Impact Analysis

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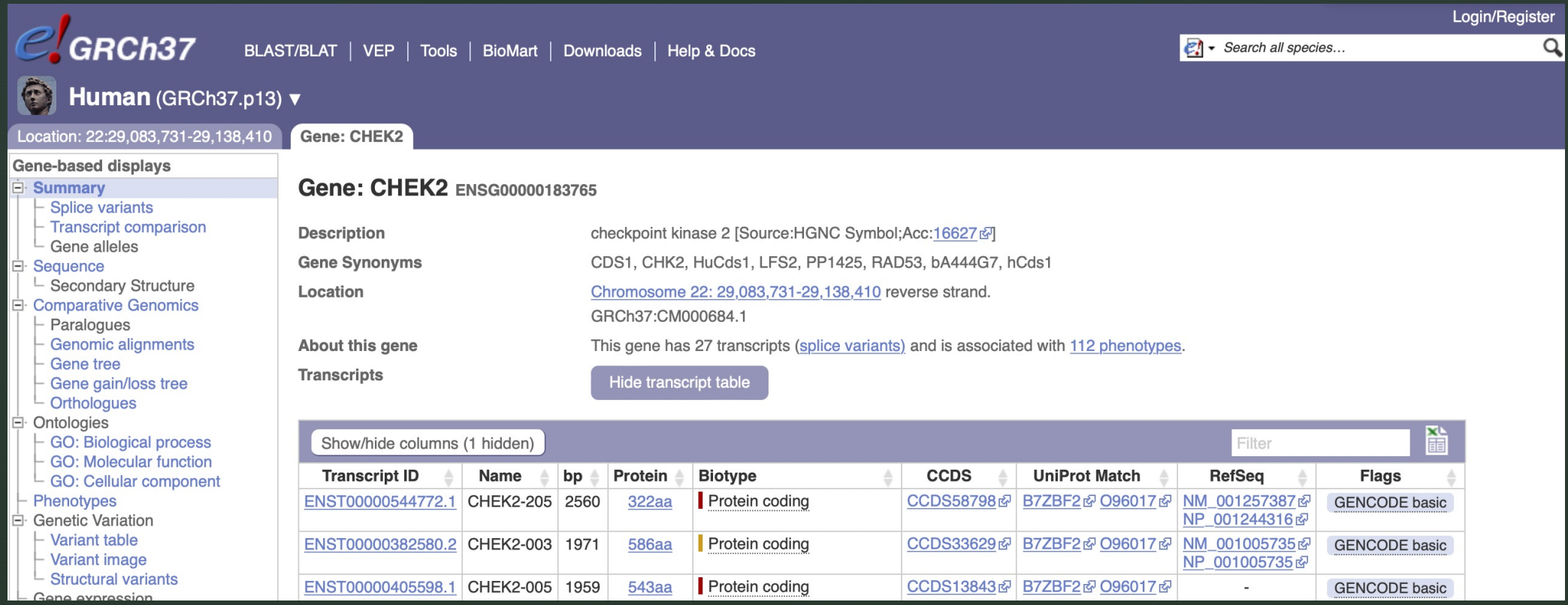
CHEK2 gene

