

Trường Đại học Khoa học tự nhiên, Đại học Quốc gia Tp. Hồ Chí Minh
Trung tâm Khoa học và Công nghệ Sinh học

Microbiome Bioinformatics Analysis

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Zalo: 0901802182

27/06/2025

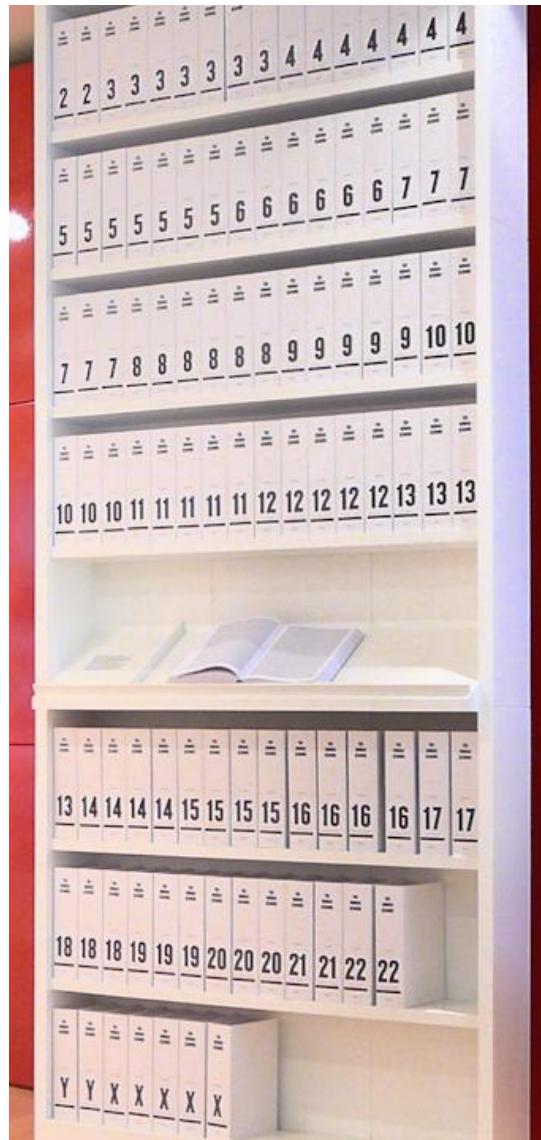
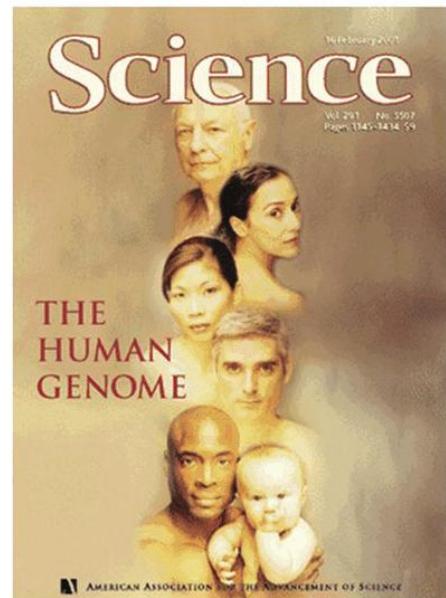
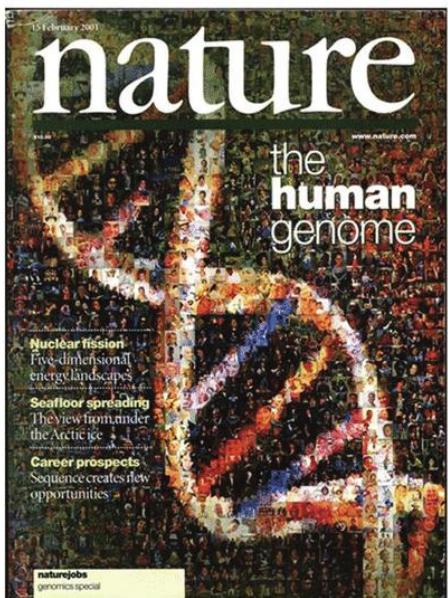
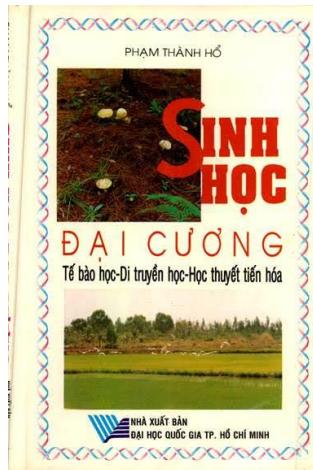
STT	Nội Dung	Giảng Viên	Thời lượng
1	Introduction to Next-Generation Sequencing (NGS), Microbiome and Applications	TS. Lưu Phúc Lợi	30 phút
2	Ảnh hưởng của việc bổ sung lysophospholipid vào khẩu phần ăn lipid thấp lên hệ vi sinh vật đường ruột của tôm thẻ chân trắng (<i>Litopenaeus vannamei</i>)	Nguyễn Quang Khải	30-45 phút
3	Nghiên cứu đặc điểm của hệ vi sinh vật trong đất nông nghiệp ở Lâm Đồng, Việt Nam	Nguyễn Mạnh Hùng	30-45 phút
4	Phát hiện hệ vi khuẩn và gen kháng kháng sinh trong nhiễm trùng huyết bằng phương pháp giải trình tự thế hệ mới	Hà Gia Huy	30-45 phút
5	Thảo luận và trả lời câu hỏi		30 phút

Nội dung

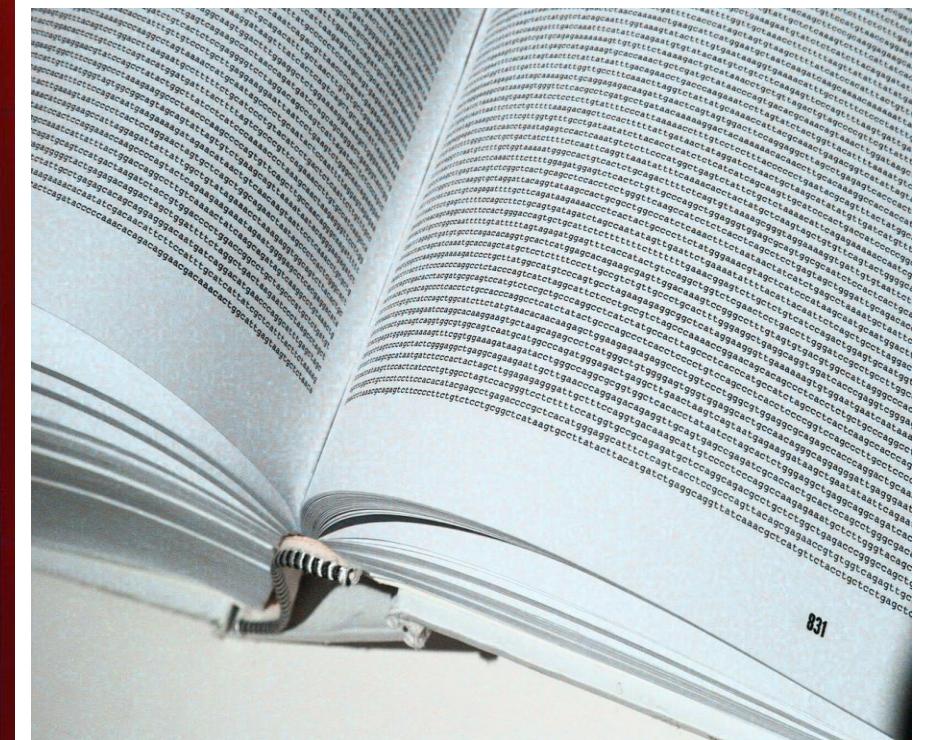
1. Giới thiệu về Dự án bộ gen người
2. Giới thiệu kỹ thuật giải trình tự thế hệ mới (NGS)
3. Quy trình **XÉT NGHIỆM** gen bằng phương pháp giải trình tự thế hệ mới
4. Giải trình tự gen toàn bộ hệ gen của một vi sinh vật
5. Giải trình tự gen toàn bộ hệ vi sinh vật

Giới thiệu về Dự án bộ gen người

Dự án hệ gen người (1990-2003)

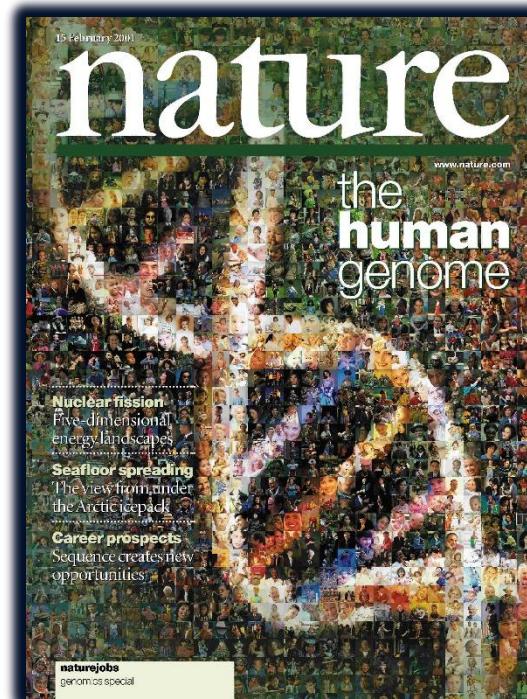


Năm 2000

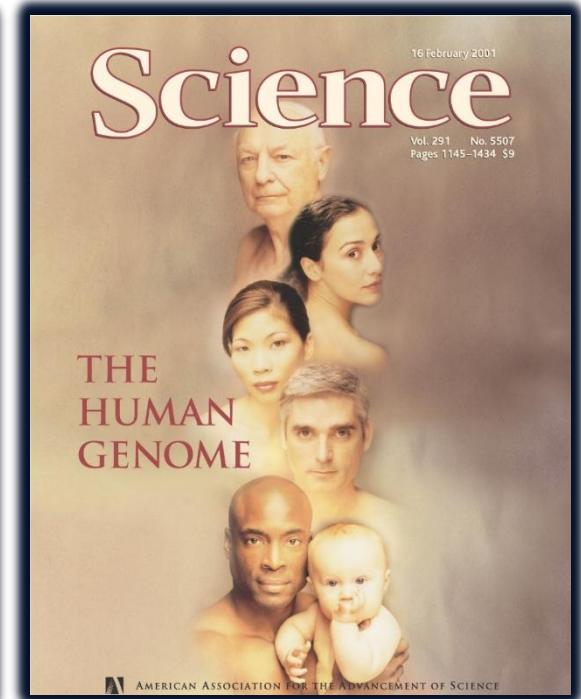


Dự án hệ gen người HGP (Oct 1990 - April 2003)

- Năm 2003, dự án hệ gen người HGP đã tạo ra một chuỗi trình tự gần 3 tỉ nucleotide chiếm hơn 90% bộ gen người.
- Đây là chuỗi gen hoàn chỉnh nhất có thể đạt được với công nghệ giải trình tự DNA thời điểm đầu những năm 2000.
=> Sự phát triển của công nghệ giải trình tự thế hệ mới (NGS).



HGP Paper



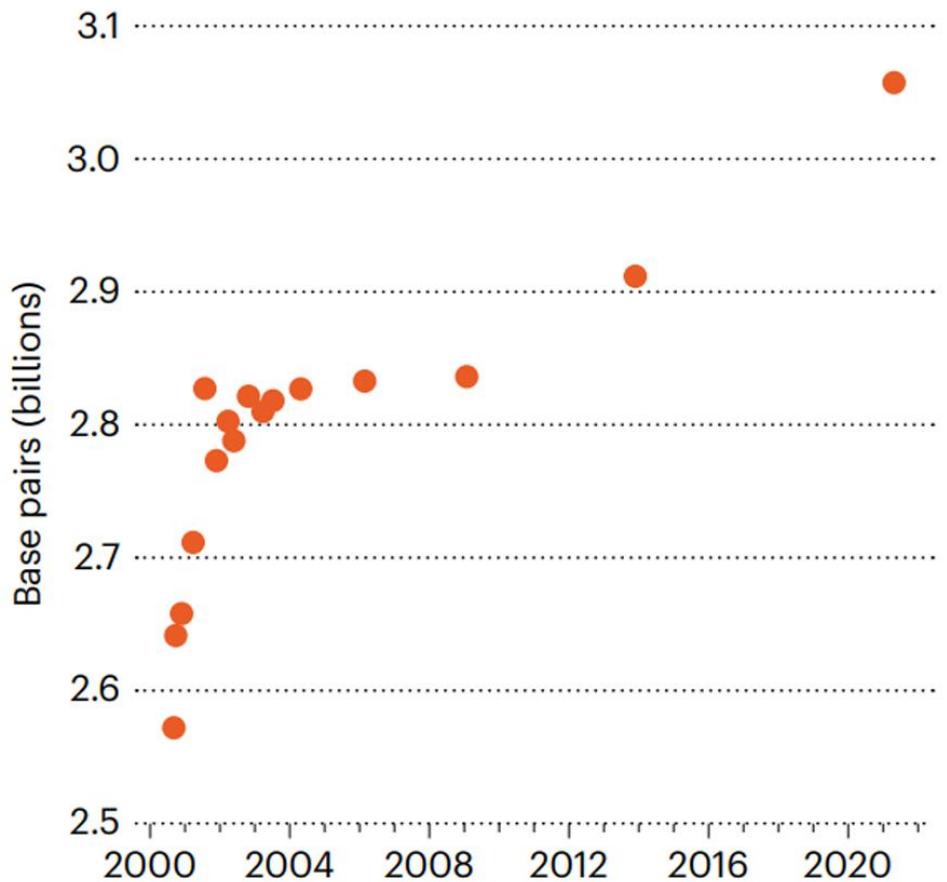
Venter/Celera Paper

A COMPLETE HUMAN GENOME IS CLOSE: HOW THE GAPS WERE FILLED

Researchers added 200 million DNA base pairs and 115 genes – but they've yet to finish the Y chromosome.

COMPLETING THE HUMAN GENOME

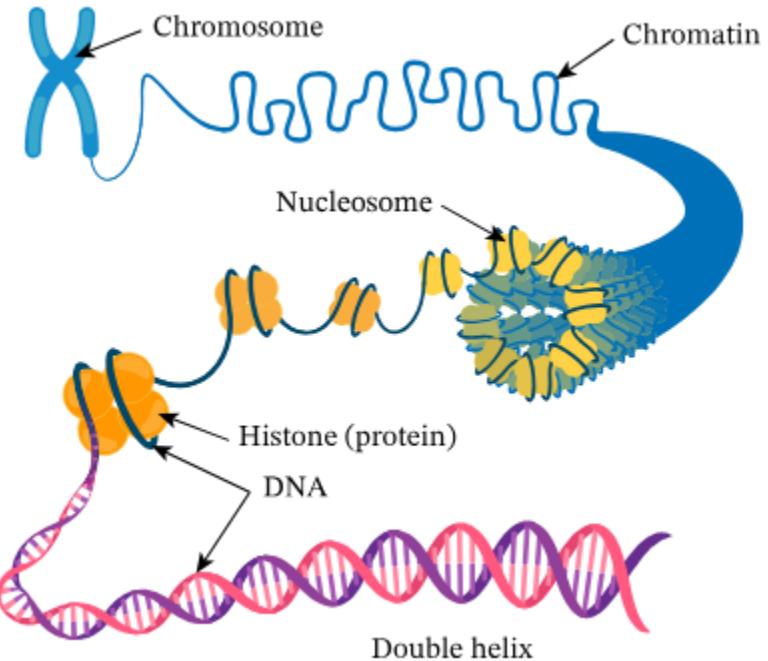
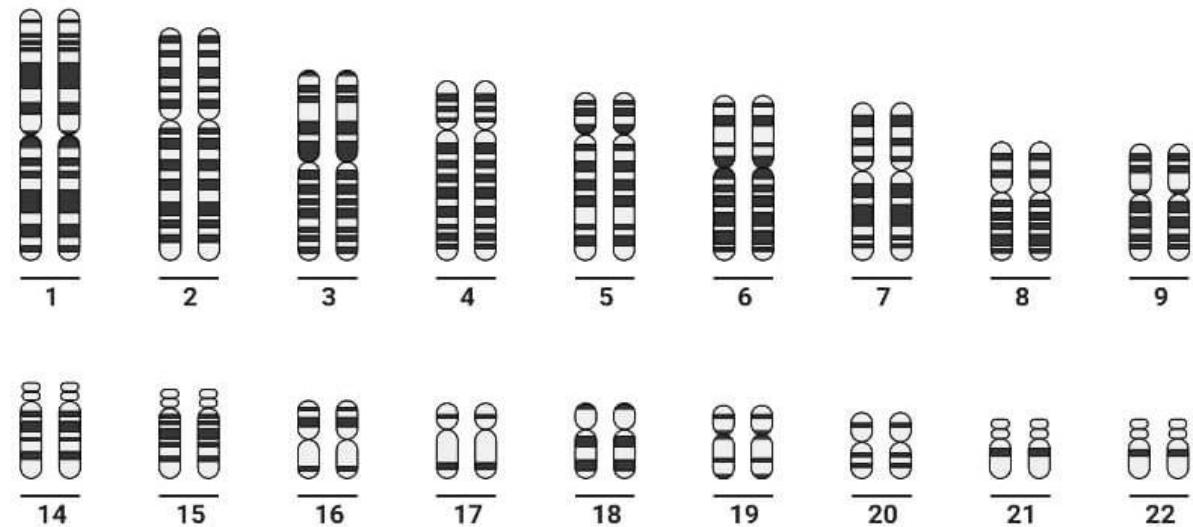
Researchers have been filling in incompletely sequenced parts of the human reference genome for 20 years, and have now almost finished it, with 3.05 billion DNA base pairs.



0.3% of sequence might still have errors. Includes X but not Y chromosome. Count excludes mitochondrial DNA.

