

Human Genome Sequencing and Interpretation

Lesson 1 - 20/01/2020

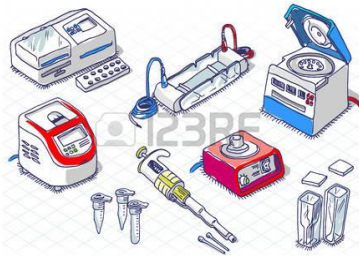
Lesson 2 - 21/01/2020

Lesson 3 - 27/01/2020

(Lesson 4 - 28/01/2020)

Prof. Massimo Delledonne
Functional Genomics lab

Library
preparation

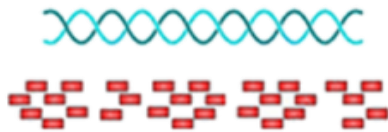


Sequencing

Bioinformatic
analysis

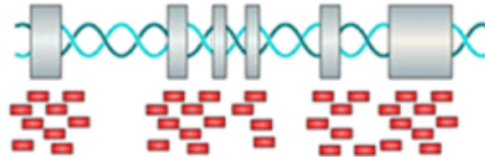


Whole genome sequencing



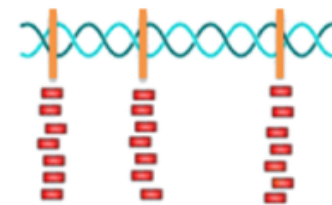
- Sequencing region : whole genome
- Sequencing Depth: >30X
- Covers everything – can identify all kinds of variants including SNPs, INDELs and SV.

Whole exome sequencing



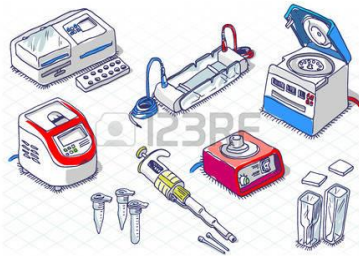
- Sequencing region: whole exome
- Sequencing Depth : >50X ~ 100X
- Identify all kinds of variants including SNPs, INDELs and SV in coding region.
- Cost effective

Targeted sequencing



- Sequencing region: specific regions (could be customized)
- Sequencing Depth : >500X
- Identify all kinds of variants including SNPs, INDELs in specific regions
- Most Cost effective

Library
preparation

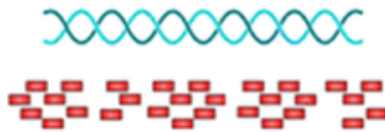


Sequencing

Bioinformatic
analysis

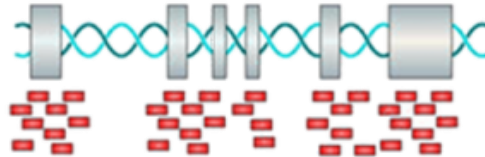


Whole genome sequencing



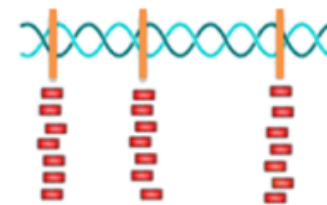
- Sequencing region : whole genome
- Sequencing Depth: >30X
- Covers everything – can identify all kinds of variants including SNPs, INDELs and SV.

Whole exome sequencing



- Sequencing region: whole exome
- Sequencing Depth : >50X ~ 100X
- Identify all kinds of variants including SNPs, INDELs and SV in coding region.
- Cost effective

Targeted sequencing

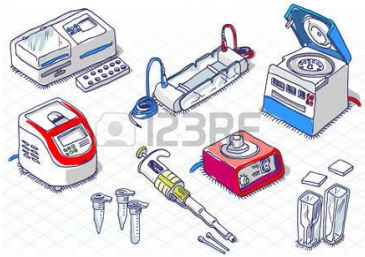


- Sequencing region: specific regions (could be customized)
- Sequencing Depth : >500X
- Identify all kinds of variants including SNPs, INDELs in specific regions
- Most Cost effective

Library
preparation



Bioinformatic
analysis



Sequencing



Data QC

Alignment

Variant
calling

Variant
prioritization

Pipeline

Data QC & Filtering

File .fastq with raw reads
for each sample

fastQC

File .fastq with raw reads
for each sample

schyte
(remove
adapters)

sickle
(trimming)

Alignment

File .fastq
with
filtered
reads

Reference
genome

bwa mem

File .bam with aligned
reads

picard

Filtered and sorted file
.bam

Variant Calling & Annotation

gatk

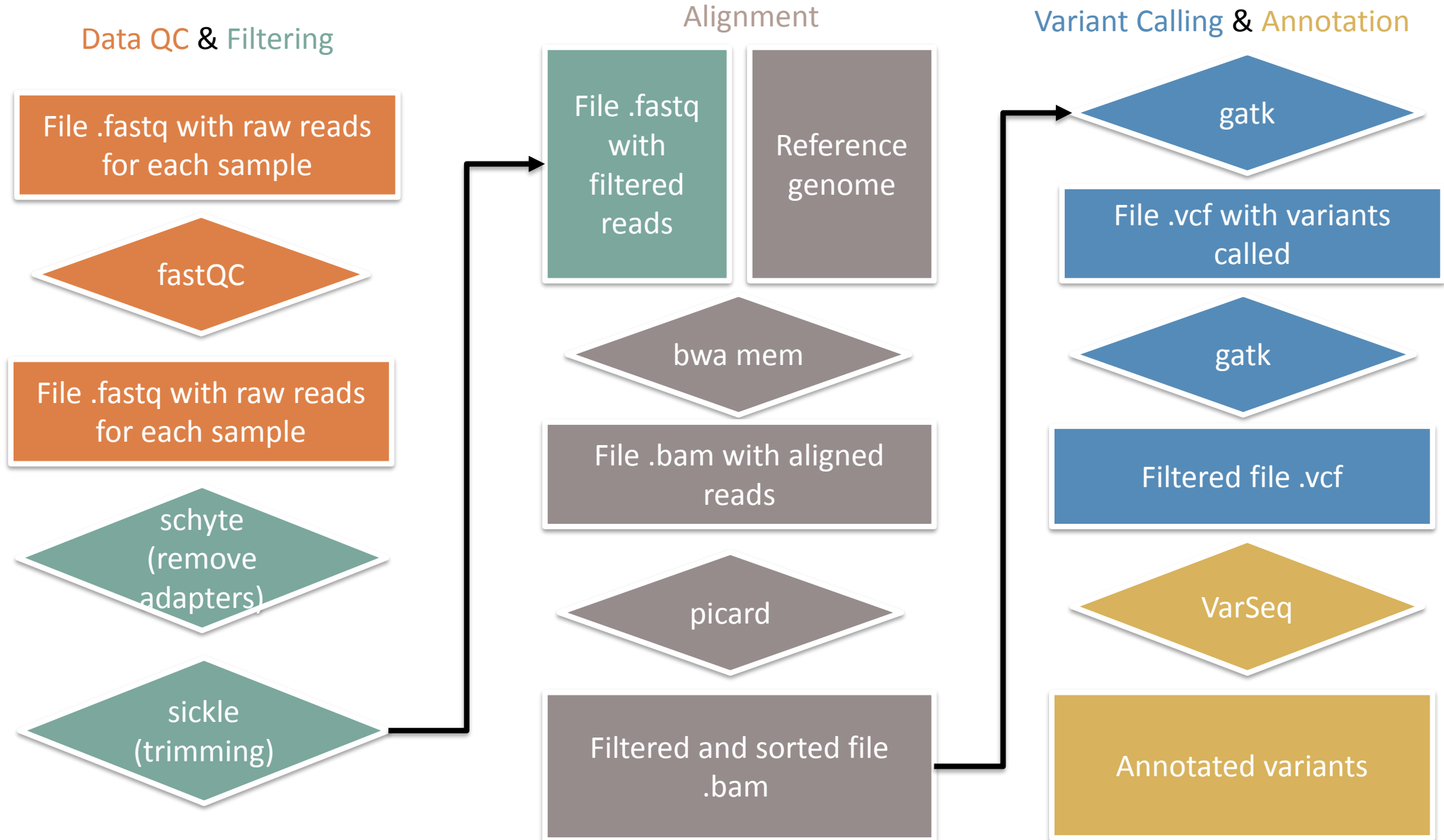
File .vcf with variants
called

gatk

Filtered file .vcf

VarSeq

Annotated variants



Shell

Windows:

- <https://mobaxterm.mobatek.net/download.html>

MAC & Linux:

- Open terminal

Connect to the server

1. Enter in the server:

a. `ssh HGSI2020@157.27.26.214`

b. Password: `hgsi2020`

2. Enter in the

folder: `cd /attachedvolume/HGSI2020`

3. Create your folder: `mkdir your_name`

4. Enter in the created folder: `cd your_name`

Work on the server

1. Create a symbolic link of the files in your folder:

```
ln -s ../example/samples/1351S/R*.fastq.gz .
```

2. Check you have copied the files: `ls`

3. Open the file to see what is inside:

```
less R1.fastq.gz
```

4. Close the visualization: `q`

.fq / .fastq file

For each sample we obtain 2 fastq files containing all the sequences generated



.fq / .fastq file

Each reads is represented by four rows...

```
@HWI-1KL152:89:C118FACXX:8:1209:20044:2901/1
CTCTGTGGCTGGGAGAGGAGTCTGGGGGGGCCCCGGGCGCCAGCCAGGGATAGCCTGATCTCTGCTCCAGTCGACAGATCCTTAACGGATTTTCTTTCTCT
+
/%53@DDAFGIDCCD;EBCE:C>DDCBB&;9<FD5=02&4?8=<CCCA<BB>FFDD=B@C98,>7:<9-9/0&74>CEBEHHBCDHA?=&A9DHB#####
```

First row identify the sequence:

The first row of a FASTQ file is a header line starting with '@'. The components of this line are identified by callouts:

- Instrument ID: @HWI-1KL152
- Flowcell ID: 89
- Tile Coordinates: C118FACXX
- Run Number: 8
- Lane: 1209
- Read ID: 20044:2901/1

```
@HWI-1KL152:89:C118FACXX:8:1209:20044:2901/1
```

Second row contains the sequence:

```
CTCTGTGGCTGGGAGAGGAGTCTGGGGGGGCCCCGGGCGCCAGCCAGGGATAGCCTGATCTCTGCTCCAGTCGACAGATCCTTAACGGATTTTCTTTCTCT
```

Thirs row contains a delimiter:

```
+
```

Fourth row indicate the quality of each base:

```
CTCTGTGGCTGGGAGAGGAGTCTGGGGGGGCCCCGGGCGCCAGCCAGGGATAGCCTGATCTCTGCTCCAGTCGACAGATCCTTAACGGATTTTCTTTCTCT
/%53@DDAFGIDCCD;EBCE:C>DDCBB&;9<FD5=02&4?8=<CCCA<BB>FFDD=B@C98,>7:<9-9/0&74>CEBEHHBCDHA?=&A9DHB#####
```

