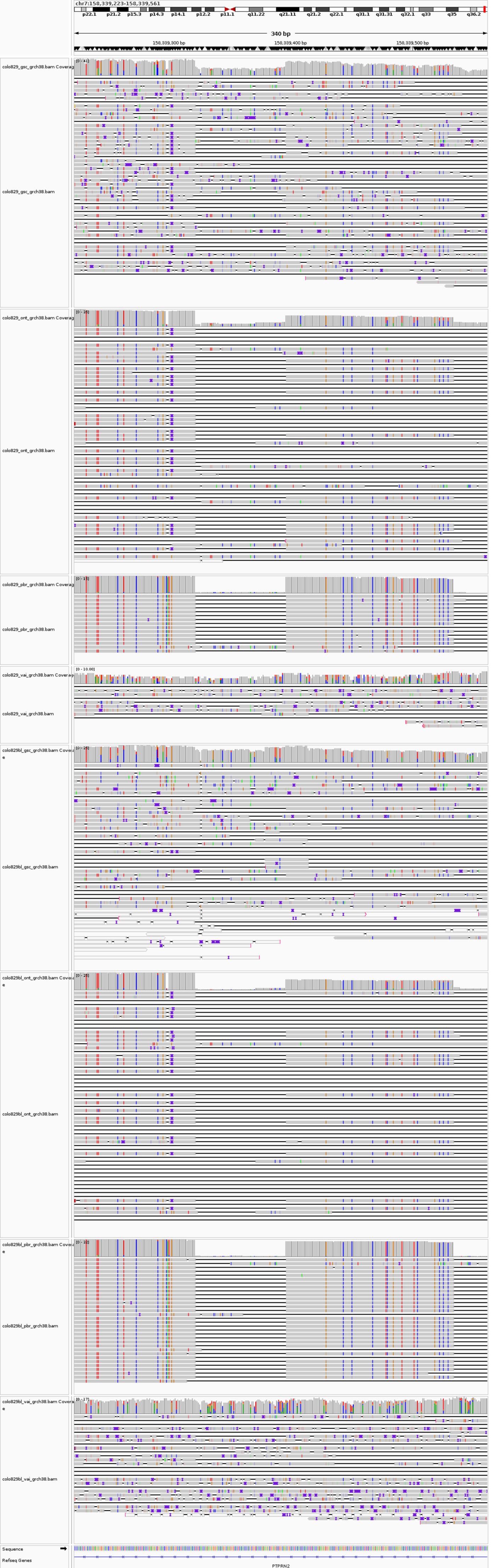


Supplementary Figure 1: IGV screenshot of the Structural Variants (Svs) labeled as False Positives (FP) according to the Valle-Inclan benchmark

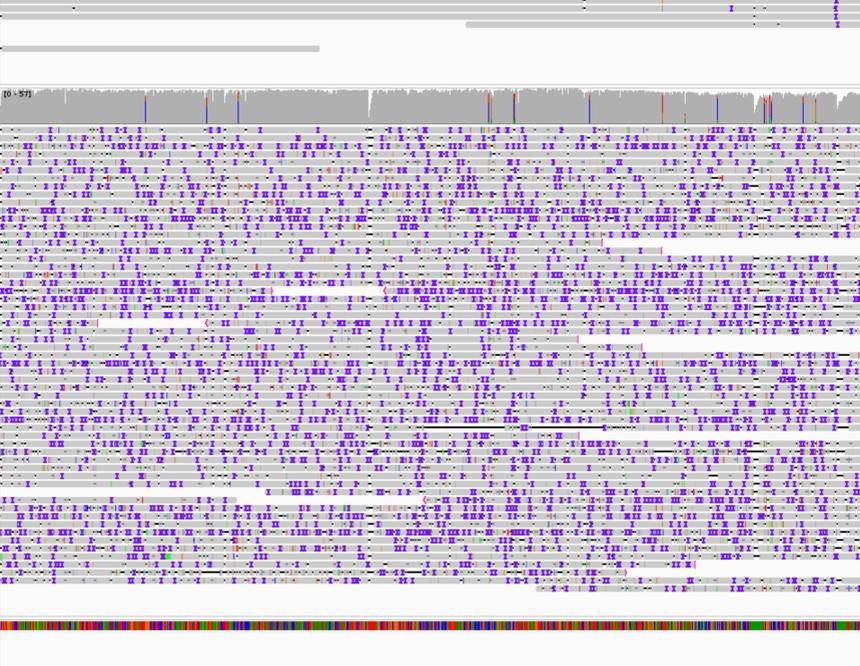
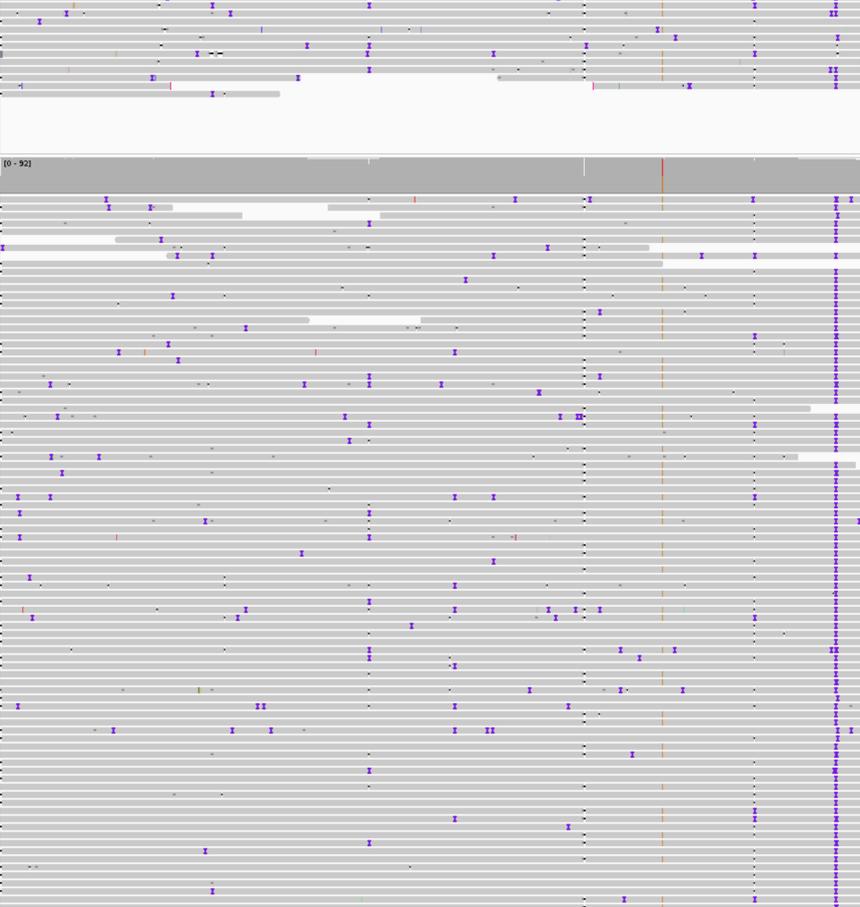
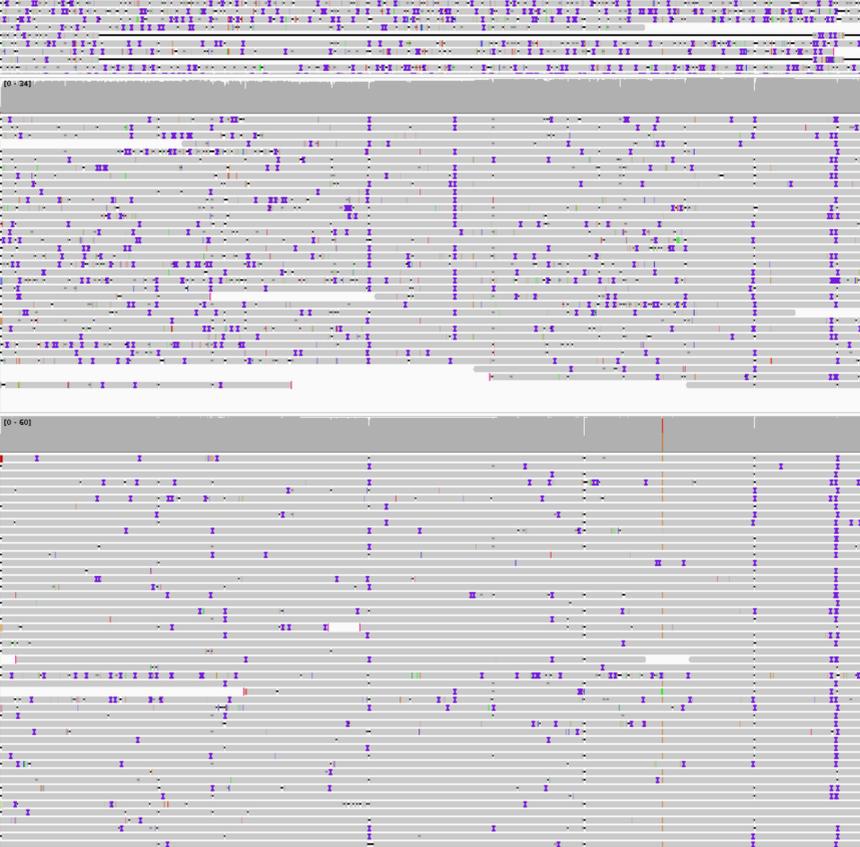
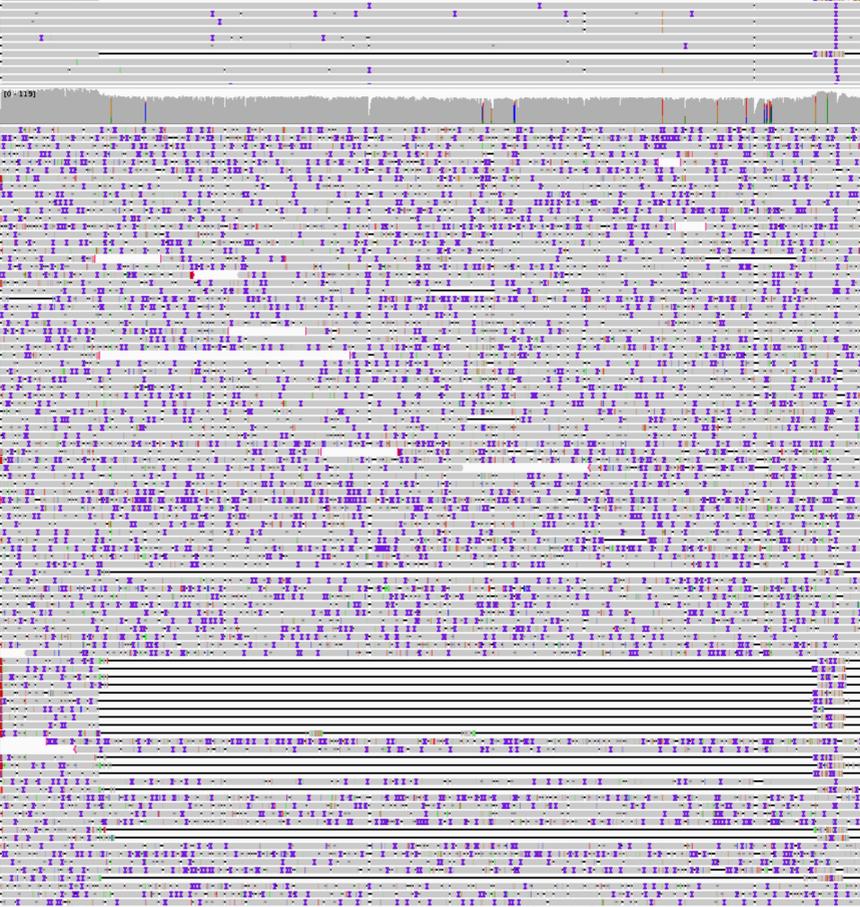
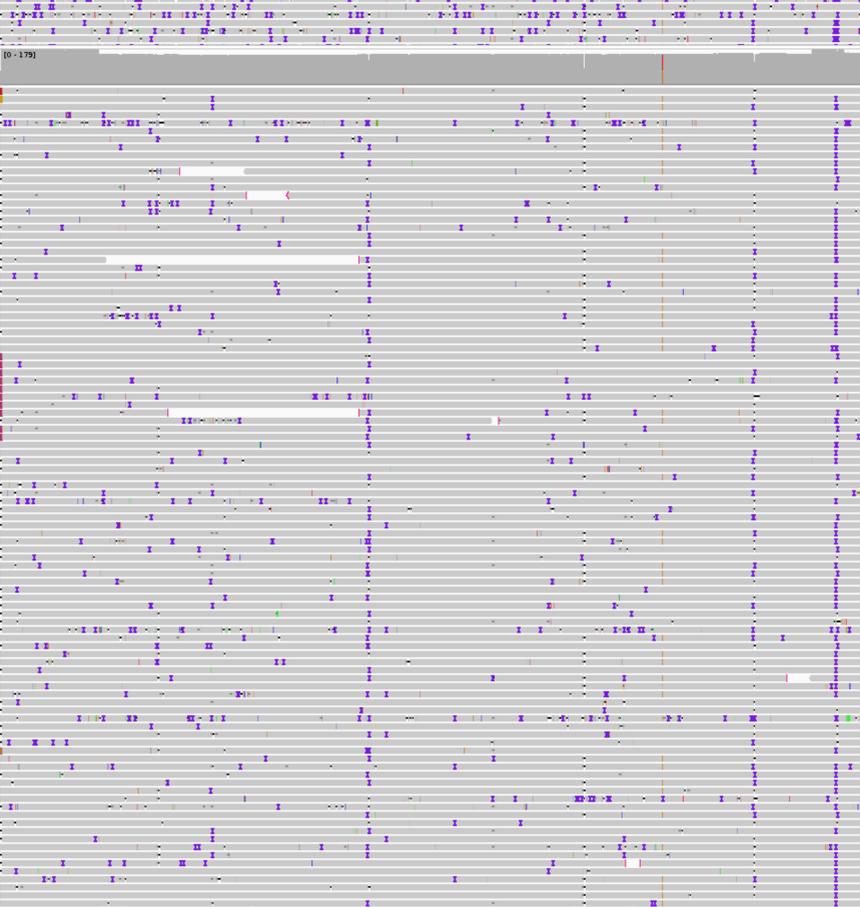
- A) An INS (SVLEN=69) located in chr7:5972497 is not a FP
- B) A DEL (SVLEN=138) located in chr7:158339323 is an actual FP
- C) An INS (SVLEN=11099) located in chr1:236097134 is not a FP
- D) An INS (SVLEN=2501) located in chr14:34545584 is not a FP
- E) A DEL (SVLEN=1419) located in chr15:23467473 is not a FP
- F) A BND located in chr18:9868620 with CHR2=20:38645825 is not a FP (CHR1)
- G) A BND located in chr18:9868620 with CHR2=20:38645825 is not a FP (CHR2)
- H) A BND located in chr19:17286003 with CHR2=20:38646024 is not a FP (CHR1)
- I) A BND located in chr19:17286003 with CHR2=20:38646024 is not a FP (CHR2)







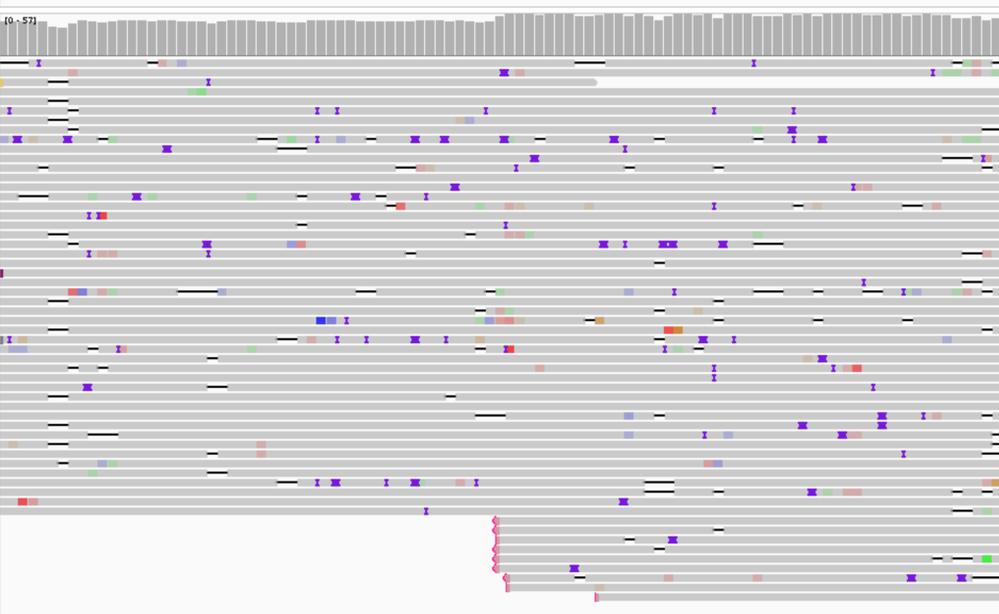




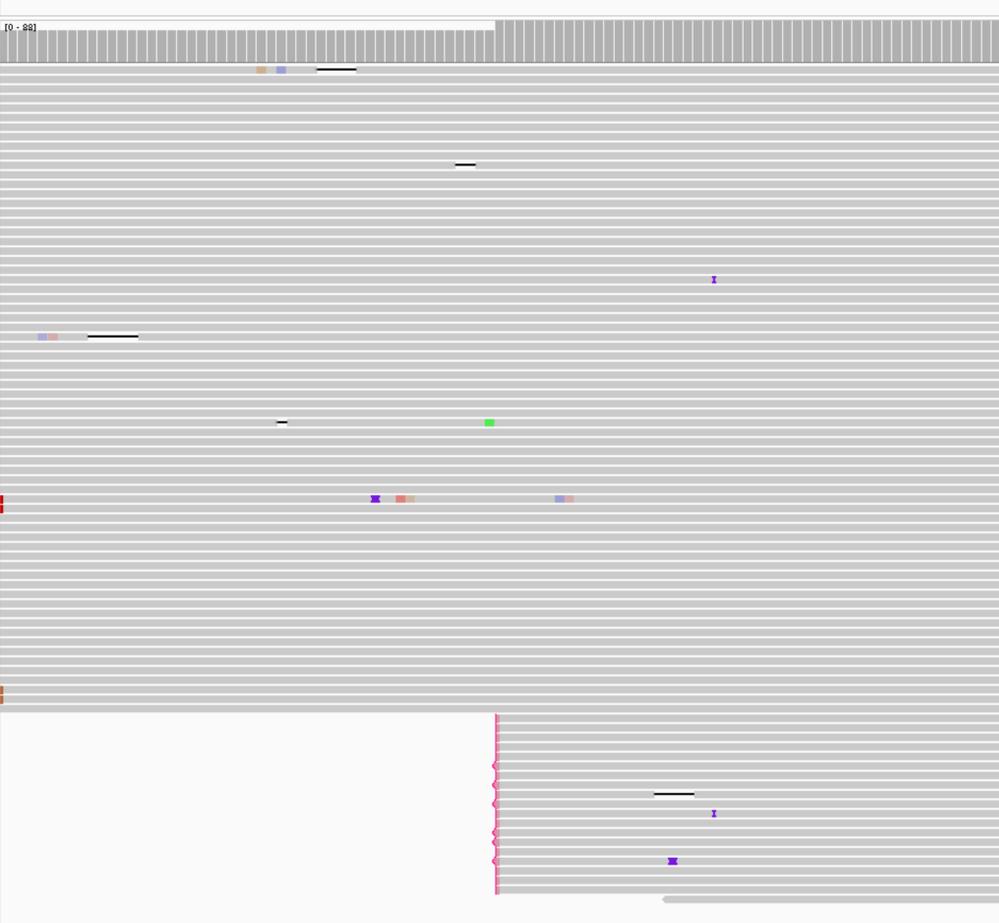
chr18:9,868,570-9,868,670  
p11.31 p11.22 p11.1 q11.2 q12.1 q12.2 q12.3 q21.1 q21.2 q21.31 q21.33 q22.1 q22.2 q23

102 bp  
9,868,580 bp 9,868,600 bp 9,868,620 bp 9,868,640 bp 9,868,660 bp

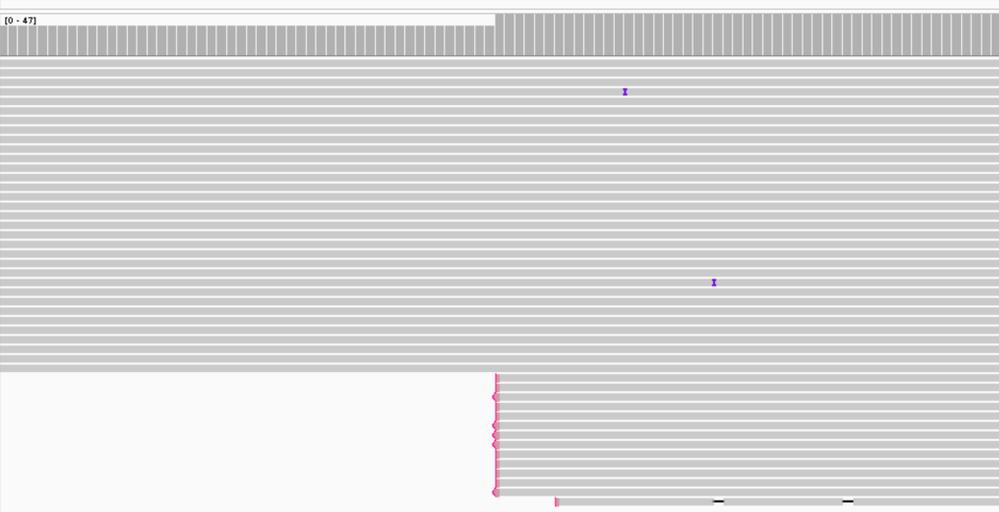
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colo829\_gsc\_grch38.bam



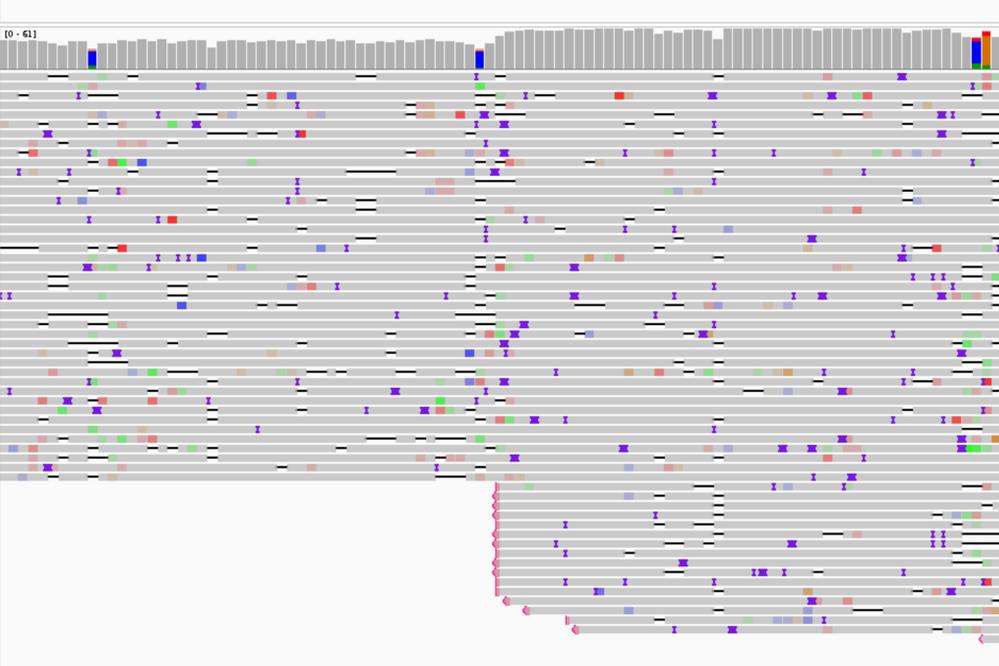
colo829\_ont\_grch38.bam Coverage  
colo829\_ont\_grch38.bam



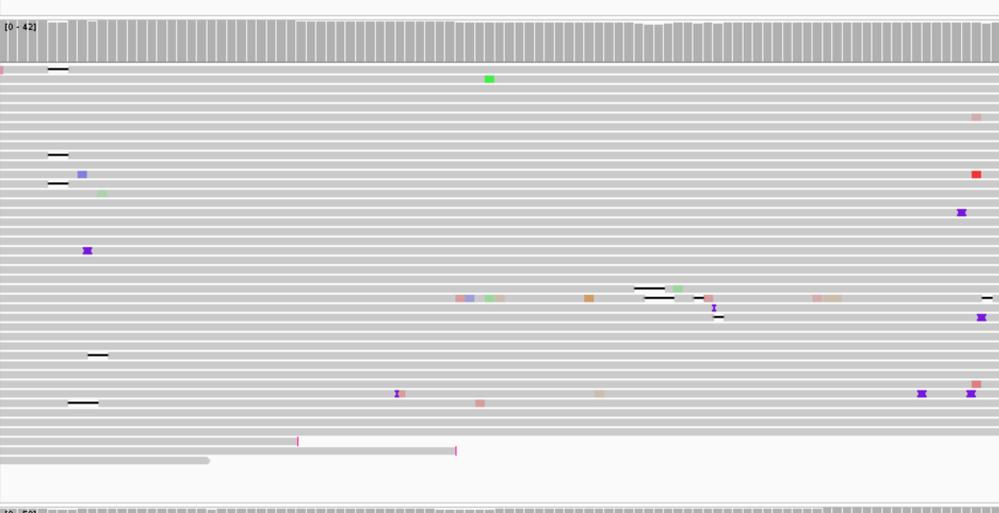
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colo829\_pbr\_grch38.bam



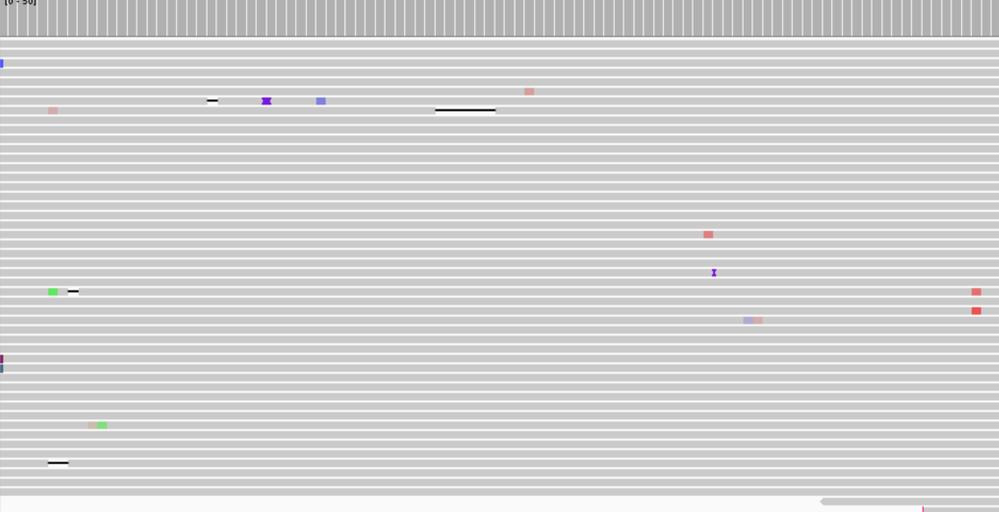
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colo829\_vai\_grch38.bam



colo829bl\_gsc\_grch38.bam Coverage  
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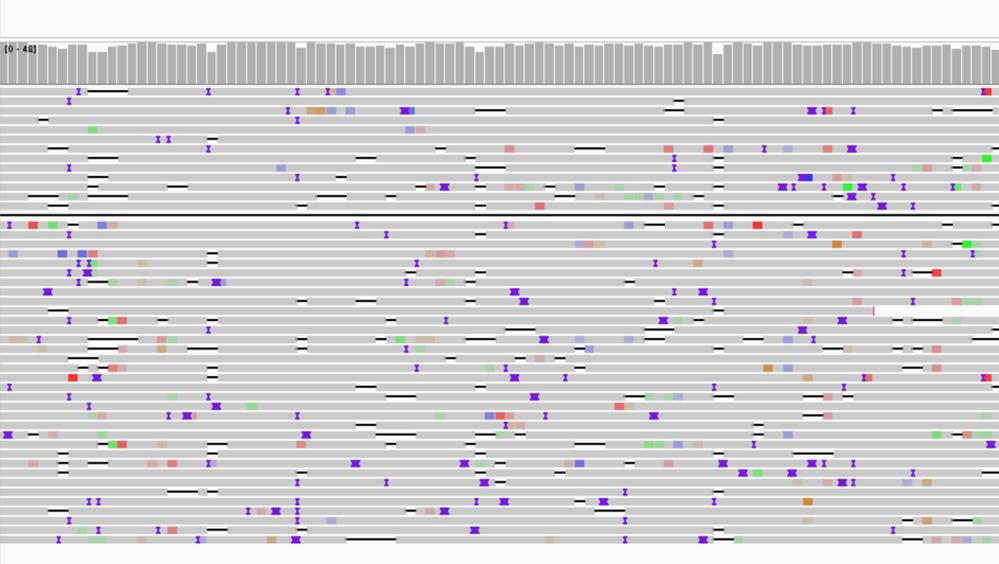
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colo829bl\_pbr\_grch38.bam



colo829bl\_vai\_grch38.bam Coverage  
colo829bl\_vai\_grch38.bam



Sequence → T T A C A C T C T C G C C A A C A G T G T A A A A G C A T T C C T A T T T C T C C A C A G C C T C G C C A G C A T C T A T T G T T T C C T G A C T T T T G A A T A A C C C A T T C T G A C T G G C G T  
RefSeq Genes

chr20:38,645,775-38,645,875

p13 p12.3 p12.2 p12.1 p11.23 p11.21 q11.1 q11.22 q12 q13.12 q13.13 q13.2 q13.31 q13.33

← 102 bp →

38,645,780 bp 38,645,800 bp 38,645,820 bp 38,645,840 bp 38,645,860 bp

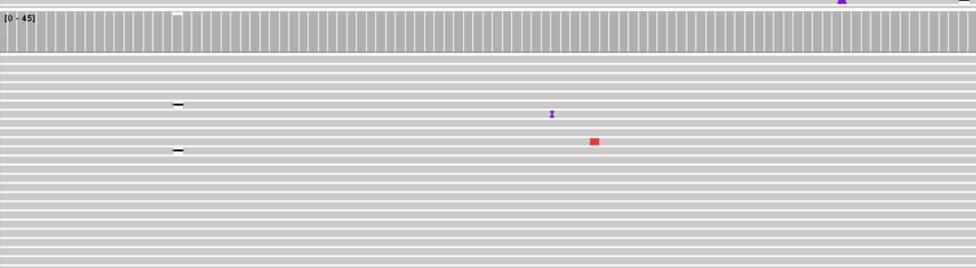
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colo829\_gsc\_grch38.bam



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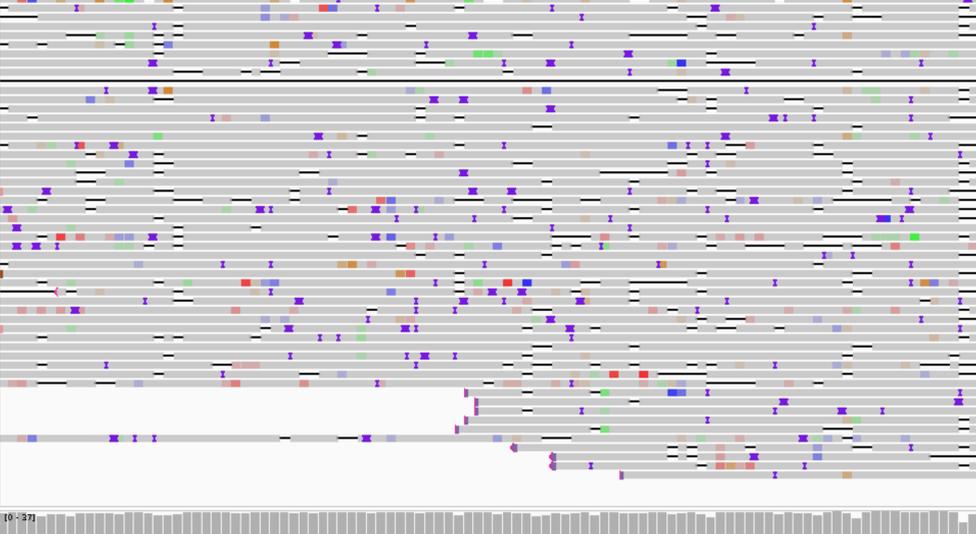
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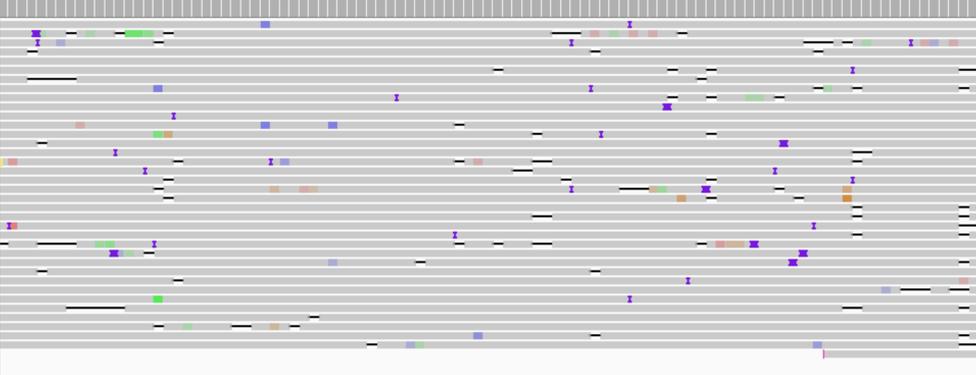
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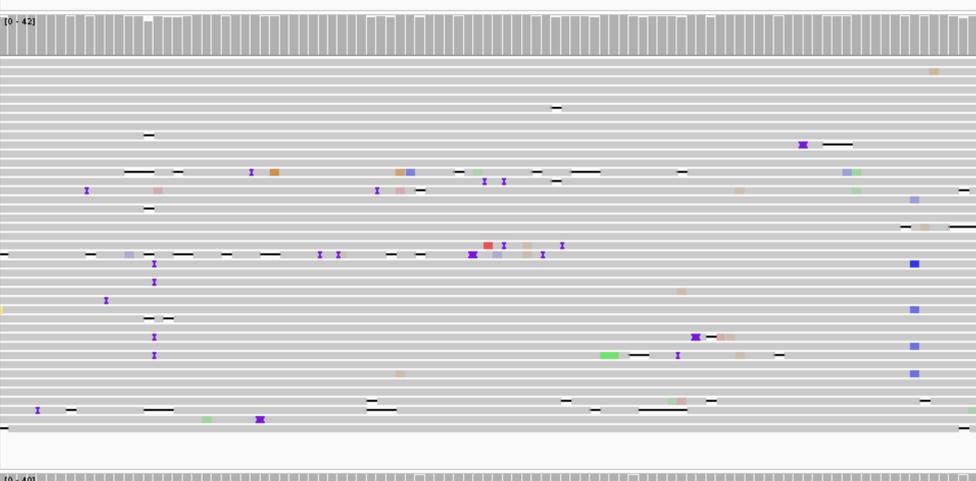
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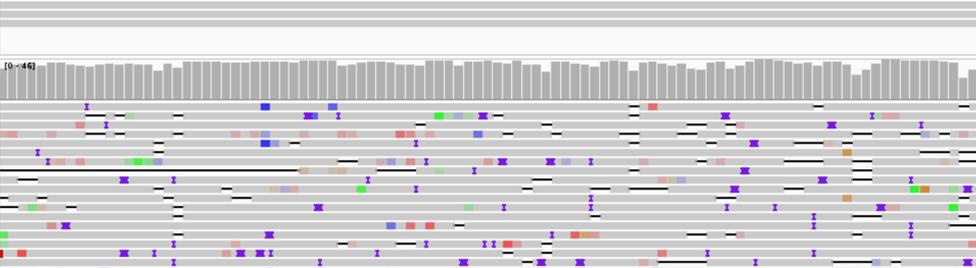
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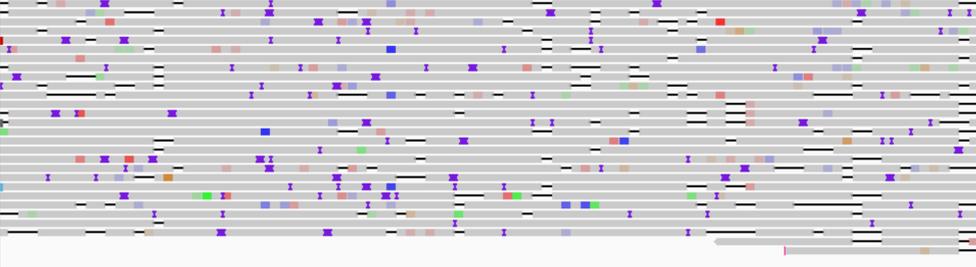
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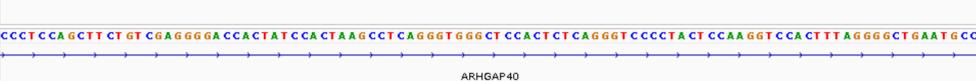
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colo829bl\_pbr\_grch38.bam



colo829bl\_vai\_grch38.bam Coverage



colo829bl\_vai\_grch38.bam



Sequence →

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Refseq Genes



ARHGAP40

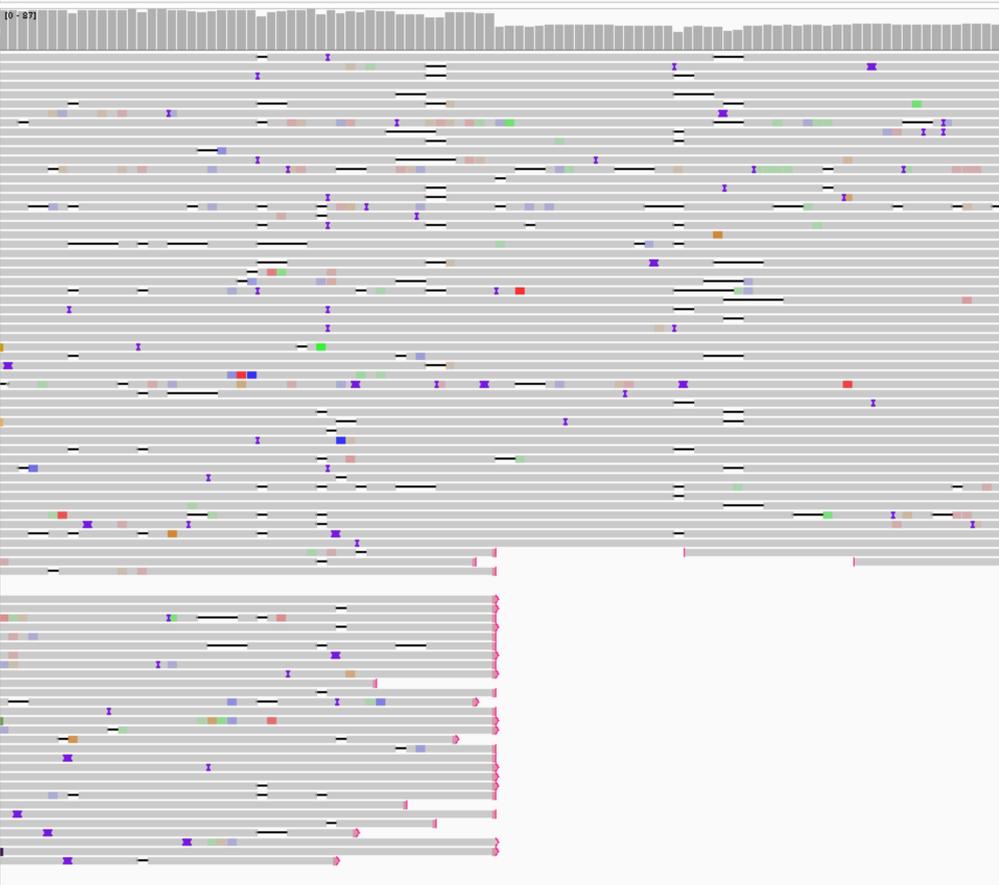
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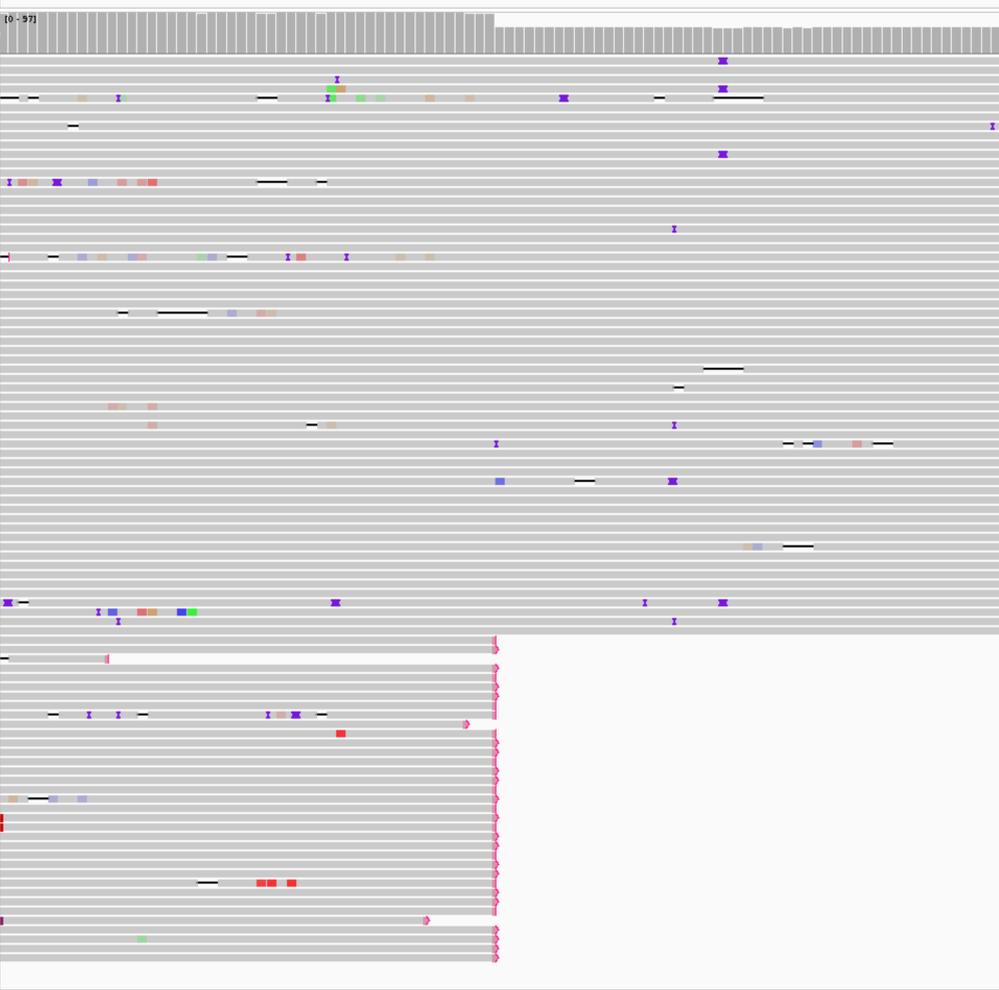
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102 bp

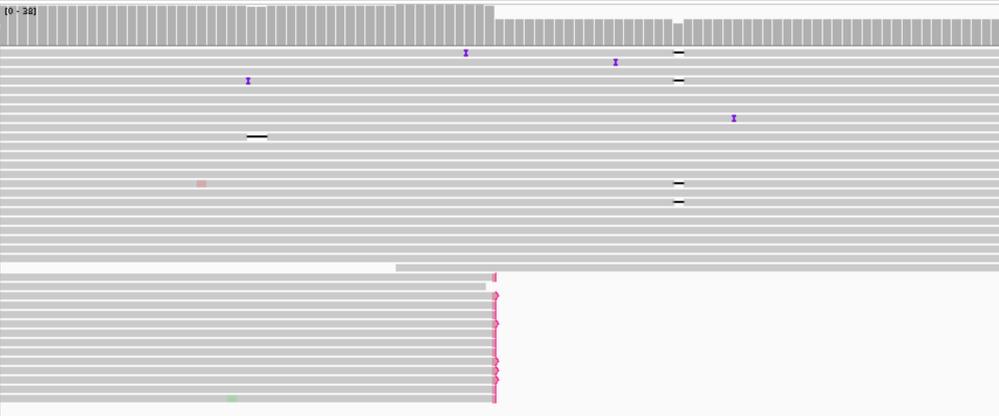
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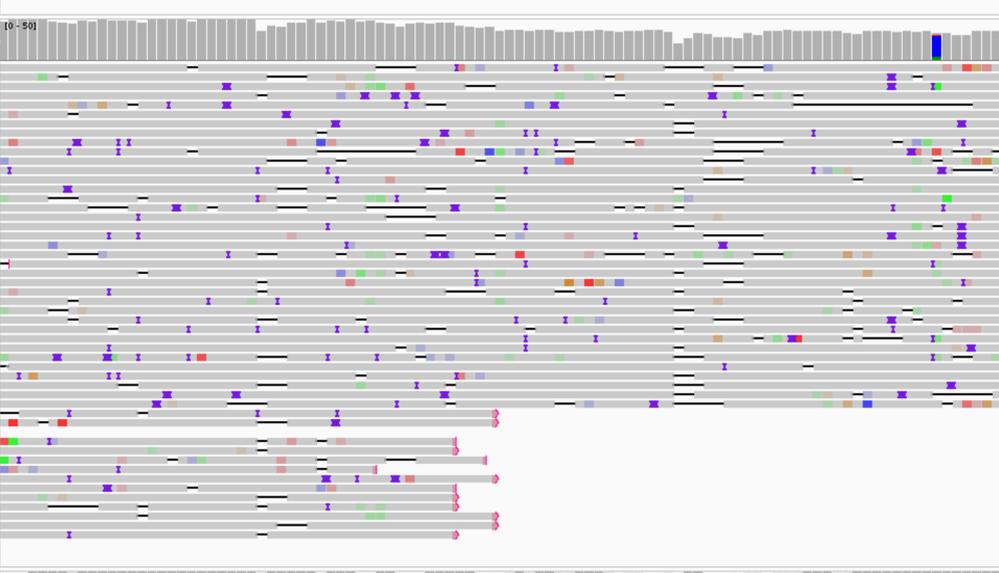
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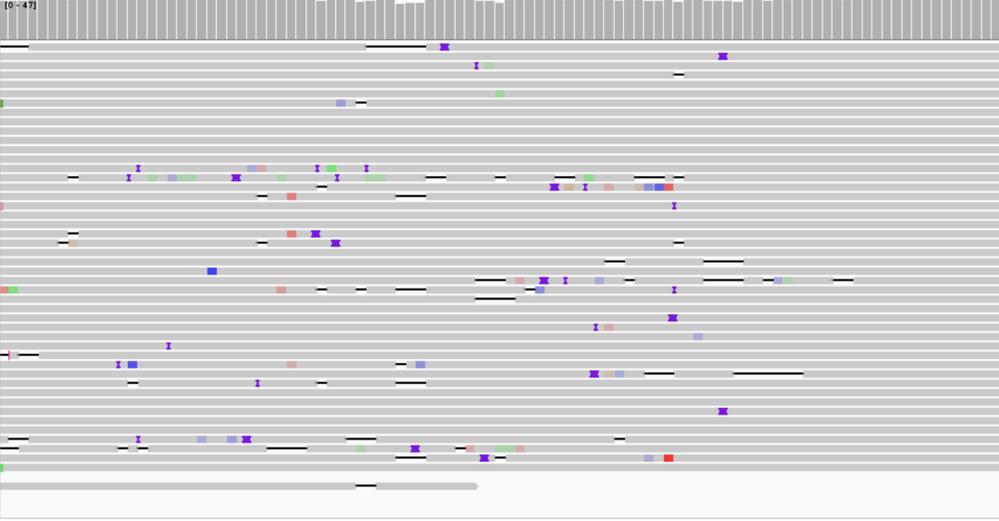
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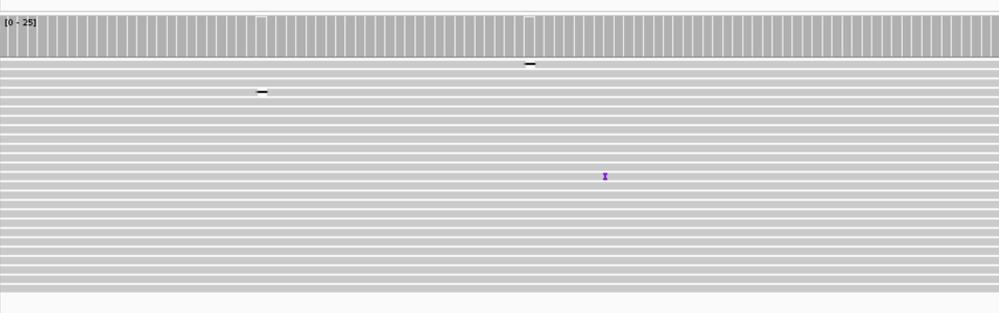
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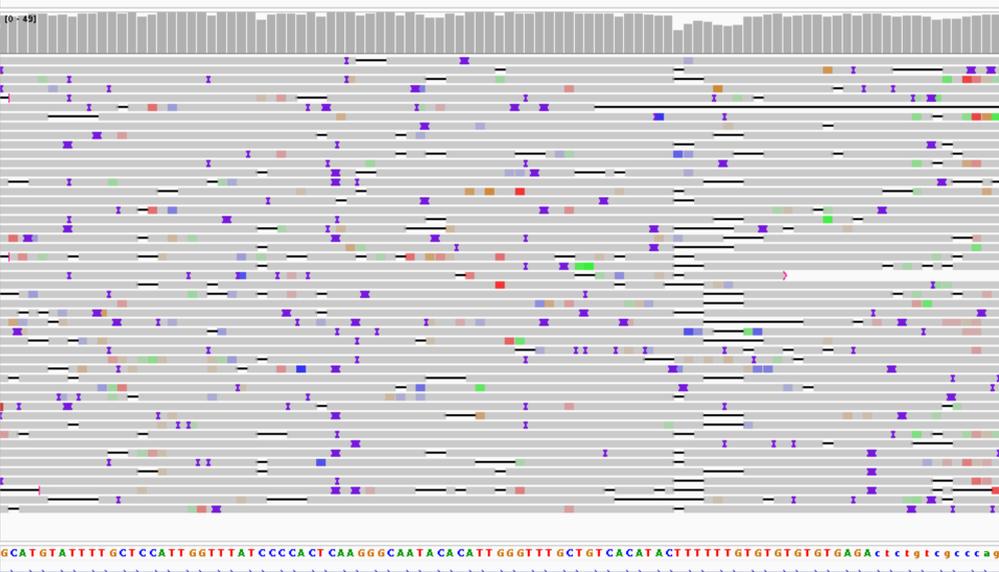
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colo829bl\_ont\_grch38.bam



colo829bl\_pbr\_grch38.bam Coverage  
colo829bl\_pbr\_grch38.bam



colo829bl\_vai\_grch38.bam Coverage  
colo829bl\_vai\_grch38.bam



Sequence → GCATGTAATTTT GCTCCATT GGTTTAT CCCCACTCAA GGGCAATACACATT GGGTTT GCTGT CACAT ACTTTTTTGTGTGTGTGAGAC t c t g t c g c c c a g  
RefSeq Genes ANKLE1

chr20:38,645,974-38,646,074

p13 p12.3 p12.2 p12.1 p11.23 p11.21 q11.1 q11.22 q12 q13.12 q13.13 q13.2 q13.31 q13.33

← 102 bp →

38,645,980 bp 38,646,000 bp 38,646,020 bp 38,646,040 bp 38,646,060 bp

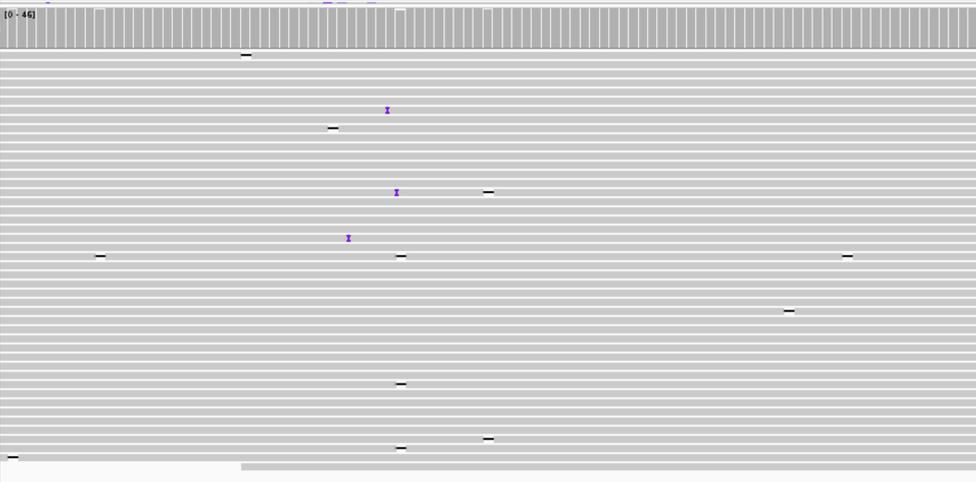
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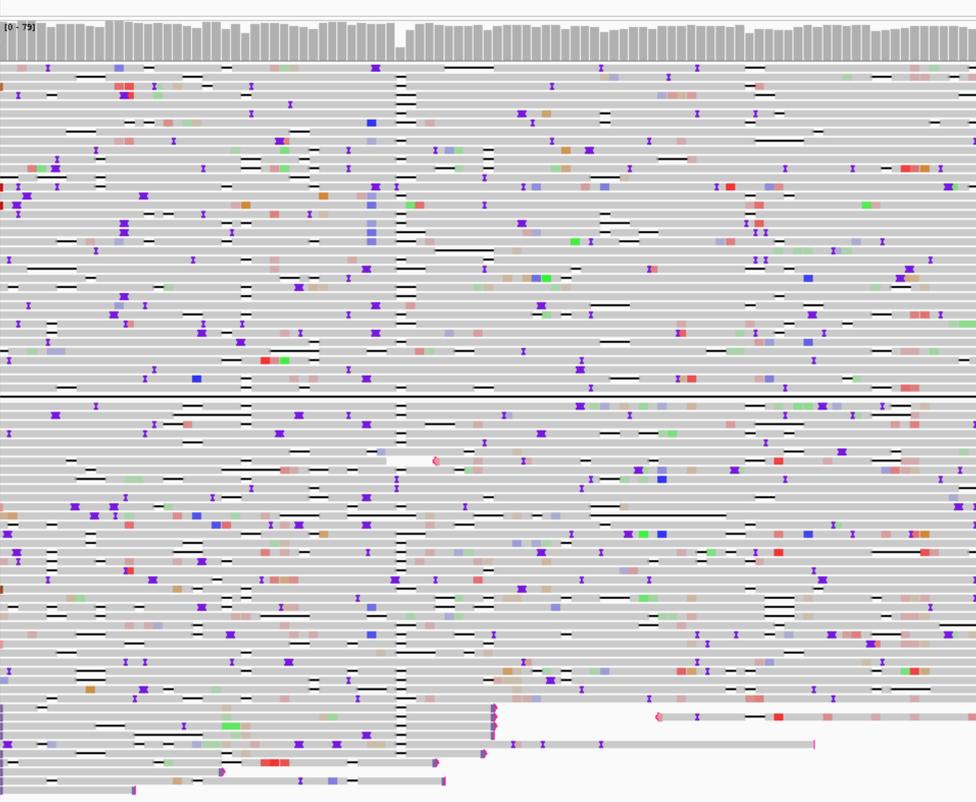
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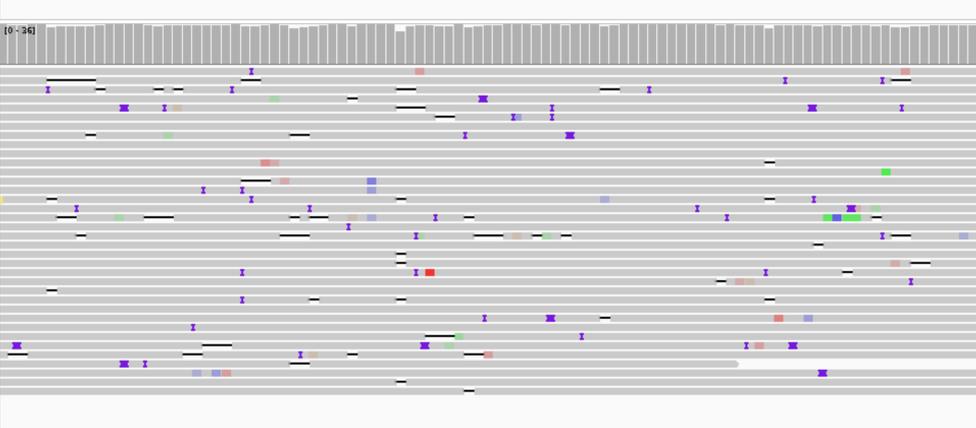
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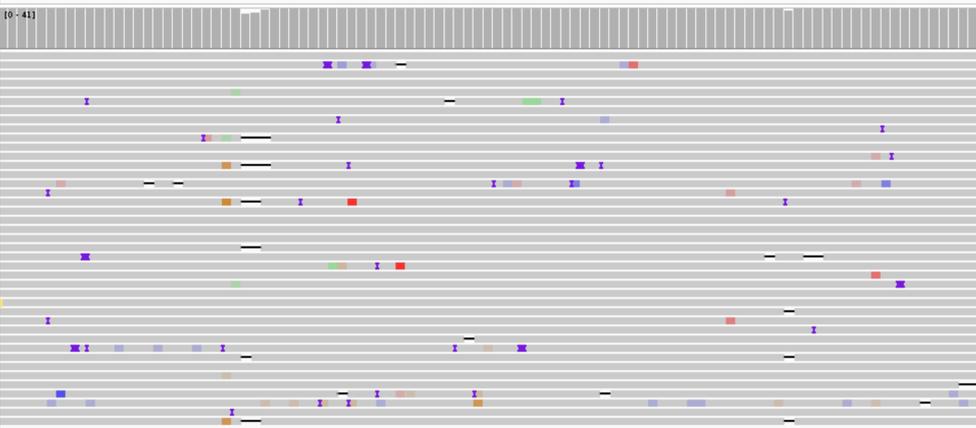
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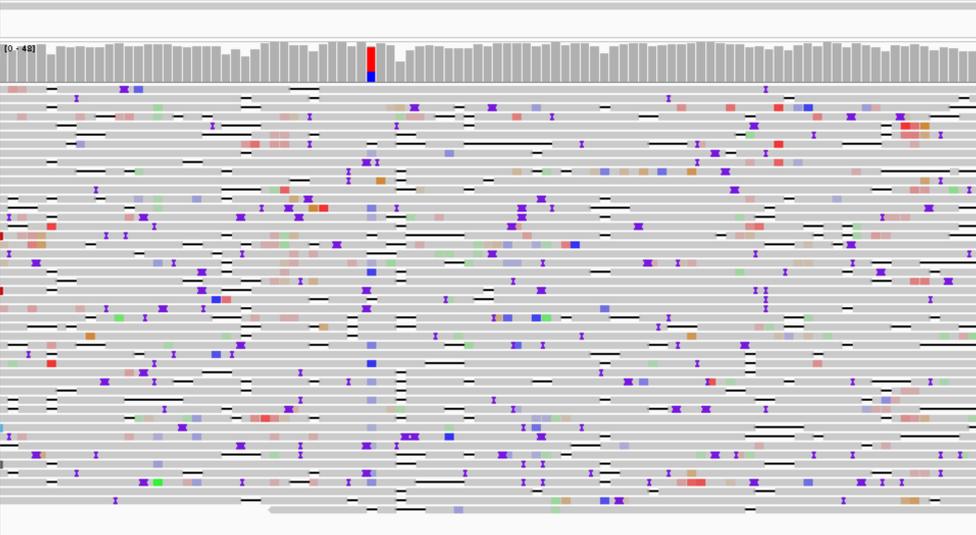
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colo829bl\_ont\_grch38.bam



colo829bl\_pbr\_grch38.bam Coverage  
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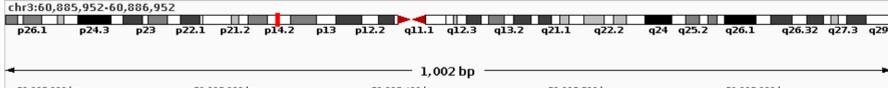
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Refseq Genes → V A S F L V A Q V  
ARHGAP40

Supplementary Figure 2: IGV screenshot of the Structural Variants (SVs) labeled as False Negatives (FN) according to the Valle-Inclan benchmark. For detail see Supplementary Table 5

- A) Evidence found, missed: a 141091 DEL located in chr3:60886452 (Valle-Inclan ID truthset\_12\_1) breakpoint 1
- B) Evidence found, missed: a 141091 DEL located in chr3:60886452 (Valle-Inclan ID truthset\_12\_1) breakpoint 2
- C) Evidence found, missed: a 100409 DEL located in chr7:78352522 (Valle-Inclan ID truthset\_23\_1) breakpoint 1
- D) Evidence found, missed: a 100409 DEL located in chr7:78352522 (Valle-Inclan ID truthset\_23\_1) breakpoint 2
- E) Evidence found, missed: a 185860 DEL located in chr10:81882050 (Valle-Inclan ID truthset\_44\_1) breakpoint 1
- F) Evidence found, missed: a 185860 DEL located in chr10:81882050 (Valle-Inclan ID truthset\_44\_1) breakpoint 2
- G) Evidence found, missed: a 7212 DUP located in chr15:41329096 (Valle-Inclan ID truthset\_55\_1)
- H) Wrongly filtered: a 55 DEL located in chr14:104093751 (Valle-Inclan ID truthset\_50\_1)
- I) Little evidence found: a 71459 DEL located in chr3:186724386 (Valle-Inclan ID truthset\_13\_1) breakpoint 1
- J) Little evidence found: a 71459 DEL located in chr3:186724386 (Valle-Inclan ID truthset\_13\_1) breakpoint 2
- K) Little evidence found: a 1163 DEL located in chr8:111050191 (Valle-Inclan ID truthset\_32\_1)
- L) Little evidence found: a 2732610 DEL located in chr10:33097525 (Valle-Inclan ID truthset\_41\_1) breakpoint 1
- M) Little evidence found: a 2732610 DEL located in chr10:33097525 (Valle-Inclan ID truthset\_41\_1) breakpoint 2
- N) Little evidence found: a 549 DEL located in chr10:53716587 (Valle-Inclan ID truthset\_42\_1)
- O) No evidence found: a 289 INV located in chr6:26193889 (Valle-Inclan ID truthset\_18\_1)
- P) No evidence found: a 100 INS located in chr14:105867667 (Valle-Inclan ID truthset\_51\_1)
- Q) No evidence found: a 100 INS located in chr14:105867667 (Valle-Inclan ID truthset\_51\_1) expanded, deletion detected

Not shown in IGV screenshots

- Some evidence found: a 1 BND located in chr3:26390427 (Valle-Inclan ID truthset\_9\_1)
- Some evidence found: a 1 BND located in chr10:58717662 (Valle-Inclan ID truthset\_43\_1)
- Little evidence found: a 1 BND located in chr3:25359111 (Valle-Inclan ID truthset\_7\_1)
- Little evidence found: a 1 BND located in chr10:7017550 (Valle-Inclan ID truthset\_38\_1)
- Little evidence found: a 1 BND located in chr15:23461732 (Valle-Inclan ID truthset\_53\_1)
- No evidence found: a 1 BND located in chr1:86871328 (Valle-Inclan ID truthset\_1\_1)
- No evidence found: a 1 BND located in chr3:25359568 (Valle-Inclan ID truthset\_8\_1)
- No evidence found: a 1 BND located in chr6:138452922 (Valle-Inclan ID truthset\_19\_1)
- No evidence found: a 1 BND located in chr6:138453044 (Valle-Inclan ID truthset\_20\_1)

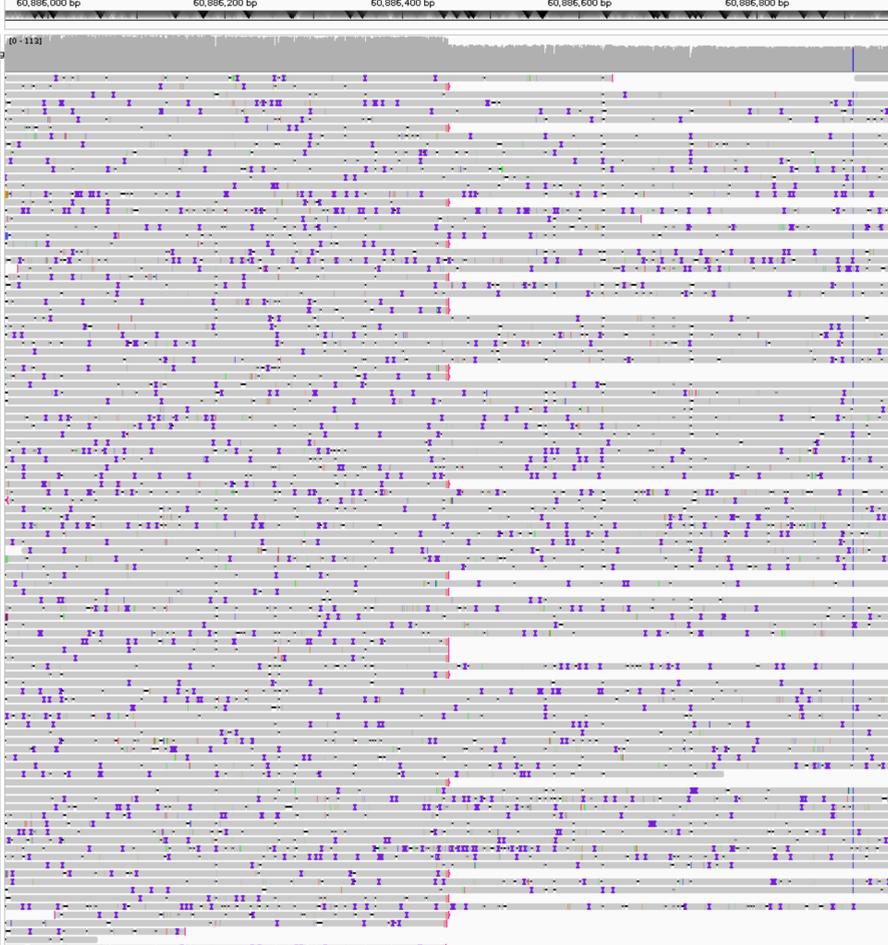


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60,886,000 bp 60,886,200 bp 60,886,400 bp 60,886,600 bp 60,886,800 bp

