



HUMAN GENOMICS

PALLE DUUN ROHDE

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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

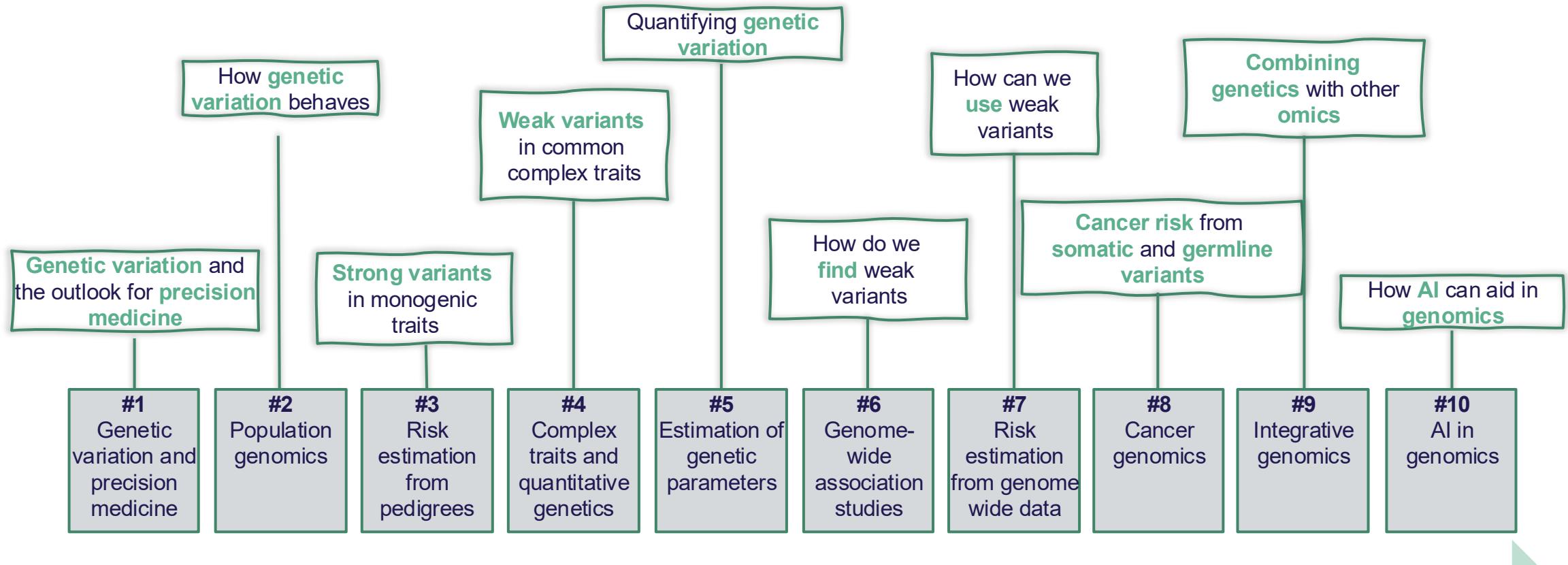
COURSE PLAN

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

#1 Genetic variation and precision 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 Cancer genomics	#9 Integrative genomics	#10 AI in genomics
4/2-26 [PDR]	6/2-26 [PDR]	11/2-26 [PDR]	25/2-26 [PDR]	3/3-26 [PDR]	10/3-26 [PDR]	13/3-26 [PDR]	16/3-26 [PDR]	24/3-26 [PDR]	30/3-26 [FFG PDR]



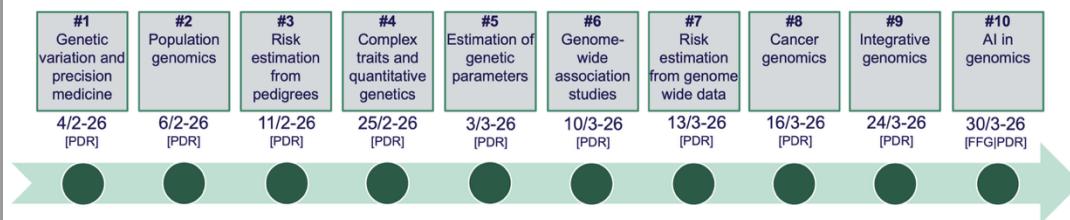
WHY THIS COURSE PLAN



COURSE MATERIAL IN MOODLE

Welcome to the 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 precision medicine.

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



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 [HERE](#).

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.

I am looking forward to see you all very soon.

Cheers

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#1 Introduction to ...



#1 Introduction to genetic variation and precision medicine

February 4th, Palle Duun Rohde

Preparation prior to the lecture

- 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 4th 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.

Lecture notes and material

Lecture notes and exercises are available at this link [[will be available before class](#)].

THE EXAM

- ❖ Individual, oral examination.
- ❖ Duration: 20 min (including assessment); 5 min presentation of known theme followed by questions across themes.
- ❖ Preparation time: 20 min with aids.
- ❖ Internal censor. Course coordinator will be responsible for the exam.
- ❖ Grading: Passed/Not passed.
- ❖ Re-examination is oral.



WHAT IS GOOD TEACHING?

- ❖ What is good teaching for you?



60 sec with the person left to you

- ❖ What format of teaching do you prefer?



60 sec with the person right to you

- ❖ What do I expect from you?

Be well-prepared for each session

- study the material from Moodle, independently study challenging concepts, and complete any unfinished exercises.

Engage actively in sessions

- take part in discussions, group work, and other interactive activities.

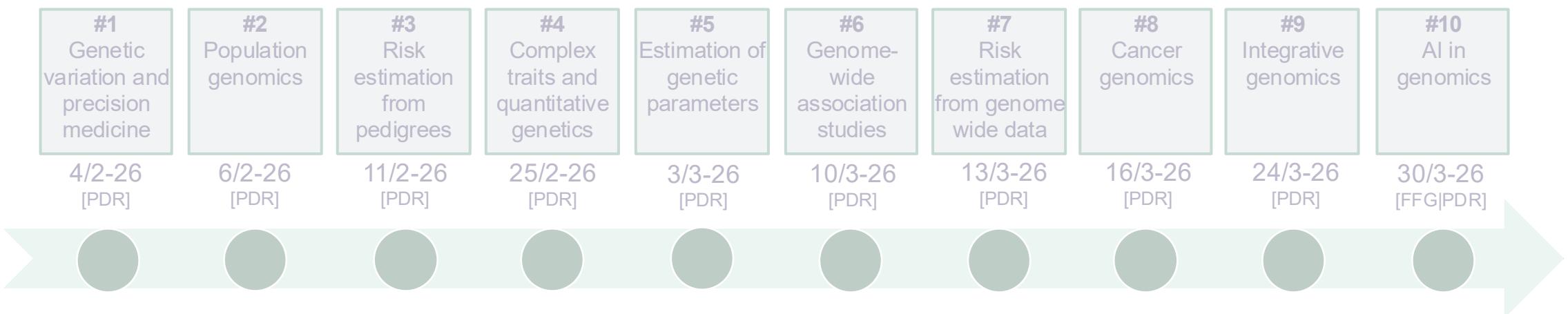
Positive and inclusive atmosphere

- ensure that everyone feels comfortable asking questions, discussing scientific topics openly, and both offering and receiving help from peers.



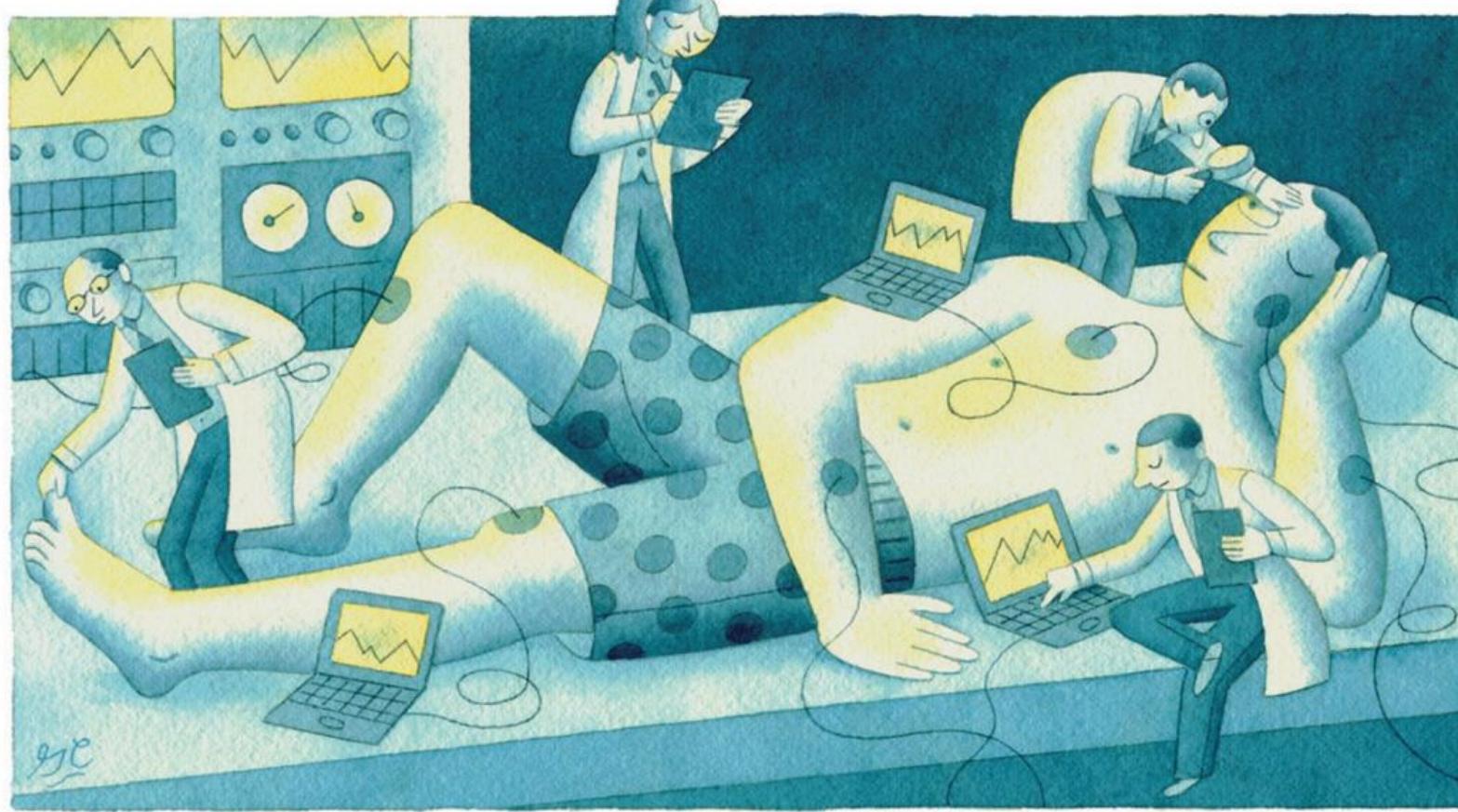
- ❖ If you need to use your Phone – please leave the room
- ❖ Minimize your usage of generative AI
- ❖ Write your own notes

