

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

Nghiên cứu và ứng dụng NGS trong lựa chọn thuốc điều trị ung thư vú, buồng trứng, tiền liệt và tụy ở người lớn

Phuc Loi Luu, PhD

Luu.p.loi@googlemail.com

29/06/2024

STT	Nội dung	Giảng viên	Thời lượng
1	Giới thiệu về công nghệ Giải trình tự bộ hệ mới và khái quát về bộ gen người		30 phút
2	Xử lý dữ liệu thô, đánh giá, sàng lọc trình tự chất lượng và sắp xếp các trình tự (BRCA1/2)	TS. Lưu Phúc Lợi BS. Nguyễn Lê Đức Minh CN. Đào Khương Duy	60 phút
3	Gọi biến thể và sàng lọc biến thể gây bệnh trên gen BRCA1/2		60 phút
4	Chú giải biến thể gây bệnh trên gen BRCA1/2 trong lâm sàng		60 phút
5	Thảo luận và trả lời câu hỏi		30 phút

Human genome structure, functions and clinical considerations

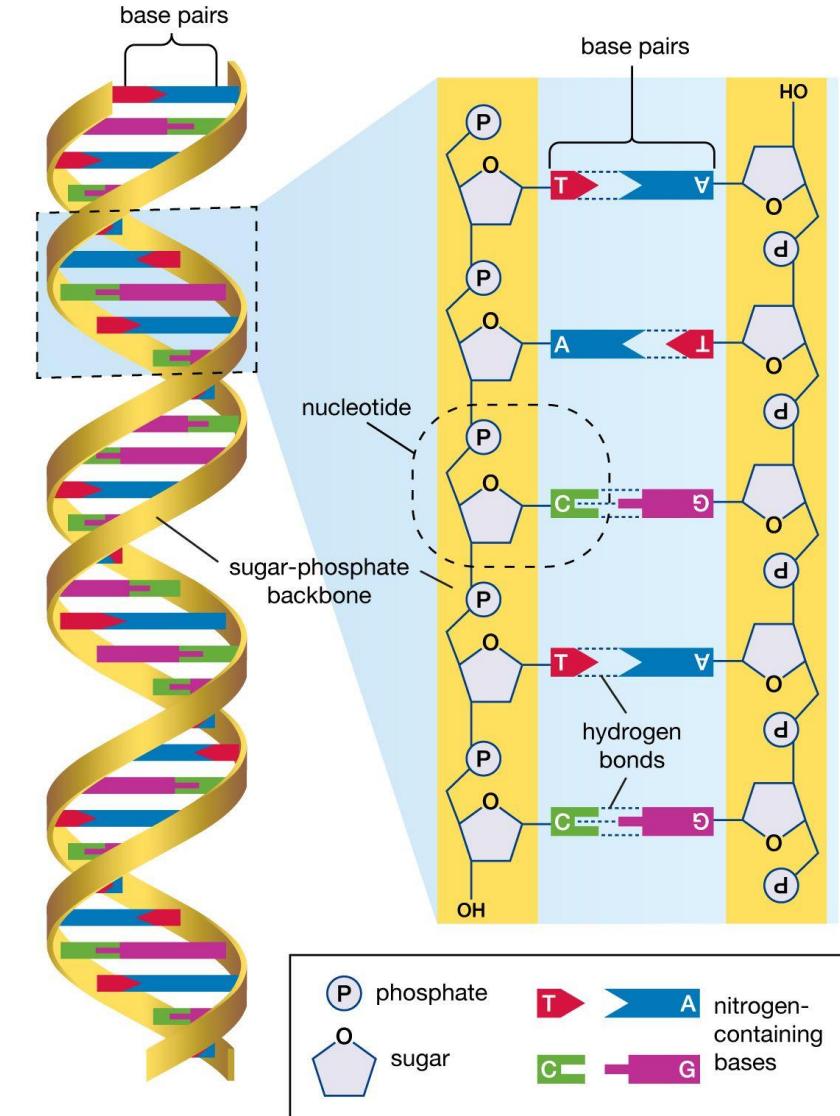
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Content

- DNA structure
- Human genome project (HGP)
- Human genome in a nutshell
- Human reference genome
- Genome components
- Genome 3D structure
- Gene structure
- Gene Annotation
- Gene conservation and mappability
- GEO NCBI
- ENA Browser EBI
- Hệ gen lâm sàng



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CpG

Race to discover
the 3D structure
of DNA

Figure 3. Rosalind Franklin at the Cabane des Evettes in the French Alps (c 1951). Photo credit: Vittorio Luzzati.



DNA Structure- Double
Helical
Watson & Crick Model

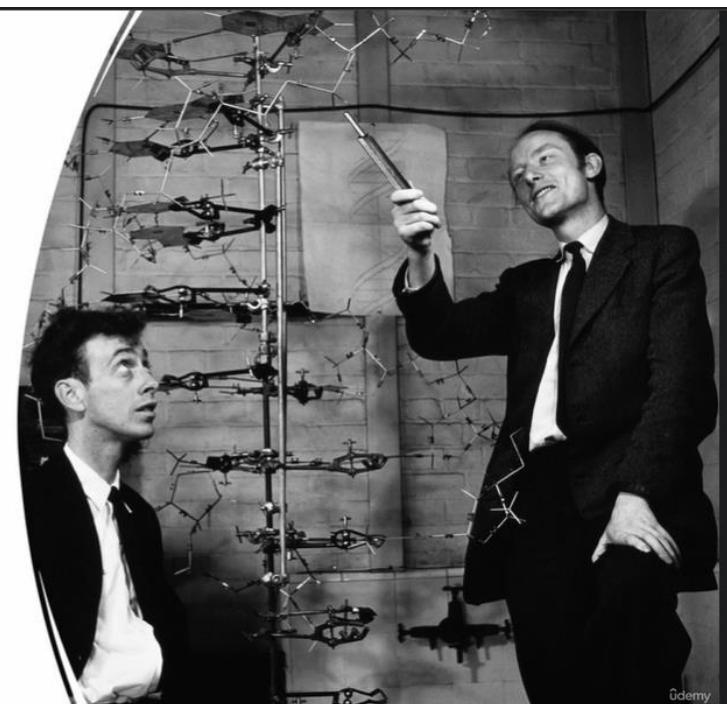
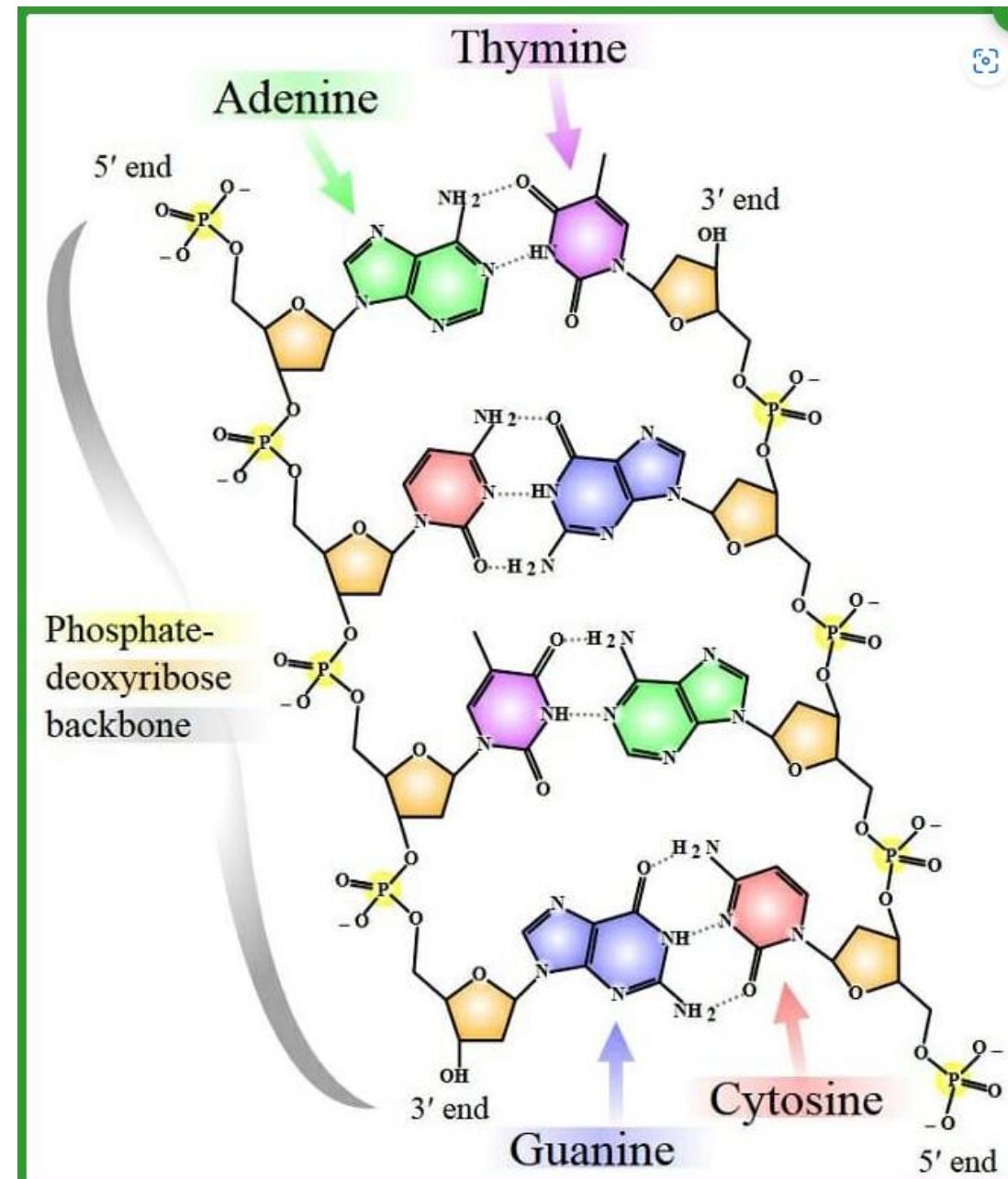


Figure 4. James Watson and Francis Crick with their DNA double helix model, 1953.

Photo credit: A. Barrington Brown/Science Photo Library



Structure of DNA

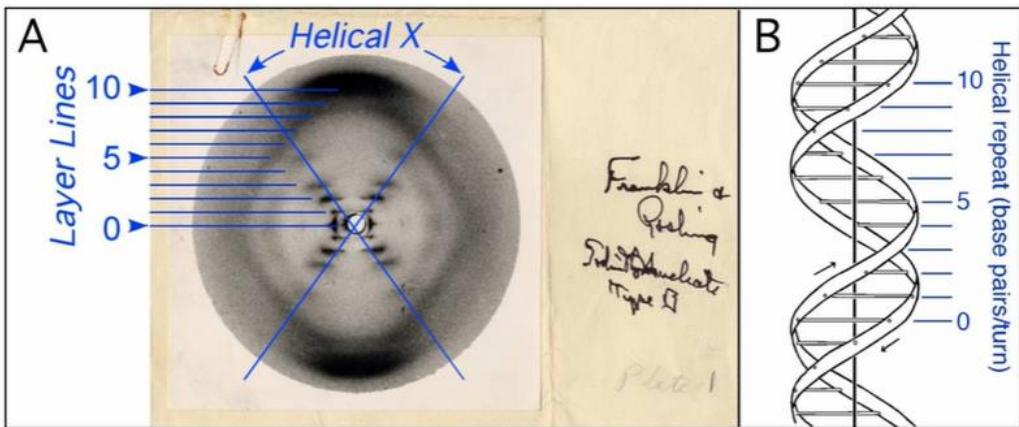


Figure 5. (A) Photo 51, the diffraction pattern from DNA in its so-called B configuration. The dimensions of DNA are: pitch $P = 3.4$ nm, radius $R = 1$ nm. Several important features include the characteristic X-shape and the ten diffracted orders per X. (B) A schematic diagram of DNA helical repeat structure. Photo is adapted from (Watson and Crick, 1953).

Structure of DNA

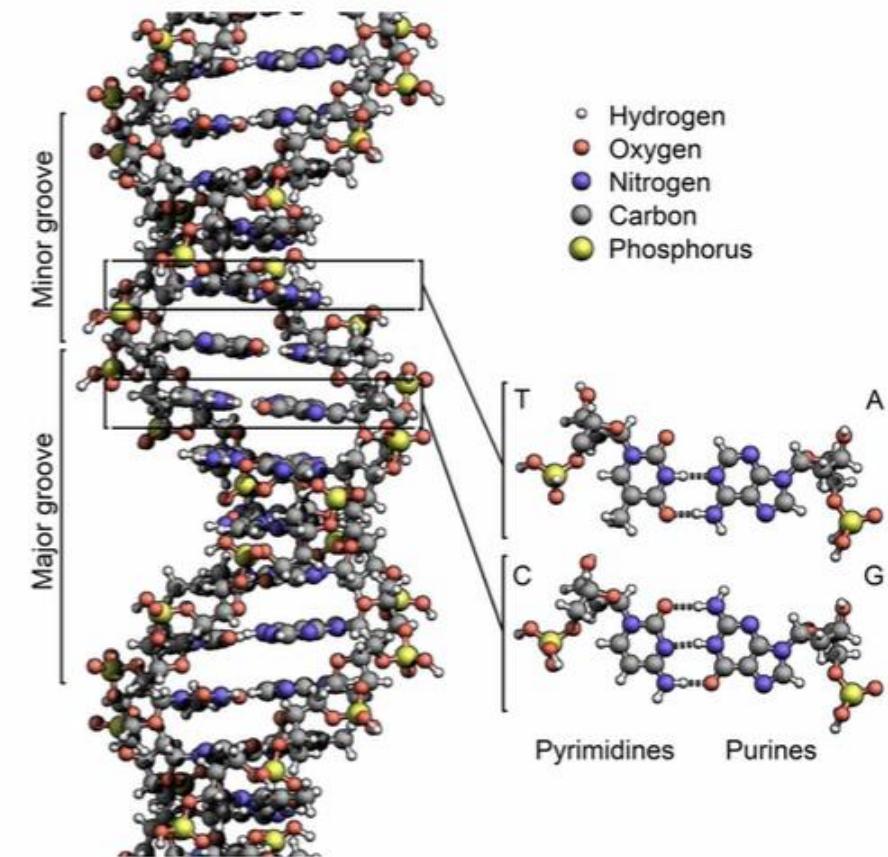
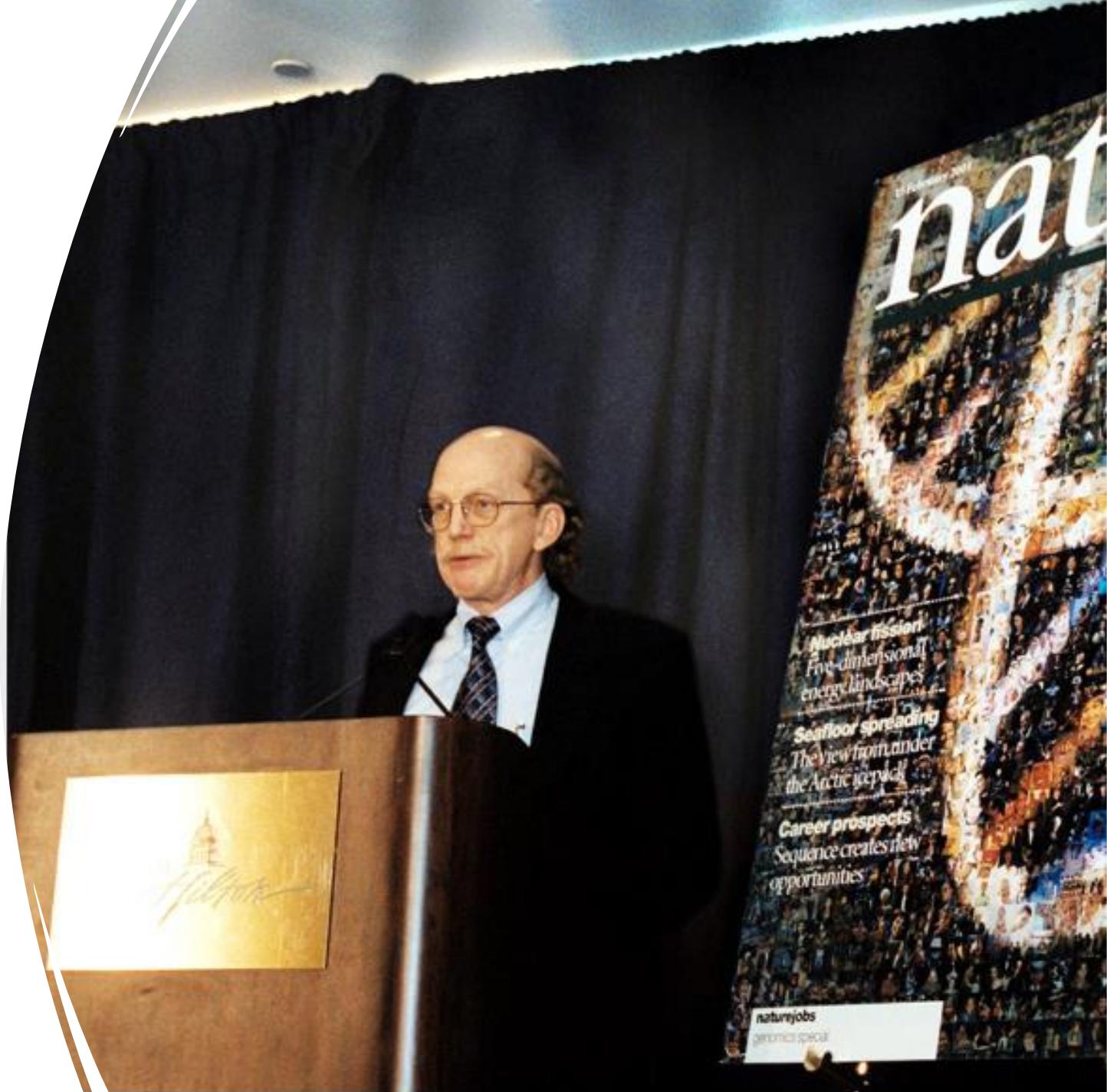


Figure 6. The structure of DNA. Image adapted from Zalli et al (2020).

