

## Column Definition

<b>Rank</b>	Rank of the gene
<b>Gene</b>	Gene Id
<b>FullName</b>	Full Name of the gene
<b>Pathway</b>	Known Pathway Associated with the Gene
<b>mRNA</b>	Transcript ID
<b>Chr</b>	Chromosome
<b>Score</b>	VAAST score of the transcript
<b>PValue</b>	VAAST p-value of the transcript
<b>LOD_Score</b>	VAAST LOD score of the transcript
<b>LOD_pValue</b>	VAAST LOD score p-value of the transcript
<b>Strand</b>	Transcript Strand
<b>#ofVar</b>	Total number of variants in the transcript
<b>VarType</b>	Type of the variant. SNVs is labeled as either "TU" or "TR". If a target variant is observed in background genomes, it is labeled "TR" (for "target"); otherwise if a variant is only observed in target genomes, it is labeled "TU" (for "target unique").
<b>VarScore</b>	Score of each variant, the sum roughly equals the transcript score ("Score") in column 8.
<b>VarLODScore</b>	VAAST LOD score of each variant
<b>VarPosition</b>	The chromosomal position of each variant
<b>AlleleCount</b>	Number of individuals in the background file containing a variant if the variant is not unique to the targets.
<b>Ref</b>	Reference nucleotide and amino-acid allele
<b>GenoType</b>	Individual genotypes following the VAAST output format. Please see the "VAAST Output" section of the VAAST users guide ( <a href="http://www.yandell-lab.org/software/VAAST/VAAST_Users_Guide.pdf">http://www.yandell-lab.org/software/VAAST/VAAST_Users_Guide.pdf</a> ) for detailed description.

## Commands

### Command - dominant

```
VAAST -gw 1e7 -m pvaast --indel --codon_bias --splice_site --fast_gp -pv_control TS0011_d.ctl -l phastcons-hg19-vertebrate.txt -o TS0011_d refGene_hg19.gff3 1kg-cgi-427_gene_only.cdr
```

### Control File - dominant (TS0011\_d.ctl)

```
#----- Basic options-----
```

```
input_ped_cdr_files:          TS0011.ped TS0011.cdr
pedigree_representatives:    3
unknown_representatives:    yes
additional_cases:
inheritance_model:           dominant
```

```
#----- Performance Tuning -----
```

```
informative_site_selection:  3
simulate_genotyping_error:   yes
genotyping_error_rate:       1.00E-04
penetrance_lower_bound:      0.6
penetrance_upper_bound:      1
```

```
#----- Gene and Variant Filtering -----
```

```
max_prevalence_filter:       0.01
lod_score_filter:            yes
clr_score_filter:            no
nocall_filter:               yes
nocall_filter_cutoff:        2
inheritance_error_filter:     yes
```

### Command - recessive

```
VAAST -gw 1e7 -m pvaast --indel --codon_bias --splice_site --fast_gp -pv_control TS0011_r.ctl -l phastcons-hg19-vertebrate.txt -o TS0011_r refGene_hg19.gff3 1kg-cgi-427_gene_only.cdr
```

### Control File - recessive (TS0011\_r.ctl)

```
#----- Basic options-----
```

```
input_ped_cdr_files:          TS0011.ped TS0011.cdr
pedigree_representatives:    3
unknown_representatives:    yes
additional_cases:
inheritance_model:           recessive
```

```
#----- Performance Tuning -----
```

```
informative_site_selection:  3
simulate_genotyping_error:   yes
genotyping_error_rate:       1.00E-04
penetrance_lower_bound:      0.6
penetrance_upper_bound:      1
```

```
#----- Gene and Variant Filtering -----
```

```
max_prevalence_filter:       0.01
lod_score_filter:            yes
clr_score_filter:            no
nocall_filter:               yes
nocall_filter_cutoff:        2
inheritance_error_filter:     yes
```





81	SLC29A3	phenylalanyl-tRNA synthetase alpha subunit (FARSA) mRNA. Homo sapiens solute carrier family 29 (nucleoside transporters) member 3 (SLC29A3) transcript variant 1 mRNA. Homo sapiens short chain dehydrogenase/reductase family 39U member 1 (SDR39U1) mRNA. Homo sapiens armadillo repeat containing 9 (ARMC9) mRNA.	NM_001174098	chr10	6.03	5.00E-03	+	1	SNP	5.98	0 0.01	73115942	NA	G V	N 0-5 A:A H:H
