



# HUMAN GENOMICS

PALLE DUUN ROHDE

Associate Professor & Research Group Leader

Genomic Medicine

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# THE COURSE TEAM



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# THE AIM OF THIS COURSE

The course provides students with a comprehensive understanding of **human genetics and genomics**, emphasizing their applications in **personalized medicine**.



# LEARNING OBJECTIVES

## KNOWLEDGE

- Explain organisation of the human genome
- Explain different types of genetic variation
- Understand the relationship between genotype and phenotype
- Understand which forces affect alleles in a population
- Explain how genetic variation regulates and affect disease with monogenic and polygenic aetiology
- Describe genetic and proteomic biomarkers in diagnostics, biomarker discovery and validation in a personalised medicine context

## SKILLS

- Apply advanced molecular methods in genetics
- Evaluate a choice of method or technology to detect and analyse genetic variation
- Choose appropriate databases, algorithms, statistics and parameters in a bioinformatics analysis
- Use bioinformatical and analytical strategies to solve problems in personalised medicine
- Understand the genetic architecture of monogenic and complex traits

## COMPETENCES

- Assess and evaluate the suitable technologies and methods for detection of genetic variants in polygenic vs monogenic diseases
- Perform simple genetic analyses
- Evaluation of scientific articles at the highest international level

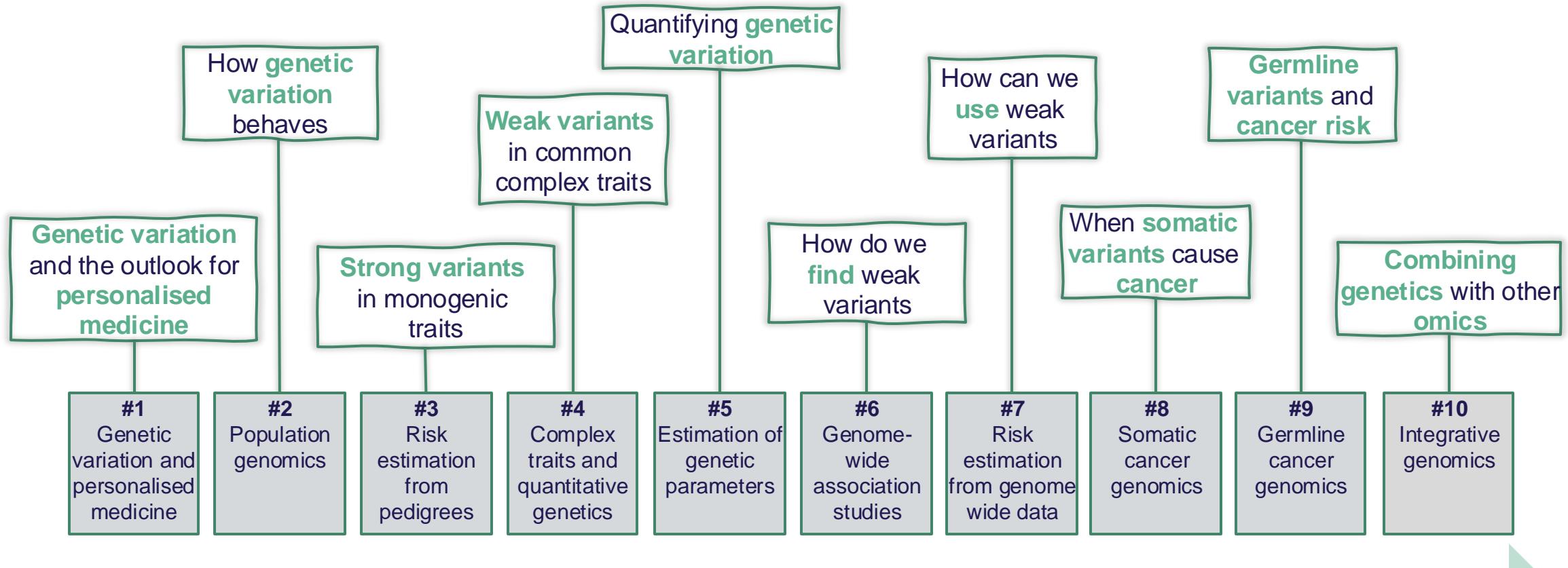
# COURSE PLAN

<b>08:15 – 08:30</b>	Recap
<b>08:30 – 08:50</b>	Lecture 1 [ <i>Introduction to population genetics</i> ]
<b>08:50 – 09:30</b>	Break + Exercises 1 [1-4]
<b>09:30 – 09:50</b>	Lecture 2 [ <i>Hardy-Weinberg</i> ]
<b>09:50 – 10:30</b>	Break + Exercises 2 [5-7]
<b>10:30 – 10:50</b>	Lecture 3 [ <i>Modulation of genetic variation</i> ]
<b>10:50 – 11:45</b>	Break + Exercises 3 [8 + computer exercise]
<b>11:45 – 12:00</b>	Padlet evaluation

#1 Genetic variation and personalised medicine	#2 Population genomics	#3 Risk estimation from pedigrees	#4 Complex traits and quantitative genetics	#5 Estimation of genetic parameters	#6 Genome-wide association studies	#7 Risk estimation from genome wide data	#8 Somatic cancer genomics	#9 Germline cancer genomics	#10 Integrative genomics
5/2-25 [PDR]	7/2-25 [PDR]	12/2-25 [PDR]	27/2-25 [PDR]	6/3-25 [PDR]	17/3-25 [PDR]	24/3-25 [PDR]	31/3-25 [AKN PDR]	7/4-25 [AKN PDR]	16/4-25 [PLM PDR]



# WHY THIS COURSE PLAN



# COURSE MATERIAL IN MOODLE

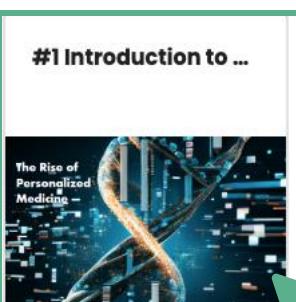
Welcome to the new module, **Human Genomics**. This module aims to provide you with insight into what shapes the human genome, and how genetics and genomics can be applied to advance our understanding of human traits, health, and disease, including their application in personalized medicine.

Below is a brief overview of the module's content:

#1 Genetic variation and personalised medicine	#2 Population genomics	#3 Risk estimation from pedigrees	#4 Complex traits and quantitative genetics	#5 Estimation of genetic parameters	#6 Genome-wide association studies	#7 Risk estimation from genome wide data	#8 Somatic cancer genomics	#9 Germline cancer genomics	#10 Integrative genomics
5/2-25 [PDR]	7/2-25 [PDR]	12/2-25 [PDR]	27/2-25 [PDR]	6/3-25 [PDR]	17/3-25 [PDR]	24/3-25 [PDR]	31/3-25 [AKN][PDR]	7/4-25 [AKN][PDR]	16/4-25 [PLM][PDR]

By the end of this module, you will have a comprehensive understanding of human genomics and its transformative role in biology and medicine. This course is designed to equip you with both theoretical knowledge and practical skills, preparing you for further research or careers in genomics-related fields.

All the material needed for this module will be made available for you below with external links for computer exercises. We will use the programming software R for many of the exercises, thus, please download R ([HERE](#)) and R studio([AND HERE](#)) before the first session.



## Preparation prior to the lecture and exercises

1) Review the following figures in **Medical Genetics (sixth edition)** by Jorde, Carey and Bamshad. The book is available via AUB. Use this [link](#), and log in under "Log in via your institution", and search after Aalborg University.

You should review them to an extent where you are able to explain the figures. All figures are part of the curriculum in the "*Advanced Biochemistry and Genetics*" course at 4<sup>th</sup> semester and represents **essential terms** that we will be building upon in this course.

- Figure 2-20, page 21 (mitosis)
- Figure 2-21, page 23 (meiosis, production of gametes)
- Figure 2-22, page 24 (crossover) + Figure 8.2, page 147
- Figure 3-3, page 27 (missense and nonsense mutations)
- Figure 3-4, page 27 (frameshift mutations)
- Figure 3-5, page 28 (splice site mutation)
- Figure 3-6, page 29 (GOF and LOF)
- Figure 3-10, page 32 (compound heterozygous)
- Figure 4-3, page 58 (pedigree symbols)
- Figure 4-6 page 59 (autosomal dominant disorder)
- Figure 4-8 page 60 (autosomal recessive disorder)
- Figure 5-8 page 82 (X-linked recessive trait)
- Figure 6-8 page 103 (meiotic nondisjunction)

2) Read "NNF white paper on precision medicine in cardiometabolic disease" [[download here](#)], and the scientific version published in 2023 in Lancet Diabetes and Endocrinology [[download here](#)].

3) Download ([here](#)) and install R-studio (and R [here](#))

## Learning outcome

After session 1 (reading the curriculum, participating in the lecture and the following exercises) you are expected to:

- Explain organisation of the human genome.
- Explain different types of genetic variation.
- Understand the relationship between genotype and phenotype

## Exercises in class

We will do two rounds of exercises in class. You do not have to look at the exercises beforehand. You will work in groups to solve the exercises. We will go through them afterwards in class. The exercises are available at [this link](#) [*will be available before the session starts*].

## Lecture notes

Lecture notes are available at [this link](#) [*will be available before the session starts*].

# THE EXAM

- ❖ Individual, oral examination.
- ❖ Duration: 20 min including assessment.
- ❖ Preparation time: 20 min with aids.
- ❖ Internal censor. Course coordinator will be responsible for the exam.
- ❖ Grading: Passed/Not passed.
- ❖ Re-examination is oral.



# LETS GET STARTED



# INTRODUCTION TO

ILLUSTRATION BY GREG CLARKE



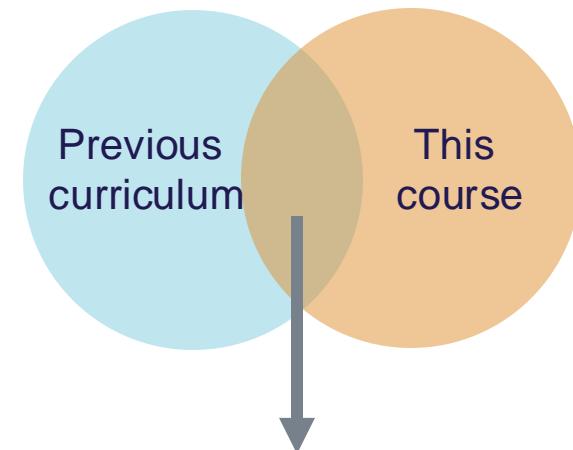
## PERSONALISED MEDICINE

## 4. Semester batchelor curriculum

- The centrale dogma
- Codons and open reading frame
- Genotype, haplotype, locus, allele
- DNA variants
  - coding, intronic, splice-site
  - synonymous and nonsynonymous
  - missense, frameshift, nonsense, stop-gain, stop-loss
  - GOF and LOF
- Monogenic disease, punnett square
- Pedigree symbols and inheritance patterns
- Penetrance-expressivity
- Ploidy and non-disjunction
- PCR-Array-Exome-Genome sequencing

Modul: Videregående  
biokemi og genetik  
MS (M 4.2)

## Where to start ?



Refresh key concepts

# OUTLINE

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# OUTLINE

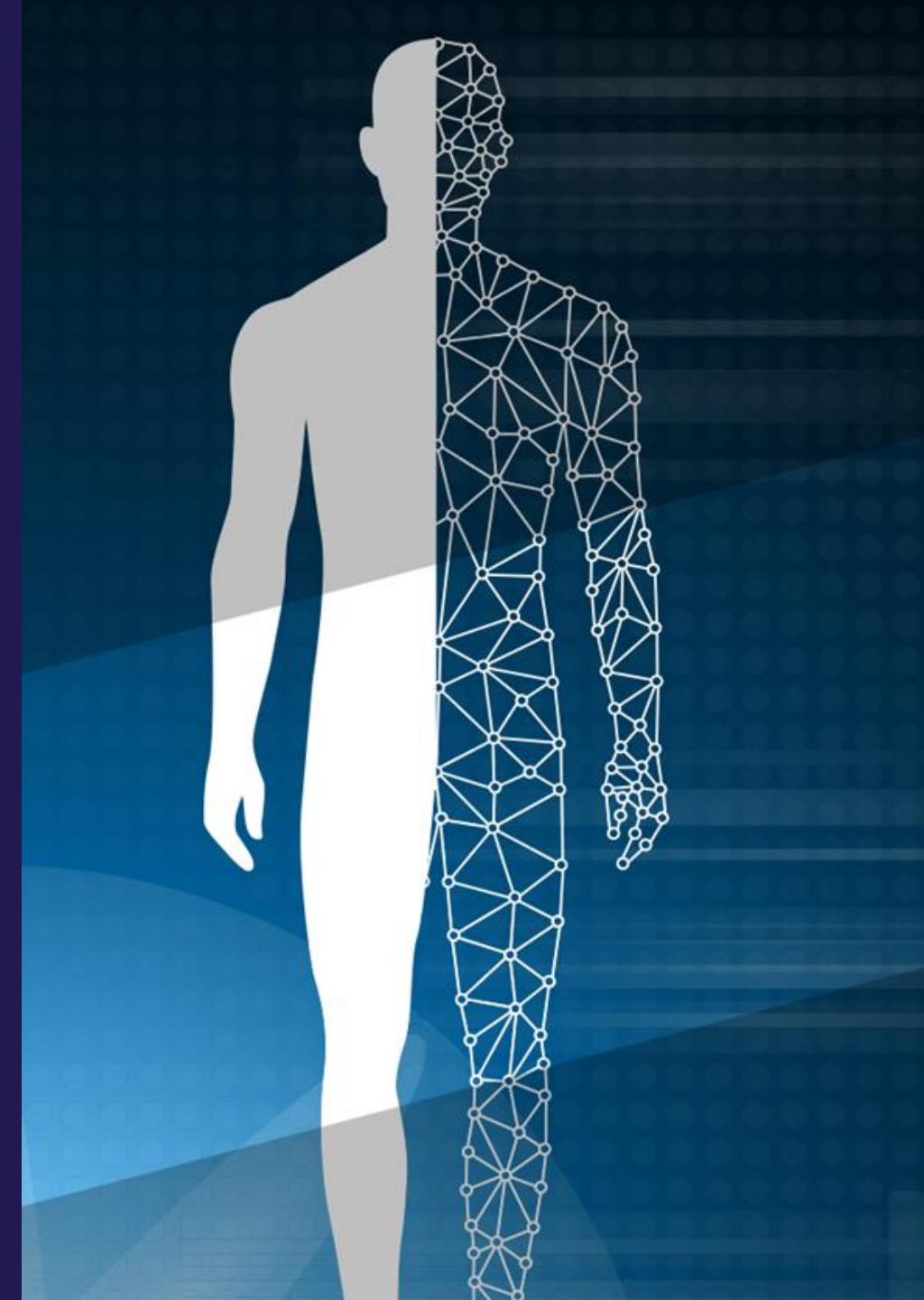
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# PERSONALISED MEDICINE

- What is Personalised Medicine?
- Why do we need it?
- Why so much focus on genetics?
- How is it presented to society?