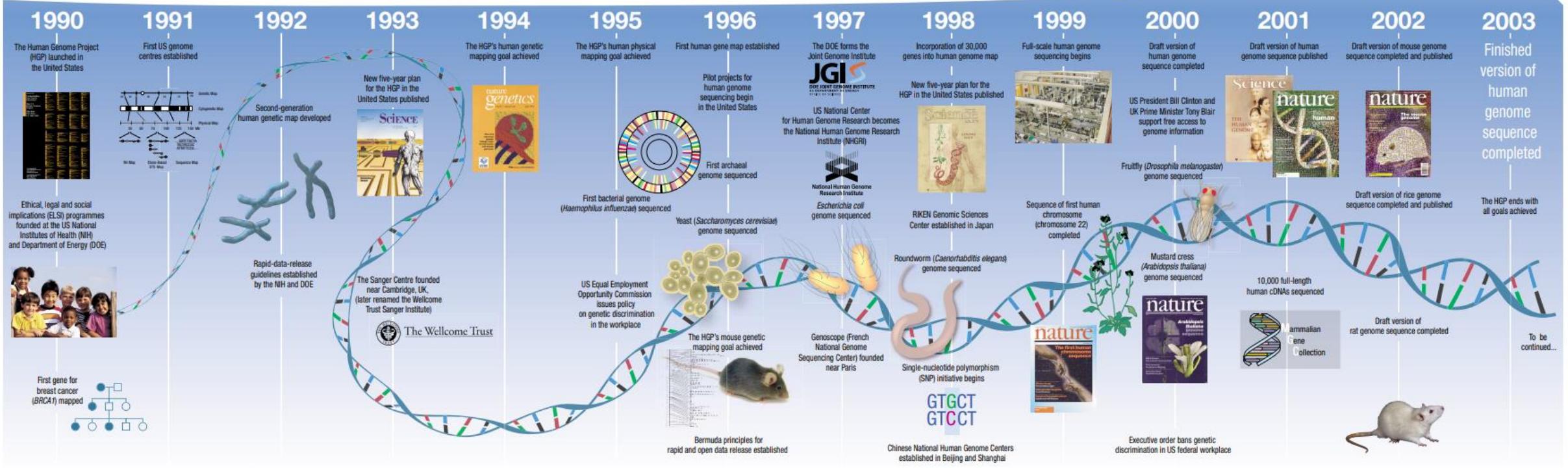
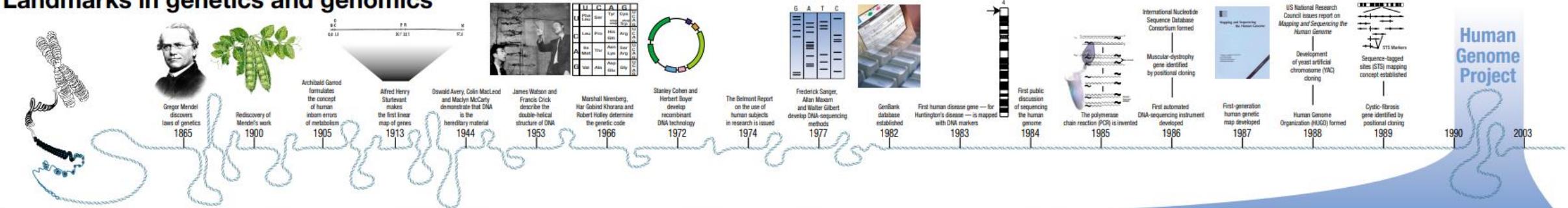
Human genome project (HGP)

- Was large, well-organized, and highly collaborative international effort that generated the **first sequence of the human genome** and that of several additional well-studied organisms.
- Carried out from 1990–2003, it was one of the most ambitious and important scientific endeavors in human history.



[Human Genome Project Timeline](#)

Landmarks in genetics and genomics



PEAS COURTESY J. BLAMIRE, CITY UNIV. NEW YORK; WATSON & CRICK COURTESY A. BARRINGTON BROWN/SPL; SCIENCE COVERS COURTESY AAAS

[Landmarks in genetics and genomics \(genome.gov\)](http://genome.gov)

Human genome project (HGP)

- The sequence of the human genome generated by the Human Genome Project was not from a single person.
- Rather, it reflects a patchwork from multiple people whose identities were intentionally made anonymous to protect their privacy.
- The project researchers used a thoughtful process to recruit volunteers, acquire their informed consent, and collect their blood samples.
- Most of the human genome sequence generated by the Human Genome Project came from blood donors in Buffalo, New York; specifically, 93% from **11 donors**, and 7% from **one donor**.

Human genome project (HGP)

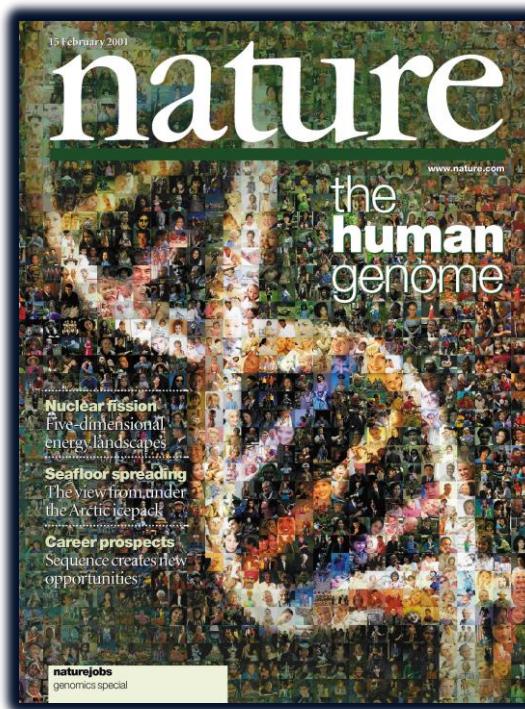
- The Human Genome Project could not have been completed as quickly and effectively without the dedicated participation of an international consortium of thousands of researchers.
- In the United States, the researchers were funded by the Department of Energy and the National Institutes of Health, which created the Office for Human Genome Research in 1988 (later renamed the National Center for Human Genome Research in 1990 and then the National Human Genome Research Institute in 1997).
- The sequencing of the human genome involved researchers from 20 separate universities and research centers across the United States, United Kingdom, France, Germany, Japan and China.
- The groups in these countries became known as the International Human Genome Sequencing Consortium.

Human genome project (HGP)

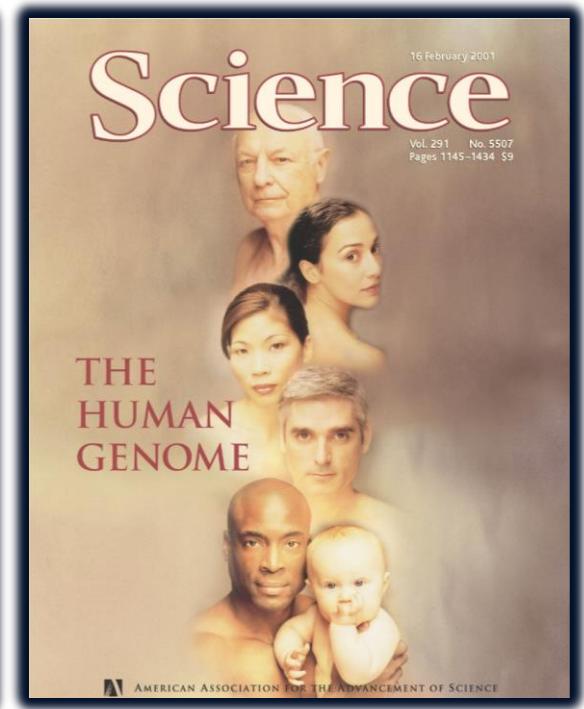
- In June 2000, the International Human Genome Sequencing Consortium [announced](#) that it had produced a draft human genome sequence that accounted for 90% of the human genome. The draft sequence contained more than 150,000 areas where the DNA sequence was unknown because it could not be determined accurately (known as gaps).
- In April 2003, the consortium [announced](#) that it had generated an essentially complete human genome sequence, which was significantly improved from the draft sequence. Specifically, it accounted for 92% of the human genome and less than 400 gaps; it was also more accurate.
- On March 31, 2022, the Telomere-to-Telomere (T2T) consortium announced that had filled in the remaining gaps and produced the [first truly complete human genome sequence](#).

Human Genome Project (Oct 1990 - April 2003)

- In 2003, the Human Genome Project produced a genome sequence that accounted for over 90% of the human genome (~3 GB).
 - It was as close to complete as the technologies for sequencing DNA allowed at the time.
- => Facilitating advancements in **next-generation sequencing (NGS)** technologies



HGP Paper

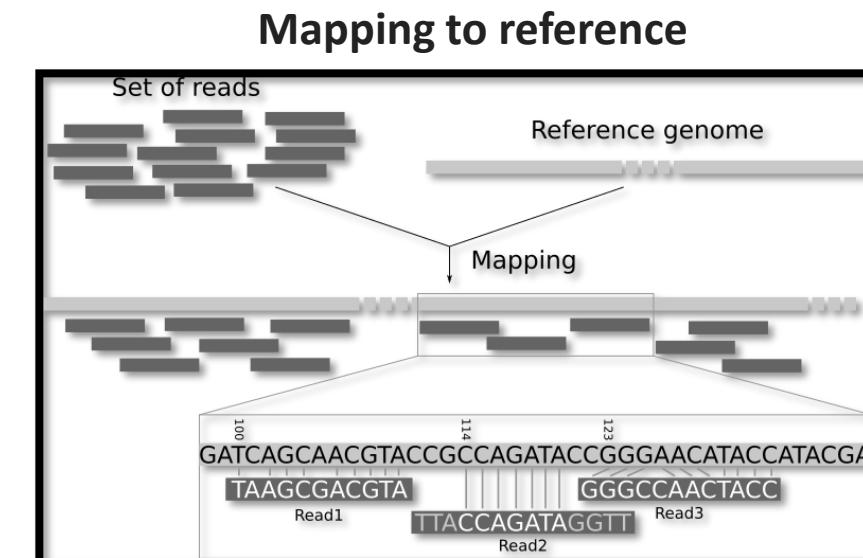
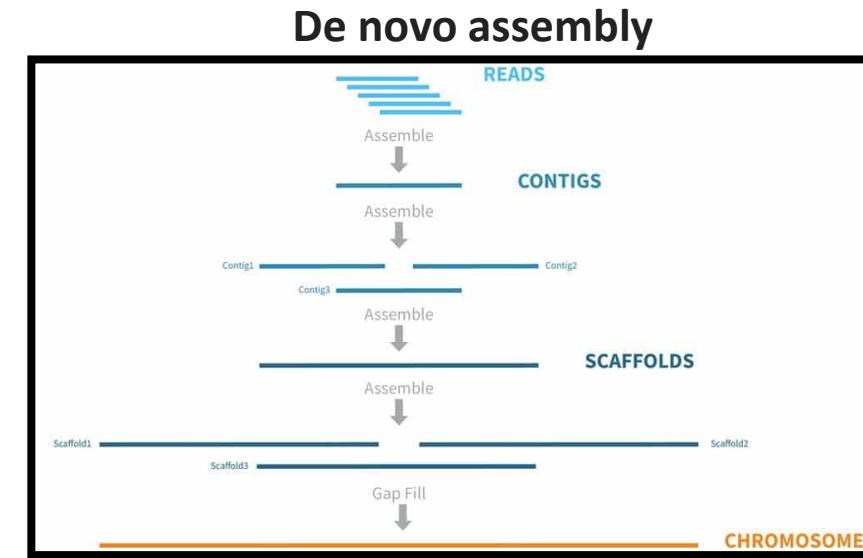
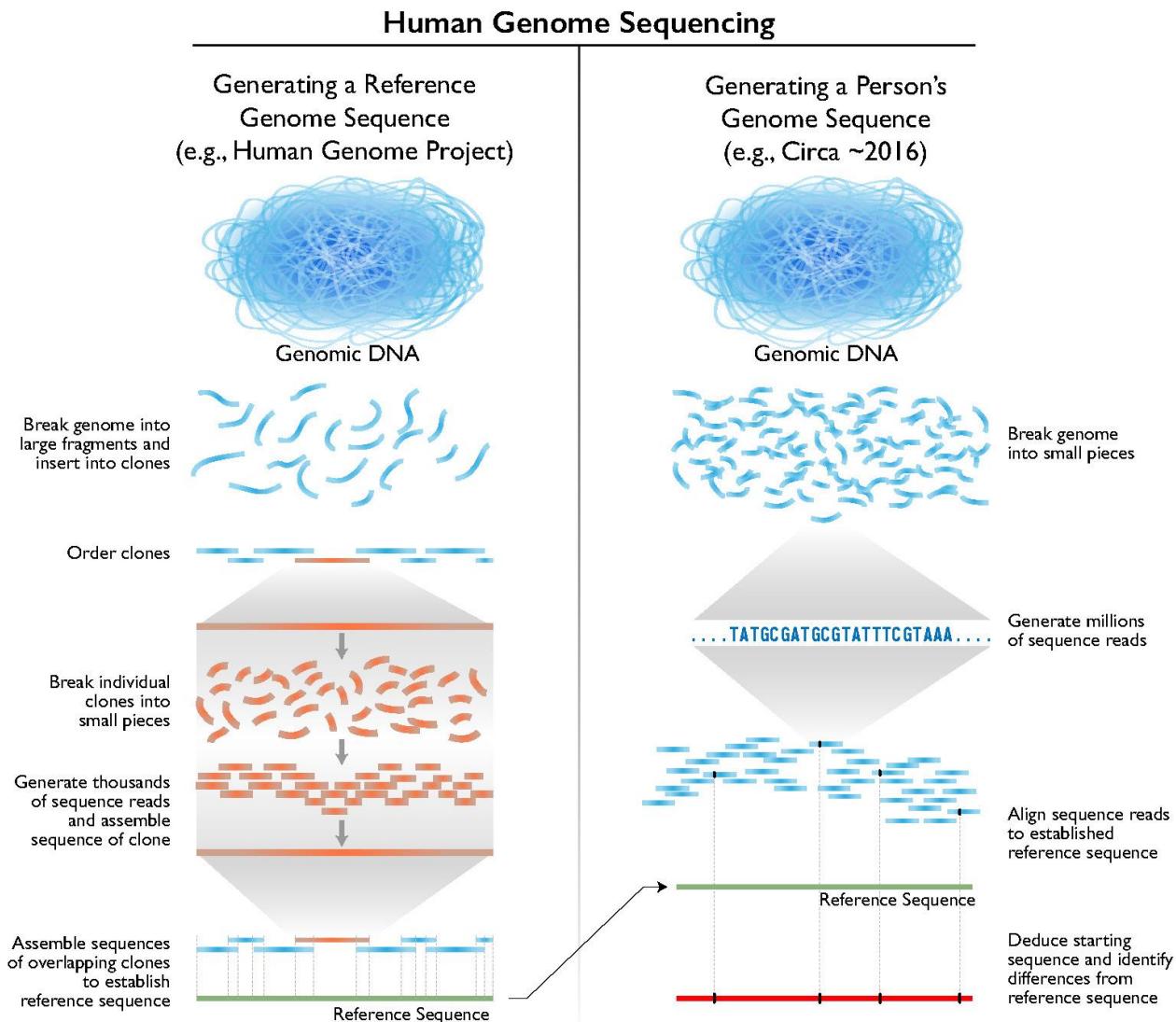


Venter/Celera Paper



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Next-Generation Sequencing (NGS)