82	SDR39U1	Homo sapiens short chain dehydrogenase/reductase family 39U member 1 (SDR39U1) mRNA.	NM_020195	chr14	9.58	5.00E-03	-	1	SNP	9.53	0 0.01	24910973	NA	T I	N 0-5 G:G L:L
83	ARMC9	Homo sapiens armadillo repeat containing 9 (ARMC9) mRNA.	NM_025139	chr2	12.88	6.00E-03	+	1	SNP	12.83	0 0.01	232087475	NA	T I	N 0-5 A:A K:K
84	CACNA1C	Homo sapiens calcium channel voltage-dependent L type alpha 1C subunit (CACNA1C) transcript variant 1 mRNA. Homo sapiens keratin associated protein 12-2 (KRTAP12-2) mRNA. Homo sapiens ataxin 3 (ATXN3) transcript variant h mRNA.	NM_001129830	chr12	9.6	6.00E-03	+	1	SNP	9.55	0 0.01	2791130	NA	C P	N 0-5 T:T L:L
85	KRTAP12-2	Homo sapiens keratin associated protein 12-2 (KRTAP12-2) mRNA.	NM_181684	chr21	8.2	6.00E-03	-	1	SNP	8.15	0 0.01	46086758	NA	C A	N 0 T:C T:A;N 1-5 T:T T:T
86	ATXN3_DUP_01	Homo sapiens ataxin 3 (ATXN3) transcript variant h mRNA.	NM_001164778	chr14	4.8	6.00E-03	-	1	SNP	4.75	0 0.01	92537354	NA	C G	N 0-5 G:G R:R
87	TMCO4	Homo sapiens transmembrane and coiled-coil domains 4 (TMCO4) mRNA. Homo sapiens enoyl CoA hydratase short chain 1 mitochondrial (ECHS1) nuclear gene encoding mitochondrial protein mRNA.	NM_181719	chr1	9.28	6.00E-03	-	1	SNP	9.23	0 0.01	20020994	NA	C R	N 0,2-4 T:C H:R;N 1,5 T:T H:H
88	ECHS1	Homo sapiens enoyl CoA hydratase short chain 1 mitochondrial (ECHS1) nuclear gene encoding mitochondrial protein mRNA.	NM_004092	chr10	0.71	6.00E-03	-	1	SNP	0.66	0 0.01	135186806	1	A V	B 1-5 A:G V:A;N 0 G:G A:A
89	ZNF844	Homo sapiens zinc finger protein 844 (ZNF844) mRNA.	NM_001136501	chr19	11.21	6.00E-03	+	1	SNP	5.72	0 1.19	12186921	NA	T V	N 0,2-5 T:C V:A
90	FZD5	Homo sapiens frizzled family receptor 5 (FZD5) mRNA. Homo sapiens MUS81 endonuclease homolog (S. cerevisiae) (MUS81) mRNA.	NM_003468	chr2	7.7	6.00E-03	-	1	SNP	5.76	0 0.42	208632817	10	G P	B 0,2-5 A:G L:P
91	MUS81	Homo sapiens frizzled family receptor 5 (FZD5) mRNA.	NM_025128	chr11	10.08	6.00E-03	+	1	SNP	10.03	0 0.01	65629934	NA	G R	N 1,3-5 C:C P:P;N 0,2 C:G P:R
92	MAP3K4	Homo sapiens mitogen-activated protein kinase kinase kinase 4 (MAP3K4) transcript variant 2 mRNA. Homo sapiens chromosome 9 open reading frame 150 (C9orf150) mRNA. Homo sapiens zinc finger and AT hook domain containing (ZFAT) transcript variant 5 mRNA.	NM_005922	chr6	13.52	6.00E-03	+	1	del	13.47	0 0.01	161519351	NA	CTG PA	0-5 :- :-
93	C9orf150	Homo sapiens mitogen-activated protein kinase kinase kinase 4 (MAP3K4) transcript variant 2 mRNA.	NM_203403	chr9	12.4	6.00E-03	+	1	ins	12.34	0 0.01	12775861	NA	- G	0-1,3-5 GGCGCGGC:GGCGCGGC GGG:GGG;2 ^: ^ ^
94	ZFAT	Homo sapiens zinc finger and AT hook domain containing (ZFAT) transcript variant 5 mRNA.	NM_020863	chr8	10.3	6.00E-03	-	1	SNP	4.81	0 1.19	135614326	7	G R	B 0,2-5 A:G W:R
95	F2RL3	Homo sapiens coagulation factor II (thrombin) receptor-like 3 (F2RL3) mRNA. Homo sapiens zinc finger protein 778 (ZNF778) transcript variant 2 mRNA.	NM_003950	chr19	9.81	6.00E-03	+	1	SNP	4.31	0 1.19	17001203	5	C P	B 0,2-5 T:C L:P
96	ZNF778	Homo sapiens zinc finger protein 778 (ZNF778) transcript variant 2 mRNA.	NM_001201407	chr16	17.41	6.00E-03	+	1	ins	17.36	0 0.01	89291210	NA	- -	0-5 GTGA:GTGA VM:VM
97	GAK	Homo sapiens cyclin G associated kinase (GAK) mRNA.	NM_005255	chr4	12.25	6.00E-03	-	1	SNP	6.76	0 1.19	862363	4	C D	B 0,2-5 A:C Y:D
98	BTNL2_DUP_07	Homo sapiens butyrophilin-like 2 (MHC class II associated) (BTNL2) mRNA.	NM_019602	chr6	10.69	6.00E-03	-	1	SNP	10.64	0 0.01	32362703	NA	G P	N 0-1,3-5 T:G Q:P;N 2 T:T Q:Q
99	MPP2	Homo sapiens membrane protein palmitoylated 2 (MAGUK p55 subfamily member 2) (MPP2) mRNA. Homo sapiens v-maf musculoaponeurotic fibrosarcoma oncogene homolog A (avian) (MAFA) mRNA.	NM_005374	chr17	10.01	6.00E-03	-	1	SNP	9.96	0 0.01	41960633	NA	C V	N 0-5 G:G L:L
100	MAFA	Homo sapiens membrane protein palmitoylated 2 (MAGUK p55 subfamily member 2) (MPP2) mRNA.	NM_201589	chr8	7.76	6.00E-03	-	1	del	7.71	0 0.01	144511954	NA	TGG HH	0-5 :- :-
