



# Review of NGS in clinic

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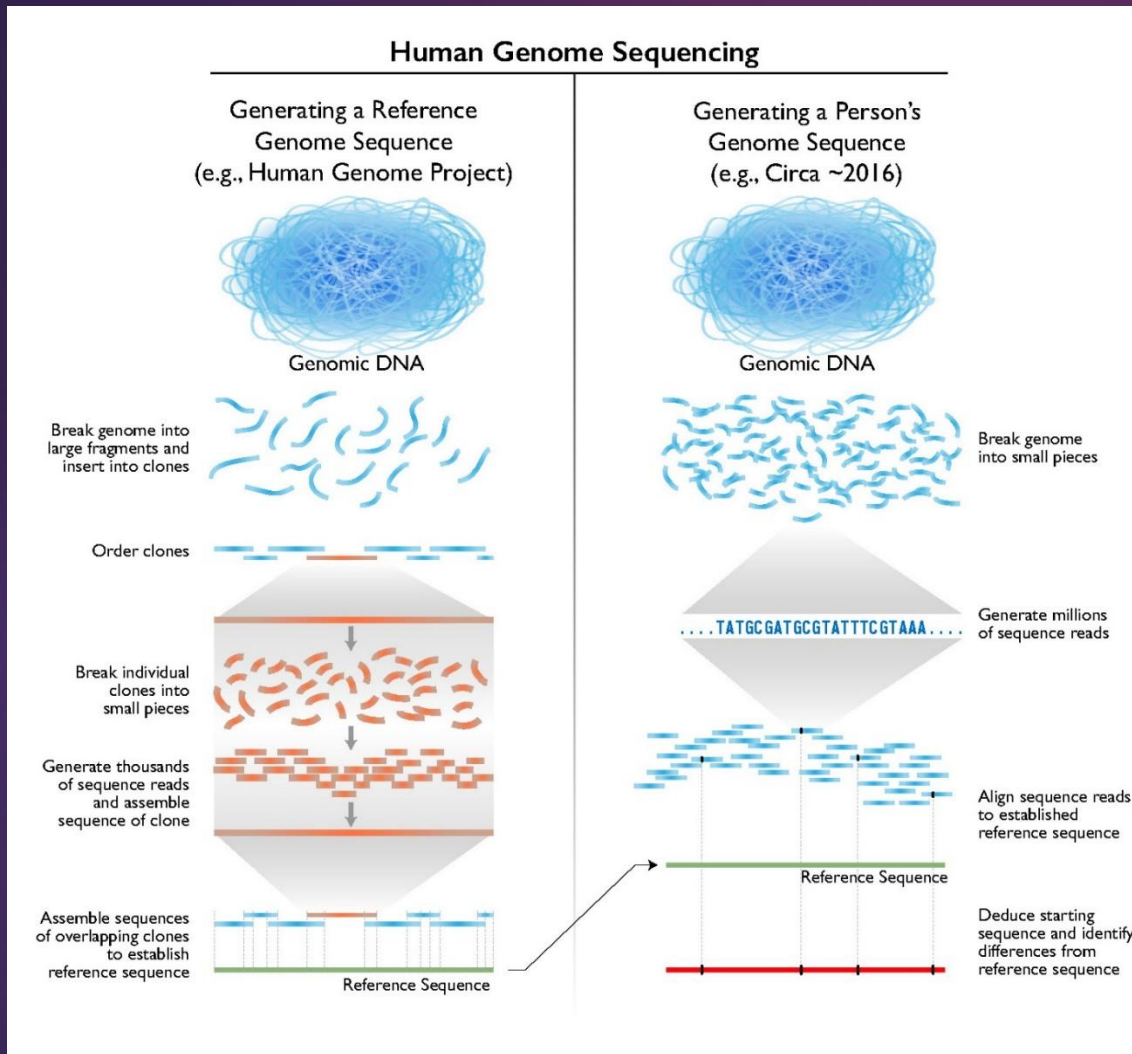
Email: [Luu.p.loi@gmail.com](mailto:Luu.p.loi@gmail.com)

