Locus 137 (rs2280774)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,900,372-32,057,972
block size	157,601 bp
variant count	19 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.353 [-2.371 – 1.344]	gene(s) hit or close-by	C2, C2-AS1, C4A, C4B, C4B-AS1, CFB, CYP21A2, DXO, MIR1236, NELFE, RNA5SP206, SKIV2L, STK19, TNXB, XXbac-BPG116M5.17
phastCons	μ = 0.032 [0 – 0.43]	eQTL gene(s)	AGPAT1, AIF1, ATF6B, BAG6, CCHCR1, CYP21A1P, CYP21A2, DXO, EHMT2, HLA-B, HLA-C, HLA-DQA2, HLA-DQB1, HLA-DQB1-AS1, HLA-DRB5, HSPA1B, HSPA1L, LST1, MSH5, MSH5-SAPCD1, NELFE, NOTCH4, PRRC2A, PSORS1C3, VARS2, XXbac-BPG248L24.12, XXbac-BPG300A18.13
GERP++	μ = -0.651 [-7.07 – 3.29]	potentially regulated gene(s)	BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, BTNL2, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, PRRT1, PRRT1, PRRT1, PRRT1, PRRT1, PRRT1, PRRT1
CADD score	μ = 4.496 [0.002 – 17.42]	disease gene(s)	C2, VARS2, CFB, SKIV2L, CYP21A2, C4A, C4B, BTNL2, HLA-DQB1, HLA-B, TNXB

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Ulcerative colitis	$<5.00\times10^{-14}$	GWAS Catalog	24837172	1
Complement C3 and C4 levels	$<1.00\times10^{-22}$	GWAS Catalog	23028341	1

Disease gene annotation

gene	trait	source DB	source entry/link
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602
CYP21A2	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 21-HYDROXYLASE [...]	OMIM	MIM:201910
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4B	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4B	COMPLEMENT COMPONENT 4B DEFICIENCY	OMIM	MIM:614379
C4B	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387
	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387

	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
BTNL2	SARCOIDOSIS, SUSCEPTIBILITY TO, 2	OMIM	MIM:612387	OMIM
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
HLA-DQB1	Bullous pemphigoid	OrphaNet	OrphaNet:703	orphanet
HLA-DQB1	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073	orphanet
HLA-DQB1	Narcolepsy without cataplexy	OrphaNet	OrphaNet:83465	orphanet
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578	orphanet
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, salt wasting form	OrphaNet	OrphaNet:315306	orphanet
CYP21A2	Nonclassic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	OrphaNet	OrphaNet:95698	orphanet
CYP21A2	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency, simple virilizing form	OrphaNet	OrphaNet:315311	orphanet
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Behcet disease	OrphaNet	OrphaNet:117	orphanet
C4B	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
TNXB	Ehlers-Danlos syndrome due to tenascin-X deficiency	OrphaNet	OrphaNet:230839	orphanet
TNXB	EHLERS-DANLOS SYNDROME, HYPERMOBILITY TYPE	OrphaNet	OrphaNet:285	orphanet
BTNL2	Sarcoidosis	OrphaNet	OrphaNet:797	orphanet
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	OMIM

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CYP21A2	splice acceptor variant	ENST00000464325	?	?					1
CYP21A2	missense variant	ENST00000478281	?	ENSP00000419572	K/R	aAg/aGg	?	?	1
CYP21A2	stop retained variant	ENST00000469053	?	ENSP00000418104	*	taA/taG	?	?	1
CYP21A2	missense variant	ENST00000418967	NM_000500.7	ENSP00000408860	K/R	aAg/aGg	?	?	1
CYP21A2	missense variant	ENST00000471671	?	ENSP00000418561	K/R	aAg/aGg	?	?	1
CYP21A2	missense variant	ENST00000435122	NM_001128590.3	ENSP00000415043	K/R	aAg/aGg	?	?	1
CYP21A2	stop retained variant	ENST00000466779	?	ENSP00000417321	*	taA/taG	?	?	1
TNXB	missense variant	ENST00000611016	?	ENSP00000483409	I/N	aTc/aAc	?	?	1
TNXB	missense variant	ENST00000451343	NM_032470.3	ENSP00000407685	I/N	aTc/aAc	?	?	1
TNXB	missense variant	ENST00000375244	NM_019105.6	ENSP00000364393	I/N	aTc/aAc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DXO	ENST00000474587	ILMN_1726990	blood	2.01×10-5 (p-value)	MuTHER consortium	6
DXO	ENST00000491327					
DXO	ENST00000473976					
DXO	ENST00000480240					
DXO	ENST00000498357					