101	LSG1	Homo sapiens large subunit GTPase 1 homolog (S. cerevisiae) (LSG1) mRNA. Homo sapiens mutated in colorectal cancers (MCC) transcript variant 2 mRNA.	NM_018385	chr3	8.54	7.00E-03	-	1	SNP	8.49	0 0.01	194373832	NA	T X	N 0-5 C:C E:E
102	MCC	Homo sapiens mutated in colorectal cancers (MCC) transcript variant 2 mRNA.	NM_001085377	chr5	12.92	7.00E-03	-	1	ins	12.86	0 0.01	112824048	NA	- S	0,2-3 :-GCC :-G;1,4-5 GCC:GCC G:G
103	DLC1	Homo sapiens deleted in liver cancer 1 (DLC1) transcript variant 3 mRNA.	NM_024767	chr8	6.93	7.00E-03	-	1	SNP	6.88	0 0.01	13356819	NA	T Q	N 0-5 G:G H:H
104	MUC6	Homo sapiens mucin 6 oligomeric mucus/gel-forming (MUC6) mRNA. Homo sapiens protocadherin alpha subfamily C 2 (PCDHAC2) transcript variant 2 mRNA.	NM_005961	chr11	13.74	7.00E-03	-	1	SNP	8.25	0 1.19	1017691	NA	C A	N 0,2-5 C:G A:P
105	PCDHAC1	Homo sapiens protocadherin alpha subfamily C 2 (PCDHAC2) transcript variant 2 mRNA.	NM_014005	chr5	13.65	7.00E-03	+	1	SNP	8.16	0 1.19	140230371	NA	A K	N 2-5 A:C K:T;N 0 C:C T:T
106	ISCU	Homo sapiens iron-sulfur cluster scaffold homolog (E. coli) (ISCU) nuclear gene encoding mitochondrial protein transcript variant 2 mRNA.	NM_213595	chr12	13.48	7.00E-03	+	1	SNP	13.43	0 0.01	108956418	NA	T F	N 0-5 G:G C:C
107	RNF169	Homo sapiens ring finger protein 169 (RNF169) mRNA. Homo sapiens family with sequence similarity 135 member B (FAM135B) mRNA.	NM_001098638	chr11	9.99	7.00E-03	+	1	SNP	4.5	0 1.19	74460148	8	C P	B 2-5 T:C S:P;N 0 T:T S:S
108	FAM135B	Homo sapiens family with sequence similarity 135 member B (FAM135B) mRNA.	NM_015912	chr8	9.55	8.00E-03	-	1	SNP	9.5	0 0.01	139165273	NA	G P	N 0,4 A:A L:L;N 1-3,5 A:G L:P
109	POTEF	Homo sapiens POTE ankyrin domain family member F (POTEF) mRNA. Homo sapiens piccolo (presynaptic cytomatrix protein) (PCLO) transcript variant 2 mRNA.	NM_001099771	chr2	9.94	8.00E-03	-	1	SNP	9.89	0 0.01	130832292	NA	T V	N 1,3-5 A:A F:F;N 0,2 A:T F:Y
110	PCLO	Homo sapiens piccolo (presynaptic cytomatrix protein) (PCLO) transcript variant 2 mRNA.	NM_014510	chr7	17.41	9.00E-03	-	1	ins	17.36	0 0.01	82581488	NA	- -	0-5 TCA:TCA *:*
111	LAMA4	Homo sapiens laminin alpha 4 (LAMA4) transcript variant 5 mRNA. Homo sapiens interferon regulatory factor 2 binding protein-like (IRF2BP1) mRNA.	NM_001105206	chr6	12.82	9.00E-03	-	1	SNP	12.77	0 0.01	112508770	NA	G A	N 0-5 T:T E:E
112	IRF2BP1	Homo sapiens interferon regulatory factor 2 binding protein-like (IRF2BP1) mRNA.	NM_024496	chr14	13.52	9.00E-03	-	1	del	13.47	0 0.01	77493792	NA	TGT QQ	0-5 :- :-
113	MICAL1	Homo sapiens microtubule associated monooxygenase calponin and LIM domain containing 1 (MICAL1) transcript variant 1 mRNA. Homo sapiens phosphatidylinositol glycan anchor biosynthesis class Q (PIGQ) transcript variant 2 mRNA.	NM_001159291	chr6	8.2	9.00E-03	-	1	SNP	8.15	0 0.01	109767931	NA	C A	N 1-2,5 T:C T:A;N 0,3-4 T:T T:T
114	PIGQ	Homo sapiens microtubule associated monooxygenase calponin and LIM domain containing 1 (MICAL1) transcript variant 1 mRNA.	NM_148920	chr16	9.52	9.00E-03	+	1	SNP	9.47	0 0.01	633354	NA	G C	N 2-4 A:A Y:Y;N 0-1,5 A:G Y:C

115	TMEM44	Homo sapiens transmembrane protein 44 (TMEM44) transcript variant 1 mRNA.		NM_001166305	chr3	7.06	9.00E-03	-	1	SNP	7.01	0 0.01	194338424	NA	G R		N 2-3 C C G;G;N 0-1,4-5 C;G G;R	
116	CUL7	Homo sapiens cullin 7 (CUL7) transcript variant 2 mRNA.		NM_014780	chr6	8.57	9.00E-03	-	1	SNP	8.52	0 0.01	43014299	NA	T Q		N 0-5 C C R;R	
117	TMEM184A	Homo sapiens transmembrane protein 184A (TMEM184A) mRNA.		NM_001097620	chr7	12.18	1.00E-02	-	1	ins	12.13	0 0.01	1586653	NA	- -		1-2,4 :-GCC :-G;0,3,5 GCC:GCC G;G	
118	PCM1	Homo sapiens pericentriolar material 1 (PCM1) mRNA.		NM_006197	chr8	10.22	1.00E-02	+	1	SNP	10.17	0 0.01	17796382	NA	A N		N 1-5 A;G N;S;N 0 G;G S;S	
119	SPAG17	Homo sapiens sperm associated antigen 17 (SPAG17) mRNA.		NM_206996	chr1	12.06	1.00E-02	-	1	SNP	6.57	0 1.19	118644480	NA	G P		N 0,2-5 C;G A;P	