SOURCE: ADAM PHILLIPY

Human Karyotype

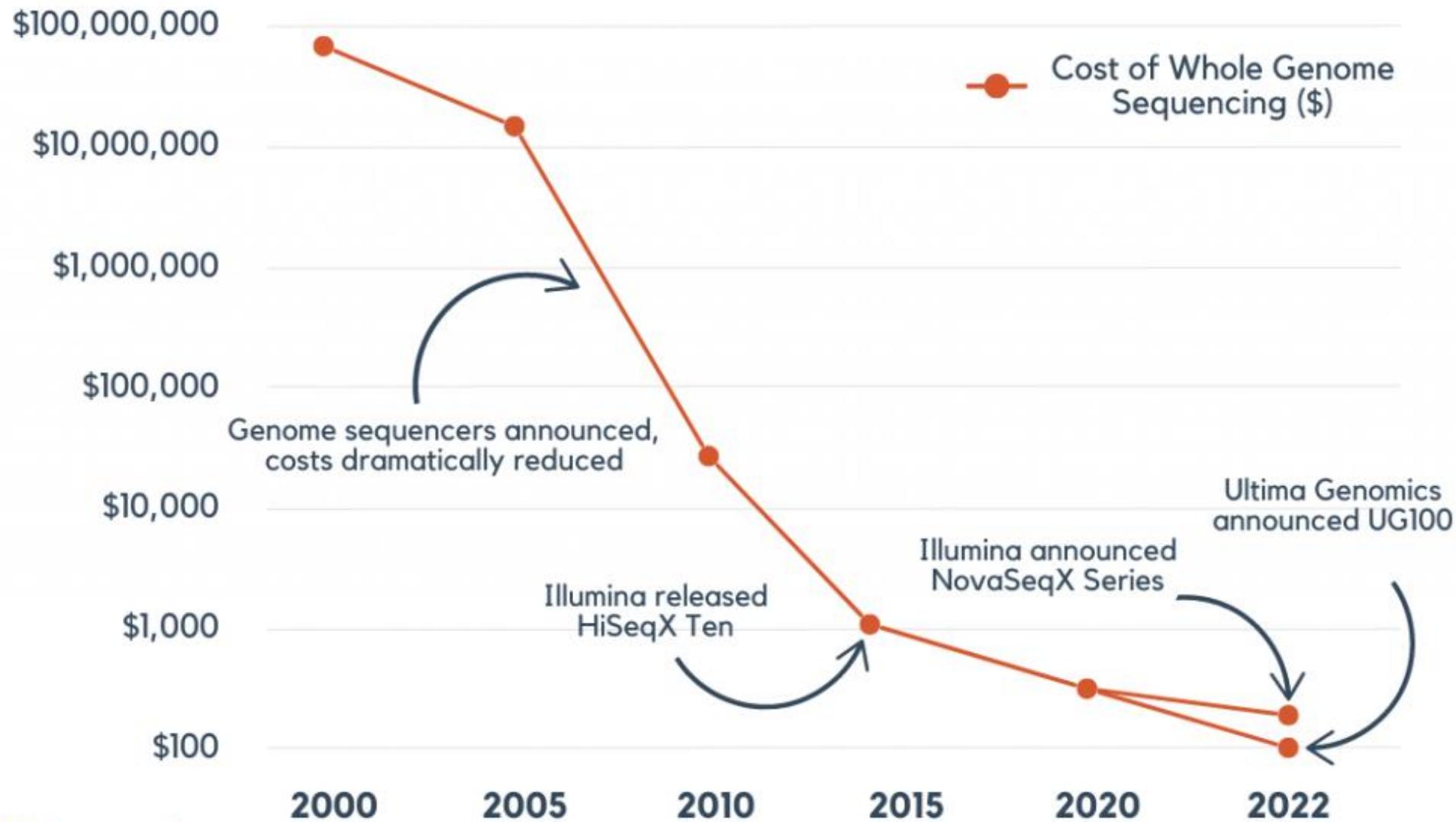


Hệ Gen người



48541 agcccttcaa agaaatgttc tcagcaggca tggagccag gacttgctcc ctttgggtag
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Decreasing Genome Sequencing Costs

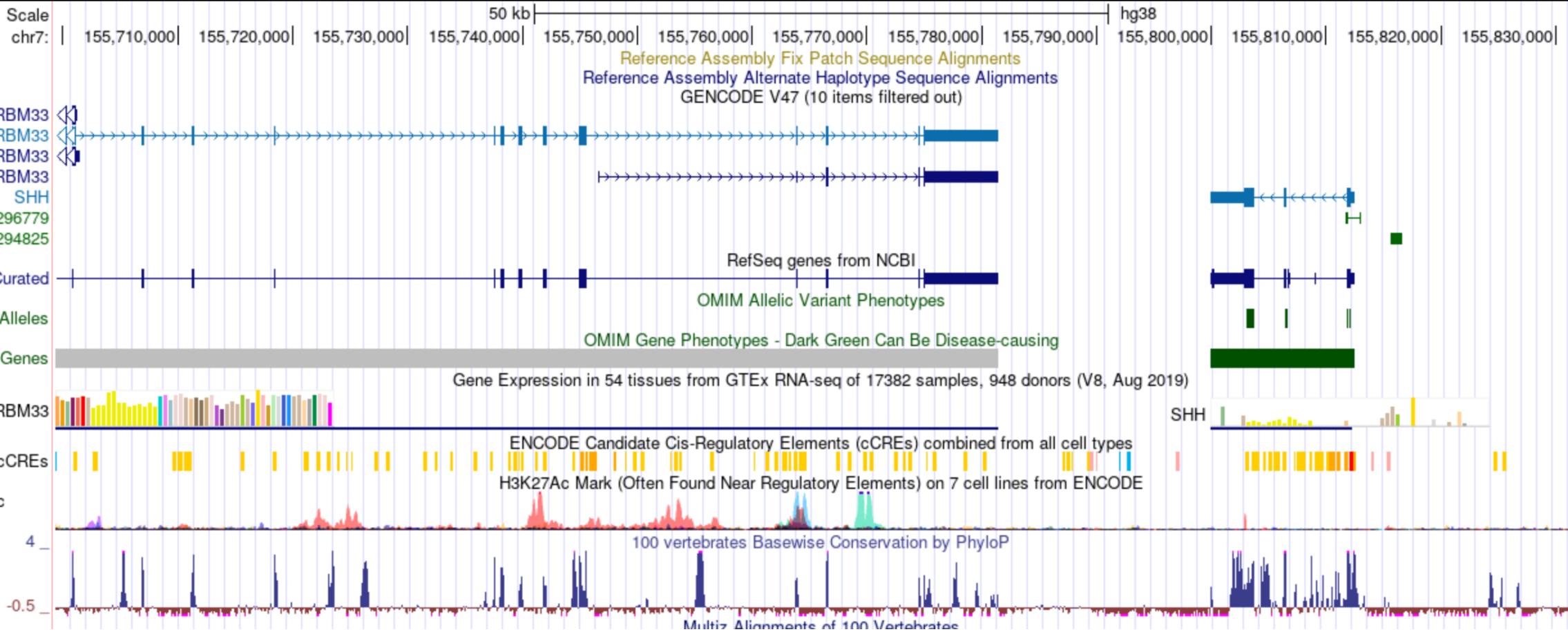
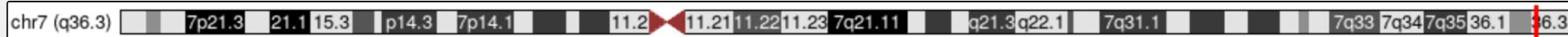


Gene Annotation

[Multi-region](#)

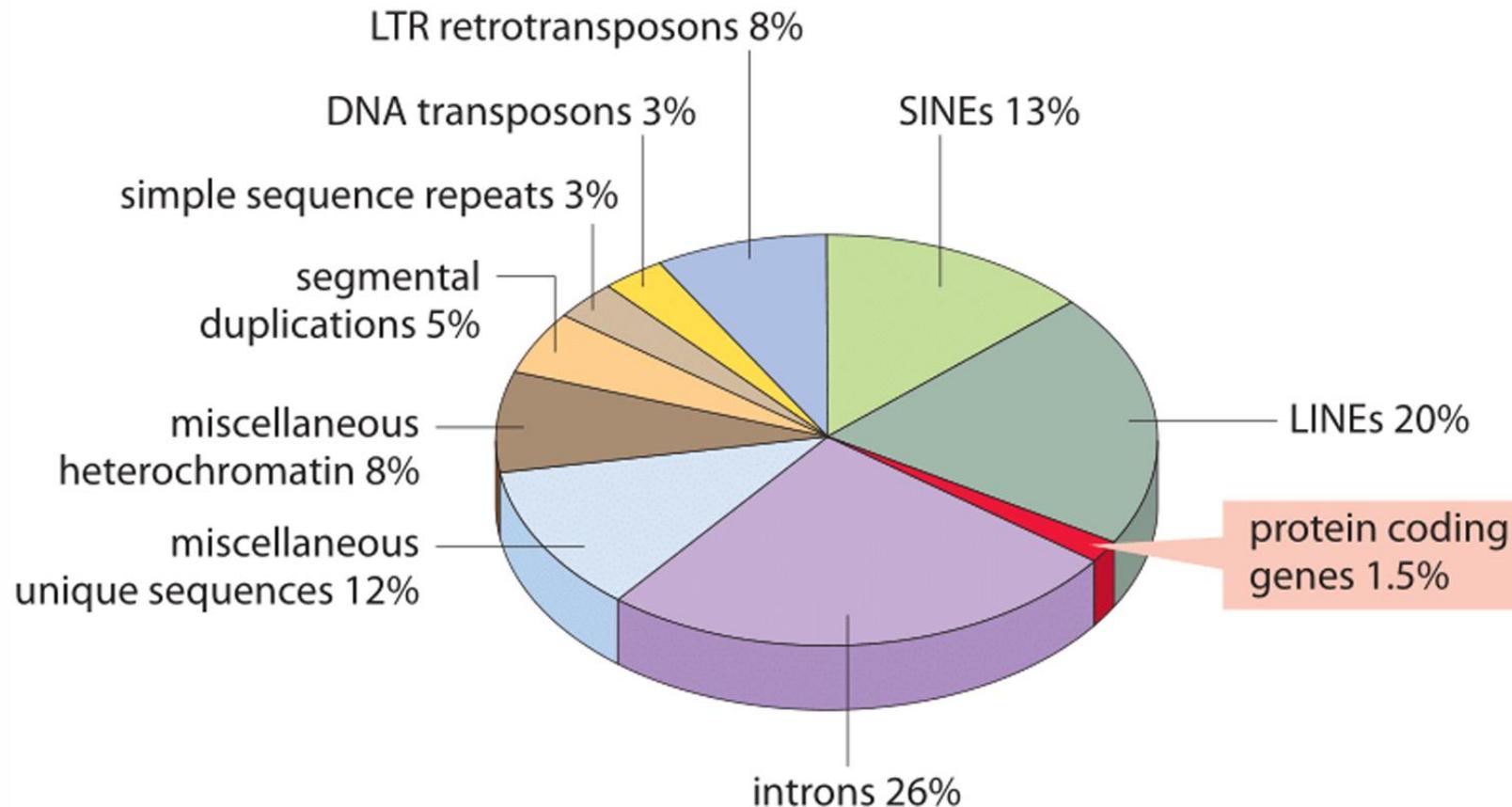
chr7:155,699,333-155,832,762 133,430 bp.

gene, chromosome range, search terms, help pages, see exam

[Search](#)[Examples](#)

Gene Annotation

main components of the human genome



GENCODE: Cơ sở dữ liệu chì giải gen

GENCODE

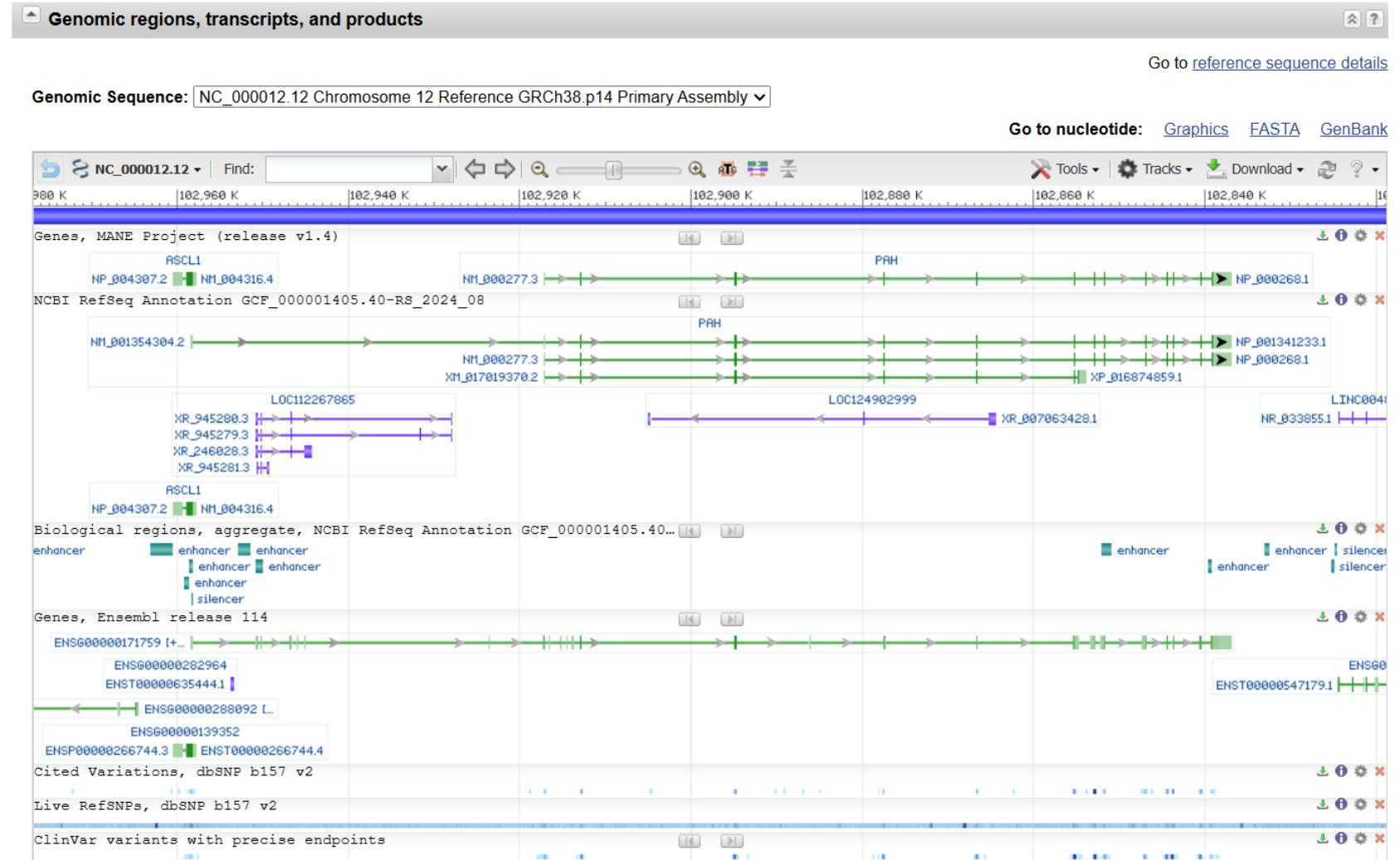
2009

Human Mouse How to access data FAQ Documentation About us

HUMAN
GENCODE 48 (May 2025)

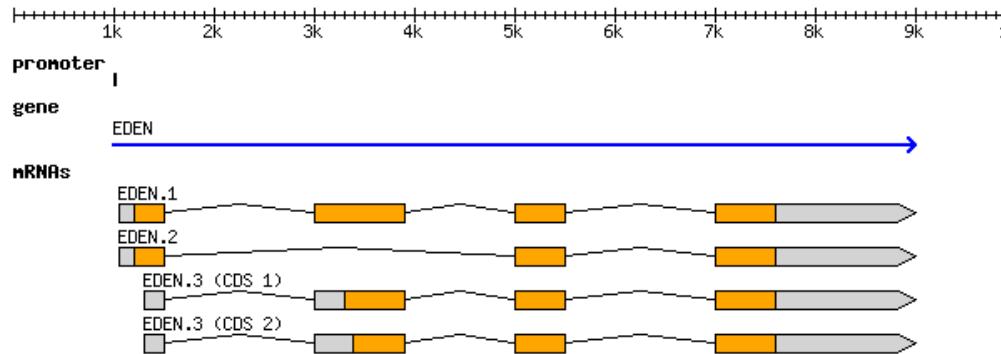


MOUSE
GENCODE M37 (May 2025)

