



Геномные браузеры UCSC & IGV

Визуализация результатов NGS

ПО и данные:

IGV:

- <http://software.broadinstitute.org/software/igv/download>

Данные:

- <https://is.gd/fyZMB8>

IGV: Integrative Genomic Viewer

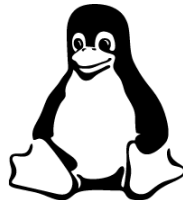


- <http://software.broadinstitute.org/software/igv/download>

Download -> Unzip

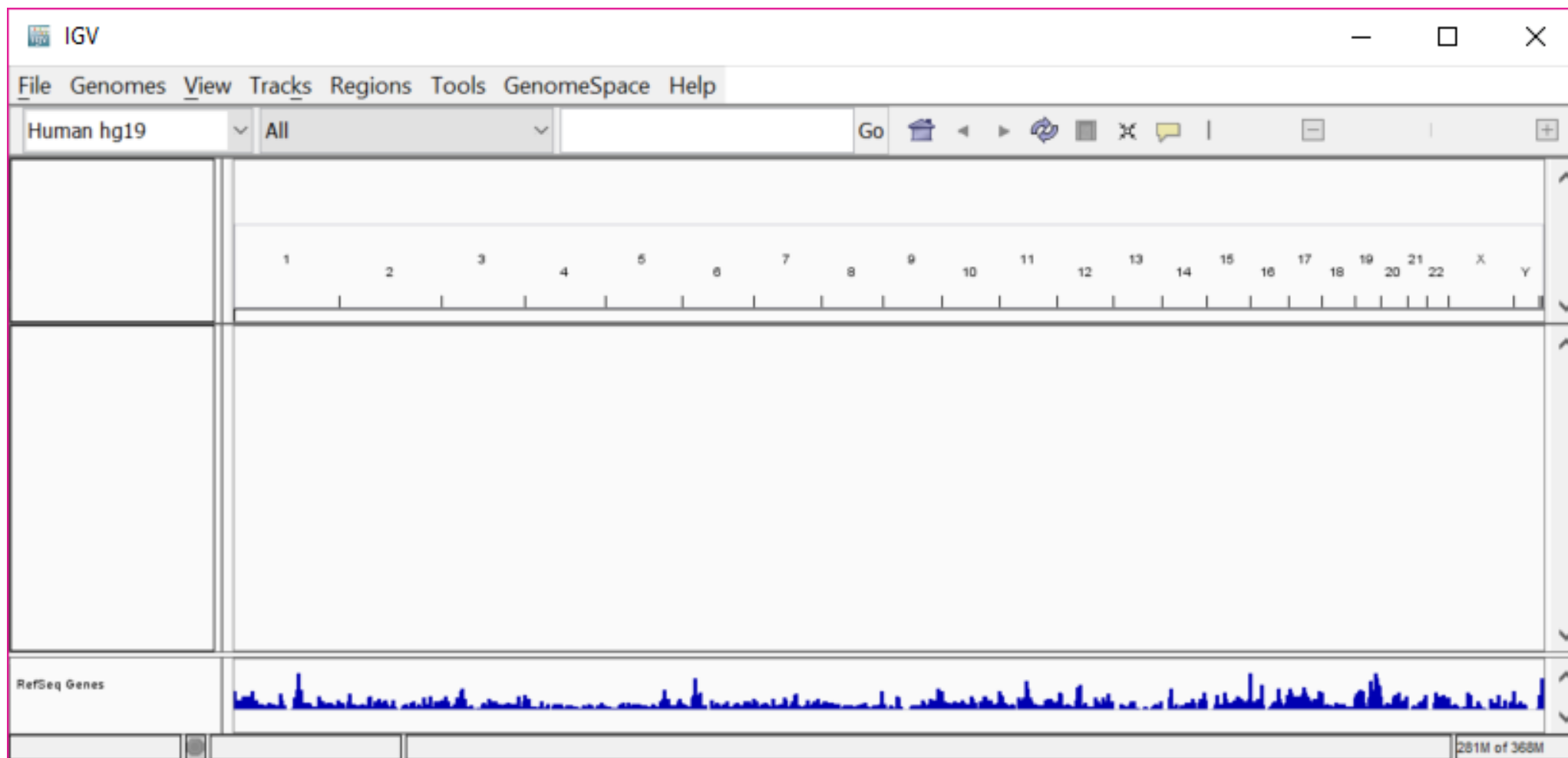