1,002 bp

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colo829\_gsc\_grch38.bam

colo829\_ort\_grch38.bam Coverage



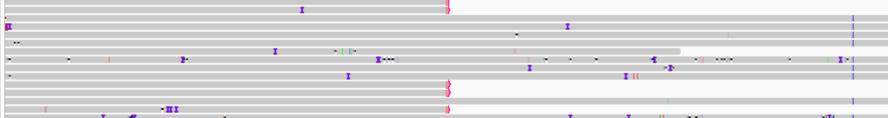
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colo829\_pbr\_grch38.bam Coverage



colo829\_pbr\_grch38.bam

colo829\_val\_grch38.bam Coverage



colo829\_val\_grch38.bam

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colo829\_l\_gsc\_grch38.bam

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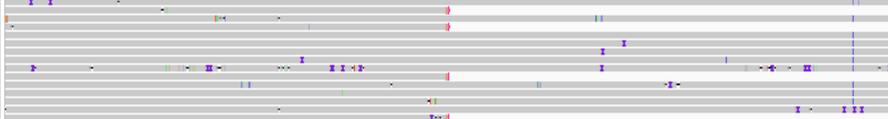
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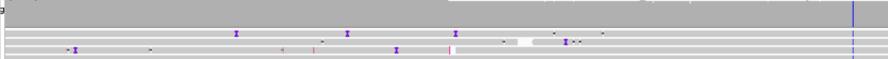
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colo829\_l\_val\_grch38.bam

Sequence

Refseq Genes



FHT

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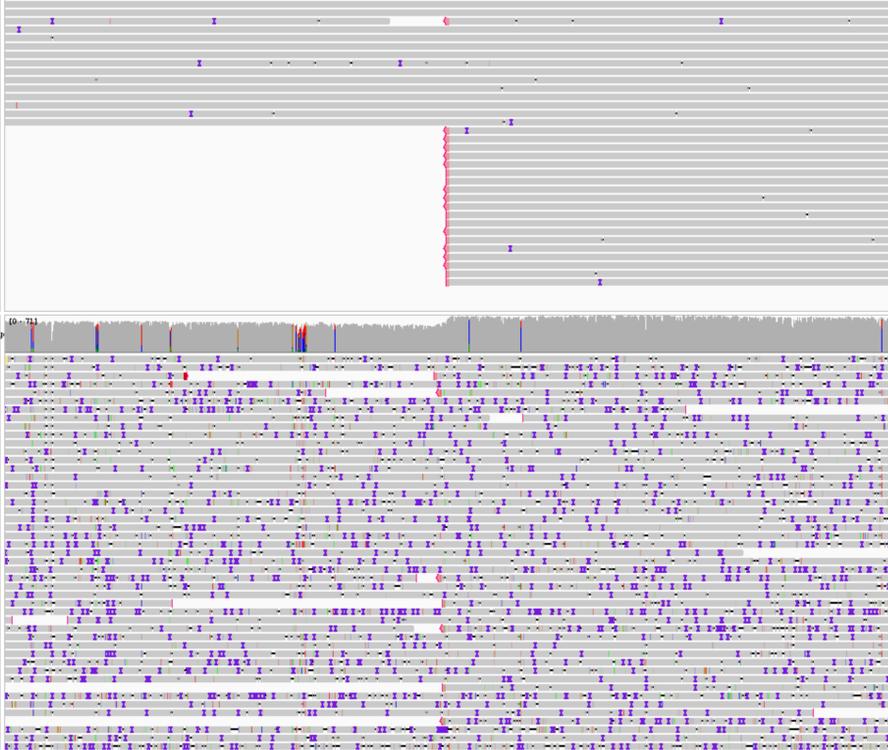
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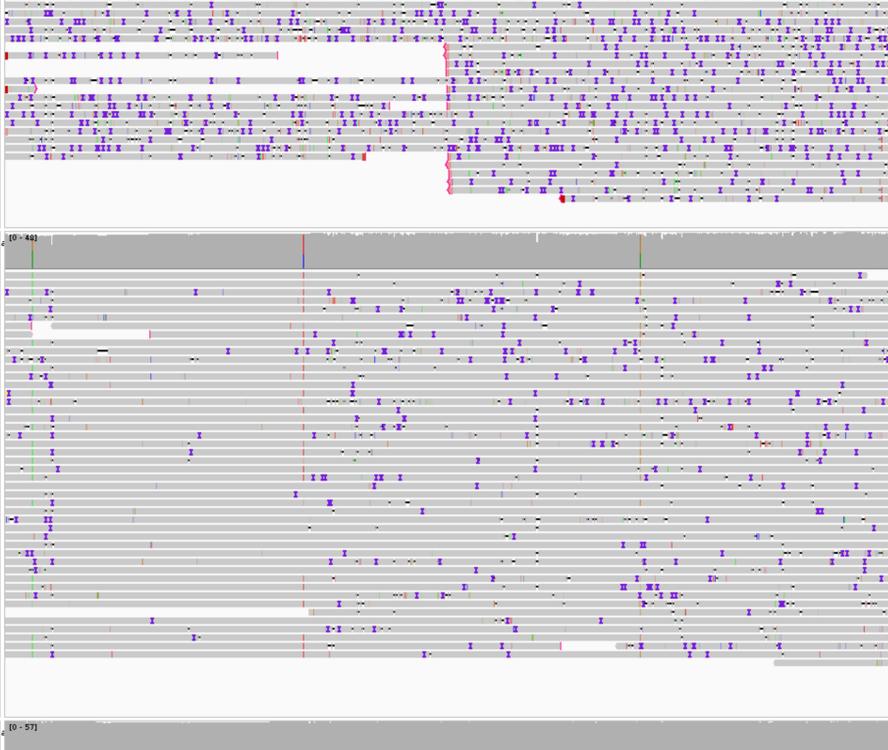
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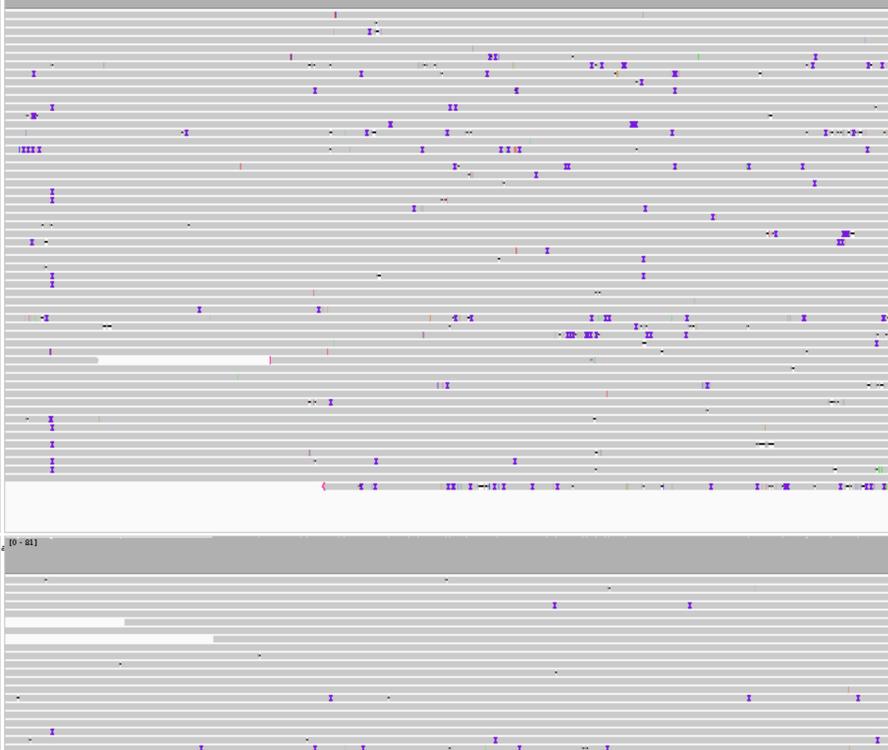
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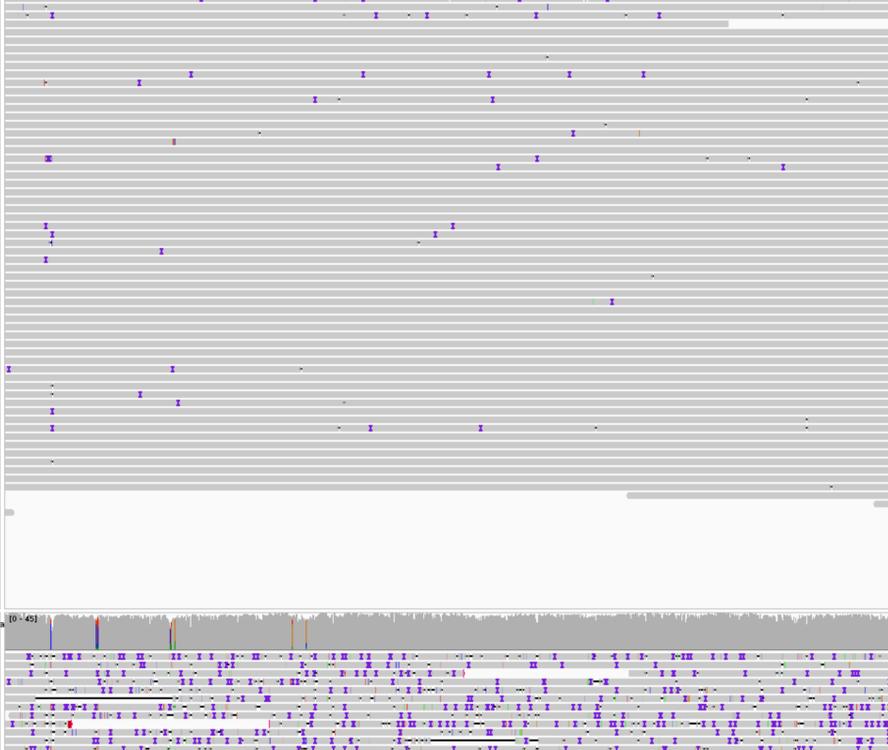
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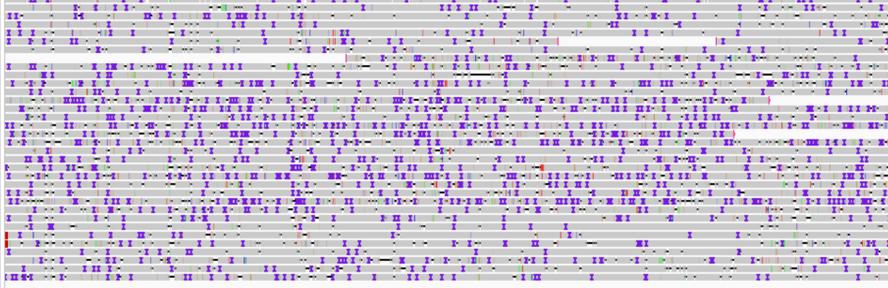
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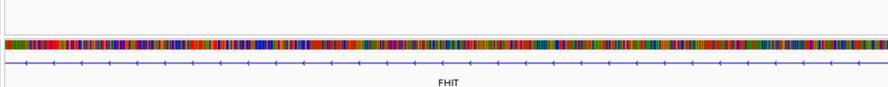
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Sequence  
Refseq Genes



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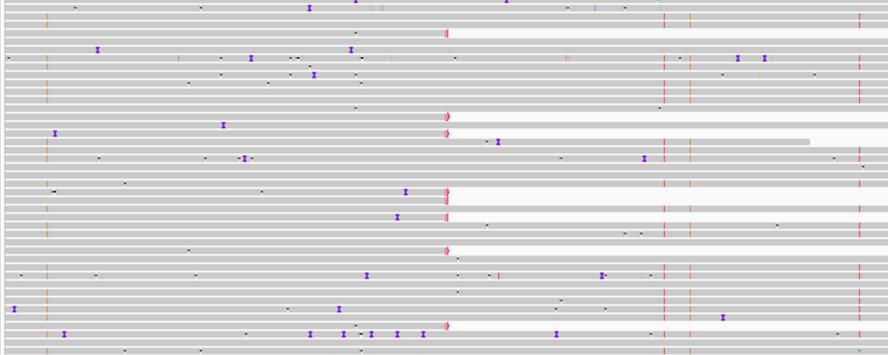
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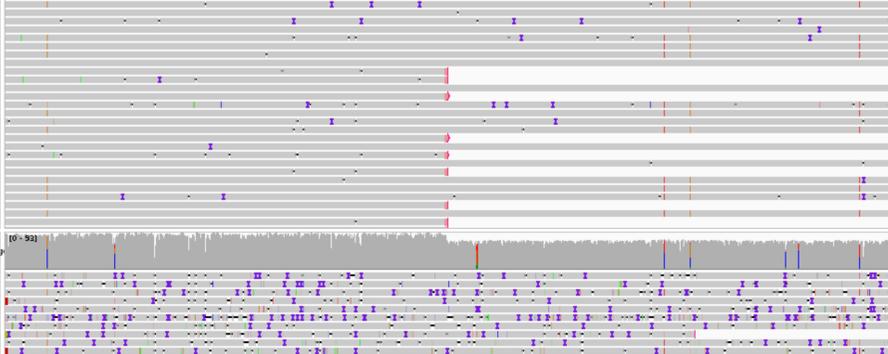
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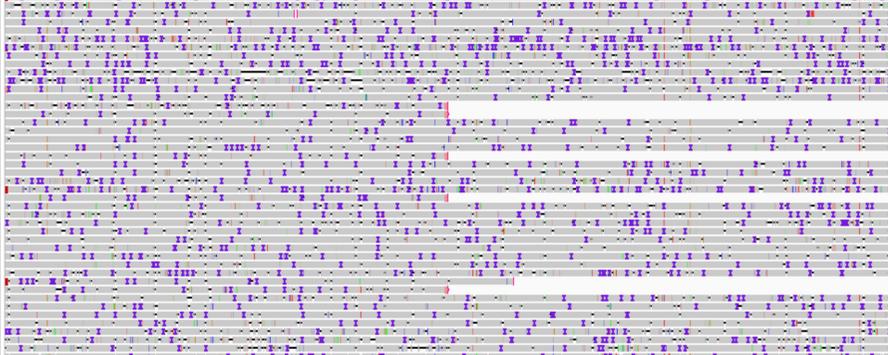
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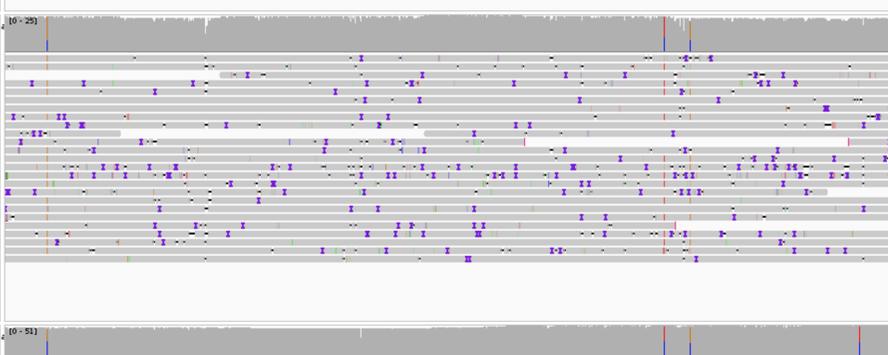
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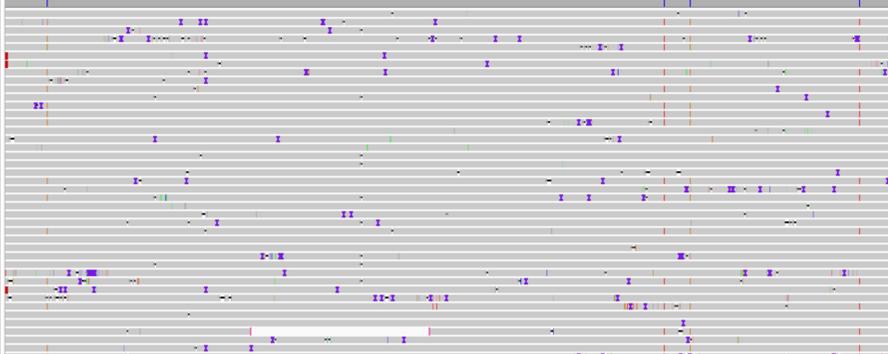
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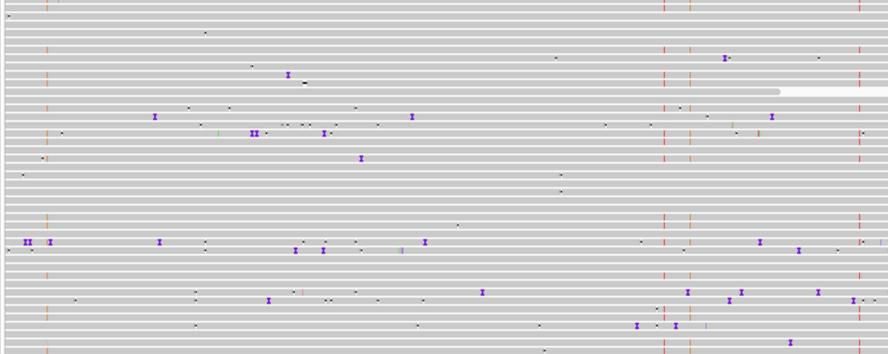
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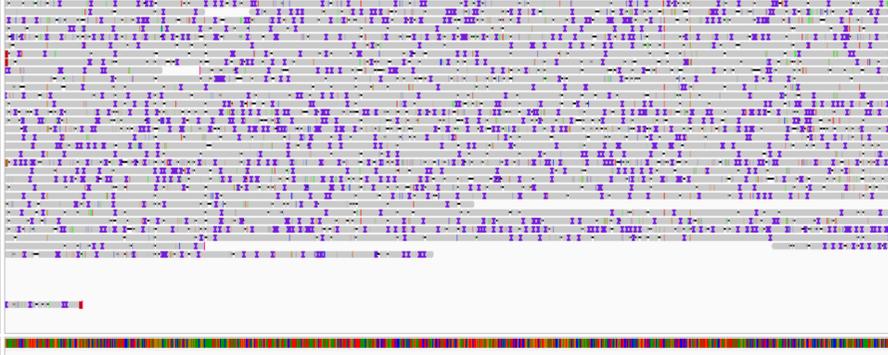
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colo829l\_pbr\_grch38.bam Coverage



colo829l\_pbr\_grch38.bam



colo829l\_val\_grch38.bam Coverage



colo829l\_val\_grch38.bam



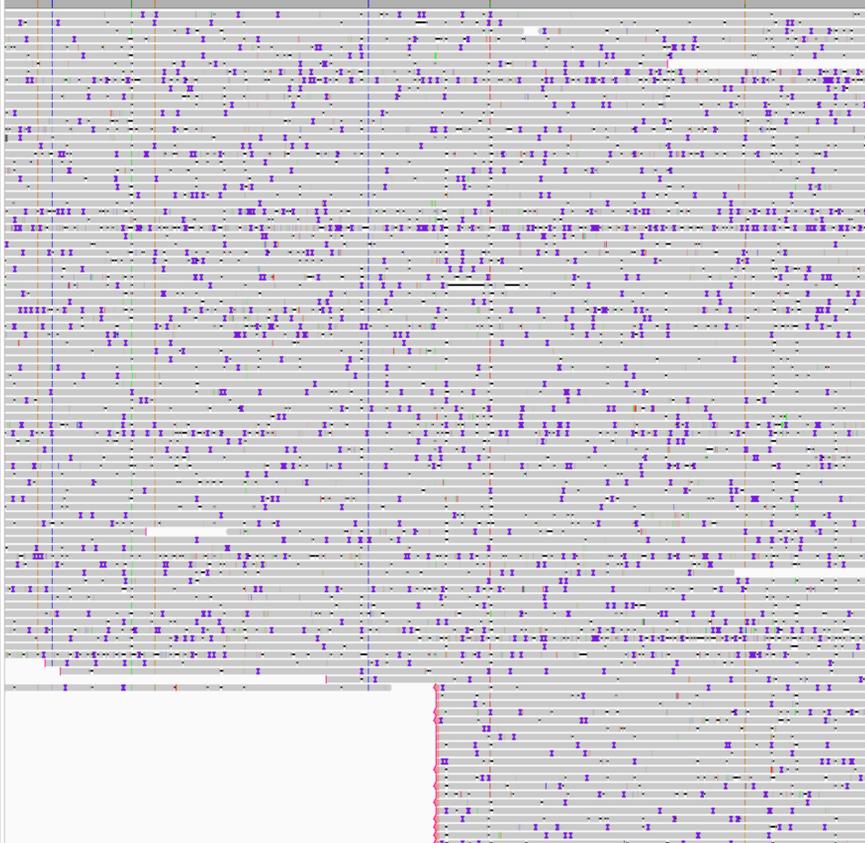
Sequence



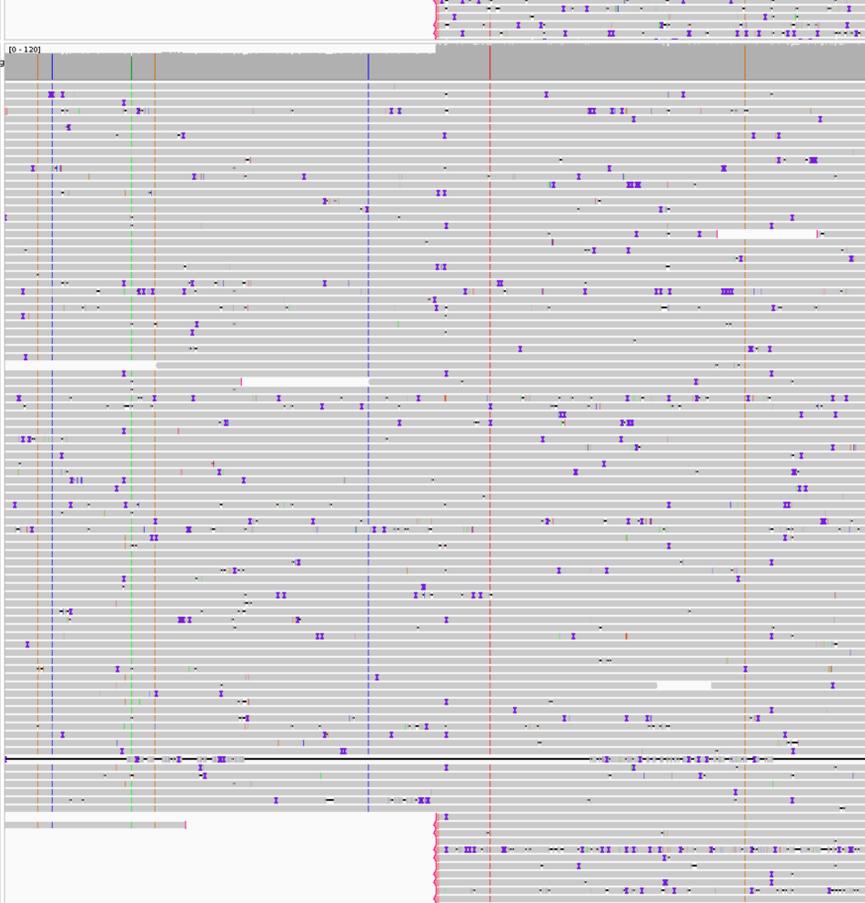
Refseq Genes



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colo829\_gsc\_grch38.bam



colo829\_ort\_grch38.bam Coverage  
colo829\_ort\_grch38.bam



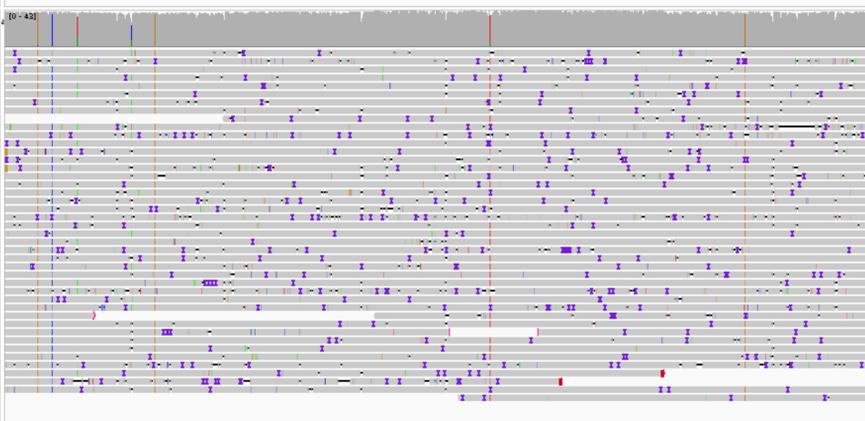
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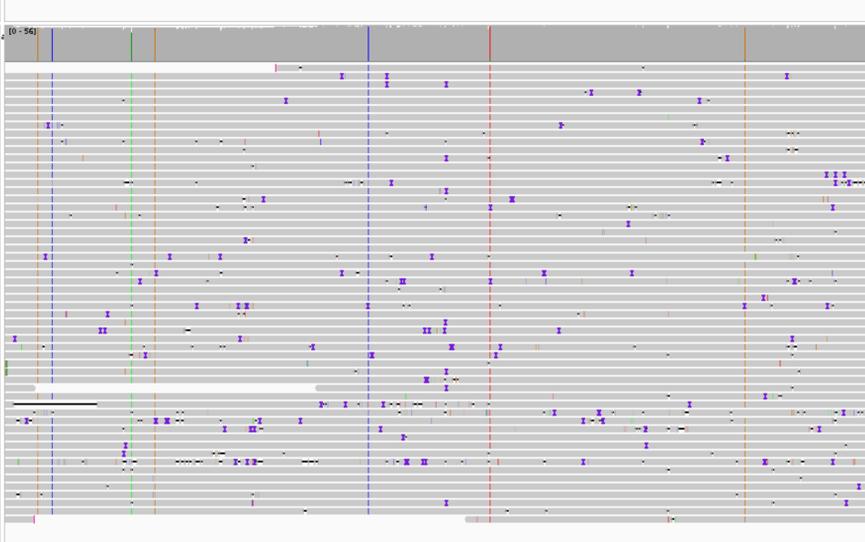
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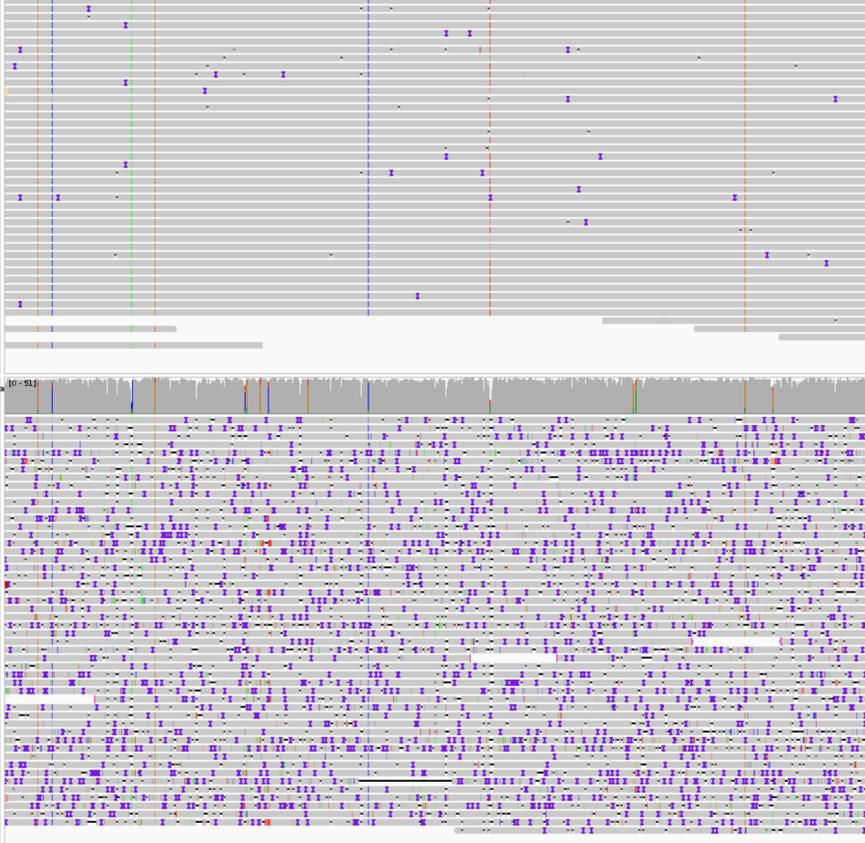
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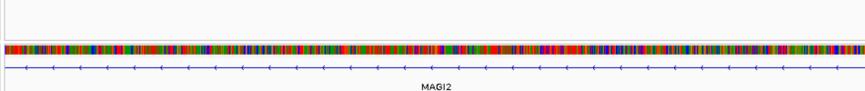
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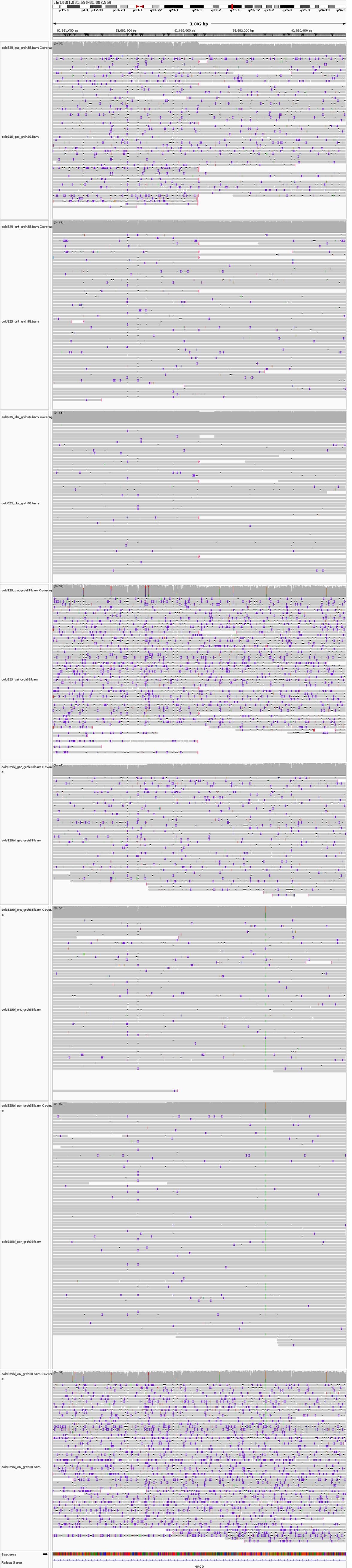


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Sequence  
Refseq Genes



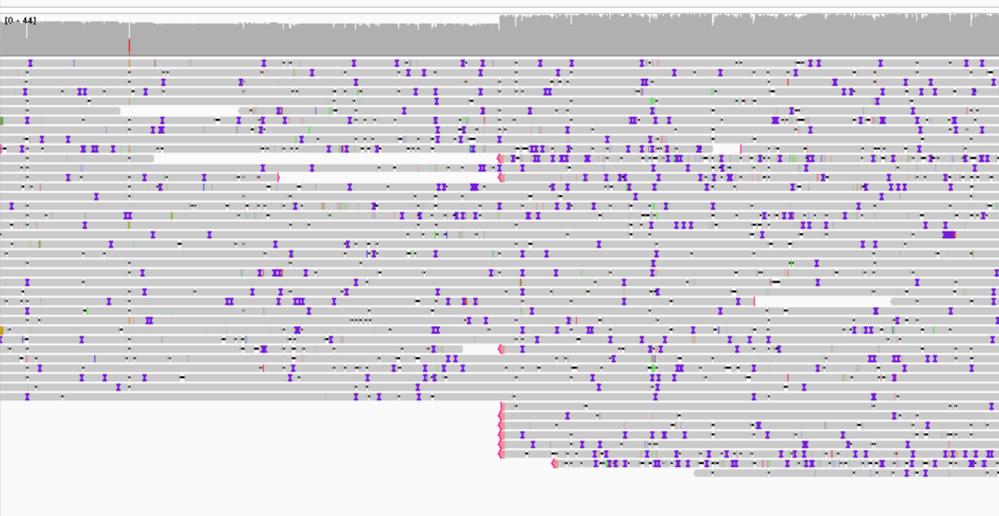


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1,002 bp

82,067,600 bp 82,067,800 bp 82,068,000 bp 82,068,200 bp 82,068,400 bp

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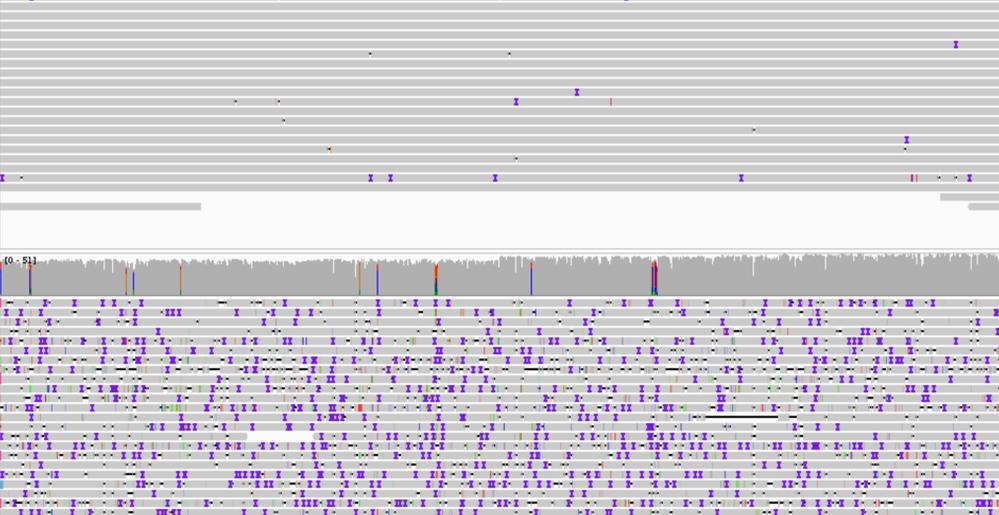
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colo829\_pbr\_grch38.bam Coverage  
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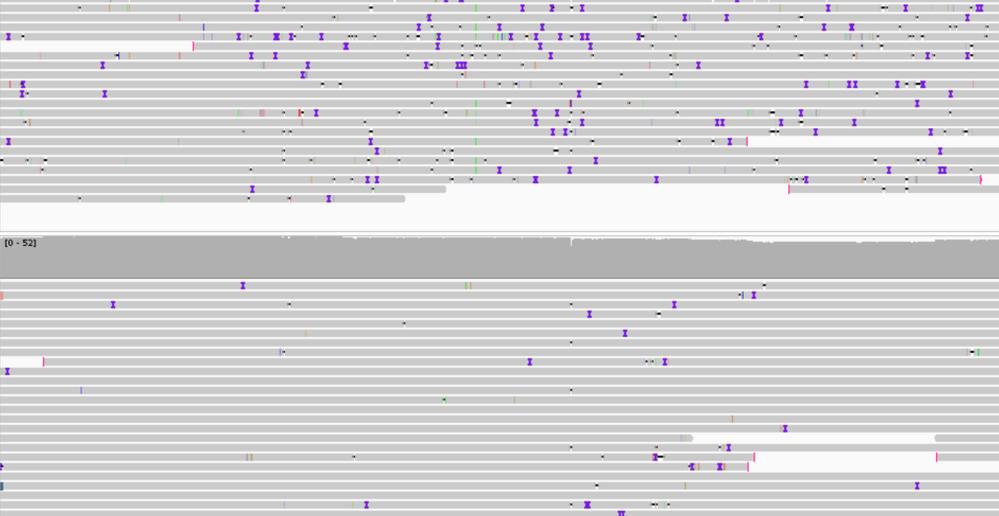
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colo829\_vai\_grch38.bam



colo829b\_gsc\_grch38.bam Coverage  
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colo829b\_ont\_grch38.bam Coverage  
colo829b\_ont\_grch38.bam



colo829b\_pbr\_grch38.bam Coverage  
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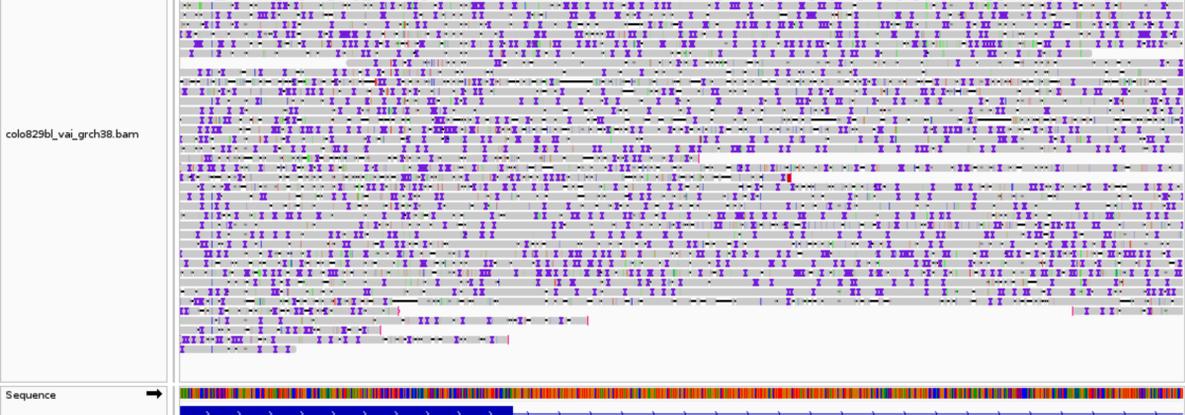
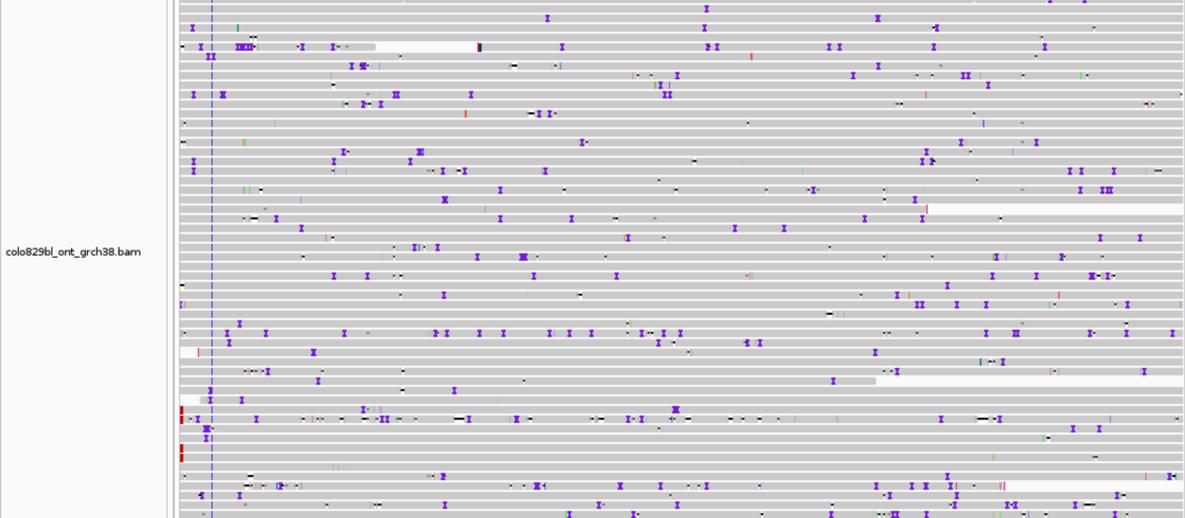
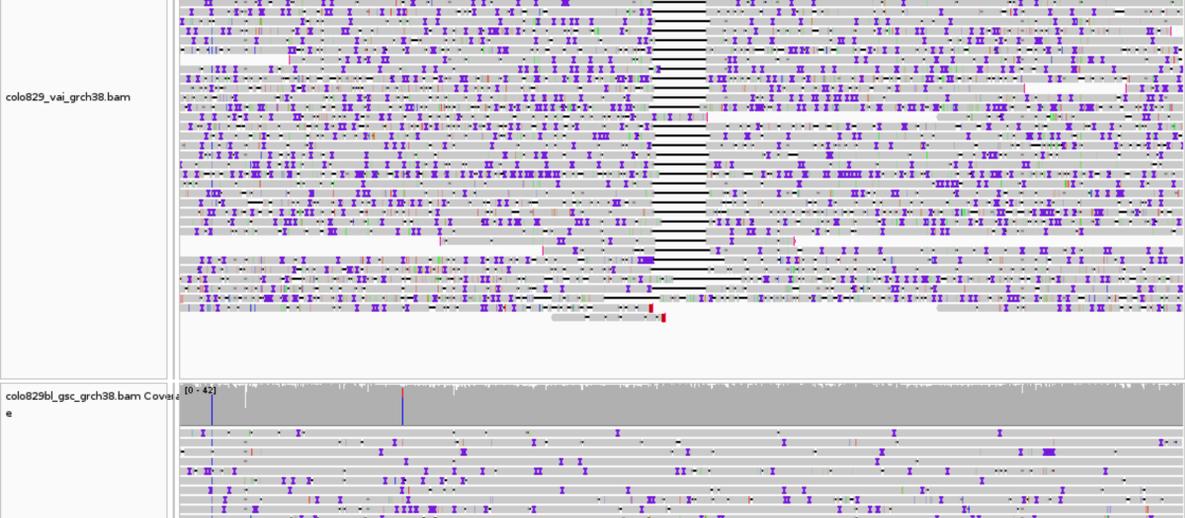
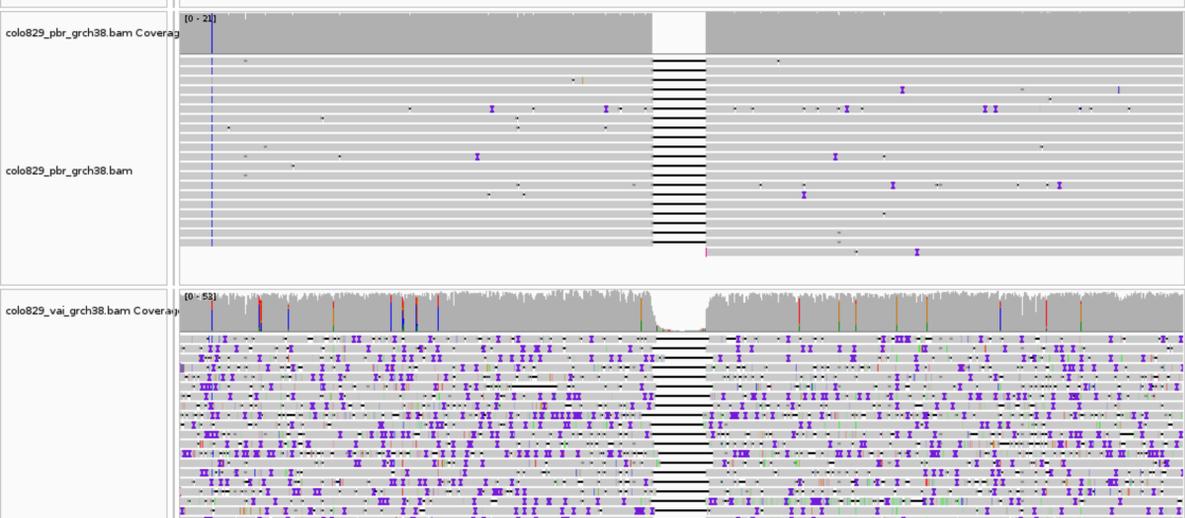
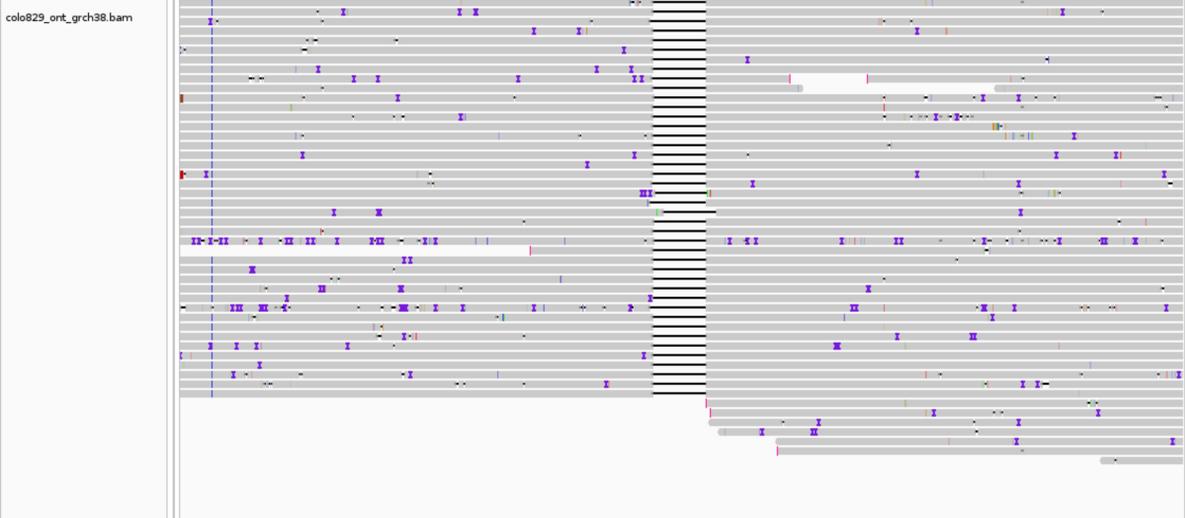
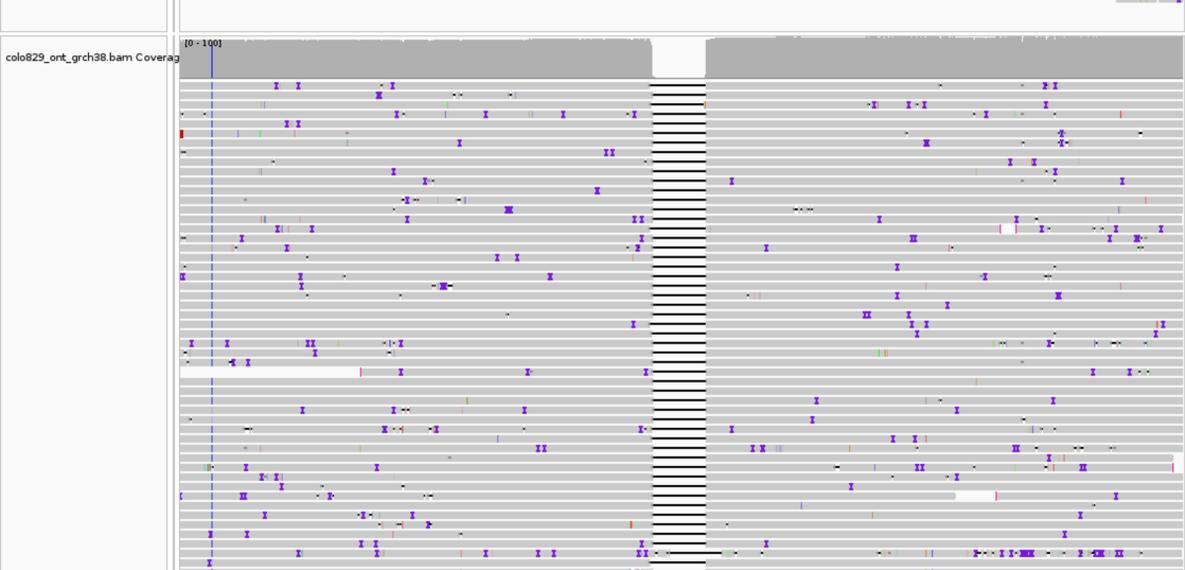
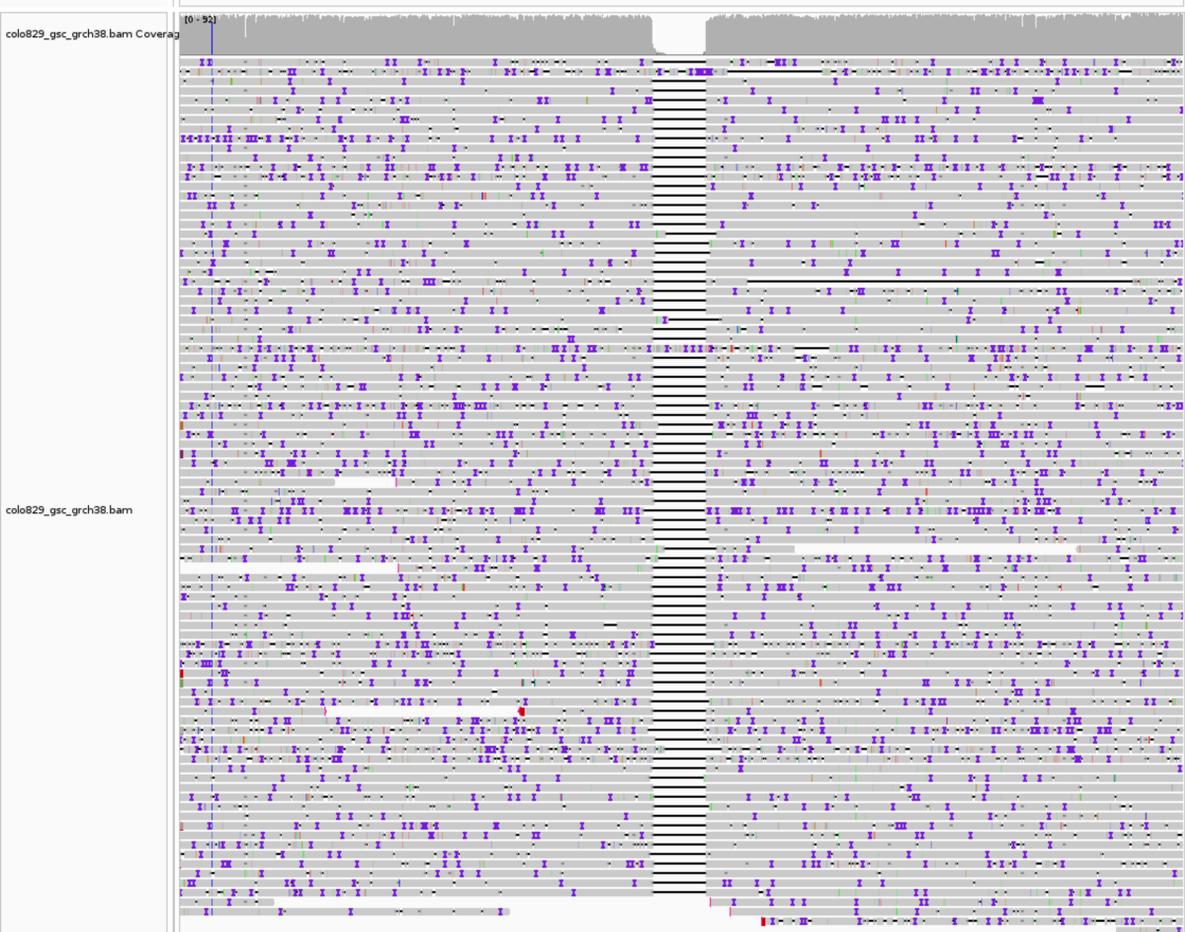


Sequence  
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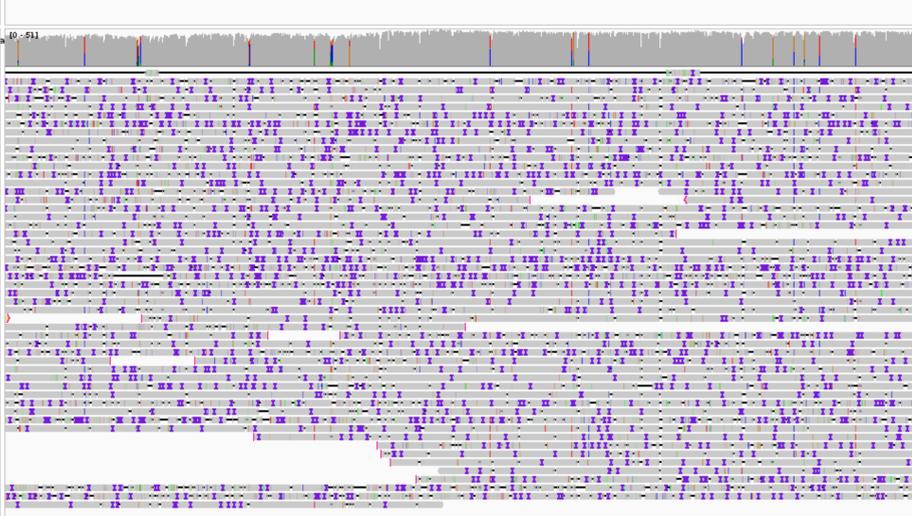
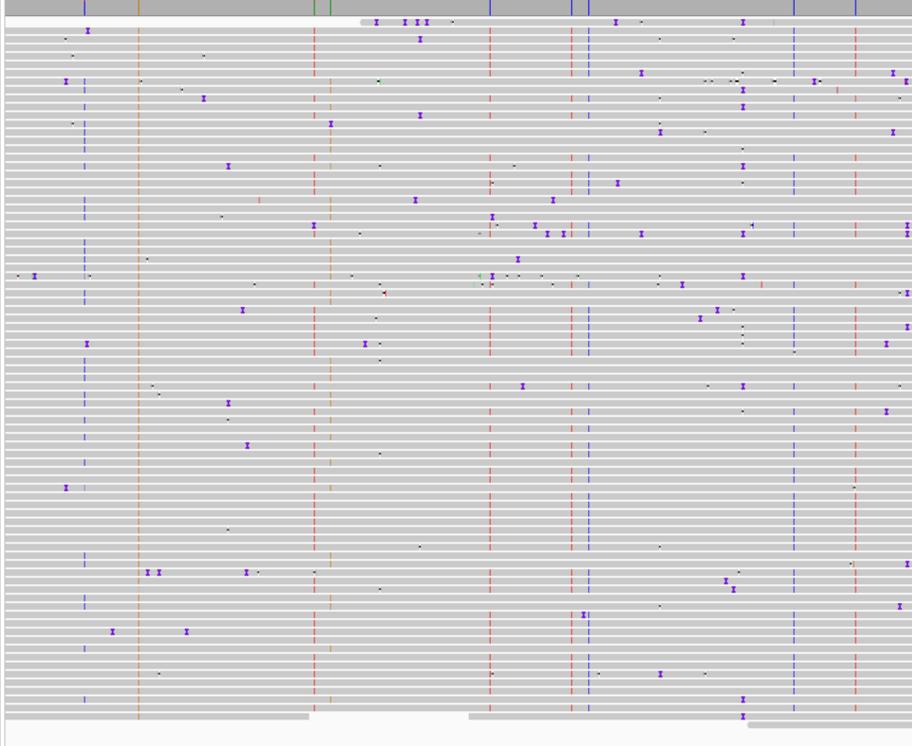
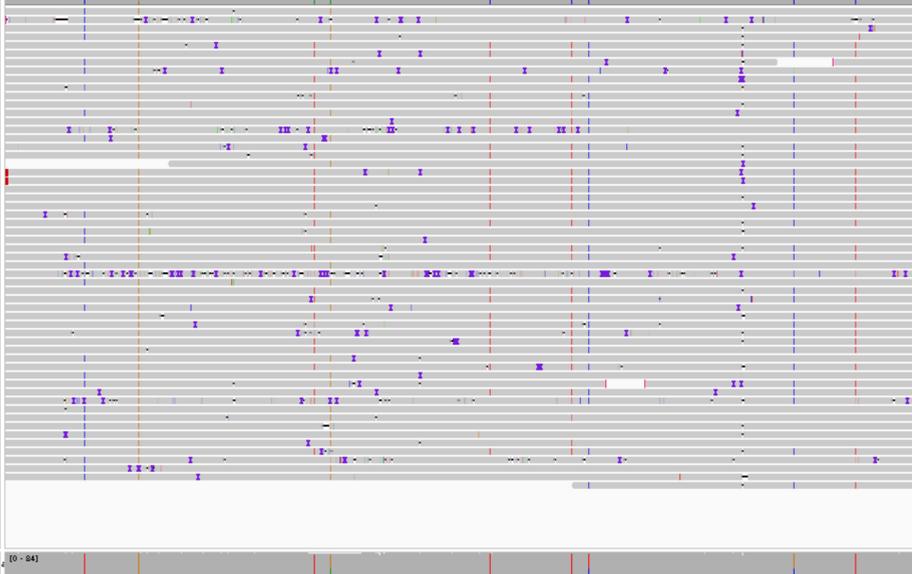
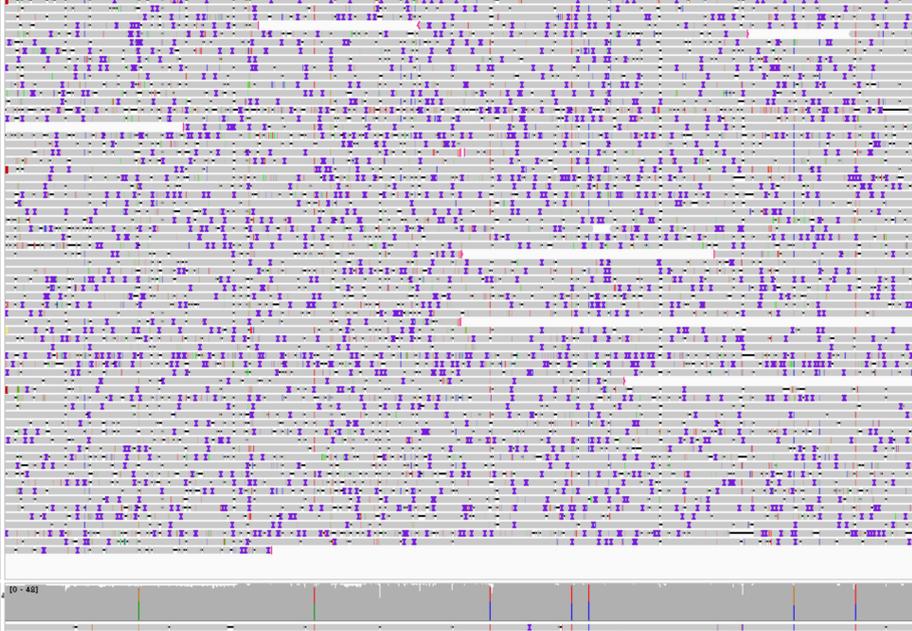
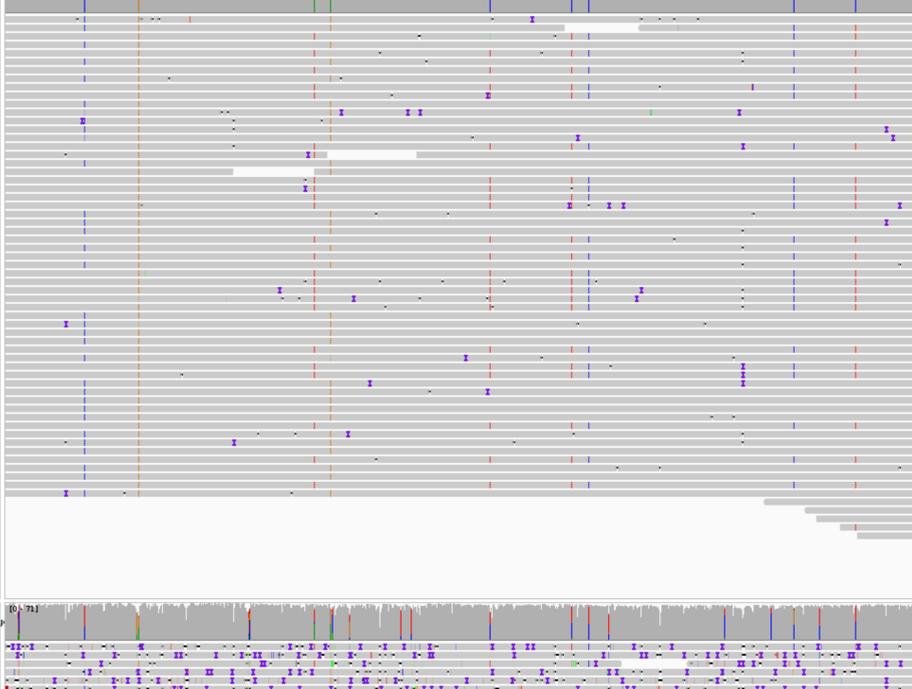
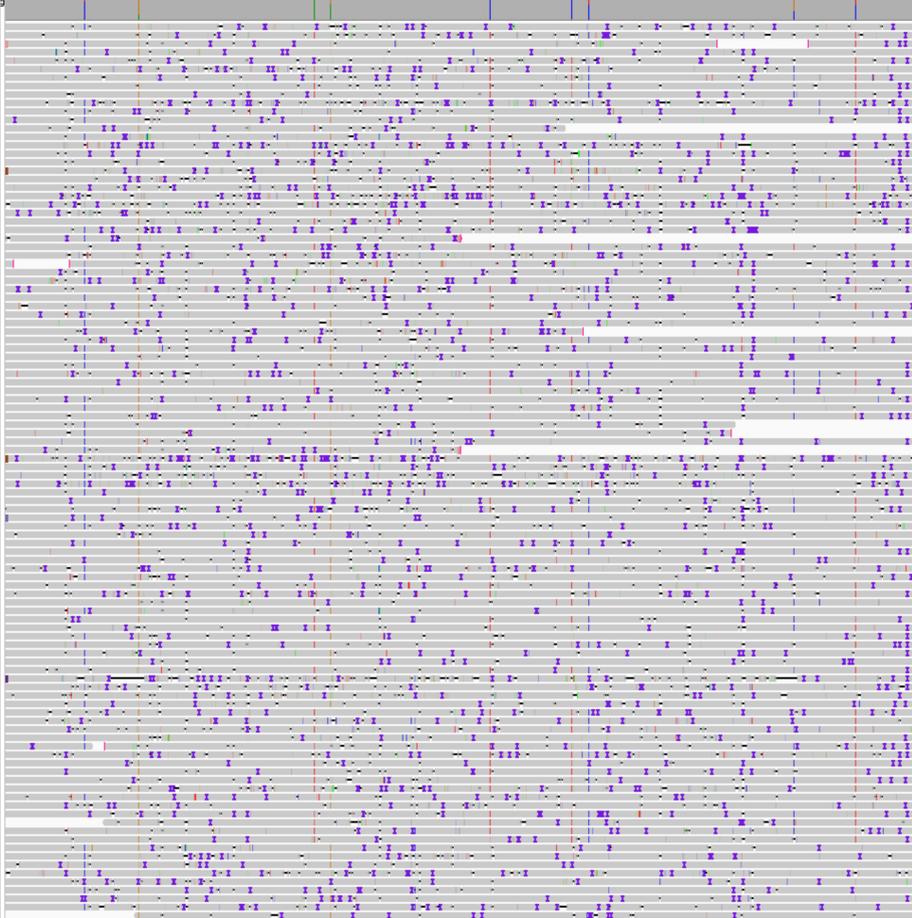
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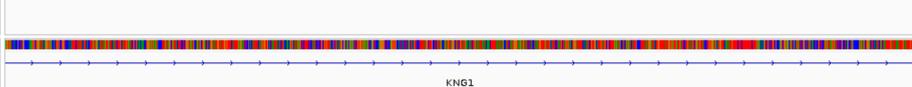




colo829\_gsc\_grch38.bam Coverage  
colo829\_gsc\_grch38.bam  
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colo829\_pbr\_grch38.bam Coverage  
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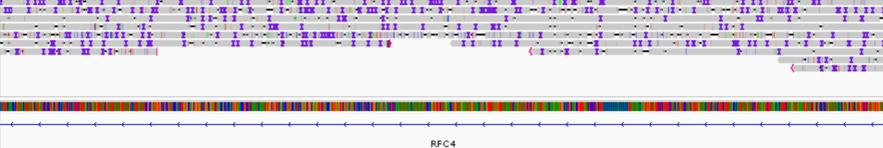
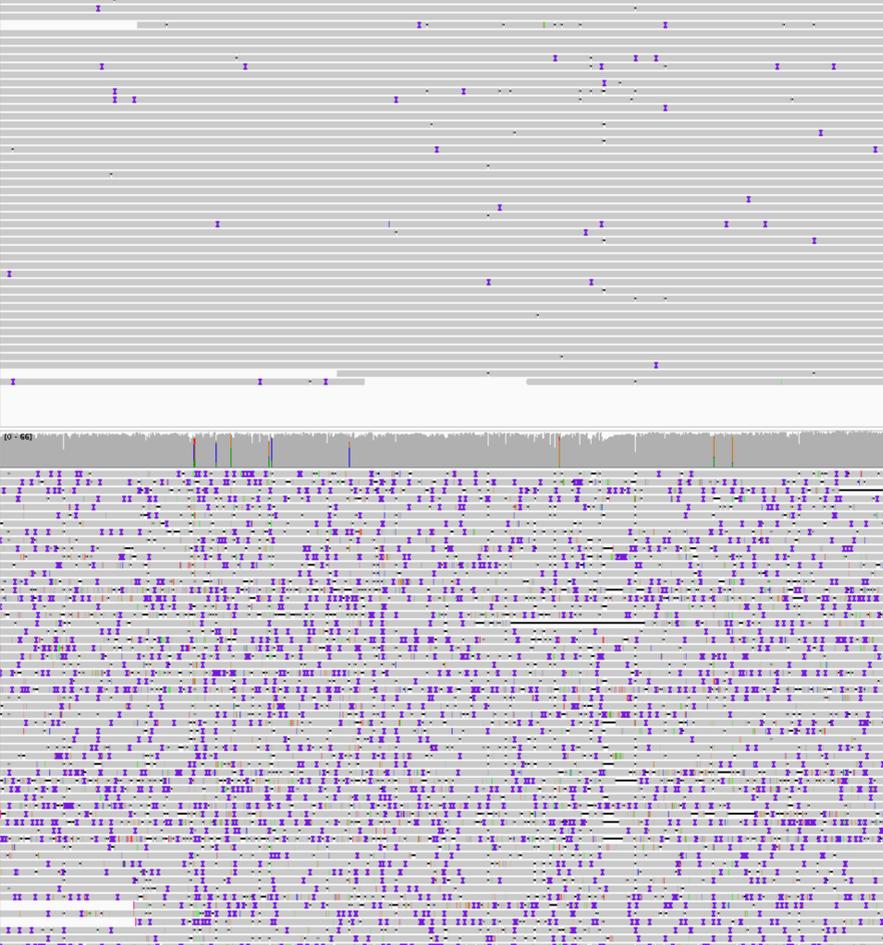
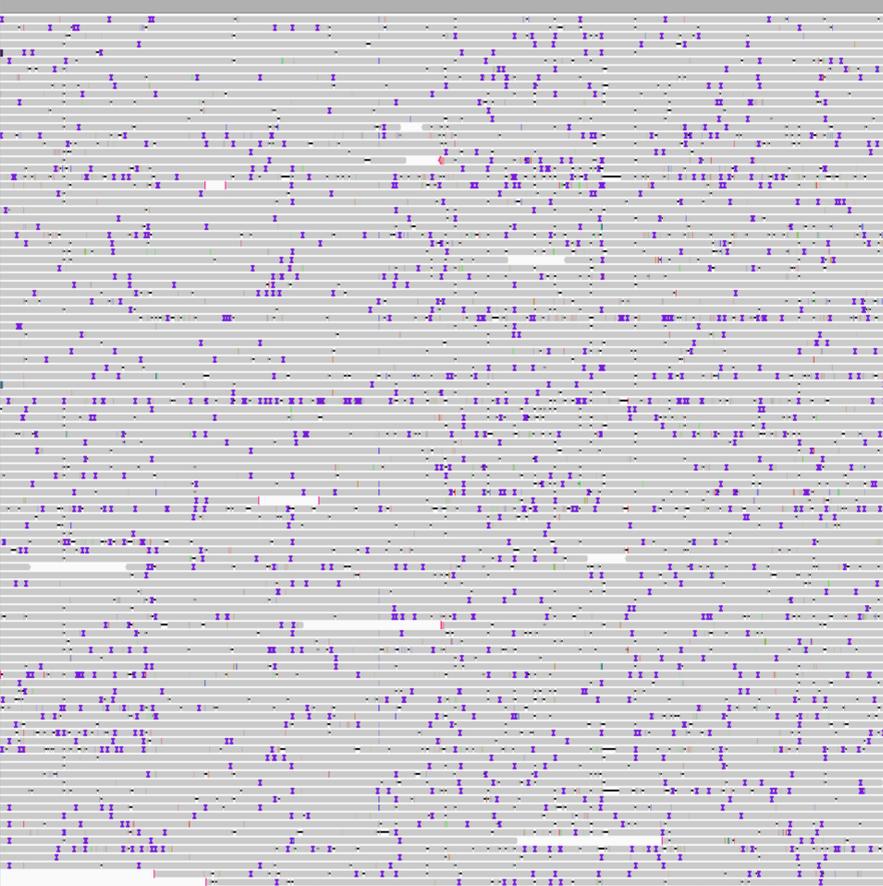


