

# NGS data visualisation on IGV

Erika Souche



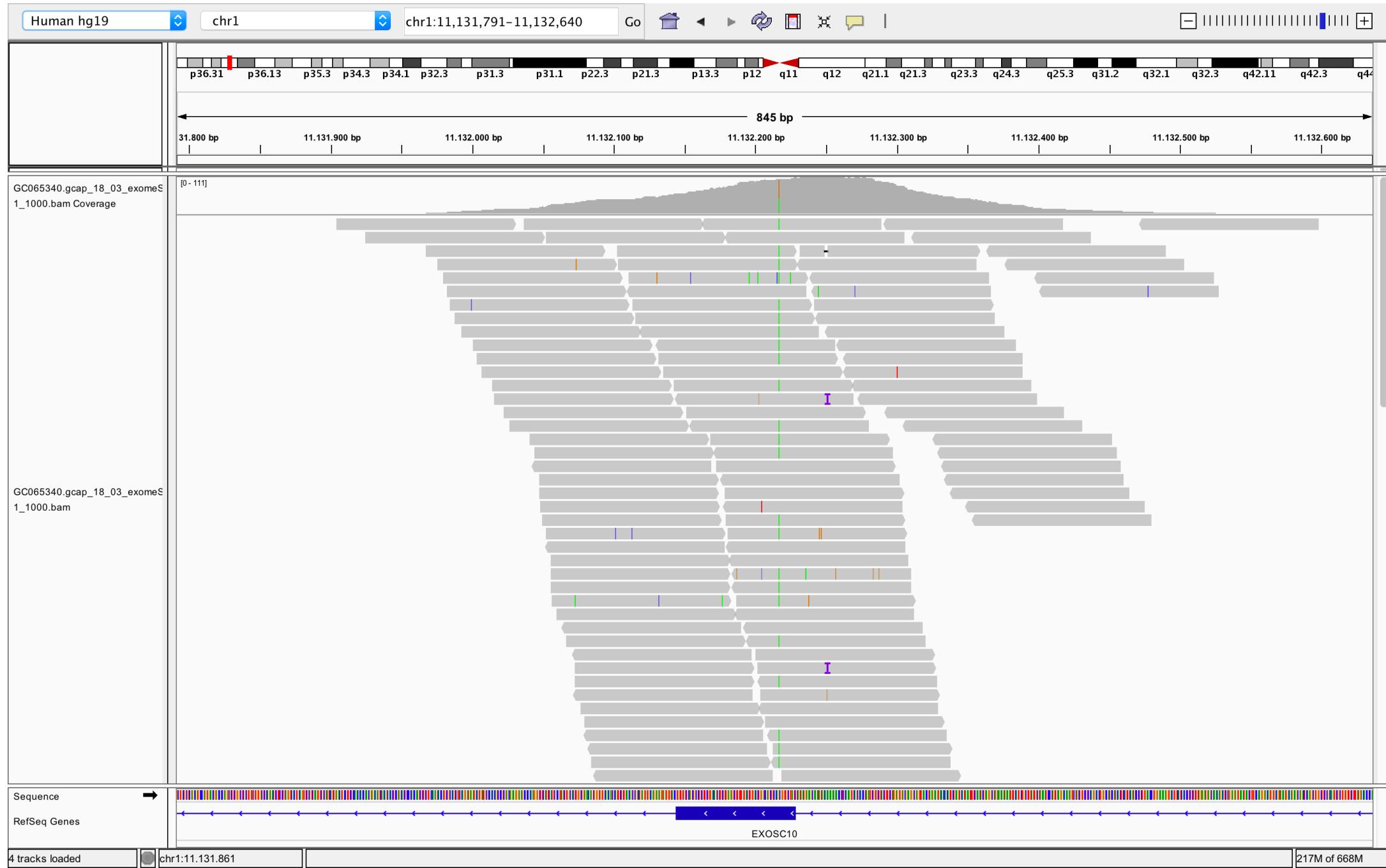
# Outline

- Same variant in various assays
- Looking at the data
- Variant analysis
- Coverage analysis

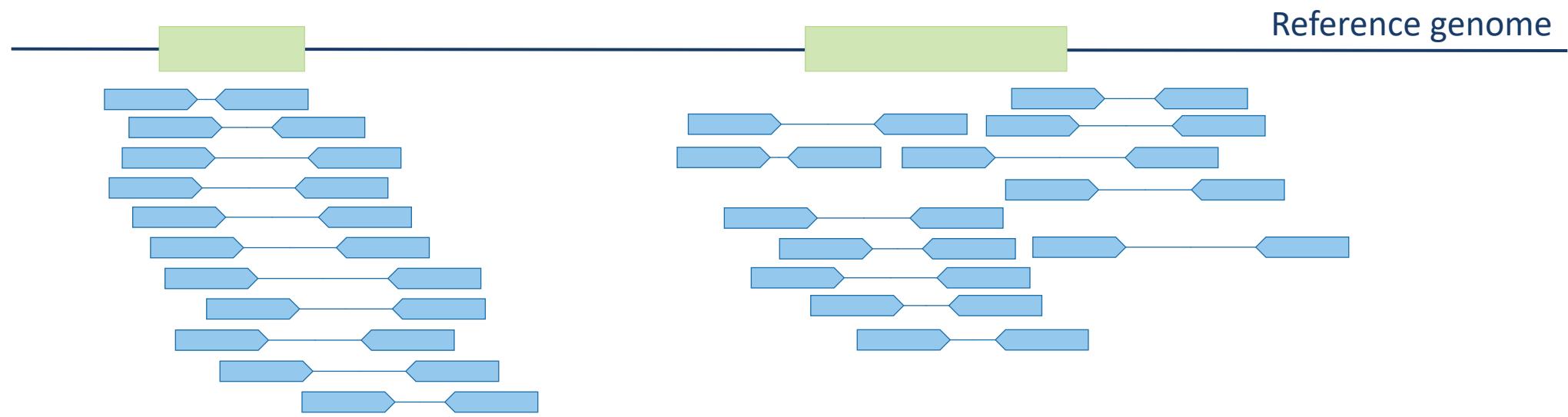
\*Focus on Illumina short read sequencing

# Outline

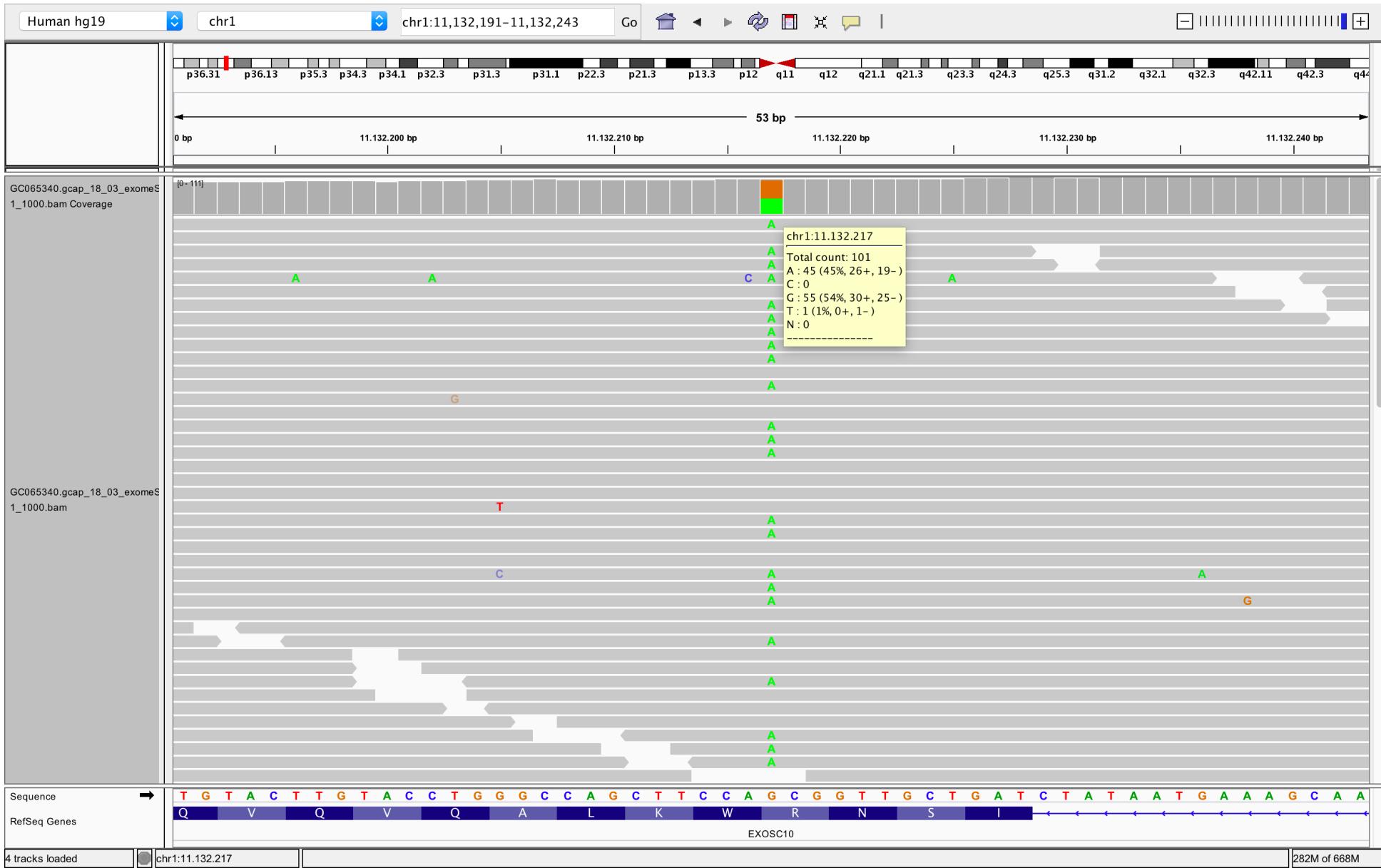
- Same variant in various assays
- Looking at the data
- Variant analysis
- Coverage analysis