**What do you think, when someone says *personalised medicine* ?**

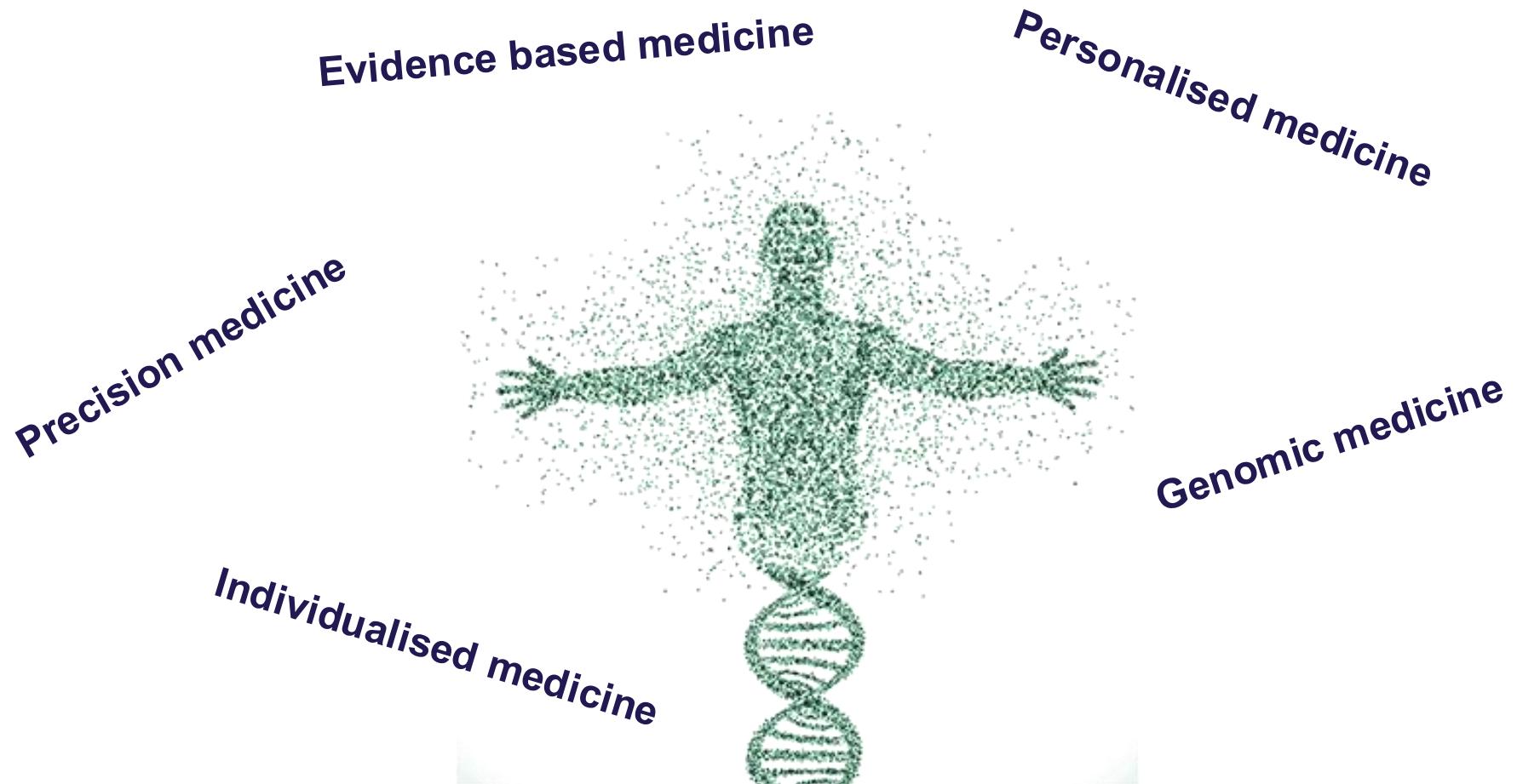
- can you think of an example?





... PM is not new

# WHAT IS PERSONALISED MEDICINE?

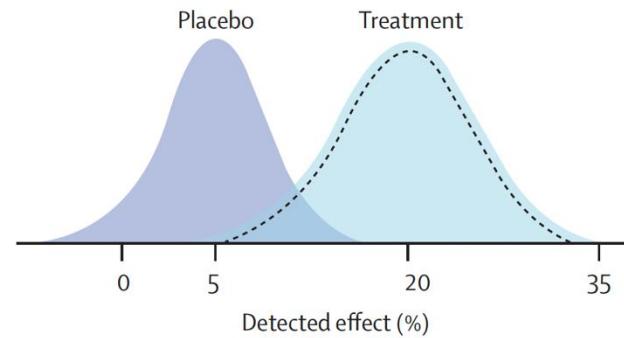


# IMPLEMENTATION OF PRECISION MEDICINE

EPPOS [evidence-based precision personalised objective subjective]

## Evidence-based Medicine

- (1) Contemporary evidence-based medicine  
Estimate average risk or response using epidemiological and clinical trial cohorts



High error

## Personalised Medicine

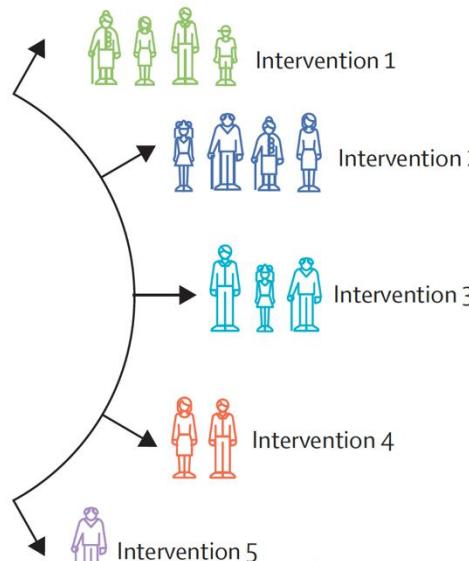
- (3) Personalisation (objective)  
Monitor response to optimise dose, timing, and delivery



Low error

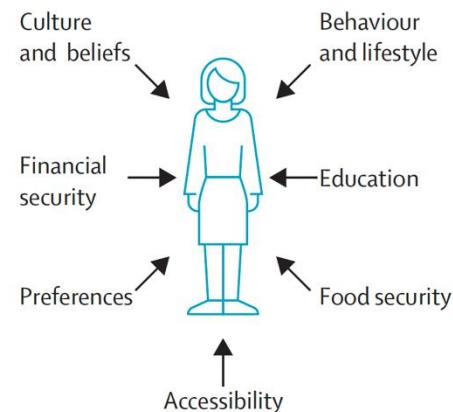
## Precision Medicine

- (2) Probability scoring and stratification  
Maximise response and minimise risk using subclassification



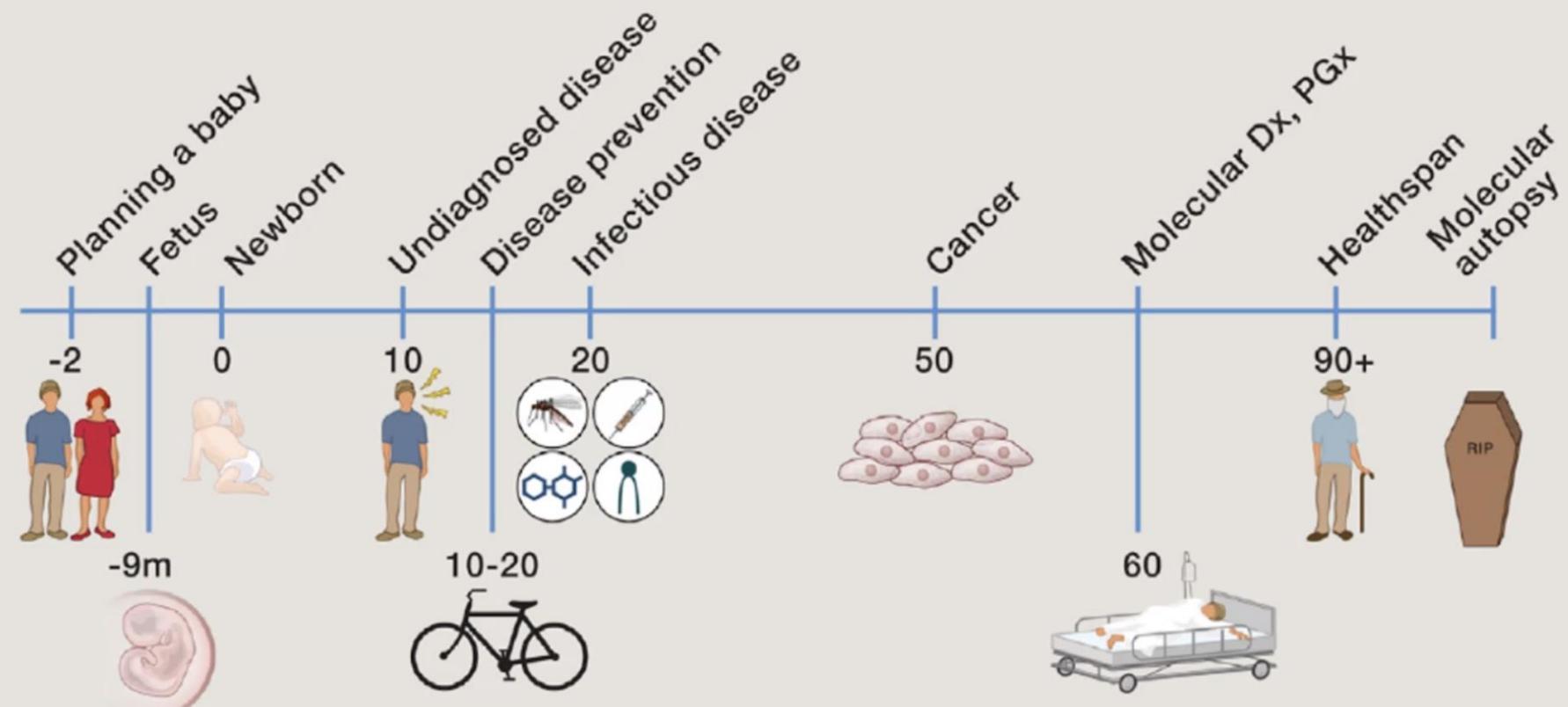
## Individualised Medicine

- (4) Personalisation (subjective)  
Adapt intervention to fit the person's needs, capabilities, and preferences



## Individualized genomic medicine

From prewomb to tomb



# WHAT IS PERSONALISED MEDICINE?

Tailoring medical decisions and interventions to the individual patient based on genomic data.

Move away from a *one-size-fits-all* policy to customising treatments for each patient.



Diagnostics, prognosis, treatment

# WHY PERSONALISED MEDICINE?

Because people are different



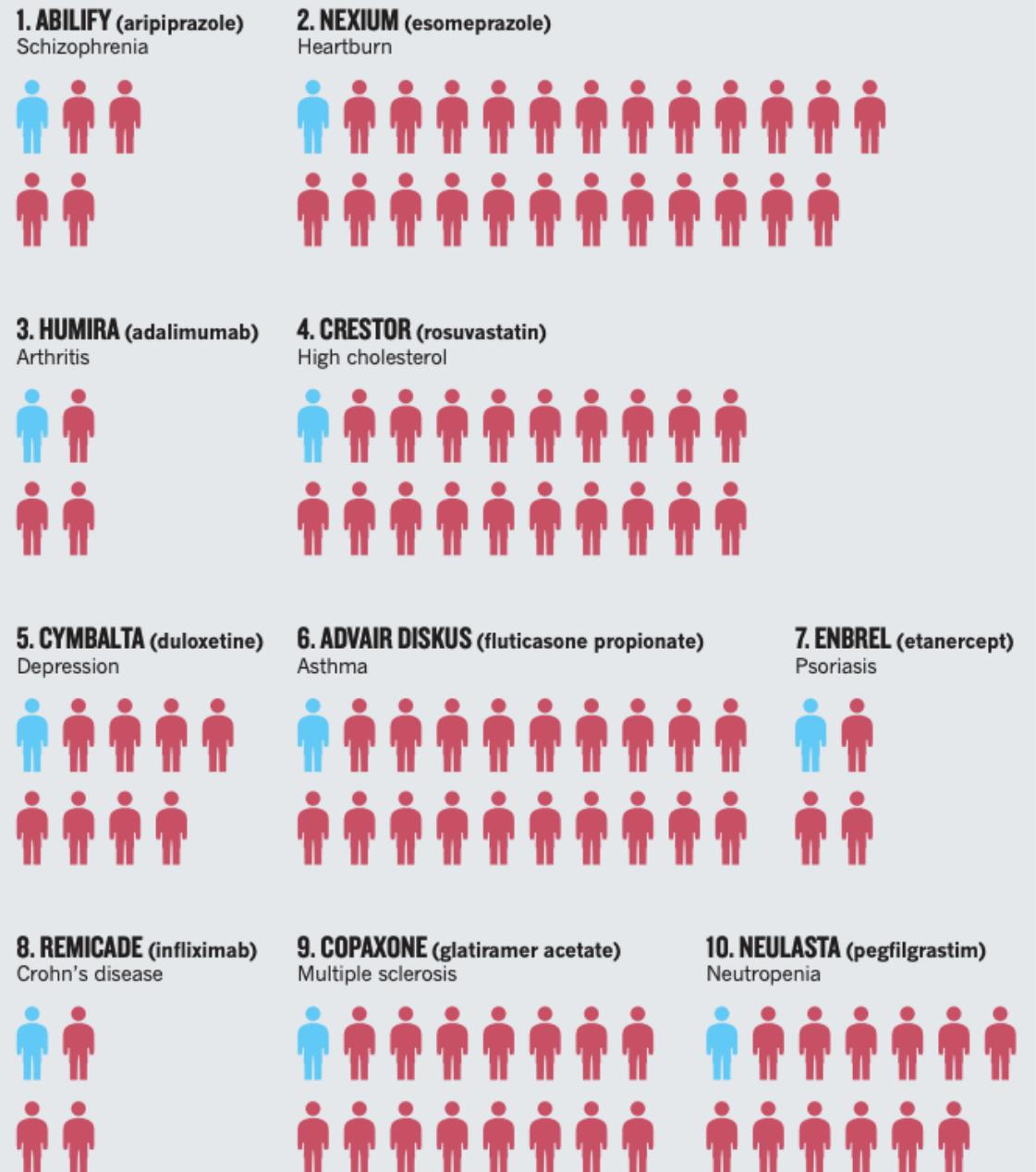
- different disease risk
- respond differently to medication
- different side effects



Diagnostics, prognosis, treatment

# IMPRECISION MEDICINE

The top ten highest-grossing drugs in the US help between 1 in 25 and 1 in 4 of the people who take them.