Sequence  
Refseq Genes





colo829\_gsc\_grch38.bam Coverage  
colo829\_gsc\_grch38.bam  
colo829\_ort\_grch38.bam Coverage  
colo829\_ort\_grch38.bam  
colo829\_pbr\_grch38.bam Coverage  
colo829\_pbr\_grch38.bam  
colo829\_val\_grch38.bam Coverage  
colo829\_val\_grch38.bam  
colo829l\_gsc\_grch38.bam Coverage  
colo829l\_gsc\_grch38.bam  
colo829l\_ort\_grch38.bam Coverage  
colo829l\_ort\_grch38.bam  
colo829l\_pbr\_grch38.bam Coverage  
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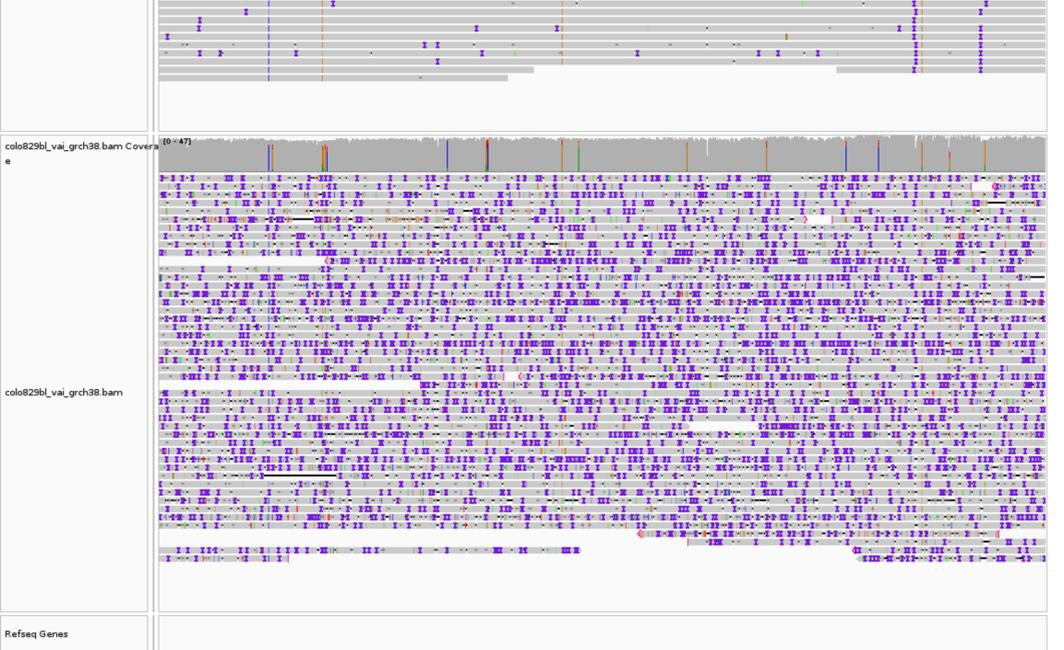
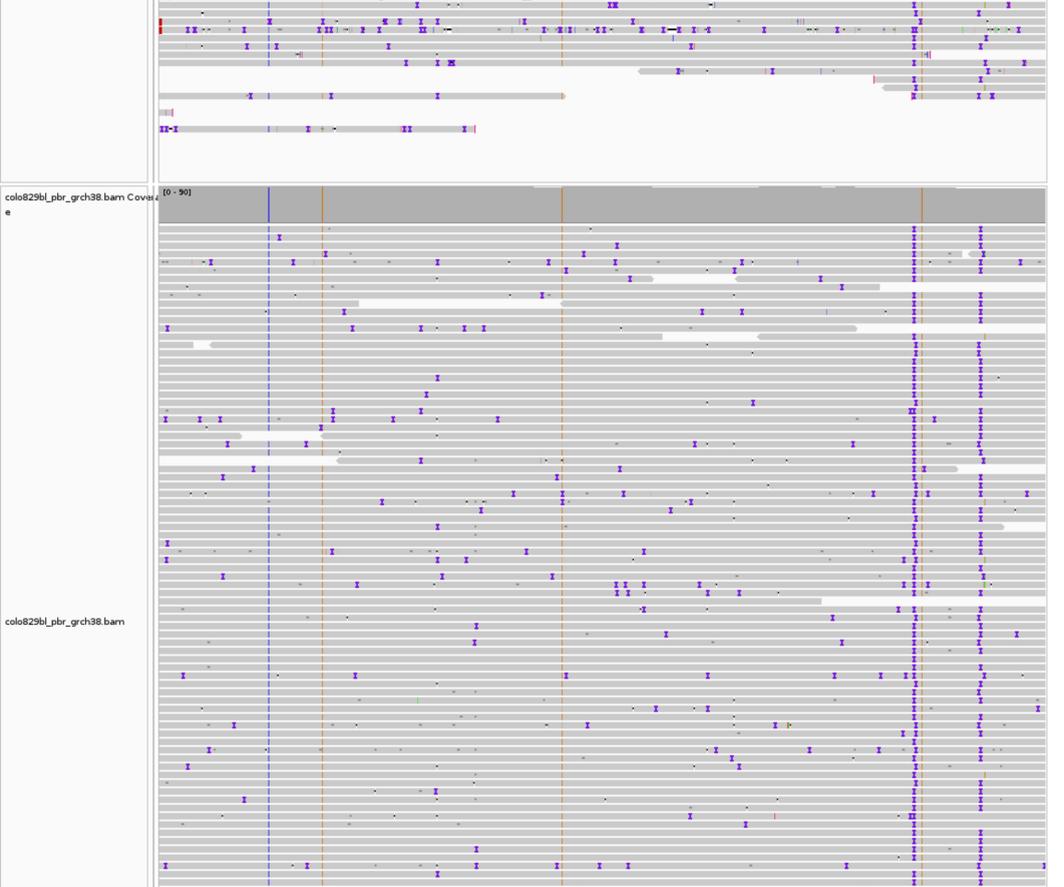
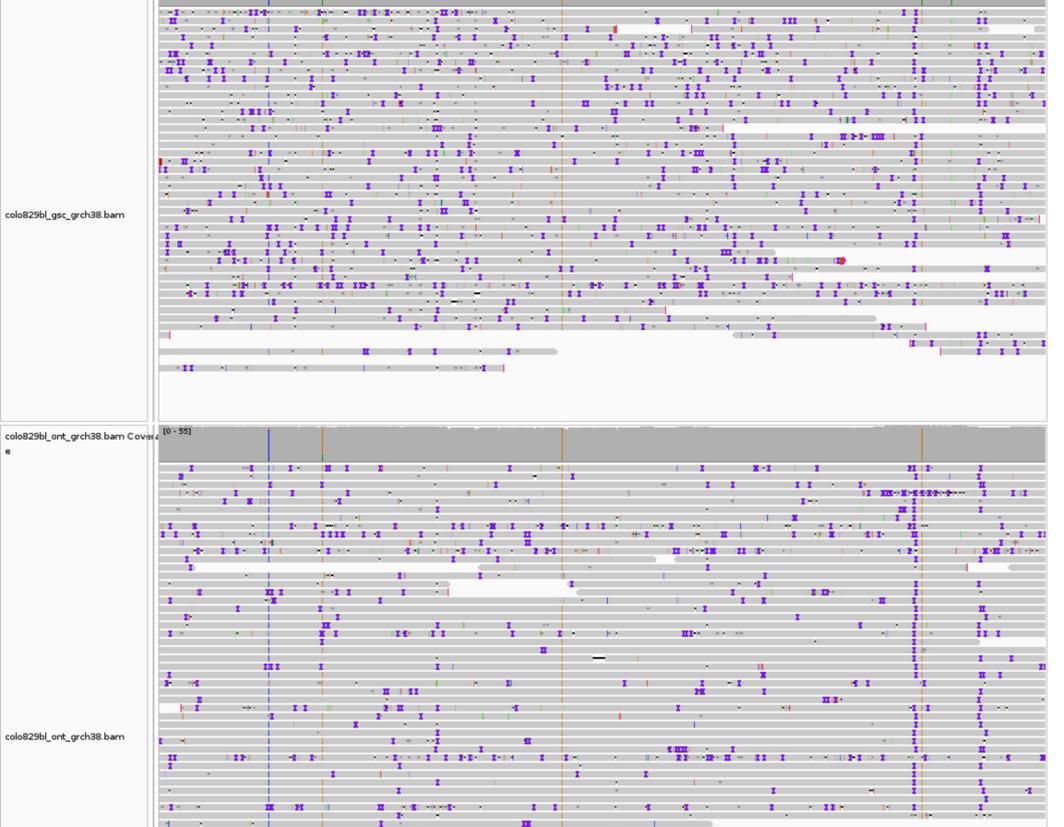
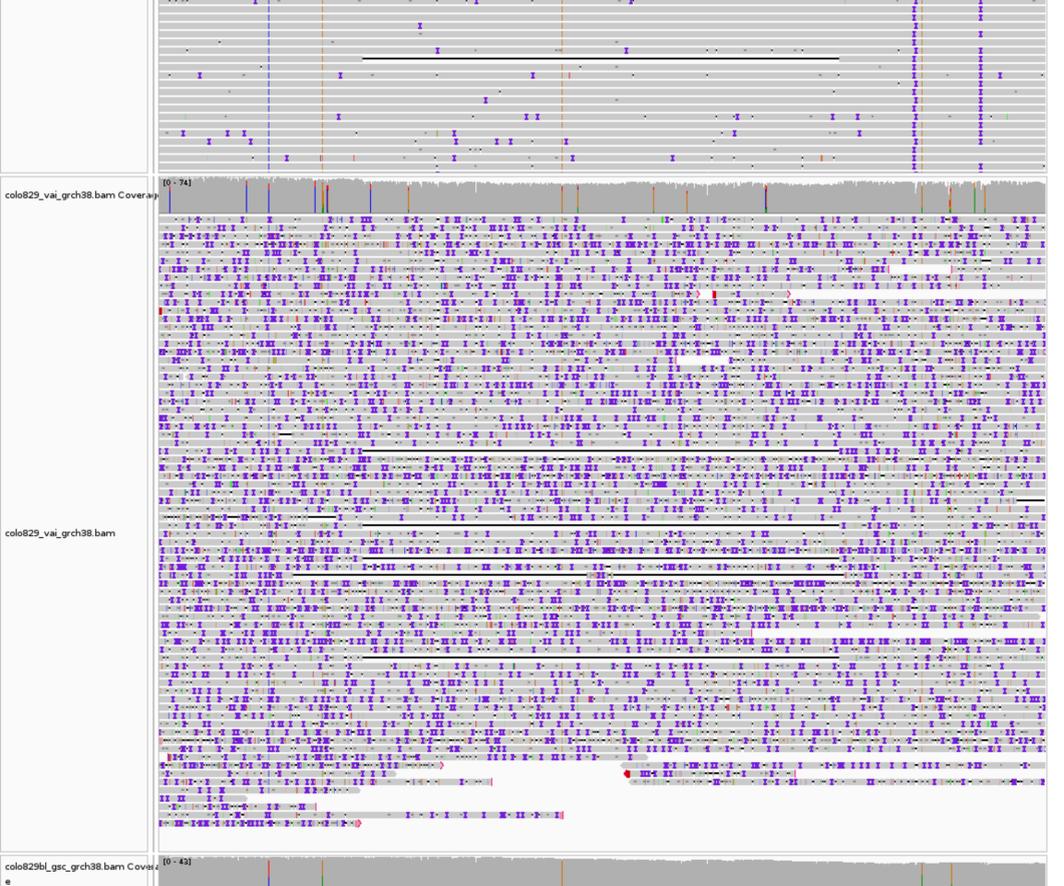
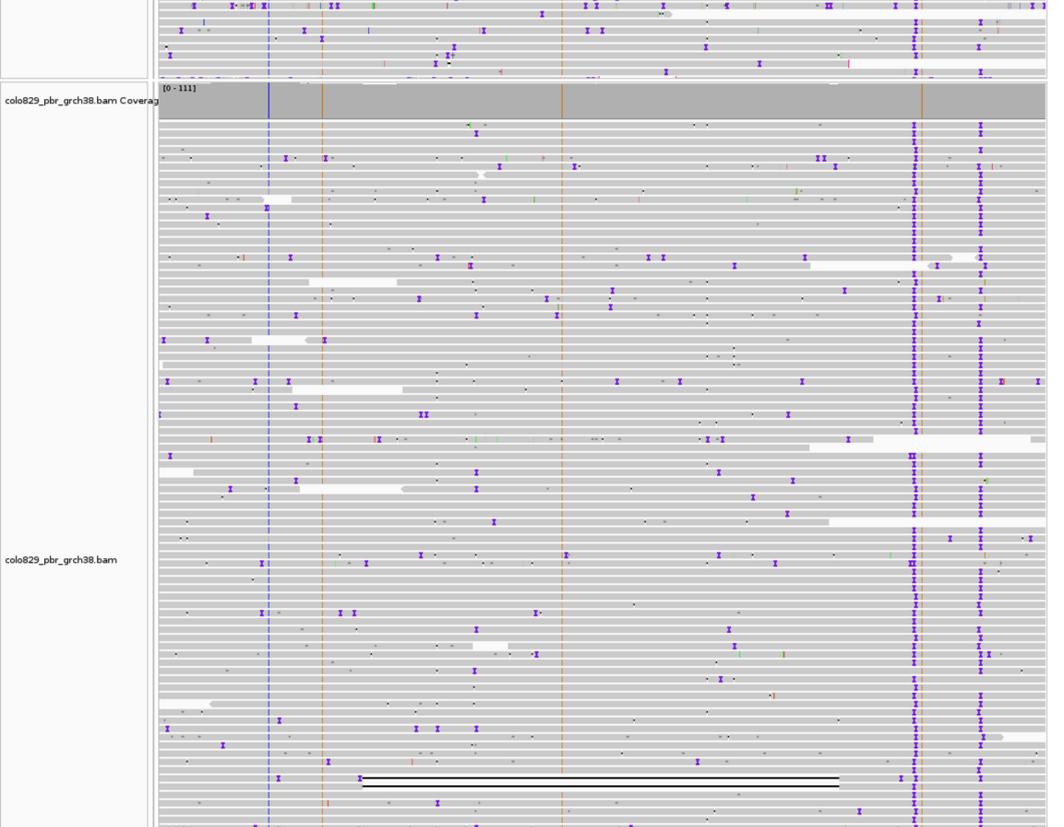
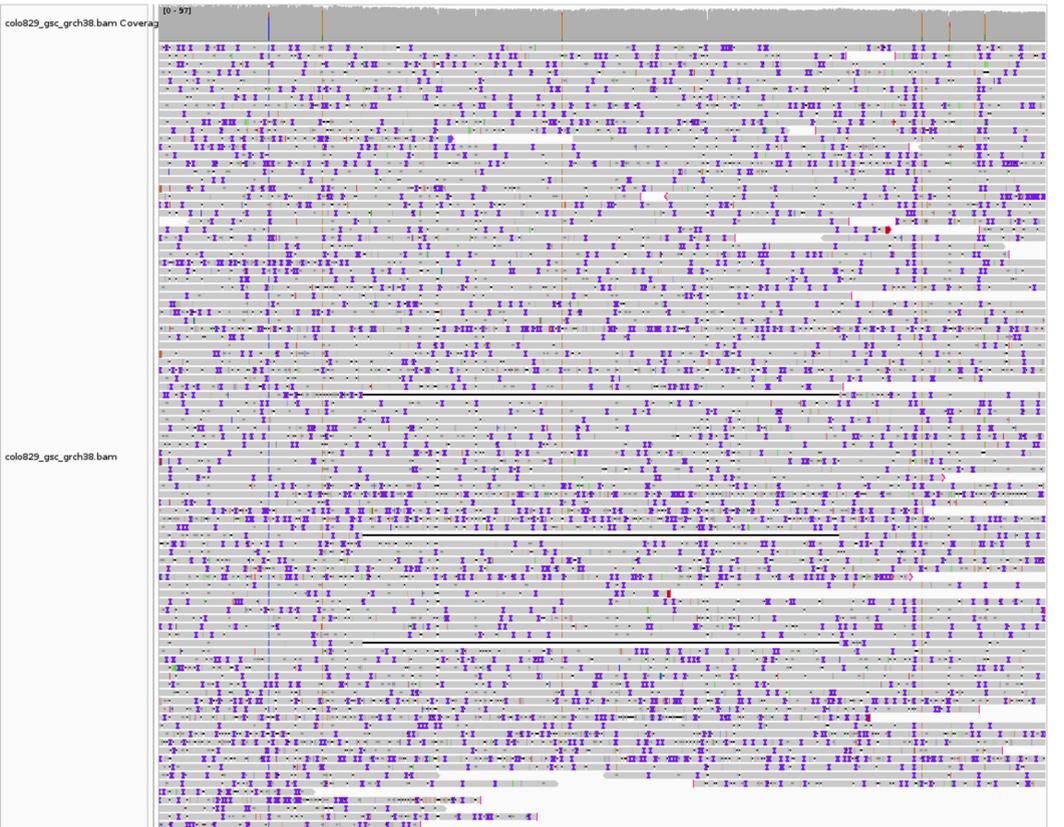


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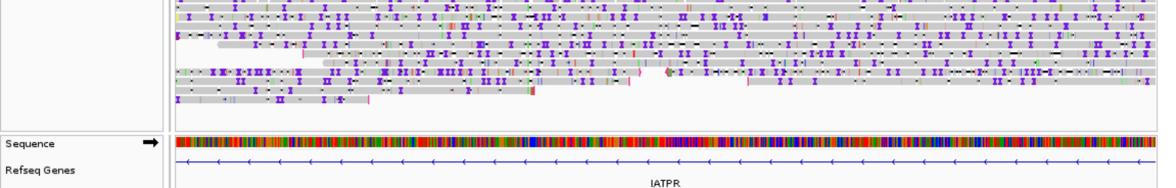
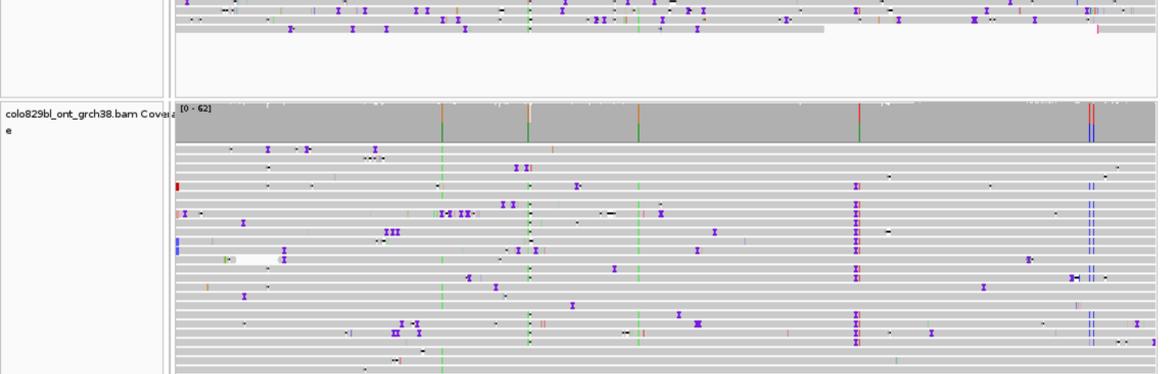
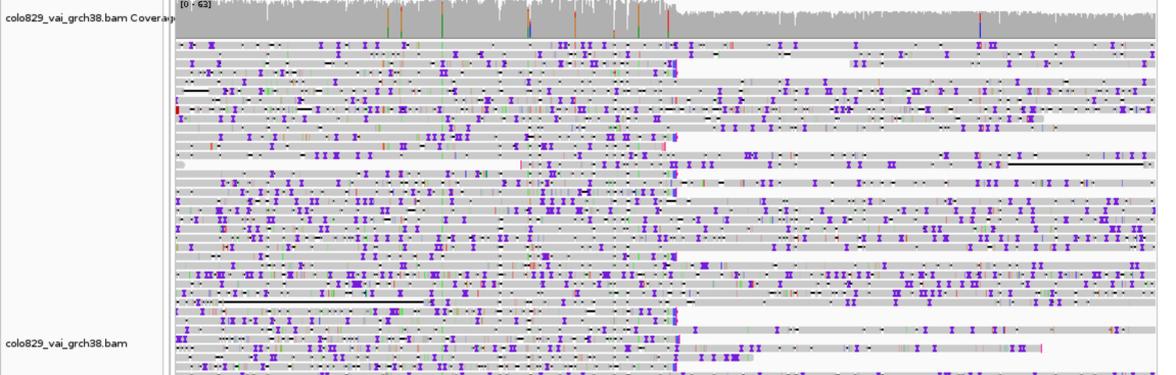
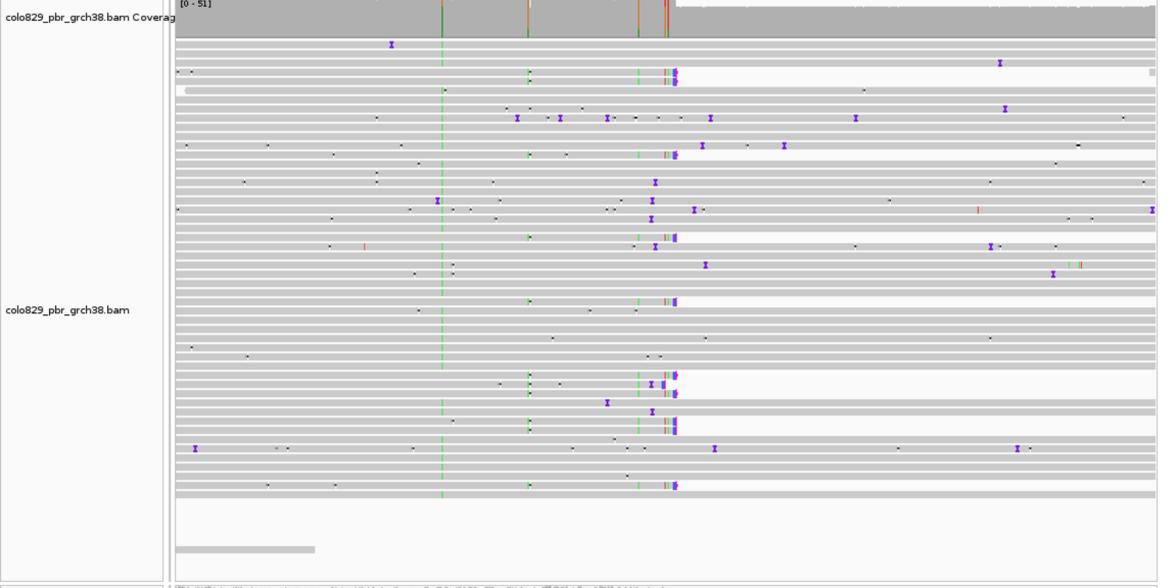
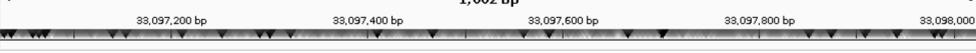
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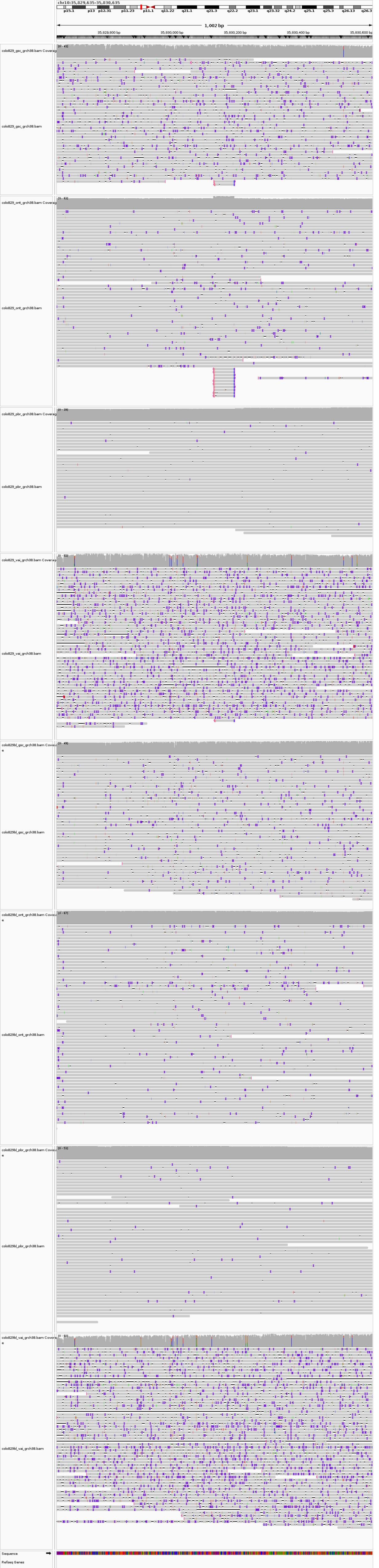


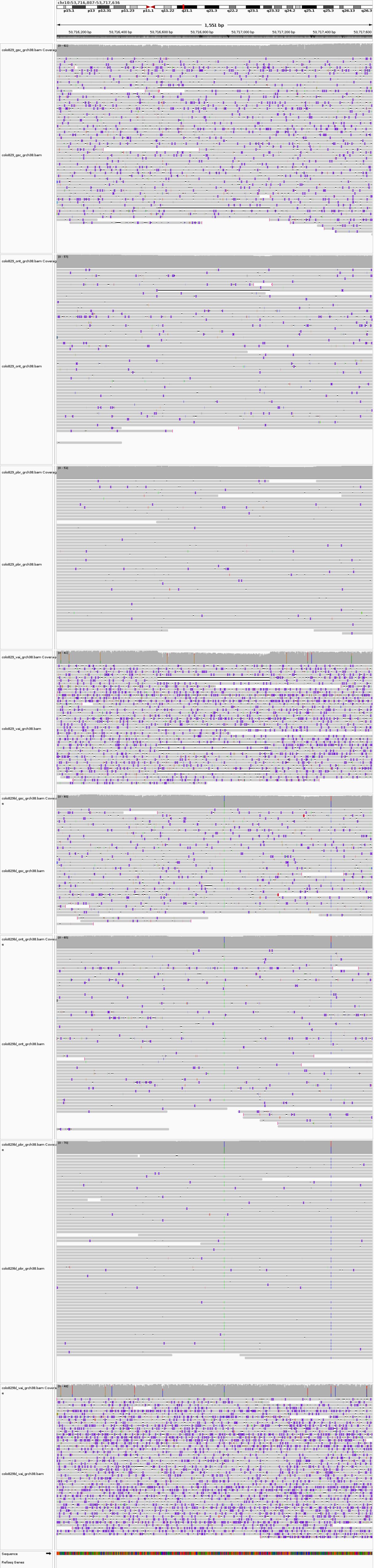
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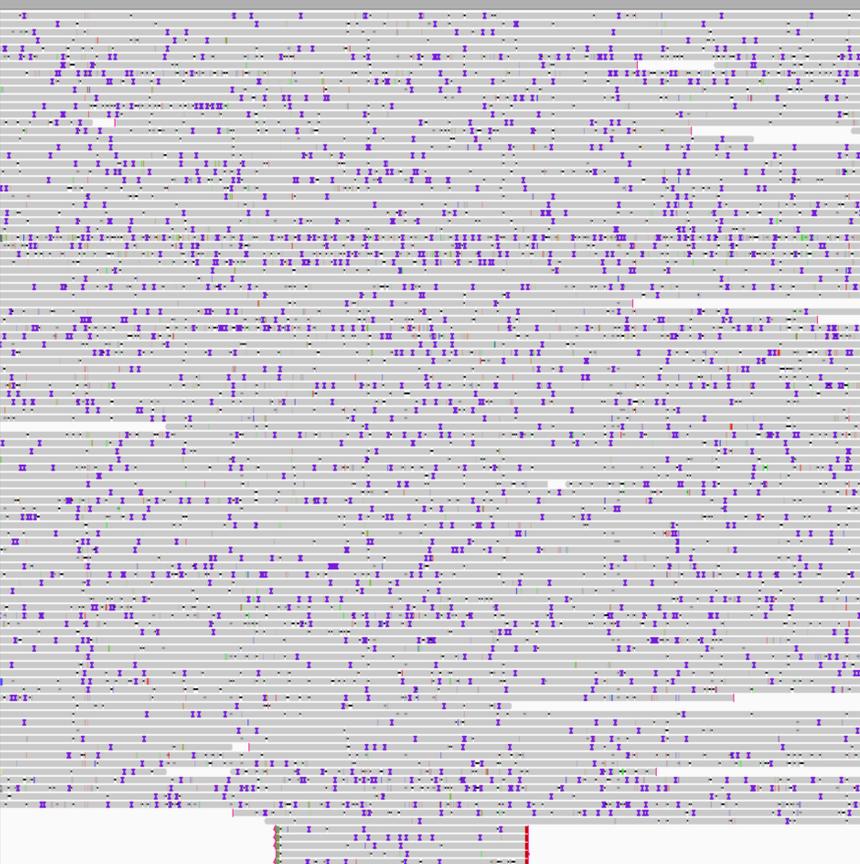
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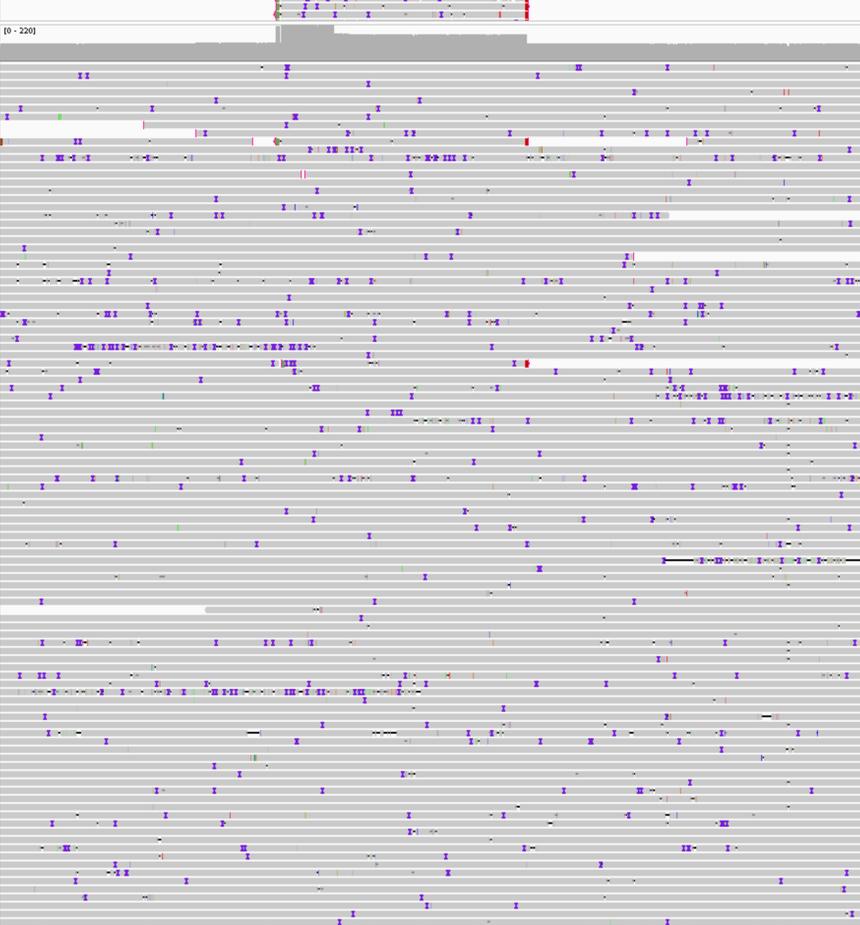




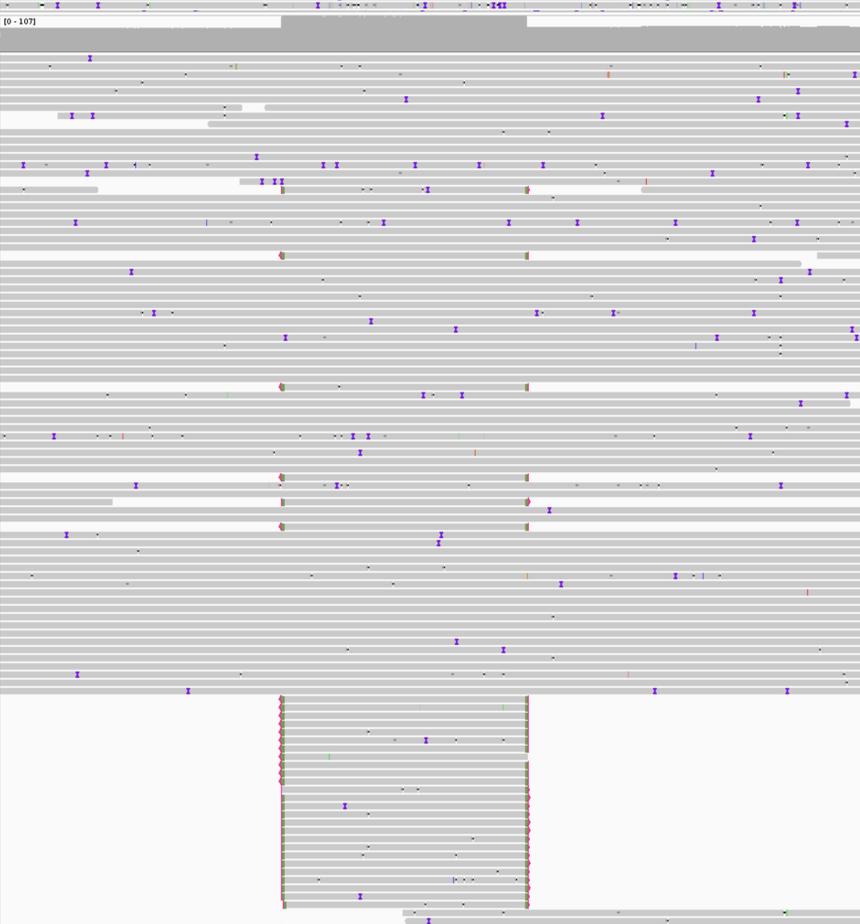
colo829\_gsc\_grch38 bam Coverage  
colo829\_gsc\_grch38 bam



colo829\_ort\_grch38 bam Coverage  
colo829\_ort\_grch38 bam



colo829\_pbr\_grch38 bam Coverage  
colo829\_pbr\_grch38 bam



colo829\_val\_grch38 bam Coverage  
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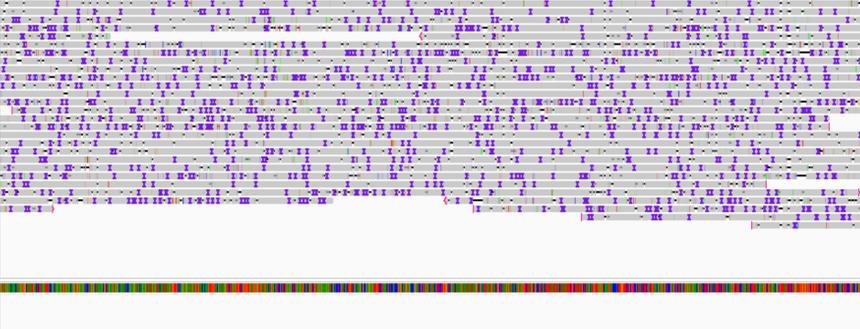
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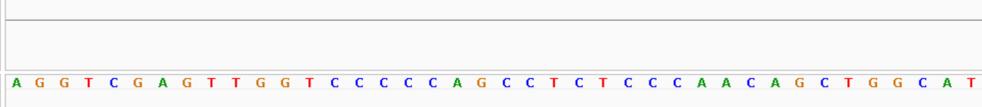
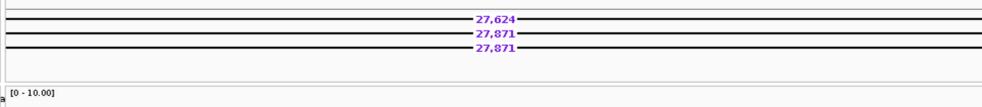
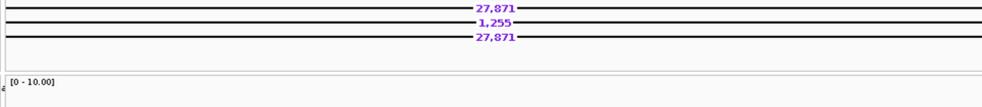
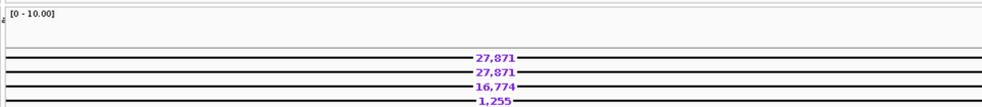
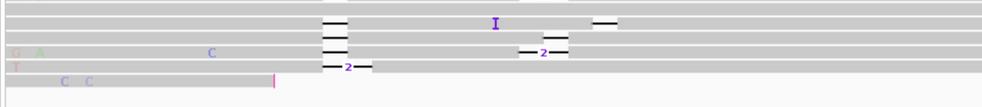
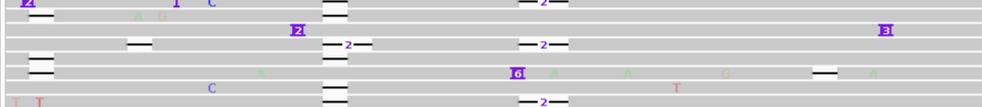
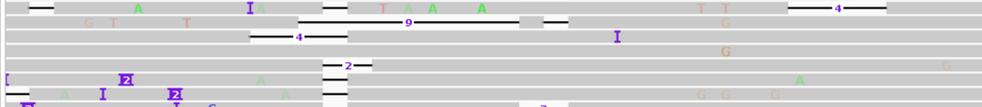
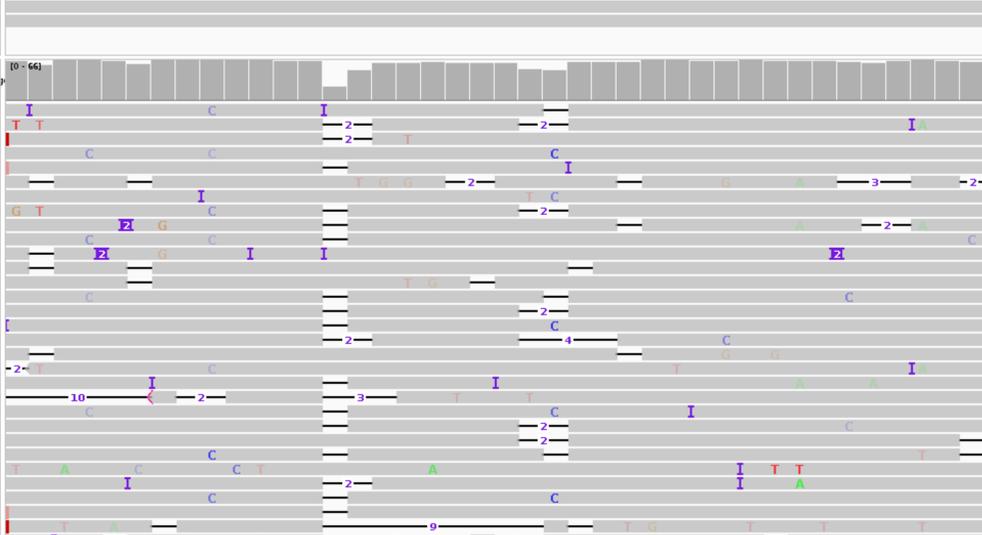
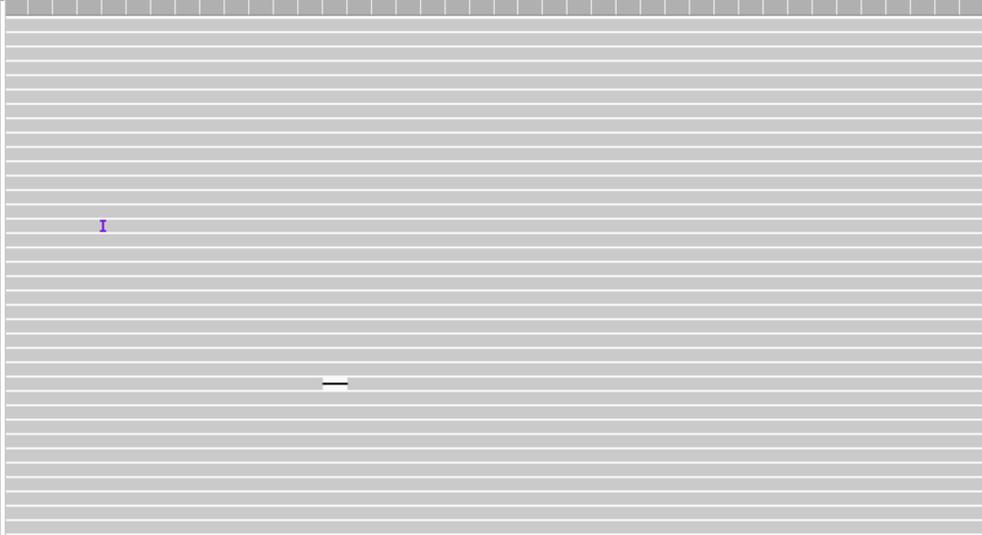
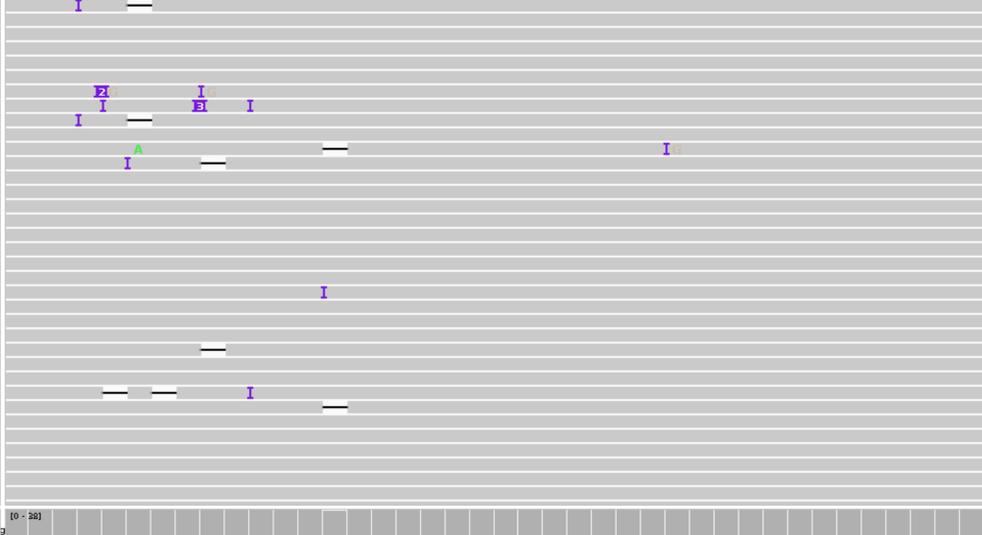
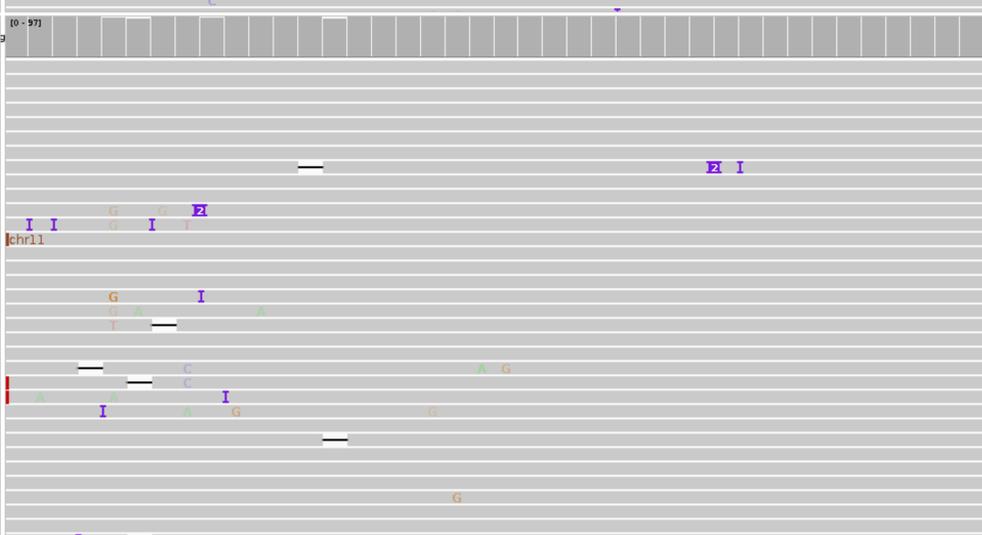
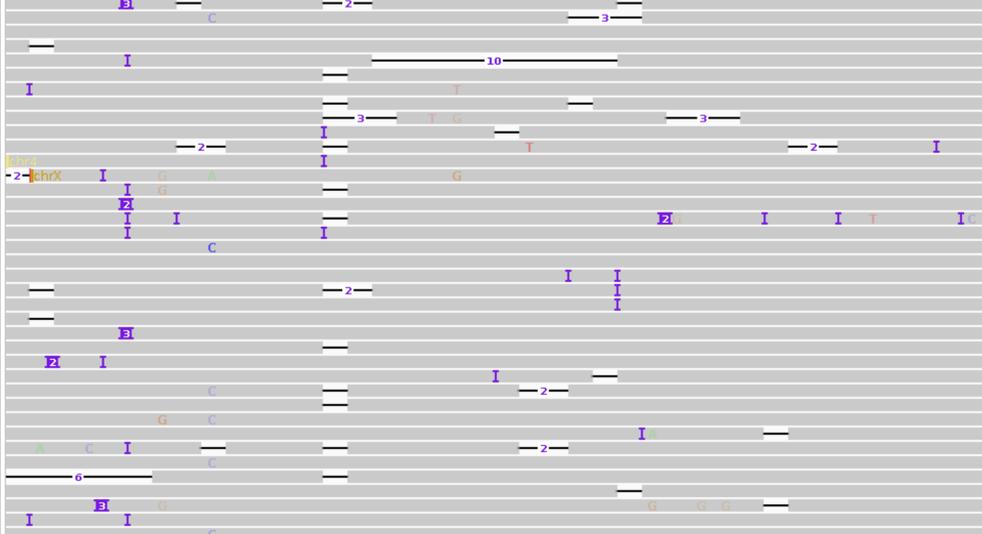
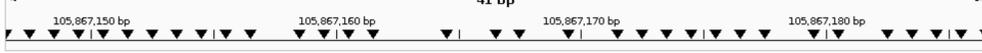


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colo829l\_pbr\_grch38 bam



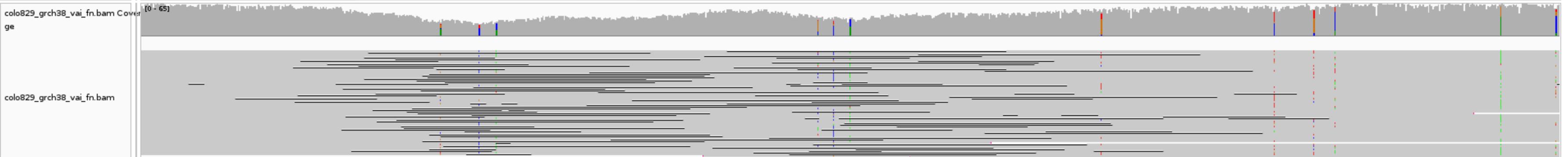
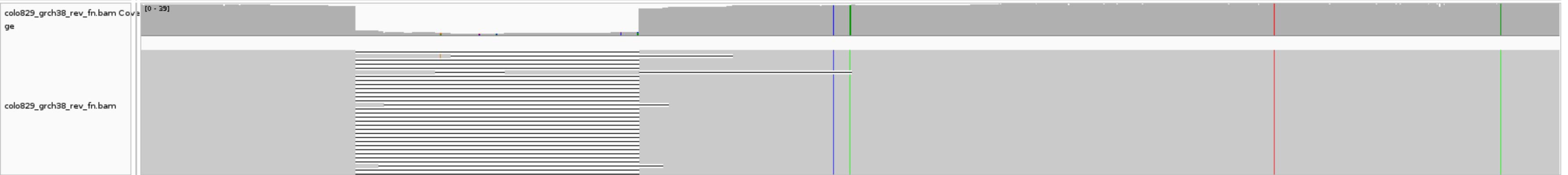
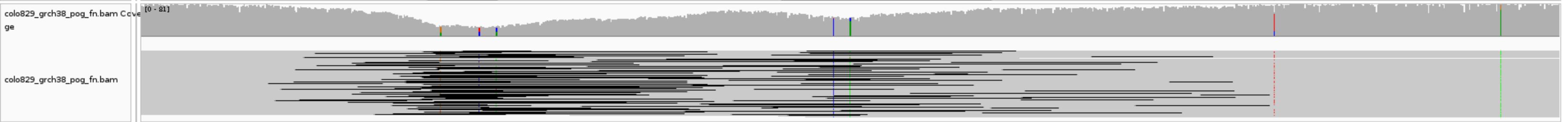
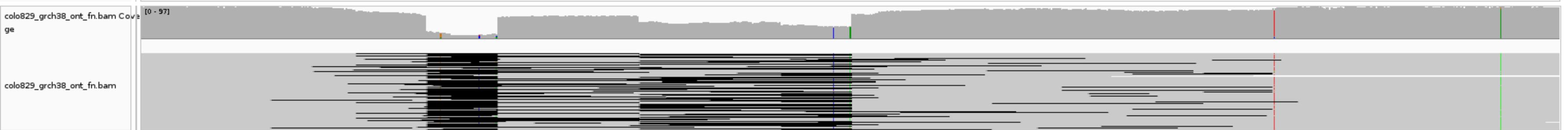
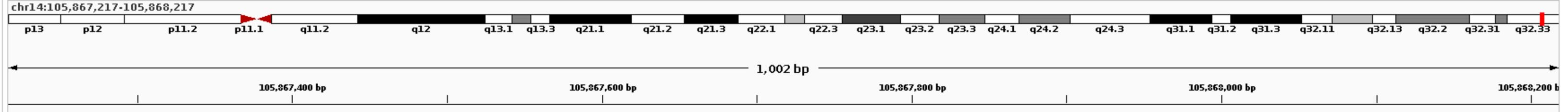
Sequence  
RefSeq Genes





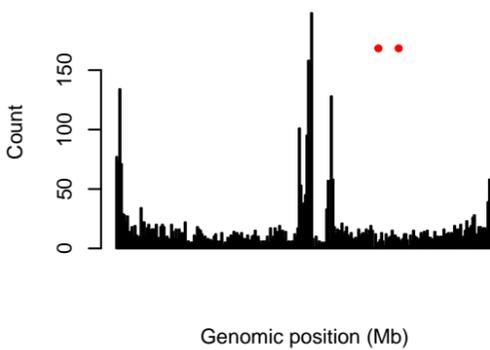
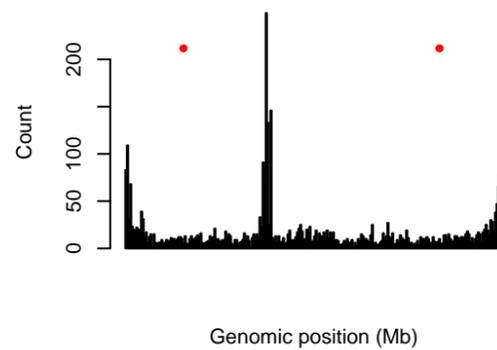
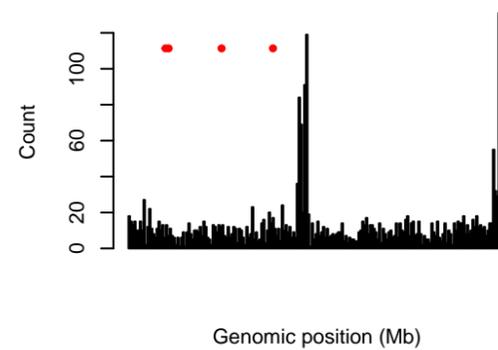
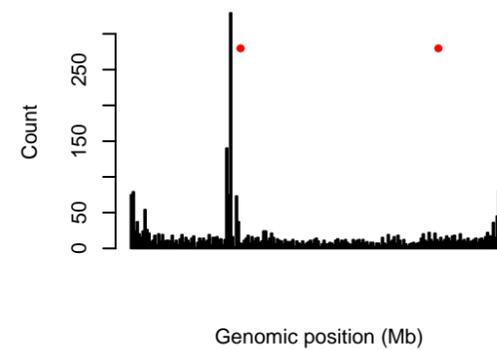
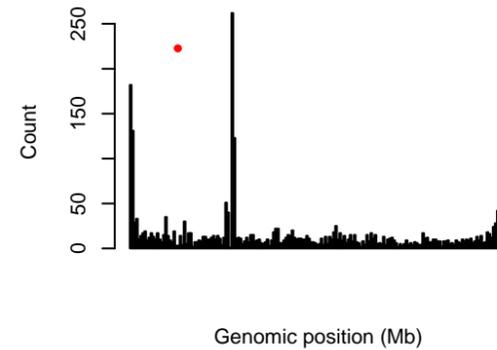
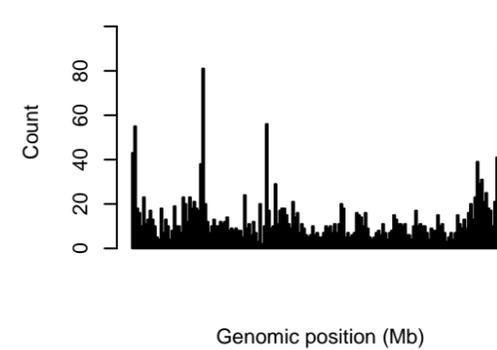
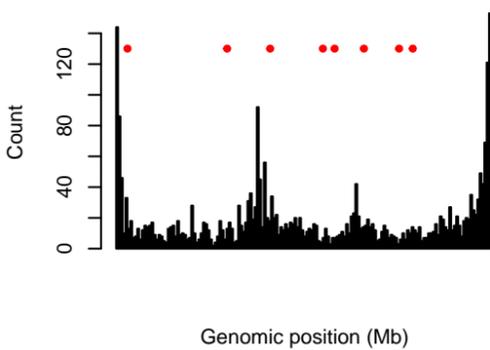
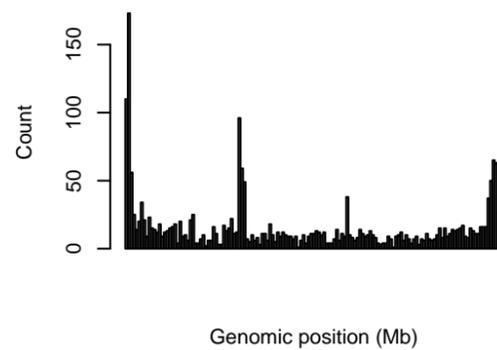
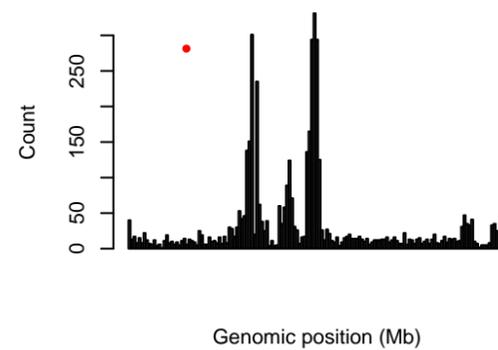
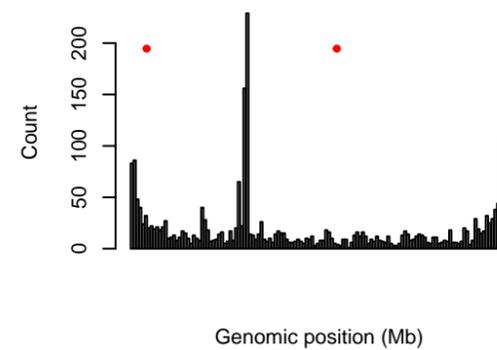
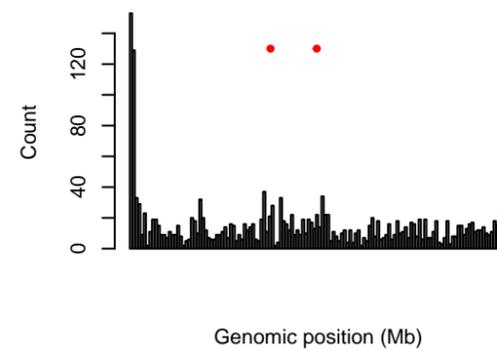
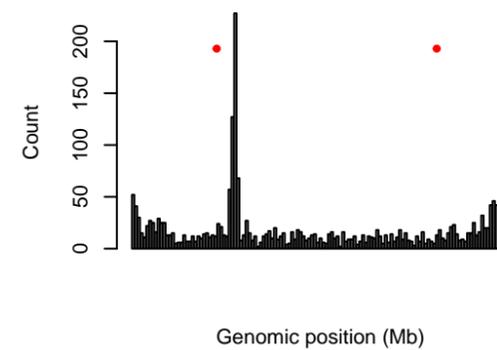
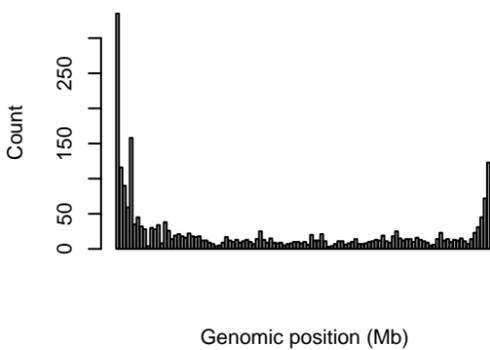
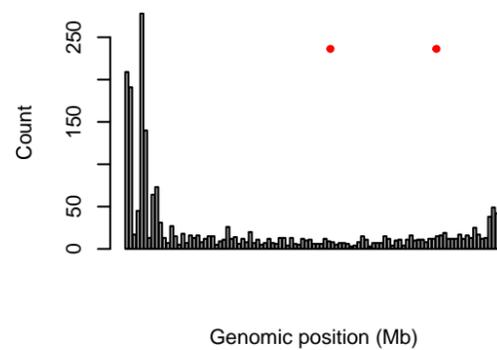
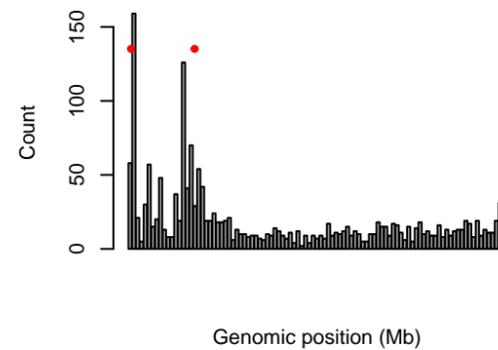
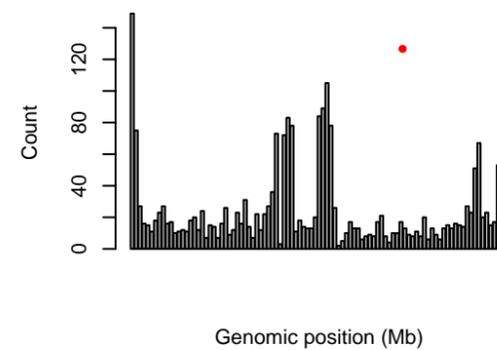
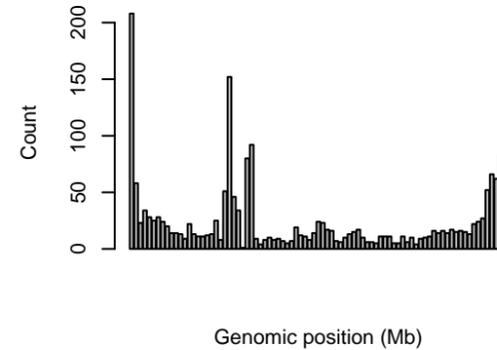
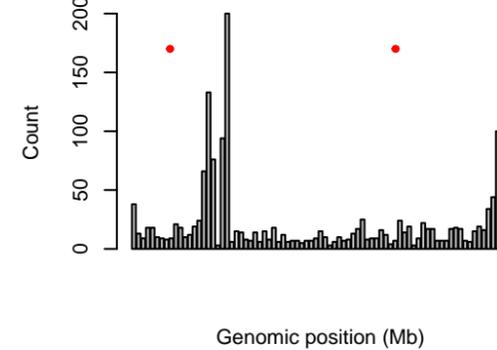
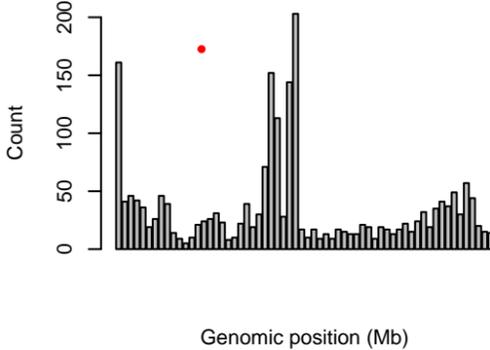
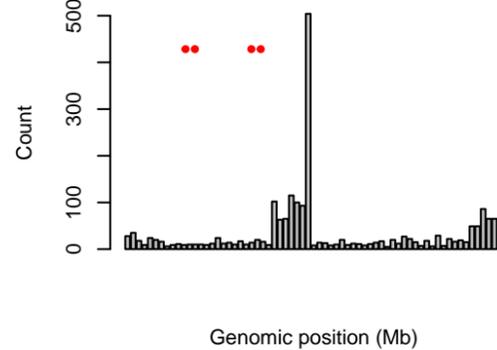
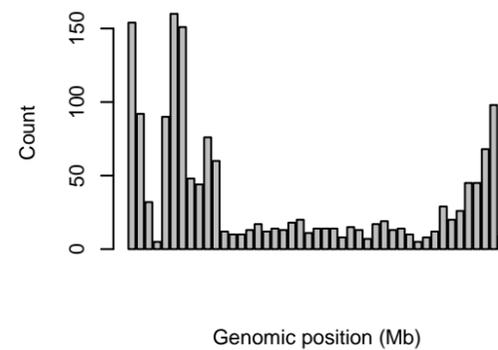
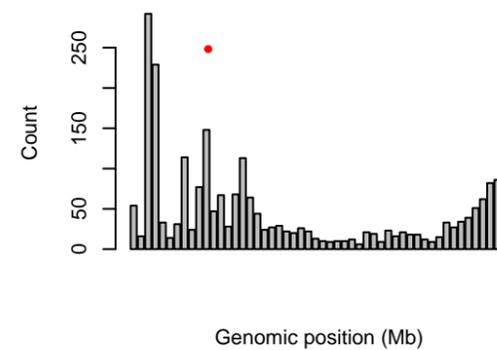
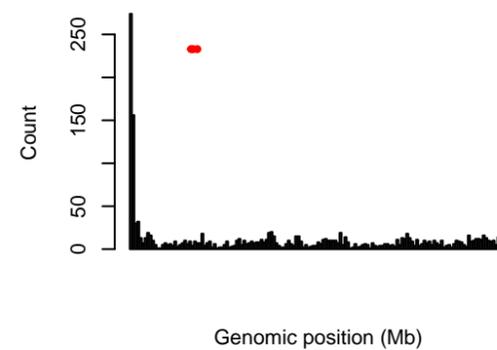
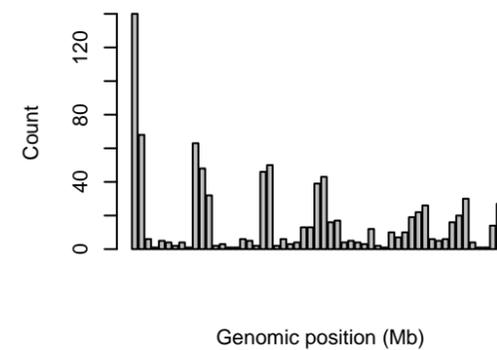
Sequence → A G G T C G A G T T G G T C C C C C A G C C T C T C C C A A C A G C T G G C A T