NGS: Reference Genome

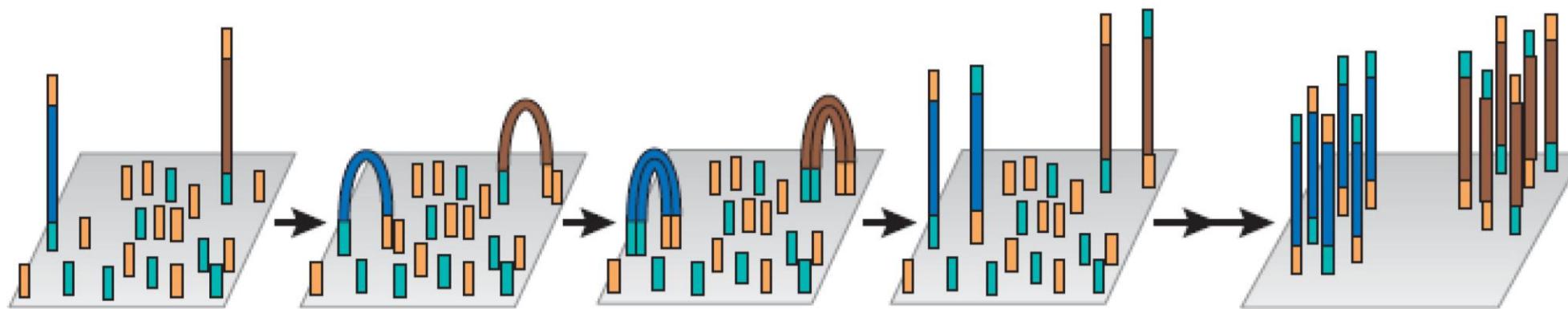
De novo assembly



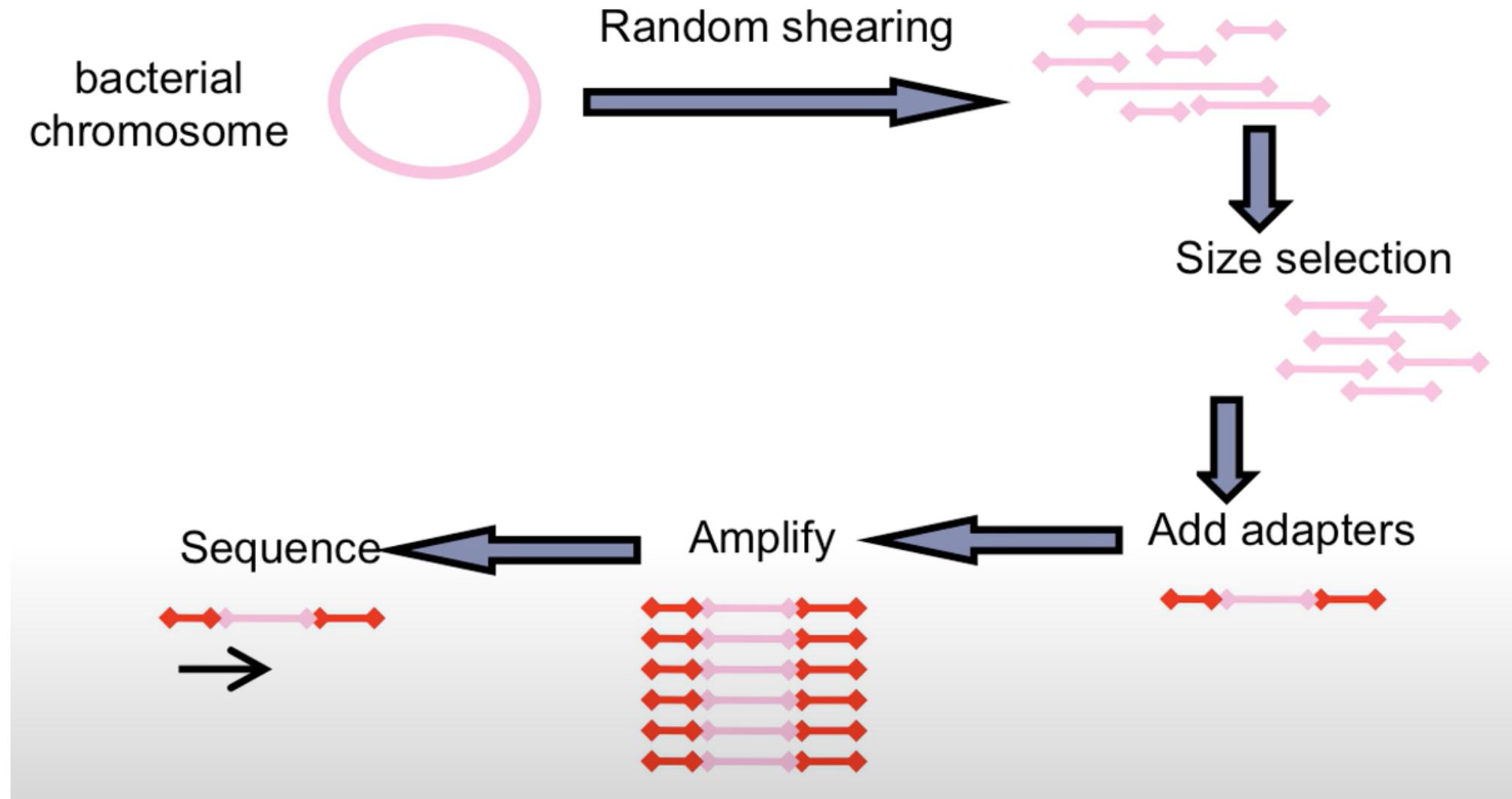
Mapping to reference



NGS: Massive Parallel Sequencing



Whole genome sequencing – Shotgun method

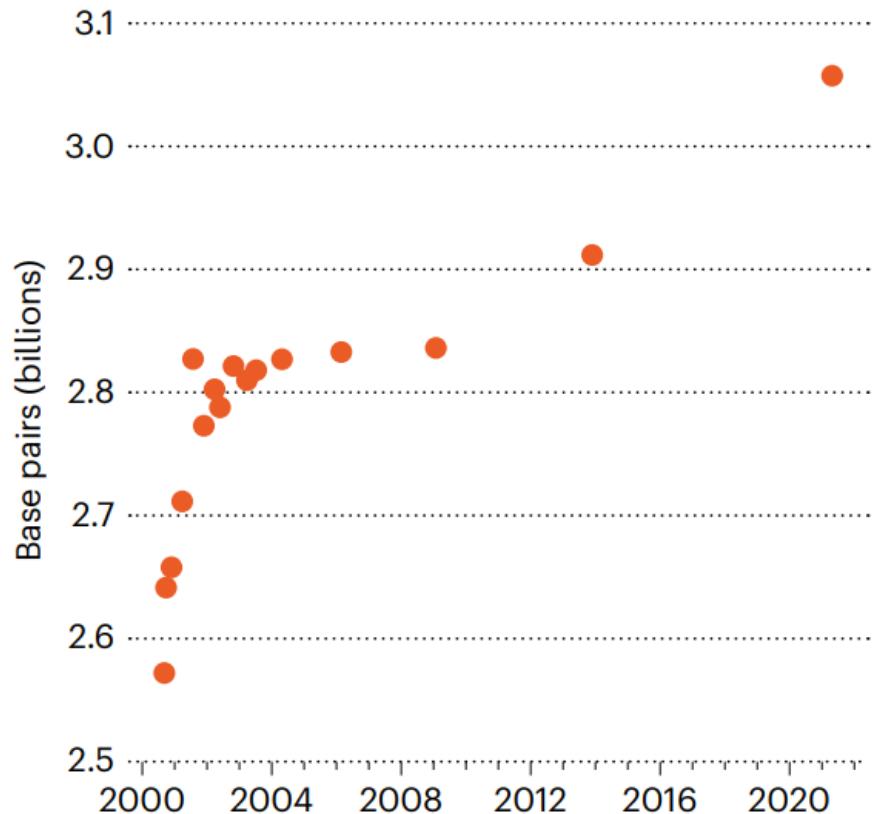


A COMPLETE HUMAN GENOME IS CLOSE: HOW THE GAPS WERE FILLED

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.

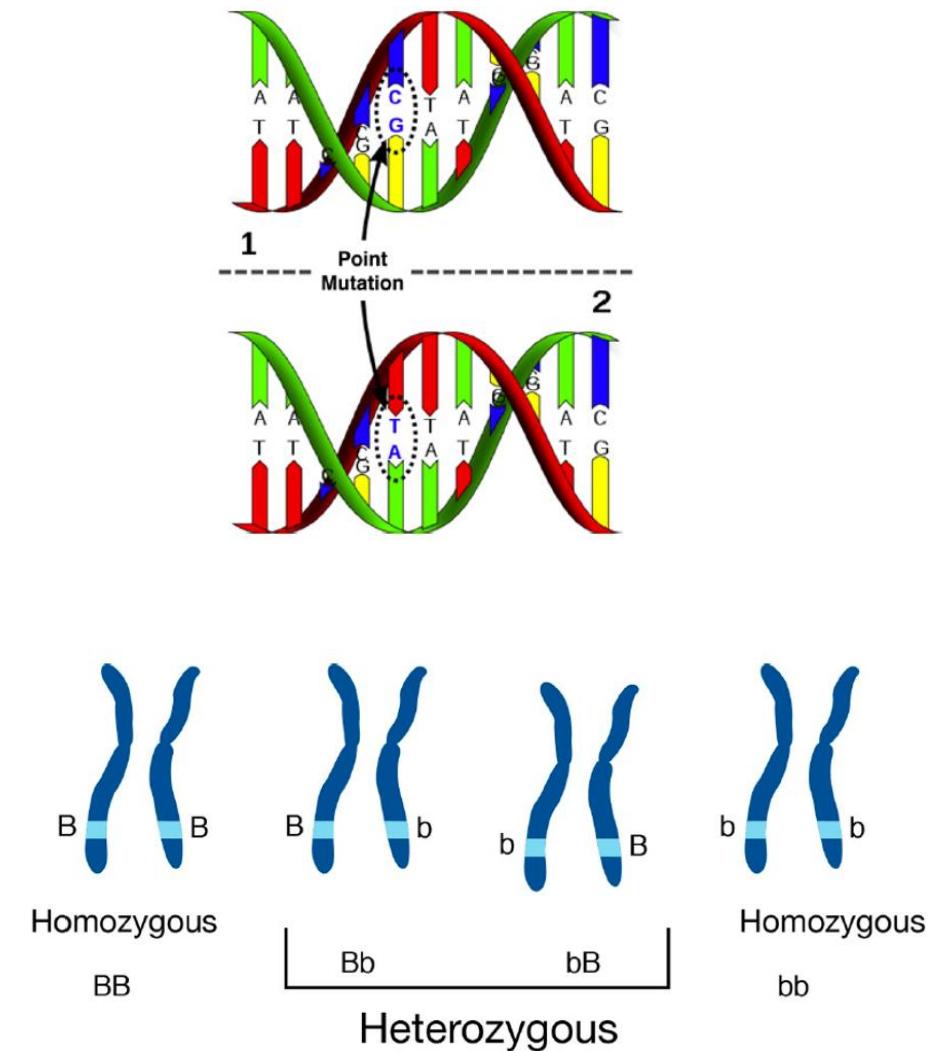
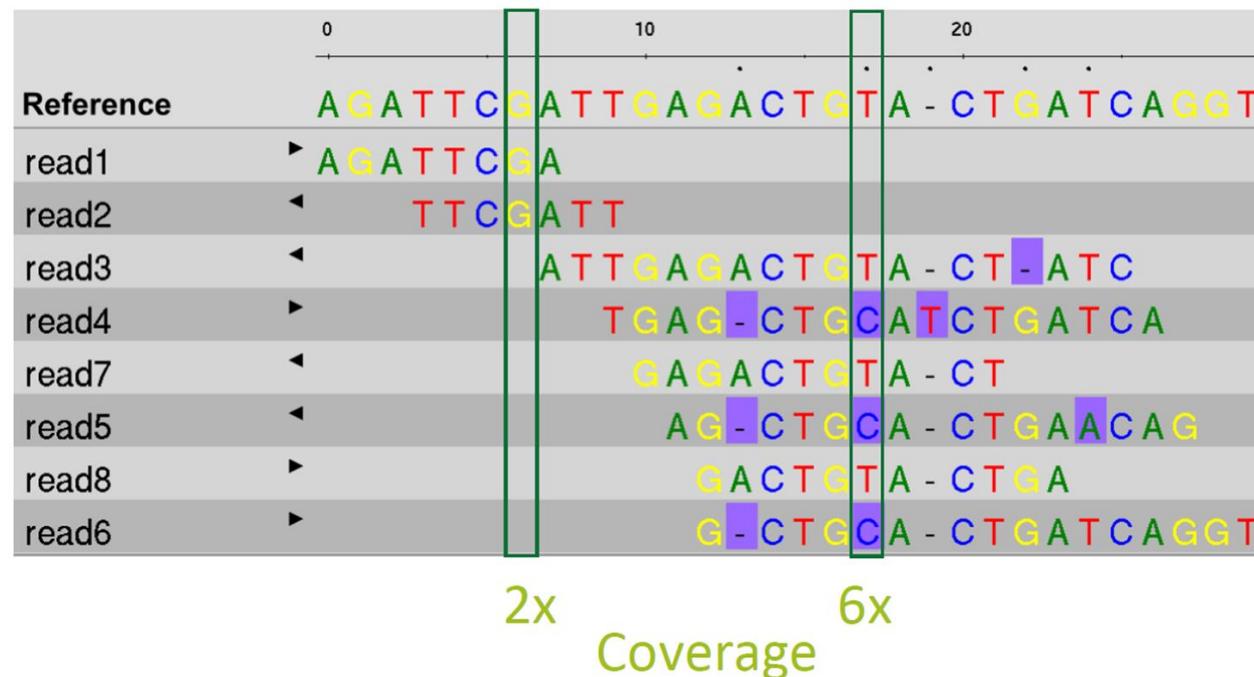


0.3% of sequence might still have errors. Includes X but not Y chromosome. Count excludes mitochondrial DNA.

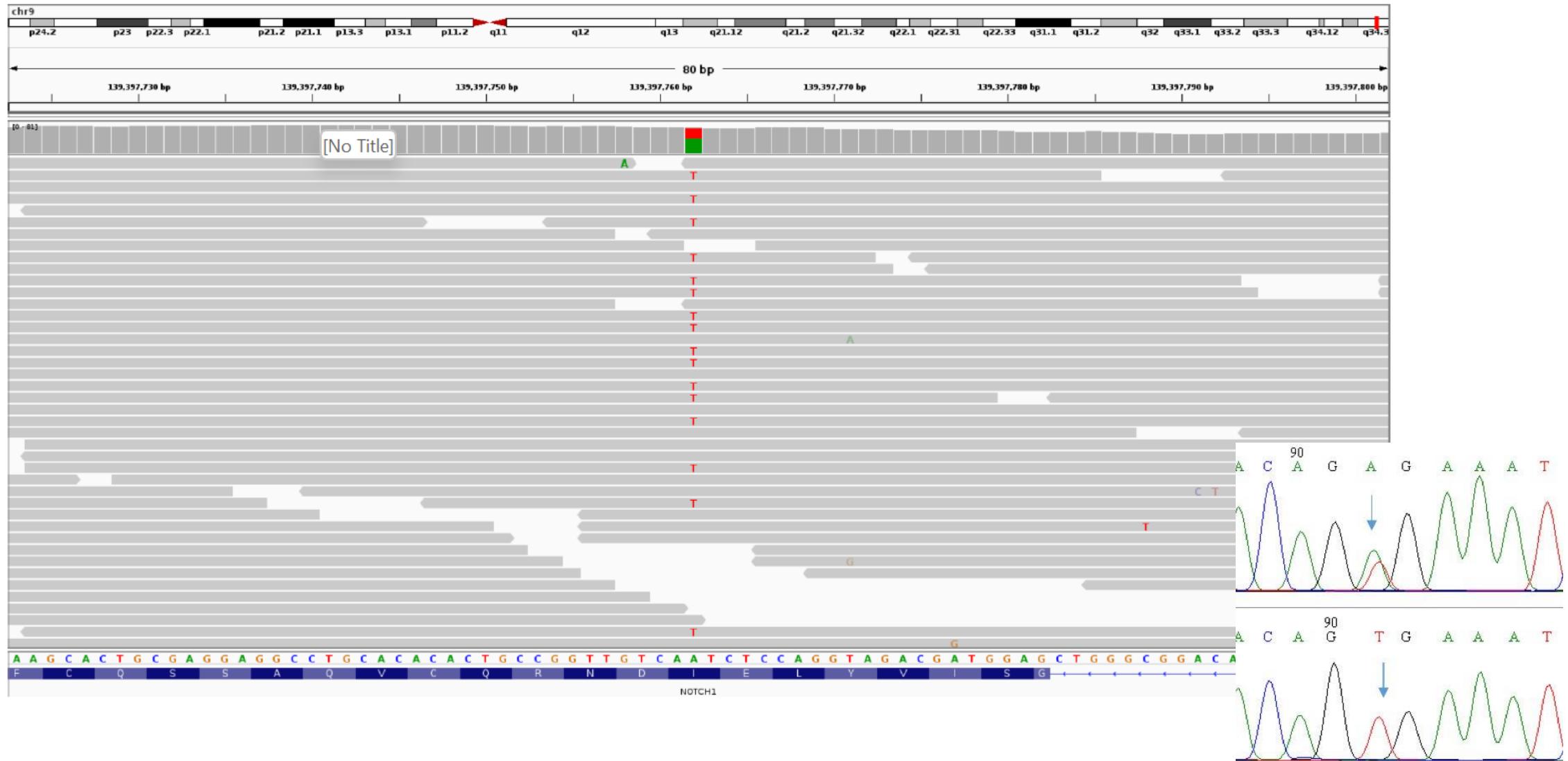
SOURCE: ADAM PHILLIPY

Human Genome Variation

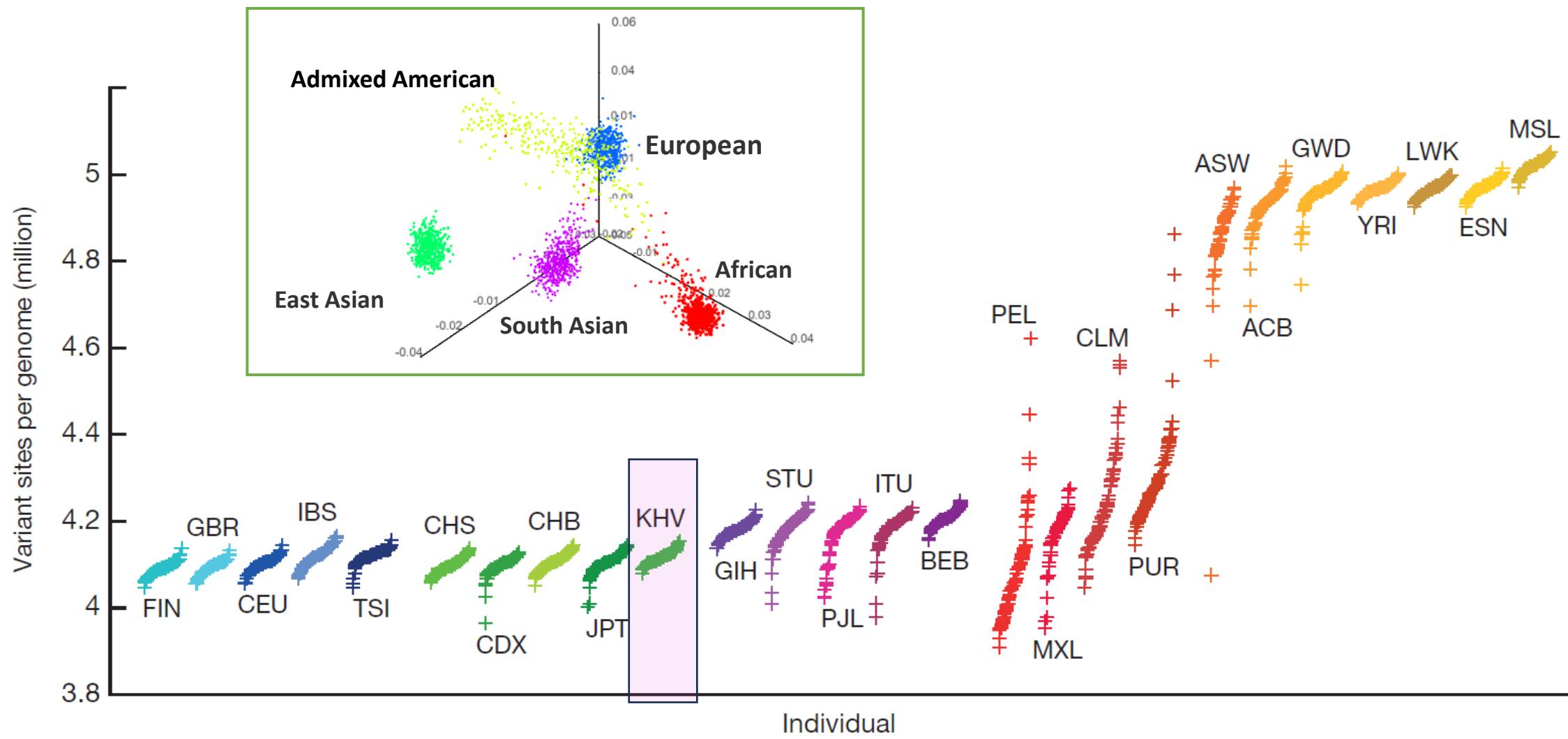
- Human genomes are > 99% similar by sequence
- A typical human genome has ~ 5 million variants with 3-4 million single nucleotide variants
- Humans are diploid



