,849	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	K2338E	chr2	108,750,341	108,750,541	chr2	110,725,324	110,728,097	1,974,783	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	K2338E	chr2	108,750,341	108,750,541	chr2	112,861,817	112,864,590	4,111,276	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	K2338E	chr2	108,750,341	108,750,541	chr2	87,862,503	87,865,276	20,885,065	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	K2338E	chr2	108,750,341	108,750,541	chr2	87,065,915	87,068,688	21,681,653	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	L2339I	chr2	108,750,344	108,750,544	chr2	109,950,390	109,953,163	1,199,846	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	L2339I	chr2	108,750,344	108,750,544	chr2	110,725,324	110,728,097	1,974,780	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	L2339F	chr2	108,750,344	108,750,544	chr2	106,405,676	106,408,449	2,341,895	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	L2339I	chr2	108,750,344	108,750,544	chr2	112,861,817	112,864,590	4,111,273	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	Q2367H	chr2	108,750,428	108,750,628	chr2	106,405,676	106,408,449	2,341,979	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	R2385T	chr2	108,750,482	108,750,682	chr2	107,853,298	107,856,071	894,411	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	G2517V	chr2	108,750,878	108,751,078	chr2	109,950,390	109,953,163	1,199,312	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	G2517V	chr2	108,750,878	108,751,078	chr2	110,725,324	110,728,097	1,974,246	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	G2517V	chr2	108,750,878	108,751,078	chr2	112,861,817	112,864,590	4,110,739	Gene	no
<i>RANBP2</i>	NP_006258	NM_006267	P2552S	chr2	108,750,983	108,751,183	chr2	107,853,298	107,856,071	894,912	Gene	no
<i>RDX</i>	NP_002897	NM_002906	D578N	chr11	109,607,721	109,607,921	chr11	4,187,906	4,189,521	105,418,200	Pseudogene	yes
<i>RDX</i>	NP_002897	NM_002906	Q567P	chr11	109,607,754	109,607,954	chr11	4,187,906	4,189,521	105,418,233	Pseudogene	no
<i>RDX</i>	NP_002897	NM_002906	K107X	chr11	109,639,940	109,640,140	chrX	27,445,989	27,446,327	na	Pseudogene	no
<i>ROCK1</i>	NP_005397	NM_005406	R1330X	chr18	16,787,507	16,787,707	chr18	108,296	108,502	16,679,005	Pseudogene	yes
<i>ROCK1</i>	NP_005397	NM_005406	C1264R	chr18	16,788,702	16,788,902	chr18	106,793	107,553	16,681,149	Pseudogene	no
<i>RPL10</i>	NP_006004	NM_006013	R116X	chrX	153,281,917	153,282,117	chr19	9,791,817	9,792,131	na	Pseudogene	no

<i>RPL10</i>	NP_006004	NM_006013	G196R	chrX	153,282,232	153,282,432	chr12	67,306,670	67,307,484	na	Pseudogene	no
<i>RPS24</i>	NP_148982	NM_033022	G59A	chr10	79,465,282	79,465,482	chr14	75,131,663	75,132,552	na	Pseudogene	no
