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1	11850750	11850750	G	A	SNP	MTHFR
1	11854476	11854476	T	G	SNP	MTHFR
1	11856378	11856378	G	A	SNP	MTHFR
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1	155880573	155880573	C	G	SNP	RIT1
1	161479745	161479745	A	G	SNP	FCGR2A
1	161518214	161518214	T	C	SNP	FCGR3A
1	161518336	161518336	C	T	SNP	FCGR3A
1	162740121	162740121	G	A	SNP	DDR2
1	179077613	179077613	T	C	SNP	ABL2
1	179112145	179112145	C	G	SNP	ABL2
1	242030151	242030151	A	G	SNP	EXO1
1	242035438	242035438	G	A	SNP	EXO1
1	242042301	242042301	G	A	SNP	EXO1
1	242042545	242042545	A	G	SNP	EXO1
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10	101563815	101563815	G	A	SNP	ABCC2
10	101977883	101977883	C	T	SNP	CHUK
10	43610119	43610119	G	A	SNP	RET
10	43620335	43620335	C	T	SNP	RET
10	88635779	88635779	C	A	SNP	BMPR1A
11	108175462	108175462	G	A	SNP	ATM
11	108183167	108183167	A	G	SNP	ATM
11	125525195	125525195	A	G	SNP	CHEK1
11	64572018	64572018	T	C	SNP	MEN1
11	67352689	67352689	A	G	SNP	GSTP1
11	92531356	92531356	A	G	SNP	FAT3
11	92534442	92534442	A	G	SNP	FAT3
11	92590448	92590448	A	G	SNP	FAT3
12	111856506	111856506	G	T	SNP	SH2B3
12	111884608	111884608	T	C	SNP	SH2B3
12	121416650	121416650	A	C	SNP	HNF1A
12	121435427	121435427	G	A	SNP	HNF1A
12	121437221	121437221	T	C	SNP	HNF1A

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12	78225374	78225374	A	G	SNP	NAV3
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12	998365	998365	G	T	SNP	WNK1
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13	103527849	103527849	G	C	SNP	ERCC5
13	103527930	103527930	G	C	SNP	ERCC5
13	110435231	110435231	C	T	SNP	IRS2
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15	67457335	67457335 A	G	SNP	SMAD3
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15	74328141	74328141 G	T	SNP	PML
15	74336633	74336633 T	C	SNP	PML
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16	89986144	89986144 C	T	SNP	MC1R
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17	1782952	1782952 A	G	SNP	RPA1
17	37884037	37884037 C	G	SNP	ERBB2
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17	56435080	56435080 G	C	SNP	RNF43
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17	8108331	8108331 A	G	SNP	AURKB
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18	42456653	42456653 G	A	SNP	SETBP1
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19	2226676	2226676 G	A	SNP	DOT1L
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19	41743861	41743861 A	G	SNP	AXL
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22	19951897	19951897 G	C	SNP	COMT
22	23627369	23627369 A	G	SNP	BCR
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22	36623920	36623920 G	A	SNP	APOL2
22	42522613	42522613 G	C	SNP	CYP2D6
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3	51978220	51978220 A	G	SNP	PARP3
4	106196951	106196951 A	G	SNP	TET2
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5	56177443	56177443 G	A	SNP	MAP3K1
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6	112382313	112382313 G	T	SNP	WISP3
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6	136582497	136582497 G	T	SNP	BCLAF1
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ENST000003:human	ensembl	70_37	-1 missense	c.3194
ENST000003:human	ensembl	70_37	-1 missense	c.3176
ENST000003:human	ensembl	70_37	-1 missense	c.3116
ENST000003:human	ensembl	70_37	-1 missense	c.2807
ENST000003:human	ensembl	70_37	-1 missense	c.2695
ENST000003:human	ensembl	70_37	-1 missense	c.2618
ENST000003:human	ensembl	70_37	-1 missense	c.680
ENST000003:human	ensembl	70_37	-1 missense	c.20
ENST000003:human	ensembl	70_37	1 missense	c.1114
ENST000003:human	ensembl	70_37	1 missense	c.7397
ENST000002:human	ensembl	70_37	1 missense	c.442

ENST000003:human	ensembl	70_37	-1 missense	c.806
ENST000003:human	ensembl	70_37	-1 missense	c.722
ENST000002:human	ensembl	70_37	-1 missense	c.131
ENST000002:human	ensembl	70_37	-1 missense	c.247
ENST000002:human	ensembl	70_37	1 missense	c.524
ENST000002:human	ensembl	70_37	1 missense	c.2632
ENST000002:human	ensembl	70_37	1 missense	c.4378
ENST000002:human	ensembl	70_37	1 missense	c.5434
ENST000005:human	ensembl	70_37	1 missense	c.1816
ENST000003:human	ensembl	70_37	1 missense	c.1051
ENST000002:human	ensembl	70_37	1 missense	c.2181
ENST000003:human	ensembl	70_37	-1 missense	c.710
ENST000003:human	ensembl	70_37	1 missense	c.830
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ENST000003{human	ensembl	70_37	-1 missense	c.232
ENST000003{human	ensembl	70_37	-1 missense	c.1004
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ENST000003{human	ensembl	70_37	-1 missense	c.3863