LETS GET STARTED



INTRODUCTION TO

ILLUSTRATION BY GREG CLARKE



PRECISION MEDICINE

OUTLINE

12:30 – 12:45	Welcome (15 min)
12:45 – 13:10	Exercise Part I (25 min) [FIGURE RECAP]
13:10 – 13:35	Lecture - <i>What is precision medicine?</i> (25 min)
13:35 – 13:50	Break
13:50 – 14:20	Exercise Part II (30 min) [E1-E6]
14:20 – 14:35	Plenum (15 min) [SOLUTIONS]
14:35 – 15:00	Lecture - <i>Genetic variation</i> (25 min)
15:00 – 15:15	Break
15:15 – 15:45	Exercise Part III (30 min) [E7-E9 + R intro]
15:45 – 16:00	Plenum (15 min) [SOLUTIONS]
16.00 – 16:15	Evaluation + Crossword

OUTLINE

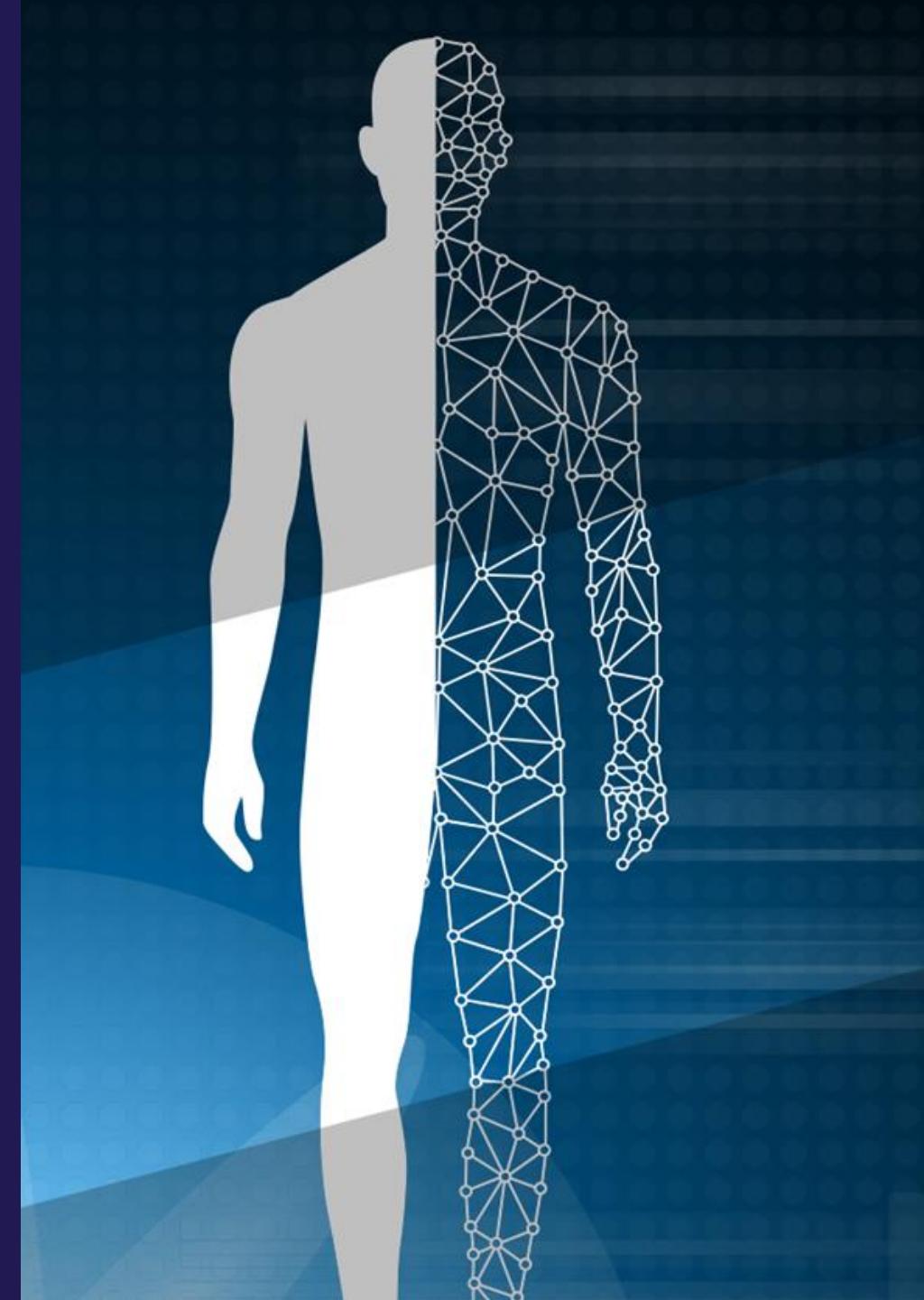
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16.00 – 16:15	Evaluation + Crossword

PRECISION MEDICINE

- What is Precision Medicine?
- Why do we need it?
- Why so much focus on genetics?



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

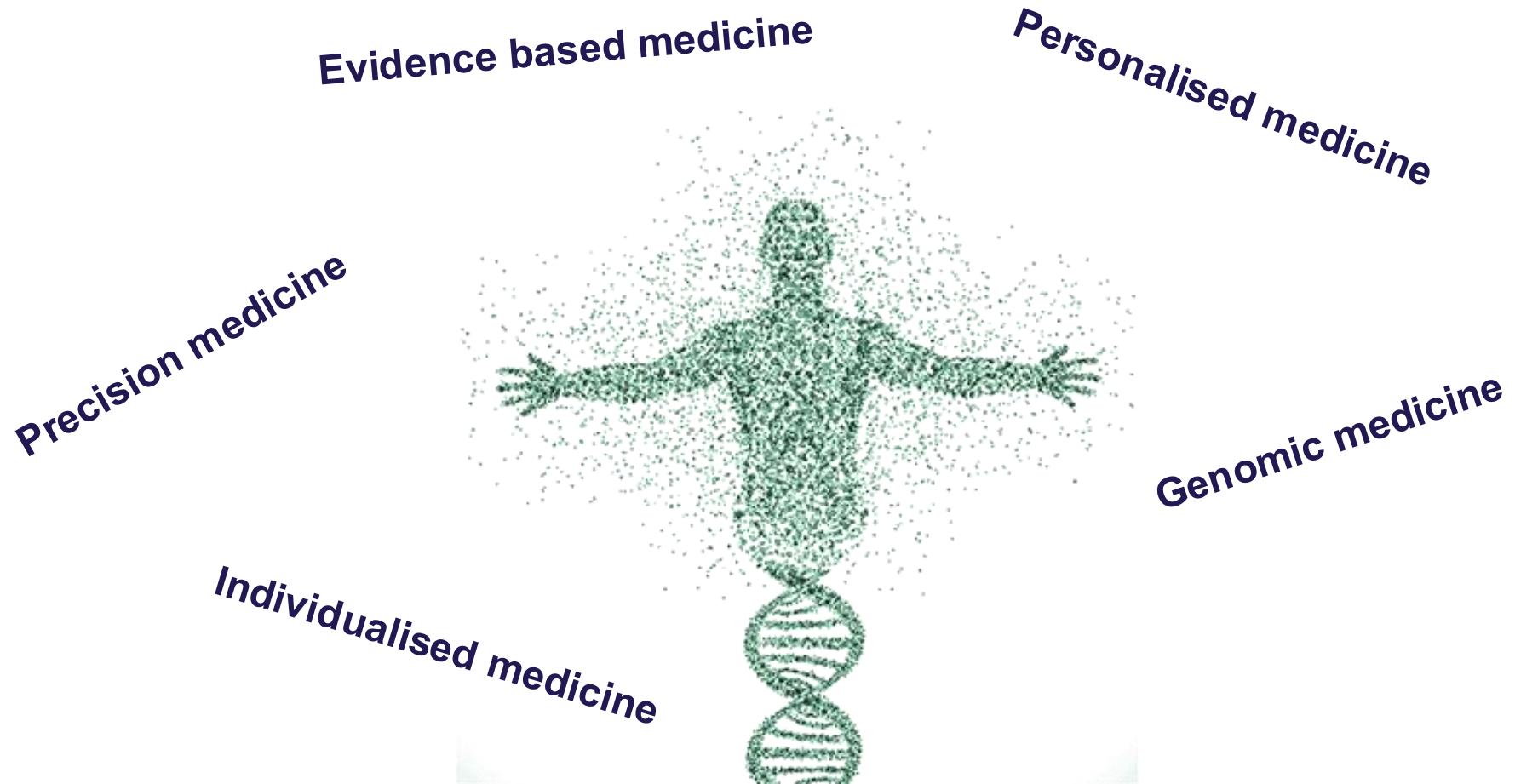
- can you think of an example?





... PM is not new

WHAT IS PRECISION MEDICINE?



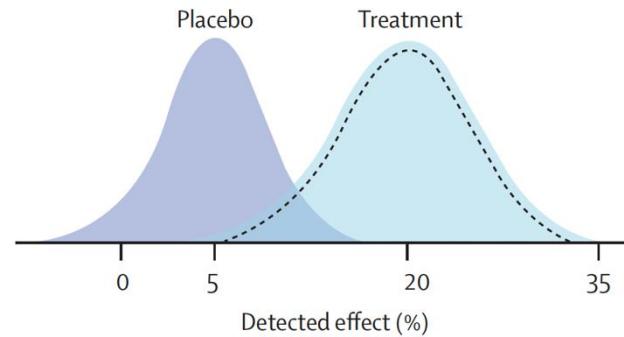
IMPLEMENTATION OF PRECISION MEDICINE

EPPOS [evidence-based precision personalised objective subjective]

Evidence-based Medicine

(1) Contemporary evidence-based medicine

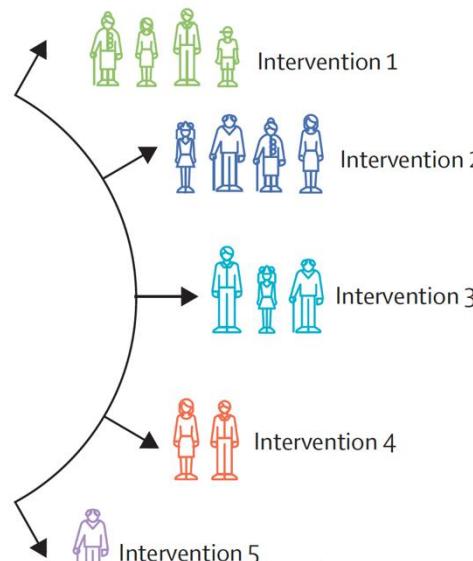
Estimate average risk or response using epidemiological and clinical trial cohorts



Precision Medicine

(2) Probability scoring and stratification

Maximise response and minimise risk using subclassification



Personalised Medicine

(3) Personalisation (objective)

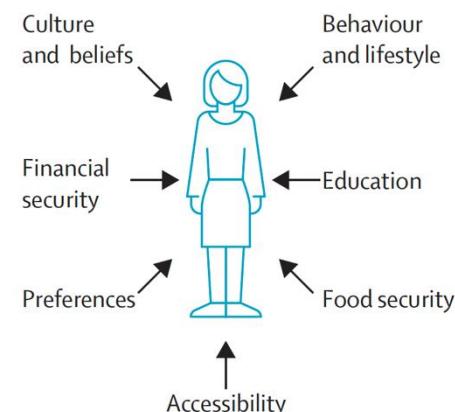
Monitor response to optimise dose, timing, and delivery



Individualised Medicine

(4) Personalisation (subjective)

Adapt intervention to fit the person's needs, capabilities, and preferences





WHAT IS PRECISION 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 PRECISION MEDICINE?