Zalo: 0901802182

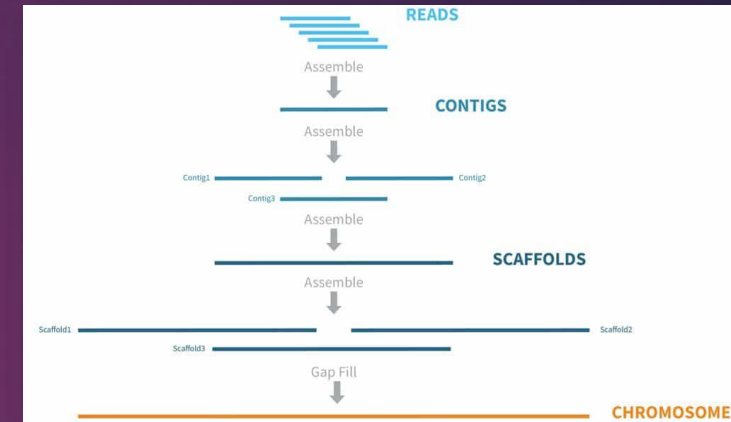
# Content of Lecture

1. Review NGS platforms
2. Review Library Prep
3. Review Genomic Testing

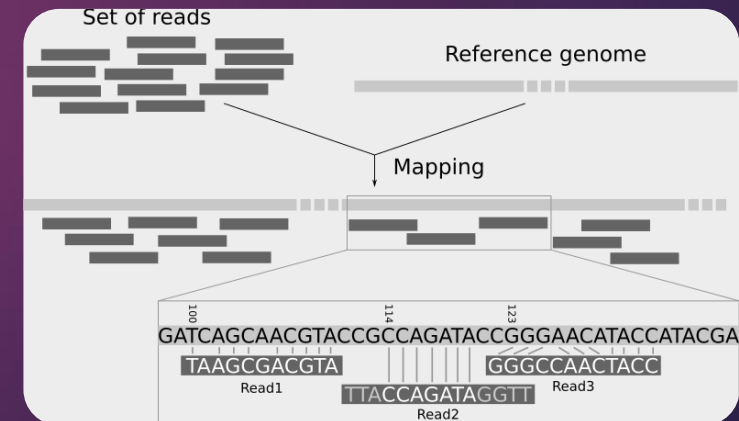
# Giải trình tự gen thế hệ mới (NGS)



