

## Whole exome sequencing in families at high risk for Hodgkin lymphoma: identification of a predisposing mutation in the *KDR* gene

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## Supplemental Methods

### Sequencing Methods

**Whole exome sequencing.** Whole exome sequencing (WES) for the HL families was performed at the Cancer Genomics Research Laboratory, National Cancer Institute (CGR, NCI), as previously described.<sup>1</sup> Briefly, 1.1 µg of genomic DNA was extracted by standard methods from blood. SeqCAP EZ Human Exome Library v3.0 (Roche NimbleGen, Madison, WI) was utilized for exome sequence capture. The captured DNA was then subject to paired-end sequencing utilizing the Illumina HiSeq2000 sequencer for 2 X 100-bp sequencing (Illumina, San Diego, CA). Exome sequencing was performed to a sufficient depth to achieve a minimum coverage of 15 reads in at least 80% of the coding sequence from the UCSC hg19 transcripts database. The actual sequencing performance in our 17 HL families was that 90% of coding sequence had > 15 reads and the average coverage was 77x. The exome data analyzed in the present paper are archived in the CGR exome build GATK\_UnifiedGenotyper\_Legacy\_Annotation dated 2014-10-20.

**Targeted sequencing.** Targeted sequencing (TS) for the HL families was performed at CGR, NCI. A targeted, multiplex PCR primer panel was designed based on the above WES data using the custom Ion Ampliseq Designer v2.2 (Thermo Fisher Scientific, Grand Island, NY). The primer panel covered 147kb and included the coding region of the selected genes, with an average coverage of 95.9%. Average amplicon size was 225bp. Sample DNA (20ng/primer pool) was amplified using this custom Ampliseq primer panel, and libraries were prepared following the manufacturer's Ion Ampliseq Library Preparation protocol (Thermo Fisher Scientific, Grand Island, NY). Individual samples were barcoded, pooled, templated, and sequenced on the Ion Torrent PGM Sequencer using the Ion PGM Template OT2 200 and Ion PGM Sequencing 200v2 kits per manufacturer's instructions. The resulting sequencing data yielded an average read depth per amplicon of 165X.

### Analyses of sequencing data

**Alignment and calling of variants.** Details of the bioinformatics pipeline for variant alignment and calling used in this study have been previously published.<sup>1</sup> Briefly, WES reads were trimmed using the Trimmomatic program<sup>2</sup>, which reported the longest high-quality stretch of each read. Only read pairs with both ends  $\geq 36$  bp were used. Reads were aligned to the hg19 reference genome using the Novoalign software version 2.07.14 (<http://www.novocraft.com>). Duplicate reads based on paired ends aligning to the same start locations due to either optical or PCR artifacts were removed from further analysis using the MarkDuplicated module of the Picard software v1.67 (<http://picard.sourceforge.net/>). Additionally, two ends of each pair had to map to the reference in complementary directions and reflect the expected fragment length ( $250 \pm 30$  bp). A local realignment around sites of insertion and deletion was performed using the RealignerTargetCreator and IndelRealigner modules from the Genome Analysis Toolkit (GATK).<sup>3</sup> Variant discovery and genotype calling of multi-allelic substitutions, insertions and deletions was performed on all individuals globally using the UnifiedGenotyper module from GATK, with minimum call quality parameter of 30 and GATK recommended quality scores recalibrations. Raw TS reads generated by the Ion Torrent sequencer were quality and adaptor trimmed by Ion Torrent Suite and then aligned to the hg19 reference sequence by TMAP

(<https://github.com/iontorrent/TS/tree/master/Analysis/TMAP>) using default parameters. Resulting BAM files were merged according to sample names and processed through an in-house quality control (QC) and coverage analysis pipeline, which generated coverage summary plots and per sample per amplicon read count heatmaps. Aligned BAM files were left aligned using GATK LeftAlignIndels module. Amplicon primers were trimmed from aligned reads by Torrent Suite. Variant calls and filtrations were made by Torrent Variant Caller 4.2 (<http://mendel.iontorrent.com/ion-docs/Torrent-Variant-Caller-Plugin.html>) and Genome Analysis Toolkit (GATK, <http://www.nature.com/ng/journal/v43/n5/full/ng.806.html>).

**Annotation of variants.** Annotation of each variant locus was made via a custom software pipeline based on public data from ANNOVAR, dbNSFP, and SnpSift integrated by CGR in-house script, including Ensembl, refGene, and UCSC KnownGene databases, the ESP6500 dataset from University of Washington's Exome Sequencing Project (<http://evs.gs.washington.edu/EVS/>), the National Center for Biotechnology Information dbSNP database<sup>4</sup> build 137, the 1000 Genomes Project<sup>5</sup>, SIFT<sup>6</sup>, PolyPhen-2 version 2.2.2<sup>7</sup>, Mutation Taster<sup>8</sup>, Mutation Assessor release 2,<sup>9</sup> FATHMM version 2.3<sup>10</sup>, and others.

**Filtering of variants.** WES variants called in discovery families were filtered and selected for additional analyses according to the following criteria: 1) present in all affected (which included both HL and NHL) or obligate carrier individuals in the pedigree and not present in unaffected married-in individuals; 2) present in  $\leq 1\%$  of families from an in-house database (CGR, NCI) of 1700 exomes in individuals from 700 families with non-hematologic cancers; 3) present in  $\leq 1\%$  of the NHLBI Exome Sequencing Project (ESP) European American population (4300 individuals) and in  $\leq 1\%$  of the phase 1 1000 Genomes Project European population (379 individuals); and 4) occurring in exonic or UTR regions or in locations linked to epigenetic findings from ENCODE.

**Prioritization of variants.** The variants in the discovery families that were kept after initial filtering were then further prioritized based on mutation type (ordered as frameshift, nonframeshift, non-synonymous, synonymous, blank); low minor allele frequency according to ESP European, 1000 Genomes and our in-house database; functional prediction for non-synonymous variants using FATHMM, Mutation Assessor, Mutation Taster, Polyphen2 Hvar, SIFT; and epigenetic findings from ENCODE. We also considered the genes' links to cancer or immune-related processes (using Ingenuity Variant Analysis, <http://www.ingenuity.com/>), and whether the variant or gene was present in multiple HL families.

**Validation and replication of variants.** A set of genes was subsequently selected based on the above prioritized list and also on amplicon length to optimize use of available resources. This gene set was re-sequenced in all available HL families using Ion Torrent as described above. We checked for technical validation in whole exome sequenced samples and for segregation in additionally sequenced family members.

**In silico analysis.** For the variants that replicated in multiple families, we conducted the following additional in silico analysis of the potential functional impact. Experimental protein structures were downloaded from the RCSB Protein Data Bank (PDB) (<http://www.rcsb.org>) and used for structure-based impact assessment analysis. The impact of the mutation on protein function was also analyzed using the Annotation, Visualization and Impact Analysis tool (AVIA) v2.0 ([avia.abcc.ncifcrf.gov](http://avia.abcc.ncifcrf.gov))<sup>11</sup>, Ensembl ([www.ensembl.org](http://www.ensembl.org)), NCBI ([www.ncbi.nlm.nih.gov](http://www.ncbi.nlm.nih.gov)) and the protein database UniProt ([www.uniprot.org](http://www.uniprot.org)). The NCBI tool Amino Acid Explorer<sup>12</sup> was

used to access the impact of the amino acid residue substitution on gene function. Discovery Studio v4.1 Visualizer (Accelrys Software Inc.) was used for analyzing the PDB files and creating protein structure figures.

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**Supplemental Table 1.** Summary of sequenced families and samples.

Family	Discovery or Replication phase	HL	NHL	Obligate Carrier	Related Unaffected	Spouse Unaffected
F1	D	3	0	2	1	1
F2	D	2	0	3	3	1
F3	D	2	1	2	7	0
F4	D	4	0	1	4	1
F5	D	3	0	1	0	1
F6*	D	3	0	1	0	0
F7	D	2	0	2	2	0
F8	D	2	0	2	0	0
F9	D	3	0	0	0	0
F10	D	3	0	1	7	1
F11	D	2	0	1	3	0
F12	D	3	0	0	0	0
F13	D	2	1	0	0	0
F14	D	3	0	0	0	0
F15	D	4	0	0	0	0
F16	D	3	0	0	0	0
F17	D	2	0	1	0	1
F18	R	2	0	0	4	0
F19	R	2	0	0	0	0
F20	R	2	0	0	0	0
F21	R	2	0	0	0	0
F22	R	2	0	0	0	0
F23	R	2	0	0	0	0
F24	R	2	0	0	0	0
F25	R	2	0	0	0	0
F26	R	2	0	0	0	0
F27	R	2	0	0	0	0
F28	R	2	0	0	0	0
F29	R	2	0	0	0	0
F30*	R	2	0	0	0	0
F31	R	2	0	0	0	0
F32	R	2	0	0	0	0
F33	R	2	0	0	0	0
F34	R	2	0	0	0	0
F35	R	2	0	0	0	0
F36	R	2	0	0	0	0
F37	R	2	0	0	0	0
F38	R	2	0	0	0	0
F39	R	2	0	0	0	0
F40	R	2	0	0	0	0
F41	R	2	0	0	0	0
F42	R	2	0	0	0	0
F43	R	2	0	0	0	0
F44	R	2	0	0	0	0
F45	R	2	0	0	0	0
F46	R	2	0	0	0	0
F47	R	2	0	0	0	0
F48	R	2	0	0	0	0
F49	R	2	0	0	0	0
F50	R	1	0	0	0	0
F51	R	1	0	0	0	0
F52	R	1	0	0	0	0
F53	R	1	0	0	0	0
F54	R	1	0	0	0	0
F56	R	1	0	0	0	0
F56	R	1	0	0	0	0
F57	R	1	0	0	0	0
F58	R	1	0	0	0	0
F59	R	1	0	0	0	0
F60	R	1	0	0	0	0
F61	R	1	0	0	0	0
F62	R	1	0	0	0	0
F63	R	1	0	0	0	0
F64	R	1	0	0	0	0
F65	R	1	0	0	0	0

\*Additional relatives were genotyped for the c.3193G>A variant in the *KDR* gene.

**Supplemental Table 2.** List of 2699 variants identified through whole exome sequencing of 17 HL discovery families according to the filtering criteria described in the methods.

CHR	Position	IDS	Reference	Variant	Gene Symbol
chr1	661906		G	C	LOC100133331
chr1	900193		G	A	
chr1	1242631		C	T	ACAP3
chr1	1904413	rs202000786	T	C	KIAA1751
chr1	2522853		G	A	FAM213B
chr1	2938416	rs137950147	T	C	ACTRT2
chr1	3662409	rs35766062	G	A	TP73-AS1
chr1	5926503	rs139022622	G	A	NPHP4
chr1	7724727	rs141259598	C	G	CAMTA1
chr1	7879366		C	A	PER3
chr1	9086387		C	T	SLC2A7
chr1	9097934	rs116780481	G	A	SLC2A5
chr1	10434267		G	A	KIF1B
chr1	11156028	rs151027964	G	C	EXOSC10
chr1	12251410		G	A	TNFRSF1B
chr1	13183837		G	C	LOC440563
chr1	16093946	rs147639515	C	T	FBLIM1
chr1	16959445		T	C	CROCCP2,FLJ00313
chr1	16972119		A	C	MST1P2
chr1	17086744		C	A	MST1P9
chr1	17257820	rs142373017	G	T	CROCC
chr1	21043941		T	G	KIF17
chr1	21071440	rs144814158	C	T	HP1BP3
chr1	21494519		T	C	EIF4G3
chr1	22030302		G	A	USP48
chr1	22173967	rs142458572	G	A	HSPG2
chr1	22419521		A	G	CDC42
chr1	23609255		G	A	HTR1D(dist_88033),HNRNPR(dist_27021)
chr1	23885773	rs146163818	A	T	ID3
chr1	25611123		C	T	RHD
chr1	25629820	rs121912763	T	G	RHD
chr1	25944238		C	T	MAN1C1
chr1	26393852		C	T	TRIM63
chr1	26608936		G	A	UBXN11
chr1	26888093		G	A	RPS6KA1
chr1	27108284		A	C	ARID1A
chr1	27224147	rs200916592	G	C	GPATCH3
chr1	27426047		T	C	SLC9A1
chr1	28293189	rs147529110	A	G	XKR8
chr1	28361983	rs112763988	T	C	EYA3
chr1	29814774		C	G	PTPRU(dist_161449),BC042538(dist_672025)
chr1	29814783		T	C	PTPRU(dist_161458),BC042538(dist_672016)
chr1	30357725		C	T	PTPRU(dist_704400),BC042538(dist_129074)
chr1	30358054	rs141499974	A	T	PTPRU(dist_704729),BC042538(dist_128745)
chr1	31349951	rs373189918	C	T	SDC3
chr1	32048902		G	A	TINAGL1
chr1	32157807		G	A	COL16A1
chr1	32204956	rs139108389	G	A	BAI2
chr1	32829219	rs374749640	C	T	TSSK3
chr1	33820711		G	A	PHC2
chr1	34684453		G	T	C1orf94
chr1	35654761	rs370753674	T	C	SFPQ
chr1	36020085	rs142411848	T	G	KIAA0319L
chr1	36367134	rs199616208	G	A	EIF2C1
chr1	36556921	rs139465196	C	T	ADPRHL2
chr1	36809771		G	A	STK40
chr1	38006163	rs147613529	G	A	SNIP1
chr1	38168805	rs187111528	G	A	CDCA8
chr1	38327788		G	T	INPP5B
chr1	39176262		C	A	LOC339442(dist_495823),RRAGC(dist_128753)
chr1	39823379	rs374553228	G	T	MACF1
chr1	39952538	rs138404184	C	T	MACF1
chr1	40209542		T	C	PPIE
chr1	40559743	rs143284035	T	C	PPT1
chr1	40723750	rs187549487	A	G	ZMPSTE24

chr1	40773387	rs144615318	C	T	COL9A2
chr1	42621305	rs201747322	C	T	GUCA2B
chr1	43826200		C	T	CDC20
chr1	43885320		C	T	SZT2
chr1	44085844	rs145235828	C	A	PTPRF
chr1	45524416	rs149382804	G	A	ZSWIM5
chr1	46309439		G	T	MAST2
chr1	46812847		C	T	NSUN4
chr1	47309993		A	G	CYP4B1(dist_24972),CYP4Z2P(dist_13913)
chr1	47882549		T	C	FOXE3
chr1	51435977	rs139867286	C	T	CDKN2C
chr1	51775076		C	G	TTC39A
chr1	51956000		G	A	EPS15
chr1	52263786		G	C	NRD1
chr1	52552811			A	BTF3L4
chr1	52882613		G		PRPF38A
chr1	52882614		G	T	PRPF38A
chr1	53377660	rs78692102	T	C	ECHDC2
chr1	53553868		C	A	SLC1A7
chr1	54056721		A	G	GLIS1
chr1	54562141	rs370390913	A	G	TCEANC2
chr1	54605242	rs41294792	G	A	CDCP2
chr1	54605698	rs147725209	C	T	CDCP2
chr1	54693723		G	A	SSBP3
chr1	55081841	rs142814457	G	C	FAM151A
chr1	55223744	rs35201073	G	C	PARS2
chr1	55282721	rs150463971	G	C	C1orf177
chr1	55617702		C	T	USP24
chr1	56989772		T	C	PPAP2B
chr1	57528562	rs141893131	C	G	DAB1
chr1	60091701	rs147949902	A	G	FGGY
chr1	62190518		C	G	TM2D1
chr1	62916781	rs62620019	T	C	USP1
chr1	63008327		G	A	DOCK7
chr1	63299472		TGT		ATG4C
chr1	65095110	rs370532570	C	G	CACHD1
chr1	65693160		A	G	AK4
chr1	65851423	rs146050826	G	A	DNAJC6
chr1	70700558			T	SRSF11
chr1	70897652	rs183429036	A	T	CTH
chr1	71478129	rs147942321	A	G	PTGER3
chr1	72748200		C	G	NEGR1
chr1	74507023	rs201361860	C	T	LRRIQ3
chr1	74819246		A		FPGT-TNNI3K,TNNI3K
chr1	75185057	rs139734636	C	T	CRYZ
chr1	76209708			AAA	ACADM
chr1	77096241		T	G	STGGALNAC3
chr1	84701660	rs144259212	C	A	PRKACB
chr1	84944975	rs145776320	C	T	RPF1
chr1	85598956	rs181947661	C	G	MIR4423
chr1	89729466		TC		GBP5
chr1	90461024		C	T	ZNF326
chr1	92177628		T	G	TGFB3
chr1	92573180	rs142006093	C	T	BTBD8
chr1	93545365		T	A	MTF2
chr1	97656890		G	A	DPYD
chr1	98186232	rs56066952	T	C	DPYD
chr1	104114315	rs12128421	C	T	AMY2B
chr1	108677689	rs569672	A	G	BC068010
chr1	109077443	rs370038770	C	G	NBPF6(dist_64183),FAM102B(dist_25528)
chr1	109534938		C	T	WDR47
chr1	109635438	rs144727464	A	G	TMEM167B
chr1	109836792	rs17036114	G	C	MYBPHL
chr1	110141590	rs377322551	G	A	MIR197
chr1	110204008	rs1063578	C	T	GSTM4
chr1	110294928	rs79849618	G	C	EPS8L3
chr1	110306433	rs72973008	C	T	EPS8L3
chr1	110655572	rs35483972	C	G	UBL4B
chr1	110917869		A	G	SLC16A4
chr1	110932065	rs188006627	T	C	SLC16A4



chr1	111136607	rs12096011	C	T	KCNA2
chr1	111147431	rs74117897	G	A	KCNA2
chr1	111437020		G	C	CD53
chr1	111492547	rs2232045	A	G	LRIF1
chr1	111825438	rs59218952	T	C	RP11-165H20.1
chr1	111891156		G	A	C1orf88
chr1	112020530		G	A	C1orf162
chr1	114380806		C	T	PTPN22
chr1	117237481		G	T	MIR320B1(dist_23032),CD2(dist_59605)
chr1	119425830	rs41301299	G	C	TBX15
chr1	119683439		G	A	AK125759,BC043601,BC063600
chr1	120049881		T	A	HSD3B1
chr1	120286604	rs148721434	C	T	PHGDH
chr1	120465262	rs17024517	C	A,T	NOTCH2
chr1	120491555		T	C	NOTCH2
chr1	120612051		C	G	NOTCH2
chr1	120839754		G	A	FAM72B
chr1	120927432		G	C,T	FCGR1B
chr1	121103817		C	A	FCGR1B(dist_167873),SRGAP2(dist_3335)
chr1	142713416		A	T	FLJ00310
chr1	142803761		C	T	BC053679
chr1	144612781		G	A	NBPF14,NBPF9
chr1	144618027	rs189951263	G	C	NBPF14,NBPF9
chr1	144822864	rs146811872	G	A	NBPF14,NBPF9,PDE4DIP
chr1	144912191	rs138598077	G	T	PDE4DIP
chr1	145039725		C	G	PDE4DIP
chr1	146736211	rs201333837	G	A	CHD1L
chr1	149369164			C	FCGR1C
chr1	149399301		A	C	Histone3
chr1	149822035		G	A	HIST2H2BC
chr1	149914601		A	C	OTUD7B
chr1	150899275	rs374583229	T	C	SETDB1
chr1	151655838	rs138859961	G	C	SNX27
chr1	151810350		A	C	C2CD4D
chr1	152276164	rs145299777	C	A	FLG
chr1	152551880			T	LCE3D
chr1	152816286	rs60975817	A	C	LCE6A
chr1	153588586		C	A	S100A14
chr1	153633660	rs201793935	C	T	SNAPIN
chr1	153766521			C	SLC27A3(dist_13888),GATAD2B(dist_10682)
chr1	153790503	rs369503904	A	G	GATAD2B
chr1	154245965	rs142150013	A	T	HAX1
chr1	154909210	rs181302437	C	T	PMVK
chr1	155109865	rs183463046	G	A	SLC50A1
chr1	155160273	rs144273480	C	A	MUC1
chr1	155194542		A	G	GBAP1
chr1	156108612		C	T	LMNA
chr1	156163860		C	A	SLC25A44
chr1	156214780	rs181264741	C	T	PAQR6
chr1	156314323		G		C1orf182
chr1	156619434		A	G	BCAN
chr1	156623110		G	A	BCAN
chr1	156698874	rs183971043	C	T	RRNAD1
chr1	156876493	rs199937671	G	A	PEAR1
chr1	156893807		C	T	LRRC71
chr1	157557685	rs374057011	T	G	FCRL4
chr1	158694850		C		OR6K3(dist_6945),OR6K6(dist_29756)
chr1	159166738	rs138675429	G	A	CADM3
chr1	159321056		C	T	BC038194
chr1	159856224		CTCTCC		CCDC19
chr1	161334734	rs370852007	C	T	C1orf192
chr1	161751865	rs72706043	G	A	ATF6
chr1	172580494		A	G	C1orf9
chr1	172633278		GTTTA		FASLG
chr1	173835293		A	G	GAS5
chr1	176833394		G	A	ASTN1
chr1	180885841	rs199998538	C	T	KIAA1614
chr1	183522050		C	T	SMG7
chr1	183602178	rs138541093	A	G	ARPC5
chr1	183907990		G	C	GLT25D2

