

## JAK2 (V617F/+) Cell line Engineering Summary

Genotype	rs #	Cosmic ID	Position			
			Chromosome Number	Chromosome	Gene	Transcript
JAK2 (V617F/+)	rs77375493	COSM12600	9q24	g.5073770G>T (GRCh37) g.5073770G>T (GRCh38)	g.93526G>T	c.1849G>T

Protein	Base change	Context	AA change
p.Val617Phe	GTC ⇒ TTC	TGGAGTATGT ( <b>G&gt;T</b> ) TCTGTGGAGA	V [Val] ⇒ F [Phe]

Reference sequence: Ensembl transcript: JAK2-001 ENST00000381652

ctatttcagggtggtatgggtcaagcctggttgactggcattattcatgattcctgtaccactcttgctctctctc  
actttgatctccatattccaggcttacacaggggttccctcagaacgttgatggcagttgcaggtccatataaag  
ggaccaagcacattgtatcctcatctatagtcctgaaagtaggagaaagtgcattctttattatggcagaga  
gaattttctgaaactattatggacaacagtcacaacaattctttgtactttttttttccttagTCTTCTTT  
GAAGCAGCAAGTATGATGAGCAAGCTTCTCACAAGCATTGGTTTAAATTATGGAGTATGT (**G>T**) TCTGT  
GGAGACGAGAgtaagtaaaactacaggctttctaattgcctttctcagagcatctgtttttgtttatatagaaaat  
tcagtttcaggatcacagctaggtgtcagtgtaaaactataatttaacaggagtttaagtatttttgaaactgaaa  
cactgt] [aggactattcagttatatctgtgaaaaaggaaagcaatgaagttaaagtagaaggttacaatgc  
ccaacaatagagtattatagtaaacaaatgtctataaacattttgtgttcatgatagcaaaagagattatggc  
aggttcaacataacattggaataactggccttttcagtacaaacttatctggaattatgaagacaaagcatat

Lowercase blue sequence indicates intron

Uppercase BLACK sequence indicates exon coding sequence

]] indicates the site of engineering

(G>T) indicates the V617F mutation

### Contact Horizon Diagnostics

Please note that primers designed upstream or across the engineering site will have a reduced efficiency and your assay may fail. Please contact us at [technical@horizondx.com](mailto:technical@horizondx.com) if you have any questions.

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