## De novo assembly



## Mapping to reference



# Giải trình tự gen thế hệ mới (NGS): Có hệ gen tham chiếu

De novo assembly



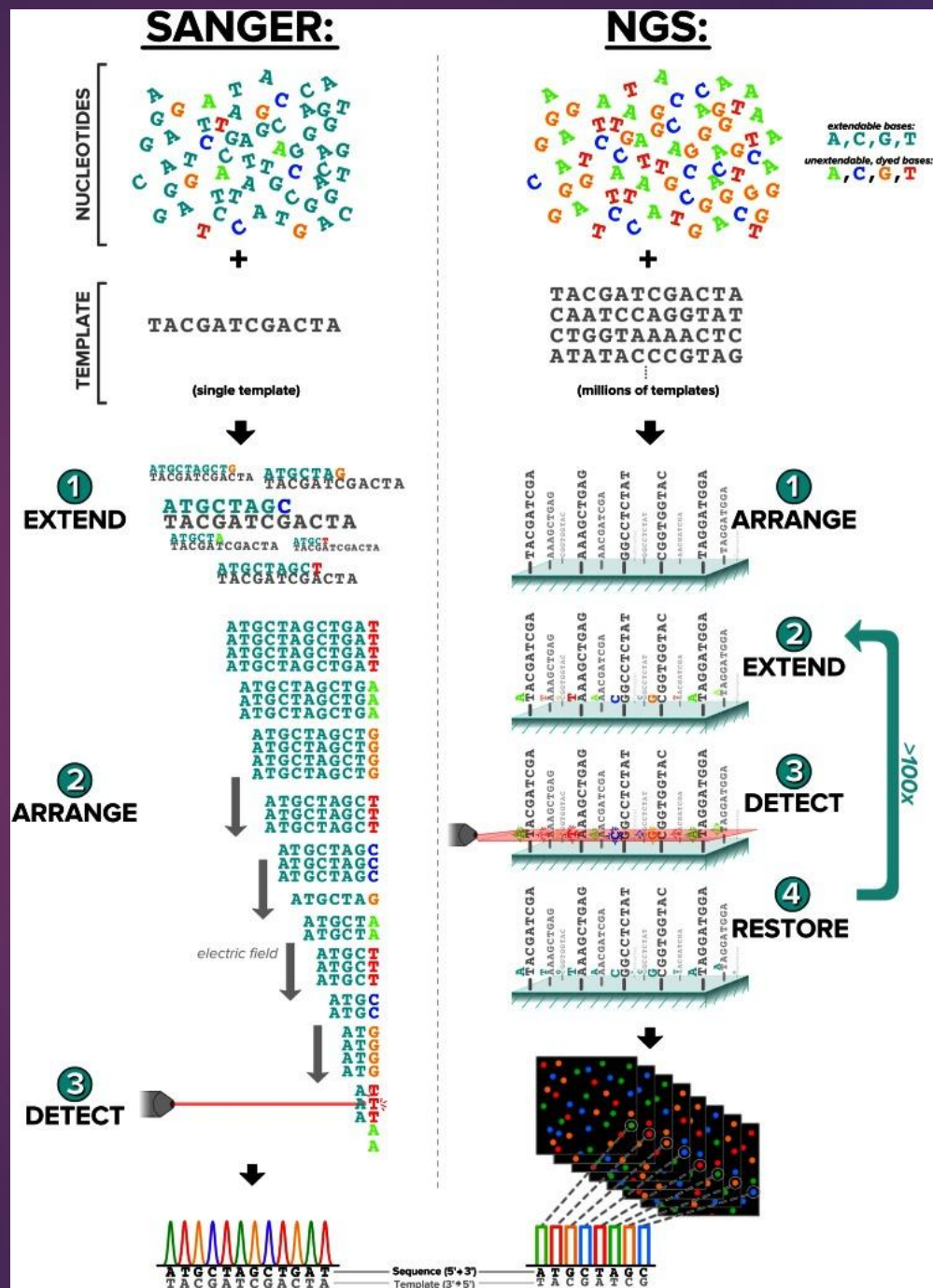
Mapping to reference



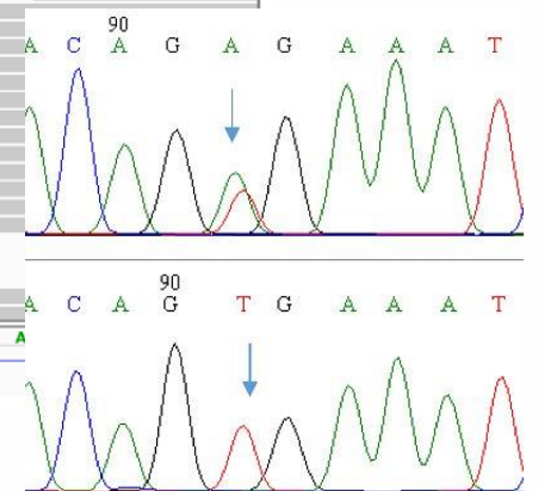


# Giải trình tự gen thế hệ mới (NGS): giải trình tự song song

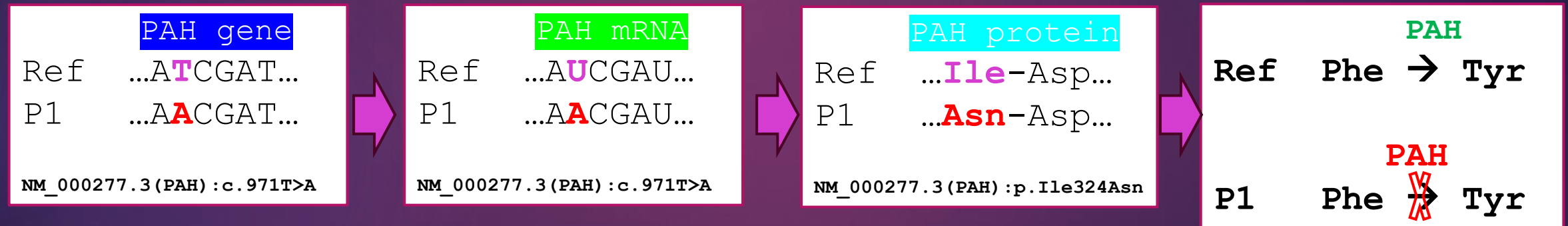
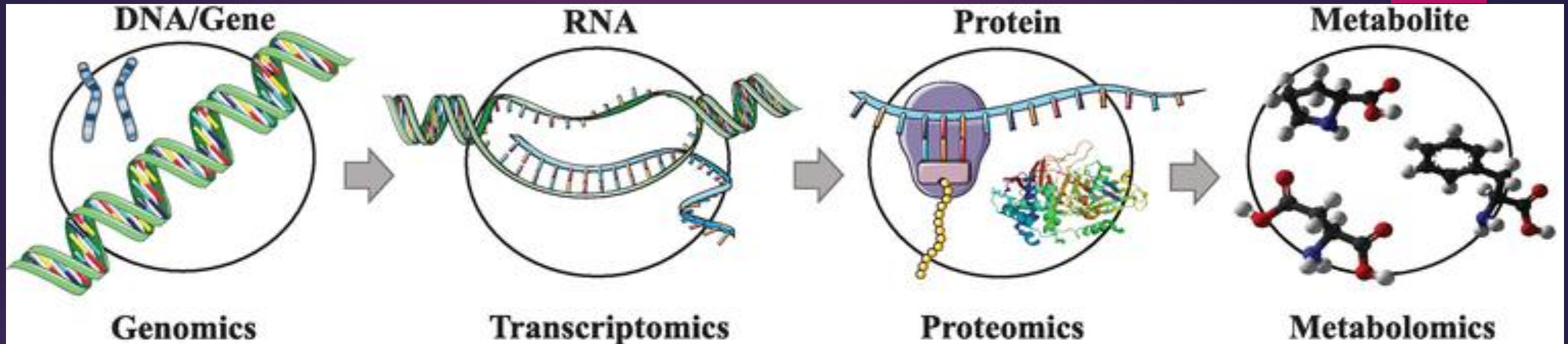
Read1 : CTCGAATACG