120	KRTAP10-2	Homo sapiens keratin associated protein 10-2 (KRTAP10-2) mRNA.		NM_198693	chr21	14.62	1.00E-02	-	1	del	9.12	0 1.19	45970760	NA	CTTGACGACAGAG PVCKK		0,2-5 :-CTTGACGACAGAG :-PVCKK	
121	CPS1	Homo sapiens carbamoyl-phosphate synthase 1 mitochondrial (CPS1) transcript variant 3 mRNA.		NM_001122633	chr2	7.76	1.00E-02	+	1	SNP	7.71	0 0.01	211456637	NA	A T		N 1-5 A;G T;A;N 0 G;G A;A	
122	RNF212	Homo sapiens ring finger protein 212 (RNF212) transcript variant 3 mRNA.		NM_001193318	chr4	17.41	1.00E-02	-	1	ins	17.36	0 0.01	1087327	NA	- -		1 CTGCCAGGCTAGAGCCAGCC:CTGCCAGGCTAGAGCCAGCC GWL*PGQ;GWL*PGQ;0,2-4 CTGCCAGGCTGGAGCCAGCC:CTGCCAGGCTGGAGCCAGCC GWLQPGQ;GWLQPGQ;5 CTGTCCAGGCTGGAGCCAGCC:CTGTCCAGGCTGGAGCCAGCC GWLQPGQ;GWLQPGQ	
123	FMNL1	Homo sapiens formin-like 1 (FMNL1) mRNA.		NM_005892	chr17	6.63	1.00E-02	+	1	SNP	6.58	0 0.01	43318779	NA	C P		N 0-5 G;G A;A	
124	TCF15	Homo sapiens transcription factor 15 (basic helix-loop-helix) (TCF15) mRNA.	Transcription Factor	NM_004609	chr20	10.03	1.00E-02	-	1	SNP	9.98	0 0.01	590542	NA	C V		N 0-5 G;G L;L	
125	RAB11FIP1	Homo sapiens RAB11 family interacting protein 1 (class I) (RAB11FIP1) transcript variant 1 mRNA.		NM_025151	chr8	6.69	1.00E-02	-	1	SNP	6.64	0 0.01	37728017		3	A M		B 0,3,5 A;G M;T;B 1-2,4 G;G T;T
126	MLL3	Homo sapiens myeloid/lymphoid or mixed-lineage leukemia 3 (MLL3) mRNA.		NM_170606	chr7	13.61	1.00E-02	-	1	ins	13.55	0 0.01	151945071	NA	- Y		0-5 :-T :-N	
127	OAS3	Homo sapiens 2',5'-oligoadenylate synthetase 3 100kDa (OAS3) mRNA.		NM_006187	chr12	3.87	1.00E-02	+	1	SNP	3.82	0 0.01	113388598		5	G R		N 4 A;A H;H;B 0-3,5 A;G H;R
128	ZDBF2	Homo sapiens zinc finger DBF-type containing 2 (ZDBF2) mRNA.		NM_020923	chr2	7.91	1.00E-02	+	1	SNP	4.64	0 0.71	207173390		9	G E		B 0,2-5 A;G K;E
129	KRTAP9-1	Homo sapiens keratin associated protein 9-1 (KRTAP9-1) mRNA.		NM_001190460	chr17	13.52	1.00E-02	+	1	del	13.47	0 0.01	39346593	NA	CCT TC		0-5 :-CCT :-TC	
130	NLRP6	Homo sapiens NLR family pyrin domain containing 6 (NLRP6) mRNA.		NM_138329	chr11	7.55	1.00E-02	+	1	SNP	7.5	0 0.01	280816	NA	A Y		N 1 A;T Y;F;N 0,2-5 T;T F;F	
131	FANCM	Homo sapiens Fanconi anemia complementation group M (FANCM) mRNA.		NM_020937	chr14	8.55	1.00E-02	+	1	SNP	3.06	0 1.19	45636328		7	A N		B 0,2-5 A;G N;S
132	TELO2	Homo sapiens telomere maintenance 2 homolog (S. cerevisiae) (TELO2) mRNA.		NM_016111	chr16	7.42	2.00E-02	+	1	SNP	7.37	0 0.01	1544302	NA	A E		N 0-4 A;G E;G;N 5 G;G G;G	
133	NCOR2	Homo sapiens nuclear receptor corepressor 2 (NCOR2) transcript variant 1 mRNA.	Transcription Factor;Huntington Disease;Notch Signaling Pathway	NM_001206654	chr12	12.18	2.00E-02	-	1	ins	12.13	0 0.01	124887058	NA	- -		0-5 GCT:GCT S;S	
134	PSD3	Homo sapiens pleckstrin and Sec7 domain containing 3 (PSD3) transcript variant 2 mRNA.		NM_015310	chr8	9.53	2.00E-02	-	1	SNP	9.48	0 0.01	18729817	NA	G T		N 0-5 A;A M;M	
135	COL24A1	Homo sapiens collagen type XXIV alpha 1 (COL24A1) mRNA.		NM_152890	chr1	3.86	3.00E-02	-	1	SNP	3.81	0 0.01	86591837		207	G A		B 0,3,5 A;A V;V;B 1-2,4 A;G V;A
136	SCARF2	Homo sapiens scavenger receptor class F member 2 (SCARF2) transcript variant 2 mRNA.		NM_182895	chr22	10.03	3.00E-02	-	1	SNP	9.98	0 0.01	20779946	NA	C V		N 0-5 G;G L;L	
137	DNAH17	Homo sapiens dynein axonemal heavy chain 17 (DNAH17) mRNA.	Huntington Disease	NM_173628	chr17	9.87	3.00E-02	-	1	SNP	9.82	0 0.01	76491127	NA	T M		N 1,4 C C V;V;N 0,2-3,5 T;C M;V	
138	PLIN4	Homo sapiens perilipin 4 (PLIN4) mRNA.		NM_001080400	chr19	8.59	3.00E-02	-	1	SNP	8.54	0 0.01	4511453	NA	G A		N 0-5 A;A V;V	
139	RNF213	Homo sapiens ring finger protein 213 (RNF213) transcript variant 2 mRNA.		NM_020914	chr17	2.76	3.00E-02	+	1	SNP	2.71	0 0.01	78346870		6	C L		B 0-5 A;C I;L
140	OBSCN	Homo sapiens obscurin cytoskeletal calmodulin and titin-interacting RhoGEF (OBSCN) transcript variant 1 mRNA.		NM_052843	chr1	10.77	3.00E-02	+	1	SNP	10.72	0 0.01	228412228	NA	G A		N 4 A;A T;T;N 0-3,5 A;G T;A	
