




Impact Analysis

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CHEK2 gene

Let us look at the following CHEK2 transcript. Note that I am in GRCh37

Note that MutationTaster had been built for GRCh37.

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Human (GRCh37.p13) ▼

Location: 22:29,083,731-29,138,410

Gene: CHEK2

Gene-based displays

- Summary
 - Splice variants
 - Transcript comparison
 - Gene alleles
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- Comparative Genomics
 - Paralogues
 - Genomic alignments
 - Gene tree
 - Gene gain/loss tree
 - Orthologues
- Ontologies
 - GO: Biological process
 - GO: Molecular function
 - GO: Cellular component
- Phenotypes
- Genetic Variation
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- Gene expression

Gene: CHEK2 ENSG00000183765

Description checkpoint kinase 2 [Source:HGNC Symbol;Acc:16627]

Gene Synonyms CDS1, CHK2, HuCds1, LFS2, PP1425, RAD53, bA444G7, hCds1

Location [Chromosome 22: 29,083,731-29,138,410](#) reverse strand.
GRCh37:CM000684.1

About this gene This gene has 27 transcripts ([splice variants](#)) and is associated with [112 phenotypes](#).

Transcripts Hide transcript table

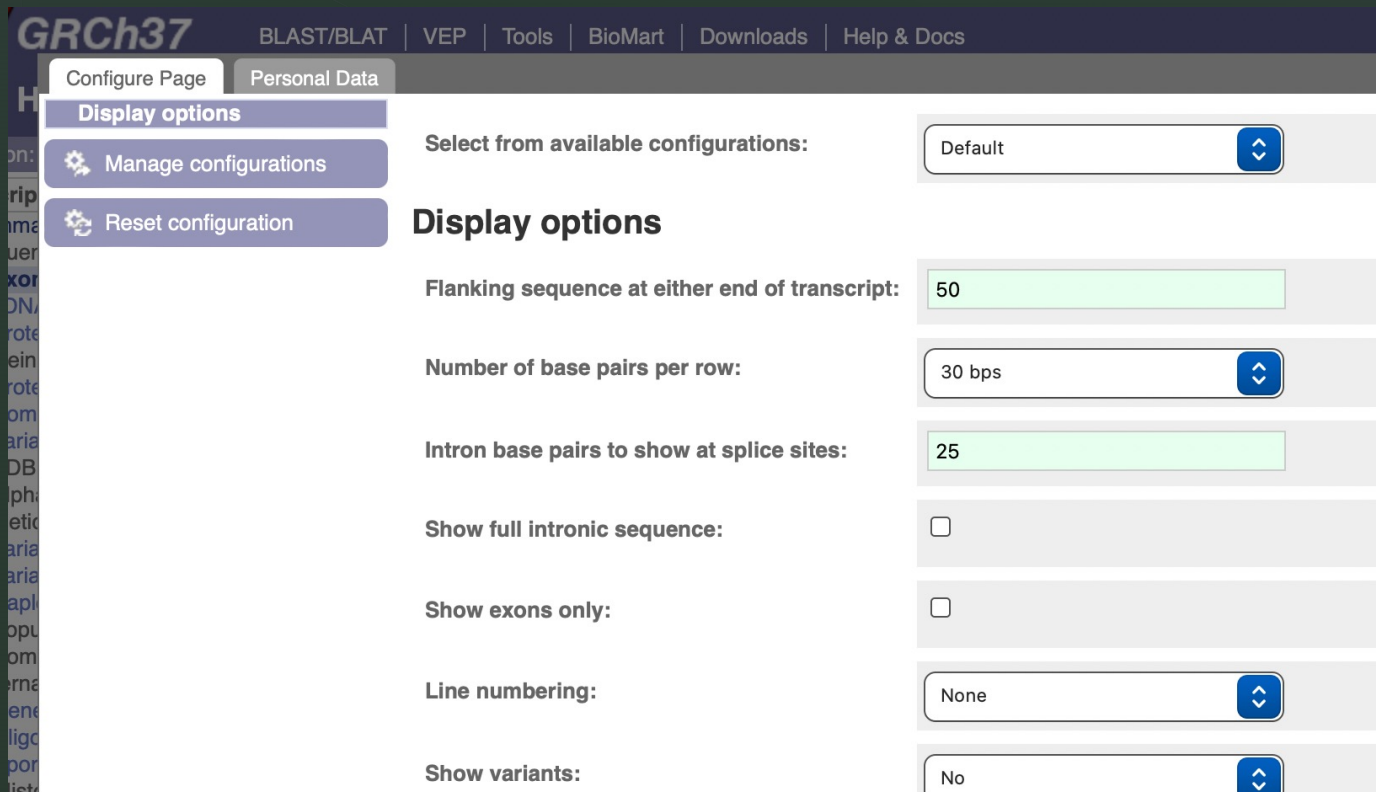
Show/hide columns (1 hidden)Filter

Transcript ID	Name	bp	Protein	Biotype	CCDS	UniProt Match	RefSeq	Flags
ENST00000544772.1	CHEK2-205	2560	322aa	Protein coding	CCDS58798	B7ZBF2 O96017	NM_001257387 NP_001244316	GENCODE basic
ENST00000382580.2	CHEK2-003	1971	586aa	Protein coding	CCDS33629	B7ZBF2 O96017	NM_001005735 NP_001005735	GENCODE basic
ENST00000405598.1	CHEK2-005	1959	543aa	Protein coding	CCDS13843	B7ZBF2 O96017	-	GENCODE basic