amino_acid_ucsc_cons	WHISP_freq	WHISP_percent	dbSNP_id	oneKGenome	oneKGenome
p.T694M	0	24	4.31 rs35737219	2	0.2
p.E470A	1	283	50.81 rs1801131	32	17
p.A263V	1	320	57.45 rs1801133	35	11
p.N46S	1	2	0.36 NA	NA	NA
p.E38K	1	3	0.54 NA	NA	NA
p.C19W	0.99	111	19.93 rs11810554	37	16
p.E11Q	0.001	477	85.64 rs493446	90	45
p.H167R	0	418	75.04 rs1801274	50	49
p.I142V	0	134	24.06 .	19	15
p.S101N	0	19	3.41 MERGED_DE	1	0.41
p.M441I	1	22	3.95 rs34722354	2	0.41
p.K930R	1	19	3.41 rs17277288	2 NA	
p.S12T	0.635	93	16.7 rs1318056	8	10
p.H354R	0.458	430	77.2 rs735943	58	60
p.V458M	0.001	230	41.29 rs4149965	25	1
p.E589K	0	342	61.4 rs1047840	36	53
p.E670G	0	485	87.07 rs1776148	65	67
p.R723C	0.218	556	99.82 rs1635498	97	87
p.I18N	1	384	68.94 rs6672420	49	40
p.V114M	0.001	25	4.49 rs12731981	5	0.2
p.R29C	0.806	532	95.51 rs1801265	78	55
p.Y39F	1	557	100 rs927344	100	98
p.V417I	0.001	183	32.85 rs2273697	20	20
p.V268I	1	429	77.02 rs2230804	52	76
p.G691S	0.049	183	32.85 rs1799939	20	9
p.R982C	0.97	18	3.23 rs17158558	2	1
p.P2T	1	239	42.91 rs11528010	25	59
p.D1853N	1	137	24.6 rs1801516	16	1
p.N1983S	0.001	557	100 rs659243	100	100
p.I471V	0.993	556	99.82 rs506504	97	99
p.T546A	0.397	541	97.13 rs2959656	99	71
p.I105V	0.001	315	56.55 rs1695	32	44
p.Q1726R	0.02	547	98.2 rs7949157	87	65
p.I2755V	0.853	549	98.56 rs3847531	88	72
p.S3812G	0.674	539	96.77 rs4753069	80	93
p.S186I	0.998 NA	NA	rs183913232	1 NA	
p.W60R	0.01	423	75.94 rs3184504	53	97
p.I27L	1	133	23.88 rs1169288	33	7
p.S487N	0.943	275	49.37 rs2464196	31	11
p.L551S	0	534	95.87 rs1169304	83	59

p.S574G	0.033	541	97.13	rs1169305	100	96
p.V109G	0.676	216	38.78	rs2066827	23	74
p.S42T	0	494	88.69	rs6486602	67	22
p.P146L	0.036	352	63.2	rs11044004	40	33
p.S112A	0.999	547	98.2	rs4149117	85	38
p.M233I	0.622	548	98.38	rs7311358	85	38
p.G256A	1	160	28.73	rs60140950	17	0.41
p.H39P	1	61	10.95	rs3759295	5	6
p.R50H	0.089	556	99.82	rs2256408	100	89
p.S1647T	0.989	133	23.88	rs11564148	33	17
p.N2081D	1	20	3.59	rs33995883	2 NA	
p.M2397T	0.003	490	87.97	rs3761863	68	46
p.S1119C	0.997	112	20.11	rs773123	11	5
p.T45A	1	557	100	rs10735309	100	91
p.L730F	1	5	0.9	rs11611231	10	14
p.T1554P	0.99	543	97.49	rs956868	84	87
p.C2004S	0.004	557	100	rs7955371	100	96
p.M2306I	0.834	348	62.48	rs12828016	38	53
p.R263Q	0.869	1	0.18	NA	NA	
p.G1053R	0.027	557	100	rs9514066	100	100
p.G1080R	0	557	100	rs9514067	100	100
p.G1057D	0.001	63	11.31	rs1805097	31	11
p.L125R	0	236	42.37	rs7338333	100	100
p.K644R	1	244	43.81	rs2302757	24	2
p.I1564T	0	506	90.84	rs1372085	96	86
p.L1550P	0	515	92.46	rs1822135	96	83
p.S1459Y	0	542	97.31	rs9318536	92	79
p.S1394A	0	556	99.82	rs9511259	96	87
p.R1108C	0.178	530	95.15	rs9318554	95	65
p.L1080R	0.839	472	84.74	rs9318558	95	83
p.V1065A	0	512	91.92	rs7334587	72	43
p.Q1059R	1	158	28.37	NA	NA	
p.I1039T	1	194	34.83	NA	NA	
p.M936T	0	556	99.82	rs4770684	97	81
p.A899T	1	515	92.46	rs2275660	77	45
p.S873N	0	190	34.11	rs7140044	18	22
p.T227M	0.954	483	86.71	rs1933437	58	30
p.D7G	0.969 NA	NA		rs12872889	25	61
p.N372H	0	268	48.11	rs144848	29	10
p.V2466A	0.016	552	99.1	rs169547	100	90
p.C148R	0	379	68.04	rs3803185	52	7

p.N269S	0	263	47.22	rs4883918	27	11
p.T241M	0	273	49.01	rs861539	40	19
p.P44L	0.261	16	2.87	rs45543740	1 NA	
p.A83T	0.994	337	60.5	rs7144658	59	40
p.S175F	0.003	71	12.75	rs10138997	8	47
p.V878L	0.006	120	21.54	rs1367580	14	46
p.I1460V	0.246	107	19.21	rs78211950	13	1
p.P1812A	0.002	113	20.29	rs3736772	13	1
p.P606S	0.998	120	21.54	rs11549465	10	4
p.C351R	0.014	243	43.63	rs8023214	56	76
p.E727D	0.003	554	99.46	rs1991517	90	93
p.V237A	0	213	38.24	rs6647	22	63
p.P277L	1	2	0.36	.	0.13 NA	
p.R363Q	0.974	508	91.2	rs1801376	70	83
p.T716S	0.988	530	95.15	rs2178004	84	50
p.Y85C	1	548	98.38	rs1065080	86	87
p.S772G	0.002	222	39.86	rs743580	53	50
p.G780V	0	161	28.9	rs743581	40	31
p.F645L	0.049	380	68.22	rs5742915	44	5
p.A86V	1	343	61.58	rs17803620	40	6
p.C742S	1	345	61.94	rs2283432	40	6
p.E7Q	0	1	0.18	rs2667660	21	17
p.E7G	0	208	37.34	rs2667661	21	17
p.Q146R	0	284	50.99	rs2235624	61	20
p.Q559R	0	94	16.88	rs152451	10	23
p.V223M	0.044	530	95.15	rs1801030	96	63
p.V777A	0.779	279	50.09	rs4788682	83	73
p.S72A	0.983	243	43.63	rs7193297	39	78
p.S858R	0.036	5	0.9	rs17233141	1	0.2
p.G501S	0	361	64.81	rs2239359	42	68
p.V6D	0	10	1.8	rs1800282	10	2
p.R160W	0.637	79	14.18	rs1805008	7	1
p.G150R	1	164	29.44	NA	NA	NA
p.T351A	0.971	114	20.47	rs5030755	11	0.41
p.P1170A	1	486	87.25	rs61552325	68	20
p.E69D	0	552	99.1	rs76372682	87	99
p.I350T	0.021	247	44.34	rs12450550	24	4
p.P686R	0.003	120	21.54	rs9652855	12	12
p.S919P	0	471	84.56	rs4986764	55	70
p.P208L	0.979	548	98.38	rs3744045	92	43
p.A67S	0	358	64.27	rs67160445	46	25