chr1	223958080		C	T	CAPN2
chr1	224926955		G	A	CNIH3
chr1	226054120		T	C	TMEM63A
chr1	228464306		G	C	OBSCN
chr1	228465031	rs376578848	G	A	OBSCN
chr1	228785822		C	T	DUSP5P
chr1	231664272		C	T	DISC1,TSNAX
chr1	233512318	rs200129562	C	T	KIAA1804
chr1	236908011	rs34785693	C	T	ACTN2
chr1	237013784		G	A	MTR
chr1	237961363	rs138498780	C	T	RYR2
chr1	240351659	rs186039551	G	T	FMN2
chr1	245018012		GTGA		HNRNPU
chr1	245247065	rs188250034	A	G	EFCAB2
chr1	247588140	rs111400208	C	T	NLRP3
chr1	248366594	rs147274825	C	T	OR2M3
chr1	248367306	rs41304018	T	A	OR2M3
chr1	248604453		T	C	OR2T1(dist_34048),OR2T2(dist_11646)
chr1	248604458		T	A	OR2T1(dist_34053),OR2T2(dist_11641)
chr1	248801976		AT		OR2T35
chr1	248801976		AT		
chr1	248814215	rs369477862	A	G	OR2T27
chr1	249105924		C	T	SH3BP5L
chr1	249150876		G	A	ZNF692
chr2	3461440		A	T	TRAPPC12
chr2	3488627		C	T	TRAPPC12
chr2	3604625		C	G	RNASEH1
chr2	3718749	rs377010333	T	C	ALLC
chr2	8370376		A	G	LINC00299
chr2	8452764		G	A	LINC00299
chr2	9545015			T	ASAP2
chr2	9558835		A	G	ITGB1BP1
chr2	10282635		G		C2orf48
chr2	10987378	rs114622572	C	T	PDIA6(dist_9275),KCNF1(dist_64685)
chr2	11943038	rs139638471	A	G	LPIN1
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chr2	20100600	rs7569132	A	G	TTC32
chr2	20133009	rs78685213	C	T	WDR35
chr2	20133113	rs16987240	A	C	WDR35
chr2	20449068	rs13394115	A	G	PUM2
chr2	21230446	rs145777339	G	A	APOB
chr2	23872341		G	A	KLHL29
chr2	24307325		G	A	TP53I3
chr2	26358452	rs116071189	C	T	RAB10
chr2	26407002		G	A	FAM59B
chr2	26534345	rs149872767	G	A	GPR113
chr2	26612523		A	G	EPT1
chr2	26618658	rs185959585	G	A	EPT1
chr2	27165501		C	T	DPYSL5
chr2	27435096	rs201195587	G	A	C2orf28
chr2	27436441		C	T	C2orf28
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chr2	29093212	rs142756911	C	T	TRMT61B
chr2	29124976	rs147302501	C	T	WDR43
chr2	29136869	rs143803602	T	C	WDR43
chr2	29259606	rs150485801	C	T	FAM179A
chr2	29404678	rs370184992	G	A	CLIP4
chr2	31560594	rs148477626	A	G	XDH
chr2	31621467	rs72549370	G	A	XDH
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chr2	32843239	rs191715743	C	T	BIRC6
chr2	33745697	rs11680495	T	C	RASGRP3
chr2	37195639		G	T	STRN(dist_2024),HEATR5B(dist_12514)
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chr2	43787691	rs74774554	G	C	THADA
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chr2	44172450	rs78949539	C	A	LRPPRC
chr2	44174906	rs200017171	G	A	LRPPRC
chr2	44512579	rs113187530	C	T	SLC3A1
chr2	44548776	rs75225457	C	T	PREPL
chr2	44549967	rs74395208	T	C	PREPL
chr2	44550435	rs111438719	T	C	PREPL
chr2	44566301	rs113272276	T	C	PREPL
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chr2	47045327		TT		LOC388948
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chr2	54175508	rs375628884	T	C	PSME4
chr2	54848701		G	T	SPTBN1
chr2	54865055		A	G	SPTBN1
chr2	55122156		C	T	EML6
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chr2	63272890		ACAG		EHBP1
chr2	70314833	rs201027527	A	G	PCBP1
chr2	71256747		G	A	OR7E91P
chr2	71780972	rs139754493	A	G	DYSF
chr2	72959997	rs116506302	A	G	EXOC6B
chr2	73195646	rs149206069	G	A	SFXN5
chr2	73715990	rs149221453	A	G	ALMS1
chr2	74425701	rs11687991	C	T	MTHFD2
chr2	74542203		G	T	SLC4A5
chr2	74650204		T	C	WDR54
chr2	74689335	rs200508287	G	T	MOGS
chr2	74901705	rs149579506	G	A	SEMA4F
chr2	79314040	rs139007123	T	G	REG1B
chr2	85626333	rs11539103	G	A	CAPG
chr2	85922478	rs149497301	G	A	GNLY
chr2	89339975	rs371315676	C	T	abParts
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chr2	96517473		C	T	AL832439(dist_24744),DKFZp667P0924(dist_4283)
chr2	96676314		T	C	LOC729234
chr2	96686643		C	T	LOC729234
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chr2	103433059		A	G	TMEM182
chr2	106016066	rs79858788	C	T	FHL2
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chr2	114472480		G	T	SLC35F5
chr2	114500349	rs150656361	C	T	SLC35F5
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chr2	122227361	rs140922400	C	T	CLASP1
chr2	122485435	rs148836478	C	T	MKI67IP
chr2	128264098		G	T	IWS1
chr2	128283956		G	C	IWS1
chr2	128703002		ATG		SAP130
chr2	130807214		T	C	LOC440905
chr2	131182409	rs150184675	T	C	AK054990
chr2	131415284		C	T	DQ580140
chr2	131809514		G	C	FAM168B
chr2	131809733		G	C	FAM168B
chr2	132202141		T	C	LOC401010
chr2	132381480		C	G	POTEKP
chr2	132905418		C	G	ANKRD30BL
chr2	133070838		C	T	AK094599
chr2	140584054	rs149085741	A	T	LOC647012(dist_927310),LRP1B(dist_404942)
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chr2	152322505		A	G	RIF1
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chr2	152536573		TAAAG		NEB
chr2	159499113	rs144866707	A	G	PKP4
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chr2	160840584	rs149133741	C	A	PLA2R1
chr2	161056502	rs142746188	C	T	ITGB6
chr2	163215632		A	G	GCA
chr2	164450219		G	A	KCNH7(dist_754962),FIGN(dist_13899)
chr2	165765194	rs141629634	C	A	SLC38A11
chr2	165801107		C	T	SLC38A11
chr2	166671089		G	A	GALNT3(dist_19920),LOC100506124(dist_42897)
chr2	166929252	rs191249847	A	C	SCN1A
chr2	169547460		T	C	CERS6
chr2	170048536		T	C	LRP2
chr2	170163703	rs16856796	T	C	LRP2
chr2	176964969		C	T	HOXD12
chr2	178416151		G	C	TTC30B
chr2	179301107		A	C	MIR548N
chr2	179319876		G	A	MIR548N
chr2	179571086		C	T	TTN
chr2	179584318		A	C	TTN
chr2	179659844	rs375448572	G	A	TTN
chr2	179997002	rs200034284	T	C	SESTD1
chr2	186657192	rs199853325	A	G	FSIP2
chr2	187361634	rs187308114	T	C	ZC3H15
chr2	191073525	rs77652701	T	C	HIBCH
chr2	191375151		G	A	TMEM194B
chr2	191766555	rs72907234	G	A	GLS
chr2	191864525		C	T	STAT1
chr2	192863715	rs147802393	T	G	TMEFF2
chr2	196681546	rs139963761	G	A	DNAH7
chr2	196728886	rs200303203	A	G	DNAH7
chr2	197631303	rs144612738	G	C	GTF3C3
chr2	197645346	rs147949431	T	C	GTF3C3
chr2	200525393		C	T	BC035629(dist_1538),FONG(dist_157690)
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chr2	203103252		A	C	SUMO1
chr2	203164943	rs200416852	T	C	NOP58
chr2	207169386		A	C	ZDBF2
chr2	207171050	rs192447754	G	A	ZDBF2
chr2	208619340		TTTTG		CCNYL1
chr2	209315484		A	G	PTH2R
chr2	210887737	rs151211217	G	A	C2orf67
chr2	211070473	rs76781609	C	T	ACADL
chr2	216237021		G	A	FN1
chr2	216297896	rs188436638	C	A	FN1
chr2	216877865		C	T	MREG
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chr2	217544110		C	G	IGFBP5
chr2	217735689		G	A	hCG_1816075
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chr2	220197953	rs377218851	A	G	RESP18
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chr2	225343040	rs61743301	G	A	CUL3
chr2	226447306	rs9917362	G	A	NYAP2
chr2	226447507	rs9917363	C	T	NYAP2
chr2	226516185		G	A	NYAP2
chr2	227773321	rs201648418		CC	RHBDD1
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chr2	227867546	rs151077907		TCTT	COL4A4
chr2	227912248	rs77277077	C	T	COL4A4
chr2	227916977	rs74768770	A	G	COL4A4

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chr2	230634186	rs199953208	A	T	
chr2	230680168		C	G	TRIP12
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chr2	232127058		G	A	ARMC9
chr2	232356279		C	A	NCL(dist_27074),LINC00471(dist_16858)
chr2	232389770		T	G	NMUR1
chr2	232651035		G	A	COPS7B
chr2	234294529	rs80287493	G	A	DGKD
chr2	234344820	rs115773423	C	G	DGKD
chr2	234590399		T	C	UGT1A10,UGT1A8,UGT1A9
chr2	234663274	rs183484892	C	T	LOC100286922
chr2	234761146		C	T	HJURP
chr2	236716551		T	C	AGAP1
chr2	238005476	rs34344319	C	T	COPS8
chr2	239894759	rs114179668	T	C	FLJ43879(dist_46794),HDAC4(dist_75105)
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chr2	242611988		C	T	AKO27332
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chr2	242814114		C	G	CXXC11
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chr3	3144191	rs76448231	C	A	IL5RA
chr3	4403383	rs114640216	T	C	SUMF1
chr3	4711151		T	G	ITPR1
chr3	4792482	rs186852629	T	C	EGOT
chr3	5257909	rs139745426	A	G	EDEM1
chr3	7732979		T	C	GRM7
chr3	9518174		A		SETD5
chr3	9862416	rs148535110	A	C	ARPC4-TTLL3,TTLL3
chr3	9918818	rs150971509	C	T	CIDEC
chr3	12858966	rs17037287	G	T	CAND2
chr3	17447837			A	TBC1D5
chr3	23847223		G	A	UBE2E1
chr3	25675431	rs370757931	A	C	TOP2B
chr3	26751947	rs147915275	C	T	LRRC3B
chr3	33406279	rs187069734	G	A	FBXL2
chr3	33430221	rs147500650	G	A	UBP1
chr3	33538614		AG		CLASP2
chr3	36897396		T	C	TRANK1
chr3	38023664		A	G	CTDSPL
chr3	38024567		A	G	CTDSPL
chr3	38025504		A	G	CTDSPL
chr3	38530980	rs62239943	G	A	ACVR2B
chr3	38603991	rs41311127	A	G	SCN5A
chr3	38793943	rs112774699	G	A	SCN10A
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chr3	41266714	rs201364335	G	C	CTNNB1
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chr3	43391833		A	G	SNRK
chr3	44688069	rs190037367	A	G	ZNF197
chr3	44967610	rs191480973	C	T	ZDHC3
chr3	45989595		A	G	CXCR6
chr3	46487942	rs146002167	C	T	LTF
chr3	46713680	rs148591844	C	A	ALS2CL
chr3	47933156	rs147606905	A	G	MAP4
chr3	48266835	rs55708841	A	G	CAMP
chr3	48556929		C	T	PFKFB4
chr3	49169005		G	A	LAMB2
chr3	49757086	rs142839075	G	C	AMIGO3
chr3	50385098		C	T	NPRL2
chr3	51312518	rs185670675	C	T	DOCK3
chr3	52521549	rs146374405	G	A	NISCH
chr3	52525027		T	A	NISCH
chr3	52548194	rs139838594	C	T	STAB1
chr3	52730184		T	C	GLT8D1
chr3	52836800	rs146710819	A	G	ITIH3
chr3	53099252		TTC		SFMBT1(dist_19161),RFT1(dist_23249)

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chr3	66431059	rs140051529	G	C	LRIG1
chr3	69071301	rs145198215	G	T	TMF1
chr3	70927654		G	A	BC015590(dist_863203),FOXP1(dist_76211)
chr3	72799481	rs144498351	C	T	SHQ1
chr3	73006571	rs184524409	A	G,T	GXYLT2
chr3	74312039	rs182251992	C	G	CNTN3
chr3	75472471	rs111651848	A	G	FAM86DP
chr3	75648291		C	A	FAM86DP(dist_164025),MIR1324(dist_31623)
chr3	75671113		G	A	FAM86DP(dist_186847),MIR1324(dist_8801)
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chr3	97711642		G	A	GABRR3
chr3	98110498	rs73854832	G	C	OR5K3
chr3	98110766	rs73854833	G	C	OR5K3
chr3	98379153	rs78372848	A	C	CPOX(dist_66698),ST3GAL6(dist_71999)
chr3	98379673		T	C	CPOX(dist_67218),ST3GAL6(dist_71479)
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chr3	100065063		C	T	NIT2
chr3	101477105		A	T	CEP97
chr3	101578209		G	T	NFKBIZ
chr3	105412384		G	A	CBLB
chr3	108391551		A	G	DZIP3
chr3	109136997	rs188917685	T	A	FLJ25363
chr3	111342669		C	T	CD96
chr3	112643668		A	G	CD200R1
chr3	113022889	rs185086524	G	A	WDR52
chr3	113530851		G	T	ATP6V1A
chr3	114018573		G	T	TIGIT
chr3	114057498		A	T	
chr3	119536003		A	G	NR1I2
chr3	120118543	rs143943080	G	A	FSTL1
chr3	120461251	rs191940068	A	C	RABL3
chr3	122598200		A	G	DIRC2
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chr3	127540479		G	A	MGLL
chr3	127770893		T	A	SEC61A1
chr3	127788409	rs147269024	C	T	SEC61A1
chr3	129815832		G	A	ALG1L2
chr3	130405297	rs188472605	C	T	PIK3R4
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chr3	138338588	rs199822647	G	A	FAIM
chr3	138431169	rs3729774	A	G	PIK3CB
chr3	139096812		C	G	COPB2
chr3	143100992	rs368676426	C	T	SLC9A9
chr3	143708818		A	G	C3orf58
chr3	145791085		T	C	PLOD2
chr3	148583269		G	A	CPA3
chr3	148757909	rs61750365	A	G	HLTF
chr3	148804319		C	A	HLTF
chr3	149212705	rs141032156	G	T	TM4SF4
chr3	149468948		T	C	COMMD2
chr3	150911475		T	A	MED12L
chr3	151165425	rs371735907	C	T	IGSF10
chr3	151491507	rs73869693	A	G	LOC201651,MIR548H2
chr3	151535146	rs73869697	A	T	MIR548H2
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chr3	155755166		C	T	GMPS(dist_99646),KCNA1(dist_83171)
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chr3	158523042		A	G	MFSD1
chr3	160939717		A	G	NMD3
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chr3	164767650		A	G	SI
chr3	164904719			TT	SLITRK3
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chr3	167322164	rs146804108	G	A	WDR49

chr3	167401811	rs190941416	C	A	PDCD10
chr3	167761315		G	C	GOLIM4
chr3	169496439		T	C	MYNN
chr3	169558000		C	T	LRR31
chr3	169894667	rs76506725	T	A	PHC3
chr3	170800024	rs200688648	C	A	TNIK
chr3	171427107	rs150190513	A	G	PLD1
chr3	178525047		T	C	AF279780,AY769439
chr3	179066350		G	A	MFN1
chr3	180041553	rs190998467	G	C	PEX5L(dist_287036),TTC14(dist_278365)
chr3	180465880		A	C	DKFZp434A128
chr3	182858293	rs373069827	G	A	LAMP3
chr3	183027276		G	A	MCF2L2
chr3	183750666		G	A	HTR3D
chr3	183899155		TTCGCTC		AP2M1
chr3	183960782			G	
chr3	184010349	rs189673882	G	A	ECE2
chr3	184019513		G	T	PSMD2
chr3	184035903	rs146749564	C	T	EIF4G1
chr3	184295518	rs144538024	C	T	EPHB3
chr3	184402299		C	T	TRNA_Asp(dist_36134),MAGEF1(dist_25856)
chr3	184797609	rs149888444	T	C	C3orf70
chr3	185407290	rs200474932	C	T	IGF2BP2
chr3	185416269	rs189077224	T	C	IGF2BP2
chr3	185979649		C	T	DGKG
chr3	186288033		T	C	DNAJB11,TBCCD1
chr3	186394861	rs143855375	C	T	HRG
chr3	186442850	rs200328706	C	G,T	KNG1
chr3	186506312		T	C	EIF4A2
chr3	186943152	rs147189329	G	A	MASP1
chr3	188464149		C	T	LPP
chr3	191075922		T	A	CCDC50
chr3	193386159	rs140403973	C	T	OPA1
chr3	194865541		G	A	XXYL1
chr3	195704017		G	A	SDHAP1
chr3	195954872	rs73212135	G	A	OSTalpha
chr3	197348388		C	A	LOC220729
chr3	197428797		G	C	KIAA0226
chr3	197907794		T	G	FAM157A
chr4	144940		G	A	ZNF718
chr4	264523		G	A	ZNF732
chr4	299412	rs146555192	C	G	ZNF732
chr4	493032		G	A	PIGG,ZNF721
chr4	843536	rs55904229	G	A	GAK
chr4	1246403		T	G	C4orf42
chr4	1645943	rs187673629	C	T	FAM53A
chr4	1696648		C	T	SLBP
chr4	1737302	rs148008952	G	A	TACC3
chr4	2252795			CGCCCCA	MXD4
chr4	2339177	rs139692789	C	T	ZFYVE28
chr4	3101010	rs371965675	A	G	HTT
chr4	3107375		C	T	HTT
chr4	4128316		G	A	BC042823(dist_51533),OTOP1(dist_62214)
chr4	4283543	rs139653868	G	A	LYAR
chr4	5733222	rs371671127	C	T	EVC
chr4	5991122		G	A	FLJ46481
chr4	5992478		A	C	FLJ46481(dist_1104),JAKMIP1(dist_35448)
chr4	6303021		A	G	WFS1
chr4	6594805	rs142055589	A	T	MAN2B2
chr4	6611542	rs201499468	A	G	MAN2B2
chr4	6616038	rs62292692	G	A,T	MAN2B2
chr4	7761424		T	A	AFAP1
chr4	7857293	rs62289316	G	C	AFAP1
chr4	8012496		C	T	ABLIM2
chr4	8358674		G	A	AX746755
chr4	9486166	rs190075635	T	G	DEFB131(dist_33926),MIR54812(dist_71623)
chr4	9495280	rs78813914	C	A	DEFB131(dist_43040),MIR54812(dist_62509)
chr4	9698274	rs200034172	T	G	AB059369(dist_29622),DQ584669(dist_9359)
chr4	9705638	rs184773942	A	G	AB059369(dist_36986),DQ584669(dist_1995)
chr4	9711237	rs138522638	T	A	DQ584669(dist_3576),DRD5(dist_72021)

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chr4	27004435		A	G	STIM2
chr4	36215002	rs189612526	A	T	ARAP2
chr4	36231081	rs147255071	C	T	ARAP2
chr4	37836235	rs113769990	T	C	PGM2
chr4	39876054		T	G	PDS5A
chr4	40133744	rs184546635	C	T	N4BP2
chr4	40426233	rs192269890	C	T	RBM47
chr4	40438690	rs185771029	T	G	RBM47
chr4	47915885		G	T	NFXL1
chr4	52928511	rs375028648	C	A	SPATA18
chr4	53497418	rs183357499	C	T	USP46
chr4	54966310		C	A	GSX2
chr4	55955969	rs56302315	C	T	KDR
chr4	56342091	rs368792480	G	A,C	CLOCK
chr4	62758596	rs76798812	C	T	LPHN3
chr4	65275108	rs200659779	G	C	TECRL
chr4	68919083		C	T	TMPRSS11F
chr4	70361341		G	T	UGT2B4
chr4	71697529		T	C	GRSF1
chr4	72934730	rs6849186	G	A	NPFFR2
chr4	72994416	rs148970744	C	G	NPFFR2
chr4	73148667		G	C	ADAMTS3
chr4	73185147	rs35860396	C	T	ADAMTS3
chr4	73205253	rs7690711	G	T	ADAMTS3
chr4	73927571	rs115487882	T	C	COX18
chr4	74283984	rs56042353	T	C	ALB
chr4	74286034	rs61332165	C	T	ALB
chr4	74321570	rs6826233	A	T	AFP
chr4	74347477	rs187320979	T	C	AFM
chr4	74846877	rs352006	G	A	PF4
chr4	74861591	rs202077518	G	A	CXCL5
chr4	74861798	rs57315103	C	T	CXCL5
chr4	76583866		G	A	G3BP2
chr4	76956917	rs146891844	T	C	ART3,CXCL11
chr4	78083150		A	G	CCNG2
chr4	78639992		T	C	CNOT6L
chr4	78722409		AG		CNOT6L
chr4	79207635	rs373093881	C	T	FRAS1
chr4	84223334	rs141973931	G	A	HPSE
chr4	84519271		G	A	AGPAT9
chr4	89318021	rs192005184	T	A	HERC6
chr4	90816361	rs139171187	A	C	MMRN1
chr4	90857588	rs41279303	G	A	MMRN1
chr4	95529236	rs180835231	C	G	PDLIM5
chr4	100125618		G	T	LOC100507053
chr4	100542119		C	T	MTTP
chr4	100543977	rs144600401	C	T	MTTP
chr4	104066273	rs61751594	T	G	CENPE
chr4	106156520	rs202152392	C	T	TET2
chr4	106569796		G	C	ARHGEF38
chr4	108831998		TTC		SGMS2
chr4	109862019		C	A	COL25A1
chr4	110980592		G	C	ELOVL6
chr4	111542095	rs146850335	G	A	PITX2
chr4	114900006	rs201161005	G	T	ARSJ
chr4	119399845		C	A	PRSS12(dist_125923),Mir_350(dist_12204)
chr4	119679003		A	G	SEC24D
chr4	119686052	rs140080517	G	A	SEC24D
chr4	122605740	rs78275252	G	A	ANXA5
chr4	122728536		G	C	EXOSC9
chr4	122738567		T	C	CCNA2
chr4	124322586		T	C	SPRY1
chr4	126402840	rs143513466	G	A	FAT4
chr4	128938556		T	C	C4orf29
chr4	129191255		C	T	PGRMC2
chr4	129776793		T	C	PHF17
chr4	134070403		A	T	BC040219,PCDH10
chr4	134073575	rs138358445	C	T	PCDH10
chr4	135122364		G	A	PABPC4L



