<u>BUZZ CHRONICLES</u> > <u>TECH</u> <u>Saved by @Mollyycolllinss</u> See On Twitter

Twitter Thread by Kevin McKernan



<u>Kevin McKernan</u> @Kevin_McKernan



https://t.co/LjpdIHa0C7 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.

Show 5 v entries Search:								
Gene ↓↑	HGVS.c ↓↑	HGVS.p ↓†	Annotation 🕼	Annotation Impact 1	Contig 🔱	Contig Pos 🎝	Ref/Alt ↓↑	Var Freq ↓
THCAS	c.1229G>A	p.Gly410Glu	missense variant	MODERATE	contig741	4416599 IGV:Start Jump	C/T	NGS:0.03
THCAS	c.373G>C	p.Val125Leu	missense variant	MODERATE	contig741	4417455 IGV:Start Jump	C/G	NGS:0.05
THCAS	c.749C>A	p.Ala250Asp	missense variant	MODERATE	contig741	4417079 IGV:Start Jump	G/T	NGS:0.12 C90:0.63
howing 1 t	o 3 of 3 entrie	25					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.



You can jump to other variants in the same gene. In this case we have one non-synonymous variant in phase with a synonymous variant in THCAS.



Cannabinoid synthase genes are cute but some folks can answer those questions with HPLC. How about flowering genes?

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

VARIANTS (SELECT GENES OF INTEREST)							
Similar to P	nilar to PHL: Protein Search:						
FLOWERIN OX=3702)	G (Arabidopsis	thaliana HGVS.c	HGVS.p ↓î	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		
	GPPs1	C 845 848delaaaG	n Glu282fs	frameshift	HIGH		

Looks to be heterozygous....

But its never this simple in cannabis.

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



contia1439:1.486.796 5 tracks loaded

7 Damaging inframe deletions or insertions (indels) in Early and Late flowering genes.

	Cono	ucve 1	HGVS.p	Annotation 🏨	Annotation Impact
Similar to El FLOWERING OX=3702)	F3: Protein E 3 (Arabidops	ARLY is thaliana 20delACAGCAACAGCAGCAGCA	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

When it comes to Indels, IGV can be very helpful to inspect the read frequency and if there appears to be any read mismapping with short reads. They all look real and diploid.



Here is another one in AAE1-2. This gene is involved in making the pre-cursors to cannabinoid synthesis.

It is not just a simple presence or absence of the cannabinoid synthase genes. One has to pay close attention to variants up stream in the pathway as well.



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