DXO	ENST00000477826					
DXO	ENST00000492946					
DXO	ENST00000375349					
DXO	ENST00000337523					
DXO	ENST00000460058					
DXO	ENST00000375356					
DXO	ENST00000487914					
DXO	ENST00000478221					
DXO	ENST00000485557					
PRRC2A	ENST00000376033	ILMN_2408179	blood	1.51×10-6 (p-value)	MuTHER consortium	7
PRRC2A	ENST00000487089					
PRRC2A	ENST00000376007					
PRRC2A	ENST00000487839					
PRRC2A	ENST00000462617					
PRRC2A	ENST00000482441					
PRRC2A	ENST00000492691					
HSPA1B	ENST00000375650	ILMN_1660436	adipocyte	2.53×10-6 (p-value)	MuTHER consortium	3
AIF1	ENST00000337917	ILMN_1792473	blood	3.51×10-10 (p-value)	MuTHER consortium	7
AIF1	ENST00000466820					
AIF1	ENST00000497362					
AIF1	ENST00000376059					
AIF1	ENST00000376049					
MSH5	ENST00000395853	ILMN_1810834	blood	6.83×10-6 (p-value)	MuTHER consortium	7
MSH5	ENST00000375750					
MSH5	ENST00000429846					
MSH5	ENST00000423982					
MSH5	ENST00000484309					
MSH5-SAPCD1	ENST00000498473					
MSH5	ENST00000375703					
MSH5	ENST00000375740					
MSH5-SAPCD1	ENST00000493662					
MSH5	ENST00000375755					
MSH5-SAPCD1	ENST00000491552					
MSH5	ENST00000463144					
LST1	ENST00000339530	ILMN_1718936	blood	6.85×10-5 (p-value)	MuTHER consortium	1
LST1	ENST00000464526					
LST1	ENST00000303757					
LST1	ENST00000419073					
LST1	ENST00000376093					
LST1	ENST00000396112					
LST1	ENST00000490742					
LST1	ENST00000433492					
LST1	ENST00000438075					
LST1	ENST00000464044					
LST1	ENST00000396101					
CYP21A1P	?	ENSG00000204338	muscularis mucosae	9.51×10-12 (p-value)	GTEx Portal V6	18
ATF6B	?	ENSG00000213676	transformed fibroblasts	2.30×10-18 (p-value)	GTEx Portal V6	18
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	5.13×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG300A18.13	?	ENSG00000273333	transformed fibroblasts	2.63×10-6 (p-value)	GTEx Portal V6	5
HLA-C	?	ENSG00000204525	blood	8.43×10-7 (p-value)	GTEx Portal V6	6
HLA-B	?	ENSG00000234745	blood	5.49×10-6 (p-value)	GTEx Portal V6	2