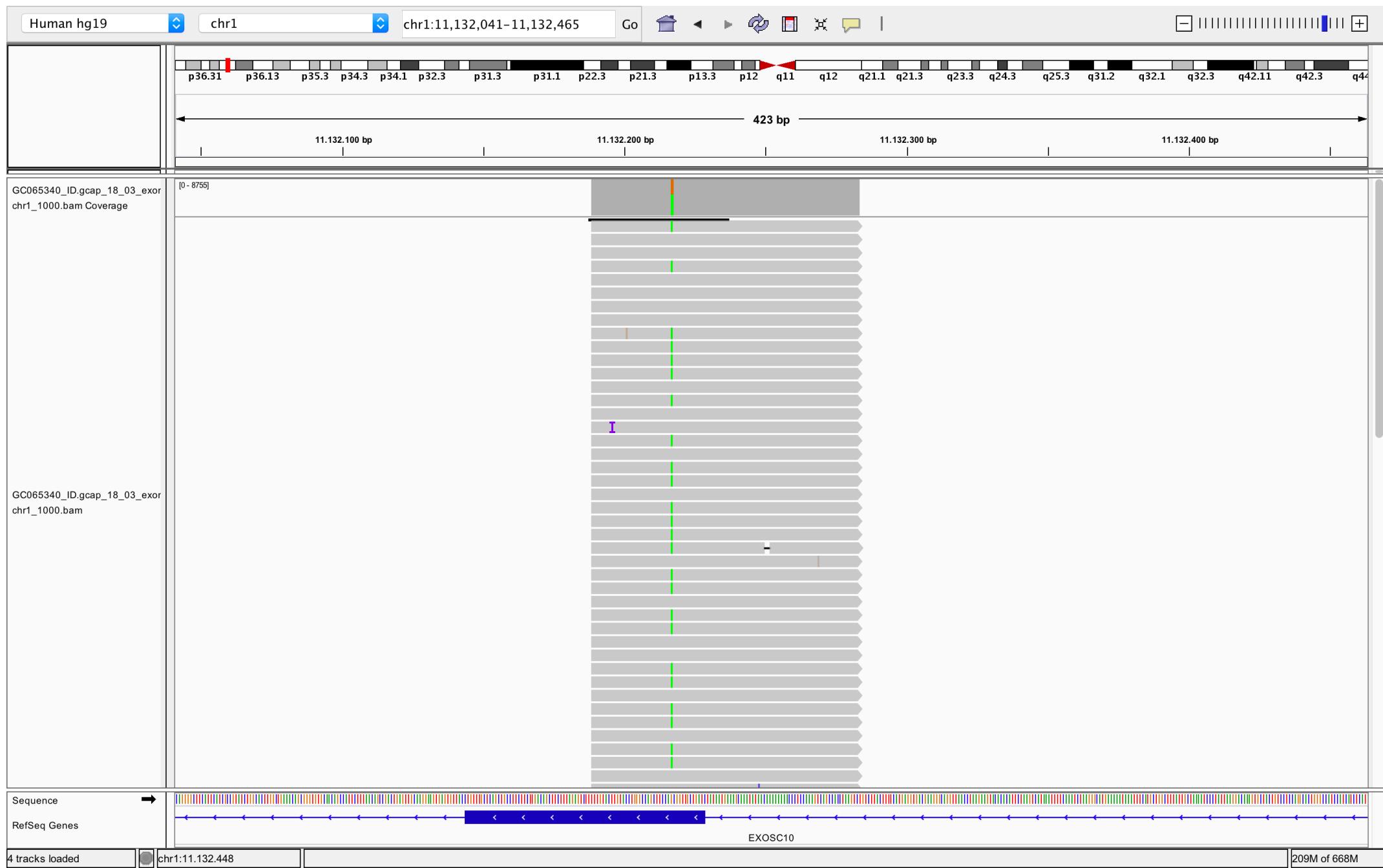


# Targeted capture sequencing

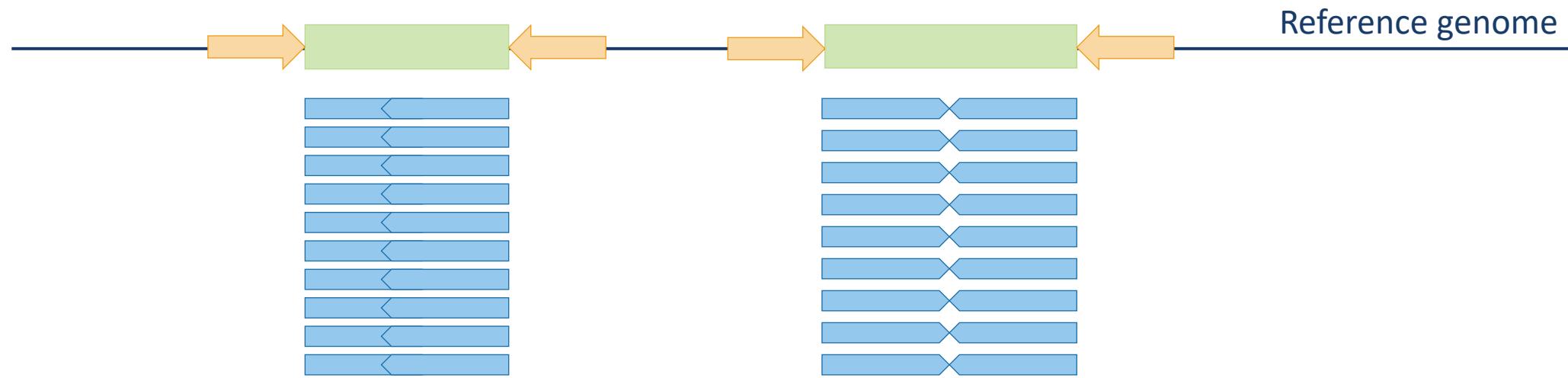


# Targeted capture sequencing

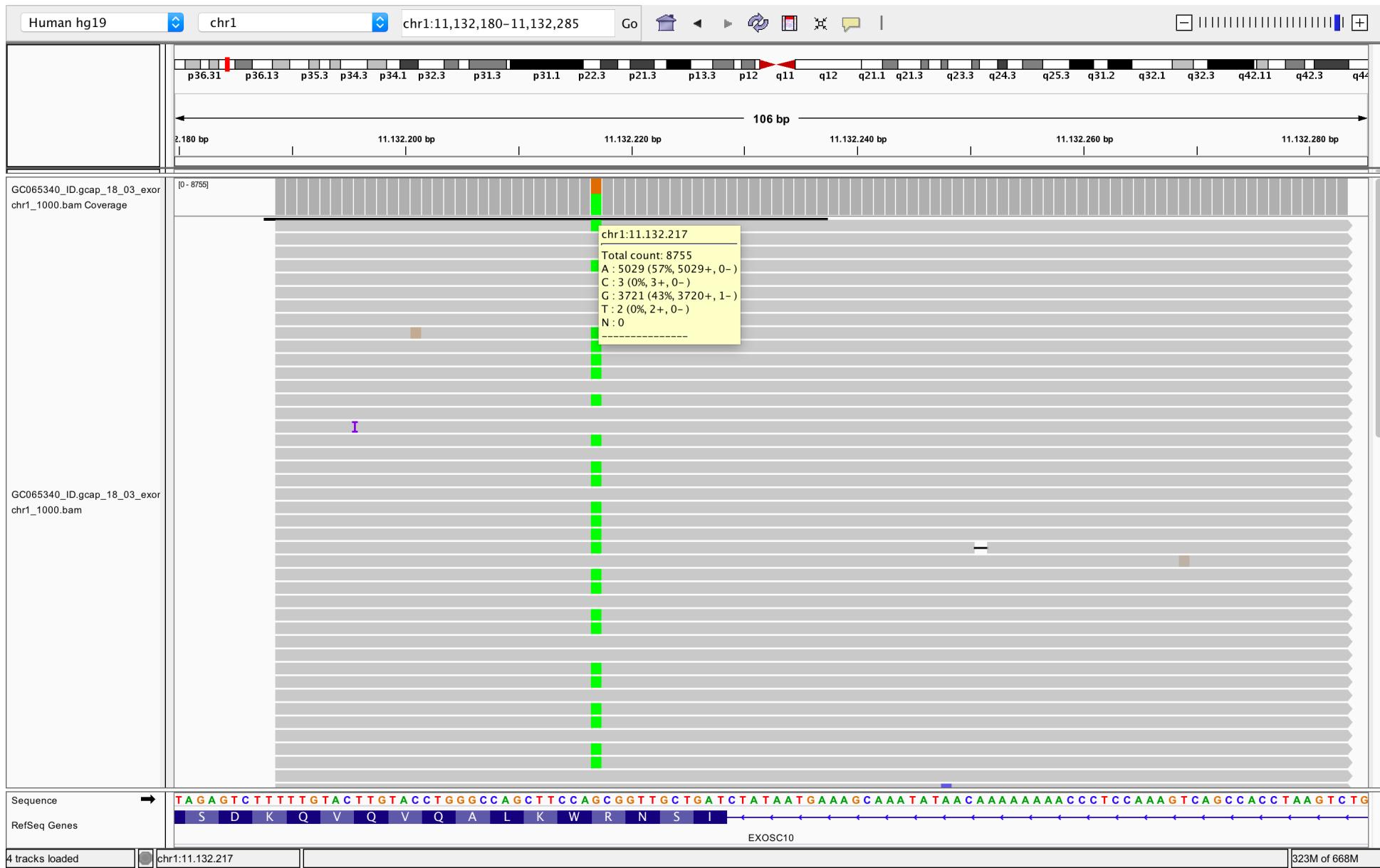