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

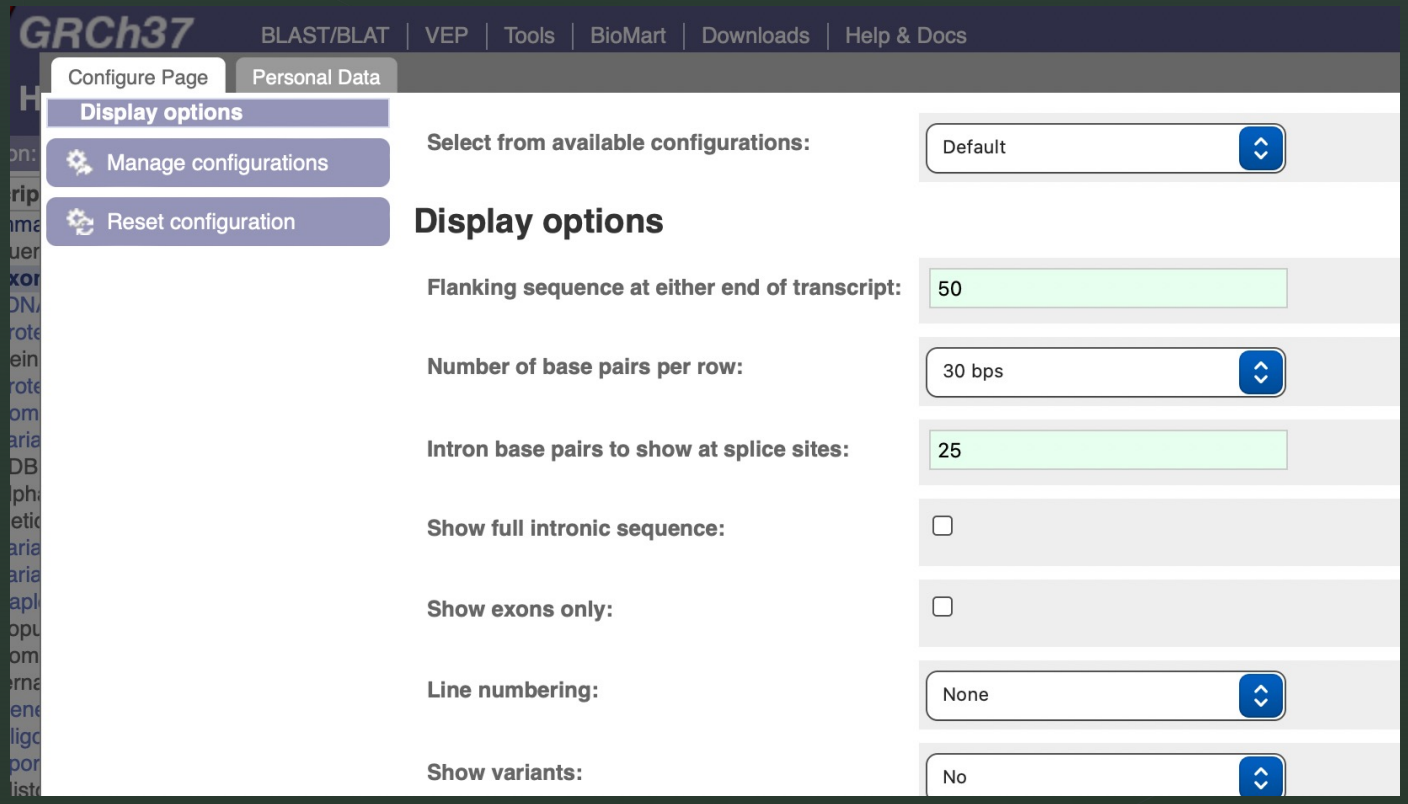
Note that MutationTaster had been built for GRCh37.



