Twitter Thread by Kevin McKernan

Kevin McKernan
@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.

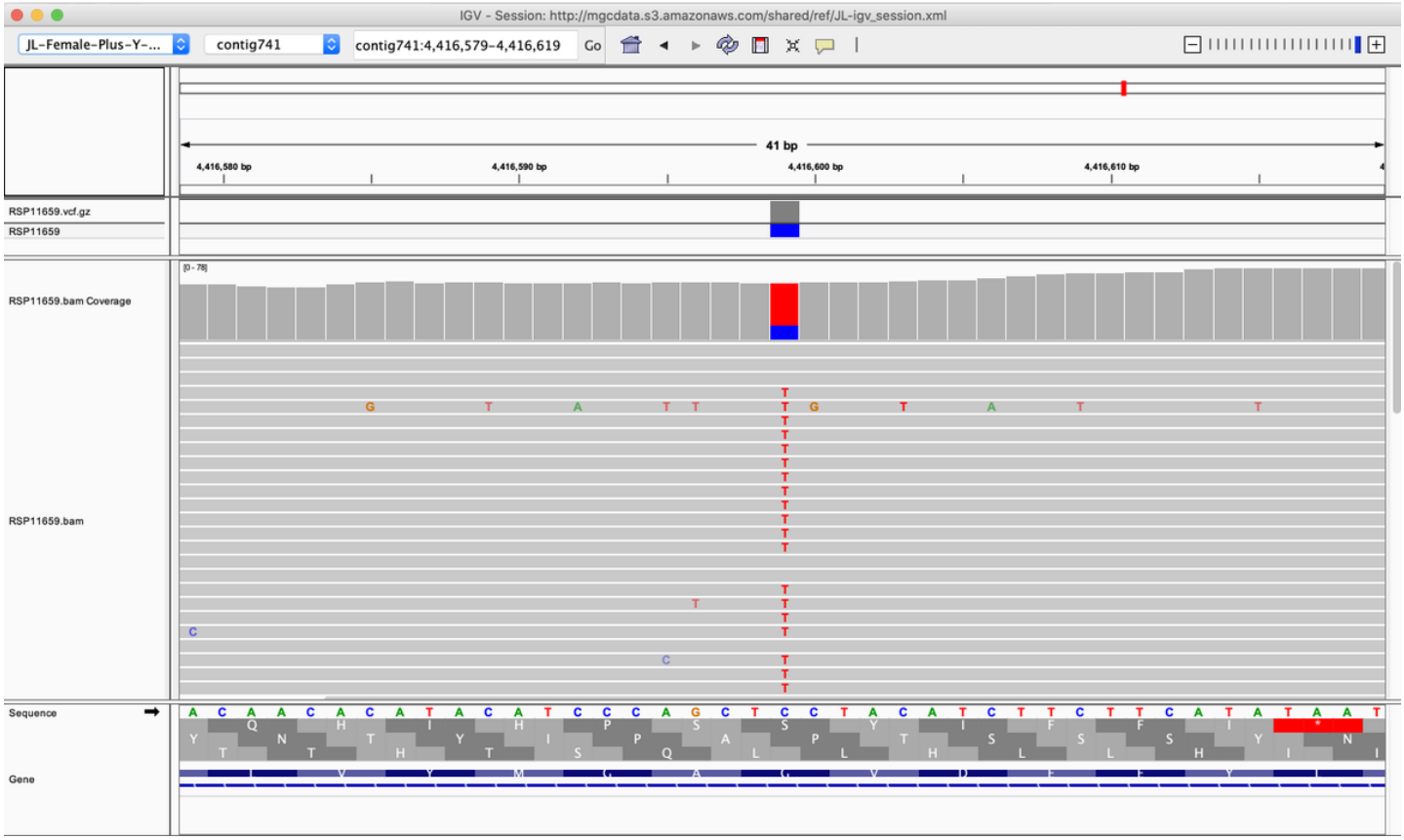
VARIANTS (THCAS, CBDAS, and CBCAS)

Show 5 entries Search:

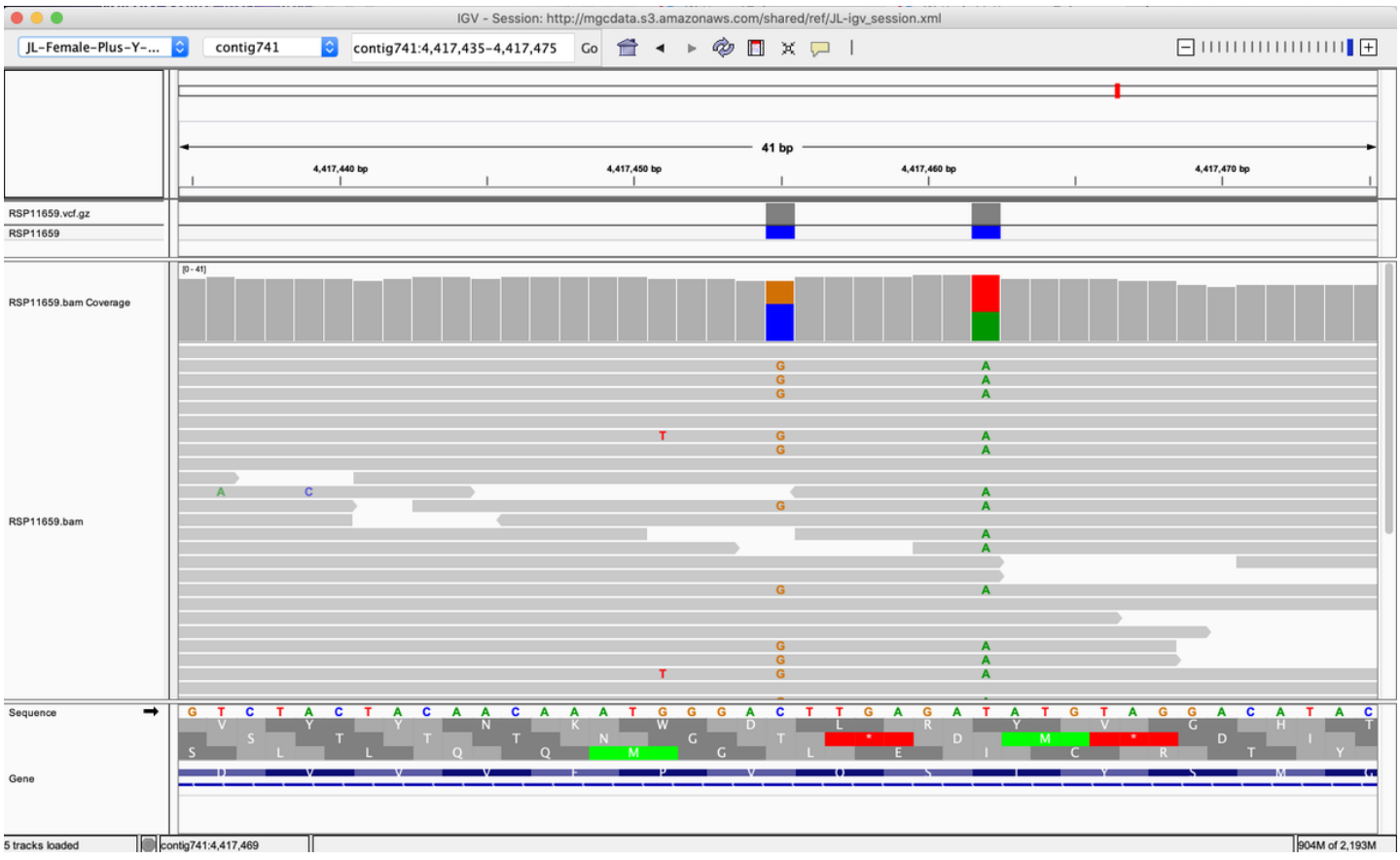
Gene	HGVS.c	HGVS.p	Annotation	Annotation Impact	Contig	Contig Pos	Ref/Alt	Var Freq
THCAS	c.1229G>A	p.Gly410Glu	missense variant	MODERATE	contig741	4416599 IGV:Start|Jump	C/T	NGS:0.039 C90:0.000
THCAS	c.373G>C	p.Val125Leu	missense variant	MODERATE	contig741	4417455 IGV:Start|Jump	C/G	NGS:0.050 C90:0.000
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.