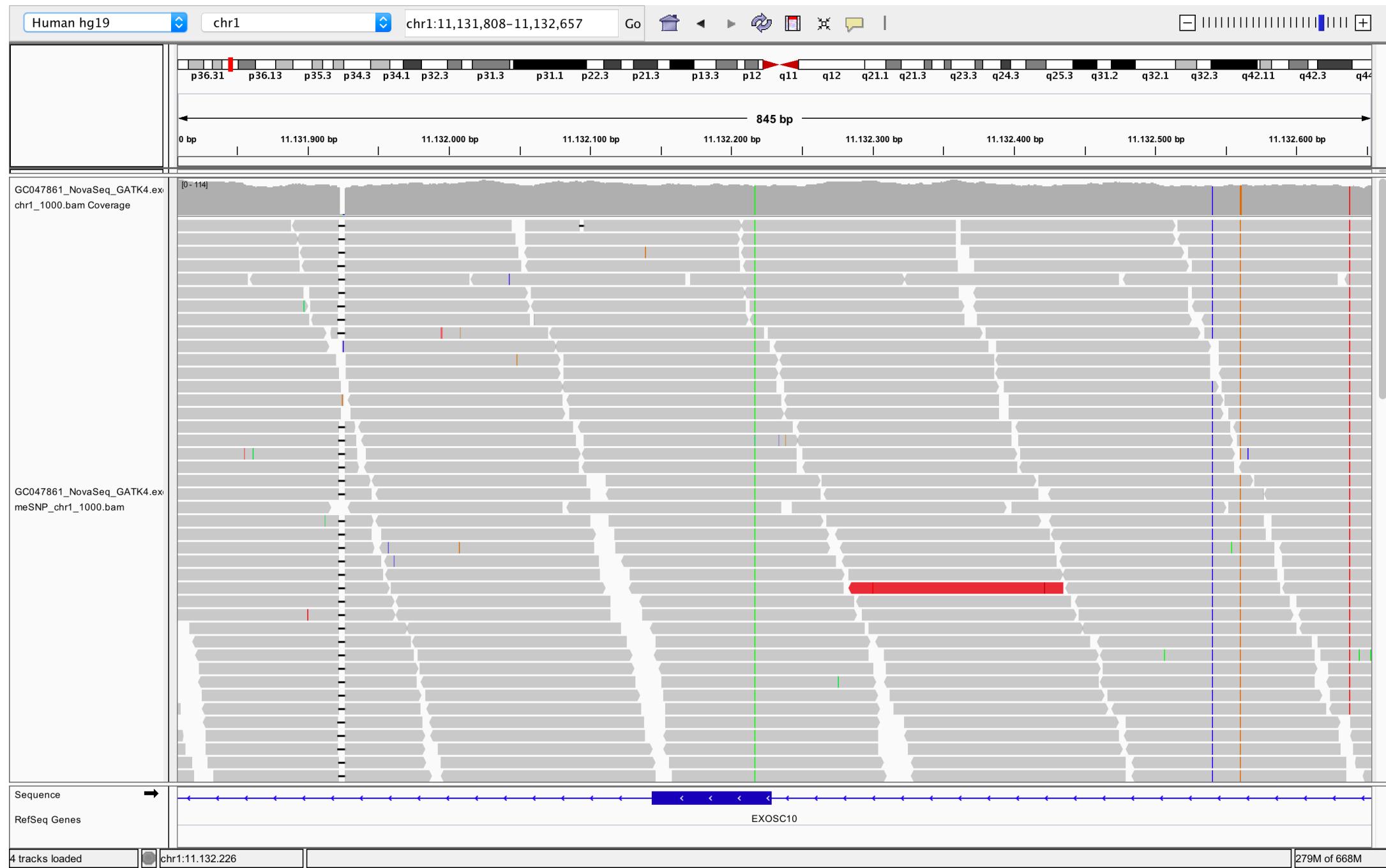


# Targeted amplicon sequencing

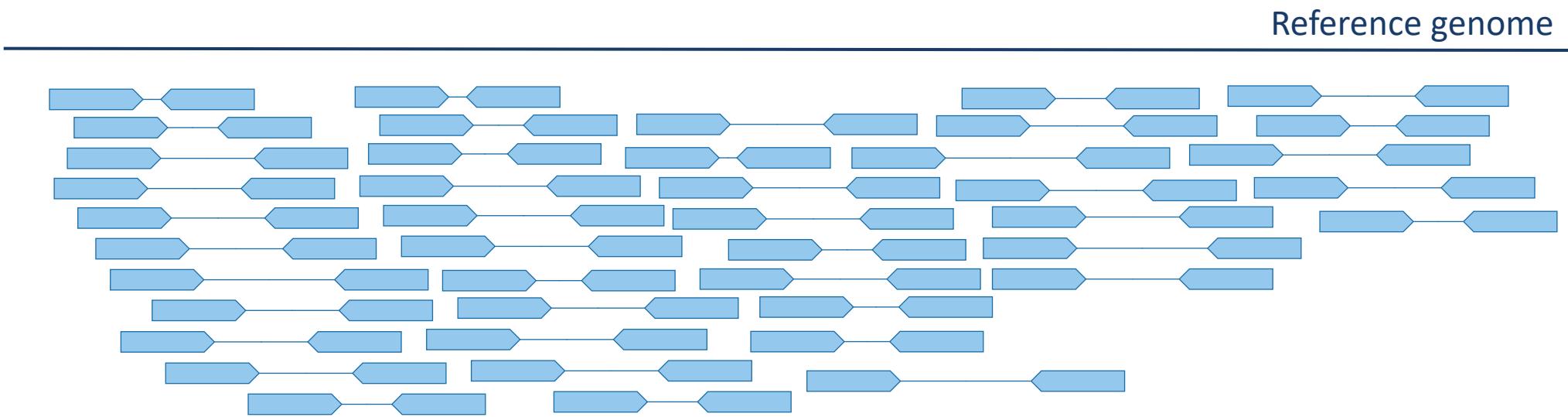


# Targeted amplicon sequencing

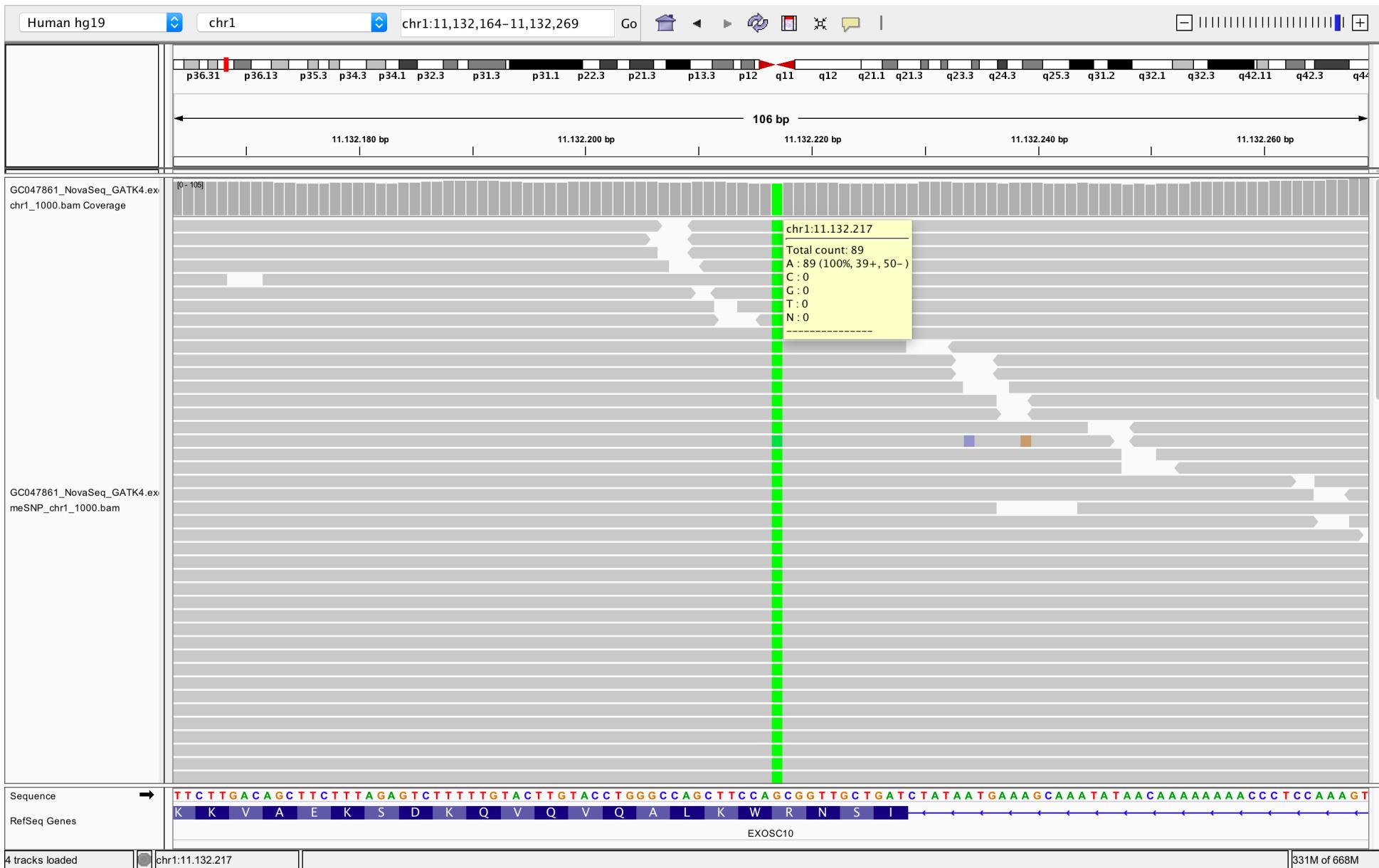




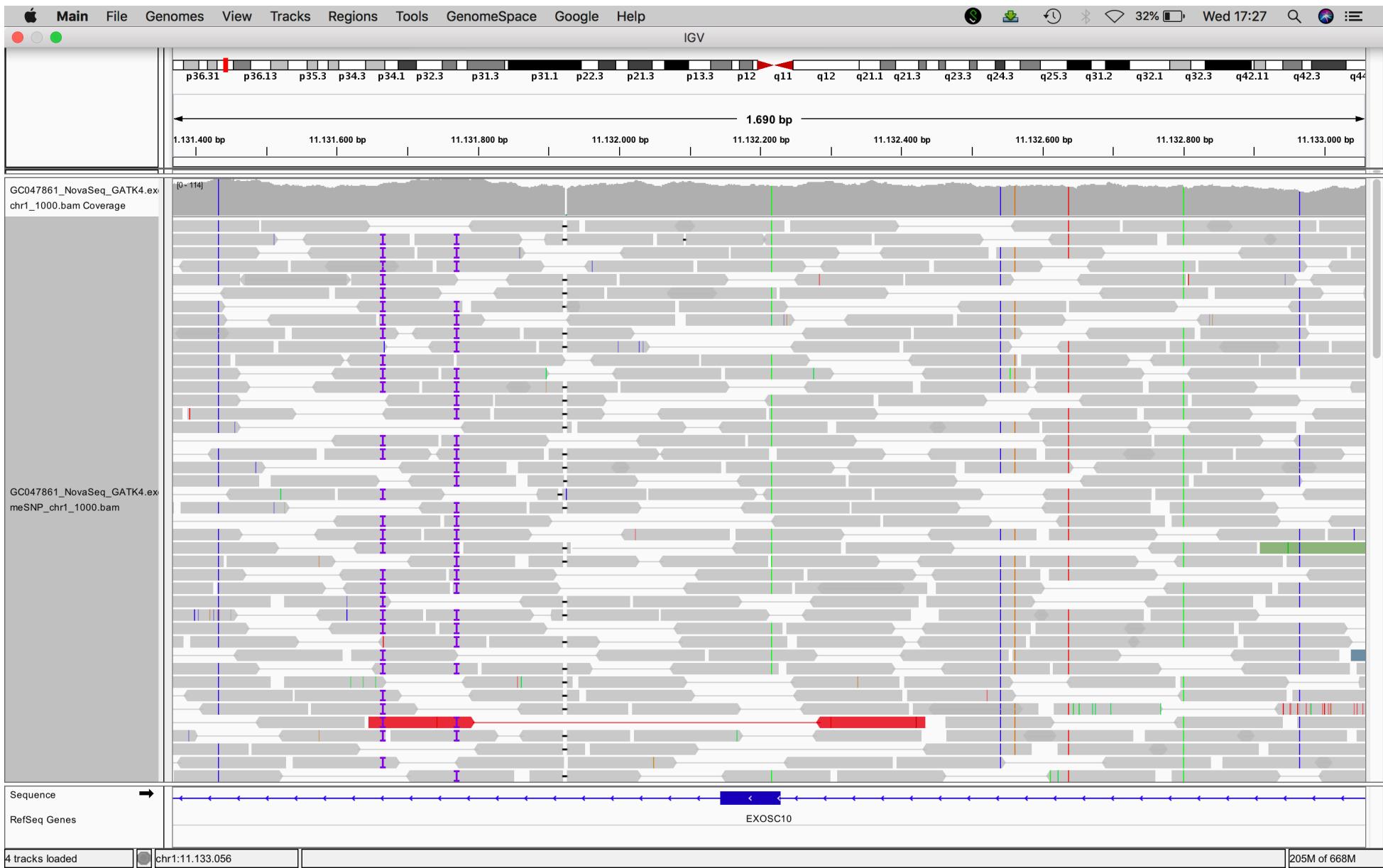