Let us look for a specific splice-site segment, AAGAGAGGC**A**gtaagtacca

Let us look at the following transcript, ENST00000544772

Let us look at the following mutation, . Here is my configuration.




The screenshot shows the 'Configure Page' for the GRCh37 VEP (Variant Effect Predictor) tool. The 'Personal Data' tab is selected. Under the 'Display options' section, there are three buttons: 'Display options', 'Manage configurations', and 'Reset configuration'. The 'Display options' section is expanded, showing the following settings:

- Select from available configurations:** Default (dropdown menu)
- Flanking sequence at either end of transcript:** 50 (text input)
- Number of base pairs per row:** 30 bps (dropdown menu)
- Intron base pairs to show at splice sites:** 25 (text input)
- Show full intronic sequence:** ☐
- Show exons only:** ☐
- Line numbering:** None (dropdown menu)
- Show variants:** No (dropdown menu)

Here is the segment of our interest; this occurs in
exon-7 end and spilling into the following intron
AAGAGAGGC[A/T]gtaagtacca

						ccatttctactctttttcttcttag
6	ENSE00003566977	29,115,473	29,115,383	-	2	91	TTTTTGTCCTTTTGTGATCTGACTGTAGATG ATCAGTCAGTTTATCCTAAGGCATTAAGAG ATGAATACATCATGTCAAAACTCTTGGAA G
	Intron 6-7	29,115,382	29,108,006			7,377	gtaaattatttttcttatataaatgc.....tcactatctttgtttttccctctag
7	ENSE00003670158	29,108,005	29,107,897	2	0	109	TGGTGCCTGTGGAGAGGTAAGCTGGCTTT CGAGAGGAAAACATGTAAGAAAGTAGCCAT AAAGATCATCAGCAAAAGGAAGTTTGCTAT TGGTTCAGCAAGAGAGGCA
	Intron 7-8	29,107,896	29,106,048			1,849	gtaagtaccaataaaaggctgatca.....aactctgttattctgtttatcaaag
8	ENSE00003566135	29,106,047	29,105,994	0	0	54	GACCCAGCTCTCAATGTTGAAACAGAAATA GAAATTTGAAAAAGCTAAATCAT
	Intron 8-9	29,105,993	29,099,555			6,439	gtaagtattattatgactcatcac.....cccttttttctccctcttcttag

Here is the submission page with details



mutation t@sting

[MutationTaster2021](#) has been released! See [changes](#).

Gene

Transcript

Position / snippet refers to

Alteration

ENST00000544772

☐ coding sequence (ORF)

☐ transcript (cDNA sequence)

☒ gene (genomic sequence)

all types by sequence

AAGAGAGGC[A/T]gtaagtacca

enter a few bases around your alteration

Format:

ACTGTC[A/T] GTGTF

ACTGTC[AG/T] GTGTF

ACTGTC[ACGT/-] GTGTF

ACTGTC[-/AA] GTGTF

A substituted by T

AG substituted by T

ACGT deleted

AA inserted

single base exchange by position

enter position


clear input

options

☒ show nucleotide alignment

Note the splice site changes summary.

Here is the output:



documentation

mutation t@sting

Prediction

Summary

analysed issue

name of alteration	no title
alteration (phys. location)	chr22:29107897T>AN/A show variant in all transcripts IGV
HGNC symbol	CHEK2
Ensembl transcript ID	ENST00000544772
Genbank transcript ID	NM_001257387
UniProt peptide	O96017
alteration type	single base exchange
alteration region	CDS
DNA changes	c.129A>T cDNA.1566A>T g.30514A>T
AA changes	no AA changes
position(s) of altered AA if AA alteration in CDS	N/A
frameshift	no

disease causing

- protein features (might be) affected
- splice site changes

analysis result

no title
chr22:29107897T>AN/A show variant in all transcripts IGV
CHEK2
ENST00000544772
NM_001257387
O96017
single base exchange
CDS
c.129A>T cDNA.1566A>T g.30514A>T
no AA changes
N/A
no

Model: *without_aae*, prob: 0.99999999926655 [\(explain\)](#)

[hyperlink](#)