- Java 8 (`java -version`)
- `igv.bat: -Xmx4g >> -Xmx1000m`

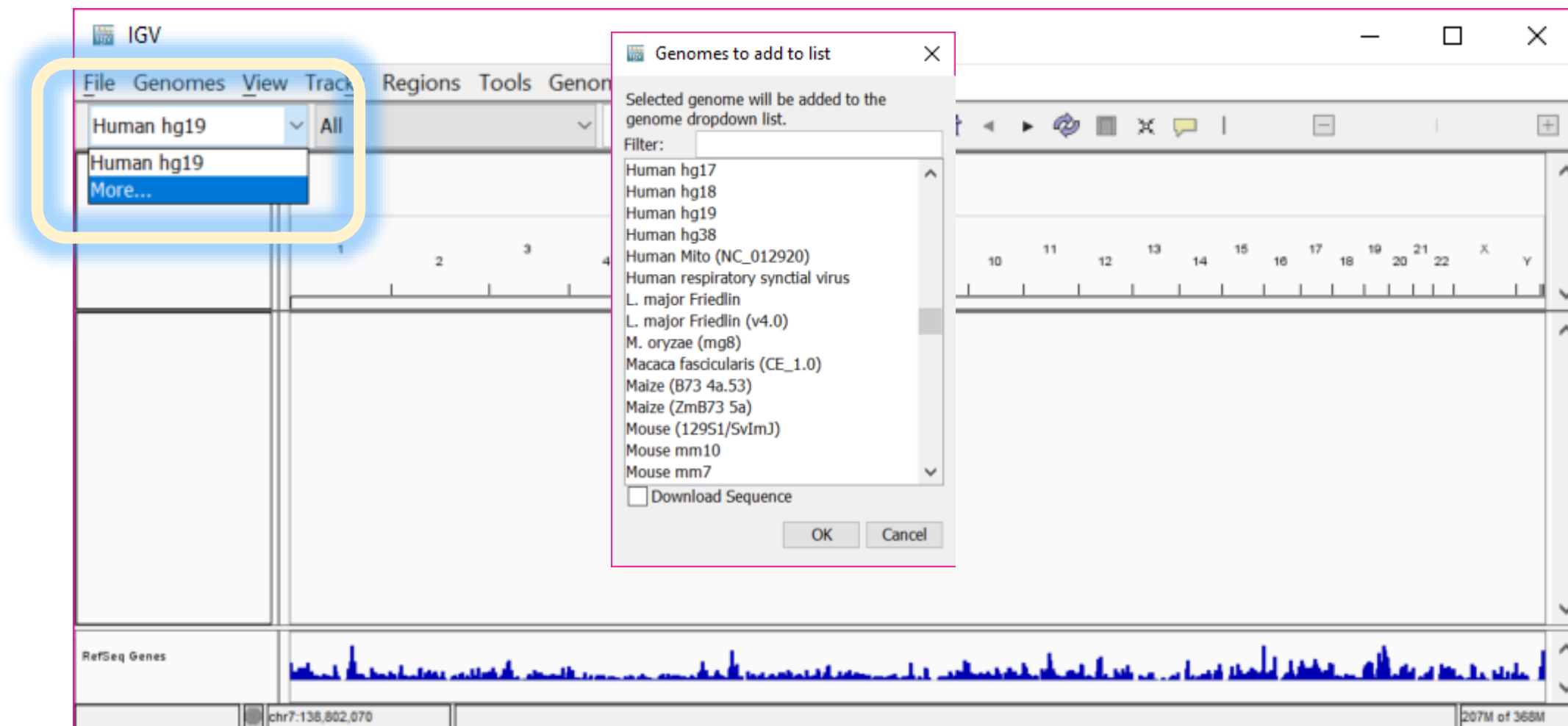


- `$ sudo apt-get install igv`

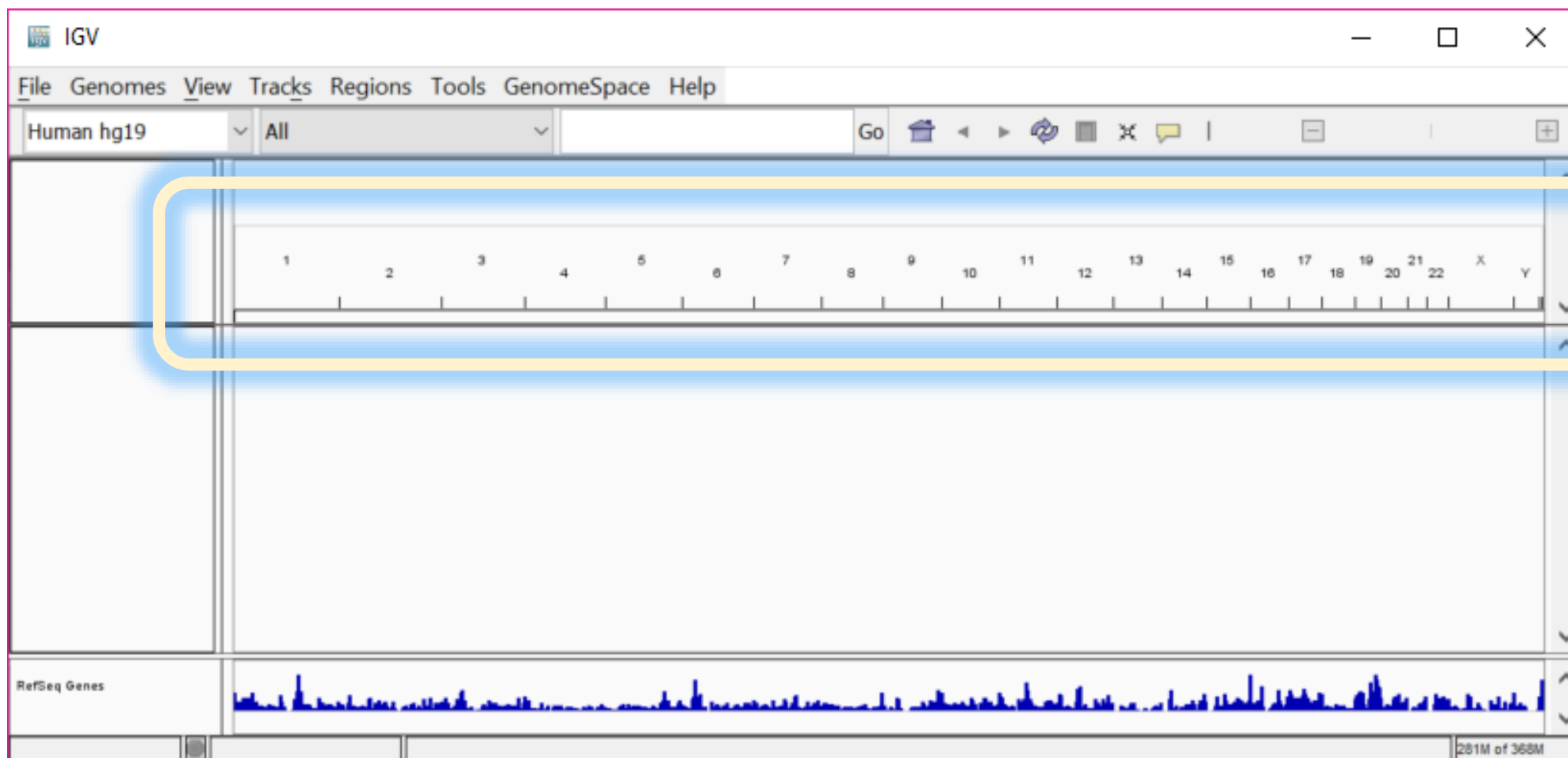
IGV: интерфейс



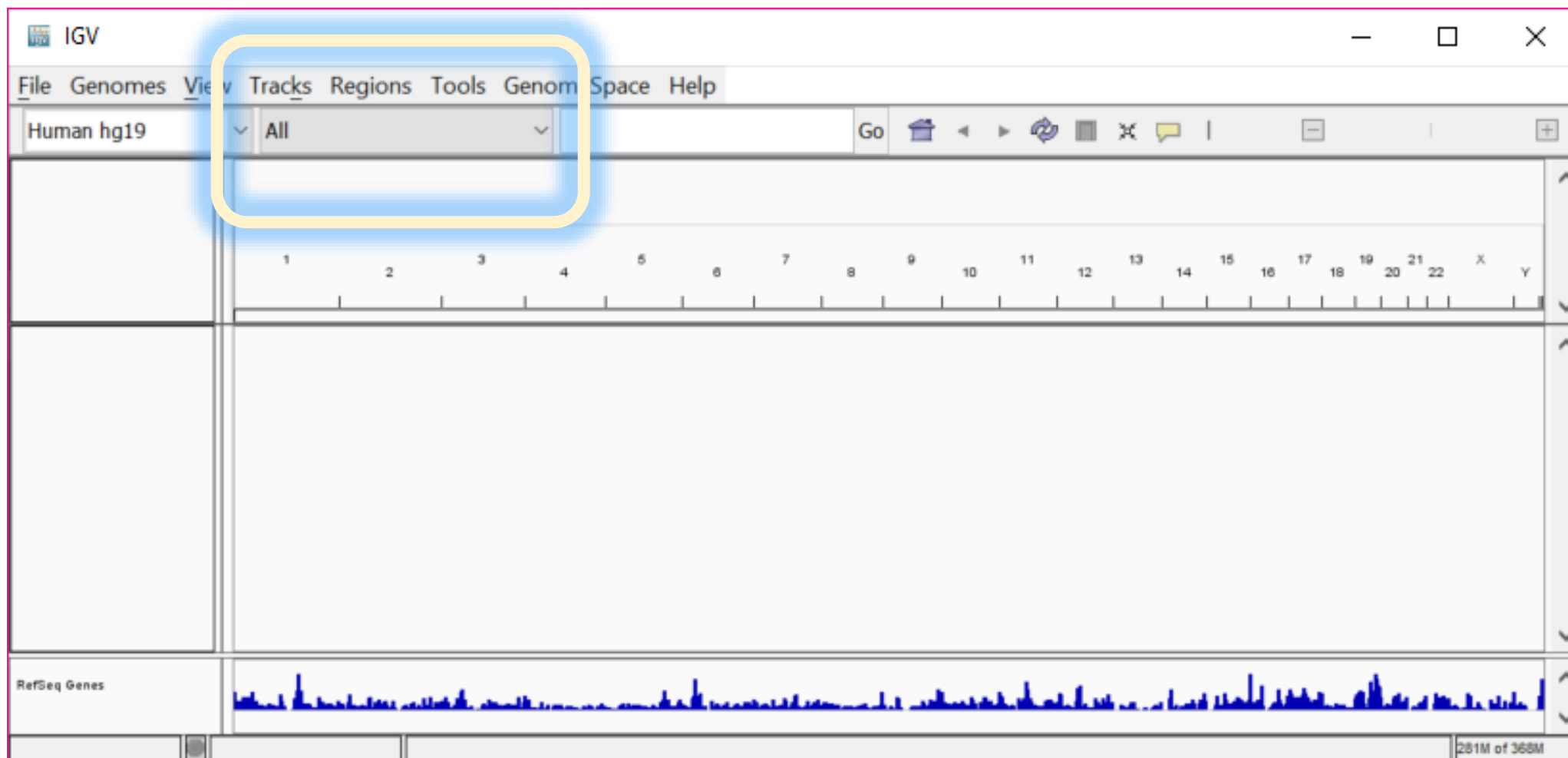
IGV: интерфейс



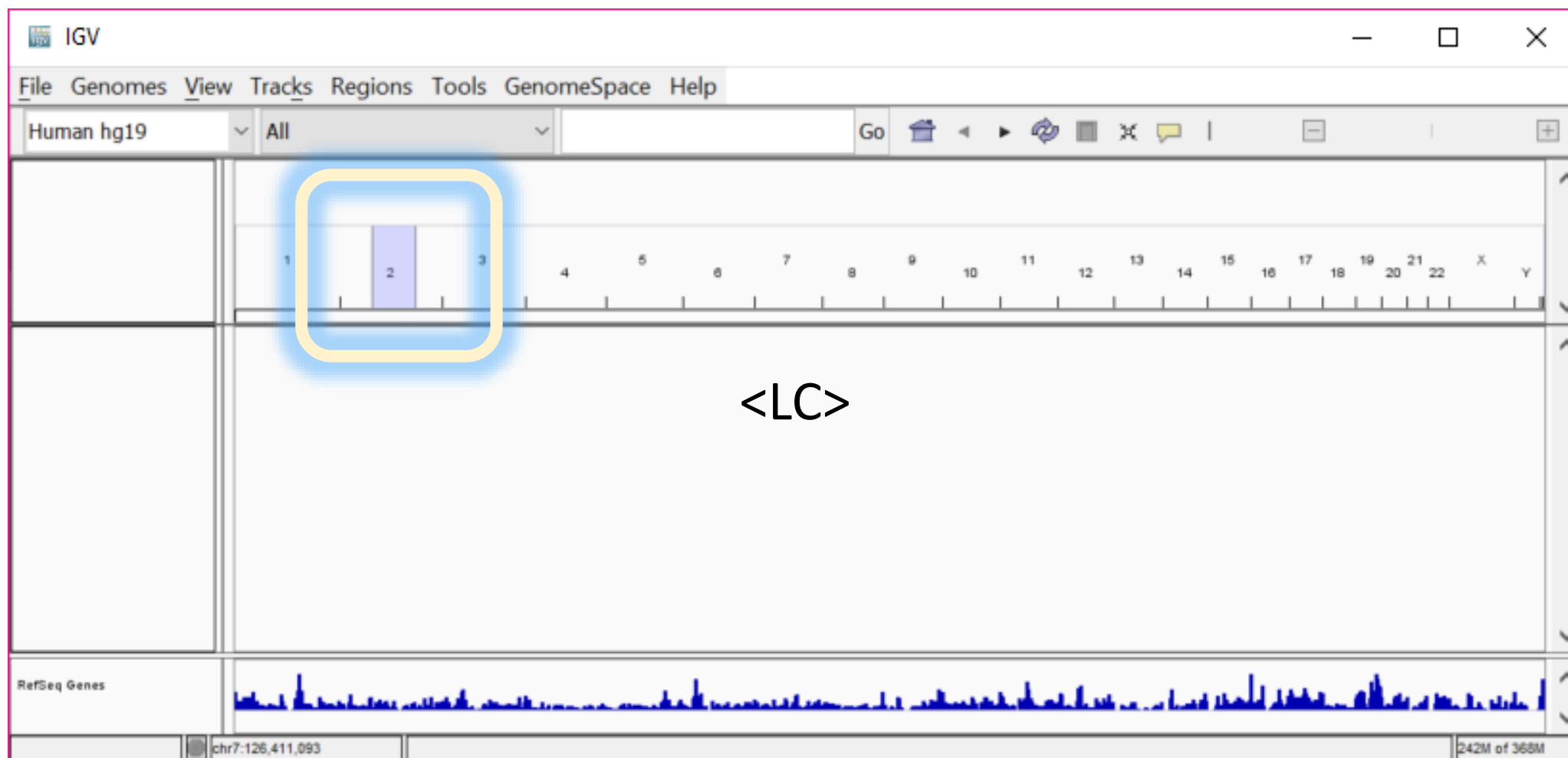
IGV: интерфейс



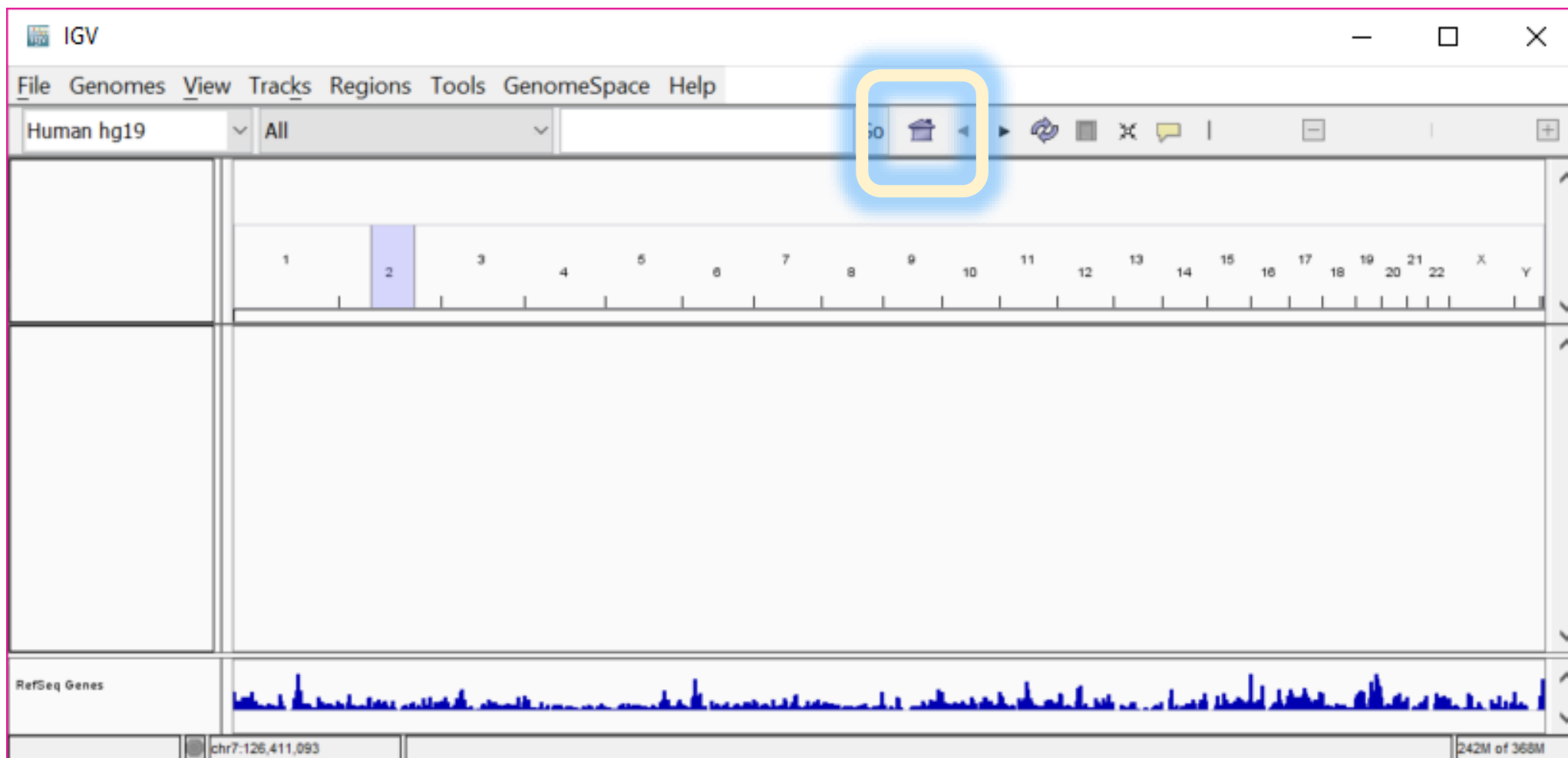