p.P72R	0.045	477	85.64	rs1042522	72	33
p.M298T	0	463	83.12	rs1059476	90	61
p.V150I	0.673	69	12.39	rs3748415	13	1
p.M1708V	0	413	74.15	rs7232237	51	72
p.A222T	0	137	24.6	rs663651	57	54
p.V231L	0.003	228	40.93	rs11082414	20	18
p.P452A	0.513	192	34.47	rs125555	19	15
p.A731T	0.746	462	82.94	rs8305	71	98
p.I2157V	0	536	96.23	rs7240666	85	90
p.H1767Y	0	144	25.85	rs7234999	11	6
p.Q1579R	0	144	25.85	rs33910491	11	2
p.H1174P	0	300	53.86	rs3809977	78	43
p.L1057V	0	530	95.15	rs3809976	78	43
p.T891I	0	550	98.74	rs3826593	91	59
p.K2T	0.638	487	87.43	rs6566987	82	97
p.G293S	0	525	94.25	rs1020694	80	94
p.A2223V	0.03	279	50.09	rs1044009	78	47
p.S313P	0.005	536	96.23	rs1011320	95	96
p.G1386S	0.023	164	29.44	rs3815308	46	4
p.A65T	1	1	0.18	rs111618794	1 NA	
p.N266D	0.087	557	100	rs7249222	100	100
p.K153T	1	44	7.9	rs35106910	4	14
p.V444A	0.995	453	81.33	rs2287622	59	54
p.I4210L	1	525	94.25	rs4667591	79	14
p.K4094E	1	523	93.9	rs2075252	78	93
p.V783I	0.999	557	100	rs2230616	100	98
p.K14R	0.005	491	88.15	rs3769823	68	61
p.R123K	1	557	100	rs849541	100	88
p.V178I	1	62	11.13	rs34218846	4	8
p.D1529E	0	287	51.53	rs1881421	40	66
p.K1491R	0.818	186	33.39	rs1881420	24	23
p.I1461V	0	557	100	rs1670283	100	97
p.N453S	0.999	96	17.24	rs1800440	20	1
p.V432L	0.001	353	63.38	rs1056836	63	19
p.L44S	0.998	553	99.28	rs7422651	91	36
p.K283E	1	557	100	rs6736913	99	100
p.I382V	1	531	95.33	rs10202624	90	98
p.Q915R	0	176	31.6	rs2303424	40	65
p.G39E	0.482	62	11.13	rs1042821	17	20
p.L815P	0	557	100	rs6058694	100	100
p.I57V	0.439	546	98.03	rs1047972	81	84

p.F31I	0	216	38.78	rs2273535	20	16
p.T1568A	0	369	66.25	rs910148	98	99
p.M544T	0	262	47.04	rs1883848	36	42
p.S535L	0	41	7.36	rs2295000	18	10
p.S511N	1	265	47.58	rs2297345	31	87
p.G8V	0.001	2	0.36	rs75603675	40	33
p.H27R	0.737	407	73.07	rs1051266	56	35
p.V158M	0.925	411	73.79	rs4680	52	31
p.K230N	0	129	23.16	rs4646315	19	15
p.N796S	1	544	97.67	rs140504	87	95
p.I245V	0	557	100	rs132760	100	100
p.R182C	0.001	83	14.9	rs7285167	9	28
p.T486S	0.003	359	64.45	rs1135840	45	32
p.R365H	1	154	27.65	rs1058172	14	2
p.C296R	0.003	472	84.74	rs16947	66	41
p.P34S	1	150	26.93	rs1065852	20	9
p.Q2513R	0	506	90.84	rs1381057	66	61
p.H1201R	0.002	152	27.29	rs3218651	17	14
p.T982R	0	479	86	rs3218649	61	52
p.R66I	0	552	99.1	rs702017	100	96
p.L496V	0.987	556	99.82	rs9833275	100	100
p.P147S	0.652	557	100	rs9840993	99	59
p.P21H	0.011	106	19.03	rs28497577	8	42
p.P161A	1	7	1.26	.	0.13	NA
p.Q939K	1	460	82.59	rs2228001	59	71
p.A499V	0.001	179	32.14	rs2228000	26	8
p.A154S	1	5	0.9	rs116535717	0.4	1
p.I219V	1	316	56.73	rs1799977	34	6
p.V515L	1 NA	NA	NA	NA	NA	NA
p.H107R	1	543	97.49	rs28547534	99	100
p.I1762V	0	132	23.7	rs2454206	39	7
p.A44T	0	267	47.94	rs79298048	36	6
p.H95R	0.001	499	89.59	rs9328733	65	89
p.H132D	0	350	62.84	NA	NA	NA
p.P142R	0	79	14.18	NA	NA	NA
p.C236R	0	308	55.3	NA	NA	NA
p.S268G	0.001	353	63.38	rs71614971	33	12
p.M286T	0	351	63.02	NA	NA	NA
p.S288G	0	228	40.93	NA	NA	NA
p.K4059N	0.964	557	100	rs1280097	100	100
p.S3554A	0.502	357	64.09	rs2637777	38	76