141	KRTAP10-7	Homo sapiens keratin associated protein 10-7 (KRTAP10-7) mRNA.		NM_198689	chr21	0.47	4.00E-02	+	1	SNP	0.42	0 0.01	46020867		2	A M		B 0-5 G;G V;V
142	EPPK1	Homo sapiens epiplakin 1 (EPPK1) mRNA.		NM_031308	chr8	11.81	4.00E-02	-	1	SNP	6.3	0 1.19	144940779		2	G R		B 0,2-5 A;G C;R
143	MUC16	Homo sapiens mucin 16 cell surface associated (MUC16) mRNA.		NM_024690	chr19	6.77	5.00E-02	-	1	SNP	6.72	0 0.01	9001833	NA	A M		N 0-5 G;G T;T	
144	HLA-DQB1_DUP_02	Homo sapiens HLA-DQB1 transcript variant 1 mRNA.		NM_002123	chr6	2.58	5.00E-02	-	1	SNP	2.53	0 0.01	32632745		4	G A		B 0,2,4 A;A V;V;B 1,3,5 A;G V;A
145	DSP	Homo sapiens desmoplakin (DSP) transcript variant 1 mRNA.		NM_004415	chr6	0.28	1.00E+00	+	1	SNP	13.77	0 0.00	7582993	NA	A E		N 1,3,5 A;T E;V	

## Recessive model

Rank	Gene	FullName	Pathway	mRNA	Chr	Score	PValue	LOD_Score	LOD_PValue	Strand	#ofVariants	VarType	VarScore	VarLODScore	VarPosition	AlleleCount	Ref	GenoType	
1	C4orf48	Homo sapiens chromosome 4 open reading frame 48 (C4orf48) transcript variant 2 mRNA.		NM_001141936	chr4	32.38	9.00E-07	1.6	9.00E-07	+	1	SNP	25	0 1.60361112696843	2044128	NA	C P	N 0-5 T;T L;L	
2	GPRIN1	Homo sapiens G protein regulated inducer of neurite outgrowth 1 (GPRIN1) mRNA.		NM_052899	chr5	44.11	2.00E-06	1.6	4.00E-04	-	2	del	19.3743	0 1.60361112696843	17602612	6	NA	CCTCCTTCCTC PRKED	0-5 :-CCTCCTTCCTC :-PRKED
3	SEPT9	Homo sapiens septin 9 (SEPT9) transcript variant 4 mRNA.		NM_001113492	chr1	29.05	3.00E-06	1.6	3.00E-06	+	1	SNP	21.6743	0 1.60361112696843	75494705	2	NA	CAAAGACCCAGGA DPGSL	0-5 :-CAAAGACCCAGGA :-DPGSL
4	KIAA0040	Homo sapiens KIAA0040 (KIAA0040) transcript variant 2 mRNA.		NM_001162893	chr1	49.5	3.00E-06	2.77	2.00E-05	-	2	del	19.3784	0 2.78390360357584	17512994	8	NA	TCTCTTG NKKK	0-5 :-TCTCTTG :-NKKK
5	ATXN3_DUP_01	Homo sapiens ataxin 3 (ATXN3) transcript variant h mRNA.		NM_001164774	chr1	24.09	9.00E-06	1.6	1.00E-05	-	1	SNP	16.7143	0 1.60361112696843	92537354	6	NA	C K	0-5 :-C :-K
6	SLC29A3	Homo sapiens solute carrier family 29 (nucleoside transporters) member 3 (SLC29A3) transcript variant 1 mRNA.		NM_001174098	chr1	28.85	1.00E-05	1.6	2.00E-05	+	1	SNP	21.4743	0 1.60361112696843	73115942	4	NA	G V	N 0-5 A;A I;I
7	GPATCH4	Homo sapiens G patch domain containing 4 (GPATCH4) transcript variant 2 mRNA.		NM_182679	chr1	40.24	1.00E-05	1.6	2.00E-05	-	1	ins	32.8643	0 1.60361112696843	15656504	9	NA	- -	0-5 AC AC V;V
8	IRF2BPL	Homo sapiens interferon regulatory factor 2 binding protein-like (IRF2BPL) mRNA.		NM_024496	chr1	36.34	3.00E-05	1.6	4.00E-05	-	1	del	28.9743	0 1.60361112696843	77493792	4	NA	TGT QQ	0-5 :- :-
9	PTGFRN	Homo sapiens prostaglandin F2 receptor negative regulator (PTGFRN) mRNA.		NM_020440	chr1	31.83	3.00E-05	1.6	9.00E-05	+	1	SNP	24.4543	0 1.60361112696843	11748771	1	NA	T S	N 0-5 A;A T;T
10	ABP1	Homo sapiens amiloride binding protein 1 (amine oxidase (copper-containing)) (ABP1) mRNA.		NM_001091	chr7	35.63	3.00E-05	1.6	1.00E-04	+	1	SNP	28.2543	0 1.60361112696843	15055605	5	NA	C T	N 0-5 G;G R;R
11	OR51M1	Homo sapiens olfactory receptor family 51 subfamily M member 1 (OR51M1) mRNA.		NM_001004756	chr1	30.23	6.00E-05	1.6	9.00E-04	+	1	SNP	22.8543	0 1.60361112696843	5411580	4	NA	T F	N 0-5 C;C L;L
12	FMNL1	Homo sapiens formin-like 1 (FMNL1) mRNA.		NM_005892	chr1	29.4	8.00E-05	1.6	1.00E-04	+	1	SNP	22.0843	0 1.60361112696843	43318779	4	NA	C P	N 0-5 G;G A;A



65	GAL3ST2	Homo sapiens galactose-3-O-sulfotransferase 2 (GAL3ST2) mRNA.		NM_022134	chr2	11.4 2	2.00E-03	0.18	4.00E-03	+	1	SNP	10.59	0 0.180112436754 885	24274353 2	NA	T L	N 0,3 T:C L:P
66	PALM2	Homo sapiens A kinase (PKA) anchor protein 2 (AKAP2) transcript variant 2 mRNA.		NM_007203	chr9	24.1 6	2.00E-03	1.6	2.00E-03	+	2	SNP	2.64 43	0 1.603611126968 43	11289936 1	NA	A K	N 0-5 C:C Q:Q
												ins	14.14 43	11290034 1	NA	- -	2-5 GAAGCT-- EA--;1 GAAGCT:GAAGCT EA:EA	