Because people are different



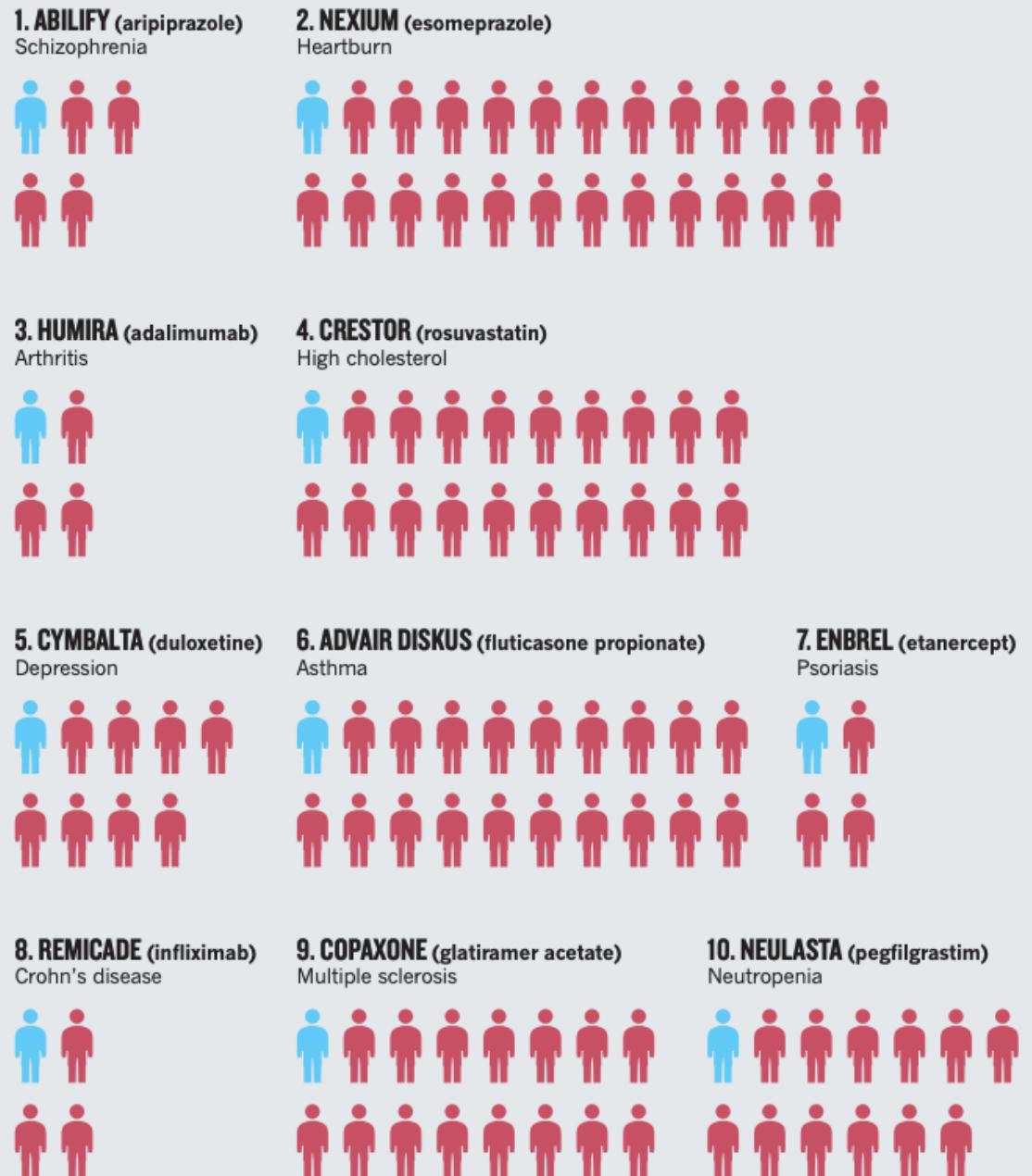
- different disease risk
- respond differently to medication
- different side effects



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.

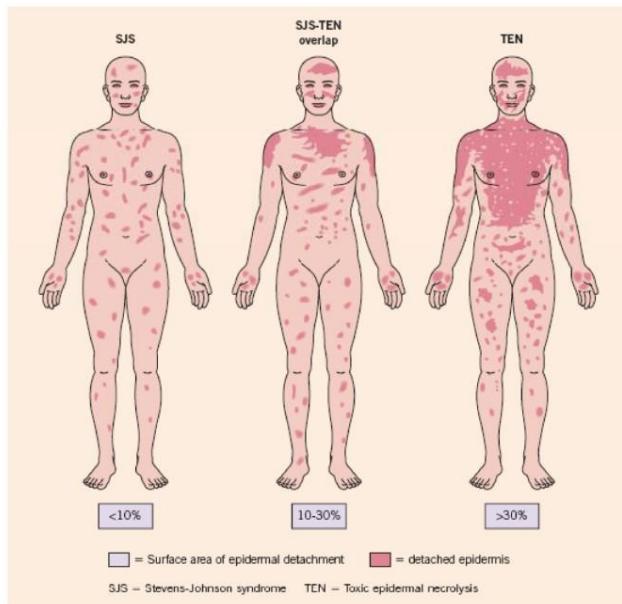


ADVERSE DRUG REACTIONS

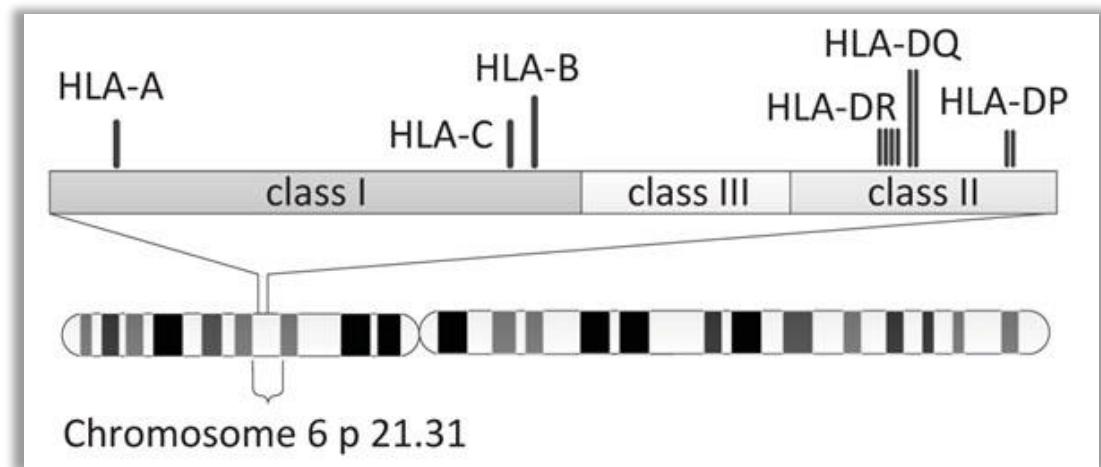


Some medications are directly harmful for certain individuals

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

A 55-YEAR-OLD MAN IN PRIMARY PREVENTION

Male, 55 years

LDL cholesterol: 4.2 mmol/L

Blood pressure: 145/90 mmHg

Former smoker

Family history of coronary artery disease

Genetic predisposition for coronary artery disease: high

Previous muscle-related side effects to medication

Patient prefers to avoid daily medication

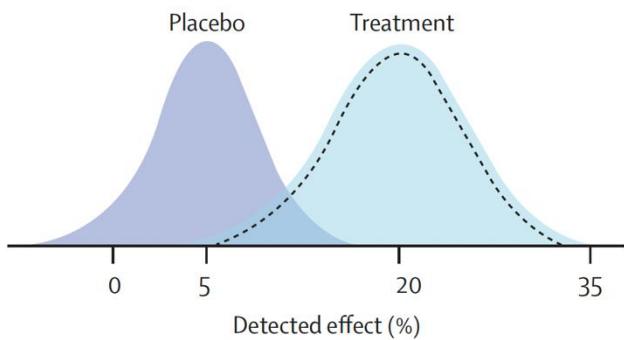
What would you do?

How should the decision about statin therapy be made for this patient?

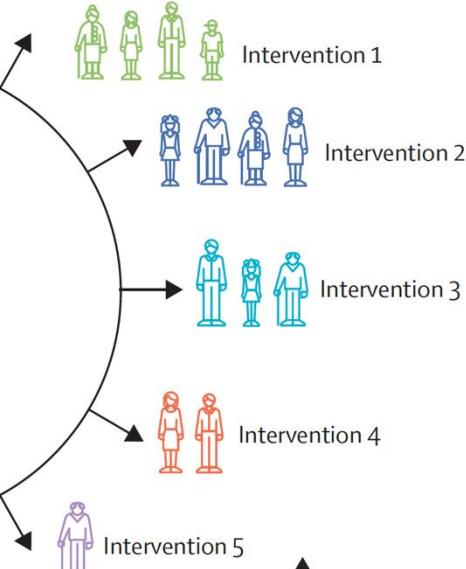
There is more than one reasonable approach

The difference lies not in the data - but in how decisions are made

Evidence-based Medicine



Precision Medicine



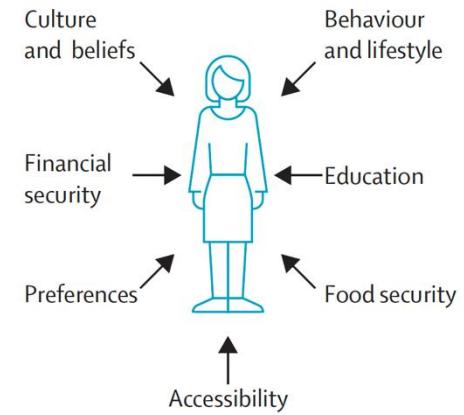
Personalised Medicine



High error

Low error

Individualised Medicine



YOUR TURN

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

Genes

Genomics



Transcripts

Transcriptomics



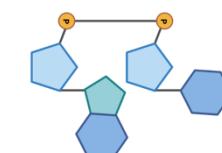
Protein

Proteomics



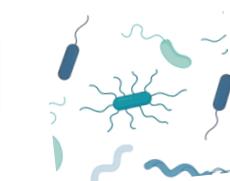
Metabolites

Metabolomics



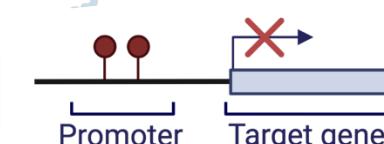
E.g., gut bacteria

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^{uk}
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 input fields. Below the header is a blue navigation bar with 'Public Health' in white. Underneath, a breadcrumb trail shows the path: European Commission > Public Health > Medicinal products > Personalised medicine. The main title 'Personalised medicine' is in large bold black font. To the left of the main content area is a sidebar with 'PAGE CONTENTS' and links to 'Legal Framework', 'Latest updates', and 'Documents'. The main content area contains a paragraph defining 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 detailed definition follows: '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.' A green box on the right labeled 'Precision medicine' contains the text: 'Personalised Medicine should be seen as an evolution of medicine, rather than a revolution, and many challenges remain before its successful application across healthcare systems.'