p.I2718V	0.94	102	18.31	rs3733406	10	6
p.H1273R	0.996	444	79.71	rs328418	55	84
p.R1064G	0.999	532	95.51	rs11939575	77	89
p.V862L	0.346	532	95.51	rs1877731	77	70
p.F614L	1	532	95.51	rs367863	77	70
p.V482I	0.97	349	62.66	rs3733413	57	19
p.S404R	0.388	490	87.97	rs3733414	65	24
p.Q472H	0.002	200	35.91	rs1870377	23	8
p.A672T	1	18	3.23	rs36050417	3 NA	
p.V1822D	0.989	512	91.92	rs459552	77	99
p.V10I	0.777	220	39.5	rs1966265	18	3
p.P136L	0.011	527	94.61	rs376618	74	71
p.H890Q	1	459	82.41	rs448012	58	46
p.I66T	0.997	493	88.51	rs1494558	70	75
p.V138I	0.014	492	88.33	rs1494555	71	90
p.I356V	0.002	256	45.96	rs3194051	29	33
p.H116Y	0	54	9.69	rs3729734	5	1
p.S837F	1	357	64.09	rs2043112	43	25
p.D806N	1	509	91.38	rs702689	70	48
p.V906I	0.347	540	96.95	rs832582	80	74
p.A99V	0.987	30	5.39	rs111840875	3	0.2
p.T1224I	0.001	532	95.51	rs462779	78	35
p.Q74H	0	253	45.42	rs1230345	23	45
p.A47T	0.933	25	4.49	rs78460560	23	19
p.T60P	0.006	11	1.97	rs9502889	97	85
p.Q61P	0	9	1.62	rs9502890	97	79
p.T888N	1	552	99.1	NA NA	NA	
p.Y459D	1 NA	NA	.		49	45
p.S209C	1	5	0.9	.	52	55
p.V16A	0.055	363	65.17	rs4880	46	41
p.S270A	0.013	548	98.38	rs316019	89	84
p.L10V	0.006	296	53.14	rs114382139	51	47
p.Q86E	0	68	12.21	NA NA	NA	
p.I166T	0.971	117	21.01	NA NA	NA	
p.R169H	0.6	144	25.85	rs111965878	36	30
p.E176V	0	233	41.83	rs112069136	66	69
p.D185E	0.064	231	41.47	rs113679013	82	93
p.G199R	0.978	3	0.54	rs146061611	16	18
p.T202M	0.002	3	0.54	rs141665107	15	17
p.D251H	1 NA	NA	NA	NA	NA	
p.V255M	0.727	55	9.87	NA NA	NA	

p.E277Q	0.846	175	31.42	NA	NA	NA	
p.L300P	0.312	283	50.81	rs116537579		60	72
p.L318F	0.002	153	27.47	rs115662401		51	62
p.T351S	0	127	22.8	NA	NA	NA	
p.E176V	0	166	29.8	rs707912		40	41
p.W171L	1	18	3.23	rs41551018		5	7
p.T167S	0.007	26	4.67	rs41541519		6	7
p.D138N	0	260	46.68	rs709055		42	48
p.S121R	0	82	14.72	NA	NA	NA	
p.T320A	0.162	306	54.94	rs116185472		45	28
p.K117Q	0.691	116	20.83	rs114346832		36	32
p.S259A	0.003	437	78.46	rs1051130		54	83
p.A464T	0.002	446	80.07	rs803064		56	65
p.S442Y	0.848	126	22.62	rs17847825		11	1
p.S100G	0.992	318	57.09	rs9639168		32	20
p.K170E	0	270	48.47	.		50	50
p.C171Y	0.978	44	7.9	NA	NA	NA	
p.R164G	0	146	26.21	rs10899750		72	94
p.N21D	0	107	19.21	rs9282564		9	0.41
p.V791M	0	412	73.97	rs532841		49	61
p.A10E	0.791	NA	NA	rs144792804		6	1
p.T260I	0	497	89.23	rs3816747		94	86
p.N255D	0.001	434	77.92	rs11203494		83	80
p.Q254H	0	396	71.1	rs11203495		79	53
p.D2378H	0.999	7	1.26	NA	NA	NA	
p.T1507A	0	417	74.87	rs6558399		77	73
p.Y744C	1	158	28.37	rs12543389		18	2
p.K544E	0	441	79.17	rs13255110		69	59
p.D202N	0.996	365	65.53	rs13260439		50	4
p.C1367R	0.394	248	44.52	rs1346044		26	16
p.R1985H	0.948	NA	NA	NA	NA	NA	
p.P1377S	0.006	21	3.77	rs61751542		2	0.41
p.N59S	0.993	420	75.4	rs968733		100	65
p.V56I	1	366	65.71	rs7023954		40	44
p.D249N	1	NA	NA	NA	NA	NA	
p.N413S	0.402	280	50.27	rs10970979		28	20
p.T293I	1	388	69.66	rs3780135		95	35
p.T1943I	1	NA	NA	NA	NA	NA	
p.N455S	0.001	494	88.69	rs1982151		73	62
p.P204H	0.805	NA	.			1	NA
p.P1315L	1	211	37.88	rs357564		34	22

p.H45L	0.998	260	46.68	rs1180895	24	5
p.T473I	0.069	380	68.22	rs6622126	43	86
p.F111L	0.241	553	99.28	rs4830173	100	100
p.G379R	1	22	3.95	NA	NA	NA
p.G115R	1	8	1.44	NA	NA	NA
p.A78T	1 NA	NA	NA	NA	NA	NA
p.G335E	0.796	87	15.62	rs41309679	8	1
p.A192T	0.006	439	78.82	rs5909299	91	16
p.T733K	0.086	130	23.34	rs2230018	12	2
p.K1288R	0.97	557	100	rs3122407	100	95