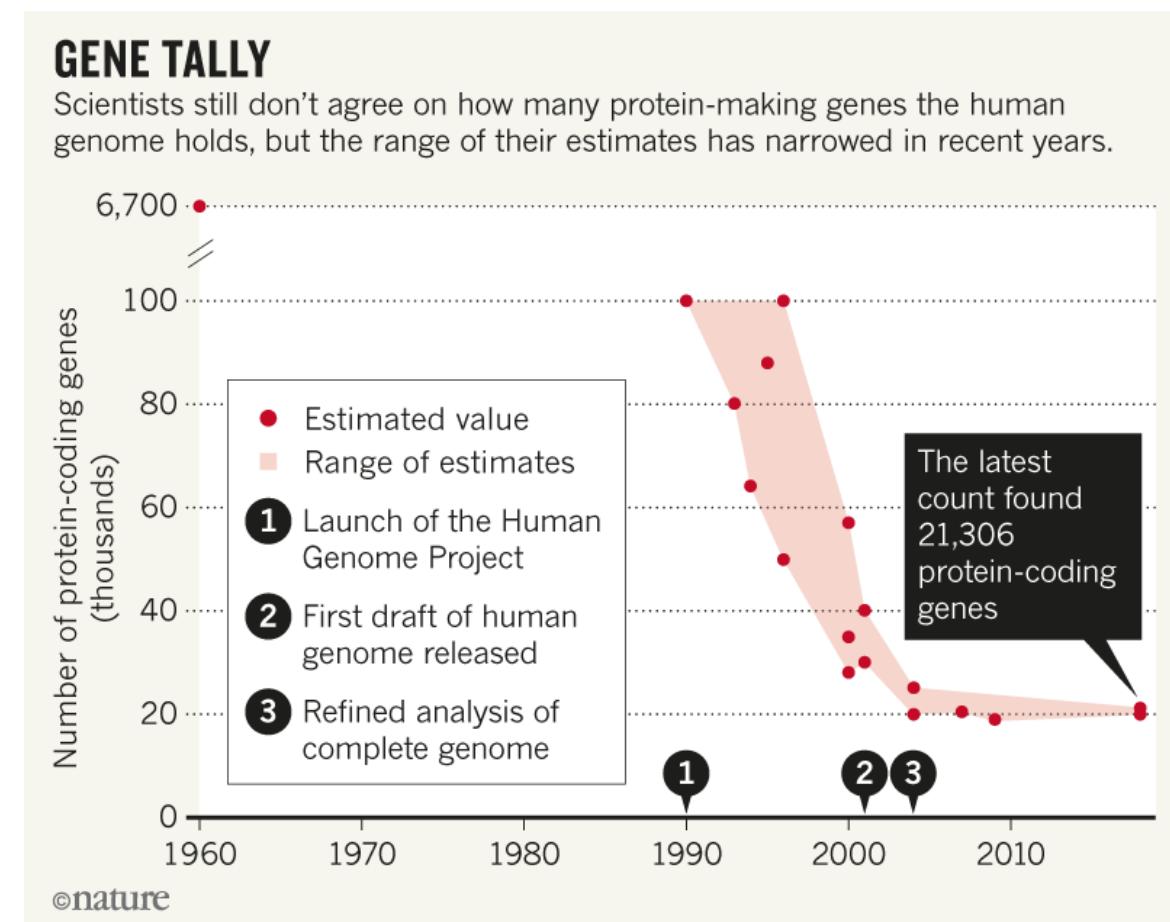



Các nhà tin sinh học chia sẻ “công thức” của họ

GTf (Gene Transfer Format) and The number of Human Genes



```
0 ##gff-version 3.2.1
1 ##sequence-region ctg123 1 1497228
2 ctg123 . gene      1000 9000 . + . ID=gene00001;Name=EDEN
3 ctg123 . TF_binding_site 1000 1012 . + . ID=tfbs00001;Parent=gene00001
4 ctg123 . mRNA      1050 9000 . + . ID=mRNA00001;Parent=gene00001;Name=EDEN.1
5 ctg123 . mRNA      1050 9000 . + . ID=mRNA00002;Parent=gene00001;Name=EDEN.2
6 ctg123 . mRNA      1300 9000 . + . ID=mRNA00003;Parent=gene00001;Name=EDEN.3
7 ctg123 . exon      1300 1500 . + . ID=exon00001;Parent=mRNA00003
8 ctg123 . exon      1050 1500 . + . ID=exon00002;Parent=mRNA00001,mRNA00002
9 ctg123 . exon      3000 3902 . + . ID=exon00003;Parent=mRNA00001,mRNA00003
10 ctg123 . exon     5000 5500 . + . ID=exon00004;Parent=mRNA00001,mRNA00002,mRNA00003
11 ctg123 . exon     7000 9000 . + . ID=exon00005;Parent=mRNA00001,mRNA00002,mRNA00003
12 ctg123 . CDS      1201 1500 . + 0 ID=cds00001;Parent=mRNA00001;Name=edenprotein.1
13 ctg123 . CDS      3000 3902 . + 0 ID=cds00001;Parent=mRNA00001;Name=edenprotein.1
14 ctg123 . CDS      5000 5500 . + 0 ID=cds00001;Parent=mRNA00001;Name=edenprotein.1
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16 ctg123 . CDS      1201 1500 . + 0 ID=cds00002;Parent=mRNA00002;Name=edenprotein.2
17 ctg123 . CDS      5000 5500 . + 0 ID=cds00002;Parent=mRNA00002;Name=edenprotein.2
18 ctg123 . CDS      7000 7600 . + 0 ID=cds00002;Parent=mRNA00002;Name=edenprotein.2
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20 ctg123 . CDS      5000 5500 . + 1 ID=cds00003;Parent=mRNA00003;Name=edenprotein.3
21 ctg123 . CDS      7000 7600 . + 1 ID=cds00003;Parent=mRNA00003;Name=edenprotein.3
22 ctg123 . CDS      3391 3902 . + 0 ID=cds00004;Parent=mRNA00003;Name=edenprotein.4
23 ctg123 . CDS      5000 5500 . + 1 ID=cds00004;Parent=mRNA00003;Name=edenprotein.4
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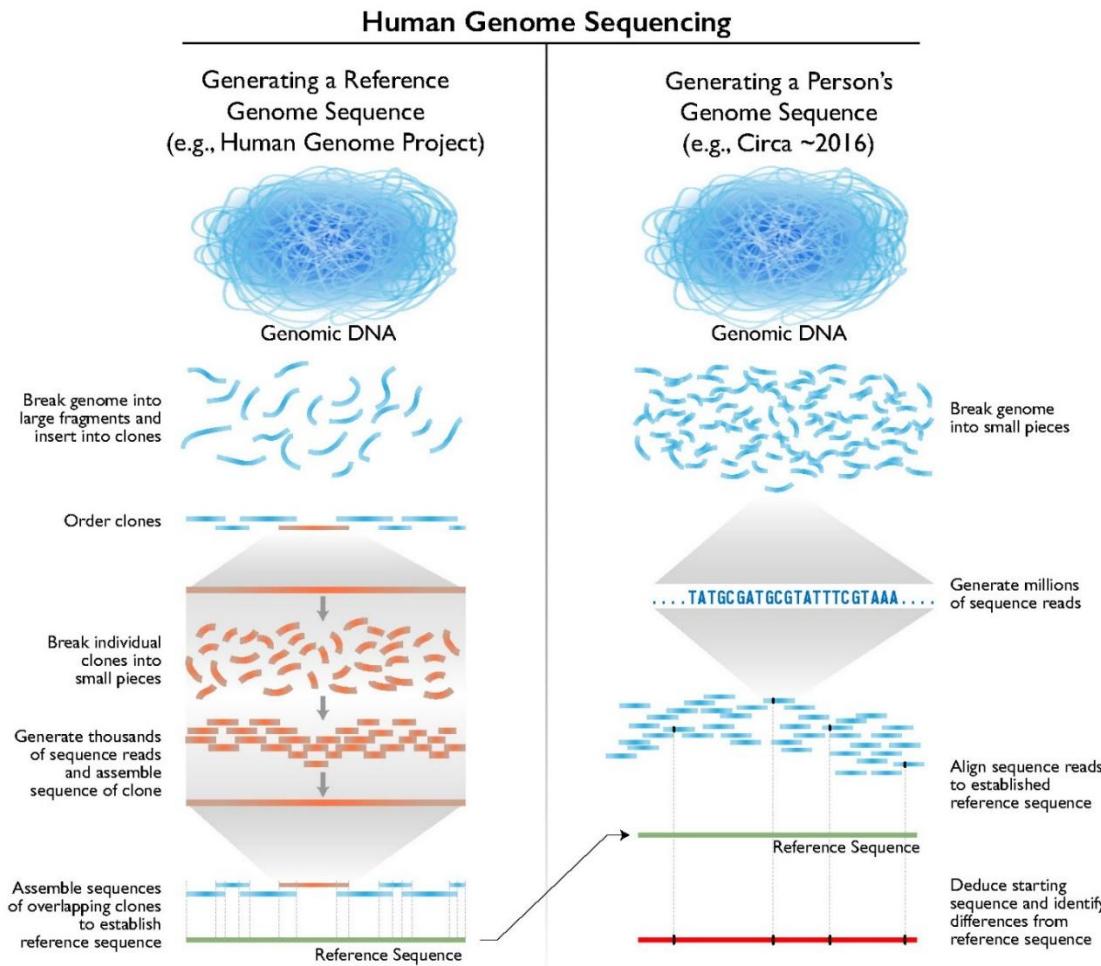


<https://www.nature.com/articles/d41586-018-05462-w>

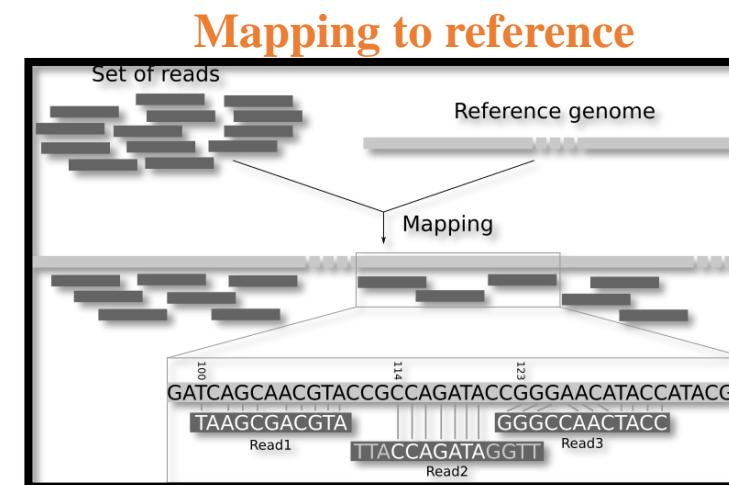
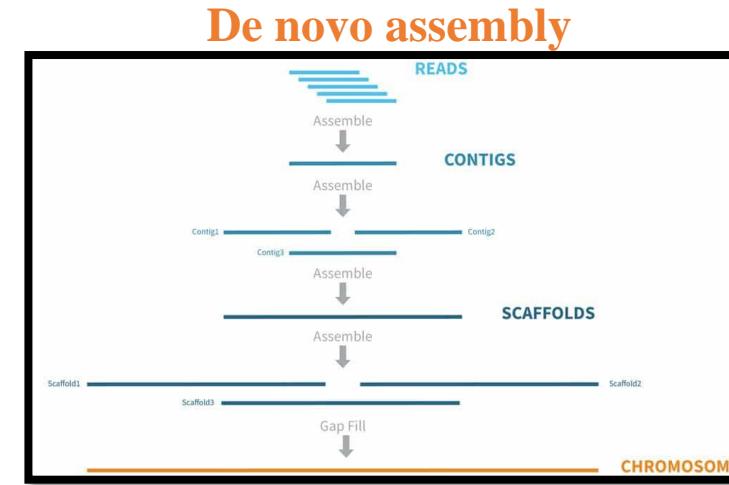
Giới thiệu về

**GIẢI TRÌNH TỰ GEN THẺ
HỆ MỚI**

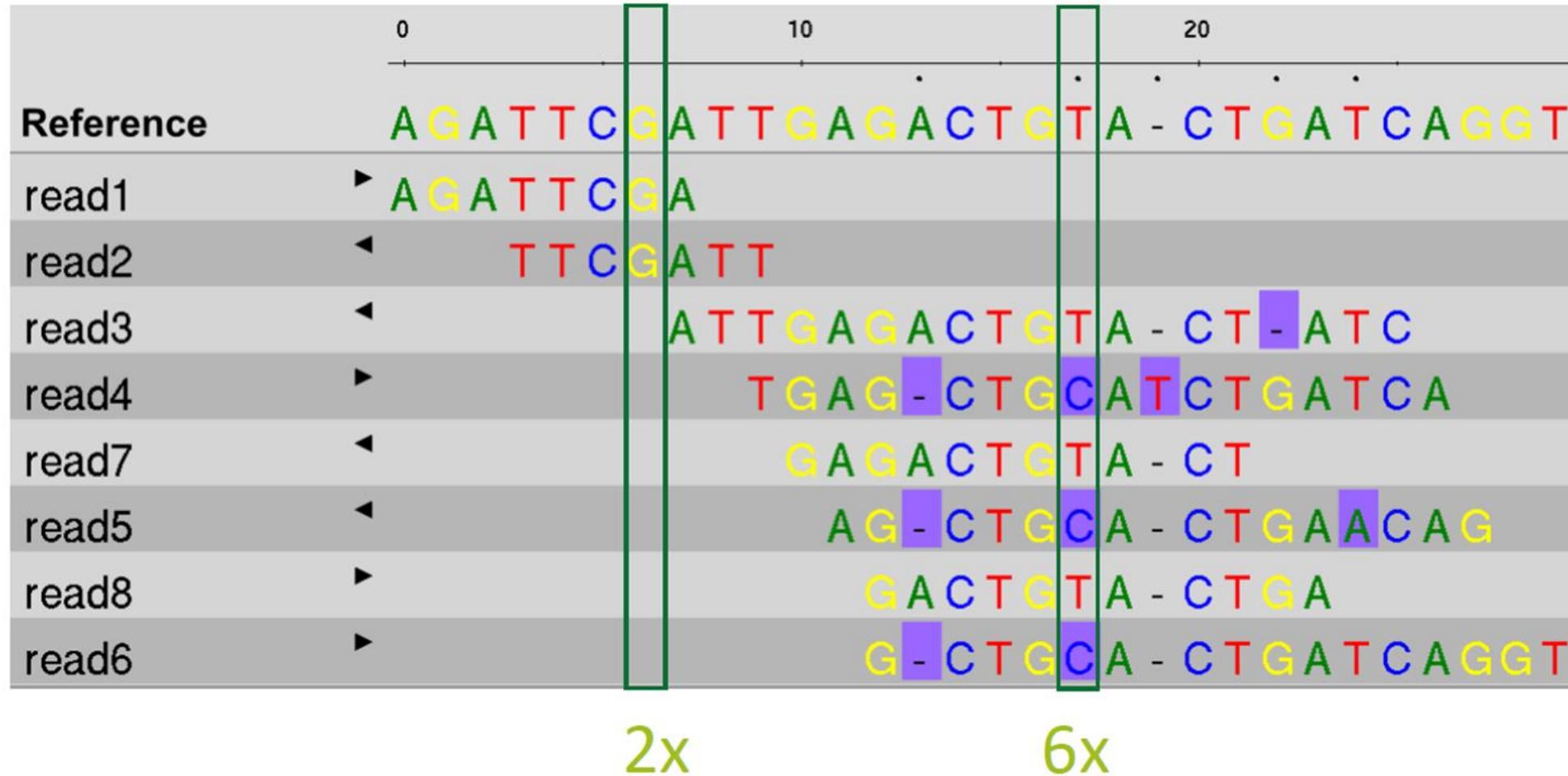
Giải trình tự gen thế hệ mới (NGS)



<https://www.genome.gov/about-genomics/fact-sheets/Sequencing-Human-Genome-cost>



Kết quả của mapping



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