Alignment and variant viewers

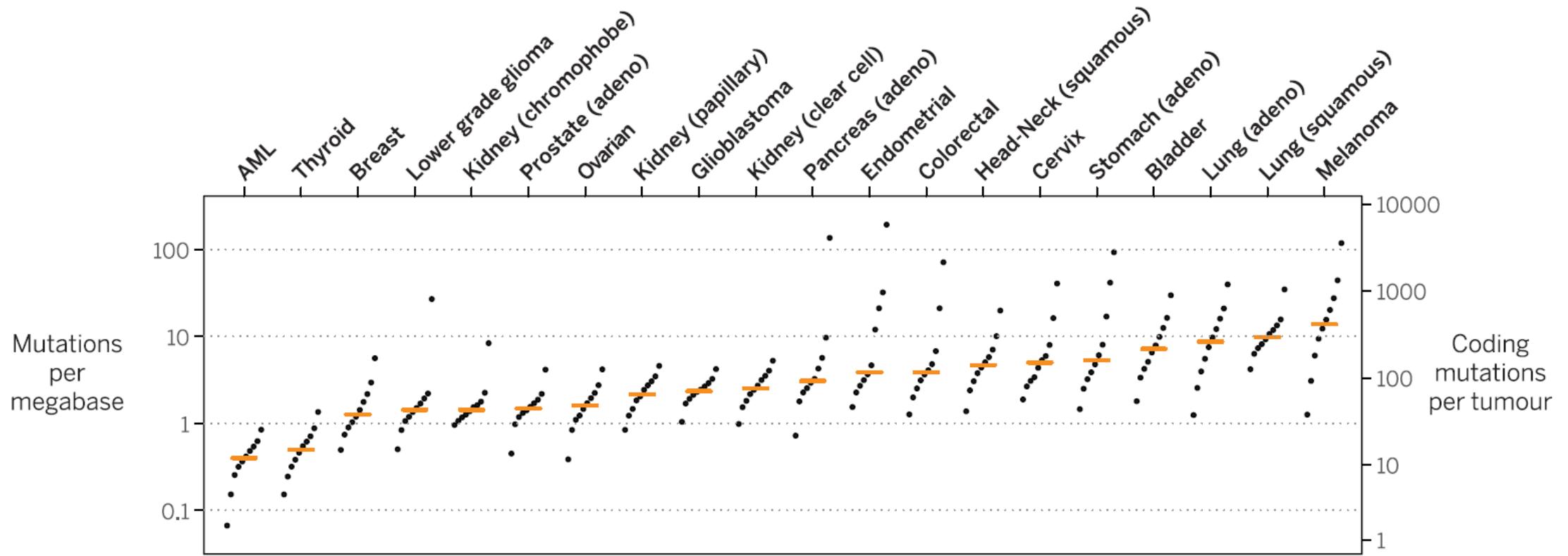


Human Genome Variation: 1000 Genomes Project



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)

(35) Lessons from the Human Genome Project - YouTube

- <https://www.youtube.com/watch?v=qOW5e4BgEa4>

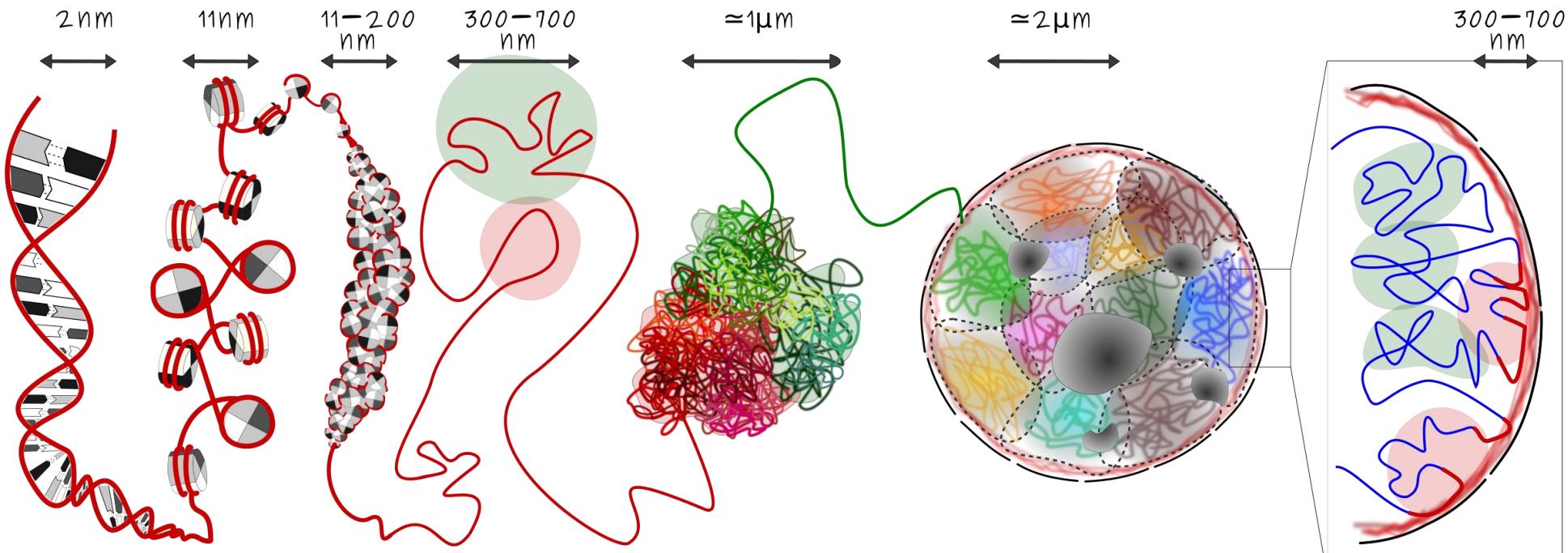
How did the Human Genome Project affect biological research in general?

- Discuss and give an example!

How did the Human Genome Project affect medical research and clinical application in general?

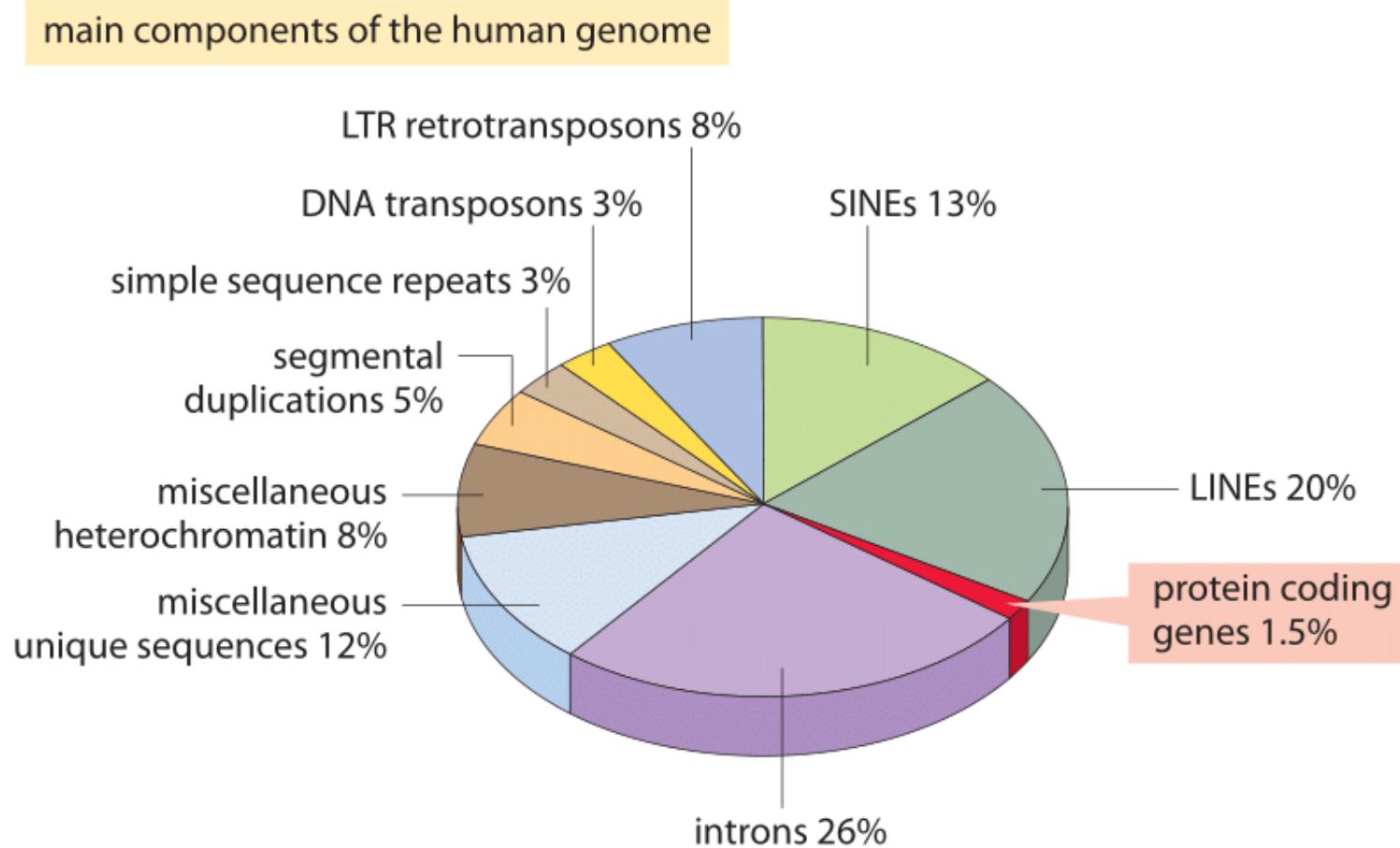
- Discuss and give an example!

DNA Chromatin fiber DNA loop & Compartment, Chromatin/TAD domains Chromosome territory inside cell nucleus Lamina-associated domain(LAD)



- Nucleobases
- Nucleosome, composed of 8 histones proteins
- Repressive chromatin domain, mainly heterochromatin
- Active chromatin domain, mainly euchromatin
- Chromosome territory
- Nucleolus
- Nuclear speckles
- Nuclear membrane, including schematic nuclear pores
- Nuclear lamina
- LAD

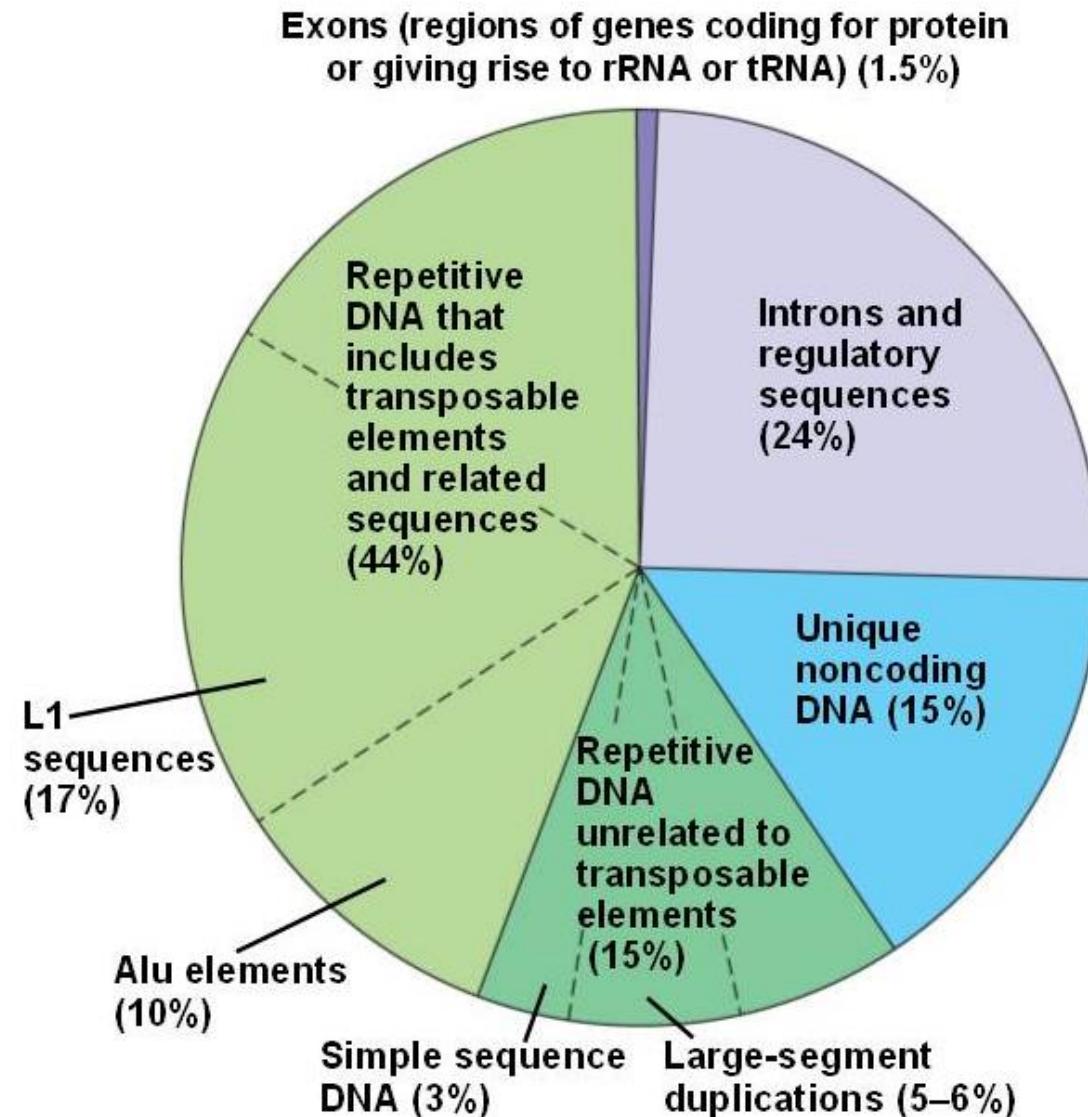
Genome components



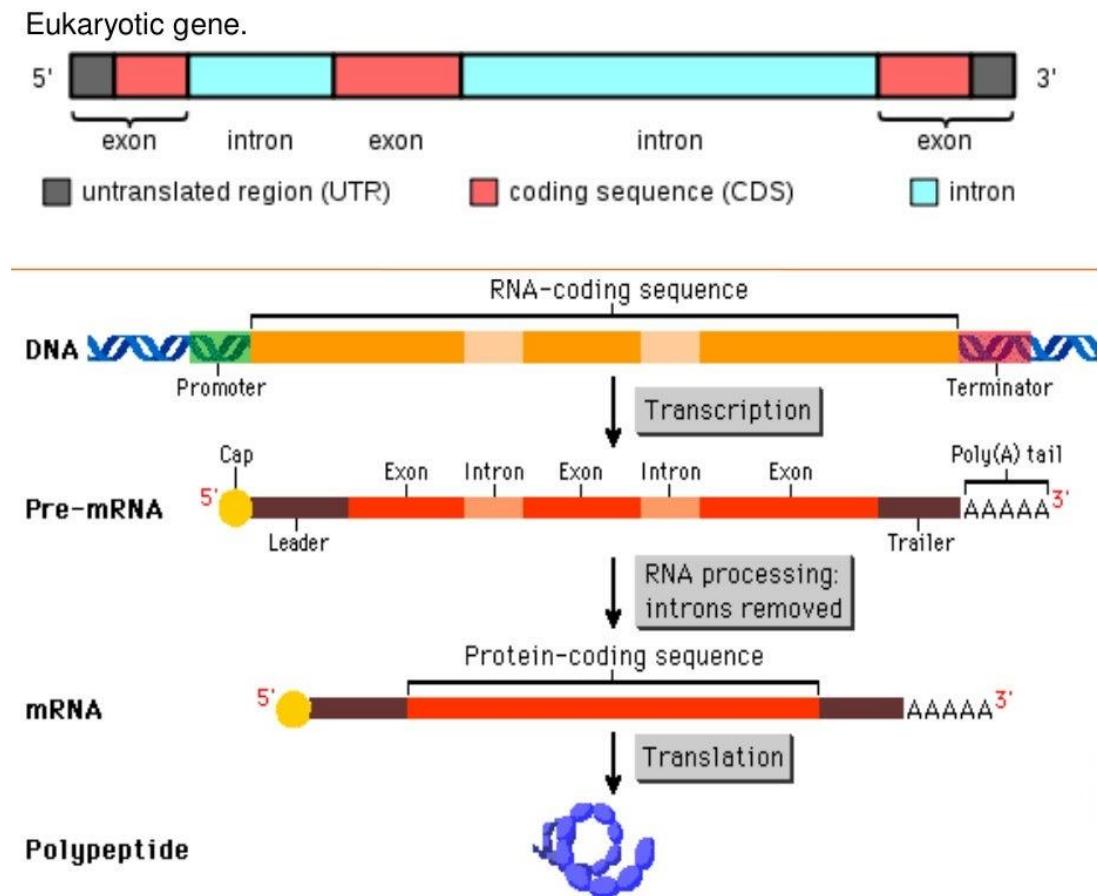
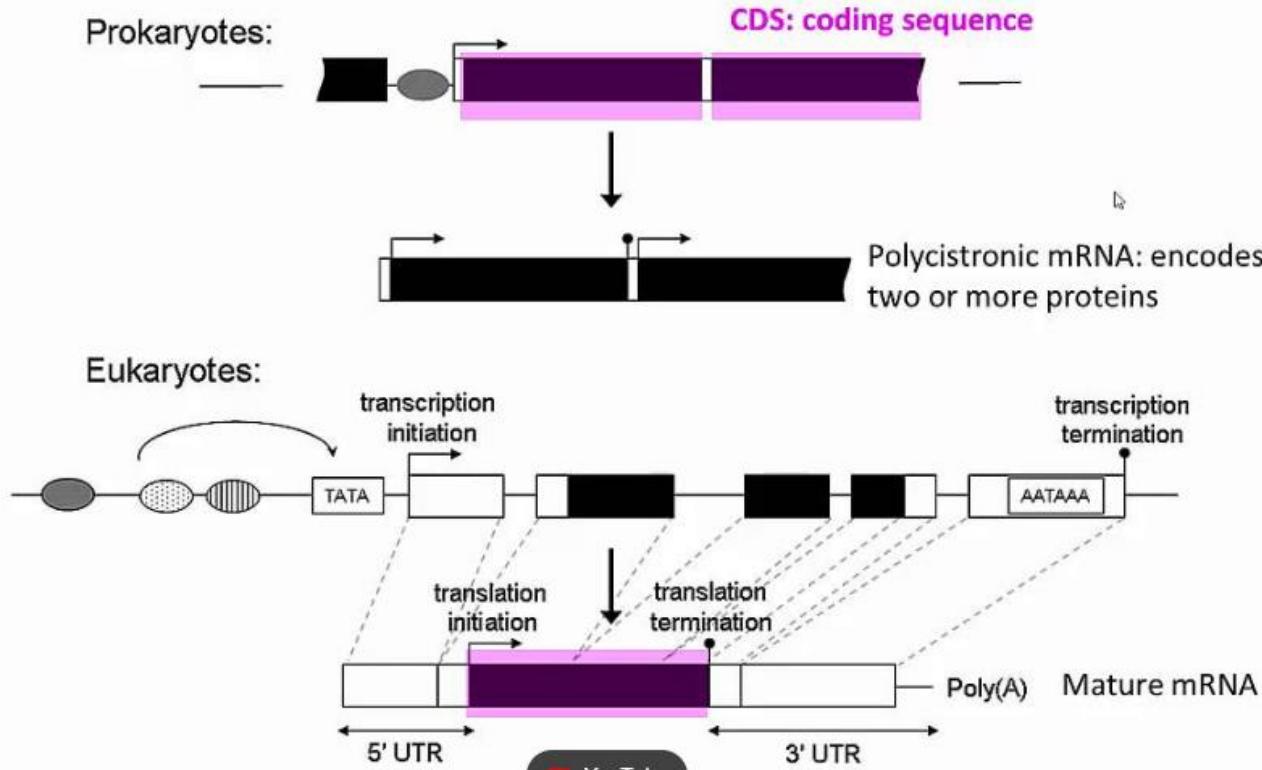
[» How many genes are in a genome? \(bionumbers.org\)](https://www.bionumbers.org)