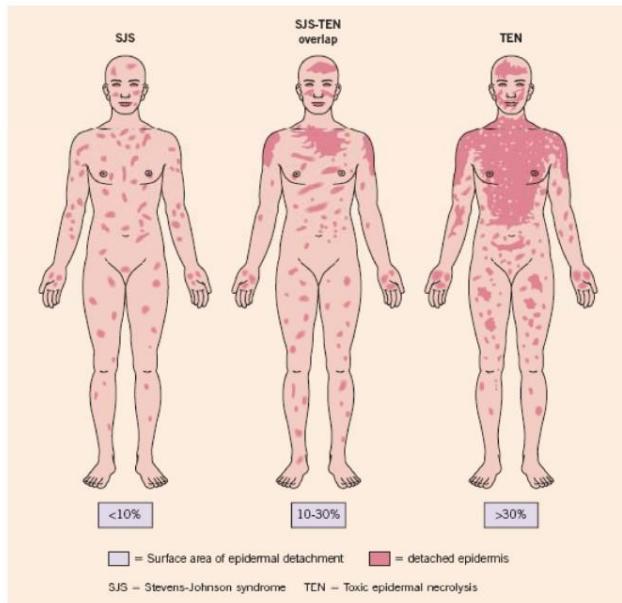


# ADVERSE DRUG REACTIONS

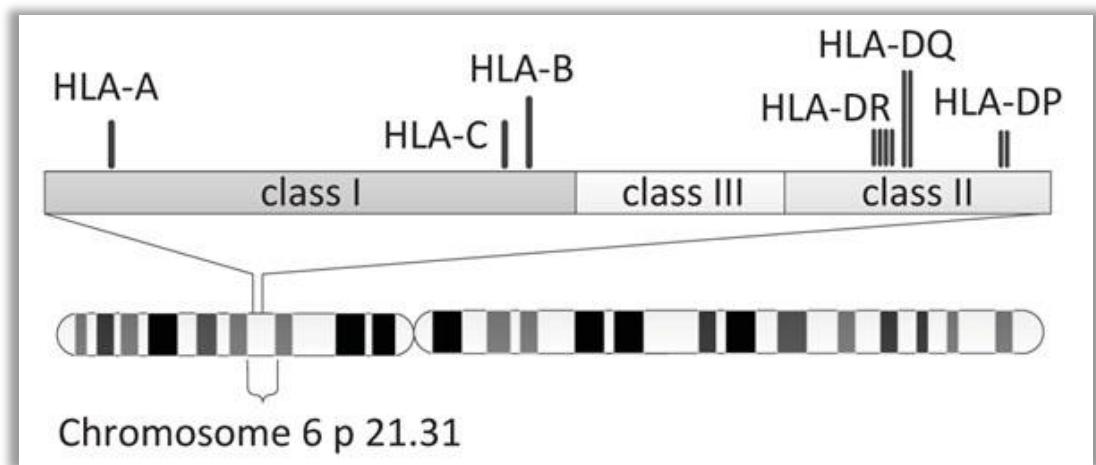


Some medications are directly harmful for certain individuals

Carbamazepine-induced [used to treat Epilepsy and nerve-pain in diabetes] **Stevens-Johnson syndrome** in patients with specific genotypes in the MHC region.



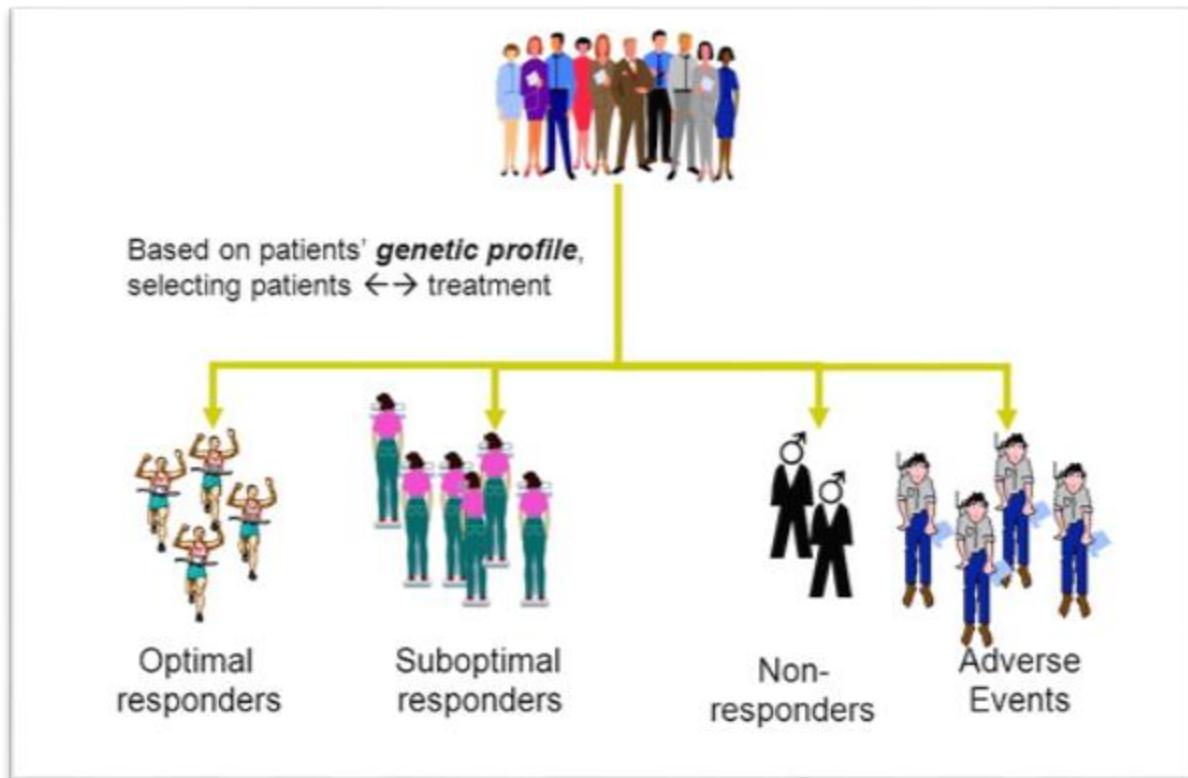
*HLA-B\*1502 in Chinese  
HLA-A\*3101 in Caucasians*



# YOUR TURN

When you see this

Is this Evidence – Precision – Personalised or Individualised medicine ?



# YOUR TURN

Which of these *omics* technologies can be used in personalised medicine?

Genes

Genomics



Transcripts

Transcriptomics



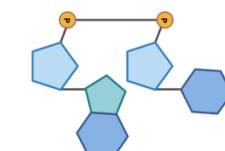
Protein

Proteomics



Metabolites

Metabolomics



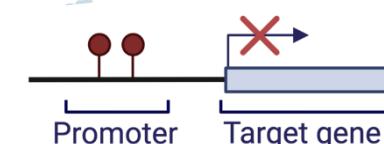
E.g., gut bacteria

Microbiomics



DNA methylation

Methylomics



# YOUR TURN

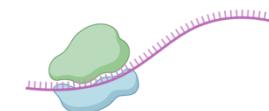
Genes

Genomics



Transcripts

Transcriptomics



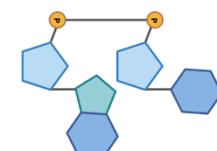
Protein

Proteomics



Metabolites

Metabolomics



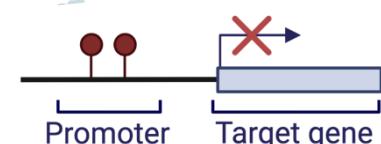
E.g., gut bacteria

Microbiomics



DNA methylation

Methylomics



# BECAUSE ...

- 1) DNA is the *Blueprint* – identical from cradle-to-grave
- 2) Driven by *technological development* – price per base
- 3) One way causation [sickle cell disease]



**biobank<sup>uk</sup>**  
Enabling scientific discoveries that improve human health

Better understanding of  
human sequence variation

Advances in genotyping and  
sequencing technologies

Sample collections of adequate size

Large-scale human studies  
linking genetic variation with  
disease susceptibility

# YOUR TURN

The screenshot shows the European Commission's Public Health website. At the top left is the European Commission logo. To its right are language selection (EN English) and search functions. A blue header bar contains the text "Public Health". Below it, a breadcrumb navigation shows the path: European Commission > Public Health > Medicinal products > Personalised medicine. The main title "Personalised medicine" is centered above a content area. On the left, under "PAGE CONTENTS", are links to "Legal Framework", "Latest updates", and "Documents". To the right, the text defines "Personalised medicine" as a medical model that aims to provide tailor-made prevention and treatment strategies for defined groups of individuals. It cites the "Council conclusions on personalised medicine for patients" from December 2015. A green-bordered box contains the definition: "A medical model using characterization of individuals' phenotypes and genotypes (e.g. molecular profiling, medical imaging, lifestyle data) for tailoring the right therapeutic strategy for the right person at the right time, and/or to determine the predisposition to disease and/or to deliver timely and targeted prevention." At the bottom, a statement notes that Personalised Medicine is an evolution of medicine, not a revolution, and challenges remain.

**Personalised medicine**

PAGE CONTENTS

Legal Framework

Latest updates

Documents

**Personalised medicine** is a medical model that aims to provide tailor-made prevention and treatment strategies for defined groups of individuals. While there is no universally accepted definition, the EU Health Ministers in their [Council conclusions on personalised medicine for patients](#), published in December 2015, defined personalised medicine as:

*A medical model using characterization of individuals' phenotypes and genotypes (e.g. molecular profiling, medical imaging, lifestyle data) for tailoring the right therapeutic strategy for the right person at the right time, and/or to determine the predisposition to disease and/or to deliver timely and targeted prevention.*

Precision medicine

- (1) Take a look at EUs definition on Personalised medicine [https://ec.europa.eu/health/medicinal-products/personalised-medicine\\_en](https://ec.europa.eu/health/medicinal-products/personalised-medicine_en)
- (2) How does this agree with our definition Evidence – Precision – Personalised or Individualised ?



**How is personalised medicine  
presented to the  
society**

***“Delivering the right treatments,  
at the right time, every time  
to the right person”***

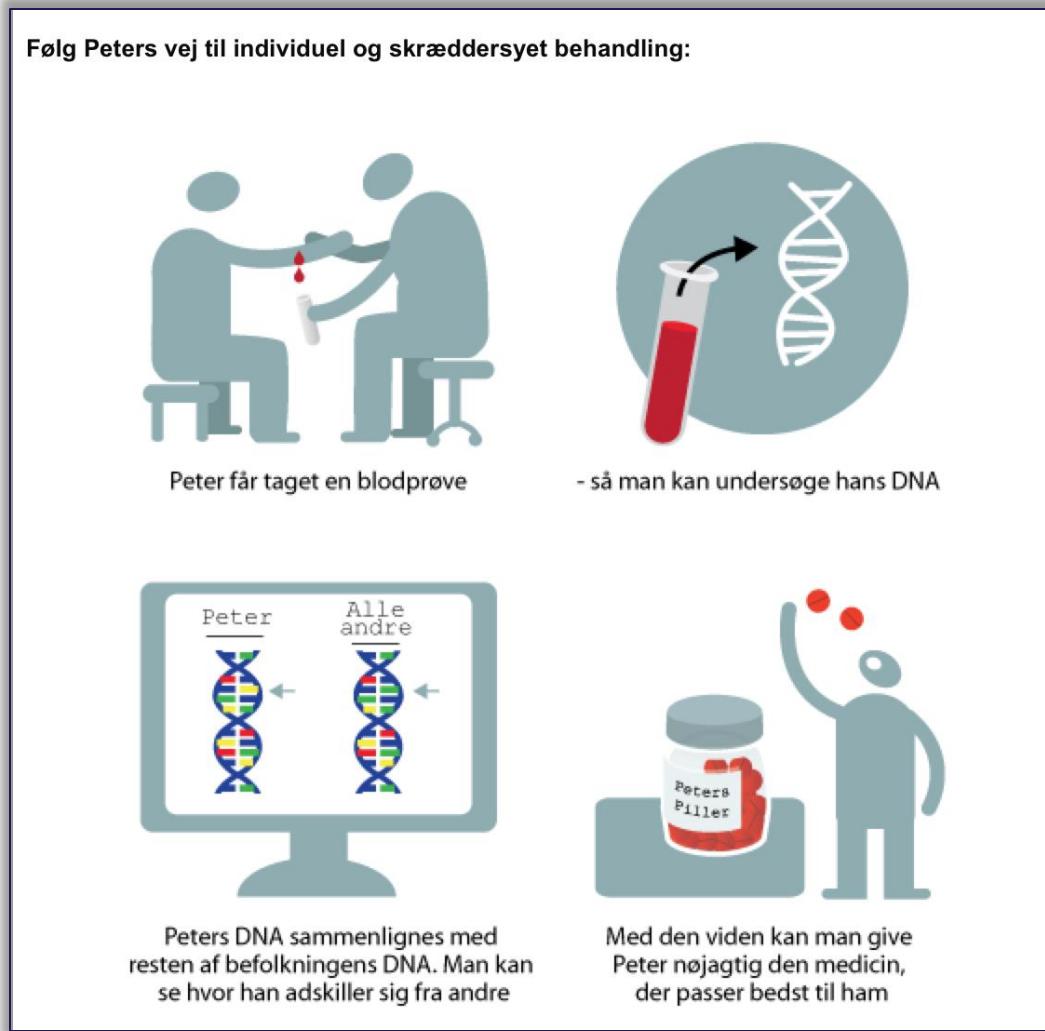


“ Sagen er ret enkel: Er vi i stand til at give patienterne den rigtige behandling fra starten af, bliver de også hurtigere raske. Og når patienterne bliver hurtigere raske, kommer de hurtigere tilbage på arbejdsmarkedet. Det er bedst for borgerne, det er bedst for samfundet. Det er en win-win.