IGV: интерфейс



IGV: интерфейс



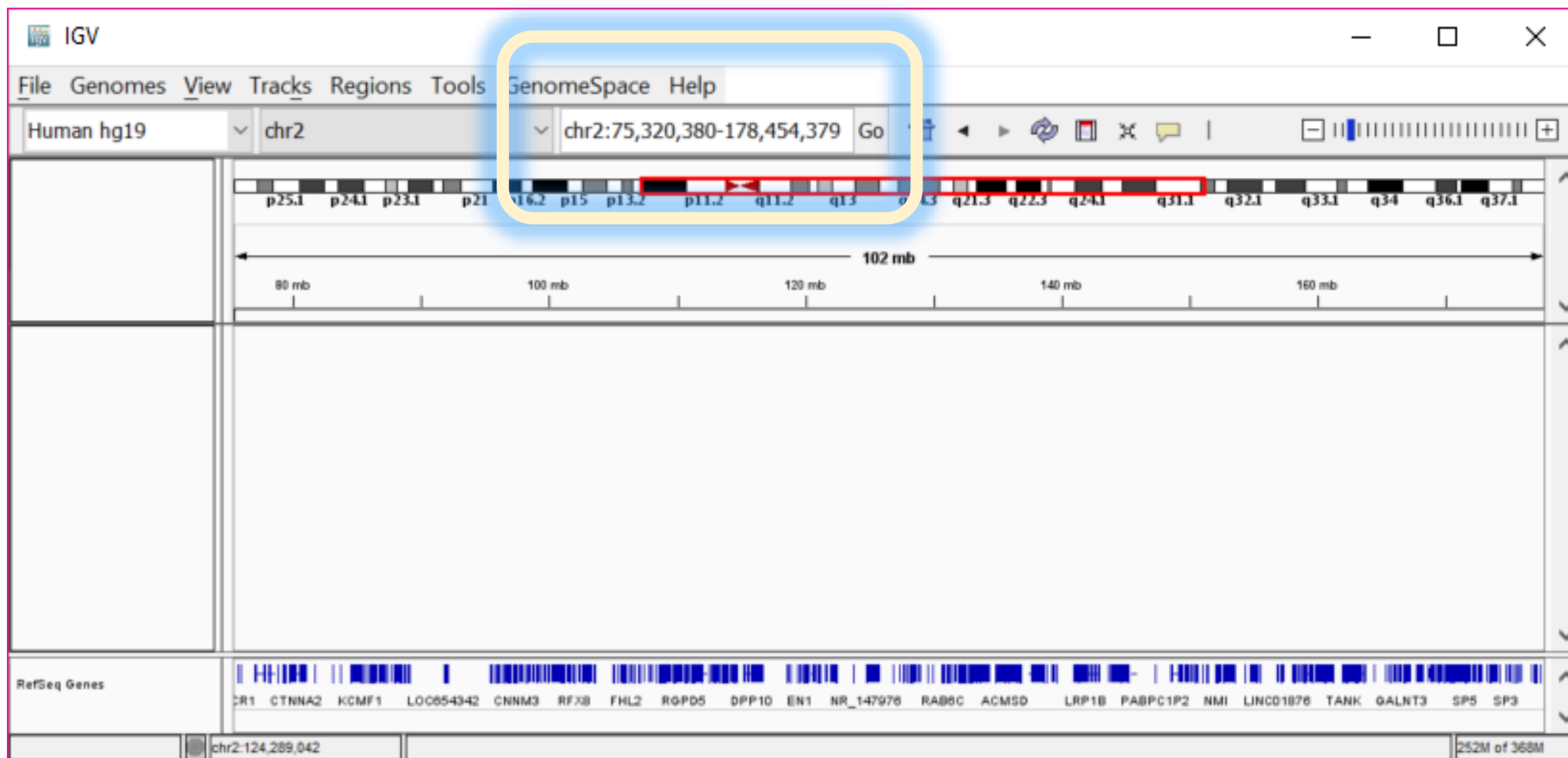
IGV: интерфейс



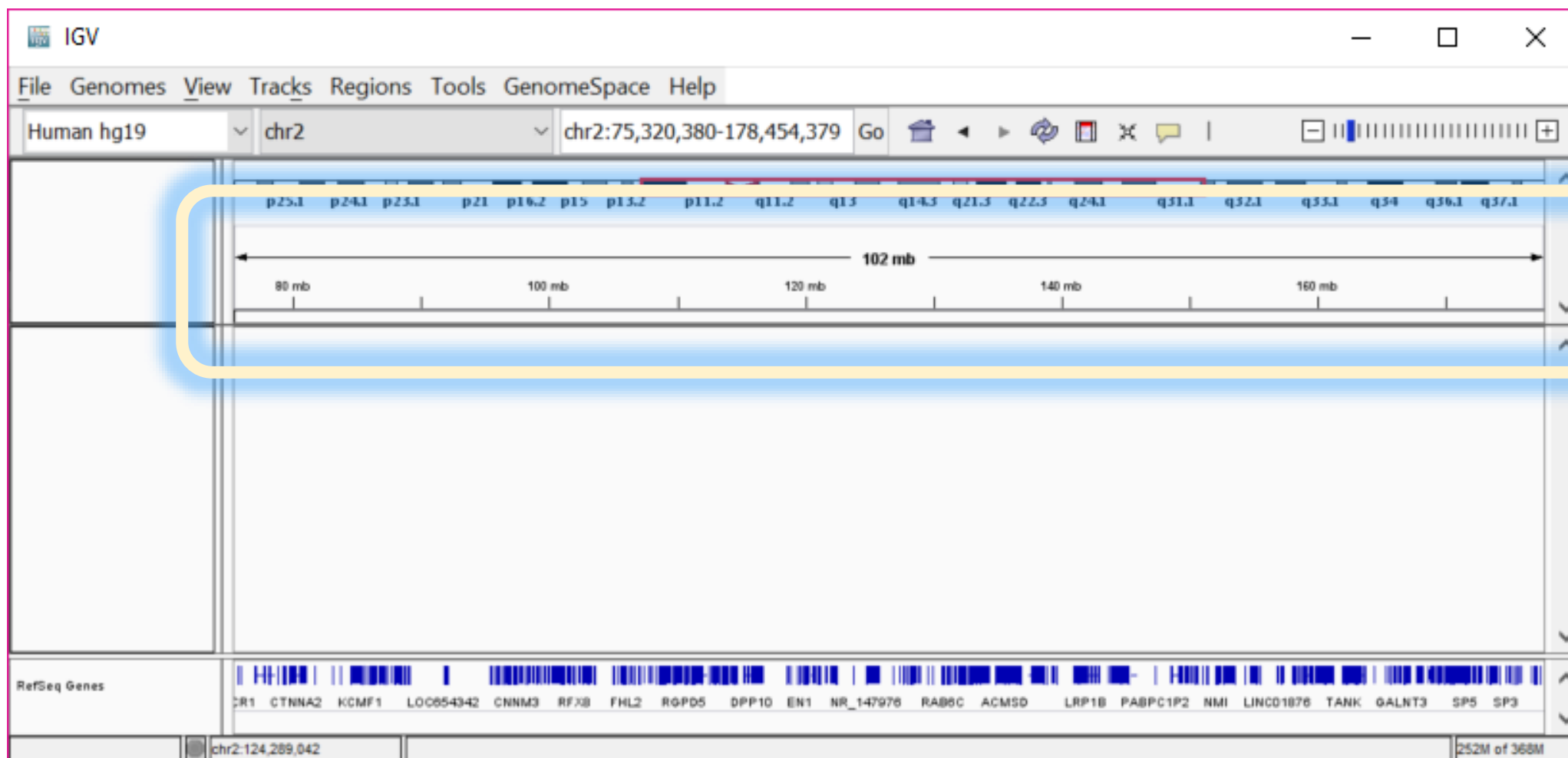
IGV: интерфейс



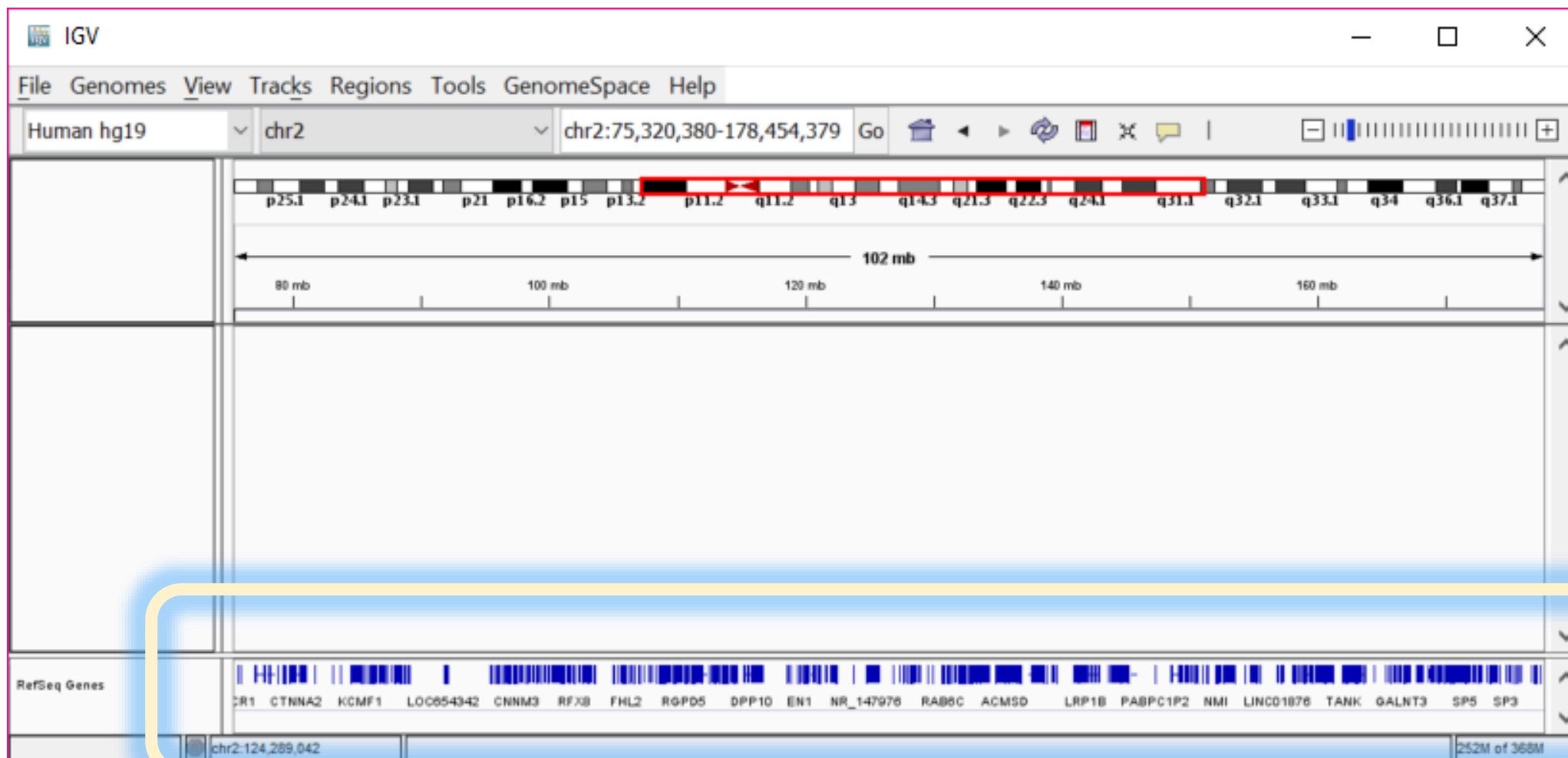
IGV: интерфейс



IGV: интерфейс



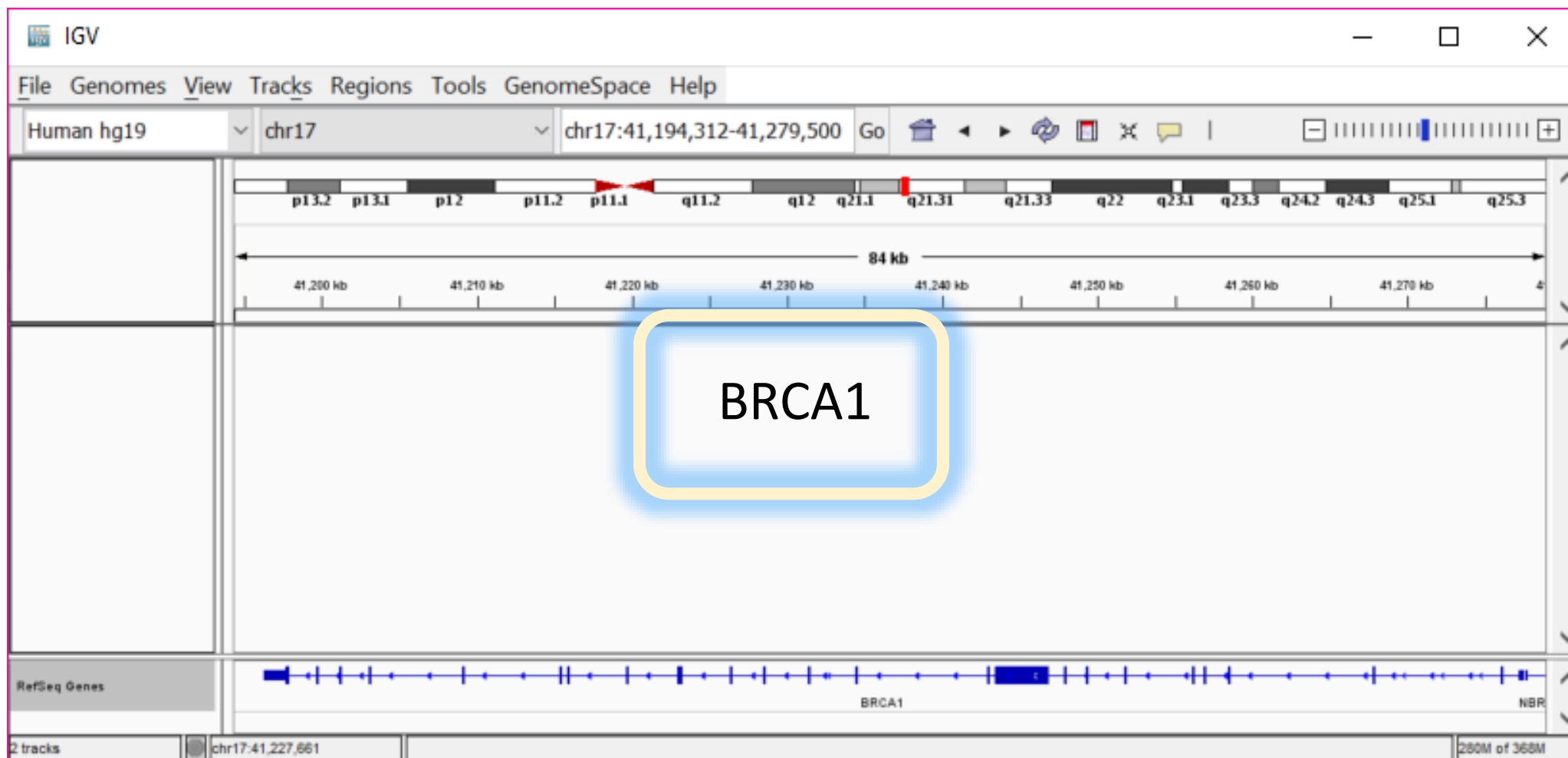
IGV: интерфейс



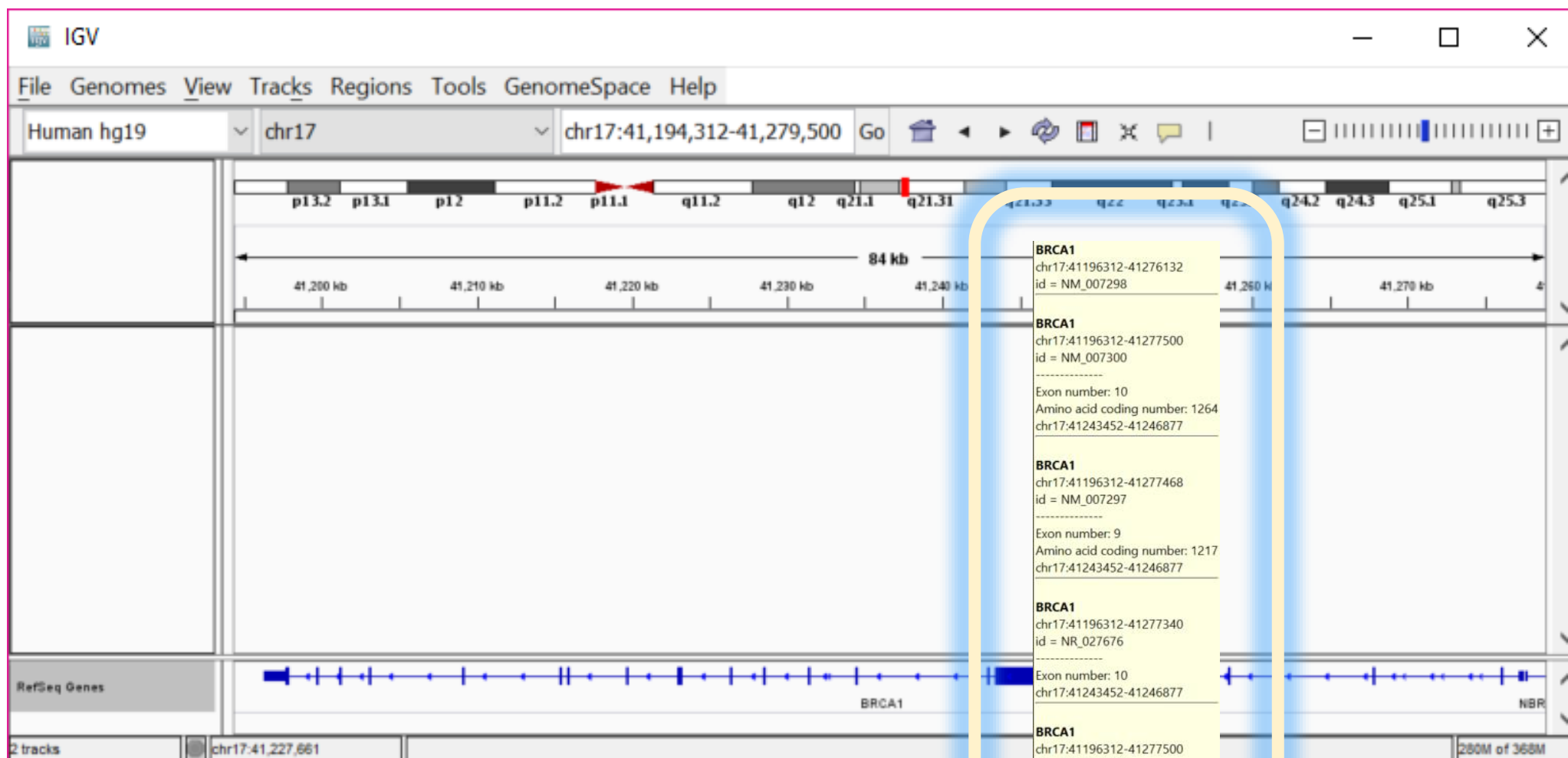
IGV: интерфейс



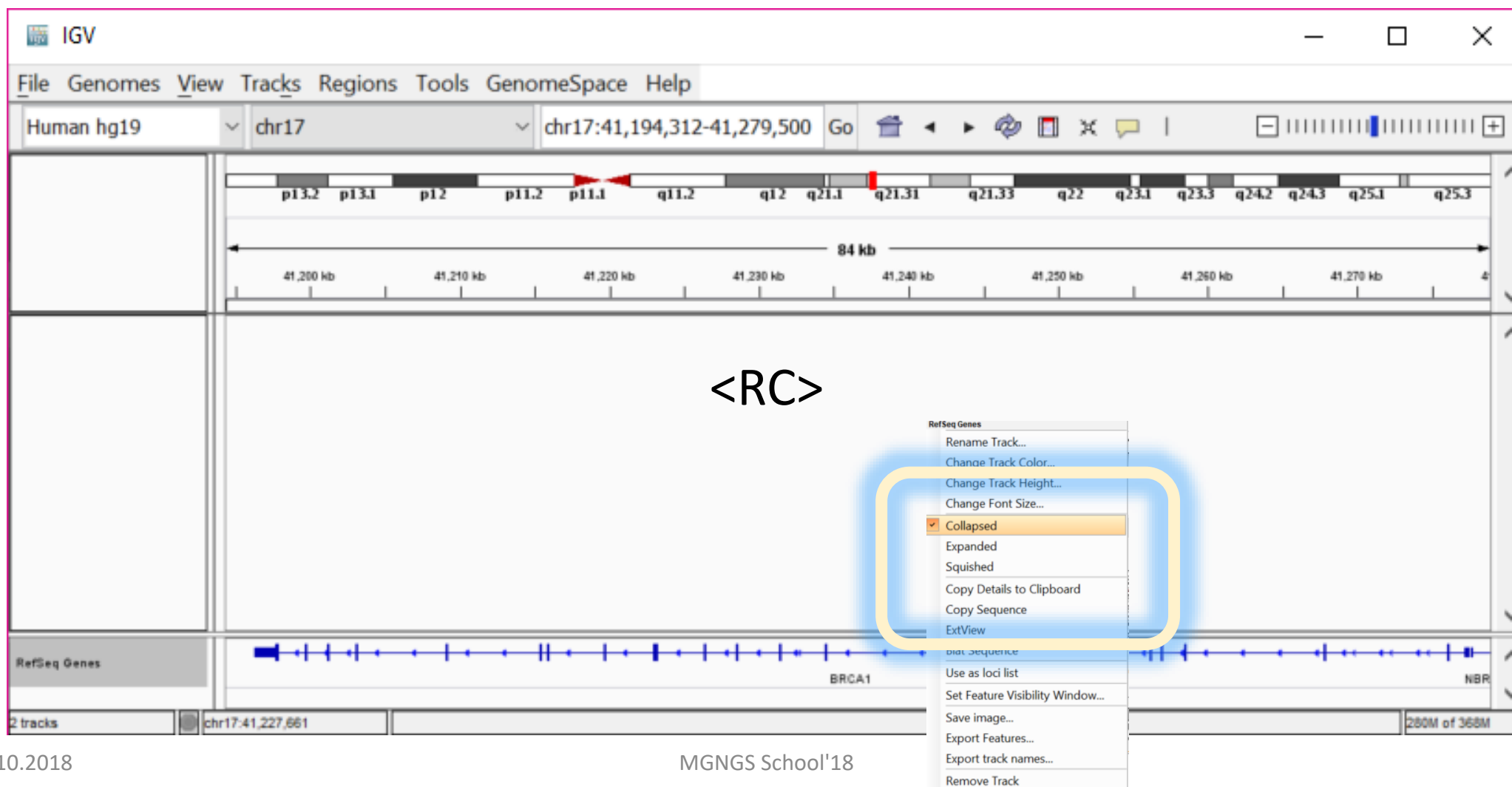