Q score as ASCII chars: "/" = 47

ASCII CODE

Dec	Hx	Oct	Char	Dec	Hx	Oct	Html	Chr	Dec	Hx	Oct	Html	Chr	Dec	Hx	Oct	Html	Chr
0	0	000	NUL (null)	32	20	040	 	Space	64	40	100	@	@	96	60	140	`	`
1	1	001	SOH (start of heading)	33	21	041	!	!	65	41	101	A	A	97	61	141	a	a
2	2	002	STX (start of text)	34	22	042	"	"	66	42	102	B	B	98	62	142	b	b
3	3	003	ETX (end of text)	35	23	043	#	#	67	43	103	C	C	99	63	143	c	c
4	4	004	EOT (end of transmission)	36	24	044	$	\$	68	44	104	D	D	100	64	144	d	d
5	5	005	ENQ (enquiry)	37	25	045	%	%	69	45	105	E	E	101	65	145	e	e
6	6	006	ACK (acknowledge)	38	26	046	&	&	70	46	106	F	F	102	66	146	f	f
7	7	007	BEL (bell)	39	27	047	'	'	71	47	107	G	G	103	67	147	g	g
8	8	010	BS (backspace)	40	28	050	((72	48	110	H	H	104	68	150	h	h
9	9	011	TAB (horizontal tab)	41	29	051))	73	49	111	I	I	105	69	151	i	i
10	A	012	LF (NL line feed, new line)	42	2A	052	*	*	74	4A	112	J	J	106	6A	152	j	j
11	B	013	VT (vertical tab)	43	2B	053	+	+	75	4B	113	K	K	107	6B	153	k	k
12	C	014	FF (NP form feed, new page)	44	2C	054	,	,	76	4C	114	L	L	108	6C	154	l	l
13	D	015	CR (carriage return)	45	2D	055	-	-	77	4D	115	M	M	109	6D	155	m	m
14	E	016	SO (shift out)	46	2E	056	.	.	78	4E	116	N	N	110	6E	156	n	n
15	F	017	SI (shift in)	47	2F	057	/	/	79	4F	117	O	O	111	6F	157	o	o
16	10	020	DLE (data link escape)	48	30	060	0	0	80	50	120	P	P	112	70	160	p	p
17	11	021	DC1 (device control 1)	49	31	061	1	1	81	51	121	Q	Q	113	71	161	q	q
18	12	022	DC2 (device control 2)	50	32	062	2	2	82	52	122	R	R	114	72	162	r	r
19	13	023	DC3 (device control 3)	51	33	063	3	3	83	53	123	S	S	115	73	163	s	s
20	14	024	DC4 (device control 4)	52	34	064	4	4	84	54	124	T	T	116	74	164	t	t
21	15	025	NAK (negative acknowledge)	53	35	065	5	5	85	55	125	U	U	117	75	165	u	u
22	16	026	SYN (synchronous idle)	54	36	066	6	6	86	56	126	V	V	118	76	166	v	v
23	17	027	ETB (end of trans. block)	55	37	067	7	7	87	57	127	W	W	119	77	167	w	w
24	18	030	CAN (cancel)	56	38	070	8	8	88	58	130	X	X	120	78	170	x	x
25	19	031	EM (end of medium)	57	39	071	9	9	89	59	131	Y	Y	121	79	171	y	y
26	1A	032	SUB (substitute)	58	3A	072	:	:	90	5A	132	Z	Z	122	7A	172	z	z
27	1B	033	ESC (escape)	59	3B	073	;	;	91	5B	133	[[123	7B	173	{	{
28	1C	034	FS (file separator)	60	3C	074	<	<	92	5C	134	\	\	124	7C	174	|	
29	1D	035	GS (group separator)	61	3D	075	=	=	93	5D	135]]	125	7D	175	}	}
30	1E	036	RS (record separator)	62	3E	076	>	>	94	5E	136	^	^	126	7E	176	~	~
31	1F	037	US (unit separator)	63	3F	077	?	?	95	5F	137	_	_	127	7F	177		DEL

Illumina Quality

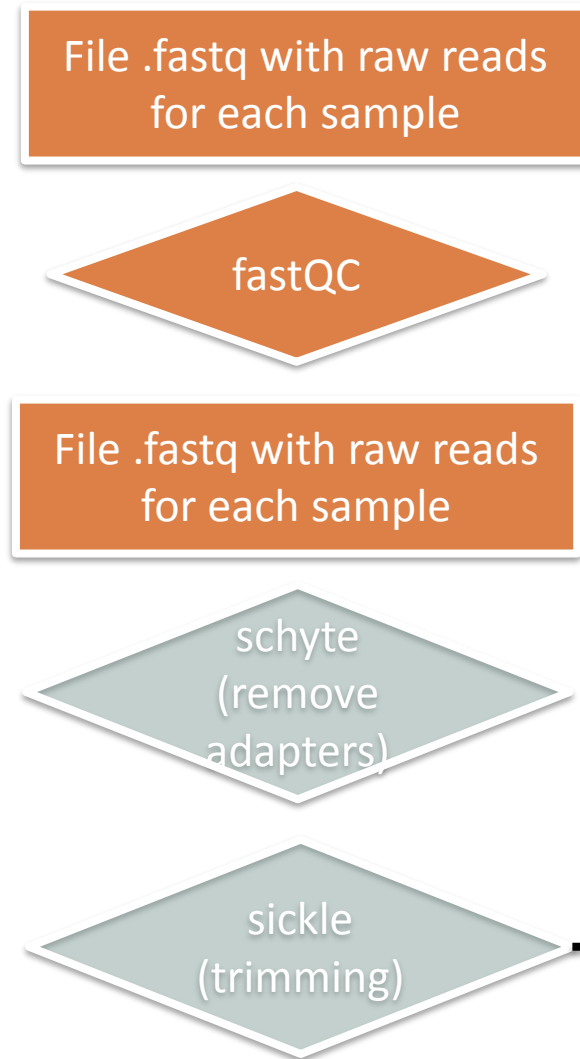
$$Q = \text{ASCII} - 33$$

ASCII_BASE=33 Illumina, Ion Torrent, PacBio and Sanger

Q	P_error	ASCII	Q	P_error	ASCII	Q	P_error	ASCII	Q	P_error	ASCII
0	1.00000	33 !	11	0.07943	44 ,	22	0.00631	55 7	33	0.00050	66 B
1	0.79433	34 "	12	0.06310	45 -	23	0.00501	56 8	34	0.00040	67 C
2	0.63096	35 #	13	0.05012	46 .	24	0.00398	57 9	35	0.00032	68 D
3	0.50119	36 \$	14	0.03981	47 /	25	0.00316	58 :	36	0.00025	69 E
4	0.39811	37 %	15	0.03162	48 0	26	0.00251	59 ;	37	0.00020	70 F
5	0.31623	38 &	16	0.02512	49 1	27	0.00200	60 <	38	0.00016	71 G
6	0.25119	39 '	17	0.01995	50 2	28	0.00158	61 =	39	0.00013	72 H
7	0.19953	40 (18	0.01585	51 3	29	0.00126	62 >	40	0.00010	73 I
8	0.15849	41)	19	0.01259	52 4	30	0.00100	63 ?	41	0.00008	74 J
9	0.12589	42 *	20	0.01000	53 5	31	0.00079	64 @	42	0.00006	75 K
10	0.10000	43 +	21	0.00794	54 6	32	0.00063	65 A			

Pipeline

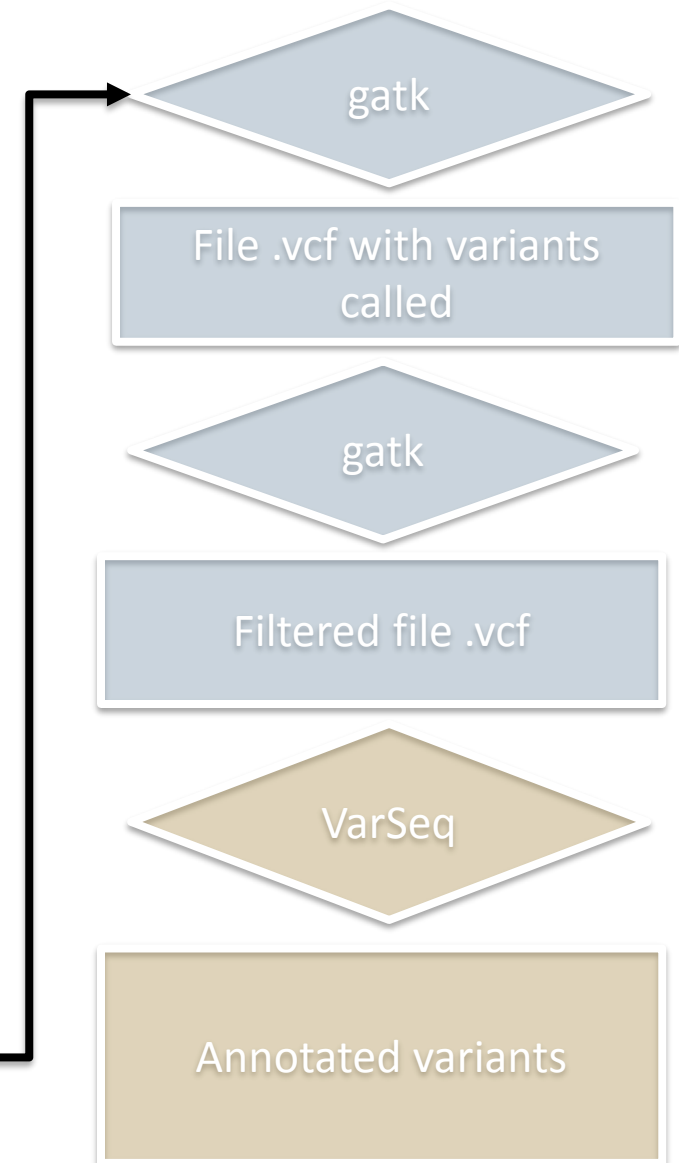
Data QC & Filtering



Alignment



Variant Calling & Annotation



**Check quality of your fastq files
with fastQC**

Fastqc command

1. In your folder, create a folder for fastqc output:

```
mkdir fastqc
```

2. Launch fastQC on both files:

```
fastqc R*.fastq.gz -o fastqc
```

Fastqc command

On the server, we don't have a graphical vision, so..