**Bent Hansen,**  
Formand, Danske Regioner



Danish society spent >100 mio kr over three years  
to establish Nationalt Genom Center



<https://www.regioner.dk/sundhed/medicin/personlig-medicin>

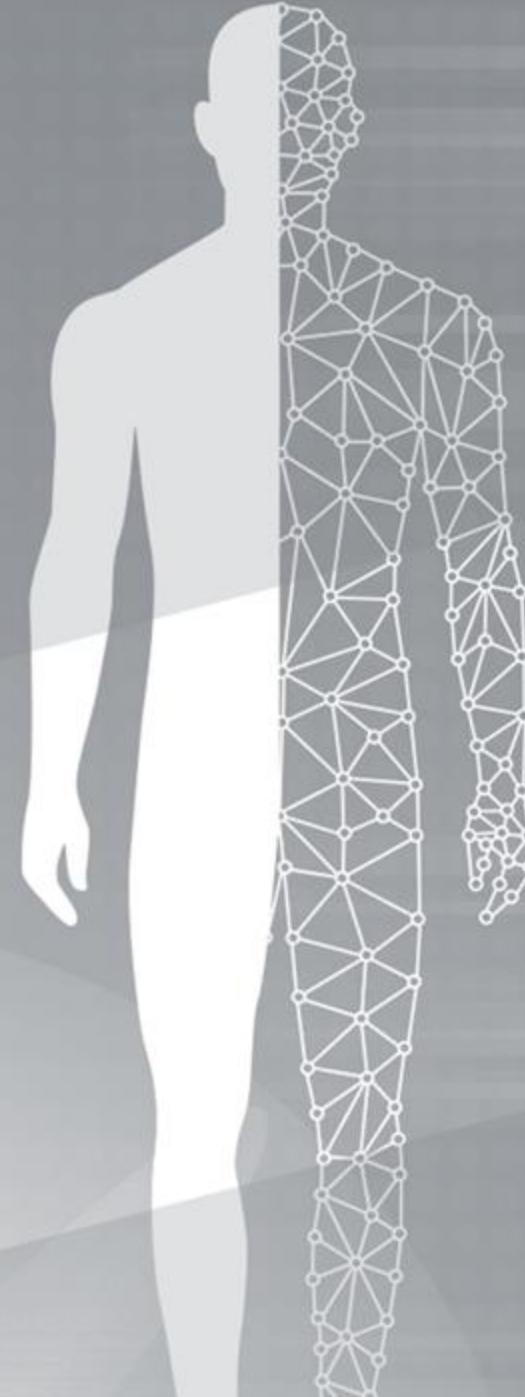


National handleplan 2021

## SUMMARY

# PERSONALISED MEDICINE

- What is Personalised Medicine?
- Why do we need it?
- Why so much focus on genetics?
- How is it presented to society?



BREAK

# OUTLINE

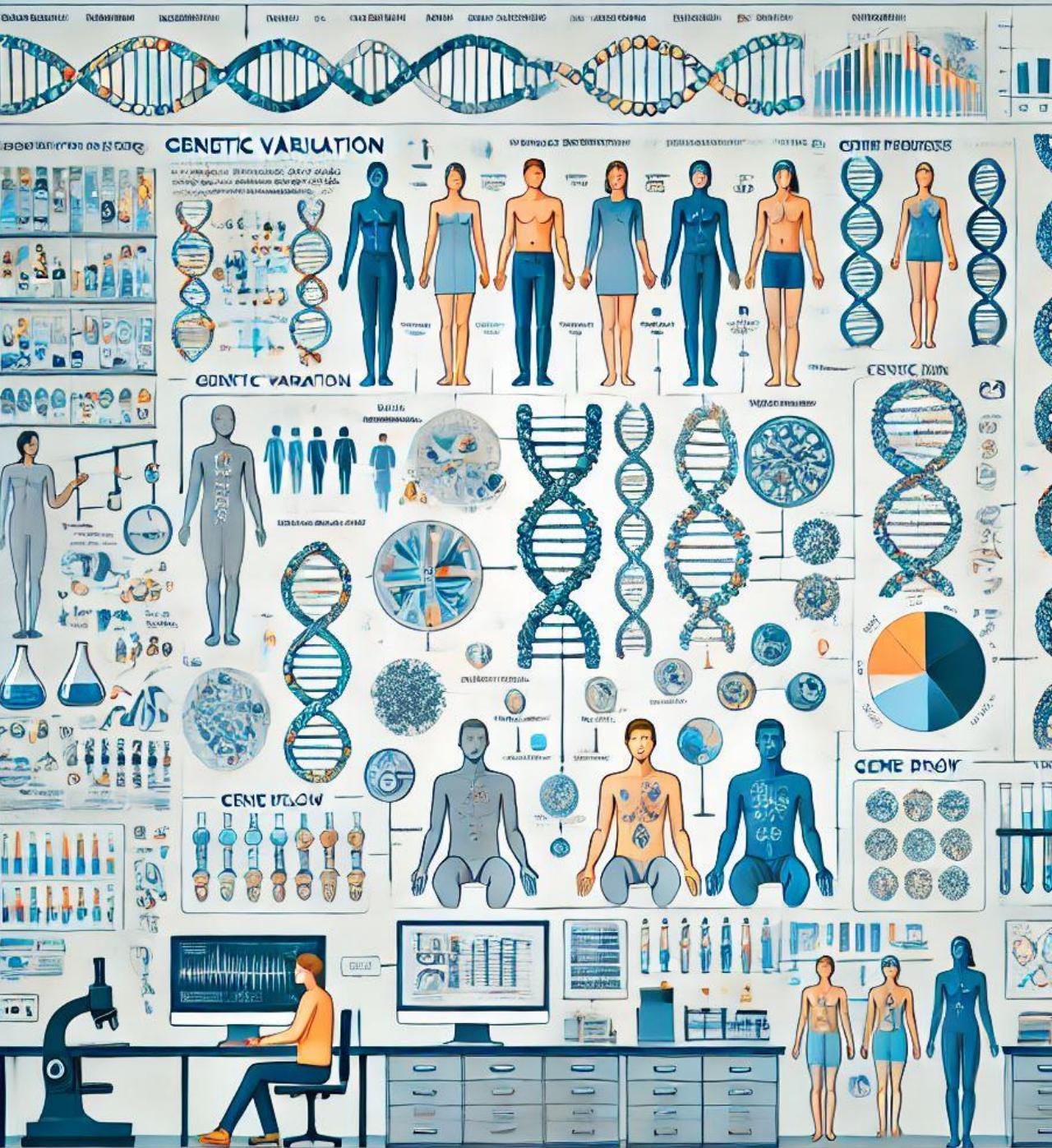
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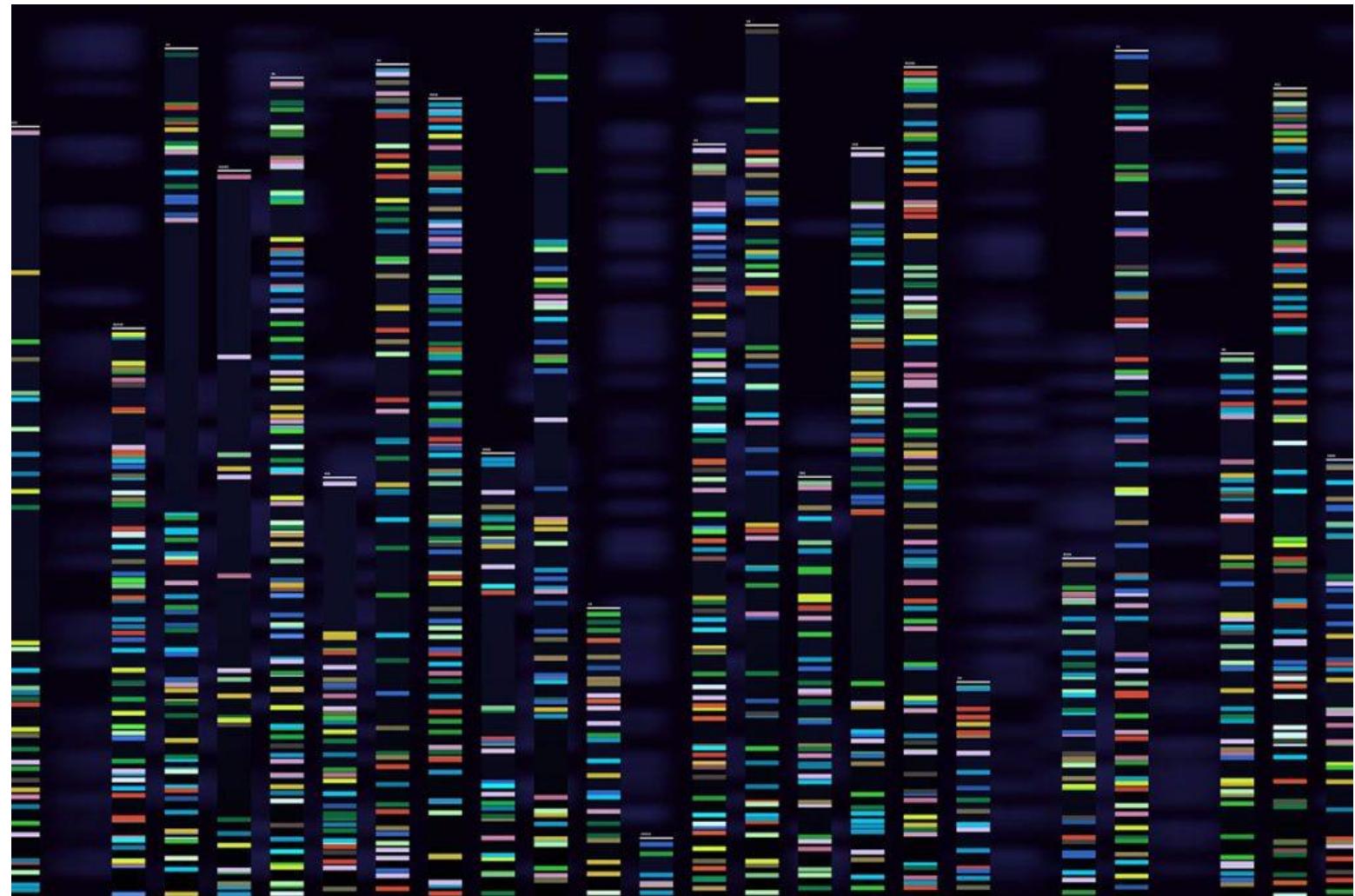
# GENETIC VARIATION

- Different types of variation
  - The genetic architecture plot
  - The journey from monogenic diseases to complex diseases