IGV: интерфейс



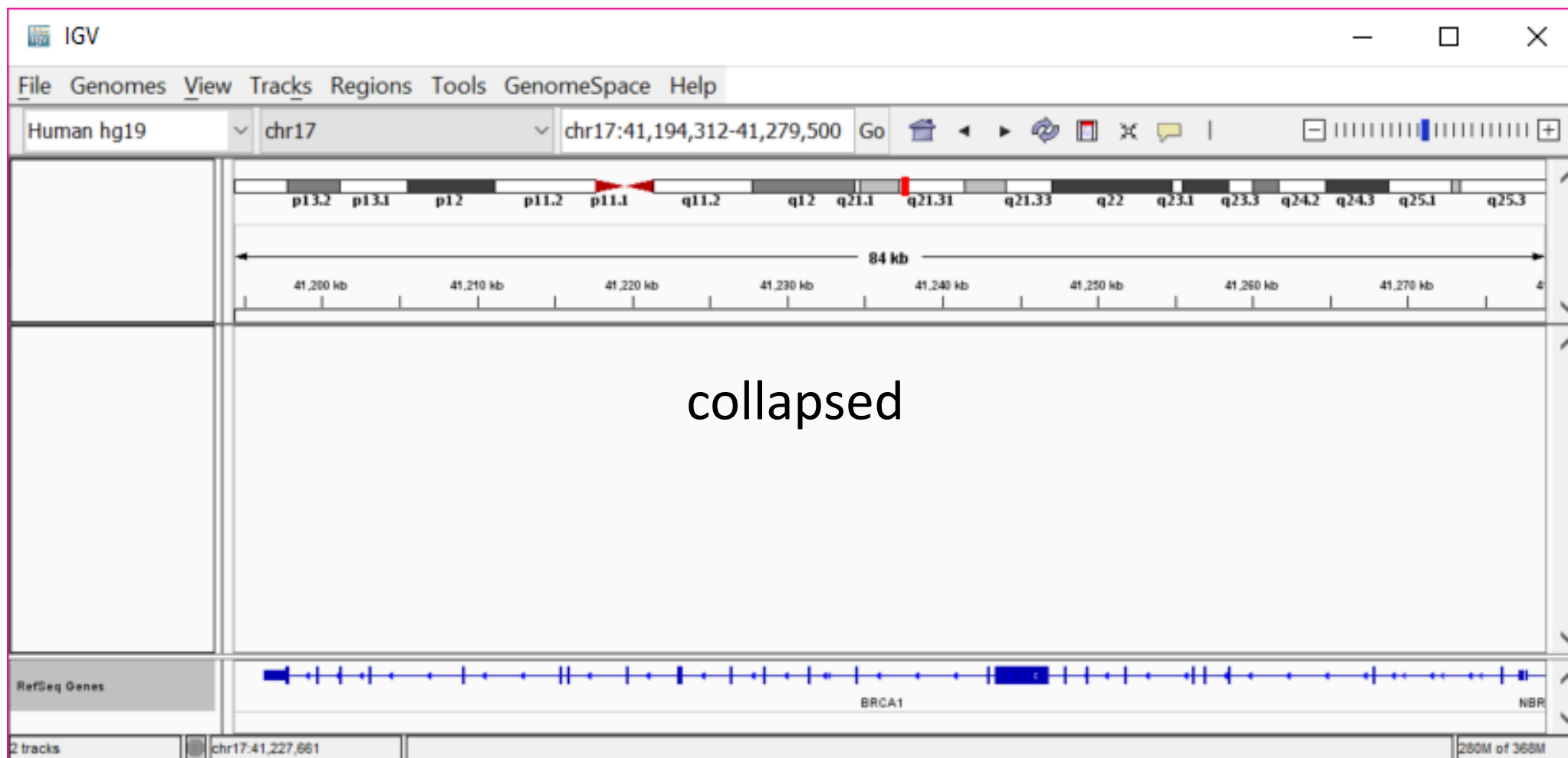
IGV: интерфейс



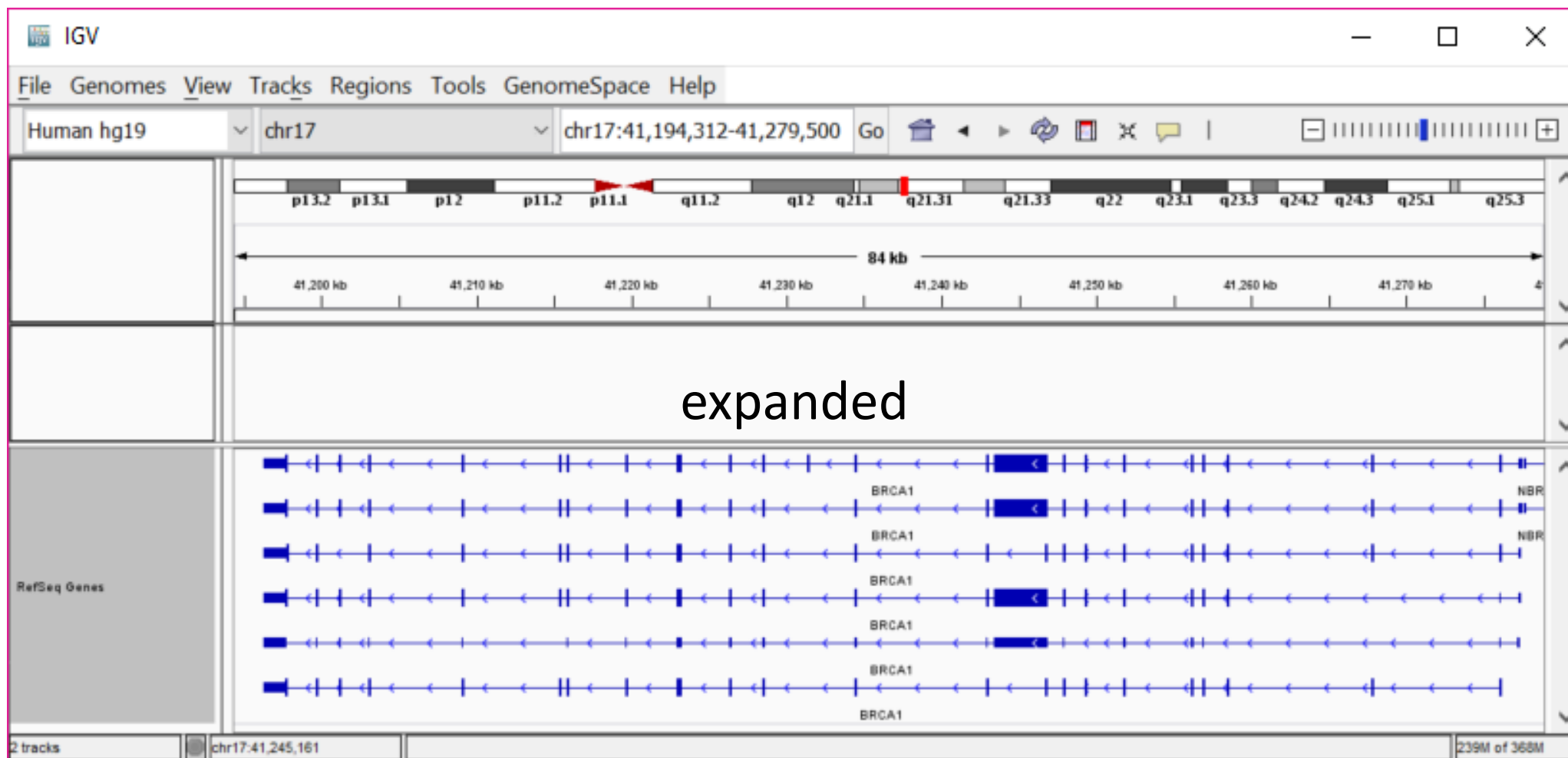
IGV: интерфейс



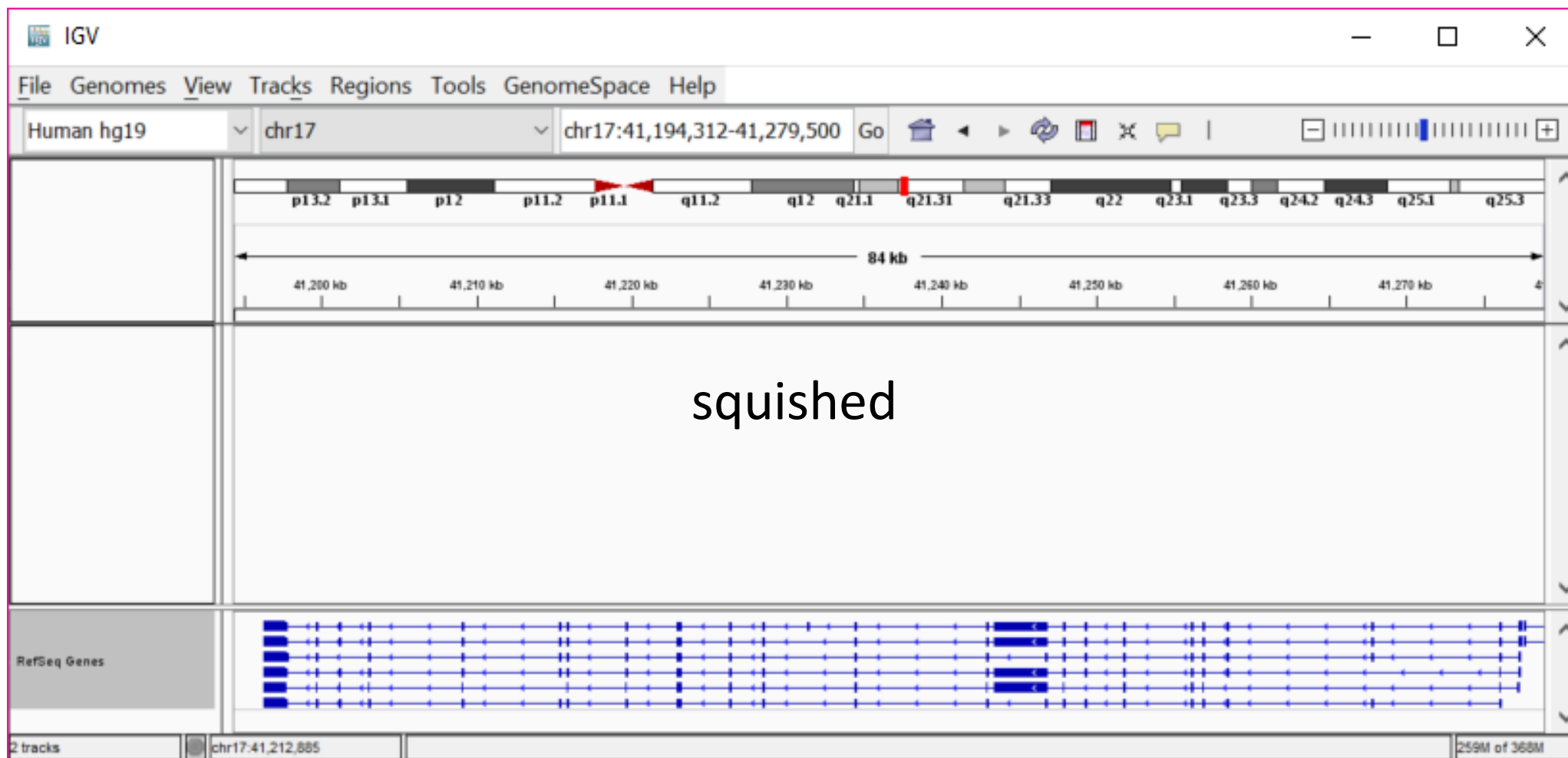
IGV: интерфейс



IGV: интерфейс



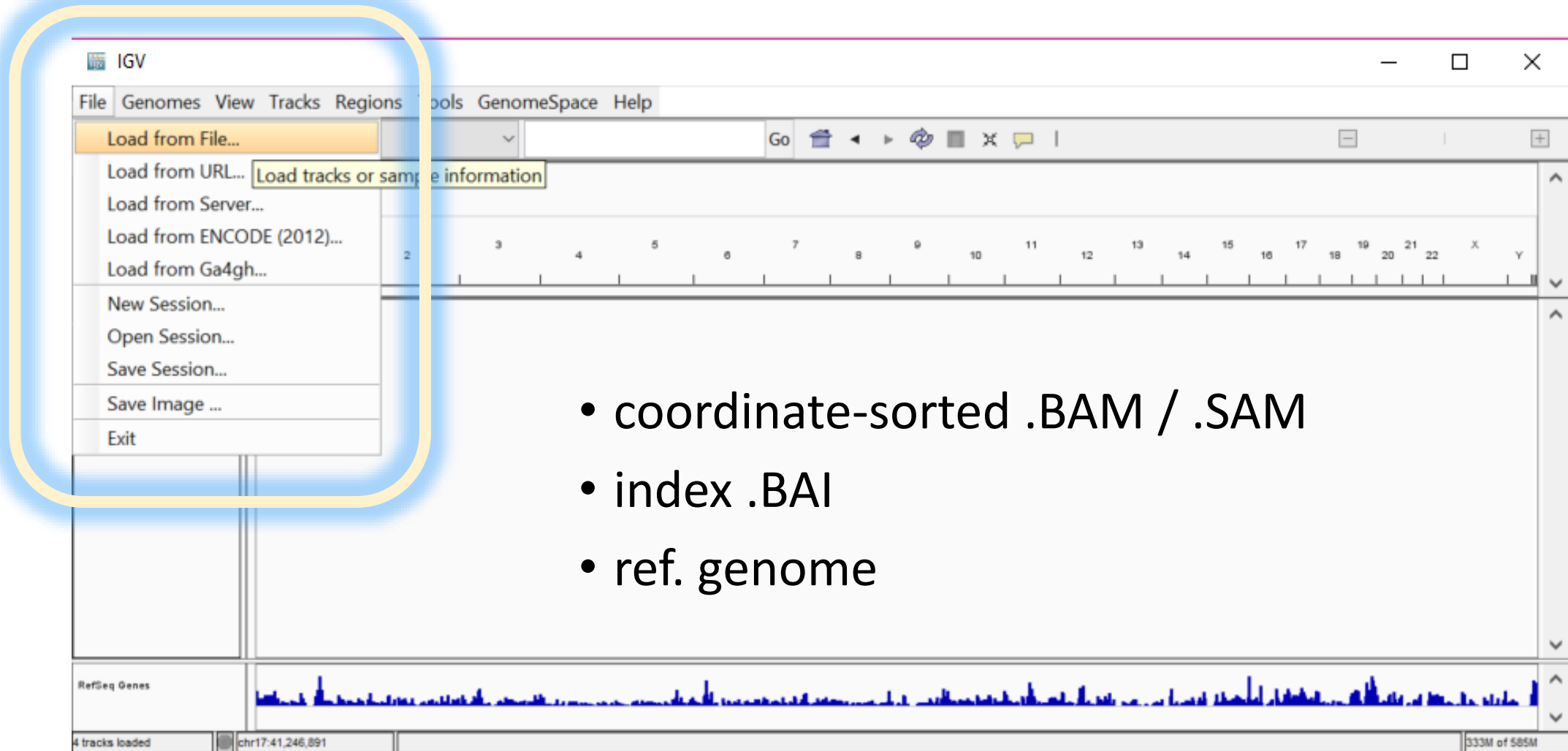
IGV: интерфейс



IGV: интерфейс

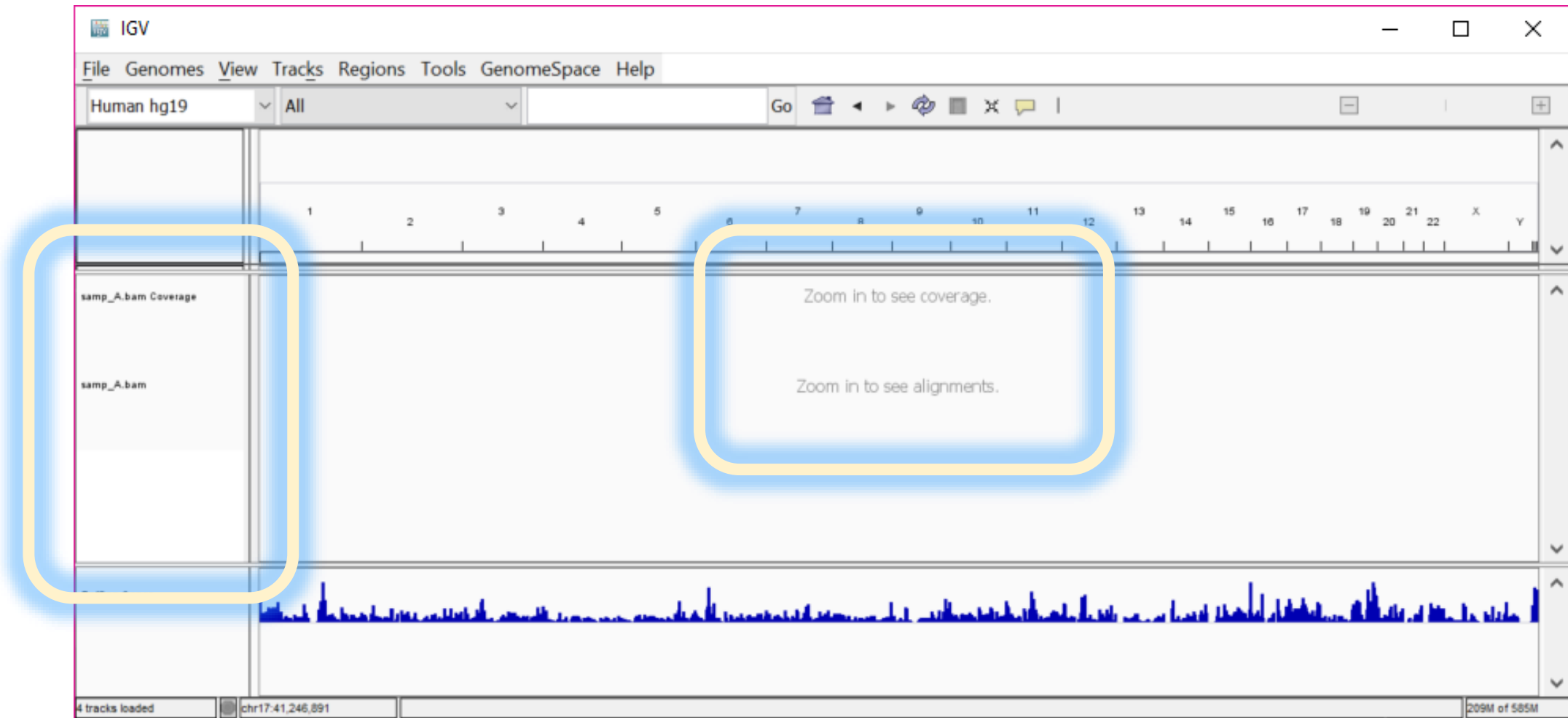


IGV: **samp_A** [open file]