1. Open a new terminal
2. Create a folder on your PC for the course: `mkdir Desktop/HGSI2020`
3. Enter in the folder: `cd Desktop/HGSI2020`
4. Download the results here:

```
rsync -auv HGSI2020@157.27.26.214:/attachedvolume/HGSI2020/Denise/fastqc/R*_fastqc.html .
```

Pass: `hgsi2020`

5. Check you have downloaded: `ls`
6. Close the shell

Download the files

1. On your desktop, open the file «HGSi2020»

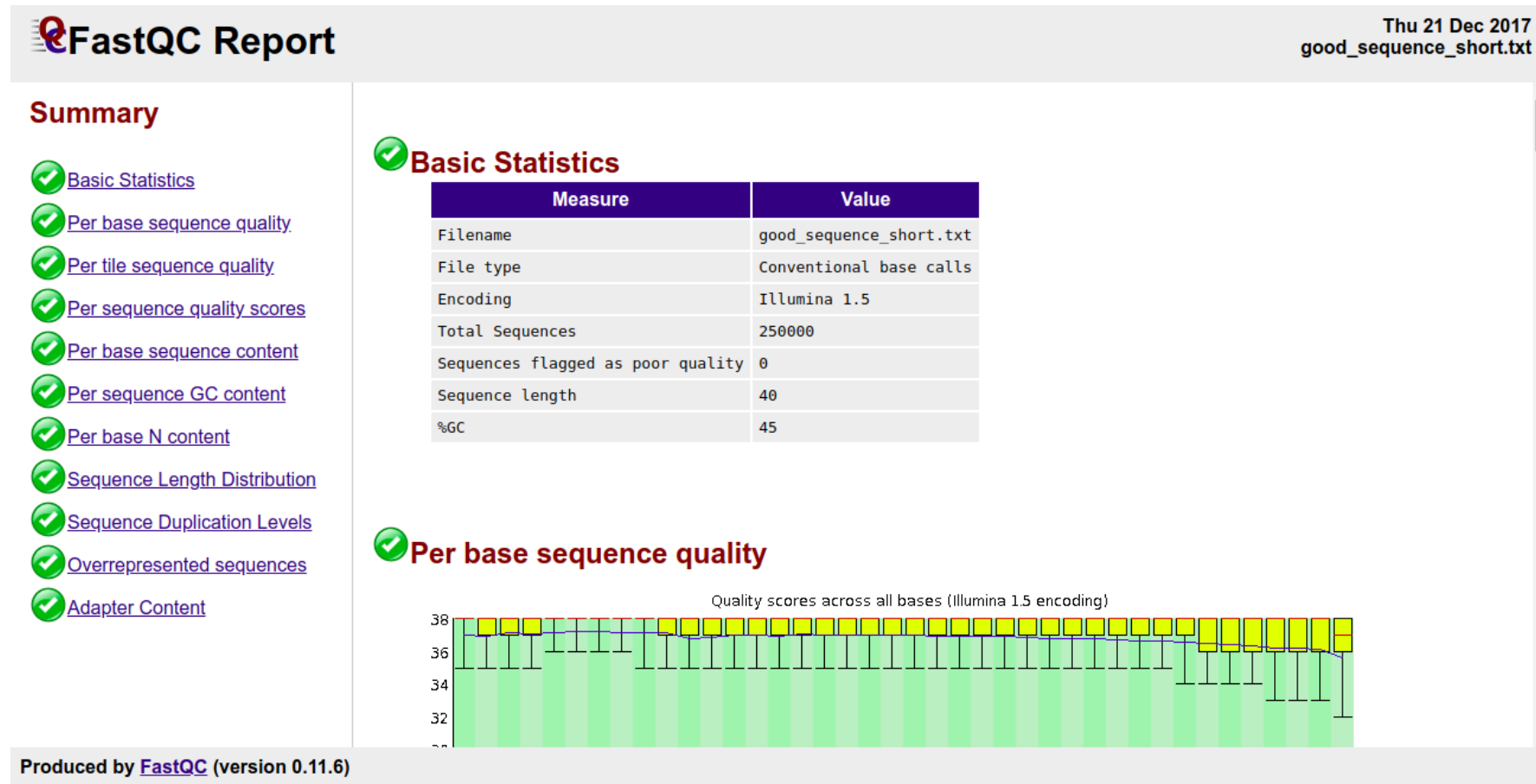


2. Open the file html

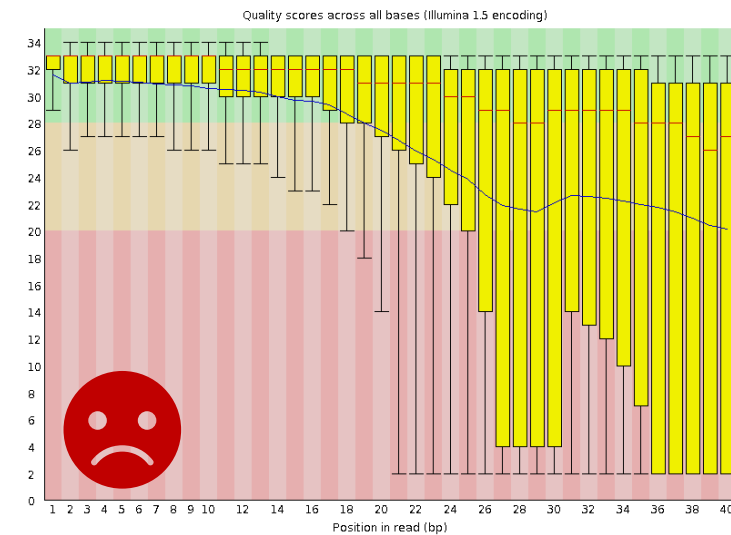
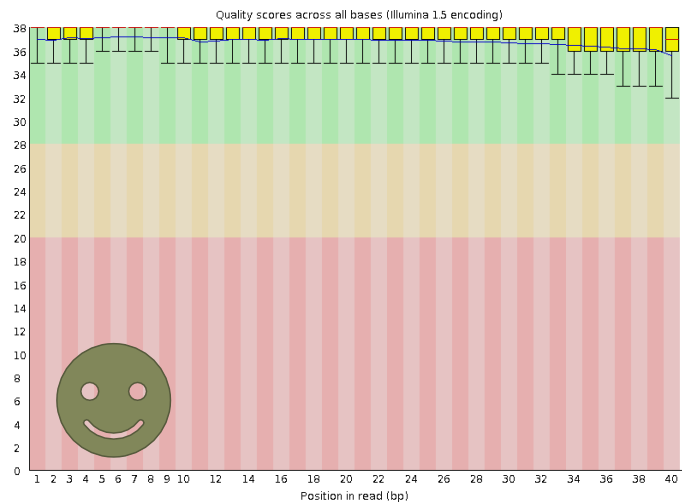
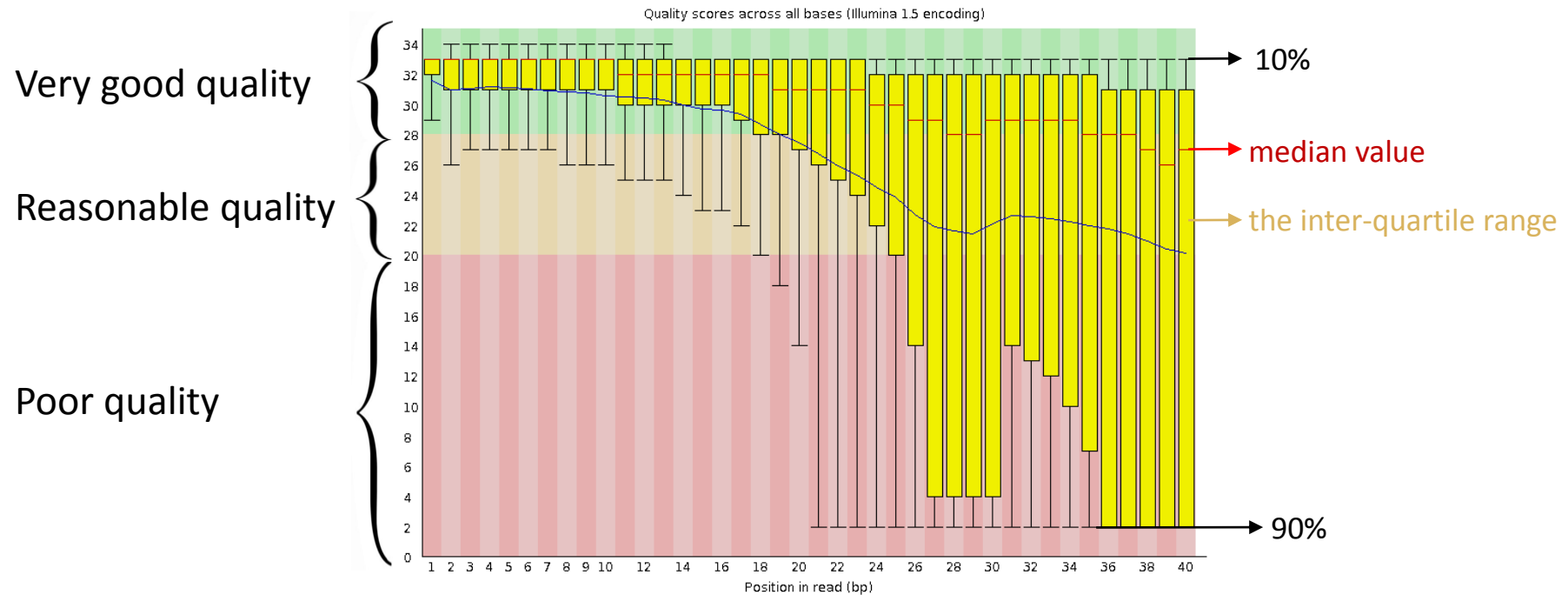
FastQC

FastQC software allows to do some quality control checks on raw sequence data coming from high throughput sequencing.

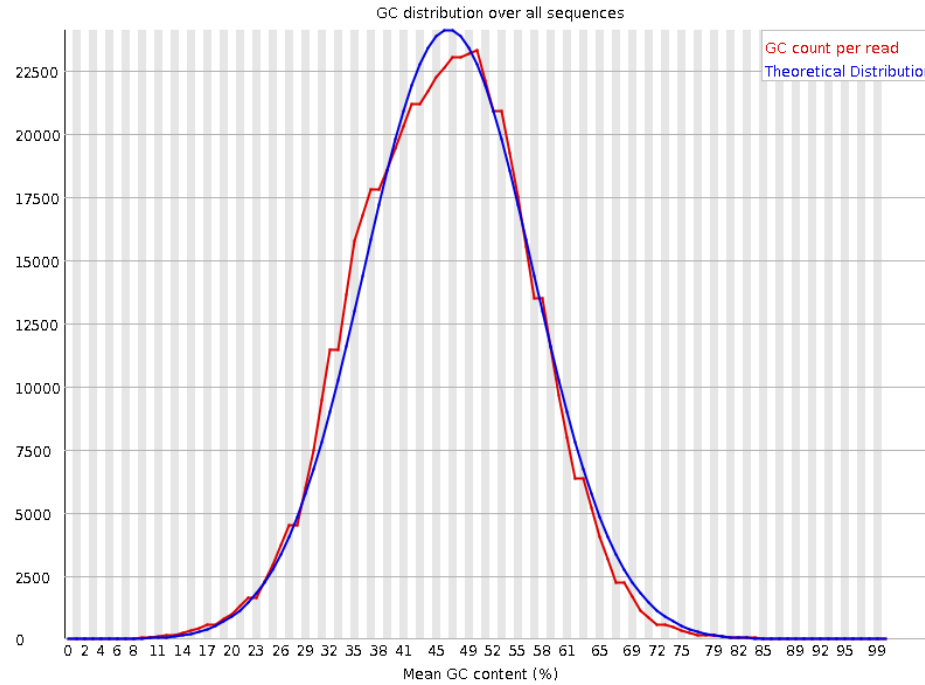
HTML Report



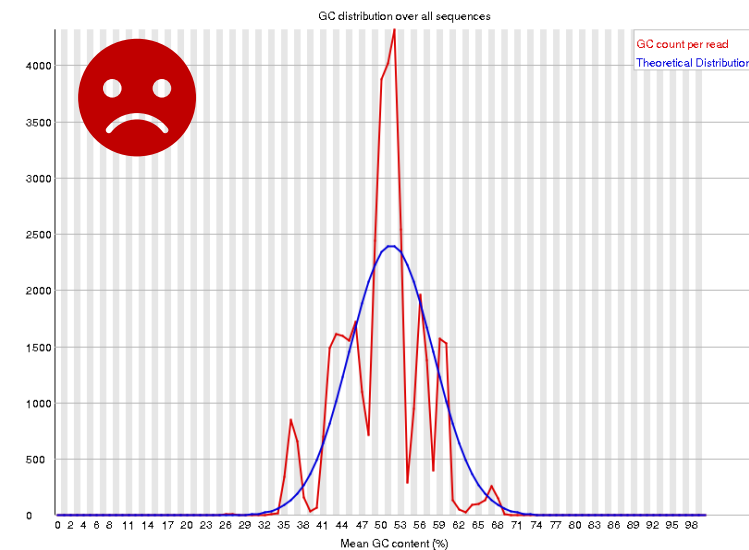
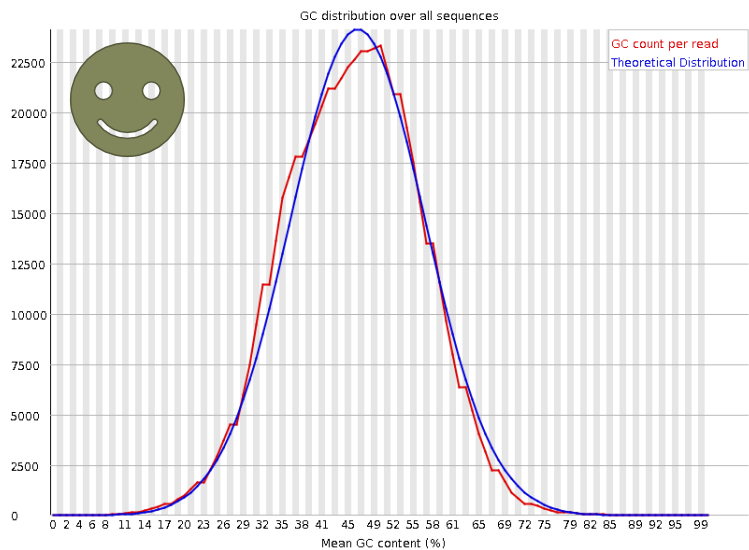
FastQC – Per base sequence quality