De novo assembly

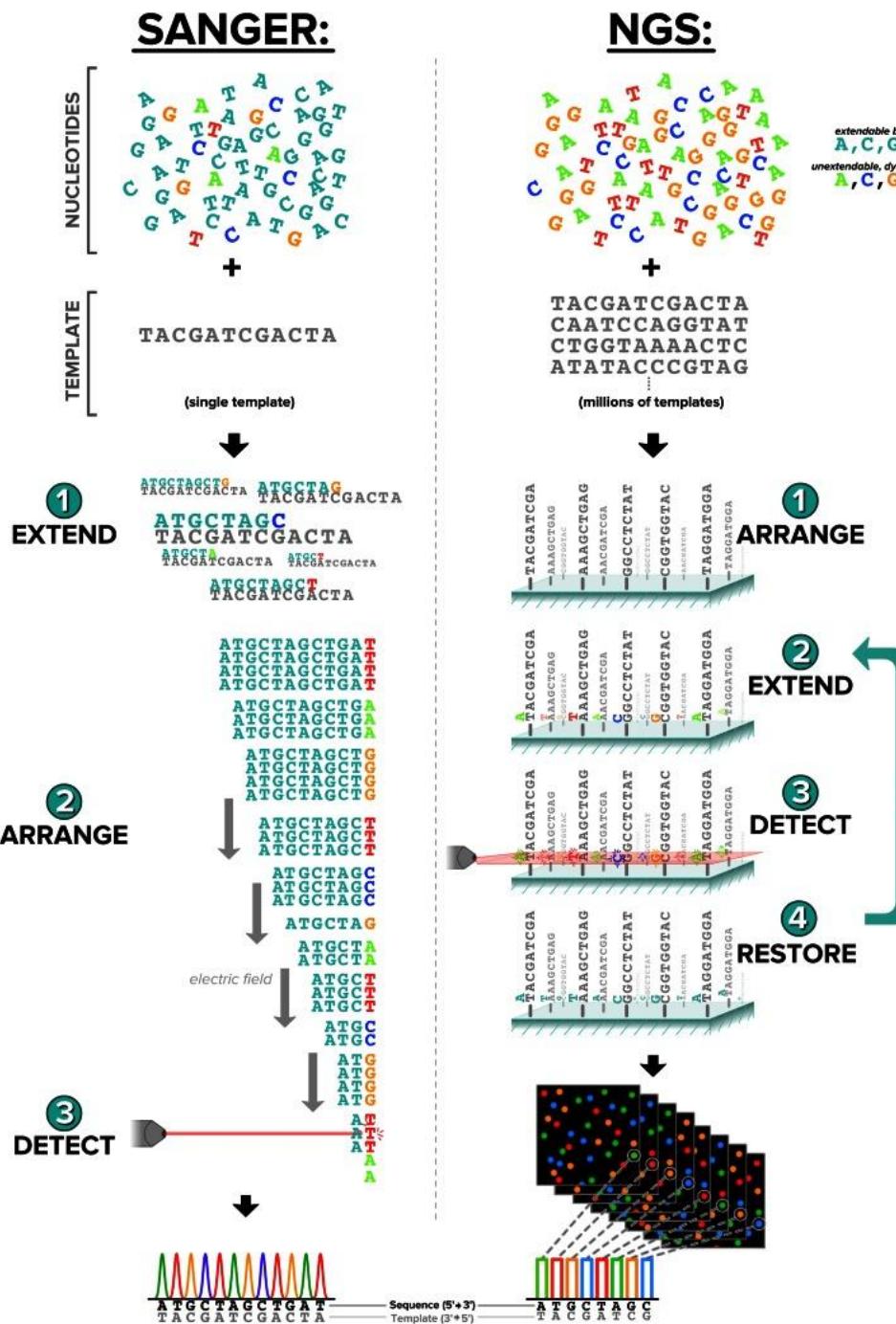


Mapping to reference



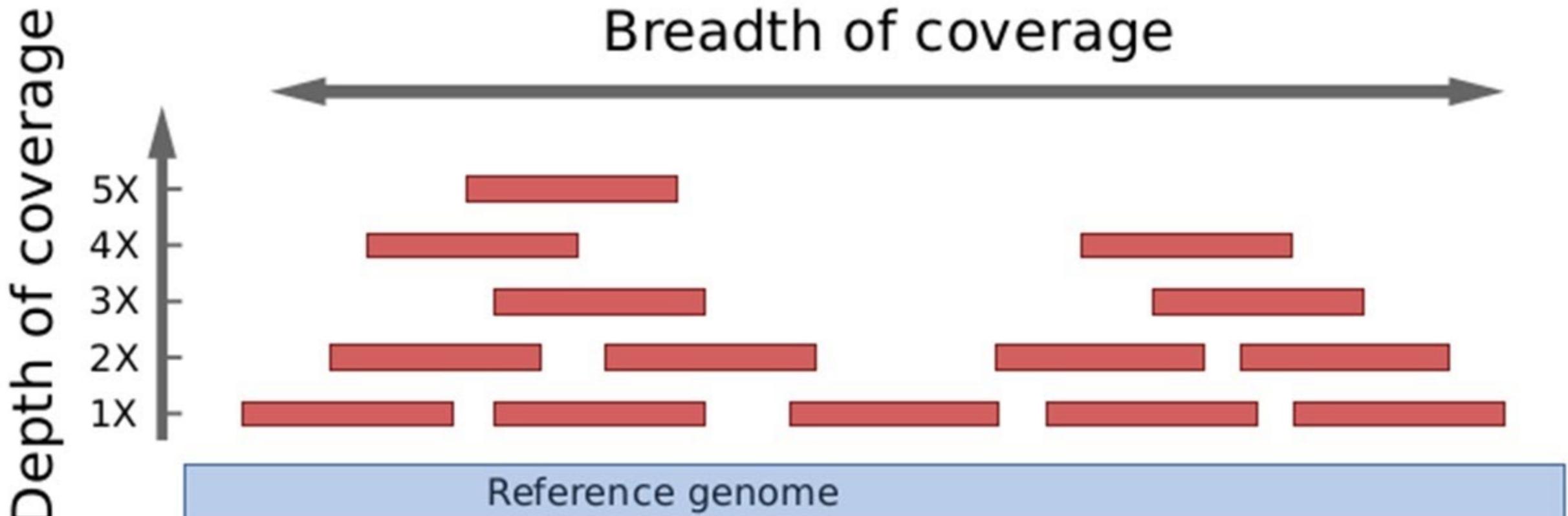
Giải trình tự gen thế hệ mới (NGS): giải trình tự song song

Read1 : CTCGAATACG

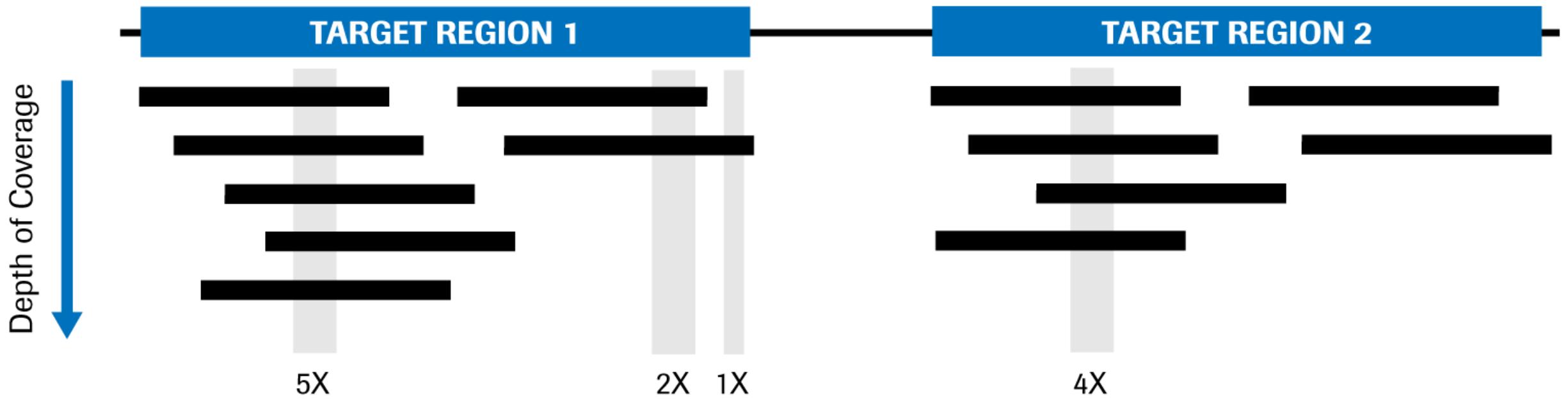


Read1 : CTCGAATACG
Read2 : CTCGAATACG
Read3 : CTCGAATACG
Read4 : CTCGAATACG
Read5 : CGCGAATACG
Read6 : CGCGAATACG
Read7 : CGCGACTACG
Read8 : CGCGAATACG

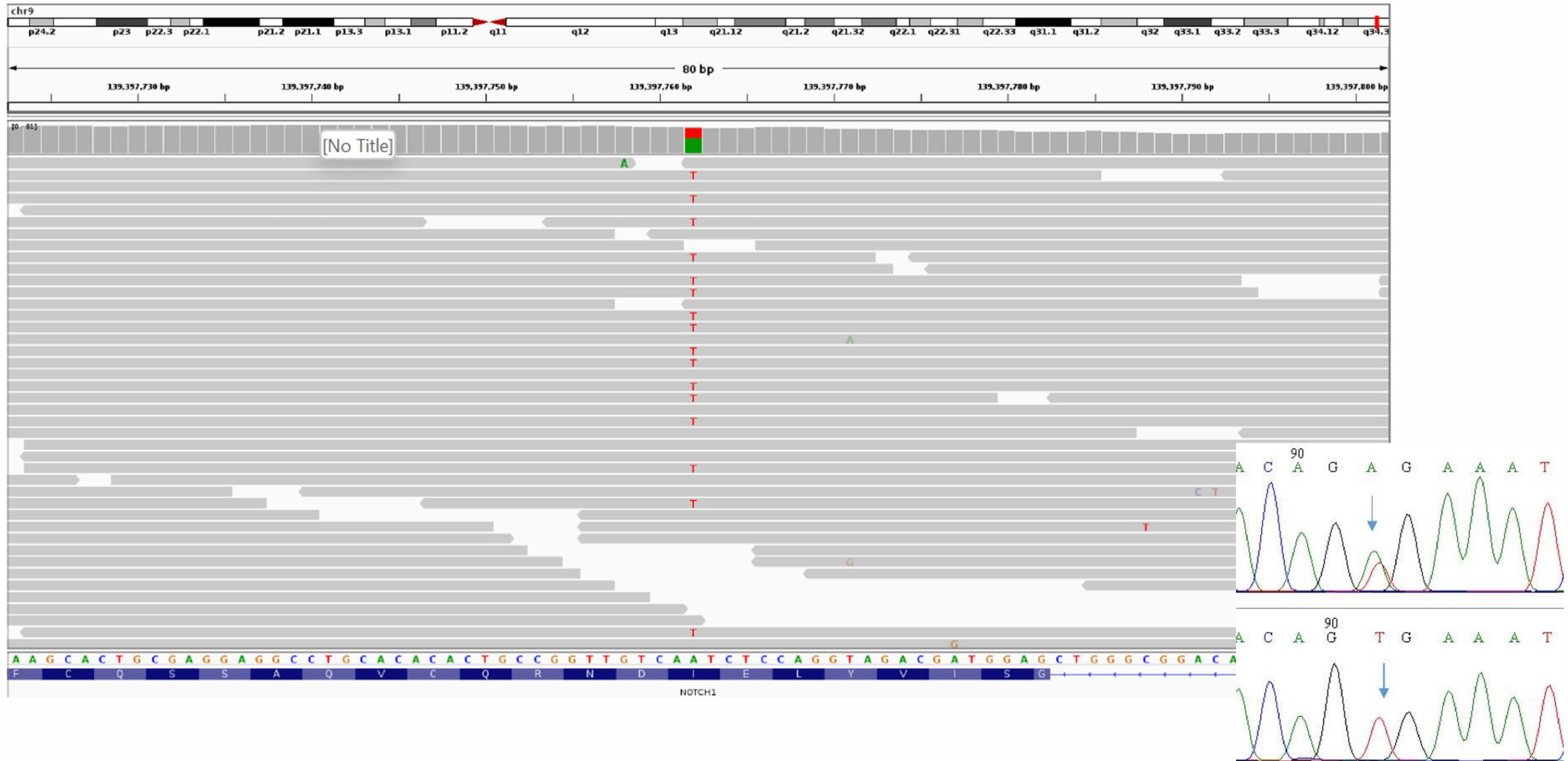
Depth vs Breadth of Coverage



Depth of Coverage



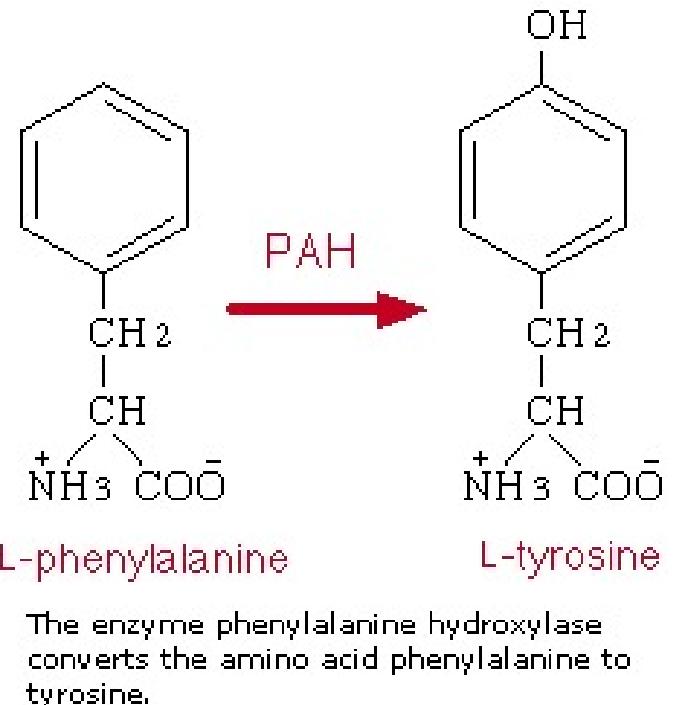
NGS vs Sanger



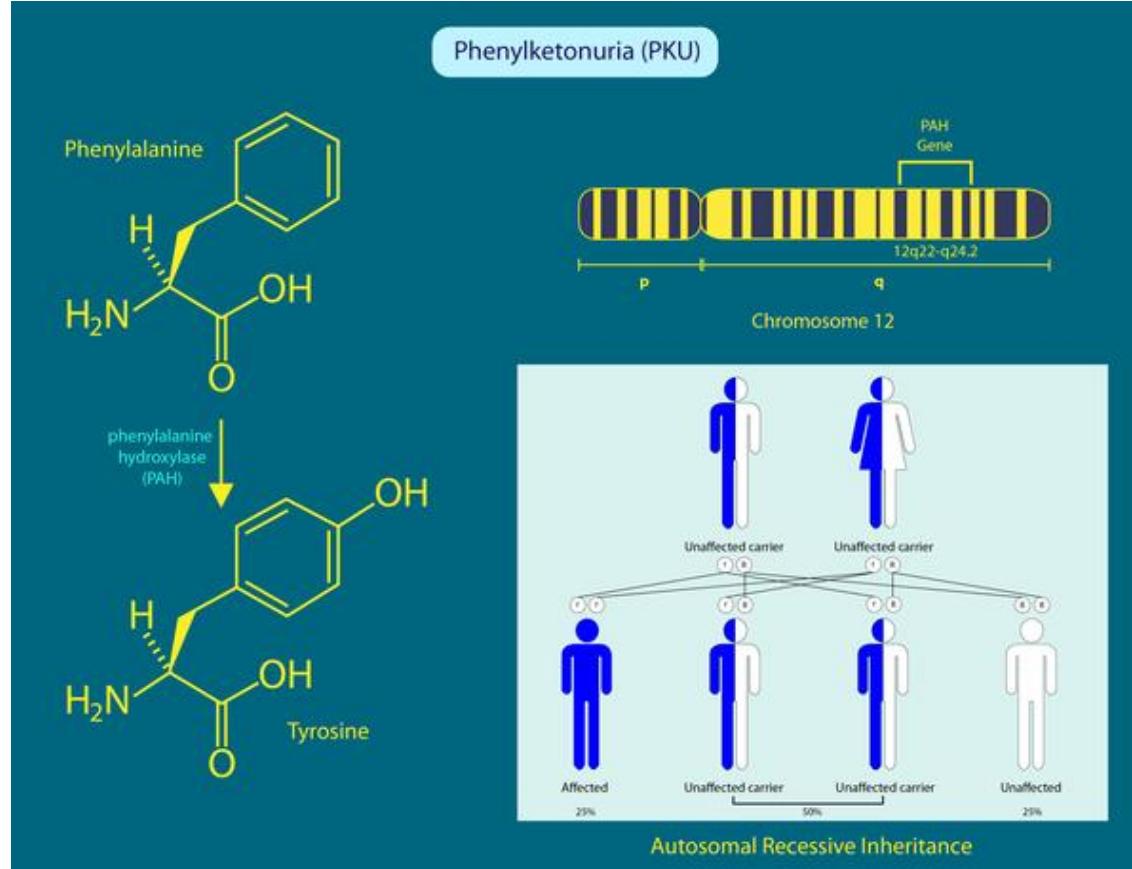
Ví dụ môi quan hệ của
BIẾN THỂ GEN và **BỆNH DI
TRUYỀN**

Phenylketonuria (PKU): Rối loạn chuyển hóa di truyền

- Nguyên nhân do thiếu hụt enzyme phenylalanine hydroxylase.
- Sự mất enzyme này dẫn đến suy giảm trí tuệ, tổn thương cơ quan, tư thế bất thường.
- Tần suất xảy ra PKU khác nhau giữa các nhóm dân tộc và các vùng địa lý trên toàn thế giới. Ở Hoa Kỳ, PKU xảy ra ở 1 trong 25.000 trẻ sơ sinh.
- Hầu hết các trường hợp PKU được phát hiện ngay sau sinh bằng sàng lọc sơ sinh và điều trị được bắt đầu ngay lập tức.



Phenylketonuria (PKU): Rối loạn chuyển hóa di truyền



Tên khác của PKU

- Folling disease
- Folling's disease
- PAH deficiency
- Phenylalanine hydroxylase deficiency
- Phenylalanine hydroxylase deficiency disease

<https://medlineplus.gov/genetics/condition/phenylketonuria/>

Trình tự gen PAH ở người - Homo sapiens (5053)

Gene (Nucleotide)

NT seq	1359 nt NT seq
	atgtccactgcggctggaaaacccaggctggcaggaaactctctgactttggacag gaaacaagcttatattgaagaacaactgaatcaaaatggccatatcactgatcttca ctcaaagaagaagtgggcattggccaaagtattgcgttatttggaggagaatgtat aacctgaccacattgaatcttagacccctcgtttaaagaaaagatgagtgatgaattttc acccatttgataaaacgttagcctgcgtctgacaaaatcatcaagatcttgggcat gacattgggtccactgtccatgagcttcacggataagaagaaaagacacagtggccctgg ttcccaagaaccattcaagagctggacagatggccaatcagattctcgtatggagcg gaactggatgctgaccaccctggtttaaagatccgtgtaccgtgcaagacggaaagcg tttgcgtacattgcctacaactaccggcatgggcagccatccctcgagtggaaatacatg gaggaagaaaaagaaaatggggcacatgttcaagactctgaagtccctgtataaaaacc catgttgcgtatgagtacaatcacatccacttgcgttgcggccatccctcgactgggttccat gaagataacatccccagctggaaagacgttctcgttccgcagacttgcactgggttcc cgccctccgacccgtggctggcctgctccctcgggatccctgggtggccatccctcg cgagtccactgcacacatcagacatggatccaagccatgtataccccgaa cctgacatctgcatgagctgtggacatgtgcccttgcgttgcacatgtgcactttgc cagtttccaggaaatggccctgcctctgggtgcacatgtgatgaaatacatgtaaaag