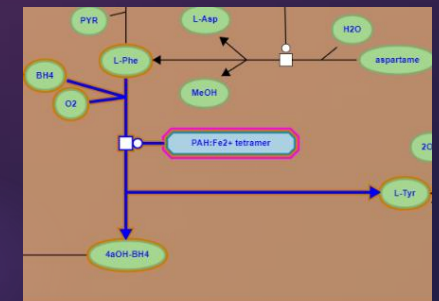
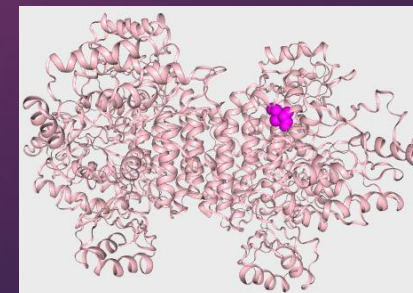
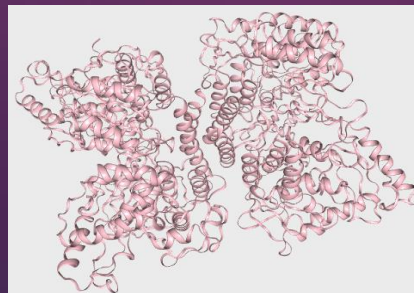
Read1 : CTCGAATACG  
 Read2 : CTCGAATACG  
 Read3 : CTCGAATACG  
 Read4 : CTCGAATACG  
 Read5 : CGCGAATACG  
 Read6 : CGCGAATACG  
 Read7 : CGCGACTACG  
 Read8 : CGCGAATACG



# Quan hệ nhân quả: Biến thể gen và bệnh di truyền



10.5772/intechopen.71769





# Introduction to NGS platforms

## ► Sanger sequencing:

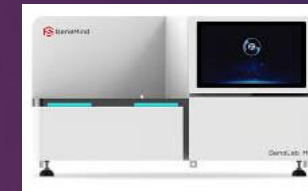
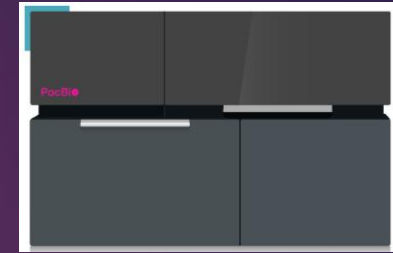
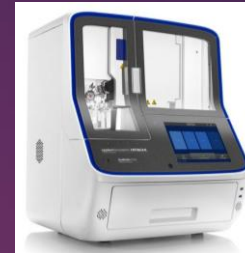
- ❑ <https://youtu.be/XtbIMQ-AI0g?si=Q0CEaWzNvXDOIQWO>

## ► Short read

- ❑ Onso (Pacbio): [https://youtu.be/tFmNO5rVoeM?si=ft4S8N9tbJ1We\\_GA](https://youtu.be/tFmNO5rVoeM?si=ft4S8N9tbJ1We_GA)
- ❑ GeneMind: [https://youtu.be/2SLemogG8v4?si=G\\_qIGS8WjtcAvwV7](https://youtu.be/2SLemogG8v4?si=G_qIGS8WjtcAvwV7)
- ❑ Illumina: <https://youtu.be/-Kw5ov-orzU?si=1izVr8sZk6wPn4ii>
- ❑ MGI: [https://youtu.be/4\\_h4wNIyfk0?si=xrMMF9EEbj-SH19](https://youtu.be/4_h4wNIyfk0?si=xrMMF9EEbj-SH19)
- ❑ Ion Torrent (Thermal Fisher): <https://youtu.be/qso3tdp0IPo?si=ACOAbgHL2dJl0cM8>
- ❑ Aviti (Element Biosciences): <https://youtu.be/pSwWofrRu3Y?si=A9Hid5fvLdcyN8rq>

## ► Long read

- ❑ Revio (Pacbio): <https://youtu.be/mXcDYKnIDE8?si=QUvHRCWHlba7uF5N>
- ❑ ONT: <https://youtu.be/MFVcj4VZHuU?si=hyRf8jU3WZRxcQF>