# WGS



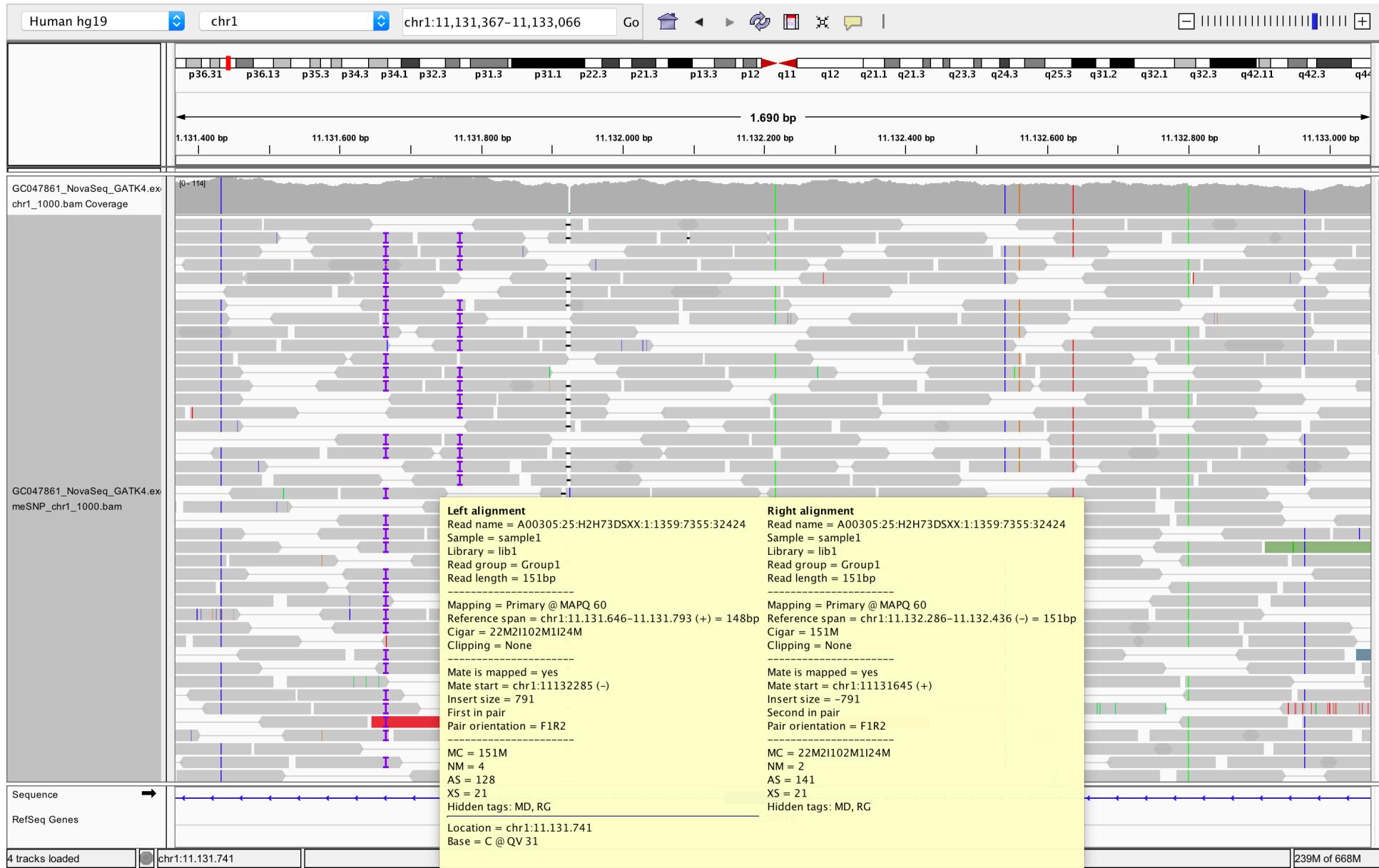
# WGS



# WGS



# WGS



# Outline

- Same variant in various assays
- Looking at the data
- Variant analysis
- Coverage analysis