<i>RPS6KB1</i>	NP_003152	NM_003161	R144W	chr17	55,358,528	55,358,728	chr17	57,678,897	57,679,546	2,320,169	Pseudogene	no
<i>RPS6KB1</i>	NP_003152	NM_003161	R144W	chr17	55,358,528	55,358,728	chr17	20,560,138	20,560,787	34,797,741	Pseudogene	no
<i>RPS6KB1</i>	NP_003152	NM_003161	P221L	chr17	55,363,740	55,363,940	chr17	20,565,673	20,566,292	34,797,448	Pseudogene	no
<i>RPS6KB1</i>	NP_003152	NM_003161	C240R	chr17	55,366,244	55,366,444	chr17	57,670,939	57,671,531	2,304,495	Pseudogene	no
<i>RPS6KB1</i>	NP_003152	NM_003161	R272C	chr17	55,366,513	55,366,713	chr17	20,568,515	20,569,104	34,797,409	Pseudogene	no
<i>RPS6KB1</i>	NP_003152	NM_003161	W276C	chr17	55,366,525	55,366,725	chr17	20,568,515	20,569,104	34,797,421	Pseudogene	no
<i>RPS6KB1</i>	NP_003152	NM_003161	W276X	chr17	55,366,525	55,366,725	chr17	57,670,930	57,671,019	2,304,205	Pseudogene	no
<i>RPS6KB1</i>	NP_003152	NM_003161	L287P	chr17	55,366,558	55,366,758	chr17	20,568,515	20,569,104	34,797,454	Pseudogene	no
<i>RPS6KB1</i>	NP_003152	NM_003161	S403T	chr17	55,372,968	55,373,168	chr17	57,663,363	57,663,985	2,290,195	Pseudogene	no
<i>RPS6KB1</i>	NP_003152	NM_003161	N405H	chr17	55,372,974	55,373,174	chr17	20,574,843	20,575,465	34,797,509	Pseudogene	no
<i>SAGE1</i>	NP_061136	NM_018666	D890G	chrX	134,822,577	134,822,777	chrX	134,576,674	134,577,272	245,305	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	L1241H	chr16	49,728,675	49,728,875	chrX	49,316,198	49,317,090	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	A1240R	chr16	49,728,678	49,728,878	chrX	49,316,198	49,317,090	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	Y1237N	chr16	49,728,687	49,728,887	chrX	49,316,198	49,317,090	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	S1022N	chr16	49,730,467	49,730,667	chrX	49,316,650	49,319,642	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	T899I	chr16	49,730,836	49,731,036	chrX	49,316,650	49,319,642	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	G895D	chr16	49,730,848	49,731,048	chrX	49,316,650	49,319,642	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	P560L	chr16	49,731,853	49,732,053	chrX	49,316,650	49,319,642	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	G556C	chr16	49,731,865	49,732,065	chrX	49,316,650	49,319,642	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	P533T	chr16	49,731,934	49,732,134	chrX	49,316,650	49,319,642	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	R498C	chr16	49,732,039	49,732,239	chrX	49,316,650	49,319,642	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	G298X	chr16	49,732,639	49,732,839	chrX	49,319,392	49,320,234	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	T284M	chr16	49,732,681	49,732,881	chrX	49,319,142	49,320,484	na	Pseudogene	no