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chr4	139981622	rs147539143	C	T	ELF2
chr4	141565037		G	A	TBC1D9
chr4	144617852		G	C	FREM3
chr4	144928192		G	C	GYPB,GYPB
chr4	146026992		C	G	ABCE1
chr4	148787968		G	A	ARHGAP10
chr4	151207180	rs202244838	G	A	LRBA
chr4	153273421		T	A	FBXW7
chr4	153547333		C	T	TMEM154
chr4	153565619			T	TMEM154
chr4	153565620		A	T	TMEM154
chr4	153565624			CTTCTGTAAC	TMEM154
chr4	153898138		C	A,G	FHDC1
chr4	155191162	rs146032523	G	A	DCHS2
chr4	158285725		G	A	GRIA2
chr4	159589343		A	C	C4orf46
chr4	162841700	rs368695376	A	G	FSTL5
chr4	164393684		G	A	TKTL2
chr4	166220988		G		KLHL2
chr4	169013923		A	C	ANXA10
chr4	169341450	rs189472897	A	C	DDX60L
chr4	175158398		G	A	FBXO8
chr4	175412398	rs140692963	T	G	HPGD
chr4	175412669	rs144520232	T	C	HPGD
chr4	177056356	rs147125405	T	C	WDR17
chr4	183836317		T	C	DCTD
chr4	185263087	rs199558624	C	T	LOC728175
chr4	185580680		G	T	CCDC111
chr4	185616601		C	T	MLF1IP
chr4	186232233	rs116003605	C	T	SNX25
chr4	186696162		C	T	SORBS2
chr4	187476365	rs144178215	A	G	MTNR1A
chr4	187509539		G	C	FAT1
chr4	187534616		G	C	FAT1
chr5	139460		G	C	PLEKHG4B
chr5	433882		G	A	AHRR
chr5	473406		C	G	SLC9A3
chr5	624587	rs62001006	C	T	CEP72
chr5	766930		C	T	TPPP(dist_73420),ZDHHC11(dist_28790)
chr5	865599	rs372573589	G	A	BRD9
chr5	1033498	rs144426465	G	A	NKD2
chr5	1052129		A	G	SLC12A7
chr5	1463694		G	C	LPCAT1
chr5	6756739		A	C	PAPD7
chr5	11397153	rs146014959	C	T	CTNND2
chr5	14498717	rs146227809	C	T	TRIO
chr5	15936900	rs375930592	G	A	FBXL7
chr5	16571809		T	C	FAM134B
chr5	16676155		C	A	MYO10
chr5	16902742		TTTC		MYO10
chr5	21802361	rs147983357	T	C	CDH12
chr5	21884340	rs112117136	A	G	CDH12
chr5	34823315		G	C	RAI14
chr5	36876673		G	A	NIPBL
chr5	37107686	rs200332492	C	A	C5orf42
chr5	38958865	rs149642899	A	G	RICTOR
chr5	40681710	rs34902673	T	C	PTGER4
chr5	40833016	rs185795402	C	A	RPL37
chr5	40834550	rs200807833	T	G	RPL37
chr5	41195939	rs114609505	G	A	C6
chr5	43122080	rs151063287	T	C	ZNF131
chr5	52235932		TG		ITGA1
chr5	52285405	rs200410974	G	A	ITGA2
chr5	53839461		A	G	SNX18
chr5	56778225	rs147643155	T	C	ACTBL2
chr5	64920604	rs186611196	A	G	C5orf44
chr5	71015536		T	C	CARTPT
chr5	74078783		T	G	FAM169A
chr5	75866423	rs192694811	C	T	IQGAP2

chr5	77298901	rs201198522	C	T	AP3B1
chr5	78301172	rs41272262	C	T	DMGDH
chr5	78326040		A	G	DMGDH
chr5	79353841		A	G	THBS4
chr5	79732577		G	A	ZFYVE16
chr5	85586899		G	A	NBPF22P
chr5	87521659		A	C	TMEM161B
chr5	89769717		G	C	MBLAC2
chr5	89854525		C	T	GPR98
chr5	89949423	rs376636949	C	T	GPR98
chr5	90664629	rs185507323	T	C	ARRDC3
chr5	94887243			A	TTC37
chr5	96117519		C	T	ERAP1
chr5	109946230		T	C	TMEM232
chr5	115298726	rs139596752	A	T	AQPEP
chr5	115909941		C	T	SEMA6A
chr5	118969974		C	A	FAM170A
chr5	120022309	rs141384895	A	T	PRR16
chr5	122758609	rs141808885	G	T	CEP120
chr5	124080382	rs61749626	T	A	ZNF608
chr5	126738168		TTC		MEGF10
chr5	131679320		G	A	LOC553103
chr5	132094826		A	C	SEP8
chr5	132568817	rs151163592	C	G	FSTL4
chr5	132569472	rs142275476	C	G	FSTL4
chr5	133309538		G	C	VDAC1
chr5	136312184		C	T	SPOCK1
chr5	137684076	rs182881841	A	G	FAM53C
chr5	138940773		G	A	UBE2D2
chr5	139682831		C	T	PFDN1
chr5	139742155	rs10054389	G	A,T	SLC4A9
chr5	139940887		G	A	
chr5	139942149		C	T	APBB3
chr5	140022257	rs367652614	G	A	TMCO6
chr5	140085215	rs143727913	G	A	ZMAT2
chr5	140181076	rs138150756	G	A	PCDHA3
chr5	140537167	rs200329533	C	T	PCDHB17
chr5	140567191	rs187473539	A	G	PCDHB9
chr5	140620802		G	A	PCDHB19P
chr5	140742664	rs182933962	G	A	PCDHGA1,PCDHGA2,PCDHGA3, PCDHGA4,PCDHGB1,PCDHGB2
chr5	140812248	rs147073234	G	A	PCDHGA12
chr5	140858014		T	A	PCDHGC3
chr5	143191869		G	A	HMHB1
chr5	145252686	rs142513740	A	G	GRXCR2
chr5	146086363	rs187931279	G	A	PPP2R2B
chr5	146970632		G	A	LOC153469
chr5	148563007	rs368655320	T	C	ABLIM3
chr5	148622226		G	C	ABLIM3
chr5	149927905	rs146085225	C	T	NDST1
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chr5	150052110		AGCCCGGGGACAGACCCGGGAGGGGCGGGA		MYOZ3
chr5	150409816	rs180904763	G	A	TNIP1
chr5	154308256		AC		GEMIN5
chr5	154315751		T	C	GEMIN5
chr5	156482446	rs141773914	T	G	HAVCR1
chr5	156934121		G	A	ADAM19
chr5	159344040	rs149660268	C	T	ADRA1B
chr5	159738849	rs200773887	G	C	CCNJL
chr5	162900937		A	G	HMMR
chr5	167841589	rs200199992	C	T	WWC1
chr5	168112707	rs72839508	G	A	SLIT3
chr5	171626549	rs200955663	C	T	EFCAB9
chr5	172750196	rs201359603	C	T	STC2
chr5	172752907	rs146441603	C	A	STC2
chr5	174953950		A	C	SFXN1
chr5	175717901	rs138565600	T	A	C5orf25
chr5	175923590	rs144969484	T	G	FAF2
chr5	176707724	rs61749654	C	G,T	NSD1
chr5	176816724		CT		SLC34A1

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chr5	177473907	rs201864338	A	C	FAM153C
chr5	177632577		A	G	HNRNPAB
chr5	178224404	rs190811678	C	A	AACSP1
chr5	179192580	rs113636707	G	A	MAML1
chr5	179563582		G	A	RASGEF1C
chr5	180432860		G	A	BTNL3
chr5	180475220		T	C	BTNL9
chr6	3137700		A	G	BPHL
chr6	4056809		C	T	PRPF4B
chr6	7247172	rs373051958	G	A	RREB1
chr6	7417816		C	T	RIOK1
chr6	7834036		G	A	BMP6
chr6	8102419	rs74880365	A	G	EEF1E1
chr6	10397062		C	T	TFAP2A
chr6	10412387		A	G	TFAP2A
chr6	10412776		G	A	LOC100130275
chr6	12749523	rs151171008	G	A	PHACTR1
chr6	13479394	rs78231241	T	C	GFOD1
chr6	15508329	rs111768285	G	A	JARID2
chr6	17611616		C	T	FAM8A1
chr6	17688442		GTTT		NUP153
chr6	17779374	rs372462124	C	T	KIF13A
chr6	18465521		A	G	RNF144B
chr6	24476437	rs61754636	C	T	GPLD1
chr6	24537130	rs182106500	A	G	ALDH5A1
chr6	24719009	rs147402940	T	C	C6orf62
chr6	25726291	rs200653429	T	G	HIST1H2AA
chr6	26017926	rs201252490	G	A	HIST1H1A
chr6	26032286		C	T	HIST1H3B
chr6	26043894	rs201553217	C	T	HIST1H2BB
chr6	26368108		T	C	BTN3A2
chr6	26406073	rs184245591	C	G	BTN3A1
chr6	26449199		T	C	BTN3A3
chr6	27879247		A	C	OR2B2
chr6	28297090	rs142223162	G	A	ZNF323
chr6	29394949	rs139568093	T	G	OR11A1
chr6	29408359	rs145441997	A	G	OR10C1
chr6	29638101	rs112913018	T	C	MOG
chr6	29974801		G	C	HLA-G,HLA-J
chr6	29977448	rs139388011	G	C	HLA-G,HLA-J
chr6	30135009	rs140385648	T	A	TRIM15
chr6	30460427		T		HLA-E
chr6	30531430		T	G	PRR3
chr6	30617401		C	T	C6orf136
chr6	30657485	rs112797927, rs114771334	G	A	NRM
chr6	30917708		G	A	DPCR1
chr6	31084108		G	A	CDSN
chr6	31516034		C	T	NFKBIL1
chr6	31544799		C	T	TNF
chr6	31598477		G	A	PRRC2A
chr6	31606749	rs145301042	T	G	BAG6
chr6	31627888		T	C	C6orf47
chr6	31670740		A	T	ABHD16A
chr6	31711749		A	T	MSH5
chr6	31750919	rs56192709, rs148267831	G	T	VAR5
chr6	31860622	rs145404362	T	C	EHMT2
chr6	31976616	rs71563401	T	C	CYP21A1P
chr6	31976619	rs71563402	C	T	CYP21A1P
chr6	32017776	rs202162469	C	T	TNXB
chr6	32133930	rs115416104	G	A	PPT2,PPT2-EGFL8
chr6	32137269	rs367933331	T	C	AGPAT1
chr6	32148497		G	T	RNF5
chr6	32361094	rs141794876	T	C	HCG23
chr6	32942687		A	G	BRD2
chr6	32975286	rs34987694, rs150629001	A	G	HLA-DOA
chr6	33167516	rs149010851	G	T	RXRB

chr6	33259601	rs182519922	A	C	RGL2
chr6	33265090		C	A	RGL2
chr6	33423341	rs150940874	C	T	ZBTB9
chr6	33863235	rs138769680	A	T	DQ589635
chr6	34215307		C	T	C6orf1
chr6	34985242	rs201532907	A	G	ANKS1A
chr6	36279473		A	G	PNPLA1(dist_3101),C6orf222(dist_4062)
chr6	36689709	rs182774229	C	G	AK125083(dist_6302),CPNE5(dist_18846)
chr6	36690275	rs193121565	C	T	AK125083(dist_6868),CPNE5(dist_18280)
chr6	36823095		C	T	PPIL1
chr6	36882169	rs371671264	C	T	C6orf89
chr6	36949471	rs149538273	G	C	MTCH1
chr6	37186674		T	C	TMEM217
chr6	37414338		G	A	FTSJD2
chr6	37615111	rs191471367	G	A	MDGA1
chr6	38100252		C	T	ZFAND3
chr6	38879373	rs201286373	GGAAT		LOC100131047
chr6	39054396	rs200276742	C	T	GLP1R
chr6	39282705	rs9471064	C	A	KCNK16
chr6	39282877	rs9471065	C	T	KCNK16
chr6	39284574	rs9462527	A	G	KCNK16
chr6	39313397	rs112535273	C	T	KIF6
chr6	42075190		T	C	C6orf132
chr6	43008764	rs36004037	C	T	CUL7
chr6	43303687	rs116070990	C	T	ZNF318
chr6	43472271		G	A	TJAP1
chr6	43485344		G	A	POLR1C
chr6	44225102		C	T	SLC35B2
chr6	45922924		T	A	CLIC5
chr6	46563647		C	T	CYP39A1
chr6	46820698		A	T	GPR116
chr6	47869147	rs28474370	G	T	C6orf138
chr6	49399340	rs113025987	C	T	MUT
chr6	49460742	rs189396607	C	G	CENPQ
chr6	49667569	rs114974767	C	A	CRISP2
chr6	49712063	rs74393864	A	G	CRISP3
chr6	49814371	rs35590668	A	G	CRISP1
chr6	50681790		C	T	TFAP2D
chr6	51640639	rs113562492	T	C	PKHD1
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chr6	53321273		T	G	7SK(dist_40868),GCLC(dist_40867)
chr6	53321278			C	7SK(dist_40873),GCLC(dist_40862)
chr6	53321278			C	
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chr6	74001984	rs144286780	G	A	C6orf147
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chr6	75875241	rs139332405	C	T	COL12A1
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chr6	76385170	rs188392670	A	G	SENP6
chr6	78400729	rs111553654	A	G	HTR1B(dist_227609),SNORD112(dist_246189)
chr6	78400734	rs9448149	C	T	HTR1B(dist_227614),SNORD112(dist_246184)
chr6	82462497	rs139573485	G	A	FAM46A
chr6	87966773	rs41273273	T	A	ZNF292
chr6	90371847	rs142038492	C	G	MDN1
chr6	96662160		A	G	FUT9
chr6	97729056		G	A	MIR548H3
chr6	99848172		G	A	BC033061
chr6	99990851	rs187227701	T	G	CCNC
chr6	105228300		T	C	HACE1
chr6	107420358	rs78776849	A	G	BEND3
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chr6	107834914		C	A	SOBP
chr6	108544237	rs371370665	T	C	SNX3
chr6	109621276		G	T	CCDC162P
chr6	109635033	rs192981785	A	G	CCDC162P(dist_5610),C6orf185(dist_24645)
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chr6	109796503	rs188884315	T	C	ZBTB24
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chr6	128812841	rs185622438	G	C	PTPRK
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chr6	129837372		G	A	LAMA2
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chr6	133073884		T	C	VNN2
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chr6	139226273		A		
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chr6	142539840		A	G	VTA1
chr6	142719150		A	G	GPR126
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chr6	144981222	rs150345409	T	C	UTRN
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chr6	146255836		T	C	SHPRH
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chr6	151673303		A	C	AKAP12
chr6	152642442	rs200796273	A	G	SYNE1
chr6	152690432	rs149091219	A	G	SYNE1
chr6	153323468		AA		MTRF1L
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chr6	160227051	rs55637742	C	T	PNLDC1
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chr6	160496705	rs141046776	C	T	IGF2R
chr6	160553407	rs36103319	G	T	SLC22A1
chr6	160898115	rs182581271	C	T	LPAL2
chr6	162406519		T	C	PARK2
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chr6	168376925	rs2516606	G	T	HGC6.3
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chr7	20371780	rs149659842	C	T	ITGB8
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chr7	23508312	rs111523797	C	T	IGF2BP3
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chr7	23731172	rs148660873	C	T	C7orf46
chr7	24727468		T	G	MPP6
chr7	26240261		A	G	HNRNPA2B1
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chr7	32907602		G	A	AVL9
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chr7	35222928	rs186164836	C	T	DPY19L2P1
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chr7	64388448	rs141427676	A	T	ZNF273
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chr7	64838916	rs367558249	G	A	ZNF92
chr7	64863093		A	G	ZNF92
chr7	65217203	rs185080334	G	C	CCT6P1
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chr7	74312917		T	C	PMS2P5
chr7	74910370	rs200338615	G	T	PMS2L14
chr7	75071929	rs2005837	T	G	POM121C
chr7	75104081		AAG		POM121C
chr7	75132411		A	G	SPDYE5
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chr7	107643149	rs147297312	G	A	LAMB1
chr7	107667270	rs1732158	C	A	LAMB4
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chr7	107684393	rs28461730	T	G	LAMB4
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chr9	111625559	rs150177974	G	A	ACTL7A
chr9	112778470		C	G	AKAP2,PALM2-AKAP2
chr9	112934277		C	A	AKAP2,PALM2-AKAP2
chr9	114156276		C	A	KIAA0368
chr9	119459904		C	T	ASTN2,TRIM32
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chr9	124222172		G	A	GGTA1P
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chr9	127638910		G	A	ARPC5L
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chr9	128104655		A	G	GAPVD1
chr9	128199718		AT		MAPKAP1
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chr9	129293503	rs139264636	GGG		Mir_1302
chr9	129455844	rs371560611	C	T	LMX1B
chr9	130258261	rs150882646	C	A	LRAM1
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chr9	130504082		G	A	SH2D3C
chr9	131709916		C	T	DOLK
chr9	131803131		C	A	FAM73B
chr9	131830316		A	T	FAM73B
chr9	131851606		T	C	DOLPP1
chr9	131891549		A	T	PPP2R4
chr9	133228484		C	T	NCS1(dist_228901),DKFZp434P0216(dist_32644)
chr9	133258210		C	G,T	NCS1(dist_258627),DKFZp434P0216(dist_2918)
chr9	133261961	rs138367364	A	T	DKFZp434P0216
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chr9	134072817	rs72768590	C	G	NUP214
chr9	134584987			A	RAPGEF1
chr9	134944629		C	T	MED27
chr9	135771506		T	C	TSC1
chr9	135802597	rs371555137	T	C	TSC1
chr9	135862356		G	A	GF1B
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chr9	136080623		G	A	OBP2B
chr9	136101924		G	A	OBP2B(dist_17296),ABO(dist_28639)
chr9	136220769	rs145615218	T	G	SURF1
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chr9	136513028	rs75215331	C	T	DBH
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chr9	138392517	rs59940288	T	G	MRPS2
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chr9	138657804	rs10118323	T	C	KCNT1
chr9	139091595	rs146827257	C	T	LHX3
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chr9	139309031	rs148940590	C	T	PMPCA
chr9	139652590		A	G	LCN8
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chr10	8093217		T	C	FLJ45983
chr10	10986247		G	C	LOC254312
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chr10	12077540	rs148365095	T	C	UPF2

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chr10	30658575		G	T	LOC729668
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chr10	51464443		T	G	AGAP7
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chr10	56367472		TAA		PCDH15
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chr10	61789270	rs80034950	C	T	ANK3
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chr10	69646948	rs35865685	G	A	SIRT1
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chr10	69676434	rs7897909	G	A	SIRT1
chr10	69991824	rs187364801	G	C	ATOH7
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chr10	71640467	rs113444400	C	T	COL13A1
chr10	71883729		T	A	AIFM2
chr10	71969131		T	A	PPA1
chr10	72300977	rs45488693	C	T	KIAA1274
chr10	73538150	rs149057462	A		CDH23
chr10	74098216		G	A	DNAJB12
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chr10	75834471	rs200699818	A	G	VCL
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chr10	81374897	rs183241359	T	C	SFTPA1
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chr10	96099644	rs368862439	T	C	NOC3L
chr10	99141549		G	A	RRP12
chr10	99161299	rs186939835	A	C	RRP12
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chr10	124097645	rs376344876	A	G	BTBD16
chr10	124348703	rs202084523	C	T	DMBT1
chr10	124608738	rs150242478	G	A	FAM24B
chr10	124909476		C	A	HMX2
chr10	126308729	rs114056130	G	A	FAM53B
chr10	126370662	rs74533612	G	T	FAM53B
chr10	126480382	rs112471520	G	C	METTL10
chr10	126524861	rs114104870	C	T	FAM175B
chr10	127408425		G	A	C10orf137
chr10	127429077		T	A	C10orf137
chr10	128823230	rs192456650	C	T	DOCK1
chr10	129535749		C	T	FOXI2
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chr11	294275	rs114112322	G	A	ATHL1
chr11	299321		A	C	IFITM5
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chr11	428749	rs114920213	G	A	ANO9
chr11	433357	rs79329594	G	C,T	ANO9
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chr11	534037		A	T	HRAS
chr11	601592	rs374545495	C	A	PHRF1
chr11	691457		C	T	DEAF1
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chr11	1078320	rs72652894	G	A	MUC2
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chr11	1264766	rs200775252	C	T	MUC5B
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chr11	1268537	rs200237369	C	T	MUC5B
chr11	1463671	rs370524119	G	A	BRSK2
chr11	1605919	rs148721177	G	T	KRTAP5-1
chr11	1628896	rs149535111	G	A	KRTAP5-3
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chr11	3062131	rs61737270	G	A	CARS
chr11	3129103	rs147555918	G	A	OSBPL5
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chr11	3430253		A	G	LOC650368
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chr11	4623157		C	T	TRIM68
chr11	4623509		T	C	TRIM68
chr11	4623688		A	C	TRIM68
chr11	5141744		T	C	DQ656008
chr11	5423977	rs77173896	G	A	HBE1,OR51B5
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chr11	5862253		A	T	OR52E6
chr11	6422101		G	A	APBB1
chr11	6478572		T	C	TRIM3
chr11	6519339	rs141467778	G	A	DNHD1
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chr11	9802945		G	A	BC073899,LOC283104
chr11	9812393	rs181706953	C	T	BC073899,LOC283104
chr11	9860893		C	T	SBF2
chr11	9917615	rs148041587	C	A	U80769
chr11	9985135			TAAT	SBF2
chr11	10315424		G	A	SBF2
chr11	10327183		G	A	ADM
chr11	10536115		A	G	RNF141
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chr11	18422408	rs200332813	A	G	LDHA
chr11	19251903	rs182770607	C	T	E2F8
chr11	20136261		G	A	NAV2
chr11	20386064		G	A	HTATIP2
chr11	20674037	rs202191558	C	T	SLC6A5
chr11	26743404		G	A	SLC5A12
chr11	32126524		C	T	RCN1
chr11	33094335		G	A	TCP11L1
chr11	33102712	rs147835239	T	C	LINC00294(dist_1712),CSTF3(dist_3418)
chr11	33375298		C	G	HIPK3
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chr11	46419153	rs137890582	C	A	AMBRA1
chr11	46783687	rs145487772	G	A	CKAP5
chr11	47350617	rs181428672	C	T	MADD
chr11	47644269	rs146359770	C	T	MTCH2
chr11	48968187		A	T	OR4A47(dist_456913),LOC120824(dist_28863)
chr11	49372757	rs199838557	G	A	FOLH1(dist_142535),LOC440040(dist_207323)
chr11	49729456		A	G	LOC440040
chr11	55038638		C	T	TRIM48
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chr11	56468916		T	A	OR8U8
chr11	56955941		T	G	LRRC55
chr11	57184021	rs143862070	A	C	PRG2,SLC43A3
chr11	57235179	rs202097987	C	T	RTN4RL2
chr11	58919922			GCAGATACTT	FAM111A
chr11	59846059	rs114855406	A	T	MS4A3(dist_7471),MS4A2(dist_10078)
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chr11	63995181	rs375245205	G	A	NUDT22
chr11	64671159	rs370161077	C	A	ATG2A
chr11	65386315		C	T	PCNXL3
chr11	65638722		T	C	EFEMP2
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chr11	65655721		G	A	FIBP
chr11	66051053		G	A	CNIH2
chr11	66392000		G	A	RBM14
chr11	66618855		T	C	PC
chr11	67187017	rs374499029	G	C	CARNS1,PPP1CA
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chr11	67219723	rs144630243	A	G	GPR152
chr11	67799526		AT		NDUF58
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chr11	70118568	rs145666130	A	G	PPFIA1
chr11	71800166	rs146942815	G	A	LRTOMT
chr11	74082033			A	PGM2L1(uc001ovb.1:exon5:c.387-2->T)
chr11	74554914		T	C	XRRA1
chr11	75111597	rs368682282	G	C	RPS3
chr11	76826540		G	A	CAPN5
chr11	76892529	rs201489714	G	A	MYO7A
chr11	77815467	rs146603801	G	A	ALG8
chr11	77907028	rs184744988	C	A	USP35
chr11	82923522		A	G	ANKRD42
chr11	89806046		T	G	TRIM49L2(dist_30853),UBTF1L1(dist_13072)
chr11	92533558	rs200944979	G	A	FAT3
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chr11	93469465		A	G	TAF1D
chr11	94278205	rs138386205	C	T	FUT4
chr11	94770875	rs181817132	G	T	KDM4DL(dist_10115),SRSF8(dist_29181)

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chr11	111921795		T	C	DLAT
chr11	113113556		A	G	NCAM1
chr11	114272291		G	A	RBM7
chr11	114451010	rs193088648	T	C	FAM55D
chr11	117095153		C	A	PCSK7
chr11	117304174	rs35399839	C	T	DSCAML1
chr11	117392079	rs373292920	C	T	DSCAML1
chr11	117693185	rs145863486	G	A	FXVD2,FXVD6-FXVD2
chr11	117695427	rs375591023	C	T	FXVD2
chr11	117947757	rs117898143	G	A	TMPRSS4
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chr11	118391845		T	C	MLL
chr11	118883002	rs148856513	C	T	CCDC84
chr11	118888915	rs186247013	A	G	RPS25
chr11	118919562	rs147081612	C	T	HYOU1
chr11	118948672		A	G	VPS11
chr11	118963869	rs150428209	G	A	HMBS
chr11	119052053		A	C	NLRX1
chr11	119227514		G	A	USP2
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chr11	121458501	rs149723506	G	A	SORL1
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chr11	122726421		T	C	CRTAM
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chr11	124121203		C	G	OR8G1
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chr12	6786555		C	T	ZNF384
chr12	6797383	rs189135169	C	T	ZNF384
chr12	6924927	rs191651672	C	T	CD4
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chr12	7053066		A	G	ATN1
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chr12	7087673	rs201219868	A	G	LPCAT3
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chr12	7290573	rs143159646	G	A	CLSTN3
chr12	7463324	rs200593237	G	T	ACSM4
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chr12	9886014		C	T	CLECL1