ctcgccacaatgttactgtggatgttgcgtctgcaaaacaggagactccata aaggcatatggctggccctgtcatccttggtaattacagactgttgcattatcagag aagccaaagcttctcccccggagctggagaagacagccatccaaaattacactgtcag gagttccagccctcttacatgtggcagagatgttgcacatgtgcaaggagaaaagtaagg aactttgctgccacaataccctcgcccttcgttgcgtacgacccatcacccaaagg attggggatcttgcgtggacaatacccagcagcttaagatggctgattccattaacagtgaa attggaaatcttgcgtggccctccagaaaataaaatgtaa

Protein (Amino Acid)

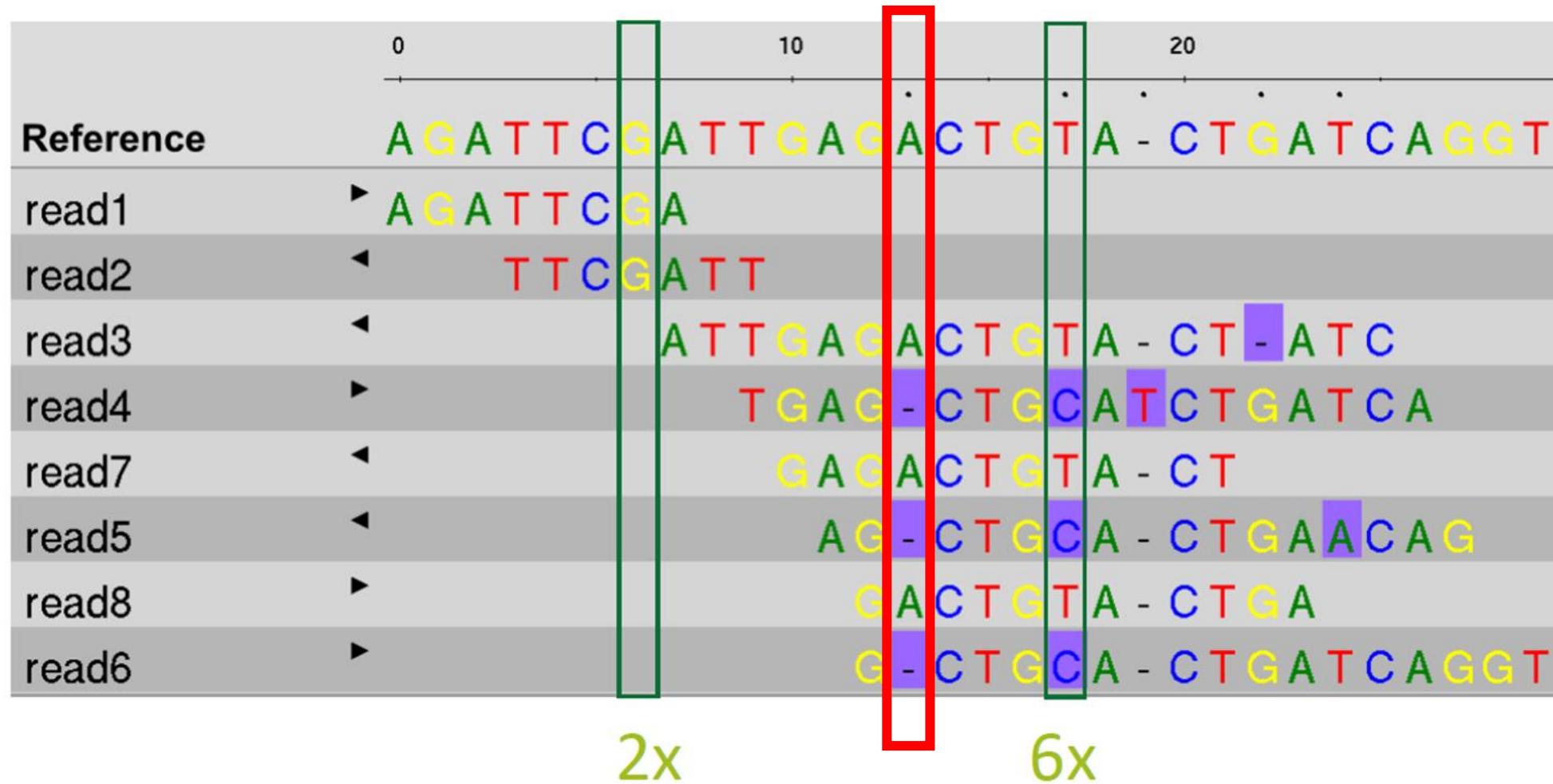
AA seq	452 aa AA seq DB search
	MSTAVLENPGLGRKLSDFGQETSYIEDNCNQNGAISLIFSLKEEVGALAKVLRLFEENDV NLTHIESRPSRLKKDEYEFFTHLDKRSLPALTNIKILRHDIGATVHELSRDKKKDTVPW FPRTIQELDRFANQILSYGAELDADHPGFKDPMVYRARRKQFADIAYNYRHGQPIPRVEYM EEEKKTWGTVFKTLKSLYKTHACYEYNHIFPLLEKYCFGHEDNIPQLEDVSQFLQTCCTGF RLRPVAGLSSRDFLGLAFRVFHCTQYIRHGSKPMYTPEPDICHELLGHVPLFSDRSFA QFSQEIGLASLGAPDEYIEKLATIYWFTVEFLCKQGDSIKAYGAGLLSSFGELQYCLSE KPKLLPLELEKTAIQNYTVTEFQPLYYVAESFNDAKEKVRNFAATIPRPFSVRYDPYTQR IEVLDNTQQLKILADSINSEIGILCSALQKIK

<https://www.genome.jp/entry/T01001:5053>

Gen PAH



Variants in PAH



ClinVar: Cơ sở dữ liệu biến thể gây bệnh

National Library of Medicine
National Center for Biotechnology Information

2013

ClinVar ClinVar Search ClinVar by gene symbols, location, HGVS expressions, c-dot, p-dot, conditions, and more Search Advanced Help

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ACTGATGGTATGGGCCAAGAGATATCT
CAGGTACGGCTGTCACTTACGGCTCAC
CAGGGCTGGGCATAAAAGTCAGGGCAGAGC
CCATGGTGCATCTGACTCCTGAGGAGAA
GCAGGTTGGTATCAAGGTTACAAGACAGGT
GGCACTGACTCTCTGCCTATTGGTCTAT

ClinVar

ClinVar aggregates information about genomic variation and its relationship to human health.

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Related Sites

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We gratefully acknowledge those who have submitted data and provided advice during the development of ClinVar.
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NM_000277.3(PAH):c.971T>A (p.Ile324Asn)

Germine Reviewed by expert panel ★★★☆ Pathogenic for Phenylketonuria Classification is based on the expert panel submission Dec 2023 by ClinGen PAH Variant Curation Expert Panel ? FDA RECOGNIZED DATABASE

Cơ sở dữ liệu ClinVar cho gen PAH

Classification type

- Germline (208)
- Somatic (0)

Germline classification

- Conflicting classifications (2)
- Benign (10)
- Likely benign (21)
- Uncertain significance (41)
- Likely pathogenic (40)
- Pathogenic (39)

Types of conflicts

- P/LP vs LB/B (0)