Genome components

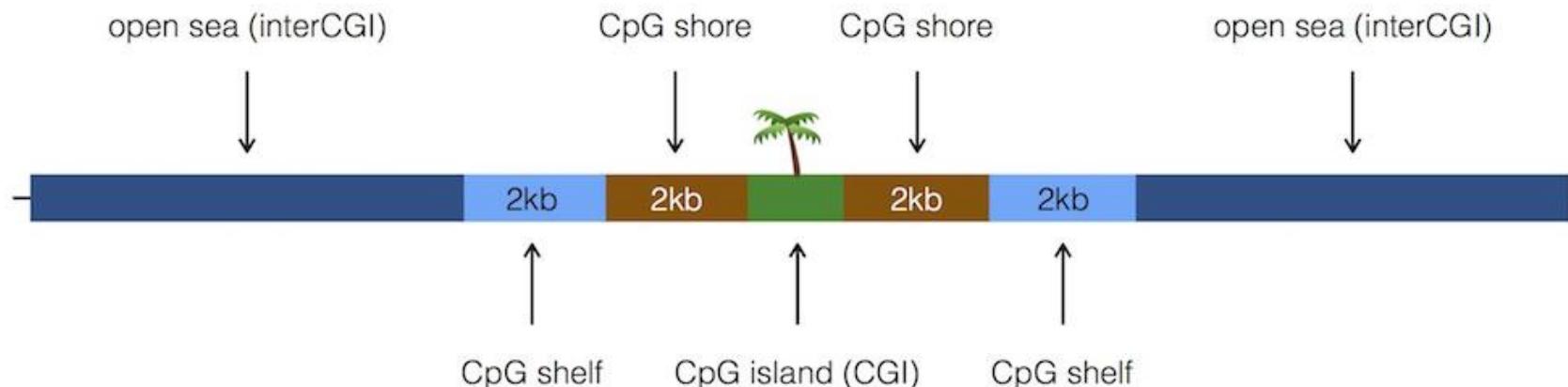
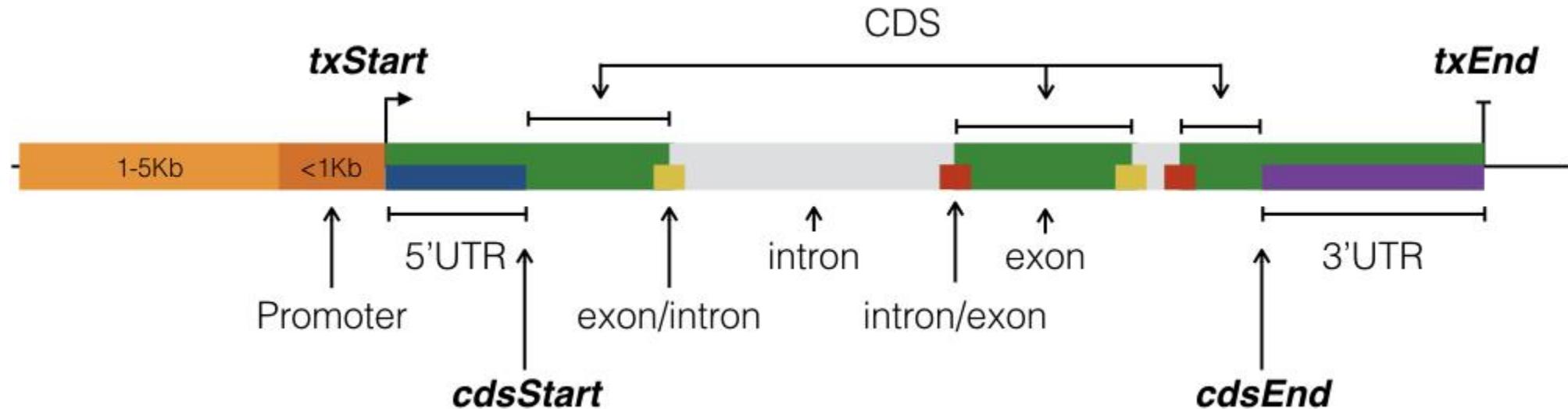
- The human genome contains less than 2% coding exons within genes.
- The remaining DNA consist of:
 - introns and regulatory sequences such as enhancers
 - unique noncoding DNA contains many pseudogenes (genes that have accumulated mutations and became nonfunctional)
 - repetitive DNA are sequences that are repeated many times; much of these are
 - transposable elements that can "jump" between chromosomes, leading to transpositions.



Gene structure



Gene structure



Understanding Gene Annotation through GENCODE

(<https://www.gencodegenes.org/>)

The screenshot shows the GENCODE website homepage. At the top, there's a navigation bar with links for Human, Mouse, How to access data, FAQ, Documentation, and About us. Below the navigation bar are two main sections: "HUMAN" and "MOUSE". The HUMAN section features a portrait of a person split vertically, with the left side in shadow and the right side in light. It is labeled "GENCODE 43 (08.02.23)". The MOUSE section features a photo of a grey mouse. It is labeled "GENCODE M32 (08.02.23)". Below these sections, a text block states the project's goal: "The goal of the GENCODE project is to identify and classify all gene features in the human and mouse genomes with high accuracy based on biological evidence, and to release these annotations for the benefit of biomedical research and genome interpretation." Three callout boxes provide additional information: 1) "GENCODE are [updating the annotation](#) of human protein-coding genes linked to SARS-CoV-2 infection and COVID-19 disease." 2) "The [Long-read RNA-seq Genome Annotation Assessment Project \(LRGASP\)](#) Consortium for systematic evaluation of different methods for transcript computational identification and quantification using long-read sequence data [has launched](#)." 3) "GENCODE are [supporting the annotation](#) of non-canonical human ORFs predicted by Ribo-seq data."

HUMAN
GENCODE 43 (08.02.23)



MOUSE
GENCODE M32 (08.02.23)



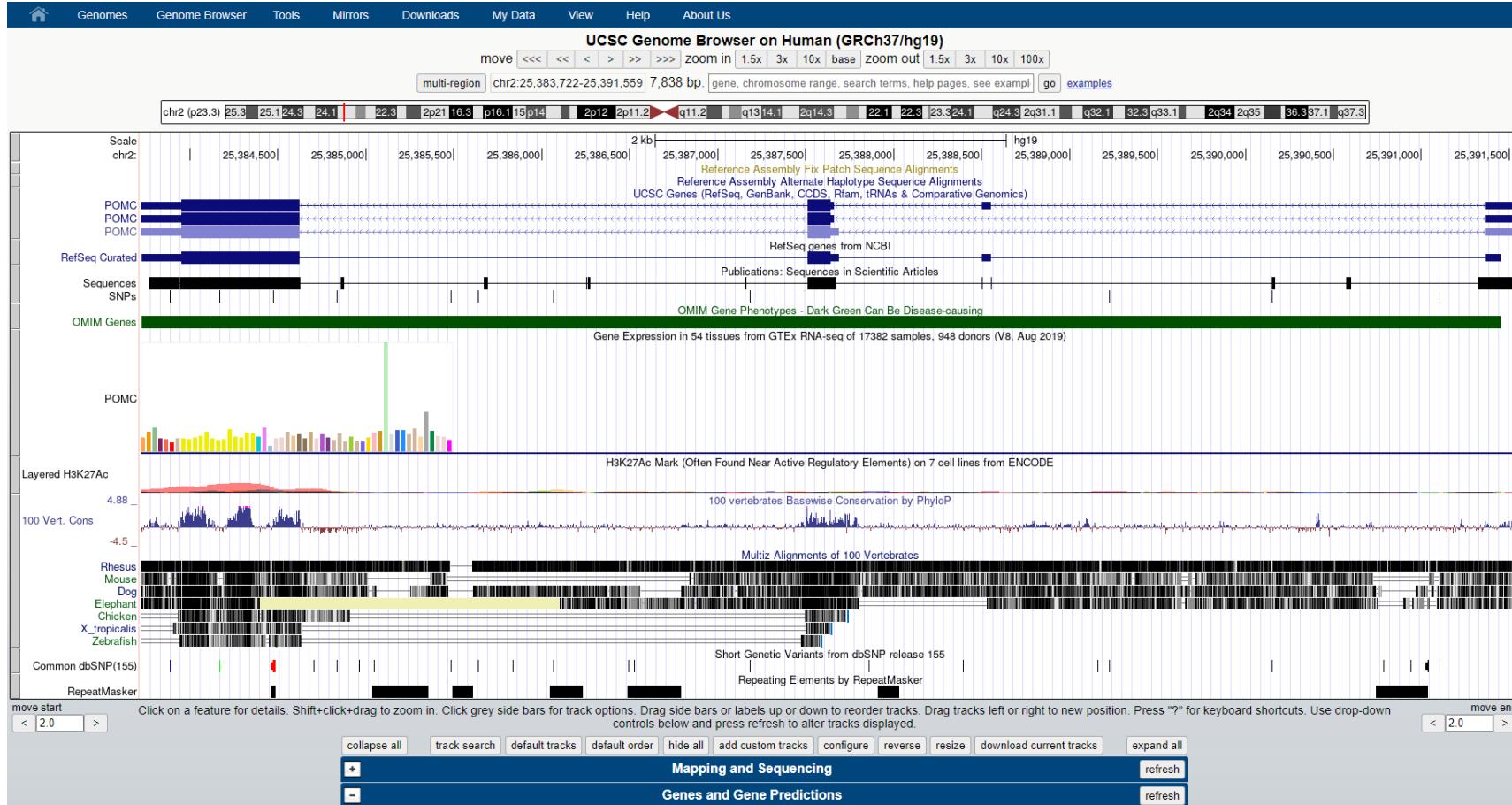
The goal of the GENCODE project is to identify and classify all gene features in the human and mouse genomes with high accuracy based on biological evidence, and to release these annotations for the benefit of biomedical research and genome interpretation.

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GENCODE are [supporting the annotation](#) of non-canonical human ORFs predicted by Ribo-seq data.

Understanding Human reference genome through the UCSC Genome Browser (<https://genome.ucsc.edu/>)



GEO NCBI (<https://www.ncbi.nlm.nih.gov/geo/>)

The screenshot shows the GEO Accession Display page for the series GSE48213. The page has a header with the NCBI logo and the GEO logo. It includes a navigation bar with links for HOME, SEARCH, SITE MAP, GEO Publications, FAQ, MIAME, and Email GEO. The main content area displays the series details for GSE48213, including its status as public on Aug 20, 2013, and its title as transcriptional profiling of a breast cancer cell line panel using RNAseq technology. The page also lists the organism (Homo sapiens), experiment type (Expression profiling by high throughput sequencing), and a summary of the study. The overall design is described as profiling cell lines in their baseline, unperturbed state. Contributors listed are Gray JW and Heiser LM, and a citation from Daemen A et al. (2013) is provided.

Scope: Format: Amount: GEO accession:

Series GSE48213 [Query DataSets for GSE48213](#)

Status	Public on Aug 20, 2013
Title	Transcriptional profiling of a breast cancer cell line panel using RNAseq technology
Organism	Homo sapiens
Experiment type	Expression profiling by high throughput sequencing
Summary	56 breast cancer cell lines were profiled to identify patterns of gene expression associated with subtype and response to therapeutic compounds.
Overall design	Cell lines were profiled in their baseline, unperturbed state.
Contributor(s)	Gray JW , Heiser LM
Citation(s)	Daemen A, Griffith OL, Heiser LM, Wang NJ et al. Modeling precision treatment of breast cancer. <i>Genome Biol</i> 2013;14(10):R110. PMID: 24176112

ENA

(<https://www.ebi.ac.uk/ena/browser/home>)

The screenshot shows the ENA (European Nucleotide Archive) browser homepage. At the top, there is a navigation bar with links to various services like MPI Mail, Garvan, Arraylog - Google S..., Machine_Learning, Slack, Welcome - Sapphire, How do you know i..., Garvan Intranet, and NSWU Mail - Phuc... There is also a search bar and a user profile icon.

The main header features the ENA logo with a DNA helix graphic and the text "European Nucleotide Archive". Below the header is a navigation menu with links to Home, Submit, Search, Rulespace, About, and Support.

The main content area displays project details for PRJNA210428. It includes a summary: "56 breast cancer cell lines were profiled to identify patterns of gene expression associated with subtype and response to therapeutic compounds." and "Overall design: Cell lines were profiled in their baseline, unperturbed state." To the right of this summary is a help icon (a question mark inside a circle).

On the right side of the page, there is a sidebar with various options:

- View:** XML (selected), XML (STUDY)
- Download:** XML (selected), XML (STUDY)
- Navigation:** Show
- Read Files:** Hide (selected)
- Publications:** Show
- Parent Projects:** Show
- Related ENA Records:** Show

Project: PRJNA210428

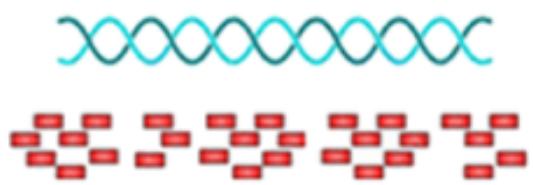
56 breast cancer cell lines were profiled to identify patterns of gene expression associated with subtype and response to therapeutic compounds.
Overall design: Cell lines were profiled in their baseline, unperturbed state.

Organism:	Homo sapiens (human)
Secondary Study	SRP026537
Accession:	
Study Title:	Transcriptional profiling of a breast cancer cell line panel using RNAseq technology
Center Name:	OHSU
Study Name:	Homo sapiens
ENA-REFSEQ:	N

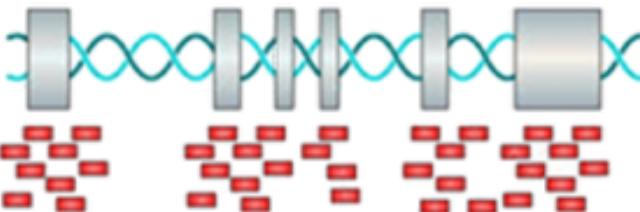
Clinical Genomics

Clinical applications of NGS

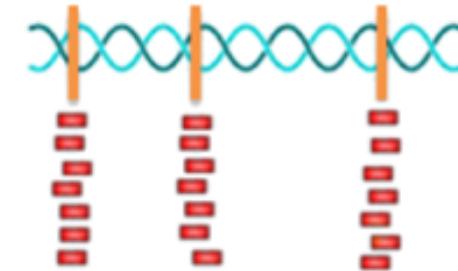
Whole genome sequencing



Whole exome sequencing



Targeted sequencing

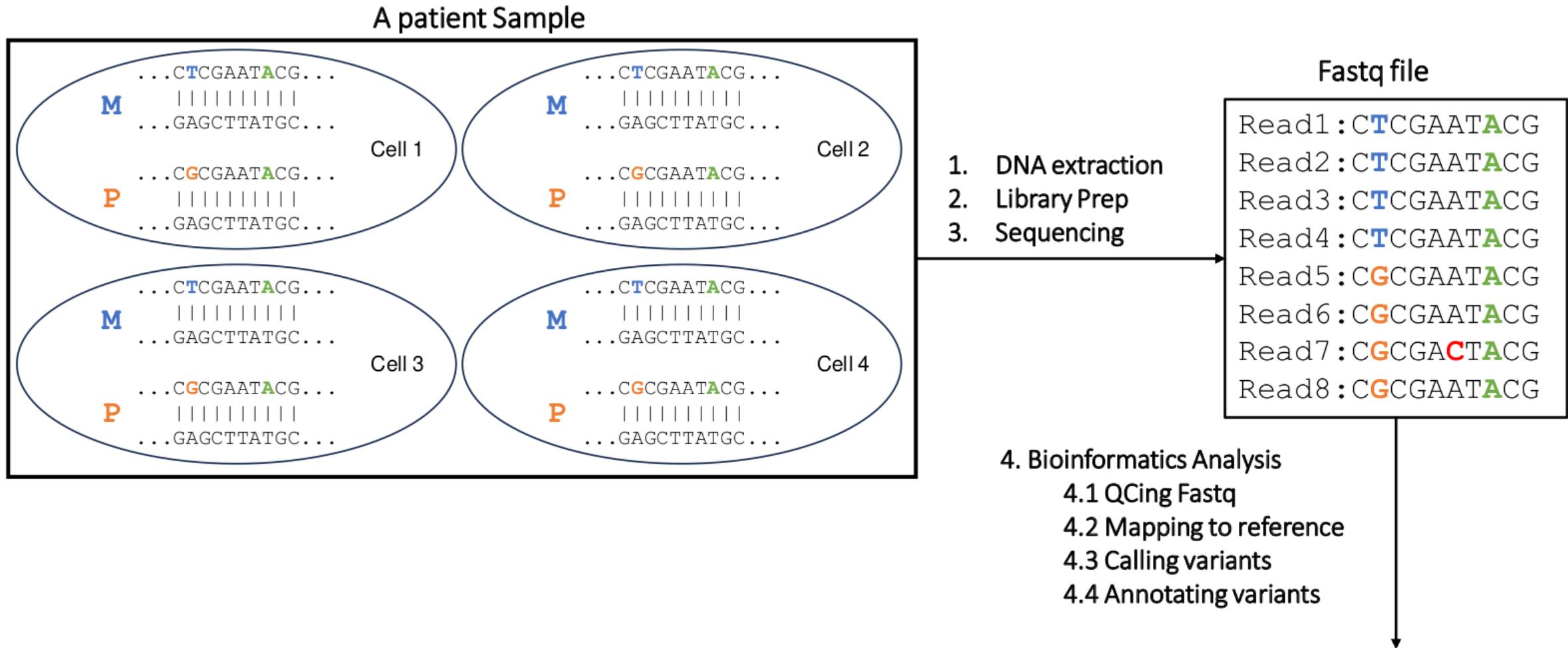


- Sequencing region : whole genome
- Sequencing Depth: >30X
- Covers everything – can identify all kinds of variants including SNPs, INDELs and SV.