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chr12	12419973		G	T	LRP6
chr12	13769570	rs199671864	A	T	GRIN2B
chr12	21445155	rs377061804	G	A	SLCO1A2
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chr12	31478155	rs117086528	C	T	FLJ13224
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chr12	33592677		C	T	SYT10
chr12	39716500	rs149075970	G	C	KIF21A
chr12	40697943	rs41286480	C	T	LRRK2
chr12	40820245	rs75634075	T	C	MUC19(dist_7210),MUC19(dist_118607)
chr12	40900203	rs191568268	G	C	MUC19(dist_87168),MUC19(dist_38649)
chr12	40913654		C	T	MUC19(dist_100619),MUC19(dist_25198)
chr12	40935566	rs76407249	C	G	MUC19(dist_122531),MUC19(dist_3286)
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chr12	43945598	rs376310421	C	T	ADAMTS20
chr12	44162311		A	G	IRAK4
chr12	44770496	rs117394252	T	C	TMEM117
chr12	45566966		G		PLEKHA8P1
chr12	45610237			A	ANO6
chr12	46246052	rs143660736	C	T	ARID2
chr12	46757119		C	A	SLC38A2
chr12	48185668		G	A	HDAC7
chr12	48238125		G	A	VDR
chr12	48397796	rs142120076	A	G	COL2A1
chr12	49259594	rs377604830	G	A	RND1
chr12	49330151	rs41291973	G	C	ARF3
chr12	49426467		TGT		MLL2
chr12	49666538		A	G	TUBA1C
chr12	49934793	rs147651812	G	A	KCNH3
chr12	50369394	rs148510811	G	A	AQP6
chr12	50821683	rs371940413	C	T	LARP4
chr12	52450405	rs140988245	C	T	NR4A1
chr12	52450644		G	T	NR4A1
chr12	52649099	rs117844128	C	A	AK300121,KRT86
chr12	52695760	rs201694571	C	G	KRT86
chr12	52758826	rs149560920	G	A	KRT85
chr12	52789643		C	T	KRT82
chr12	53068303		C	G	KRT1
chr12	53162233		C	T	KRT76
chr12	53413619		T		EIF4B
chr12	53931274		TCT		ATF7
chr12	54379816		G	T	HOXC10
chr12	54757171	rs140650044	G	C	GPR84
chr12	54929999	rs375303752	C	T	NCKAP1L
chr12	55248412	rs201867480	C	T	MUCL1
chr12	55361695		T	C	KIAA0748
chr12	55421745	rs191166307	G	A	NEUROD4
chr12	55423523	rs188286259	G	A	NEUROD4
chr12	55693540		C	A,T	OR6C6(dist_4524),OR6C1(dist_20844)
chr12	55714478	rs200476888	C	T	OR6C1
chr12	55806407		G	A	OR6C65(dist_11156),OR6C76(dist_13631)
chr12	55806408		G	T	OR6C65(dist_11157),OR6C76(dist_13630)
chr12	55846809	rs200822200	G	A	OR6C2
chr12	56031464	rs143887647	G	A	OR10P1
chr12	56214080	rs145457304	T	A	ORMDL2
chr12	56215175		G	A	DNAJC14
chr12	56294425		C	T	WIBG
chr12	56335404		C	T	DGKA
chr12	56388307	rs187146028	C	T	RAB5B
chr12	56435644		G	A	RPS26
chr12	56436465		CT		RPS26

chr12	56490174		C	T	ERBB3
chr12	56495339	rs55699040	C	A	ERBB3
chr12	56522618		CT		ESYT1
chr12	56553920		C	T	MYL6
chr12	56577635		C	G	SMARCC2
chr12	56755246	rs373590737	T	G	APOF
chr12	56864843	rs190064727	C	G	GLS2
chr12	57030158		C	T	BAZ2A
chr12	57110000	rs183828032	G	A	NACA
chr12	57441314		C	A	MYO1A
chr12	57490429	rs146670318	G	C	STAT6
chr12	57995171	rs377089363	A	G	PIP4K2C
chr12	58120726	rs199648267	A	G	AGAP2,LOC100130776
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chr12	64521467		T	C	SRGAP1
chr12	65130583		T	C	GNS
chr12	66698597		C	T	HELB
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chr12	70949213	rs189340950	C	T	PTPRB
chr12	70963665	rs139698072	G	A	PTPRB
chr12	71960487	rs199507313	A	G	LGR5
chr12	72004091	rs17227269	T	C	ZFC3H1
chr12	72089963	rs191922663	T	A	TMEM19
chr12	72317081		TT		TBC1D15
chr12	72317489		T	C	TBC1D15
chr12	72318138		T	A	TBC1D15
chr12	80171313		C	T	PPP1R12A
chr12	80611599		G	C	OTOGL
chr12	88530661		C	T	CEP290
chr12	89742834		C	G	DUSP6
chr12	91347324		A	T	C12orf12
chr12	94975829	rs149007412	C	T	TMCC3
chr12	99076643		AT		APAF1
chr12	100552894		G	A	GOLGA2P5
chr12	100733911		G	A	SCYL2
chr12	104142874	rs201554631	C	A	STAB2
chr12	104160024		C	T	STAB2
chr12	104391269	rs142096211	G	A	GLT8D2
chr12	104697533		G	T	EID3
chr12	109246399	rs182266049	T	A	SSH1
chr12	112184086	rs150349412	G	A	ACAD10
chr12	112578821	rs114172135	G	A	TRAFD1
chr12	113424858		C	T	OAS2
chr12	113440757		C	T	OAS2
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chr12	120156811	rs73215362	G	C,T	CIT
chr12	120780266		G	T	MSI1
chr12	122186346	rs201256957	C	T	TMEM120B
chr12	122817561	rs143777418	A	C	CLIP1
chr12	123950176	rs148264502	C	T	SNRNP35
chr12	124841318	rs111850964	G	A	NCOR2
chr12	124968492		C	T	BC017209
chr12	129180623		G	A	TMEM132C
chr12	131498784	rs140426880	G	A	GPR133
chr12	133198337		T	C	P2RX2
chr12	133212485	rs5744991	C	T	POLE
chr12	133237415	rs5744846	A	C	POLE
chr12	133297495	rs11548882	A	C	PGAM5
chr12	133347659	rs181747546	T	C	GOLGA3
chr12	133350692	rs75861584	G	A	GOLGA3
chr12	133352827		T	C	GOLGA3
chr12	133352828		G	A	GOLGA3
chr12	133352933		C	T	GOLGA3
chr12	133363117	rs148429683	C	T	GOLGA3
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chr13	20398534		C	A	ZMYM5
chr13	20625641		G	C	ZMYM2



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chr13	25425647	rs34209003	A	G	RNF17
chr13	25478209		C	T	CENPJ
chr13	25671714	rs61739066	A	G	PABPC3
chr13	28155529	rs78036110	A	T	LNK2
chr13	29274938		G	A	SLC46A3
chr13	30084663		C	T	SLC7A1
chr13	31496144	rs190065125	A	G	LOC100507064
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chr13	41646777		C	T	WBP4
chr13	45694783		G	T	GTF2F2
chr13	46946172		AG		KIAA0226L
chr13	47224167		A	G	LRCH1
chr13	48708381		T	C	MED4(dist_39130),ITM2B(dist_98893)
chr13	49281386	rs139045217	C	A,T	CYSLTR2
chr13	49841707	rs139520607	A	G	CDADC1
chr13	49841879	rs149790234	T	C	CDADC1
chr13	53157044	rs187049885	A	G	TPTE2P3
chr13	61987500	rs142199876	T	C	PCDH20
chr13	73302184	rs191478196	T	A	BORA
chr13	73346652		T	C	DIS3
chr13	73636962	rs182192000	C	T	KLF5
chr13	74269689		C	T	KLF12
chr13	74545115		TGAC		KLF12
chr13	75936050		AAAT		TBC1D4
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chr13	78133858		G	A	SCEL
chr13	78146303	rs141416270	C	T	SCEL
chr13	79213134	rs144588084	T	G	RNF219
chr13	92051171		C	G	GPC5
chr13	95055605	rs187678452	A	G	GPC6
chr13	98086988		G	A	RAP2A
chr13	98116786		G	A	RAP2A
chr13	98633801		G	A	IPO5
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chr13	110408258		C	A	IRS2
chr13	111279995		G	A	CARKD
chr13	111893629		C	T	ARHGEF7
chr13	113813005	rs148527063	C	G	PROZ
chr13	114265307	rs139701699	G	A	TFDP1
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chr14	20228761		G	A	OR4Q3(dist_12233),OR4M1(dist_19721)
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chr14	20296550		A	T	OR4N2
chr14	20842603		G	A	TEP1
chr14	20897414	rs200807217	G	A	KLHL33
chr14	22933328	rs376871927	C	T	TCRDV2,TRDC
chr14	23395920	rs370955151	G	A	PRMT5
chr14	23416713		G	A	HAUS4
chr14	24036169		G	A	AX747770
chr14	24572424		C	T	PCK2
chr14	24607804	rs184683192	G	A	PSME1
chr14	24909979	rs112432580	C	T	KHNYN
chr14	24910742			A	SDR39U1
chr14	25042909	rs61737118	G	A	CTSG
chr14	25078609		C	T	GZMH
chr14	25100364	rs142875748	A	G	GZMB
chr14	29237718	rs34654108	G	A	FOXG1
chr14	31081610	rs201971360	G	A	G2E3
chr14	31353798	rs370307569	A	G	COCH
chr14	33684466		A	G	NPAS3
chr14	35231242	rs146357991	G	T	BAZ1A
chr14	35234340		T	C	BAZ1A

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chr14	55604146	rs199699629	C	T	LGALS3
chr14	55820088		T	C	FBXO34
chr14	55903340	rs145550690	T	C	TBPL2
chr14	56108061		T	C	KTN1
chr14	57674030		T	A,C	EXOC5
chr14	57863373		C	T	NAA30
chr14	58669637	rs117148720	A	C	ACTR10
chr14	59792755	rs61755642	G	A	DAAM1
chr14	59830610		G	A	DAAM1
chr14	59988189		A	C	C14orf38
chr14	60193957		T	C	RTN1
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chr14	64587777	rs140277551	C	G	SYNE2
chr14	64746787		G	A	ESR2
chr14	64892725	rs200604984	A	G	MTHFD1
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chr14	65211097		G	A	PLEKHG3
chr14	66209166	rs199509807	G	A	FUT8
chr14	68280822	rs145402701	T	C	ZFYVE26
chr14	68758867		T	C	RAD51B
chr14	71514912			A	PCNX
chr14	71522342	rs144807065	A	G	PCNX
chr14	71995911		G	A	SIPA1L1
chr14	72139429	rs192663868	G	A	SIPA1L1
chr14	73637535		G	C	PSEN1
chr14	73958735		C	G	C14orf169
chr14	73973254	rs143649458	C	T	HEATR4
chr14	74402757	rs151325166	A	G	FAM161B
chr14	74413070		G	A	FAM161B
chr14	74424813		C	G	ENTPD5
chr14	74763023		A	T	ABCD4
chr14	74983553	rs145851939	G	A	LTBP2
chr14	76525445		C	T	IFT43
chr14	76640134	rs183315508	G	T	C14orf118
chr14	77018586	rs370980830	G	A	ESRRB(dist_50406),Mir_384(dist_172080)
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chr14	81646290		G	A	DL490427
chr14	90722781		G	C	PSMC1
chr14	91655448	rs12895348	G	A	C14orf159
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chr14	91739969	rs77154172	A	G	CCDC88C
chr14	91774440		G	T	CCDC88C
chr14	93406372		C	A	ITPK1
chr14	93406373		A	G	ITPK1
chr14	94953836	rs200583511	G	A	SERPINA12
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chr14	96706594			GCG	BDKRB2
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chr14	96772075	rs146028674	G	A	ATG2B
chr14	100992304		G	A	WDR25
chr14	101346808		C	T	MIR431
chr14	101347588	rs199947359	A	G	RTL1
chr14	101349996		C	T	RTL1
chr14	102025029		C	T	DIO3AS,DIO3OS
chr14	102392992		TTAATG		PPP2R5C
chr14	102550733	rs118043158	T	C	HSP90AA1
chr14	103413209		C		CDC42BPB
chr14	103571473	rs193228003	A	C	EXOC3L4
chr14	103803954	rs189972171	C	A	EIF5
chr14	103805970	rs72708831	T	C	EIF5
chr14	105269029	rs143538688	A	G	ZBTB42
chr14	105643144	rs142630749	C	T	NUDT14
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chr14	106204274		C	T	IGH@,abParts
chr14	106208002		A	G	IGHG1
chr14	106208003		T	C	IGHG1
chr14	106237741	rs12586349	A	G	FLJ00385,abParts
chr14	106370373	rs376428626	A	C	abParts
chr14	106436097		G	A	ADAM6
chr14	106780450		C	G	abParts
chr14	106940028	rs201358096	T	C	LINC00221,abParts
chr14	107131048	rs201182063	C	T	abParts
chr14	107198945	rs201899598	T	C	abParts
chr14	107210868		A	G	abParts
chr15	23441738		G	A	GOLGA8E
chr15	25472362		G	A	SNORD115-15,SNURF-SNRPN
chr15	25472490		G	C	SNORD115-15,SNURF-SNRPN
chr15	25924571	rs376091391	C	T	ATP10A
chr15	30384318		G	A	DQ596686,JA429730
chr15	30773259		T	C	FLJ00278(dist_69597),FLJ00278(dist_73726)
chr15	31090172		A	G	DQ588973
chr15	31669112		T	C	KLF13
chr15	31779609		A	G	OTUD7A
chr15	32738693		C	T	JA429730
chr15	34671342		A	C	GOLGA8A
chr15	34673974		G	A	GOLGA8A
chr15	38228620	rs149517482	G	T	TMCO5A
chr15	39883924	rs58176920	T	C	THBS1
chr15	39885069	rs41305270	A	C	THBS1
chr15	41037150	rs375972281	A	G	FAM82A2
chr15	41230734		G	T	DLL4
chr15	42058091			G	MGA
chr15	42060226		G	A	MGA
chr15	42149677	rs373519127	G	A	SPTBN5
chr15	42644568		C	T	GANC
chr15	42681214	rs141948992	A	G	CAPN3
chr15	43212881		C	T	TTBK2
chr15	43569193	rs138055873	C	T	TGM7
chr15	43668969		G	A	TUBGCP4
chr15	43893734	rs138763871	G	A	STRC
chr15	43896820	rs202169022	A	C	STRC
chr15	44962119	rs376897489	C	G	PATL2
chr15	45389453	rs143471358	G	C	DUOX2
chr15	45567590	rs187388323	G	A	SLC28A2
chr15	45783196	rs146241104	T	C	SLC30A4
chr15	50399012	rs144367909	G	A	ATP8B4
chr15	51783975	rs143971162	G	A	DMXL2
chr15	53177816		C	A	ONECUT1(dist_95607),WDR72(dist_628122)
chr15	55912391	rs79244725	G	A	PRTG
chr15	57809027	rs149779988	C	T	CGNL1
chr15	58471357	rs142159680	G	A	AQP9
chr15	59465975	rs145459485	G	A	MYO1E
chr15	59548377		C	T	MYO1E
chr15	63618467	rs371148738	C	T	CA12
chr15	64976976		T	C	ZNF609
chr15	67473820		C	G	SMAD3
chr15	67985586		G	T	MAP2K5
chr15	68502055	rs144507672	G	A	CLN6
chr15	68582325	rs142755987	G	A	FEM1B
chr15	72704090		T	A	TMEM202(dist_3382),ARIH1(dist_62577)
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chr15	73064328		A	T	ADPGK
chr15	73617673	rs138714806	C	G	HCN4
chr15	74364560		G	A	GOLGA6A
chr15	74709012		A	C	SEMA7A
chr15	75541808	rs186427718	A	G	C15orf39(dist_37298),GOLGA6C(dist_9091)
chr15	75586816		G	C	GOLGA6D
chr15	75595302		C	T	DNM1P34
chr15	75659784		G		MAN2C1
chr15	76077930		C	T	DNM1P35
chr15	79586212	rs143968951	C	T	ANKRD34C
chr15	80464566		A	C	FAH

chr15	82069601		G	A	TMC3(dist_403183),MEX3B(dist_264527)
chr15	82069602		C	T	TMC3(dist_403184),MEX3B(dist_264526)
chr15	84858994		G	A	LOC100505679(dist_8009),LOC440300(dist_1606)
chr15	84950568		G	C	DQ574758
chr15	84958507		T	G	DQ574758(dist_7052),DQ576060(dist_21421)
chr15	85788463		C	T	AK301968
chr15	85804016		C	G	AK301968(dist_13606),AKAP13(dist_119855)
chr15	86213096	rs144636854	T	G	AKAP13
chr15	86273756	rs149781236	A	G	AKAP13
chr15	86290973		C	T	AKAP13
chr15	87066060	rs372004187	G	A	AGBL1
chr15	89007767		A	T	MRPL46
chr15	91447670		G	A	MAN2A2
chr15	91474511			GTC	HDDC3
chr15	93429716		A	G	LOC100507217
chr15	93522524	rs199828490	G	A	CHD2
chr15	93616331		G	A	RGMA
chr15	98517089		T	C	ARRDC4
chr15	99675659	rs187659522	C	G	SYNM
chr15	99768850		G	C	TTC23
chr15	100346890		G	A	DNM1P46
chr15	101821635	rs181805503	G	A	SNRPA1
chr15	102346417	rs199599374	C	A	OR4F6
chr16	103784		C	A	POLR3K,SNRNP25
chr16	105802	rs181031563	A	G	SNRNP25
chr16	278894		A	T	LUC7L
chr16	840044		T	C	CHTF18
chr16	840580	rs139538455	C	T	CHTF18
chr16	1035290		C	T	SOX8
chr16	1291548	rs143043553	C	T	TPSAB1
chr16	1312658	rs202160427	T	C	TPSD1(dist_4164),UBE2I(dist_46496)
chr16	1379563		A	G	UBE2I(dist_2544),BAIAP3(dist_4043)
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chr16	1396050	rs139257128	G	A	BAIAP3
chr16	1396557		G	A	BAIAP3
chr16	1748888	rs34233915	G	A	HN1L
chr16	1825321	rs141898850	G	A	EME2
chr16	2011696	rs145006994	C	T	NDUFB10
chr16	2012677	rs143347500	G	A	RPS2
chr16	2013347		G	A	RPS2
chr16	2015009	rs144198199	T		SNHG9
chr16	2055670	rs116852870	G	A	ZNF598
chr16	2284291	rs374527304	C	T	E4F1
chr16	2296910	rs140831866	G	C	ECI1
chr16	2338092	rs370076414	T	G	ABCA3
chr16	2764324	rs368697889	A	G	PRSS27
chr16	3041113		G	C	LINC00514
chr16	3117911	rs191418775	C	A	IL32
chr16	3554660	rs139983845	A	C	CLUAP1
chr16	4410470		G	A	CORO7,CORO7-PAM16
chr16	5140298		T	A	FAM86A
chr16	8898738		A	C	PMM2
chr16	11023194		G	T	DEXI
chr16	11051409	rs80252423	G	A	CLEC16A
chr16	13329390		C	A	SHISA9
chr16	15188229		C	G	PDXDC1
chr16	15221963	rs202086144	C	T	FLJ00285
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chr16	16358994	rs202085202	G	A	NOMO3
chr16	16409974		T	C	MIR3180-1(dist_6149),PKD1P1(dist_8538)
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chr16	23491121	rs200560596	A	C	GGA2
chr16	23767741		G		CHP2
chr16	27224776		T	C	JMJD5
chr16	28112999	rs199516479	C	T	XPO6
chr16	28844673	rs141936417	T	G	ATXN2L
chr16	28856191	rs145900455	T	A	NPIPL1,TUFM
chr16	29565975		C	A	BOLA2

chr16	30290490		G	A	LOC595101
chr16	30295957		C	T	LOC595101
chr16	30583859	rs182307643	G	A	ZNF688
chr16	30970601		A	G	SETD1A
chr16	55844909	rs146595460	T	C	CES1
chr16	55883627	rs181438833	G	T	CES5A
chr16	55886804	rs147727760	C	G	CES5A
chr16	56484834	rs143897621	G	T	NUDT21
chr16	56670306		CTT		MT1JP
chr16	56917997	rs79351185	C	T	SLC12A3
chr16	56970692		C	T	HERPUD1
chr16	57005363	rs35915925	G	A	CETP
chr16	57101834		A	G	NLRC5
chr16	57101835		C	T	NLRC5
chr16	57186986		G	C	FAM192A
chr16	57282311		C	T	ARL2BP
chr16	58031898		G	A	ZNF319
chr16	58312470	rs201240120	C	T	CCDC113
chr16	58537661	rs200745405	G	A	NDRG4
chr16	58554814		GGT		CNOT1
chr16	64981902		A	G	CDH11
chr16	66998387		G	C	CES3
chr16	67178592		C	T	C16orf70
chr16	67979573		G	A	SLC12A4
chr16	68772235		C	T	CDH1
chr16	68868902		G	A	CDH1
chr16	69458780		A	G	CYB5B
chr16	69790591		C	T	NOB1(dist_1762),WWP2(dist_5683)
chr16	70507153		G	A	FUK
chr16	70699180		G	A	MTSS1L
chr16	70712972		C	G	MTSS1L
chr16	71690256		G	A	PHLPP2
chr16	71692464		C	T	PHLPP2
chr16	71884209	rs77031689	C	A	ATXN1L
chr16	71976301		G		PKD1L3
chr16	75018756		G	T	WDR59
chr16	75140233		A	G	ZNRF1
chr16	75268923	rs61736821	G	A	BCAR1
chr16	75446269		C	T	CFDP1
chr16	75576580	rs201412708	A	G	TMEM231
chr16	77468360	rs199523966	C	T	ADAMTS18
chr16	82033722		C	T	SDR42E1
chr16	82033735		G	C	SDR42E1
chr16	84211808		C		TAF1C
chr16	84216440	rs142844957	C	T	TAF1C
chr16	85705886		C	T	KIAA0182
chr16	85743811		G	T	C16orf74
chr16	87678075	rs138715600	C	T	JPH3
chr16	88573964	rs111592605	C	T	ZFPM1
chr16	88808404		G	A	PIEZO1
chr16	89763405		C	T	SPATA2L
chr16	90234394		C	T	AK302511
chr17	14164		C	T	DOC2B
chr17	295729	rs185363014	C	T	FAM101B
chr17	440468	rs138877126	G	A	VPS53
chr17	489661	rs73277147	G	T	VPS53
chr17	650156	rs182380523	A	G	GEMIN4
chr17	708345	rs76916305	C	T	NXN
chr17	909317		G	A	ABR
chr17	1395657	rs56374015	C	G	MYO1C
chr17	1538682	rs139029063	C	G	SCARF1
chr17	2147818		GAAGA		SMG6
chr17	2195946	rs183768144	G	A	SMG6
chr17	2210962		T	C	SRR
chr17	2211063	rs141716829	G	C	SRR
chr17	2341705		A	C	METTL16
chr17	2586856		T	G	PAFAH1B1
chr17	3336325	rs148101354	G	A	OR1E2
chr17	3550792	rs144751390	C	T	CTNS
chr17	3832661	rs138830033	C	T	ATP2A3

chr17	3977494		C	T	ZZEF1
chr17	3994329		A	T	ZZEF1
chr17	4068131		A	G	ANKFY1
chr17	4109761	rs201117792	T	A	ANKFY1
chr17	4174688	rs186461618	G	A	UBE2G1
chr17	4446216	rs145116086	C	T	MYBBP1A
chr17	4500575		A	G	SMTNL2
chr17	4689412	rs200181773	C	T	VMO1
chr17	4693253	rs199986886	T	C	BC150535,GLTPD2
chr17	4836093		C	T	GP1BA
chr17	4856741		C	T	ENO3
chr17	5461726	rs140677797	C	G,T	NLRP1
chr17	6023882	rs148883242	G	A	WSCD1
chr17	6331702	rs16955851	T	A	AIPL1
chr17	6908625	rs147158964	G	A	ALOX12
chr17	7386116			TATG	SLC35G6
chr17	7531164		G	A	SAT2
chr17	7657928		C	T	RPL29P2
chr17	7760260		G	T	LSMD1
chr17	7960801	rs138366666	A	G	AX747630
chr17	8272563		T	C	KRBA2
chr17	10427981		G	C	MYH2
chr17	10706104	rs189037055	C	T	LOC100289255
chr17	12647908		G	C	MYOCD
chr17	16557161		C	T	ZNF624
chr17	16692501	rs201049683	A	C	FAM106CP
chr17	16704378	rs200556865	A	G	USP32P1
chr17	16753359		T	G	KRT16P2(dist_17212),FLJ00050(dist_72870)
chr17	16753486		G	A	KRT16P2(dist_17339),FLJ00050(dist_72743)
chr17	16855992		C	T	TNFRSF13B
chr17	16893649		G	T	TNFRSF13B(dist_18247),MPRIP(dist_52425)
chr17	17226505	rs190487721	C	T	NT5M
chr17	17397831			A	RASD1
chr17	17495065		C	A	PEMT
chr17	18011924		G	C	MYO15A
chr17	18012031		C	T	MYO15A
chr17	18022937	rs183969516	G	C	MYO15A
chr17	18058985		C	G	MYO15A
chr17	18301508		T	C	DQ585853,DQ595299
chr17	18314910		CCT		AX748015
chr17	18314916		C	T	AX748015
chr17	18315242			CTCTCC	AX748015
chr17	18333987		C	T	AK296148
chr17	18430070		G	A	FAM106A
chr17	18457379		G	A	USP32P2(dist_12145),CCDC144B(dist_49223)
chr17	18528463		A	C	CCDC144B
chr17	18965270			GTTTCTCTGAAC	GRAP(dist_14934),DL490867(dist_50314)
chr17	19246736		GAA		B9D1
chr17	19246739		GAG		B9D1
chr17	19449780	rs34012597	C	T	SLC47A1
chr17	19463685	rs8081204	A	G	SLC47A1
chr17	19528339	rs73310719	C	T	TRNA_Pseudo(dist_21681),ALDH3A2(dist_23725)
chr17	19645412	rs57633837	C	T	ALDH3A1
chr17	19844126	rs61655182	T	C	AKAP10
chr17	20278522		G	A	CCDC144C
chr17	20304049	rs115465086	C	T	CCDC144C
chr17	20405885		G	A,C	KRT16P3
chr17	20425160		G	A	AK127974
chr17	20435824			GGGAGA	AK127974(dist_3824),DQ600835(dist_10458)
chr17	20483817		C	T	CDRT15L2
chr17	20483818	rs116532204	G	A	CDRT15L2
chr17	20483901	rs142233315	C	T	CDRT15L2
chr17	20492390		C		CDRT15L2(dist_8166),Mir_652(dist_228509)
chr17	20492531		T	G	CDRT15L2(dist_8307),Mir_652(dist_228368)
chr17	20492642	rs2619248	A	G	CDRT15L2(dist_8418),Mir_652(dist_228257)
chr17	20493135		T	C	CDRT15L2(dist_8911),Mir_652(dist_227764)
chr17	20493194		AC		CDRT15L2(dist_8969),Mir_652(dist_227705)
chr17	20493509	rs147441312	C	T	CDRT15L2(dist_9285),Mir_652(dist_227390)
chr17	20624048		G	A	CDRT15L2(dist_139824),Mir_652(dist_96851)
chr17	20634332	rs149922239	T	C	CDRT15L2(dist_150108),Mir_652(dist_86567)