- P/LP vs VUS (0)
- VUS vs LB/B (2)

Molecular consequence

- Frameshift (20)
- Missense (96)
- Nonsense (7)
- Splice site (9)
- ncRNA (0)
- Near gene (0)
- UTR (27)

Variation type

- Deletion (39)

Links from Gene

[Display options](#) ▾ [Sort by Relevance](#) ▾ [Download](#) ▾

Items: 1 to 100 of 209

<< First < Prev Page of 3 Next > Last >>

Variation	Gene (Protein Change)	Type (Consequence)	Condition	Classification, Review status
<input type="checkbox"/> NM_004316.4(ASCL1):c.51G>T (p.Gln17His)	ASCL1, PAH (Q17H)	Single nucleotide variant (missense variant +1 more)	not specified	 Uncertain significance ★
<input type="checkbox"/> NC_000012.11:g.(?_103232953)_(1_03240749_?)del	PAH	Deletion	Phenylketonuria	 Pathogenic ★
<input type="checkbox"/> NC_000012.11:g.(?_103288493)_(1_03310908_?)del	PAH	Deletion	Phenylketonuria	 Pathogenic ★
<input type="checkbox"/> NC_000012.11:g.(?_103248894)_(1_03249131_?)del	PAH	Deletion	Phenylketonuria	 Pathogenic ★
<input type="checkbox"/> NC_000012.12:g.(?_102894715)_(102894938_?)del	PAH	Deletion	Phenylketonuria	 Pathogenic ★
<input type="checkbox"/> NC_000012.11:g.(?_103306549)_(1_03306696_?)del	PAH	Deletion	Phenylketonuria	 Pathogenic ★
<input type="checkbox"/> NM_000277.3(PAH):c.1179_1180del(p.Asn393fs)	PAH (N393fs)	Deletion (frameshift variant)	Phenylketonuria	 Likely pathogenic ★

https://www.ncbi.nlm.nih.gov/clinvar?LinkName=gene_clinvar&from_uid=5053

Biến thể gây bệnh - Pathogenic variant in PAH

NM_000277.3(PAH):c.971T>A (p.Ile324Asn)

ClinVar Genomic variation as it relates to human health

Search by gene symbols, location, HGVS expressions, c-dot, p-dot, conditions, : [Search ClinVar](#) [?](#)

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NM_000277.3(PAH):c.971T>A (p.Ile324Asn)

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We've updated the ClinVar website to better support classifications of somatic variants!

Read more about changes to the website in our [web release notes](#); more information about somatic variants in ClinVar is available on [GitHub](#).

Germline

Top reviewed classifications are shown here. Submission summary: **1 submission 1 submitter 1 condition**

Reviewed by expert panel **Pathogenic** for Phenylketonuria

Dec 2023 by [ClinGen PAH Va...](#) [FDA RECOGNIZED DATABASE](#)

Somatic

No data submitted for somatic clinical impact

No data submitted for oncogenicity

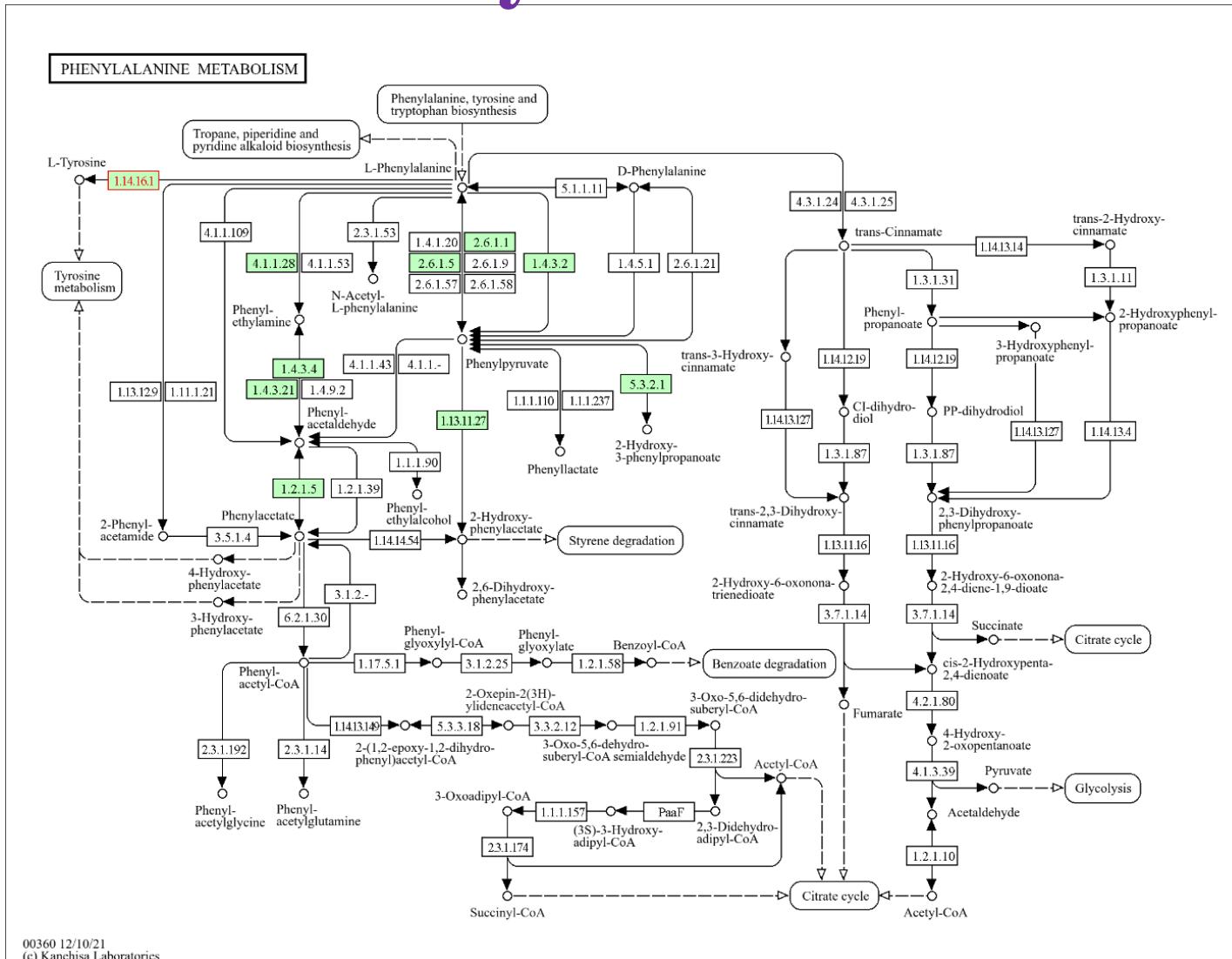