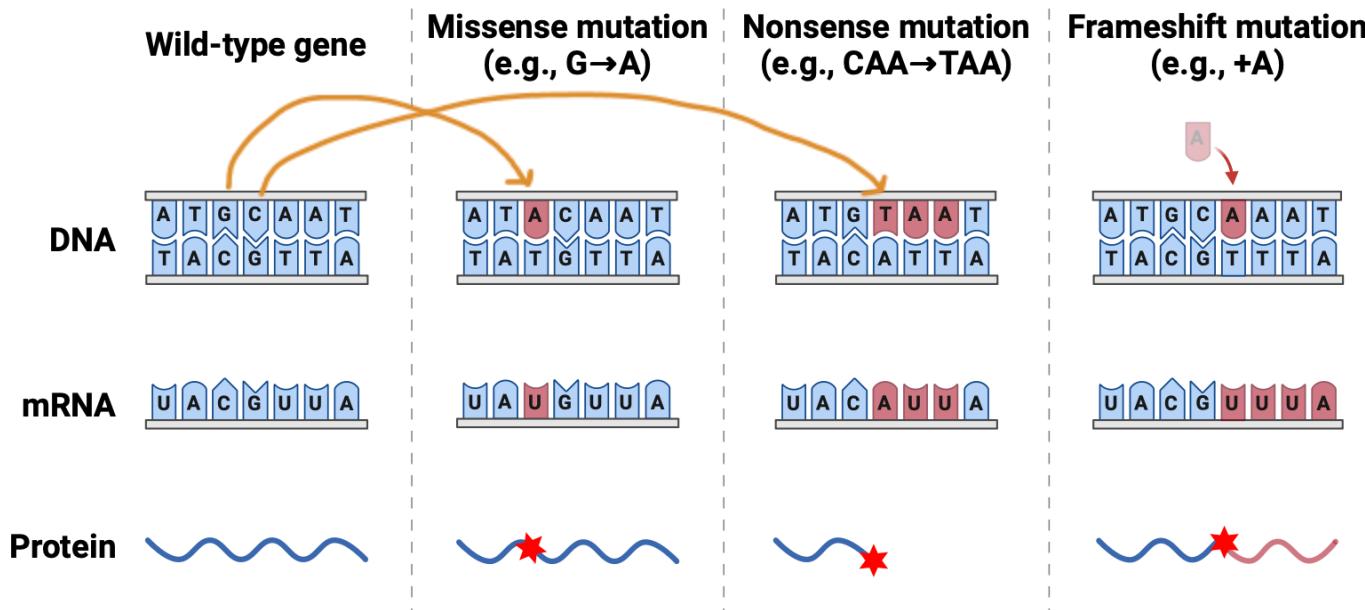


# YOUR TURN

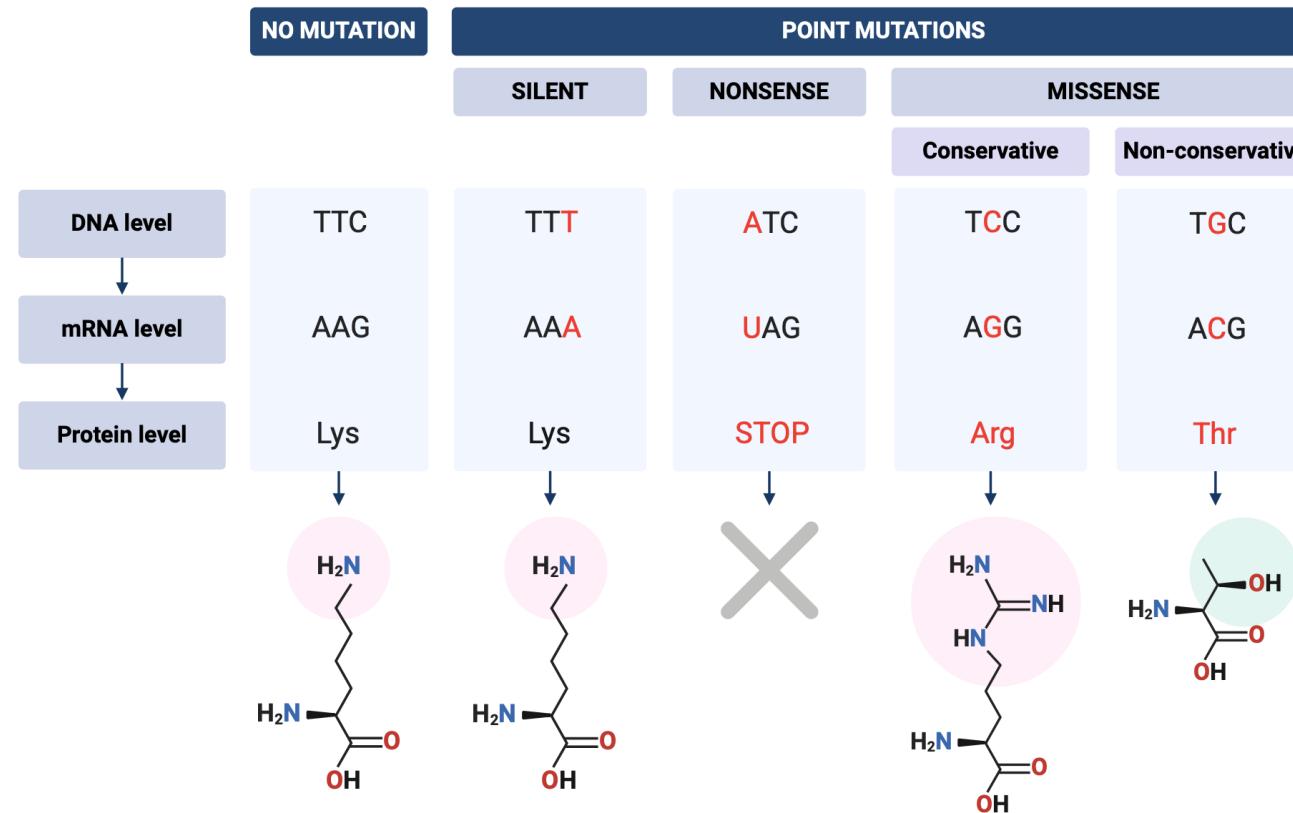
What generates genetic variation?



# MUTATIONS GENERATE GENETIC VARIATION



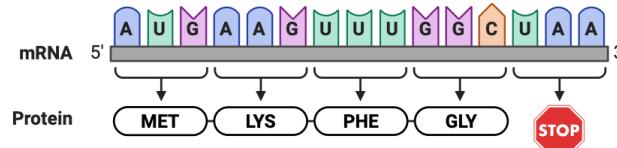
# MUTATIONS GENERATE GENETIC VARIATION



# MUTATIONS GENERATE GENETIC VARIATION

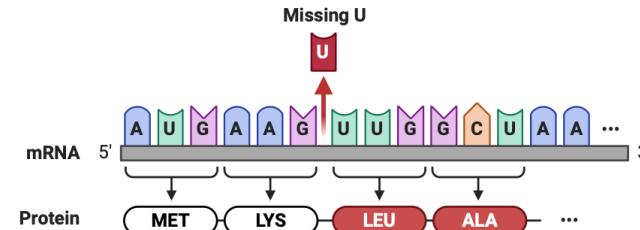
## Wild type

mRNA sequence without any mutation



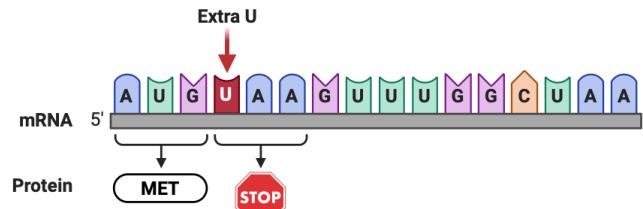
## Base-pair deletion

Frameshift causing extensive missense



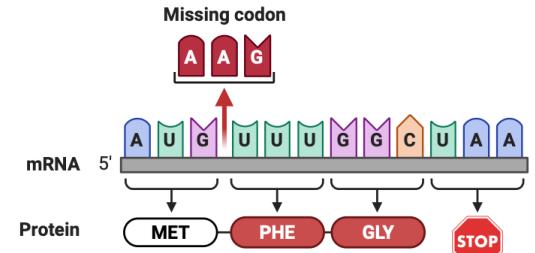
## Base-pair insertion

Frameshift causing immediate nonsense



## Three-nucleotide insertion/deletion

Extra/missing amino acids



# GENETIC DIVERSITY

Human evolution is driven by several different (evolutionary) factors

- ❖ Genetic mutations
- ❖ Migration
- ❖ Natural selection
- ❖ Genetic drift

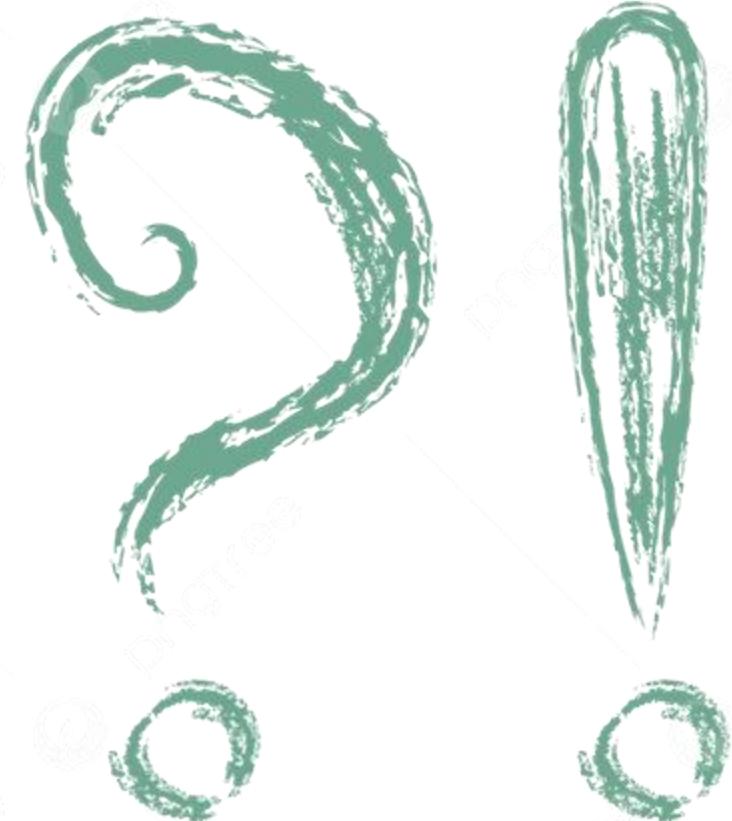
The product is genetic diversity within a population.

**Understanding the genetic diversity and how it has arisen is a necessary precursor to understand the genetics of complex traits.**



# GENETIC TERMINOLOGY 1/2

- ❖ The **genome** refers to the complete set of genetic information found in a cell and includes 22 pairs of autosomal chromosomes plus XX or XY.
- ❖ The human genome is made up of >20,000 **genes**, and the location of a gene is referred to as a **locus**.
- ❖ Genetic variation at a locus is referred to as **allelic variation**, where the different forms are known as **alleles**.
- ❖ Underlying genetic variation are **changes in DNA sequence**.

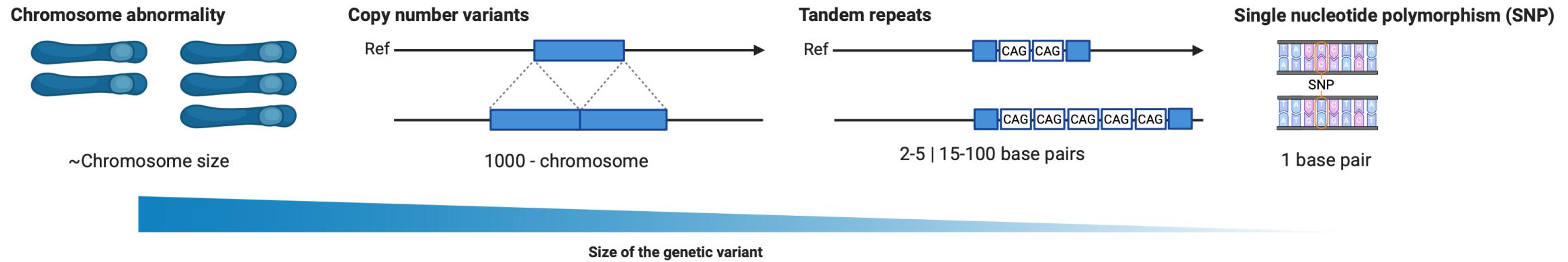


# GENETIC TERMINOLOGY 2/2

- ❖ Traditionally, the term **mutation** has been used in two ways;
  - an event that produces a change in the base sequence
  - the outcome of the process, the altered DNA sequence.
- ❖ The great majority of mutations are neutral on the phenotype, thus more neutral terms are now preferred:
  - **DNA variant** or **genetic variant**
- ❖ In population genomics, a DNA variant is classified as common (>5%), low frequent (0.5-5%), or rare (<0.5%).
  - Single Nucleotide Polymorphisms (**SNPs**, DNA variant freq of >1%)
  - Single Nucleotide Variants (**SNVs**)



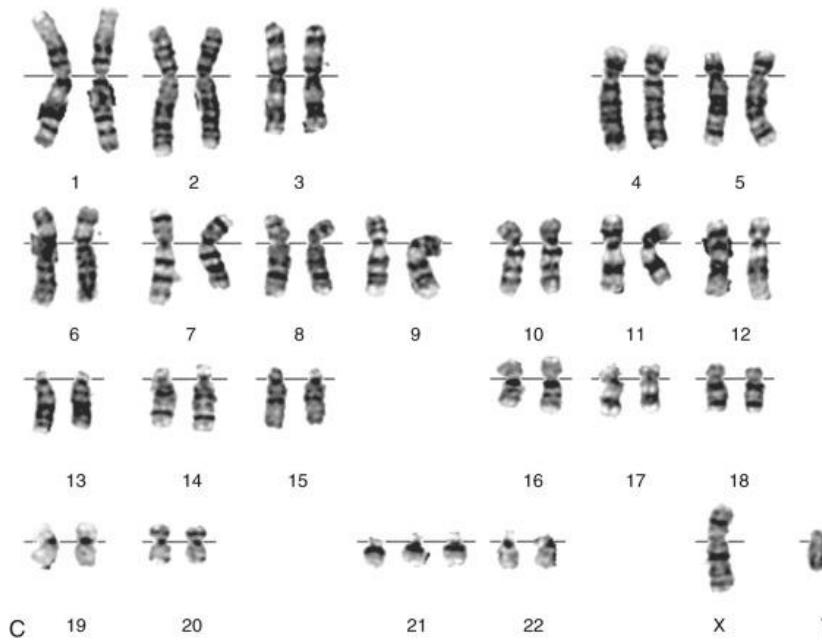
# SIZES OF GENETIC VARIATION



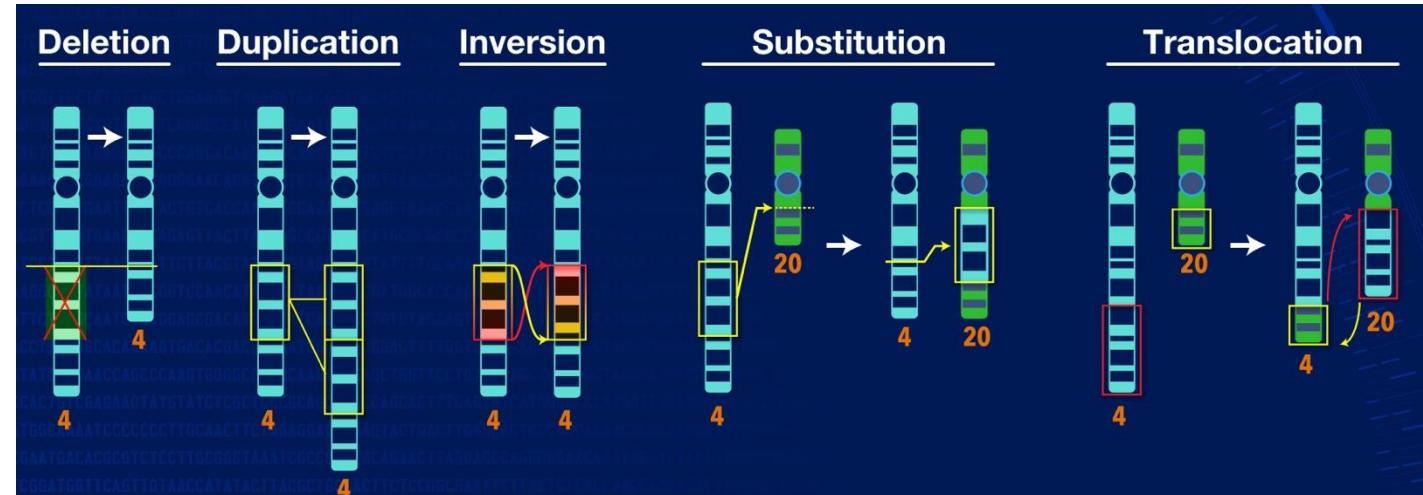
# GENETIC VARIATION

## CHROMOSOME ABNORMALITIES

Chromosome abnormalities can be numerical or structural.