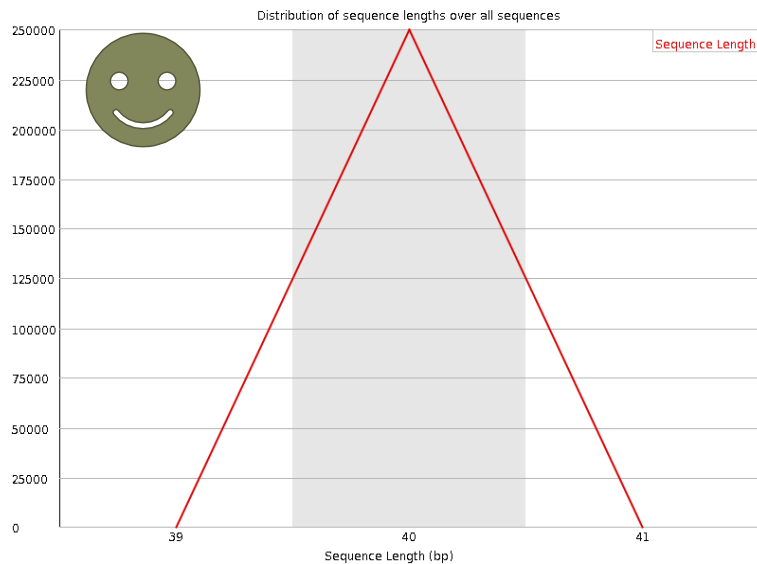
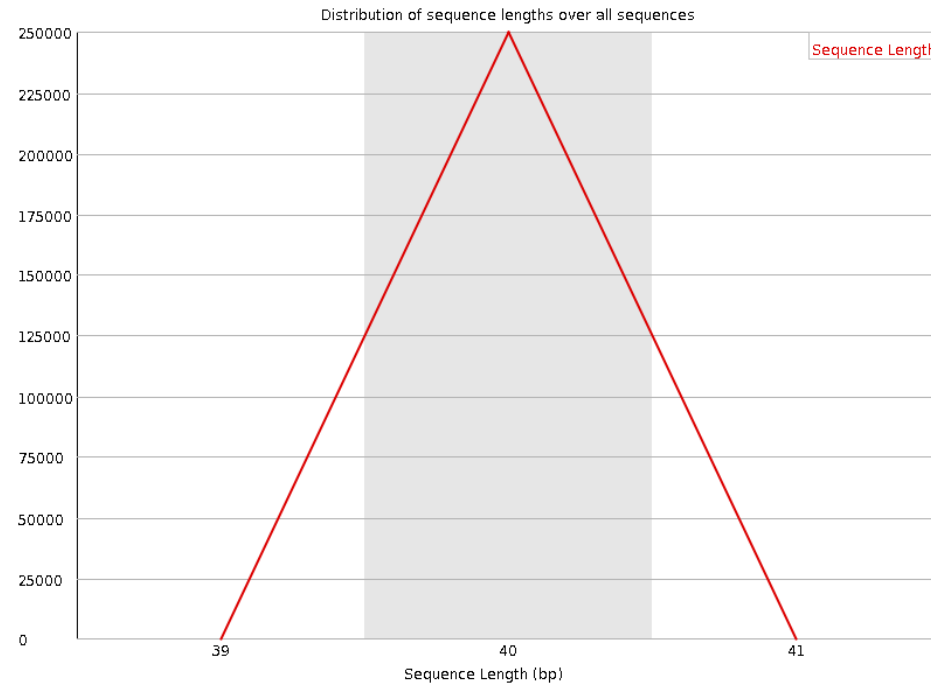
FastQC – Per sequence GC content



Measure of the GC content across the whole length of each sequence and compares it to a modelled normal distribution of GC content

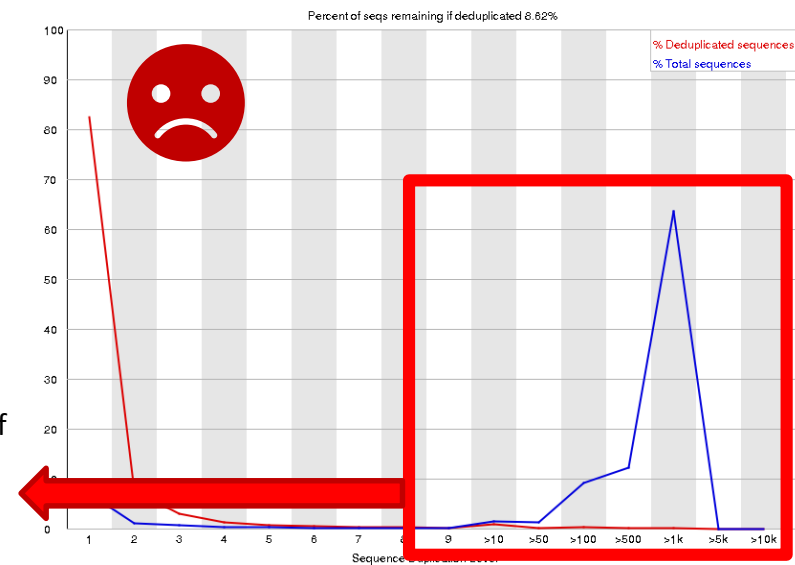
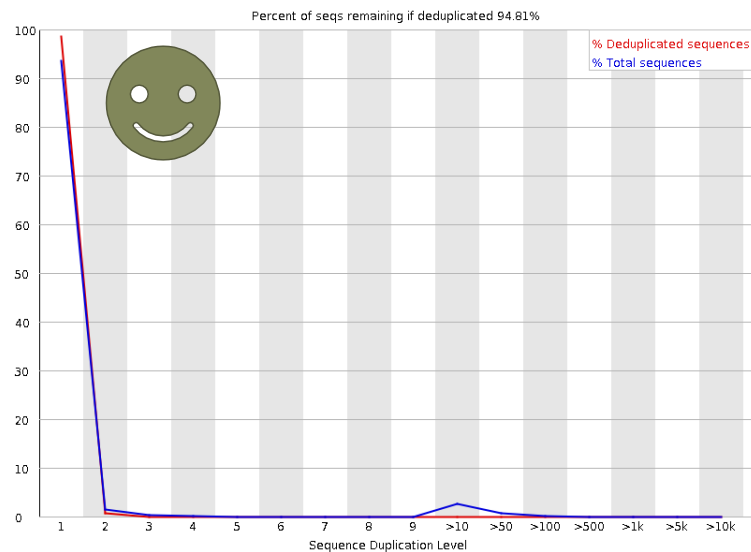
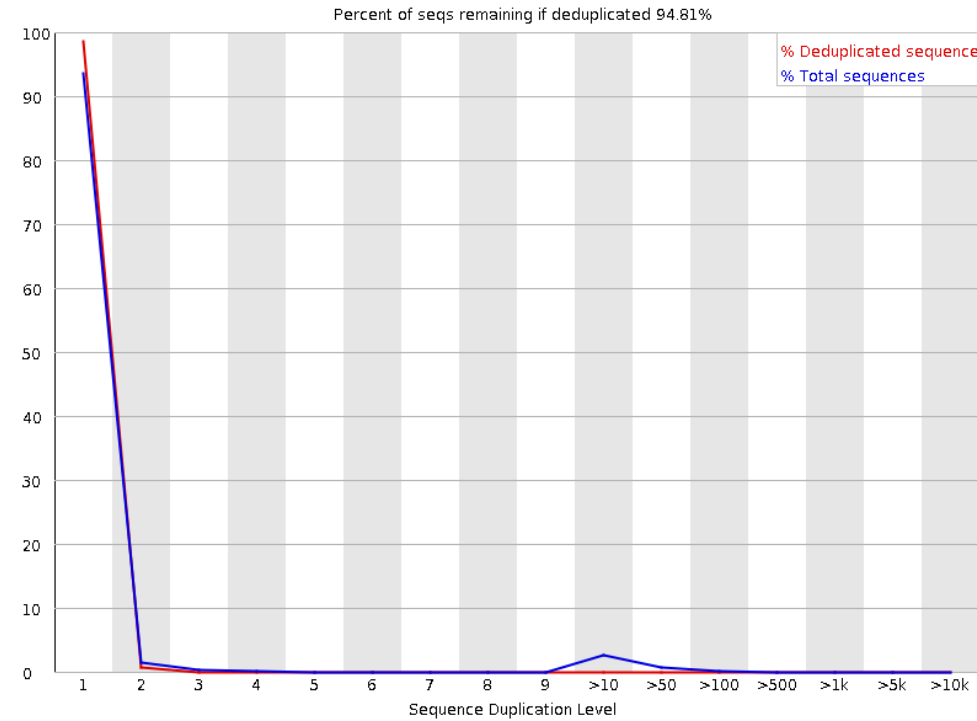