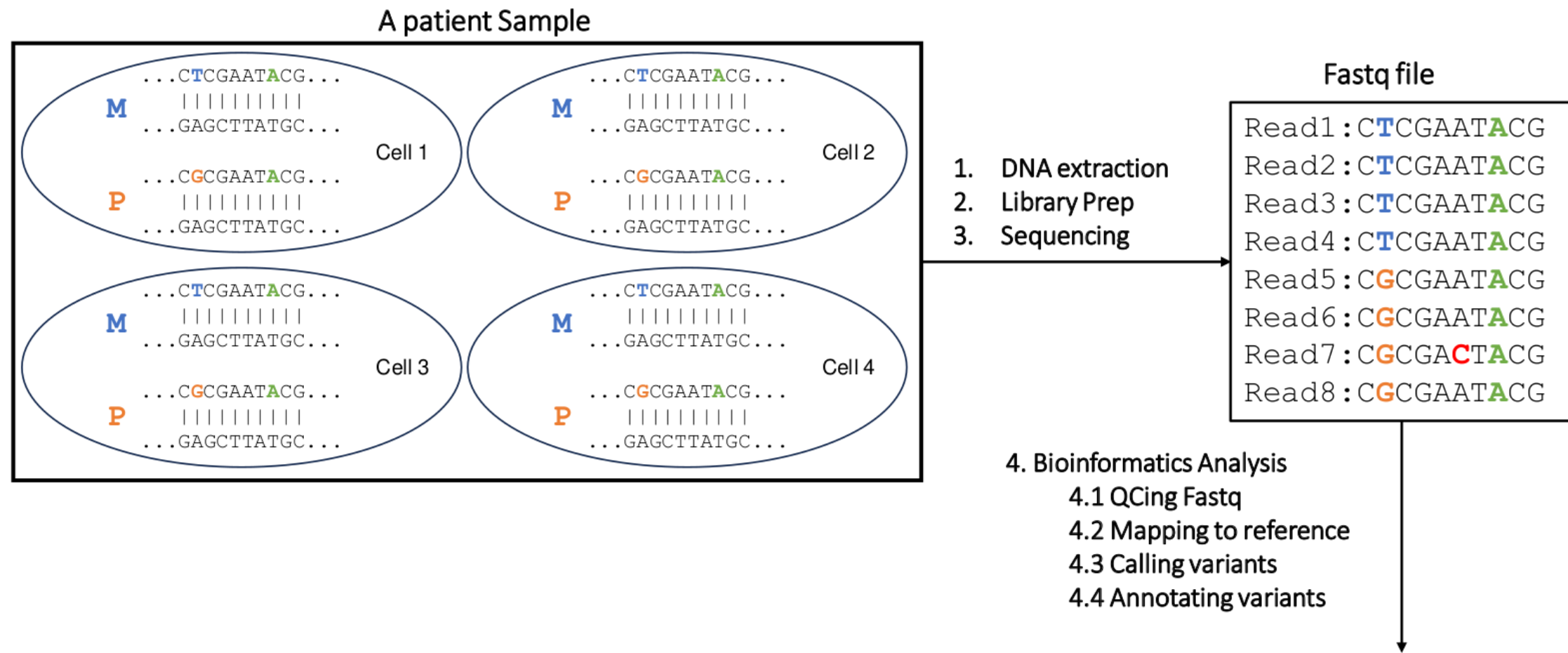




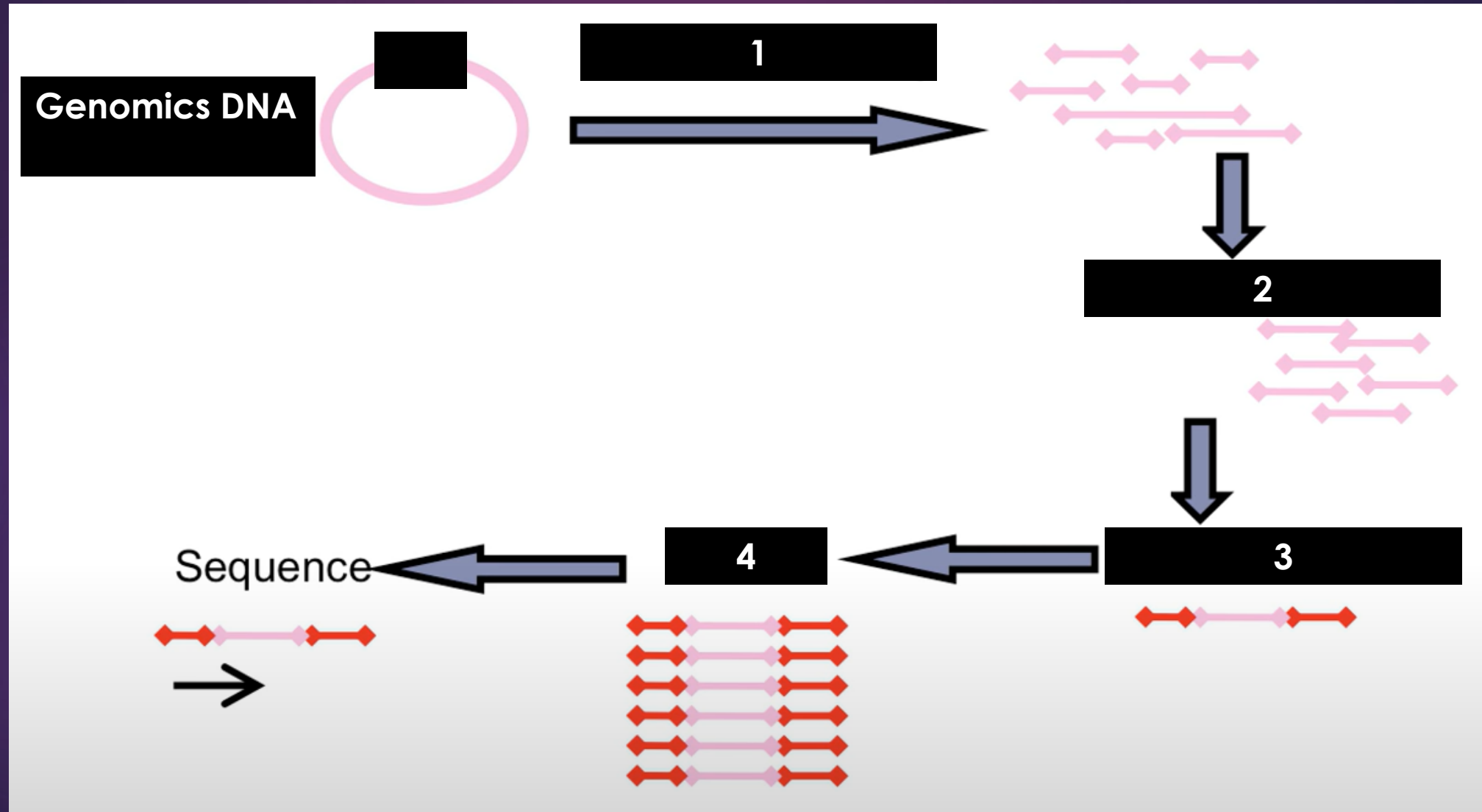


# Quy trình XÉT NGHIỆM gen bằng phương pháp giải trình