

A

Mutation Taster

Prediction

disease causing

Model: *simple_aae*, prob: 0.9999999999999999 (explain)

Summary

- amino acid sequence changed
- protein features (might be) affected
- splice site changes

[hyperlink](#)

analysed issue	analysis result												
name of alteration	no title												
alteration (phys. location)	chr5:149435680C>G show variant in all transcripts IGV												
HGNC symbol	CSF1R												
Ensembl transcript ID	ENST00000286301												
Genbank transcript ID	NM_005211												
UniProt peptide	P07333												
alteration type	single base exchange												
alteration region	CDS												
DNA changes	c.2463G>C cDNA.2755G>C g.57256G>C												
AA changes	W821C Score: 215 explain score(s)												
position(s) of altered AA if AA alteration in CDS	821												
frameshift	no												
known variant	Variant was neither found in ExAC nor 1000G. Search ExAC												
regulatory features	H3K36me3, Histone, Histone 3 Lysine 36 Tri-Methylation												
phyloP / phastCons	<table><tr><td></td><td>PhyloP</td><td>PhastCons</td></tr><tr><td>(flanking)</td><td>4.737</td><td>1</td></tr><tr><td></td><td>5.734</td><td>1</td></tr><tr><td>(flanking)</td><td>5.734</td><td>1</td></tr></table>		PhyloP	PhastCons	(flanking)	4.737	1		5.734	1	(flanking)	5.734	1
		PhyloP	PhastCons										
	(flanking)	4.737	1										
		5.734	1										
	(flanking)	5.734	1										
	explain score(s) and/or inspect your position(s) in in UCSC Genome Browser												

B

Mutation Assessor

	Mutation	AA variant	Gene	MSA	PDB	Func. Impact	FI score	Uniprot	Refseq	MSA height	Codon start position	Func. region	Protein bind.site	DNA/RNA bind.site	small.mol bind.site
1	CSF1R_HUMAN W821C	W821C	CSF1R	msa	pdb	medium	2.93	CSF1R_HUMAN	NP_005202	1303	isoforms hg19:chr5:149435680		1		MES EPE