Refseq Genes



### **Supplement figure 3**

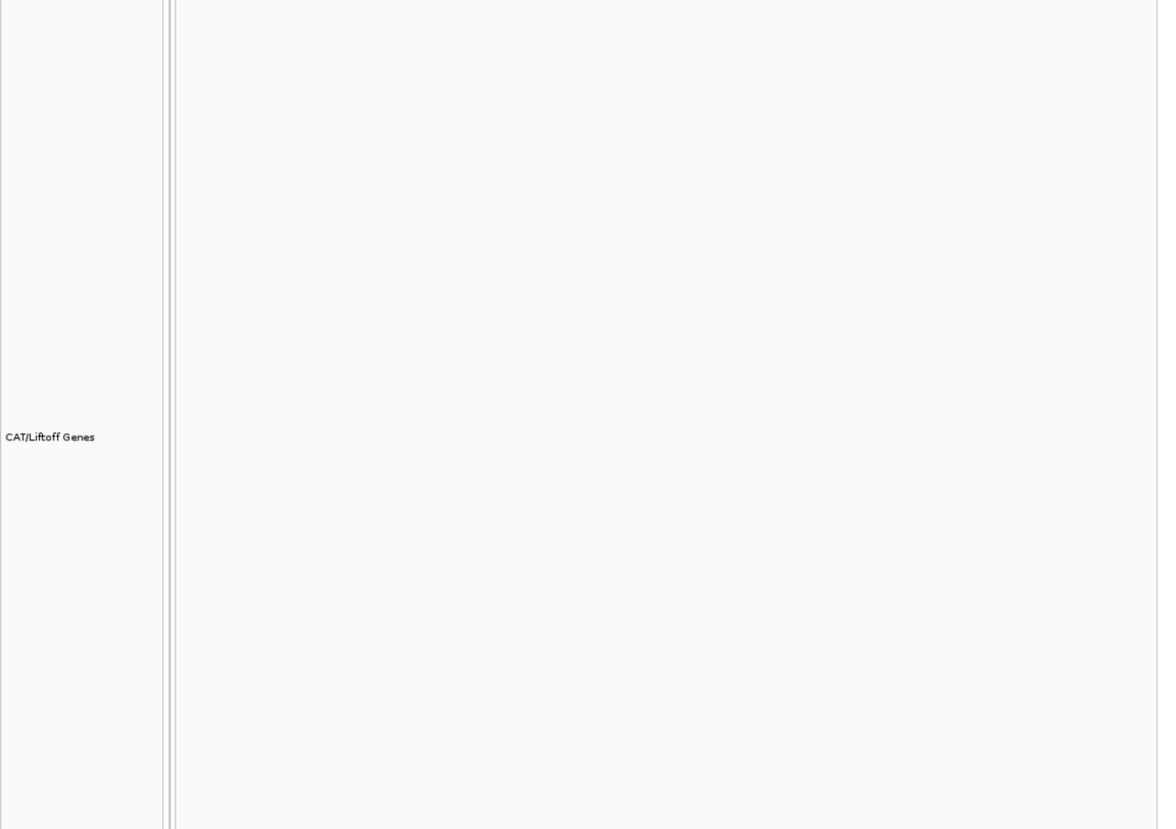
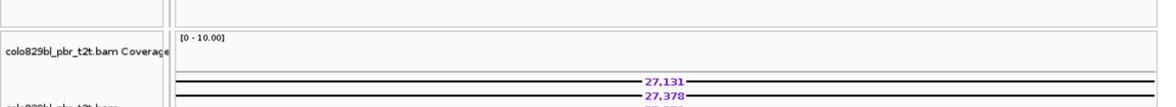
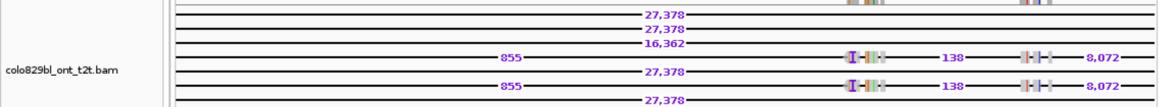
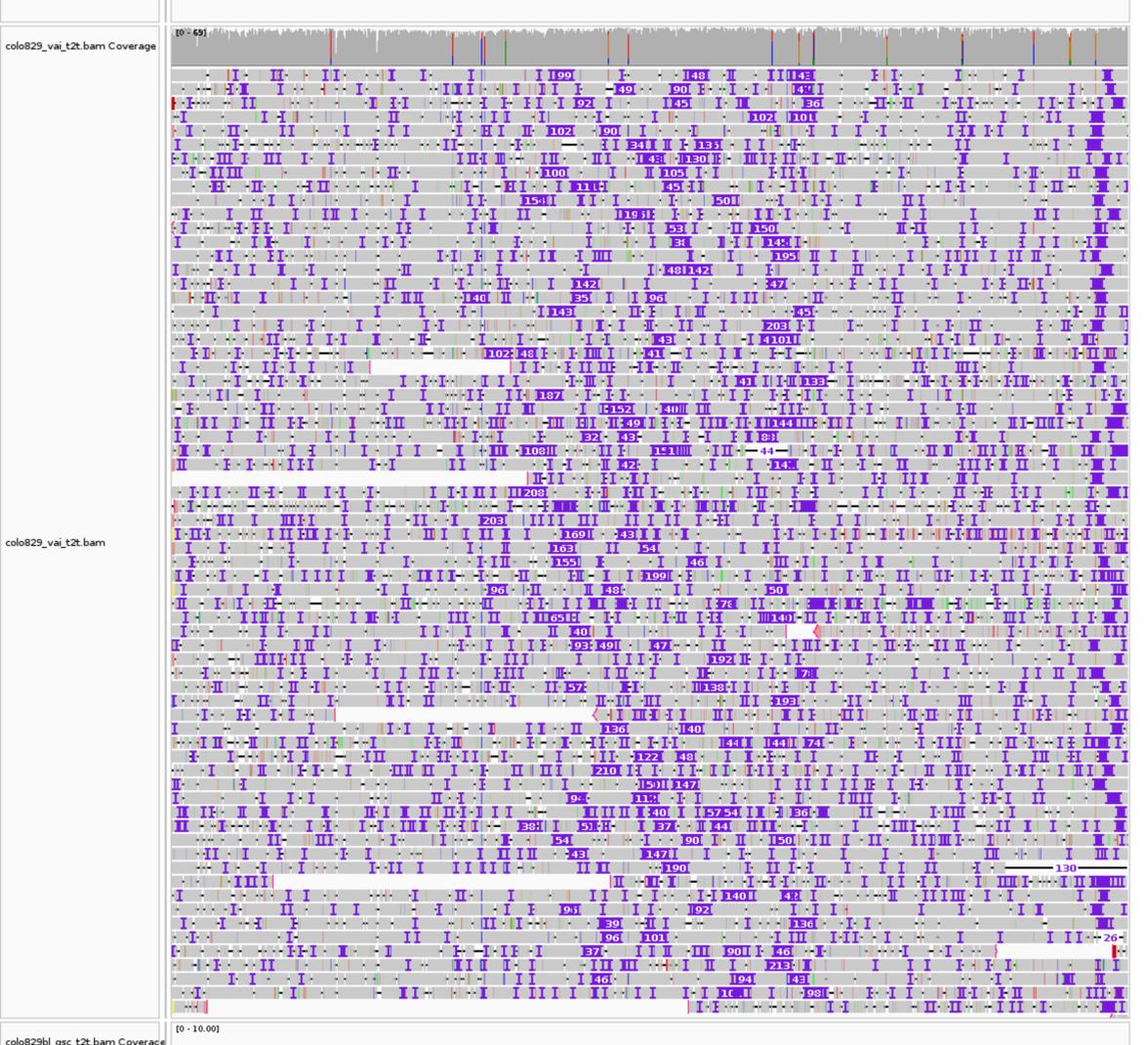
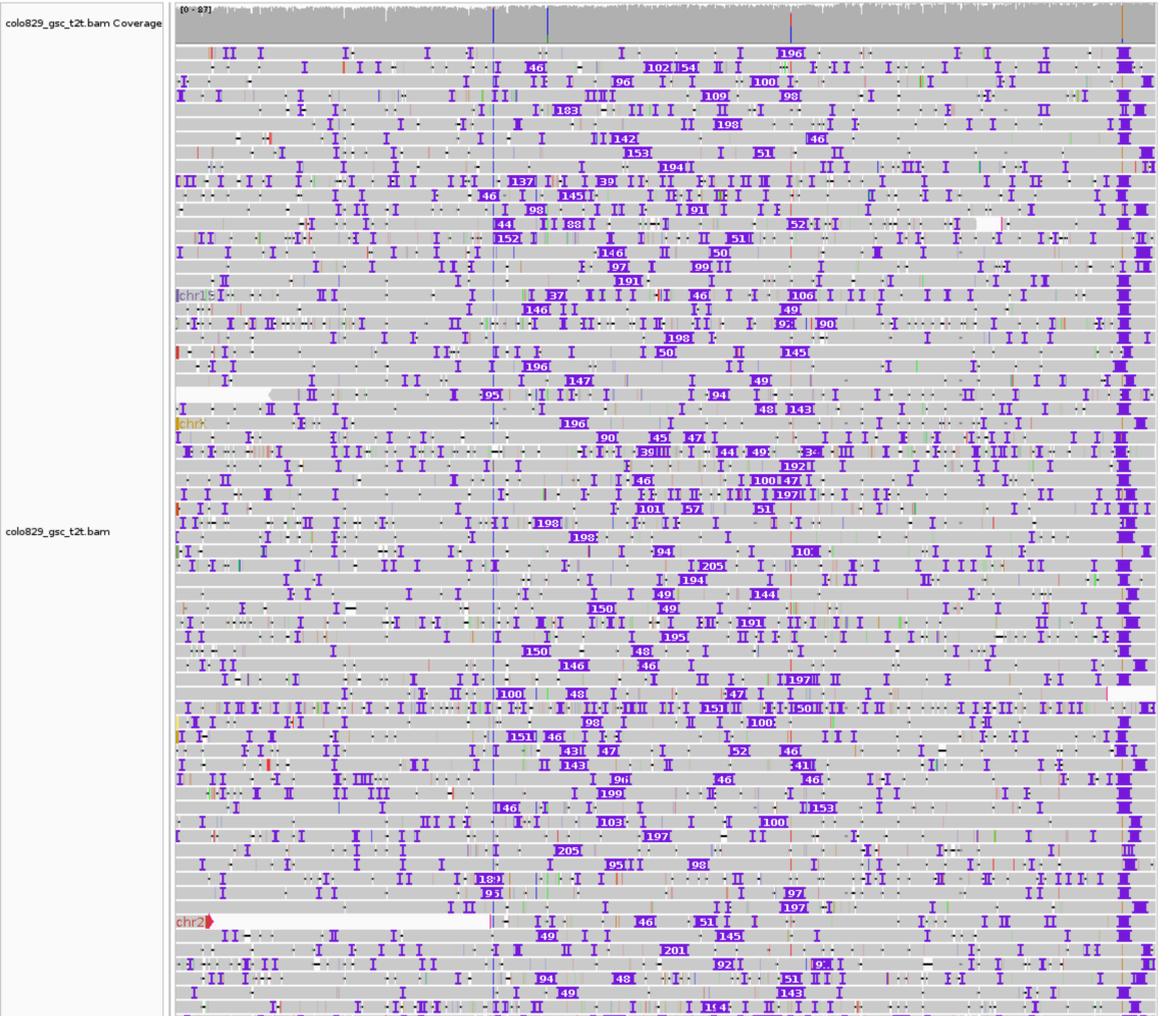
Distribution of SVs detected in the COLO829-T2T analysis. All 22 autosomes, and two sex chromosomes (X and Y) are shown. Beneath the genomic position the ideogram for each chromosome is shown. The red dots depict 43 somatic SVs described in Supplementary table 7.

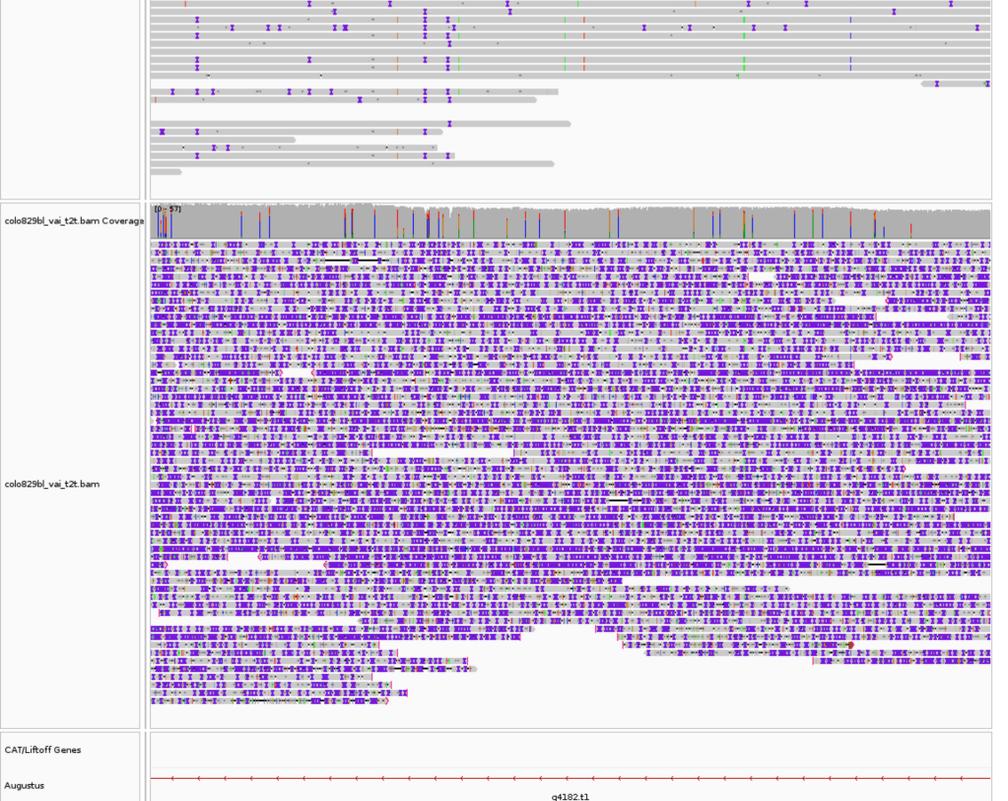
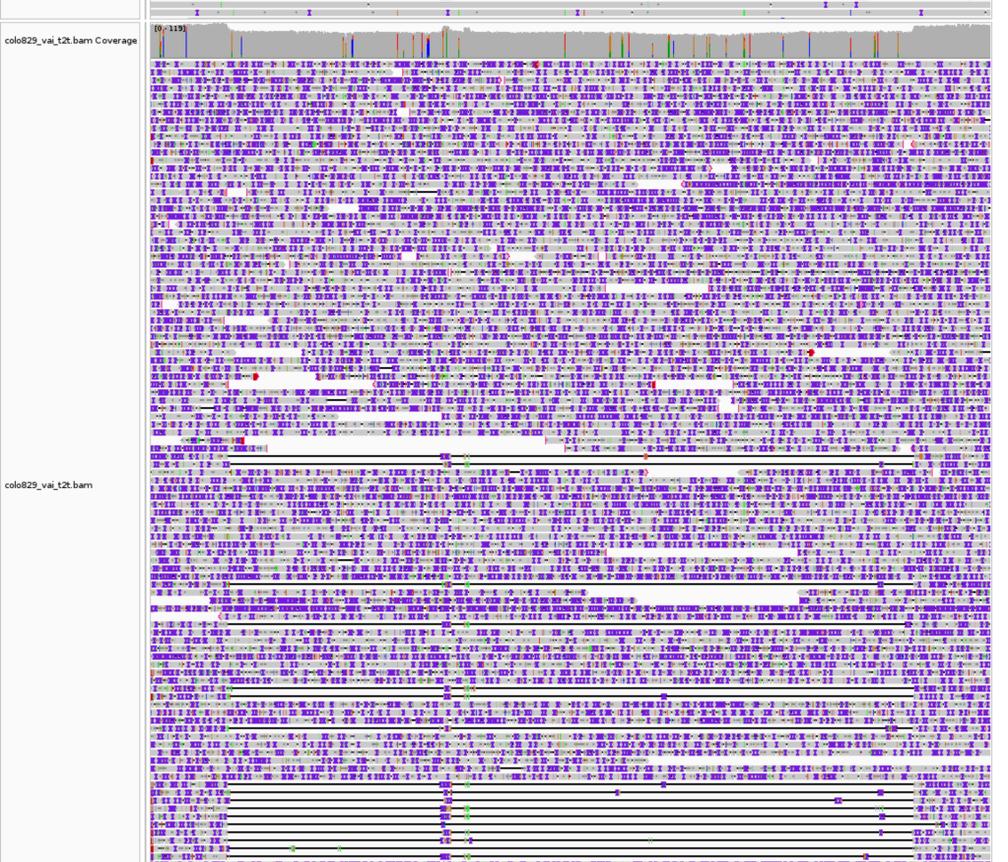
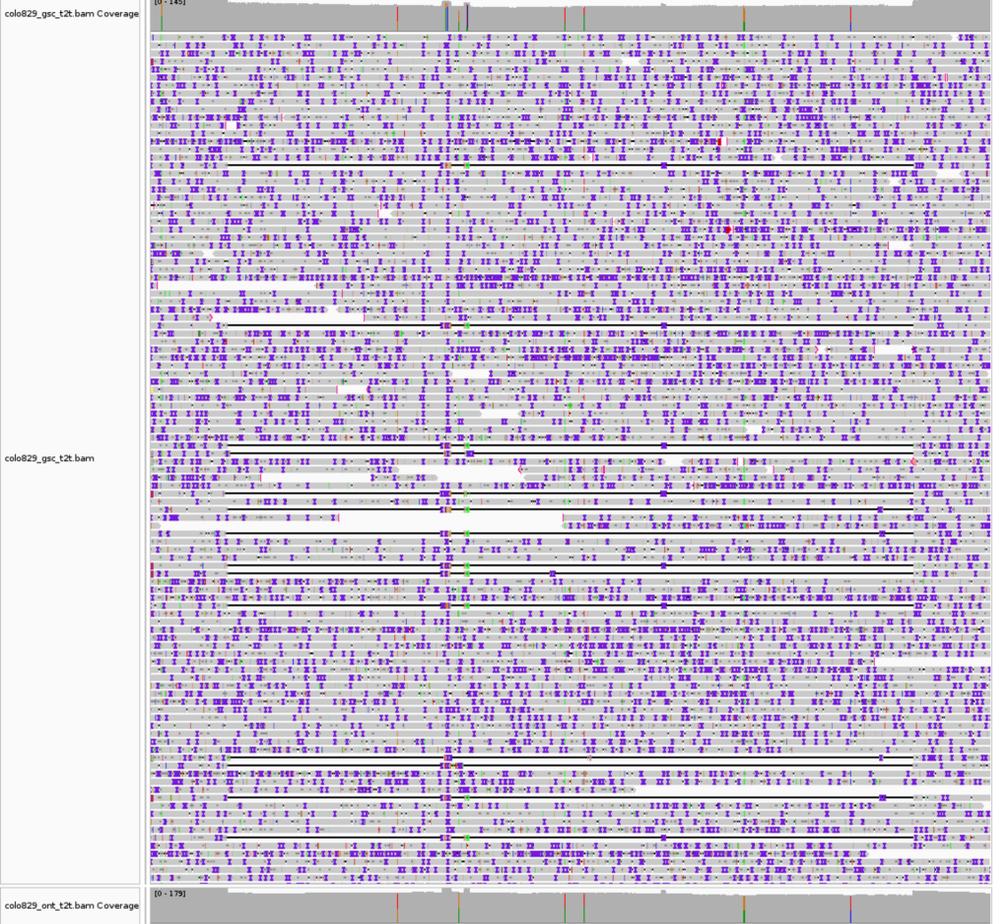
**SV distribution in CHM13-T2T chr1****SV distribution in CHM13-T2T chr2****SV distribution in CHM13-T2T chr3****SV distribution in CHM13-T2T chr4****SV distribution in CHM13-T2T chr5****SV distribution in CHM13-T2T chr6****SV distribution in CHM13-T2T chr7****SV distribution in CHM13-T2T chr8****SV distribution in CHM13-T2T chr9****SV distribution in CHM13-T2T chr10****SV distribution in CHM13-T2T chr11****SV distribution in CHM13-T2T chr12****SV distribution in CHM13-T2T chr13****SV distribution in CHM13-T2T chr14****SV distribution in CHM13-T2T chr15****SV distribution in CHM13-T2T chr16****SV distribution in CHM13-T2T chr17****SV distribution in CHM13-T2T chr18****SV distribution in CHM13-T2T chr19****SV distribution in CHM13-T2T chr20****SV distribution in CHM13-T2T chr21****SV distribution in CHM13-T2T chr22****SV distribution in CHM13-T2T chrX****SV distribution in CHM13-T2T chrY**

Supplementary Figure 4: IGV screenshot of the Structural Variants (SVs) labeled as False Positives (FP) according to the Valle-Inclan benchmark (based on read-names as the coordinates are in CHM13-T2T)

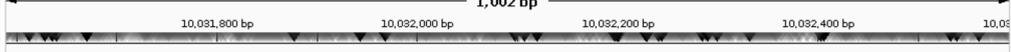
- A) A 57 bp INS located in chr2:45420200 wrongly labeled as FP
- B) A 71 bp INS located in chr7:6090247 wrongly labeled as FP
- C) A 1,357 bp DEL located in chr12:36018420 wrongly labeled as FP
- D) A 197 bp INS located in chr14:100139043 wrongly labeled as FP
- E) A 4,570 bp DEL located in chr15:21201145 wrongly labeled as FP
- F) A BND located in chr18:10032090 (2<sup>nd</sup> breakpoint in chr10:7593331) wrongly labeled as FP, breakpoint 1
- G) A BND located in chr18:10032090 (2<sup>nd</sup> breakpoint in chr10:7593331) wrongly labeled as FP, breakpoint 2
- H) A BND located in chr19:17420891 (2<sup>nd</sup> breakpoint in chr10:7017345) wrongly labeled as FP, breakpoint 1
- I) A BND located in chr19:17420891 (2<sup>nd</sup> breakpoint in chr10:7017345) wrongly labeled as FP, breakpoint 2
- J) A 1,878 bp DEL located in chr20:29423649 wrongly labeled as FP







chr18:10,031,590-10,032,590  
p11.31 p11.22 p11.1 q11.2 q12.1 q12.2 q12.3 q21.1 q21.2 q21.31 q21.33 q22.1 q22.2 q23



colo829\_gsc\_t2t.bam Coverage  
colo829\_gsc\_t2t.bam



colo829\_ont\_t2t.bam Coverage  
colo829\_ont\_t2t.bam



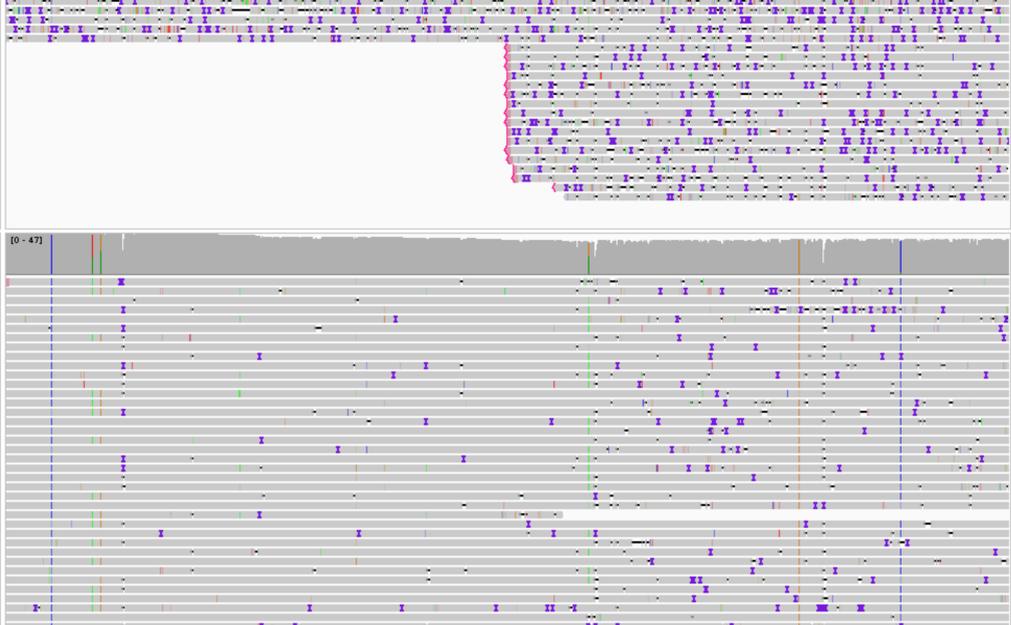
colo829\_pbr\_t2t.bam Coverage  
colo829\_pbr\_t2t.bam



colo829\_val\_t2t.bam Coverage  
colo829\_val\_t2t.bam



colo829b\_gsc\_t2t.bam Coverage  
colo829b\_gsc\_t2t.bam



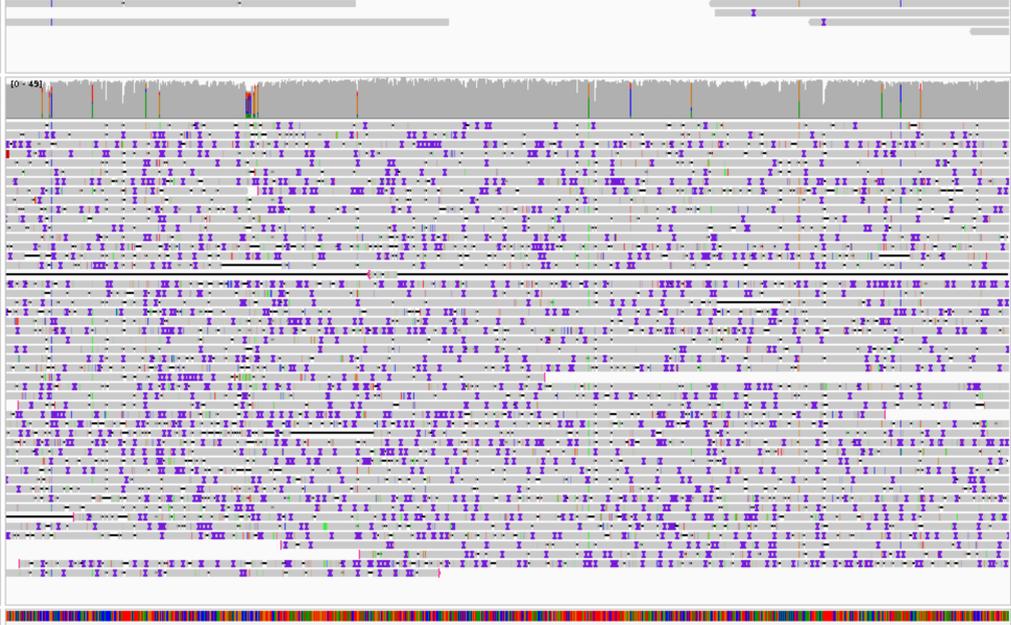
colo829b\_ont\_t2t.bam Coverage  
colo829b\_ont\_t2t.bam



colo829b\_pbr\_t2t.bam Coverage  
colo829b\_pbr\_t2t.bam



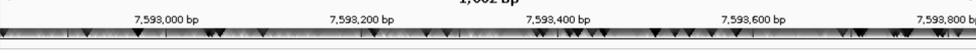
colo829b\_val\_t2t.bam Coverage  
colo829b\_val\_t2t.bam



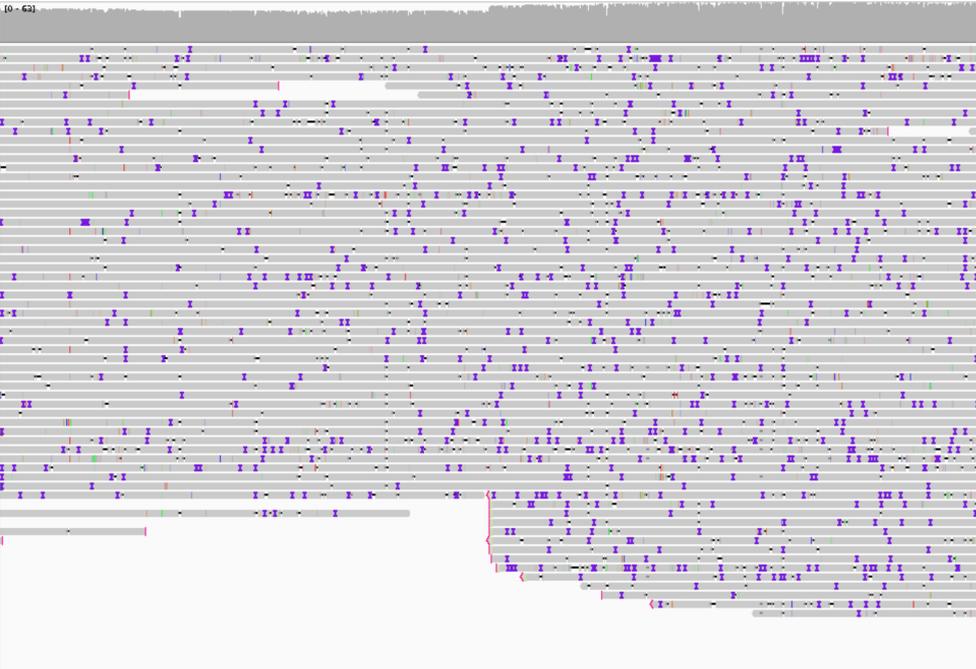
Sequence  
CAT/Liftoff Genes  
Augustus



chr10:7,592,831-7,593,831  
p15.1 p13 p12.2 p11.23 p11.1 q11.22 q21.1 q21.3 q22.2 q23.1 q23.32 q24.2 q25.1 q25.3 q26.13 q26.3



colo829\_gsc\_t2t.bam Coverage  
colo829\_gsc\_t2t.bam



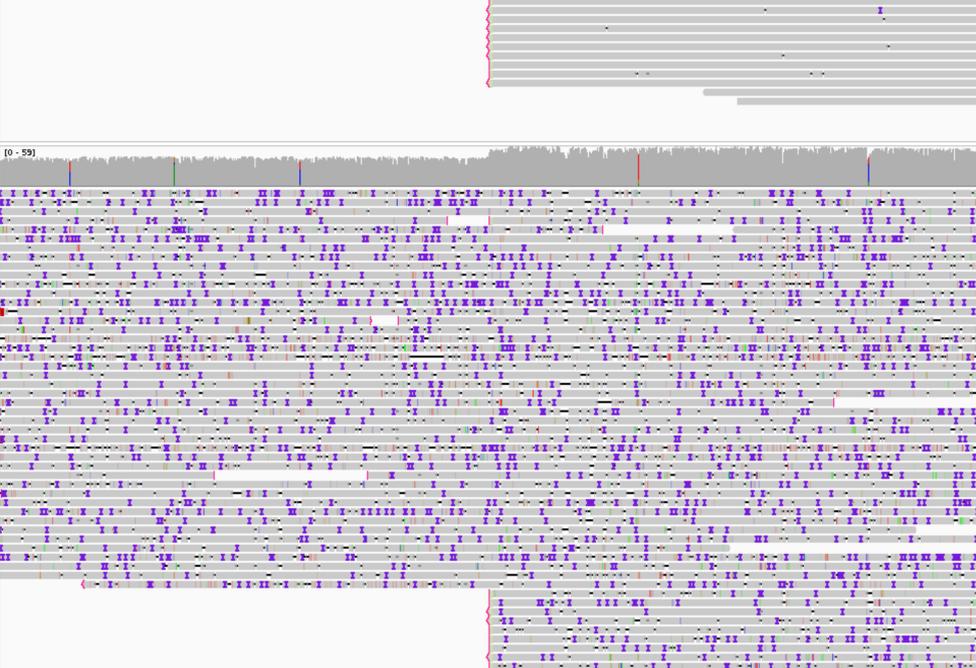
colo829\_ort\_t2t.bam Coverage  
colo829\_ort\_t2t.bam



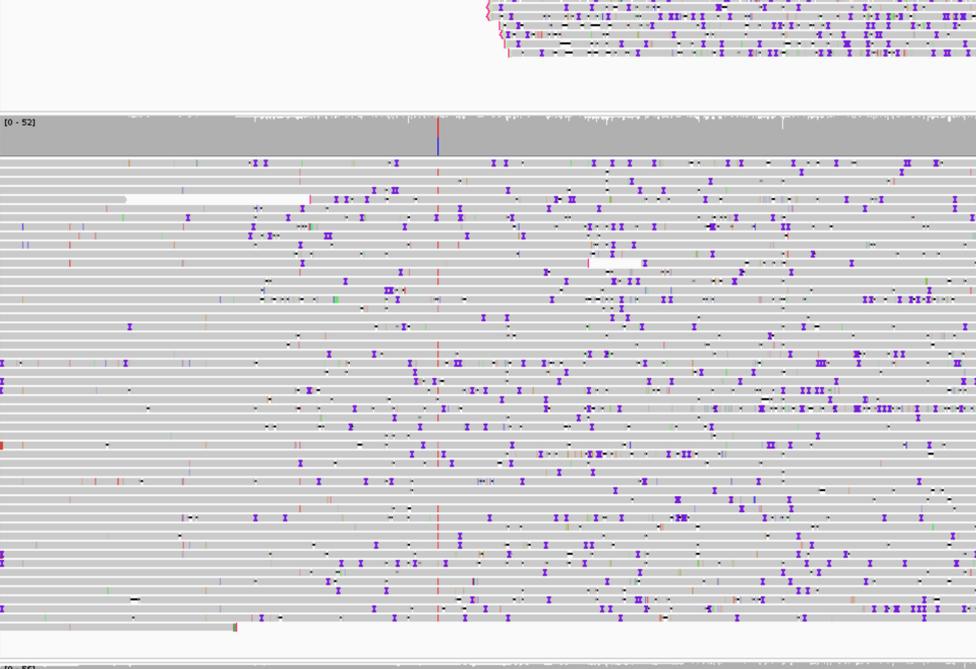
colo829\_pbr\_t2t.bam Coverage  
colo829\_pbr\_t2t.bam



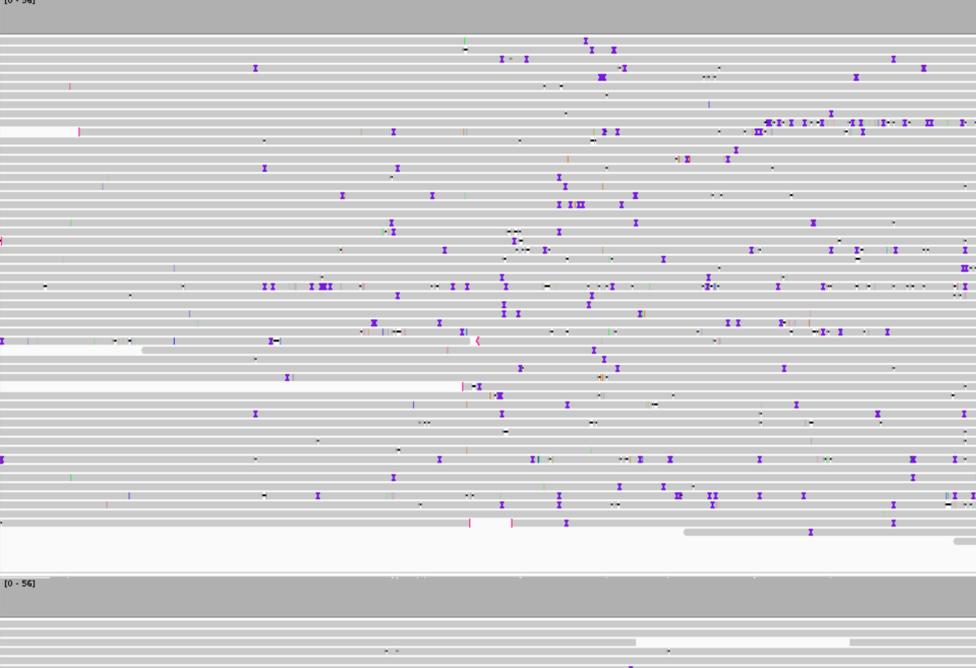
colo829\_val\_t2t.bam Coverage  
colo829\_val\_t2t.bam



colo829bl\_gsc\_t2t.bam Coverage  
colo829bl\_gsc\_t2t.bam



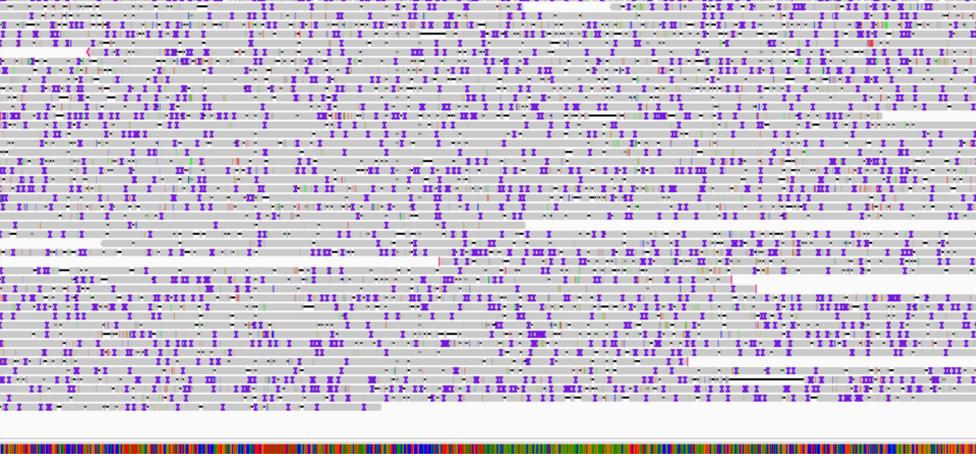
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colo829bl\_ort\_t2t.bam



colo829bl\_pbr\_t2t.bam Coverage  
colo829bl\_pbr\_t2t.bam



colo829bl\_val\_t2t.bam Coverage  
colo829bl\_val\_t2t.bam



Sequence  
CAT/Liftoff Genes  
Augustus



chr19:17,420,391-17,421,391

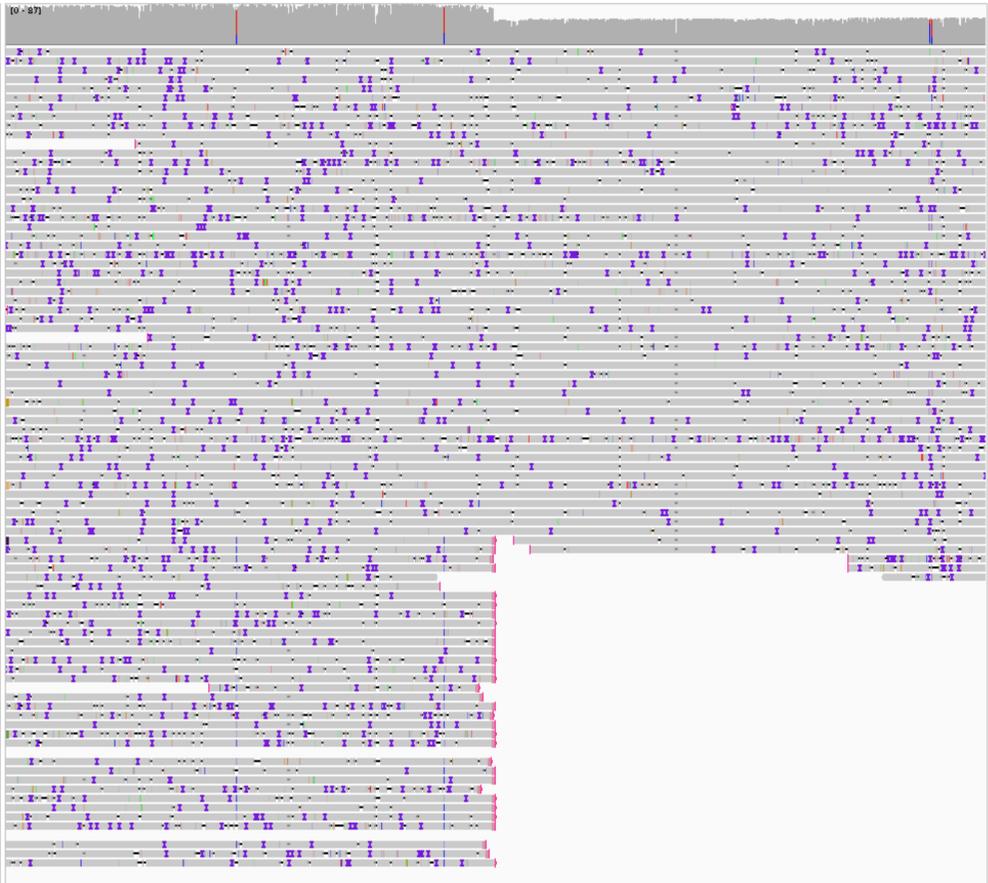
p13.3	p13.2	p13.13	p13.11	p12	p11	q12	q13.11	q13.13	q13.31	q13.33	q13.42
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← 1,002 bp →

17,420,600 bp      17,420,800 bp      17,421,000 bp      17,421,200 bp      17,421,400 bp

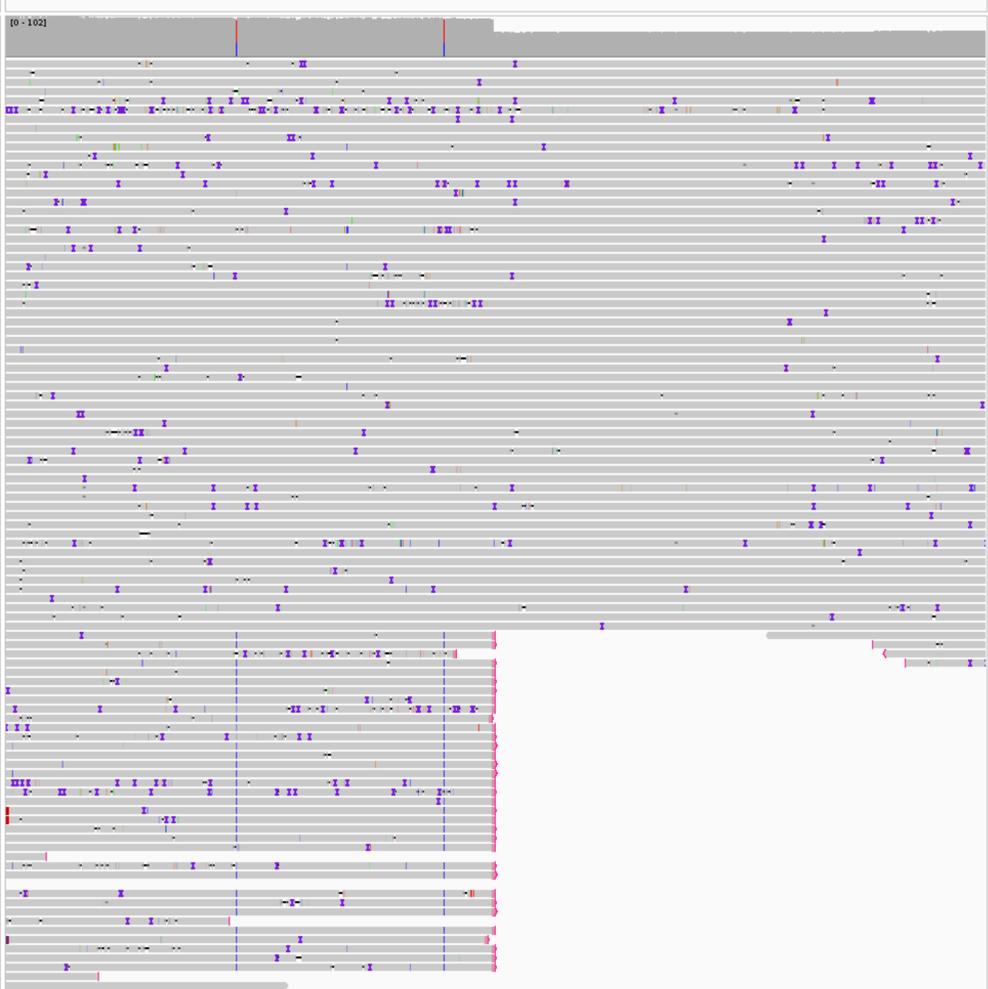
colo829\_gsc\_t2t.bam Coverage

colo829\_gsc\_t2t.bam



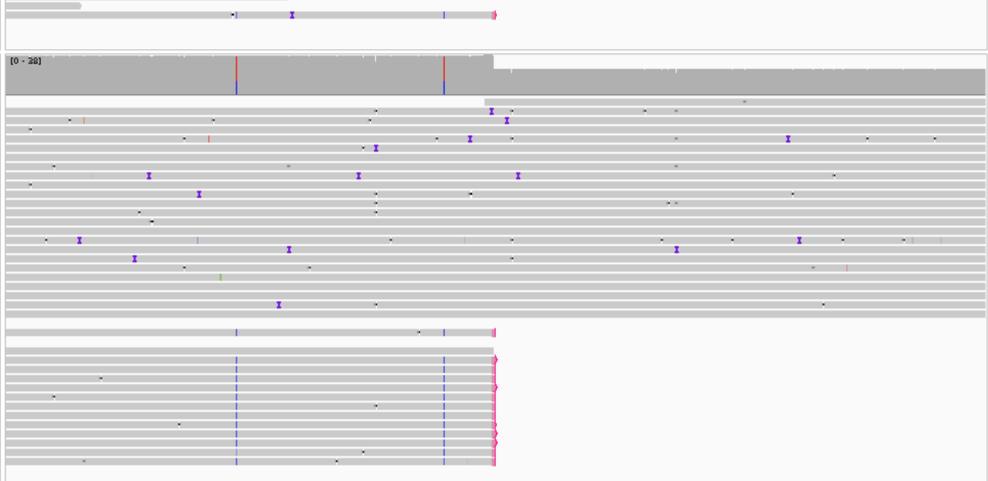
colo829\_ort\_t2t.bam Coverage

colo829\_ort\_t2t.bam



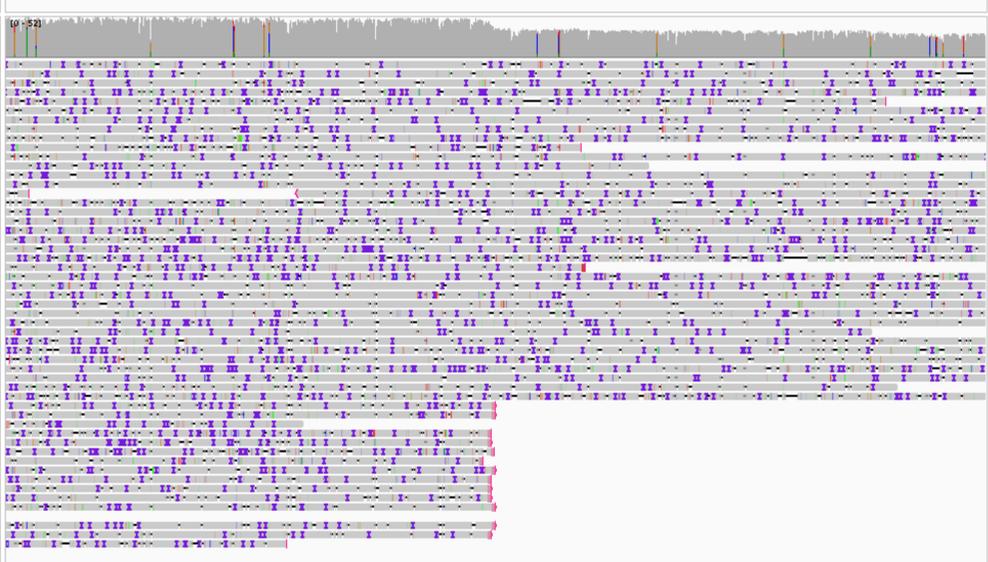
colo829\_pbr\_t2t.bam Coverage

colo829\_pbr\_t2t.bam



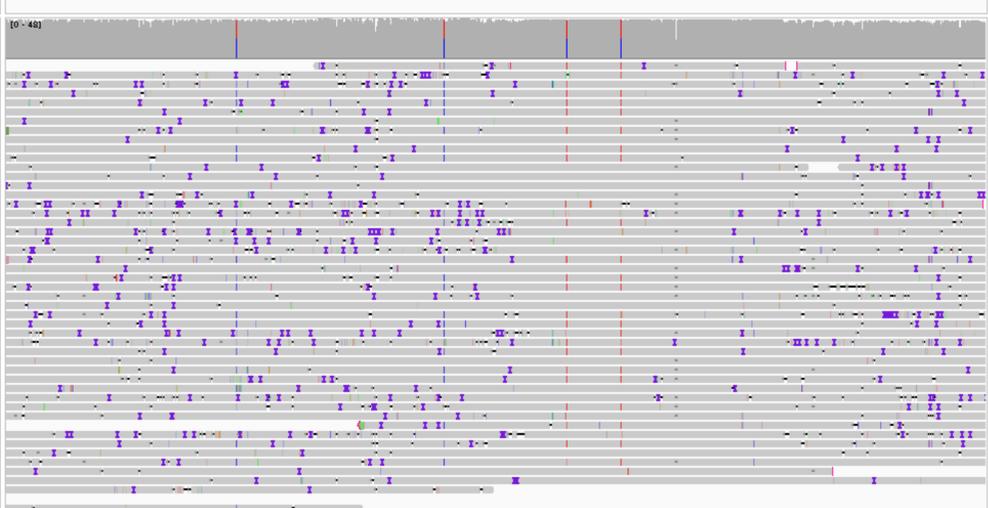
colo829\_val\_t2t.bam Coverage

colo829\_val\_t2t.bam



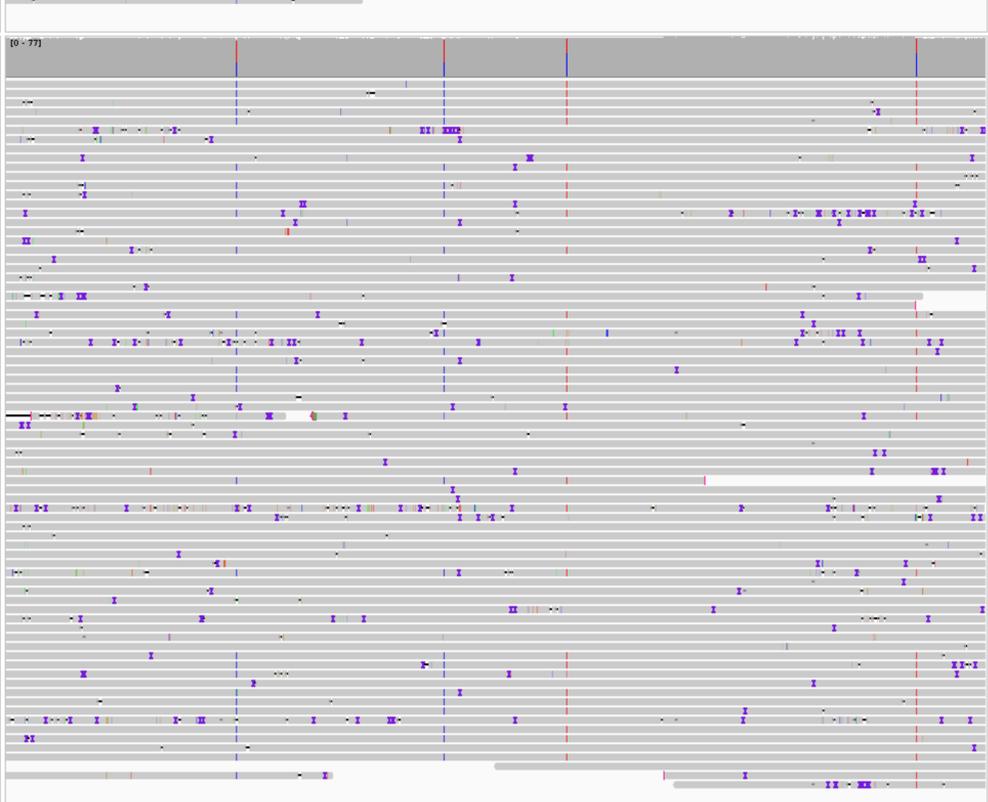
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colo829bl\_gsc\_t2t.bam



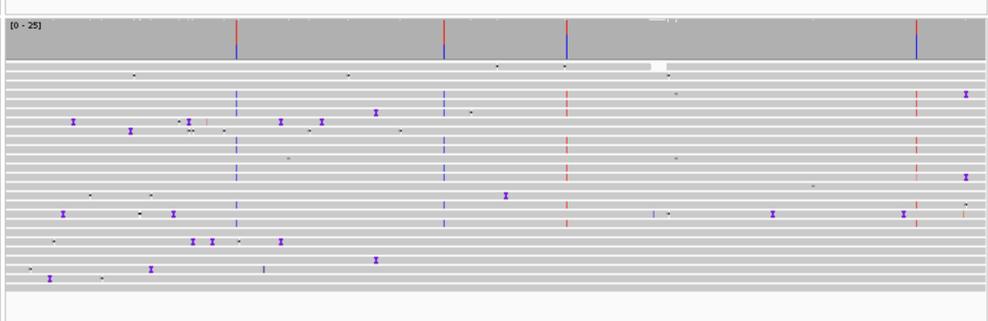
colo829bl\_ort\_t2t.bam Coverage

colo829bl\_ort\_t2t.bam



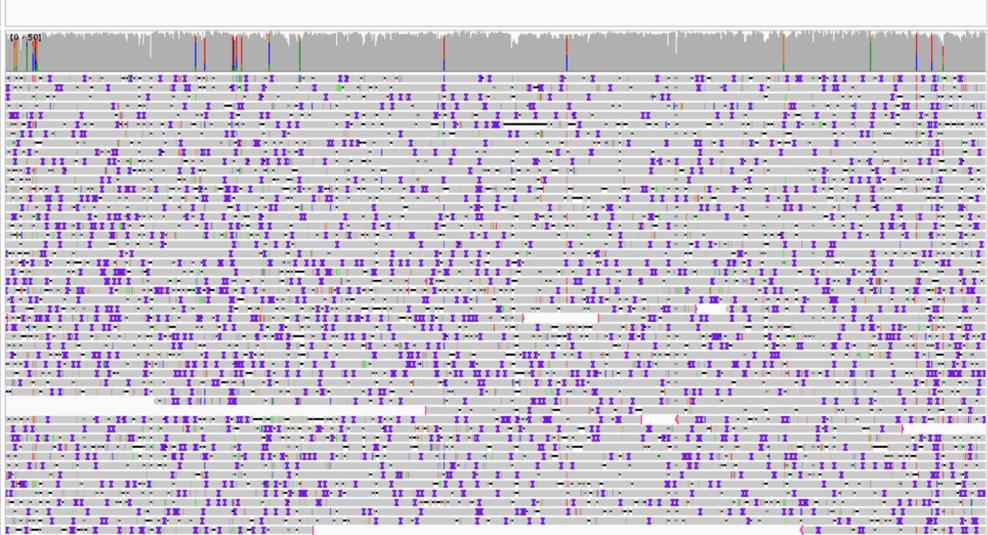
colo829bl\_pbr\_t2t.bam Coverage

colo829bl\_pbr\_t2t.bam



colo829bl\_val\_t2t.bam Coverage

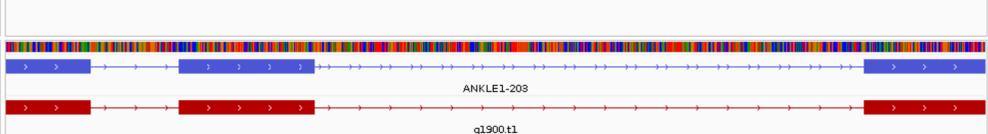
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Sequence →

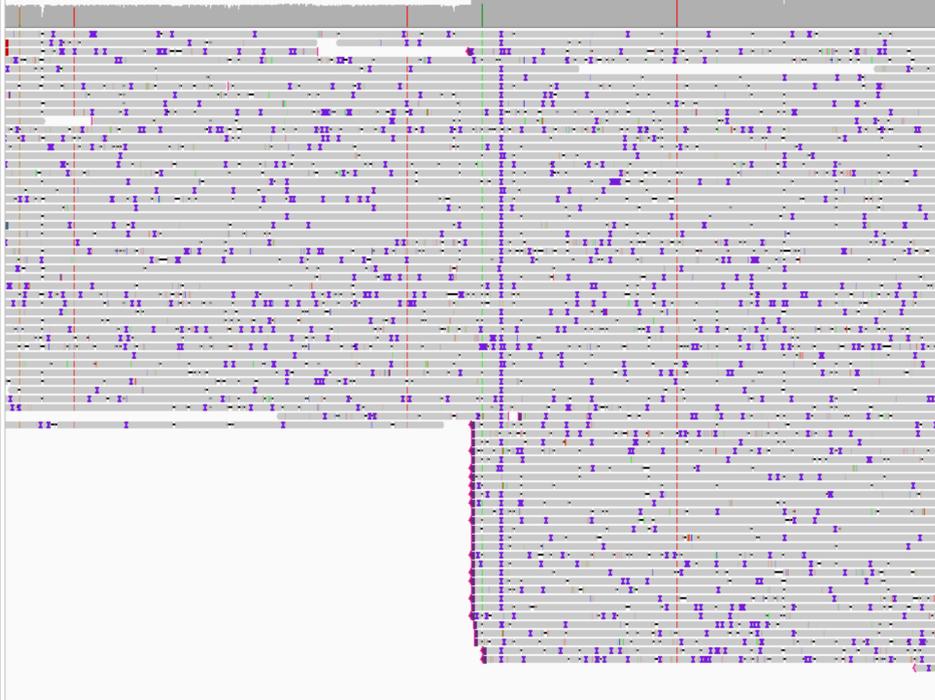
CAT/Liftoff Genes

Augustus





colo829\_gsc\_t2t.bam Coverage  
colo829\_gsc\_t2t.bam



colo829\_ont\_t2t.bam Coverage  
colo829\_ont\_t2t.bam



colo829\_pbr\_t2t.bam Coverage  
colo829\_pbr\_t2t.bam



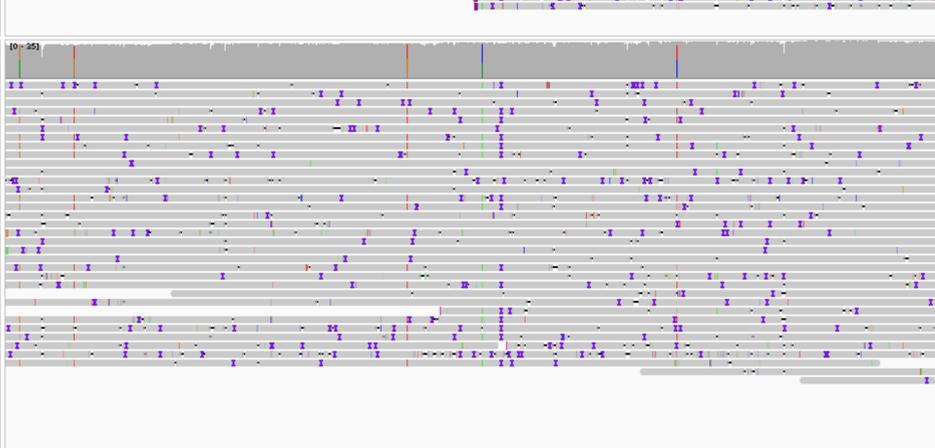
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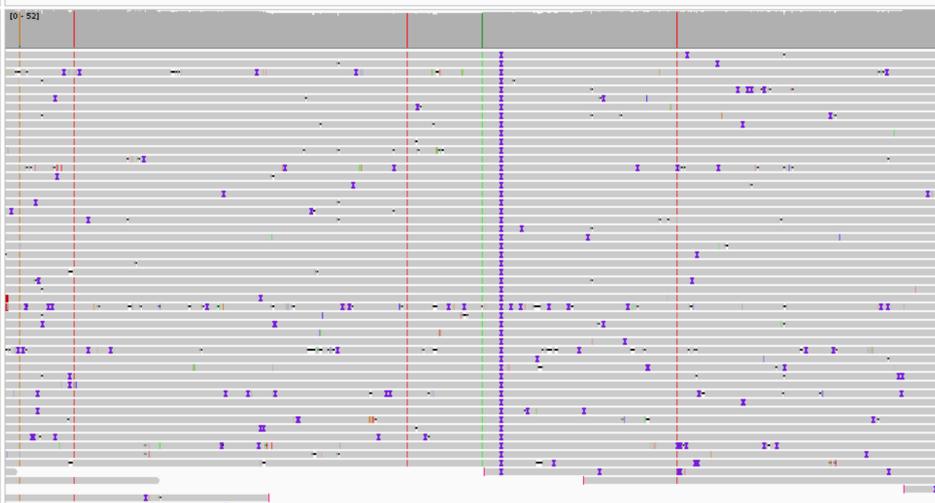
colo829b\_gsc\_t2t.bam Coverage  
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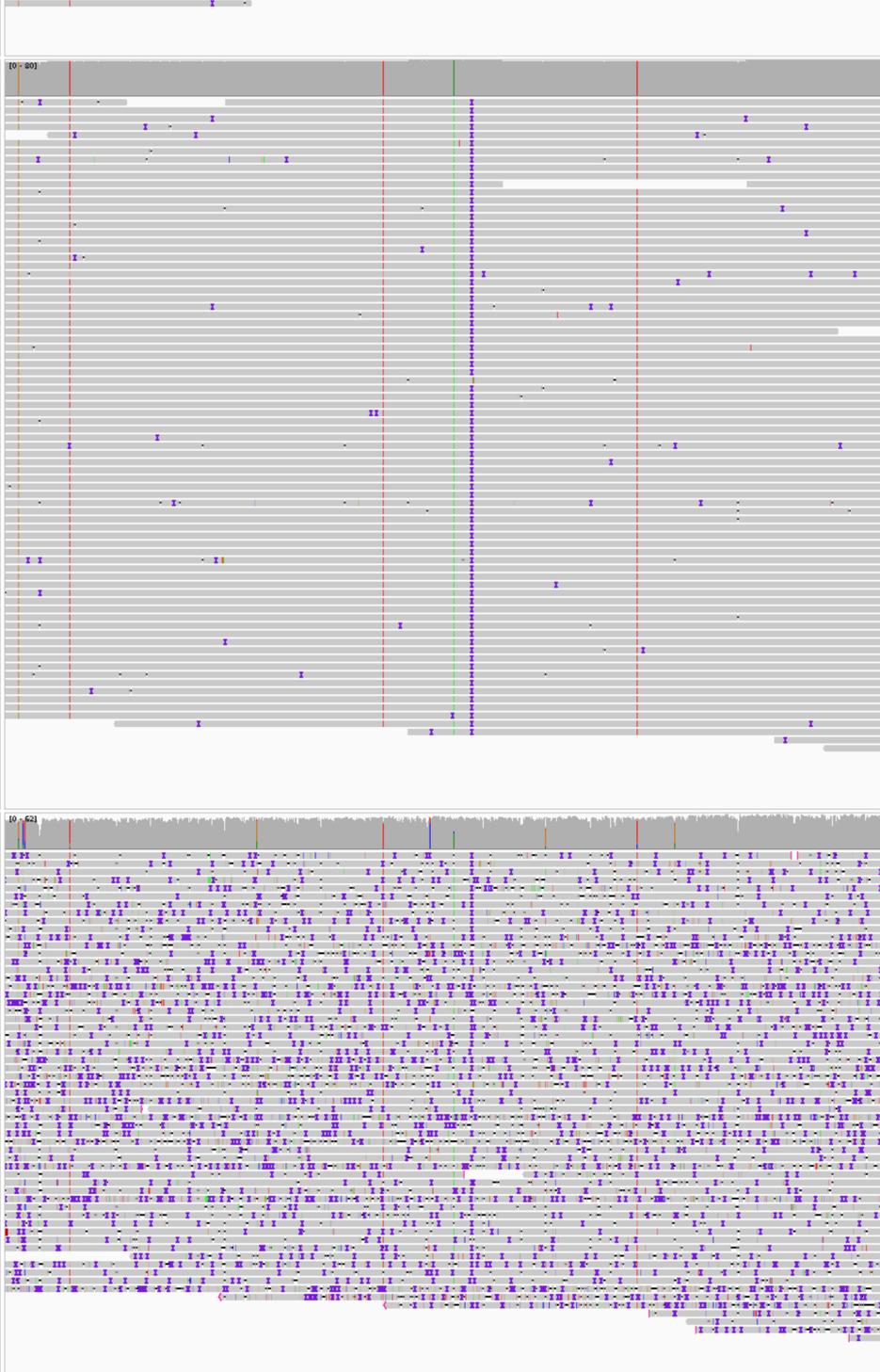
colo829b\_ont\_t2t.bam Coverage  
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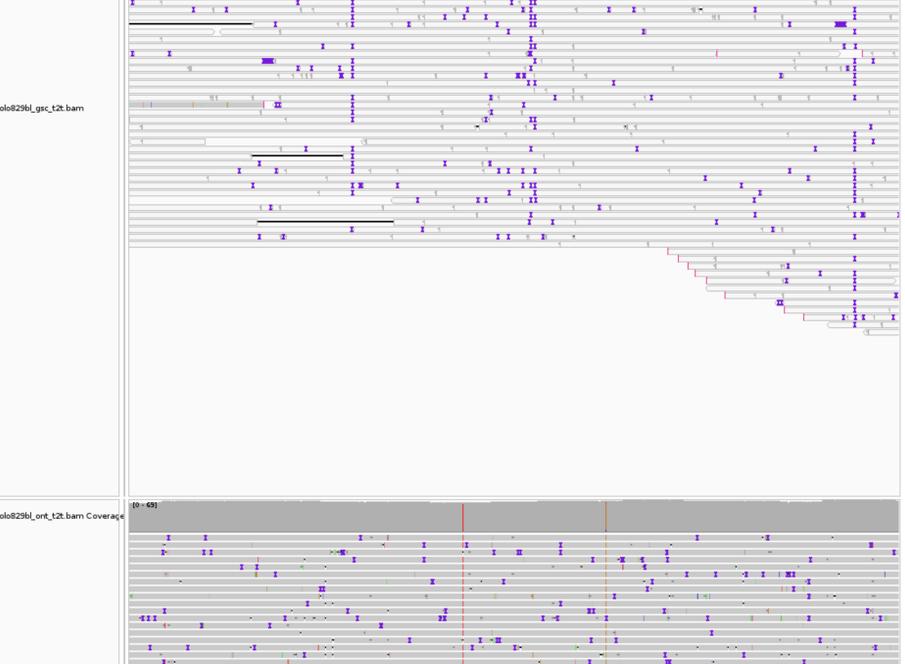
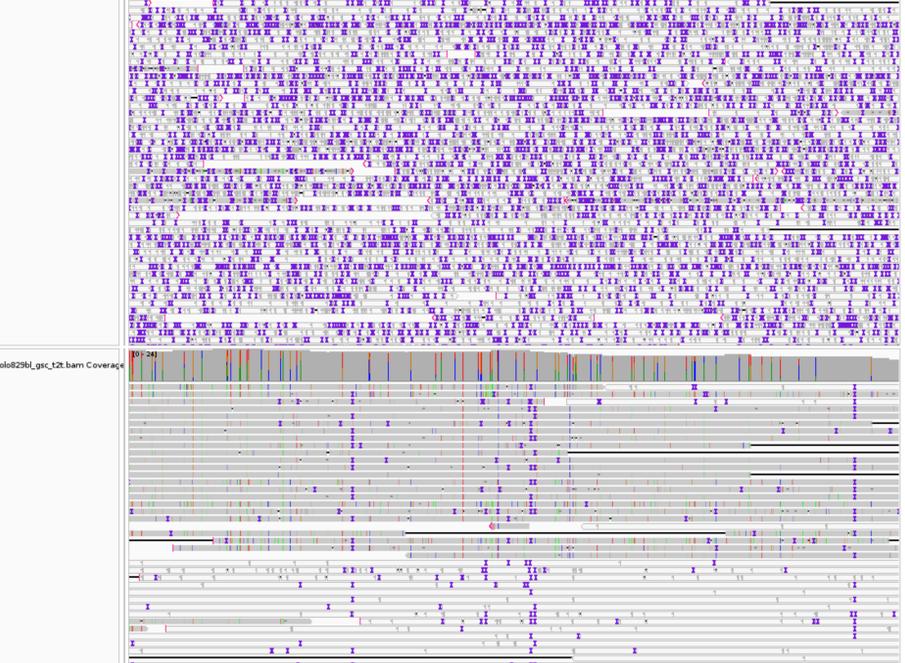
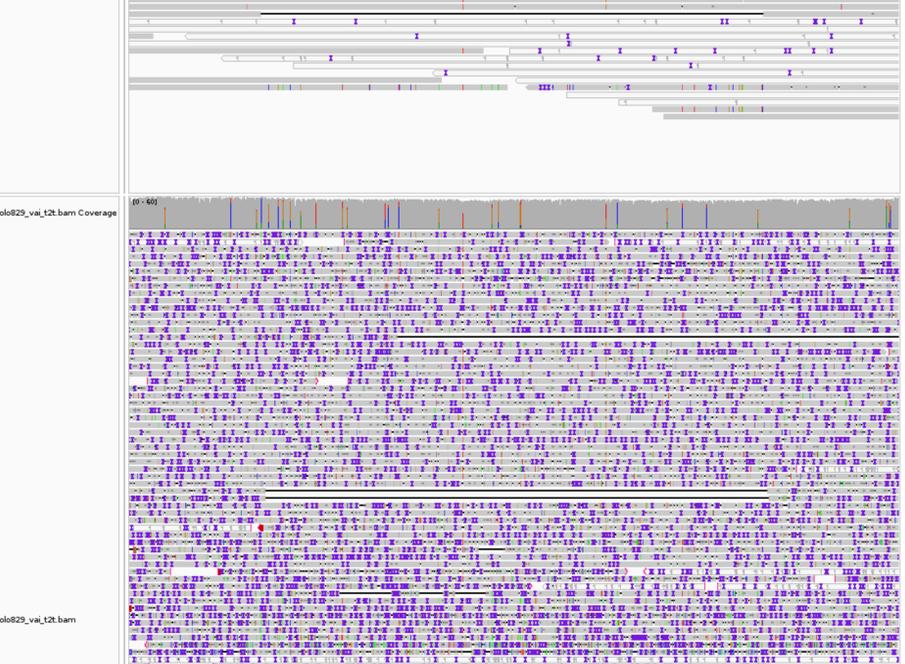
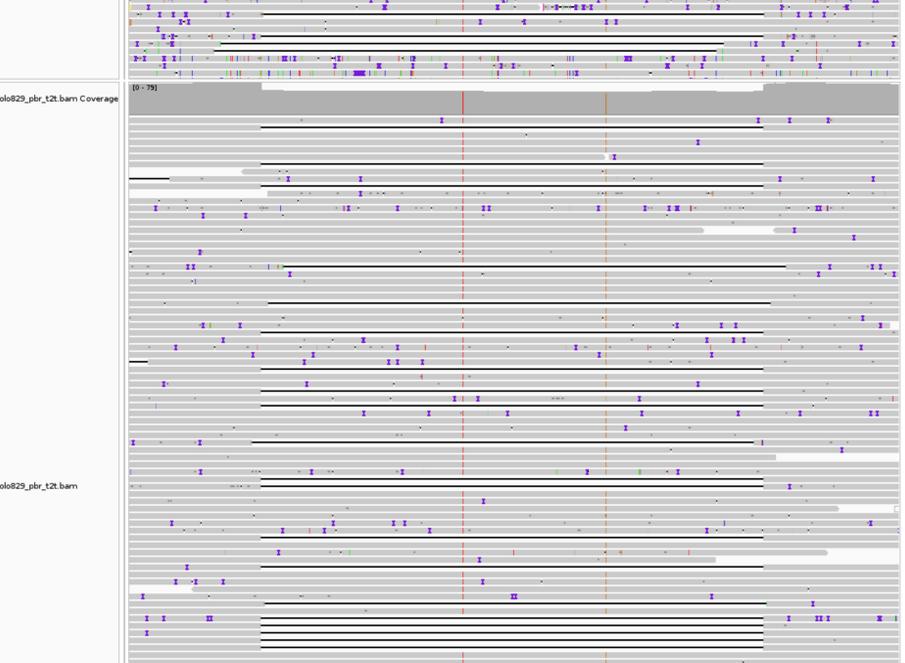
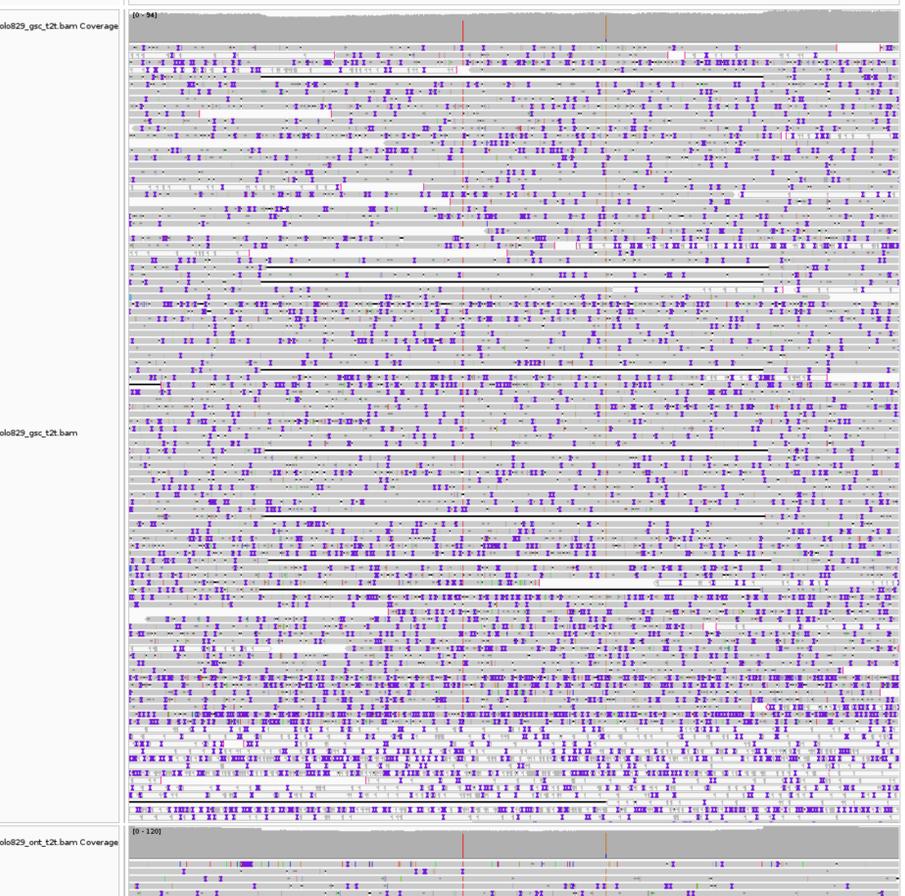


colo829b\_pbr\_t2t.bam Coverage  
colo829b\_pbr\_t2t.bam



colo829b\_vai\_t2t.bam Coverage  
colo829b\_vai\_t2t.bam  
Sequence  
CAT/Liftoff Genes  
Augustus