FastQC – Sequence Length Distribution



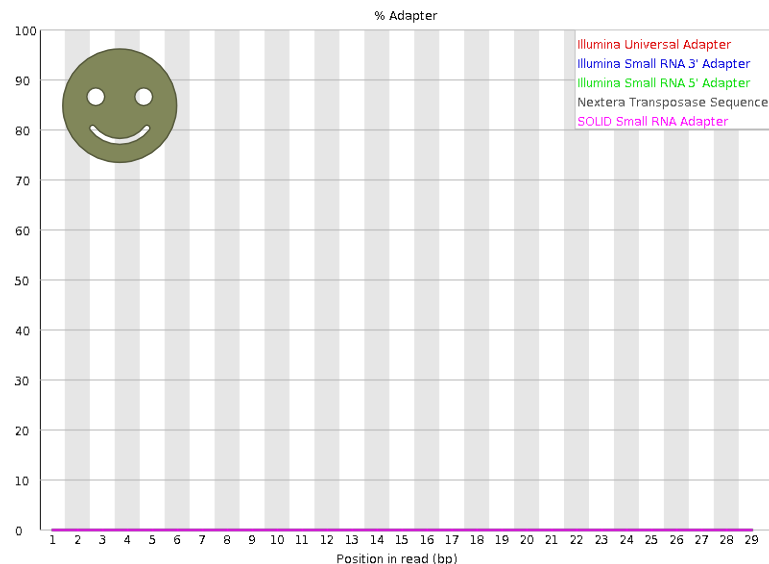
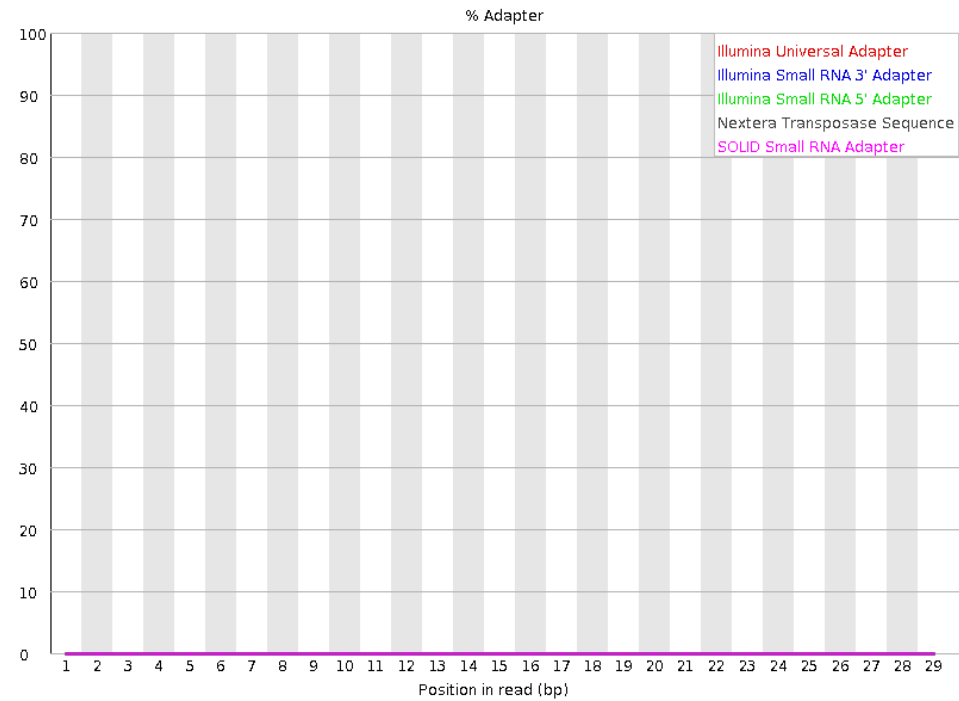
Some sequences
have different length
or zero length

FastQC – Sequence Duplication Levels

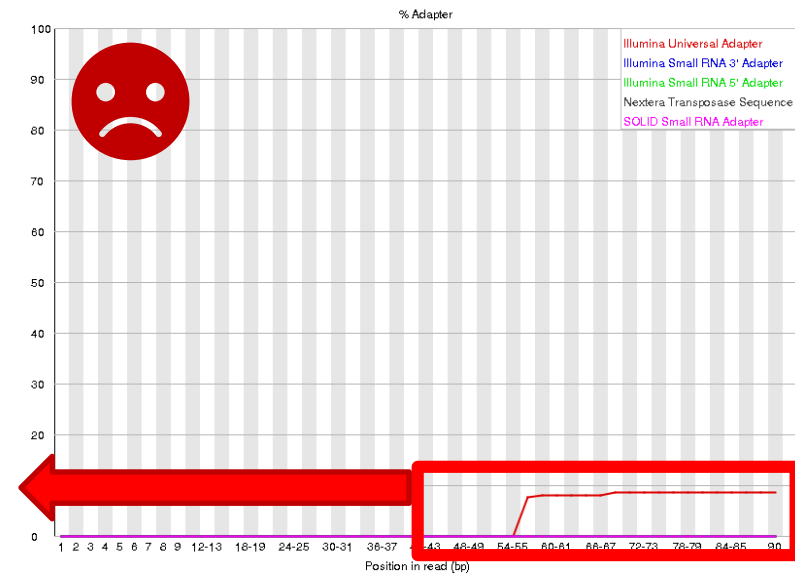


Very large number of sequences with high levels of duplication

FastQC – Adapter Content



Some sequences
contains adapters



Pipeline

Data QC & Filtering

File .fastq with raw reads
for each sample

fastQC

File .fastq with raw reads
for each sample

schyte
(remove
adapters)

sickle
(trimming)