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

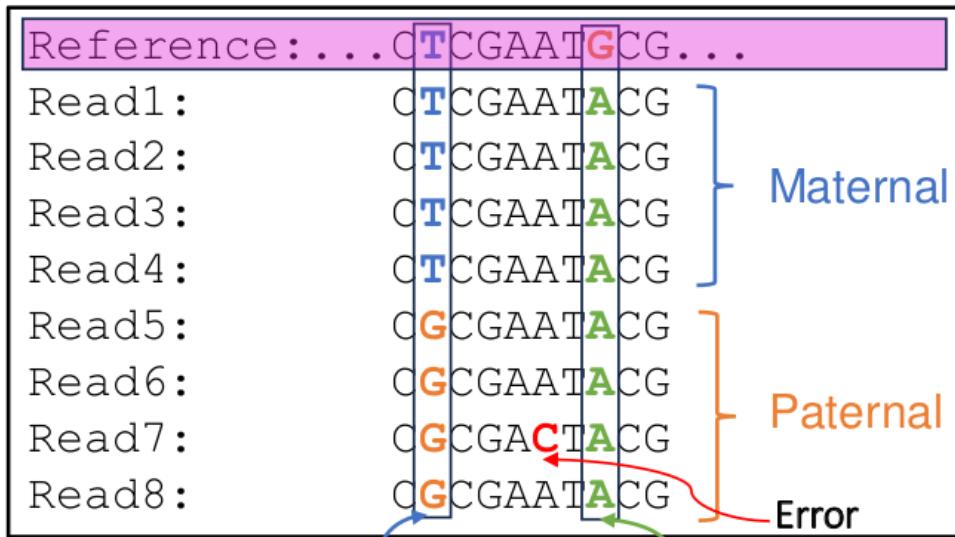
- Sequencing region: specific regions (could be customized)
- Sequencing Depth : >500X
- Identify all kinds of variants including SNPs, INDELs in specific regions
- Most Cost effective