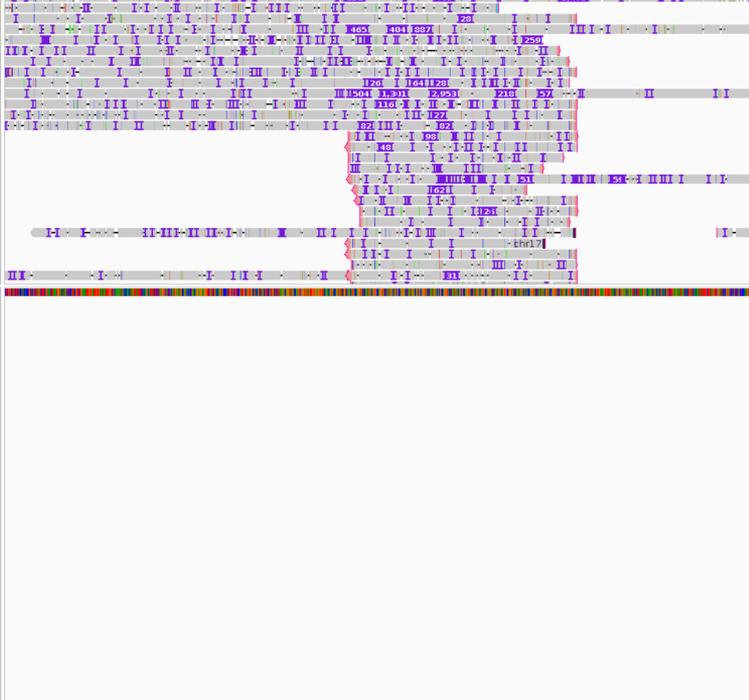
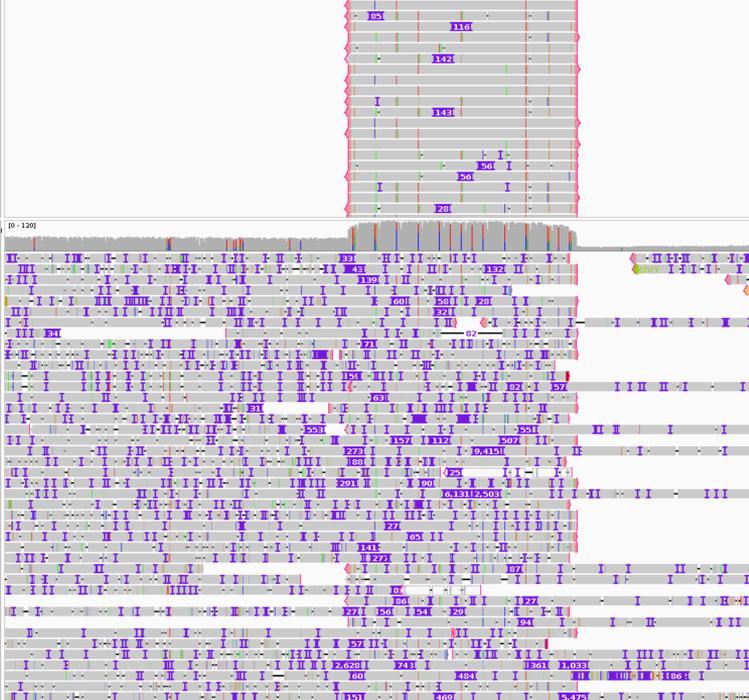
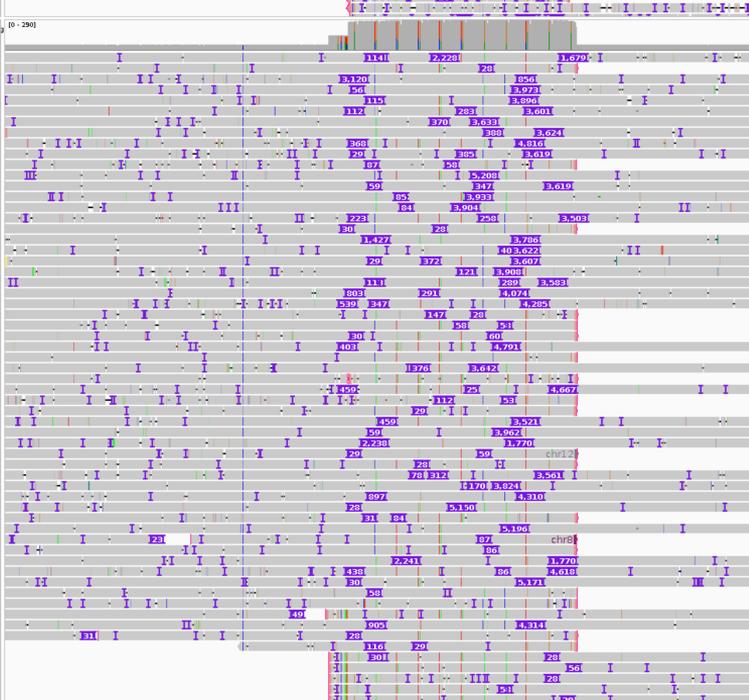
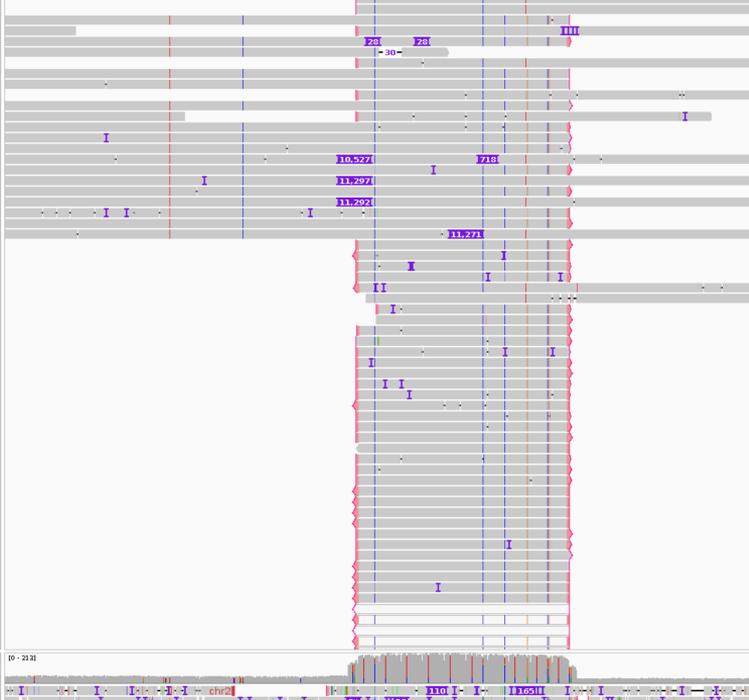
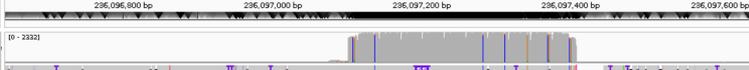
CATL/Rhoff Genes  
Augustus





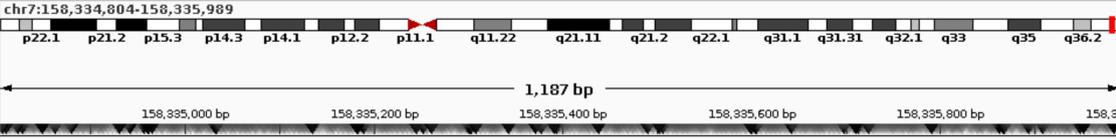
Supplementary Figure 5: IGV screenshot of the Structural Variants (SVs) labeled as False Positives (FP) according to the Valle-Inclan benchmark

- A) A 11,260 bp INS located in chr1:236097142 wrongly labeled as FP according to the benchmark of Valle-Inclan
- B) A 54 bp INS located in chr7:75595250 wrongly labeled as FP according to the benchmark of Valle-Inclan
- C) A 185 bp DEL located in chr7:158335304 wrongly labeled as FP according to the benchmark of Valle-Inclan
- D) A 2,397 bp INS located in chr14:34545553 wrongly labeled as FP according to the benchmark of Valle-Inclan
- E) A 199 bp INS located in chr14:105230166 wrongly labeled as FP according to the benchmark of Valle-Inclan
- F) A 4,567 bp DEL located in chr15:23467473 wrongly labeled as FP according to the benchmark of Valle-Inclan
- G) A BND located in chr19:17286003 (second breakpoint in chr10:7017548) wrongly labeled as FP according to the benchmark of Valle-Inclan, breakpoint 1
- H) A BND located in chr19:17286003 (second breakpoint in chr10:7017548) wrongly labeled as FP according to the benchmark of Valle-Inclan, breakpoint 2

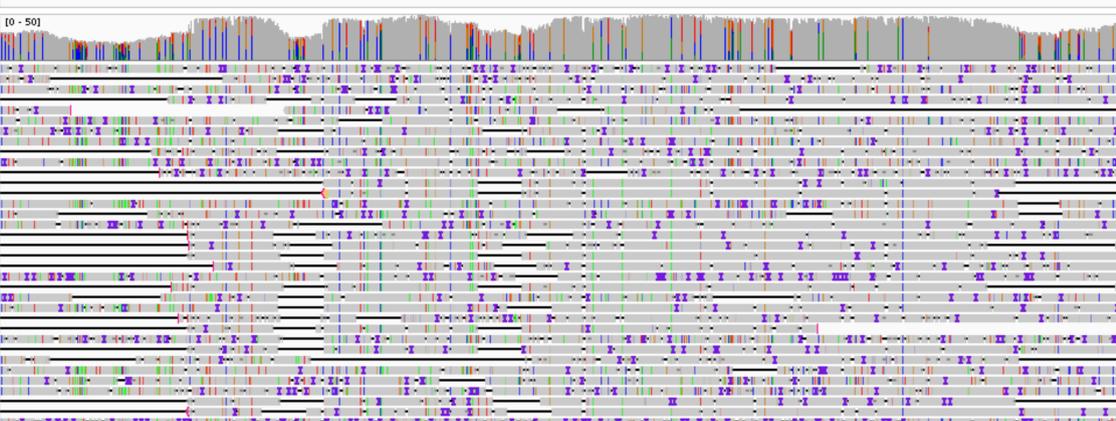


Refseq Genes

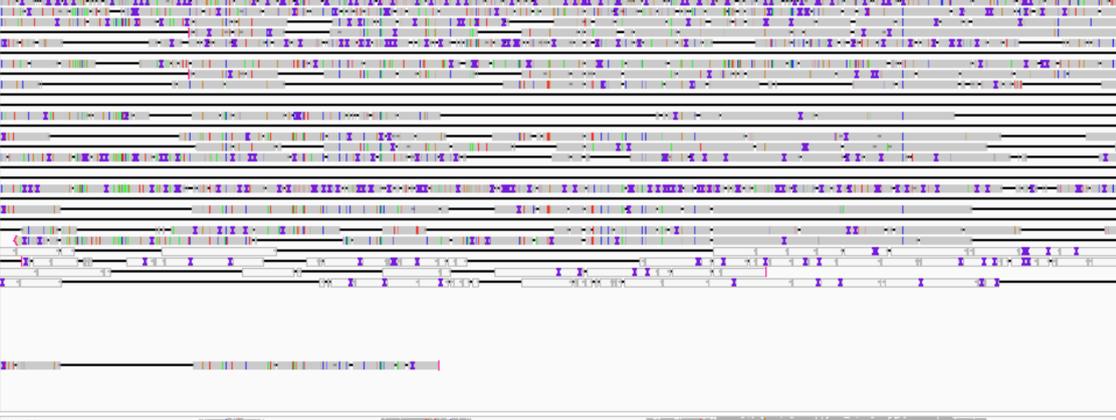




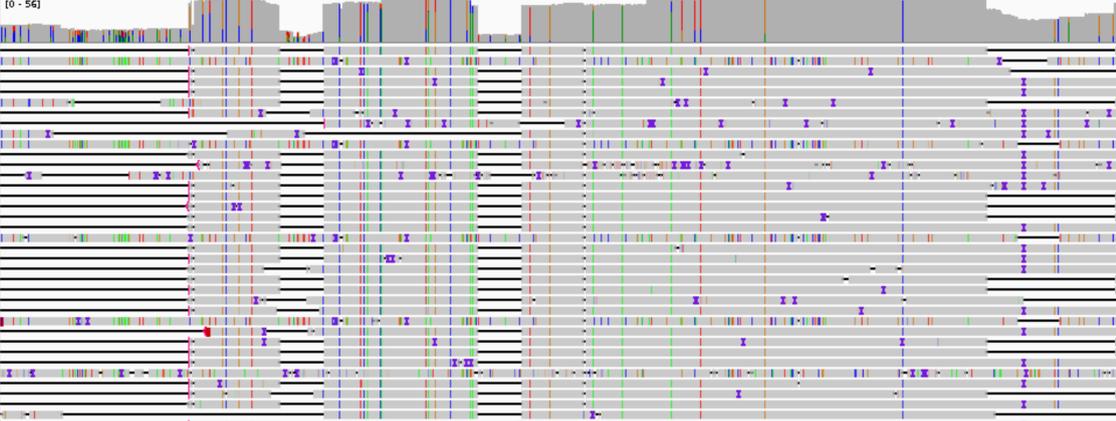
colo829\_gsc\_lifted.bam Coverage



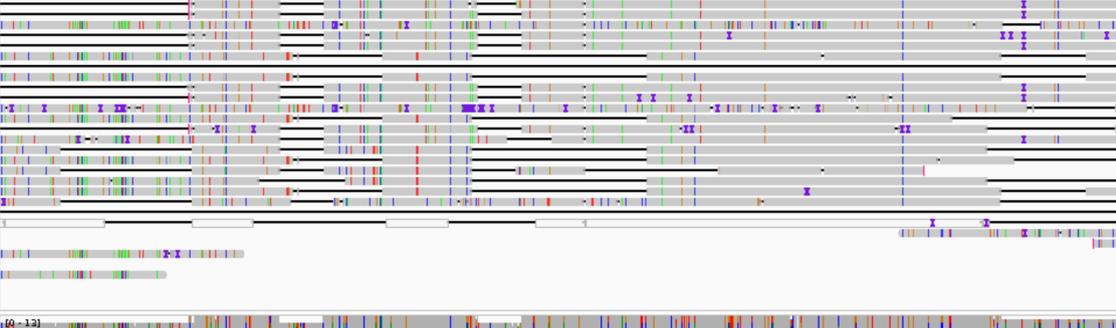
colo829\_gsc\_lifted.bam



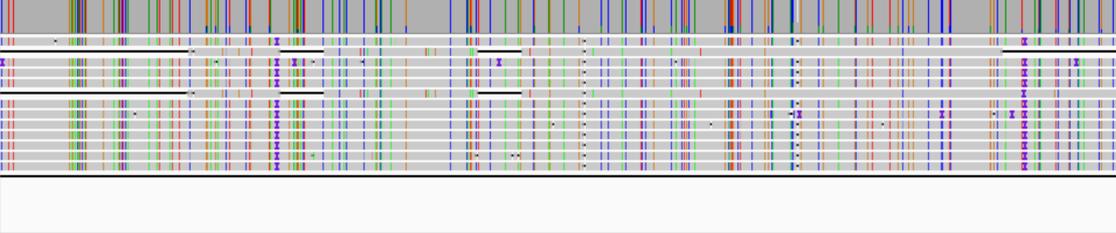
colo829\_ont\_lifted.bam Coverage



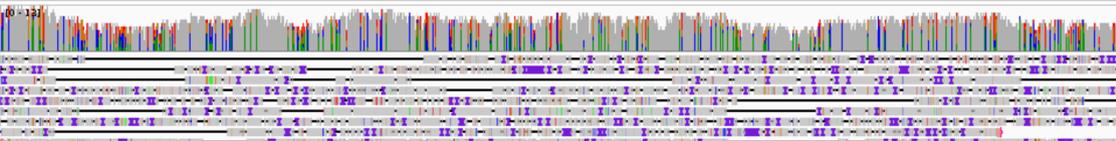
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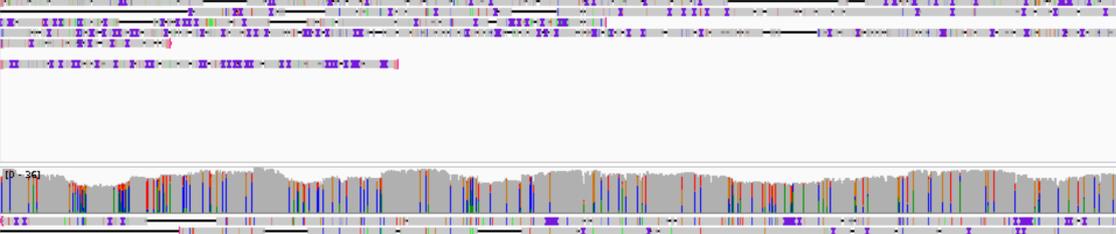
colo829\_pbr\_lifted.bam Coverage



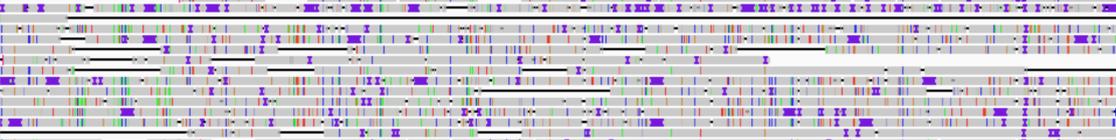
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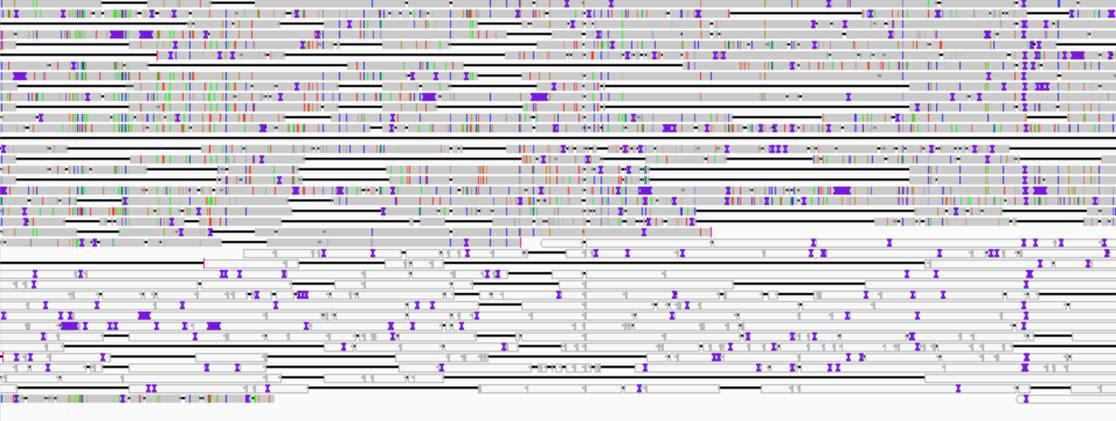
colo829\_vai\_lifted.bam Coverage



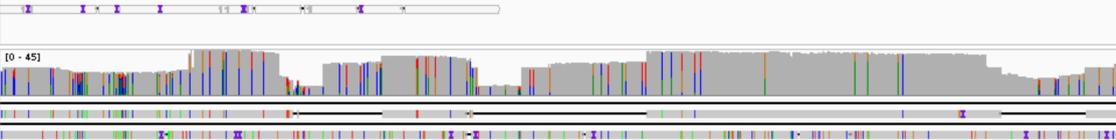
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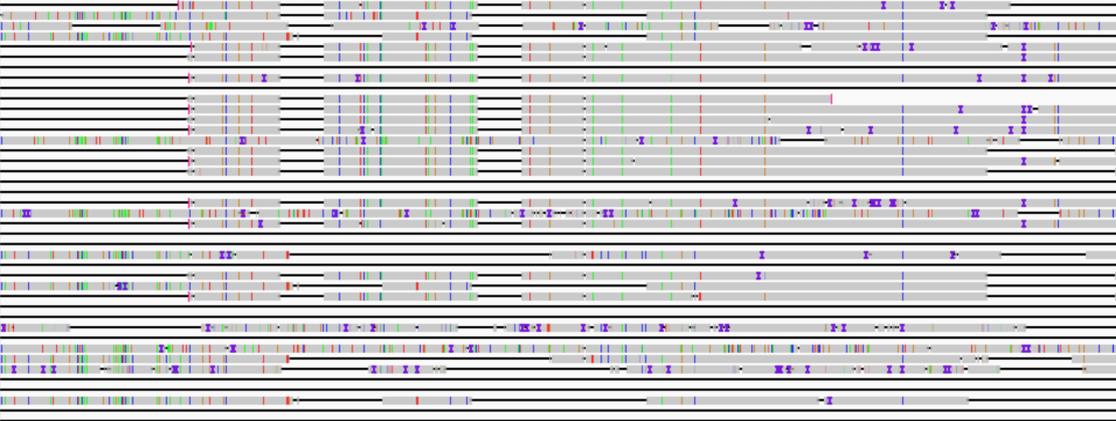
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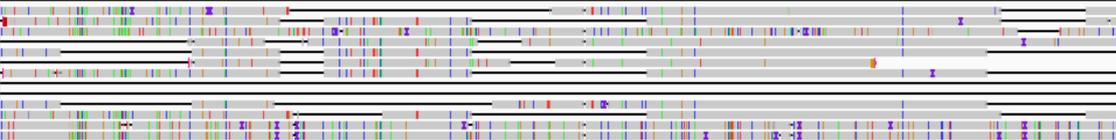
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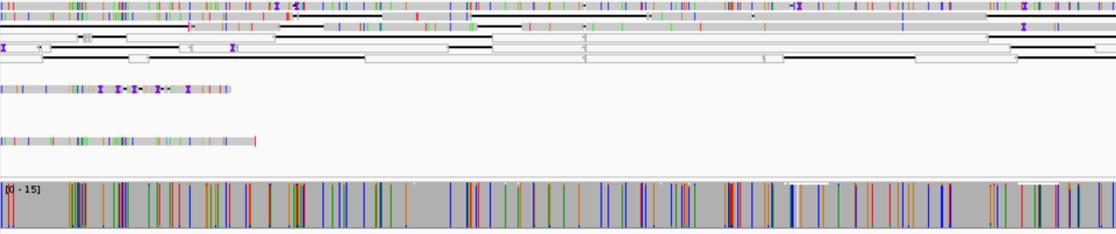
colo829bl\_ont\_lifted.bam Coverage



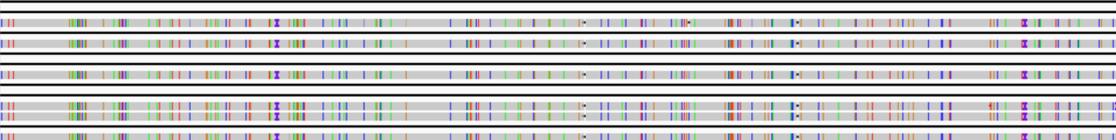
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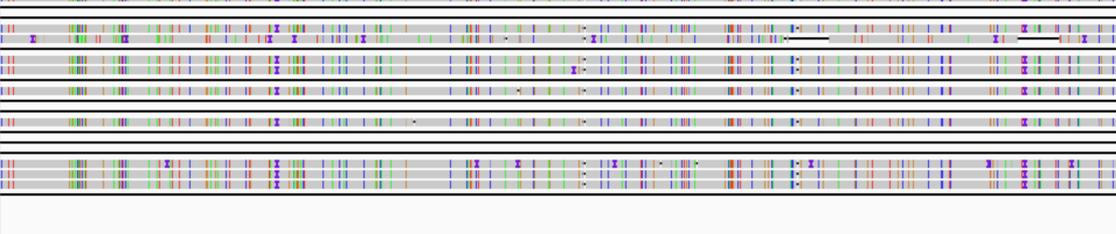
colo829bl\_pbr\_lifted.bam Coverage



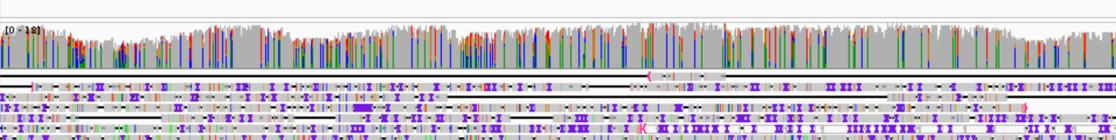
colo829bl\_pbr\_lifted.bam



colo829bl\_vai\_lifted.bam Coverage



colo829bl\_vai\_lifted.bam



Sequence

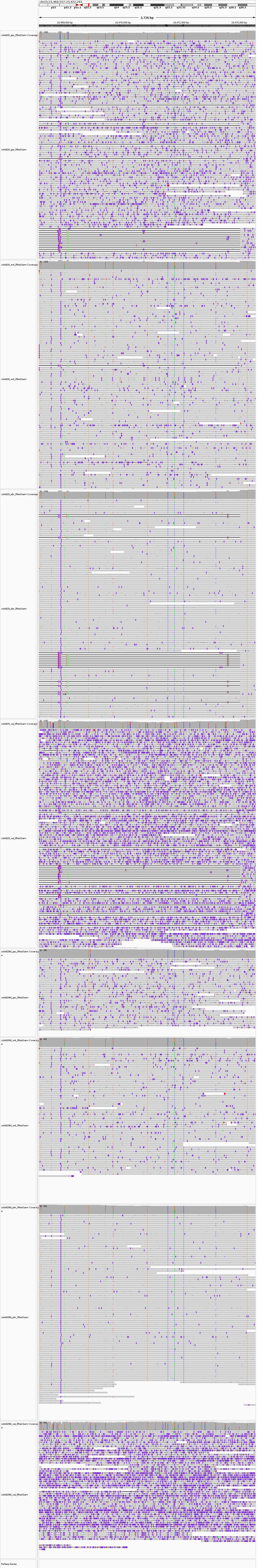


RefSeq Genes

PTPRN2







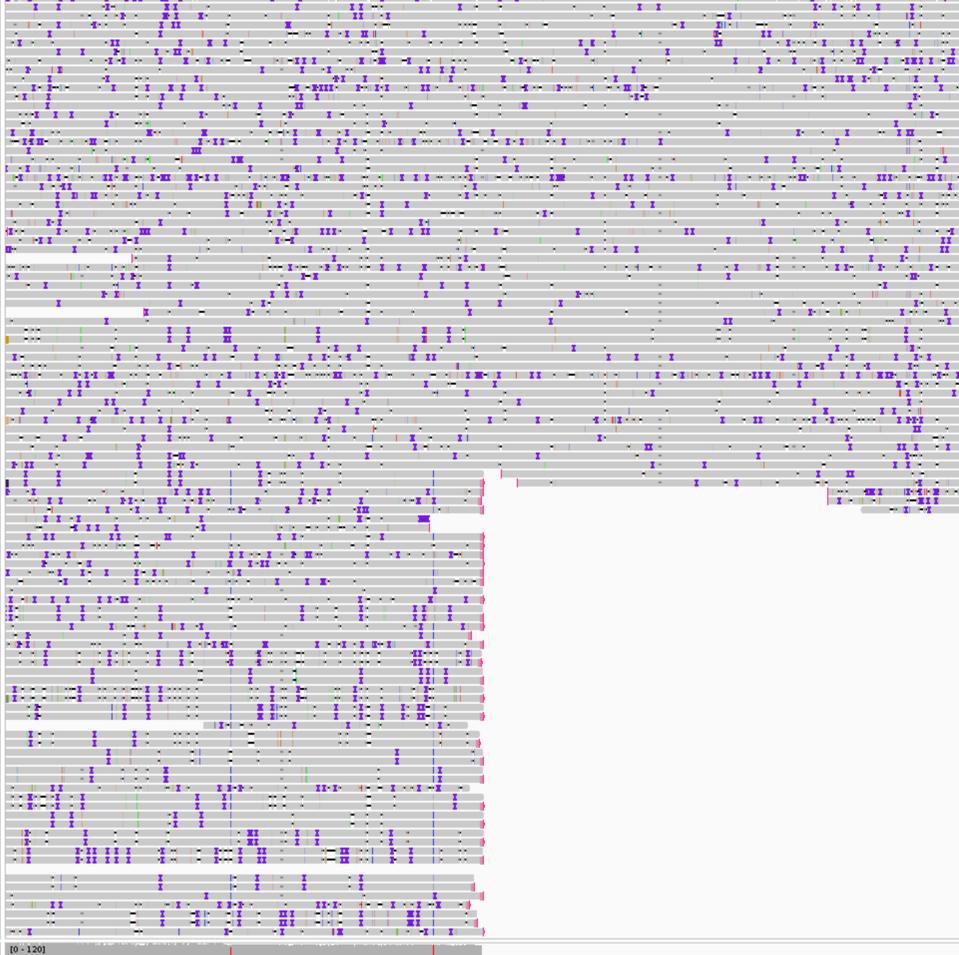
chr19:17,285,503-17,286,503

p13.3 p13.2 p13.13 p13.11 p12 p11 q11 q12 q13.11 q13.13 q13.31 q13.33 q13.42

17,285,600 bp 17,285,800 bp 17,286,000 bp 17,286,200 bp 17,286,400 bp

← 1,002 bp →

colo829\_gsc\_lifted.bam Coverage



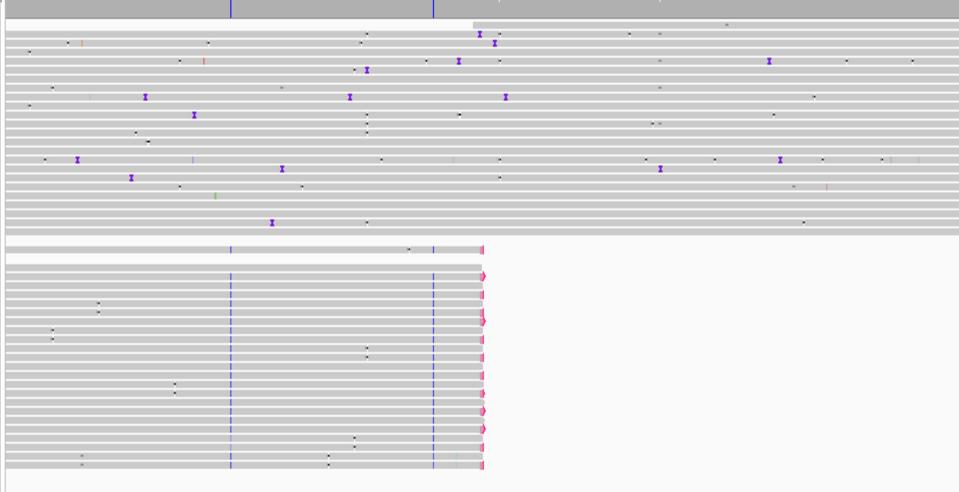
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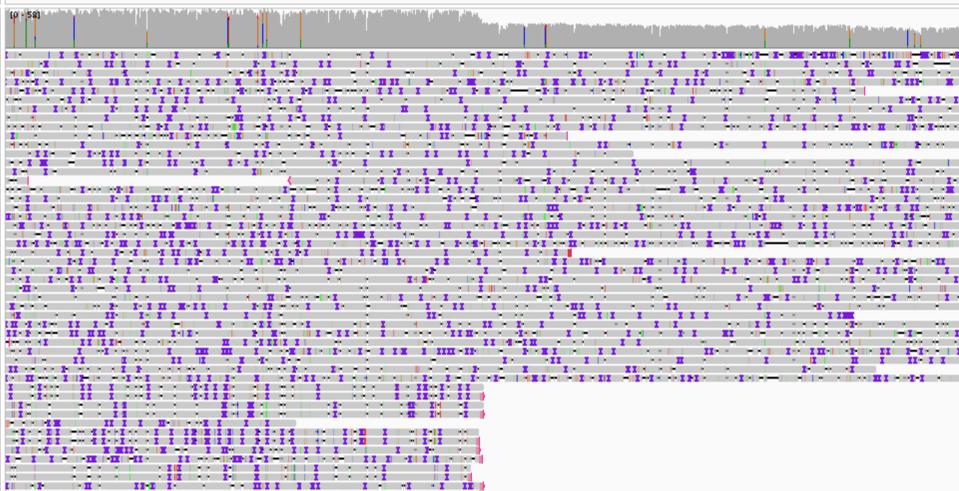
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colo829\_ont\_lifted.bam



colo829\_pbr\_lifted.bam Coverage



colo829\_pbr\_lifted.bam



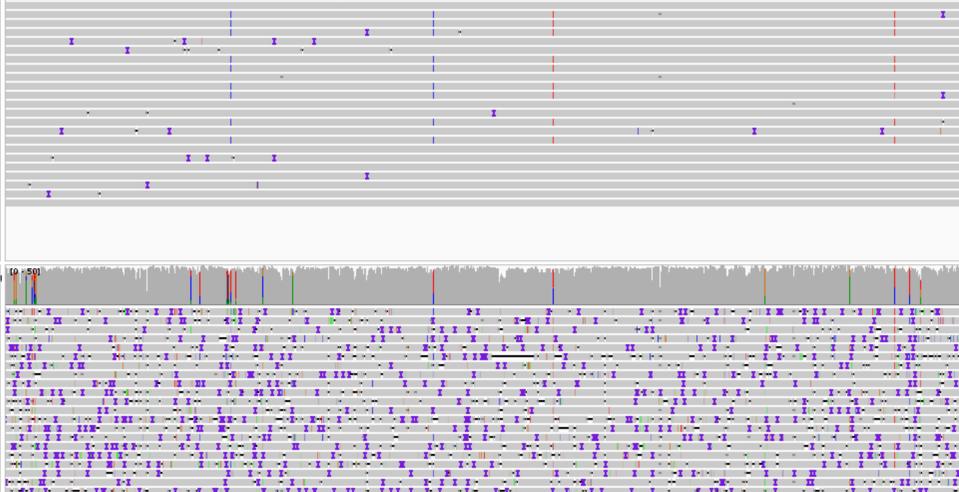
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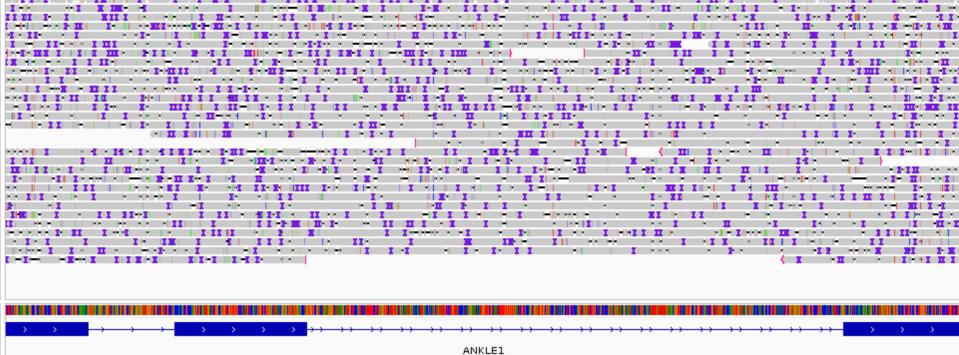
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colo829b1\_gsc\_lifted.bam Coverage



colo829b1\_gsc\_lifted.bam



colo829b1\_ont\_lifted.bam Coverage



colo829b1\_ont\_lifted.bam



colo829b1\_pbr\_lifted.bam Coverage



colo829b1\_pbr\_lifted.bam



colo829b1\_vai\_lifted.bam Coverage



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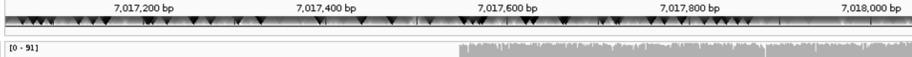
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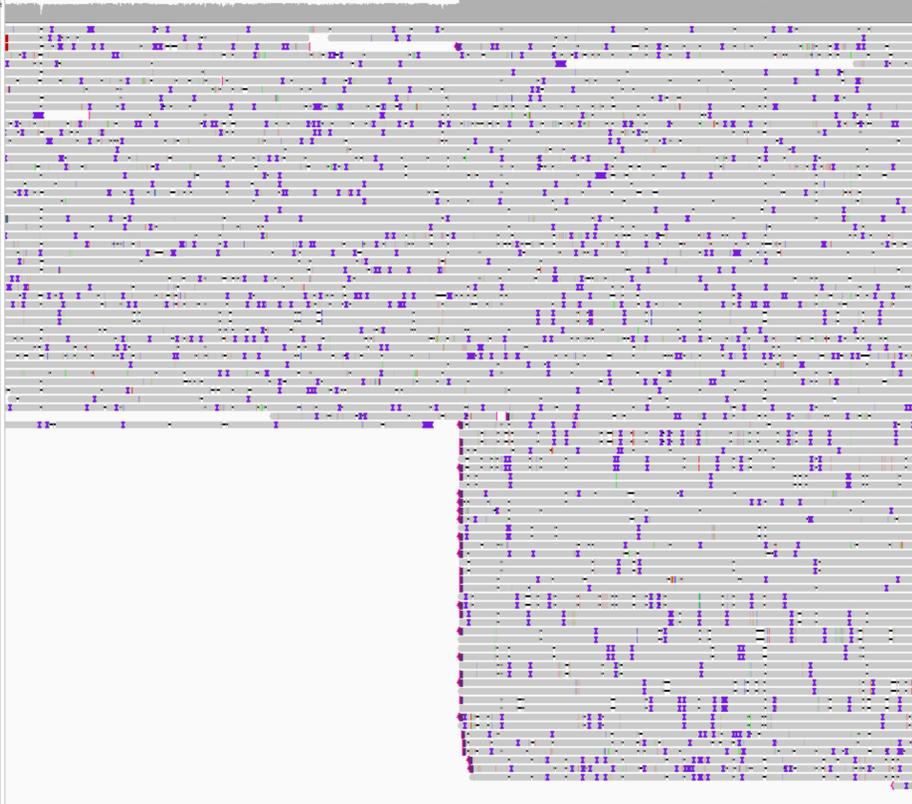
RefSeq Genes



ANKLE1



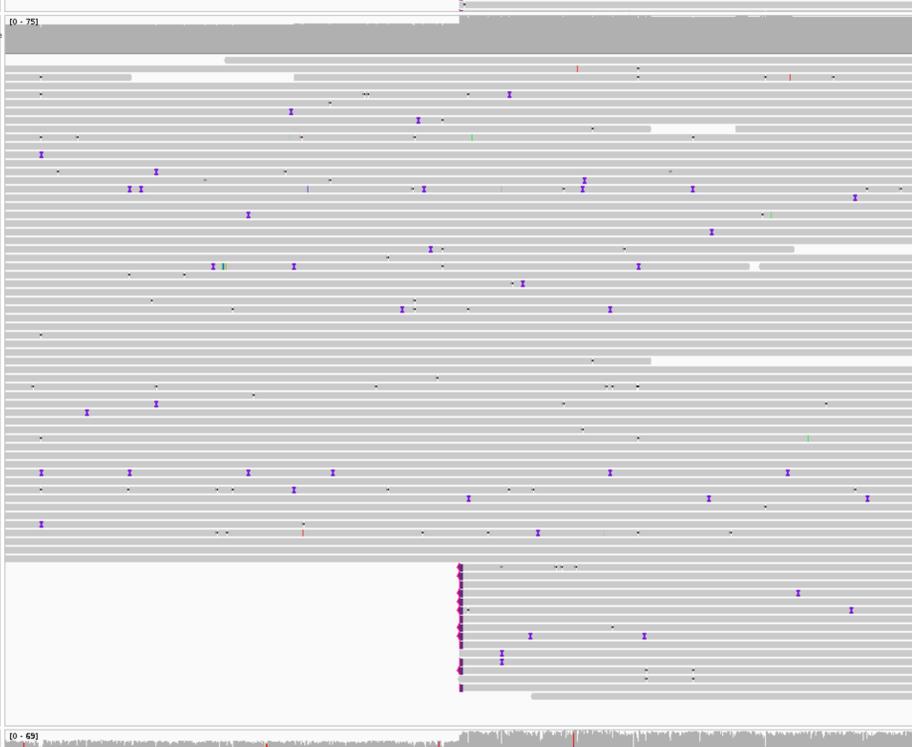
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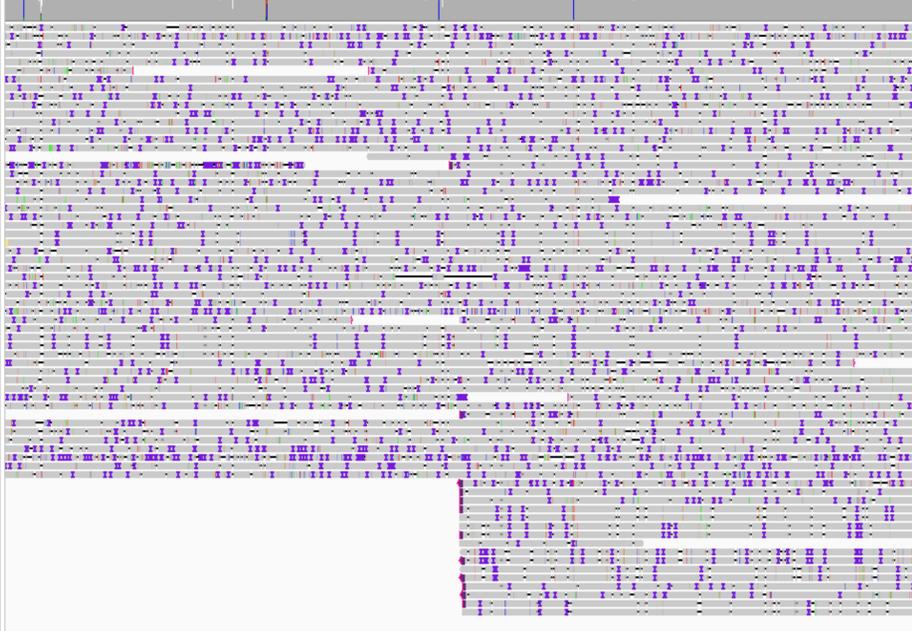
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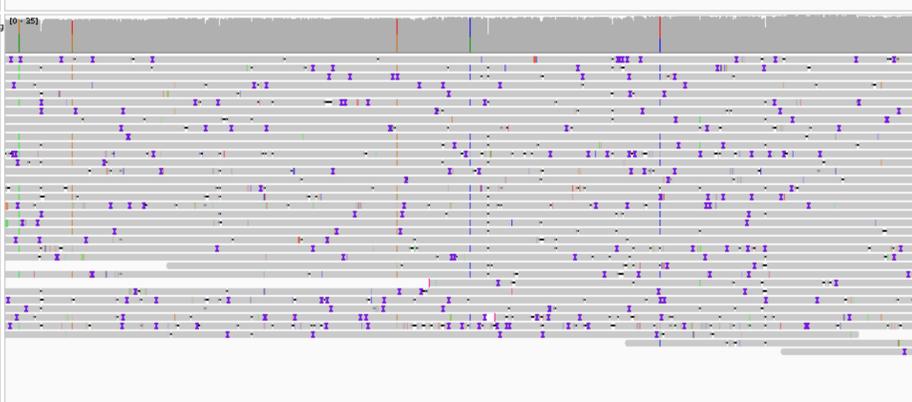
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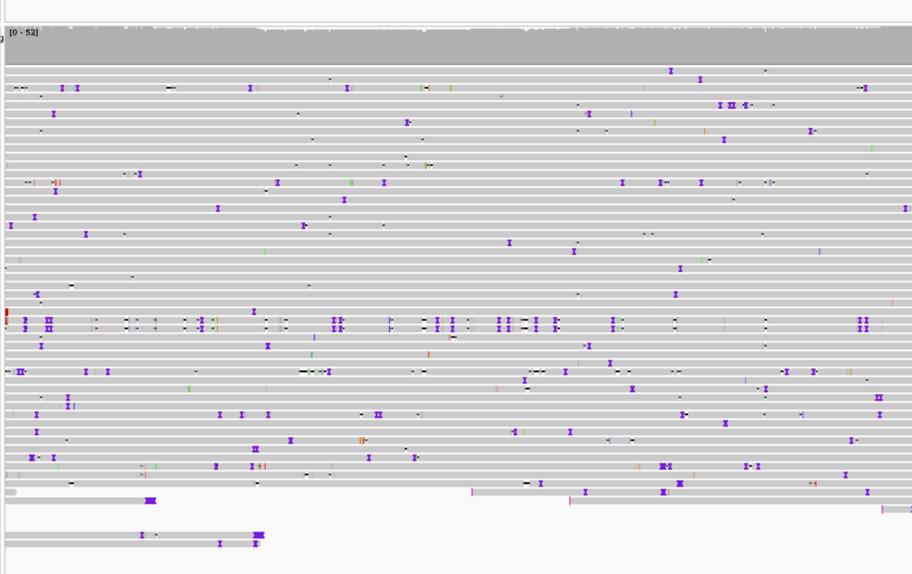
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colo829\_vai\_lifted bam



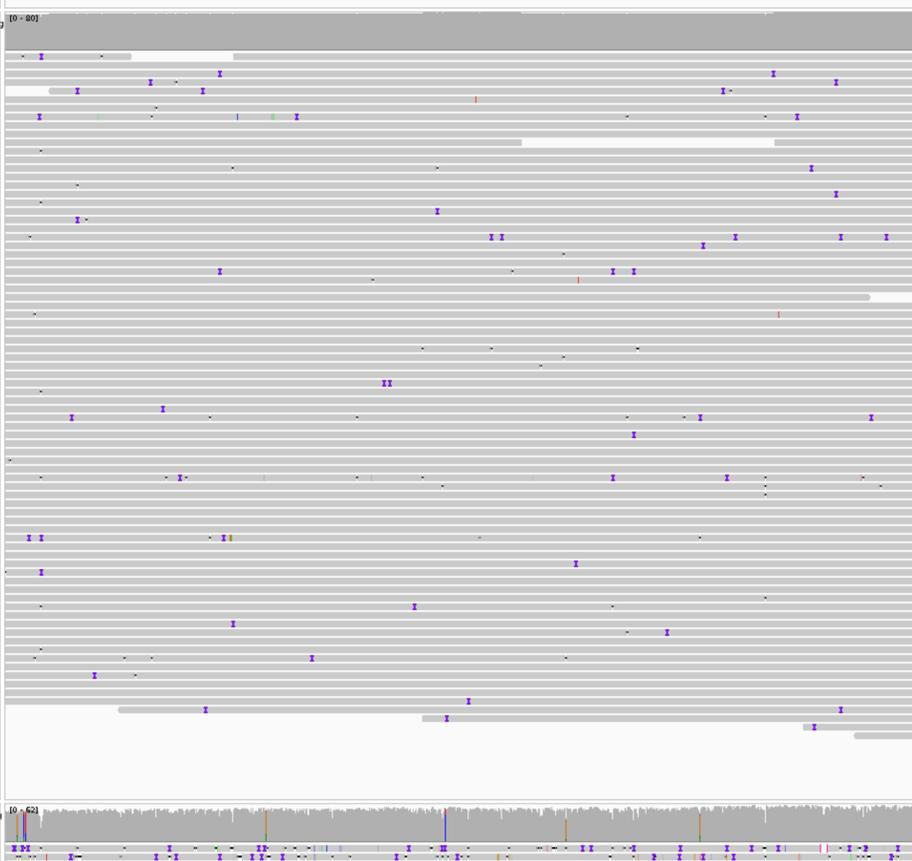
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e



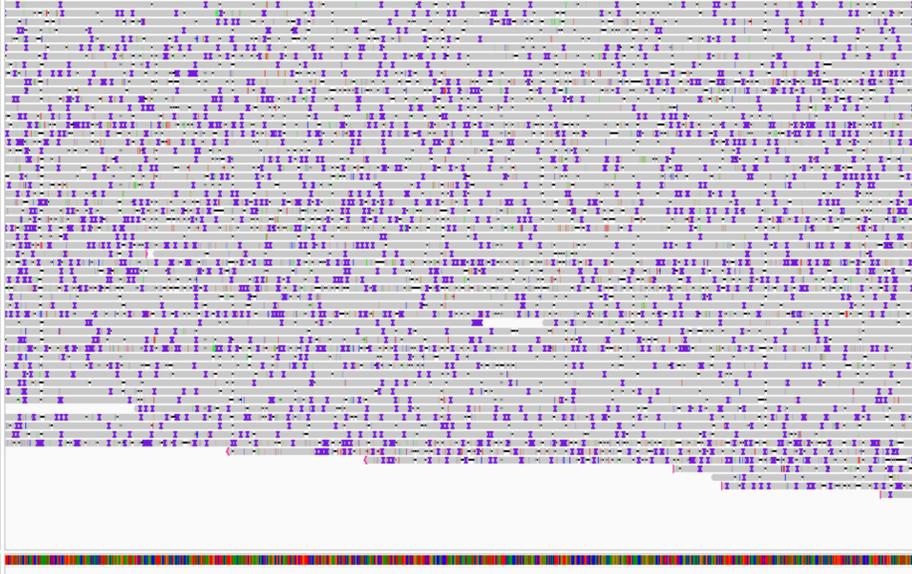
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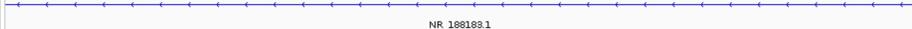
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colo829b\_vai\_lifted bam Coverage  
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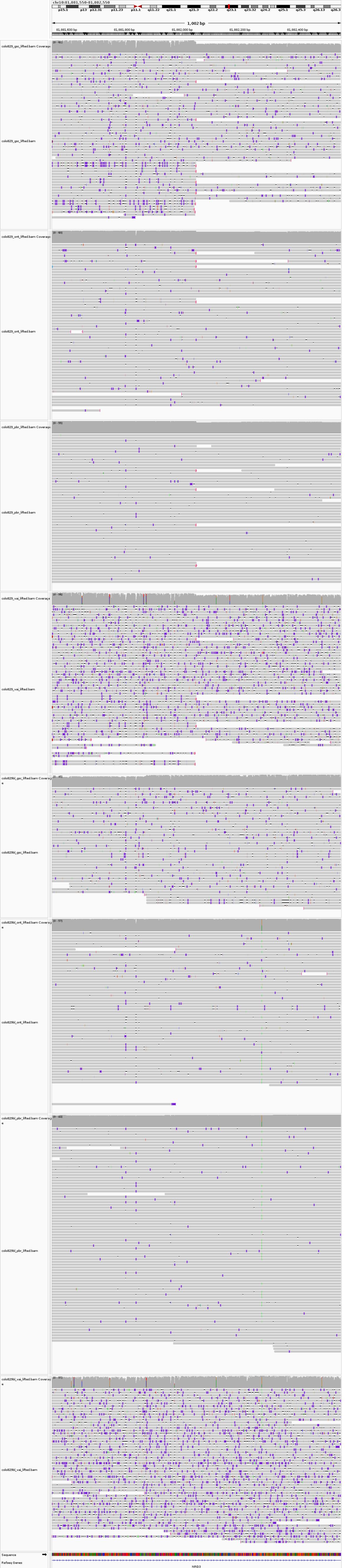


Sequence  
Refseq Genes

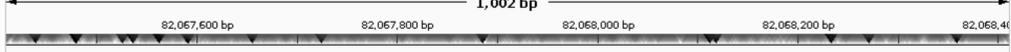


Supplementary Figure 6: IGV screenshot of the Structural Variants (SVs) labeled as False Negative (FN) according to the Valle-Inclan benchmark

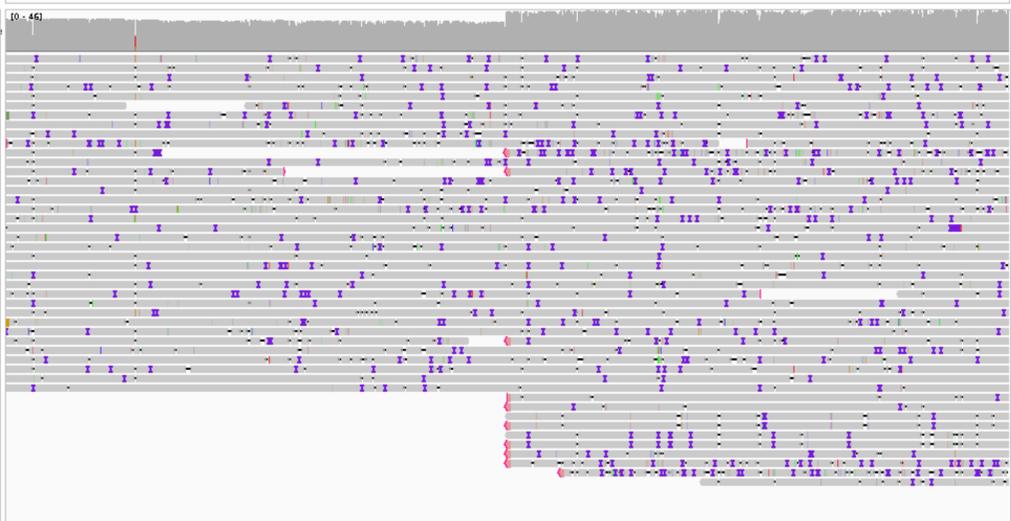
- A) Evidence found, missed: a 185,860 bp DEL located in chr10:81882050 (Valle-Inclan ID truthset\_44\_1), breakpoint 1
- B) Evidence found, missed: a 185,860 bp DEL located in chr10:81882050 (Valle-Inclan ID truthset\_44\_1), breakpoint 2
- C) No evidence found, filtered: a BND located in chr1:86871328 (Valle-Inclan ID truthset\_1\_1)
- D) No evidence found, filtered: a BND located in chr3:25359111 (Valle-Inclan ID truthset\_7\_1)
- E) No evidence found, filtered: a BND located in chr3:25359568 (Valle-Inclan ID truthset\_8\_1)
- F) Little evidence found, filtered: a BND located in chr3:26390427 (Valle-Inclan ID truthset\_9\_1)
- G) Little evidence found, filtered: a 71,459 bp DEL located in chr3:186724386 (Valle-Inclan ID truthset\_13\_1), breakpoint 1
- H) Little evidence found, filtered: a 71,459 bp DEL located in chr3:186724386 (Valle-Inclan ID truthset\_13\_1), breakpoint 2
- I) No evidence found, filtered: a 289 bp INV located in chr6:26193889 (Valle-Inclan ID truthset\_18\_1)
- J) No evidence found, filtered: a BND located in chr6:138452922 (Valle-Inclan ID truthset\_19\_1)
- K) No evidence found, filtered: a BND located in chr6:138453044 (Valle-Inclan ID truthset\_20\_1)
- L) Little evidence found, filtered: a 1,163 bp DEL located in chr8:111050191 (Valle-Inclan ID truthset\_32\_1)
- M) Some evidence found, filtered: a BND located in chr10:7017549 (Valle-Inclan ID truthset\_38\_1)
- N) No evidence found, filtered: a 2,732,610 bp DEL located in chr10:33097525 (Valle-Inclan ID truthset\_41\_1), breakpoint 1
- O) No evidence found, filtered: a 2,732,610 bp DEL located in chr10:33097525 (Valle-Inclan ID truthset\_41\_1), breakpoint 2
- P) Little evidence found, filtered: a 549 bp DEL located in chr10:53716587 (Valle-Inclan ID truthset\_42\_1)
- Q) Little evidence found, filtered: a BND located in chr10:58717662 (Valle-Inclan ID truthset\_43\_1)
- R) No evidence found, filtered: a 100 bp INS located in chr14:105867667 (Valle-Inclan ID truthset\_51\_1)
- S) Little evidence found, filtered: a BND located in chr15:23461732 (Valle-Inclan ID truthset\_53\_1)