tự thế hệ mới (NGS)

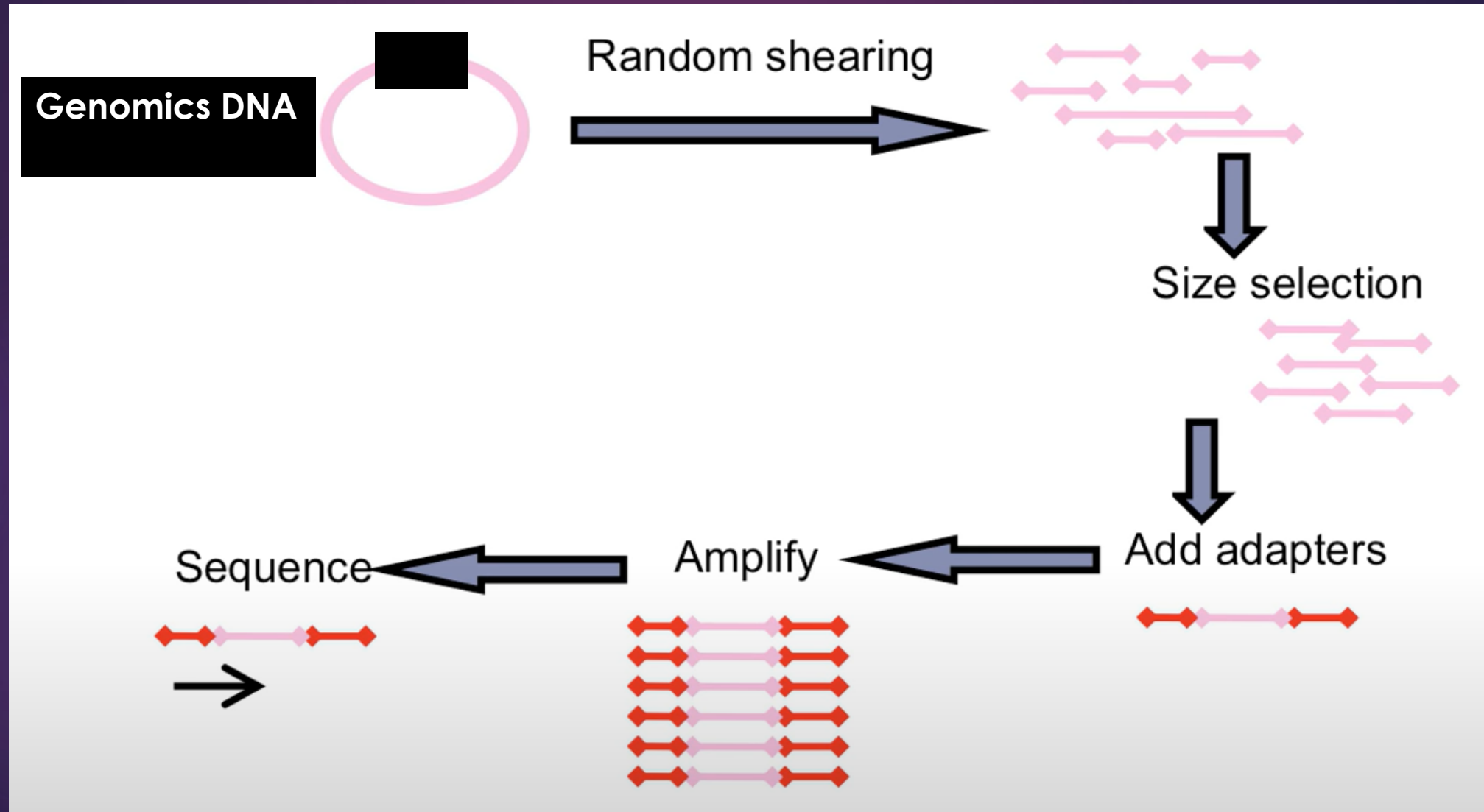
# Các bước trong XÉT NGHIỆM gen bằng phương pháp giải trình tự thế hệ mới (1)