HLA-DQA2	?	ENSG00000237541	blood	2.78×10-7 (p-value)	GTEx Portal V6	4
CYP21A1P	?	ENSG00000204338	blood	9.80×10-10 (p-value)	GTEx Portal V6	15
PRRC2A	?	ENSG00000204469	blood	7.42×10-7 (p-value)	GTEx Portal V6	8
ATF6B	?	ENSG00000213676	breast	1.64×10-10 (p-value)	GTEx Portal V6	18
HLA-B	?	ENSG00000234745	breast	1.39×10-6 (p-value)	GTEx Portal V6	3
BAG6	?	ENSG00000204463	tibial artery	7.76×10-12 (p-value)	GTEx Portal V6	18
XXbac-BPG300A18.13	?	ENSG00000273333	tibial artery	1.30×10-9 (p-value)	GTEx Portal V6	18
HLA-C	?	ENSG00000204525	tibial artery	2.02×10-6 (p-value)	GTEx Portal V6	8
CYP21A1P	?	ENSG00000204338	tibial artery	1.82×10-14 (p-value)	GTEx Portal V6	18
VARS2	ENST00000321897	ILMN_1737585	monocyte	1.34×10-4 (p-value)	Fairfax et al.	7
VARS2	ENST00000625423		b-cell	1.90×10-5 (p-value)	Fairfax et al.	7
VARS2	ENST00000542001					
VARS2	ENST00000541562					
VARS2	ENST00000469358					
VARS2	ENST00000477288					
VARS2	ENST00000473916					
VARS2	ENST00000476162					
ATF6B	?	ENSG00000213676	skeletal muscle	8.95×10-19 (p-value)	GTEx Portal V6	18
HLA-C	?	ENSG00000204525	skeletal muscle	1.45×10-6 (p-value)	GTEx Portal V6	3
CYP21A1P	?	ENSG00000204338	skeletal muscle	1.12×10-7 (p-value)	GTEx Portal V6	12
HLA-C	?	ENSG00000204525	sun exposed skin	5.42×10-7 (p-value)	GTEx Portal V6	8
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	1.03×10-6 (p-value)	GTEx Portal V6	3
EHMT2	?	ENSG00000204371	sun exposed skin	4.15×10-6 (p-value)	GTEx Portal V6	1
CYP21A1P	?	ENSG00000204338	sun exposed skin	7.27×10-7 (p-value)	GTEx Portal V6	14
XXbac-BPG300A18.13	?	ENSG00000273333	sun exposed skin	9.84×10-7 (p-value)	GTEx Portal V6	2
ATF6B	?	ENSG00000213676	left ventricle	2.03×10-13 (p-value)	GTEx Portal V6	18
CYP21A1P	?	ENSG00000204338	left ventricle	3.41×10-7 (p-value)	GTEx Portal V6	11
XXbac-BPG300A18.13	?	ENSG00000273333	aorta	4.87×10-7 (p-value)	GTEx Portal V6	18
XXbac-BPG248L24.12	?	ENSG00000271581	aorta	3.80×10-6 (p-value)	GTEx Portal V6	1
HLA-C	?	ENSG00000204525	aorta	1.02×10-5 (p-value)	GTEx Portal V6	1
CYP21A1P	?	ENSG00000204338	aorta	1.09×10-7 (p-value)	GTEx Portal V6	15
HLA-DQB1	?	ENSG00000179344	subcutaneous adipocytes	1.82×10-6 (p-value)	GTEx Portal V6	4
HLA-DQB1-AS1	?	ENSG00000223534	subcutaneous adipocytes	6.49×10-7 (p-value)	GTEx Portal V6	15
CYP21A1P	?	ENSG00000204338	subcutaneous adipocytes	8.10×10-16 (p-value)	GTEx Portal V6	18
HLA-C	?	ENSG00000204525	subcutaneous adipocytes	7.08×10-7 (p-value)	GTEx Portal V6	4
ATF6B	?	ENSG00000213676	subcutaneous adipocytes	2.38×10-7 (p-value)	GTEx Portal V6	14
PSORS1C3	?	ENSG00000204528	stomach	5.16×10-8 (p-value)	GTEx Portal V6	3
ATF6B	?	ENSG00000213676	stomach	6.65×10-7 (p-value)	GTEx Portal V6	1

