

# SNiPACard

## Block annotations

### Block info










genomic range	chr3:46,596,264-46,631,058 <i>e!</i>
block size	34,795 bp
variant count	9 variants

### Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.261$ [-1.057 – 0.212]	gene(s) hit or close-by	AC104304.1 <i>e!</i> , LRRC2 <i>e!</i> , LRRC2-AS1 <i>e!</i> , TDGF1 <i>e!</i>
phastCons	$\mu = 0.047$ [0 – 0.328]	eQTL gene(s)	ALS2CL <i>e!</i> , LRRC2 <i>e!</i> , LRRC2-AS1 <i>e!</i> , TDGF1 <i>e!</i>
GERP++	$\mu = -0.249$ [-4.4 – 1.74]	potentially regulated gene(s)	AC104304.1 <i>e!</i> , CCR2 <i>e!</i> , LRRC2 <i>e!</i> , LTF <i>e!</i> , PRSS44 <i>e!</i> , PRSS50 <i>e!</i>
CADD score	$\mu = 4.291$ [1.295 – 8.84]	disease gene(s)	TDGF1 <i>e!</i> , LTF <i>e!</i> , CCR2 <i>e!</i>













## Trait annotations

### Disease gene annotation

gene	trait	source DB	source entry/link
TDGF1 <i>e!</i>	TERATOCARCINOMA-DERIVED GROWTH FACTOR 1	OMIM	MIM:187395 
LTF <i>e!</i>	SPECIFIC GRANULE DEFICIENCY	OMIM	MIM:245480 
CCR2 <i>e!</i>	HUMAN IMMUNODEFICIENCY VIRUS TYPE 1, SUSCEPTIBILITY TO	OMIM	MIM:609423 
TDGF1 <i>e!</i>	Semilobar holoprosencephaly	OrphaNet	OrphaNet:220386 
TDGF1 <i>e!</i>	Alobar holoprosencephaly	OrphaNet	OrphaNet:93925 
TDGF1 <i>e!</i>	Microform holoprosencephaly	OrphaNet	OrphaNet:280200 
TDGF1 <i>e!</i>	Midline interhemispheric variant of holoprosencephaly	OrphaNet	OrphaNet:93926 
TDGF1 <i>e!</i>	Septopreoptic holoprosencephaly	OrphaNet	OrphaNet:280195 
TDGF1 <i>e!</i>	Lobar holoprosencephaly	OrphaNet	OrphaNet:93924 

## Direct effect on regulation

### cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	atrial appendage	6.64×10 <sup>-21</sup> (p-value)	GTEx Portal V6 	9
ALS2CL <i>e!</i>	?	ENSG00000178038 <i>e!</i>	transformed fibroblasts	9.66×10 <sup>-7</sup> (p-value)	GTEx Portal V6 	5
LRRC2 <i>e!</i>	?	ENSG00000163827 <i>e!</i>	transformed fibroblasts	7.45×10 <sup>-33</sup> (p-value)	GTEx Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	breast	1.41×10 <sup>-27</sup> (p-value)	GTEx Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	skeletal muscle	1.27×10 <sup>-57</sup> (p-value)	GTEx Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	putamen	2.37×10 <sup>-10</sup> (p-value)	GTEx Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	unexposed skin	1.15×10 <sup>-20</sup> (p-value)	GTEx Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	sun exposed skin	1.16×10 <sup>-19</sup> (p-value)	GTEx Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	ovary	5.21×10 <sup>-18</sup> (p-value)	GTEx Portal V6 	9
LRRC2 <i>e!</i>	?	ENSG00000163827 <i>e!</i>	ovary	2.35×10 <sup>-6</sup> (p-value)	GTEx Portal V6 	1
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	left ventricle	2.50×10 <sup>-37</sup> (p-value)	GTEx Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	subcutaneous adipocytes	6.28×10 <sup>-55</sup> (p-value)	GTEx Portal V6 	9

TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	caudate basal ganglia	2.40×10 <sup>-12</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	visceral adipocytes	2.25×10 <sup>-32</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	cortex	5.06×10 <sup>-15</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	tibial nerve	8.12×10 <sup>-48</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	pituitary	2.18×10 <sup>-9</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	anterior cingulate cortex	1.80×10 <sup>-10</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	esophagus mucosa	2.71×10 <sup>-27</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	testis	4.56×10 <sup>-17</sup> (p-value)	GTEX Portal V6 	9
LRRC2 <i>e!</i>	?	ENSG00000163827 <i>e!</i>	testis	1.17×10 <sup>-7</sup> (p-value)	GTEX Portal V6 	9
LRRC2-AS1 <i>e!</i>	?	ENSG00000268324 <i>e!</i>	testis	1.42×10 <sup>-5</sup> (p-value)	GTEX Portal V6 	4
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	hypothalamus	1.44×10 <sup>-10</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	frontal cortex	7.78×10 <sup>-15</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	uterus	3.21×10 <sup>-13</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	muscularis mucosae	1.23×10 <sup>-40</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	lung	1.26×10 <sup>-27</sup> (p-value)	GTEX Portal V6 	9
LRRC2 <i>e!</i>	?	ENSG00000163827 <i>e!</i>	tibial artery	1.66×10 <sup>-8</sup> (p-value)	GTEX Portal V6 	7
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	prostate	2.11×10 <sup>-13</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	transverse colon	3.56×10 <sup>-7</sup> (p-value)	GTEX Portal V6 	5
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	terminal ileum	2.23×10 <sup>-7</sup> (p-value)	GTEX Portal V6 	5
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	nucleus accumbens	1.76×10 <sup>-12</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	spleen	2.86×10 <sup>-21</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	coronary artery	1.44×10 <sup>-14</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	sigmoid colon	1.13×10 <sup>-25</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	vagina	2.24×10 <sup>-9</sup> (p-value)	GTEX Portal V6 	7
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	gastroesophageal junction	2.89×10 <sup>-25</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	adrenal gland	1.93×10 <sup>-27</sup> (p-value)	GTEX Portal V6 	9
LRRC2 <i>e!</i>	?	ENSG00000163827 <i>e!</i>	adrenal gland	8.32×10 <sup>-27</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	cerebellar hemisphere	5.18×10 <sup>-8</sup> (p-value)	GTEX Portal V6 	9
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	cerebellum	8.08×10 <sup>-9</sup> (p-value)	GTEX Portal V6 	9
LRRC2 <i>e!</i>	?	ENSG00000163827 <i>e!</i>	cerebellum	9.29×10 <sup>-7</sup> (p-value)	GTEX Portal V6 	5
TDGF1 <i>e!</i>	?	ENSG00000241186 <i>e!</i>	hippocampus	2.99×10 <sup>-9</sup> (p-value)	GTEX Portal V6 	9

### ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000337772 <i>e!</i>	1	ENCP00000036261	LRRC2 <i>e!</i>
		ENCP00000036273	PRSS50 <i>e!</i>
		ENCP00000036248	CCR2 <i>e!</i>
		ENCP00000036263	AC104304.1 <i>e!</i>
		ENCP00000036262	AC104304.1 <i>e!</i>
		ENCP00000036277	PRSS44 <i>e!</i>
		ENCP00000036258	LTF <i>e!</i>

### Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001363001 <i>e!</i> (TF binding site)	3	embryonic stem cell (H1ESC) endothelium (HUVEC) blood (K562)	Rad21, TAF7, H3K27ac, HDAC2, H3K4me2, SP1, H3K4me3, DNase1, PolII, H3K27me3, H3K36me3 H3K27me3 H3K27me3
ENSR00001363008 <i>e!</i> (TF binding site)	2	embryonic stem cell (H1ESC) lung (IMR90) blood (K562)	DNase1, H3K36me3, PolII H3K27me3 H3K27me3

### Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC104304.1 <i>e!</i>	upstream gene variant	4217	ENST00000583198 <i>e!</i> ?	?	?	1
LRRC2-AS1 <i>e!</i>	upstream gene variant	2624	ENST00000599511 <i>e!</i> ?	?	?	1
LRRC2-AS1 <i>e!</i>	upstream gene variant	2630	ENST00000614743 <i>e!</i> ?	?	?	1
TDGF1 <i>e!</i>	upstream gene variant	1394	ENST00000459867 <i>e!</i> ?	?	?	4
TDGF1 <i>e!</i>	upstream gene variant	3402	ENST00000505797 <i>e!</i> ?	?	?	2
TDGF1 <i>e!</i>	upstream gene variant	1519	ENST00000493282 <i>e!</i> ?	?	?	3
TDGF1 <i>e!</i>	upstream gene variant	17	ENST00000471721 <i>e!</i> ?	?	?	4
TDGF1 <i>e!</i>	upstream gene variant, downstream gene variant	1008	ENST00000296145 <i>e!</i>	NM_003212.3	ENSP00000296145 <i>e!</i>	5
TDGF1 <i>e!</i>	downstream gene variant	4493	ENST00000542931 <i>e!</i>	NM_001174136.1	ENSP00000446375 <i>e!</i>	2

### Putative effect on transcript

#### Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LRRC2 <i>e!</i>	ENST00000296144 <i>e!</i>	?	ENSP00000296144 <i>e!</i>	5
LRRC2 <i>e!</i>	ENST00000496388 <i>e!</i>	?	?	1
LRRC2 <i>e!</i>	ENST00000395905 <i>e!</i>	NM_024512.4	ENSP00000379241 <i>e!</i>	1
LRRC2 <i>e!</i>	ENST00000469912 <i>e!</i>	?	?	1
TDGF1 <i>e!</i>	ENST00000542931 <i>e!</i>	NM_001174136.1	ENSP00000446375 <i>e!</i>	4
TDGF1 <i>e!</i>	ENST00000505797 <i>e!</i>	?	?	4

#### 5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TDGF1 <i>e!</i>	ENST00000296145 <i>e!</i>	NM_003212.3	ENSP00000296145 <i>e!</i>	1

#### Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
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