# Whole genome sequencing – Shotgun Library Prep



# Whole genome sequencing – Shotgun Library Prep





# Các bước trong XÉT NGHIỆM gen bằng phương pháp giải trình tự thế hệ mới (2)

## 4.2 Mapping reads to reference

Reference: ...CTCGAATGCG...				
Read1:	C	T	C	Maternal
Read2:	C	T	C	
Read3:	C	T	C	
Read4:	C	T	C	
Read5:	C	G	C	Paternal
Read6:	C	G	C	
Read7:	C	G	C	
Read8:	C	G	C	

Heterozygous

Homozygous

4.3 Calling variants

ANN=G|stop\_gained|HIGH|OR4F5|ENSG00000186092|transcript|ENST0000641515.2|protein\_coding|3/3|c.822T>G|p.Trp274\*|882/2618|822/981|274/326||Pathogenic

ANN=A|frameshift\_variant|HIGH|ZSWIM2|ENSG00000163012|transcript|ENST00000295131.3|protein\_coding|9/9|c.1238G>A|p.Ile413|1293/2451|1238/1902|413/633|;;LOF=(ZSWIM2|ENSG00000163012|1|1.00)

4.4 Annotating variants

```
##fileformat=VCFv4.3
##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">
##FORMAT=<ID=GQ,Number=1,Type=Integer,Description="Genotype Quality">
##FORMAT=<ID=DP,Number=1,Type=Integer,Description="Read Depth">
##FORMAT=<ID=AD,Number=2,Type=Integer,Description="Read depth for each allele">
```

#CHROM	POS	ID	REF	ALT	QUAL	FILTER	FORMAT	Sample1
20	14370	rs6054257	T	G	129	PASS	GT:GQ:DP:AD	0/1:48:8:4,4
20	17330	.	G	A	150	PASS	GT:GQ:DP:AD	1/1:49:8:8,8