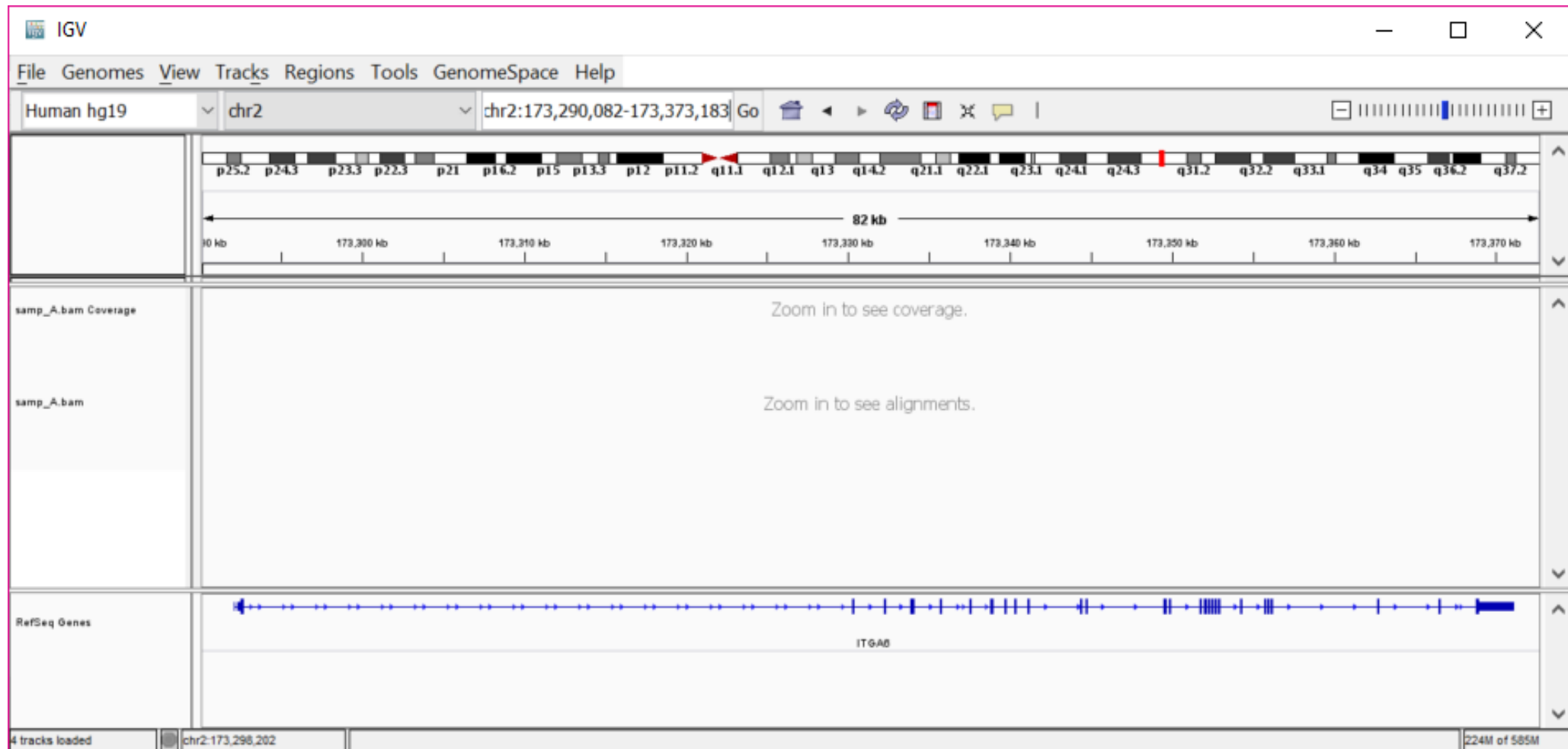


- coordinate-sorted .BAM / .SAM
- index .BAI
- ref. genome

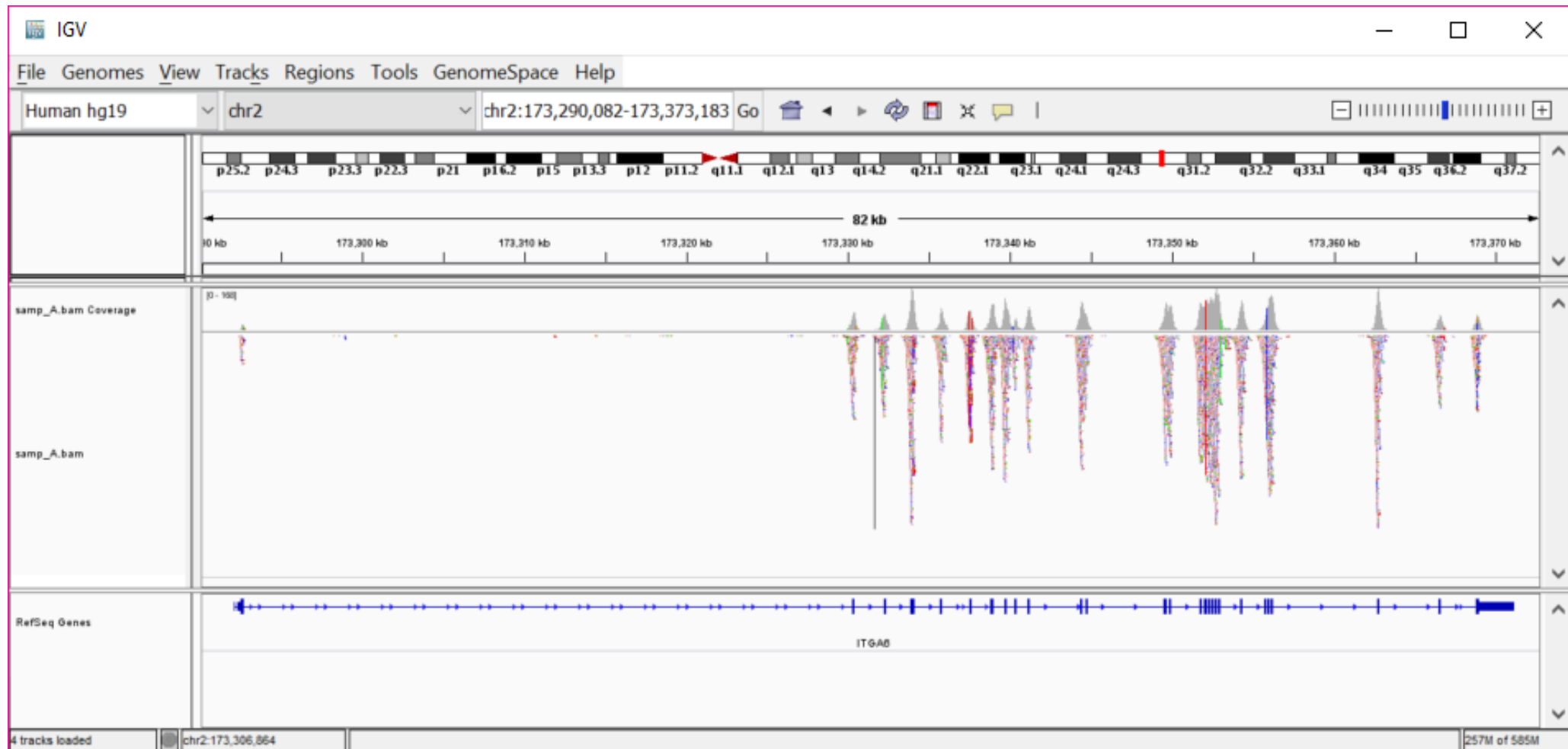
IGV: samp_A



IGV: **samp_A** [go to ITGA6]



IGV: **samp_A** [go to ITGA6]



IGV: **samp_A** [visibility range 30 > 100]

The screenshot displays the IGV interface with the 'Preferences' dialog box open for the 'Alignments' track. The 'Alignments' track is highlighted in blue. The 'Preferences' dialog box is open, showing various options for the 'Alignments' track. The 'Visibility range threshold (bp)' is set to 100. The 'Mapping quality threshold' is set to 0. The 'Coverage Track Options' section shows 'Coverage allele-fraction threshold' set to 0.2 and 'Quality weight allele fraction' checked. The 'Splice Junction Track Options' section shows 'Show flanking regions' unchecked, 'Min flanking width' set to 0, and 'Min junction coverage' set to 1. The 'Insert Size Options' section shows 'Compute' checked, 'Minimum (bp)' set to 50, 'Maximum (bp)' set to 1000, 'Minimum (percentile)' set to 0.5, and 'Maximum (percentile)' set to 99.5. The 'OK' button is highlighted in blue.

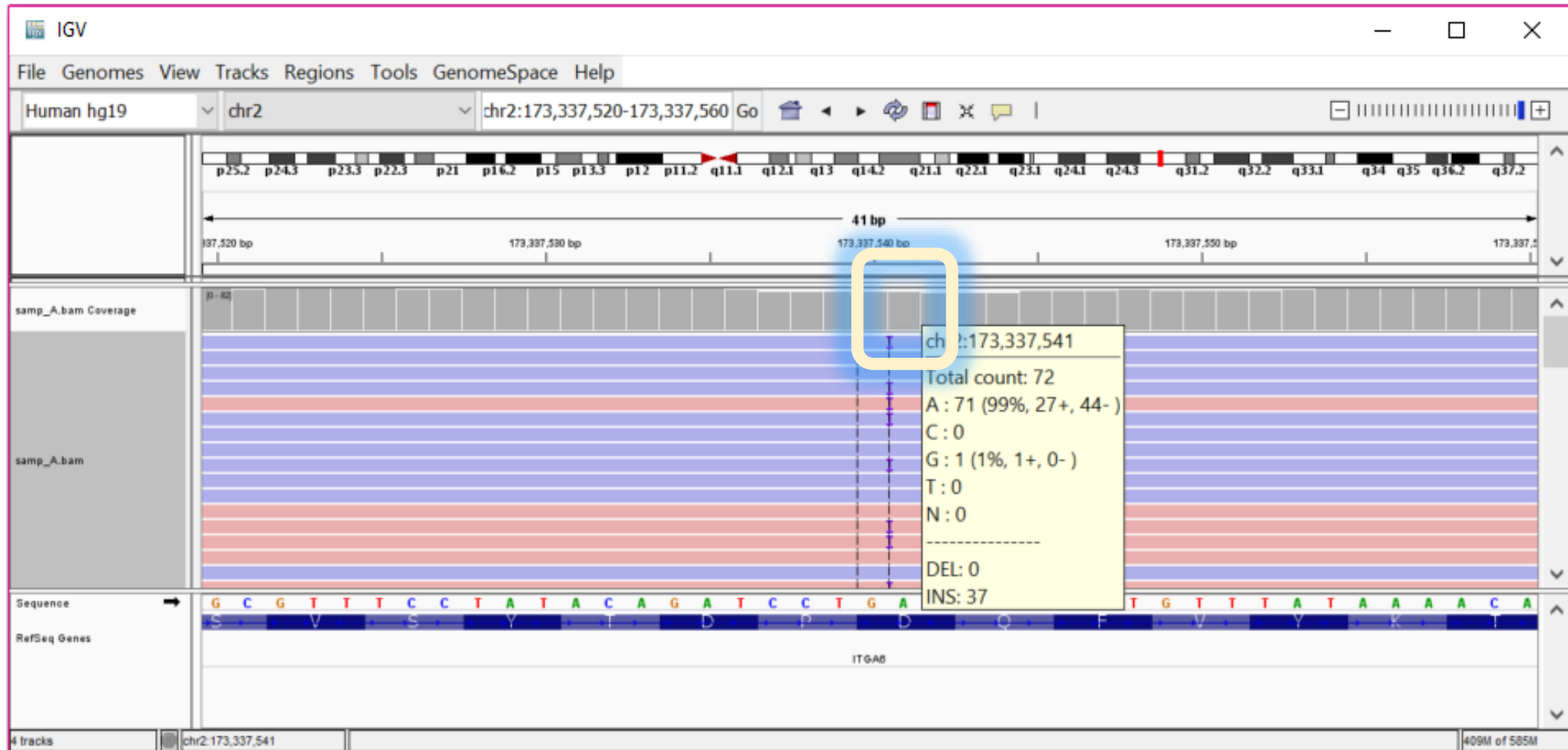
IGV: **samp_A** [chr2:173337540 InsA]

1	Chr	Start	End	Ref	Alt	Func.refG	Gene.refG
2	chr2	173292709	173292709	A	G	intronic	ITGA6
3	chr2	173292713	173292713	C	T	intronic	ITGA6
4	chr2	173330549	173330549	T	G	intronic	ITGA6
5	chr2	173332115	173332115	G	A	intronic	ITGA6
6	chr2	173334161	173334161	-	ATGATTTAGTACC	intronic	ITGA6
7	chr2	173337495	173337495	C	T	intronic	ITGA6
8	chr2	173337540	173337540	-	A	exonic	ITGA6
9	chr2	173337681	173337681	G	T	intronic	ITGA6
10	chr2	173340198	173340198	A	C	intronic	ITGA6
11	chr2	173352103	173352103	C	T	exonic	ITGA6
12	chr2	173353060	173353060	G	A	intronic	ITGA6
13	chr2	173353318	173353318	G	A	intronic	ITGA6
14	chr2	173355907	173355907	A	C	intronic	ITGA6
15	chr2	173368897	173368897	G	C	exonic	ITGA6

IGV: **samp_A** [chr2:173337540 InsA]



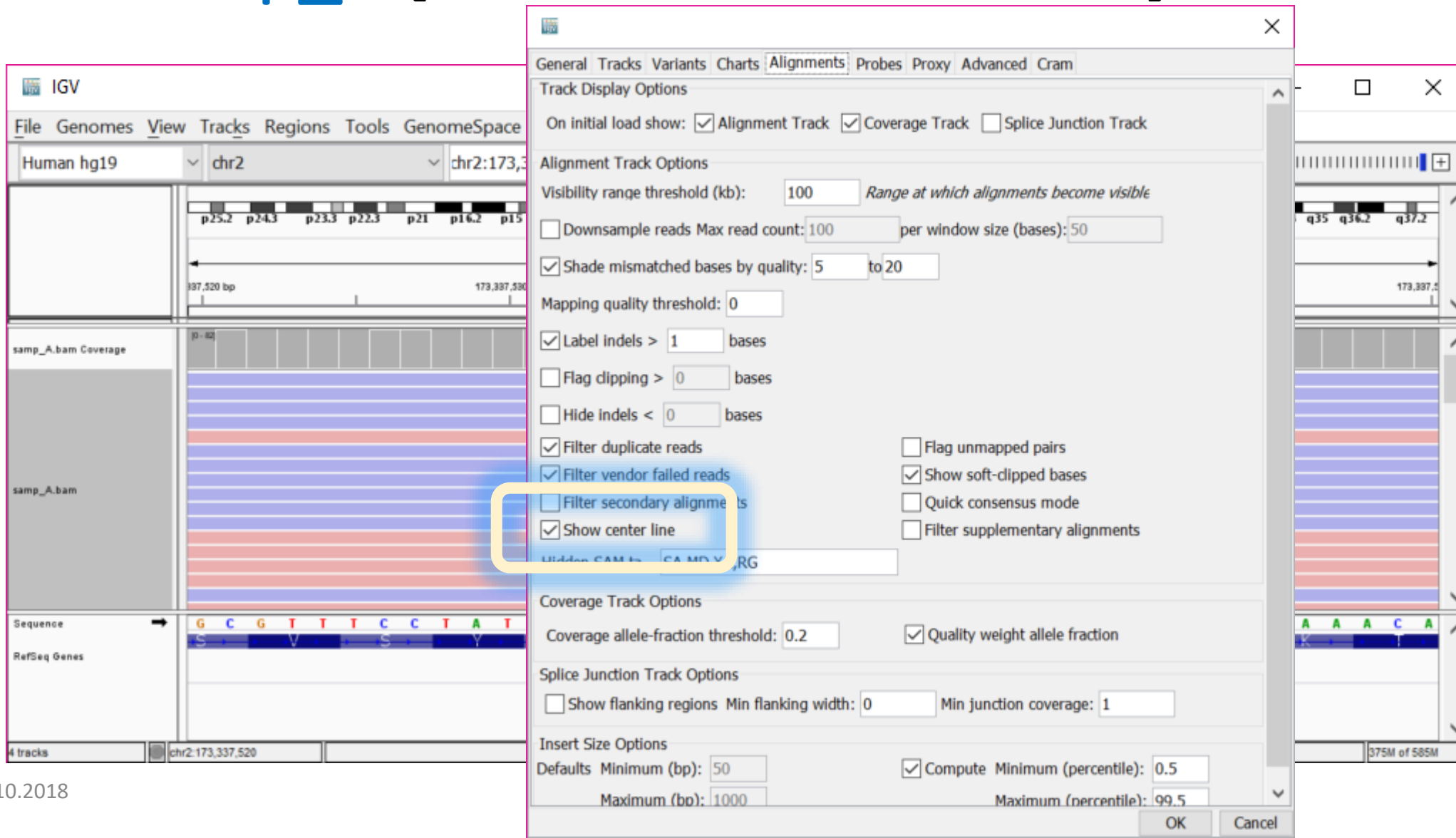
IGV: **samp_A** [chr2:173337540 InsA]



IGV: **samp_A** [chr2:173337540 InsA]



IGV: **samp_A** [chr2:173337540 InsA]



The screenshot displays the IGV interface with the 'Alignments' track selected. The 'Alignments' track options are visible, including 'Filter secondary alignments' and 'Show center line'. The 'Alignments' track is highlighted with a yellow box. The 'Alignments' track options are visible, including 'Filter secondary alignments' and 'Show center line'.