67	IRF2BP2	Homo sapiens interferon regulatory factor 2 binding protein 2 (IRF2BP2) transcript variant 1 mRNA.		NM_001077397	chr1	13.3 7	2.00E-03	1.6	1.00E-03	-	1	SNP	5.98	0 1.602893807354 75	23474500 9	25	A S	B 0-5 G:G P:P
68	ISCU	Homo sapiens iron-sulfur cluster scaffold homolog (E. coli) (ISCU) nuclear gene encoding mitochondrial protein transcript variant 2 mRNA.		NM_213595	chr1	36.3 2	2.00E-03	1.6	3.00E-03	+	1	SNP	28.93	0 0	10895641 8	NA	T F	N 0-5 G:G C:C
69	PSD3	Homo sapiens pleckstrin and Sec7 domain containing 3 (PSD3) transcript variant 2 mRNA.		NM_015310	chr8	32.3 5	2.00E-03	1.6	1.00E-02	-	1	SNP	24.97	0 1.602893807354 75	18729817	NA	G T	N 0-5 A:A M:M
70	OR9G9	Homo sapiens olfactory receptor family 9 subfamily G member 9 (OR9G9) mRNA.		NM_001013358	chr1	21.1 2	2.00E-03	1.6	6.00E-03	+	2	SNP	4.21 43	0 1.603611126968 43	56468020	2	T C	B 0-5 T:C C:R
												SNP	9.79 43	56468021	NA	G C	N 0-5 A:G Y:C	
71	TELO2	Homo sapiens TEL2 telomere maintenance 2 homolog (S. cerevisiae) (TELO2) mRNA.		NM_016111	chr1	30.2 6	2.00E-03	1.6	2.00E-02	+	1	SNP	22.86 43	0 1.603611126968 43	1544302	NA	A E	N 0-4 A:G E:G;N 5 G:G G:G
72	NEURL	Homo sapiens neuralized homolog (Drosophila) (NEURL) mRNA.	Notch Signaling Pathway;Nervous System Development	NM_004210	chr1	6.58 0	3.00E-03	0.18	2.00E-03	+	1	SNP	5.74	0 0.180112436754 885	10534445 1	115	G G	B 0,3-5 A:A S:5;B 1-2 A:G S:G
73	PTPLA	Homo sapiens protein tyrosine phosphatase-like (proline instead of catalytic arginine) member A (PTPLA) mRNA.		NM_014241	chr1	25.3 0	3.00E-03	1.6	4.00E-03	-	1	SNP	17.94	0 0	17659149	2	C E	B 0-5 T:T K:K
74	CDC42EP1	Homo sapiens CDC42 effector protein (Rho GTPase binding) 1 (CDC42EP1) mRNA.		NM_152243	chr2	18.7 2	3.00E-03	1.6	1.00E-03	+	1	del	11.31	0 1.602893807354 75	37964409	NA	CAGCGCGCTGCTGCAACCCT PAPAANPS	0-5 :- :-
75	EMR3	Homo sapiens egf-like module containing mucin-like hormone receptor-like 3 (EMR3) mRNA.	Neurological system process;Neurontransmitter Secretion	NM_032571	chr1	12.9 9	3.00E-03	1.6	2.00E-03	-	1	SNP	5.59	0 1.602893807354 75	14769339	16	C E	B 0-5 G:G Q:Q
76	HOMER2	Homo sapiens homeobox and leucine zipper encoding (HOMER2) mRNA.	Transcription Factor	NM_020834	chr1	25.7 4	3.00E-03	2.77	3.00E-03	-	1	del	12.96	0 2.783903603575 84	23744827	NA	TCC EE	2 :- :-;0-1,3-5 :-TCC :-EE
77	TRNP1	Homo sapiens TMF1-regulated nuclear protein 1 (TRNP1) mRNA.		NM_001013642	chr1	10.7 0	3.00E-03	1.6	2.00E-03	+	1	SNP	3.3	0 1.602893807354 75	27320356	27	T W	B 0-5 C:C R:R
78	SLC6A15	Homo sapiens solute carrier family 6 (neutral amino acid transporter) member 15 (SLC6A15) transcript variant 2 mRNA.	Neurological system process	NM_018057	chr1	16.5 2	3.00E-03	1.6	4.00E-03	-	1	SNP	9.2	0 1.602893807354 75	85277561	70	T D	B 0-5 A:A V:V
79	SAMD11	Homo sapiens sterile alpha motif domain containing 11 (SAMD11) mRNA.		NM_152486	chr1	23.8 5	3.00E-03	1.6	5.00E-03	+	1	SNP	16.87	0 1.603611126968 43	877831	4	T W	B 0-5 C:C R:R
80	COG3	Homo sapiens component of oligomeric golgi complex 3 (COG3) mRNA.		NM_031431	chr1	17.7 3	3.00E-03	1.6	3.00E-03	+	1	SNP	10.32	0 1.602893807354 75	46108853	27	T L	B 0-5 C:C S:S
81	TNFRSF10C	Homo sapiens tumor necrosis factor receptor superfamily member 10c decoy without an intracellular domain (TNFRSF10C) mRNA.	MAPKKK cascade;Nervous System Development;NF-KappaB Cascade	NM_003841	chr8	13.9 7	3.00E-03	1.6	4.00E-03	+	1	SNP	6.58	0 1.602893807354 75	22974450	62	T I	B 0-5 C:C T:T
82	NOL10	Homo sapiens nucleolar protein 10 (NOL10) mRNA.		NM_024894	chr2	9.86 0	4.00E-03	0.18	4.00E-03	-	1	SNP	9.02	0 0.180112436754 885	10717806	81	C D	B 1,5 T:C N:D;B 0,2-4 T:T N:N
83	RFX1	Homo sapiens regulatory factor X 1 (influences HLA class II expression) (RFX1) mRNA.	Transcription Factor	NM_002918	chr1	2.87 9	4.00E-03	0.18	4.00E-03	-	1	SNP	2.03	0 0.180112436754 885	14083761	56	T T	B 0,2-3,5 C:C A:A;B 1,4 T:C T:A