On this page

[Classification Summary](#) [Variant Details](#) [Genes](#) [Germline](#) [Conditions](#) [Submissions](#) [Citations](#) [Text mined Citations](#)

[Feedback](#)

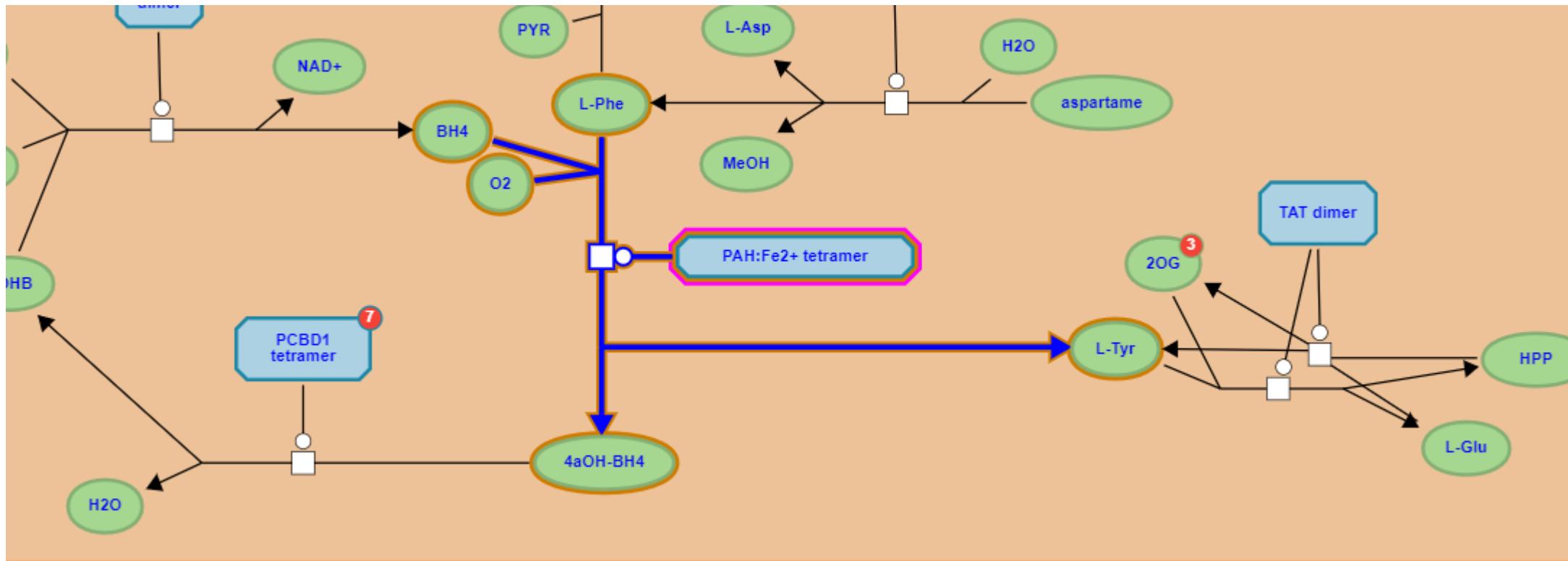
<https://www.ncbi.nlm.nih.gov/clinvar/variation/2682170/>

PAH: chuyển hóa Phenylalanine thành Tyrosine



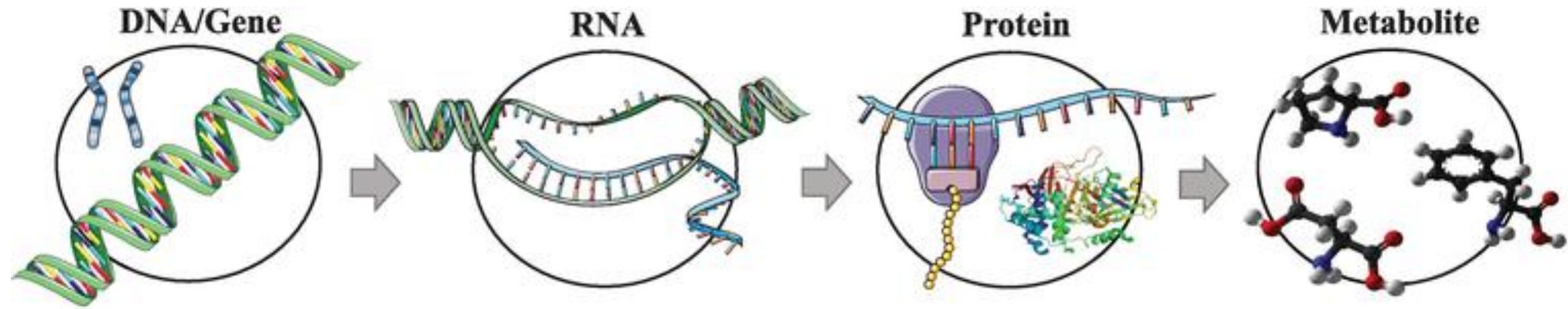
00360 12/10/21
(c) Kanchisa Laboratories

PAH: chuyển hóa Phenylalanine thành Tyrosine



<https://reactome.org/PathwayBrowser/#/R-HSA-8963691&SEL=R-HSA-71118&PATH=R-HSA-1430728,R-HSA-71291&FLG=UniProt:P00439>

Mối liên kết: Biến thể gen và bệnh di truyền



Genomics

PAH gene
Ref ...ATCGAT...
P1 ...AACGAT...

NM_000277.3(PAH):c.971T>A

Transcriptomics

PAH mRNA
Ref ...AUCGAU...
P1 ...AACGAU...

NM_000277.3(PAH):c.971T>A

Proteomics

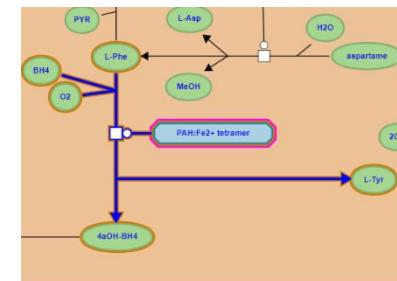
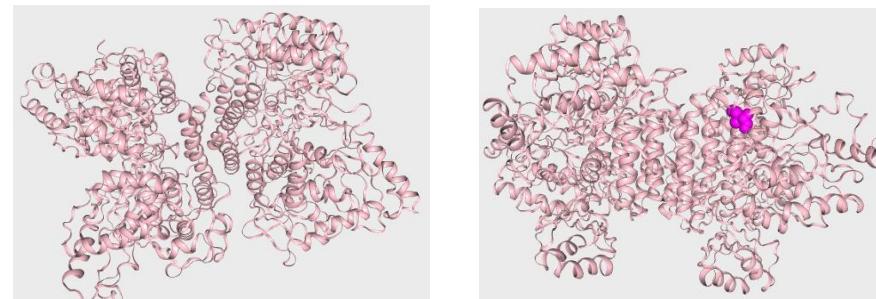
PAH protein
Ref ...Ile-Asp...
P1 ...Asn-Asp...

NM_000277.3(PAH):p.Ile324Asn

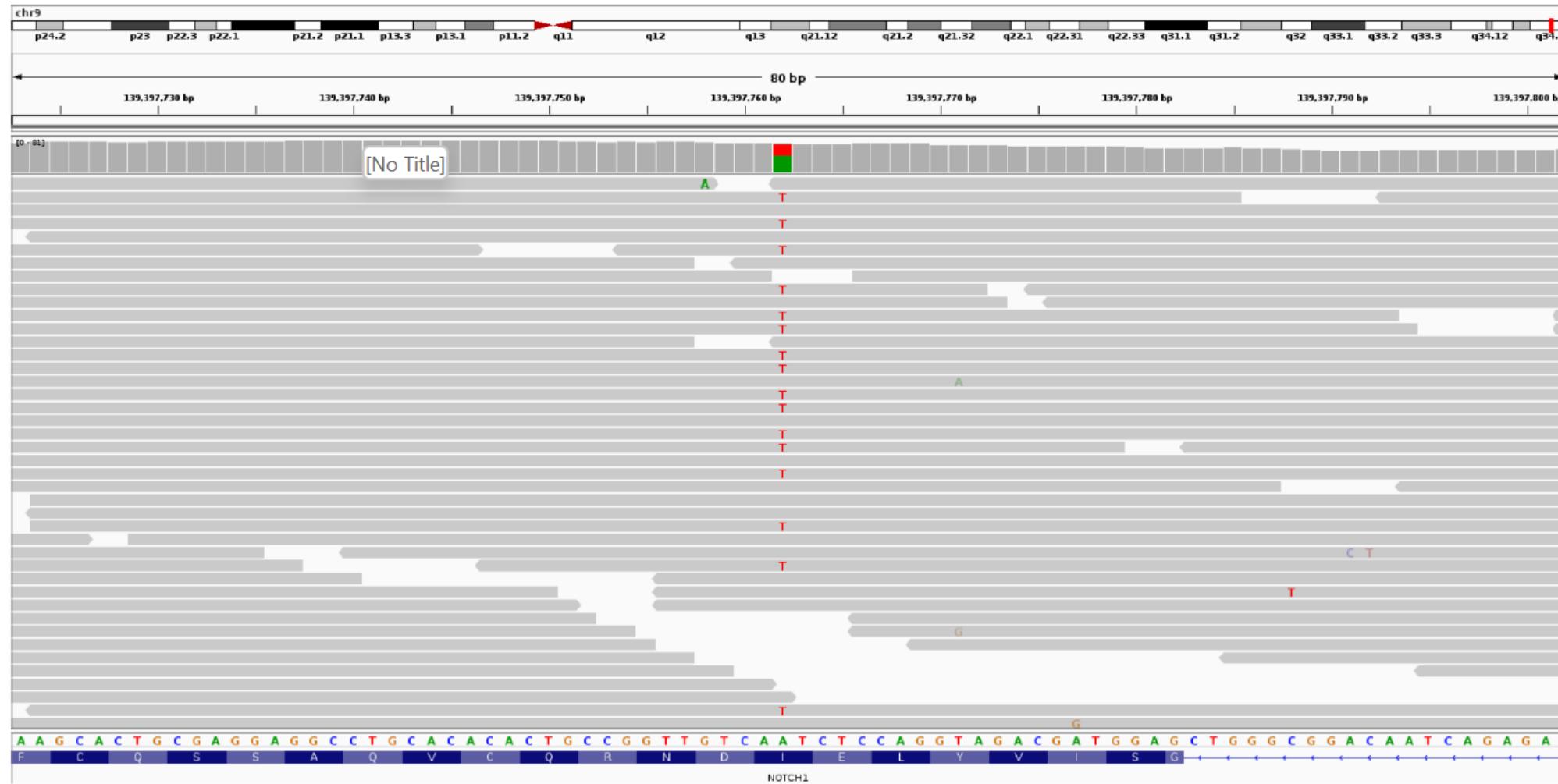
Metabolomics

PAH
Ref Phe → Tyr

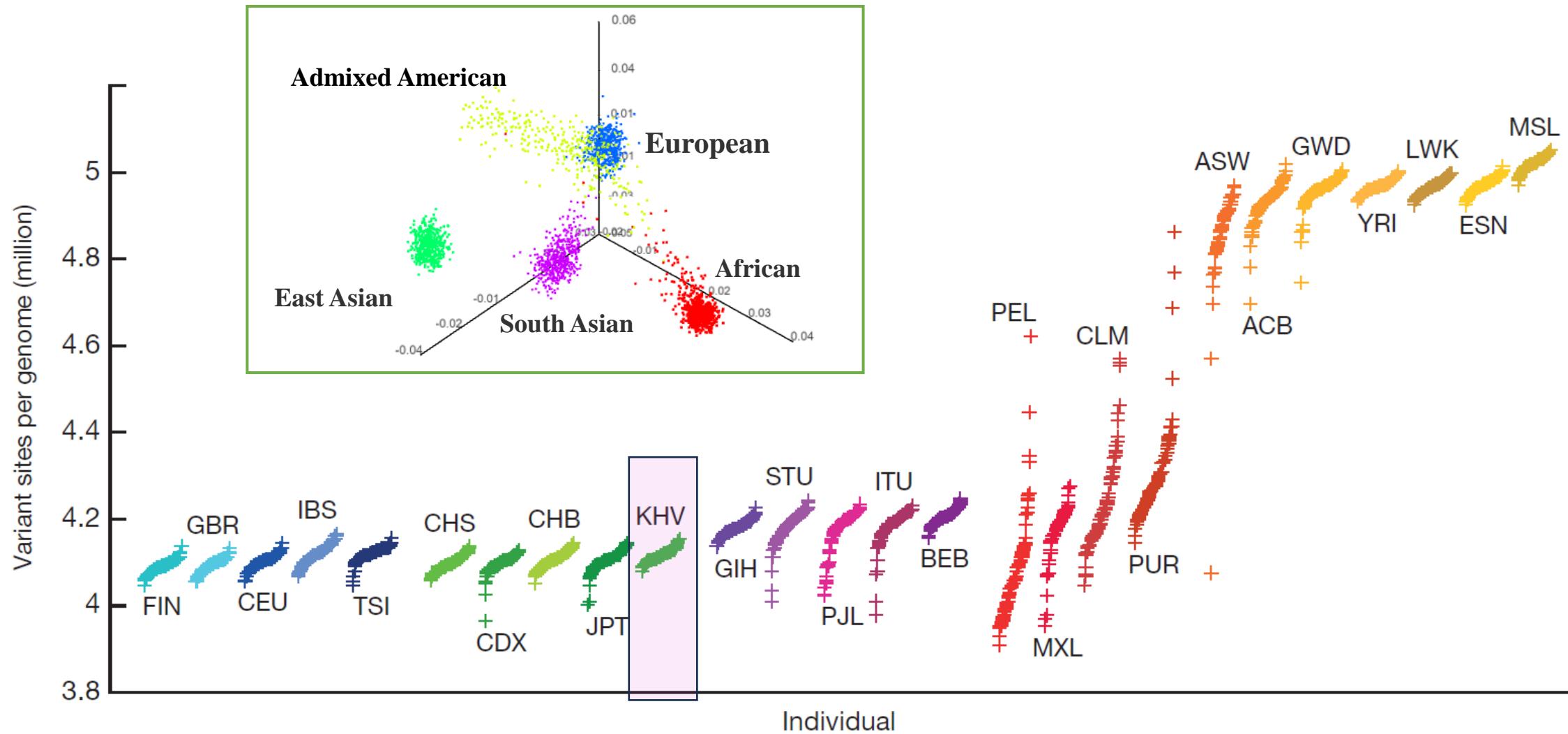
P1 Phe ~~→~~ Tyr



Alignment and variant viewers

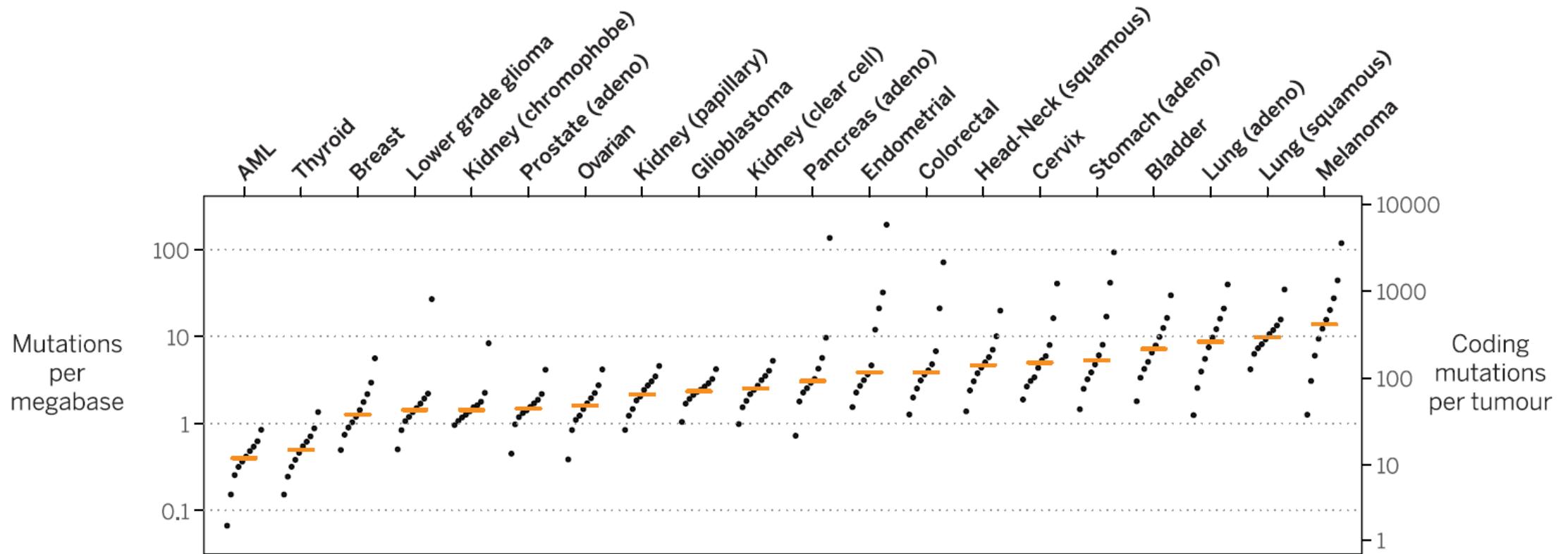


Human Genome Variation: 1000 Genomes Project



The number of variant sites per genome of 1K human genomes project (2015)
Kinh in Ho Chi Minh City, Vietnam (KHV)

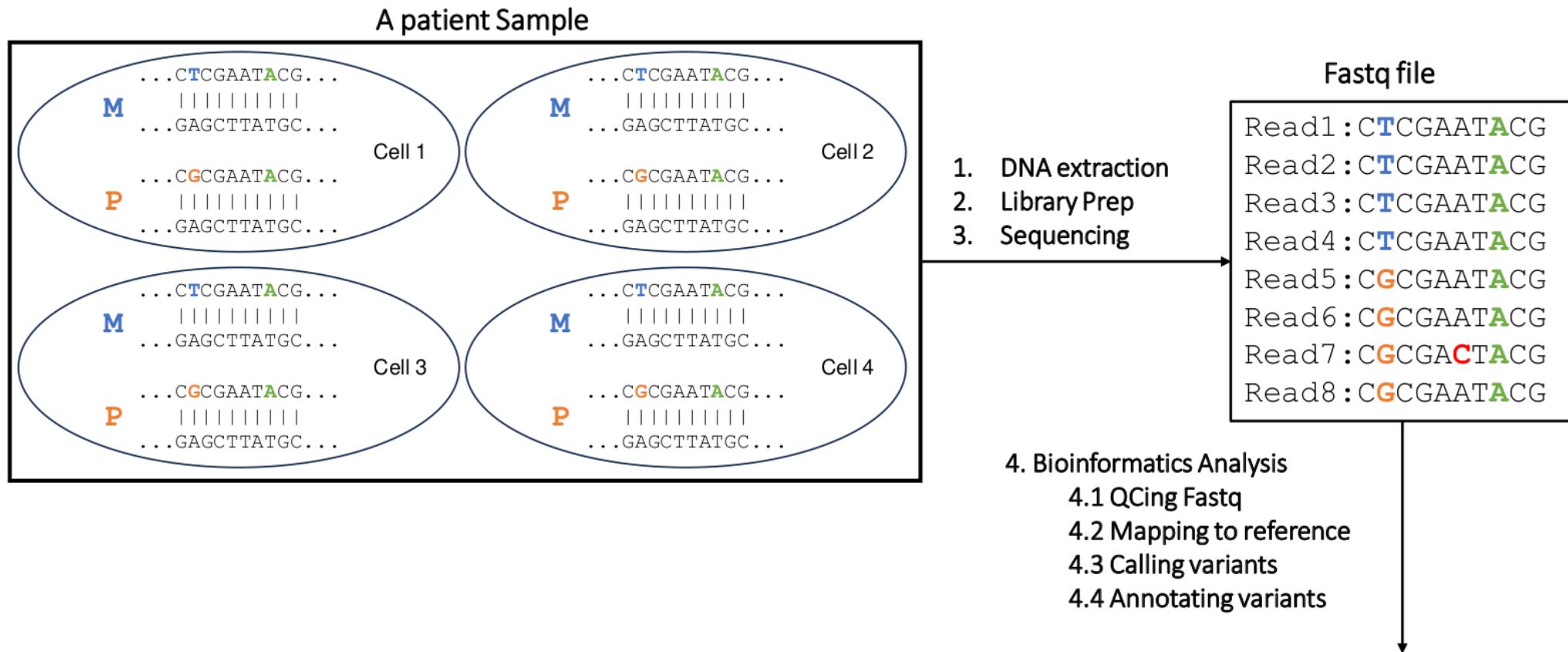
Cancer Genome Somatic Variation



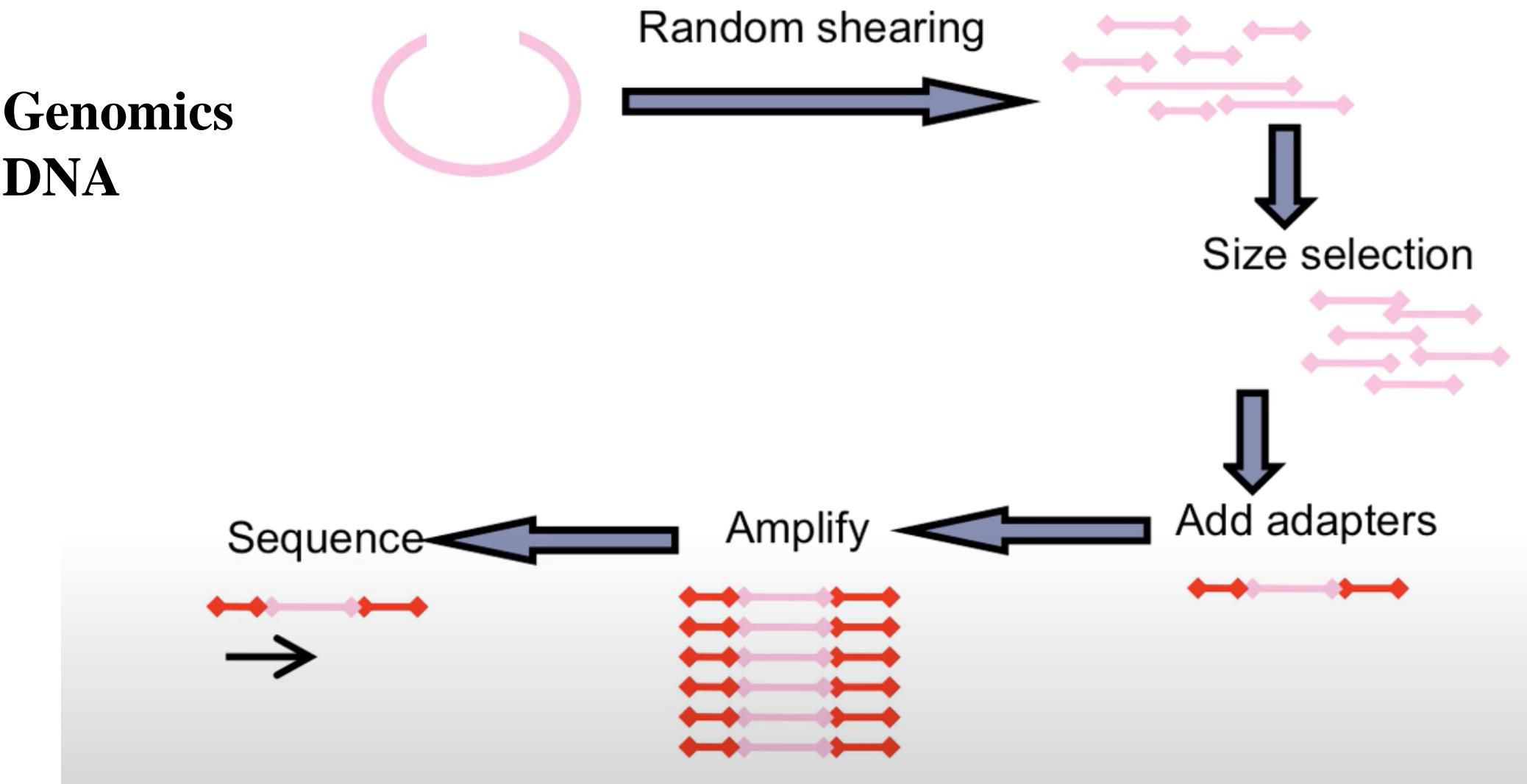
Mutation burden in 20 tumor types and relative contribution of different mutational processes.
For each tumor type, samples were divided into deciles on the basis of their mutation burden. (2015)

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)

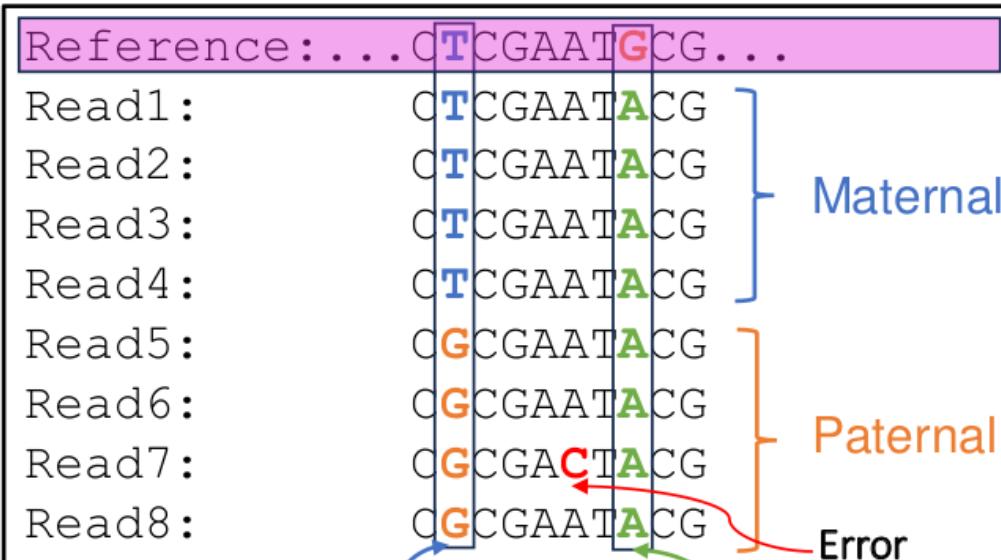


Whole genome sequencing – Shotgun Library Prep



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

4.2 Mapping reads to reference



Heterozygous

Homozygous

4.3 Calling 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

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)

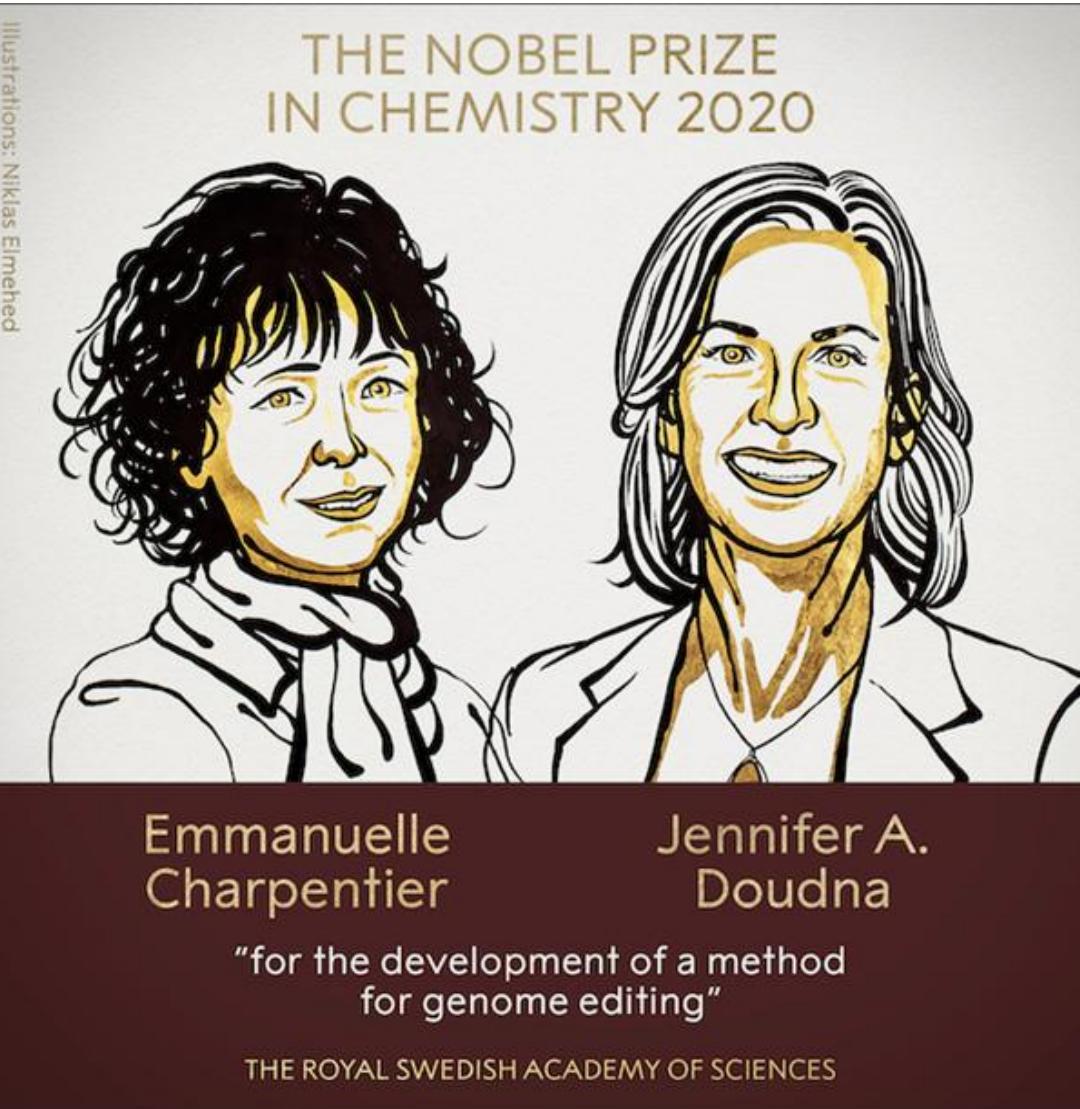
4.4 Annotating variants

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



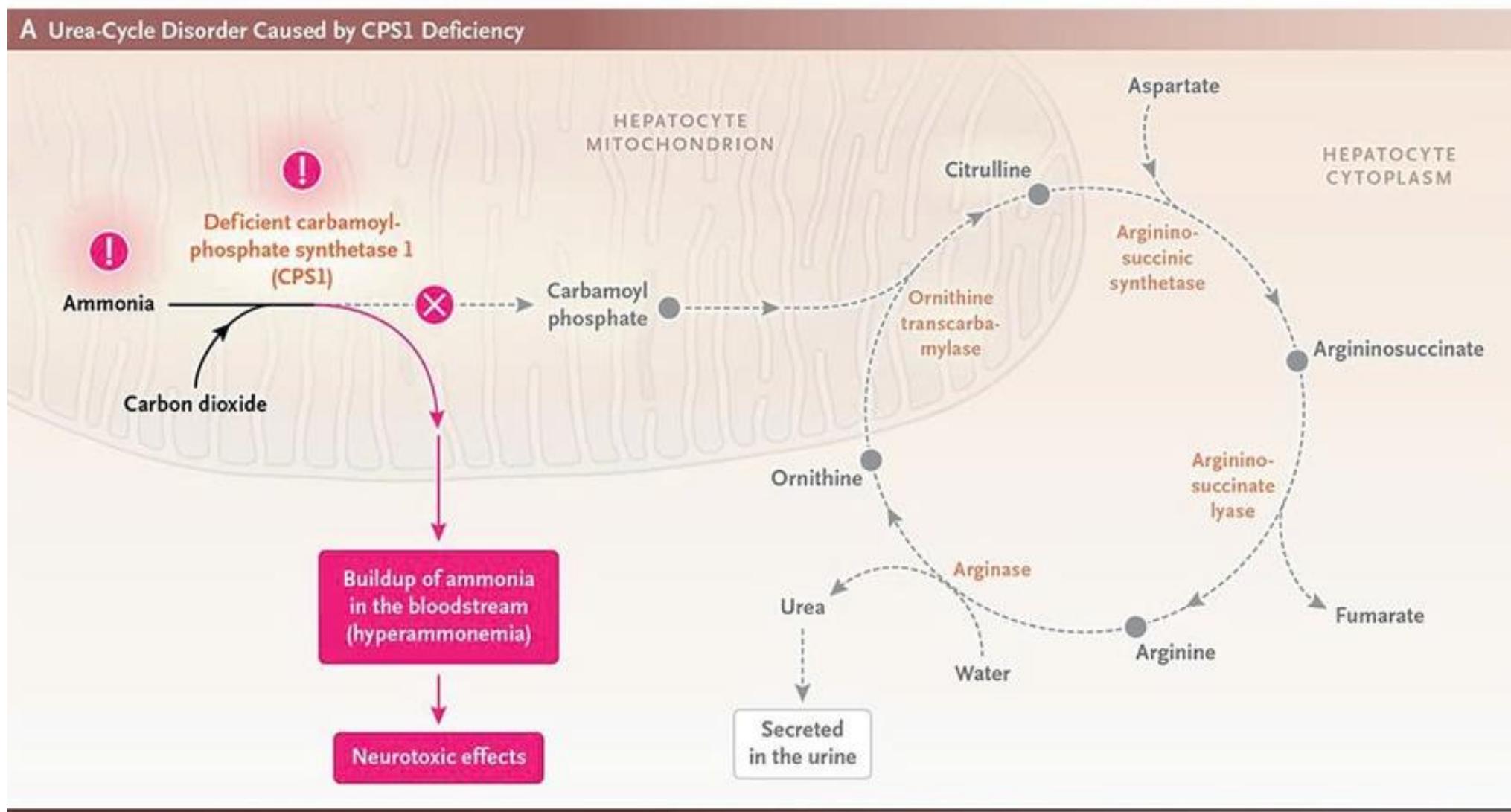
<https://ggba.swiss/en/the-first-medical-genomics-center-opens-in-geneva/>

**LIỆU CÓ GIÚP
ÍCH?**

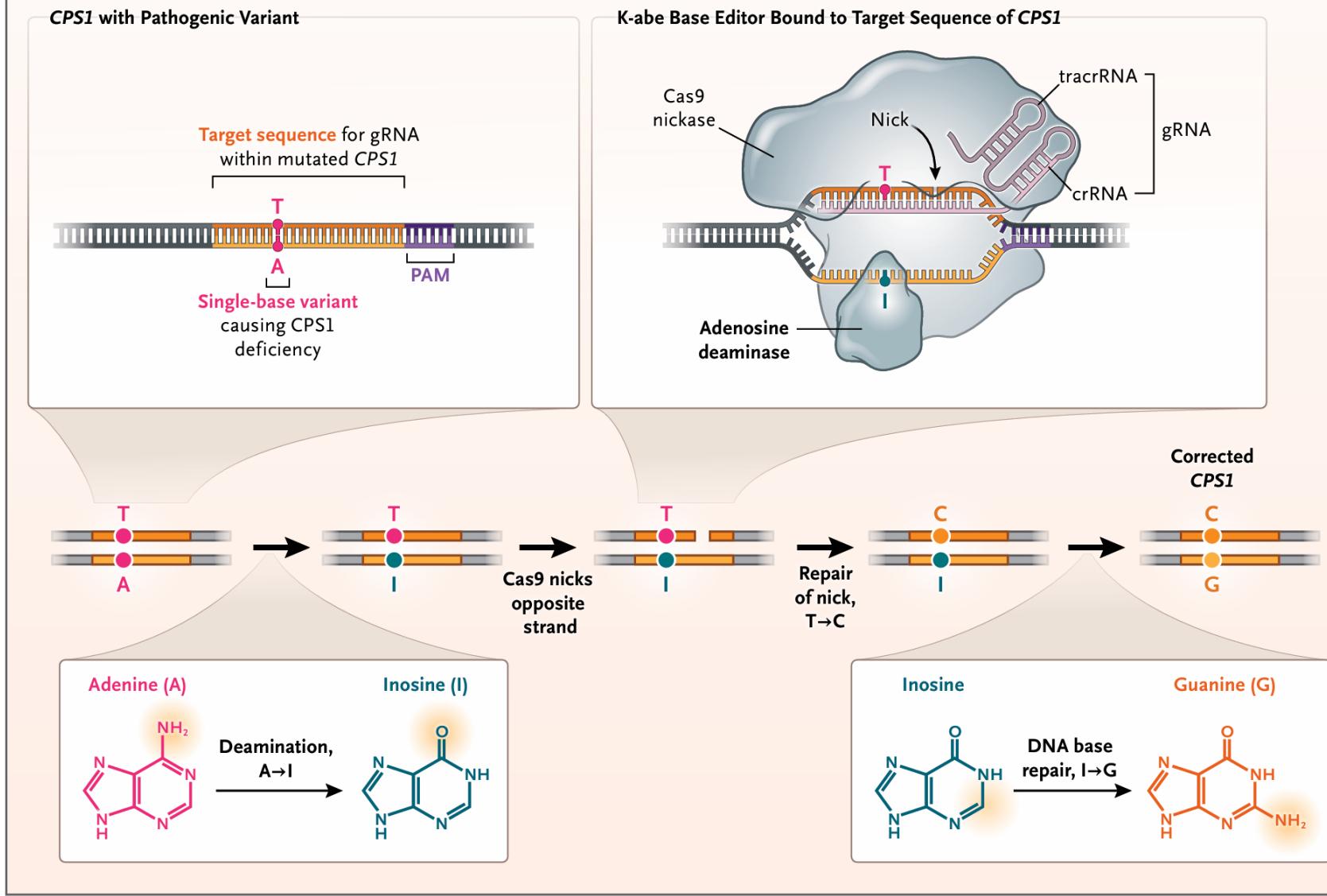


CRISPR-CAS9

New England Journal of Medicine (NEJM), May 2025



B Adenine Base Editor–Mediated Repair of CPS1 Variant