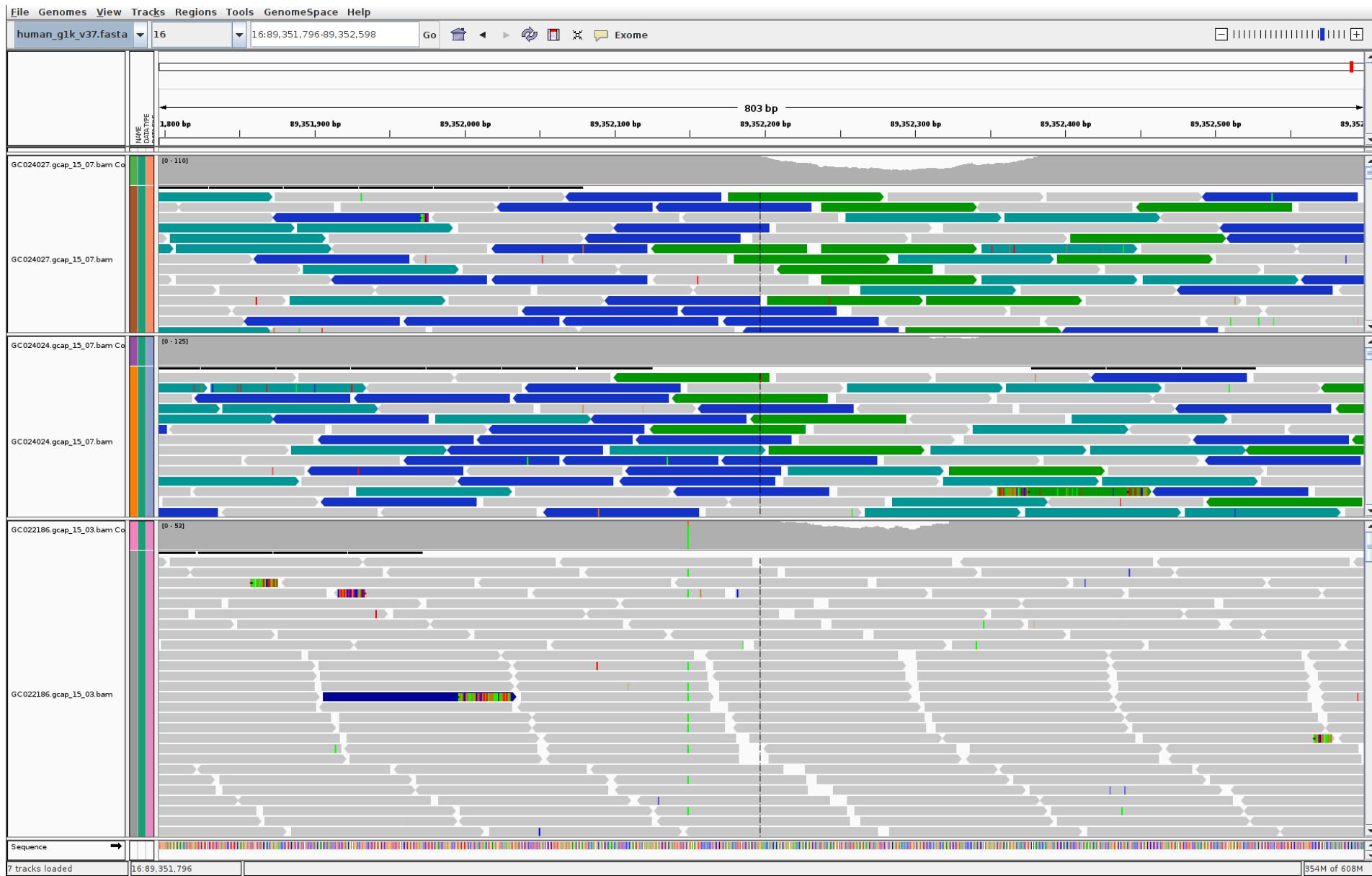
# Viewing a gene



# Mapping quality



# Read pairs



# Outline

- Looking at the data
- Same variants in various assays
- Variant analysis
- Coverage analysis