PAGE CONTENTS

[Legal Framework](#)

[Latest updates](#)

[Documents](#)

Personalised medicine

[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](#) (EN | ***), 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

Personalised Medicine should be seen as an evolution of medicine, rather than a revolution, and many challenges remain before its successful application across healthcare systems.

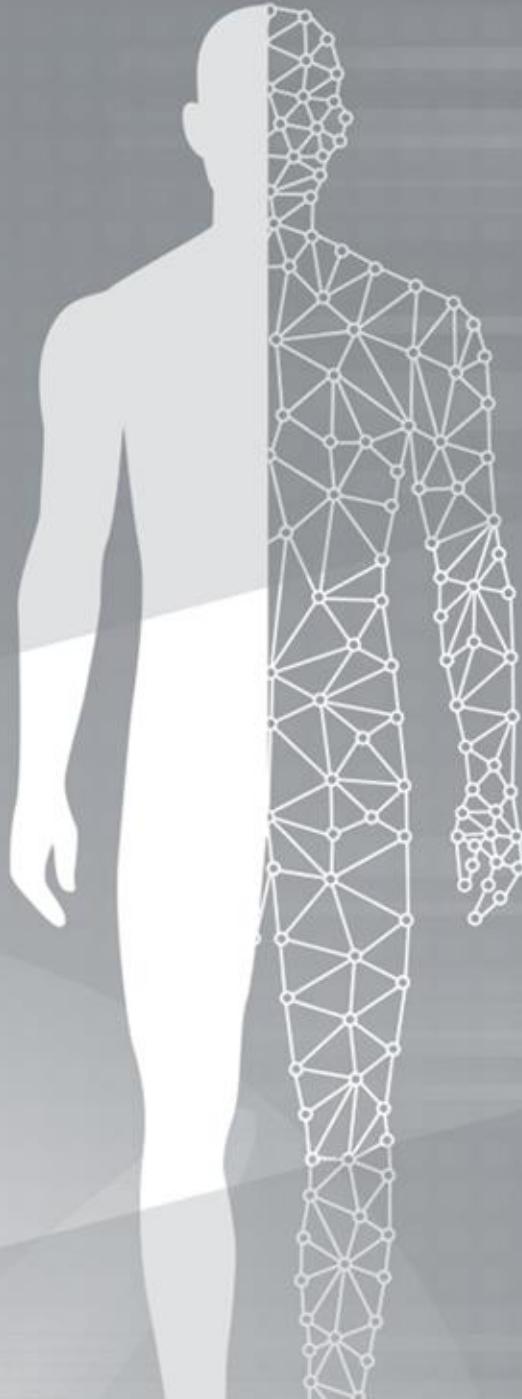
(1) Take a look at EUs definition on Personalised medicine https://ec.europa.eu/health/medicinal-products/personalised-medicine_en

(2) How does this agree with our definition Evidence – Precision – Personalised or Individualised ?

SUMMARY

PERSONALISED MEDICINE

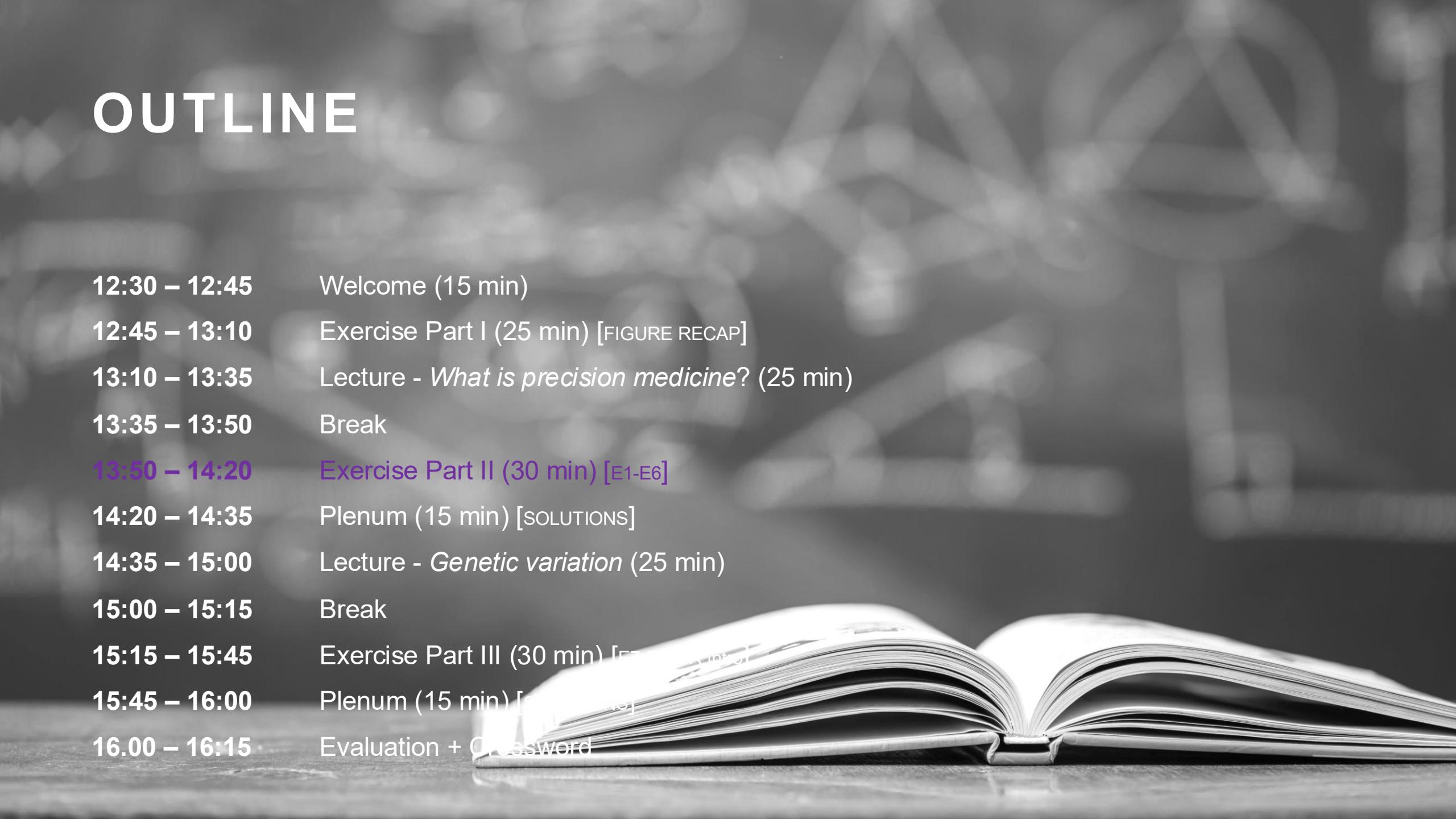
- What is Personalised Medicine?
- Why do we need it?
- Why so much focus on genetics?





BREAK

OUTLINE

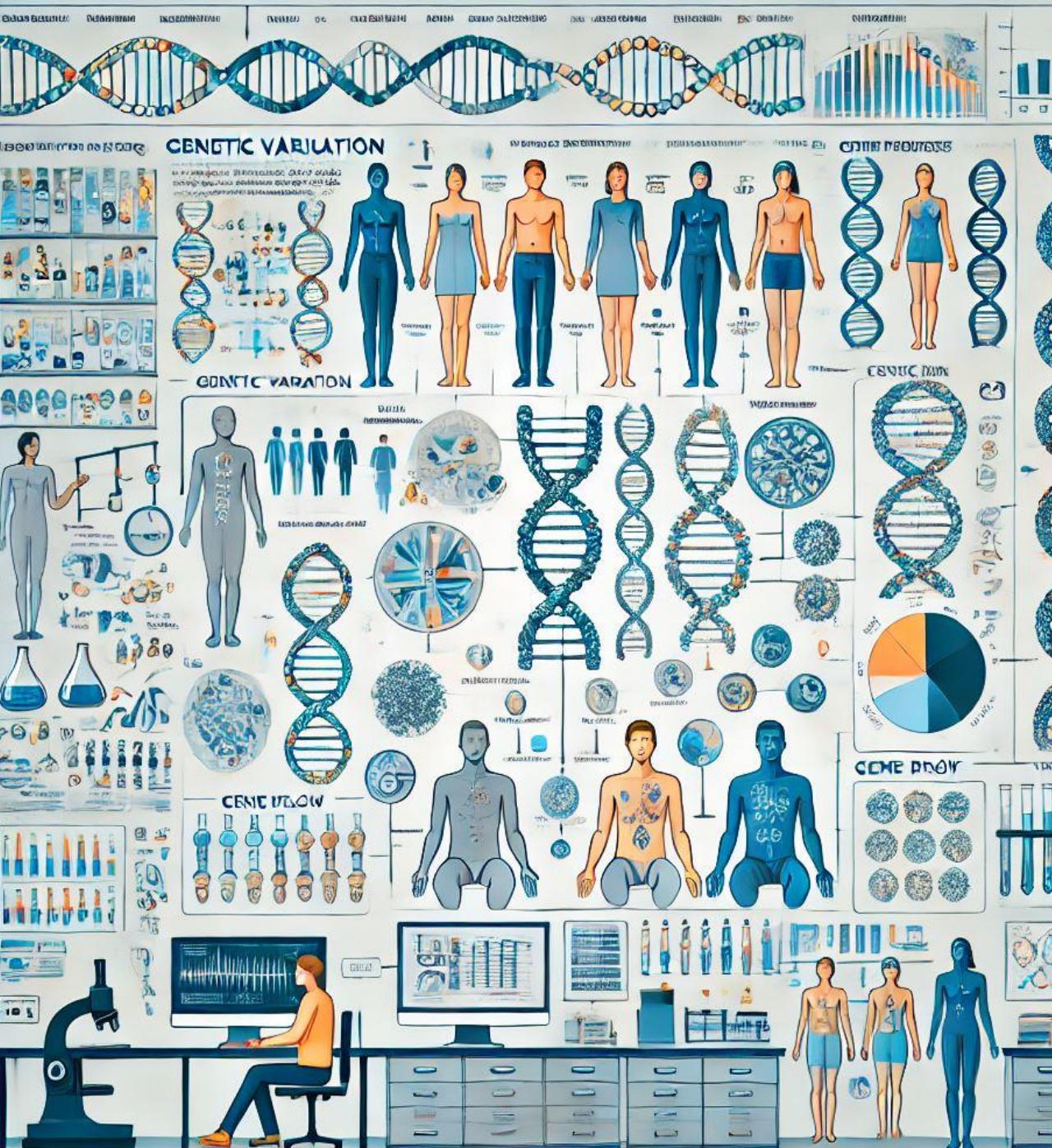
- 
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 - 13:10 – 13:35** Lecture - *What is precision medicine?* (25 min)
 - 13:35 – 13:50** Break
 - 13:50 – 14:20** Exercise Part II (30 min) [E1-E6]
 - 14:20 – 14:35** Plenum (15 min) [SOLUTIONS]
 - 14:35 – 15:00** Lecture - *Genetic variation* (25 min)
 - 15:00 – 15:15** Break
 - 15:15 – 15:45** Exercise Part III (30 min) [E7-E12]
 - 15:45 – 16:00** Plenum (15 min) [DISCUSSION]
 - 16.00 – 16:15** Evaluation + Crossword