HLA-C	?	ENSG00000204525	tibial nerve	3.59×10-7 (p-value)	GTEx Portal V6	8
CYP21A1P	?	ENSG00000204338	tibial nerve	4.93×10-14 (p-value)	GTEx Portal V6	18
ATF6B	?	ENSG00000213676	tibial nerve	2.15×10-8 (p-value)	GTEx Portal V6	12
CCHCR1	?	ENSG00000204536	tibial nerve	2.05×10-6 (p-value)	GTEx Portal V6	12
XXbac-BPG300A18.13	?	ENSG00000273333	tibial nerve	2.25×10-6 (p-value)	GTEx Portal V6	10
XXbac-BPG300A18.13	?	ENSG00000273333	esophagus mucosa	1.02×10-8 (p-value)	GTEx Portal V6	18
ATF6B	?	ENSG00000213676	esophagus mucosa	8.01×10-7 (p-value)	GTEx Portal V6	11
DXO	ENST00000474587	ILMN_1726990	blood	1.56×10-5 (p-value)	Westra et al.	2
DXO	ENST00000491327					
DXO	ENST00000473976					
DXO	ENST00000480240					
DXO	ENST00000498357					
DXO	ENST00000477826					
DXO	ENST00000492946					
DXO	ENST00000375349					
DXO	ENST00000337523					
DXO	ENST00000460058					
DXO	ENST00000375356					
DXO	ENST00000487914					
DXO	ENST00000478221					
DXO	ENST00000485557					
HSPA1L	ENST00000375654	ILMN_1654566	blood	3.16×10-6 (p-value)	Westra et al.	2
HSPA1B	ENST00000375650	ILMN_1660436	blood	2.45×10-6 (p-value)	Westra et al.	1
NELFE	ENST00000492185	ILMN_1765532	blood	2.23×10-6 (p-value)	Westra et al.	4
NELFE	ENST00000488426					
NELFE	ENST00000481121					
NELFE	ENST00000375429					
NOTCH4	ENST00000474612	ILMN_1711157	blood	3.19×10-5 (p-value)	Westra et al.	3
NOTCH4	ENST00000375023					
NOTCH4	ENST00000491215					
AGPAT1	ENST00000375107	ILMN_1679520	blood	1.09×10-4 (p-value)	Westra et al.	2
AGPAT1	ENST00000395499					
AGPAT1	ENST00000375104					
AGPAT1	ENST00000395496					
AGPAT1	ENST00000395497					
AGPAT1	ENST00000490711					
AGPAT1	ENST00000336984					
CYP21A1P	?	ENSG00000204338	adrenal gland	4.42×10-10 (p-value)	GTEx Portal V6	18
CYP21A1P	?	ENSG00000204338	testis	6.35×10-11 (p-value)	GTEx Portal V6	18
EHMT2	?	ENSG00000204371	testis	3.76×10-6 (p-value)	GTEx Portal V6	2
CYP21A1P	?	ENSG00000204338	lung	3.80×10-12 (p-value)	GTEx Portal V6	17
CYP21A1P	?	ENSG00000204338	thyroid	4.47×10-12 (p-value)	GTEx Portal V6	17
CYP21A2	?	ENSG00000231852	thyroid	4.28×10-6 (p-value)	GTEx Portal V6	11
ATF6B	?	ENSG00000213676	atrial appendage	7.92×10-9 (p-value)	GTEx Portal V6	14
CYP21A1P	?	ENSG00000204338	atrial appendage	7.65×10-7 (p-value)	GTEx Portal V6	2

CYP21A1P	?	ENSG00000204338	transverse colon	6.50×10-8 (p-value)	GTEx Portal V6	15
CYP21A1P	?	ENSG00000204338	unexposed skin	5.25×10-7 (p-value)	GTEx Portal V6	1
CYP21A1P	?	ENSG00000204338	cortex	5.11×10-9 (p-value)	GTEx Portal V6	15
CYP21A1P	?	ENSG00000204338	sigmoid colon	4.18×10-8 (p-value)	GTEx Portal V6	13
CYP21A1P	?	ENSG00000204338	cerebellum	3.20×10-8 (p-value)	GTEx Portal V6	13
CYP21A1P	?	ENSG00000204338	caudate basal ganglia	1.05×10-6 (p-value)	GTEx Portal V6	1
CYP21A1P	?	ENSG00000204338	cerebellar hemisphere	3.59×10-7 (p-value)	GTEx Portal V6	12
HLA-DQB1	?	ENSG00000179344	blood	4.04×10-2 (q-value)	SeeQTL DB (HapMap)	2
HLA-DRB5	ENST00000374975	ILMN_1697499	monocyte	1.61×10-12 (p-value)	Zeller et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445144	1	ENCP00000048200	BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2 BTNL2
		ENCP00000048071	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000048171	PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1 PRRT1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001703098 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001703099 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
ENSR00000662352 (promoter flanking region)	1	NHLF	DNase1, H3K4me1, H3K27ac
		HSMMtube	DNase1, H3K27me3
		Osteobl	H3K4me2, H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3, Max, DNase1
		skin (NHDF-AD)	H3K4me1, DNase1, H3K27ac
		muscle (HSMM)	DNase1, H3K27me3
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3, DNase1