Trisomi 21



# GENETIC VARIATION

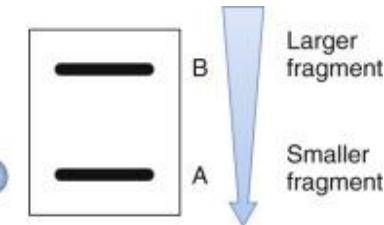
## TANDEM REPEATS



Advantage:  
many many  
alleles exists

Micro satellites  
(2-5 bp) - STRs

Mini satellites  
(15-100 bp) -  
VNTR



### Forensic DNA analysis

United States, 13 autosomal Short Tandem Repeat (STR) loci are now accepted as the system used for forensic purposes.

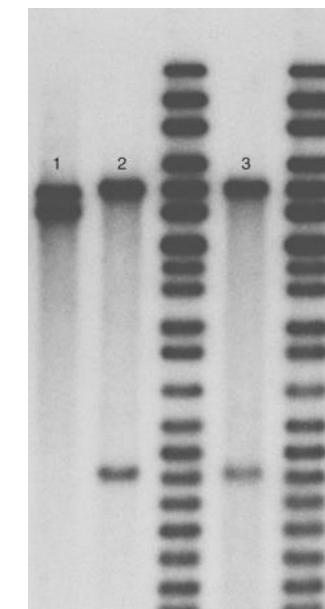
Short tandem repeats	8 repeats
Participant 1	CTAGAGATAGATAGATAGATAGATAGATAGACTAGACTAG
Participant 2	CTAGAGATAGATAGATAGATAGATAGATAGATAGACTAGA
Participant 3	CTAGAGATAGATAGATAGATAGATAGATAGATAGACTAGA
Participant 4	CTAGAGATAGATAGATAGATAGATAGATAGATAGATAGACTAGAC

Short tandem repeats	9 repeats
Participant 1	CTAGAGATAGATAGATAGATAGATAGATAGACTAGACTAG

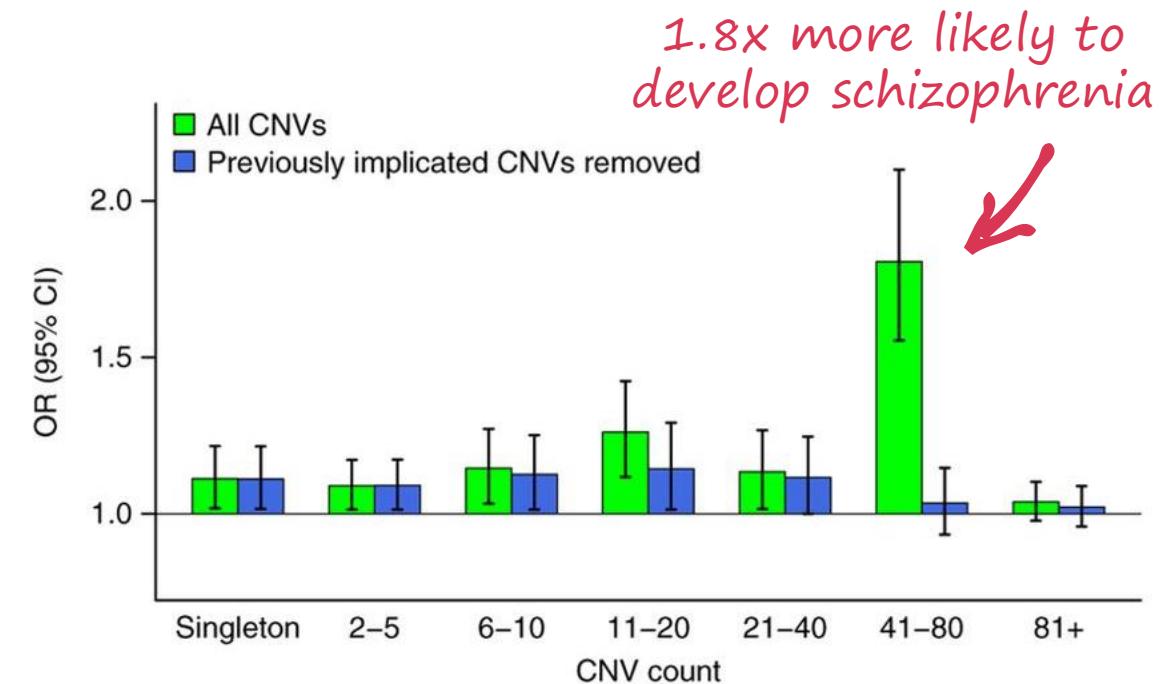
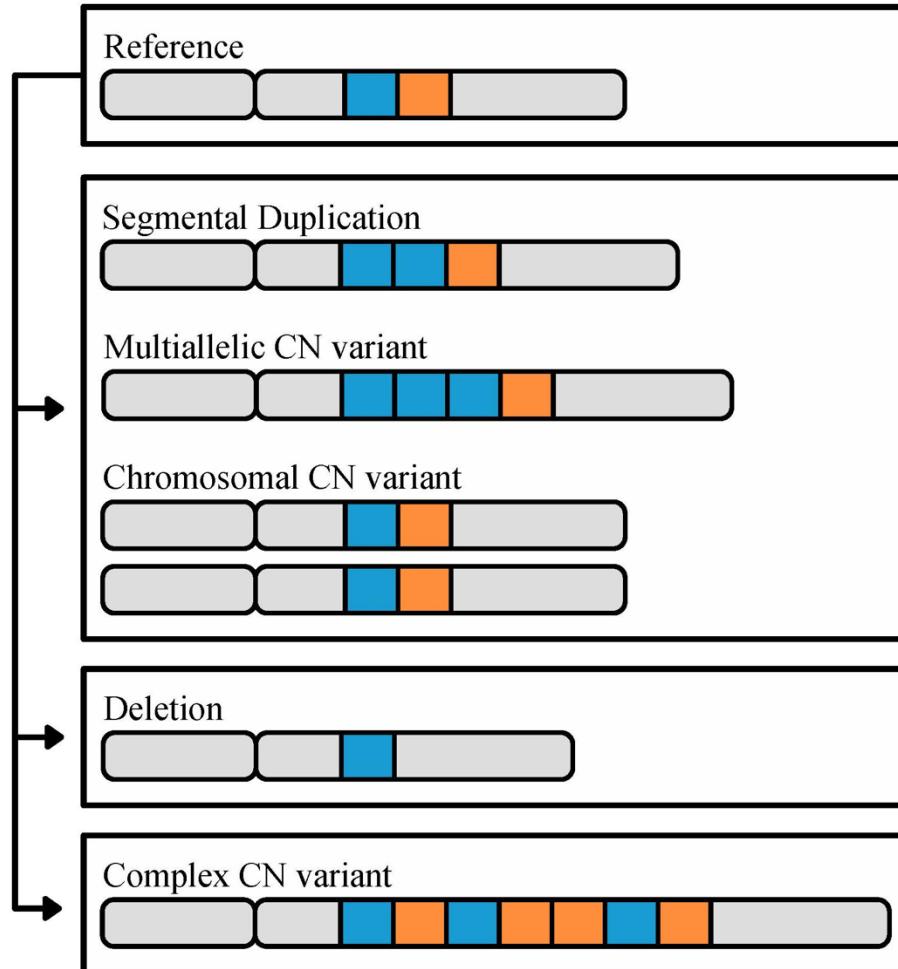
Short tandem repeats	10 repeats
Participant 4	CTAGAGATAGATAGATAGATAGATAGATAGATAGATAGACTAGAC



# GENETIC VARIATION

## COPY NUMBER VARIANTS (CNV)

50 bp - kromosom

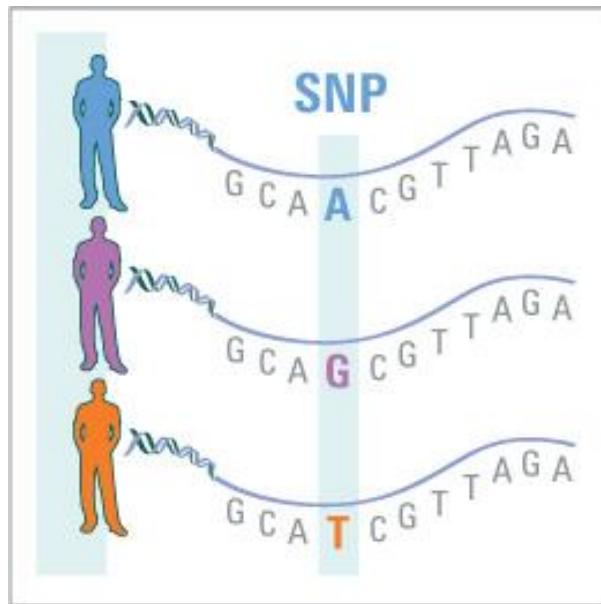


# GENETIC VARIATION

## SINGLE NUCLEOTIDE POLYMORPHISMS (SNPs)

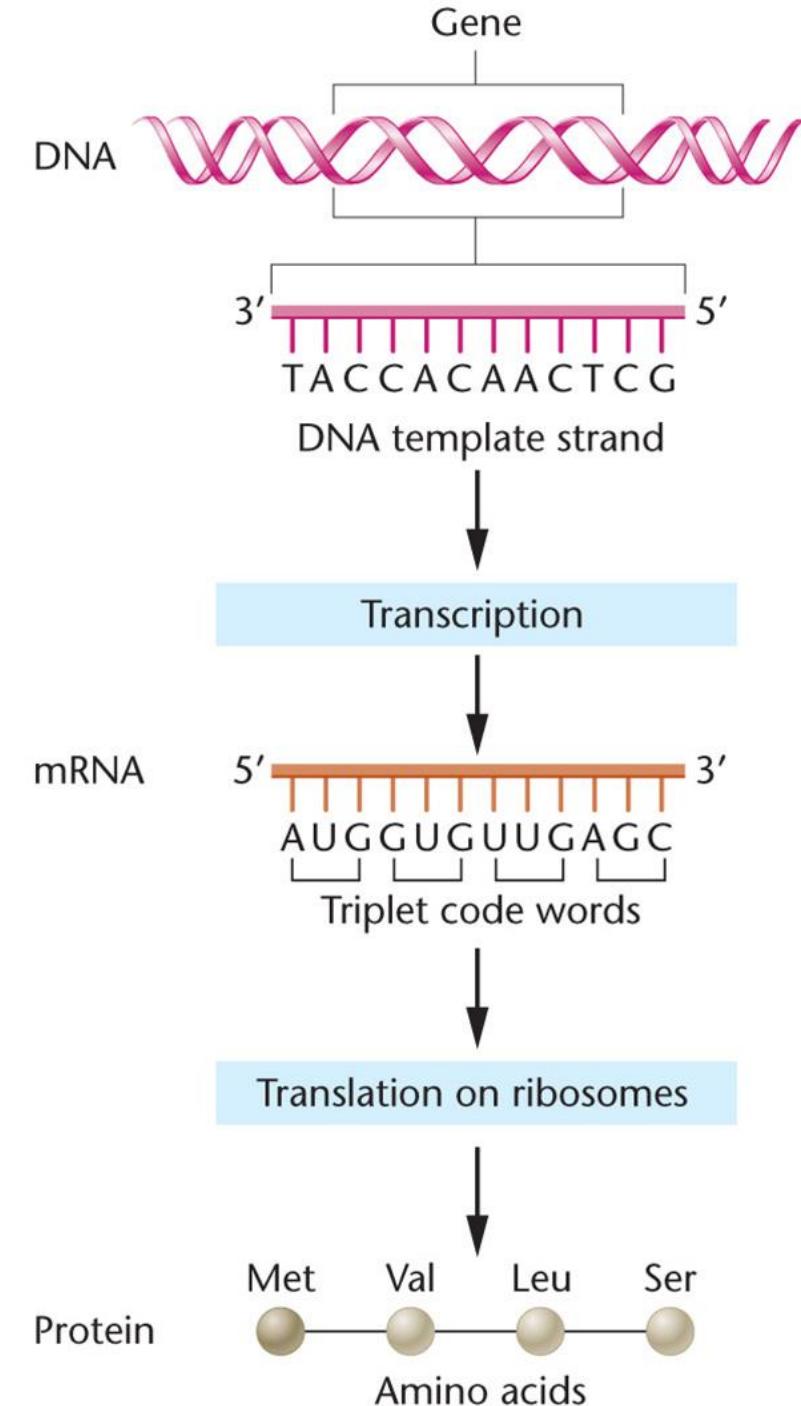
A common change in a single base pair; ~1/1000 bp