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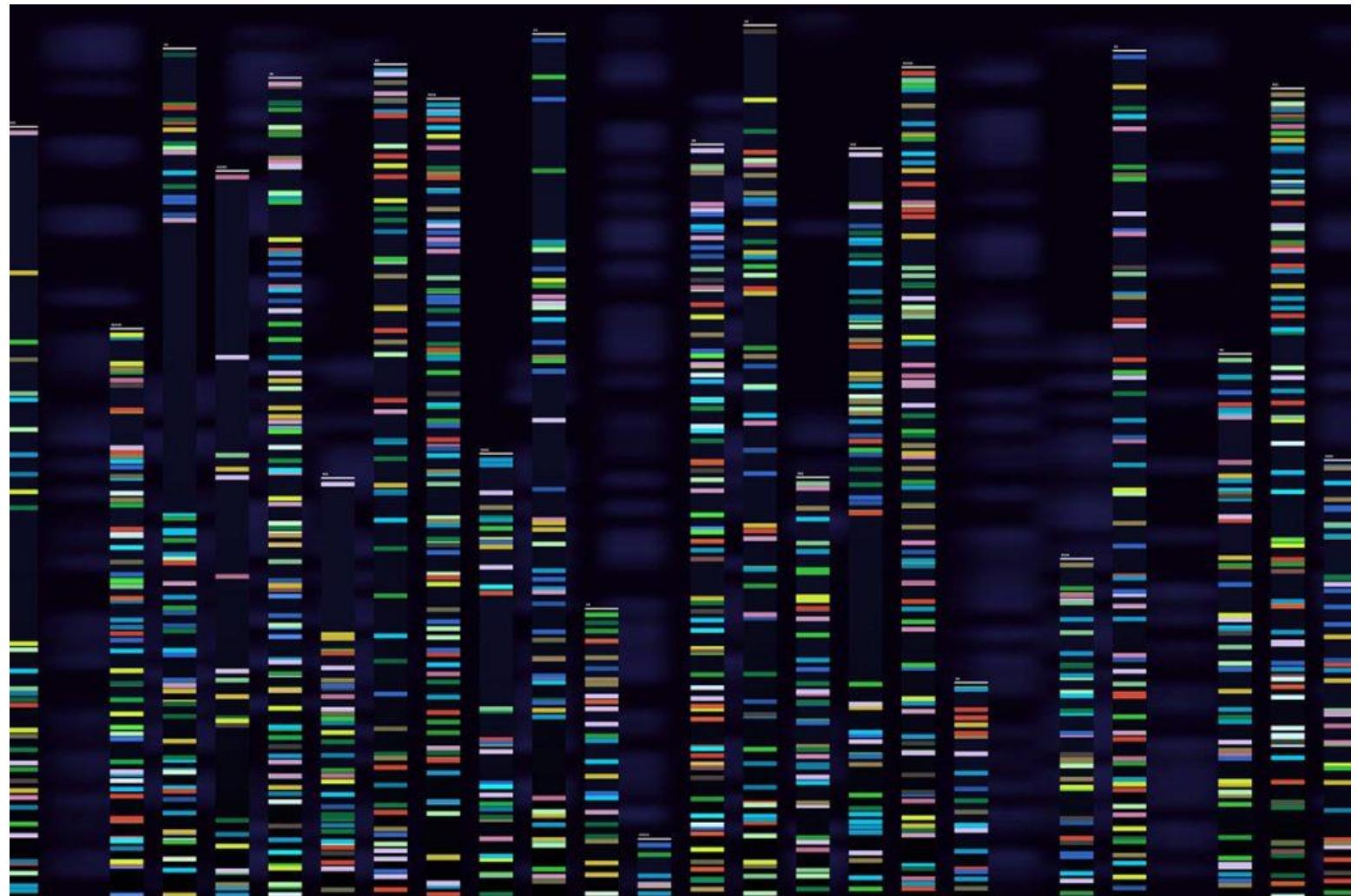
GENETIC VARIATION

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

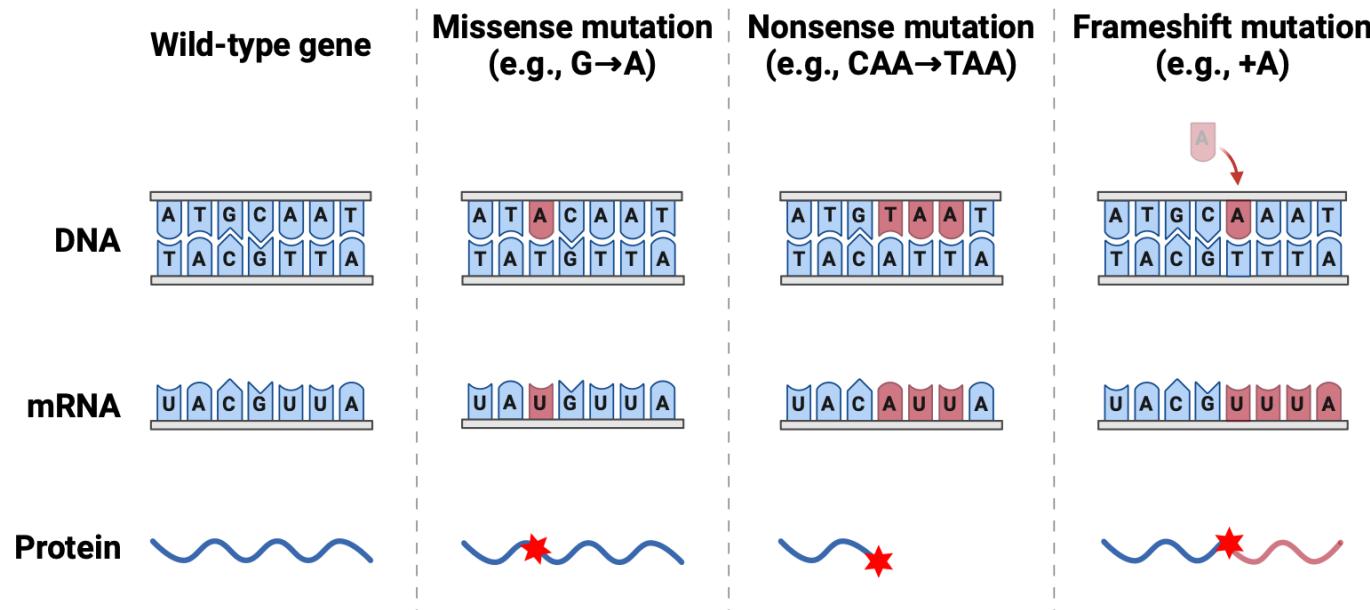


YOUR TURN

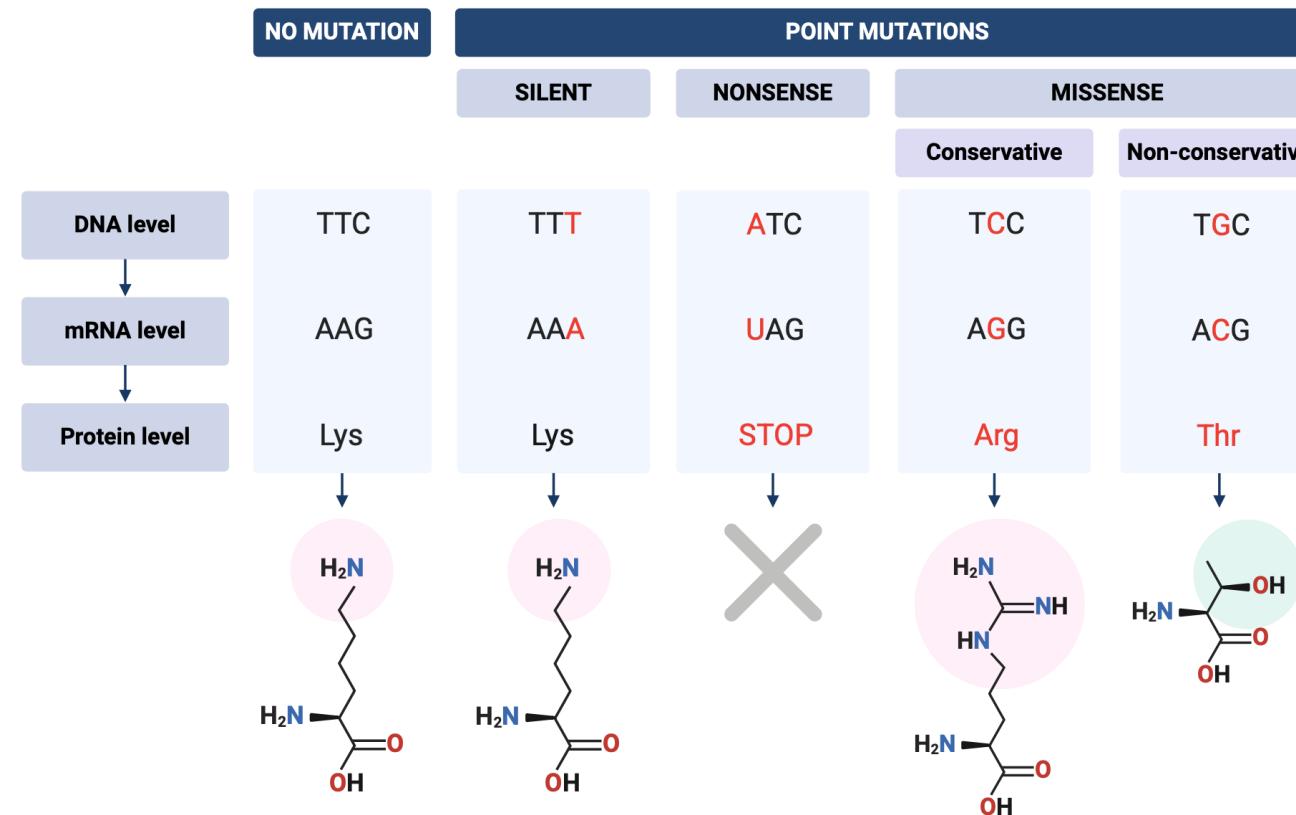
What generates genetic variation?