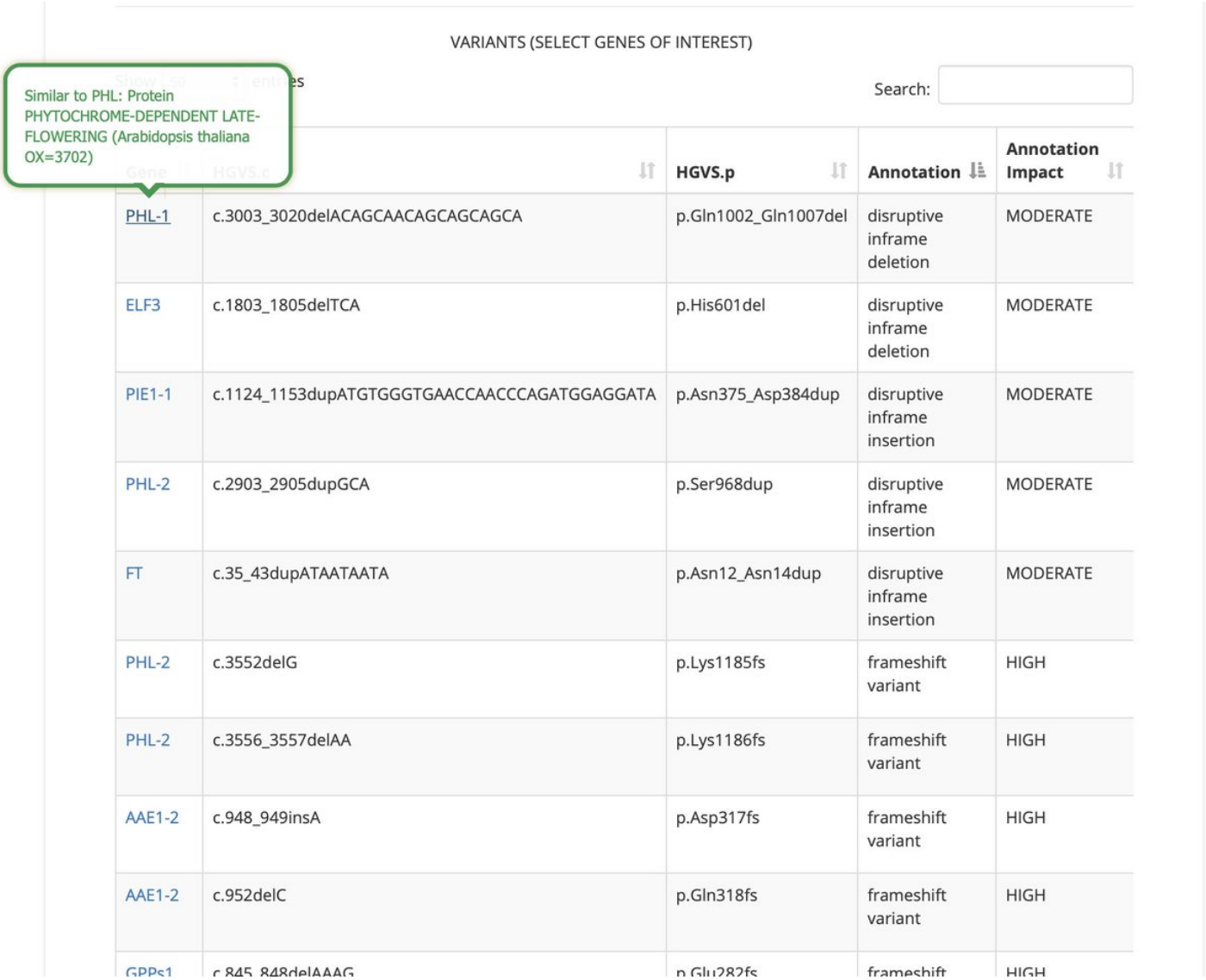


You can jump to other variants in the same gene. In this case we have one non-synonymous variant in phase with a synonymous variant in THCAS.