# Checking variants

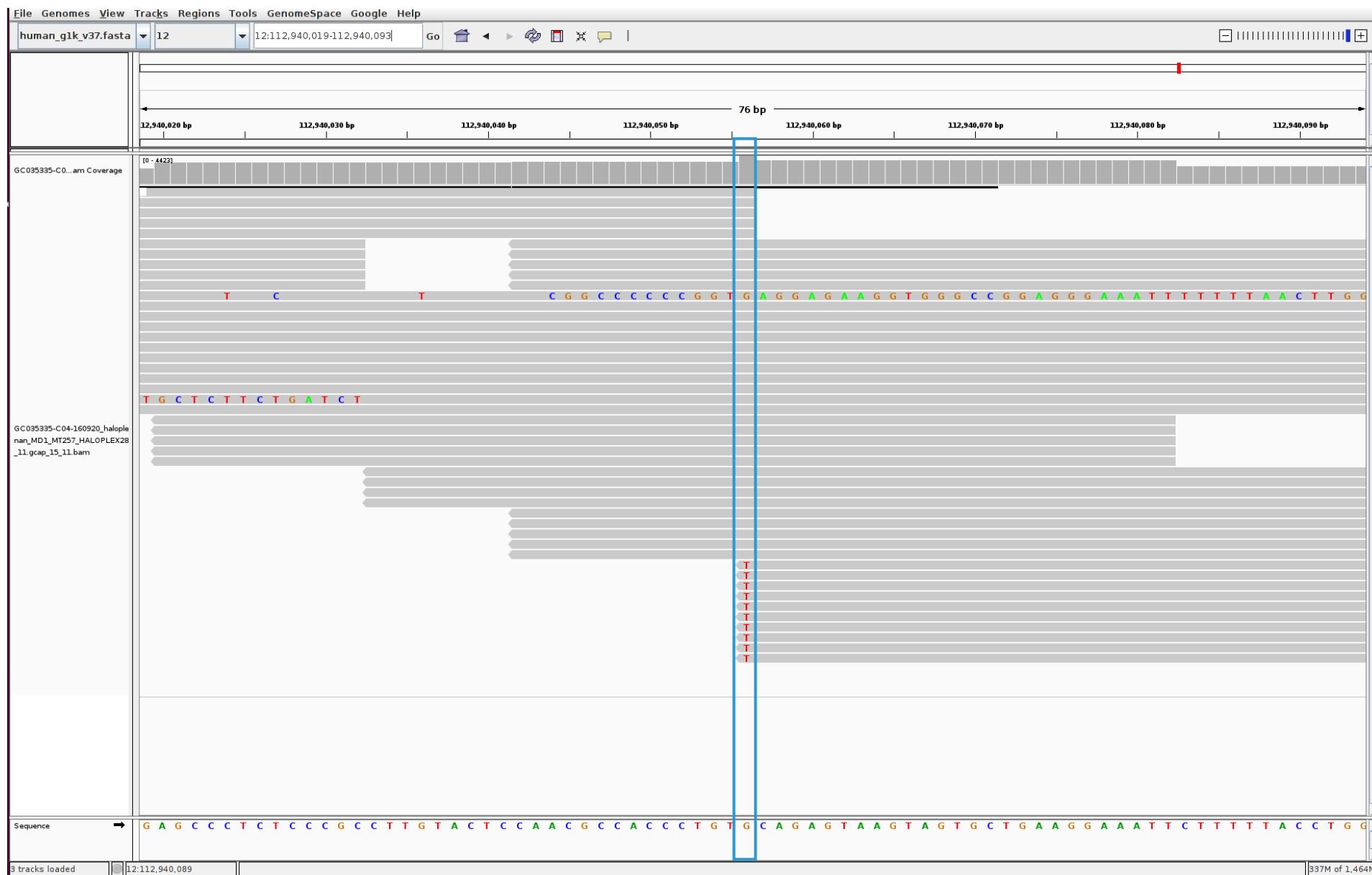
## (1) variant detected with unbalance allelic depth

Amplicon based assay

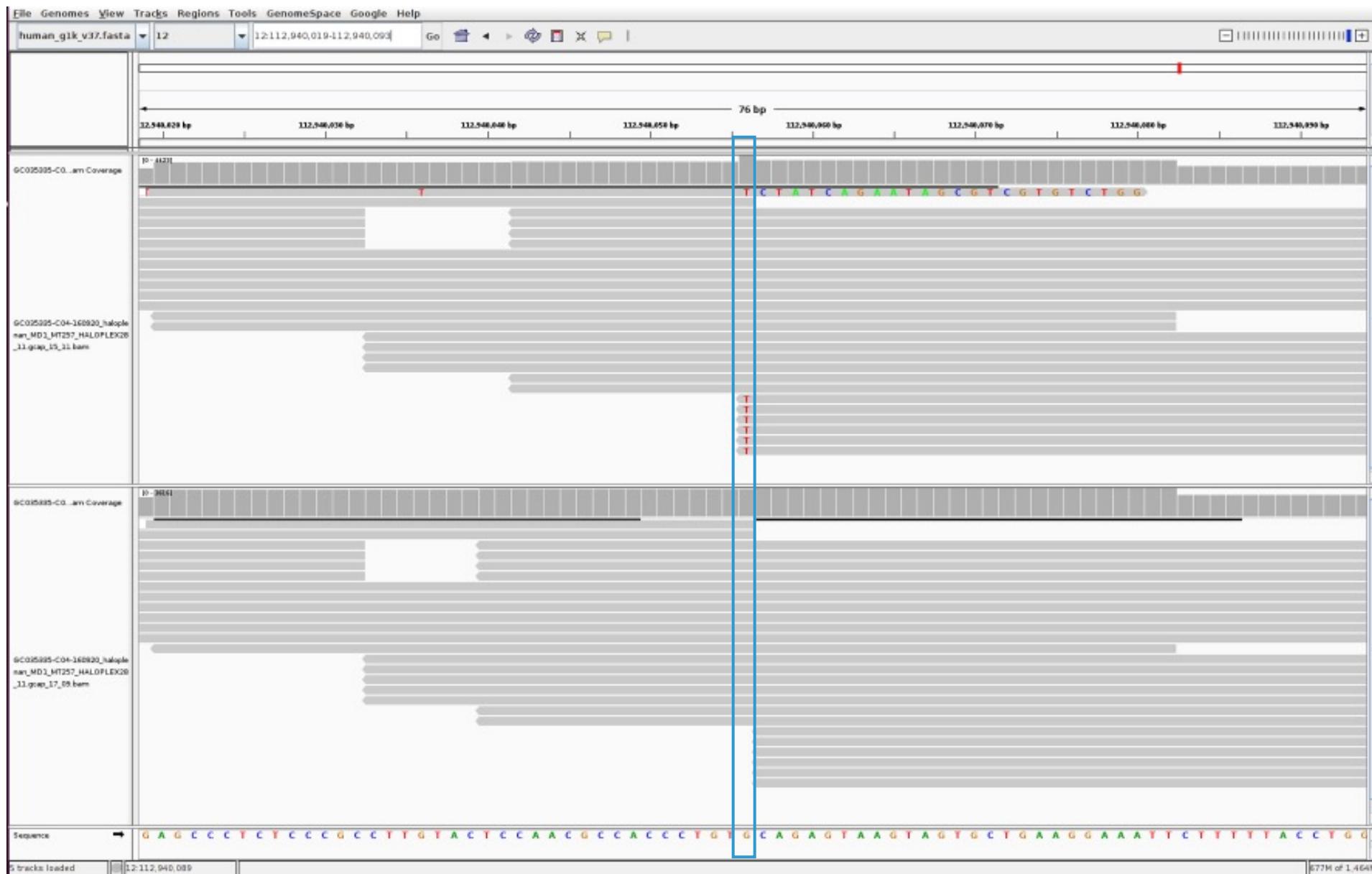
Heterozygous variant G/T

Format	Value
GT	0/1
AD	<b>3791,1108</b>
DP	4899
GQ	99

# (1) unbalance AD



# (1) unbalance AD



# Checking variants

(2) variants on same allele ?