Let us look for a specific splice-site segment, AAGAGAGGCAgtaagtacca

Let us look at the following transcript, ENST00000544772

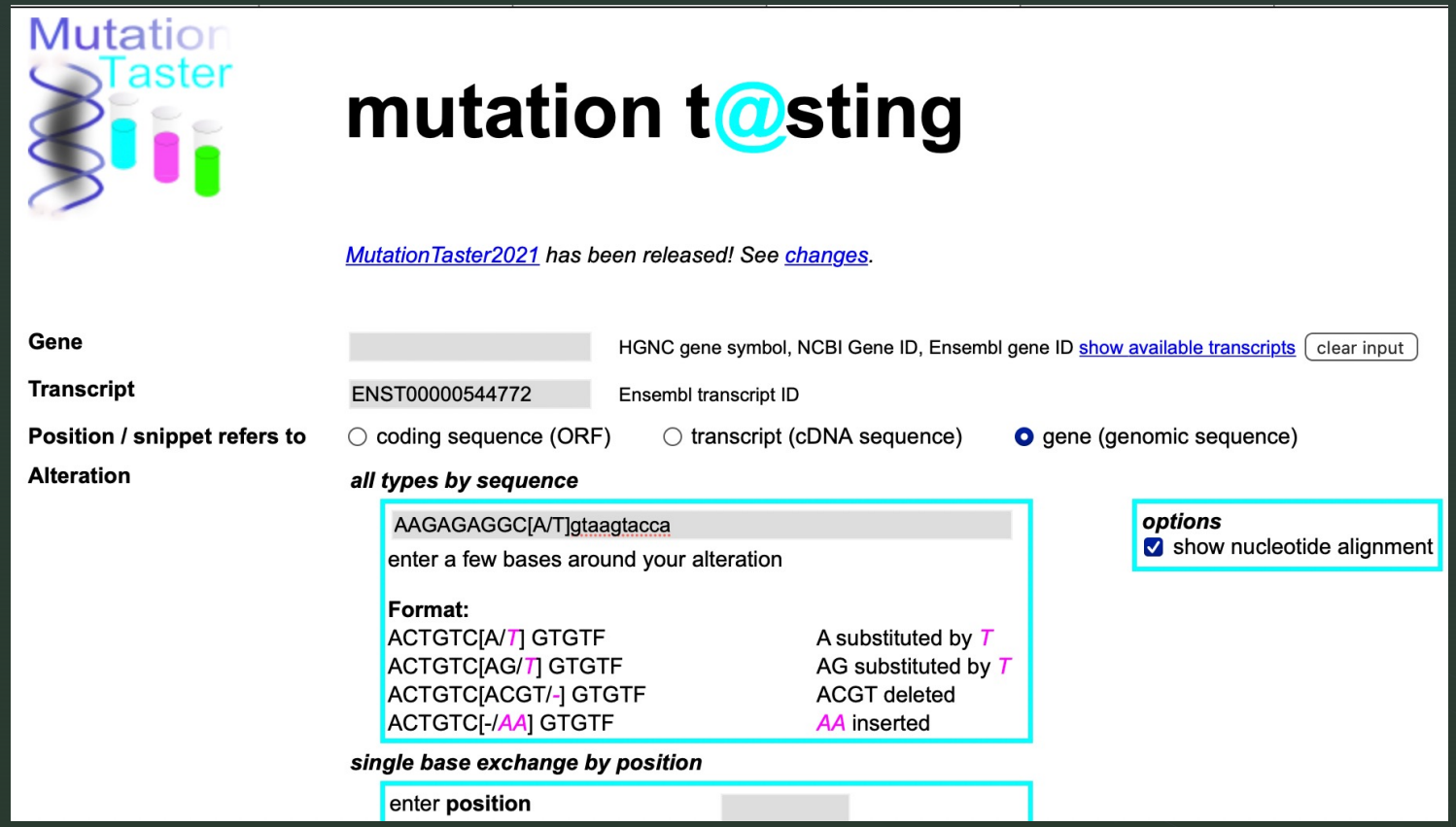
Let us look at the following mutation, . Here is my configuration.