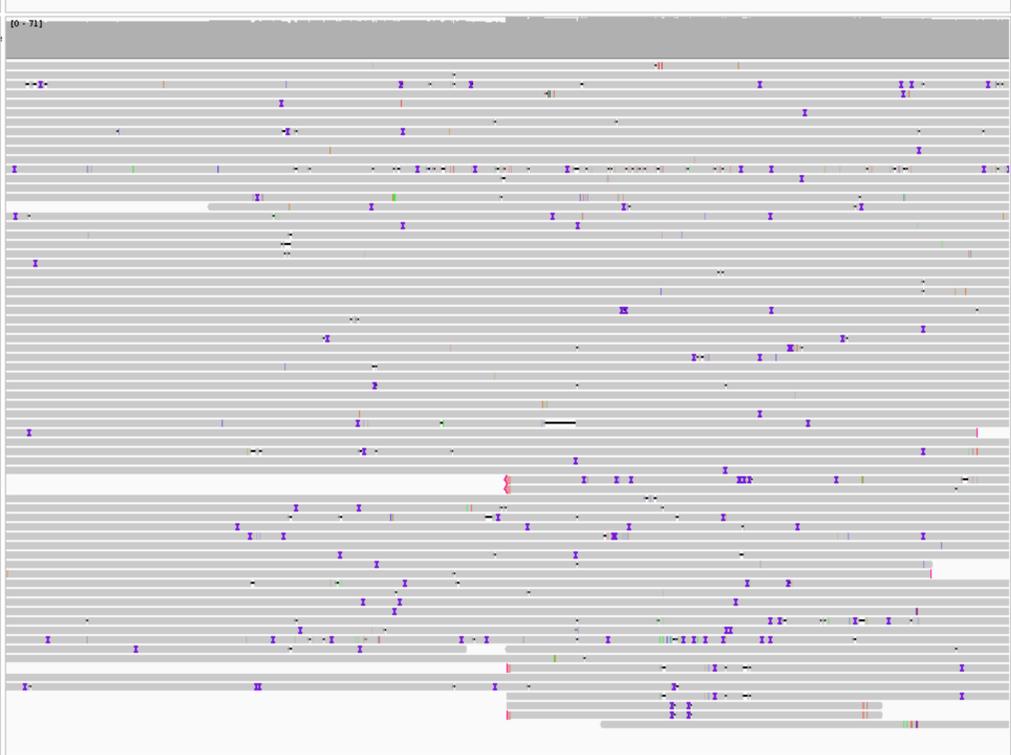
chr10:82,067,410-82,068,410  
p15.1 p13 p12.31 p11.23 p11.1 q11.22 q21.1 q21.3 q22.2 q23.1 q23.32 q24.2 q25.1 q25.3 q26.13 q26.3



colo829\_gsc\_lifted.bam Coverage  
colo829\_gsc\_lifted.bam



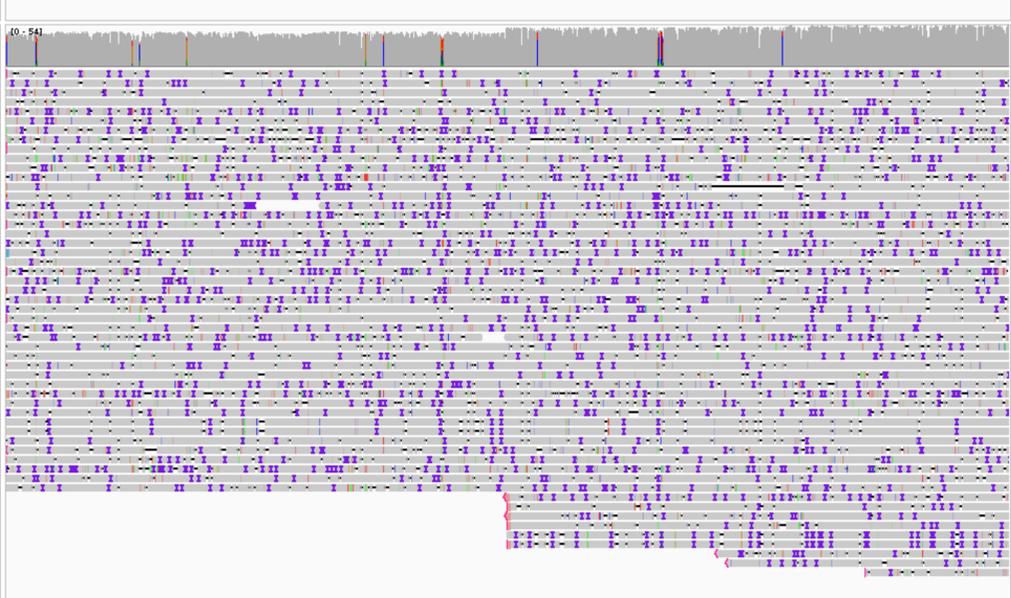
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colo829\_ort\_lifted.bam



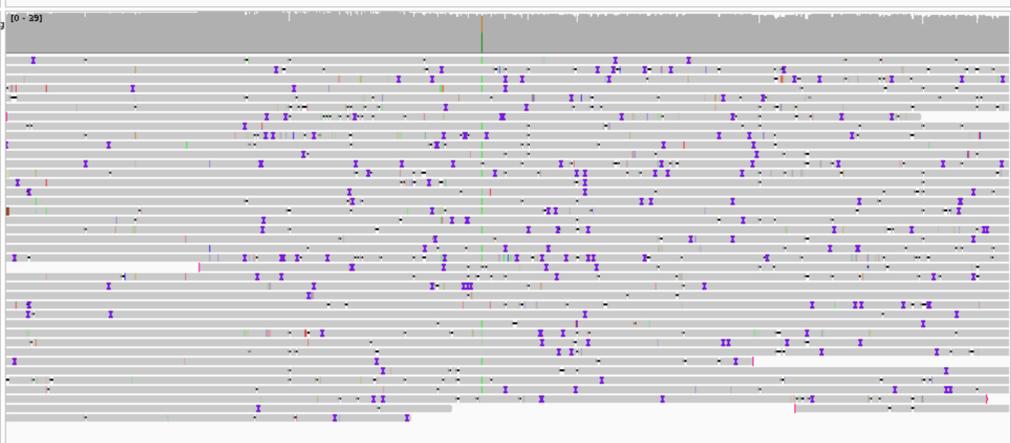
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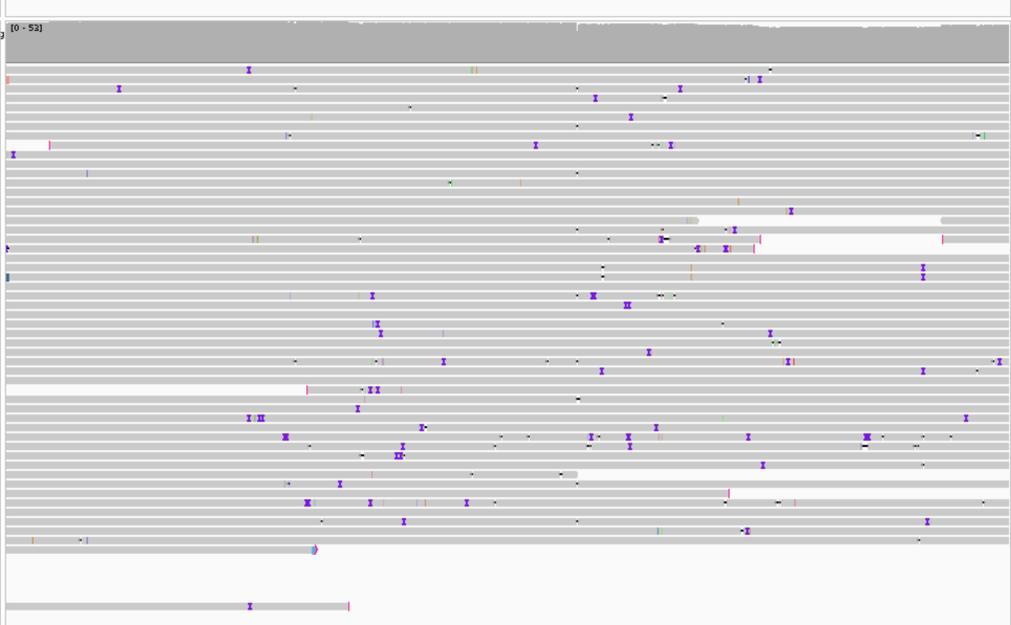
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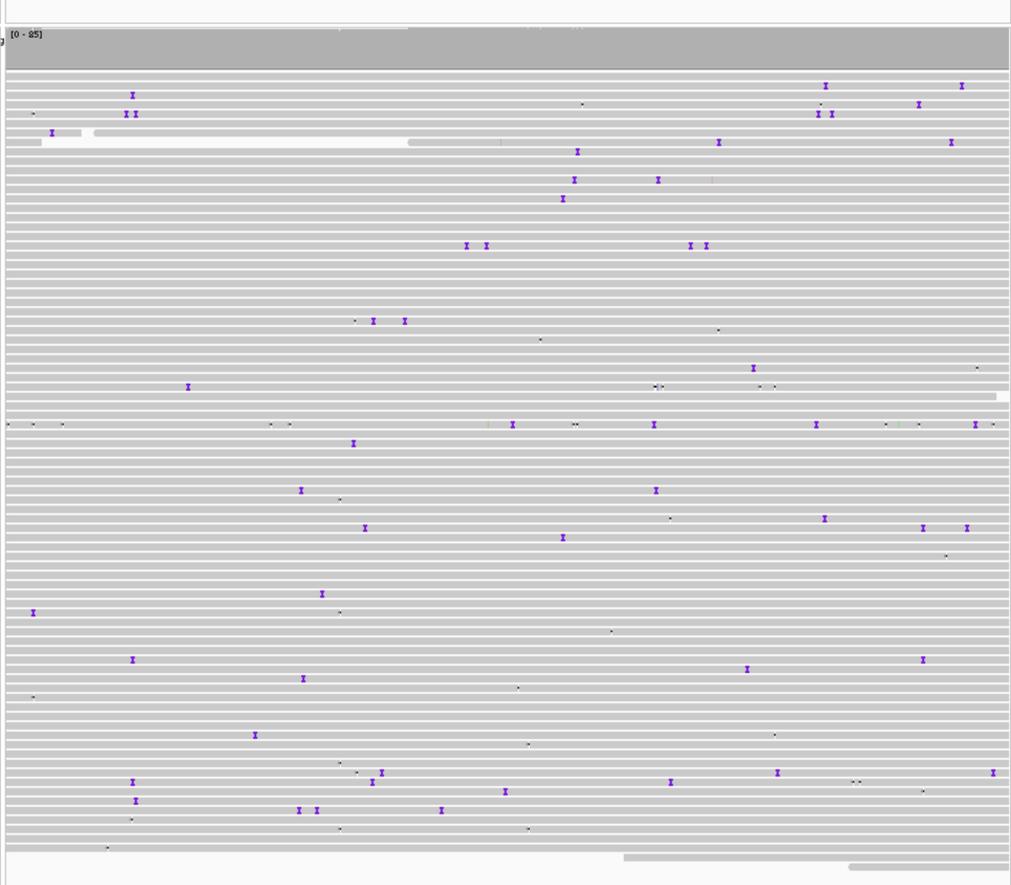
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colo829bl\_ort\_lifted.bam Coverage  
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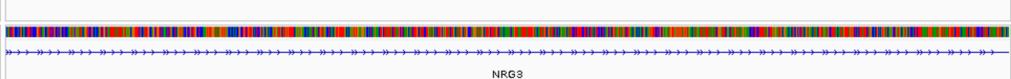
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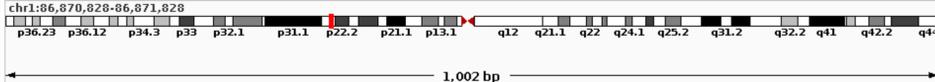


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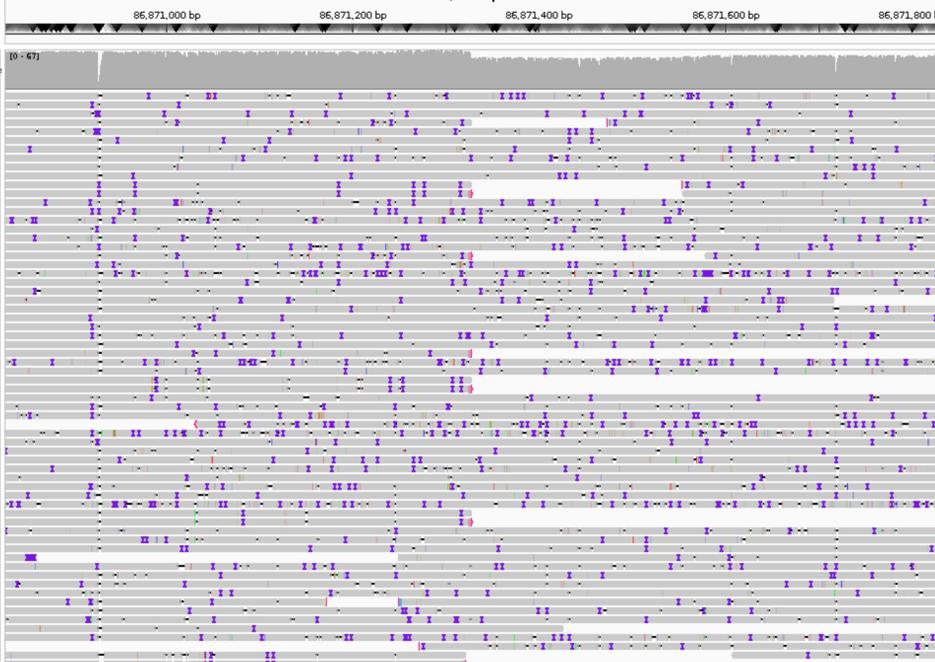


Sequence  
RefSeq Genes





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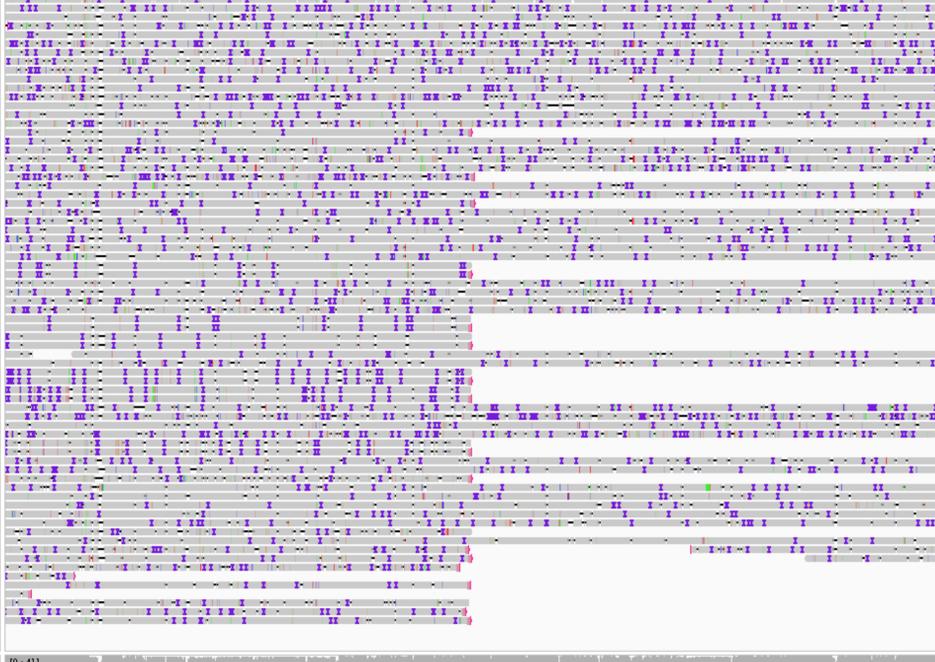
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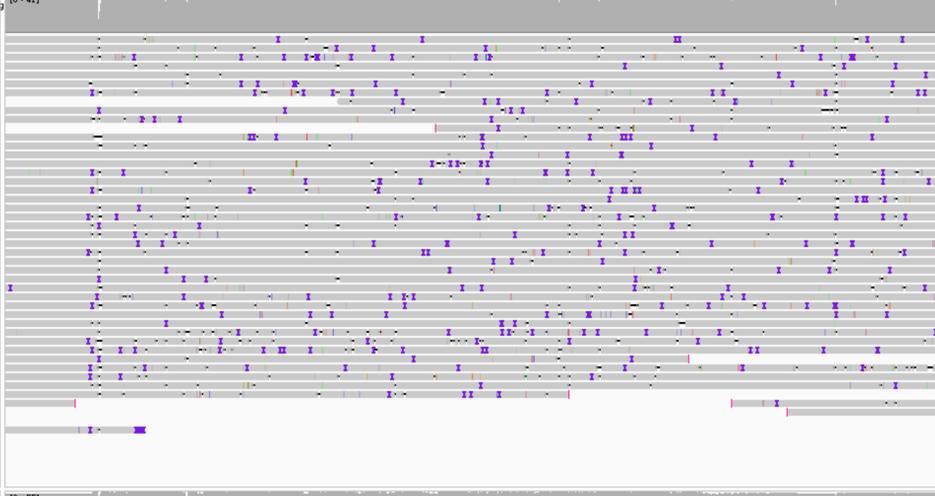
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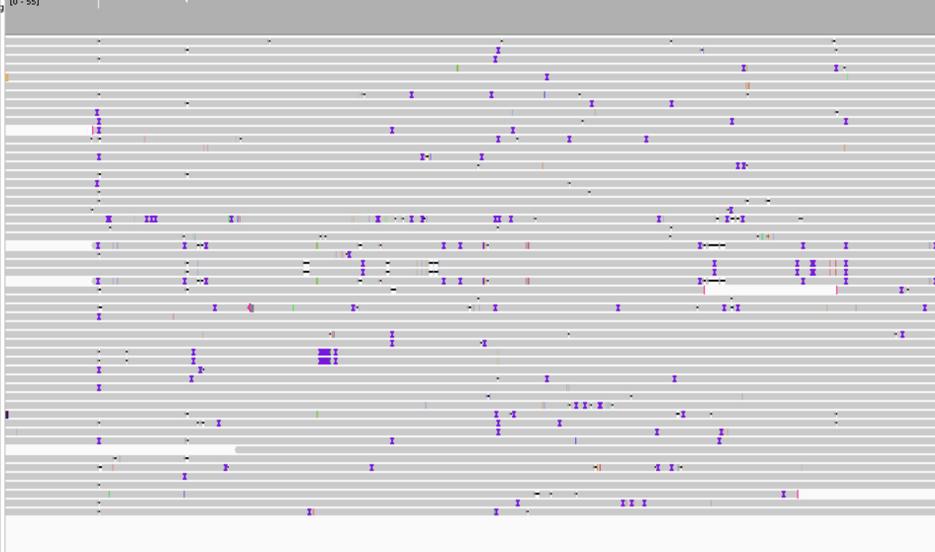
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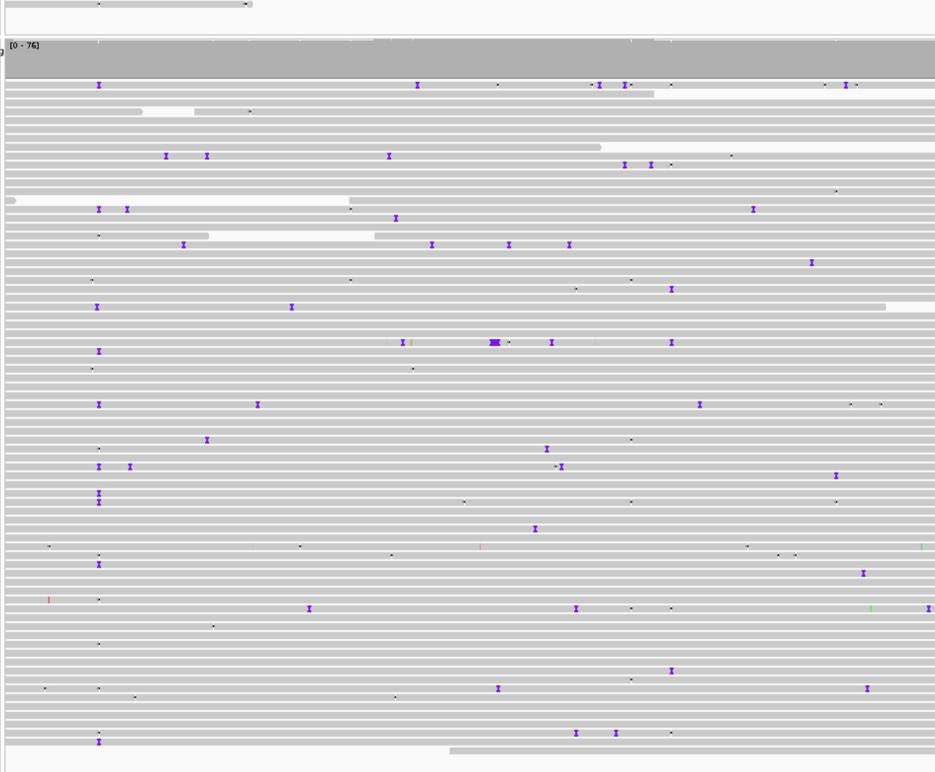
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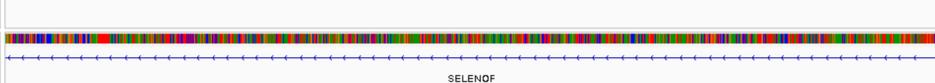
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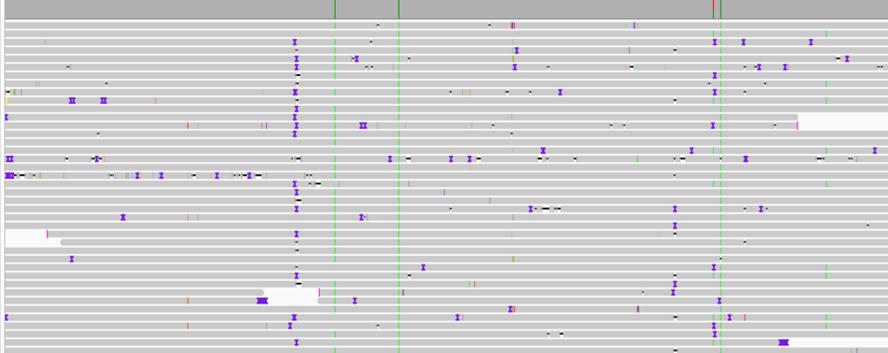
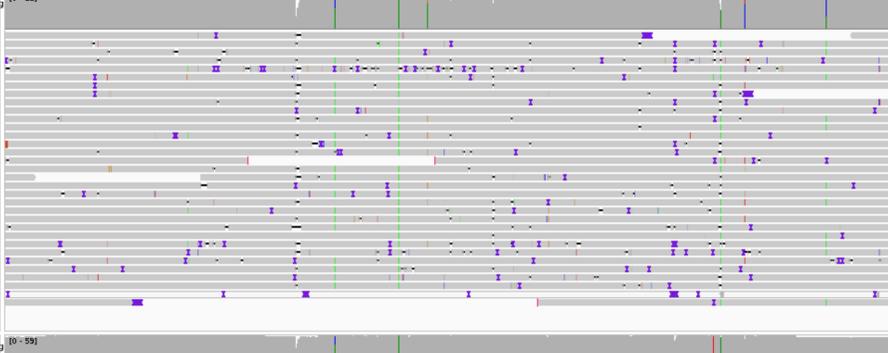
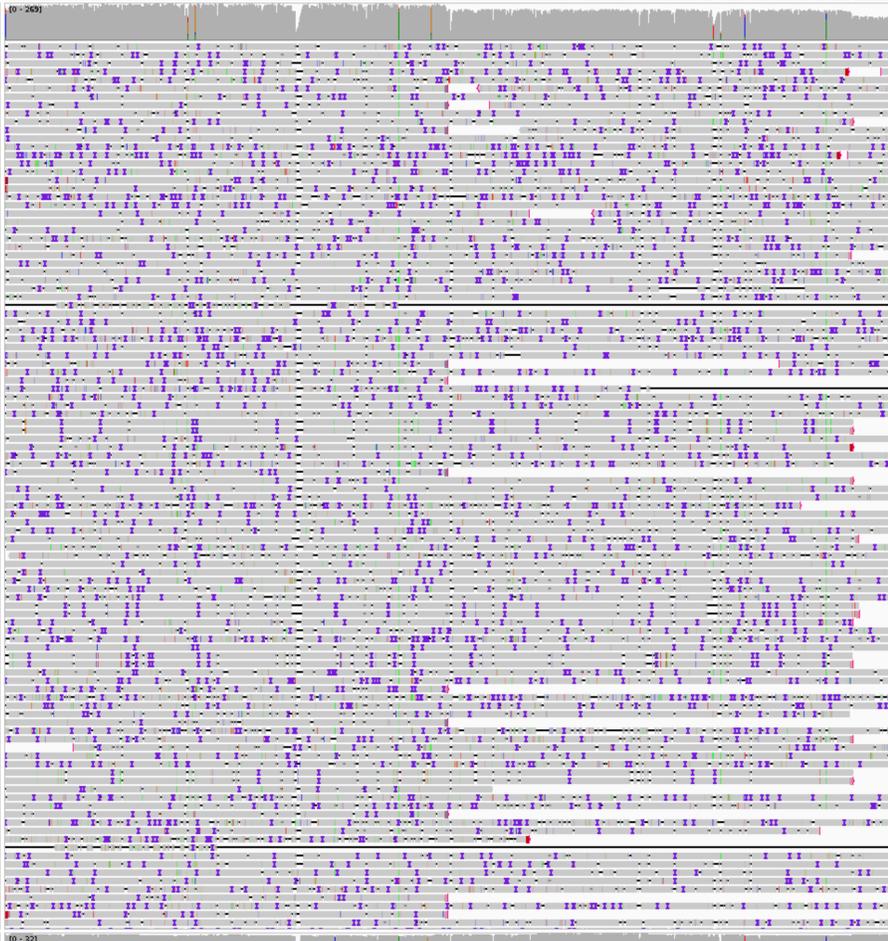
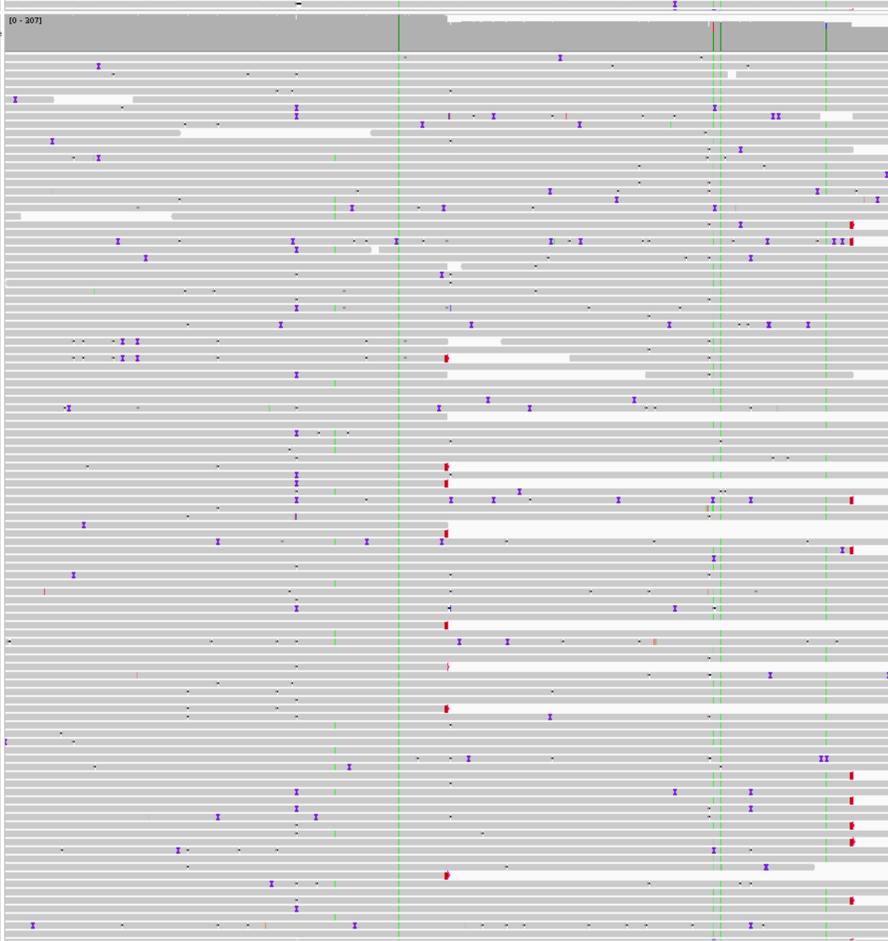


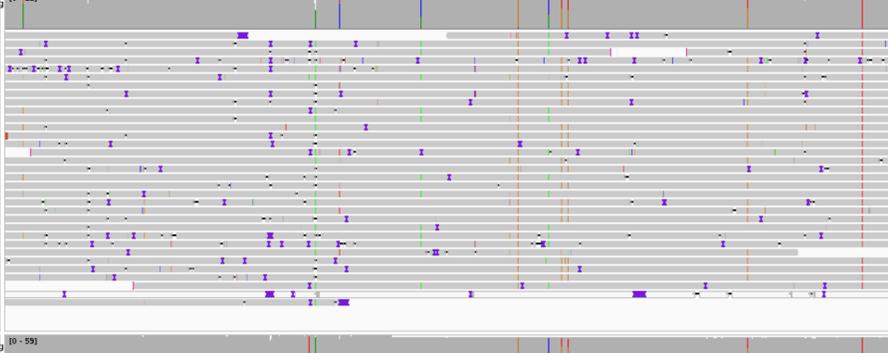
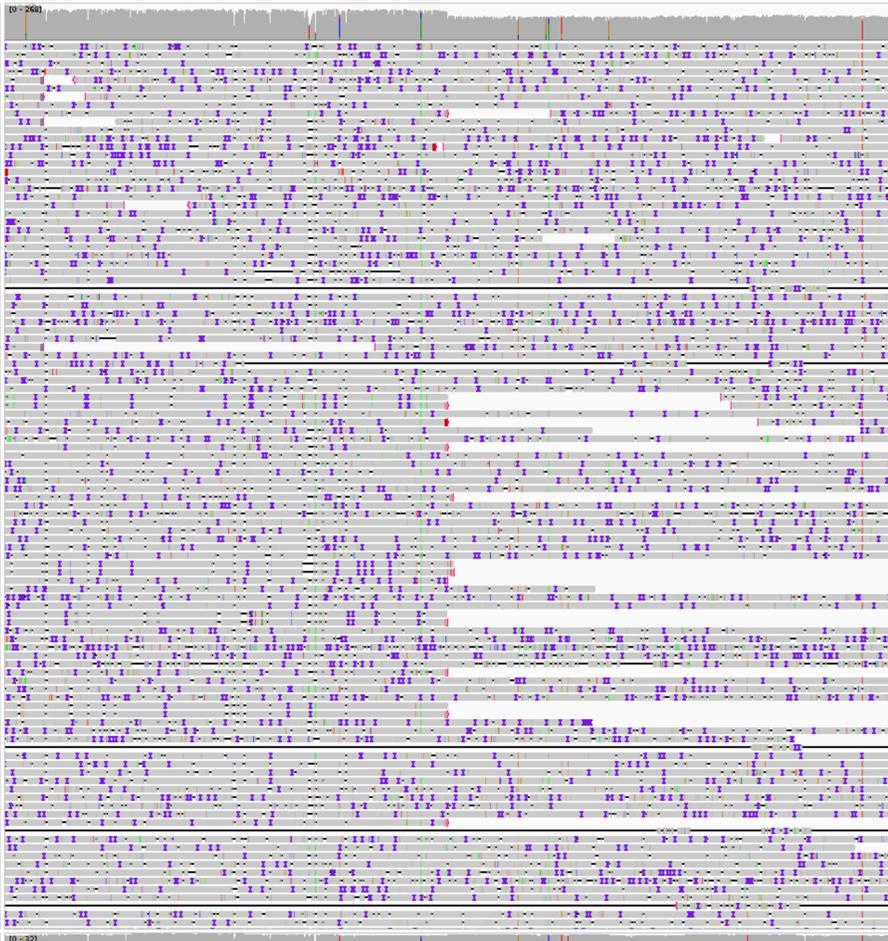
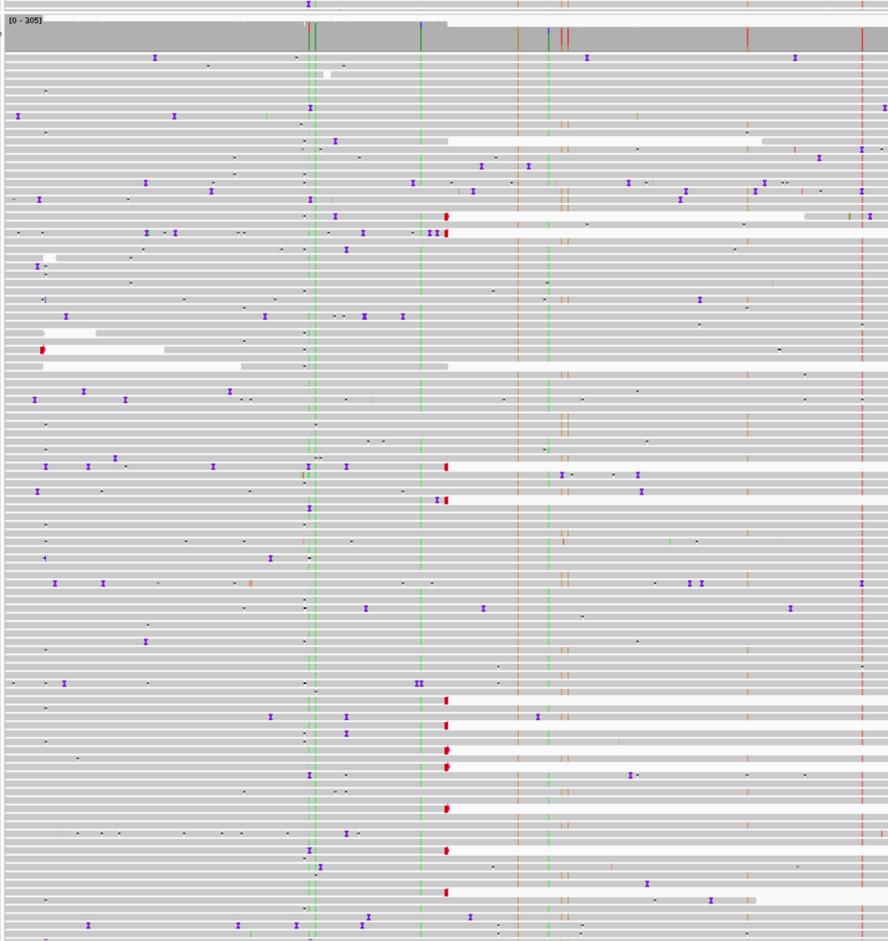
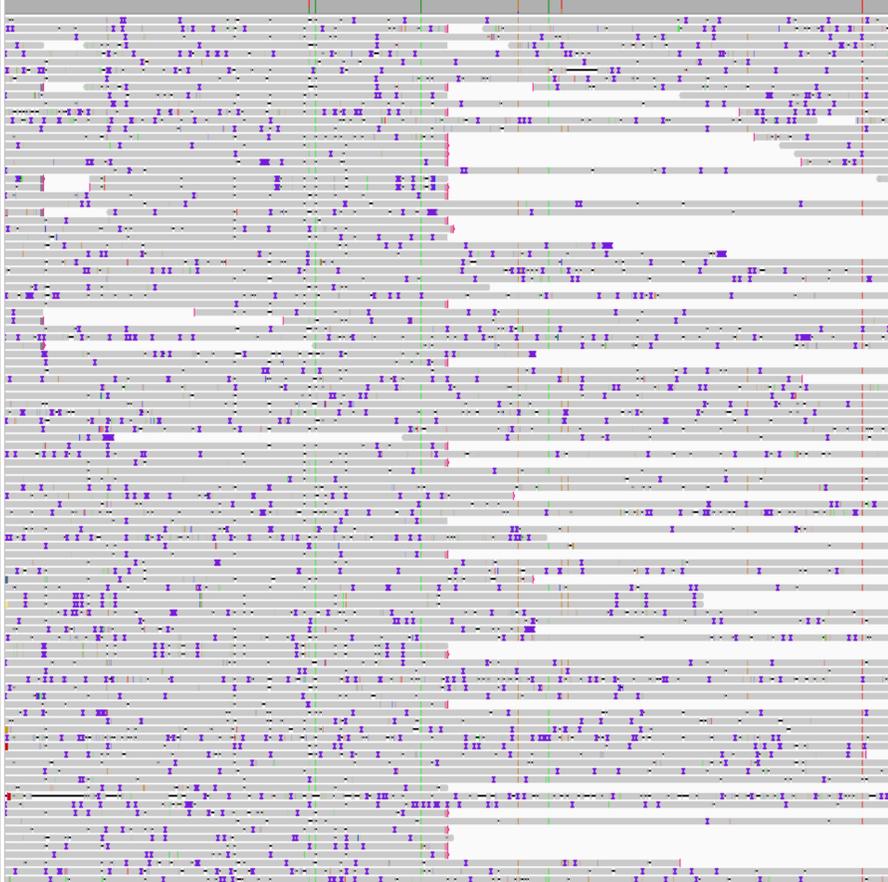
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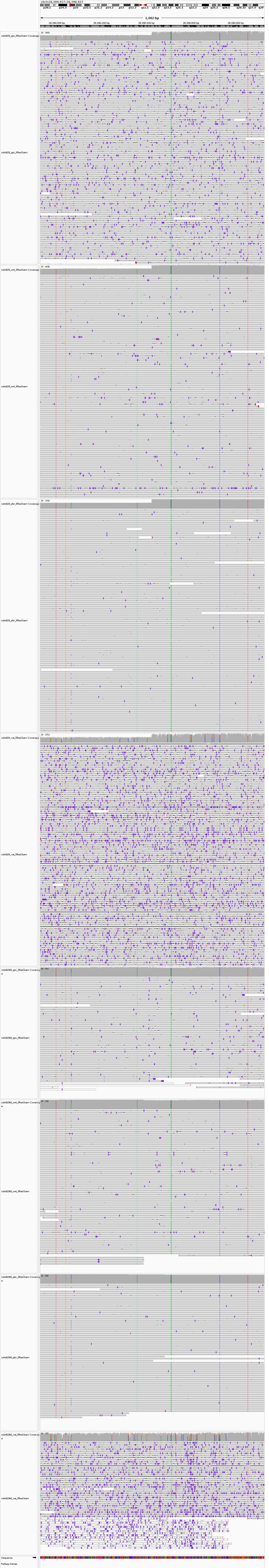


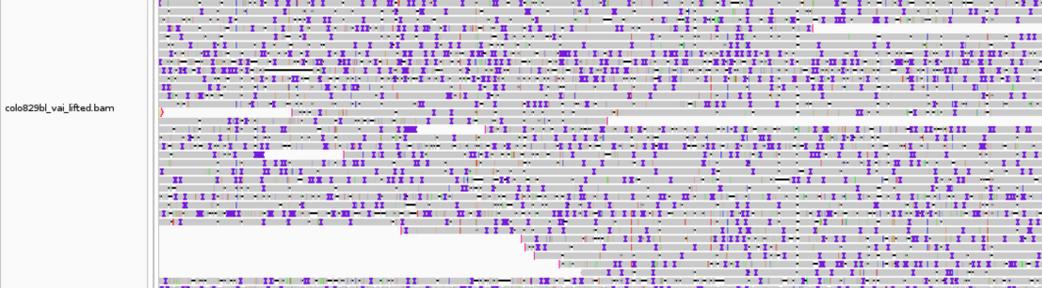
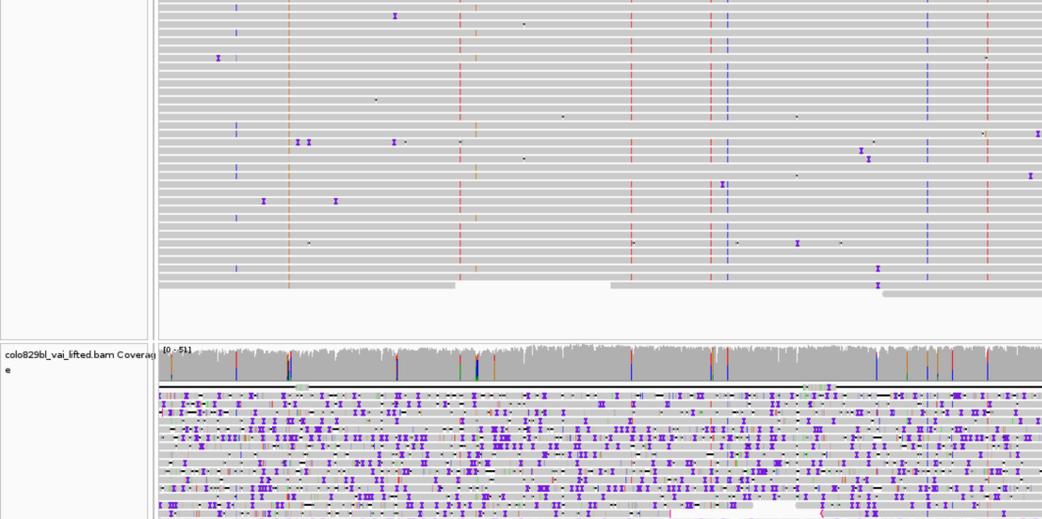
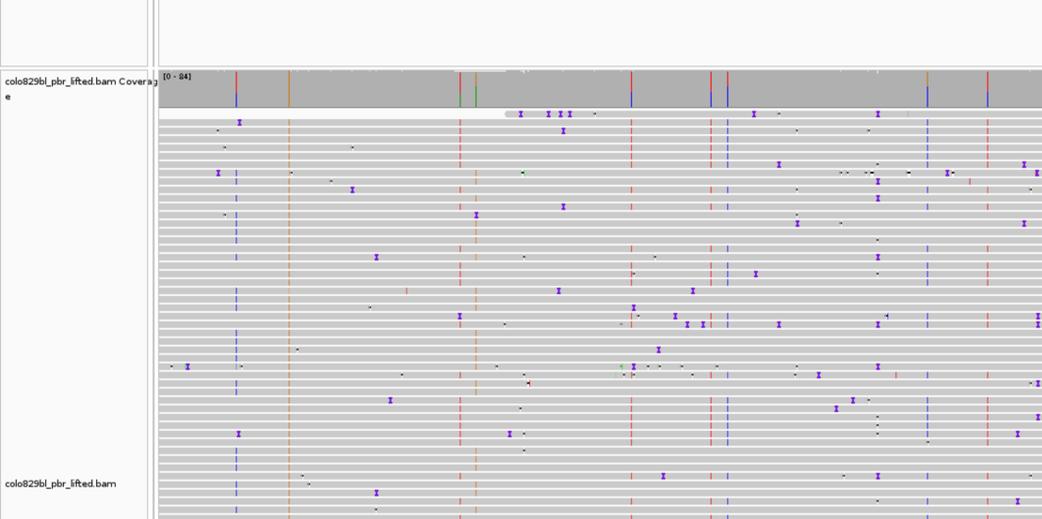
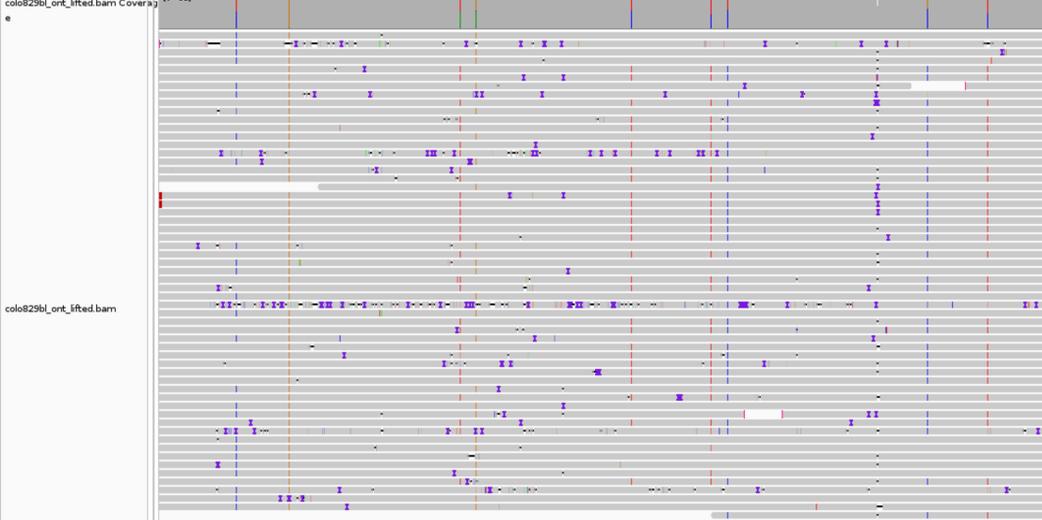
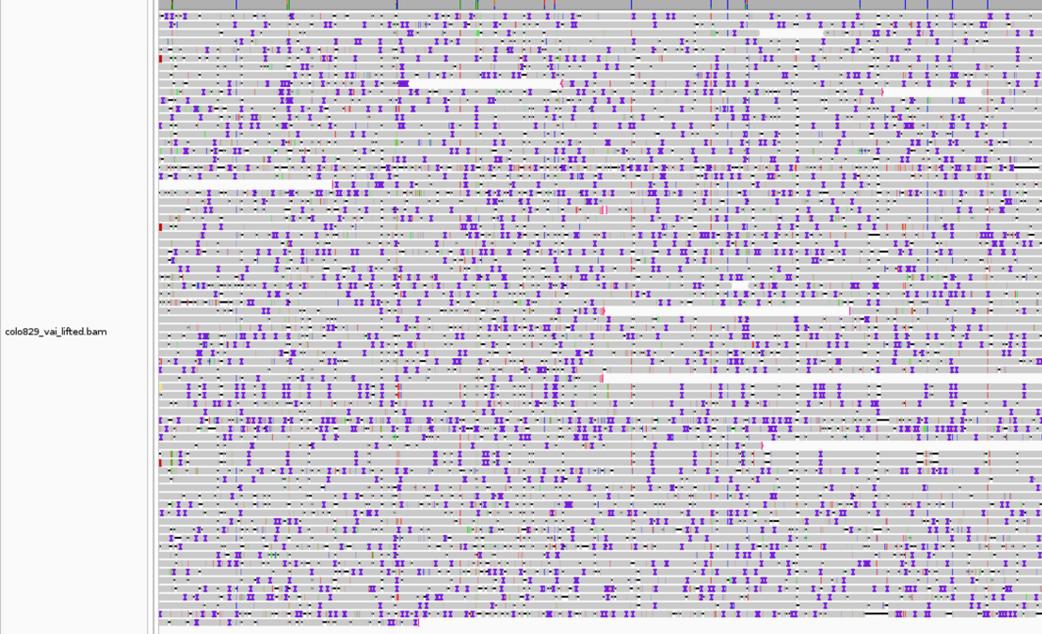
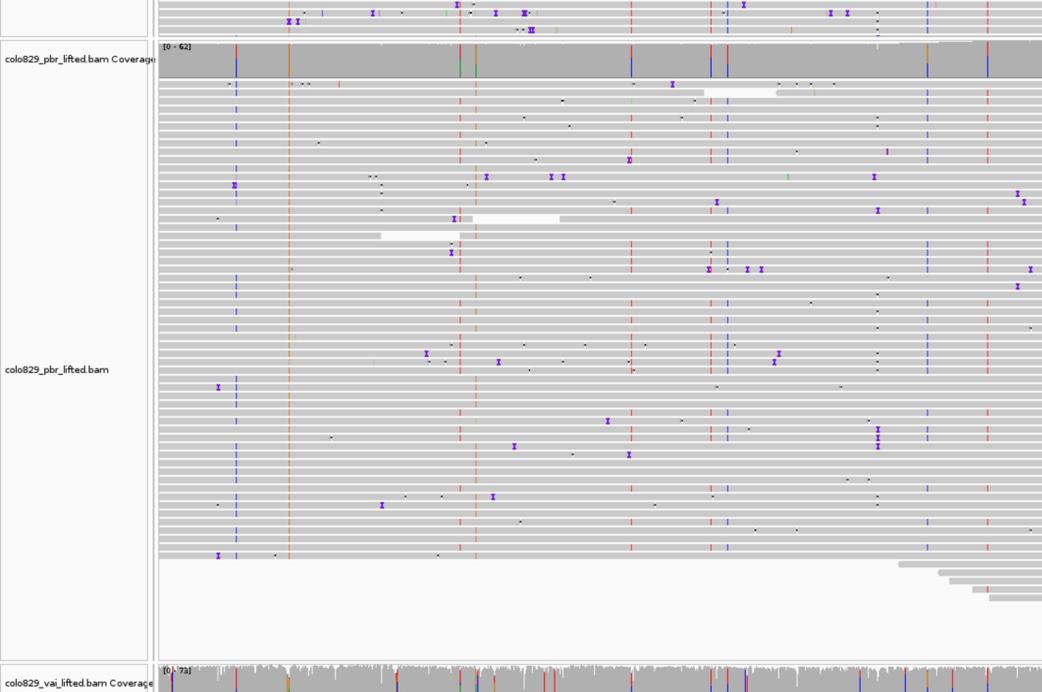
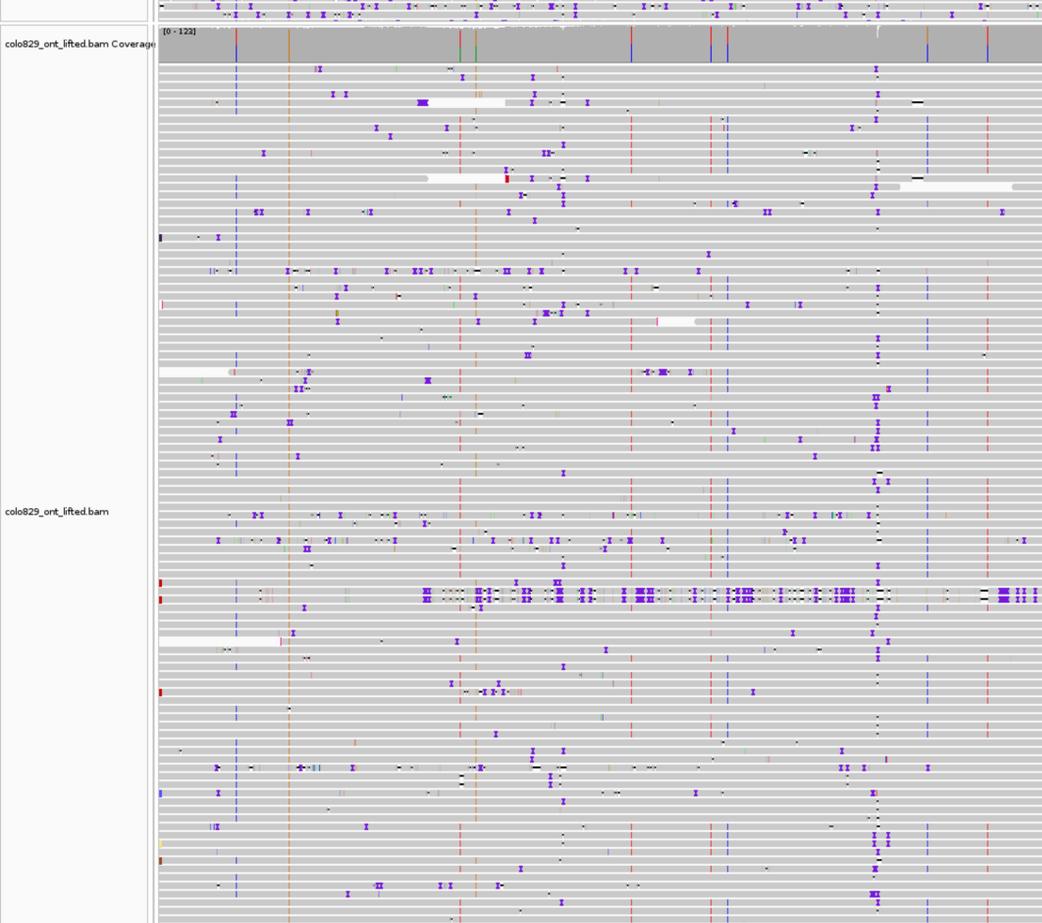
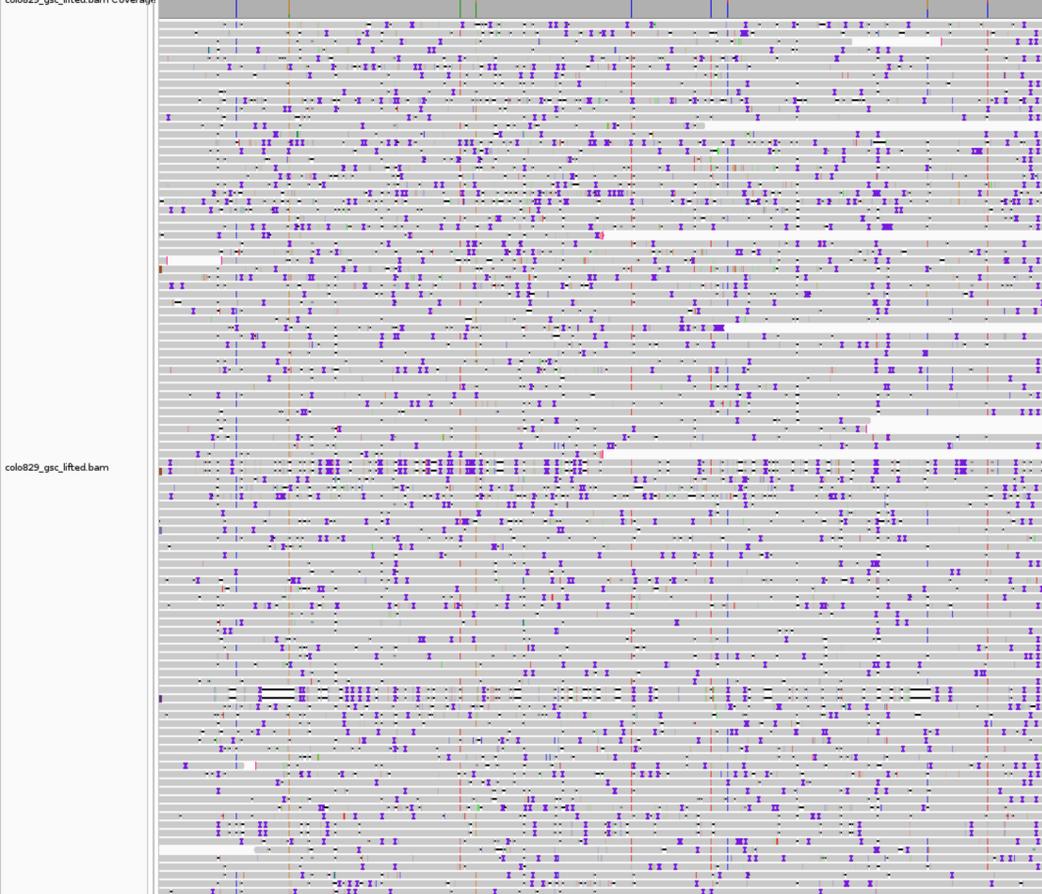
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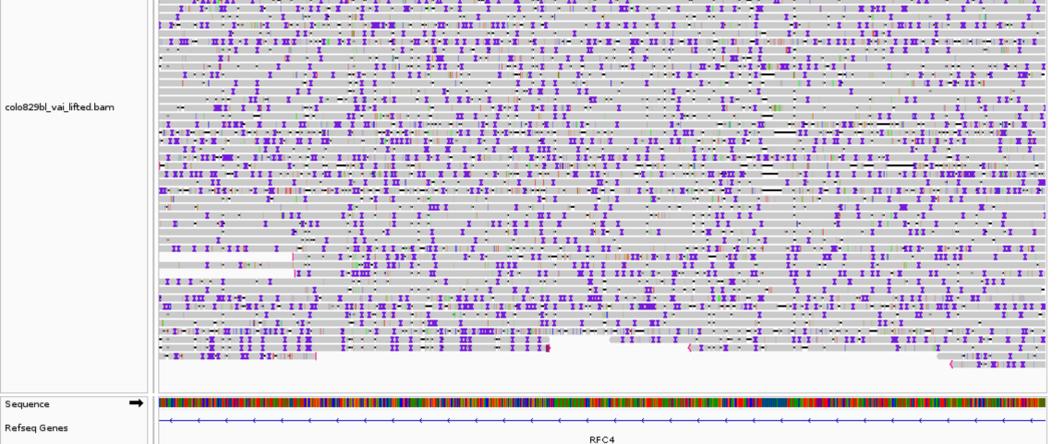
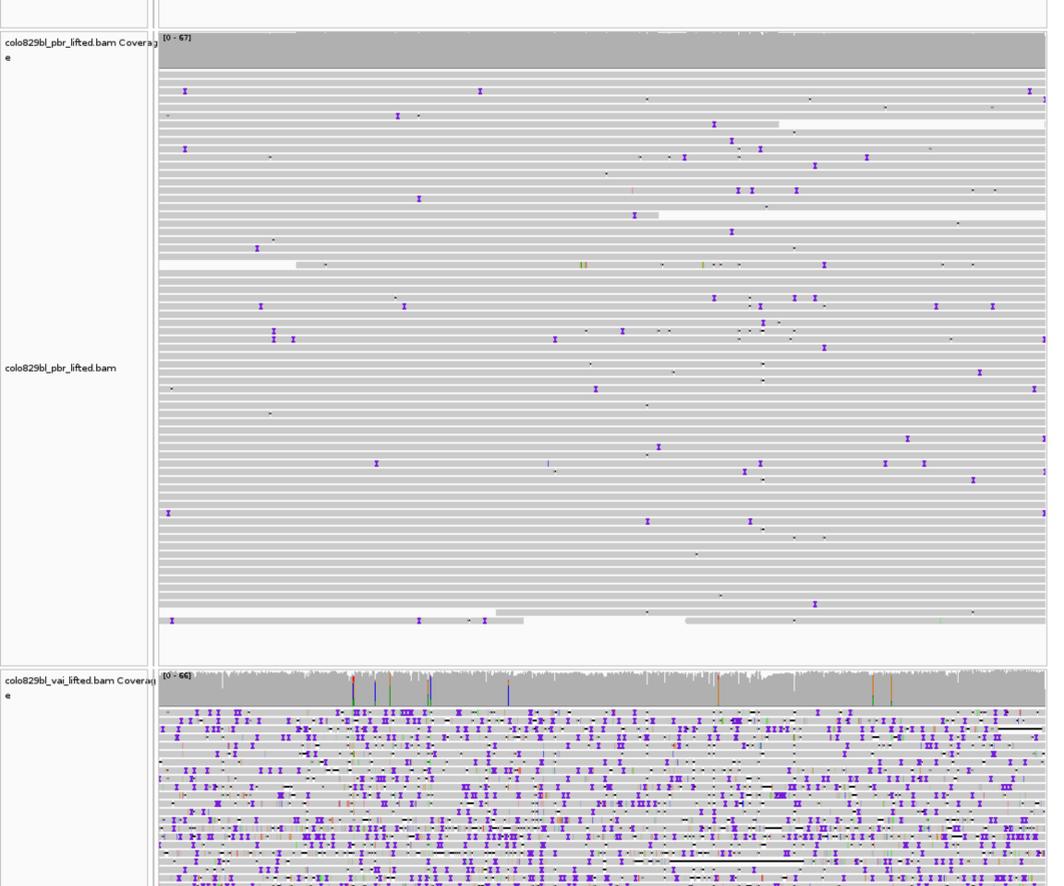
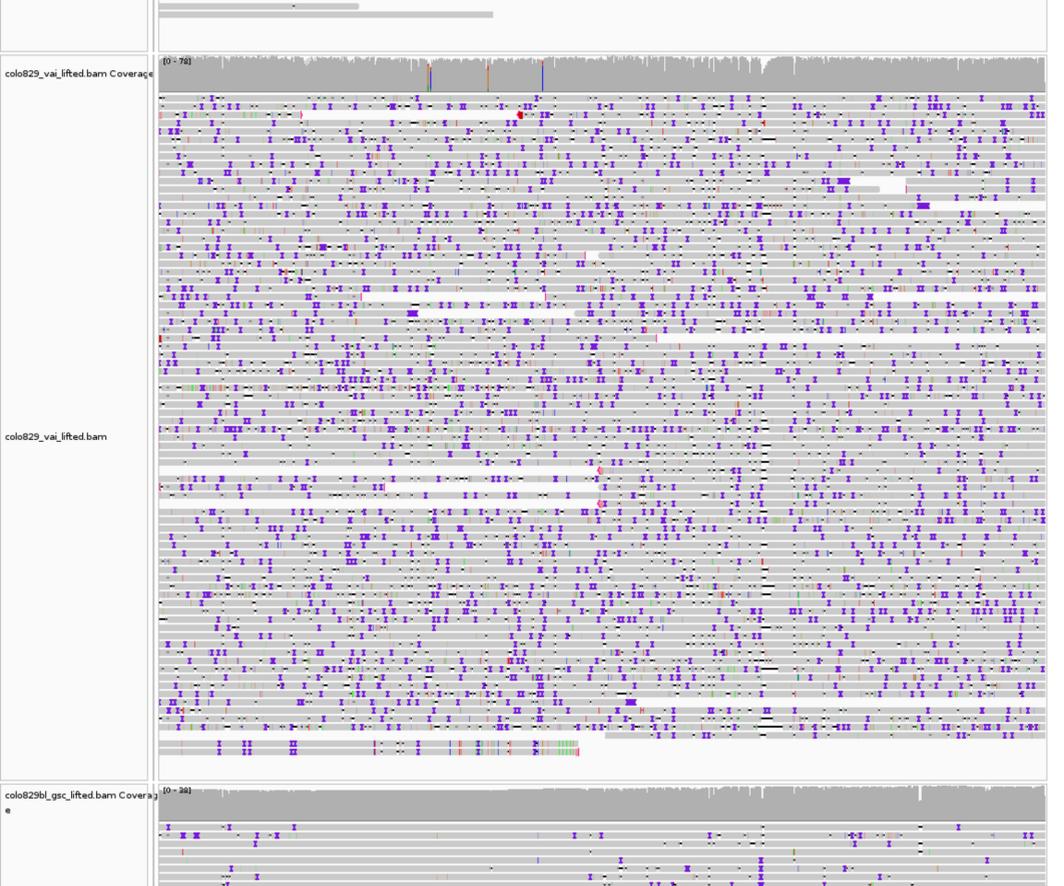