<i>SALL1</i>	NP_002959	NM_002968	A206V	chr16	49,732,915	49,733,115	chrX	49,319,142	49,320,484	na	Pseudogene	no
<i>SCN2A</i>	NP_066287	NM_021007	A202C	chr2	165,874,008	165,874,208	chr2	165,728,291	165,728,928	145,080	Gene	no
<i>SCN9A</i>	NP_002968	NM_002977	N1665D	chr2	166,764,266	166,764,466	chr2	166,555,757	166,557,477	206,789	Gene	no
<i>SDHA</i>	NP_004159	NM_004168	G84E	chr5	277,476	277,676	chr3	196,874,434	196,875,035	na	Pseudogene	no
<i>SDHA</i>	NP_004159	NM_004168	G84E	chr5	277,476	277,676	chr3	197,196,645	197,197,246	na	Pseudogene	no
<i>SDHA</i>	NP_004159	NM_004168	G84E	chr5	277,476	277,676	chr3	198,834,238	198,834,839	na	Pseudogene	no
<i>SDHA</i>	NP_004159	NM_004168	G184R	chr5	278,993	279,193	chr3	196,875,885	196,876,588	na	Gene	no
<i>SDHA</i>	NP_004159	NM_004168	G184R	chr5	278,993	279,193	chr3	198,832,686	198,833,395	na	Pseudogene	no

SDHA	NP_004159	NM_004168	R188G	chr5	279,005	279,205	chr3	197,195,094	197,195,797	na	Pseudogene	no
SDHA	NP_004159	NM_004168	L204S	chr5	279,053	279,253	chr3	198,832,686	198,833,395	na	Pseudogene	no
SDHA	NP_004159	NM_004168	Y215C	chr5	281,223	281,423	chr3	196,877,874	196,878,538	na	Gene	no
SDHA	NP_004159	NM_004168	Y215C	chr5	281,223	281,423	chr3	197,193,158	197,193,822	na	Pseudogene	no
SDHA	NP_004159	NM_004168	R246H	chr5	281,316	281,516	chr3	198,830,736	198,831,400	na	Pseudogene	no
SDHA	NP_004159	NM_004168	S346W	chr5	286,634	286,834	chr3	196,882,933	196,883,603	na	Gene	no
SDHA	NP_004159	NM_004168	S346W	chr5	286,634	286,834	chr3	197,188,100	197,188,770	na	Pseudogene	no
SDHA	NP_004159	NM_004168	R379P	chr5	288,231	288,431	chr3	196,884,284	196,884,987	na	Pseudogene	no
SDHA	NP_004159	NM_004168	R379P	chr5	288,231	288,431	chr3	197,186,717	197,187,420	na	Pseudogene	no
SDHA	NP_004159	NM_004168	A449V	chr5	289,529	289,729	chr3	196,885,617	196,886,260	na	Gene	no
SDHA	NP_004159	NM_004168	A449V	chr5	289,529	289,729	chr3	197,185,444	197,186,087	na	Pseudogene	no
SDHA	NP_004159	NM_004168	S509X	chr5	293,467	293,667	chr5	1,642,362	1,642,481	1,348,695	Pseudogene	no
SDHA	NP_004159	NM_004168	R585G	chr5	304,444	304,644	chr3	196,893,337	196,893,977	na	Gene	no
SDHA	NP_004159	NM_004168	R585G	chr5	304,444	304,644	chr3	197,178,523	197,179,163	na	Gene	no
SDHA	NP_004159	NM_004168	A591T	chr5	304,462	304,662	chr5	1,629,041	1,629,675	1,324,379	Pseudogene	no
SDHA	NP_004159	NM_004168	R594G	chr5	304,471	304,671	chr3	196,893,337	196,893,977	na	Gene	no
SDHA	NP_004159	NM_004168	R594G	chr5	304,471	304,671	chr3	197,178,523	197,179,163	na	Gene	no
SDHA	NP_004159	NM_004168	V644I	chr5	309,372	309,572	chr3	196,897,617	196,898,203	na	Pseudogene	no
SDHA	NP_004159	NM_004168	V644I	chr5	309,372	309,572	chr3	197,174,356	197,174,942	na	Gene	no
SERPINH1	NP_001226	NM_001235	L114P	chr11	74,955,284	74,955,484	chr9	34,308,197	34,309,497	na	Pseudogene	no
