## Twitter Thread by Kevin McKernan





https://t.co/LjpdlHa0C7 is getting a good face-lift.

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

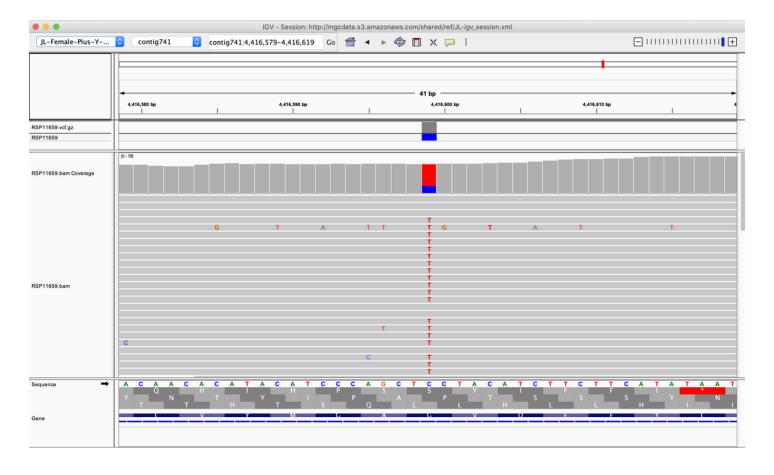
There is also an IGV integration. You need IGV installed locally but the Start|Jump links will open up you BAM file in IGV.

show 5 ventries Search						n: [		
Gene ↓↑	HGVS.c ↓↑	HGVS.p ↓↑	Annotation 🕸	Annotation Impact J↑	Contig ↓↑	Contig Pos I↑	Ref/Alt 🎼	Var Freq ↓
THCAS	c.1229G>A	p.Gly410Glu	missense variant	MODERATE	contig741	4416599 IGV:Start Jump	С/Т	NGS:0.039
THCAS	c.373G>C	p.Val125Leu	missense variant	MODERATE	contig741	4417455 IGV:Start Jump	C/G	NGS:0.050
THCAS	c.749C>A	p.Ala250Asp	missense variant	MODERATE	contig741	4417079 IGV:Start Jump	G/T	NGS:0.12

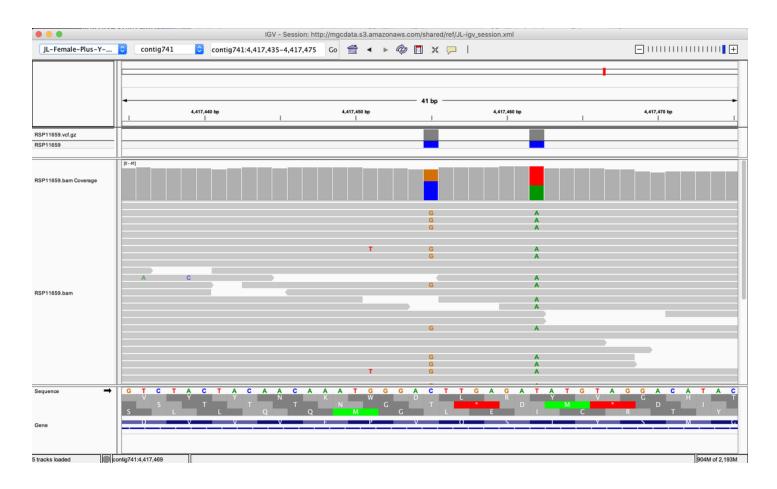
You must have IGV open and then click the start link.

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?

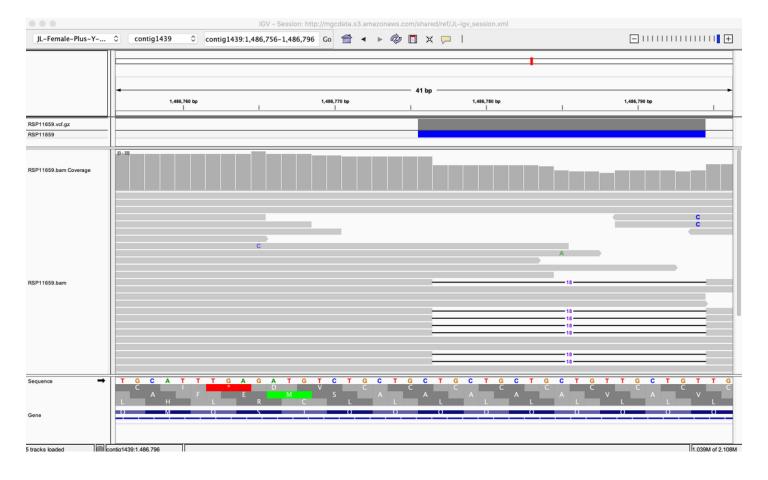
This sample has a disruptive in frame deletion on a late flowering gene. And it's very rare in the population.

PHL: Protein ROME-DEPEND	ENT LATE-		Search:		
(NG (Arabidopsis	thaliana 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_43dupATAATA	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 848delAAG	n Glu282fs	frameshift	нісн	

Looks to be heterozygous....

But its never this simple in cannabis.

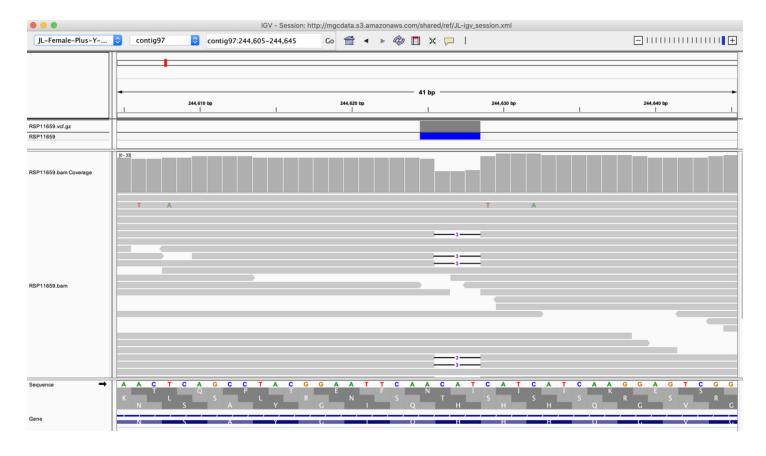
Let's look at what other damaging variations it has.



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

Co	no It u	11	HGVS.p ↓↑	Annotation ↓≟	Annotation Impact	
	ar to ELF3: Protein EARLY WERING 3 (Arabidopsis thaliana 3702)  20delACAGCAACAGCAGCA			disruptive inframe deletion	MODERATE	
ELF	F <u>3</u> c.	1803_1805delTCA	p.His601del	disruptive inframe deletion	MODERATE	
PIE	:1-1 c.	1124_1153dupATGTGGGTGAACCAACCCAGATGGAGGATA	p.Asn375_Asp384dup	disruptive inframe insertion	MODERATE	
PH	L-2 c.	2903_2905dupGCA	p.Ser968dup	disruptive inframe insertion	MODERATE	
FT	c.	35_43dupATAATAATA	p.Asn12_Asn14dup	disruptive inframe insertion	MODERATE	
PH	L-2 c.	3552delG	p.Lys1185fs	frameshift variant	HIGH	
PH	L-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.