IGV

File Genomes View Tracks Regions Tools GenomeSpace

Human hg19 chr2 chr2:173,337,520

samp_A.bam Coverage

samp_A.bam

Sequence RefSeq Genes

4 tracks chr2:173,337,520

Alignments

General Tracks Variants Charts Alignments Probes Proxy Advanced Cram

Track Display Options

On initial load show: ☒ Alignment Track ☒ Coverage Track ☐ Splice Junction Track

Alignment Track Options

Visibility range threshold (kb): 100 Range at which alignments become visible

☐ Downsample reads Max read count: 100 per window size (bases): 50

☒ Shade mismatched bases by quality: 5 to 20

Mapping quality threshold: 0

☒ Label indels > 1 bases

☐ Flag clipping > 0 bases

☐ Hide indels < 0 bases

☒ Filter duplicate reads

☒ Filter vendor failed reads

☐ Filter secondary alignments

☒ Show center line

Hidden SAM tags: SAM MD, RG

Coverage Track Options

Coverage allele-fraction threshold: 0.2 ☒ Quality weight allele fraction

Splice Junction Track Options

☐ Show flanking regions Min flanking width: 0 Min junction coverage: 1

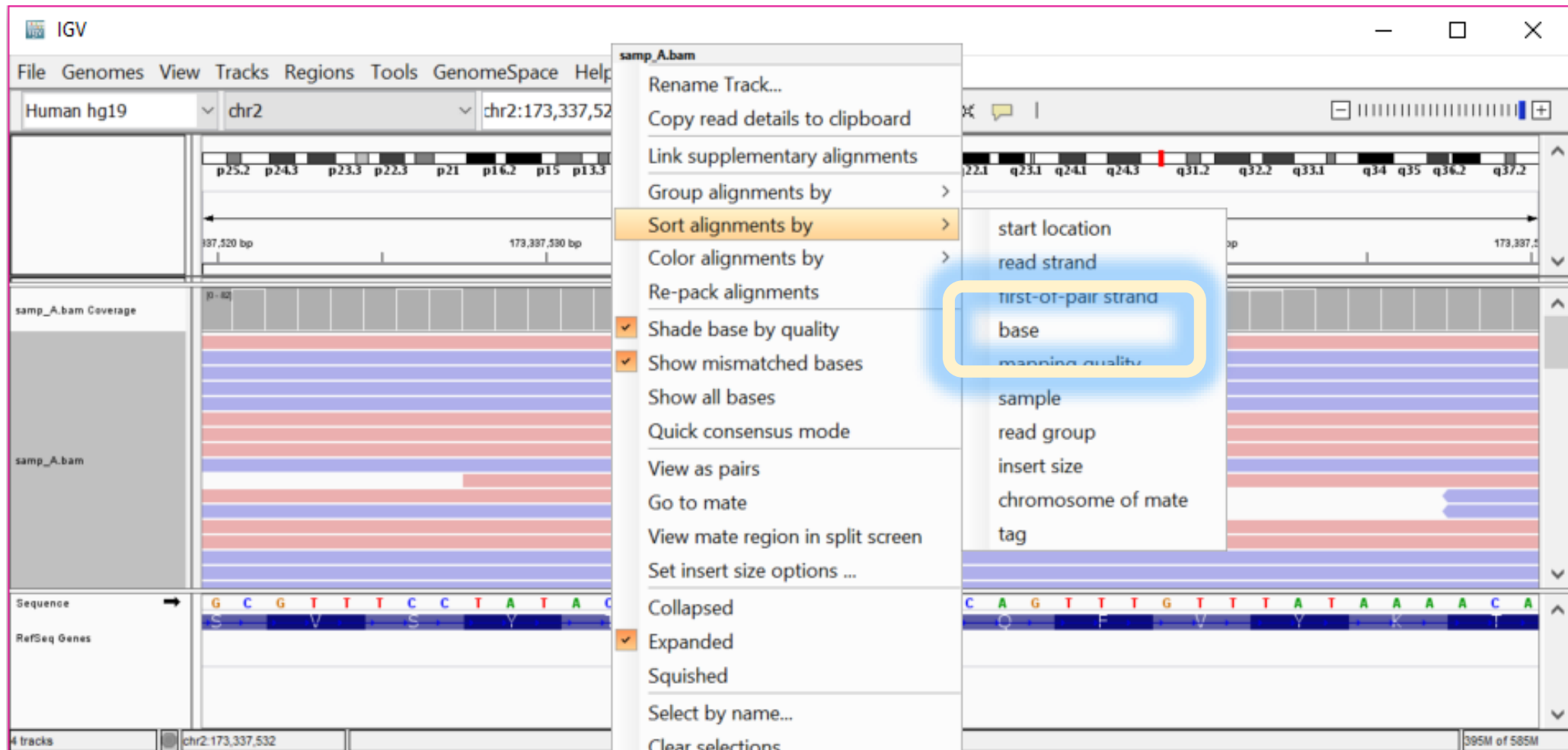
Insert Size Options

Defaults Minimum (bp): 50 Maximum (bp): 1000

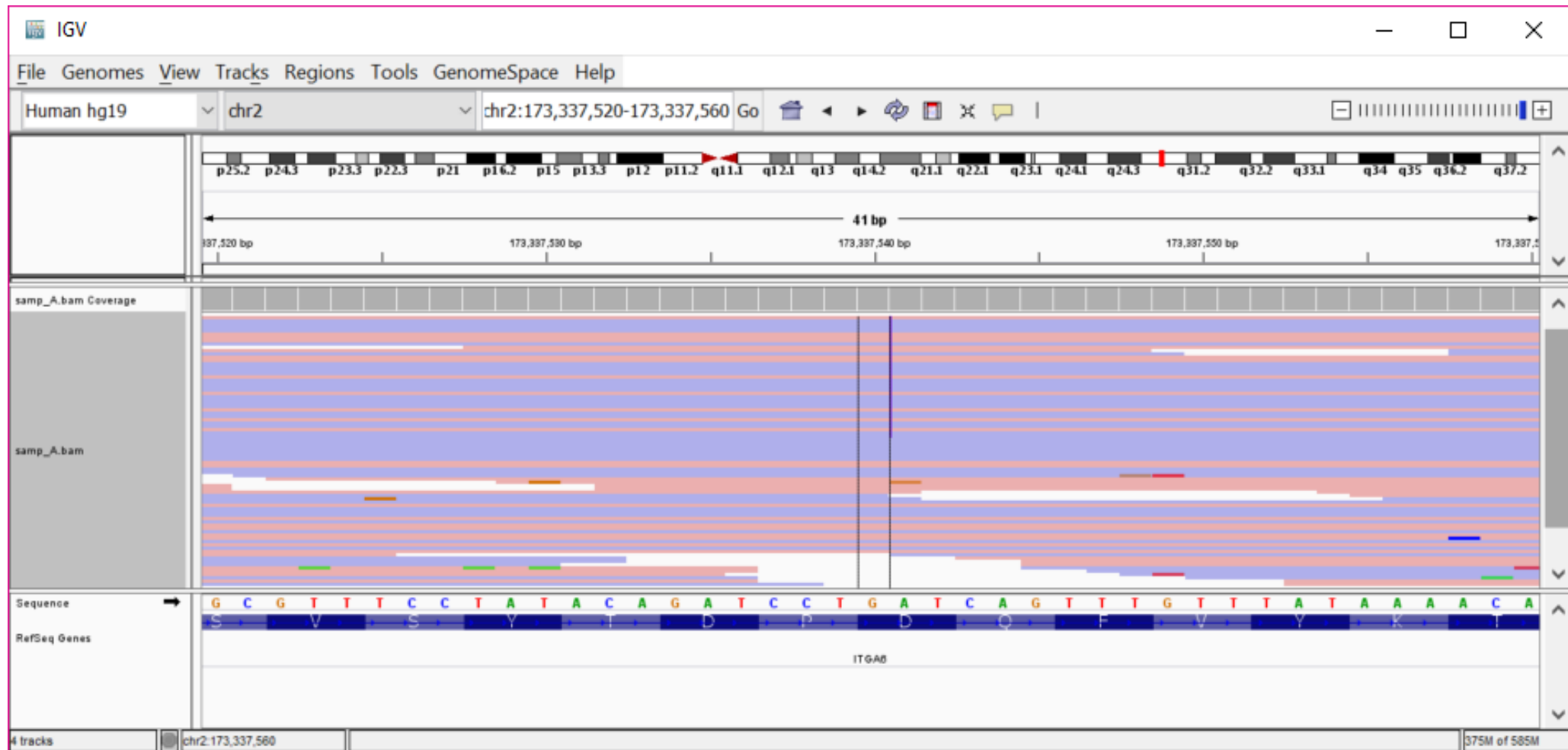
☒ Compute Minimum (percentile): 0.5 Maximum (percentile): 99.5

OK Cancel

IGV: **samp_A** [chr2:173337540 InsA]



IGV: **samp_A** [chr2:173337540 InsA]



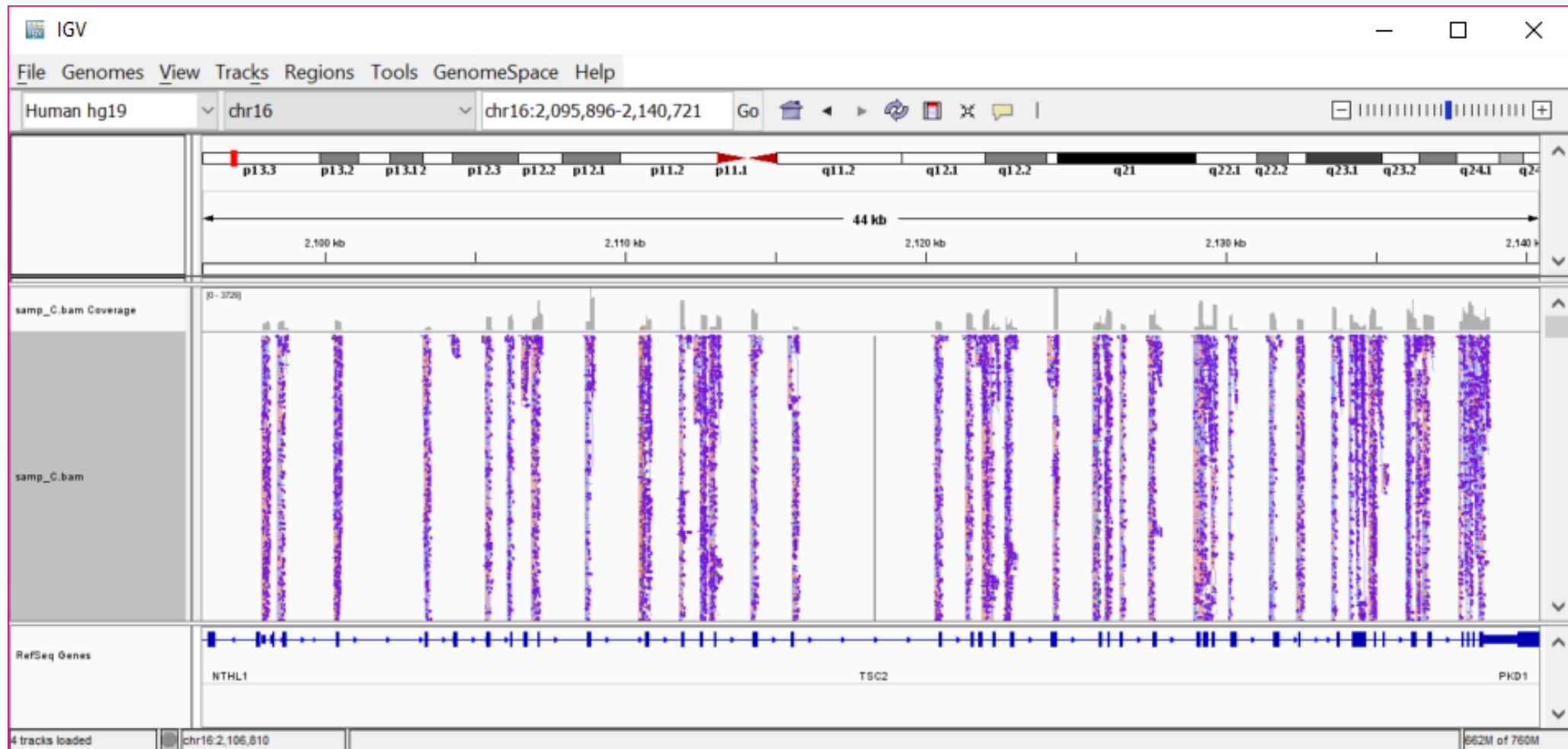
IGV: **samp_A** [chr2:173368897 G>C]

1	Chr	Start	End	Ref	Alt	Func.refG	Gene.refG
2	chr2	173292709	173292709	A	G	intronic	ITGA6
3	chr2	173292713	173292713	C	T	intronic	ITGA6
4	chr2	173330549	173330549	T	G	intronic	ITGA6
5	chr2	173332115	173332115	G	A	intronic	ITGA6
6	chr2	173334161	173334161	-	ATGATTTAGTACC	intronic	ITGA6
7	chr2	173337495	173337495	C	T	intronic	ITGA6
8	chr2	173337540	173337540	-	A	exonic	ITGA6
9	chr2	173337681	173337681	G	T	intronic	ITGA6
10	chr2	173340198	173340198	A	C	intronic	ITGA6
11	chr2	173352103	173352103	C	T	exonic	ITGA6
12	chr2	173353060	173353060	G	A	intronic	ITGA6
13	chr2	173353318	173353318	G	A	intronic	ITGA6
14	chr2	173355907	173355907	A	C	intronic	ITGA6
15	chr2	173368897	173368897	G	C	exonic	ITGA6