MUTATIONS GENERATE GENETIC VARIATION



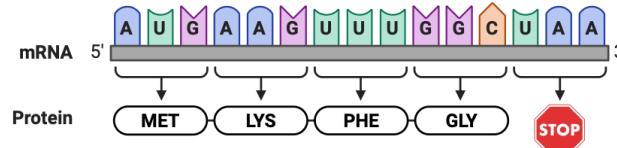
MUTATIONS GENERATE GENETIC VARIATION



MUTATIONS GENERATE GENETIC VARIATION

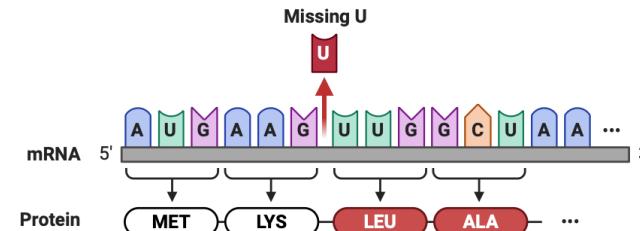
Wild type

mRNA sequence without any mutation



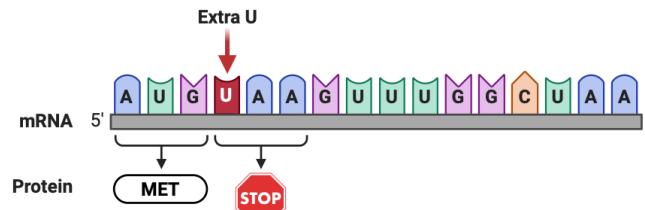
Base-pair deletion

Frameshift causing extensive missense



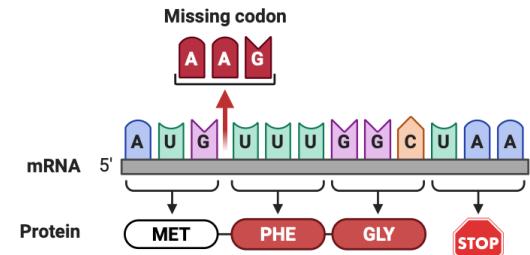
Base-pair insertion

Frameshift causing immediate nonsense



Three-nucleotide insertion/deletion

Extra/missing amino acids



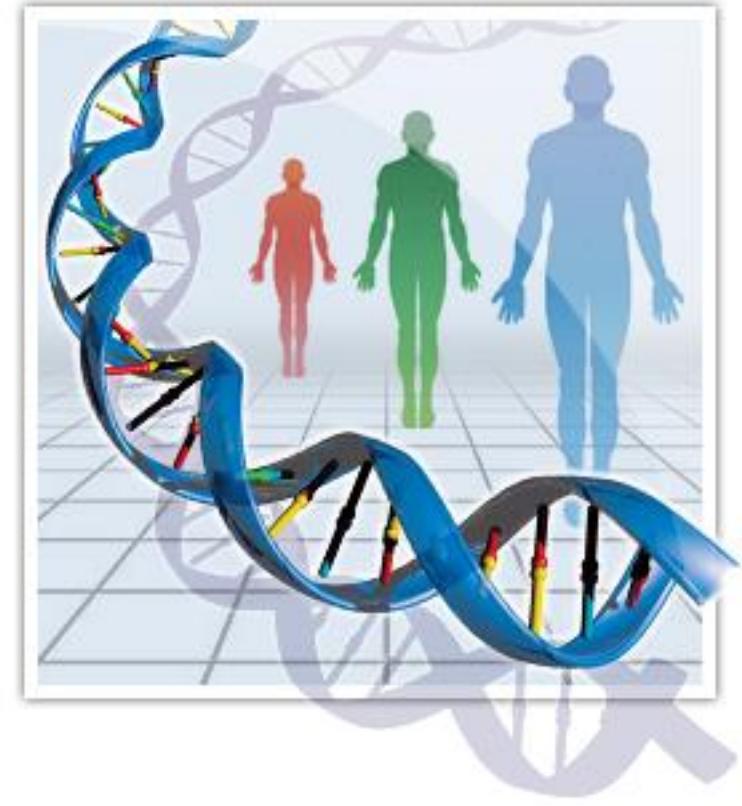
GENETIC DIVERSITY

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.



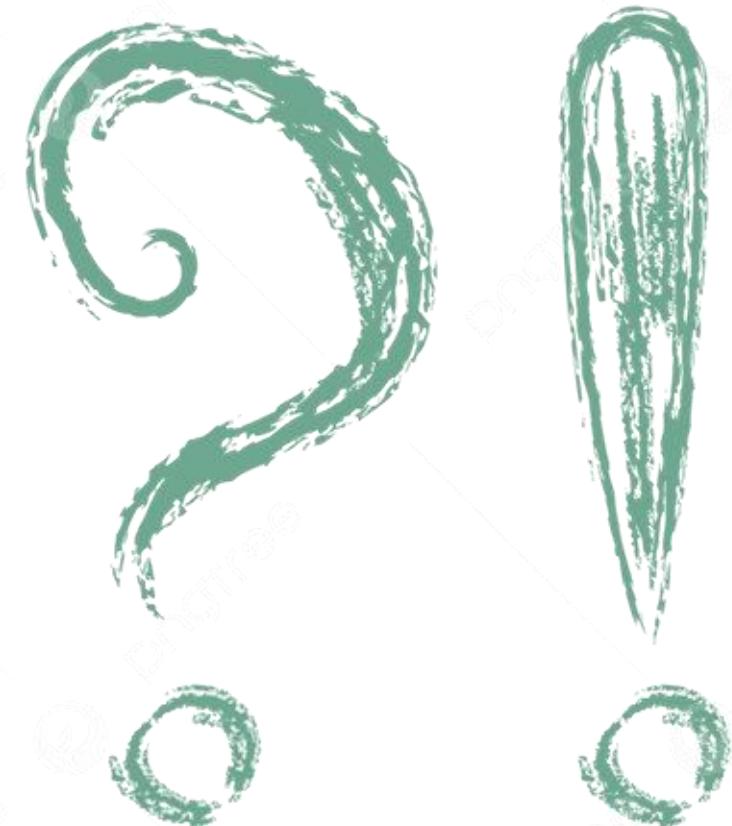
GENETIC TERMINOLOGY 1/2

- ❖ 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**.

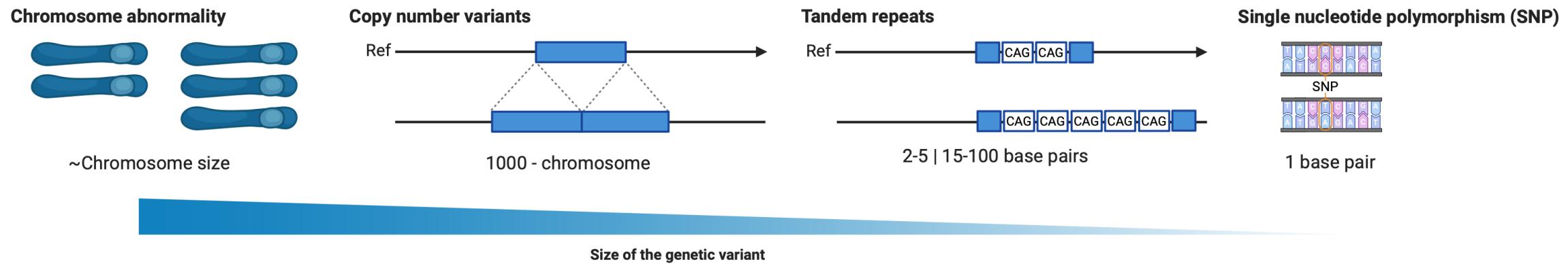


GENETIC TERMINOLOGY 2/2

- ❖ 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**)



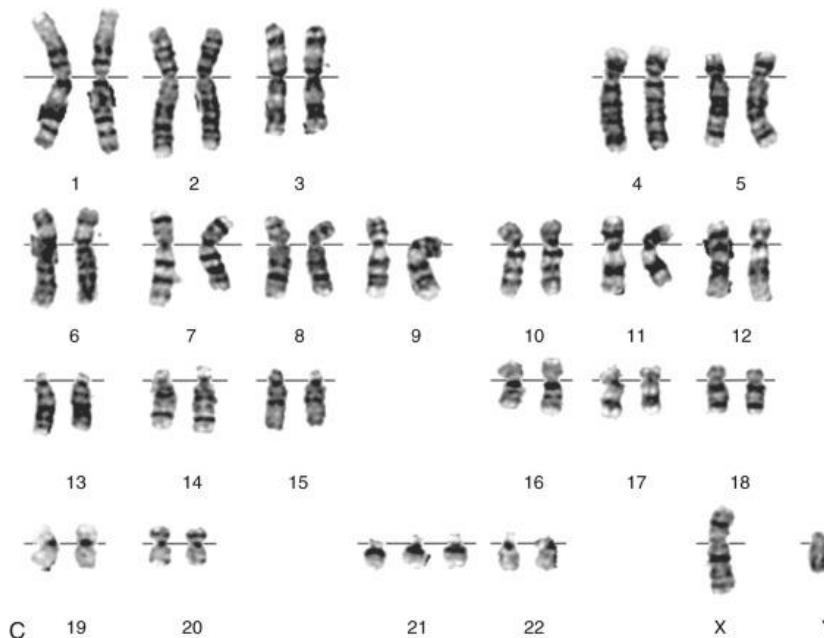
SIZES OF GENETIC VARIATION



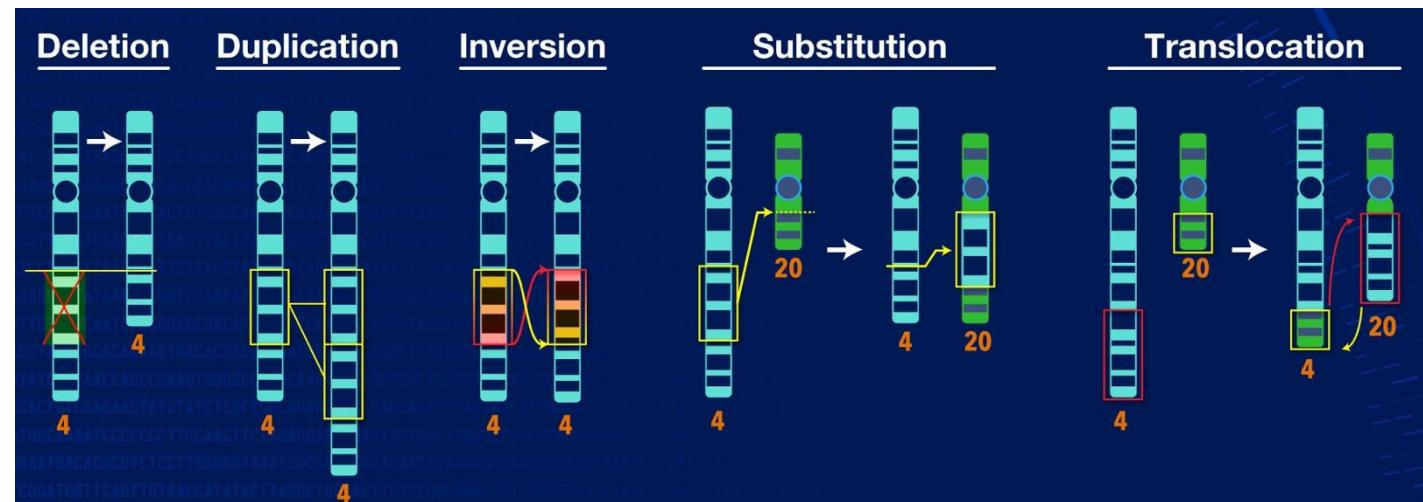
GENETIC VARIATION

CHROMOSOME ABNORMALITIES

Chromosome abnormalities can be numerical or structural.