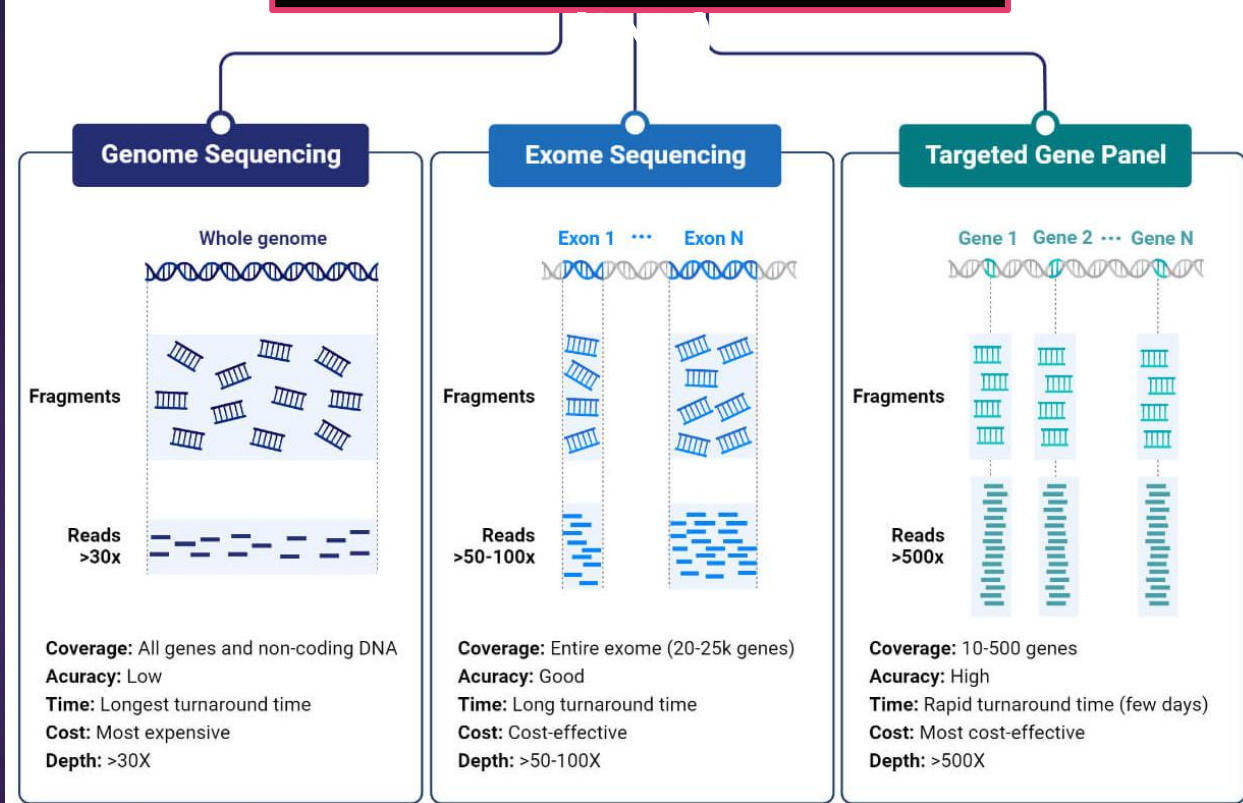
# Hội đồng Hệ Gen: phiên giải và hội chẩn những biến thể trong báo cáo kết quả NGS



<https://ggba.swiss/en/the-first-medical-genomics-center-opens-in-geneva/>

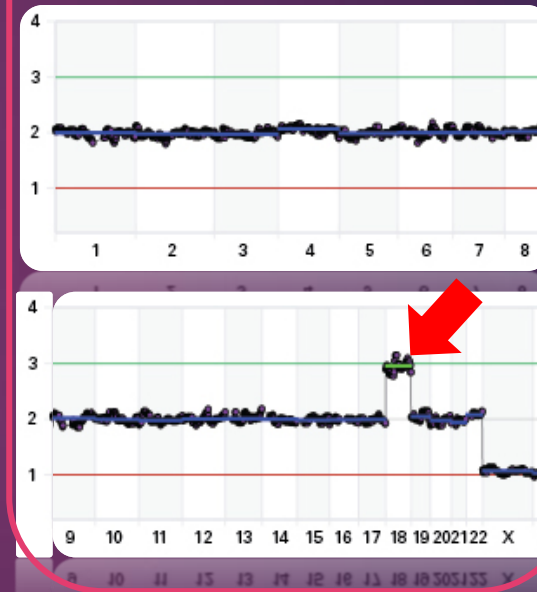
# Ứng dụng giải trình tự gen thế hệ mới trong lâm sàng

## Single Nucleotide Variant

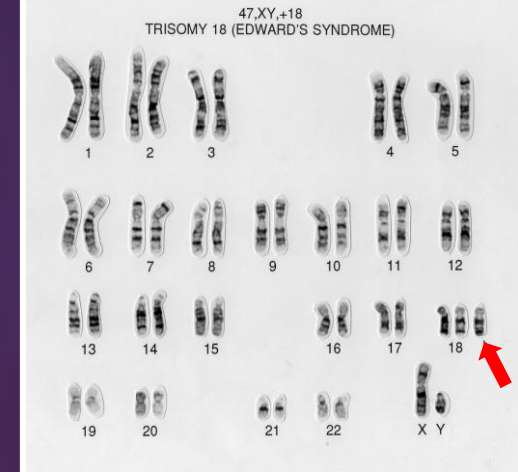


<https://microbenotes.com/next-generation-sequencing-ngs/>

## Copy Number Variation (CNV) Low-Pass Genome Sequencing



## Karyotype



<https://wellcomecollection.org/search/images?query=eacahzt2u#>





# Xin chân thành cảm ơn!

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