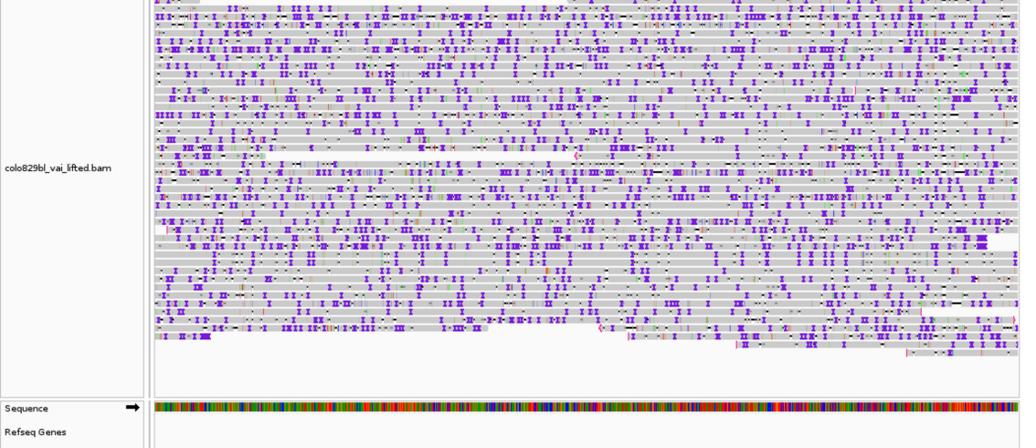
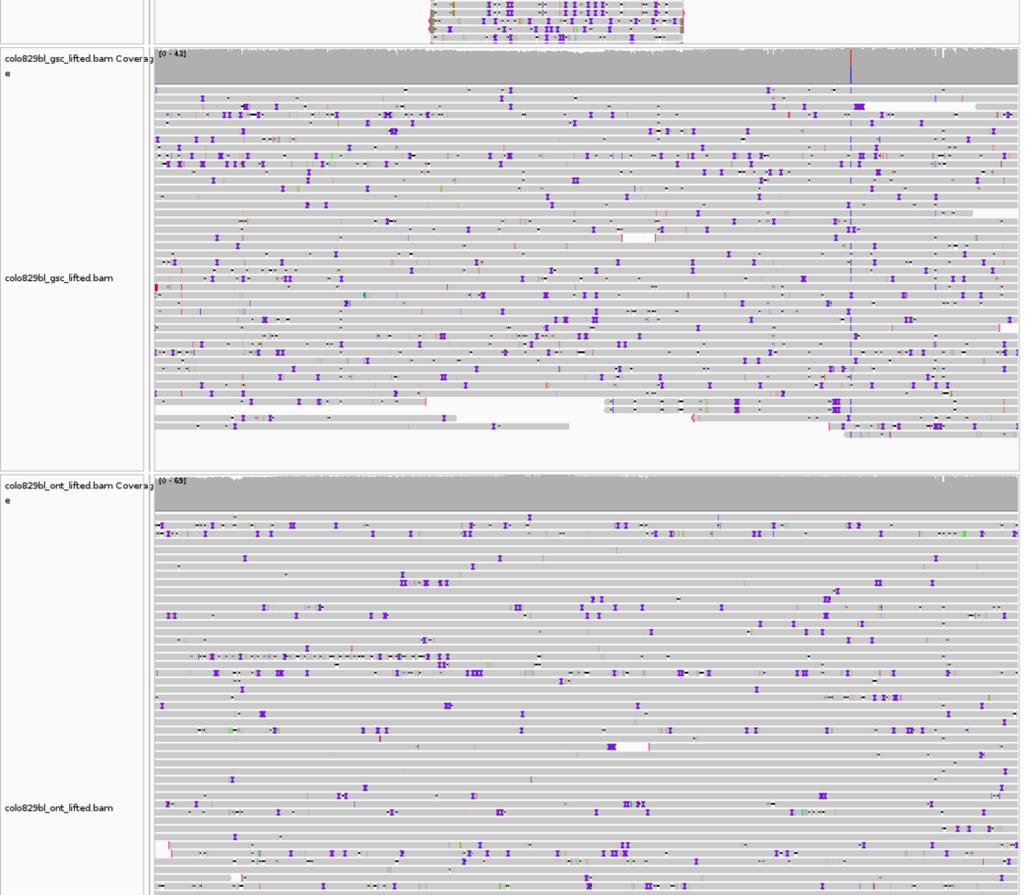
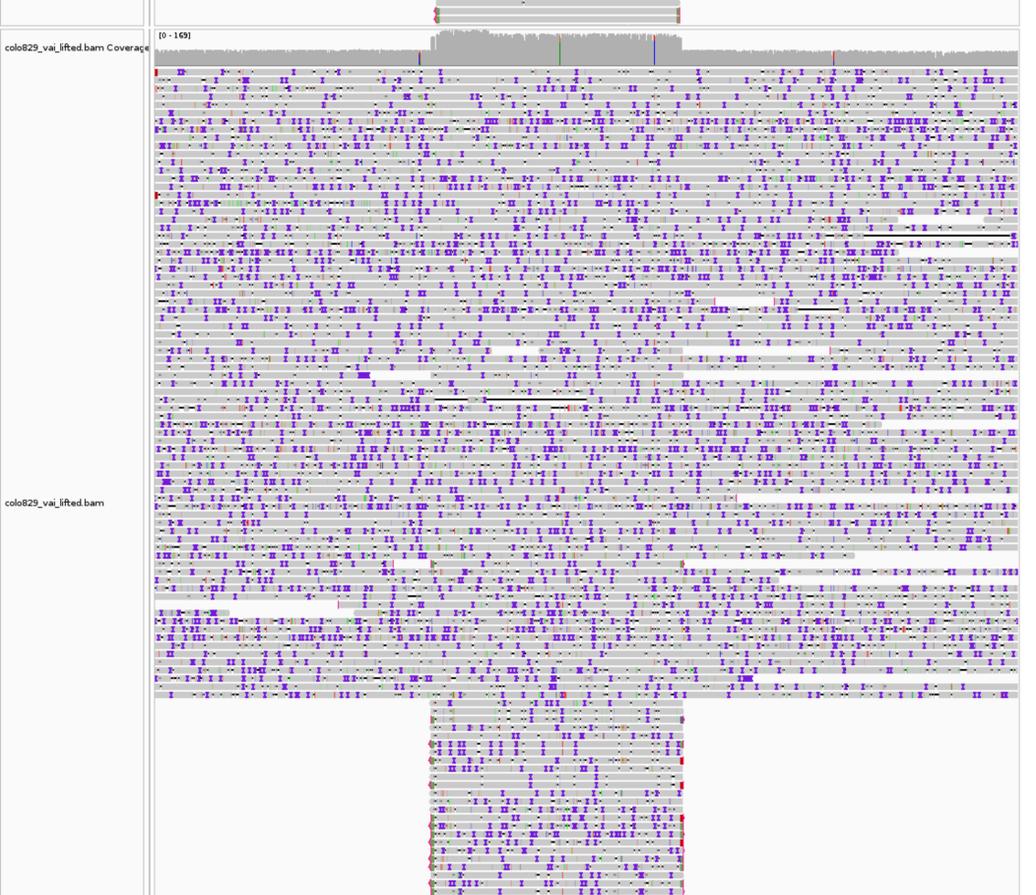
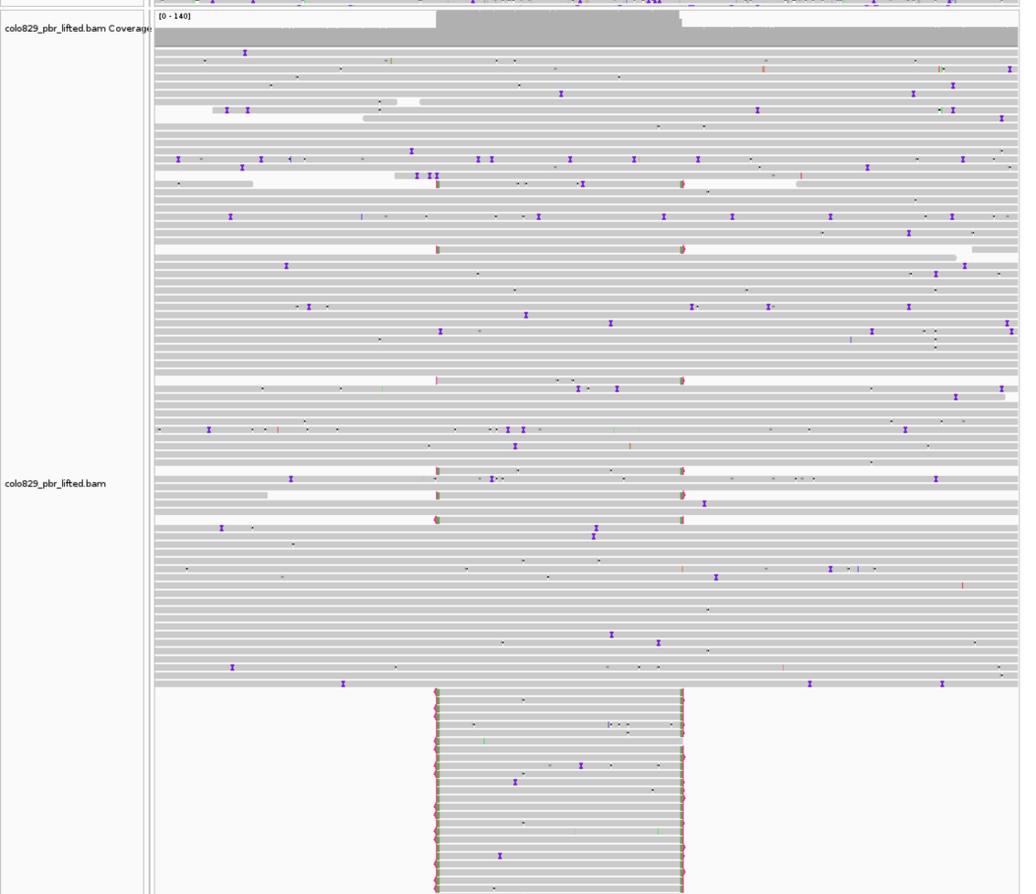
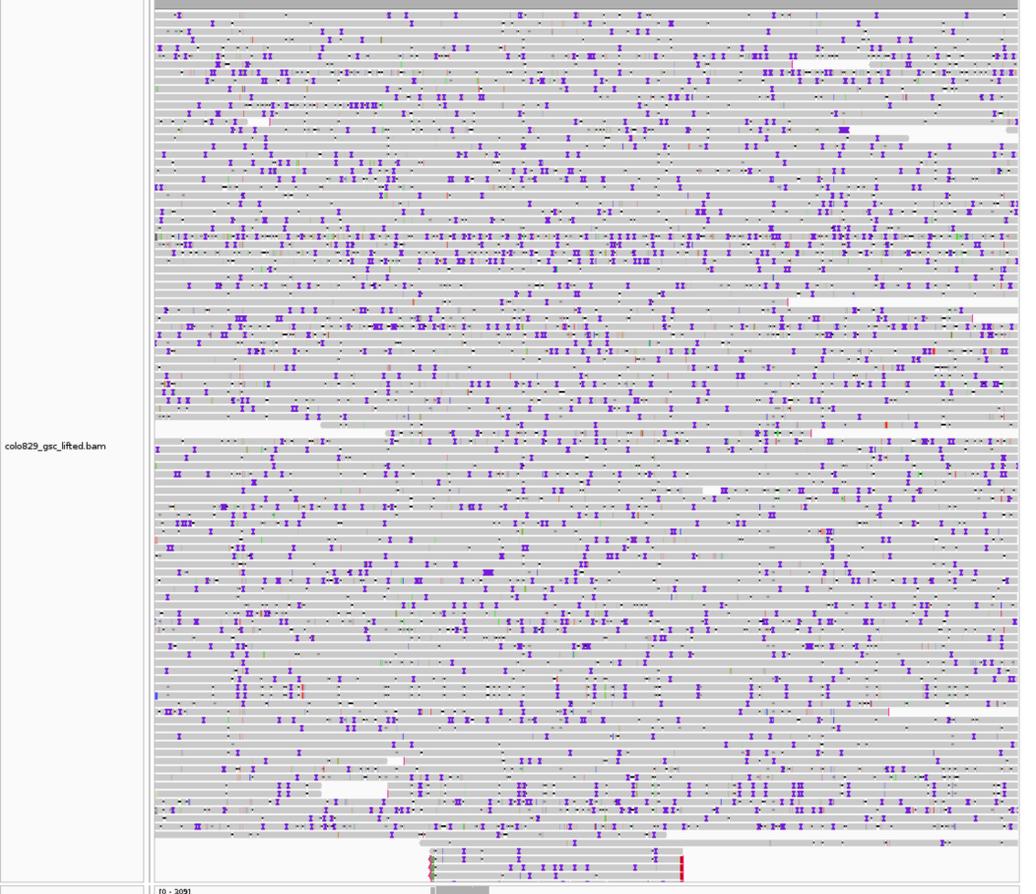






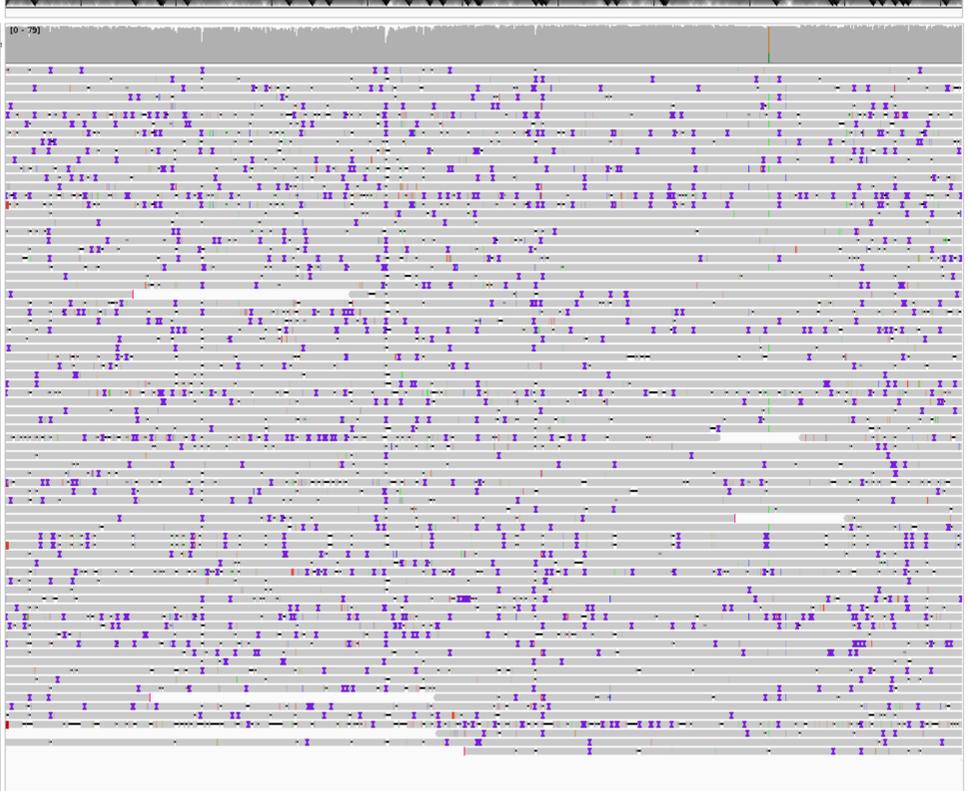


RFC4





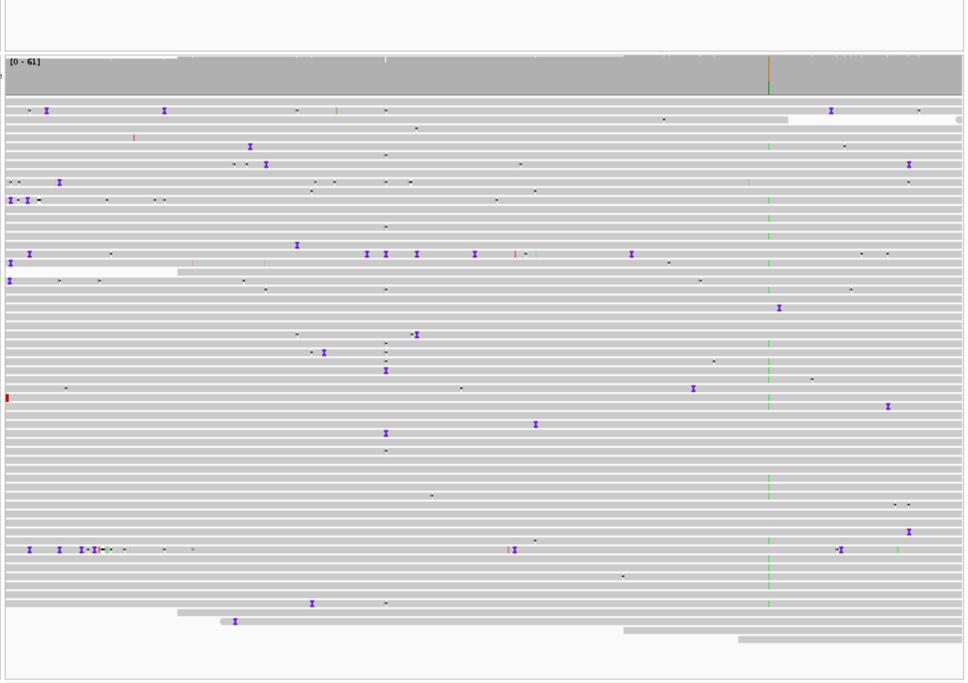
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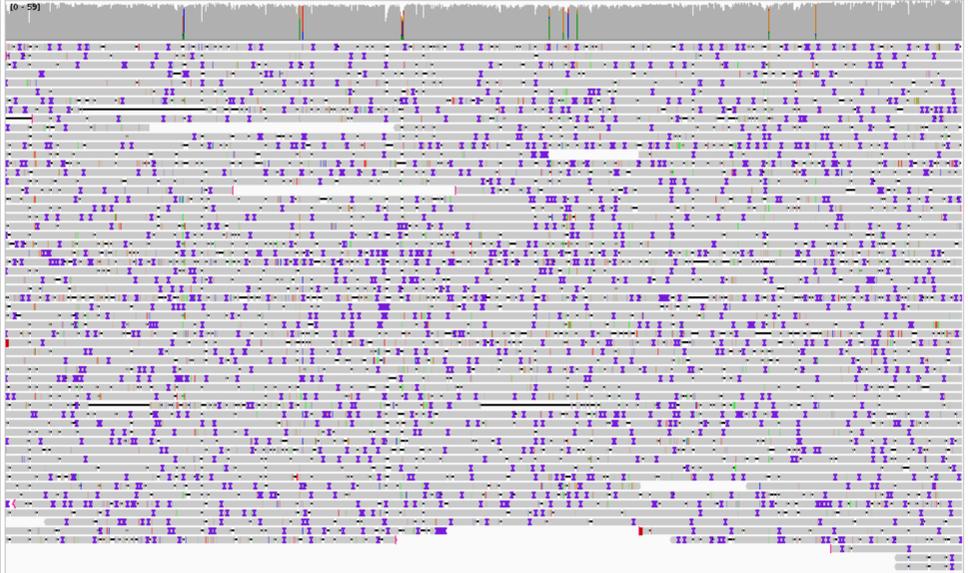
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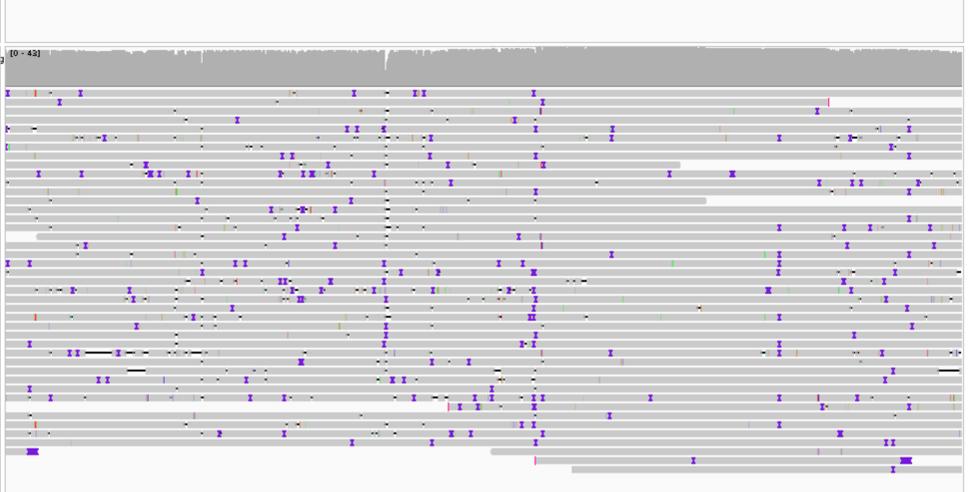
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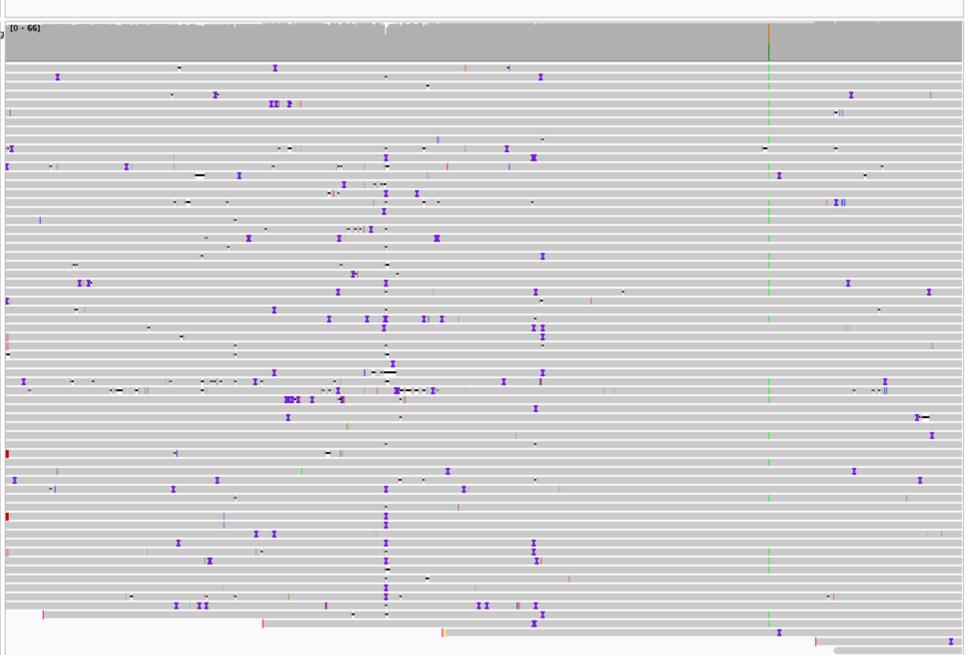
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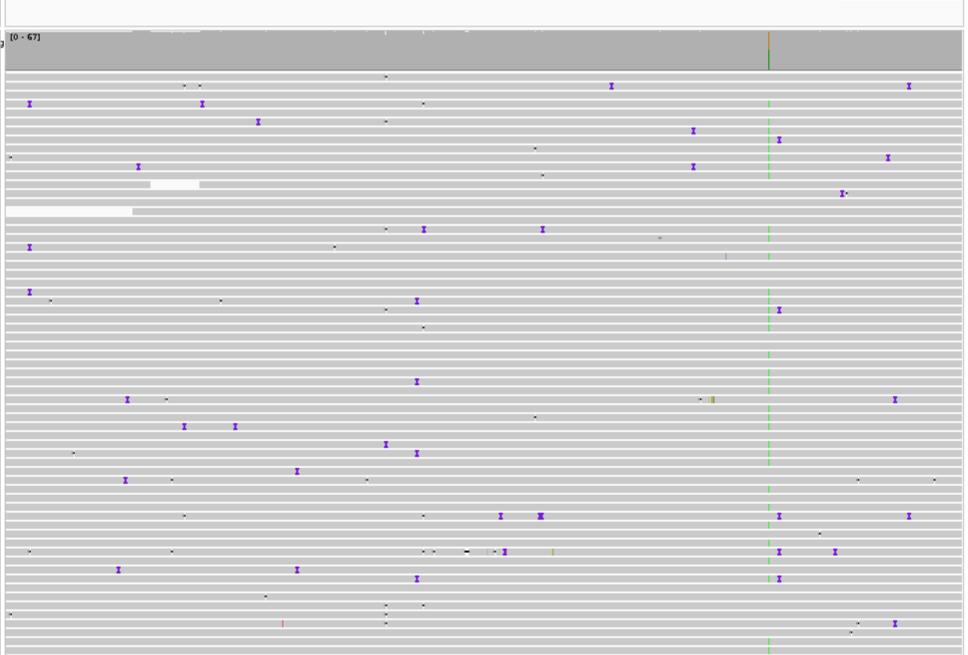
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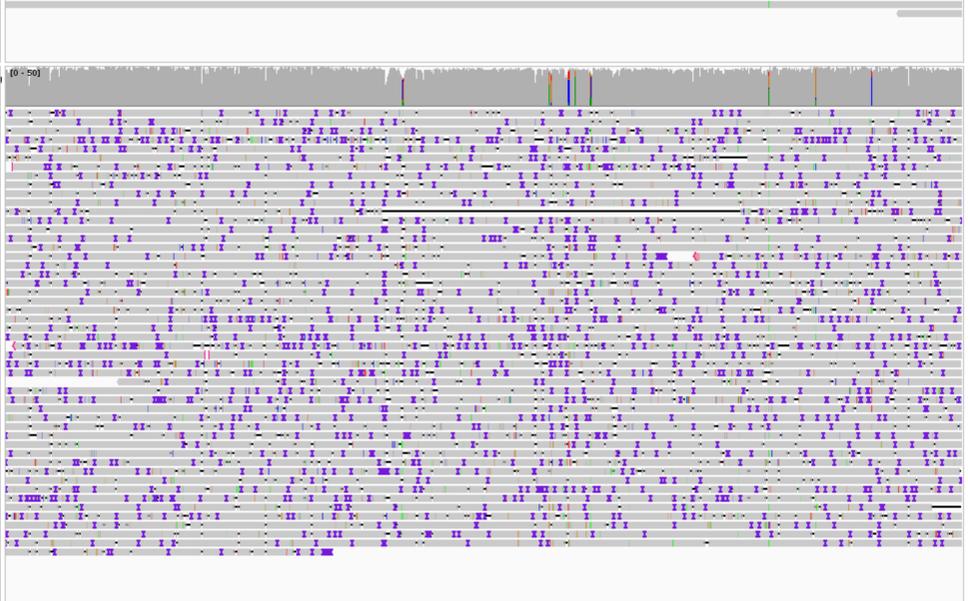
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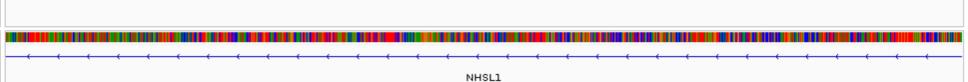
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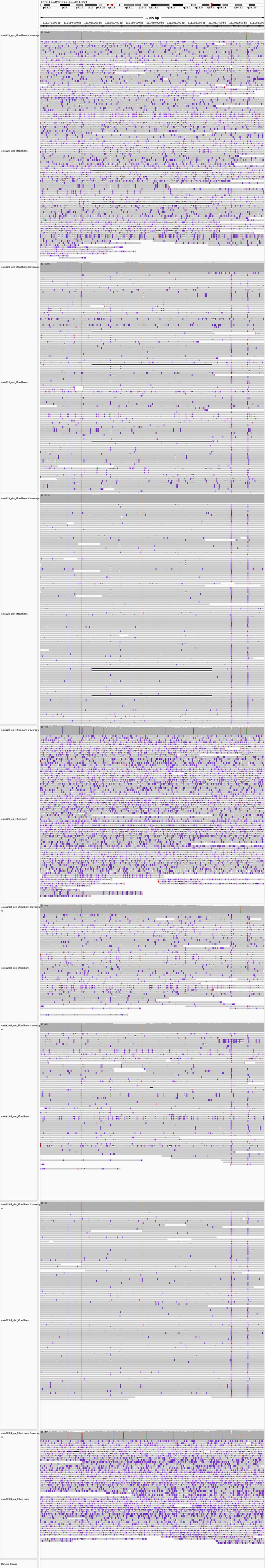
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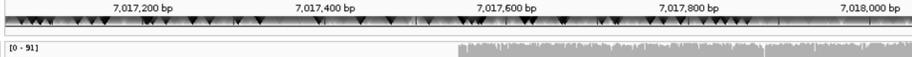


Sequence  
Refseq Genes









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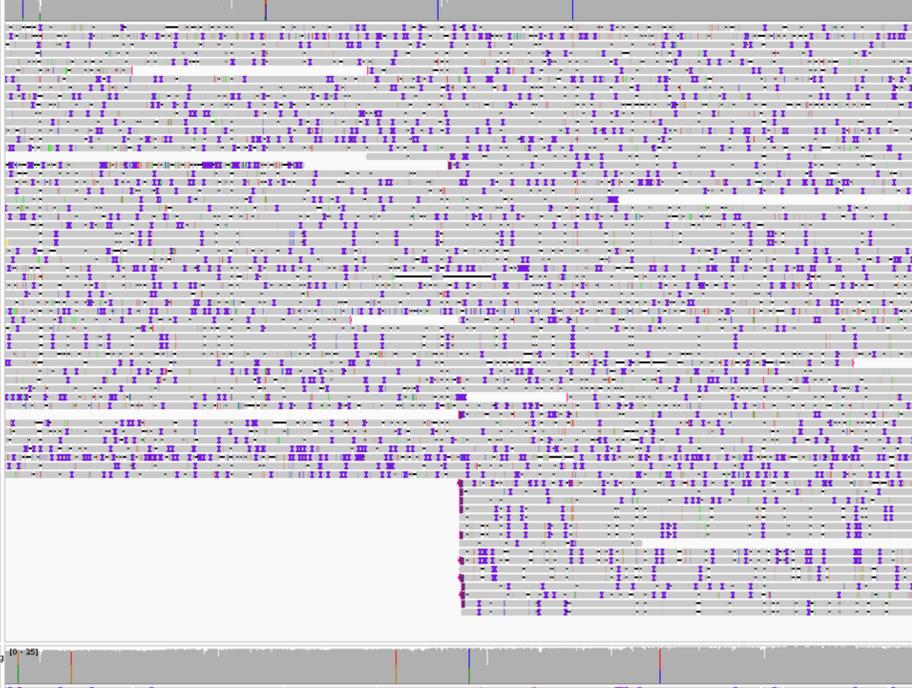
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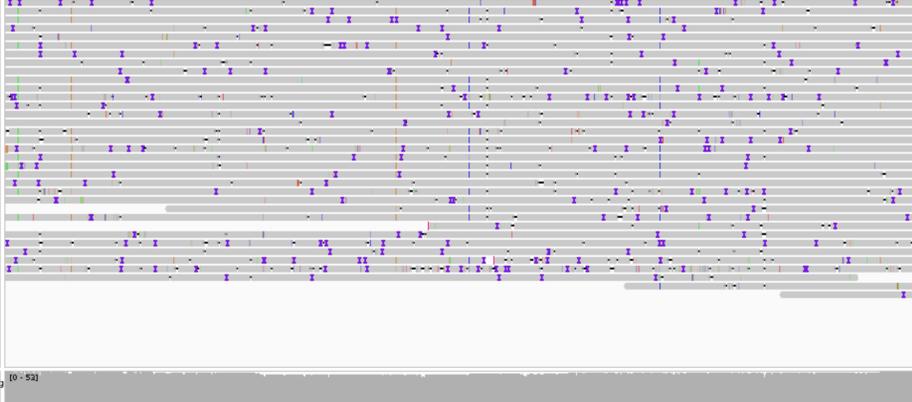
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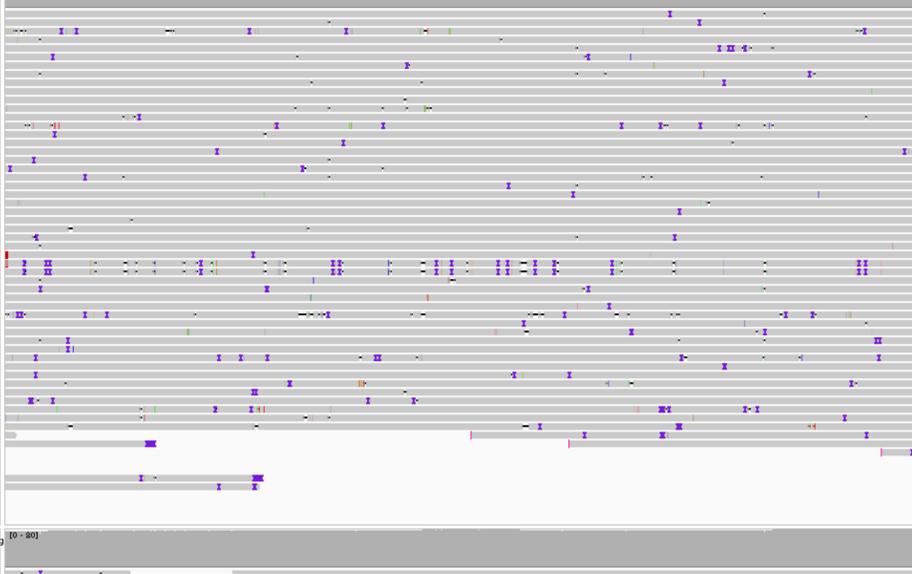
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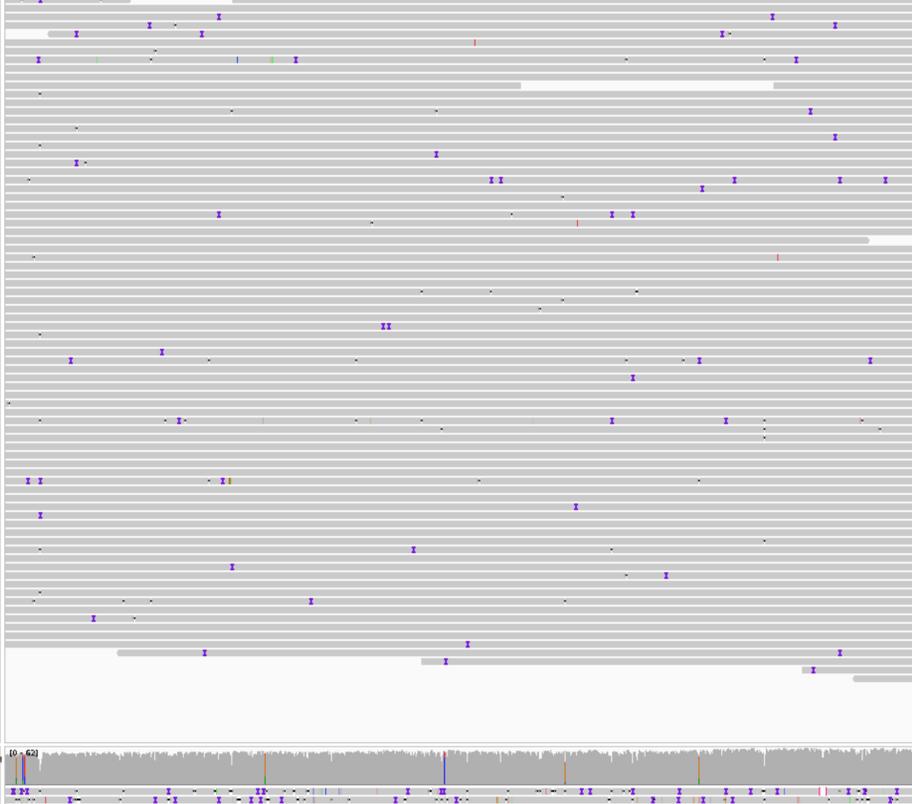
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e  
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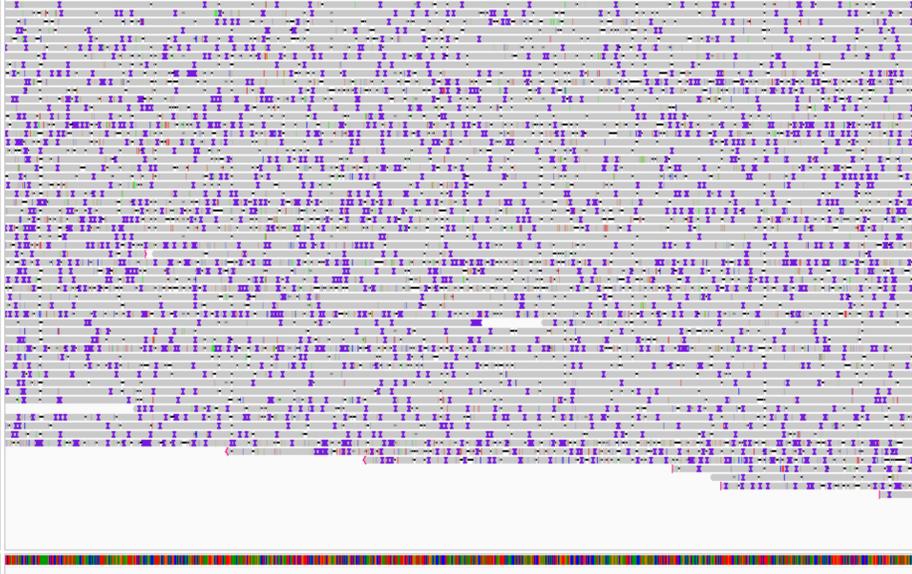
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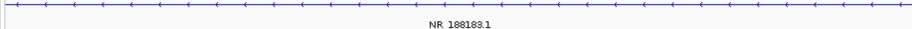
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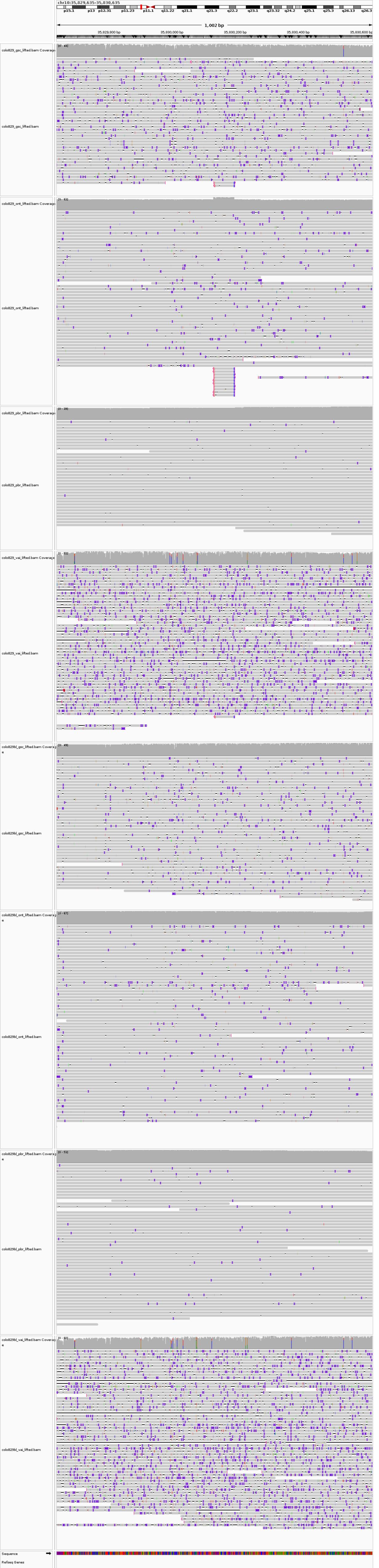
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Sequence  
Refseq Genes

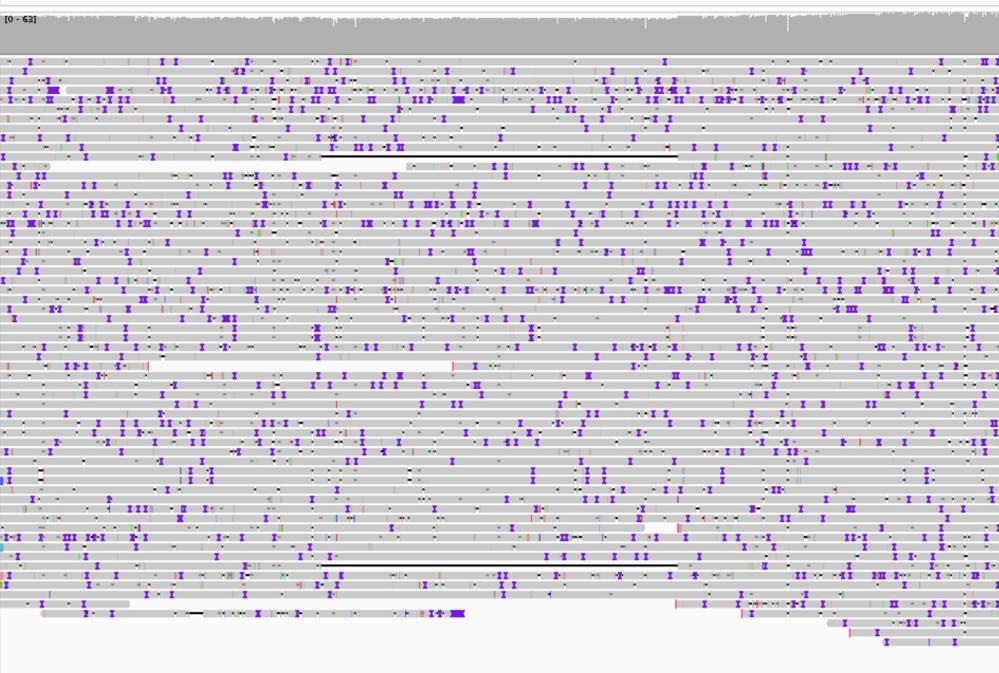








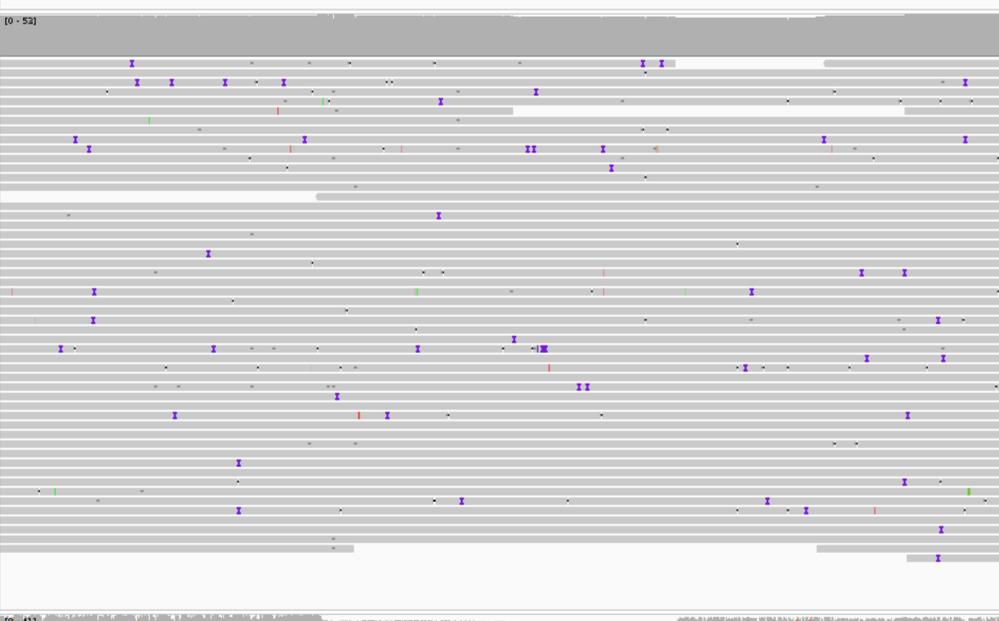
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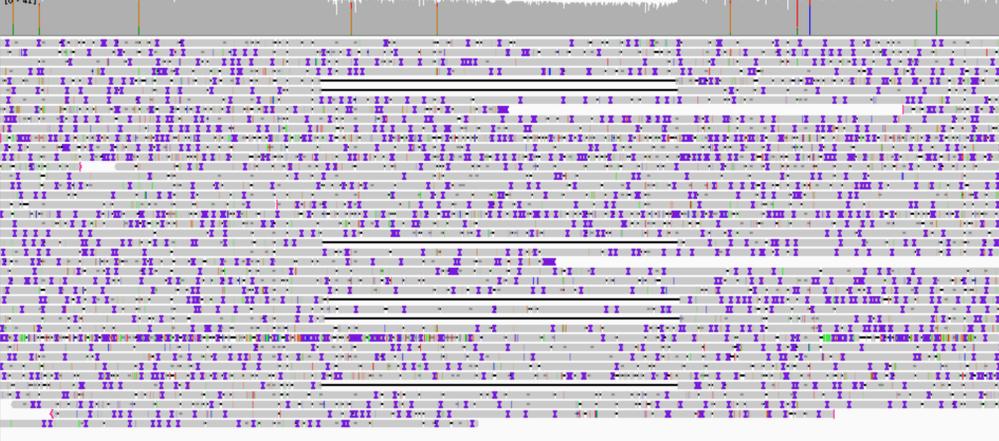
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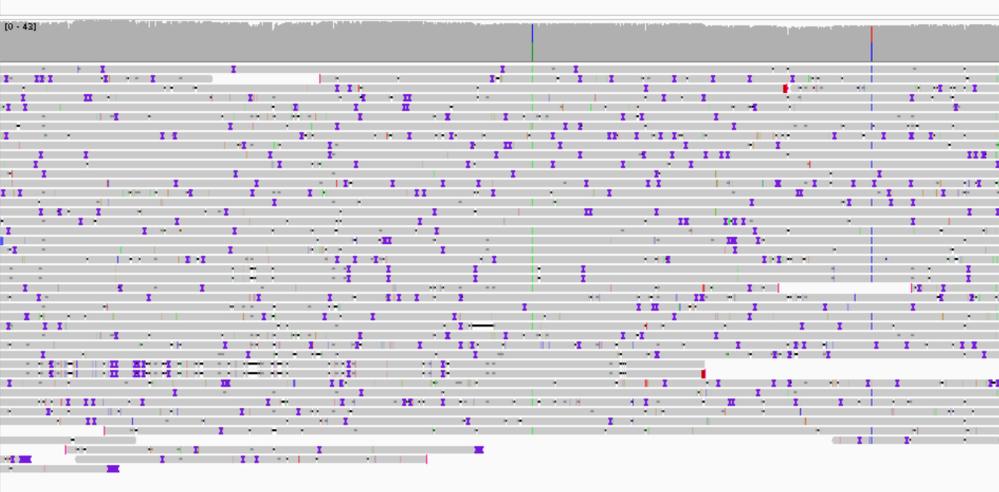
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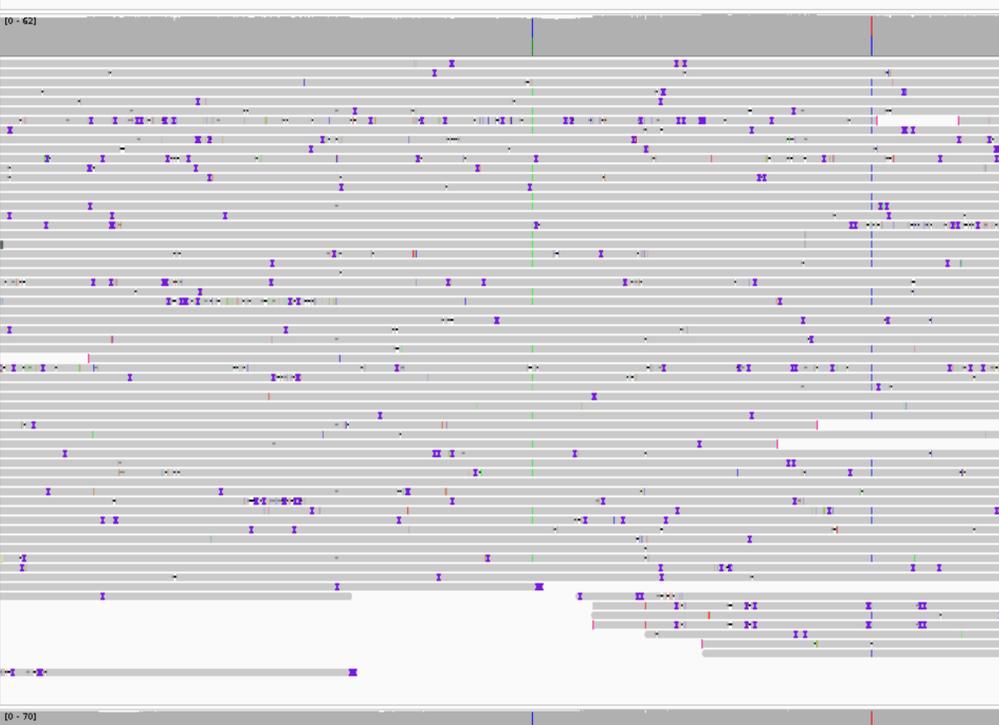
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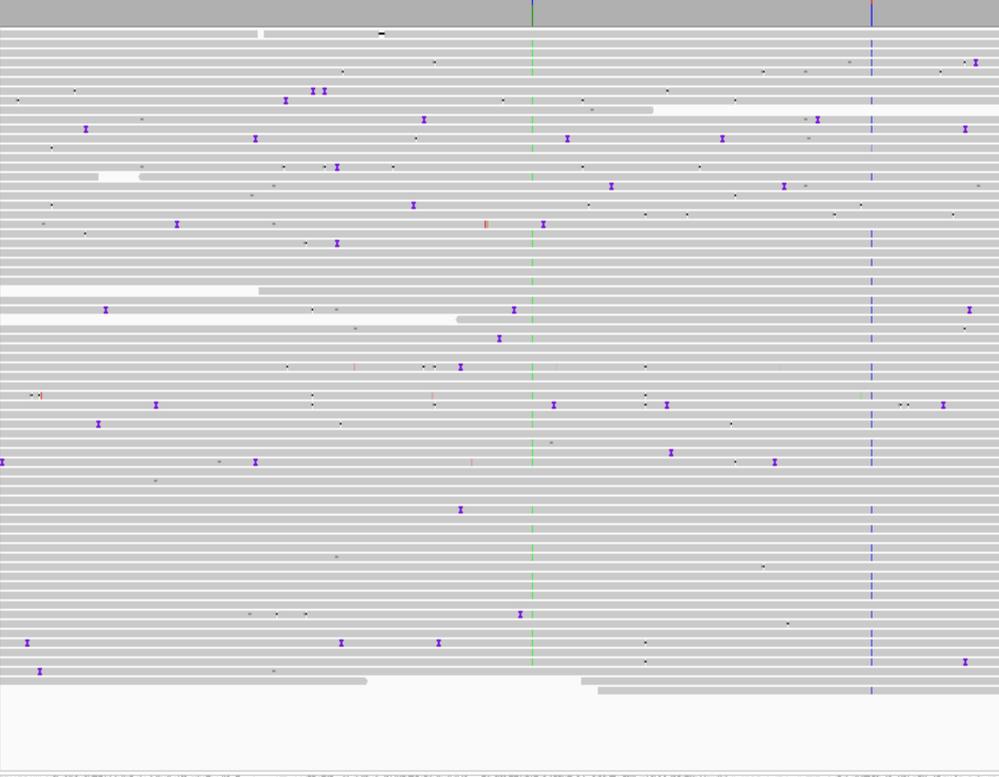
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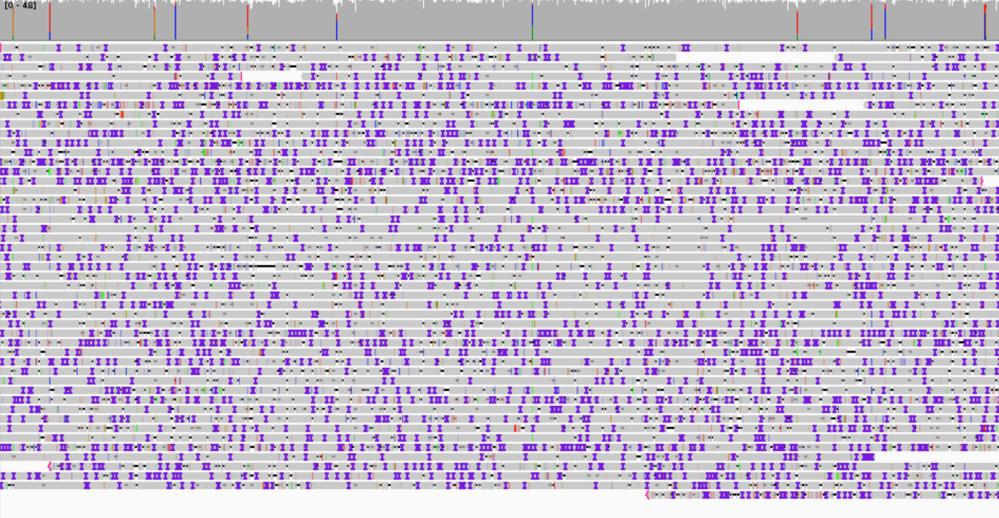
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colo829bl\_vai\_lifted.bam Coverage  
colo829bl\_vai\_lifted.bam

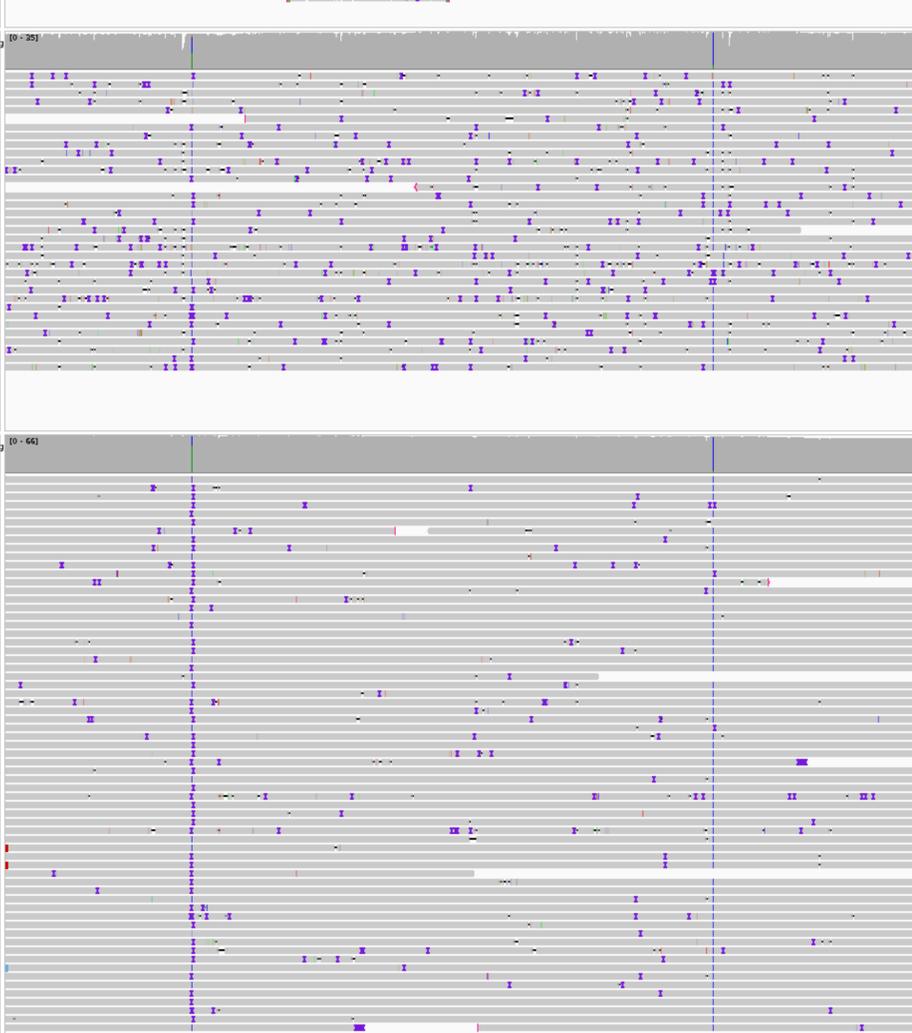
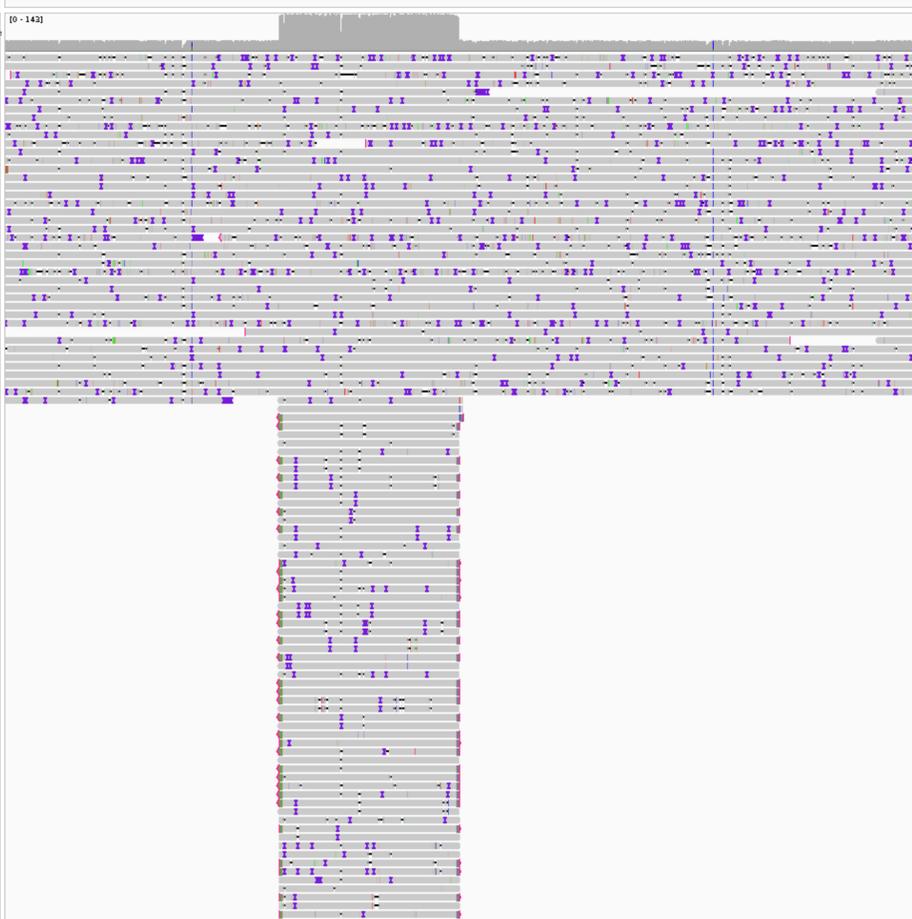


Sequence  
RefSeq Genes

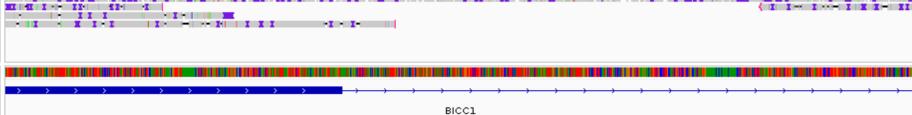


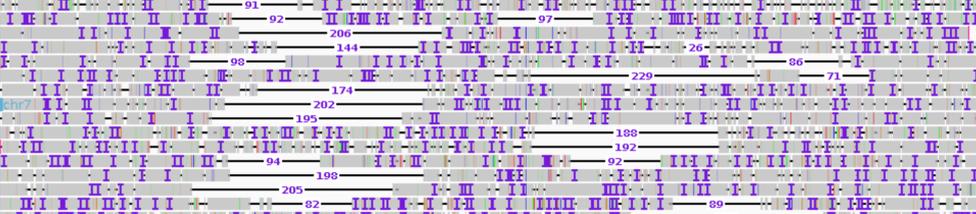
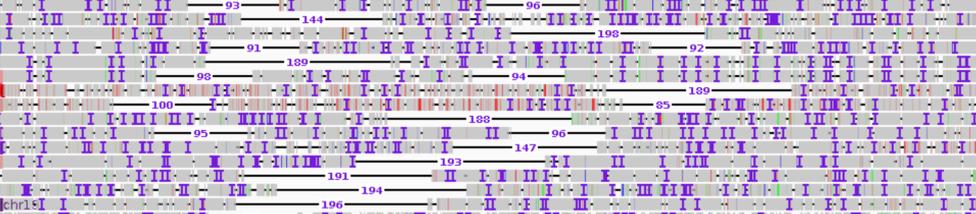
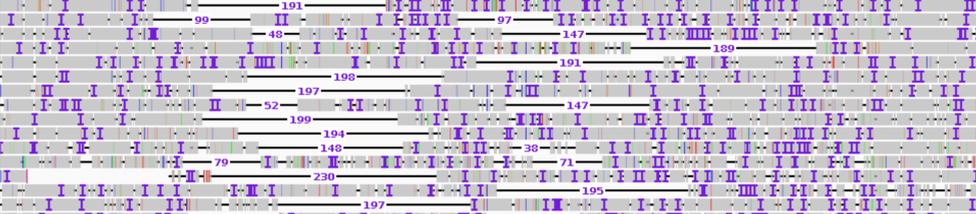
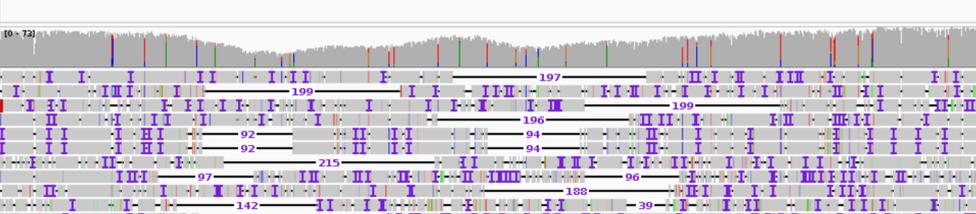
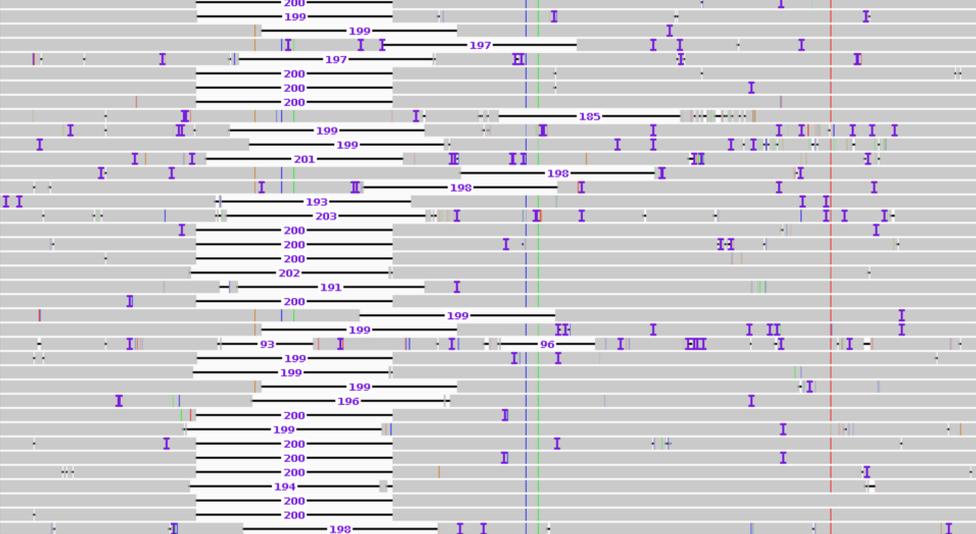
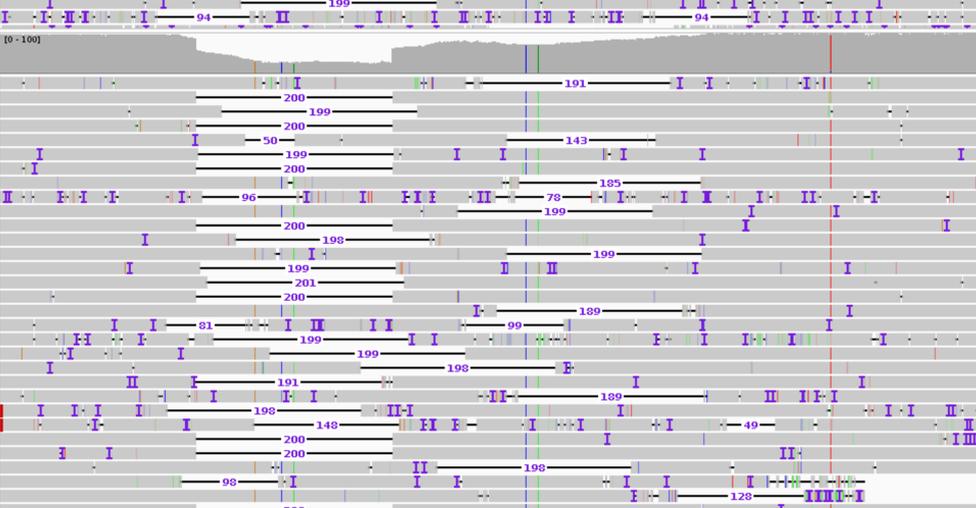
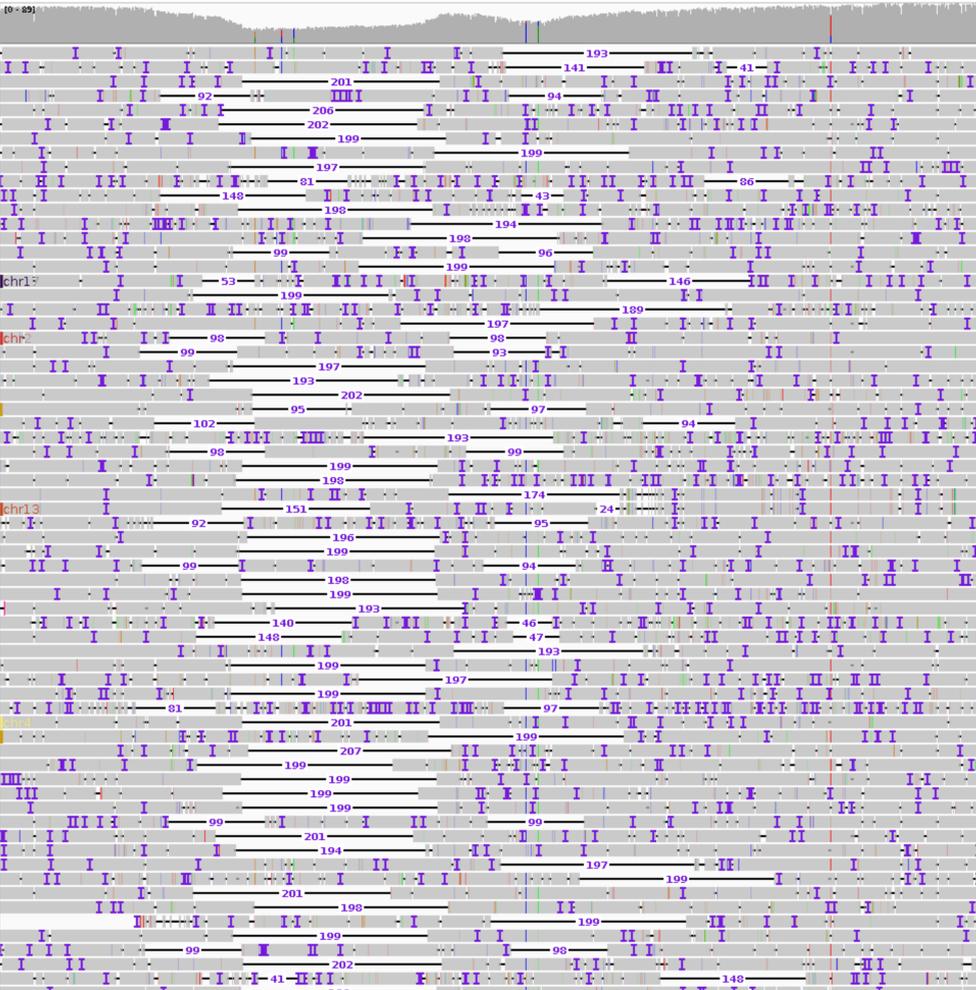