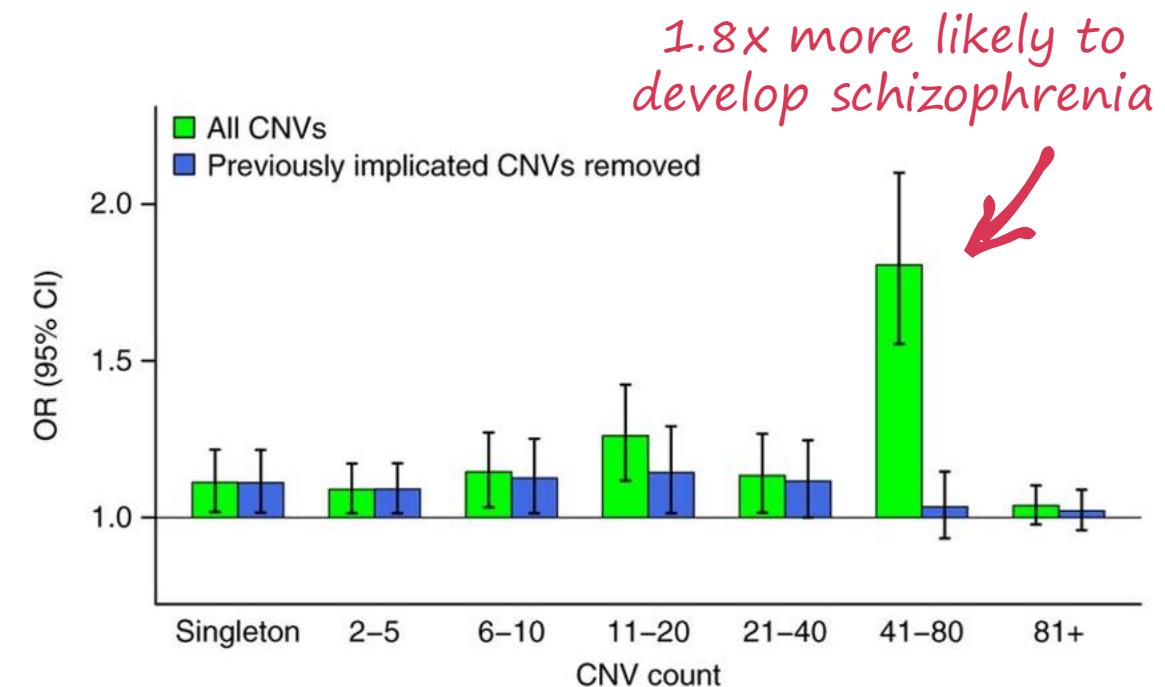
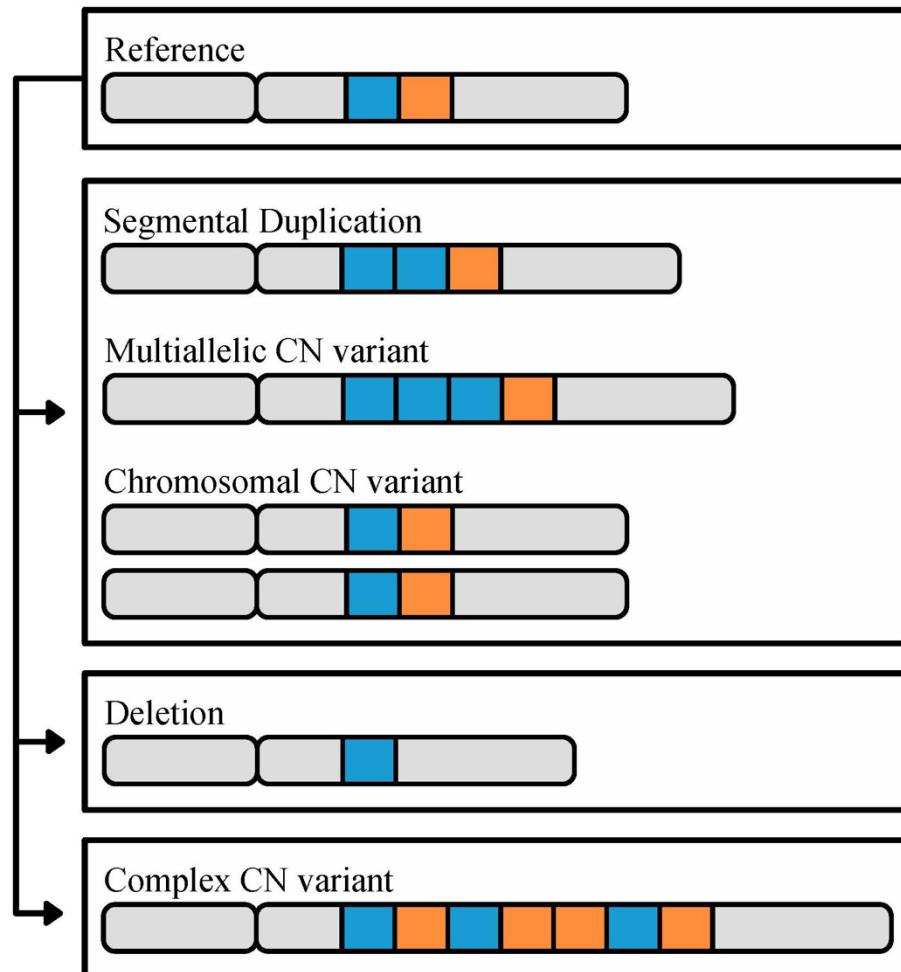
Trisomi 21



GENETIC VARIATION

COPY NUMBER VARIANTS (CNV)

50 bp - kromosom



1.8x more likely to develop schizophrenia

GENETIC VARIATION

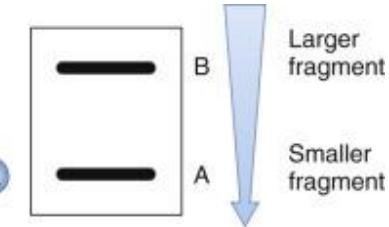
TANDEM REPEATS



Advantage:
many many
alleles exists

Micro satellites
(2-5 bp) - STRs

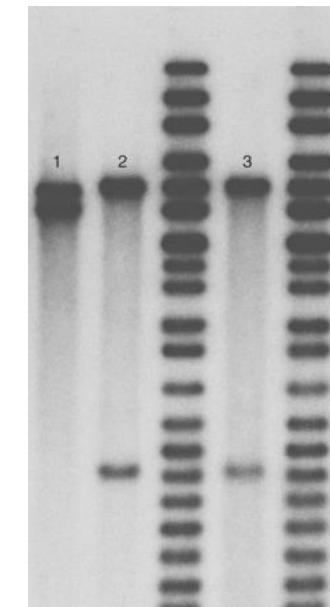
Mini satellites
(15-100 bp) -
VNTR



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
	9 repeats
	10 repeats

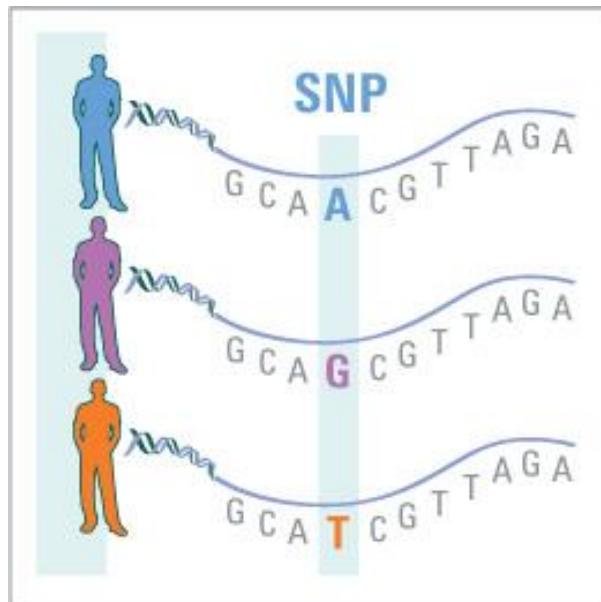


GENETIC VARIATION

SINGLE NUCLEOTIDE POLYMORPHISMS (SNPs)

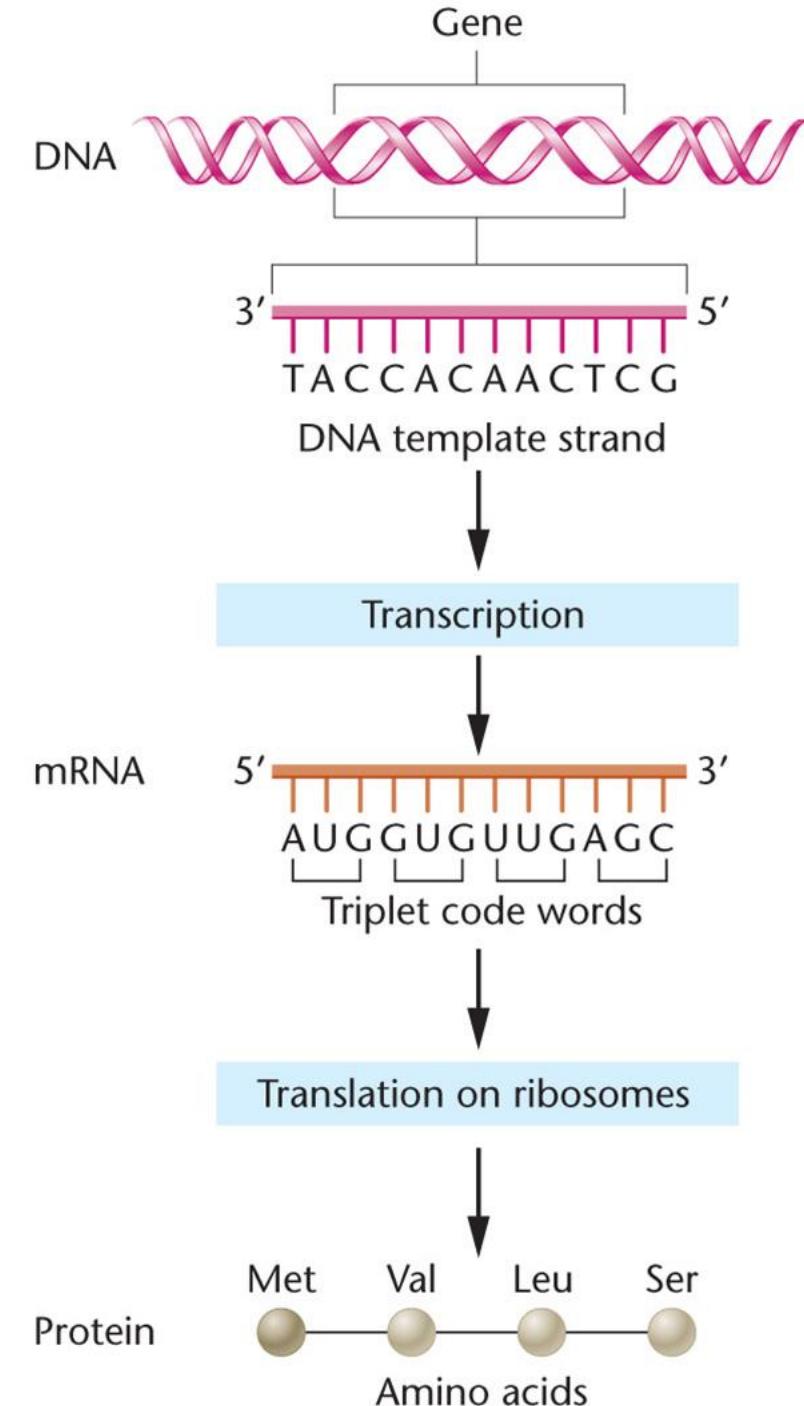
A common change in a single base pair; ~1/1000 bp