84	WWC2	Homo sapiens WW and C2 domain containing 2 (WWC2) mRNA.		NM_024949	chr4	16.3 0	4.00E-03	1.6	3.00E-03	+	1	SNP	8.91	0 1.602893807354 75	18419023 3	49	G A	B 2-5 T:G S:A;B 0 T:T S:S
85	MLLT6	Homo sapiens myeloid/lymphoid or mixed-lineage leukemia (trithorax homolog Drosophila); translocated to 6 (MLLT6) mRNA.	Transcription Factor	NM_005937	chr1	16.6 7	4.00E-03	1.6	2.00E-03	+	1	SNP	9.44	0 1.603611126968 43	36861983	6	T V	B 0-5 C:C A:A
86	PCLO	Homo sapiens piccolo (presynaptic cytomatrix protein) (PCLO) transcript variant 2 mRNA.	Neurological system process;Neurontransmitter Secretion	NM_033026	chr7	40.2 4	4.00E-03	1.6	1.00E-01	-	1	ins	32.86	0 1.603611126968 43	82581488	NA	- -	0-5 TCA:TC A**
87	PARP9	Homo sapiens poly (ADP-ribose) polymerase family member 9 (PARP9) transcript variant 6 mRNA.		NM_031458	chr3	21.2 9	4.00E-03	2.78	6.00E-03	-	1	SNP	8.46	0 2.783903603575 84	12227726 8	51	G S	B 0 A:A L:L;B 2-5 A:G L:L
88	ZP3	Homo sapiens zona pellucida glycoprotein 3 (sperm receptor) (ZP3) transcript variant 1 mRNA.		NM_001110354	chr7	14.4 6	4.00E-03	1.6	5.00E-03	+	1	SNP	7.07	0 1.602893807354 75	76054372	89	G G	B 0-5 A:A R:R
89	ZNF534	Homo sapiens zinc finger protein 534 (ZNF534) transcript variant 2 mRNA.	Transcription Factor	NM_001143939	chr1	20.6 9	4.00E-03	1.6	5.00E-03	+	1	SNP	13.22	0 1.603611126968 43	52942445	10	C H	B 0-5 T:T Y:Y
90	SOBP	Homo sapiens sine oculis binding protein homolog (Drosophila) (SOBP) mRNA.		NM_018013	chr6	13.8 0	5.00E-03	1.6	3.00E-03	+	1	SNP	6.41	0 1.602893807354 75	10795609 5	32	A S	B 0-5 G:G G:G
91	BCLAF1	Homo sapiens BCL2-associated transcription factor 1 (BCLAF1) transcript variant 1 mRNA.	Transcription Factor	NM_001077440	chr6	20.4 7	5.00E-03	1.6	7.00E-03	-	2	SNP	8.61 75	0 1.602893807354 75	13659069 8	NA	C R	N 0,4-5 T:C H:R
												SNP	0.62 75	13659728 8	19	A Y	B 0-5 A:C Y:D	
92	TXNDC2	Homo sapiens thioredoxin domain containing 2 (spermatzoa) (TXNDC2) transcript variant 1 mRNA.	Oxidative stress response;Stress Response	NM_001098529	chr1	18.5 8	5.00E-03	1.6	1.00E-02	+	1	SNP	11.15	0 1.602893807354 75	9887497	6	G E	B 0-5 A:A K:K
93	AOX1	Homo sapiens aldehyde oxidase 1 (AOX1) mRNA.		NM_001159	chr2	11.8 9	5.00E-03	0.18	7.00E-03	+	1	SNP	11.06	0 0.180112436754 885	20147859 8	NA	C A	N 0,2-3 T:C V:A
94	MEGF10	Homo sapiens multiple EGF-like-domains 10 (MEGF10) mRNA.	Nervous System Development	NM_032446	chr5	18.4 6	6.00E-03	2.78	6.00E-03	+	1	SNP	5.63	0 2.783903603575 84	12679128 2	69	G R	B 0 A:A K:K;B 2-5 A:G K:R
95	ARID3A	Homo sapiens AT rich interactive domain 3A (BRIGHT-like) (ARID3A) mRNA.	Transcription Factor	NM_005224	chr1	20.1 9	6.00E-03	1.6	6.00E-03	+	1	SNP	12.76	0 1.603611126968 43	971949	NA	G G	N 0-5 A:A S:S
96	NCOR2	Homo sapiens nuclear receptor corepressor 2 (NCOR2) transcript variant 1 mRNA.	Transcription Factor;Notch Signaling Pathway;Huntington Disease	NM_001077261	chr1	35.0 2	6.00E-03	1.6	6.00E-02	-	1	ins	27.63	0 1.603611126968 43	12488705 8	NA	- -	0-5 GCT:GCT S:S
97	MLL3	Homo sapiens myeloid/lymphoid or mixed-lineage leukemia 3 (MLL3) mRNA.		NM_170606	chr7	31.1 0	7.00E-03	1.6	3.00E-02	-	2	SNP	12.42 43	0 1.603611126968 43	15194500 1	NA	C G	N 1-2,5 T:C S:G
												ins	13.55 43	15194507 1	NA	- Y	0-5 :-T :-N	
98	ZNRF4	Homo sapiens zinc and ring finger 4 (ZNRF4) mRNA.		NM_181710	chr1	16.6 9	7.00E-03	2.78	7.00E-03	+	1	SNP	3.83	0 2.783903603575 84	5455735	128	G R	B 2,4 A:A Q:Q;B 0-1,3,5 A:G Q:R
99	PLIN4	Homo sapiens perilipin 4 (PLIN4) mRNA.		NM_001080400	chr1	31.4 9	7.00E-03	1.6	1.00E-01	-	1	SNP	24.04	0 1.603611126968 43	4511453	NA	G A	N 0-5 A:A V:V
100	KIF27	Homo sapiens kinesin family member 27 (KIF27) mRNA.		NM_017576	chr9	18.4 2	8.00E-03	2.78	8.00E-03	-	1	SNP	5.59	0 2.783903603575 84	86474115	96	T N	B 0 C D:D;B 2-5 T:C N:D
101	GPR44	Homo sapiens prostaglandin D2 receptor 2 (PTGDR2) mRNA.	Neurological system process;G-protein coupled receptor	NM_004778	chr1	8.36 1	8.00E-03	1.6	6.00E-03	-	1	SNP	0.96	0 1.602893807354 75	60620585	40	A V	B 0-5 G:G A:A