liver (HepG2)	H3K4me1, H3K27me3
lung (IMR90)	H3K36me3, H3K27me3, DNase1
blood (GM12878)	H3K27me3
nervous (NH-A)	H3K27me3, DNase1
skin (NHEK)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C2	upstream gene variant	1688	ENST00000485690	?	ENSP00000417790	1
C2	upstream gene variant, downstream gene variant	3460	ENST00000486124	?	?	2
C2	downstream gene variant	4910	ENST00000497706	?	ENSP00000417482	1
C2	downstream gene variant	4139	ENST00000383177	?	ENSP00000372664	1
C2	downstream gene variant	4092	ENST00000442278	NM_001145903.2	ENSP00000395683	1
C2	downstream gene variant	3701	ENST00000413154	?	ENSP00000403325	1
C2	downstream gene variant	4242	ENST00000469372	NM_001282457.1	ENSP00000418923	1
C2	downstream gene variant	4092	ENST00000452323	NM_001178063.2	ENSP00000392322	1
C2	downstream gene variant	4091	ENST00000468407	?	?	1
C2	downstream gene variant	4372	ENST00000482060	?	ENSP00000418332	1
C2	downstream gene variant	4091	ENST00000299367	NM_001282458.1, NM_000063.5	ENSP00000299367	1
C2-AS1	downstream gene variant	1879	ENST00000630806	?	?	1
C4A	upstream gene variant	4155	ENST00000428956	NM_007293.2	ENSP00000396688	2
C4A	upstream gene variant	4974	ENST00000467749	?	?	1
C4A	upstream gene variant	4199	ENST00000498271	NM_001252204.1	ENSP00000420212	2
C4B	downstream gene variant	3441	ENST00000468936	?	?	2
C4B	downstream gene variant	3427	ENST00000425700	?	ENSP00000391933	2
C4B	downstream gene variant	3426	ENST00000435363	NM_001002029.3	ENSP00000415941	2
C4B	downstream gene variant	3842	ENST00000496065	?	?	2
C4B	downstream gene variant	3443	ENST00000468237	?	?	2
C4B	downstream gene variant	4513	ENST00000473957	?	?	2
C4B	downstream gene variant	3610	ENST00000463249	?	?	2
C4B	downstream gene variant	4239	ENST00000496560	?	?	2
C4B-AS1	upstream gene variant	2586	ENST00000415626	?	?	2
CFB	downstream gene variant	165	ENST00000497841	?	?	2
CFB	upstream gene variant, downstream gene variant	1111	ENST00000482312	?	?	2
CFB	downstream gene variant	506	ENST00000482886	?	?	2
CFB	upstream gene variant, downstream gene variant	1614	ENST00000498317	?	ENSP00000418833	2
CFB	downstream gene variant	1794	ENST00000460718	?	ENSP00000417793	1
CFB	downstream gene variant	2622	ENST00000475617	?	ENSP00000420090	1
CFB	downstream gene variant	212	ENST00000461483	?	?	2
CFB	downstream gene variant	2643	ENST00000472581	?	?	1