oneKGenome	ESP_ALL_MAF	ESP_EA_MAF	ESP_AA_MAF	Ref_reads	Var_reads	VAF	
	1	2.6512	0.2724	1.8453	33	41	55.41
	23	31.314	15.5016	25.9573	69	45	39.47
	32	34.686	12.1652	27.0567	50	59	54.13
NA	NA	NA	NA	36	10	21.74	
NA	NA	NA	NA	32	11	25.58	
	32	NA	NA	148	36	19.57	
	81	7.8372	45.143	20.4752	0	61	100
	43	48.3023	44.167	46.9014	164	160	49.38
	24	NA	NA	106	66	38.37	
	0.46	NA	NA	135	43	24.16	
	1	2.186	0.4085	1.5839	429	416	49.23
	1	1.407	0.3858	1.061	82	54	39.71
	7	9.2105	9.5189	9.3094	71	89	55.62
	65	46.0349	37.8348	43.257	83	81	49.39
	12	24.1279	4.4031	17.4458	66	62	48.44
	36	37.4767	47.5034	40.8734	84	84	50
	71	36.8256	29.9591	34.4995	0	69	100
	92	2.422	11.4441	5.4803	0	354	100
	55	48.3953	42.3706	46.355	104	114	52.29
	2	3.314	0.5447	2.3758	51	38	42.7
	77	22.4884	39.9682	28.41	0	287	100
	100	0	1.9292	0.6535	0	106	100
	17	19.4767	18.7018	19.2142	92	56	37.84
	56	49.8953	25.6922	41.6961	59	75	55.97
	15	18.4535	10.3268	15.7004	44	52	54.17
	2	1.6395	1.5887	1.6223	52	68	56.67
	45	25.6163	35.0204	38.9513	88	62	41.33
	8	14.0181	2.8396	10.2323	51	46	47.42
	100	NA	NA	NA	0	281	100
	98	3.6288	0.6361	2.6154	0	163	100
	85	0.4073	26.2381	9.1567	0	84	100
	32	33.1667	41.9626	36.0837	68	51	42.86
	79	15.2278	30.6443	20.1759	0	96	100
	86	14.5056	24.6214	17.7123	0	190	100
	80	19.2354	8.7642	15.8212	0	143	100
	0.46	0.5132	0.1187	0.3968	14	17	54.84
	78	49.4651	9.3963	36.5985	24	24	50
	30	33.5156	12.0744	26.2498	34	50	59.52
	33	31.1163	12.2787	24.7347	23	24	51.06
	80	18.2558	39.9682	25.6113	0	224	100

	99	0.035	3.8076	1.3113	0	32	100
	30	23.7785	30.687	39.1857	65	80	55.17
	62	34.2907	27.6214	47.1936	55	53	49.07
	34	39.9312	35.5764	38.5896	94	79	45.66
	71	13.6194	44.8025	27.7069	0	376	100
	71	13.6163	44.7571	27.718	0	161	100
	8	15.6279	3.2456	11.4332	188	138	42.33
	8	6.0465	5.4017	5.8281	82	71	46.41
	97	0.0233	10.1453	3.4523	0	159	100
	28	29.8068	17.8945	25.7737	93	77	45.29
	1	1.907	0.2724	1.3532	94	71	43.03
	55	34.4884	47.139	38.7727	34	45	56.96
	6	10.6512	8.7154	9.9954	37	16	30.19
	98	0.0485	8.3996	2.6688	0	123	99.19
	14	NA	NA	NA	46	41	47.13
	85	14.8721	14.4803	14.7394	1	121	99.18
	99	0	4.108	1.3917	0	36	100
	39	39.1047	48.9787	43.1416	45	50	52.63
NA		0.0581	0	0.0384	75	82	52.23
	100	0	0.749	0.2537	0	109	100
	100	0	0.749	0.2537	0	84	100
	30	29.0306	11.6877	23.2613	0	32	96.97
	100	0.0123	0.3002	0.1069	0	58	95.08
	22	23.0698	4.9478	16.9306	112	112	50
	95	4.4767	10.985	6.6815	0	105	100
	94	4.4884	11.5297	6.8738	0	109	100
	90	NA	NA	NA	0	142	99.3
	95	4.4767	10.8942	6.6508	0	111	100
	89	5.5116	29.2556	13.5553	0	38	100
	93	4.5846	11.1893	6.8231	0	124	100
	65	NA	NA	NA	23	124	84.35
NA		NA	NA	NA	121	24	16.55
NA		NA	NA	NA	96	22	18.64
	94	3.3023	13.232	6.6662	0	120	100
	66	22.9884	49.8184	32.0775	78	51	39.53
	23	18.8605	20.6537	19.4679	41	40	49.38
	57	38.5116	34.3395	47.7087	301	228	43.1
	34	NA	NA	NA	49	63	56.25
	24	28.5898	12.8928	23.3209	91	78	46.15
	98	0.0582	6.4714	2.2304	0	91	100
	31	48.604	14.5937	38.9248	41	26	38.81