Alignment

File .fastq
with
filtered
reads

Reference
genome

bwa mem

File .bam with aligned
reads

picard

Filtered and sorted file
.bam

Variant Calling & Annotation

gatk

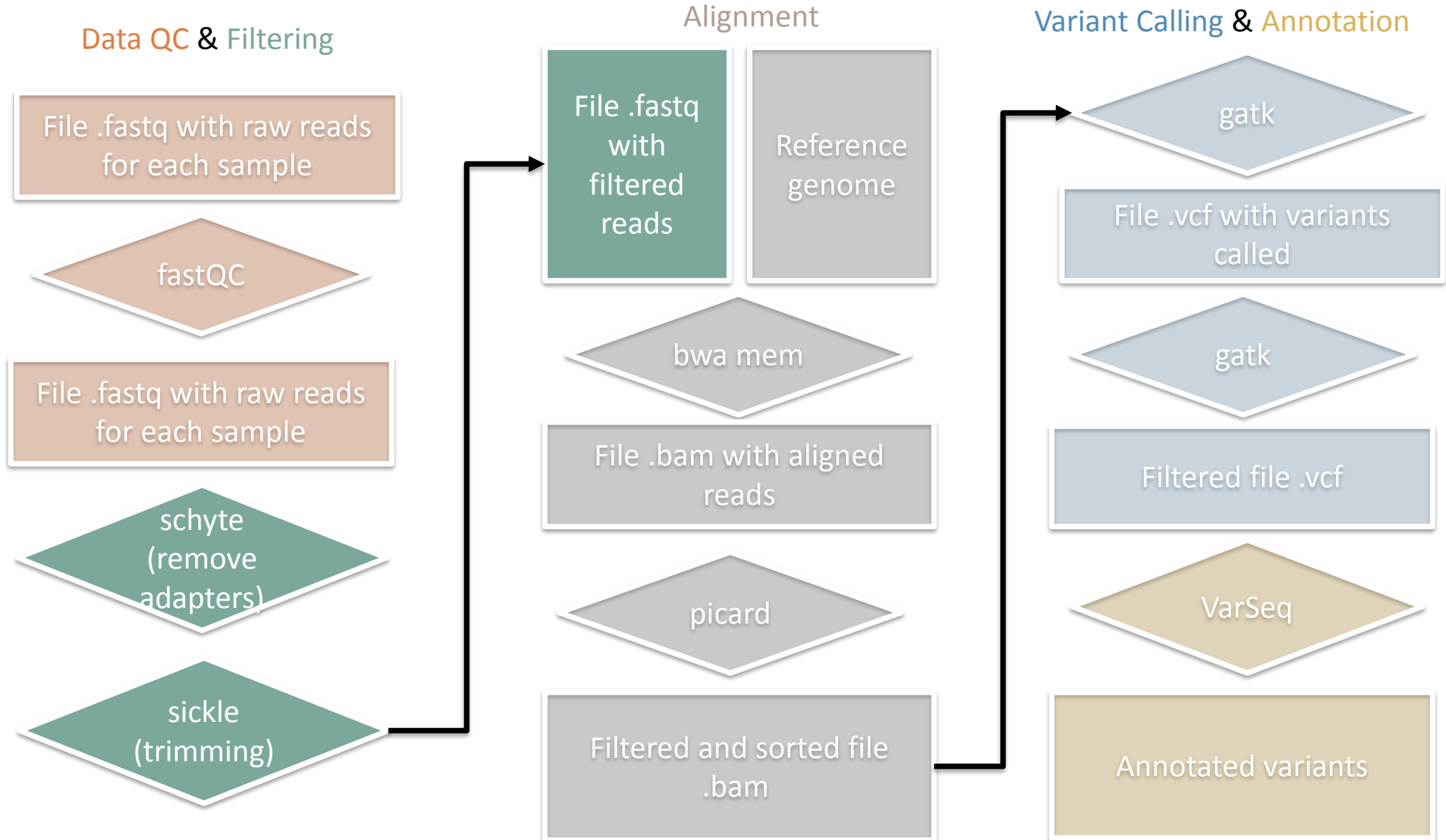
File .vcf with variants
called

gatk

Filtered file .vcf

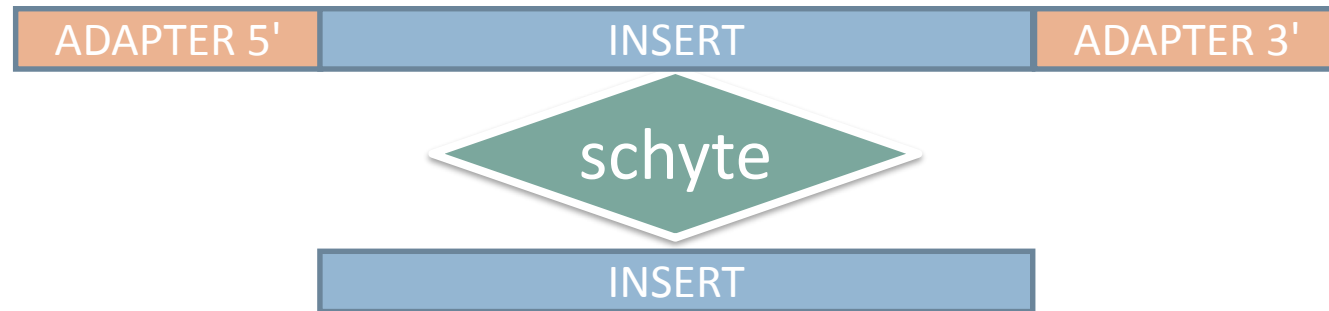
VarSeq

Annotated variants



Remove adapters

Input: RAW fastQ read

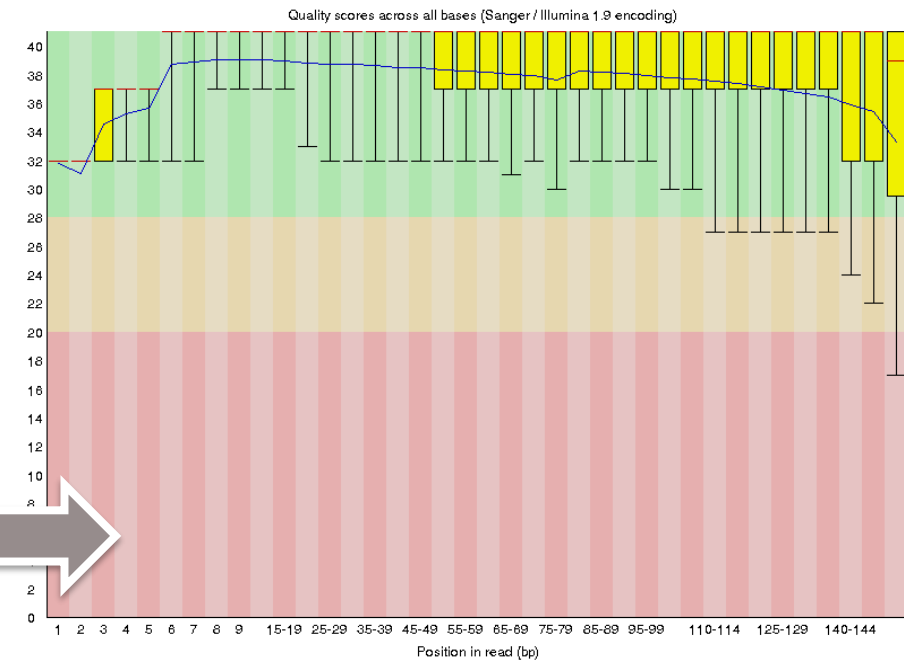
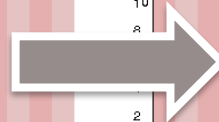
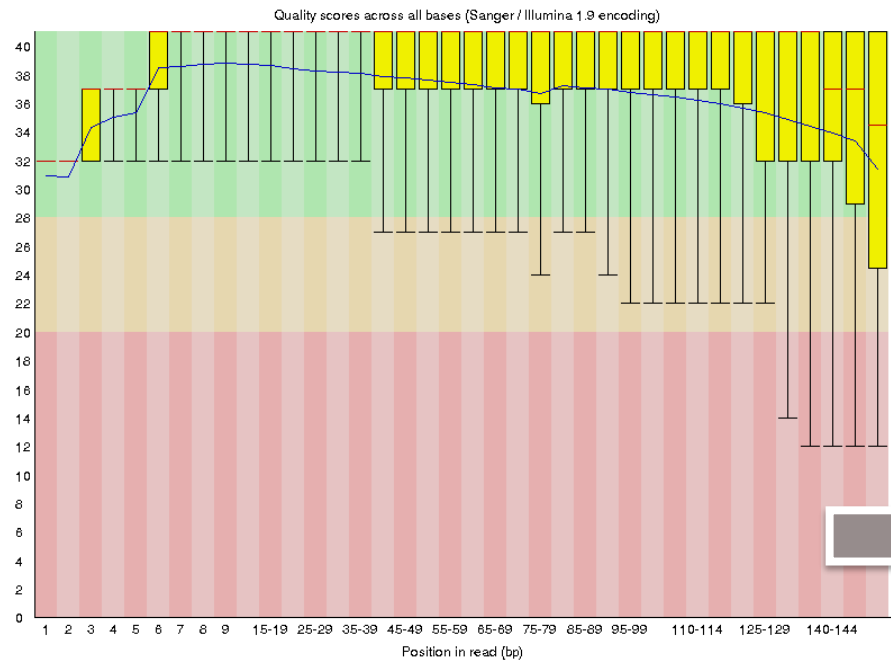


Reads trimming

Input: RAW fastQ read without adapters



Low quality
bases



Filter command

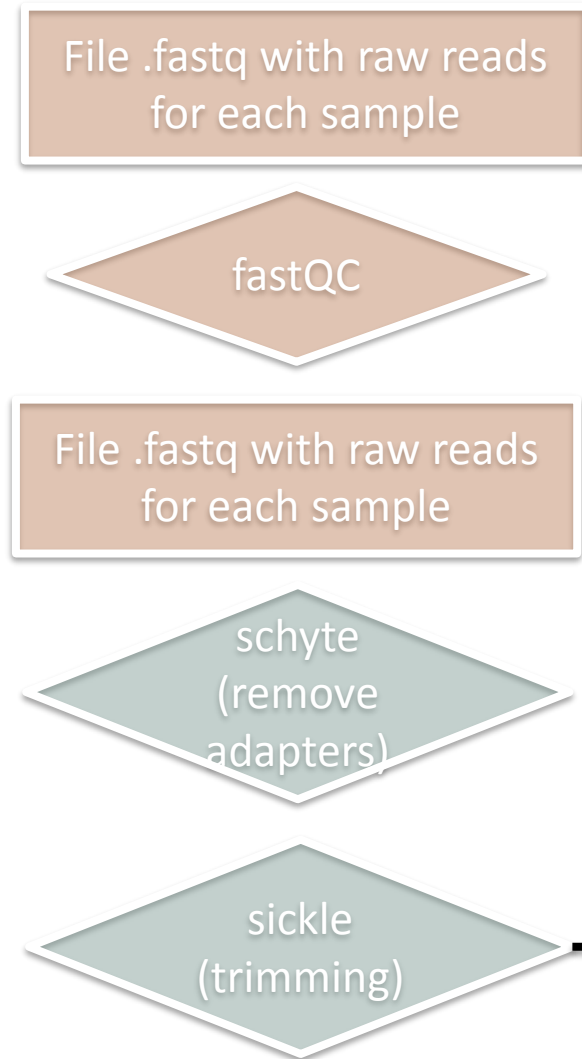
Remove adapters from both reads and trimm reads:

```
sickle pe -g -t sanger  
-f <( scythe -a ../example/reference/illumina_adapters.fa -q sanger R1.fastq.gz)  
-r <( scythe -a ../example/reference/illumina_adapters.fa -q sanger R2.fastq.gz )  
-o trimmed1.fastq.gz -p trimmed2.fastq.gz -s /dev/null
```

Execute the script: `bash /attachedvolume/HGSI2020/example/scripts/step0.trimming.sh`

Pipeline

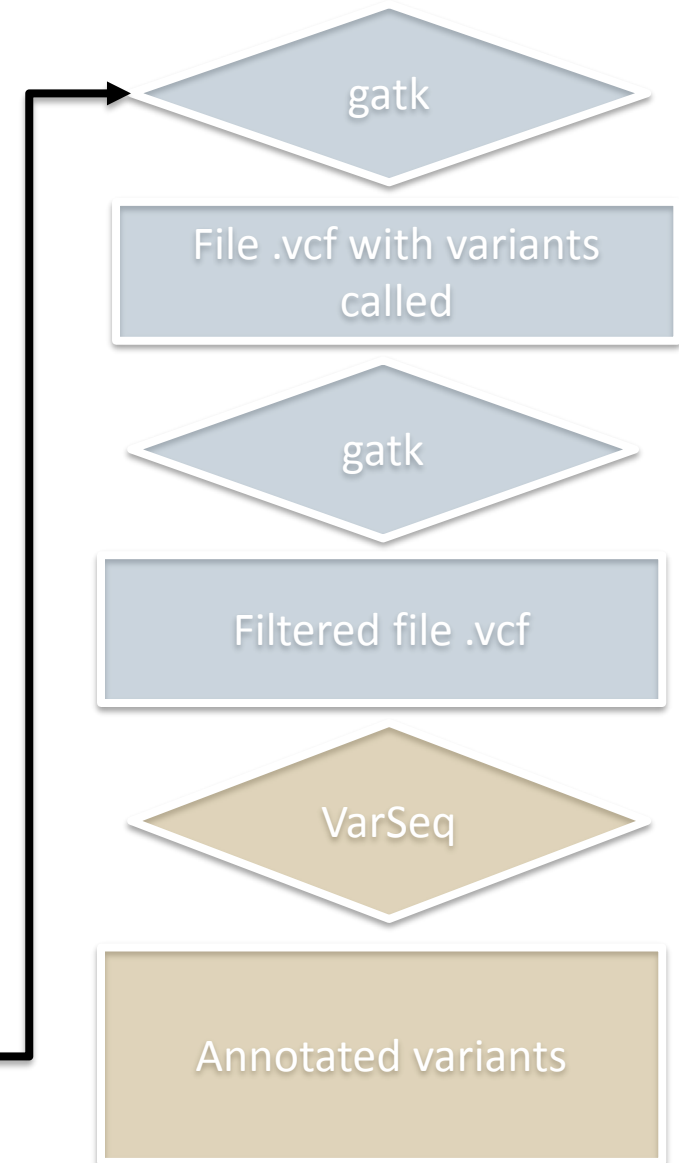
Data QC & Filtering



Alignment



Variant Calling & Annotation



Alignment

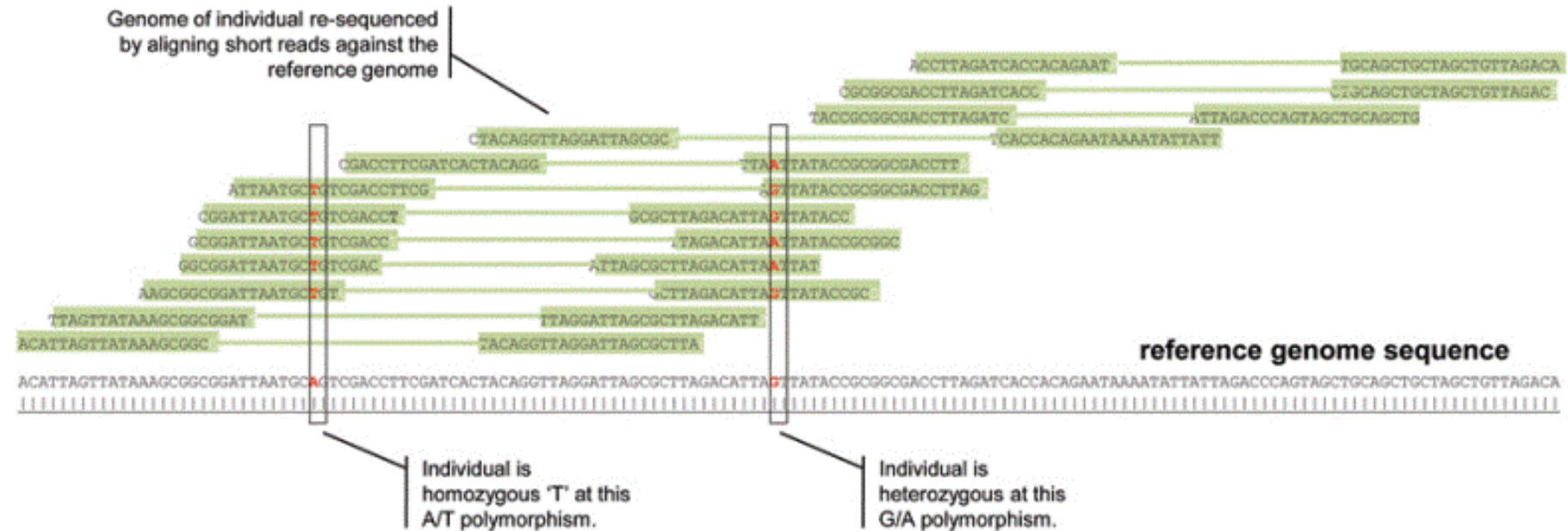
Many of the next-generation sequencing projects begin with a known, or so-called 'reference', genome. In this case, to make sense of the reads, their positions within the reference sequence must be determined. This process is known as aligning or 'mapping' the read to the reference.