CFB	downstream gene variant	165	ENST00000467150	?	?	2
CFB	downstream gene variant	3053	ENST00000452035	?	?	1
CFB	downstream gene variant	1645	ENST00000483004	?	ENSP00000419887	1
CFB	downstream gene variant	1614	ENST00000425368	NM_001710.5	ENSP00000416561	1
CFB	downstream gene variant	165	ENST00000465750	?	?	2
CFB	downstream gene variant	2454	ENST00000467360	?	?	1
CYP21A2	downstream gene variant	3062	ENST00000478281	?	ENSP00000419572	1
CYP21A2	downstream gene variant	825	ENST00000418967	NM_000500.7	ENSP00000408860	1
CYP21A2	downstream gene variant	3011	ENST00000488465	?	?	1
CYP21A2	downstream gene variant	2855	ENST00000469053	?	ENSP00000418104	1
CYP21A2	downstream gene variant	853	ENST00000435122	NM_001128590.3	ENSP00000415043	1
CYP21A2	downstream gene variant	2672	ENST00000466879	?	?	1
CYP21A2	downstream gene variant	853	ENST00000479074	?	?	1
CYP21A2	downstream gene variant	1309	ENST00000483041	?	?	1
CYP21A2	downstream gene variant	2889	ENST00000471671	?	ENSP00000418561	1
CYP21A2	downstream gene variant	853	ENST00000486063	?	?	1
CYP21A2	downstream gene variant	853	ENST00000479730	?	?	1
CYP21A2	downstream gene variant	2739	ENST00000464325	?	?	1
CYP21A2	downstream gene variant	3380	ENST00000480027	?	?	1
CYP21A2	upstream gene variant, downstream gene variant	105	ENST00000462278	?	?	3
CYP21A2	downstream gene variant	2671	ENST00000466779	?	ENSP00000417321	1
DXO	upstream gene variant	4609	ENST00000487914	?	?	1
DXO	upstream gene variant	4462	ENST00000480240	?	ENSP00000418632	1
DXO	upstream gene variant	4503	ENST00000473976	?	?	1
DXO	upstream gene variant	4346	ENST00000477826	?	?	1
DXO	upstream gene variant	4436	ENST00000492946	?	?	1
DXO	upstream gene variant	4915	ENST00000498357	?	?	1
DXO	upstream gene variant	4500	ENST00000474587	?	?	1
DXO	upstream gene variant	4389	ENST00000485557	?	?	1
DXO	upstream gene variant	4360	ENST00000478221	?	?	1
DXO	upstream gene variant	4306	ENST00000337523	NM_005510.3	ENSP00000337759	1
DXO	upstream gene variant	4589	ENST00000460058	?	?	1
DXO	upstream gene variant	4614	ENST00000375356	?	ENSP00000364505	1
DXO	upstream gene variant	4913	ENST00000491327	?	?	1
DXO	upstream gene variant	4308	ENST00000375349	?	ENSP00000364498	1
MIR1236	downstream gene variant, upstream gene variant	3141	ENST00000408340	?	?	2
NELFE	downstream gene variant, upstream gene	1075	ENST00000492539	?	ENSP00000419183	2

	variant					
NELFE	downstream gene variant, upstream gene variant	1804	ENST00000375429	NM_002904.5	ENSP00000364578	2
NELFE	downstream gene variant, upstream gene variant	60	ENST00000441998	?	ENSP00000397914	3
NELFE	downstream gene variant, upstream gene variant	2062	ENST00000375425	?	ENSP00000364574	2
NELFE	downstream gene variant, upstream gene variant	691	ENST00000436289	?	ENSP00000414029	3
NELFE	downstream gene variant, upstream gene variant	606	ENST00000454913	?	ENSP00000409389	3
NELFE	downstream gene variant, upstream gene variant	1386	ENST00000494956	?	ENSP00000420205	2
NELFE	downstream gene variant, upstream gene variant	2468	ENST00000625905	?	ENSP00000486884	2
NELFE	downstream gene variant, upstream gene variant	1122	ENST00000426722	?	ENSP00000394340	2
NELFE	downstream gene variant, upstream gene variant	1952	ENST00000481121	?	?	2
NELFE	downstream gene variant, upstream gene variant	1367	ENST00000491139	?	?	2
NELFE	downstream gene variant, upstream gene variant	1995	ENST00000488426	?	?	2
NELFE	downstream gene variant, upstream gene variant	1954	ENST00000444811	?	ENSP00000388400	2
NELFE	downstream gene variant, upstream gene variant	1876	ENST00000492185	?	?	2
RNA5SP206	upstream gene variant	4248	ENST00000516703	?	?	1
SKIV2L	downstream gene variant	956	ENST00000492900	?	?	1
SKIV2L	upstream gene variant	583	ENST00000466290	?	?	1
STK19	downstream gene variant	4179	ENST00000460018	?	ENSP00000418350	1
STK19	downstream gene variant	3222	ENST00000479644	?	?	3
STK19	upstream gene variant	2522	ENST00000490822	?	?	3
TNXB	downstream gene variant	2046	ENST00000490077	?	?	2
TNXB	downstream gene variant	3019	ENST00000479795	?	ENSP00000418248	1
TNXB	downstream gene variant	3157	ENST00000498094	?	?	2
TNXB	downstream gene variant	2045	ENST00000375244	NM_019105.6	ENSP00000364393	2
TNXB	downstream gene variant	2046	ENST00000451343	NM_032470.3	ENSP00000407685	2
TNXB	downstream gene variant	653	ENST00000613214	?	ENSP00000480067	2
TNXB	downstream gene variant, upstream gene variant	23	ENST00000611016	?	ENSP00000483409	5
XXbac-BPG116M5.17	downstream gene variant	1650	ENST00000456570	?	ENSP00000410815	1
XXbac-BPG116M5.17	downstream gene variant	1668	ENST00000477310	?	ENSP00000418996	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C2	ENST00000442278	NM_001145903.2	ENSP00000395683	1
C2	ENST00000411571	?	ENSP00000388727	1
C2	ENST00000447952	?	ENSP00000391354	1
C2	ENST00000494905	?	ENSP00000419048	1
C2	ENST00000452323	NM_001178063.2	ENSP00000392322	1