Capture assay

Recessive disorder

Two heterozygous variants: GACT/G and G/A

Format	Variant 1	Variant 2
GT	0/1	0/1
AD	50,39	42,44
DP	89	86
GQ	99	99

## (2) Variants on same allele ?



# Checking variants

## (3) Two variants detected with unbalance allelic depth

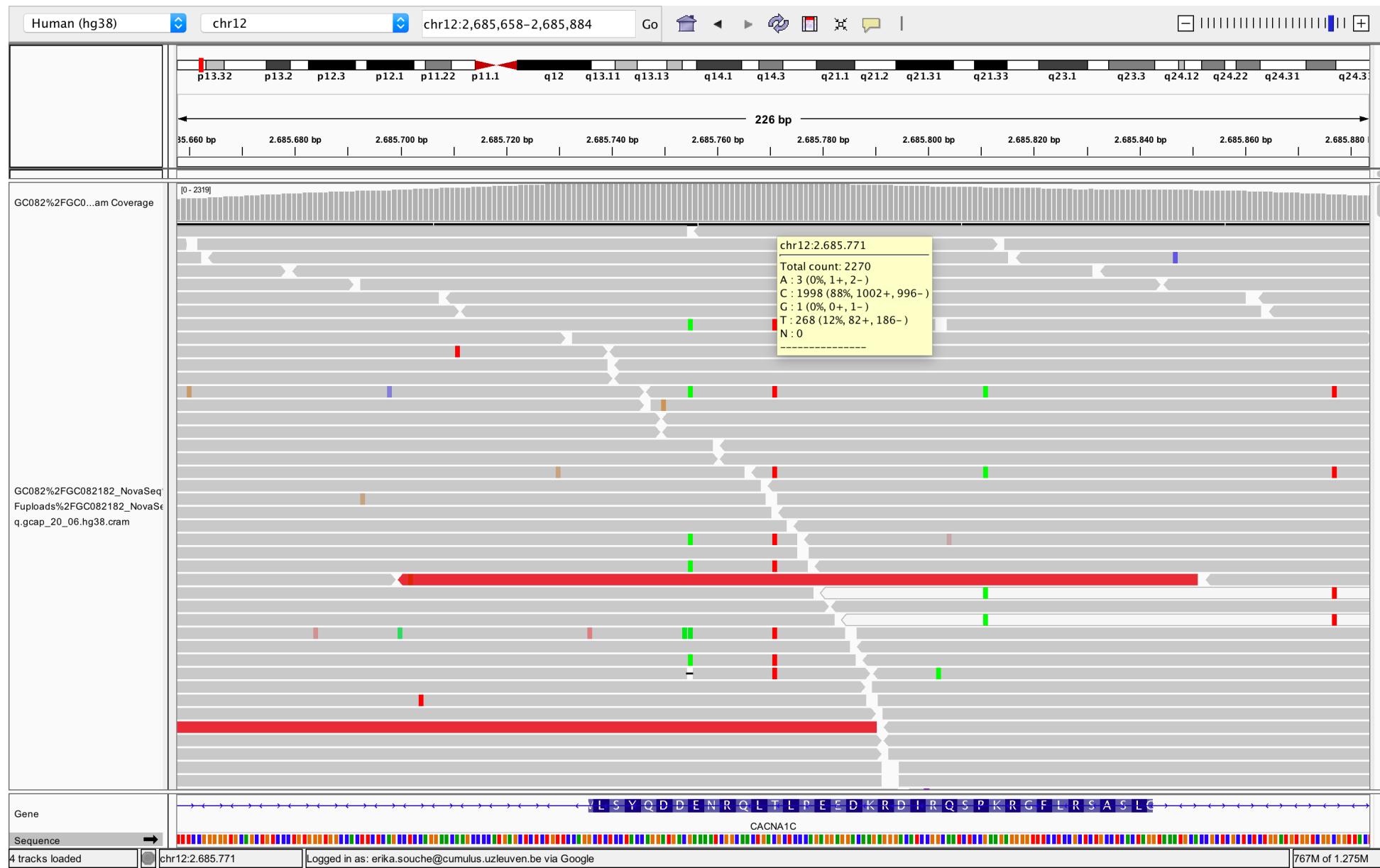
Capture assay

Heterozygous variants

rs200231105 & rs201777030

Format	Variant 1	Variant 2
GT	0/1	0/1
AD	<b>1939,256</b>	<b>1995,217</b>
DP	2209	2222
GQ	99	99

# (3) unbalance AD



### (3) unbalance AD

# BLAT reads supporting variants

## CACNA1C gene

# LINC02371 gene

```
0000001 ctcagcctccaggaacaagccccatgagctctgttccaggcttccta 0000050
>>>>> ||||| ||||| ||||| ||||| ||||| ||||| ||||| ||||| ||||| >>>>>
2742033 ctcagcctccaggaacaagccccatgagctctgttccaggcttccta 2742082

0000051 ccaggatgacaaaaatcgcaactgatgctcccagaggaggacaagaggg 0000100
>>>>> ||||| ||||| ||||| ||||| ||||| ||||| ||||| ||||| >>>>>
2742083 ccaggatgacaaaaatcgcaactgatgctcccagaggaggacaagaggg 2742132

0000101 a 0000101
>>>>> | >>>>>
2742133 a 2742133
```

# Checking variants

(4) real *de novo* variant ?

Capture assay

Trio *i.e.* child & parents

Heterozygous deletion TA/T in child

Format	Value
GT	0/1
AD	43,51
DP	94
GQ	99

# (4) real *de novo* ?



# Checking variants in IGV

(4) real *de novo* variant ?

Targeted assay

Trio *i.e.* child & parents

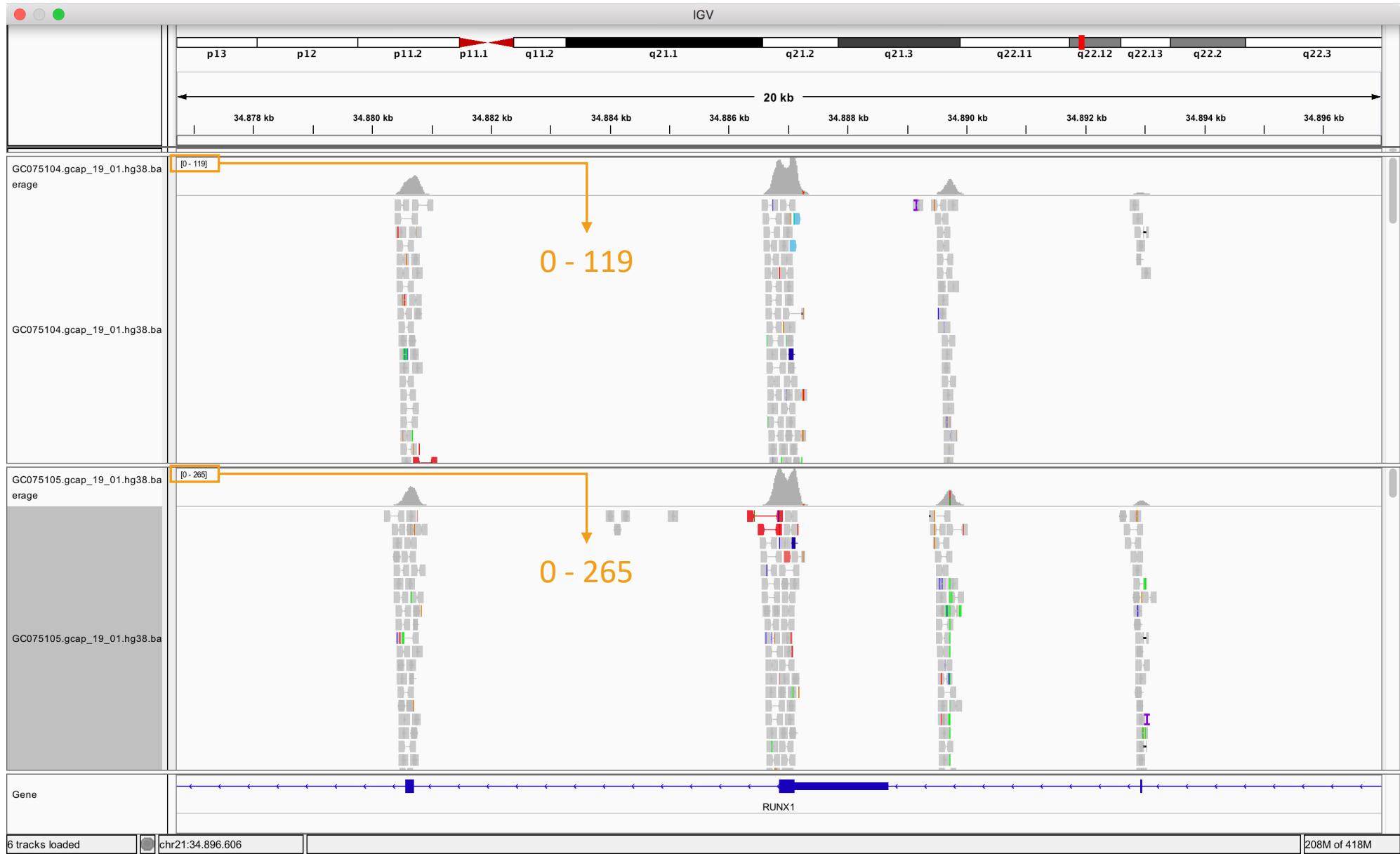
Heterozygous deletion TA/T in child

Format	Index	Mother	Father
GT	0/1	0/0	0/0
AD	43,51	103,0	81,0
DP	94	103	81
GQ	99	-	-