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

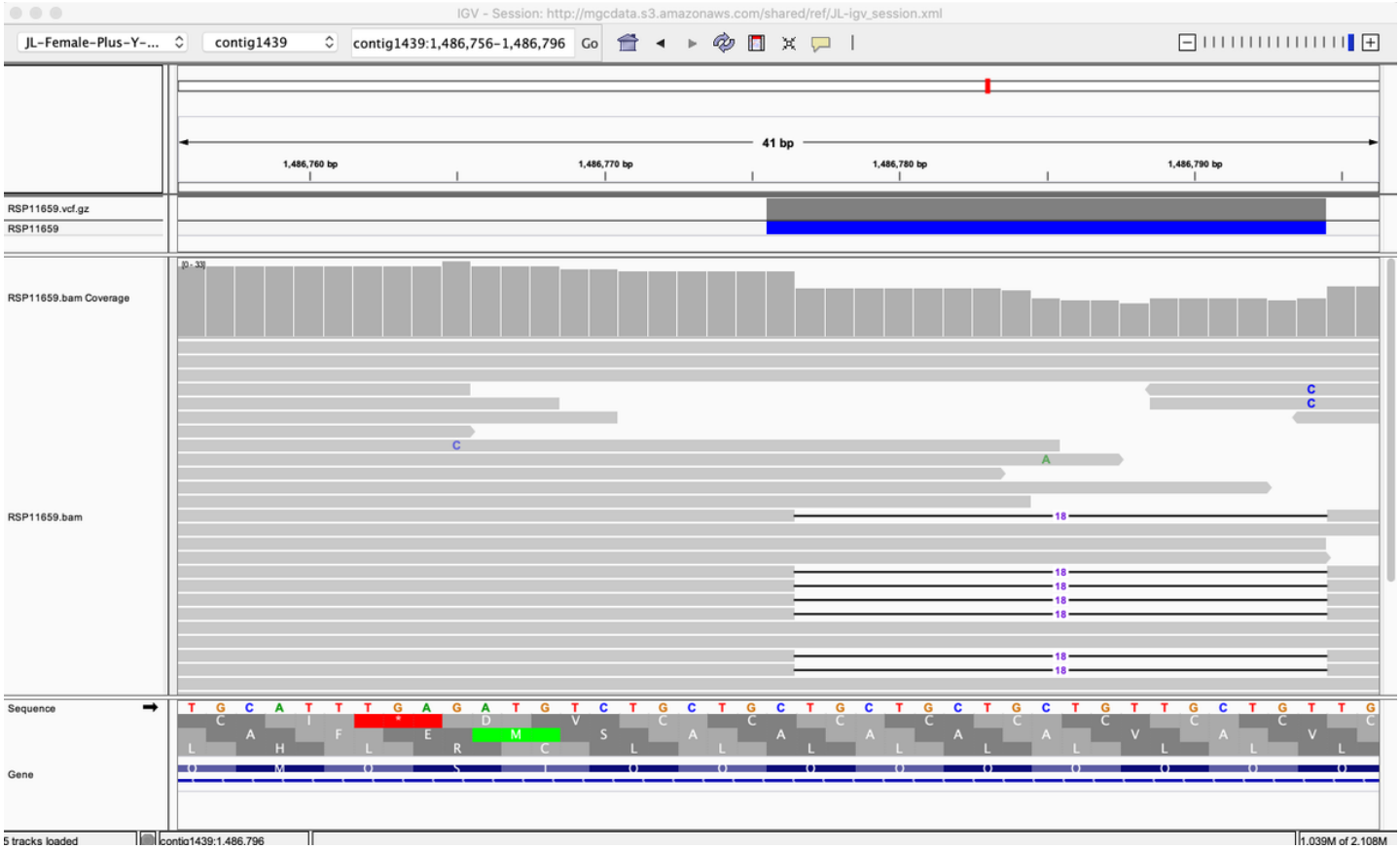
This sample has a disruptive in frame deletion on a late flowering gene. And it's very rare in the population.



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
GPPs1	c.845_848delAAAG	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.