Gene-editing therapy made in just 6 months helps baby with life-threatening disease

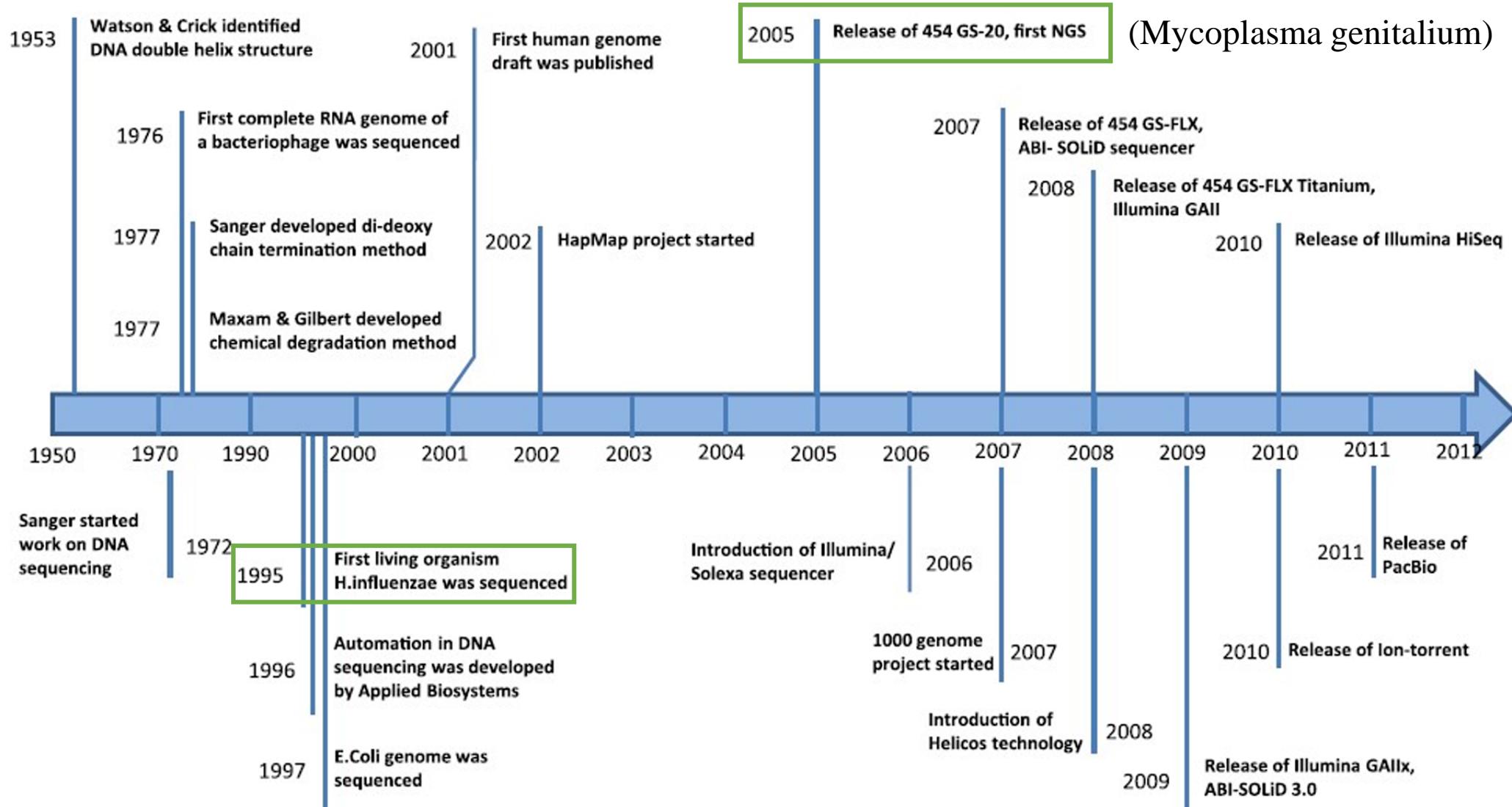
Custom CRISPR paves the way for treating genetic disorders in tailor-made ways

13 MAY 2023 · 1:00 PM ET · BY JOCelyn KAISER

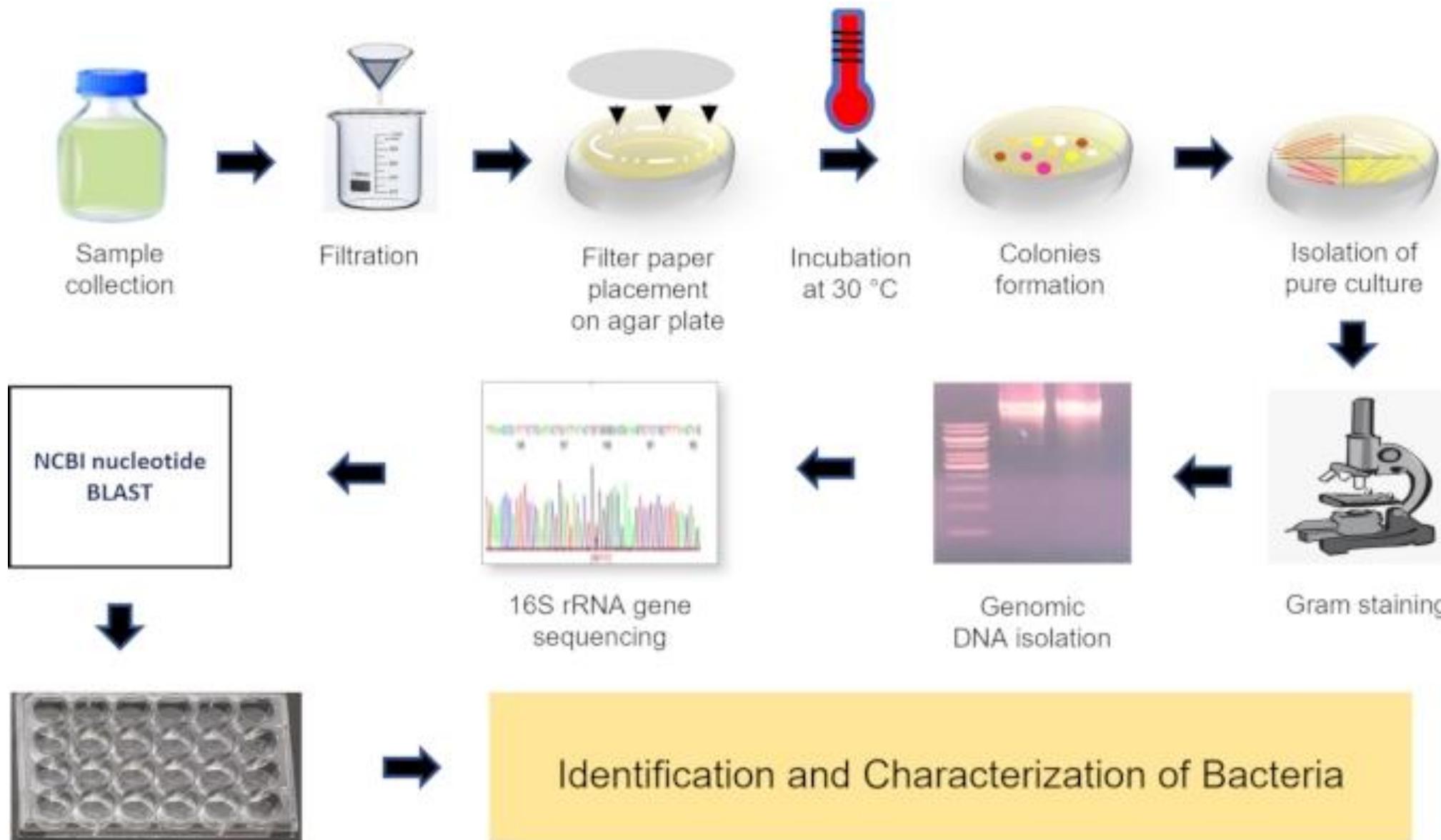


An infant who received a customized gene-editing treatment, shown here with researchers Kiran Musunuru (left) and Rebecca Ahrens-Nicklas, now needs less medicine to defuse a blood buildup of ammonia. From "Gene-editing therapy made in just 6 months helps baby with life-threatening disease" by Jocelyn Kaiser, 2023. <https://www.science.org/content/article/gene-editing-therapy-made-just-6-months-helps-baby-life-threatening-disease>

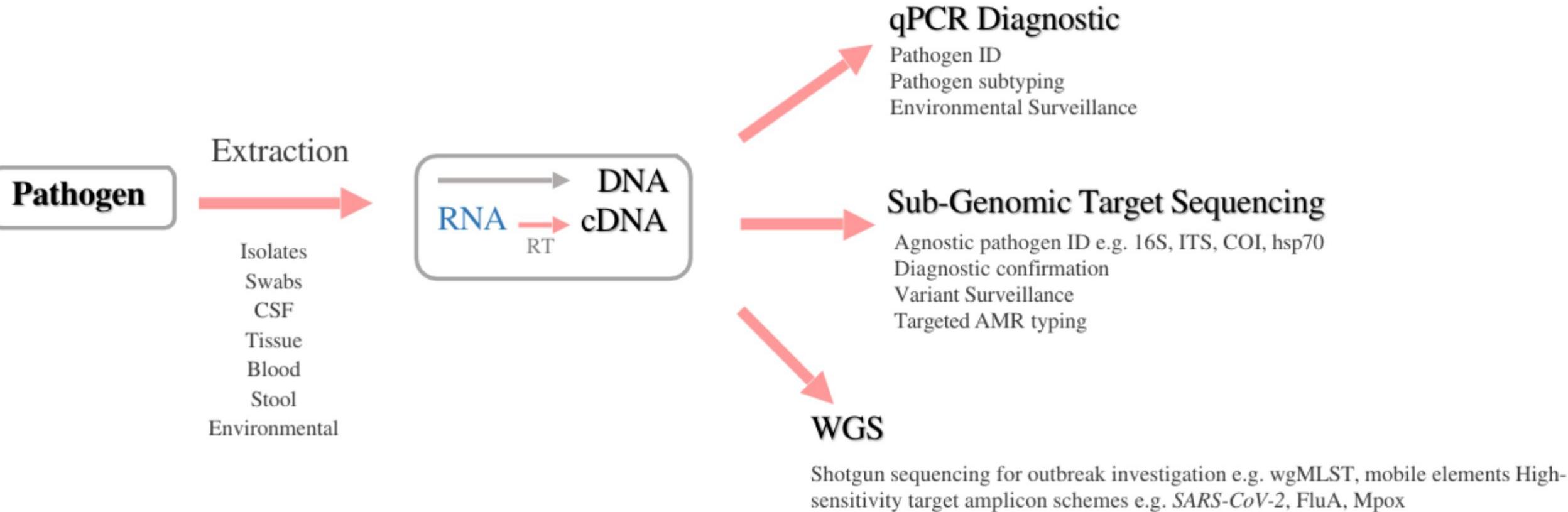
Giải trình tự genome vi sinh vật



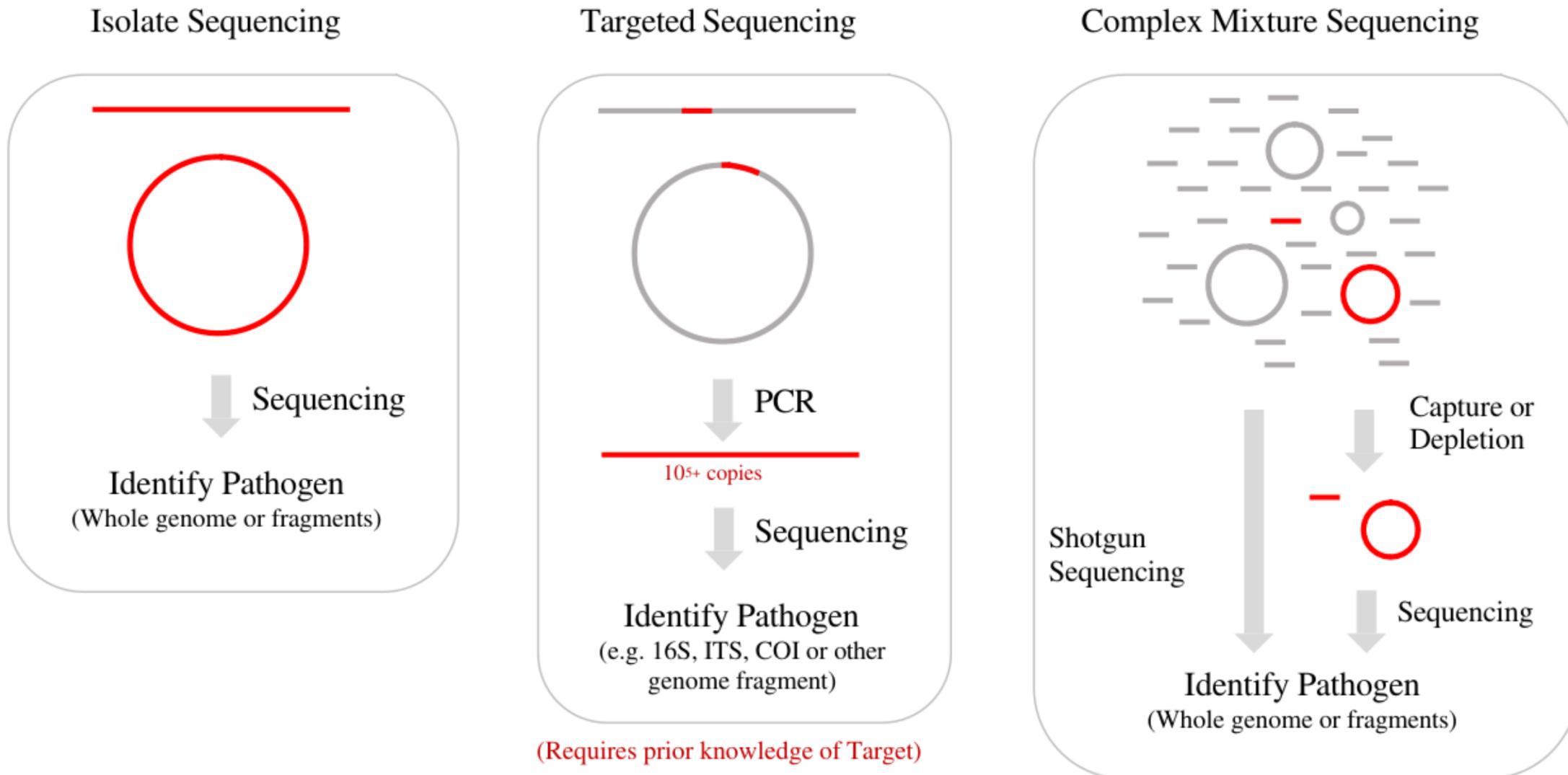
Pathogen Genetic testing and typing (1)



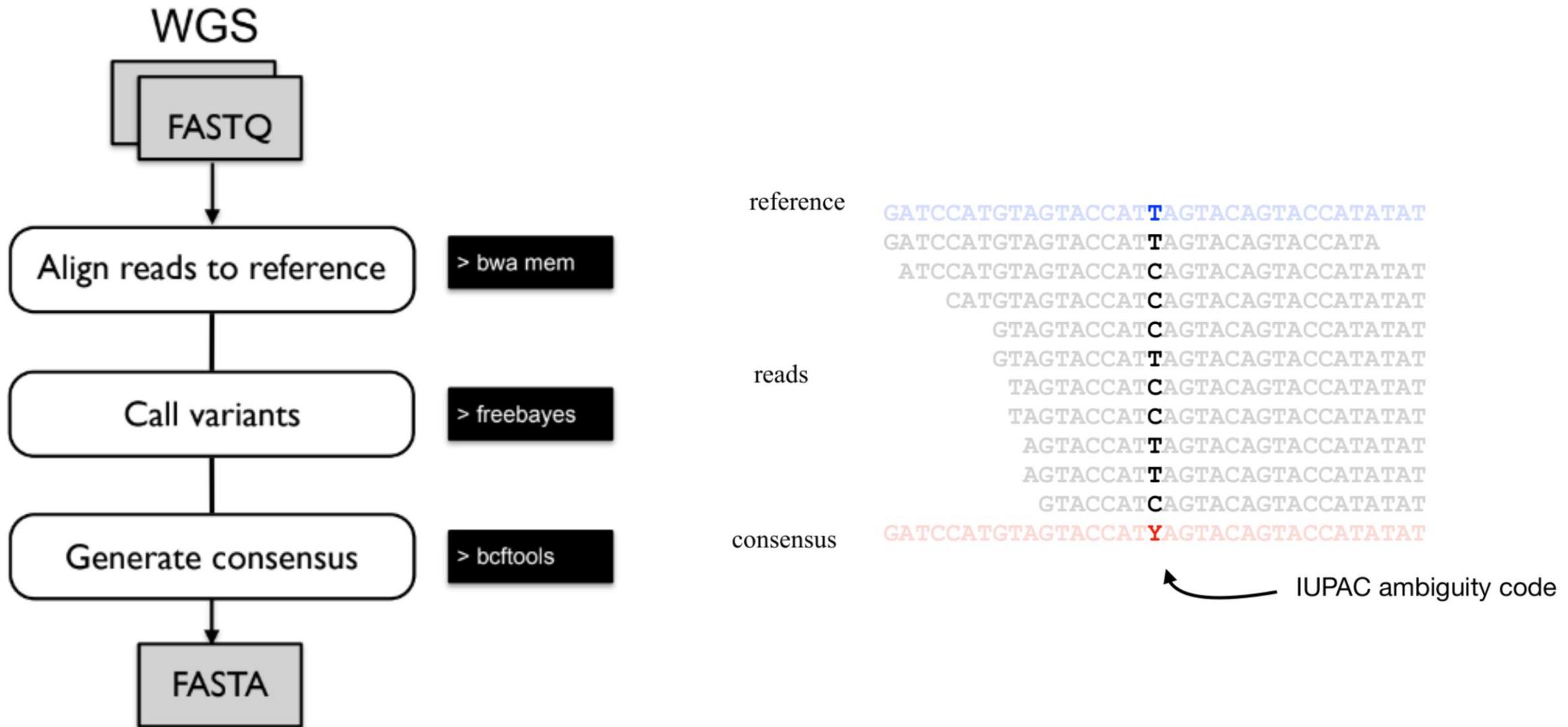
Pathogen Genetic testing and typing (2)



Pathogen Genetic testing and typing (3)

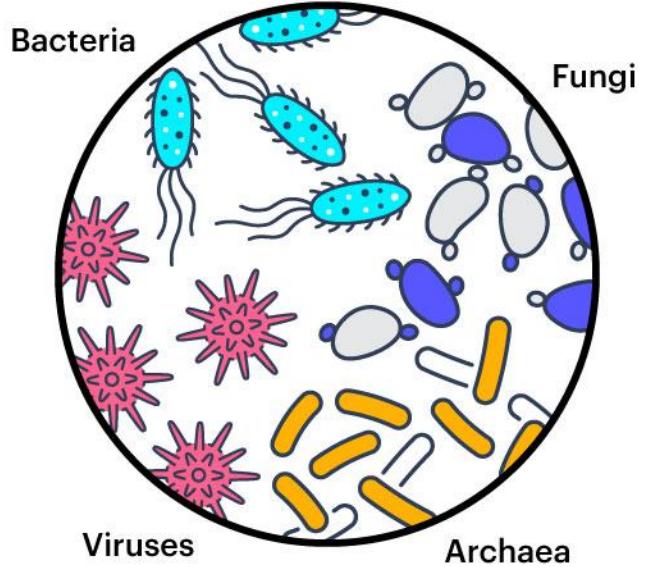


Pathogen Genetic testing and typing (4)

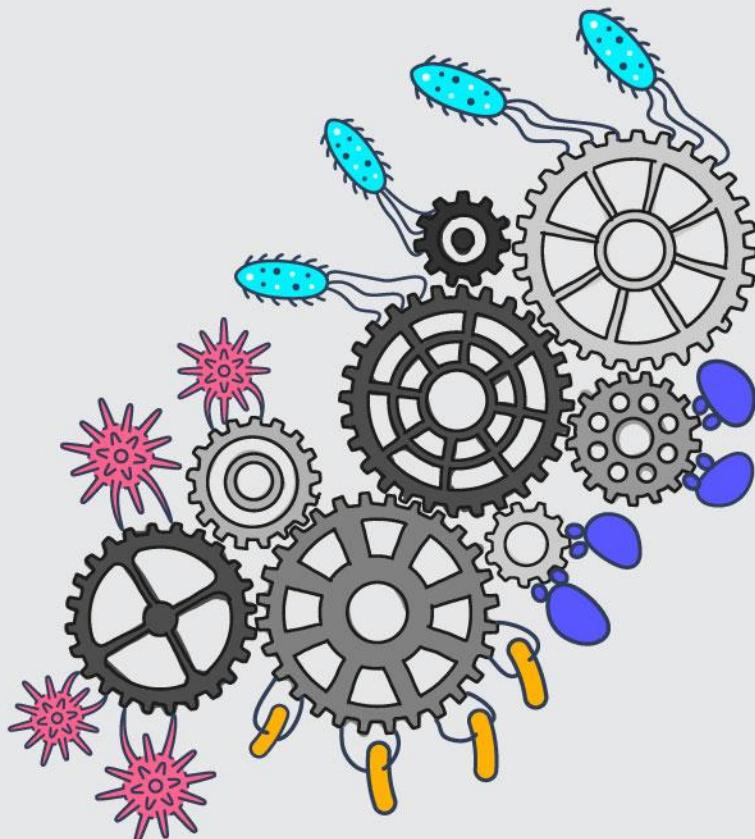


What IF (5)

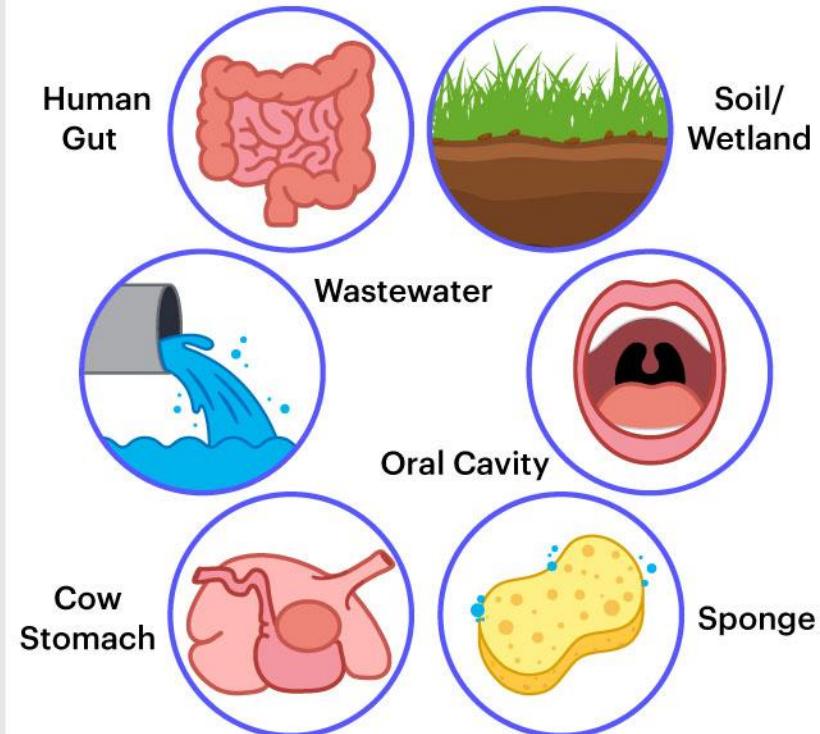
Microbiomes are communities of diverse microbes



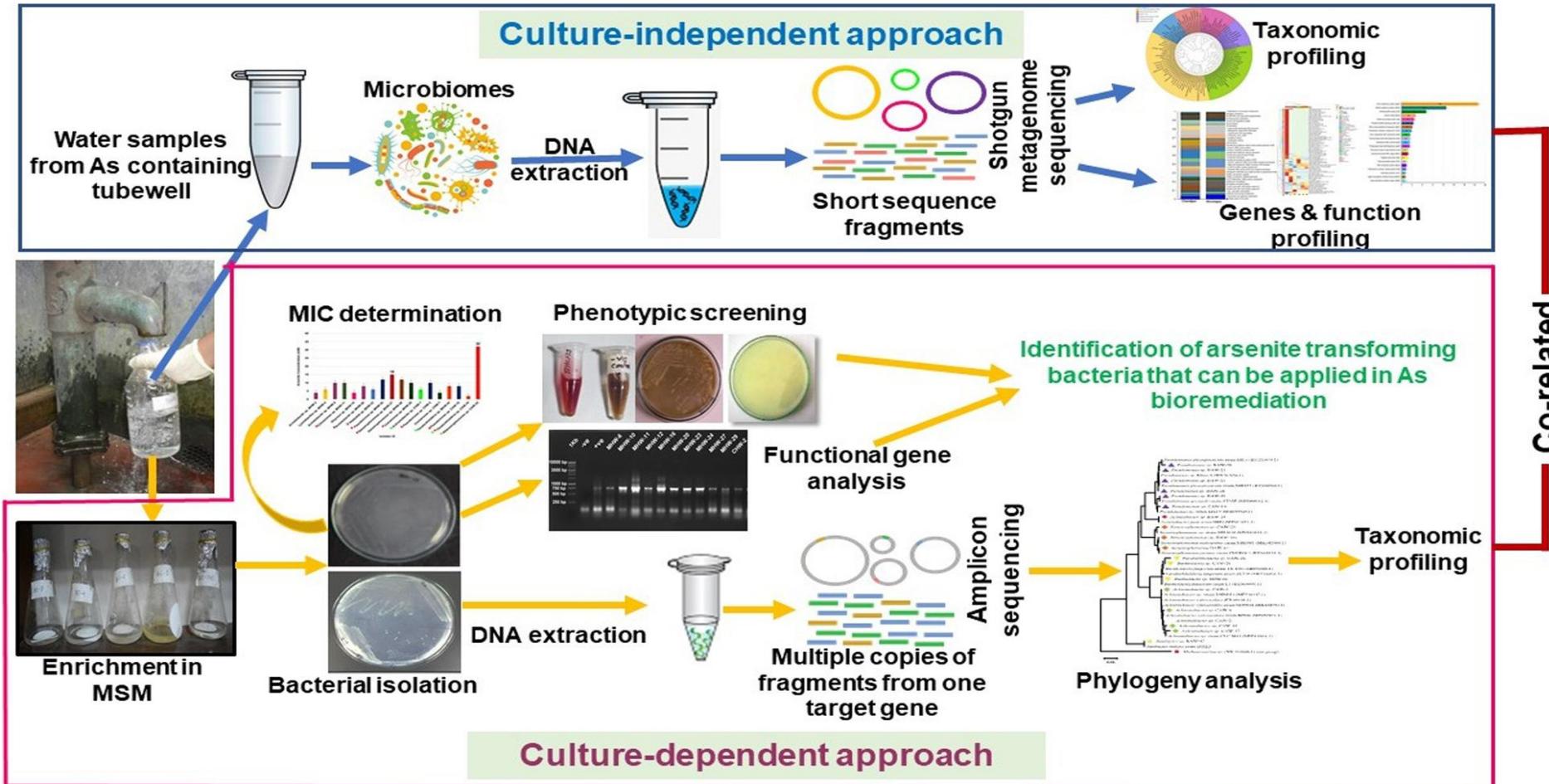
Each kind of microbe has its own role in the community



Microbiomes are everywhere

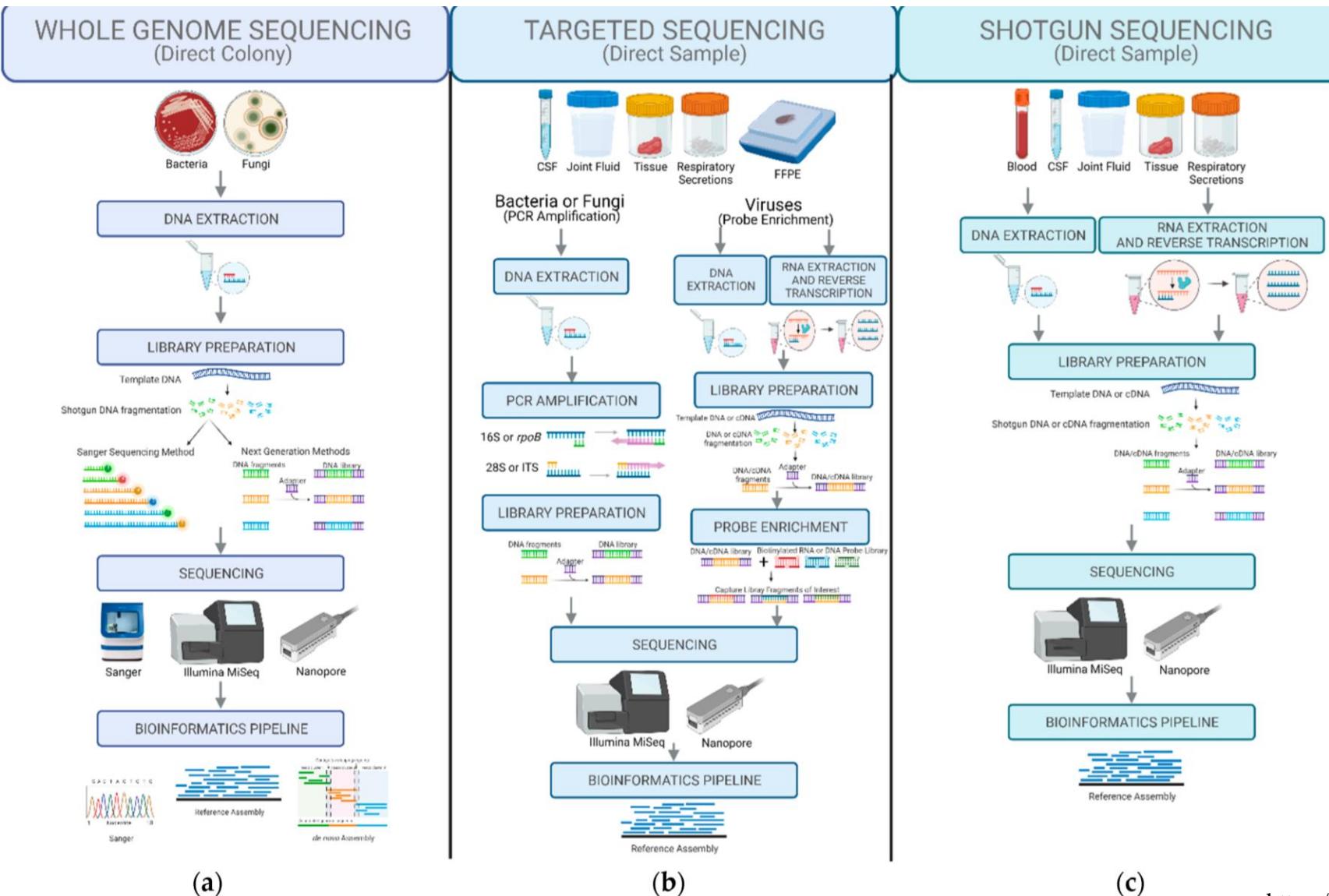


Metagenomics vs culture-dependent

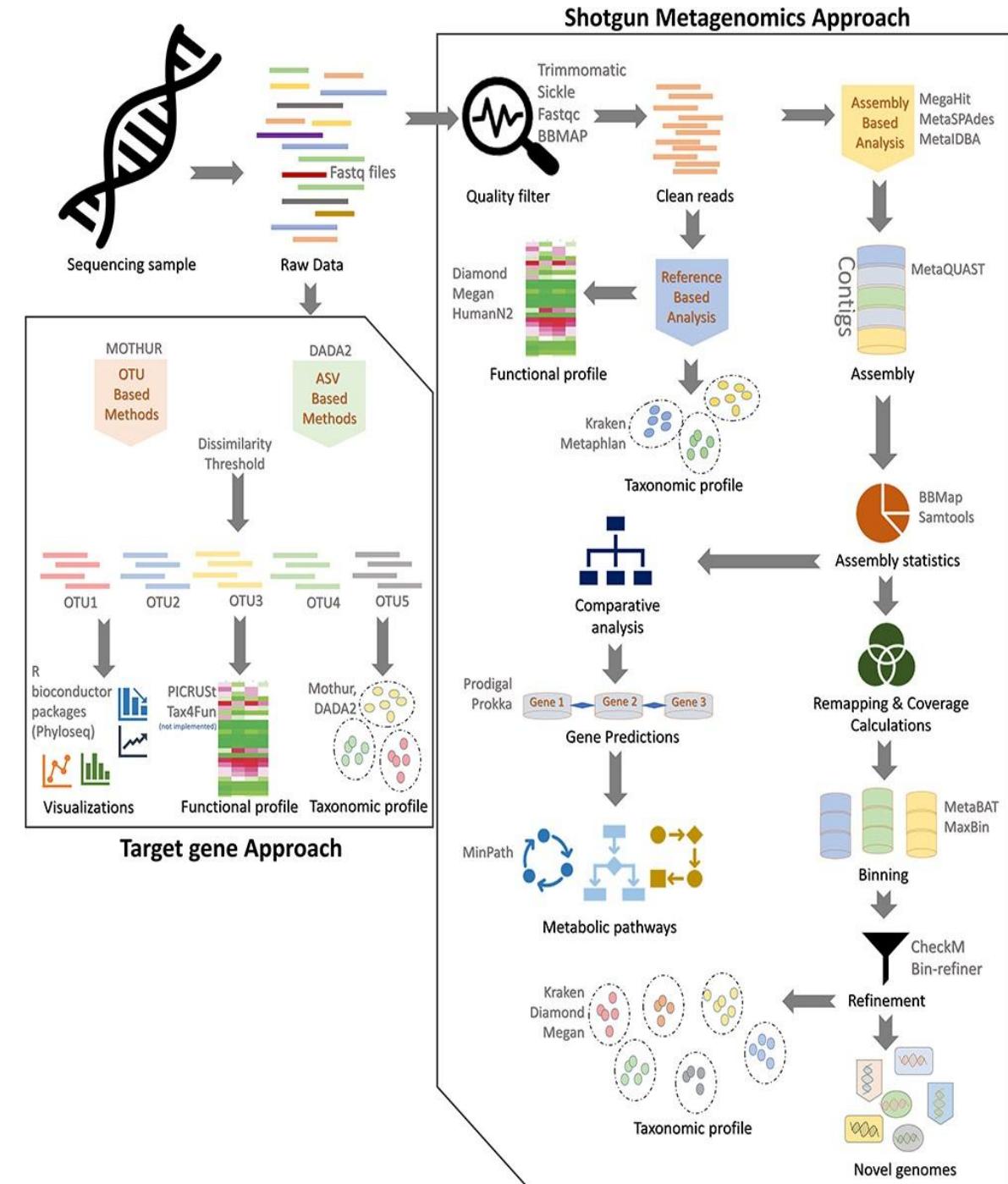
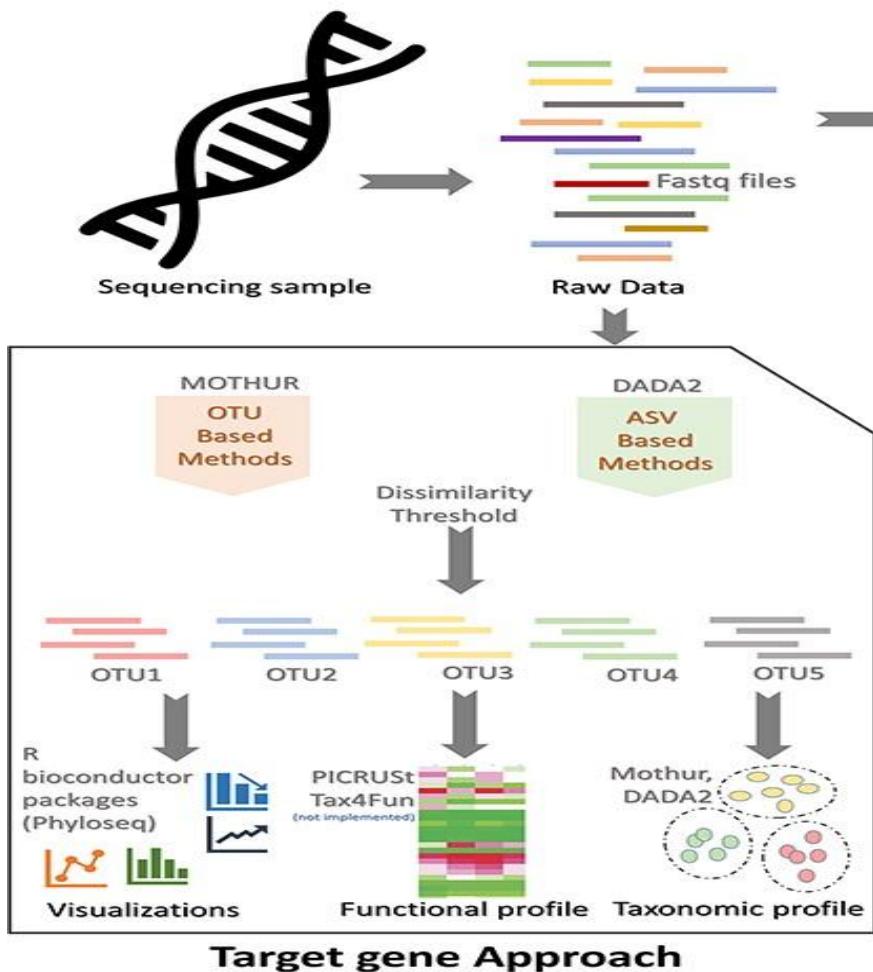


This study was carried out to assess the diversity and transformation potentials of the arsenic-affected groundwater microbiomes using both culture-dependent and independent (shotgun metagenomics) approaches.

Genomics vs Metagenomics Analysis



Target vs Shotgun



Xin gửi lời cảm ơn đến

Nhóm nghiên cứu

- **Bác sĩ Nguyễn Lê Đức Minh**
- **Thạc sỹ Đào Khương Duy**
- **Thạc sỹ Ngô Đại Phú**
- **Thạc sỹ Trần Thị Mỹ Qui**
- **Bác sĩ NGUYEN Thuy-Minh-Thu**
- **Thạc sỹ, bác sĩ Nguyễn Huy Thịnh**
- **Thạc sỹ Như**
- **KS Nguyễn Minh Hoàng**

Nhóm cộng tác nghiên cứu

- **Trần Quang Hải**
- **TS Nguyễn Anh Thư**
- **Hoàng Kim**
- **Huy Hà**
- **Hoàng Sơn**
- **TS Bác sĩ Nguyễn Thị Kim Nhi**
- **Bác sĩ Hậu**
- **PGS TS Bác sĩ Phạm Lê An**

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Xin chân thành cảm ơn!

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