Clinical applications of NGS



Clinical applications of NGS

4.2 Mapping reads to reference



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

```
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##FORMAT=<ID=GQ,Number=1,Type=Integer,Description="Genotype Quality">
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##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

Analyzing and Interpreting genomic records

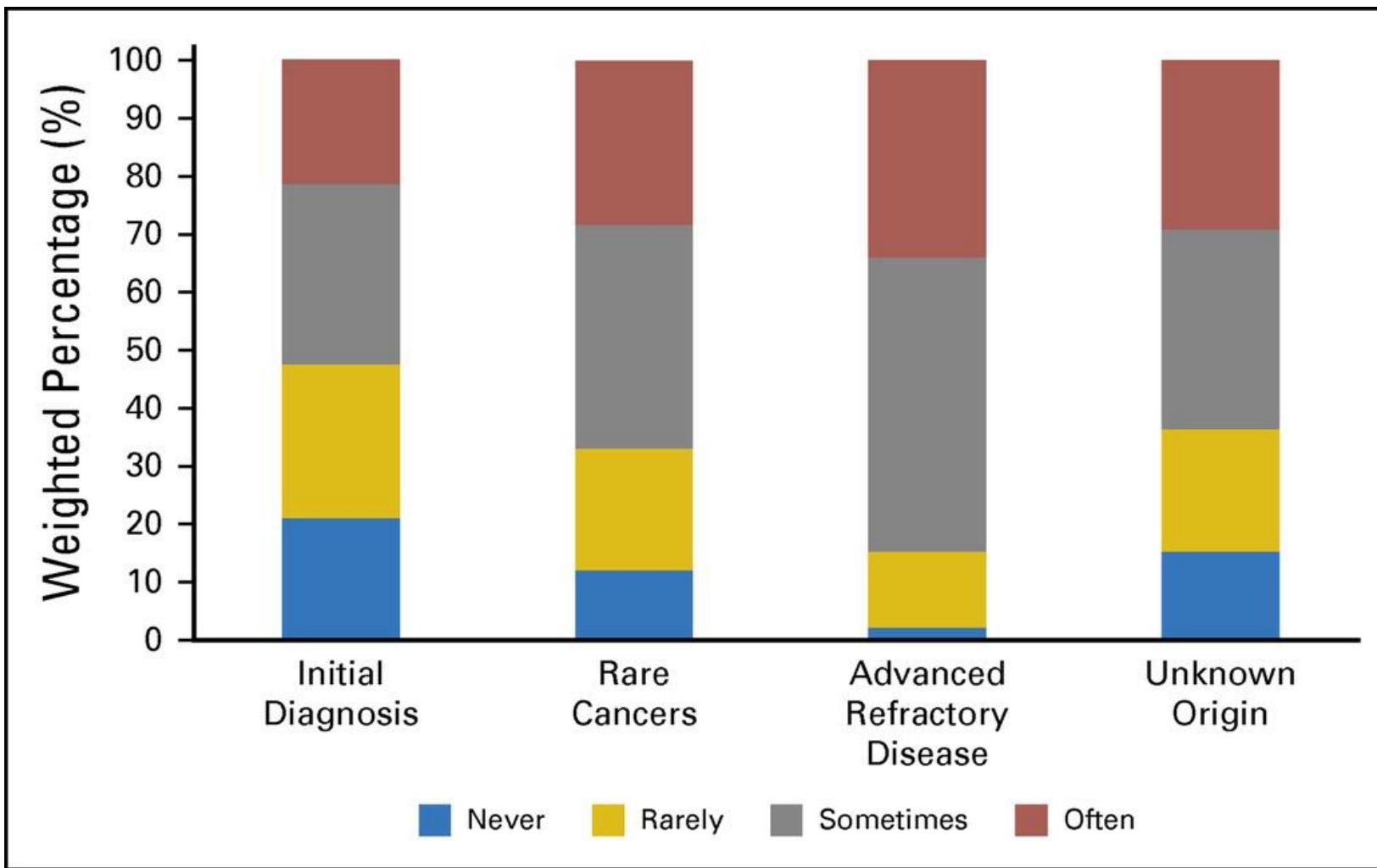
Genome board

- A team of doctors, biologists, bioinformaticians and geneticists
- Analyze and interpret patient records to detect the rare conditions they suffer from and improve their treatment

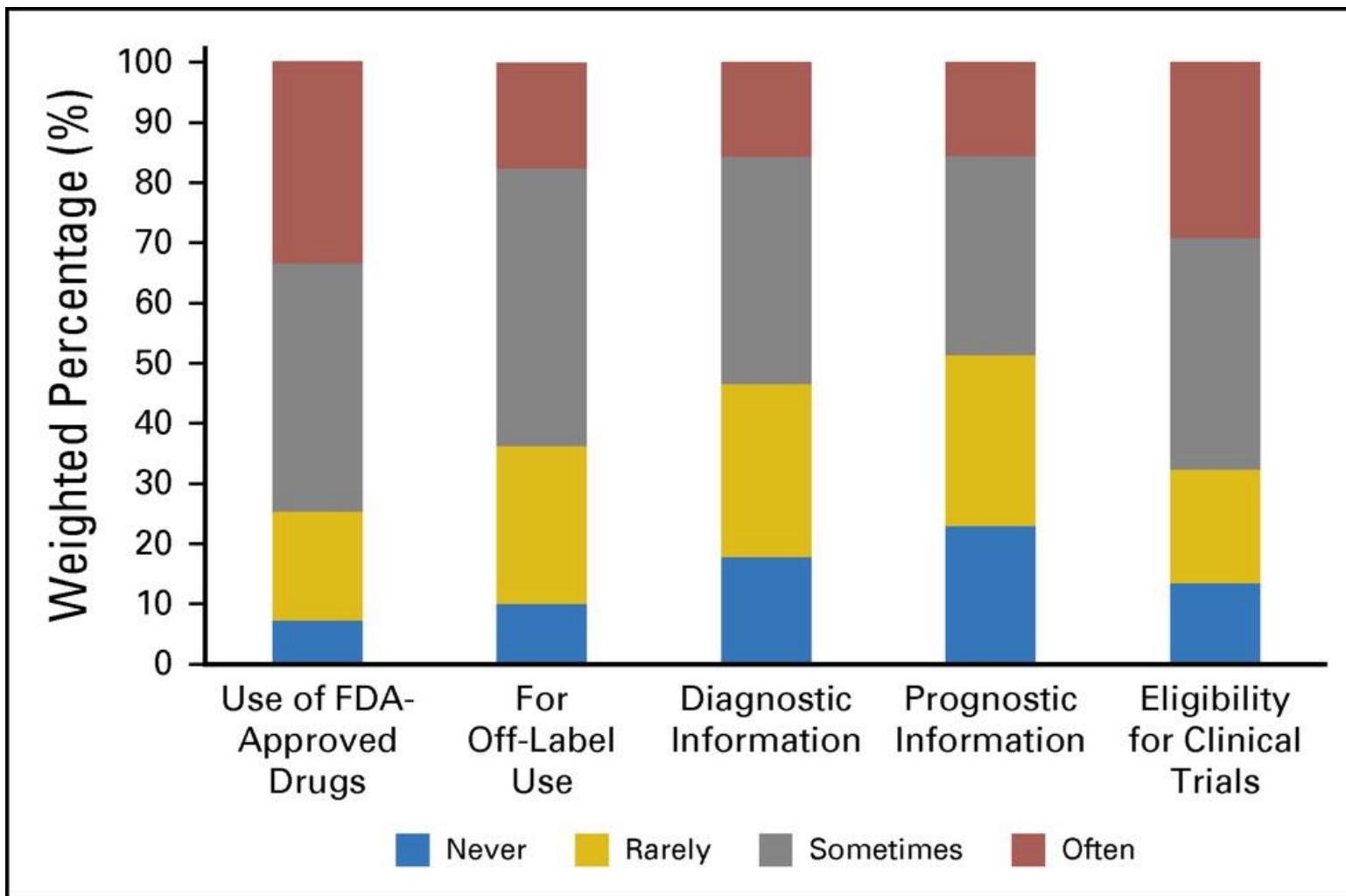


<https://ggba.swiss/en/the-first-medical-genomics-center-opens-in-geneva/>

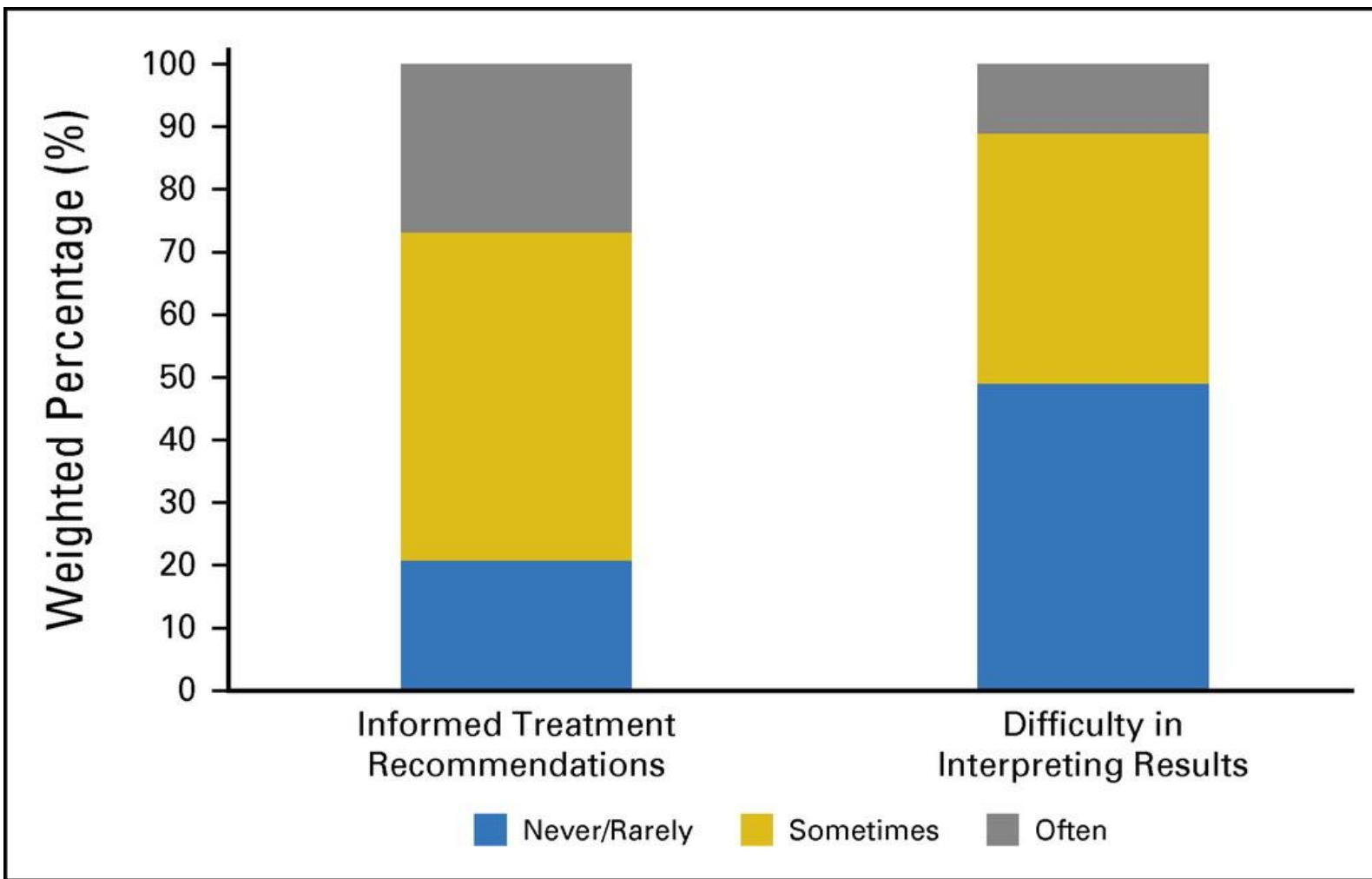
NGS is used in clinical cancer care



NGS is used in clinical cancer care



NGS is used in clinical cancer care



Whole Exome Sequencing (WES): epilepsy and autism

DEE001

Gene	Amino acid change	cDNA	Variant type	Allele frequency	Transcript	Variant effect	ClinVar significance
CPT2	p.Arg631Cys	c.1891C>T	SNP	0.5	ENST00000371486.4	Missense variant	Pathogenic
CDKL5	p.Gln881Ter	c.2641C>T	SNP	0.5	ENST00000623535.2	Stop gained (Nonsense)	Pathogenic
GALC		c.1162-4del	DEL (1bp)	1	ENST00000261304.7	Intron variant	Conflicting interpretations of pathogenicity
TUBB2B	p.Ala248Val	c.743C>T	SNP	0.5	ENST00000259818.8	Missense variant	Conflicting interpretations of pathogenicity



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University of Medicine and Pharmacy, HCM city
Neuro-pediatrician, Children hospital 2- HCM city



Nguyen Le Duc Minh, MD
CPH, HCM city, Vietnam

DEE001 – CDKL5 (Xp22.13)

rs1057519541

Current Build 156
Released September 21, 2022

Organism	Homo sapiens	Clinical Significance	Reported in ClinVar
Position	chrX:18628515 (GRCh38.p14)	Gene : Consequence	CDKL5 : Stop Gained
Alleles	C>T	Publications	1 citation
Variation Type	SNV Single Nucleotide Variation	Genomic View	See rs on genome
Frequency	None		

Variant Details Clinical Significance HGVS Submissions History Publications Flanks

Allele: T (allele ID: 362353)

ClinVar Accession ▲ Disease Names ◇ Clinical Significance

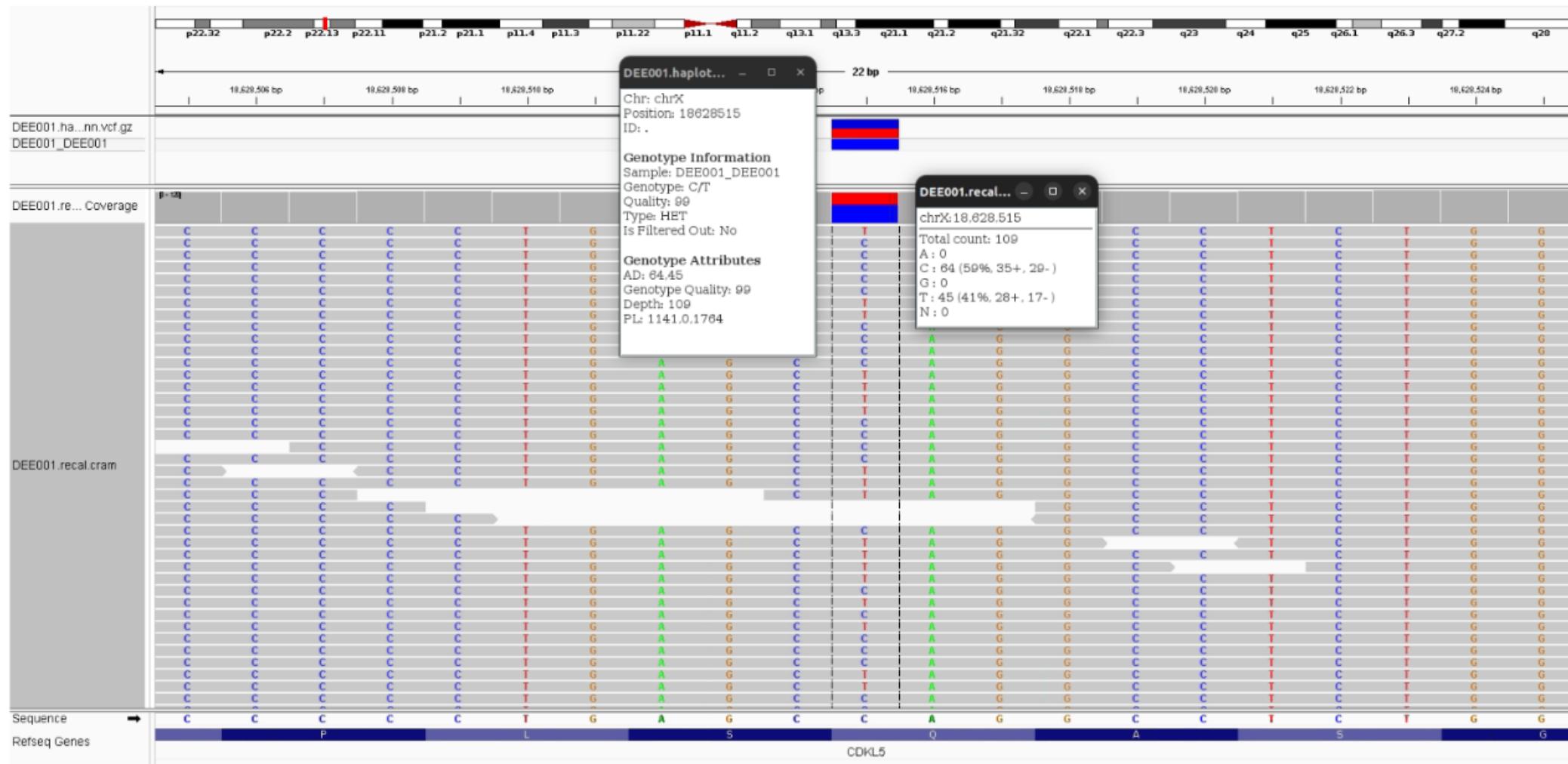
RCV000416943.1 Focal epilepsy Pathogenic

https://www.ncbi.nlm.nih.gov/snp/rs1057519541#clinical_significance

Diagnosis, confirmation and interpretation

Whole Exome Sequencing (WES): epilepsy and autism

IGV - DEE001 – CDKL5

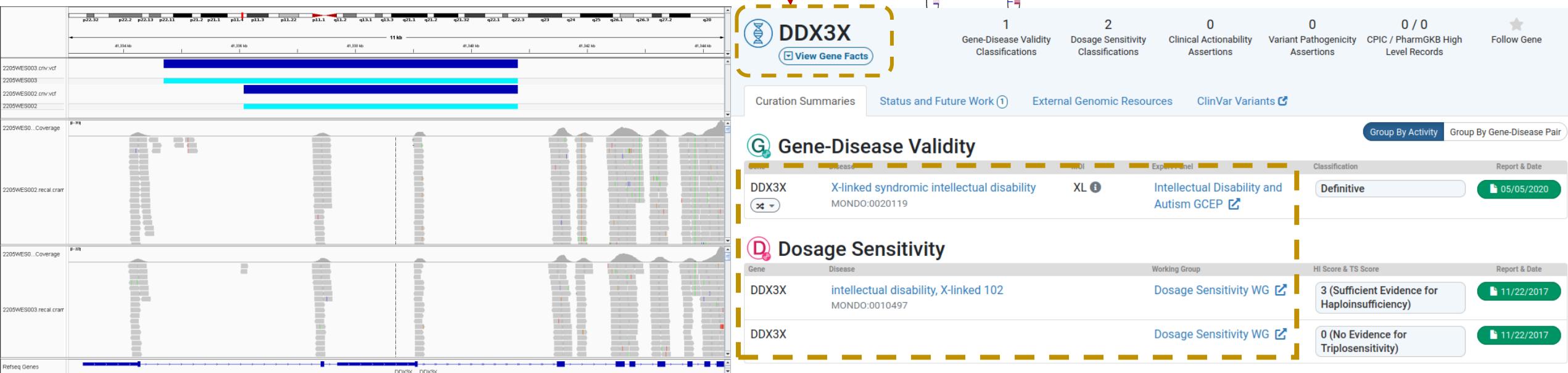


Whole exome sequencing (WES): Microcephaly–related disease

Sample	Variant ID	Chromosome	Gene	CNV type	Clinical Significance
WES002	chrX_41336082_41340814	Xp11.4	DDX3X	DEL	Likely pathogenic
WES003	chrX_41334705_41340814	Xp11.4	DDX3X	DEL	Likely pathogenic



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Cancer Hotspot Gene Panel covered 53 genes

Gene	Amino Acid Change	Coding	Variant Type	Allele Frequency	Transcript	Variant effect	ClinVar Significance
CTNNB1	p.Ser33Tyr	c.98C>A	SNP	0.5	ENST00000349496	MISSENSE	Pathogenic/ Likely_pathogenic
PIK3CA	p.Gly914Arg	c.2740G>A	SNP	0.5	ENST00000263967	MISSENSE	Pathogenic
KRAS	p.Gly12Asp	c.35G>A	SNP	0.5	ENST00000256078	MISSENSE	Pathogenic
BRCA2	p.Ile2675AspfsTer6	c.8021dup	INS	0.5	ENST00000544455	FRAMESHIFT	Pathogenic



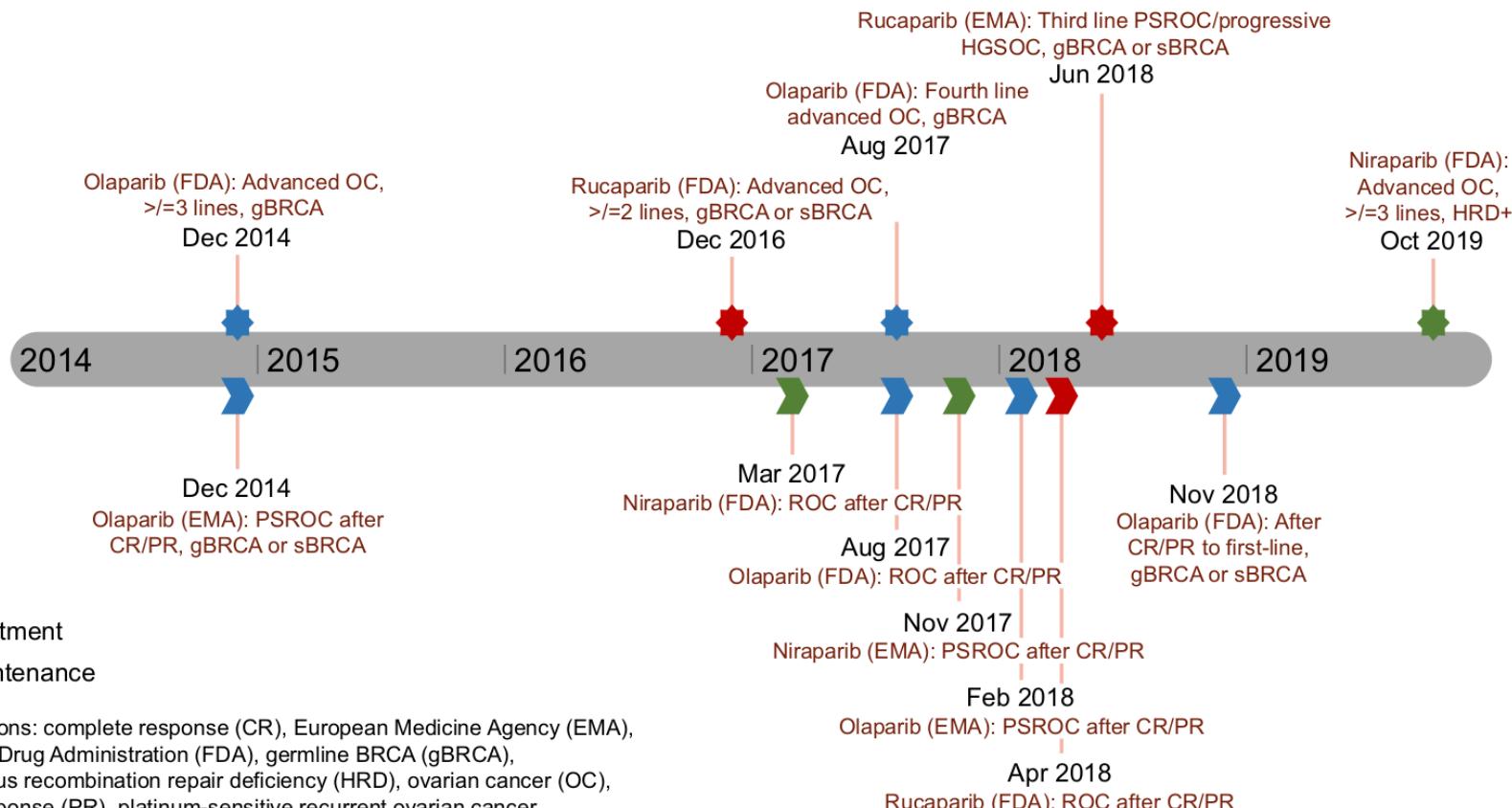
Cancer Screening



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Gene Panel: BRCA1 and BRCA2

Olaparib (AstraZeneca) is a medication for the maintenance treatment of **BRCA-mutated** advanced breast, ovarian, prostate, and pancreatic cancer in adults.



Abbreviations: complete response (CR), European Medicine Agency (EMA), Food and Drug Administration (FDA), germline BRCA (gBRCA), homologous recombination repair deficiency (HRD), ovarian cancer (OC), partial response (PR), platinum-sensitive recurrent ovarian cancer (PSROC), somatic BRCA (sBRCA), recurrent ovarian cancer (ROC).

DOI:10.1007/s11912-020-0873-4

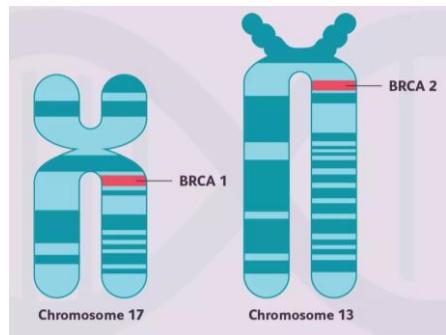
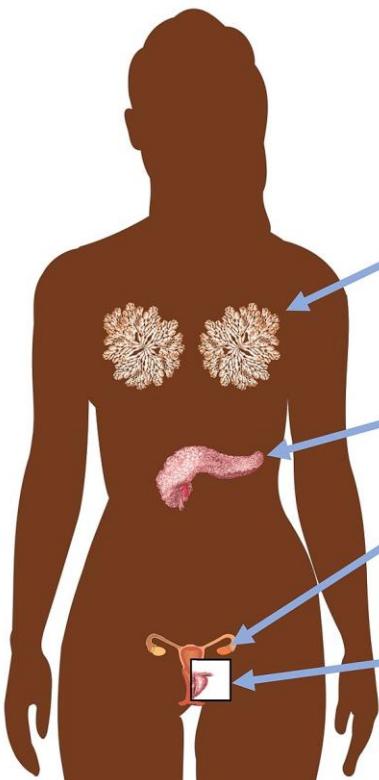


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Thinh Nguyen Huy, MD
HCM City Oncology Hospital, Vietnam

Gene Panel: BRCA1 and BRCA2



	BRCA1	BRCA2
Breast cancer:	50% to 65% Males: 1.2%	40% to 55% Males: Up to 9%
Pancreas cancer:	1-3%	2-7%
Ovarian cancer:	40% to 65%	15% to 25%
Prostate cancer:	9%	15%



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03 Nơ Trang Long, P.7, Q. Bình Thạnh, TP. HCM

BRCA.01

KẾT QUẢ XÉT NGHIỆM BRCA1/2

Họ và tên : TRẦN THỊ X. Tuổi : 1956 Giới tính : NỮ

Số hồ sơ:

KHOA:

Bệnh phẩm : Mô vúi nến

BS điều trị:

Số block: XXXX

Yêu cầu: Xét nghiệm giải trình tự gen trên hệ thống MiSeq [02 gen BRCA1 và BRCA2]

Ngày nhận chỉ định: 20/10/2022 Ngày thực hiện: 25/10/2022

Chẩn đoán lâm sàng: Ung thư buồng trứng đích trong grade cao/ Ung thư vú trái

Chất lượng mẫu: MẪU ĐẶT (kích thước 17mm x 16mm, thành phần buồu 70%)

Phương pháp: Giải trình tự gen bằng phương pháp NGS cho 02 gen BRCA1 và BRCA2

- Hệ thống xét nghiệm Illumina MiSeqDx (CE/US-IVD)
- Bộ xét nghiệm: NGeneBio BRCAaccuTest™Plus (CE-IVD)
- Phần mềm hỗ trợ phân tích kết quả: NGeneBio NGeneAnalySys™ (CE-IVD)