Computationally difficult

- Short Reads
- Lots of repeats
- Presence of mismatch

Different algorithm solution:

- Bowtie
- BWA
- ISAAC Aligner



Alignment command

- Align your reads to the reference genome (chr6 hg38):

```
/attachedvolume/HGSI2020/example/bin/bwa-0.7.12 mem  
/attachedvolume/HGSI2020/example/reference/chr6.hg38.fa  
trimmed1.fastq.gz trimmed2.fastq.gz > sample.sam
```

- Turn your file sam into file bam:

```
samtools view -bT /attachedvolume/HGSI2020/example/reference/chr6.hg38.fa -o  
sample.bam sample.sam
```

Alignment command

- Sort your file:

```
samtools sort sample.bam -o sample.sorted.bam
```

- Create index for your bam file:

```
samtools index sample.sorted.bam
```

- Open the file:

```
samtools view sample.sorted.bam | less -S
```

Alignment output – BAM file

Header

Body

```
@HD VN:1.3 SO:coordinate
@SQ SN:chr2 LN:243199373
@PG ID:bwa PN:bwa VN:0.7.15-r1140 CL:bwa mem chr2.fasta read1.trimmed.fastq.gz read2.trimmed.fastq.gz
NB500897:75:H5NWWBGXY:3:13608:21224:10540 2147 chr2 12323980 0 57H38M21H = 69870261 57546346 GGCTGAGGTGGGAG
NB500897:78:H5T5FBGXY:4:11401:24478:5392 2131 chr2 25275067 0 108H33M9H = 69650885 44375787 AAAAATAAAAAATAA
NB500897:75:H5NWWBGXY:3:12601:23290:7800 2115 chr2 33141345 0 94H56M = 68269866 35128522 GGGGGGGGGGGGGGGGGGGGGG
NB500897:78:H5T5FBGXY:3:12609:16449:18291 2115 chr2 33141438 0 120H30M = 68547068 35405631 GGGGGGGGGGGGGGGGGGGGGG
NB500897:75:H5NWWBGXY:1:13104:6732:16100 2115 chr2 33141478 0 120H30M = 68547068 35405591 GGGGGGGGGGGGGGGGGGGGGG
NB500897:78:H5T5FBGXY:4:12411:14105:19392 2147 chr2 33141574 0 100H50M = 69871302 36729810 GGGGGGGGGGGGGGGGGGGGGG
NB500897:75:H5NWWBGXY:2:23311:23906:6768 2115 chr2 33141581 0 87H43M = 68269811 35128231 GGGGGGGGGGGGGGGGGGGGGG
NB500897:75:H5NWWBGXY:1:12106:7875:15761 2115 chr2 33141594 0 120H30M = 68547068 35405475 GGGGGGGGGGGGGGGGGGGGGG
NB500897:78:H5T5FBGXY:3:22601:12050:10673 161 chr2 50626928 0 2S64M = 69890571 19263716 AAAAAAAAAAAAAAAAAAAAAA
NB500897:75:H5NWWBGXY:4:12607:26302:14475 81 chr2 52194430 0 78M = 69688660 17494154 ATGTTGGCGAGGCTGGTCTCCA
NB500897:78:H5T5FBGXY:4:23607:18581:7865 163 chr2 58355595 0 26M = 58355665 151 TAGCTGGGATTACAGGTGTGTGCCAC
NB500897:78:H5T5FBGXY:4:23607:18581:7865 83 chr2 58355665 0 4S81M13S = 58355595 -151 CTCACCATGTTGCCAGGCTGG
NB500897:75:H5NWWBGXY:3:13608:21224:10540 2179 chr2 65437823 0 63M53H = 69870261 4432439 ACTATGCTGACCAGGTTGGTTTCAAATTCC
NB500897:75:H5NWWBGXY:2:11103:13114:1410 99 chr2 67999995 60 66M = 68000128 259 CTCACAATAAATTTATTTTTTCAAAGCAG
NB500897:75:H5NWWBGXY:2:11103:13114:1410 147 chr2 68000128 60 126M = 67999995 -259 CATCTAGATAGCTATCTTTCCAGACTTTTC
NB500897:78:H5T5FBGXY:1:21201:19536:13168 99 chr2 68000446 60 150M = 68000452 156 CCTCAGAGTATTAAGACCACATAGTATAT
NB500897:78:H5T5FBGXY:4:23607:24808:1972 99 chr2 68000446 60 150M = 68000452 156 CCTCAGAGTATTAAGACCACATAGTATAT
NB500897:78:H5T5FBGXY:1:21201:19536:13168 147 chr2 68000452 60 150M = 68000446 -156 AGTATTAAGACCACATAGTATATATTTTC
NB500897:78:H5T5FBGXY:4:23607:24808:1972 147 chr2 68000452 60 150M = 68000446 -156 AGTATTAAGACCACATAGTATATATTTTC
NB500897:75:H5NWWBGXY:4:12408:9893:6566 163 chr2 68001096 60 150M = 68001096 150 AGATTCCTGGCTAAATTCACCATTGAAAGAAATTGA
NB500897:75:H5NWWBGXY:4:12408:9893:6566 83 chr2 68001096 60 150M = 68001096 -150 AGATTCCTGGCTAAATTCACCATTGAAAGAAATTGA
NB500897:78:H5T5FBGXY:2:12107:7865:18634 99 chr2 68001412 60 150M = 68001549 287 TTCCTAACTAAATACTGACTAGAACAGTGA
NB500897:78:H5T5FBGXY:2:12107:7865:18634 147 chr2 68001549 60 150M = 68001412 -287 CCCAACTCCTGCCACTCTAGCCACATCAAG
NB500897:78:H5T5FBGXY:1:12109:14488:19284 99 chr2 68001927 60 150M = 68002195 418 ACGTAGTAGAAATTCACAGAATACTTGTA
NB500897:78:H5T5FBGXY:1:12305:16077:11340 163 chr2 68001976 60 150M = 68002289 462 AATTCCTCACCAGCTTCAGCAGCTTAAGGA
NB500897:75:H5NWWBGXY:1:22202:17880:17856 99 chr2 68001995 60 150M = 68002066 221 CAGCTTAAGGATAAAGAATCTTGCATCTAA
```

Read ID

Mapping
Tag

Mapping
position

Mapping quality

CIGAR
string

Pair
information

Cigar

Op	Description
M	alignment match (can be a sequence match or mismatch)
I	insertion to the reference
D	deletion from the reference
N	skipped region from the reference
S	soft clipping (clipped sequences present in SEQ)
H	hard clipping (clipped sequences NOT present in SEQ)
P	padding (silent deletion from padded reference)
=	sequence match
X	sequence mismatch



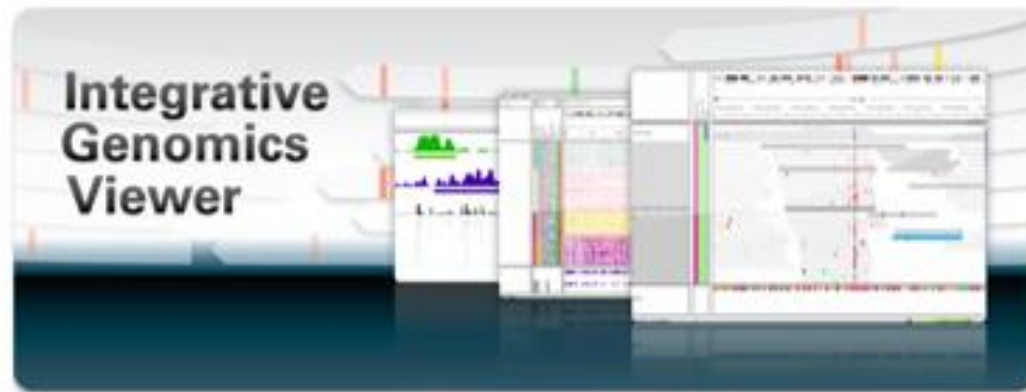
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Overview

The **Integrative Genomics Viewer (IGV)** is a high-performance visualization tool for interactive exploration of large, integrated genomic datasets. It supports a wide variety of data types, including array-based and next-generation sequence data, and genomic annotations.

IGV is available in multiple forms, including:

- the original **IGV** - a Java desktop application,
- IGV-Web** - a web application,
- igv.js** - a JavaScript component that can be embedded in web pages (for developers)

This site is focused on the IGV desktop application. See <https://igv.org> for links to all forms of IGV.

Download IGV



Download the IGV desktop application and igvtools.

Note that the IGV-Web application at <https://igv.org/app> runs in a web browser and requires no downloads. Click on the Help link in the app for more information.

Citing IGV

To cite your use of IGV in your publication, please reference one or more of:

James T. Robinson, Helga Thorvaldsdóttir, Wendy Winckler, Mitchell Guttman, Eric S. Lander, Gad Getz, Jill P. Mesirov. [Integrative Genomics Viewer](#). *Nature Biotechnology* 29, 24–26 (2011). (Free PMC article [here](#)).

Helga Thorvaldsdóttir, James T. Robinson, Jill P. Mesirov. [Integrative Genomics Viewer \(IGV\): high-performance genomics data visualization and exploration](#). *Briefings in Bioinformatics* 14, 178–192 (2013).

James T. Robinson, Helga Thorvaldsdóttir, Aaron M. Wenger, Ahmet Zehir, Jill P. Mesirov. [Variant Review with the Integrative Genomics Viewer \(IGV\)](#). *Cancer Research* 77(21):31–34 (2017).

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IGV participates in the [GenomeSpace](#) initiative, which is funded by the [National Human Genome Research Institute](#).

IGV

<https://software.broadinstitute.org/software/igv/>

Download IGV

Install IGV 2.8.x

See the [Release Notes](#) for what's new in each release.



IGV Mac App

Download and unzip the Mac App Archive, then double-click the IGV application to run it. You can move the app to the *Applications* folder, or anywhere else.

MacOS Catalina users: We sign our Mac App as a trusted Apple developer, but it is not yet notarized by Apple (a new requirement in Catalina). To run it, right-click on the downloaded IGV app; select "Open" from the menu; and click the "Open" button in the window that pops up. After that, double-clicking on the app will also work.



IGV for Windows

Download and run the installer.
An IGV shortcut will be created on the Desktop; double-click it to run the application.



IGV for Linux

Download and unzip the Archive.
See the downloaded *readme.txt* for further instructions.



IGV and igvtools to run on the command line (all platforms)

Download and unzip the Archive. **Requires Java 11.**
See the downloaded *readme.txt* and *igvtools_readme.txt* for further instructions.