SLC25A15	NP_055067	NM_014252	G244X	chr13	40,280,583	40,280,783	chr21	9,920,983	9,921,117	na	Pseudogene	no
SLC25A15	NP_055067	NM_014252	A279D	chr13	40,281,634	40,281,834	chr13	24,416,263	24,416,885	15,864,749	Pseudogene	no
SLC25A15	NP_055067	NM_014252	A279D	chr13	40,281,634	40,281,834	chr13	18,887,994	18,888,580	21,393,054	Pseudogene	no
SLC25A15	NP_055067	NM_014252	A279D	chr13	40,281,634	40,281,834	chr22	15,506,332	15,506,954	na	Pseudogene	no
SLC25A15	NP_055067	NM_014252	A279D	chr13	40,281,634	40,281,834	chrY	27,141,930	27,142,552	na	Pseudogene	no
SLC6A14	NP_009162	NM_007231	A316D	chrX	115,496,552	115,496,752	chrX	115,691,294	115,691,973	194,542	Pseudogene	no
SLC6A8	NP_005620	NM_005629	Y94C	chrX	152,608,943	152,609,143	chr16	32,802,142	32,802,770	na	Pseudogene	no
SLC6A8	NP_005620	NM_005629	Y94C	chrX	152,608,943	152,609,143	chr16	33,687,556	33,688,184	na	Gene	no
SLC6A8	NP_005620	NM_005629	A495V	chrX	152,612,985	152,613,185	chr16	32,798,188	32,798,831	na	Gene	no
SLC6A8	NP_005620	NM_005629	Y551N	chrX	152,613,324	152,613,524	chr16	32,797,750	32,798,420	na	Gene	no
SLC6A8	NP_005620	NM_005629	V572M	chrX	152,613,387	152,613,587	chr16	32,797,750	32,798,420	na	Gene	no
SLC6A8	NP_005620	NM_005629	V572M	chrX	152,613,387	152,613,587	chr16	33,691,722	33,692,575	na	Gene	no
SLC7A5	NP_003477	NM_003486	I139V	chr16	86,460,012	86,460,212	chr16	29,531,676	29,532,712	56,927,300	Pseudogene	no

<i>SLC7A5</i>	NP_003477	NM_003486	V88M	chr16	86,460,165	86,460,365	chr16	29,531,676	29,532,712	56,927,453	Pseudogene	no
<i>SMG1</i>	NP_057097	NM_016013	W136X	chr15	39,476,041	39,476,241	chr19	61,545,809	61,546,600	na	Pseudogene	no
<i>SMG1</i>	NP_057097	NM_016013	W125X	chr15	39,476,074	39,476,274	chr19	61,545,809	61,546,600	na	Pseudogene	no
<i>SMG1</i>	NP_055907	NM_015092	L1278X	chr16	18,778,397	18,778,597	chr16	21,367,268	21,367,435	2,588,671	Pseudogene	no
<i>SMG1</i>	NP_055907	NM_015092	L1278X	chr16	18,778,397	18,778,597	chr16	21,799,528	21,799,695	3,020,931	Pseudogene	no
<i>SMG1</i>	NP_055907	NM_015092	L1278X	chr16	18,778,397	18,778,597	chr16	22,409,113	22,409,280	3,630,516	Pseudogene	no
<i>SMG1</i>	NP_055907	NM_015092	L1278X	chr16	18,778,397	18,778,597	chr16	29,447,757	29,447,924	10,669,160	Pseudogene	no
<i>SMG1</i>	NP_055907	NM_015092	L1278X	chr16	18,778,397	18,778,597	chr16	30,187,742	30,187,909	11,409,145	Pseudogene	no
<i>SMG1</i>	NP_055907	NM_015092	R164X	chr16	18,810,997	18,811,197	chr16	29,480,334	29,480,474	10,669,137	Pseudogene	no
<i>SMG1</i>	NP_055907	NM_015092	R164X	chr16	18,810,997	18,811,197	chr16	30,220,041	30,220,181	11,408,844	Pseudogene	no
<i>SNX19</i>	NP_055573	NM_014758	P507S	chr11	130,289,423	130,289,623	chr13	18,365,348	18,366,450	na	Pseudogene	no
<i>SNX19</i>	NP_055573	NM_014758	P507S	chr11	130,289,423	130,289,623	chr18	14,619,754	14,622,101	na	Pseudogene	no