102	RIN3	Homo sapiens Ras and Rab interactor 3 (RIN3) mRNA.		NM_024832	chr1	19.5 4	8.00E-03	1.6	1.00E-02	+	1	SNP	12.12	0 1.602893807354 75	93118229	44	C R	B 0-5 T:T C:C
103	ZMYM5	Homo sapiens zinc finger MYM-type 5 (ZMYM5) transcript variant 1 mRNA.	Transcription Factor	NM_001039649	chr1	17.9 3	9.00E-03	1.6	1.00E-02	-	1	SNP	10.54	0 1.602816540651 69	20425911	72	C C	N 4 A:A F:F;B 0-3,5 A:C F:C
104	BPIL1	Homo sapiens BPI fold containing family B member 2 (BPFB2) mRNA.	Stress Response	NM_025227	chr2	0 5	1.00E-02	0.18	8.00E-03	+	1	SNP	4.31	0 0.180112436754 885	31596472	97	A K	B 1,5 A:T K:M;B 0,2-4 T:T M:M
105	CDC27	Homo sapiens cell division cycle 27 homolog (S. cerevisiae) (CDC27) transcript variant 2 mRNA.		NM_001114091	chr1	36.9 7	1.00E-02	1.58	5.00E-02	-	2	SNP	15.6 43	0 1.603611126968 43	45234360	2	A L	B 0-5 A:C L:*
												SNP	14.63 43	45249316 1	NA	T Y	N 0-5 T:C Y:C	
106	CDHR3	Homo sapiens cadherin-related family member 3 (CDHR3) mRNA.		NM_152750	chr7	15.9 6	1.00E-02	1.6	1.00E-02	+	1	SNP	8.57	0 1.602816540651 69	10565845 1	126	G C	B 0-5 A:A Y:Y
107	PRIM2	Homo sapiens primase DNA polypeptide 2 (58kDa) (PRIM2) mRNA.		NM_000947	chr6	18.8 6	1.00E-02	1.6	1.00E-02	+	2	SNP	6.42 0 0	57398270	416	C Q	B 0,2-5 T:C *Q	
												ins	5.06 0 0	57398186	NA	- E	0-5 A:- K:-	
108	SSCS5D	Homo sapiens scavenger receptor cysteine rich domain containing (5 domains) (SSCS5D) transcript variant 2 mRNA.		NM_001144950	chr1	31.2 9	1.00E-02	0.78	5.00E-02	+	1	ins	27.63	0 0.779126465217 891	56029616	NA	- -	0-1,4 :-CCA :-P;2-3 CCA:CCA P:P
109	C10orf92	Homo sapiens tetraatricopeptide repeat domain 40 (TTC40) mRNA.		NM_001200049	chr1	23.9 0	1.00E-02	2.78	4.00E-02	-	2	SNP	1.91 84	0 2.783903603575 84	13464827 9	38	C E	B 0-1,3,5 T:C K:E;B 2,4 T:T K:K
												SNP	9.23 84	13466080 7	NA	C G	N 0,3,5 T:C S:G	
110	C20orf151	Homo sapiens chromosome 20 open reading frame 151 (C20orf151) mRNA.	Transcription Factor	NM_080833	chr2	19.2 0	1.00E-02	1.6	2.00E-02	-	1	SNP	11.86	0 1.602893807354 75	60987737	29	C E	B 0-1,4-5 T:C K:E;B 2-3 T:T K:K
111	RP1L1	Homo sapiens retinitis pigmentosa 1-like 1 (RP1L1) mRNA.	Neurological system process	NM_178857	chr8	38.4 8	1.00E-02	1.58	1.00E-01	-	2	ins	19.36 43	0 1.603611126968 43	10467685	NA	- -	1-3,5



													75							
163	ZNF217	Homo sapiens zinc finger protein 217 (ZNF217) mRNA.	Transcription Factor	NM_006526	chr2	0	5.86	8.00E-01	0.83	2.00E-02	-	1	SNP	2.76	0 0	52192595	10	C R	B 0,2-3 T:C Q:R	
164	HLA-C_DUP_06	NA		NM_002117_DUP_03	chr6	3.49		8.00E-01	0.18	4.00E-01	-	1	SNP	23.81	0 1.60289380735475	31239776	1	A C	B 0,2-3,5 A:C C:G;N 1,4 C:C G:G	
165	SVEP1	Homo sapiens sushi von Willebrand factor type A EGF and pentraxin domain containing 1 (SVEP1) mRNA.		NM_153366	chr9	1.47		9.00E-01	0.14	1.00E-01	-	2	SNP	1.13	0 1.60289380735475	11316679	109	A F	B 1,4-5 A:T F:I	
166	MUC5B	Homo sapiens mucin 5B oligomeric mucus/gel-forming (MUC5B) mRNA.		NM_002458	chr1	1	3.34	9.00E-01	0.28	3.00E-01	+	1	SNP	20.56	0 1.60289380735475	11325195	20	T I	B 0,2-3,5 T:C I:V	
167	TTN	Homo sapiens titin (TTN) transcript variant novex-3 mRNA.		NM_133378	chr2	0.72		1.00E+00	0.05	3.00E-01	-	2	SNP	11.23	0 1.60289380735475	1268608	NA	A S	N 0 G:G G:G	
	HLA-DQB1_DUP_02	Homo sapiens major histocompatibility complex class II DQ beta 1 (HLA-DQB1) transcript variant 1 mRNA.		NM_002123	chr6	0.65		1.00E+00	0.05	4.00E-01	-	1	SNP	14.44	0 1.60289380735475	17940037	9	NA	A F	N 0,2 A:G F:L
169	HLA-A_DUP_07	NA		NM_002116_DUP_03	chr6	0.56		1.00E+00	0.03	3.00E-01	+	2	SNP	9.64	0 0	17944491	5	3	A S	B 0,2 A:G S:P
													SNP	5.15	0 1.60289380735475	32632745	4	G A	B 0,2,4 A:A V:V;B 1,3,5 A:G V:A	
													SNP	12.82	0 1.60281654065169	29911271	NA	G E	N 1,3,5 C:G D:E	
													SNP	12.82	0 1.60281654065169	29911272	NA	T W	N 1,5 T:G W:G	