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chr17	21320850		G	A	KCNJ12,KCNJ18
chr17	25932545		G	A	KSR1
chr17	25974373	rs149003631	G	A	LGALS9
chr17	26666928		C	T	TNFAIP1
chr17	26961387		A	T	KIAA0100
chr17	26982679		G	A	SDF2
chr17	27047215		T	G	RPL23A
chr17	27061065	rs147141279	G	C	NEK8
chr17	27225513		A	G	DHRS13
chr17	27448659	rs371862120	C	T	MYO18A
chr17	27899111		G	A	TP53I13
chr17	27963197	rs138448323	T	C	SSH2
chr17	28534704		G	A	SLC6A4
chr17	31261127		A	G	TMEM98
chr17	33884688	rs199663871	G	A	SLFN14
chr17	34093309	rs182659305	G	A	MMP28
chr17	34883376	rs367752659	C	A	MYO19
chr17	36214816	rs183830426	A	G	LOC284100
chr17	36613782	rs144494500	G	A	ARHGAP23
chr17	36891266		G	A	CISD3,PCGF2
chr17	37034485	rs150792531	G	A	LASP1
chr17	37826849		A	G	PGAP3,PNMT
chr17	37886770		C	A	MIEN1
chr17	38027207		T	C	ZBPB2
chr17	38064436		A	T	GSDMB
chr17	38251358	rs140172310	T	C	NR1D1
chr17	38927984	rs140996904	G	A	KRT26
chr17	39081638	rs144893068	C	T	KRT23
chr17	40699346			G	NAGLU(dist_2880),HSD17B1(dist_4638)
chr17	40724668		G	C	MLX,PSMC3IP
chr17	41585386		A	G	DHX8
chr17	42462301	rs374469971	G	A	ITGA2B
chr17	44049429		G	T	MAPT
chr17	48264289		C	A	COL1A1
chr17	56699380		C	T	TEX14
chr17	58132920		G	C	HEATR6
chr17	59885856	rs28997570	T	C	BRIP1
chr17	60038278		T	C	MED13
chr17	60690994		T	C	TLK2
chr17	60802372	rs111291268	G	A	MAR10
chr17	60827878	rs112201730	G	A	MAR10
chr17	61772692		T	C	MAP3K3
chr17	61903283		T	C	FTSJ3
chr17	61912422		G	A	SMARCD2
chr17	61972753		A	T	CSH1
chr17	61996176	rs368843010	G	A	GH1
chr17	62530438	rs141992513	C	T	CEP95
chr17	65027942	rs185149010	C	T	CACNG4
chr17	65105363	rs200486206	T	G	HELZ
chr17	65936654	rs200180767	G	A	BPTF
chr17	67109643		T	C	ABCA6
chr17	70643662	rs183190153	T	C	SLC39A11
chr17	72842224	rs41282061	G	C	GRIN2C
chr17	77681534		G	A	HP09025
chr17	77808866	rs77181887	C	T	CBX4
chr17	78226557		G	A	SLC26A11
chr17	78307917	rs113139767	G	A	RNF213
chr17	79226014	rs137924826	G	A	SLC38A10
chr17	79525134		C	T	NPLOC4
chr17	79671148	rs117968022	G	A	MRPL12,SLC25A10
chr17	80008329		G	A	RFNG
chr17	80430508		G	A	NARF
chr17	80561003		A	G	FOXK2
chr18	8829020		C	T	CCDC165
chr18	8831735		G	A	CCDC165
chr18	11610762		C	T	PIEZO2(dist_462001),DQ594439(dist_43191)
chr18	11853614		T	G	CHMP1B
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chr18	12859422	rs117614942	C	G	PTPN2

chr18	19447400	rs142654284	G	A	MIB1
chr18	21118754	rs149795082	C	T	NPC1
chr18	21120398		AAG		NPC1
chr18	21120398		AG		NPC1
chr18	21453038	rs145044428	C	T	LAMA3
chr18	46446675		G	A	SMAD7
chr18	47322244		C	A	ACAA2
chr18	47927506		TCT		SKA1(dist_6966),MAPK4(dist_158978)
chr18	48190511	rs55800528	C	T	MAPK4
chr18	54629727	rs143674934	G	A	WDR7
chr18	55273928	rs370440566	G	A	NARS
chr18	57567520		G	T	PMAIP1
chr18	60807115	rs138259755	A	T	BCL2
chr18	61089598		T	G	VPS4B
chr18	67231857	rs139324250	G	A,C	DOK6
chr18	67721409	rs35374168	T	C	RTTN
chr18	71740524		C		FBXO15
chr18	74208258		CTT		AK126293
chr18	74637173	rs183899517	C	T	ZNF236
chr18	74692253		C	T	AK094611
chr18	77171164	rs76525142	G	A	NFATC1
chr18	77475372		G	A	CTDP1
chr19	239219		A	C	DQ593908(dist_29863),PPAP2C(dist_41825)
chr19	362268	rs112813656	C	T	THEG
chr19	841082	rs201472135	C	T	PRTN3
chr19	871248		C	T	MED16
chr19	1078260		A	C	HMHA1
chr19	1086060	rs190907000	C	G	HMHA1
chr19	1111958		C	T	SBNO2
chr19	1226640		G	A	STK11
chr19	1924398		G	A	SCAMP4
chr19	1989953	rs373290178	C	G	BTBD2
chr19	2099372	rs372167894	G	A	IZUMO4
chr19	2270037		G	T	OAZ1,SPPL2B
chr19	2431477			TG	
chr19	2737187	rs371495611	G	A	SLC39A3
chr19	2808456	rs200160673	C	T	
chr19	2841436		G	C	ZNF555
chr19	3005425	rs144033921	C	T	TLE2
chr19	3005689	rs181337463	G	C	TLE2
chr19	3179427	rs143964699	G	A	S1PR4
chr19	3590375		G	A	GIPC3
chr19	4207293	rs199801260	T	C	ANKRD24
chr19	4433466	rs149907294	C	T	CHAF1A
chr19	4502326		C	T	PLIN4
chr19	4795688	rs34393409	A	G	AK126532
chr19	4947257	rs201350936	A	G	UHRF1
chr19	5641697	rs139125544	G	A	SAFB
chr19	5995598	rs201301974	G	A	RFX2
chr19	6719034		G	A	C3
chr19	7032766		C	T	MBD3L5
chr19	7420322		C	T	INSR(dist_126311),ARHGEF18(dist_27398)
chr19	7616183	rs186340734	G	A	PNPLA6
chr19	7986401	rs145231464	G	A	SNAPC2
chr19	9014463		C		
chr19	9071401	rs182936649	G	A	MUC16
chr19	9074922		A		MUC16
chr19	9271071	rs141277819	C	T	ZNF317
chr19	9938722		G	C	UBL5
chr19	10105963		G	A	COL5A3
chr19	10292808	rs140385241	G	A	DNMT1
chr19	10292907	rs79902277	A	G	DNMT1
chr19	11312612		C	A	DOCK6
chr19	11326221	rs146084897	G	C	DOCK6
chr19	11664044		G	A	ELOF1
chr19	12267288		G		ZNF20,ZNF625
chr19	12511858	rs199800367	G	A	ZNF799
chr19	12811602		G	A	TNPO2
chr19	12969179		C	T	MAST1
chr19	13888892		G	T	C19orf53



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chr19	15295265	rs190177286	G	C	NOTCH3
chr19	18375511	rs375229949	G	A	KIAA1683
chr19	18554086		C	T	ELL
chr19	21240227		A	G	ZNF430
chr19	22778979	rs188334423	C	T	LOC440518
chr19	23543329	rs199921854	T	C	ZNF91
chr19	29698188		T	A	UQCRFS1
chr19	33300908		T	G	TDRD12
chr19	33517549		A	C	RHPN2
chr19	33695885	rs139036056	T	G	LRP3
chr19	33697128		C	T	LRP3
chr19	34856723		G	A	GPI
chr19	35612329		C	T	FXD3
chr19	35835659		A	T	CD22
chr19	36037223	rs181445157	A	C	TMEM147
chr19	37038224	rs370748331	A	G	ZNF529
chr19	37309643	rs199662090	C	T	ZNF790
chr19	37854273	rs141902493	G	A	HKR1
chr19	39109784	rs2232993	A	G	EIF3K
chr19	39321709	rs201526955	G	C	ECH1,HNRNPL
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chr19	40827216		G	C	C19orf47
chr19	41133059		C	T	LTBP4
chr19	41211091		G	A	ADCK4
chr19	41248551	rs144724170	G	A	C19orf54
chr19	41698002	rs187440090	G	A	DQ590318(dist_56040),CYP2S1(dist_1113)
chr19	42027478		T	C	LOC100505495(dist_20924),CEACAM21(dist_55053)
chr19	42219659	rs144819038	C	T	CEACAM5
chr19	42880522	rs370578585	G	A	MEGF8
chr19	42907292		TG		AK311181
chr19	44536123	rs74659740	A	G	ZNF222
chr19	44635702	rs200227995	G	A	ZNF225
chr19	45028231	rs36053277	G	A	CEACAM20
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chr19	45411110	rs769452	T	C	APOE
chr19	45578863		C	G	ZNF296
chr19	45641929		A	G	PPP1R37
chr19	45922382		G	A	ERCC1
chr19	45977823		G	T	FOSB
chr19	46032441	rs201888844	T	A	OPA3
chr19	46094063	rs147607317	C	T	GPR4
chr19	46195192		T	G	SNRPD2
chr19	46303572		G	A	RSPH6A
chr19	49132108		A	C	SPHK2
chr19	49248578		C	T	IZUMO1
chr19	49403954		G	A	NUCB1
chr19	49589897	rs143820545	A	G	SNRNP70
chr19	49843687			CC	CD37
chr19	50166918	rs183177527	C	T	IRF3
chr19	50482369	rs140365927	G	T	VRK3
chr19	50655899		T		IZUMO2
chr19	50794283		T	C	MYH14
chr19	50839885	rs142503912	G	T	NAPSB
chr19	51165654	rs374127192	C	T	SHANK1
chr19	51507337	rs183694524	C	T	KLK9
chr19	51559597			GG	KLK13
chr19	51671579	rs200616536	G	A	SIGLECP3
chr19	51689680	rs372327052	A	C	SIGLECP3(dist_12900),CD33(dist_38655)
chr19	51729630	rs368789174	C	T	CD33
chr19	51742917	rs148758925	G	A	CD33
chr19	52395150	rs150082546	T	C	ZNF649
chr19	52795363	rs7248487	A	G	ZNF766
chr19	53553068		A	G	ERVV-2
chr19	53994611	rs117322260	A	G	ZNF813
chr19	54177310	rs148036416	T	C	MIR498
chr19	54191721	rs59229577	T	C	MIR1283-1
chr19	54194015	rs145528706	T	G	MIR520A
chr19	54201728	rs182668166	A	G	MIR523,MIR525

chr19	54214432	rs141856595	G	C	MIR524
chr19	54215346	rs104151111	A	G	MIR517A
chr19	54215828	rs7257536	G	A	MIR519D
chr19	54219798	rs115691838	C	A	MIR521-2
chr19	54545281		G	A	VSTM1
chr19	54598563		C	G	OSCAR
chr19	54618552		C	T	TFPT
chr19	54684467		A	G	MBOAT7
chr19	55105359		C	T	LILRA1,LILRA2
chr19	55175710	rs150571856	G	A	LILRB4
chr19	55258713		C	T	KIR2DL2,KIR2DL3
chr19	55266576		T	C	KIR2DL2,KIR2DL3,KIR2DS4
chr19	55644082		C	G	TNNT1
chr19	55711704	rs142951925	C	T	PTPRH
chr19	55739485		C	T	TMEM86B
chr19	55918770		G	A	UBE2S
chr19	57293351	rs139421959	G	T	ZIM2
chr19	57323878	rs191672288	A	T	PEG3-AS1
chr19	58004993	rs201067474	C	T	ZNF419
chr19	58557728		G	C	ZSCAN1
chr19	58755406	rs138031572	T	C	ZNF544
chr20	982903		T	C	RSPO4
chr20	2320535	rs76528811	C	T	TGM3
chr20	2464237		C	T	ZNF343
chr20	2637237	rs55904186	G	A	NOP56
chr20	3025358	rs368671021	T	C	GNRH2
chr20	3103393	rs193292020	T	C	LOC100134015
chr20	3734564		A	C	C20orf27
chr20	5903010		A	G	CHGB
chr20	8864472	rs202098034		TCTT	PLCB1
chr20	17639597		C	T	RRBP1
chr20	17956347	rs143417446	C	T	C20orf72
chr20	18167778	rs143936715	A	G	CSRP2BP
chr20	18424032	rs41276434	G	C	DZANK1
chr20	18446029		T	C	DZANK1
chr20	18470482		A	T	RBBP9
chr20	21492236	rs145444905	C	T	NKX2-2
chr20	21686344		C	T	PAX1
chr20	23335299	rs2683	C	T	NXT1
chr20	24565429	rs182971715	A	G	SYNDIG1
chr20	25061845		G	C	VSX1
chr20	25288786		G	A	ABHD12
chr20	25666773		C	A	ZNF337
chr20	29948832	rs6057613	C	A,T	DEFB116(dist_52444),DEFB118(dist_7589)
chr20	30432724	rs201517755	C	T	FOXS1
chr20	32266060	rs199745489	C	T	E2F1
chr20	32957010		C	T	ITCH
chr20	34246710		A	C	CPNE1,RBM12
chr20	34443174		A	G	PHF20
chr20	34638472		T	C	LOC647979
chr20	35242164		A	G	SLA2
chr20	36488536		C	T	CTNBL1
chr20	39989932		C	T	EMILIN3
chr20	39989937		C	T	EMILIN3
chr20	40980902	rs201153898	C	T	PTPRT
chr20	42208627	rs35793869	C	T	SGK2
chr20	43257934		G	A	ADA
chr20	43600842	rs200616256	T	C	STK4
chr20	44035268		C	A	DBNDD2
chr20	44463024			CGAGGACGA	SNX21
chr20	44463037			GACGAGGACGACGAG	SNX21
chr20	44468987		C	G	SNX21
chr20	44472152	rs41282772	G	A	ACOT8
chr20	45315157	rs45505297	G	A	TP53RK
chr20	45891386		A	C	ZMYND8
chr20	48844933	rs78566106	G	A	CEBPB(dist_35706),AK307192(dist_39102)
chr20	49126945		G	C	PTPN1
chr20	49200434		C	T	PTPN1
chr20	49231183		A	C	FAM65C
chr20	50217816	rs138952924	G	C	ATP9A

chr20	52198285	rs146782561	C	T	ZNF217
chr20	52700219	rs112306370		G	BCAS1(dist_12915),CYP24A1(dist_69769)
chr20	53208527	rs7272626	A	G	DOK5
chr20	54572731		G	T	CBLN4
chr20	54956620		T	C	AURKA
chr20	55750060	rs61733438	T	C	BMP7
chr20	57242132		T	C	STX16
chr20	57269565	rs370807734	C	T	NPEPL1
chr20	60847361		C	T	OSBPL2
chr20	60884813		G	C	LAMA5
chr20	60902342	rs371271948	C	T	LAMA5
chr20	61039710		C	T	GATA5
chr20	61162228	rs199563235	C	T	C20orf166
chr20	61296677		C	T	LOC100127888
chr20	61449904	rs149582526	C	T	COL9A3
chr20	61945147	rs376343023	C	T	COL20A1
chr20	62367256	rs144346262	G	T	ZGPAT; LIME1
chr20	62550796	rs370651852	C	T	MIR941-1
chr20	62563657		G	A	DNAJC5
chr20	62844979	rs140907107	C	G	MYT1
chr20	62904715	rs144264619	G	A	PCMTD2
chr21	10914438	rs141919406	A	C	TPTE
chr21	11013244		T	C	TPTE(dist_22324),BAGE2(dist_7569)
chr21	14424104	rs1054925	G	C	ANKRD30BP2
chr21	17681751		A	G	LINC00478
chr21	27107620		T	G	ATP5J,GABPA
chr21	32639257		T	C	TIAM1
chr21	33755761	rs185028523	C	A	URB1
chr21	40193616	rs185172042	C	T	ETS2
chr21	41301136		C	T	PCP4
chr21	42837287		G	A	pp9284
chr21	43230669	rs146699277	C	T	PRDM15
chr21	43319355	rs150987090	C	T	C2CD2
chr21	43410876	rs113350151	A	G	ZNF295
chr21	43720172	rs182781701	C	G	AL355711
chr21	43770042	rs141463867	G	A	TFF2
chr21	43857549		A	T	UBASH3A
chr21	44568002	rs148729434	C	T	U2AF1(dist_40314),CRYAA(dist_21139)
chr21	44589747		C	G	CRYAA
chr21	45166165		A	G	PDXK
chr21	45168985		TTTATTTTAC		
chr21	45389040	rs75532875	C	T	AGPAT3
chr21	45429152	rs147943694	T	G	AGPAT3(dist_21677),TRAPPC10(dist_3054)
chr21	45429457		T	G	AGPAT3(dist_21982),TRAPPC10(dist_2749)
chr21	45528760	rs111704236	A	G	PWP2
chr21	45540962	rs138954845	A	G	PWP2
chr21	45544621		C	T	PWP2
chr21	45547103	rs182880150	C	T	PWP2
chr21	45557143	rs61737073	C	T	C21orf33
chr21	45565624	rs140876259	G	T	C21orf33
chr21	45648978		T	C	ICOSLG
chr21	46058114		C	A	KRTAP10-10
chr21	46058277		G	C	KRTAP10-10
chr21	46271414		A	G	PTTG1IP
chr21	46308800	rs2230531	C	T	ITGB2
chr21	46330190	rs199948899	G	A	ITGB2
chr21	46914760		C	T	COL18A1
chr21	46918394		G	A	SLC19A1
chr21	47602854		G	A	C21orf56
chr21	47686037	rs144904756	A	C	MCM3AP
chr22	16255824		T	C	P704P
chr22	17119251		G	A	TPTEP1
chr22	17589509	rs369912474	T	C	IL17RA
chr22	25498937		C	A	LOC100128531
chr22	26886069	rs114904777	G	A	SRRD
chr22	29140559	rs73170629	T	C	HSCB
chr22	31042606	rs148304964	G	A	SLC35E4
chr22	32808249		G	A	C22orf28
chr22	32924024	rs146298573	A	G	SYN3
chr22	36655614		G	A	APOL1

chr22	38120712	rs186620158	C	A,G	TRIOBP
chr22	38201496	rs186139362	C	T	H1F0
chr22	39473490		G	C	APOBEC3G
chr22	39910061		G	A	SMCR7L
chr22	40661068		C	A	TNRC6B
chr22	41591639		C	T	BC040700
chr22	41609673		T		AK057177
chr22	41636806		G	C	CHADL
chr22	41864052	rs61736535	T	C	PHF5A
chr22	41926757	rs35463585	G	A	POLR3H
chr22	41940075	rs148608238	C	T	POLR3H
chr22	42016757		C	T	PPPDE2
chr22	42463715		G	T	NAGA
chr22	44285371		G	A	PNPLA5
chr22	45277072		G	T	PHF21B
chr22	45691510	rs143237351	G	A	UPK3A
chr22	45802410	rs377173833	T	G	SMC1B
chr22	45939294	rs140586412	C	T	FBLN1
chr22	46508887		A	G	DM119500,MIR4763,MIRLET7B
chr22	46684178		C	T	TTC38
chr22	46758391		C	T	CELSR1
chr22	46769075		C	G	CELSR1
chr22	47185269		G	A	TBC1D22A
chr22	48972450		C	G	FAM19A5
chr22	50278031	rs146151193	G	A	ZBED4
chr22	50644876	rs377092978	G	A	SELO
chr22	50718513	rs202091361	G	A	PLXNB2
chr22	50895458	rs115032856	G	C	SBF1
chr22	50925232		C	T	MIOX
chr22	50944491	rs141408392	G	A	LMF2
chr22	51207190	rs376882402	C	G	RABL2B
chr22	51207620		A	T	RPL23AP82
chrM	7023		T	C	BC018860
chrX	3238115	rs140695513	T	C	MXRA5
chrX	7811752	rs369525999	C	A	VCX
chrX	8138177		G	T	VCX2
chrX	9915107		A	G	SHROOM2
chrX	9916899		C		SHROOM2
chrX	12739059	rs41297281	C	G	FRMPD4
chrX	13338658		A	G	ATXN3L
chrX	14549268		C	T	GLRA2
chrX	17579425	rs191448394	T	A	NHS
chrX	23689967	rs148808015	A	G	PRDX4
chrX	24304528	rs193138333	G	A	ZFX(dist_70156),FAM48B2(dist_24451)
chrX	24330928	rs200275949	G	C	FAM48B2
chrX	27840405	rs146965052	G	A	MAGEB10
chrX	29974783			AT	
chrX	31198768		T	A	DMD
chrX	36162739		A	G	CXorf59
chrX	37029074		A	G	FAM47C
chrX	39910743		T	C	BCOR
chrX	40597472		C	T	BC039399
chrX	41530851				ATAAGGATTCATTGAAGATGA
chrX	44382909				TGTT
chrX	44870417	rs41309721	G	A	FUNDC1
chrX	46950859		C	G	KDM6A
chrX	47046040		TCC		RGN
chrX	47046043	rs200521605	G	T	RBM10
chrX	47445282		G	A	RBM10
chrX	47445366	rs150231209	G	A	TIMP1
chrX	47584922		G	C	TIMP1
chrX	47919343		T	C	CXXC1P1
chrX	48814964		G	A	ZNF630
chrX	49031294		C	A	OTUD5
chrX	49061358		C		PLP2
chrX	57622549				TTCCTCTACTGAATAATT
chrX	62917140		G	A	CACNA1F
chrX	65242554	rs182374510	C	T	ZXDB
chrX	67283825	rs143713841	G	T	ARHGEF9
chrX	68058970		A	G	VSIG4
					OPHN1
					EFNB1

chrX	69644899		A	G	GDPD2
chrX	69723852		C	T	DLG3
chrX	70535990		A	G	BCYRN1
chrX	70794373		TCAAGCCTGAT		OGT
chrX	71525279	rs190894379	G	A	CITED1
chrX	73955082		T	C	KIAA2022
chrX	77160840	rs41304464	G	T	COX7B
chrX	84526516	rs138675194	A	G	ZNF711
chrX	84532913		A	G	POF1B
chrX	85218753	rs374880482	T	C	CHM
chrX	86888870	rs149752456	A	T	KLHL4
chrX	101087536		C	T	NXF5
chrX	101969860	rs137939858	G	C	ARMCX5-GPRASP2,GPRASP2
chrX	103494779	rs188475442	G	T	ESX1
chrX	105189958	rs200616257	T	C	NRK
chrX	107288267		G	A	VSIG1
chrX	108917879			GG	ACSL4
chrX	108917880			GT	ACSL4
chrX	108917880		A	G,T	
chrX	109441790	rs140247782	A	G	AMMECR1
chrX	109582509	rs151041387	T	C	AMMECR1
chrX	110924386		G	T	ALG13
chrX	111018789		GTA		TRPC5
chrX	111625429	rs188300399	A	G	ZCCHC16
chrX	111699585		T	G	ZCCHC16
chrX	119514807	rs78375633	A	G	ATP1B4
chrX	120183640		A	G	GLUD2
chrX	123043602		T	C	XIAP
chrX	130215563		C	T	ARHGAP36
chrX	130223863		A	G	ARHGAP36
chrX	131351458		G	C	RAP2C
chrX	131759883		G	A	HS6ST2
chrX	134494878	rs141878135	C	T	ZNF449
chrX	140269913		C	G	LDOC1
chrX	149657999	rs192797759	G	A	MAMLD1
chrX	152134019	rs190679591	G	A	ZNF185
chrX	152160081	rs370233185	A	T	PNMA5
chrX	152612801	rs61999320	G	A	ZNF275
chrX	152771326	rs146322407	C	T	BGN
chrX	152954356		G	A	SLC6A8
chrX	153184771	rs74328477	G	A	ARHGAP4
chrX	153191716	rs12834662	G	A	ARHGAP4
chrX	153247356		T	G	TMEM187
chrX	153581375	rs371611462	C	T	FLNA
chrX	153718446		C	G	SLC10A3
chrX	153877495		C	G	IKBKG(dist_6478),CTAG2(dist_2751)
chrX	153997389		A	G	DKC1
chrY	28716110		G	A	U6(dist_208871),NONE(dist_NONE)

**Supplemental Table 3.** Results in genes selected for Ion Torrent validation and follow-up study (A=affected, C=obligate carrier, U=unaffected).