Accounts for ~90% of all variation in the human genome



All (known) SNPs have a unique identifier  
(independent of alleles)

*rsXXX – Ref-*SNP cluster ID number**



# GENETIC VARIATION

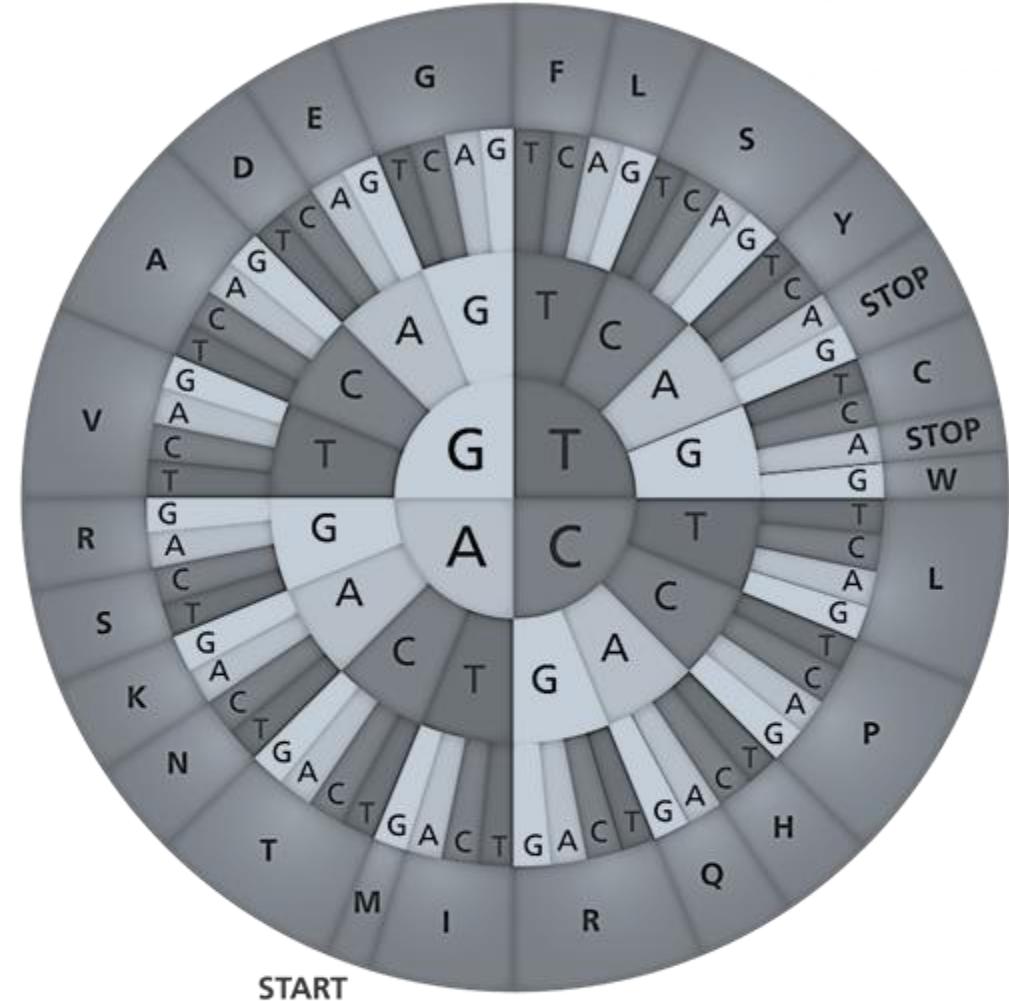
Genetic variation arise as a consequence  
of changes in the DNA sequence



Can change the order of amino acids



The structure of the protein is changed



Amino acid code			
A - Alanine	G - Glycine	M - Methionine	S - Serine
C - Cysteine	H - Histidine	N - Asparagine	T - Threonine
D - Aspartic acid	I - Isoleucine	P - Proline	V - Valine
E - Glutamic acid	K - Lysine	Q - Glutamine	W - Tryptophan
F - Phenylalanine	L - Leucine	R - Arginine	Y - Tyrosine

# GENOTYPE TO PHENOTYPE

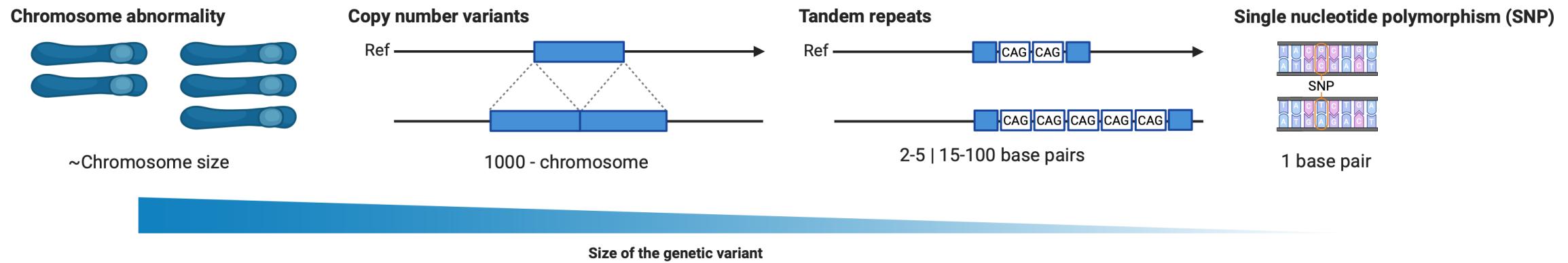
NORMAL $\beta$ -GLOBIN				
DNA.....	TGA	GGA	CTC	CTC.....
mRNA.....	ACU	CCU	GAG	GAG.....
Amino acid.....	Thr	Pro	Glu	Glu.....
	4	5	6	7
MUTANT $\beta$ -GLOBIN				
DNA.....	TGA	GGA	CAC	CTC.....
mRNA.....	ACU	CCU	GUG	GAG.....
Amino acid.....	Thr	Pro	Val	Glu.....
	4	5	6	7

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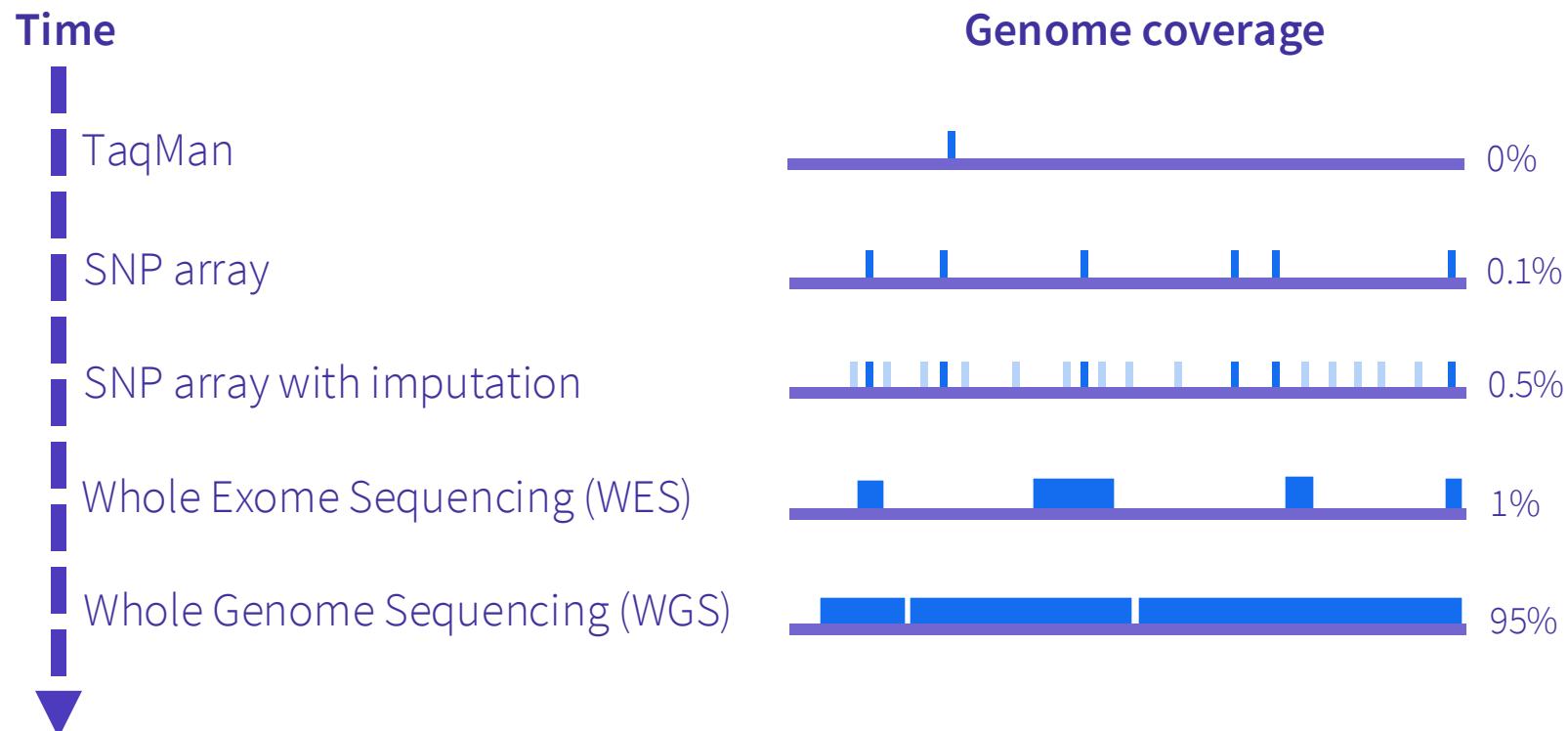


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# SIZES OF GENETIC VARIATION



# GENOMIC COVERAGE



Adapted from Uitterlinden A. (2016) An Introduction to Genome-Wide Association Studies: GWAS for Dummies. Seminars in Reproductive Medicine, 34(4): 196-204.

# GENOTYPING VS SEQUENCING

# GENOTYPING VS SEQUENCING

# Genotyping

# GENOTYPING VS SEQUENCING

# Genotyping

# WES

# GENOTYPING VS SEQUENCING

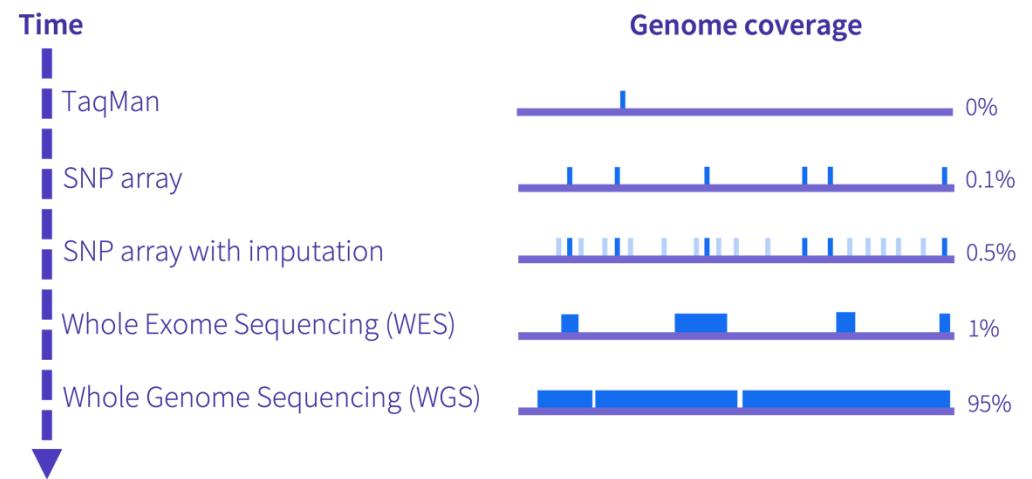
## Genotyping

WES

WGS

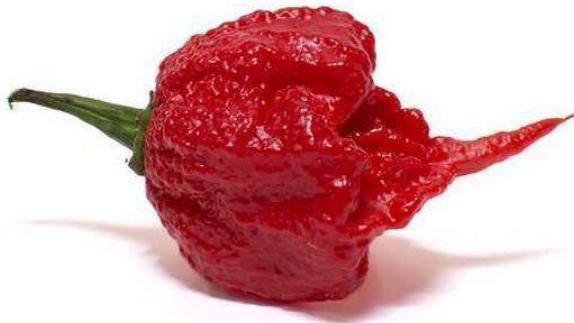
# WHICH TECHNOLOGY?

The choice of technology for detecting single nucleotide polymorphisms (SNPs) depends upon the application.