Download the bam and the bai

- Download the bam file and the index file on your pc:

- Open new terminal:

```
cd Desktop/HGSI2020
```

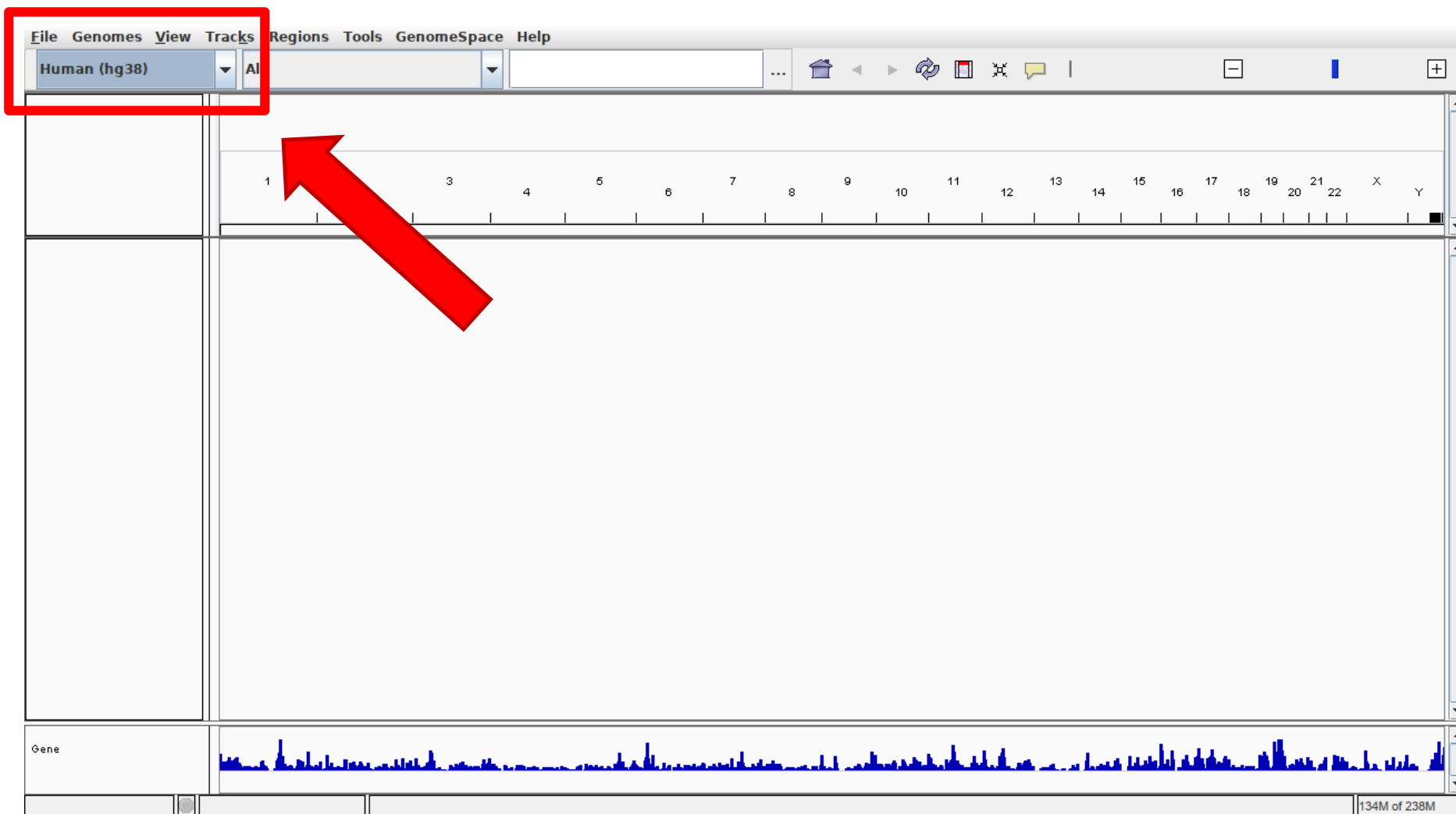
```
rsync -auv HGSI2020@157.27.26.214:/attachedvolume/HGSI2020/Denise/sample.sorted.bam* .
```

- Check if you have downloaded: `ls`

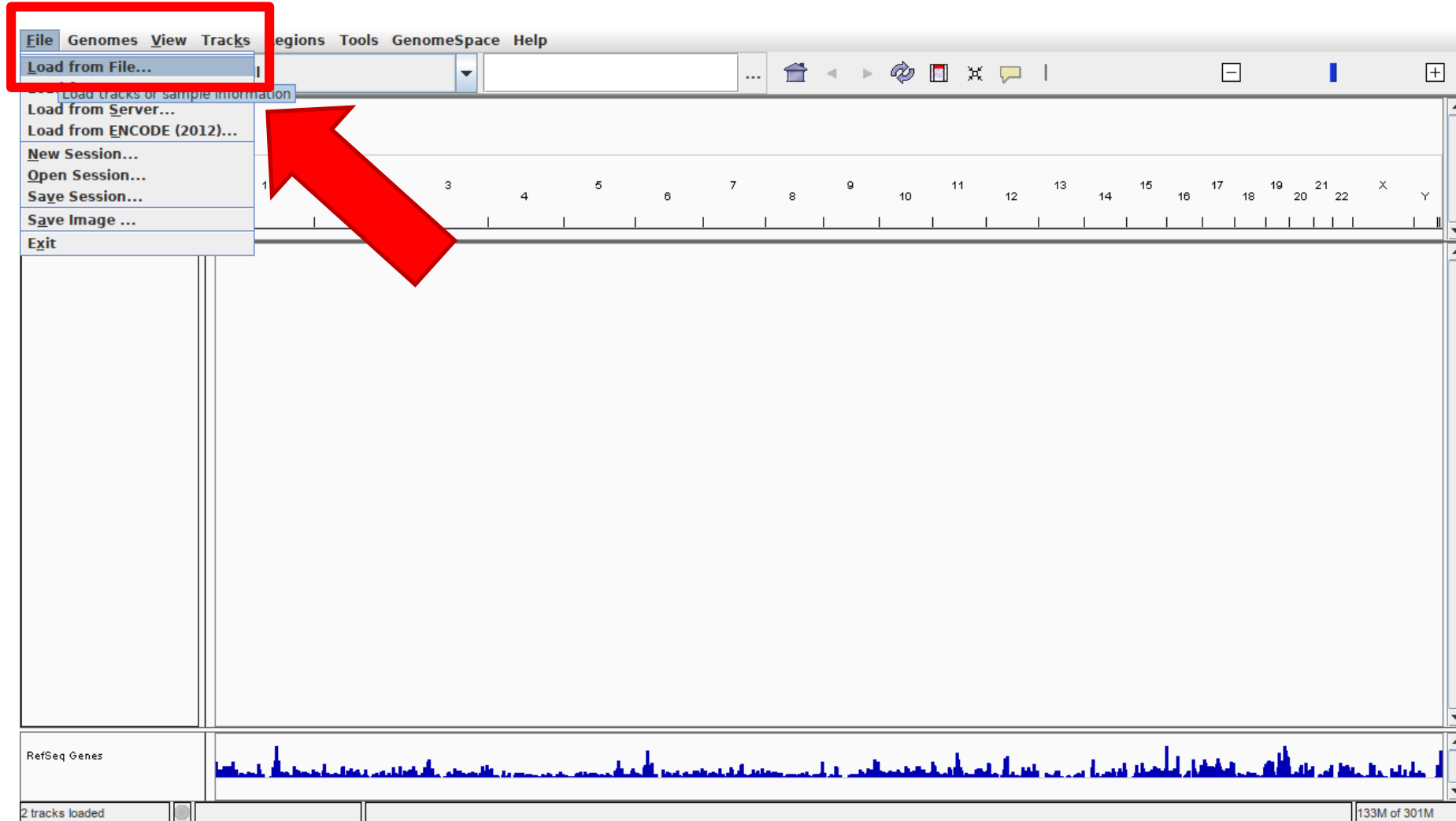
- Open IGV

```
./igv.sh for Ubuntu
```

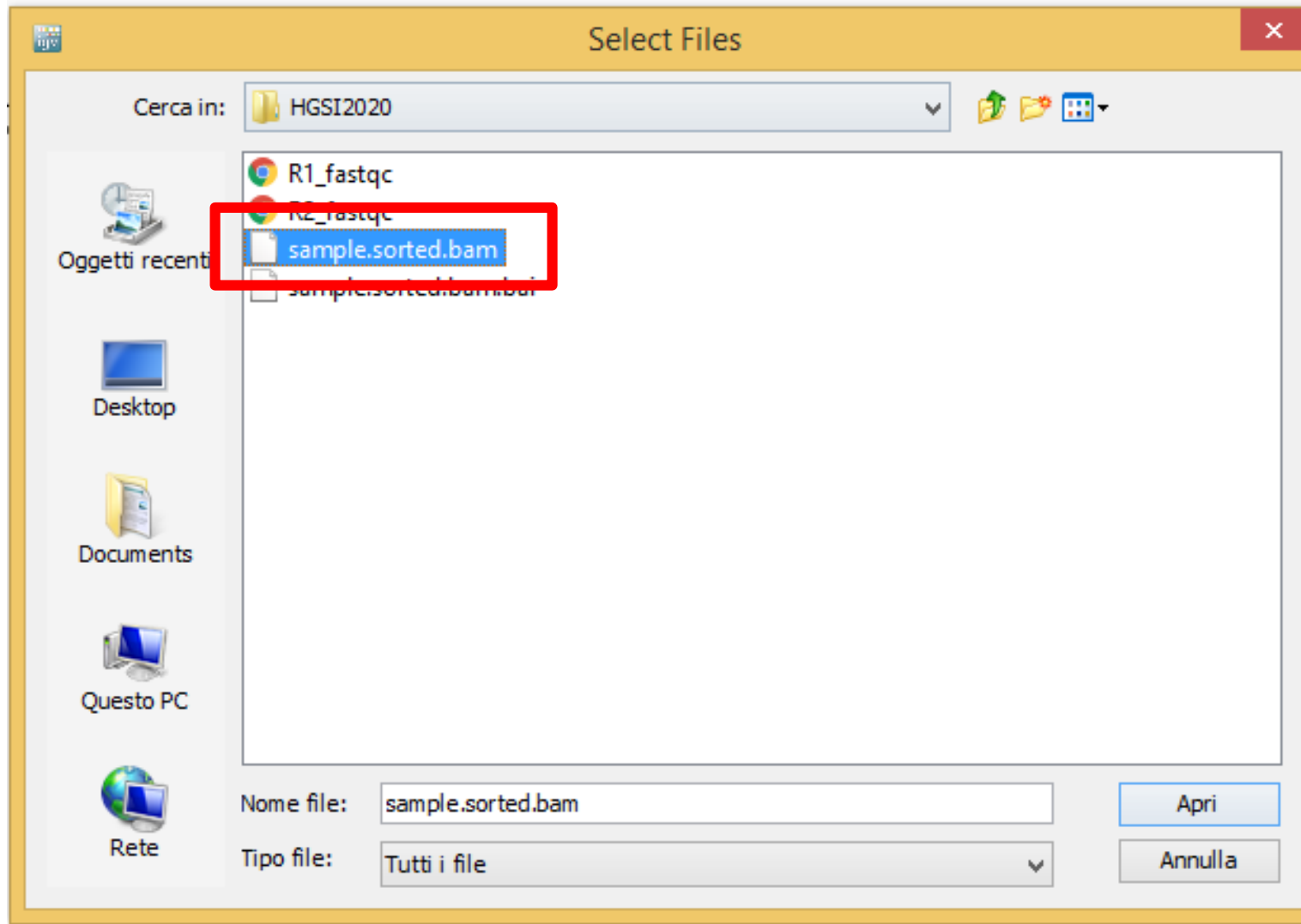
Choose the right genome



Upload the bam



Go into the folder "HGS12020",
choose the file bam and open it



Search a specific region

The screenshot displays a genomic browser interface with a menu bar at the top containing 'File', 'Genomes', 'View', 'Tracks', 'Regions', 'Tools', 'GenomeSpace', and 'Help'. Below the menu, a dropdown menu is set to 'Human (hg38)', and another dropdown is set to 'All'. A text input field contains the search query 'chr6:289,015-307,482', which is highlighted by a red rectangular box. To the right of the input field is a 'Go' button and a series of navigation icons. Below the navigation bar is a chromosome ideogram showing chromosomes 1 through 22, X, and Y. A large red arrow points from the text 'We search the region: chr6:289,015-307,482' to the search input field. The main panel of the browser is divided into three tracks: 'sample.sorted.bam Coverage', 'sample.sorted.bam', and 'Gene'. The 'Gene' track at the bottom shows a blue signal across the genome. The status bar at the bottom left indicates '4 tracks' and the bottom right shows '154M of 291M'.

Human (hg38) All chr6:289,015-307,482 Go

1 2 3 4 5 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y

sample.sorted.bam Coverage

sample.sorted.bam

Gene

4 tracks 154M of 291M

We search the region:
chr6:289,015-307,482

Results



Difference between genome and exome sequencing