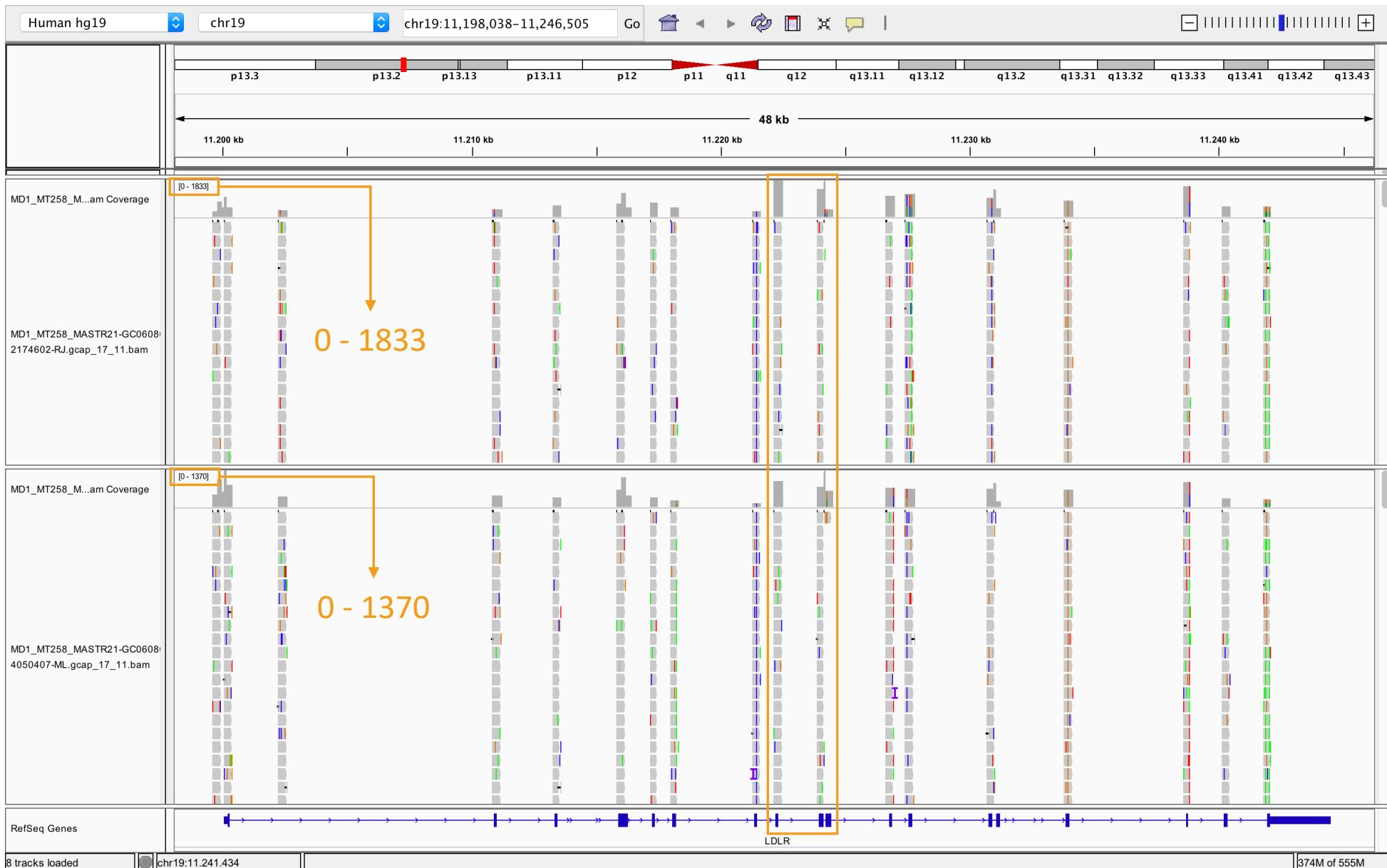
# Outline

- Looking at the data
- Same variants in various assays
- Variant analysis
- Coverage analysis

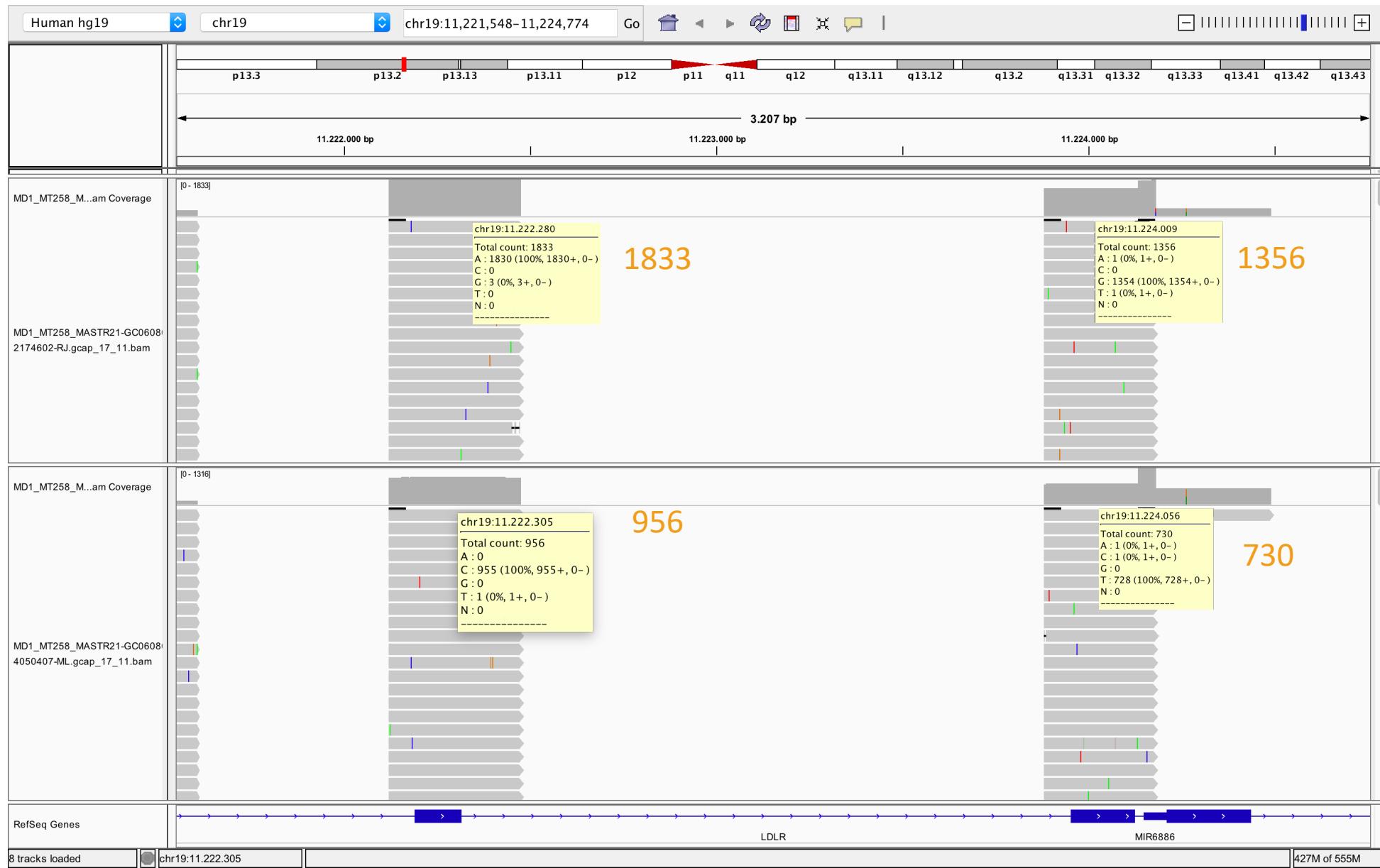
# Coverage analysis



# Coverage analysis



# Coverage analysis



# Summary

- Viewing the BAM file may help
  - Identifying processing issues
  - Understanding the results
  - Have a feeling of the data
- Viewing the BAM file is not always necessary
  - Joint genotyping
  - Structural variant detection software
  - Etc.

# Questions?

Erika Souche