<i>STRC</i>	NP_714544	NM_153700	T1709A	chr15	41,679,461	41,679,661	chr15	41,779,014	41,779,663	99,353	Pseudogene	no
<i>STRC</i>	NP_714544	NM_153700	L1640F	chr15	41,679,996	41,680,196	chr15	41,779,587	41,780,245	99,391	Pseudogene	no
<i>STRC</i>	NP_714544	NM_153700	L1506P	chr15	41,682,658	41,682,858	chr15	41,781,995	41,782,662	99,137	Pseudogene	no
<i>STRC</i>	NP_714544	NM_153700	Q1353X	chr15	41,684,107	41,684,307	chr15	41,783,619	41,783,816	99,312	Pseudogene	no
<i>TARDBP</i>	NP_031401	NM_007375	T30M	chr1	10,996,361	10,996,561	chr13	59,746,684	59,747,414	na	Pseudogene	no
<i>TARDBP</i>	NP_031401	NM_007375	Y43C	chr1	10,996,400	10,996,600	chr2	238,805,011	238,805,813	na	Pseudogene	no
<i>TARDBP</i>	NP_031401	NM_007375	A326P	chr1	11,004,931	11,005,131	chr2	238,805,740	238,806,386	na	Pseudogene	no
<i>TARDBP</i>	NP_031401	NM_007375	A329V	chr1	11,004,940	11,005,140	chr2	238,805,740	238,806,386	na	Pseudogene	no
<i>TARDBP</i>	NP_031401	NM_007375	A329V	chr1	11,004,940	11,005,140	chr8	64,211,394	64,212,289	na	Pseudogene	no
<i>TARDBP</i>	NP_031401	NM_007375	G335C	chr1	11,004,958	11,005,158	chr2	238,805,740	238,806,386	na	Pseudogene	no
<i>TARDBP</i>	NP_031401	NM_007375	G338C	chr1	11,004,967	11,005,167	chr13	59,747,519	59,748,393	na	Pseudogene	no
<i>TARDBP</i>	NP_031401	NM_007375	G351C	chr1	11,005,006	11,005,206	chr8	64,211,394	64,212,289	na	Pseudogene	no
<i>TARDBP</i>	NP_031401	NM_007375	Y374C	chr1	11,005,075	11,005,275	chr2	238,805,862	238,806,586	na	Pseudogene	no
<i>TBX20</i>	NP_001071121	NM_001077653	T444M	chr7	35,208,478	35,208,678	chr12	62,364,543	62,365,381	na	Pseudogene	no
<i>TBX20</i>	NP_001071121	NM_001077653	Y309N	chr7	35,210,582	35,210,782	chr12	62,368,175	62,368,791	na	Pseudogene	no
<i>TBX20</i>	NP_001071121	NM_001077653	F256L	chr7	35,246,960	35,247,160	chr12	62,404,564	62,405,219	na	Pseudogene	no
<i>TIMM8A</i>	NP_004076	NM_004085	R80X	chrX	100,488,096	100,488,296	chr2	162,642,116	162,642,430	na	Pseudogene	no
<i>TLL1</i>	NP_036596	NM_012464	G288E	chr4	167,148,497	167,148,697	chr12	11,560,376	11,560,983	na	Pseudogene	no
<i>TLR2</i>	NP_003255	NM_003264	P631H	chr4	154,845,302	154,845,502	chr4	154,819,661	154,820,937	24,365	Pseudogene	yes
<i>TLR2</i>	NP_003255	NM_003264	P669L	chr4	154,845,416	154,845,616	chr4	154,819,661	154,820,937	24,479	Pseudogene	no
<i>TLR2</i>	NP_003255	NM_003264	R677W	chr4	154,845,440	154,845,640	chr4	154,819,661	154,820,937	24,503	Pseudogene	yes

TRIP11	NP_004230	NM_004239	L1955W	chr14	91,505,744	91,505,944	chr12	105,088,779	105,090,592	na	Pseudogene	no
TUBA1A	NP_006000	NM_006009	K370M	chr12	47,865,205	47,865,405	chr11	89,655,224	89,656,842	na	Pseudogene	no
TUBA1A	NP_006000	NM_006009	R320H	chr12	47,865,355	47,865,555	chr11	89,655,224	89,656,842	na	Pseudogene	no