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

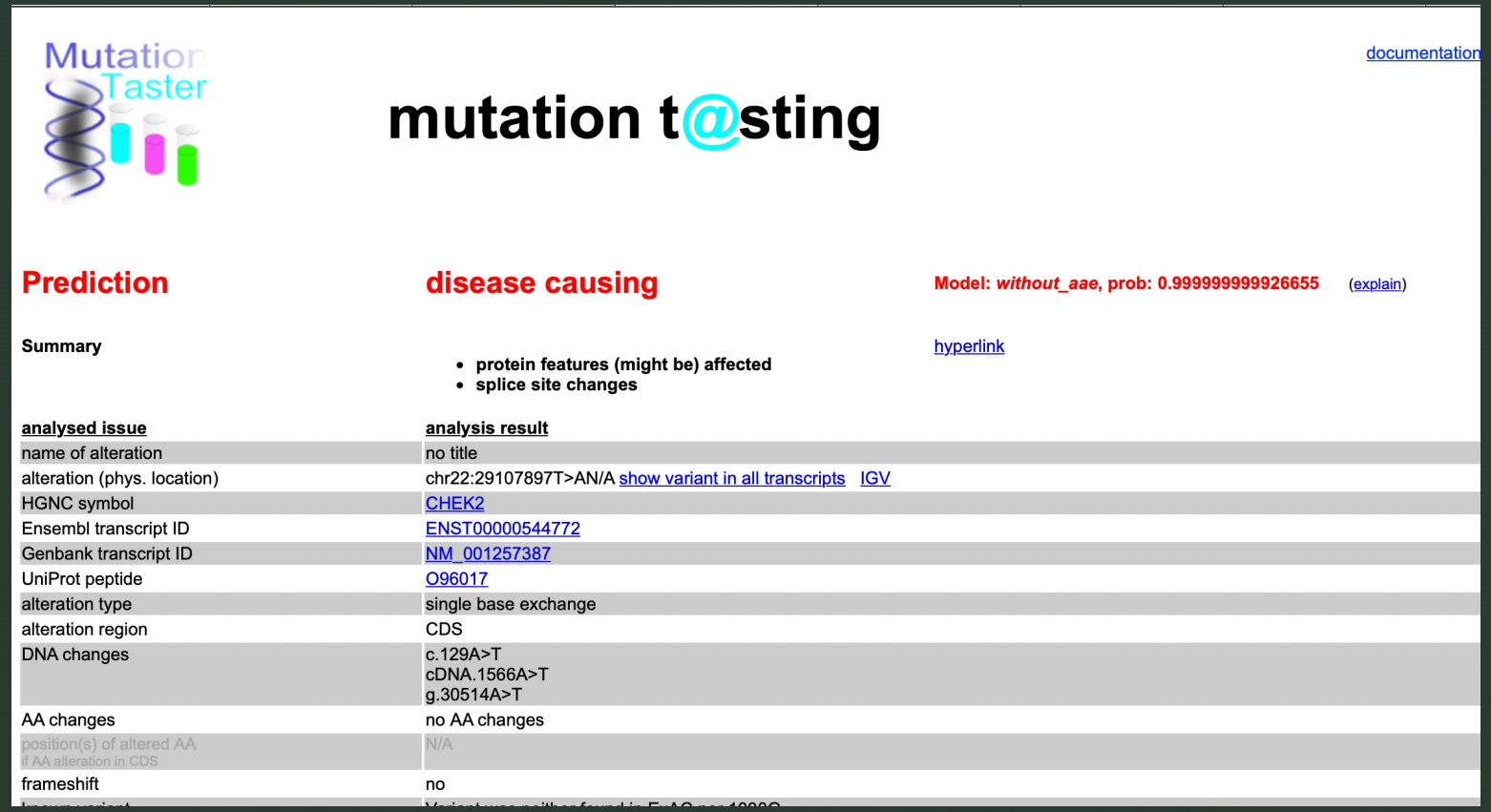
							.....ccatttctactcttttcttccttag
6	ENSE00003566977	29,115,473	29,115,383	-	2	91	TTTTTGTCTTTTTTGATCTGACTGTAGATG ATCAGTCAGTTTATCCTAAGGCATTAAGAG ATGAATACATCATGTCAAAAACTCTTGGAA G
	Intron 6-7	29,115,382	29,108,006			7,377	gtaaattattttcttatataaatgc..... .....tcactatctttgtttttccctctag
7	ENSE00003670158	29,108,005	29,107,897	2	0	109	TGGTGCCTGTGGAGAGGTAAAGCTGGCTTT 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 GAAATTTTGAAAAAGCTAAATCAT
	Intron 8-9	29,105,993	29,099,555			6,439	gtaagtattattatatgactcatac..... .....ccctttttttctcccccttcttcttag

Here is the submission page with details



Note the splice site changes summary.

Here is the output:



Mutation Taster

documentation

mutation t@sting

Prediction **disease causing** **Model: *without_aae*, prob: 0.999999999926655** (explain)

Summary hyperlink

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

analysed issue	analysis result
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