32	29.6417	11.3987	23.4615	362	339	48.36
25	37.8227	21.5811	32.3231	36	60	62.5
1	1.907	0.3408	1.3767	146	151	50.84
63	41.0554	44.1163	46.1312	29	30	50.85
18	5.5233	39.5143	17.0383	62	77	55.4
21	11.8937	33.2726	19.1299	83	84	50
9	10.3284	2.2923	7.6035	101	76	42.94
9	10.5814	2.3377	7.7887	54	68	55.74
7	11.1395	4.1307	8.7652	59	72	54.96
51	41.2607	30.3653	37.9316	89	71	44.38
89	8.6628	5.7195	7.6657	97	99	50.51
24	21.1744	45.9374	32.3159	53	34	39.08
0.05	0.0729	0.0526	0.0665	42	41	48.81
62	30.407	18.5883	26.4032	76	54	41.54
77	17.1611	43.6581	25.3888	90	67	42.68
85	11.6306	12.7215	12	60	58	49.15
55	49.2553	48.8171	49.9076	202	197	49.25
39	35.932	32.1201	34.642	204	228	52.53
23	46.195	10.6233	34.157	0	77	100
29	38.4275	9.4773	28.6275	0	114	100
29	38.9509	9.5271	28.9935	0	120	99.17
26	1.5581	1.7735	1.631	60	40	40
26	1.314	1.5007	1.3771	61	40	39.6
51	40.4773	26.5435	48.3431	52	41	44.09
15	9.093	22.0756	13.4831	43	31	41.89
89	0.2094	37.1812	12.7137	0	249	100
77	15.0602	23.1231	17.7912	1	99	99
43	37.0979	32.4108	47.3904	0	187	98.94
1	0.7209	0.1592	0.5309	79	88	52.69
61	38.1047	31.9609	48.2302	1	137	99.28
5	6.6423	1.4321	4.9961	0	22	100
3	7.7147	1.3452	5.5624	32	26	44.07
NA	NA	NA	NA	750	240	24.19
4	11.0116	2.1108	7.9963	28	23	45.1
49	34.3488	23.4453	48.6468	0	156	99.36
74	10.5814	2.5874	7.8733	0	183	100
16	26.093	8.1026	19.9985	80	58	42.03
8	12.5814	11.8248	12.3251	135	145	51.6
67	41.1047	33.8856	38.6591	92	74	44.31
80	6.1047	48.5247	20.4752	0	106	100
46	46.0214	29.4598	40.4092	197	164	45.18

60	25.4767	40.5086	36.9963	212	231	51.91
75	11.7093	29.7322	17.8149	1	205	99.51
10	12.0233	2.4512	8.7806	0	171	100
68	48.6583	26.2162	41.3465	57	42	42.42
53	NA	NA	NA	669	558	45.48
14	20.4419	14.798	18.5299	395	444	52.73
18	19.6744	13.7086	17.6534	79	65	45.14
77	30.107	6.7635	22.1966	38	44	53.66
90	19.5	11.3936	16.7538	37	44	54.32
7	13.6279	6.8997	11.3486	55	63	53.39
6	13.6279	3.064	10.0492	52	32	38.1
70	23.2442	48.3432	31.7469	58	45	43.69
70	23.1628	48.2978	31.6777	24	19	44.19
81	7.3721	31.7295	15.6236	0	85	100
92	20.6186	5.0302	15.8069	54	60	52.63
85	20.8256	6.9905	16.1387	0	189	100
63	19.8716	39.7016	26.5104	0	57	100
96	5.1885	3.1108	4.4846	114	108	48.65
28	43.2499	9.3624	32.1217	0	70	100
0.18	0.1695	0	0.115	71	70	49.65
100	NA	NA	NA	0	84	100
7	4.2209	12.1198	6.8968	36	38	51.35
59	39.9635	41.4309	40.4225	69	70	50.36
54	20.9884	24.217	39.551	1	220	99.55
74	24.1628	11.4843	19.8678	0	306	100
99	0.0233	2.6555	0.915	0	155	100
65	29.4118	40.8385	33.0529	0	155	100
97	0.1047	10.5084	3.6291	0	140	100
4	5.314	8.0799	6.251	368	309	45.58
57	34.3837	39.1512	43.3492	37	37	50
40	21.3605	24.2397	22.3358	103	83	44.62
99	0.0349	2.7916	0.9688	0	206	100
10	18.6977	3.3364	13.4938	80	59	42.45
61	44.407	23.0141	44.5564	79	61	43.57
74	10.3328	41.6704	26.6	0	189	100
99	1.686	0.3631	1.2379	0	176	100
85	9.6534	5.5606	8.2667	0	145	100
50	36.1627	41.7237	42.9194	58	61	51.26
19	17.2432	19.3961	17.965	52	41	43.62
100	0	0.0227	0.0077	0	824	100
84	16.2442	16.1825	16.2233	0	136	100

	32	21.686	13.232	18.8221	80	73	47.71
	99	1.3594	0.3872	1.0308	0	45	100
	47	41.2963	42.8507	41.8231	131	97	42.54
	20 NA	NA	NA		92	71	43.56
	52	30.8605	20.9487	47.1859	84	70	45.16
	27 NA	NA	NA		0	31	100
	49	43.0533	39.1473	49.0678	0	207	99.52
	39	47.8488	31.4798	45.1484	0	148	100
	17 NA	NA	NA		58	42	42
	80	15.407	6.128	12.2636	0	189	100
	100 NA	NA	NA		0	89	100
	11	7.7032	28.2546	14.4764	26	55	67.9
	40	43.1131	36.3595	40.832	186	170	47.75
	7 NA	NA	NA		180	92	33.82
	66	34.2718	48.6566	40.0447	0	119	100
	26	22.3773	11.9536	18.853	94	118	55.66
	72	31.7558	35.5424	33.0386	0	152	100
	28	14.6628	10.4857	13.2477	49	36	42.35
	67	37.1365	44.7751	39.7231	0	87	100
	99	0	3.3591	1.1379	0	121	100
	100	0	0.4085	0.1384	0	150	100
	88	0.7093	36.2687	12.7557	0	170	100
	16	9.3837	36.2687	18.4915	91	88	49.16
	0.14	1.2558	0.3177	0.938	177	166	48.4
	66	39.8517	27.0121	35.7938	0	64	100
	25	24.8185	9.1837	20.0583	27	25	48.08
	0.41	0.2908	0.371	0.316	54	60	52.63
	17	32.0233	9.2147	24.2965	15	25	62.5
NA	NA	NA	NA		109	110	50.23
	100	1.0924	0.193	0.7957	0	146	100
	24	37.335	8.9595	28.7341	314	301	48.94
	20	40.0233	11.5297	30.3706	318	266	43.61
	73	31.6047	8.4884	23.7736	322	233	40.17
NA	NA	NA	NA		362	250	40.65
NA	NA	NA	NA		458	176	27.63
NA	NA	NA	NA		379	199	34.31
	21	37.2732	14.0263	29.3955	314	302	48.71
NA	NA	NA	NA		341	260	42.83
NA	NA	NA	NA		321	266	45.08
	100	0.6409	0.1305	0.4793	0	184	100
	55	36.1723	29.236	47.1603	0	155	100