TUBA1A	NP_006000	NM_006009	Y319C	chr12	47,865,358	47,865,558	chr11	89,655,224	89,656,842	na	Pseudogene	no
TUBA1A	NP_006000	NM_006009	R308C	chr12	47,865,391	47,865,591	chr1	47,130,273	47,131,339	na	Pseudogene	no
TUBA1A	NP_006000	NM_006009	P268R	chr12	47,865,511	47,865,711	chr5	43,267,702	43,268,636	na	Pseudogene	no
TUBA1A	NP_006000	NM_006009	Y210C	chr12	47,865,685	47,865,885	chr11	89,655,224	89,656,842	na	Pseudogene	no
TUBA1A	NP_006000	NM_006009	I171N	chr12	47,865,802	47,866,002	chr11	89,655,224	89,656,842	na	Pseudogene	no
TUBB2B	NP_821080	NM_178012	A248V	chr6	3,170,477	3,170,677	chr6	3,122,025	3,122,746	47,731	Pseudogene	no
TUFM	NP_003312	NM_003321	T433I	chr16	28,761,765	28,761,965	chr17	22,433,596	22,434,461	na	Pseudogene	no
UBE2NL	NP_001013007	NM_001012989	L89X	chrX	142,795,035	142,795,235	chr4	173,537,824	173,538,213	na	Pseudogene	no
UBE3A	NP_570853	NM_130838	D438H	chr15	23,166,939	23,167,139	chr21	31,355,993	31,356,822	na	Pseudogene	no
UBE3A	NP_570853	NM_130838	R401X	chr15	23,167,050	23,167,250	chr21	31,356,243	31,356,572	na	Pseudogene	no
UGT1A7	NP_061950	NM_019077	T240M	chr2	234,255,942	234,256,142	chr2	234,221,901	234,222,661	33,281	Pseudogene	no
UGT1A7	NP_061950	NM_019077	R254X	chr2	234,255,984	234,256,184	chr2	234,158,837	234,159,676	96,308	Pseudogene	yes
UGT1A8	NP_061949	NM_019076	R254X	chr2	234,191,754	234,191,954	chr2	234,177,598	234,177,786	13,968	Pseudogene	no
UGT2A3	NP_079019	NM_024743	F460S	chr4	69,830,223	69,830,423	chr4	69,687,240	69,688,033	142,190	Pseudogene	no
UGT2A3	NP_079019	NM_024743	V459D	chr4	69,830,226	69,830,426	chr4	70,150,579	70,151,372	320,153	Pseudogene	no
UGT2A3	NP_079019	NM_024743	V459N	chr4	69,830,226	69,830,426	chr4	70,258,018	70,258,733	427,592	Pseudogene	no
UGT2A3	NP_079019	NM_024743	R457Q	chr4	69,830,232	69,830,432	chr4	70,150,579	70,151,372	320,147	Pseudogene	no
UGT2A3	NP_079019	NM_024743	P454T	chr4	69,830,241	69,830,441	chr4	69,687,240	69,688,033	142,208	Pseudogene	no
UGT2A3	NP_079019	NM_024743	R445G	chr4	69,830,268	69,830,468	chr4	70,257,941	70,258,518	427,473	Pseudogene	no
UGT2A3	NP_079019	NM_024743	N439D	chr4	69,830,286	69,830,486	chr4	69,687,240	69,688,033	142,253	Pseudogene	no
UGT2A3	NP_079019	NM_024743	N439D	chr4	69,830,286	69,830,486	chr4	70,150,579	70,151,372	320,093	Pseudogene	no
UGT2A3	NP_079019	NM_024743	N439Y	chr4	69,830,286	69,830,486	chr4	70,257,941	70,258,518	427,455	Pseudogene	no
UGT2A3	NP_079019	NM_024743	A426V	chr4	69,830,778	69,830,978	chr4	70,149,931	70,150,646	318,953	Pseudogene	no
UGT2A3	NP_079019	NM_024743	M404R	chr4	69,830,844	69,831,044	chr4	69,686,592	69,687,274	143,570	Pseudogene	no
UGT2A3	NP_079019	NM_024743	N400A	chr4	69,830,856	69,831,056	chr4	70,149,931	70,150,646	318,875	Pseudogene	no