Count	Gene symbol	No amplicons	Chromosome	Position	Reference Allele	Sample Allele	Gene Region	Translation Impact	Protein Variant	Family members with variant	Summary of FU results
1	AURKA	13	20	54956620	T	C	Exonic	missense	p.N192D	4/4 A, 1/1 C, 1/5 U;	Segregation in F4;
2	BCAR1	16	16	75268923	G	A	Exonic	missense	p.P671L	3/3 A;	Segregation in F13;
3	CD33	12	19	51742917	G	A	Exonic	missense	p.E230K; p.E357K	3/3 A;	Segregation in F6;
3	CD33	12	19	51729630	C	T	Intronic	NA		1/2 A;	Nonsegregation in F20;
4	COX7A2L	4	2	42578483	G	A	Exonic	missense	p.P74L	3/3 A;	Segregation in F14;
5	CYSLTR2	8	13	49281386	C	T	Exonic	missense	p.R145W	3/3 A;	Segregation in F6;
6	EMILIN3	16	20	39989937	C	T	Exonic	missense	p.E758K	2/2 A, 2/2 C;	Segregation in F8;
6	EMILIN3	16	20	39989932	C	T	Exonic	synonymous	p.Q759Q	2/2 A, 3/3 C, 1/3 U;	Segregation in F2;
6	EMILIN3	16	20	39990710	C	T	Exonic	missense	p.R500Q	2/2 A, 3/3 C, 1/3 U;	Segregation in F2;
6	EMILIN3	16	20	39993740	C	T	Exonic	missense	p.R500Q	2/2 A;	Segregation in F36;
6	EMILIN3	16	20	39993740	C	T	Exonic	synonymous	p.A75A	2/2 A;	Segregation in F41;
7	FAM86A	10	16	5140298	T	A	Exonic	stop gain	p.K143*; p.K177*	2/2 A, 1/2 C, 2/2 U;	Nonsegregation in F7;
8	FSTL5	32	4	162841700	A	G	Exonic	missense	p.C89R; p.C88R	2/2 A, 2/2 C, 1/2 U;	Segregation in F7;
8	FSTL5	32	4	162376067	C	T	Intronic	NA		2/2 A;	Segregation in F30;
9	GPT	21	8	145730071	C	T	Exonic	missense	p.R83C	4/4 A, 1/1 C, 4/5 U;	Segregation in F4;
9	GPT	21	8	145732151	G	A	Exonic	missense	p.R442H	2/2 A;	Segregation in F47;
10	HLTF	49	3	148757909	A	G	Exonic	missense	p.I804T	3/3 A, 1/1 U;	Segregation in F1;
10	HLTF	49	3	148750300	A	T	Intronic	NA		1/2 A, 2/2 C, 0/2 U;	Nonsegregation in F7;
10	HLTF	49	3	148757805	AT		Intronic	NA		2/2 A;	Segregation in F45;
10	HLTF	49	3	148778374	G	A	Intronic	NA		2/2 A;	Segregation in F45;
10	HLTF	49	3	148782649	T	C	Exonic	missense	p.Y332C	2/2 A;	Segregation in F41;
10	HLTF	49	3	148782649	T	C	Exonic	missense	p.Y332C	2/2 A;	Segregation in F45;
10	HLTF	49	3	148786021	C	T	Intronic	NA		2/2 A;	Segregation in F29;
11	IL17RA	25	22	17589509	T	C	Exonic	missense	p.L467P	2/2 A, 1/2 C, 2/2 U;	Nonsegregation in F7;
11	IL17RA	25	22	17577965	C	T	Exonic	missense	p.T51M	2/2 A;	Segregation in F41;
11	IL17RA	25	22	17578011	C	T	Intronic	NA		1/1 A;	Single in F61;
11	IL17RA	25	22	17589639	C	T	Exonic	synonymous	p.D510D	2/2 A;	Segregation in F46;
11	IL17RA	25	22	17589928	G	A	Exonic	missense	p.E607K	2/2 A;	Segregation in F31;
11	IL17RA	25	22	17589928	G	A	Exonic	missense	p.E607K	2/2 A;	Segregation in F39;
12	ITGB2	28	21	46308800	C	T	Exonic	missense	p.E630K	2/2 A, 3/3 C, 0/3 U;	Segregation in F2;
12	ITGB2	28	21	46308800	C	T	Exonic	missense	p.E630K	2/4 A, 0/0 U;	Nonsegregation in F8;
13	KDR	49	4	55955969	C	T	Exonic	missense	p.A1065T	3/3 A;	Segregation in F6;
13	KDR	49	4	55963956	C		Intronic	NA		2/2A;	Segregation in F30;
13	KDR	49	4	55972081	C	T	Exonic	synonymous	p.A521A	2/2 A;	Segregation in F33;
13	KDR	49	4	55953744	CCCCG	CCCCG	Intronic	NA		2/2 A;	Segregation in F21;
13	KDR	49	4	55955475		CT	Intronic	NA		1/1 A;	Single in F59;
14	KRT82	15	12	52789643	C	T	ncRNA, Intron	NA		1/1 A;	Single in F57;
14	KRT82	15	12	52797534	C	G	Exonic	missense	p.E191Q	2/2 A, 3/3 C, 2/3 U;	Segregation in F2;
15	MYL6	8	12	56553920	C	T	Exonic	missense	p.L113F	1/1 A;	Single in F50;
16	NKD2	15	5	1033498	G	A	Exonic	missense	p.A72T	3/3 A, 1/1 C, 0/1 U;	Segregation in F5;
16	NKD2	15	5	1038278	C	T	Exonic; 3'UTR	synonymous	p.H382H	2/2 A, 3/3 C, 2/3 U;	Segregation in F2;
16	NKD2	15	5	1038278	C	T	Exonic; 3'UTR	synonymous	p.H382H	1/2 A;	Nonsegregation in F9;
17	PPP1R32	14	11	61249376	A	G	Exonic	missense	p.Y32C	2/2 A;	Segregation in F36;
17	PPP1R32	14	11	61249804	G	A	Exonic	missense	p.R44H	2/2 A, 3/3 C, 2/3 U;	Segregation in F2;
17	PPP1R32	14	11	61252200	A	G	Exonic	missense	p.Q141R	2/2 A;	Segregation in F24;
17	PPP1R32	14	11	61252303	A	G	Exonic	missense	p.Q141R	2/2 A;	Segregation in F20;
17	PPP1R32	14	11	61252303	A	G	Intronic	NA		2/2 A;	Segregation in F20;
17	PPP1R32	14	11	61252303	A	G	Intronic	NA		2/2 A;	Segregation in F45;
18	RHOG	4	11	3849350	C	A	Exonic	missense		2/2 A;	Segregation in F45;
19	SDR42E1	9	16	82033722	C	T	Exonic	missense	p.R59H	3/3 A, 1/1 C, 0/1 U;	Segregation in F1;
19	SDR42E1	9	16	82033735	G	C	Exonic	missense	p.Q55E	3/3 A, 1/1 C, 0/1 U;	Segregation in F5;
20	SMTNL2	13	17	4500575	A	G	Exonic	missense	p.T406A; p.T262A	4/4 A, 1/1 C, 3/5 U;	Segregation in F5;
21	SUDS3	15	12	118821819	T	C	Exonic	missense	p.M72T	3/3 A;	Segregation in F4;
22	VNN2	16	6	133073884	T	C	Exonic	missense	p.H181R; p.H128R	4/4 A, 1/1 C, 3/5 U;	Segregation in F4;
22	VNN2	16	6	133073884	T	C	Exonic	missense	p.H181R; p.H128R	3/3 A;	Segregation in F6;
22	VNN2	16	6	133073884	T	C	Exonic	missense	p.H181R; p.H128R	2/2 A, 2/2 C, 0/2 U;	Segregation in F7;
23	ZGPAT; LIME1	10	20	62367256	G	T	Exonic	missense	p.K507N; p.K498N;	3/3 A;	Segregation in F6;
23	ZGPAT; LIME1	10	20	62370169	C	T	Exonic	synonymous	p.S268S	2/2 A;	Segregation in F23;

**Supplemental Table 4.** Clinical characteristics of the KDR p.A1065T mutation segregating families (U=Unaffected, HL=Hodgkin lymphoma, NHL=non-Hodgkin lymphoma, DLBCL=diffuse large B-cell lymphoma, NS= Nodular sclerosis).

Family	Member	Diagnosis	Method of Diagnosis	Sex	Age at diagnosis or exam	Presence of p.A1065T KDR variant
F6	I.1	U		M	NA	NA
F6	I.2	U		F	NA	NA
F6	II.1	U		M	48	no
F6	II.2	U, obligate carrier		F	47	yes
F6	II.3	NHL, DLBCL	NIH slide review	M	26	NA
F6	II.4	U		F	NA	NA
F6	III.1	HL	Path report	M	13	yes
F6	III.2	HL, NS	NIH slide review	M	16	yes
F6	III.3	U		M	14	no
F6	III.4	HL, NS	NIH slide review	M	18	yes
F6	III.5	U		M	NA	NA
F30	I.1	Lymphoma	Death certificate	M	82	NA
F30	I.2	U		F	NA	NA
F30	I.3	U		M	NA	NA
F30	I.4	U		F	NA	NA
F30	II.1	U		F	NA	NA
F30	II.2	U		M	NA	NA
F30	II.3	U		F	80	no
F30	II.4	Lymphoma	Death certificate	M	44	NA
F30	III.1	HL, NS	Path report	M	40	yes
F30	III.2	U		F	49	NA
F30	IV.1	HL, NS	Path report	F	14	yes
F30	IV.2	U		F	18	yes
F30	IV.3	U		M	13	yes

**Supplemental Table 5.** Predictions of the effect of the KDR variant p.A1065T based on various impact assessment programs available in AVIA v2.

Impact Assessment Program	Prediction
SIFT	Tolerated
Polyphen2 (Human Var)	Damaging
Polyphen2 (Human Div)	Damaging
Mutation Taster	Disease Causing
Mutation Accessor	Neutral
CADD	Damaging
FATHMM	Deleterious
VEST	Damaging



**Supplemental Table 6A.** Ingenuity Variant Analysis: top enriched pathways based on 2699 selected variants.

Name	p-value	#Genes	#Variants
Role of Macrophages, Fibroblasts and Endothelial Cells in Rheumatoid Arthritis	1.73E-04	17	19
LPS/IL-1 Mediated Inhibition of RXR Function	2.70E-04	11	11
Altered T Cell and B Cell Signaling in Rheumatoid Arthritis	7.20E-04	4	4
Hematopoiesis from Pluripotent Stem Cells	7.36E-04	1	1
Allograft Rejection Signaling	7.62E-04	3	3
OX40 Signaling Pathway	1.19E-03	3	3
Communication between Innate and Adaptive Immune Cells	1.36E-03	5	5
IL-10 Signaling	1.86E-03	1	1
Autoimmune Thyroid Disease Signaling	2.05E-03	2	2
CCR5 Signaling in Macrophages	2.19E-03	6	8
Type I Diabetes Mellitus Signaling	3.19E-03	6	6
Superpathway of Cholesterol Biosynthesis	3.25E-03	2	4
Oxidative Ethanol Degradation III	3.94E-03	1	1
Lipid Antigen Presentation by CD1	4.28E-03	1	1
Regulation of IL-2 Expression in Activated and Anergic T Lymphocytes	5.33E-03	5	5
Role of NFAT in Regulation of the Immune Response	5.60E-03	13	15
Hepatic Cholestasis	6.23E-03	11	11
Role of Tissue Factor in Cancer	1.06E-02	6	6
Dendritic Cell Maturation	1.13E-02	15	16
TREM1 Signaling	1.16E-02	3	4
Type II Diabetes Mellitus Signaling	1.19E-02	11	13
Bile Acid Biosynthesis, Neutral Pathway	1.24E-02	1	1
SAPK/JNK Signaling	1.38E-02	7	7
G Protein Signaling Mediated by Tubby	1.58E-02	1	1
Pyrimidine Ribonucleotides De Novo Biosynthesis	1.58E-02	1	1
LPS-stimulated MAPK Signaling	1.73E-02	3	3
Role of NFAT in Cardiac Hypertrophy	1.74E-02	14	16
Toll-like Receptor Signaling	1.85E-02	3	3
Human Embryonic Stem Cell Pluripotency	1.92E-02	11	15
Crosstalk between Dendritic Cells and Natural Killer Cells	1.93E-02	5	5
Chondroitin Sulfate Biosynthesis	1.94E-02	2	2
ILK Signaling	1.95E-02	31	34
iCOS-iCOSL Signaling in T Helper Cells	2.02E-02	8	10
Lymphotoxin I <sup>2</sup> Receptor Signaling	2.08E-02	2	2
Cdc42 Signaling	2.25E-02	15	16
Cytotoxic T Lymphocyte-mediated Apoptosis of Target Cells	2.40E-02	2	2
Chondroitin Sulfate Biosynthesis (Late Stages)	2.54E-02	1	1
Role of Osteoblasts, Osteoclasts and Chondrocytes in Rheumatoid Arthritis	2.70E-02	17	17
Sperm Motility	2.74E-02	10	13
B Cell Receptor Signaling	2.79E-02	13	14
IL-1 Signaling	3.24E-02	5	5
PPAR Signaling	3.27E-02	6	7
CD27 Signaling in Lymphocytes	3.66E-02	2	2
FAK Signaling	3.70E-02	20	21
Telomerase Signaling	3.71E-02	17	17
PKC $\delta$ Signaling in T Lymphocytes	3.75E-02	13	15
Granulocyte Adhesion and Diapedesis	3.78E-02	12	13
CD40 Signaling	3.79E-02	3	3
Relaxin Signaling	3.94E-02	9	9
NRF2-mediated Oxidative Stress Response	3.96E-02	17	18
UVC-Induced MAPK Signaling	4.07E-02	1	1
Hepatic Fibrosis / Hepatic Stellate Cell Activation	4.09E-02	31	38
Actin Cytoskeleton Signaling	4.10E-02	37	41
CCR3 Signaling in Eosinophils	4.27E-02	8	10
GPCR-Mediated Nutrient Sensing in Enteroendocrine Cells	4.40E-02	7	9
Integrin Signaling	4.40E-02	30	34
Cardiac Hypertrophy Signaling	4.48E-02	18	18
Cholesterol Biosynthesis I	4.76E-02	1	3
Cholesterol Biosynthesis II (via 24,25-dihydrolanosterol)	4.76E-02	1	3
Cholesterol Biosynthesis III (via Desmosterol)	4.76E-02	1	3
iNOS Signaling	4.83E-02	2	2
G Beta Gamma Signaling	4.95E-02	9	11

**Supplemental Table 6B.** Ingenuity Variant Analysis: top enriched groups based on 2699 selected variants.

Name	p-value	#Genes	#Variants
collagen	2.26E-06	15	20
CYP	3.05E-05	1	1
UGT	1.80E-03	6	3
Collagen type I	3.27E-03	4	5
SWI-SNF	3.58E-03	6	7
PCAF-SWI/SNF	6.40E-03	6	7
Ncor2-Nr1i2	7.92E-03	2	3
xanthine dehydrogenase	7.92E-03	2	3
AC1/8	7.92E-03	2	2
ARID1	7.92E-03	2	2
glutaminase	7.92E-03	2	2
Î±2M-TNFÎ±	7.92E-03	2	2
Dgk	8.47E-03	4	5
alpha-glucosidase	1.15E-02	3	3
Dok	1.87E-02	3	3
LDL	2.00E-02	6	7
glucuronosyltransferase	2.00E-02	6	3
HDL	2.12E-02	9	10
ITPR	2.23E-02	2	4
Laminin2	2.23E-02	2	4
ABLIM	2.23E-02	2	3
Collagen type XVIII	2.23E-02	2	3
ethanolaminephosphotransferase	2.23E-02	2	3
methylenetetrahydrofolate dehydrogenase (NAD)	2.23E-02	2	3
ADRA1	2.23E-02	2	2
Collagen type ix	2.23E-02	2	2
DKC1-TEP1-TERT	2.23E-02	2	2
Eif4g	2.23E-02	2	2
FNG	2.23E-02	2	2
SSH	2.23E-02	2	2
transketolase	2.23E-02	2	2
Vdac	2.23E-02	2	2
HDL-cholesterol	2.72E-02	9	10
IntegrinÎ²	2.80E-02	3	4
ETS	2.80E-02	3	3
PTPase	2.97E-02	12	14
AKAP	3.03E-02	4	6
Collagen(s)	3.03E-02	4	5
MHC	3.22E-02	1	1
Mucin	3.85E-02	4	4
ATPase	3.89E-02	12	15
LRP	3.93E-02	3	5
eIF4A-eIF4E-eIF4Î³	3.93E-02	3	3
Laminin1	4.21E-02	2	5
Rnr	4.21E-02	2	5
Notch	4.21E-02	2	4
CTNNÎ²-CDHE/N	4.21E-02	2	3
methenyltetrahydrofolate cyclohydrolase	4.21E-02	2	3
methylenetetrahydrofolate dehydrogenase (NADP)	4.21E-02	2	3
1-acylglycerophosphocholine O-acyltransferase	4.21E-02	2	2
IFN alpha/beta	4.21E-02	2	2
Karyopherin beta	4.21E-02	2	2
Ltbp	4.21E-02	2	2
Ptger	4.21E-02	2	2
SMC	4.21E-02	2	2
SMC1-SA2	4.21E-02	2	2
Troponin t	4.21E-02	2	2
Vps/Vta1 Complex	4.21E-02	2	2
peptidylprolyl isomerase	4.83E-02	5	6

**Supplemental Table 6C.** Ingenuity Variant Analysis: top enriched processes based on 2699 selected variants.

Name	p-value	#Genes	#Variants
cell biology	1.41E-57	1339	1472
physiology	3.99E-27	786	851
mutation	6.91E-22	17	21
olfactory response	2.64E-21	19	20
cellular metabolism	7.36E-21	608	700
cell signaling	6.15E-19	618	702
metabolism of drug	6.04E-17	172	205
olfaction	9.12E-16	50	55
olfactory system function	9.12E-16	50	55
metabolism of lipid	3.75E-15	346	399
regulation of immune response	5.57E-15	510	580
disorder of genitourinary system	6.32E-15	1259	1422
metabolic process of reactive oxygen species	4.54E-13	118	142
metabolism of reactive oxygen species	4.76E-13	118	142
immune stimulation, inflammation	1.28E-12	382	435
synthesis of reactive oxygen species	1.41E-12	118	142
immunology	2.35E-11	772	898
cell death and survival	5.02E-10	661	731
cell death	5.14E-10	615	680
sensory system function	6.50E-10	114	129
quantity of synapse	2.02E-09	26	9
peroxidation	5.05E-09	24	27
peroxidation of lipid	5.37E-09	24	27
production of reactive oxygen species	1.21E-08	94	112
quantity of gap junctions	1.26E-08	27	10
immune response	1.99E-08	402	460
quantity of glutathione in liver	4.79E-08	1	1
signal transduction	5.93E-08	165	189
communication of cells	7.98E-08	197	227
communication	8.46E-08	203	234
innate immune system (antigen non-specific)	9.62E-08	272	314
conversion of lipid	9.86E-08	41	47
growth of bacteria	9.86E-08	13	14
quantity of reactive oxygen species	1.85E-07	39	52
anti-microbial molecule based immunity	2.05E-07	87	108
modification of retinoid	2.39E-07	1	1
necrosis	2.51E-07	494	550
morphology of organ	3.01E-07	339	395
neurobiology	3.23E-07	609	674
modification of lipid	3.33E-07	85	96
abnormal morphology of cells	3.60E-07	214	252
function of blood vessel	4.98E-07	27	39
nitric oxide signaling	5.50E-07	86	102
quantity of intercellular junctions	6.79E-07	28	12
release of monoamines	9.20E-07	27	34
formation of reactive oxygen species	1.03E-06	23	32
concentration of glutathione	1.05E-06	19	22
concentration of lipid	1.59E-06	139	164
nucleic acid metabolism	1.69E-06	256	303
heavy metal transport	1.91E-06	120	135
quantity of cell-cell contacts	2.40E-06	28	12
abnormal morphology of cardiovascular system	2.46E-06	111	132
quantity of metal	2.91E-06	118	133
synthesis of nitric oxide	3.95E-06	66	78
metabolism of nucleic acid component or derivative	4.15E-06	104	125
cell damage	4.34E-06	113	130
metabolic process of aromatic chemical	6.32E-06	19	18
metabolism of aromatic chemical	6.69E-06	19	18
morphology of brain	7.30E-06	86	101
conversion of sterol	8.23E-06	1	1
conversion of cholesterol	8.98E-06	1	1
morphology of nervous system	1.01E-05	143	165
morphology of body region	1.02E-05	285	327
synthesis of nucleotide	1.28E-05	80	93
binding of guanosine 5'-O-(3-thiotriphosphate)	1.38E-05	5	6
secretion of thromboxane	1.41E-05	15	20

morphology of axial skeleton	1.50E-05	41	47
adaptive immune system (antigen specific)	1.62E-05	290	326
modification of 22:6(n-3) fatty acids	1.66E-05	1	1
modification of n-3 fatty acids	1.66E-05	1	1
modification of docosahexaenoic acid	1.81E-05	1	1
abnormal morphology of brain	1.82E-05	80	95
macrophages	1.87E-05	185	213
conversion of 22:6(n-3) fatty acids	1.97E-05	1	1
conversion of docosahexaenoic acid	1.97E-05	1	1
morphology of central nervous system	1.99E-05	93	108
abnormal morphology of shoulder girdle	2.06E-05	11	13
morphology of tissue	2.08E-05	266	305
metabolic process of nucleotide	2.13E-05	88	105
metabolic process of midazolam	2.15E-05	1	1
metabolic process of paclitaxel	2.15E-05	1	1
metabolism of midazolam	2.15E-05	1	1
metabolism of paclitaxel	2.15E-05	1	1
quantity of metal ion	2.33E-05	114	128
synthesis of terpenoid	2.34E-05	55	69
metabolic process of triazolam	2.35E-05	1	1
metabolism of triazolam	2.35E-05	1	1
activation of acetaminophen	2.56E-05	1	1
metabolic process of alprazolam	2.56E-05	1	1
metabolic process of cyclophosphamide	2.56E-05	1	1
metabolic process of tramadol	2.56E-05	1	1
metabolism of alprazolam	2.56E-05	1	1
metabolism of cyclophosphamide	2.56E-05	1	1
metabolism of tramadol	2.56E-05	1	1
modification of imipramine	2.56E-05	1	1
metabolism of nucleotide	2.70E-05	88	105
abnormal morphology of axial skeleton	2.73E-05	39	44
metabolic process of acetaminophen	2.80E-05	1	1
metabolic process of diazepam	2.80E-05	1	1
metabolism of acetaminophen	2.80E-05	1	1