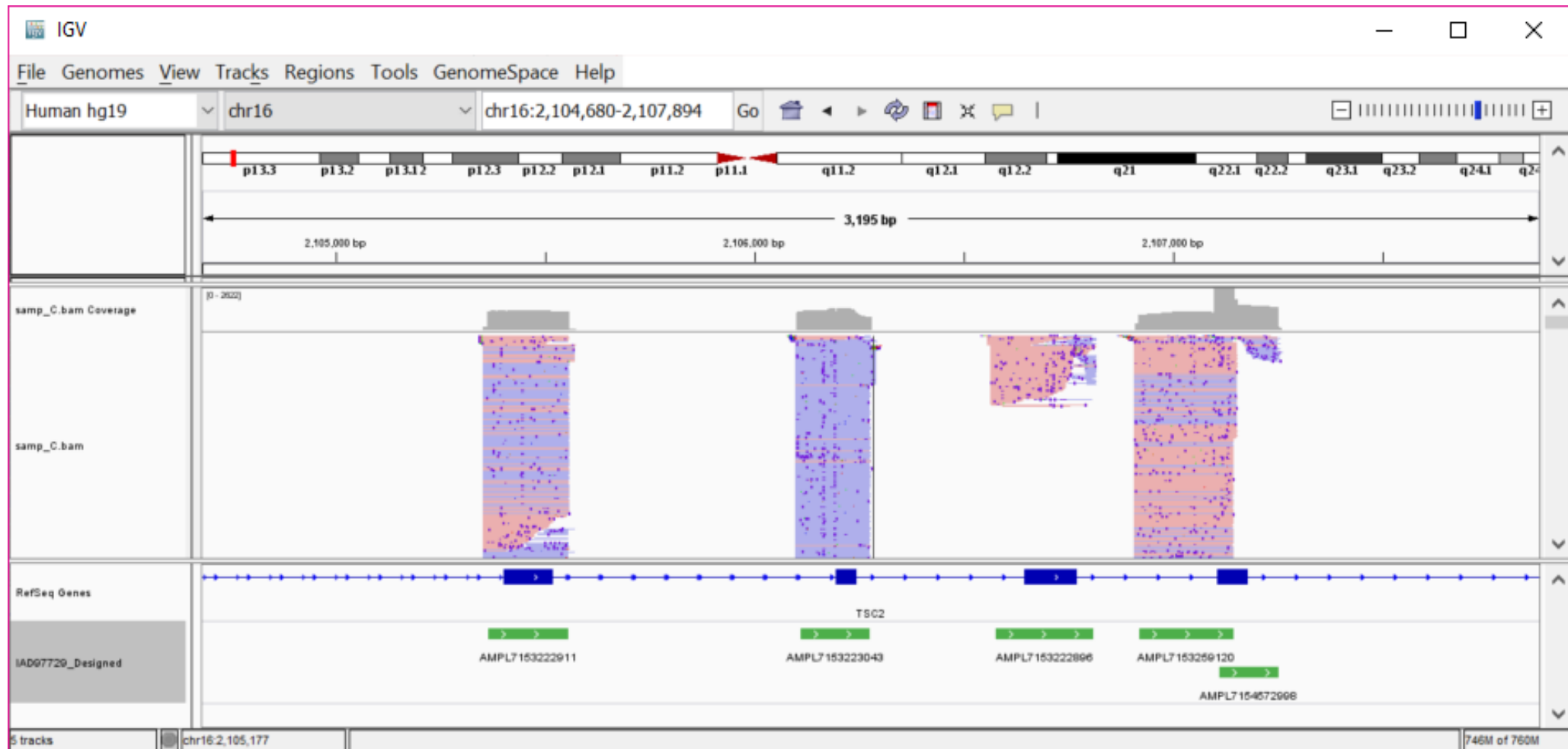
IGV: **samp_A** [chr2:173368897 G>C]



IGV: **samp_C** [TSC 2]



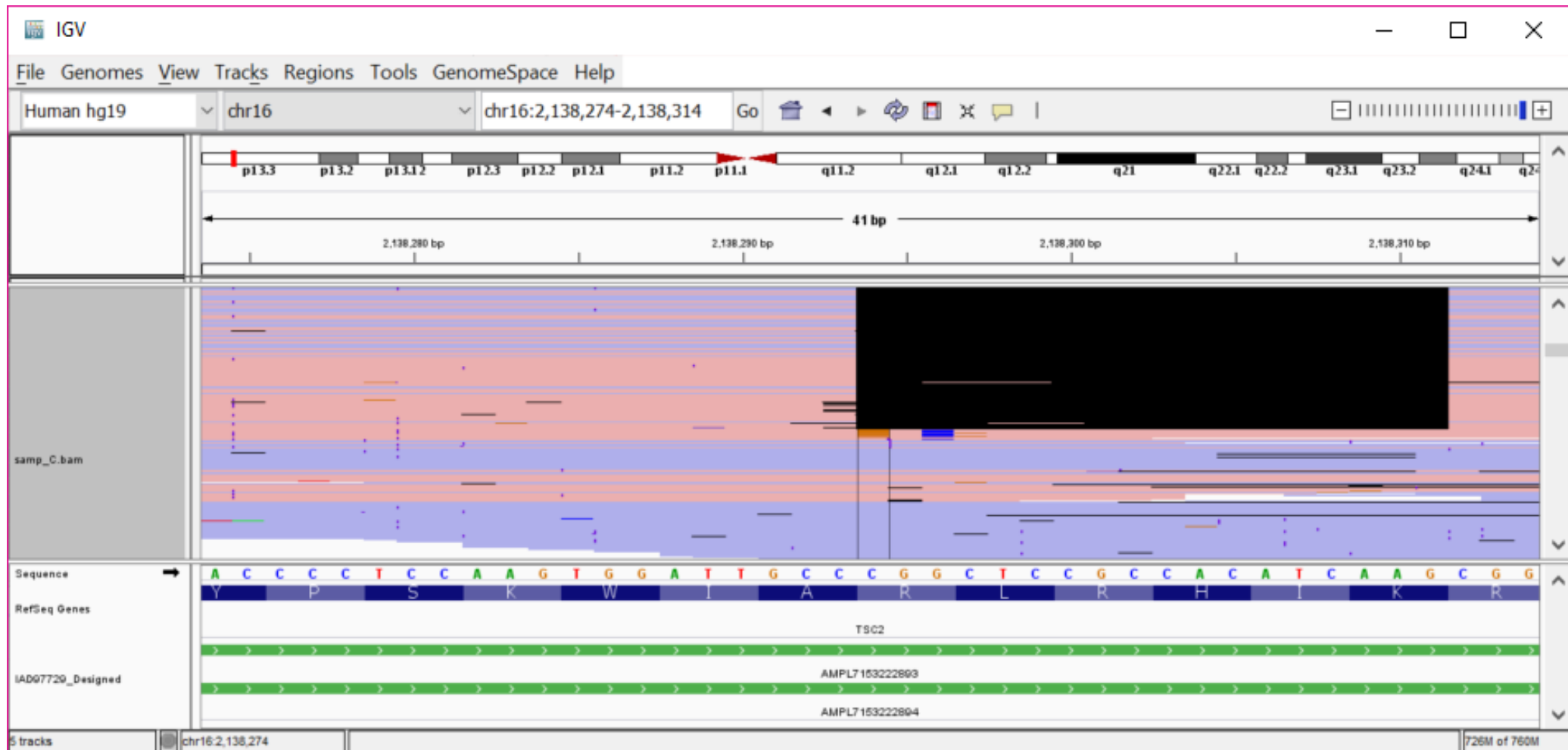
IGV: **samp_C** [BAM + BED]



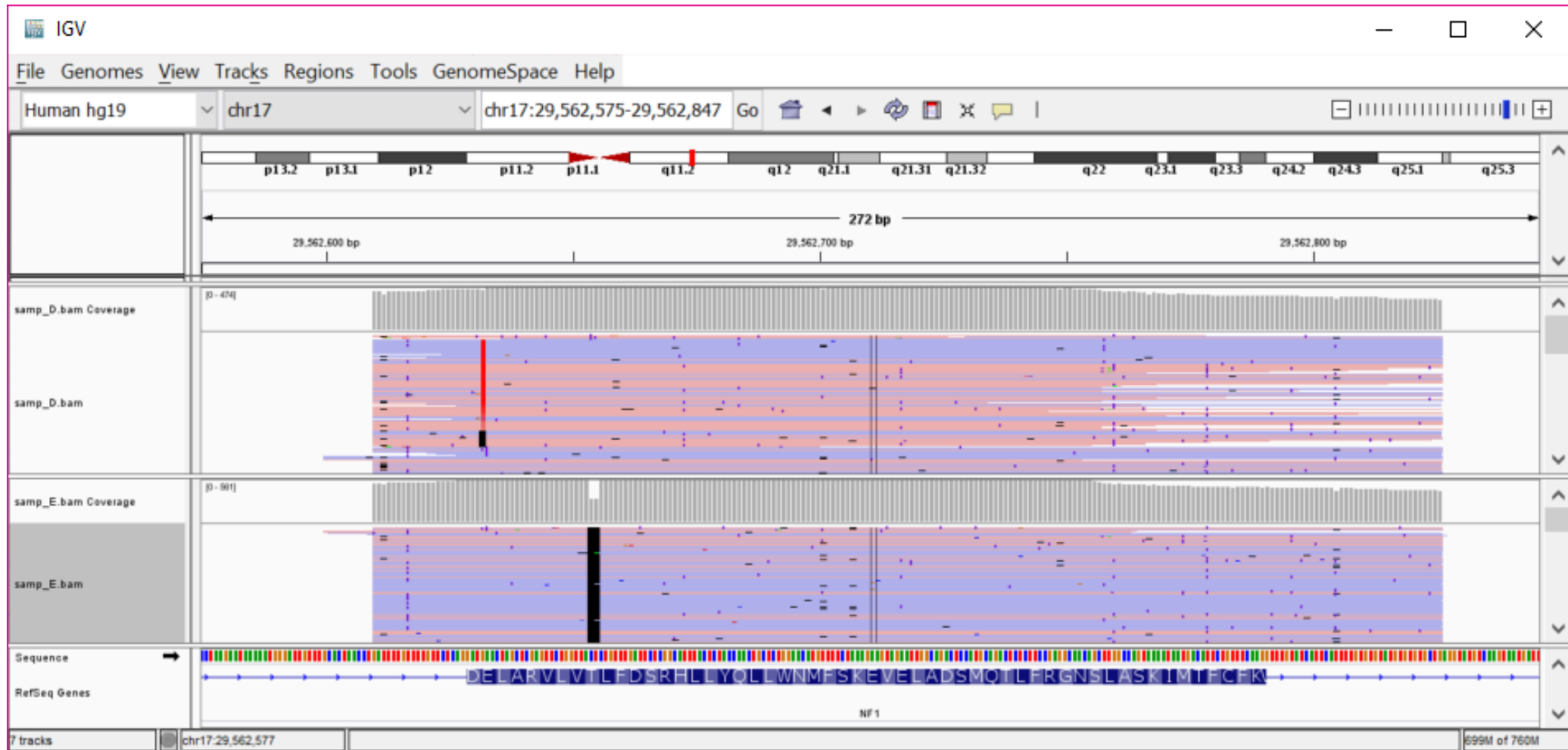
IGV: **samp_C** [chr16:2138294 Del_18bp]

1	Chr	Start	End	Ref	Alt	Func.refGene	Gene.refGene
2	chr16	2110571	2110571	C	G	intronic	TSC2
3	chr16	2125769	2125769	G	A	intronic	TSC2
4	chr16	2138084	2138084	A	C	exonic	TSC2
5	chr16	2138087	2138087	G	-	exonic	TSC2
6	chr16	2138086	2138086	C	G	exonic	TSC2
7	chr16	2138294	2138311	CGGCTCCGCCACATCAAG	-	exonic	TSC2

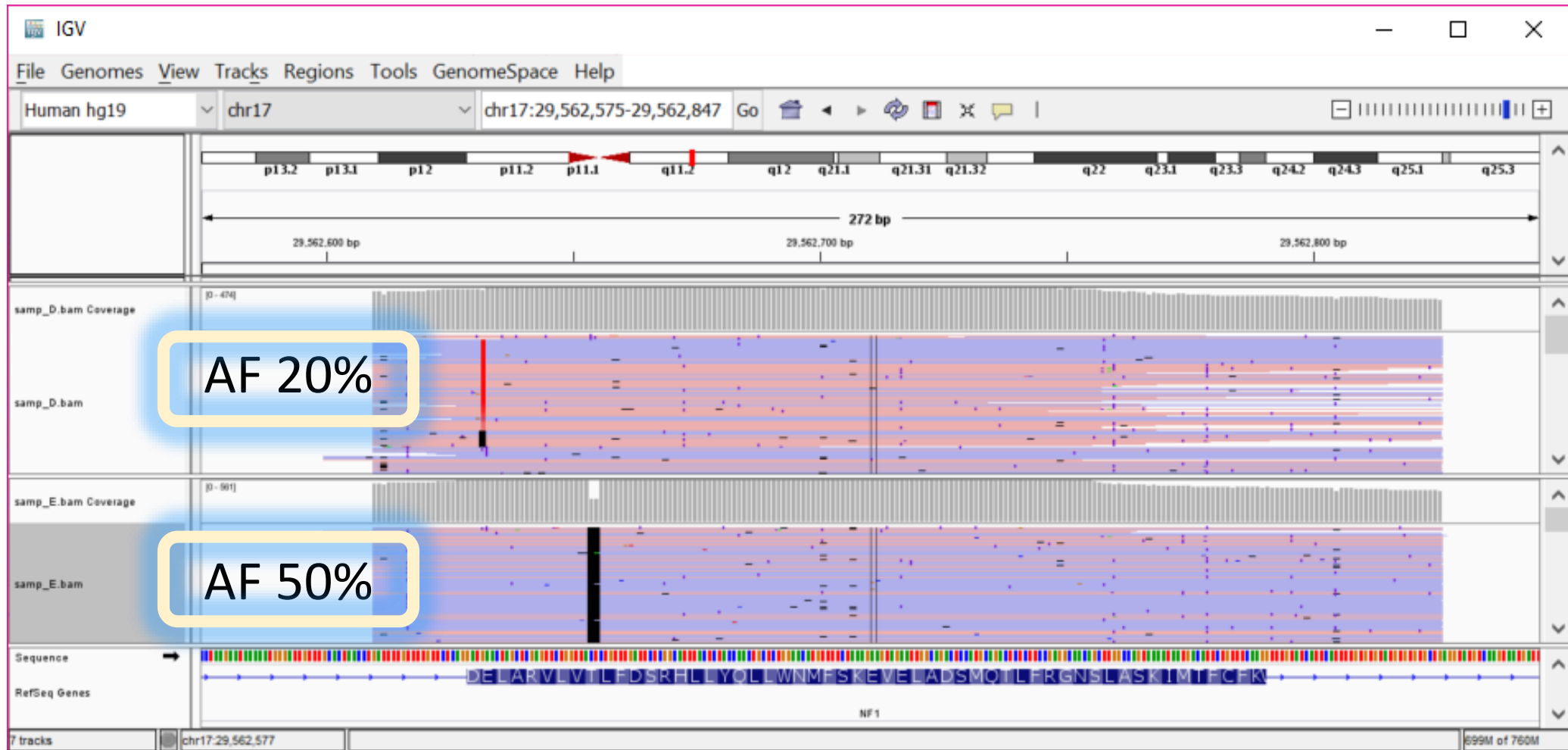
IGV: samp_C [del]



IGV: samp_D & samp_E [NF1 exon28]