UGT2A3	NP_079019	NM_024743	G385D	chr4	69,830,901	69,831,101	chr4	69,686,592	69,687,274	143,627	Pseudogene	no
UGT2A3	NP_079019	NM_024743	G385E	chr4	69,830,901	69,831,101	chr4	70,149,931	70,150,646	318,830	Pseudogene	no
UGT2A3	NP_079019	NM_024743	G385E	chr4	69,830,901	69,831,101	chr4	70,257,299	70,258,014	426,198	Pseudogene	no
UGT2A3	NP_079019	NM_024743	V303G	chr4	69,832,921	69,833,121	chr4	70,148,020	70,148,654	314,899	Pseudogene	no
UGT2A3	NP_079019	NM_024743	G298W	chr4	69,832,936	69,833,136	chr4	69,684,672	69,685,309	147,627	Pseudogene	no

<i>UGT2A3</i>	NP_079019	NM_024743	L170P	chr4	69,851,457	69,851,657	chr4	70,082,040	70,082,845	230,383	Pseudogene	no
<i>UGT2A3</i>	NP_079019	NM_024743	L170P	chr4	69,851,457	69,851,657	chr4	70,206,521	70,207,503	354,864	Pseudogene	no
<i>UGT2A3</i>	NP_079019	NM_024743	C154R	chr4	69,851,505	69,851,705	chr4	70,082,040	70,082,845	230,335	Pseudogene	no
<i>UGT2A3</i>	NP_079019	NM_024743	C154R	chr4	69,851,505	69,851,705	chr4	70,143,035	70,143,900	291,330	Pseudogene	no
<i>UGT2A3</i>	NP_079019	NM_024743	C154H	chr4	69,851,505	69,851,705	chr4	70,206,521	70,207,503	354,816	Pseudogene	no
<i>UGT2A3</i>	NP_079019	NM_024743	C154R	chr4	69,851,505	69,851,705	chr4	70,246,990	70,247,795	395,285	Pseudogene	no
<i>UGT2A3</i>	NP_079019	NM_024743	C23X	chr4	69,851,898	69,852,098	chr4	70,246,917	70,247,264	394,819	Pseudogene	no
<i>UGT2B10</i>	NP_001066	NM_001075	P357S	chr4	69,726,688	69,726,888	chr4	70,298,652	70,299,256	571,764	Pseudogene	no
<i>UGT2B7</i>	NP_001065	NM_001074	V493M	chr4	70,012,832	70,013,032	chr4	69,814,982	69,815,820	197,012	Pseudogene	no
<i>UNC93A</i>	NP_061847	NM_018974	Y387H	chr6	167,648,617	167,648,817	chr6	167,541,862	167,542,697	105,920	Pseudogene	no
<i>VMA21</i>	NP_001017980	NM_001017980	A87V	chrX	150,324,043	150,324,243	chr7	43,773,297	43,773,970	na	Pseudogene	no
<i>VMA21</i>	NP_001017980	NM_001017980	W95X	chrX	150,324,067	150,324,267	chr7	43,773,547	43,773,720	na	Pseudogene	no
<i>XIAP</i>	NP_001158	NM_001167	C471R	chrX	122,868,531	122,868,731	chr2	113,327,126	113,328,417	na	Pseudogene	no
<i>XIAP</i>	NP_001158	NM_001167	C481R	chrX	122,868,561	122,868,761	chr10	111,429,940	111,431,039	na	Pseudogene	no
<i>XRCC6</i>	NP_001460	NM_001469	C150R	chr22	40,362,481	40,362,681	chrX	149,149,702	149,152,028	na	Pseudogene	no

ProteinID and GeneID are from the RefSeq database. Site: wild-type amino acid followed by the codon position and the deleterious amino acid (X: stop codon). Chr: chromosome. The "r" the end of the chromosome number indicates an unmapped scaffold on that chromosome. When available, the distance between donor and acceptor sequences is given in base-pairs. In HGMD: amino acid replacement reported in the HGMD missense/nonsense mutations data set.

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