**Supplemental Table 6D.** Ingenuity Variant Analysis: top enriched diseases based on 2699 selected variants.

Name	p-value	#Genes	#Variants
abdominal adenocarcinoma	9.57E-53	1193	1346
abdominal carcinoma	9.57E-53	1193	1346
adenocarcinoma	6.55E-52	1286	1446
epithelial cancer	9.56E-52	1597	1786
melanoma	1.80E-49	1209	1367
melanocytic lesion	1.92E-49	1209	1367
neoplasia of epithelial tissue	2.50E-48	1610	1800
abdominal cancer	4.73E-48	1580	1768
abdominal neoplasm	2.72E-44	1588	1774
digestive system cancer	3.60E-43	1352	1516
tumorigenesis of tissue	4.70E-43	1634	1823
female genital tract adenocarcinoma	3.53E-42	769	883
digestive organ tumor	3.97E-40	1357	1523
gastrointestinal adenocarcinoma	1.68E-37	821	918
female genital tract cancer	5.69E-36	842	964
conventional melanoma	3.13E-35	797	905
malignant cutaneous melanoma cancer	3.13E-35	797	905
malignant solid tumor	6.07E-34	1810	2007
gastrointestinal carcinoma	2.32E-33	870	965
tumorigenesis of reproductive tract	2.90E-33	858	978
hepatocellular carcinoma	3.48E-33	801	919
genital tract cancer	1.07E-32	910	1043
urogenital cancer	1.36E-32	992	1136
skin cancer	5.52E-32	814	924
female genital neoplasm	2.22E-31	869	989
pelvic cancer	2.53E-31	972	1113
tumorigenesis of genital organ	1.01E-30	928	1059
colorectal adenocarcinoma	1.48E-30	687	772
liver cancer	7.17E-30	811	931
skin tumor	1.34E-29	834	944
solid tumor	1.50E-29	1837	2034
pelvic tumor	1.87E-29	987	1127
colon adenocarcinoma	1.91E-29	652	736
colon carcinoma	2.28E-29	666	750
skin lesion	2.41E-29	841	950
hepatobiliary system cancer	2.64E-29	814	933
adenocarcinoma in endometrium	6.80E-29	519	602
genital tumor	1.24E-28	939	1070
endometrioid cancer	1.99E-28	519	602
endometrioid carcinoma	1.99E-28	519	602
endometrioid neoplasm	1.99E-28	519	602
disease of reproductive tract	2.06E-28	949	1081
gastrointestinal tract cancer	2.47E-28	953	1068
colorectal carcinoma	2.71E-28	719	803
abnormal growth in endometrium	5.98E-28	560	644
endometrium tumor	5.98E-28	560	644
endometrial cancer	6.18E-28	559	643
neoplasia of endometrium	6.18E-28	559	643
liver tumor	1.70E-27	835	959
liver lesion	2.16E-27	836	960
endometrial carcinoma	2.52E-27	525	608
Gastrointestinal Tract Cancer and Tumors	3.42E-27	960	1076
neoplasia of colon	4.62E-27	700	785
tumorigenesis of large intestine	4.62E-27	700	785
Female Reproductive Disorders	5.56E-27	943	1081
colon cancer	6.37E-27	696	781
colon tumor	8.87E-27	701	787
disease of colon	1.04E-26	704	790
tumorigenesis of intestine	2.34E-26	701	786
breast or colorectal cancer	2.79E-26	971	1096
colorectal cancer	1.00E-25	816	920
malignant neoplasm of large intestine	1.16E-25	816	920
intestinal cancer	1.44E-25	818	923
uterine carcinoma	2.30E-25	537	620
colorectal neoplasia	2.71E-25	821	926
intestinal tumor	3.07E-25	825	931

disease of large intestine	7.30E-25	822	927
uterine cancer	1.17E-23	576	659
Uterine Cancer and Tumors	1.17E-23	576	659
Cancer	8.26E-23	1829	2029
disease of genital organ	8.27E-23	1024	1170
malignancy	8.65E-23	1829	2029
uterine tumor	8.12E-22	592	675
gastroesophageal adenocarcinoma	8.53E-22	301	344
disease of uterus	1.38E-21	594	677
serous neoplasm	1.90E-21	400	474
gastro-esophageal carcinoma	3.26E-21	317	363
disease of gastrointestinal tract	1.82E-20	998	1120
breast or ovarian carcinoma	4.44E-20	498	597
gastric carcinoma	7.31E-20	259	305
gastrointestinal disturbance	1.78E-19	995	1118
gastric adenocarcinoma	2.08E-19	249	292
breast or ovarian cancer	2.70E-19	657	775
serous adenocarcinoma	4.18E-19	366	435
Reproductive System Disease	4.27E-19	1147	1303
ovarian adenocarcinoma	4.87E-18	363	432
disease of intestine	1.54E-17	847	957
epithelial ovarian cancer	1.87E-17	379	452
epithelial ovarian tumor	1.87E-17	379	452
cecum carcinoma	3.92E-17	256	279
cecal cancer	4.22E-17	256	279
cecal neoplasm	4.54E-17	256	279
cecum adenocarcinoma	1.14E-16	254	275
female genital tract serous cancer	2.05E-16	350	413
ovarian cancer	2.14E-16	415	497
disease of skin	4.18E-16	945	1069
Dermatological Disorders	7.60E-16	946	1070
Gastrointestinal Diseases	1.15E-15	1449	1619
Endocrine System Disorders	1.25E-15	962	1125
gastroesophageal cancer	1.97E-15	334	382

**Supplemental Table 6E.** GoMiner: top enriched biological processes based on 2699 selected variants.

<a href="#">HYPERLINKED GO CATEGORY</a>	<a href="#">TOTAL GENES</a>	<a href="#">CHANGED GENES</a>	<a href="#">ENRICHMENT</a>	<a href="#">LOG10(p)</a>	<a href="#">FALSE DISCOVERY RATE</a>
<a href="#">GO:0007155 cell adhesion</a>	834	149	1.468286	-6.273083	0
<a href="#">GO:0022610 biological adhesion</a>	834	149	1.468286	-6.273083	0
<a href="#">GO:0030030 cell projection organization</a>	671	123	1.506513	-5.839069	0
<a href="#">GO:0035023 regulation of Rho protein signal transduction</a>	108	31	2.359002	-5.525564	0
<a href="#">GO:0048666 neuron development</a>	616	112	1.494265	-5.195735	0.002
<a href="#">GO:0000902 cell morphogenesis</a>	605	110	1.494265	-5.112849	0.003333
<a href="#">GO:0051056 regulation of small GTPase mediated signal transduction</a>	347	70	1.657902	-4.932922	0.004286
<a href="#">GO:0009653 anatomical structure morphogenesis</a>	1489	232	1.280512	-4.674465	0.005
<a href="#">GO:0007266 Rho protein signal transduction</a>	147	36	2.012684	-4.587214	0.004444
<a href="#">GO:0031175 neuron projection development</a>	542	98	1.485994	-4.518023	0.005
<a href="#">GO:0032989 cellular component morphogenesis</a>	638	112	1.442739	-4.499056	0.004545
<a href="#">GO:0000904 cell morphogenesis involved in differentiation</a>	532	96	1.48303	-4.402986	0.004167
<a href="#">GO:0007265 Ras protein signal transduction</a>	341	67	1.61477	-4.370129	0.003846
<a href="#">GO:0048858 cell projection morphogenesis</a>	507	92	1.491318	-4.337109	0.003571
<a href="#">GO:0007010 cytoskeleton organization</a>	538	96	1.466491	-4.21052	0.005333
<a href="#">GO:0032990 cell part morphogenesis</a>	511	92	1.479644	-4.206019	0.005
<a href="#">GO:0048812 neuron projection morphogenesis</a>	471	86	1.50061	-4.191101	0.005294
<a href="#">GO:0016043 cellular component organization</a>	3220	455	1.161304	-4.13262	0.006111
<a href="#">GO:0007160 cell-matrix adhesion</a>	120	30	2.054614	-4.102865	0.005789
<a href="#">GO:0006996 organelle organization</a>	1578	239	1.244747	-3.972504	0.0075
<a href="#">GO:0016337 cell-cell adhesion</a>	320	62	1.592326	-3.910029	0.008571
<a href="#">GO:0071840 cellular component organization or biogenesis</a>	3327	466	1.151128	-3.858482	0.008182
<a href="#">GO:0048667 cell morphogenesis involved in neuron differentiation</a>	468	84	1.475108	-3.845485	0.008261
<a href="#">GO:0046578 regulation of Ras protein signal transduction</a>	233	48	1.693073	-3.803742	0.010417
<a href="#">GO:0030182 neuron differentiation</a>	765	126	1.353628	-3.711293	0.0128
<a href="#">GO:0030029 actin filament-based process</a>	330	62	1.544074	-3.527346	0.019231
<a href="#">GO:0022008 neurogenesis</a>	864	138	1.31267	-3.411651	0.023333
<a href="#">GO:0048041 focal adhesion assembly</a>	25	10	3.287383	-3.397163	0.023571
<a href="#">GO:0007264 small GTPase mediated signal transduction</a>	574	97	1.388833	-3.365902	0.027241
<a href="#">GO:0007156 homophilic cell adhesion</a>	138	31	1.846175	-3.321119	0.030333
<a href="#">GO:0031589 cell-substrate adhesion</a>	151	33	1.796087	-3.268478	0.031935
<a href="#">GO:0007409 axonogenesis</a>	440	77	1.43823	-3.233745	0.03125
<a href="#">GO:0048699 generation of neurons</a>	819	130	1.304517	-3.134996	0.038788
<a href="#">GO:0035024 negative regulation of Rho protein signal transduction</a>	8	5	5.136536	-2.966448	0.071176
<a href="#">GO:0048468 cell development</a>	1025	156	1.250809	-2.833322	0.084286
<a href="#">GO:0050954 sensory perception of mechanical stimulus</a>	105	24	1.878505	-2.812329	0.085833
<a href="#">GO:0001958 endochondral ossification</a>	12	6	4.109229	-2.810659	0.087368
<a href="#">GO:0006911 phagocytosis engulfment</a>	12	6	4.109229	-2.810659	0.087368
<a href="#">GO:0007044 cell-substrate junction assembly</a>	39	12	2.528756	-2.772719	0.094615
<a href="#">GO:0030036 actin cytoskeleton organization</a>	292	53	1.491706	-2.754501	0.09375
<a href="#">GO:0051128 regulation of cellular component organization</a>	717	113	1.295238	-2.688734	0.098293
<a href="#">GO:0007399 nervous system development</a>	1367	200	1.202408	-2.631206	0.116905
<a href="#">GO:0071842 cellular component organization at cellular level</a>	2167	304	1.152935	-2.59805	0.119535
<a href="#">GO:0001782 B cell homeostasis</a>	13	6	3.793134	-2.588741	0.125455
<a href="#">GO:0032469 endoplasmic reticulum calcium ion homeostasis</a>	6	4	5.478972	-2.572962	0.142609
<a href="#">GO:0034983 peptidyl-lysine deacetylation</a>	6	4	5.478972	-2.572962	0.142609
<a href="#">GO:0040011 locomotion</a>	951	144	1.244435	-2.568705	0.140426
<a href="#">GO:0043068 positive regulation of programmed cell death</a>	494	81	1.347561	-2.532768	0.14125
<a href="#">GO:0071841 cellular component organization or biogenesis at cellular level</a>	2259	315	1.146	-2.52051	0.140612
<a href="#">GO:0033043 regulation of organelle organization</a>	279	50	1.472842	-2.515081	0.139
<a href="#">GO:0007411 axon guidance</a>	328	57	1.428208	-2.478058	0.145098
<a href="#">GO:0043065 positive regulation of apoptosis</a>	491	80	1.339056	-2.434408	0.157692
<a href="#">GO:0010942 positive regulation of cell death</a>	499	81	1.334058	-2.414044	0.157736
<a href="#">GO:0007229 integrin-mediated signaling pathway</a>	59	15	2.089438	-2.411219	0.156481
<a href="#">GO:0016311 dephosphorylation</a>	176	34	1.587657	-2.392978	0.166545
<a href="#">GO:0051271 negative regulation of cellular component movement</a>	77	18	1.921198	-2.356638	0.175357
<a href="#">GO:0002576 platelet degranulation</a>	83	19	1.881334	-2.350369	0.174561
<a href="#">GO:0034394 protein localization at cell surface</a>	15	6	3.287383	-2.217302	0.241724
<a href="#">GO:0030334 regulation of cell migration</a>	234	42	1.475108	-2.215896	0.237797
<a href="#">GO:0051493 regulation of cytoskeleton organization</a>	156	30	1.580473	-2.148994	0.266
<a href="#">GO:0007605 sensory perception of sound</a>	99	21	1.743309	-2.133837	0.274426
<a href="#">GO:0035601 protein deacylation</a>	30	9	2.465537	-2.123399	0.276129
<a href="#">GO:0006917 induction of apoptosis</a>	362	60	1.362175	-2.11941	0.276825
<a href="#">GO:2000145 regulation of cell motility</a>	237	42	1.456436	-2.118806	0.272969
<a href="#">GO:0012501 programmed cell death</a>	1274	183	1.180516	-2.117538	0.269846
<a href="#">GO:0030155 regulation of cell adhesion</a>	157	30	1.570406	-2.109074	0.269848

<a href="#">GO:0006915 apoptosis</a>	1268	182	1.179621	-2.095584	0.267015
<a href="#">GO:0043583 ear development</a>	106	22	1.705718	-2.095003	0.263235
<a href="#">GO:0012502 induction of programmed cell death</a>	363	60	1.358423	-2.094114	0.25942
<a href="#">GO:0051270 regulation of cellular component movement</a>	259	45	1.427917	-2.073983	0.260143
<a href="#">GO:0046580 negative regulation of Ras protein signal transduction</a>	16	6	3.081922	-2.059696	0.268169
<a href="#">GO:0060350 endochondral bone morphogenesis</a>	21	7	2.739486	-2.015277	0.289861
<a href="#">GO:0050974 detection of mechanical stimulus involved in sensory perception</a>	12	5	3.424357	-1.998246	0.31589
<a href="#">GO:0042471 ear morphogenesis</a>	71	16	1.852047	-1.996106	0.313649
<a href="#">GO:0001867 complement activation lectin pathway</a>	8	4	4.109229	-1.991665	0.333816
<a href="#">GO:0060119 inner ear receptor cell development</a>	8	4	4.109229	-1.991665	0.333816
<a href="#">GO:0032956 regulation of actin cytoskeleton organization</a>	102	21	1.692035	-1.984205	0.331039
<a href="#">GO:0009966 regulation of signal transduction</a>	898	132	1.208058	-1.967463	0.332051
<a href="#">GO:0051179 localization</a>	3523	468	1.091751	-1.964963	0.328354
<a href="#">GO:0007015 actin filament organization</a>	148	28	1.554843	-1.947226	0.333125
<a href="#">GO:0030336 negative regulation of cell migration</a>	72	16	1.826324	-1.937514	0.328171
<a href="#">GO:2000146 negative regulation of cell motility</a>	72	16	1.826324	-1.937514	0.328171
<a href="#">GO:0034329 cell junction assembly</a>	142	27	1.562665	-1.92476	0.333735
<a href="#">GO:0043242 negative regulation of protein complex disassembly</a>	43	11	2.102396	-1.922758	0.331786
<a href="#">GO:0016477 cell migration</a>	566	87	1.263261	-1.918336	0.330118
<a href="#">GO:0051058 negative regulation of small GTPase mediated signal transduction</a>	17	6	2.900632	-1.917011	0.333837
<a href="#">GO:0071844 cellular component assembly at cellular level</a>	797	118	1.216785	-1.903269	0.337126
<a href="#">GO:0043067 regulation of programmed cell death</a>	978	142	1.193273	-1.900515	0.334432
<a href="#">GO:0042981 regulation of apoptosis</a>	971	141	1.193412	-1.892295	0.341236
<a href="#">GO:0023051 regulation of signaling process</a>	903	132	1.201369	-1.891973	0.337444
<a href="#">GO:0050905 neuromuscular process</a>	55	13	1.942545	-1.884603	0.336703
<a href="#">GO:0001764 neuron migration</a>	61	14	1.886203	-1.878114	0.337174
<a href="#">GO:0006470 protein dephosphorylation</a>	137	26	1.559707	-1.862974	0.337849
<a href="#">GO:0035303 regulation of dephosphorylation</a>	33	9	2.241398	-1.838056	0.346277
<a href="#">GO:0044087 regulation of cellular component biogenesis</a>	171	31	1.489896	-1.836821	0.342632
<a href="#">GO:0051647 nucleus localization</a>	13	5	3.160945	-1.832546	0.353646
<a href="#">GO:0031061 negative regulation of histone methylation</a>	5	3	4.931075	-1.827736	0.402376
<a href="#">GO:0032486 Rap protein signal transduction</a>	5	3	4.931075	-1.827736	0.402376
<a href="#">GO:0051016 barbed-end actin filament capping</a>	5	3	4.931075	-1.827736	0.402376
<a href="#">GO:0051552 flavone metabolic process</a>	5	3	4.931075	-1.827736	0.402376
<a href="#">GO:0060117 auditory receptor cell development</a>	5	3	4.931075	-1.827736	0.402376
<a href="#">GO:0035148 tube formation</a>	62	14	1.855781	-1.816986	0.406863
<a href="#">GO:0022607 cellular component assembly</a>	1068	153	1.177363	-1.811687	0.403981
<a href="#">GO:0015931 nucleobase nucleoside nucleotide and nucleic acid transport</a>	132	25	1.556526	-1.800989	0.408077
<a href="#">GO:0032970 regulation of actin filament-based process</a>	106	21	1.628185	-1.799379	0.404857
<a href="#">GO:0010941 regulation of cell death</a>	986	142	1.183591	-1.787913	0.407264
<a href="#">GO:0042596 fear response</a>	18	6	2.739486	-1.787144	0.411589
<a href="#">GO:0016575 histone deacetylation</a>	23	7	2.50127	-1.785609	0.413796
<a href="#">GO:0010922 positive regulation of phosphatase activity</a>	9	4	3.652648	-1.779918	0.421681
<a href="#">GO:0010953 regulation of protein maturation by peptide bond cleavage</a>	9	4	3.652648	-1.779918	0.421681
<a href="#">GO:0031060 regulation of histone methylation</a>	9	4	3.652648	-1.779918	0.421681
<a href="#">GO:0070373 negative regulation of ERK1 and ERK2 cascade</a>	9	4	3.652648	-1.779918	0.421681
<a href="#">GO:0070613 regulation of protein processing</a>	9	4	3.652648	-1.779918	0.421681
<a href="#">GO:0048870 cell motility</a>	596	90	1.241042	-1.77462	0.416
<a href="#">GO:0051674 localization of cell</a>	596	90	1.241042	-1.77462	0.416
<a href="#">GO:0048562 embryonic organ morphogenesis</a>	146	27	1.519852	-1.77256	0.414138
<a href="#">GO:0042472 inner ear morphogenesis</a>	57	13	1.874385	-1.756944	0.416923
<a href="#">GO:0045995 regulation of embryonic development</a>	34	9	2.175474	-1.752768	0.416949
<a href="#">GO:0006476 protein deacetylation</a>	29	8	2.267161	-1.713196	0.432353
<a href="#">GO:0001662 behavioral fear response</a>	14	5	2.935164	-1.685636	0.446423
<a href="#">GO:0002920 regulation of humoral immune response</a>	14	5	2.935164	-1.685636	0.446423
<a href="#">GO:0032856 activation of Ras GTPase activity</a>	14	5	2.935164	-1.685636	0.446423
<a href="#">GO:0051043 regulation of membrane protein ectodomain proteolysis</a>	14	5	2.935164	-1.685636	0.446423
<a href="#">GO:0007275 multicellular organismal development</a>	3286	434	1.085457	-1.684157	0.443226
<a href="#">GO:0030835 negative regulation of actin filament depolymerization</a>	24	7	2.39705	-1.682793	0.44664
<a href="#">GO:0019318 hexose metabolic process</a>	231	39	1.387532	-1.681283	0.443254
<a href="#">GO:0051276 chromosome organization</a>	594	89	1.231385	-1.680683	0.439764
<a href="#">GO:0042059 negative regulation of epidermal growth factor receptor signaling pathway</a>	35	9	2.113318	-1.671826	0.438295
<a href="#">GO:0060349 bone morphogenesis</a>	35	9	2.113318	-1.671826	0.438295
<a href="#">GO:0008219 cell death</a>	1387	193	1.143592	-1.670894	0.435769
<a href="#">GO:0070534 protein K63-linked ubiquitination</a>	19	6	2.595303	-1.668409	0.439389
<a href="#">GO:0051651 maintenance of location in cell</a>	71	15	1.736294	-1.655364	0.440606
<a href="#">GO:0009887 organ morphogenesis</a>	596	89	1.227253	-1.646859	0.442932
<a href="#">GO:0033044 regulation of chromosome organization</a>	47	11	1.923469	-1.637887	0.446045
<a href="#">GO:0016265 death</a>	1390	193	1.141124	-1.637377	0.444593
<a href="#">GO:0008654 phospholipid biosynthetic process</a>	110	21	1.568978	-1.629989	0.447059