	7	7.3848	6.9341	7.2431	54	54	50
	68	44.7759	18.7333	36.6237	0	189	100
	73	20.4219	10.6928	17.2777	0	152	100
	68	20.4268	26.5817	22.4477	0	63	100
	68	20.4734	25.3209	21.9923	0	131	100
	36	42.2764	22.2086	46.0717	0	55	100
	43	34.7093	30.172	45.6843	0	158	100
	23	23.686	11.0077	19.3911	286	195	40.46
	3	2.9651	0.6128	2.1682	113	99	46.7
	86	23.5	5.4066	17.3743	46	56	54.9
	25	22.6977	6.1053	17.0767	60	60	50
	81	24.8023	33.931	27.8948	0	132	100
	53	37.6629	45.4504	40.2987	26	24	48
	61	32.6395	24.5801	29.9093	82	84	50.6
	67	31.9884	12.5965	25.419	131	127	49.22
	23	26.407	34.9977	29.3172	50	55	52.38
	2	4.8301	1.1818	3.5945	124	147	54.24
	39	40.1047	28.5293	36.1833	87	82	48.52
	50	27.9161	49.4635	34.9883	0	112	100
	70	17.7177	25.4627	20.153	0	131	100
	2	2.8605	0.7944	2.1605	126	151	54.51
	59	18.7209	44.0282	31.3365	0	85	100
	30	27.686	45.1884	33.6153	86	82	48.81
	24	16.7988	13.2434	15.6164	48	50	51.02
	84	2.4335	12.3197	5.5983	0	67	100
	83	2.6414	13.1225	5.9524	0	64	100
NA	NA	NA	NA		145	77	34.68
	48	NA	NA	NA	130	86	39.45
	53	NA	NA	NA	152	53	25.85
	37	49.9535	41.9655	47.3085	0	26	100
	88	10.7907	14.6164	12.0867	0	175	100
	52	48.1024	47.5261	47.907	1	413	99.76
NA	NA	NA	NA		59	198	77.04
NA	NA	NA	NA		12	125	91.24
	33	NA	NA	NA	15	127	89.44
	74	NA	NA	NA	26	131	82.91
	90	16.395	9.8143	14.0361	0	185	99.46
	14	NA	NA	NA	132	96	42.11
	14	NA	NA	NA	132	108	45
NA	NA	NA	NA		157	70	30.84
NA	NA	NA	NA		176	73	29.32

NA	NA	NA	NA	29	328	91.88	
	67	39.7195	30.3441	36.3626	0	604	100
	53	46.4747	47.5812	48.6012	0	461	100
NA	NA	NA	NA	0	368	100	
	41	NA	NA	NA	140	91	39.39
	7	4.4463	4.516	4.4699	149	83	35.62
	8	4.9443	5.343	5.0786	154	83	35.02
	47	42.6689	45.6187	43.649	130	94	41.96
NA		40.3573	40.033	40.2551	80	97	54.8
	33	44.9612	31.1714	40.0237	41	54	56.25
	38	36.9419	37.7667	37.2213	61	72	54.14
	58	45.3488	22.3786	37.5673	120	101	45.5
	56	43.3837	40.7853	42.5035	0	47	100
	11	10.7326	2.5647	7.9656	41	45	52.33
	32	33.7646	19.4697	29.2889	0	136	100
	50	NA	NA	NA	248	345	58.08
NA	NA	NA	NA	NA	447	137	23.42
	75	NA	NA	NA	0	226	99.12
	4	9.9651	2.7009	7.5042	111	91	45.05
	47	49.2558	41.103	47.4781	350	332	48.54
	3	NA	NA	NA	132	167	55.85
	81	6.6977	13.4589	8.9882	0	863	100
	87	3.5349	9.0104	5.3898	0	766	100
	66	7.0116	26.3504	13.563	0	757	99.47
NA		0.2572	1.1612	0.564	2667	556	17.23
	65	22.7802	23.3936	22.9843	237	214	47.24
	11	16.4911	4.4282	12.514	444	415	48.2
	56	30.3159	34.8386	31.831	0	570	100
	32	49.2921	10.0696	37.0092	173	167	48.97
	18	25.5814	17.1811	22.7357	121	104	46.22
NA		0.0872	0	0.0603	139	115	45.1
	1	2.3414	0.3019	1.7	111	106	48.85
	92	0.2597	24.8158	8.5844	0	39	100
	41	41.5	43.4862	42.1728	61	52	46.02
NA		0.0116	0	0.0077	93	98	51.31
	19	30.9186	22.9006	28.2024	64	57	46.72
	79	6.1886	42.0824	23.6879	0	107	100
NA		0.0465	0	0.0308	57	52	47.71
	71	26.3458	41.3817	31.4312	62	72	53.73
	0.23	0.186	0.0454	0.1384	95	94	49.74
	38	32.9818	23.8873	29.9252	270	298	52.37

	13	27.17	7.9531	20.1931	0	156	100
	71	42.9548	18.6701	43.1128	0	38	100
	100	NA	NA	NA	0	113	100
NA		NA	NA	NA	91	104	53.33
NA		NA	NA	NA	77	22	22.22
NA		NA	NA	NA	51	12	19.05
	4	8.4164	1.4863	5.8996	0	241	100
	72	11.3314	24.0959	32.2092	0	67	100
	16	11.8014	4.8501	9.2777	0	374	100
	99	0	2.6858	0.9751	0	98	100