C2	ENST00000452202	?	ENSP00000406121	1
C2	ENST00000482060	?	ENSP00000418332	1
C2	ENST00000484636	?	ENSP00000420305	1
C2	ENST00000497706	?	ENSP00000417482	1
C2	ENST00000418949	NM_001282459.1	ENSP00000406190	1
C2	ENST00000383177	?	ENSP00000372664	1
C2	ENST00000469372	NM_001282457.1	ENSP00000418923	1
C2	ENST00000299367	NM_001282458.1, NM_000063.5	ENSP00000299367	1
CFB	ENST00000425368	NM_001710.5	ENSP00000416561	1
CFB	ENST00000452035	?	?	1
CFB	ENST00000483004	?	ENSP00000419887	1
CYP21A2	ENST00000479730	?	?	1
CYP21A2	ENST00000464325	?	?	1
CYP21A2	ENST00000488465	?	?	1
CYP21A2	ENST00000471671	?	ENSP00000418561	1
CYP21A2	ENST00000418967	NM_000500.7	ENSP00000408860	1
CYP21A2	ENST00000479074	?	?	1
CYP21A2	ENST00000478281	?	ENSP00000419572	1
CYP21A2	ENST00000483041	?	?	1
CYP21A2	ENST00000486063	?	?	1
CYP21A2	ENST00000469053	?	ENSP00000418104	1
CYP21A2	ENST00000435122	NM_001128590.3	ENSP00000415043	1
CYP21A2	ENST00000466779	?	ENSP00000417321	1
NELFE	ENST00000492185	?	?	1
NELFE	ENST00000481121	?	?	1
NELFE	ENST00000375429	NM_002904.5	ENSP00000364578	1
NELFE	ENST00000444811	?	ENSP00000388400	1
NELFE	ENST00000375425	?	ENSP00000364574	1
NELFE	ENST00000488426	?	?	1
SKIV2L	ENST00000375394	NM_006929.4	ENSP00000364543	1
SKIV2L	ENST00000461073	?	ENSP00000419905	1
SKIV2L	ENST00000465703	?	?	1
SKIV2L	ENST00000474839	?	ENSP00000420470	1
SKIV2L	ENST00000628157	?	ENSP00000485707	1
SKIV2L	ENST00000488648	?	?	1
STK19	ENST00000484540	?	?	3
STK19	ENST00000492583	?	?	3
STK19	ENST00000491861	?	?	3
STK19	ENST00000478486	?	?	3
STK19	ENST00000375333	NM_032454.1	ENSP00000364482	3
STK19	ENST00000466336	?	?	3

STK19	ENST00000469907	?	?	3
STK19	ENST00000473983	?	?	3
STK19	ENST00000463823	?	?	3
STK19	ENST00000375331	NM_004197.1	ENSP00000364480	3
STK19	ENST00000466132	?	?	3
STK19	ENST00000471028	?	?	3
STK19	ENST00000519179	?	ENSP00000454870	3
STK19	ENST00000483801	?	ENSP00000418866	3
TNXB	ENST00000613214	?	ENSP00000480067	4
TNXB	ENST00000375244	NM_019105.6	ENSP00000364393	9
TNXB	ENST00000611016	?	ENSP00000483409	3
XXbac-BPG116M5.17	ENST00000456570	?	ENSP00000410815	2
XXbac-BPG116M5.17	ENST00000477310	?	ENSP00000418996	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CFB	ENST00000467360	?	1
CYP21A2	ENST00000479730	?	2
CYP21A2	ENST00000488465	?	2
CYP21A2	ENST00000466879	?	2
CYP21A2	ENST00000479074	?	2
CYP21A2	ENST00000483041	?	2
CYP21A2	ENST00000486063	?	2
CYP21A2	ENST00000480027	?	2
TNXB	ENST00000498094	?	1
TNXB	ENST00000490077	?	1