<a href="#">GO:0046486 glycerolipid metabolic process</a>	205	35	1.403151	-1.627833	0.445328
<a href="#">GO:0034330 cell junction organization</a>	157	28	1.465712	-1.623223	0.448261
<a href="#">GO:0031099 regeneration</a>	91	18	1.625629	-1.608092	0.448489
<a href="#">GO:0006707 cholesterol catabolic process</a>	10	4	3.287383	-1.60137	0.451156
<a href="#">GO:0008340 determination of adult lifespan</a>	10	4	3.287383	-1.60137	0.451156
<a href="#">GO:0016127 sterol catabolic process</a>	10	4	3.287383	-1.60137	0.451156
<a href="#">GO:0032862 activation of Rho GTPase activity</a>	10	4	3.287383	-1.60137	0.451156
<a href="#">GO:0033057 multicellular organismal reproductive behavior</a>	10	4	3.287383	-1.60137	0.451156
<a href="#">GO:0034446 substrate adhesion-dependent cell spreading</a>	10	4	3.287383	-1.60137	0.451156
<a href="#">GO:0051044 positive regulation of membrane protein ectodomain proteolysis</a>	10	4	3.287383	-1.60137	0.451156
<a href="#">GO:0055117 regulation of cardiac muscle contraction</a>	10	4	3.287383	-1.60137	0.451156
<a href="#">GO:0006970 response to osmotic stress</a>	36	9	2.054614	-1.59494	0.44953
<a href="#">GO:0008637 apoptotic mitochondrial changes</a>	36	9	2.054614	-1.59494	0.44953
<a href="#">GO:0033135 regulation of peptidyl-serine phosphorylation</a>	25	7	2.301168	-1.586942	0.452252
<a href="#">GO:0048538 thymus development</a>	25	7	2.301168	-1.586942	0.452252
<a href="#">GO:0009790 embryo development</a>	675	99	1.205374	-1.580715	0.452368
<a href="#">GO:0001656 metanephros development</a>	42	10	1.956776	-1.579301	0.451176
<a href="#">GO:0006642 triglyceride mobilization</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0006658 phosphatidylserine metabolic process</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0010955 negative regulation of protein maturation by peptide bond cleavage</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0016090 prenol metabolic process</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0016093 polyprenol metabolic process</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0042159 lipoprotein catabolic process</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0043374 CD8-positive alpha-beta T cell differentiation</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0045071 negative regulation of viral genome replication</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0051569 regulation of histone H3-K4 methylation</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0051894 positive regulation of focal adhesion assembly</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0060561 apoptosis involved in morphogenesis</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0072111 cell proliferation involved in kidney development</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0072215 regulation of metanephros development</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0085029 extracellular matrix assembly</a>	6	3	4.109229	-1.567615	0.473174
<a href="#">GO:0005975 carbohydrate metabolic process</a>	601	89	1.217043	-1.564419	0.47131
<a href="#">GO:0030278 regulation of ossification</a>	92	18	1.607959	-1.564072	0.469112
<a href="#">GO:0007017 microtubule-based process</a>	278	45	1.330326	-1.563339	0.466471
<a href="#">GO:0031102 neuron projection regeneration</a>	20	6	2.465537	-1.55943	0.471871
<a href="#">GO:0002209 behavioral defense response</a>	15	5	2.739486	-1.554336	0.476724
<a href="#">GO:0010259 multicellular organismal aging</a>	15	5	2.739486	-1.554336	0.476724
<a href="#">GO:0015813 L-glutamate transport</a>	15	5	2.739486	-1.554336	0.476724
<a href="#">GO:0006921 cellular component disassembly involved in apoptosis</a>	73	15	1.688724	-1.553998	0.474914
<a href="#">GO:0006820 anion transport</a>	112	21	1.540961	-1.550667	0.473807
<a href="#">GO:0050885 neuromuscular process controlling balance</a>	31	8	2.120892	-1.54384	0.476836
<a href="#">GO:0030335 positive regulation of cell migration</a>	139	25	1.47814	-1.543617	0.471732
<a href="#">GO:2000147 positive regulation of cell motility</a>	139	25	1.47814	-1.543617	0.471732
<a href="#">GO:0006639 acylglycerol metabolic process</a>	86	17	1.624579	-1.543499	0.470222
<a href="#">GO:0044085 cellular component biogenesis</a>	1190	166	1.14644	-1.536516	0.470552
<a href="#">GO:0040012 regulation of locomotion</a>	265	43	1.333561	-1.532341	0.468571
<a href="#">GO:0019722 calcium-mediated signaling</a>	61	13	1.751475	-1.526663	0.468033
<a href="#">GO:0030031 cell projection assembly</a>	106	20	1.550652	-1.52649	0.463297
<a href="#">GO:0043254 regulation of protein complex assembly</a>	106	20	1.550652	-1.52649	0.463297
<a href="#">GO:0006641 triglyceride metabolic process</a>	80	16	1.643692	-1.523902	0.461667
<a href="#">GO:0001843 neural tube closure</a>	37	9	1.999084	-1.521847	0.459415
<a href="#">GO:0034332 adherens junction organization</a>	37	9	1.999084	-1.521847	0.459415
<a href="#">GO:0043244 regulation of protein complex disassembly</a>	55	12	1.793118	-1.517283	0.458942
<a href="#">GO:0010769 regulation of cell morphogenesis involved in differentiation</a>	113	21	1.527324	-1.512282	0.45822
<a href="#">GO:0022604 regulation of cell morphogenesis</a>	113	21	1.527324	-1.512282	0.45822
<a href="#">GO:0001841 neural tube formation</a>	49	11	1.84496	-1.512096	0.457917
<a href="#">GO:0035466 regulation of signaling pathway</a>	1170	163	1.144965	-1.50011	0.459585
<a href="#">GO:0006638 neutral lipid metabolic process</a>	87	17	1.605906	-1.49937	0.458247
<a href="#">GO:0030834 regulation of actin filament depolymerization</a>	26	7	2.212662	-1.497407	0.462103
<a href="#">GO:0022603 regulation of anatomical structure morphogenesis</a>	281	45	1.316123	-1.492553	0.460969
<a href="#">GO:0051129 negative regulation of cellular component organization</a>	210	35	1.369743	-1.488134	0.461269
<a href="#">GO:0048589 developmental growth</a>	182	31	1.399847	-1.487799	0.459091
<a href="#">GO:0051494 negative regulation of cytoskeleton organization</a>	62	13	1.723225	-1.473838	0.462965
<a href="#">GO:0051272 positive regulation of cellular component movement</a>	148	26	1.443783	-1.470804	0.462
<a href="#">GO:0030035 microspike assembly</a>	32	8	2.054614	-1.466215	0.464179
<a href="#">GO:0048568 embryonic organ development</a>	225	37	1.35148	-1.465253	0.46198
<a href="#">GO:0001838 embryonic epithelial tube formation</a>	56	12	1.761098	-1.461605	0.462118
<a href="#">GO:0050848 regulation of calcium-mediated signaling</a>	21	6	2.348131	-1.459069	0.468529
<a href="#">GO:0051261 protein depolymerization</a>	50	11	1.808061	-1.452896	0.47039
<a href="#">GO:0006464 protein modification process</a>	1846	249	1.108557	-1.452808	0.468301

<a href="#">GO:0060606 tube closure</a>	38	9	1.946477	-1.452309	0.468068
<a href="#">GO:0001783 B cell apoptosis</a>	11	4	2.98853	-1.448146	0.485571
<a href="#">GO:0007020 microtubule nucleation</a>	11	4	2.98853	-1.448146	0.485571
<a href="#">GO:0032026 response to magnesium ion</a>	11	4	2.98853	-1.448146	0.485571
<a href="#">GO:0018209 peptidyl-serine modification</a>	69	14	1.667513	-1.439784	0.484387
<a href="#">GO:0030832 regulation of actin filament length</a>	69	14	1.667513	-1.439784	0.484387
<a href="#">GO:0015800 acidic amino acid transport</a>	16	5	2.568268	-1.436214	0.486267
<a href="#">GO:0019098 reproductive behavior</a>	16	5	2.568268	-1.436214	0.486267
<a href="#">GO:0032006 regulation of TOR signaling cascade</a>	16	5	2.568268	-1.436214	0.486267
<a href="#">GO:0050982 detection of mechanical stimulus</a>	16	5	2.568268	-1.436214	0.486267
<a href="#">GO:0051568 histone H3-K4 methylation</a>	16	5	2.568268	-1.436214	0.486267
<a href="#">GO:0009792 embryo development ending in birth or egg hatching</a>	393	60	1.254726	-1.434496	0.484587
<a href="#">GO:0010639 negative regulation of organelle organization</a>	122	22	1.482017	-1.428869	0.484521
<a href="#">GO:0006913 nucleocytoplasmic transport</a>	248	40	1.325558	-1.42494	0.482864
<a href="#">GO:0006814 sodium ion transport</a>	129	23	1.465306	-1.4215	0.481126
<a href="#">GO:0048705 skeletal system morphogenesis</a>	129	23	1.465306	-1.4215	0.481126
<a href="#">GO:0001836 release of cytochrome c from mitochondria</a>	27	7	2.130711	-1.413622	0.477401
<a href="#">GO:0006509 membrane protein ectodomain proteolysis</a>	27	7	2.130711	-1.413622	0.477401
<a href="#">GO:0007632 visual behavior</a>	27	7	2.130711	-1.413622	0.477401
<a href="#">GO:0030042 actin filament depolymerization</a>	27	7	2.130711	-1.413622	0.477401
<a href="#">GO:0050773 regulation of dendrite development</a>	27	7	2.130711	-1.413622	0.477401
<a href="#">GO:0072175 epithelial tube formation</a>	57	12	1.730202	-1.407922	0.479737
<a href="#">GO:0051169 nuclear transport</a>	249	40	1.320234	-1.401093	0.480917
<a href="#">GO:0042058 regulation of epidermal growth factor receptor signaling pathway</a>	51	11	1.772609	-1.396005	0.481435
<a href="#">GO:0006325 chromatin organization</a>	447	67	1.231849	-1.394485	0.47974
<a href="#">GO:0050890 cognition</a>	123	22	1.469968	-1.394179	0.478448
<a href="#">GO:0016331 morphogenesis of embryonic epithelium</a>	83	16	1.584281	-1.391119	0.480043
<a href="#">GO:0040013 negative regulation of locomotion</a>	83	16	1.584281	-1.391119	0.480043
<a href="#">GO:0032507 maintenance of protein location in cell</a>	64	13	1.669374	-1.373383	0.492723
<a href="#">GO:0044262 cellular carbohydrate metabolic process</a>	441	66	1.229973	-1.369435	0.491568
<a href="#">GO:0031532 actin cytoskeleton reorganization</a>	22	6	2.241398	-1.366374	0.493875
<a href="#">GO:0034968 histone lysine methylation</a>	22	6	2.241398	-1.366374	0.493875
<a href="#">GO:0050803 regulation of synapse structure and activity</a>	22	6	2.241398	-1.366374	0.493875
<a href="#">GO:0051693 actin filament capping</a>	22	6	2.241398	-1.366374	0.493875
<a href="#">GO:0008653 lipopolysaccharide metabolic process</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0009103 lipopolysaccharide biosynthetic process</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0016556 mRNA modification</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0032229 negative regulation of synaptic transmission GABAergic</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0033137 negative regulation of peptidyl-serine phosphorylation</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0040023 establishment of nucleus localization</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0042711 maternal behavior</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0048525 negative regulation of viral reproduction</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0050858 negative regulation of antigen receptor-mediated signaling pathway</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0050860 negative regulation of T cell receptor signaling pathway</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0051095 regulation of helicase activity</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0051323 metaphase</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0051818 disruption of cells of other organism involved in symbiotic interaction</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0051883 killing of cells in other organism involved in symbiotic interaction</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0060122 inner ear receptor stereocilium organization</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0060746 parental behavior</a>	7	3	3.522196	-1.365186	0.500469
<a href="#">GO:0043412 macromolecule modification</a>	1928	258	1.099773	-1.355666	0.502568
<a href="#">GO:0009146 purine nucleoside triphosphate catabolic process</a>	152	26	1.405789	-1.346544	0.504147
<a href="#">GO:0043149 stress fiber assembly</a>	28	7	2.054614	-1.335095	0.51112
<a href="#">GO:0006066 alcohol metabolic process</a>	503	74	1.209077	-1.332609	0.510308
<a href="#">GO:0001933 negative regulation of protein phosphorylation</a>	46	10	1.786621	-1.329803	0.511533
<a href="#">GO:0006541 glutamine metabolic process</a>	17	5	2.417194	-1.329361	0.517689
<a href="#">GO:0010623 developmental programmed cell death</a>	17	5	2.417194	-1.329361	0.517689
<a href="#">GO:0051220 cytoplasmic sequestering of protein</a>	17	5	2.417194	-1.329361	0.517689
<a href="#">GO:0016568 chromatin modification</a>	347	53	1.255269	-1.328413	0.515925
<a href="#">GO:0010563 negative regulation of phosphorus metabolic process</a>	65	13	1.643692	-1.325625	0.514045
<a href="#">GO:0045936 negative regulation of phosphate metabolic process</a>	65	13	1.643692	-1.325625	0.514045
<a href="#">GO:0001952 regulation of cell-matrix adhesion</a>	34	8	1.933755	-1.323407	0.51653
<a href="#">GO:0014020 primary neural tube formation</a>	40	9	1.849153	-1.323043	0.516059
<a href="#">GO:0016044 cellular membrane organization</a>	459	68	1.217549	-1.317058	0.517815
<a href="#">GO:0032204 regulation of telomere maintenance</a>	12	4	2.739486	-1.31489	0.526218
<a href="#">GO:0050856 regulation of T cell receptor signaling pathway</a>	12	4	2.739486	-1.31489	0.526218
<a href="#">GO:0051893 regulation of focal adhesion assembly</a>	12	4	2.739486	-1.31489	0.526218
<a href="#">GO:0090109 regulation of cell-substrate junction assembly</a>	12	4	2.739486	-1.31489	0.526218

**Supplemental Table 6F.** GoMiner: top enriched molecular functions based on 2699 selected variants.

<a href="#">HYPERLINKED GO CATEGORY</a>	<a href="#">TOTAL GENES</a>	<a href="#">CHANGED GENES</a>	<a href="#">ENRICHMENT</a>	<a href="#">LOG10(p)</a>	<a href="#">FALSE DISCOVERY RATE</a>
<a href="#">GO:0005085 guanyl-nucleotide exchange factor activity</a>	156	40	2.085897	-5.436726	0
<a href="#">GO:0005089 Rho guanyl-nucleotide exchange factor activity</a>	72	22	2.485694	-4.4927	0.01
<a href="#">GO:0008092 cytoskeletal protein binding</a>	527	96	1.481898	-4.387942	0.006667
<a href="#">GO:0005088 Ras guanyl-nucleotide exchange factor activity</a>	89	25	2.285112	-4.329437	0.005
<a href="#">GO:0030695 GTPase regulator activity</a>	446	83	1.513913	-4.201803	0.004
<a href="#">GO:0060589 nucleoside-triphosphatase regulator activity</a>	455	83	1.483967	-3.895688	0.003333
<a href="#">GO:0051020 GTPase binding</a>	113	28	2.015752	-3.722937	0.005714
<a href="#">GO:0019899 enzyme binding</a>	675	113	1.361859	-3.483866	0.01375
<a href="#">GO:0005083 small GTPase regulator activity</a>	292	56	1.560137	-3.360733	0.018889
<a href="#">GO:0005509 calcium ion binding</a>	638	106	1.351583	-3.183721	0.031
<a href="#">GO:0046872 metal ion binding</a>	3730	514	1.121016	-3.082103	0.033636
<a href="#">GO:0019902 phosphatase binding</a>	56	16	2.324286	-3.068972	0.030833
<a href="#">GO:0043169 cation binding</a>	3773	519	1.119021	-3.0417	0.03
<a href="#">GO:0043167 ion binding</a>	3783	520	1.118213	-3.018945	0.033571
<a href="#">GO:0003779 actin binding</a>	328	60	1.48811	-3.013994	0.032
<a href="#">GO:0005201 extracellular matrix structural constituent</a>	82	20	1.984146	-2.73819	0.073125
<a href="#">GO:0019903 protein phosphatase binding</a>	50	14	2.2778	-2.661879	0.070588
<a href="#">GO:0030169 low-density lipoprotein binding</a>	21	8	3.099048	-2.624301	0.077778
<a href="#">GO:0030020 extracellular matrix structural constituent conferring tensile strength</a>	6	4	5.423333	-2.556128	0.099474
<a href="#">GO:0005080 protein kinase C binding</a>	31	10	2.624194	-2.528755	0.0975
<a href="#">GO:0005539 glycosaminoglycan binding</a>	148	30	1.648986	-2.425479	0.109524
<a href="#">GO:0001871 pattern binding</a>	162	32	1.606914	-2.368875	0.125217
<a href="#">GO:0030247 polysaccharide binding</a>	162	32	1.606914	-2.368875	0.125217
<a href="#">GO:0031267 small GTPase binding</a>	100	22	1.7897	-2.354092	0.122083
<a href="#">GO:0071813 lipoprotein particle binding</a>	29	9	2.524655	-2.20017	0.163462
<a href="#">GO:0071814 protein-lipid complex binding</a>	29	9	2.524655	-2.20017	0.163462
<a href="#">GO:0005515 protein binding</a>	6787	884	1.059576	-2.184198	0.162593
<a href="#">GO:0005041 low-density lipoprotein receptor activity</a>	11	5	3.697727	-2.167713	0.166071
<a href="#">GO:0003995 acyl-CoA dehydrogenase activity</a>	16	6	3.050625	-2.03804	0.196207
<a href="#">GO:0005178 integrin binding</a>	64	15	1.906641	-2.022183	0.193
<a href="#">GO:0051721 protein phosphatase 2A binding</a>	12	5	3.389583	-1.979443	0.217097
<a href="#">GO:0017016 Ras GTPase binding</a>	90	19	1.717389	-1.913764	0.24375
<a href="#">GO:0005543 phospholipid binding</a>	194	35	1.467655	-1.907042	0.237879
<a href="#">GO:0030246 carbohydrate binding</a>	368	60	1.326359	-1.88188	0.247647
<a href="#">GO:0008201 heparin binding</a>	110	22	1.627	-1.858237	0.251714
<a href="#">GO:0008307 structural constituent of muscle</a>	44	11	2.03375	-1.815674	0.259722
<a href="#">GO:0004614 phosphoglucomutase activity</a>	5	3	4.881	-1.815298	0.302632
<a href="#">GO:0004957 prostaglandin E receptor activity</a>	5	3	4.881	-1.815298	0.302632
<a href="#">GO:0004721 phosphoprotein phosphatase activity</a>	165	30	1.479091	-1.754309	0.358974
<a href="#">GO:0001948 glycoprotein binding</a>	45	11	1.988556	-1.743565	0.355
<a href="#">GO:0015248 sterol transporter activity</a>	14	5	2.905357	-1.667787	0.378095
<a href="#">GO:0030228 lipoprotein receptor activity</a>	14	5	2.905357	-1.667787	0.378095
<a href="#">GO:0019901 protein kinase binding</a>	188	33	1.427952	-1.665333	0.369302
<a href="#">GO:0001540 beta-amyloid binding</a>	19	6	2.568947	-1.648217	0.377955
<a href="#">GO:0017048 Rho GTPase binding</a>	30	8	2.169333	-1.601805	0.385111
<a href="#">GO:0000287 magnesium ion binding</a>	163	29	1.447331	-1.596257	0.378261
<a href="#">GO:0016634 oxidoreductase activity acting on the CH-CH group of donors oxygen as acceptor</a>	10	4	3.254	-1.586402	0.39383
<a href="#">GO:0000166 nucleotide binding</a>	2244	304	1.102068	-1.558157	0.397083
<a href="#">GO:0004558 alpha-glucosidase activity</a>	6	3	4.0675	-1.555614	0.403019
<a href="#">GO:0004559 alpha-mannosidase activity</a>	6	3	4.0675	-1.555614	0.403019
<a href="#">GO:0004972 N-methyl-D-aspartate selective glutamate receptor activity</a>	6	3	4.0675	-1.555614	0.403019
<a href="#">GO:0005521 lamin binding</a>	6	3	4.0675	-1.555614	0.403019
<a href="#">GO:0015288 porin activity</a>	6	3	4.0675	-1.555614	0.403019
<a href="#">GO:0019838 growth factor binding</a>	111	21	1.539054	-1.546367	0.400185
<a href="#">GO:0019904 protein domain specific binding</a>	420	65	1.258988	-1.538866	0.405636
<a href="#">GO:0004143 diacylglycerol kinase activity</a>	15	5	2.711667	-1.536958	0.403684
<a href="#">GO:0015923 mannosidase activity</a>	15	5	2.711667	-1.536958	0.403684
<a href="#">GO:0017124 SH3 domain binding</a>	107	20	1.520561	-1.446422	0.437241
<a href="#">GO:0030234 enzyme regulator activity</a>	889	127	1.162143	-1.435156	0.439661
<a href="#">GO:0015450 P-P-bond-hydrolysis-driven protein transmembrane transporter activity</a>	11	4	2.958182	-1.43363	0.437903
<a href="#">GO:0016868 intramolecular transferase activity phosphotransferases</a>	11	4	2.958182	-1.43363	0.437903
<a href="#">GO:0042805 actinin binding</a>	11	4	2.958182	-1.43363	0.437903
<a href="#">GO:0030165 PDZ domain binding</a>	56	12	1.743214	-1.431678	0.433492

<a href="#">GO:0005217 intracellular ligand-gated ion channel activity</a>	16	5	2.542188	-1.419301	0.447031
<a href="#">GO:0015296 anion:cation symporter activity</a>	33	8	1.972121	-1.370065	0.464
<a href="#">GO:0042826 histone deacetylase binding</a>	45	10	1.807778	-1.361993	0.462727
<a href="#">GO:0008270 zinc ion binding</a>	1954	264	1.099099	-1.361972	0.45597
<a href="#">GO:0035091 phosphoinositide binding</a>	116	21	1.472716	-1.361096	0.451765
<a href="#">GO:0031432 titin binding</a>	7	3	3.486429	-1.353614	0.484286
<a href="#">GO:0045294 alpha-catenin binding</a>	7	3	3.486429	-1.353614	0.484286
<a href="#">GO:0008093 cytoskeletal adaptor activity</a>	22	6	2.218636	-1.347608	0.485493
<a href="#">GO:0005319 lipid transporter activity</a>	64	13	1.652422	-1.342847	0.481389
<a href="#">GO:0004553 hydrolase activity hydrolyzing O-glycosyl compounds</a>	90	17	1.536611	-1.337908	0.476301
<a href="#">GO:0042162 telomeric DNA binding</a>	17	5	2.392647	-1.312908	0.49027
<a href="#">GO:0016763 transferase activity transferring pentosyl groups</a>	46	10	1.768478	-1.304143	0.4872

Supplement Figure Legend. Conformational mobility of the activation loop is depicted using two representative group PDB structures of wild-type VEGFR2. Group1 representative wild-type structure (PDB ID: 3VO3) is shown on the left and the group 2 representative wild-type structure (PDB ID: 2OH4) is displayed on the right. The protein structure is set to solid ribbon and colored in N(blue)-to-C(red) terminal coloring style. The active site residue locations are colored in green and the activation loop segment in cyan. The inhibitor molecule is shown in ball and stick style and highlighted with a transparent closed surface. The amino acid residue A1065 is shown in CPK (solid spheres) style. The two structures were aligned and then separated for clarity

**Supplemental Figure 1. Inhibitor bound VEGFR2 structures PDB IDs 3VO3 (left) and 2OH4 (right).** The protein structure display is set to solid ribbon and colored using N(blue)-to-C(red) terminal coloring style. The active site residue locations are colored in green and the activation loop segment is colored in cyan. The inhibitor molecule is shown in ball and stick style and highlighted with a transparent closed surface. The modified amino acid residue, A1065, is shown in CPK style. The two structures were aligned and then separated for clarity.