# GENETIC VARIATION IS LIKE CHILI

Carolina reaper



Strong effect on phenotype

Habanero Lemon



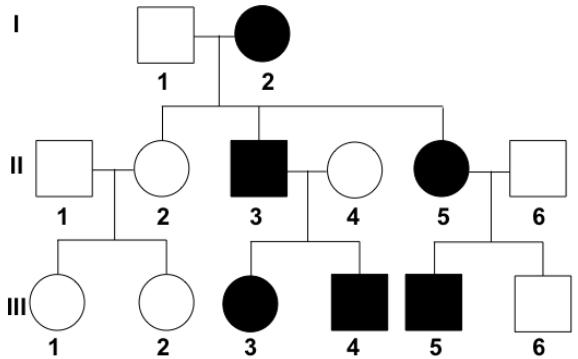
Moderate effect on phenotype

Bell pepper

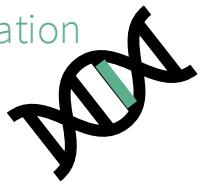


Weak effect on phenotype  
(or none at all)

# GENETIC VARIATION IS LIKE CHILI



Mutation



Each genetic variant is **both** necessary and sufficient



Each genetic variant is **neither** necessary nor sufficient



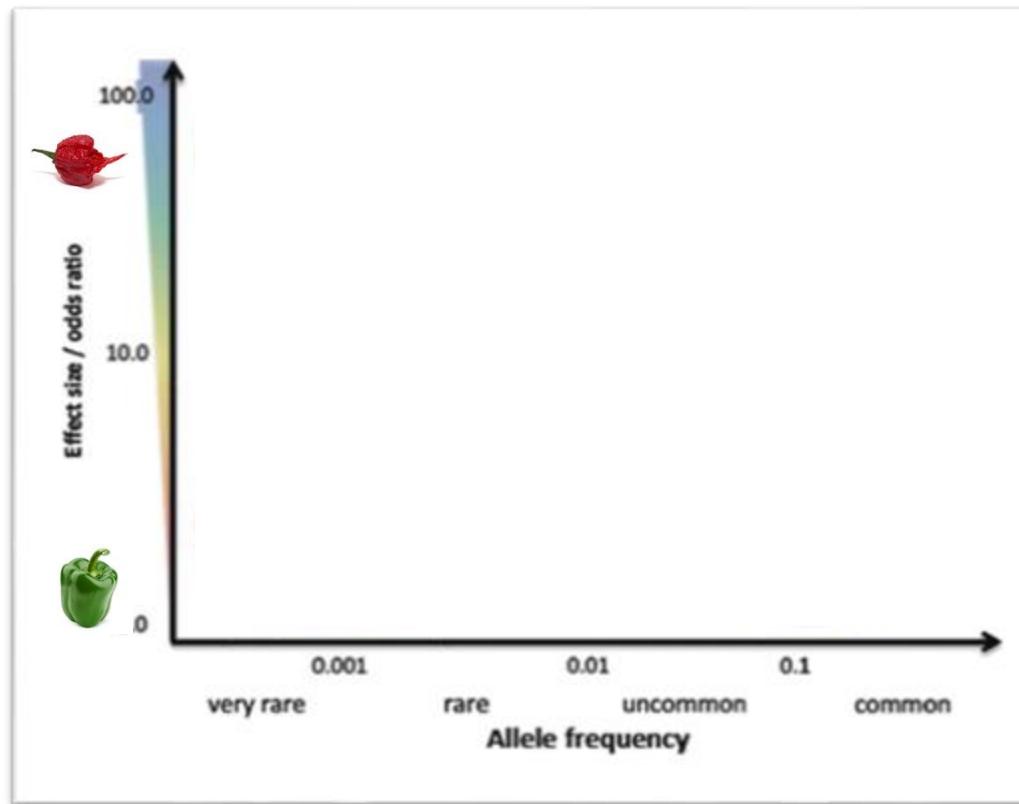
# ARCHITECTURE PLOT

All genetic variants have a spot in the architecture plot (for a certain phenotype)

## YOUR TURN

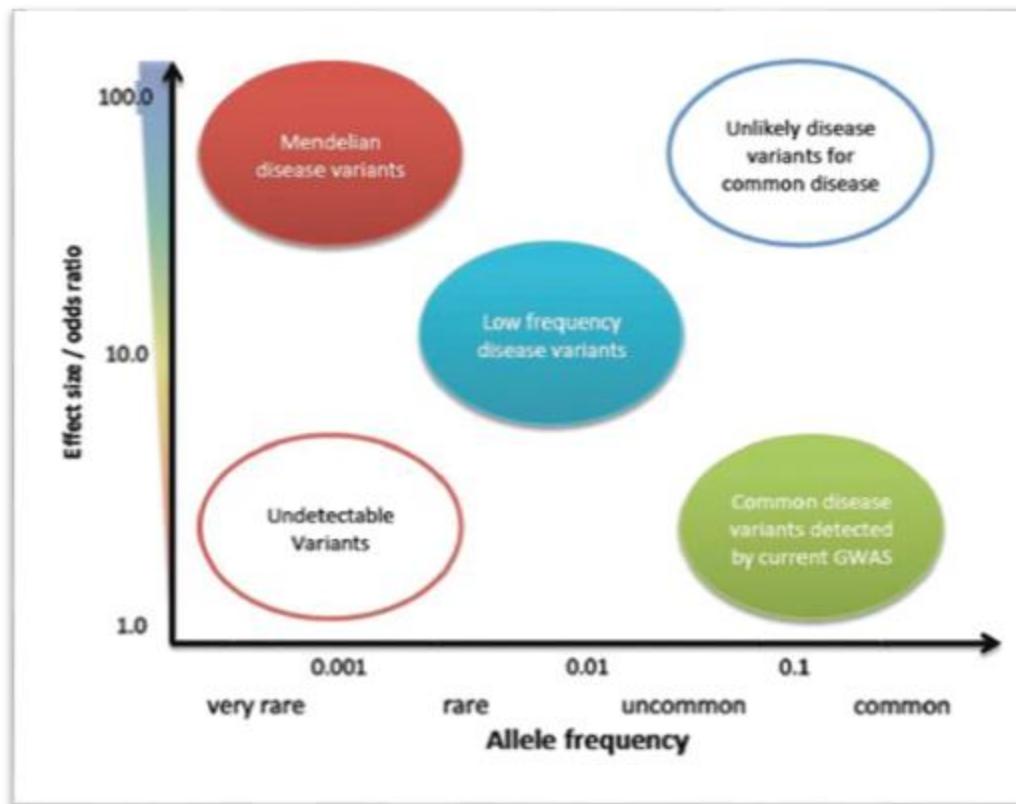
Where are variants causing monogenic disease?

Where are the variants causing complex disease?



Monogenic and complex diseases have different genetic architectures

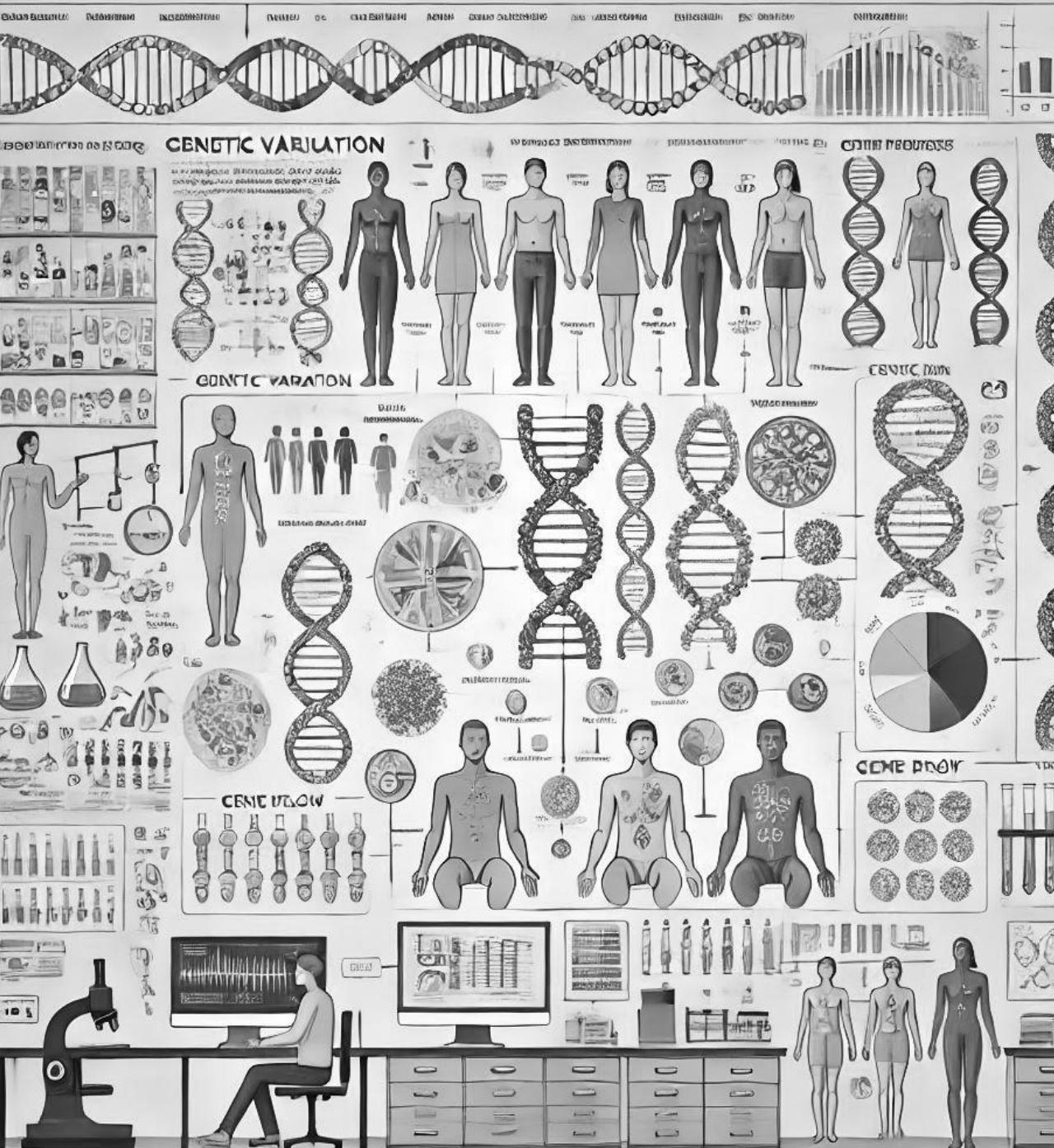
# IN THIS COURSE WE LOOK AT THE FULL SPECTRUM



Monogenic and complex diseases  
have different genetic architectures

# SUMMARY GENETIC VARIATION

- Different types of variation
- The genetic architecture plot
- The journey from monogenic diseases to complex diseases





**BREAK**

# OUTLINE

<b>08:15 – 08:30</b>	Welcome (15 min)
<b>08:30 – 08:55</b>	Exercises 1 (25 min) [ <i>figure recap</i> ]
<b>08:55 – 09:20</b>	Lecture - <i>What is personalised medicine?</i> (25 min)
<b>09:20 – 09:35</b>	Break
<b>09:35 – 10:05</b>	Exercises 2 (30 min) [1-6]
<b>10:05 – 10:20</b>	Plenum (15 min) [ <i>solutions</i> ]
<b>10:20 – 10:45</b>	Lecture - <i>Genetic variation</i> (25 min)
<b>10:45 – 11:00</b>	Break
<b>11:00 – 11:30</b>	<b>Exercises 3 (30 min) [7-11]</b>
<b>11:30 – 11:45</b>	Plenum (15 min) [ <i>genetic variation</i> ]
<b>11:45 – 12:00</b>	eBoard evaluation + Crossword

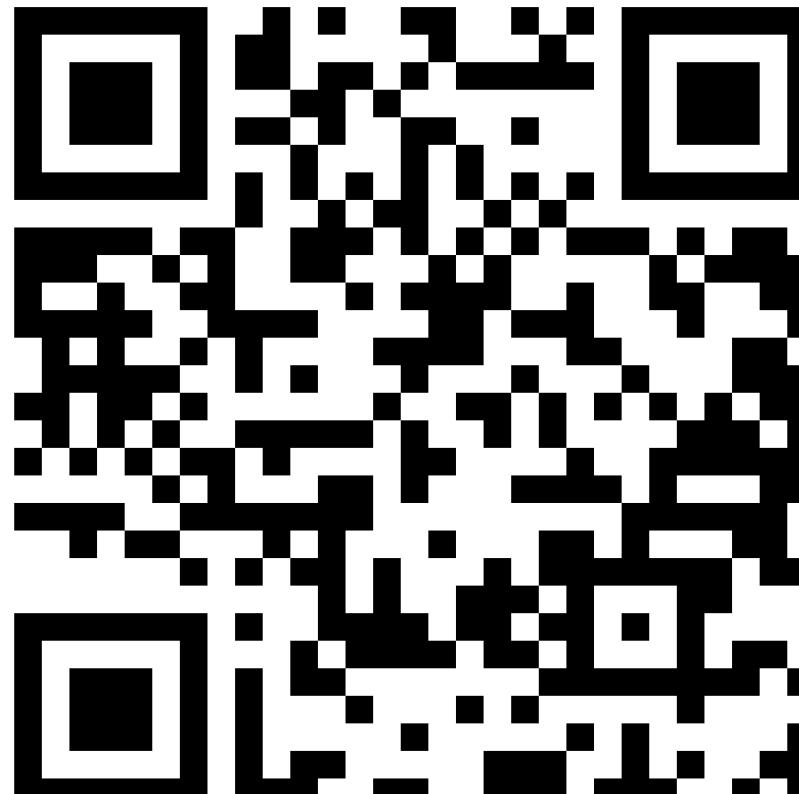
# SUMMARY

- ⌚ 10 sessions - read the readme - prepare before class – **this is important**
- ⌚ Personalised medicine is not new
- ⌚ We need it because people are different – different response and side effects
- ⌚ Evidence-Precision-Personalised-Individualised
- ⌚ Strong focus on genetics because of blueprint and causality (and price per base)
- ⌚ All genetic variants have a place in the architecture plot
- ⌚ This course will look at ***all*** variants – strong and weak

# OUTLINE

08:15 – 08:30	Welcome (15 min)
08:30 – 08:55	Exercises 1 (25 min) [In pairs <i>figure recap</i> ]
08:55 – 09:20	What is personalised medicine? (25 min)
09:20 – 09:35	Break
09:35 – 10:05	Exercises 2 (30 min) [1-6]
10:05 – 10:20	Plenum (15 min) [ <i>solutions</i> ]
10:20 – 10:45	Genetic variation (25 min)
10:45 – 11:00	Break
11:00 – 11:30	Exercises 3 (30 min) [7-11]
11:30 – 11:45	Plenum (15 min) [ <i>solutions</i> ]
<b>11.45 – 12:00</b>	<b>eBoard evaluation + Crossword</b>

# eBOARD EVALUATION



[LINK](#)

# GENOMICS CROSSWORD

<https://crosswordlabs.com/embed/pm-genomics-brushup>

*Hint: Use “-” to indicate a space*