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colo829\_gsc\_lifted.bam  
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colo829\_ont\_lifted.bam  
colo829\_pbr\_lifted.bam Coverage  
0 - 42  
colo829\_pbr\_lifted.bam  
colo829\_val\_lifted.bam Coverage  
0 - 101  
colo829\_val\_lifted.bam  
colo829b\_gsc\_lifted.bam Coverage  
0 - 251  
colo829b\_gsc\_lifted.bam  
colo829b\_ont\_lifted.bam Coverage  
0 - 66  
colo829b\_ont\_lifted.bam  
colo829b\_pbr\_lifted.bam Coverage  
0 - 63  
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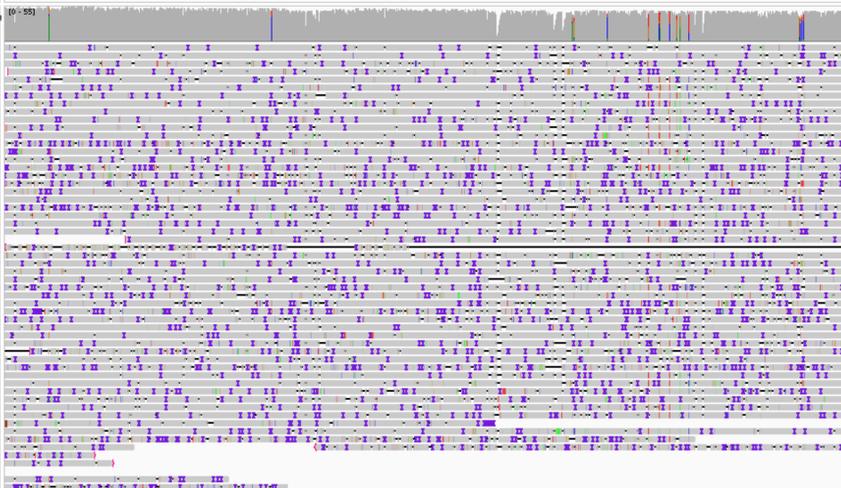
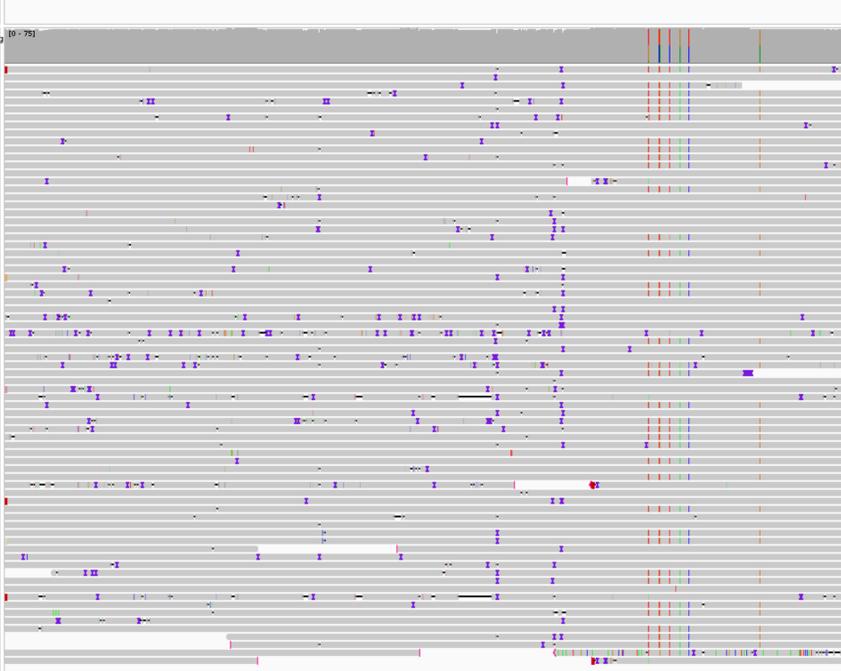
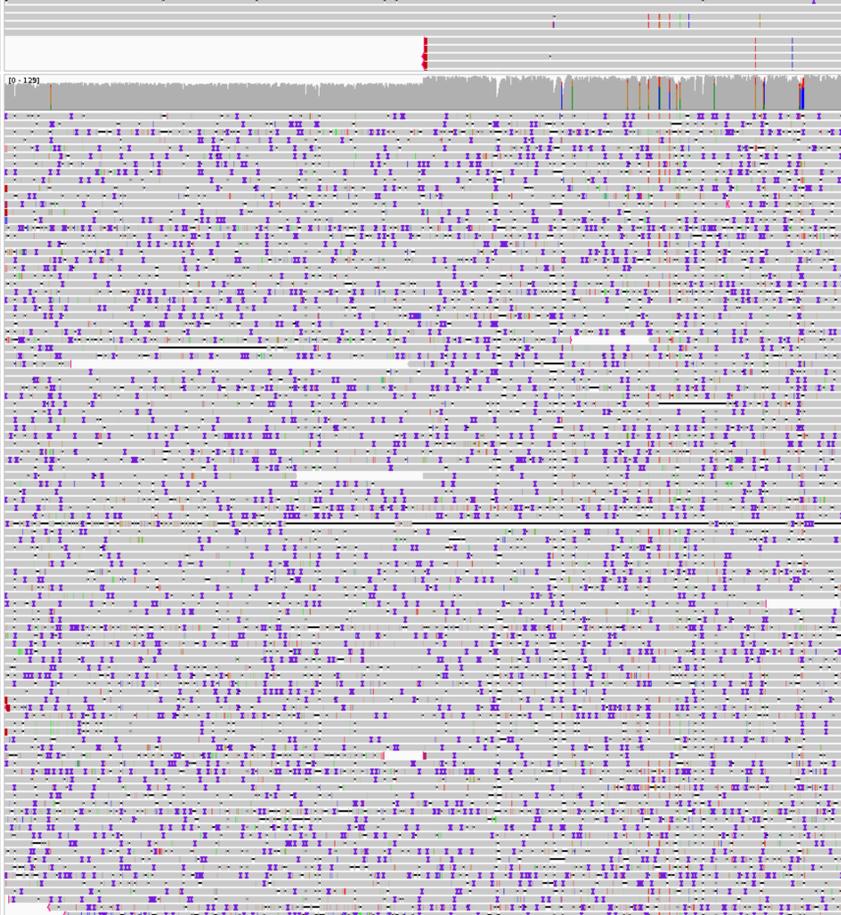
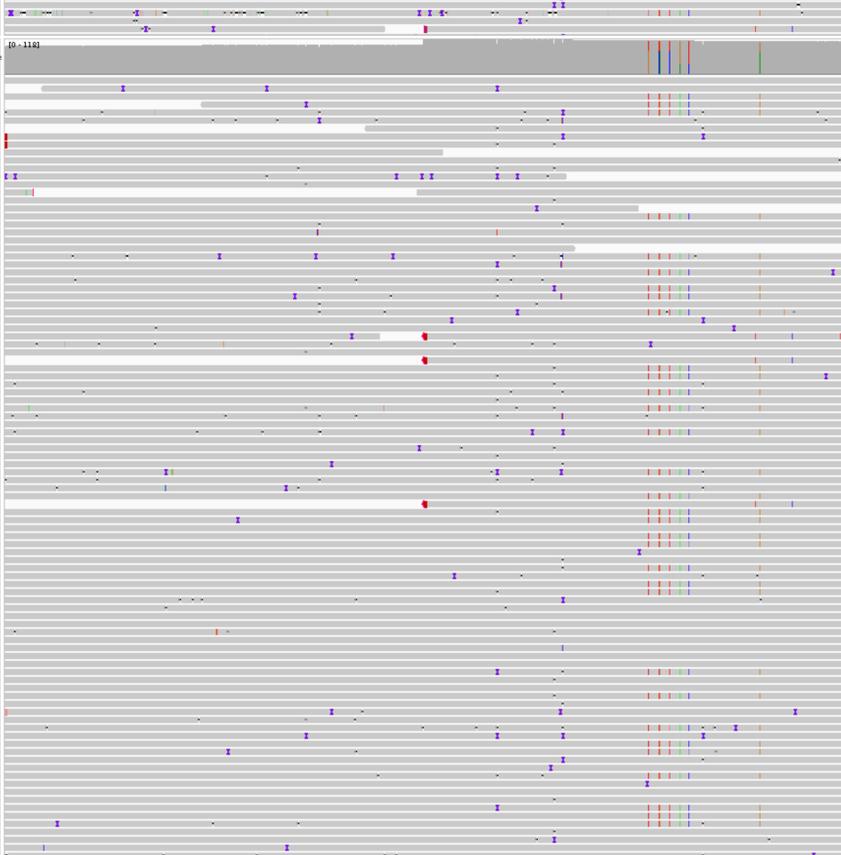
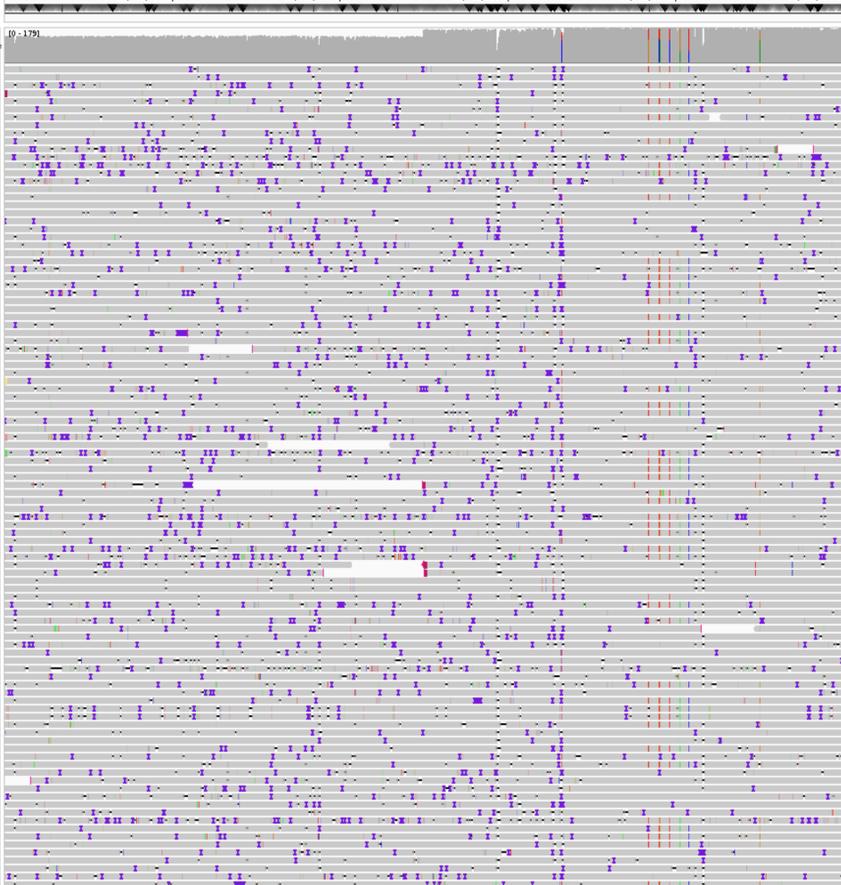


Sequence  
Refseq Genes





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colo829\_ort\_lifted.bam  
colo829\_pbr\_lifted.bam Coverage  
colo829\_pbr\_lifted.bam  
colo829\_val\_lifted.bam Coverage  
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colo829\_gsc\_lifted.bam Coverage  
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colo829\_val\_lifted.bam Coverage  
colo829\_val\_lifted.bam



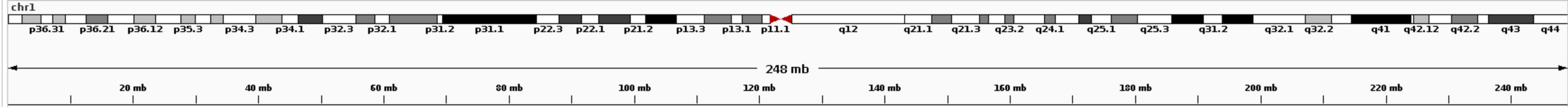
Sequence  
RefSeq Genes



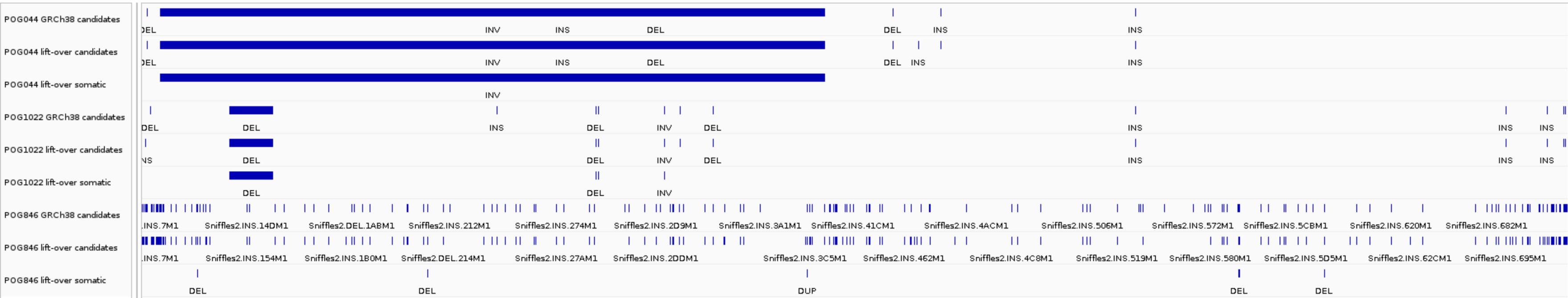
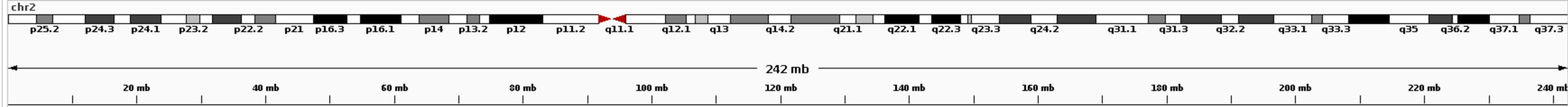
Supplementary Figure 7: IGV screenshot of the somatic candidate and somatic curated Structural Variants (SVs) for the POG samples POG044, POG1022 and POG846 in both the GRCh38 and lift-over analysis.

A-W) Chromosomes 1-22 and X. Rows showed are:

1. POG044 GRCh38 candidates
2. POG044 Lift-over candidates
3. POG044 Lift-over somatic
4. POG1022 GRCh38 candidates
5. POG1022 Lift-over candidates
6. POG1022 Lift-over somatic
7. POG846 GRCh38 candidates
8. POG846 Lift-over candidates
9. POG846 Lift-over somatic

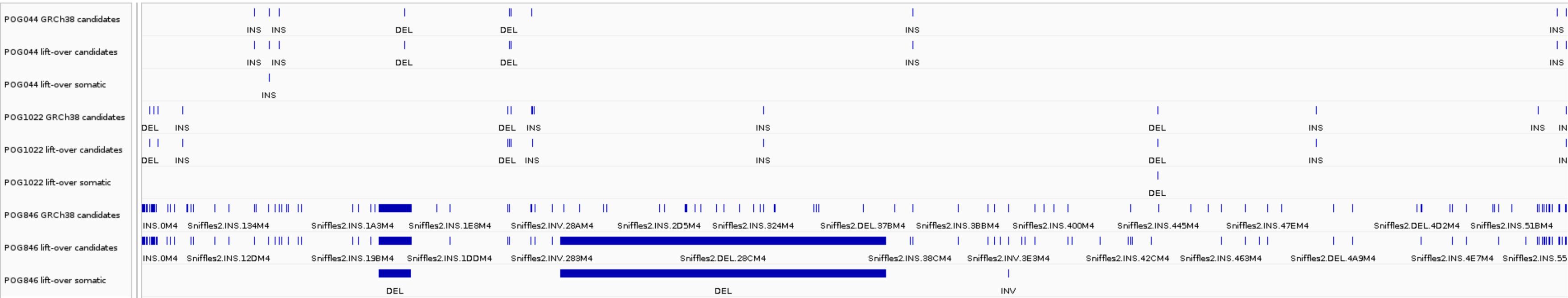
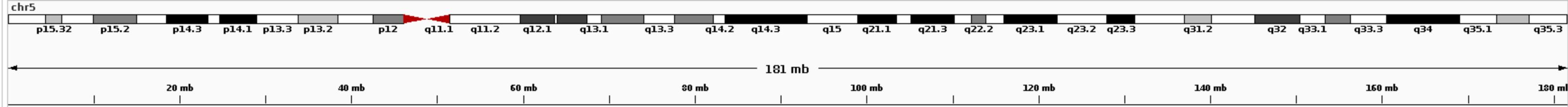


Category	Genomic Features
POG044 GRCh38 candidates	Vertical lines representing structural variants. Labels: NS, INS, INS, DEL, INS, INS, DEL, INS.
POG044 lift-over candidates	Vertical lines representing structural variants. Labels: NS, INS, INS, DEL, INS, INS, DEL, INS.
POG044 lift-over somatic	Vertical lines representing structural variants. Labels: N]chrX:2227331], DEL, DEL, DEL.
POG1022 GRCh38 candidates	Vertical lines representing structural variants. Labels: IS, INS, DEL, INS, DEL, DEL, INS, INS, INS, DEL, DEL.
POG1022 lift-over candidates	Vertical lines representing structural variants. Labels: IS, INS, DEL, INS, DEL, DEL, INS, INS, INS, DEL, DEL.
POG1022 lift-over somatic	Vertical lines representing structural variants. Labels: INS, DEL.
POG846 GRCh38 candidates	Vertical lines representing structural variants. Labels: INS.5M0, Sniffles2.INS.131M0, Sniffles2.INS.1D8M0, Sniffles2.INS.245M0, Sniffles2.INS.295M0, Sniffles2.INS.2F6M0, Sniffles2.INS.376M0, Sniffles2.INS.415M0, Sniffles2.INS.498M0, Sniffles2.DEL.4F8M0, Sniffles2.INS.568M0, Sniffles2.INS.5DFM0, Sniffles2.DEL.668M0.
POG846 lift-over candidates	Vertical lines representing structural variants. Labels: INS.5M0, Sniffles2.INS.137M0, Sniffles2.INS.1E2M0, Sniffles2.INS.24DM0, Sniffles2.INS.2B7M0, Sniffles2.INS.320M0, Sniffles2.INS.380M0, Sniffles2.INS.423M0, Sniffles2.INS.4A9M0, Sniffles2.DEL.507M0, Sniffles2.INS.570M0, Sniffles2.INS.5D6M0, Sniffles2.INS.649M0, Sniffles2.INS.
POG846 lift-over somatic	Vertical lines representing structural variants. Labels: DEL, DEL, DEL, DEL, DUP, DEL.



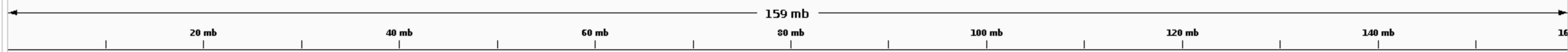






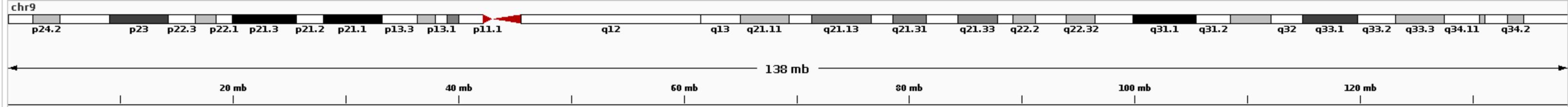


chr7



Category	Genomic Region	Variant Type
POG044 GRCh38 candidates	~15.5-15.8	INS
	~11.2-11.5	INS, INS
	~11.2-11.5	INS
POG044 lift-over candidates	~15.5-15.8	INS
	~11.2-11.5	INS, INS
	~11.2-11.5	INS
POG044 lift-over somatic	~15.5-15.8	INS
POG1022 GRCh38 candidates	~11.2-11.5	DEL
	~21.1-21.2	DEL
	~31.1-31.2	INS
POG1022 lift-over candidates	~11.2-11.5	DEL
	~21.1-21.2	DEL
	~31.1-31.2	INS
POG1022 lift-over somatic	~21.1-21.2	DEL
POG846 GRCh38 candidates	~15.5-15.8	INS.DM6
	~11.2-11.5	Sniffles2.INV.D2M6
	~31.1-31.2	Sniffles2.INV.D9M6
POG846 lift-over candidates	~15.5-15.8	INS.DM6
	~11.2-11.5	Sniffles2.INV.D9M6
	~31.1-31.2	Sniffles2.INV.D9M6
POG846 lift-over somatic	~15.5-15.8	INV
	~31.1-31.2	DEL





Category	Genomic Region	Variant Type
POG044 GRCh38 candidates	20-25 Mb	DEL, BND
	30-35 Mb	DUP, DEL
POG044 lift-over candidates	20-25 Mb	DEL, INS
	30-35 Mb	DUP, DEL
POG044 lift-over somatic	20-25 Mb	DEL, INS
	30-35 Mb	DUP
POG1022 GRCh38 candidates	20-25 Mb	INS, DEL
	30-35 Mb	INS
POG1022 lift-over candidates	20-25 Mb	INS, DEL
	30-35 Mb	INS
POG1022 lift-over somatic	20-25 Mb	DEL
	30-35 Mb	DEL
POG846 GRCh38 candidates	20-25 Mb	Sniffles2.INS.24M8, Sniffles2.INS.62M8, Sniffles2.DEL.88M8, Sniffles2.INS.BDM8
	30-35 Mb	Sniffles2.INS.16DM8, Sniffles2.INS.211M8, Sniffles2.INS.25AM8
POG846 lift-over candidates	20-25 Mb	2.INS.AM8, Sniffles2.INS.62M8, Sniffles2.DEL.89M8, Sniffles2.INS.F5M8
	30-35 Mb	Sniffles2.INS.172M8, Sniffles2.INS.224M8, Sniffles2.INS.265M8, Sniffles2.INS.2C7M8, Sniffles2.INS.2F6M8, Sniffles2.INS.33FM8, Sniffles2.INS.382M8
POG846 lift-over somatic	20-25 Mb	DEL
	30-35 Mb	DEL

BND/TRA\_N[chr11:64489844[

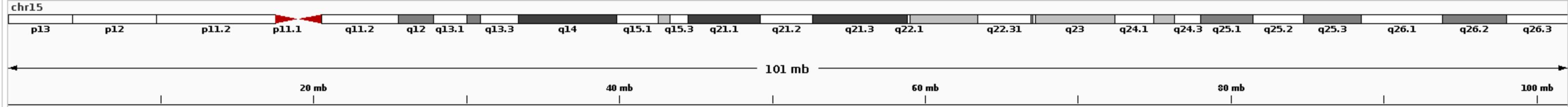




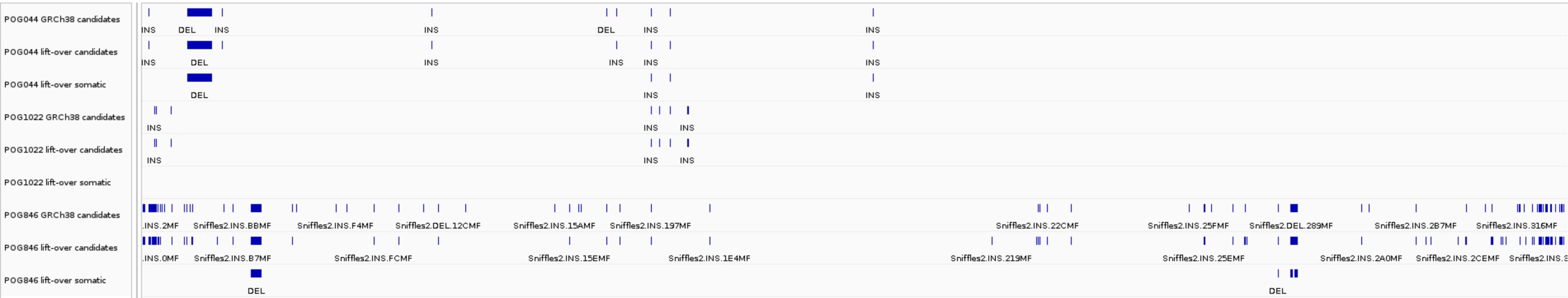
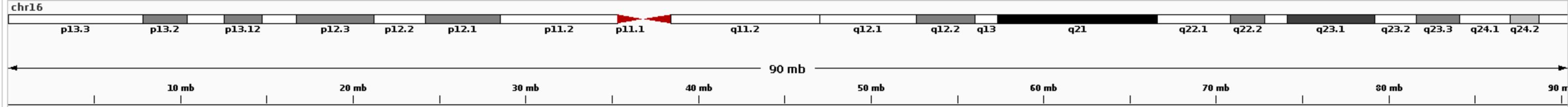








Category	Genomic Region	Variant Type
POG044 GRCh38 candidates	p11.1	INS
	p11.1	DEL
	q13.3	INS
POG044 lift-over candidates	p11.1	INS
	p11.1	DEL
	q13.3	INS
POG044 lift-over somatic	p11.1	DEL
POG1022 GRCh38 candidates	q11.2	INS
	q14	DEL
	q25.1	INS
POG1022 lift-over candidates	q11.2	INS
	q14	DEL
	q25.1	INS
POG1022 lift-over somatic	q25.1	DEL
POG846 GRCh38 candidates	p11.1	Sniffles2.INS.11ME
	q11.2	Sniffles2.INS.C1ME
POG846 lift-over candidates	p11.1	Sniffles2.INS.10ME
	q11.2	Sniffles2.INS.B3ME
POG846 lift-over somatic	q22.31	DEL











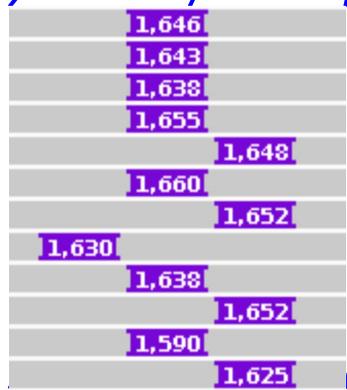
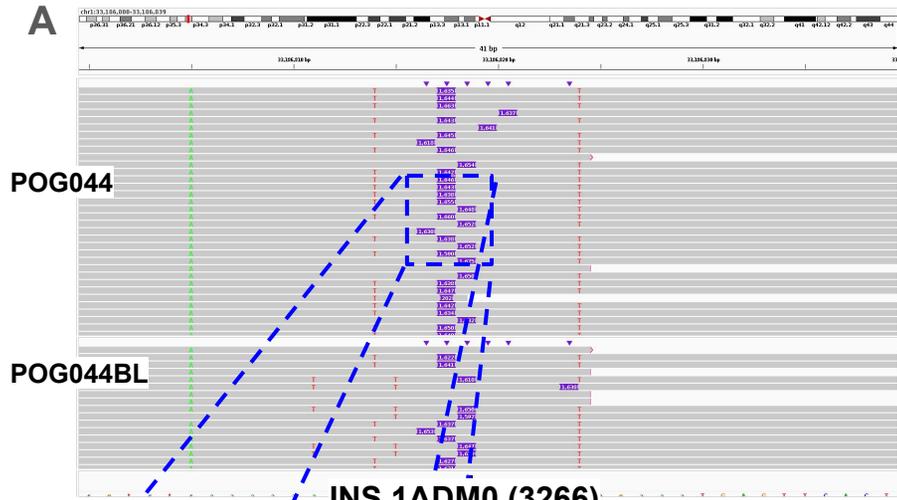






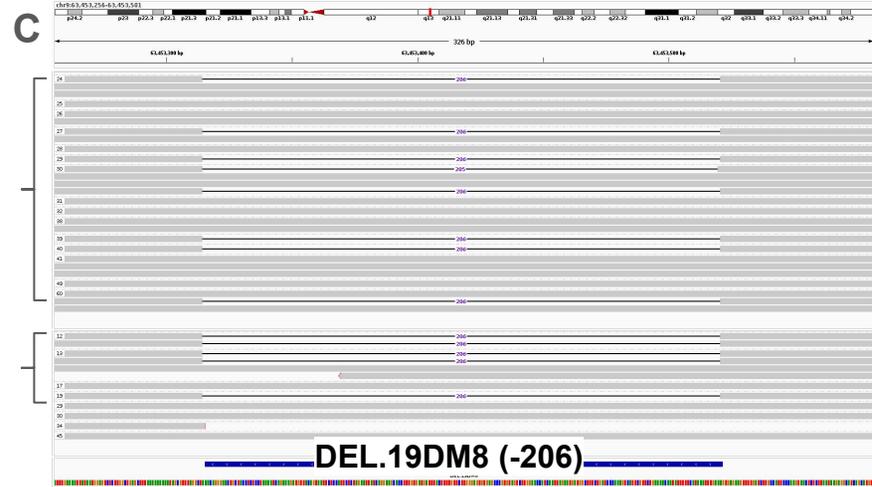
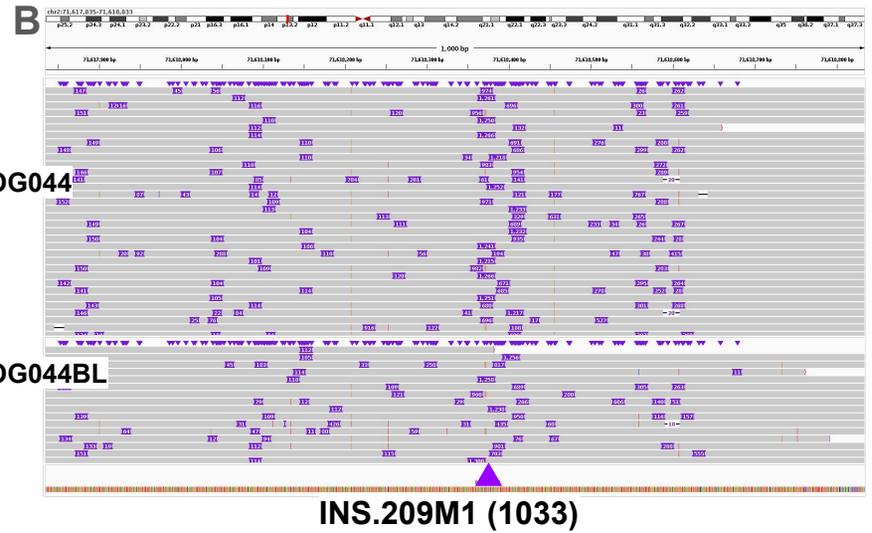
### **Supplement figure 8**

IGV screenshot of predicted somatic events that are wrongly labeled in sample POG044. Closer inspection shows support in the control samples. A) collapsed insertion in the tumor sample which causes a difference in size between tumor and normal. B) Insertion detected in a low complexity region, alongside multiple SVs of the same type. C) SV not detected in the control samples filtered by read quality.



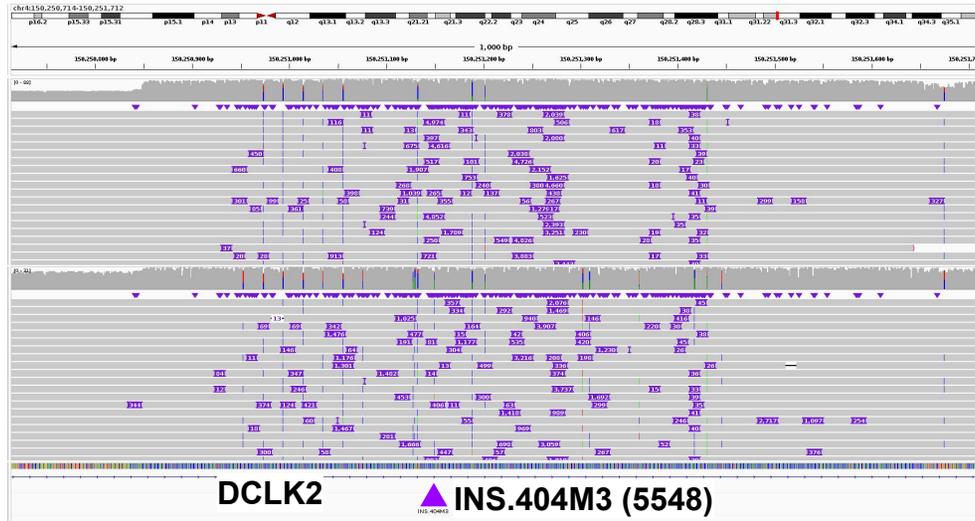
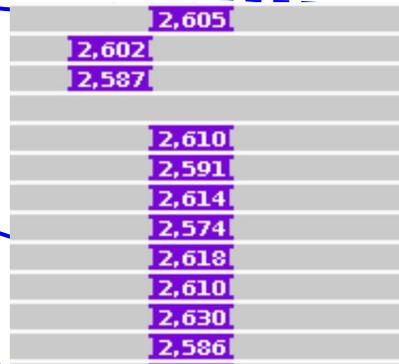
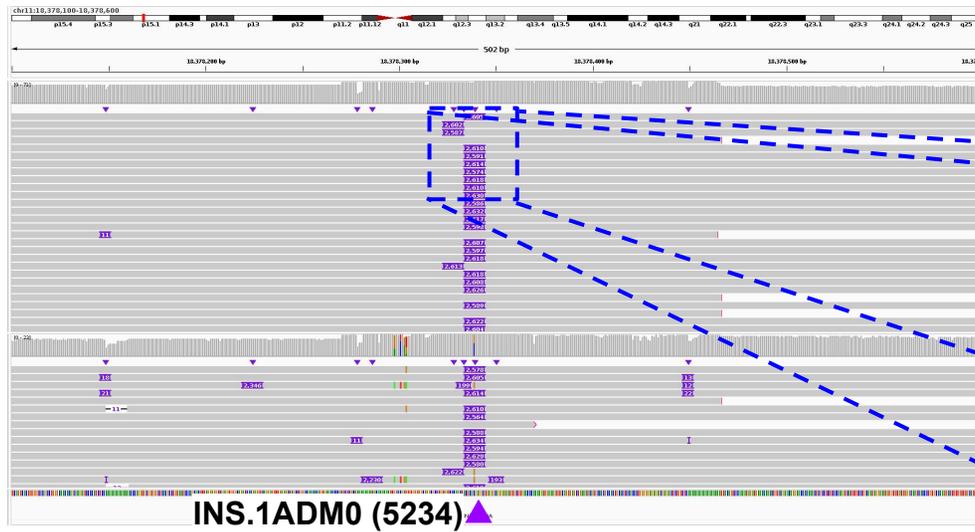
POG044  
MQ (24-60)

POG044BL  
MQ (12-19)



### **Supplement figure 9**

IGV screenshot of predicted somatic events that are wrongly labeled in sample POG1022. Closer inspection shows support in the control samples. A) Insertion detected in a low complexity region, alongside multiple SVs of the same type. B) collapsed insertion in the tumor sample which causes a difference in size between tumor and normal.

**A****B**

### **Supplement figure 10**

IGV screenshot of a predicted non-synonymous deletion on chr1 of POG044 which is overlapping the gene NBPF20. This region is affected by several deletion events.

chr1:145,317,425-145,392,444

p36.31 p36.21 p36.12 p35.3 p34.3 p34.1 p32.3 p32.1 p31.2 p31.1 p22.3 p22.1 p21.2 p13.3 p13.1 p11.1 q12 q21.1 q21.3 q23.2 q24.1 q25.1 q25.3 q31.2 q32.1 q32.2 q41 q42.12 q42.2

145,320 kb 145,330 kb 145,340 kb 145,350 kb 145,360 kb 145,370 kb 145,380 kb

75 kb

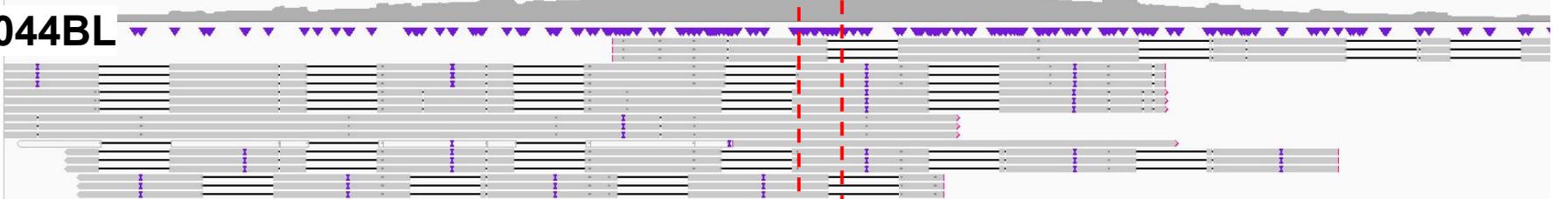
[0 - 2705]

**POG044**



[0 - 682]

**POG044BL**



**NBPF20**

NBPF20

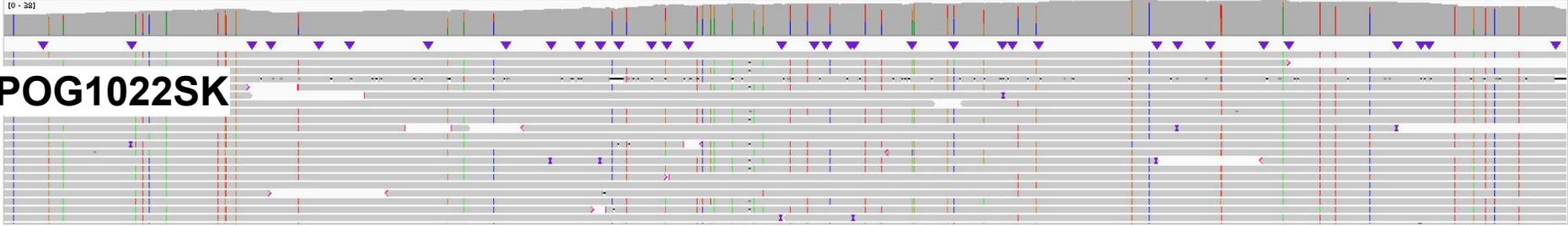
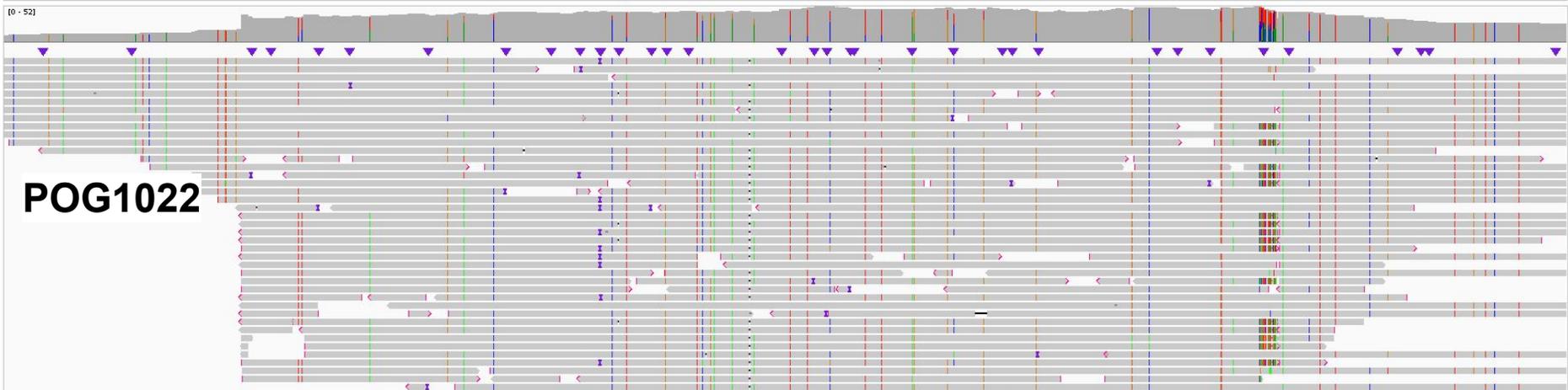
**DEL.3C4M0**

**Supplement figure 11**

IGV screenshot of a predicted non-synonymous SV (inversion) on chr2 of POG1022 which is overlapping the gene IGKV3-11.

chr2:88,856,365-88,892,855  
p25.2 p24.3 p24.1 p23.2 p22.2 p21 p16.3 p16.1 p14 p13.2 p12 p11.2 q11.1 q12.1 q13 q14.2 q21.1 q22.1 q22.3 q23.3 q24.2 q31.1 q31.3 q32.2 q33.1 q33.3 q35 q36.2 q37.1 q37.3

36 kb  
88,860 kb 88,870 kb 88,880 kb 88,890 kb

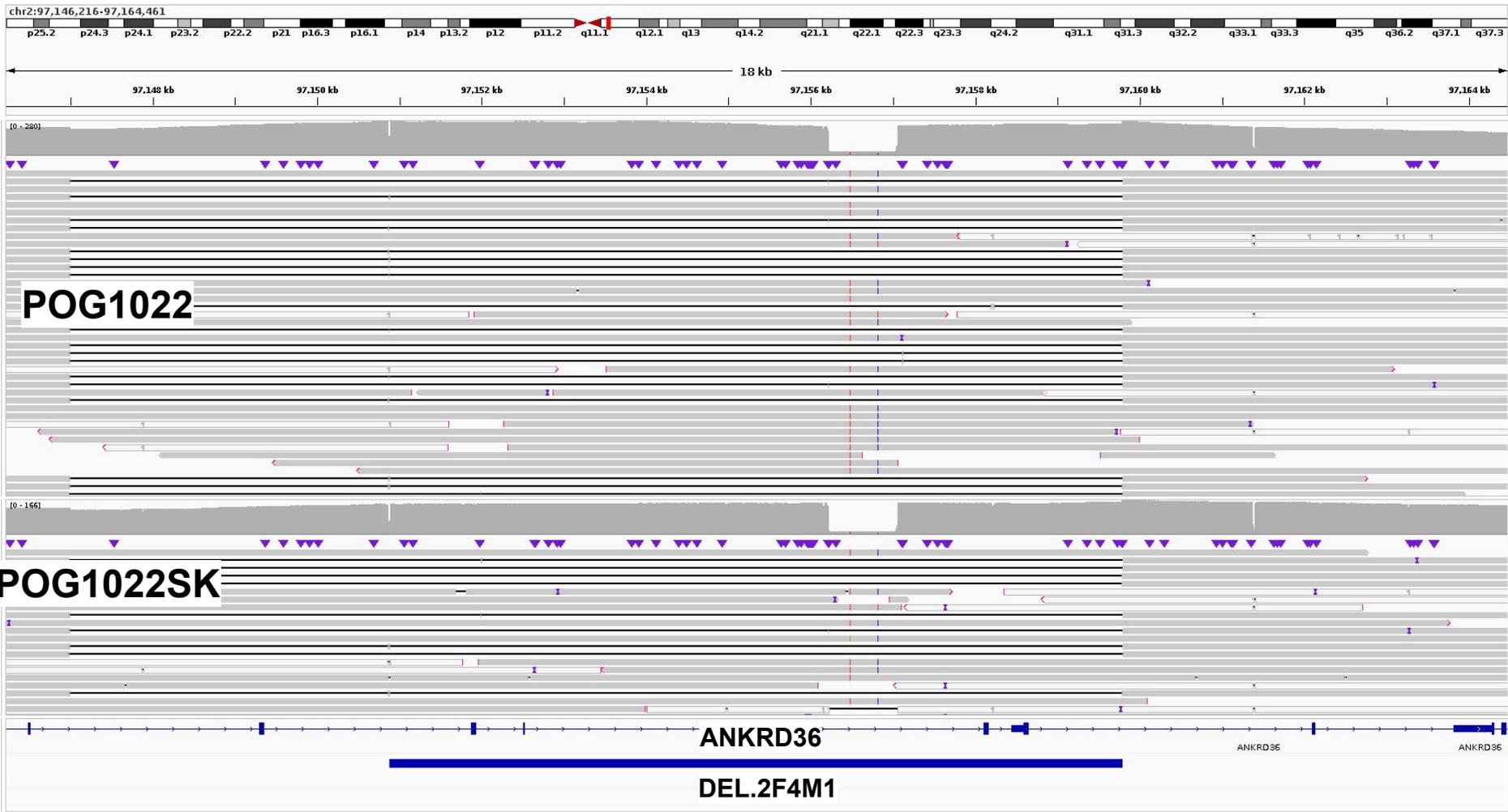


IGKV3-11

INV.299M1

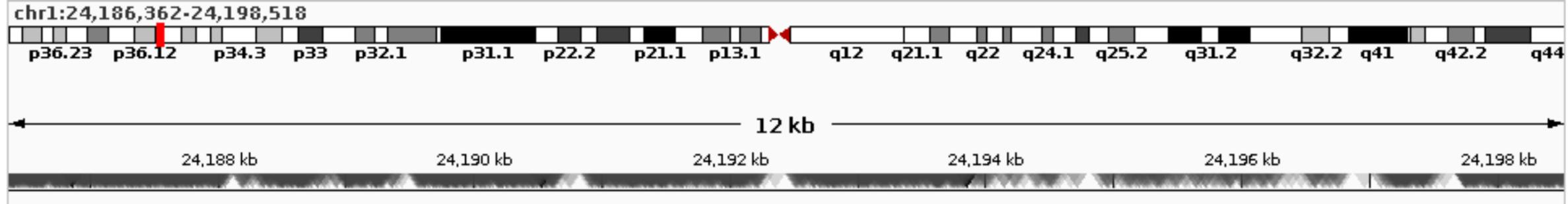
**Supplement figure 12**

IGV screenshot of a predicted non-synonymous SV (deletion) on chr2 of POG1022 which is overlapping the gene ANKRD36. Closer inspection shows a larger deletion in the same region (12kb) with some reads supporting the SV in the control.

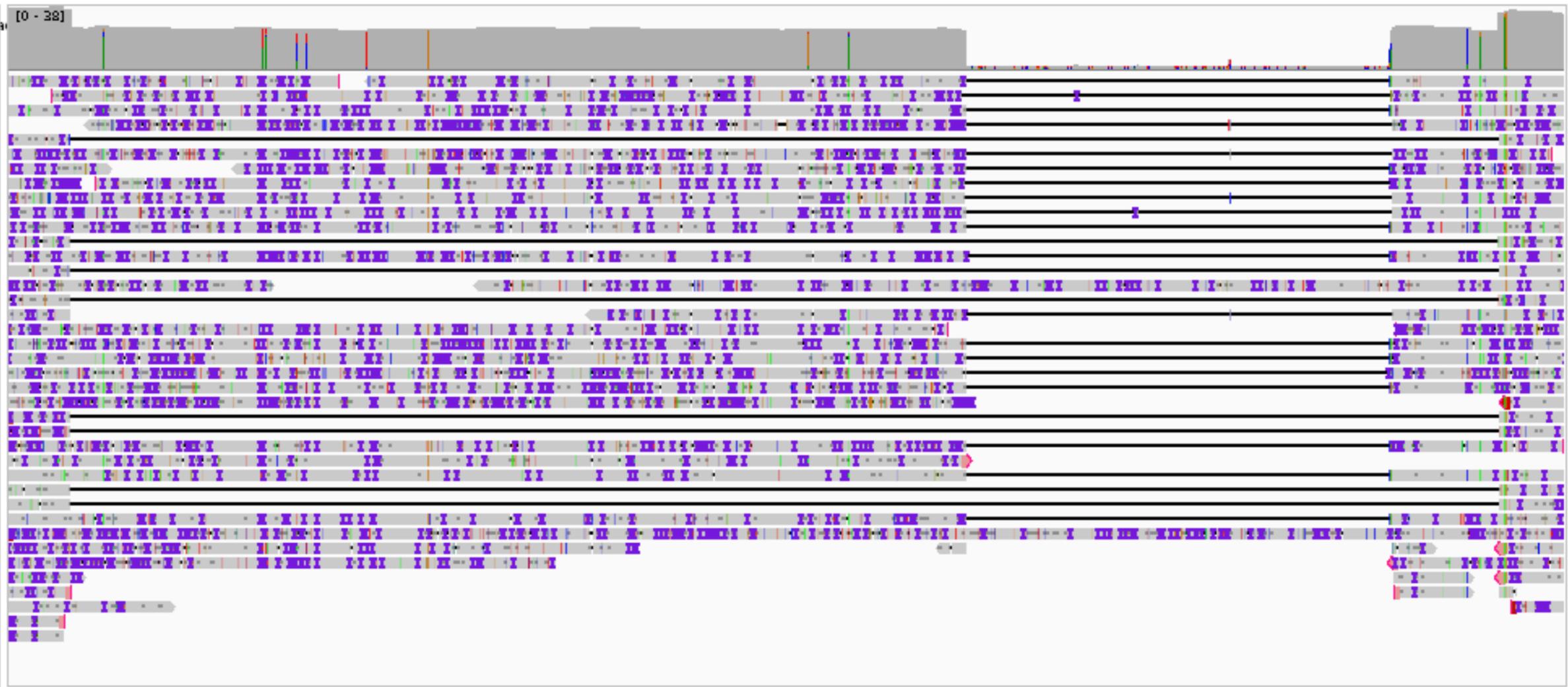


Supplementary Figure 13: IGV screenshot of the Structural Variants (SVs) affecting genes in the POG846 sample. For detail see Supplementary Table 16

- A) A 11,156 deletion affecting *IFNLR1*
- B) A 7,600 deletion affecting *RAB11A*
- C) A 1,645 deletion affecting *CBFA2T2*
- D) A 851 deletion affecting *BOD1L1*



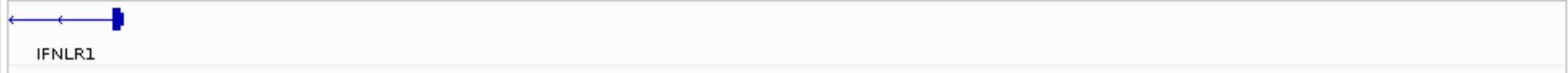
pog846\_tumor\_lifted.bam Coverage

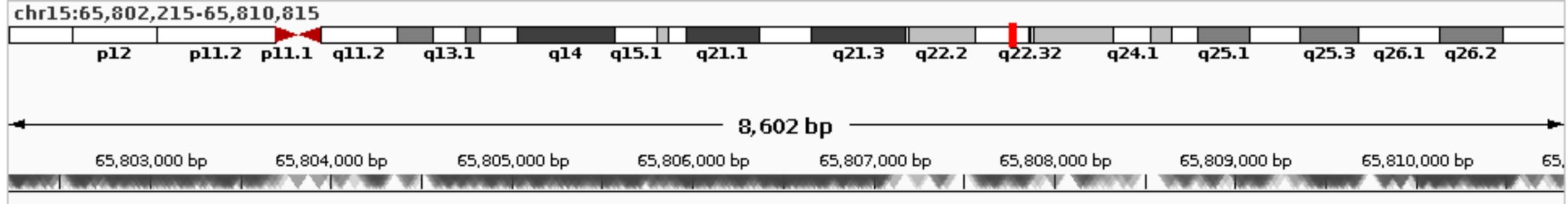


pog846\_control\_lifted.bam Coverage

pog846\_control\_lifted.bam

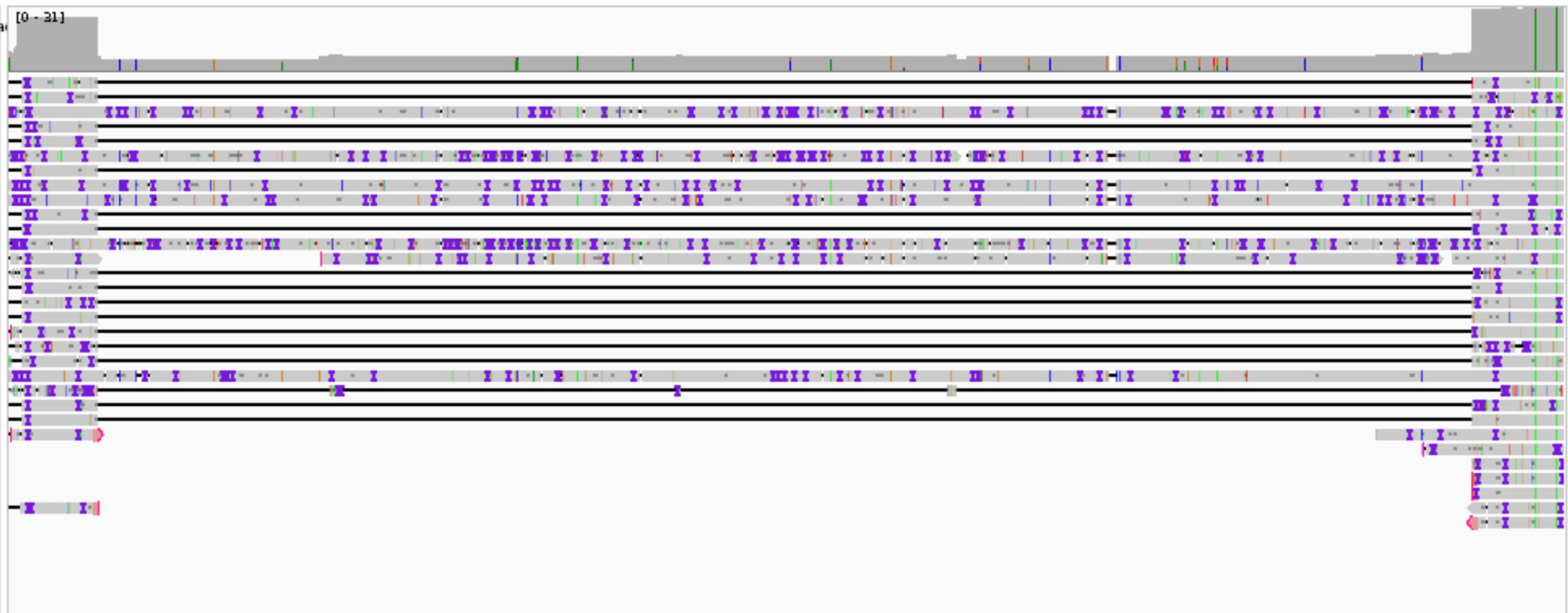
Refseq Genes





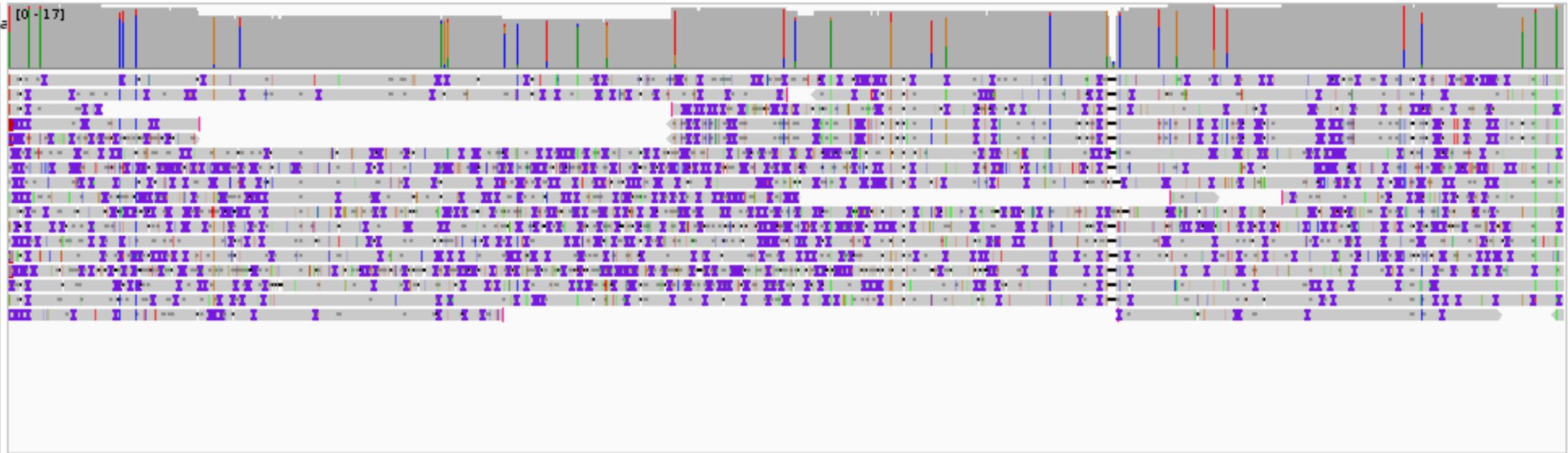
pog846\_tumor\_lifted.bam Coverage

pog846\_tumor\_lifted.bam

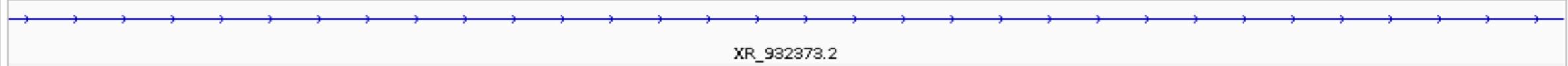


pog846\_control\_lifted.bam Coverage

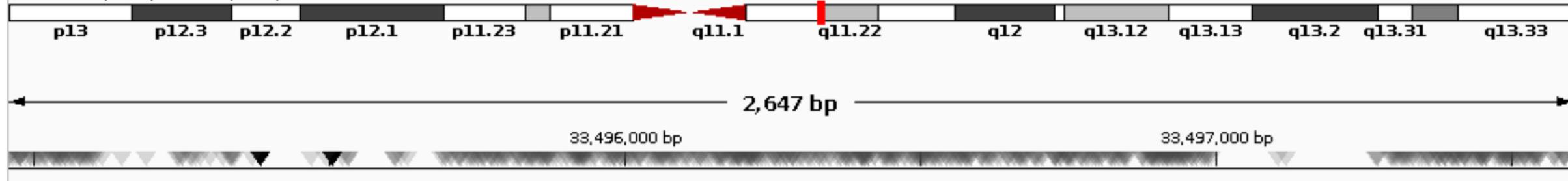
pog846\_control\_lifted.bam



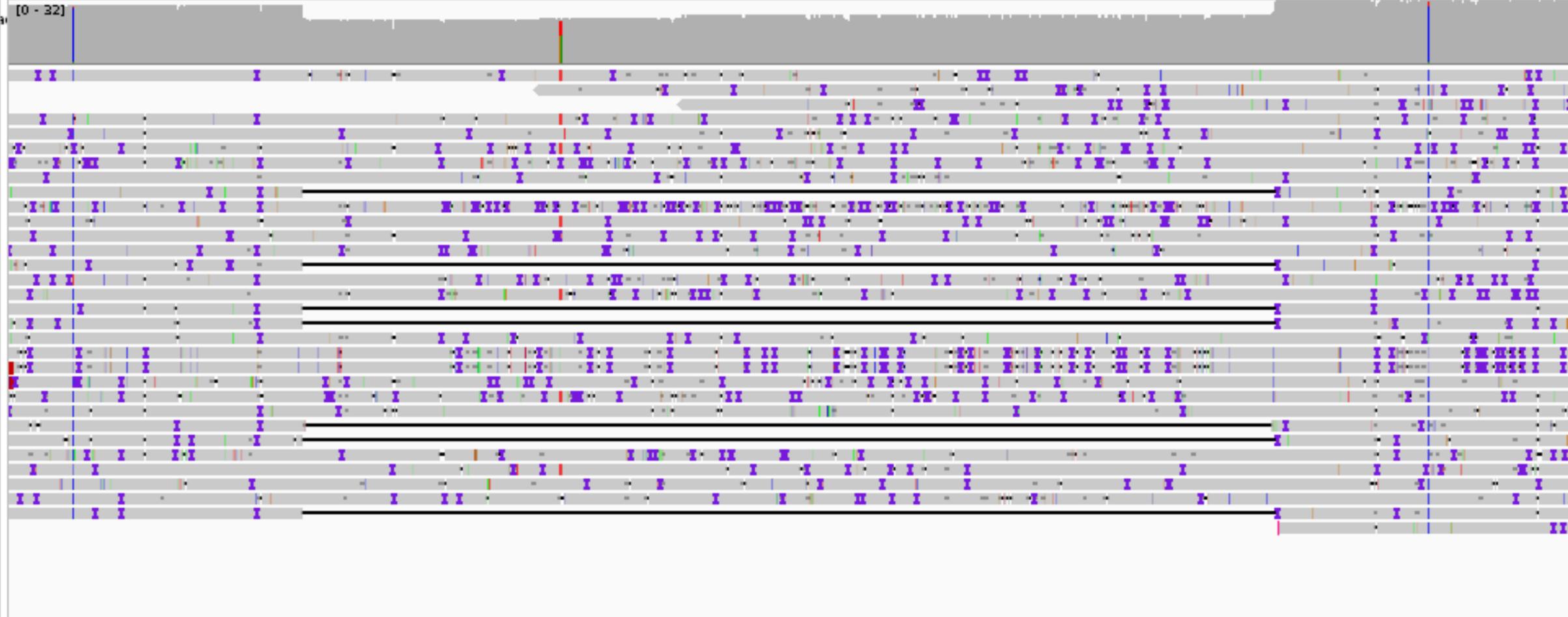
Refseq Genes



chr20:33,494,959-33,497,604

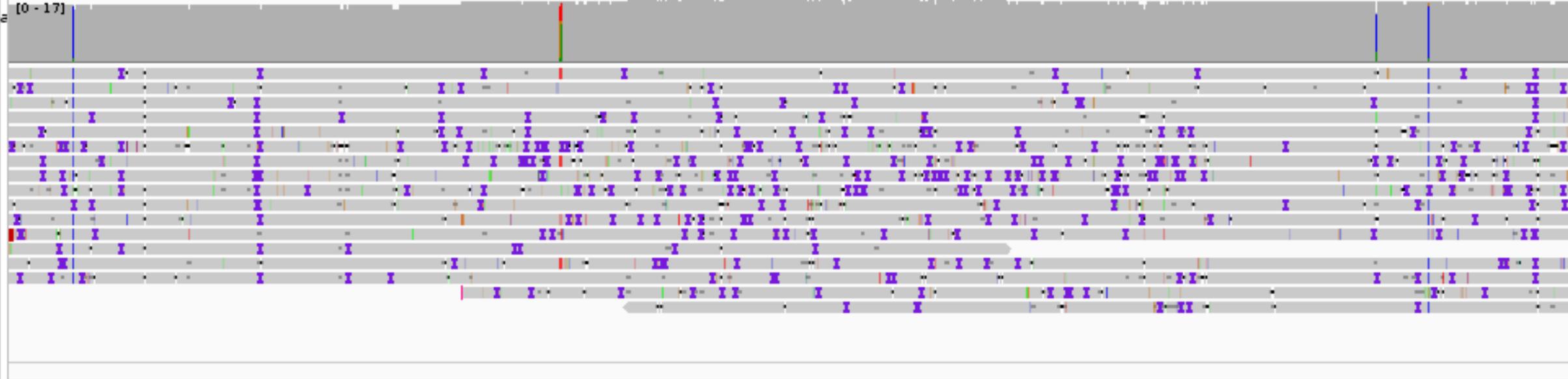


pog846\_tumor\_lifted.bam Coverage



pog846\_tumor\_lifted.bam

pog846\_control\_lifted.bam Coverage

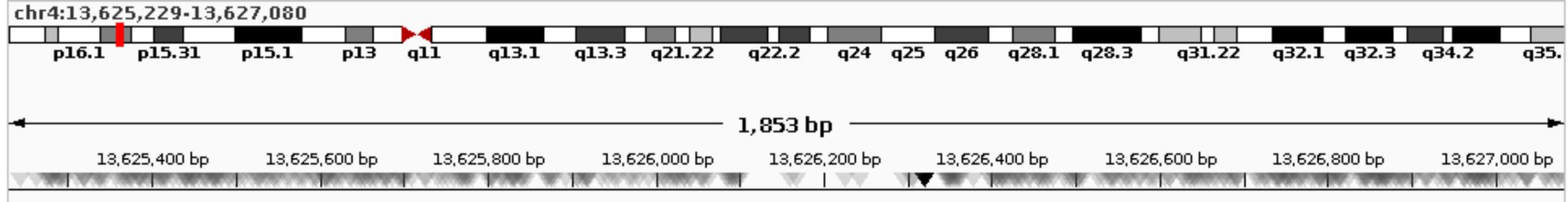


pog846\_control\_lifted.bam

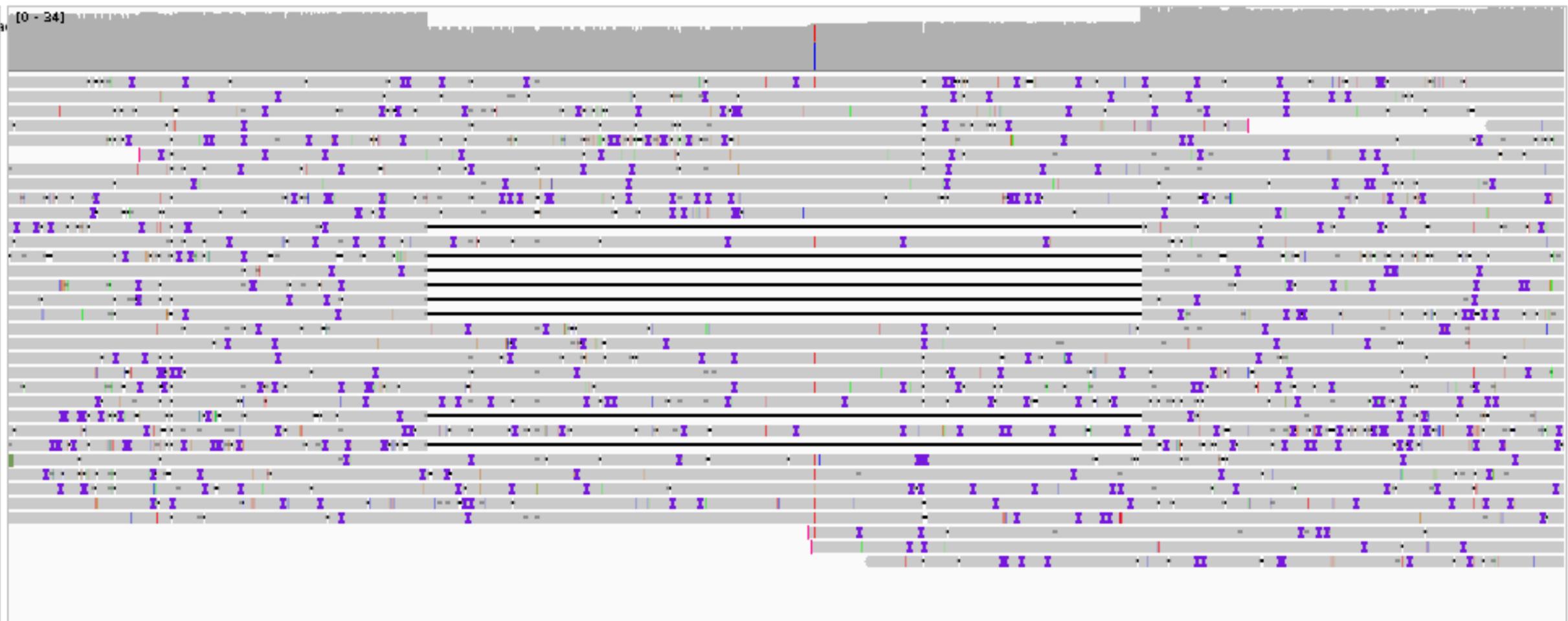
Refseq Genes



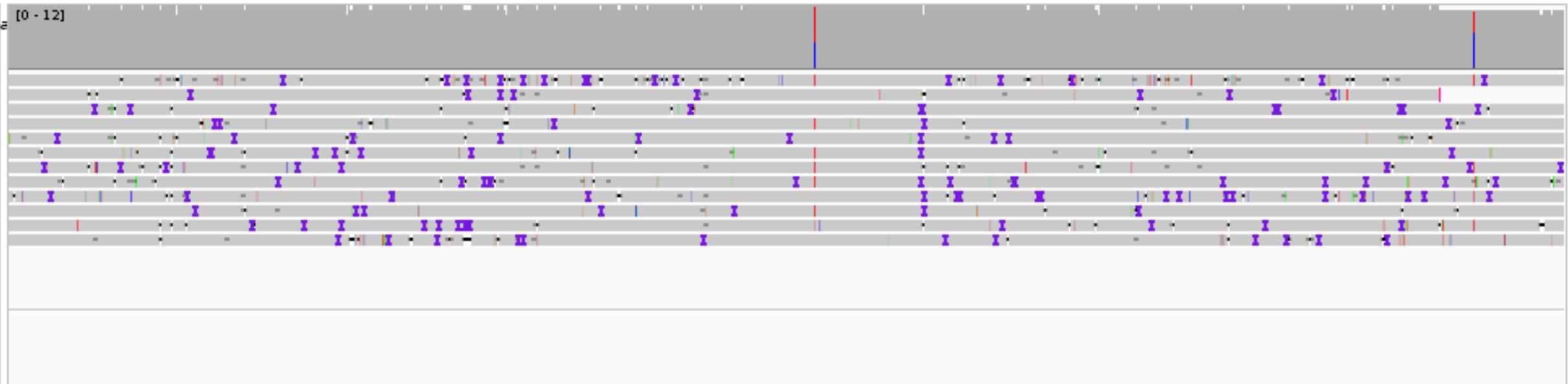
CBFA2T2



pog846\_tumor\_lifted.bam Coverage  
pog846\_tumor\_lifted.bam



pog846\_control\_lifted.bam Coverage  
pog846\_control\_lifted.bam



Sequence  
Refseq Genes



BOD1L1

### **Supplement figure 14**

Example of instability of the COLO829 cancer cell-lines. An inversion detected in three out of four replicates with noticeable duplication (not detected by SV callers) in all four