Kết quả: PHÁT HIỆN 1 BIẾN THẾ MẤT ĐOẠN NUCLEOTIDE NHỎ (DEL) GÂY BỆNH TRÊN GEN BRCA1

MÔ TẢ KẾT QUẢ			
Gen	Biến thể gây bệnh/có khả năng gây bệnh	Tỷ lệ	Phân loại
BRCA1	c.5335del (p.Gln1779AsnfsTer14)	82.03%	Gây bệnh (Pathogenic)
BRCA2	Không phát hiện	Không	Không

Ngày 01 tháng 11 năm 2022

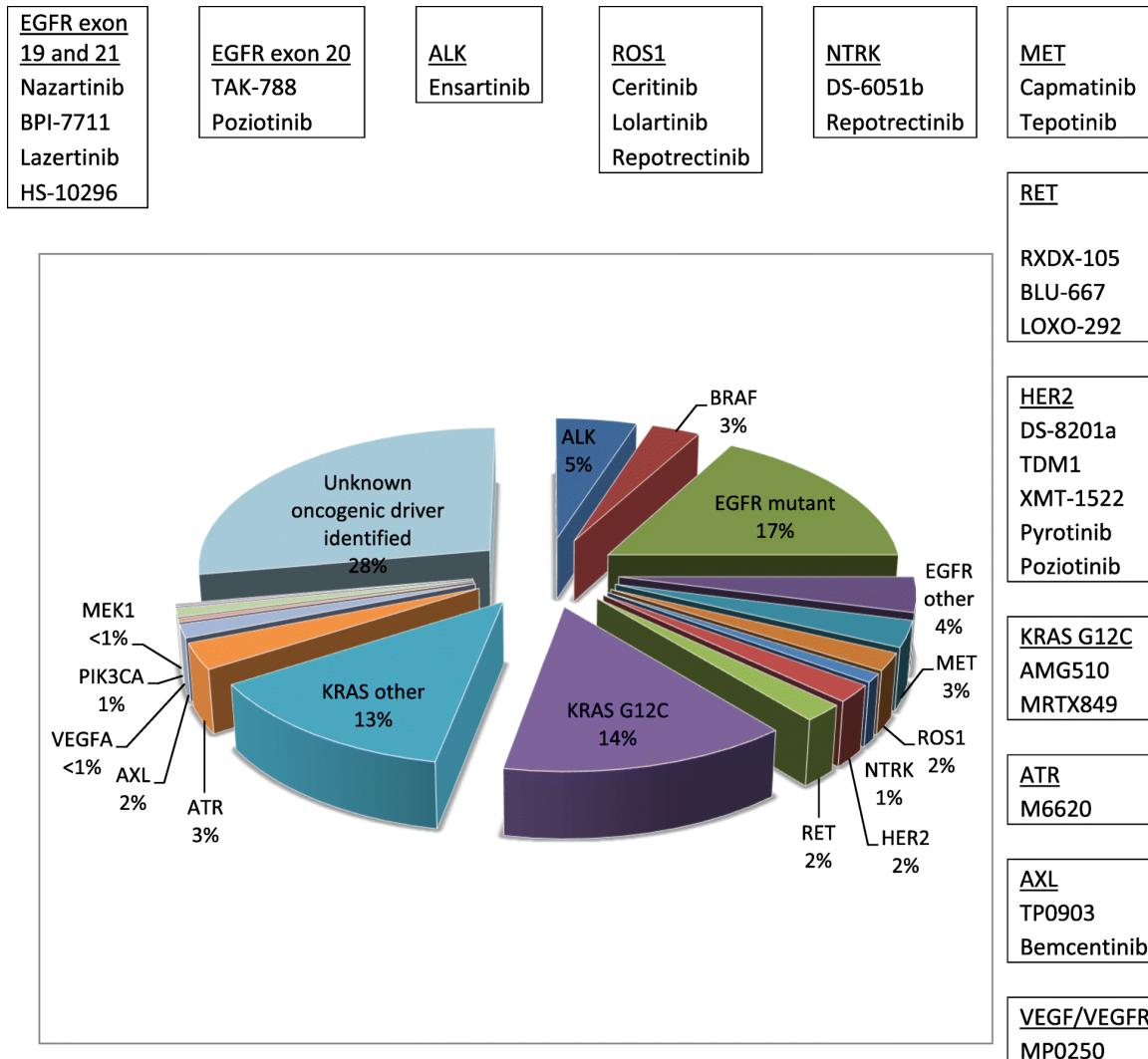
Phụ trách kỹ thuật

Bác sĩ đọc kết quả

CN. NGUYỄN THỊ B

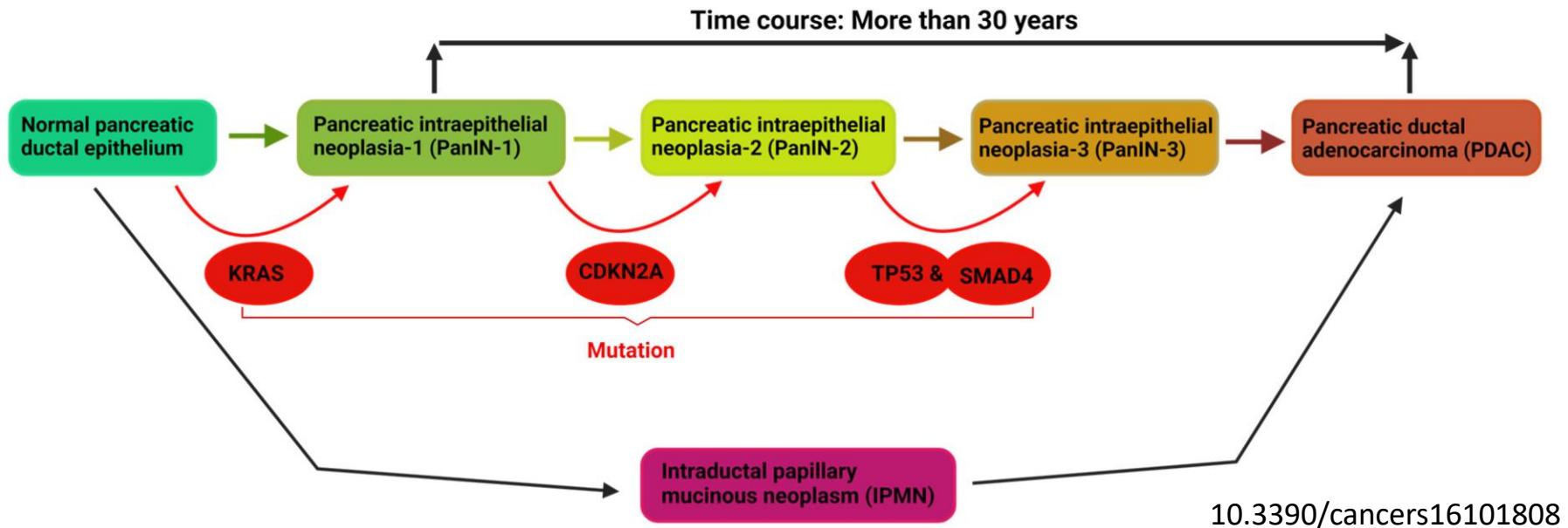
BS. NGUYỄN VĂN A

Gene Panel: EGFR, ALK, ROS1 NTRK, MET, RET, HER2, KRAS, ATR, AXL and VEGFR



Gene Panel: KRAS

- Pancreatic cancer progression



- FDA approval of sotorasib for targeting the KRAS-G12C mutation in PDAC patients
- KRAS-G12C mutation is only found in less than 2% of PDAC cases

Spatial Transcriptomics Analysis of Non-small Cell Lung Cancer: **is immunotherapy effective for this lung cancer patient?**



Tissue section



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CPH, HCM City, Vietnam

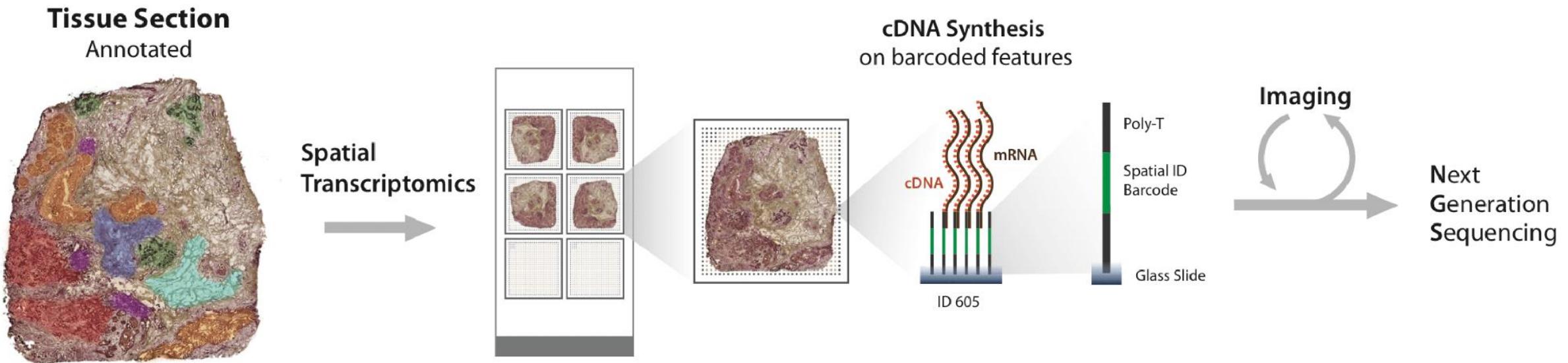


Thi-My-Hanh Luong,
MD, PhD
Karolinska Institutet,
Sweden



Kim-Anh Ly, Bsc
Gene Smart,
Vietnam

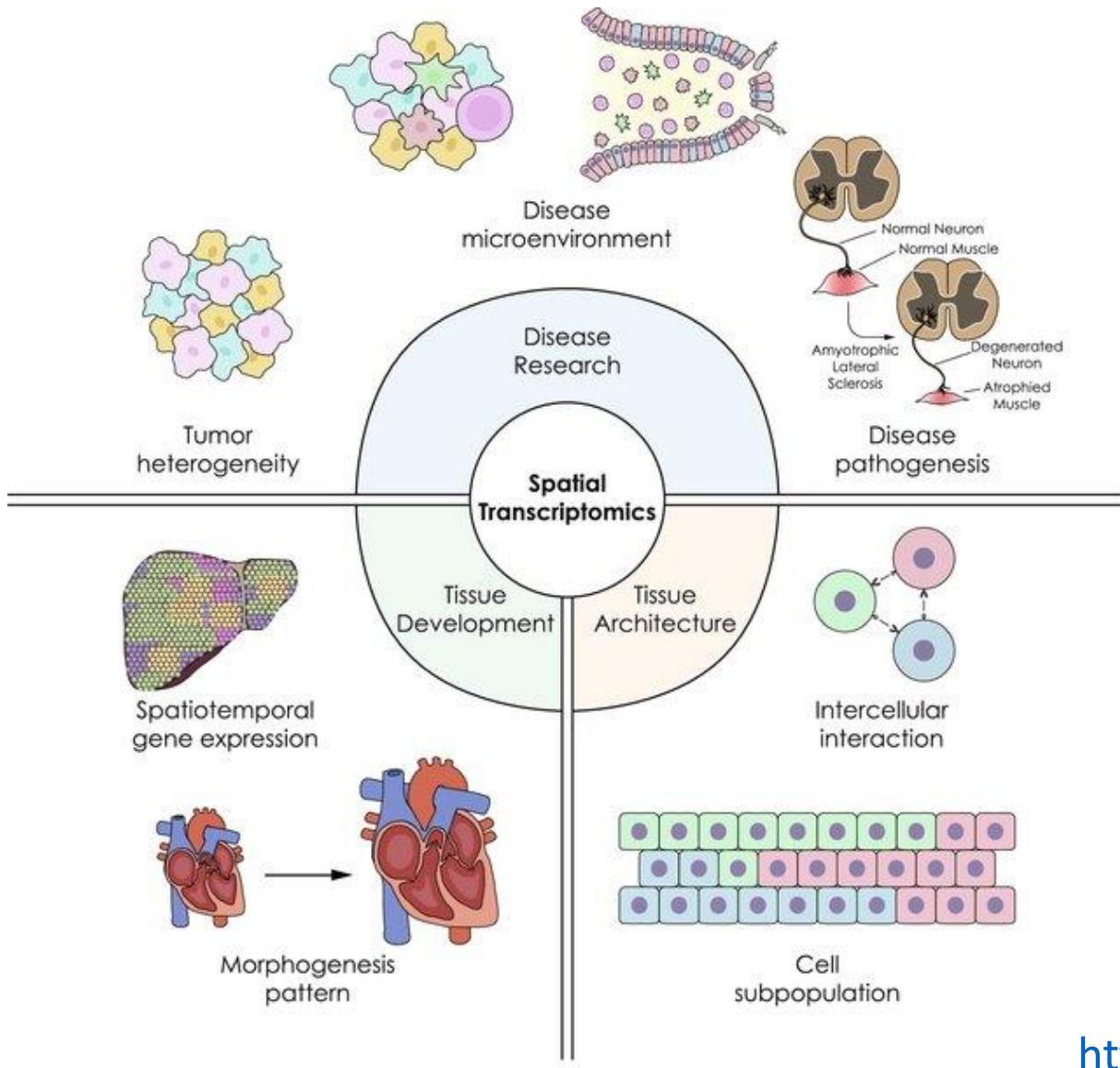
Spatial Transcriptomics



Visium Spatial Transcriptomics Technology

<https://www.scilifelab.se/units/spatial-transcriptomics/>

Spatial Transcriptomics (Visium)



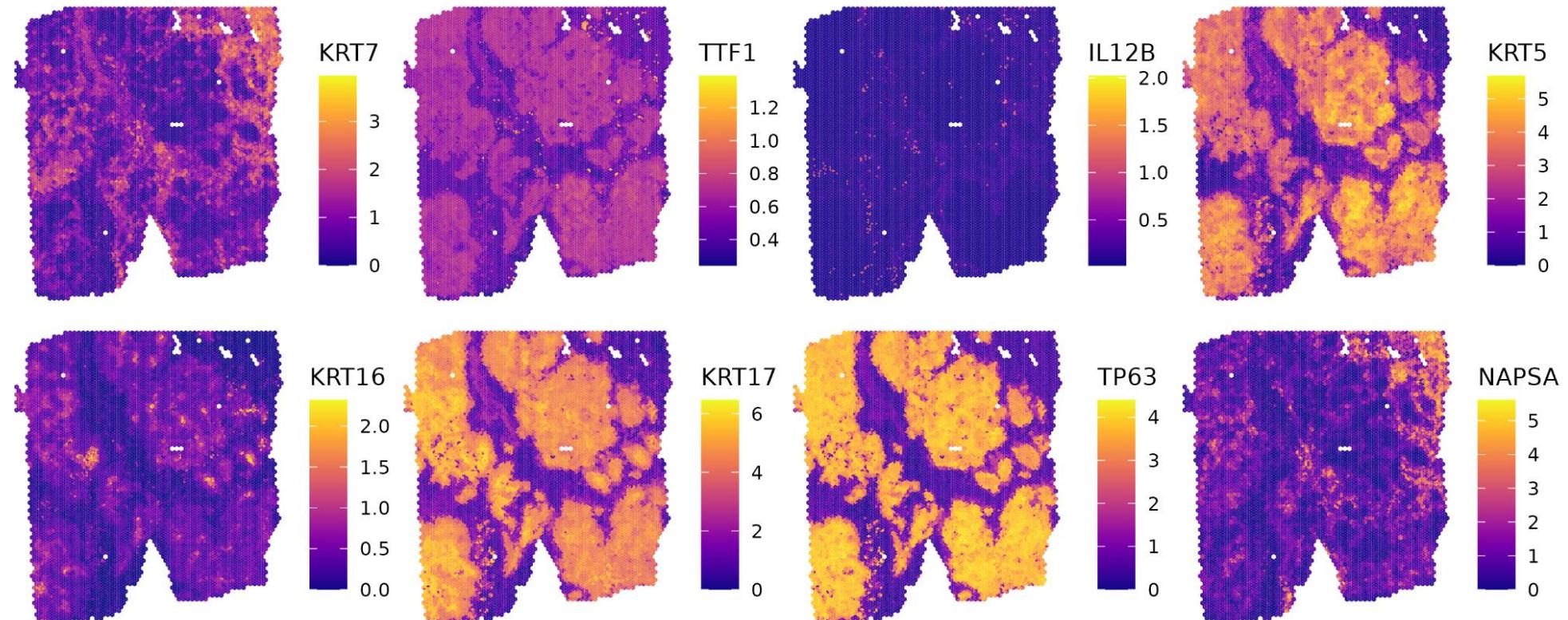
Applications:

- Tumor heterogeneity
- Disease microenvironment
- Disease pathogenesis
- Morphogenesis patterns
- Intercellular interaction
- Identify drug targets
- Immune profiling

<http://dx.doi.org/10.1186/s12967-023-04150-2>

Visualization of marker genes in spatial location

Lung cancer marker genes

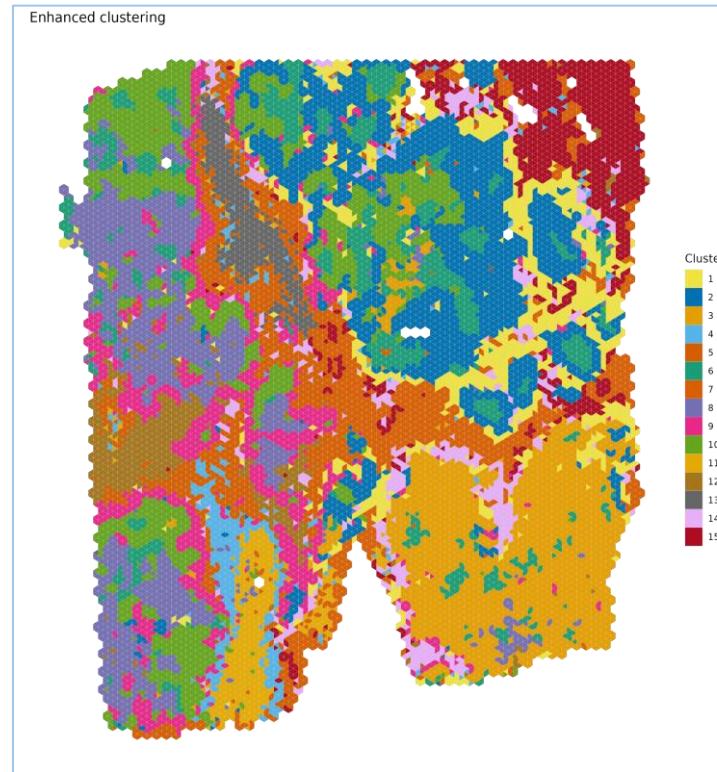


→ Identify tumor subtypes and tumor regions

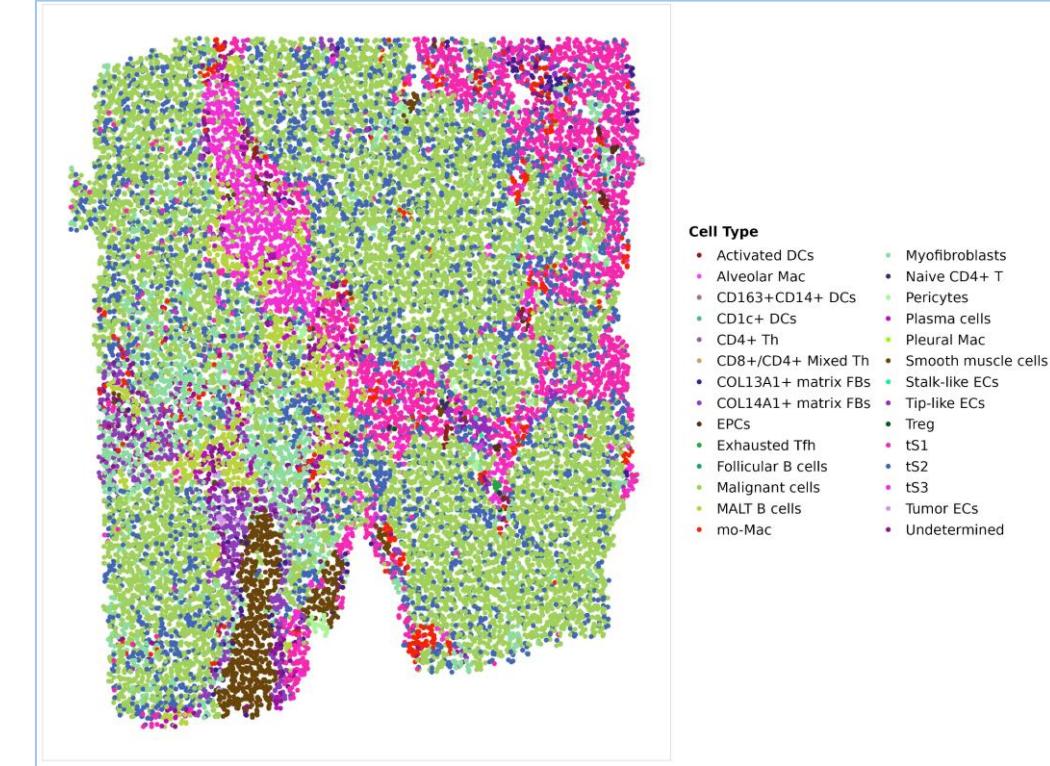
Spatial expression patterns and cell-type identification



Tissue section



Spatial clusters



Cell-type in single-cell resolution

==> Explore tumor microenvironments

Precision microbiome testing: STI, HPV and AMR

Women's Health Test

Page 1 of 4

Patient Name:	nan	Provider:	Mony Sary, Yap Chew, Wendy Ullmer	Patient ID:	VS8
Gender:	nan	Provider NPI:	nan	Specimen ID:	KH20.45474
DOB:	nan	Order Date:	nan	Specimen Type:	nan
Age:	nan	Health Status Reported:	nan	Collection Date:	nan

Sexually Transmitted Infections

Name	Associated Condition	Result
<i>Neisseria gonorrhoeae</i>	Gonorrhea, urethritis, pelvic inflammatory disease, gonococcemia, gonococcal ophthalmia neonatorum	Detected
<i>Chlamydia trachomatis</i>	Chlamydia, cervicitis, urethritis, pelvic inflammatory disease	Not Detected
<i>Mycoplasma genitalium</i>	Urethritis, cervicitis, pelvic inflammatory disease	Not Detected
<i>Treponema pallidum</i>	Syphilis	Not Detected
<i>Haemophilus ducreyi</i>	Chancroid	Not Detected
<i>Trichomonas vaginalis</i>	Trichomoniasis	Not Detected
<i>Human papillomavirus</i>	Cervical and anogenital cancers, genital warts	Detected
<i>Herpes simplex virus</i>	Genital herpes, oral herpes	Not Detected

Viruses Detected

Name	Associated Condition
<i>Human papillomavirus 62 (HPV 62)</i>	Unknown risk for cervical cancer

Note: Human papillomavirus (HPV) 16, 18, 31, 33, 35, 39, 45, 51, 56, 58, 59, and 68 are considered high-risk or probable high-risk due to their association with cervical cancer. HPV 6, 11, 42, 43, and 44 are considered low-risk for cervical cancer but may cause genital warts. Other HPV genotypes found in the sample may have intermediate or unknown risk for cervical cancer.

Antimicrobial Resistance Genes Detected

AMR Gene Name	Function	Drug Class
<i>Neisseria.gonorrhoeae.folP</i>	Dihydropteroate synthase (mutated)	Sulfonamide

Women's Health Test

Page 1 of 4

Patient Name:	nan	Provider:	Yap Chew	Patient ID:	nan
Gender:	nan	Provider NPI:	nan	Specimen ID:	202122865
DOB:	nan	Order Date:	nan	Specimen Type:	nan
Age:	nan	Health Status Reported:	nan	Collection Date:	nan

Sexually Transmitted Infections

Name	Associated Condition	Result
<i>Neisseria gonorrhoeae</i>	Gonorrhea, urethritis, pelvic inflammatory disease, gonococcemia, gonococcal ophthalmia neonatorum	Not Detected
<i>Chlamydia trachomatis</i>	Chlamydia, cervicitis, urethritis, pelvic inflammatory disease	Not Detected
<i>Mycoplasma genitalium</i>	Urethritis, cervicitis, pelvic inflammatory disease	Not Detected
<i>Treponema pallidum</i>	Syphilis	Not Detected
<i>Haemophilus ducreyi</i>	Chancroid	Not Detected
<i>Trichomonas vaginalis</i>	Trichomoniasis	Not Detected
<i>Human papillomavirus</i>	Cervical and anogenital cancers, genital warts	Detected
<i>Herpes simplex virus</i>	Genital herpes, oral herpes	Not Detected

Viruses Detected

Name	Associated Condition
<i>Human papillomavirus 52 (HPV 52)</i>	High-risk for cervical cancer
<i>Human papillomavirus 68 (HPV 68)</i>	High-risk for cervical cancer

Note: Human papillomavirus (HPV) 16, 18, 31, 33, 35, 39, 45, 51, 56, 58, 59, and 68 are considered high-risk or probable high-risk due to their association with cervical cancer. HPV 6, 11, 42, 43, and 44 are considered low-risk for cervical cancer but may cause genital warts. Other HPV genotypes found in the sample may have intermediate or unknown risk for cervical cancer.

Thank you!