

Twitter Thread by Kevin McKernan



Kevin McKernan

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<https://t.co/LjpdIH0C7> is getting a good face-lift.

Samples now have variant tables with annotation on their impact to the gene.

There is also an IGV integration. You need IGV installed locally but the Start|Jump links will open up you BAM file in IGV.

VARIANTS (THCAS, CBDAS, and CBCAS)								
Show	5	▼	entries	Search:				
Gene ↕	HGVS.c ↕	HGVS.p ↕	Annotation ↕	Annotation Impact ↕	Contig ↕	Contig Pos ↕	Ref/Alt ↕	Var Freq ↕
THCAS	c.1229G>A	p.Gly410Glu	missense variant	MODERATE	contig741	4416599 IGV:Start Jump	C/T	NGS : 0.039 C90 : 0.000
THCAS	c.373G>C	p.Val125Leu	missense variant	MODERATE	contig741	4417455 IGV:Start Jump	C/G	NGS : 0.050 C90 : 0.000
THCAS	c.749C>A	p.Ala250Asp	missense variant	MODERATE	contig741	4417079 IGV:Start Jump	G/T	NGS : 0.127 C90 : 0.632
Showing 1 to 3 of 3 entries							Previous	1 Next

You must have IGV open and then click the start link.

The sample's BAM will show up for viewing in IGV.

Once open, you can use the jump link to move swiftly around to each variant. This is a variant in THCAS.

This sample has a disruptive in frame deletion on a late flowering gene. And it's very rare in the population.

Similar to PHL: Protein PHYTOCHROME-DEPENDENT LATE-FLOWERING (Arabidopsis thaliana OX=3702)

VARIANTS (SELECT GENES OF INTEREST)

Search:

Gene	HGVS.c	HGVS.p	Annotation	Annotation Impact
PHL-1	c.3003_3020delACAGCAACAGCAGCAGCA	p.Gln1002_Gln1007del	disruptive inframe deletion	MODERATE
ELF3	c.1803_1805delTCA	p.His601del	disruptive inframe deletion	MODERATE
PIE1-1	c.1124_1153dupATGTGGGTGAACCAACCCAGATGGAGGATA	p.Asn375_Asp384dup	disruptive inframe insertion	MODERATE
PHL-2	c.2903_2905dupGCA	p.Ser968dup	disruptive inframe insertion	MODERATE
FT	c.35_43dupATAATAATA	p.Asn12_Asn14dup	disruptive inframe insertion	MODERATE
PHL-2	c.3552delG	p.Lys1185fs	frameshift variant	HIGH
PHL-2	c.3556_3557delAA	p.Lys1186fs	frameshift variant	HIGH
AAE1-2	c.948_949insA	p.Asp317fs	frameshift variant	HIGH
AAE1-2	c.952delC	p.Gln318fs	frameshift variant	HIGH
GPPc1	c.845_848delAAAAG	p.Glu282fs	frameshift	HIGH

Looks to be heterozygous....
But its never this simple in cannabis.

Let's look at what other damaging variations it has.