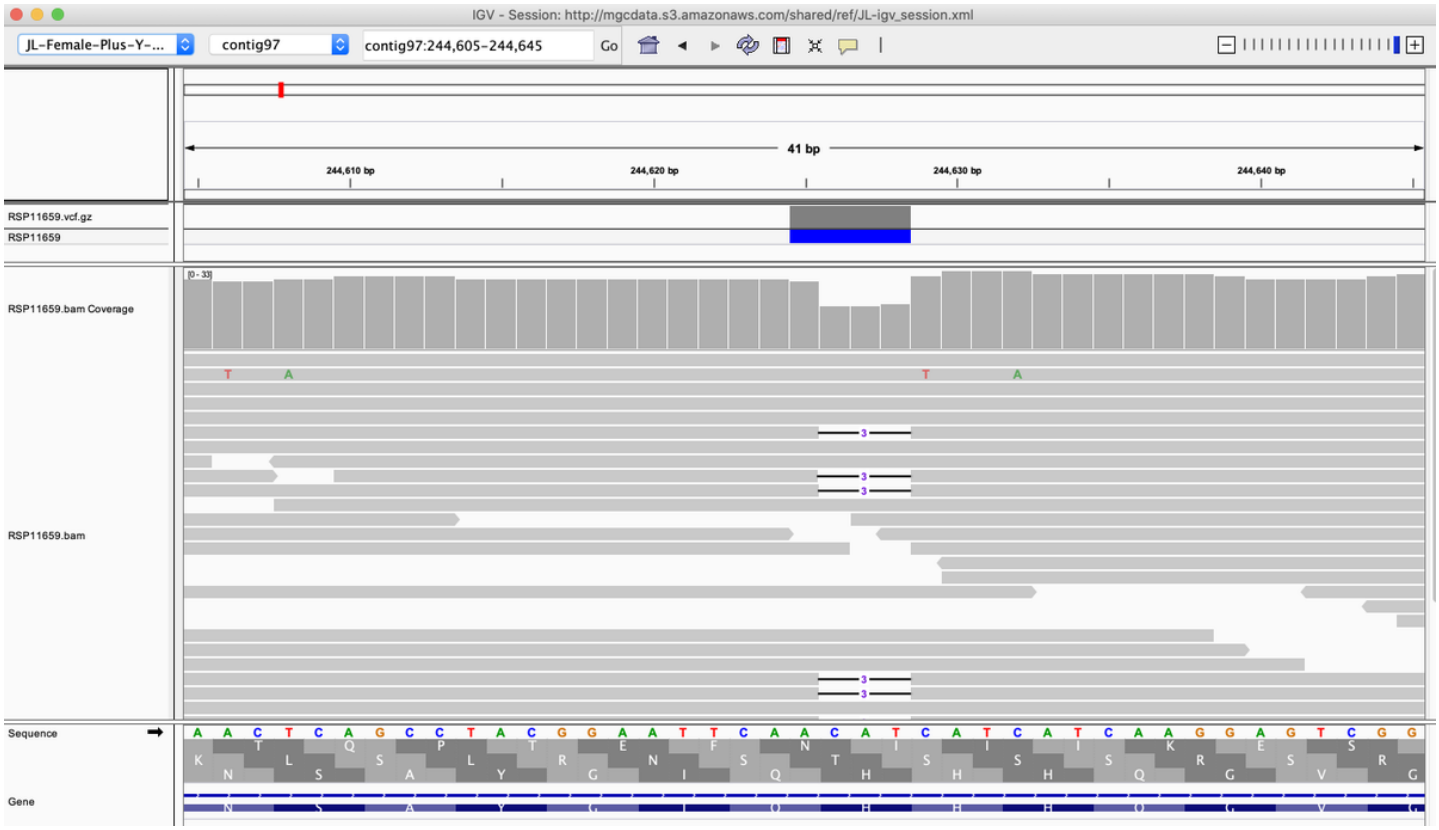


7 Damaging inframe deletions or insertions (indels) in Early and Late flowering genes.

Similar to ELF3: Protein EARLY FLOWERING 3 (Arabidopsis thaliana OX=3702)