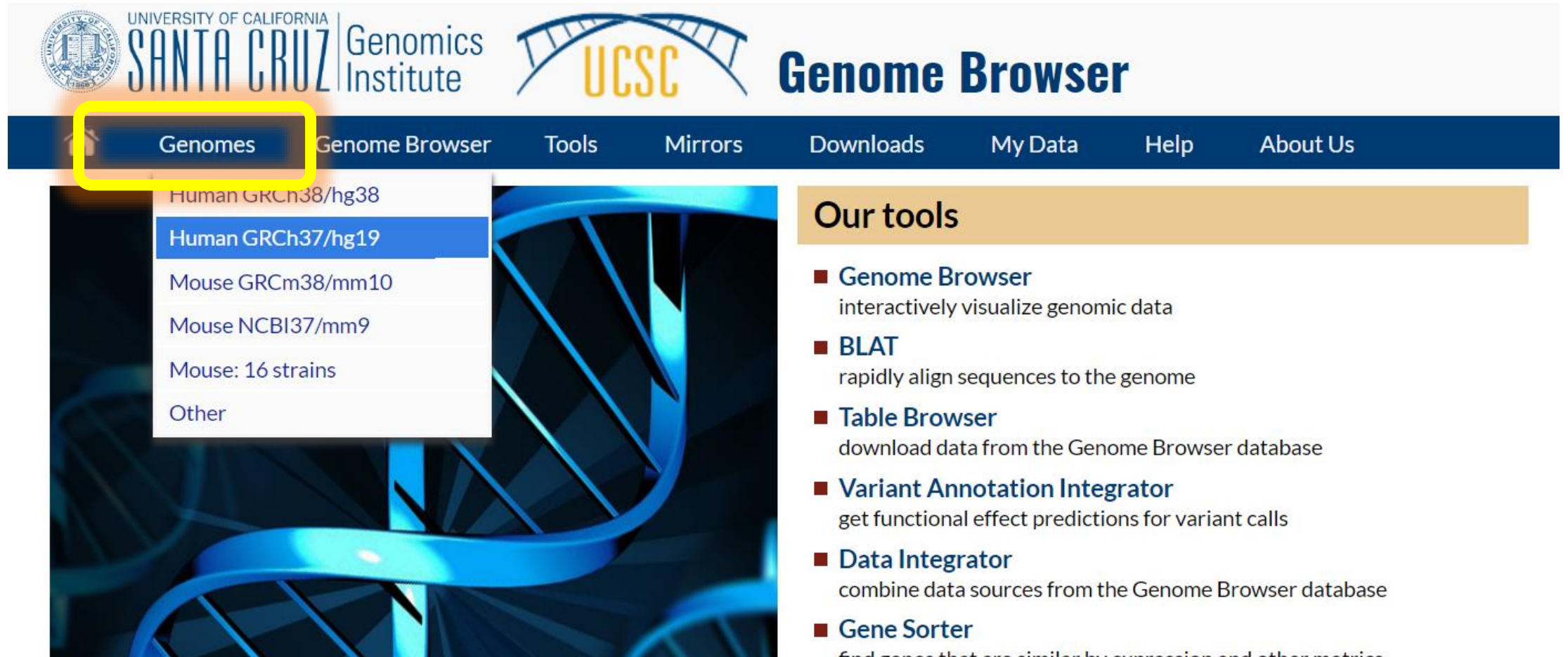
IGV: **samp_D** & **samp_E** [allele frequency]



UCSC Genome Browser

- <https://genome.ucsc.edu/>

UCSC Genome Browser



UNIVERSITY OF CALIFORNIA
SANTA CRUZ Genomics
Institute

UCSC

Genome Browser

Genomes Genome Browser Tools Mirrors Downloads My Data Help About Us

- Human GRCh38/hg38
- Human GRCh37/hg19**
- Mouse GRCm38/mm10
- Mouse NCBI37/mm9
- Mouse: 16 strains
- Other

Our tools

- **Genome Browser**
interactively visualize genomic data
- **BLAT**
rapidly align sequences to the genome
- **Table Browser**
download data from the Genome Browser database
- **Variant Annotation Integrator**
get functional effect predictions for variant calls
- **Data Integrator**
combine data sources from the Genome Browser database
- **Gene Sorter**
find genes that are similar by expression and other metrics

[illegible]

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<https://genome.ucsc.edu/>
 UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly



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<https://genome.ucsc.edu/>
 UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly

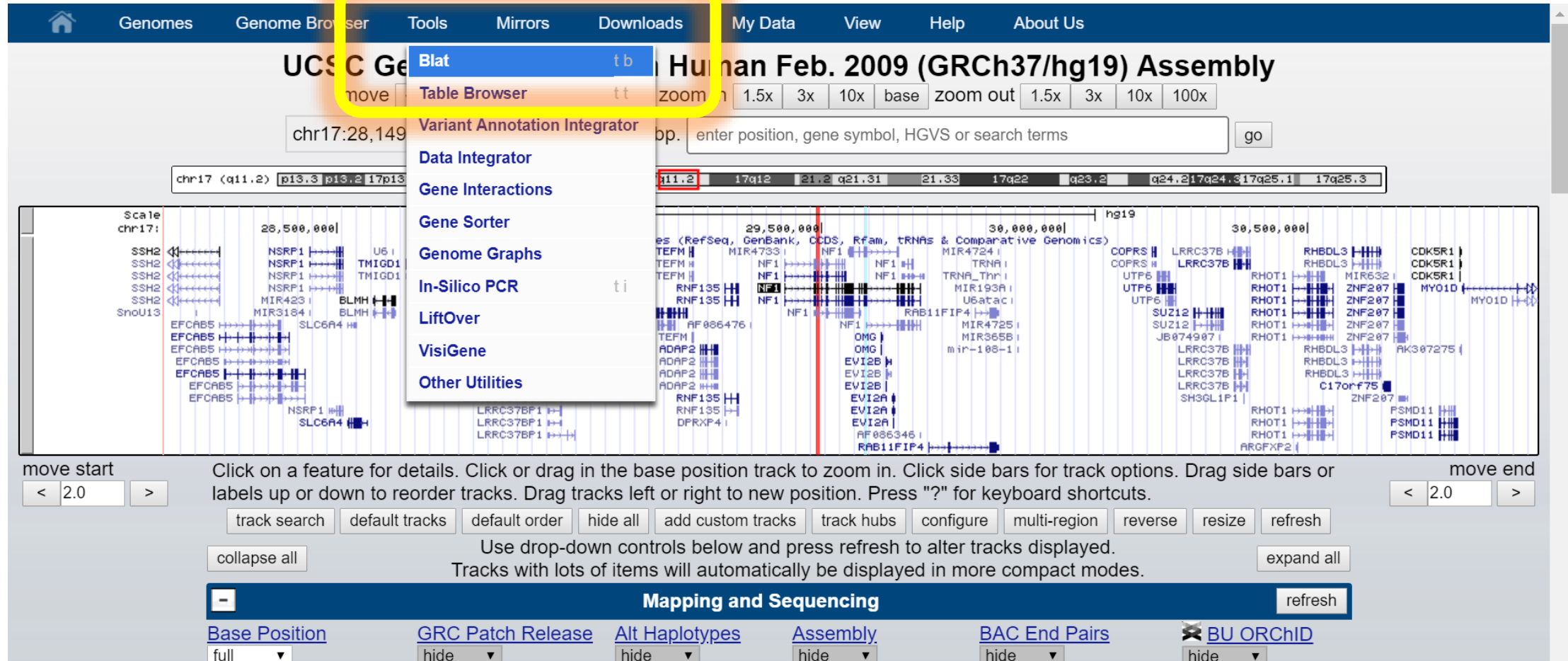


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 UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly



UCSC Genome Browser



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Human BLAT Search

BLAT Search Genome

Genome: Assembly: Query type: Sort output: Output type:

```
GATTTTGTTCAGGATTAAGTCTCGAGTTCTGGTTACTCTGTTTATTCTCGGCATTTACTCTACCAACTGCT
CTGGAACATGTTTCTAAAGAAGTAGAATTGGCAGACTCCATGCAGACTCTCTCCGAGGCAACAGCTTGCCAGTAAAT
AATGACATTCTGTTTCAAGGTTGTATCATTCTTTGTGTGTATGTGTGTGCTG
```

Paste in a query sequence to find its location in the genome. Multiple sequences may be searched if separated by lines starting with '>' followed by the sequence name.

File Upload: Rather than pasting a sequence, you can choose to upload a text file containing the sequence.

UCSC Genome Browser

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Human BLAT Results

BLAT Search Results

Go back to [chr17:29562610-29562826](#) on the Genome Browser.

Custom track name:

Custom track description:

ACTIONS	QUERY	SCORE	START	END	QSIZE	IDENTITY	CHRO	STRAND	START	END	SPAN
browser details	YourSeq	215	1	217	217	99.6%	17	+	29562610	29562826	217
browser details	YourSeq	189	1	217	217	94.0%	15	-	22143079	22143297	219
browser details	YourSeq	187	1	217	217	93.5%	15	-	21131902	21132120	219
browser details	YourSeq	26	179	210	217	96.5%	5	-	178118008	178118051	44
browser details	YourSeq	24	1	25	217	100.0%	12	+	21087736	21088232	497
browser details	YourSeq	22	179	201	217	100.0%	X	+	110987110	110987134	25
browser details	YourSeq	22	194	215	217	100.0%	5	+	149668850	149668871	22

UCSC Genome Browser

UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x

chr17:29,562,594-29,562,840 247 bp.

chr17 (q11.2) p13.3 p13.2 17p13.1 17p12 17p11.2 17q11.2 17q12 21.2 q21.31 21.33 17q22 q23.2 q24.2 17q24.3 17q25.1 17q25.3

Scale chr17: 100 bases

29,562,650 29,562,700 29,562,750 29,562,800 hg19

UCSC Genes (RefSeq, GenBank, CCDS, Rfam, tRNAs & Comparative Genomics)

NF1 NF1 NF1

YourSeq Your Sequence from Blat Search

move start Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options. Drag side bars or move end labels up or down to reorder tracks. Drag tracks left or right to new position. Press "?" for keyboard shortcuts. move end

< 2.0 >

track search default tracks default order hide all add custom tracks track hubs configure multi-region reverse resize refresh

collapse all Use drop-down controls below and press refresh to alter tracks displayed. expand all

Tracks with lots of items will automatically be displayed in more compact modes.

Mapping and Sequencing

Base Position full ▾	GRC Patch Release hide ▾	Alt Haplotypes hide ▾	Assembly hide ▾	BAC End Pairs hide ▾	BU ORChID hide ▾
Chromosome Band hide ▾	deCODE Recomb hide ▾	ENCODE Pilot [No data-chr17]	FISH Clones hide ▾	Fosmid End Pairs hide ▾	Gap hide ▾
GC Percent hide ▾	GRC Incident hide ▾	GRC Map Contigs hide ▾	Hg18 Diff hide ▾	Hg38 Diff hide ▾	Hi Seq Depth hide ▾

UCSC Genome Browser

The screenshot displays the UCSC Genome Browser interface for the human genome assembly (GRCh37/hg19). The top navigation bar includes links for Genomes, Genome Browser, Tools, Mirrors, Downloads, My Data, View, Help, and About Us. The main title is "UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly". Below the title, there are navigation controls for moving and zooming. The current view is centered on chromosome 17, specifically the region chr17:29,562,594-29,562,840 (247 bp). A search bar is present with the text "enter position, gene symbol, HGVS or search terms" and a "go" button. The main display area shows a genomic track with a yellow box highlighting a specific region. Below the track, there are instructions for using the browser, including "Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position. Press '?' for keyboard shortcuts." and a "refresh" button. The bottom section, titled "Mapping and Sequencing", contains a grid of track options with dropdown menus for each track.

UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x

chr17:29,562,594-29,562,840 247 bp. enter position, gene symbol, HGVS or search terms go

chr17 (q11.2) p13.3 p13.2 17p13.1 17p12 17p11.2 17q11.2 17q12 21.2 q21.31 21.33 17q22 q23.2 q24.2 17q24.3 17q25.1 17q25.3

Scale chr17: 100 bases

29,562,650 29,562,700 29,562,750 hg19 29,562,800

UCSC Genes (RefSeq, GenBank, CCDS, Rfam, tRNAs & Comparative Genomics)

NF1 NF1 NF1

YourSeq Your Sequence from Blat Search

move start < 2.0 > move end < 2.0 >

Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position. Press "?" for keyboard shortcuts.

track search default tracks default order hide all add custom tracks track hubs configure multi-region reverse resize refresh

collapse all Use drop-down controls below and press refresh to alter tracks displayed. expand all

Tracks with lots of items will automatically be displayed in more compact modes.

Mapping and Sequencing refresh

Base Position full ▼	GRC Patch Release hide ▼	Alt Haplotypes hide ▼	Assembly hide ▼	BAC End Pairs hide ▼	BU ORChID hide ▼
Chromosome Band hide ▼	deCODE Recomb hide ▼	ENCODE Pilot [No data-chr17]	FISH Clones hide ▼	Fosmid End Pairs hide ▼	Gap hide ▼
GC Percent hide ▼	GRC Incident hide ▼	GRC Map Contigs hide ▼	Hg18 Diff hide ▼	Hg38 Diff hide ▼	Hi Seq Depth hide ▼

UCSC Genome Browser

UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x

chr17:29,562,626-29,562,637 12 bp. enter position, gene symbol, HGVS or search terms go

chr17 (q11.2) p13.3 p13.2 17p13.1 17p12 17p11.2 17q11.2 17q12 21.2 q21.31 21.33 17q22 q23.2 q24.2 17q24.3 17q25.1 17q25.3

Scale chr17: 29,562,626| 29,562,627| 29,562,628| 29,562,629| 29,562,630| 29,562,631| 29,562,632| 29,562,633| 29,562,634| 29,562,635| 29,562,636| hg19

UCSC Genes (RefSeq, GenBank, CCDS, Rfam, tRNAs & Comparative Genomics)

Base Position: full

Chromosome Band: hide

GC Percent: hide

GRC Patch Release: hide

deCODE Recomb: hide

GRC Incident: hide

Alt Haplotypes: hide

ENCODE Pilot: [No data-chr17]

GRC Map Contigs: hide

Assembly: hide

FISH Clones: hide

Hg18 Diff: hide

BAC End Pairs: hide

Fosmid End Pairs: hide

Hg38 Diff: hide

BU ORChID: hide

Gap: hide

Hi Seq Depth: hide

Mapping and Sequencing refresh

UCSC Genome Browser

Phenotype and Literature

Publications full ▼	ClinGen CNVs hide ▼	ClinVar Variants full ▼	Coriell CNVs hide ▼	COSMIC Regions hide ▼	DECIPHER CNVs hide ▼
DECIPHER SNVs hide ▼	Development Delay hide ▼	GAD View hide ▼	Gene Interactions hide ▼	GeneReviews hide ▼	GWAS Catalog hide ▼
HGMD Variants full ▼	Lens Patents hide ▼	LOVD Variants full ▼	¹⁸ MGI Mouse QTL hide ▼	OMIM Alleles full ▼	OMIM Genes hide ▼
OMIM Pheno Loci hide ▼	¹⁸ RGD Human QTL hide ▼	¹⁸ RGD Rat QTL hide ▼	SNPedia hide ▼	UniProt Variants hide ▼	Web Sequences hide ▼

refresh

UCSC Genome Browser

UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x

chr17:29,562,626-29,562,637 12 bp. enter position, gene symbol, HGVS or search terms go

chr17 (q11.2) p13.3 p13.2 17p13.1 17p12 17p11.2 17q11.2 17q12 21.2 q21.31 21.33 17q22 q23.2 q24.2 17q24.3 17q25.1 17q25.3

Scale chr17: 29,562,626| 29,562,627| 29,562,628| 29,562,629| 29,562,630| 29,562,631| 29,562,632| 29,562,633| 29,562,634| 29,562,635| 29,562,636| hg19

UCSC Genes (RefSeq, GenBank, CCDS) Comparative Genomics

NF1 D 1237 E 1238 L 1239

NF1 D 1237 E 1238 L 1239

NF1 D 1097 E 1098 L 1099

NF1 D 270 E 271 L 272

YourSeq Your Sequence from Blat Search

ClinVar Short Variants <= 100bp

Human Gene Mutation Database Public Variants

move start < 2.0 > move end < 2.0 >

track search default tracks default order hide all add custom tracks track hubs configure multi-region reverse resize refresh

collapse all expand all

Use drop-down controls below and press refresh to alter tracks displayed. Tracks with lots of items will automatically be displayed in more compact modes.

Mapping and Sequencing refresh

Base Position GRC Patch Release Alt Haplotypes Assembly BAC End Pairs BU ORCID

Спасибо за внимание

User guides

IGV

- <http://software.broadinstitute.org/software/igv/userguide>

UCSC

<https://genome.ucsc.edu/goldenpath/help/hgTracksHelp.html>