chromosome	start	stop	reference	variant	type	gene_name
11	94225807	94225807	C	T	SNP	MRE11A
22	42524947	42524947	C	T	SNP	CYP2D6
12	121434630	121434631	-	TCATTCAT	INS	HNF1A
12	18435399	18435401	CCC	-	DEL	PIK3C2G
12	974308	974309	-	C	INS	WNK1
12	974355	974356	-	C	INS	WNK1
15	100252710	100252712	CAG	-	DEL	MEF2A
15	31776211	31776213	GGC	-	DEL	OTUD7A
17	48452978	48452979	-	AGC	INS	EME1
18	42456670	42456671	-	TCTT	INS	SETBP1
18	51795958	51795960	CGA	-	DEL	POLI
18	56203253	56203255	AAG	-	DEL	ALPK2
20	32664864	32664865	-	CAG	INS	RALY
5	56177849	56177851	CAA	-	DEL	MAP3K1
5	74886294	74886296	TTT	-	DEL	POLK
6	29912029	29912029	G	-	DEL	HLA-A
6	32191658	32191659	-	AGCAGC	INS	NOTCH4
7	90894459	90894460	-	CCG	INS	FZD1
8	48805816	48805817	-	G	INS	PRKDC
X	135956571	135956572	-	GG	INS	RBMX
X	135960146	135960147	-	AA	INS	RBMX
X	66766356	66766357	-	GGC	INS	AR

transcript_id	transcript_sp	transcript_so	transcript_ve	strand	trv_type	c_position
ENST00000040	human	ensembl	70_37		-1 splice_site	c.29+1
ENST00000036	human	ensembl	70_37		-1 splice_site	c.506-1
ENST00000054	human	ensembl	70_37		1 frame_shift_ic	c.1043_1044
ENST00000055	human	ensembl	70_37		1 in_frame_del	c.384_386
ENST00000055	human	ensembl	70_37		1 frame_shift_ic	c.2172_2173
ENST00000055	human	ensembl	70_37		1 frame_shift_ic	c.2219_2220
ENST00000035	human	ensembl	70_37		1 in_frame_del	c.1255_1257
ENST00000038	human	ensembl	70_37		-1 in_frame_del	c.2088_2086
ENST00000039	human	ensembl	70_37		1 in_frame_ins	c.409_410
ENST00000042	human	ensembl	70_37		1 frame_shift_ic	c.681_682
ENST00000057	human	ensembl	70_37		1 in_frame_del	c.42_44
ENST00000036	human	ensembl	70_37		-1 in_frame_del	c.4166_4164
ENST00000024	human	ensembl	70_37		1 in_frame_ins	c.689_690
ENST00000039	human	ensembl	70_37		1 in_frame_del	c.2822_2824
ENST00000051	human	ensembl	70_37		1 in_frame_del	c.1385_1387
ENST00000037	human	ensembl	70_37		1 frame_shift_ic	c.750
ENST00000037	human	ensembl	70_37		-1 in_frame_ins	c.48_47
ENST00000028	human	ensembl	70_37		1 in_frame_ins	c.264_265
ENST00000031	human	ensembl	70_37		-1 frame_shift_ic	c.3730_3729
ENST00000032	human	ensembl	70_37		-1 frame_shift_ic	c.906_905
ENST00000032	human	ensembl	70_37		-1 frame_shift_ic	c.316_315
ENST00000037	human	ensembl	70_37		1 in_frame_ins	c.1368_1369

amino_acid_ucsc_cons	WHISP_freq	WHISP_percent	dbSNP_id	oneKGenome	oneKGenome	
e1+1	0.004	165	29.62	rs496797	45	66
e4-1	1	102	18.31	rs3892097	19	6
p.F352fs	0.006:0.017	42	7.54	.	NA	NA
p.P129in_frame	0.001:0.000:C	346	62.12	.	40	18
p.I725fs	0.999:1.000	6	1.08	.	25	21
p.F741fs	1.000:1.000	5	0.9	.	61	48
p.Q422in_frame	1.000:1.000:1	NA	NA	.	NA	NA
p.A696in_frame	0.061:0.001:C	NA	NA	.	64	72
p.138in_frame	0.140:0.176	378	67.86	.	NA	NA
p.T227fs	0.000:0.000	89	15.98	.	NA	NA
p.D17in_frame	0.000:0.000:C	74	13.29	.	70	92
p.F1389in_frame	0.000:0.000:C	142	25.49	.	12	6
p.231in_frame	0.001:0.001	12	2.15	.	66	80
p.T945in_frame	0.958:0.952:C	470	84.38	.	NA	NA
p.F463in_frame	0.021:0.046:C	70	12.57	.	NA	NA
p.D251fs	1	192	34.47	.	NA	NA
p.17in_frame	0.002:0.003	16	2.87	.	NA	NA
p.92in_frame	0.006:0.934	74	13.29	.	37	50
p.R1243fs	0.179:0.000	551	98.92	.	100	100
p.S303fs	1.000:1.000	NA	NA	.	NA	NA
p.P105fs	1.000:1.000	26	4.67	.	NA	NA
p.460in_frame	0.027:0.039	NA	NA	.	NA	NA

oneKGenome	ESP_ALL_MAF	ESP_EA_MAF	ESP_AA_MAF	Ref_reads	Var_reads	MAF	MAF
53	NA	NA	NA	0	245	99.59	
11	19.0723	7.2854	15.1047	346	207	37.43	
NA	NA	NA	NA	19	28	59.57	
30	39.9156	21.6393	34.2379	136	82	37.61	
20	NA	NA	NA	39	35	47.3	
55	NA	NA	NA	55	48	46.6	
NA	34.7315	18.2568	29.3189	160	134	45.58	
73	37.8378	21.0526	32.1429	8	5	38.46	
NA	47.3346	32.106	45.6622	101	93	47.94	
NA	46.0321	47.575	46.56	782	465	37.29	
75	31.4791	8.8722	23.8832	49	25	33.78	
7	13.8236	6.9418	11.4795	51	33	39.29	
71	NA	NA	NA	28	79	73.83	
NA	NA	NA	NA	37	122	76.73	
NA	9.1713	1.7112	6.6294	67	60	47.24	
NA	36.8474	36.6587	36.7827	161	70	30.3	
NA	NA	NA	NA	55	15	21.43	
39	39.1085	38.5961	38.9349	47	16	25.4	
100	0.038	0.1621	0.0776	24	222	90.24	
NA	NA	NA	NA	89	34	27.64	
NA	NA	NA	NA	72	33	31.43	
NA	NA	NA	NA	80	23	22.33	