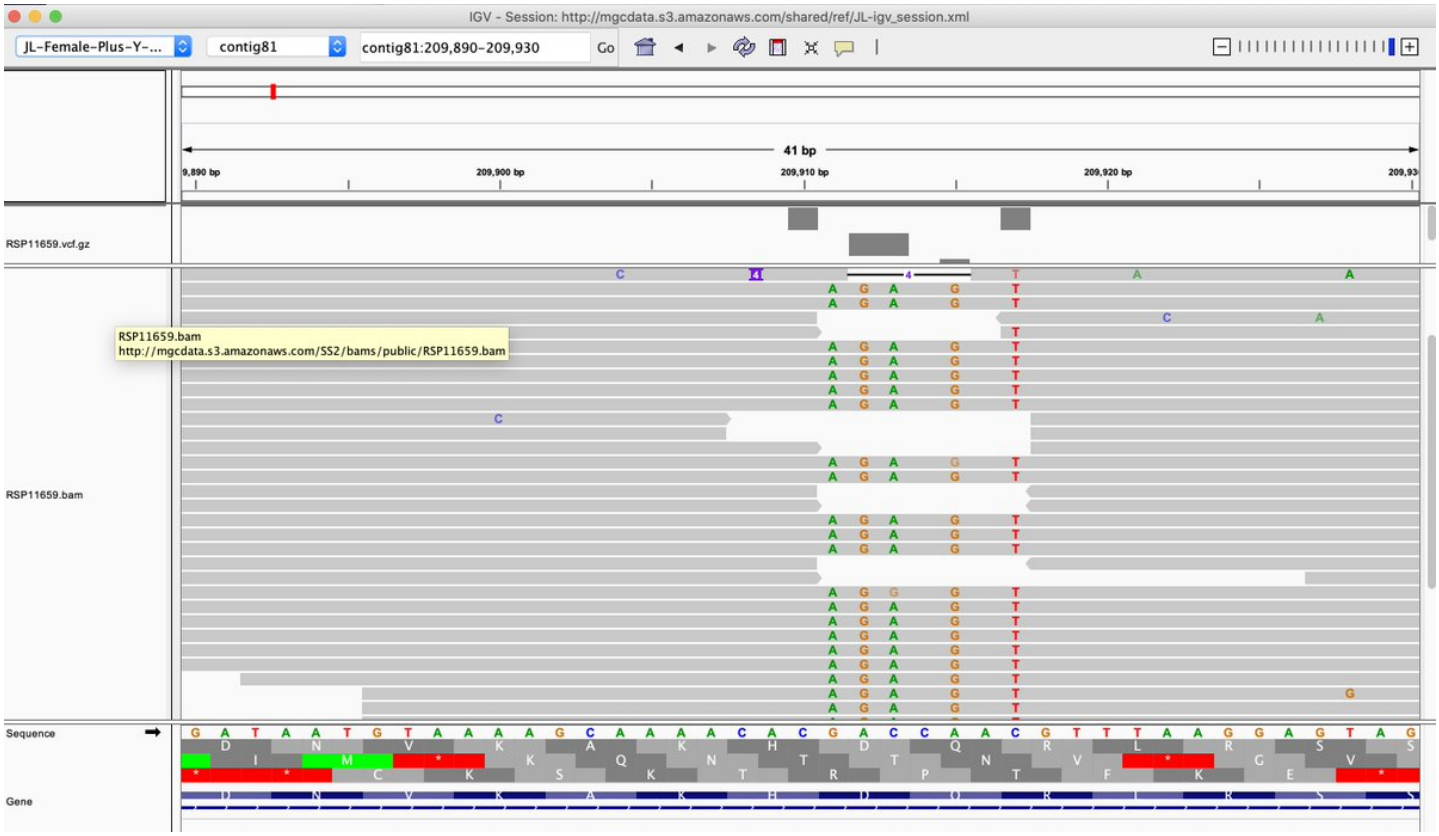
Gene	HGVS.c	HGVS.p	Annotation	Annotation Impact
	20delACAGCAACAGCAGCAGCA	p.Gln1002_Gln1007del	disruptive inframe deletion	MODERATE
ELF3	c.1803_1805delTCA	p.His601del	disruptive inframe deletion	MODERATE
PIE1-1	c.1124_1153dupATGTGGGTGAACCAACCCAGATGGAGGATA	p.Asn375_Asp384dup	disruptive inframe insertion	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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