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

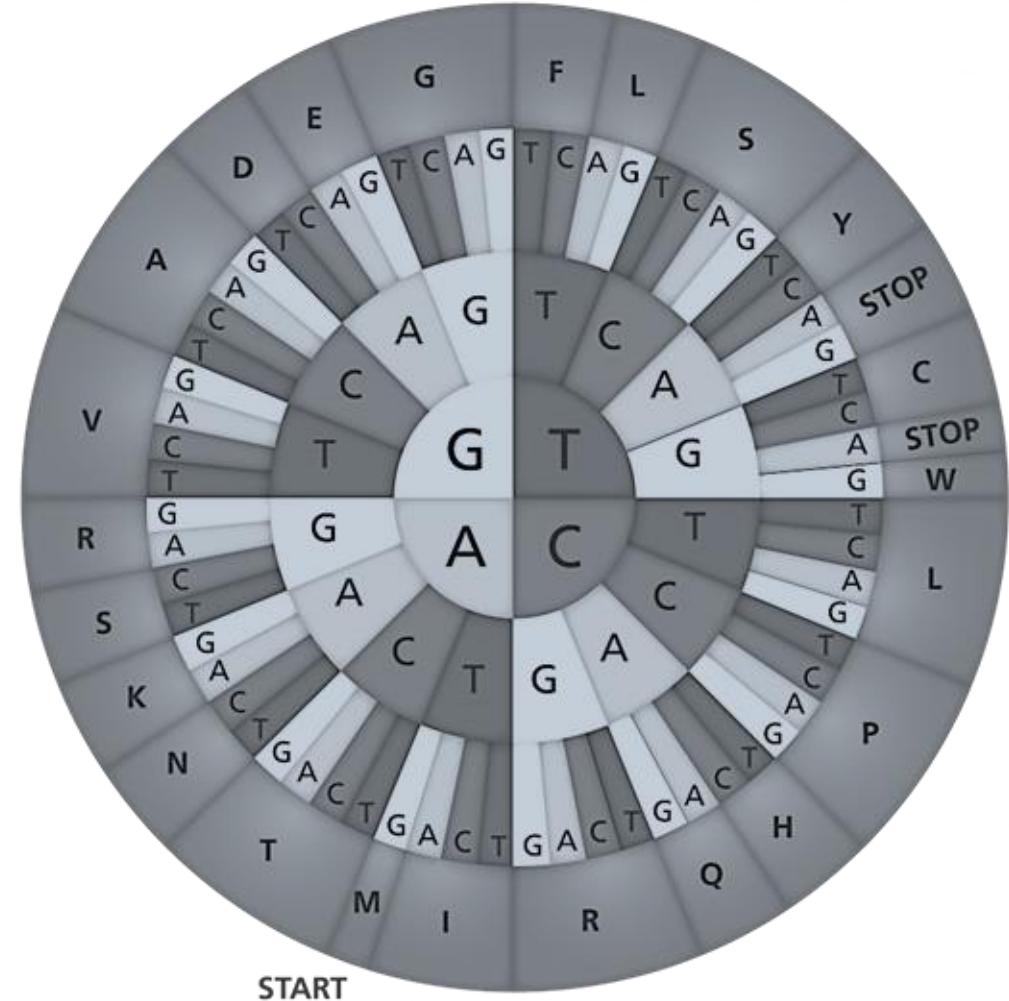
Genetic variation arise as a consequence
of changes in the DNA sequence



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 β -GLOBIN

DNA.....	TGA	GGA	CTC	CTC.....
mRNA.....	ACU	CCU	GAG	GAG.....
Amino acid.....	Thr	Pro	Glu	Glu.....

4 5 6 7

MUTANT β -GLOBIN

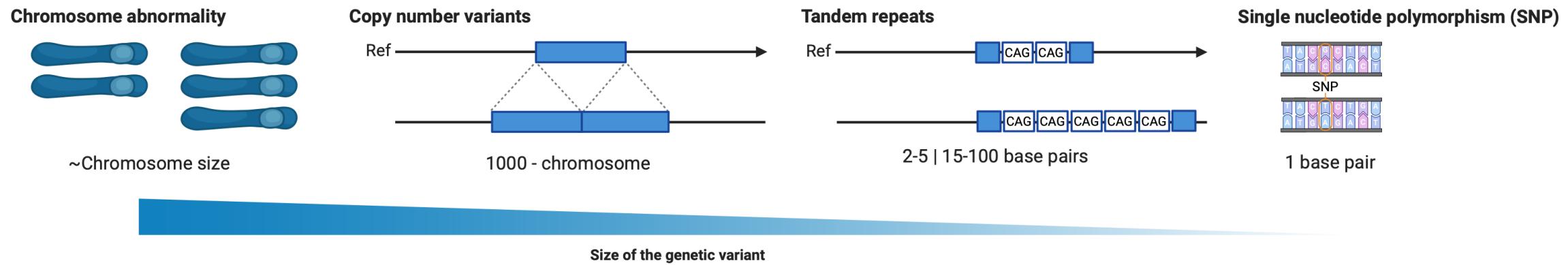
DNA.....	TGA	GGA	CAC	CTC.....
mRNA.....	ACU	CCU	GUG	GAG.....
Amino acid.....	Thr	Pro	Val	Glu.....

4 5 6 7

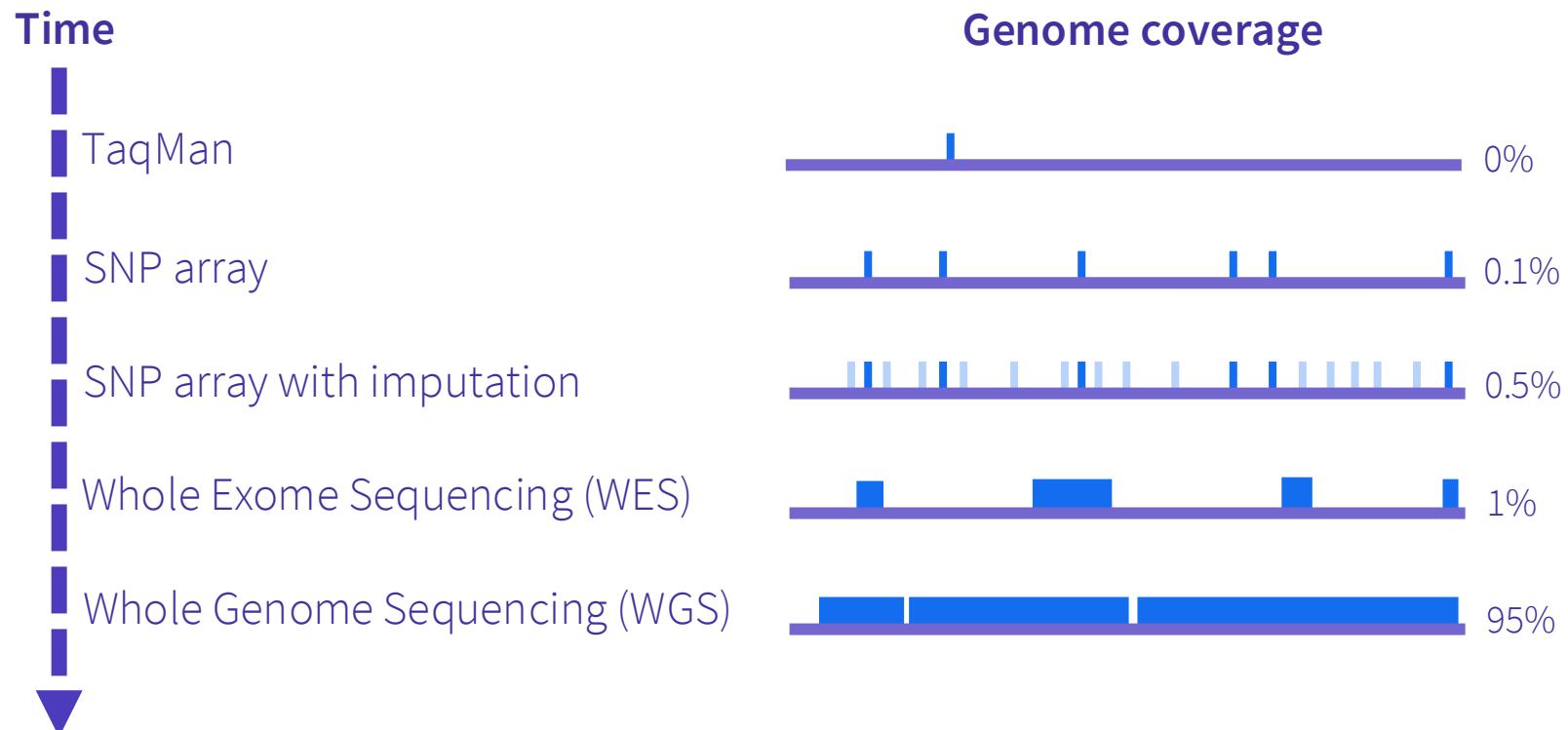
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SIZES OF GENETIC VARIATION



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

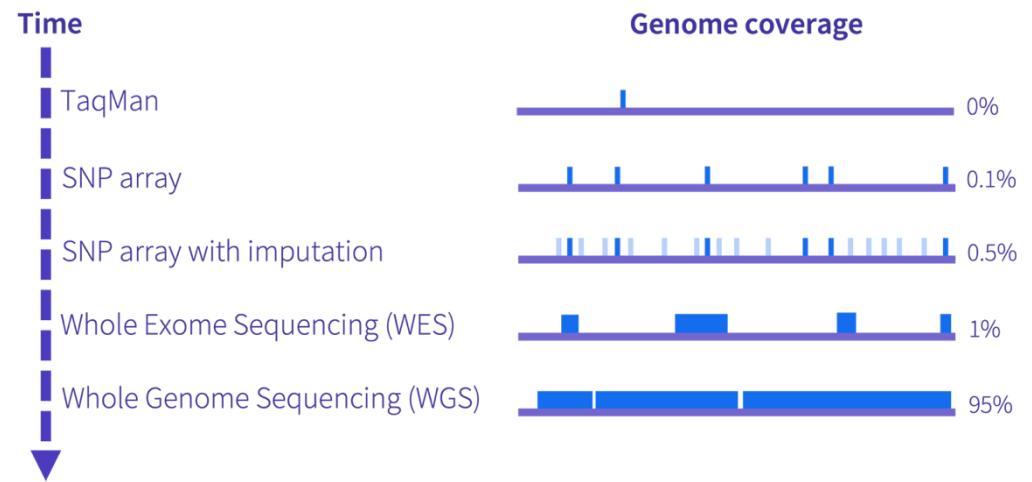
Genotyping

WES

WGS

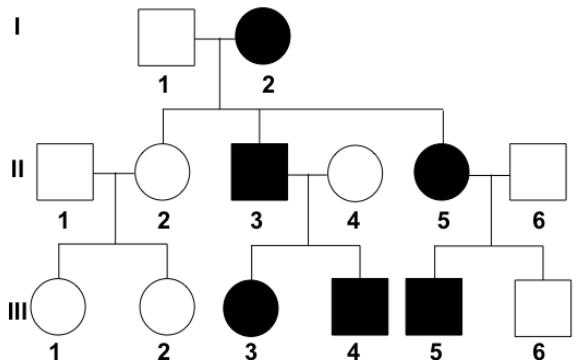
WHICH TECHNOLOGY?

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



THE GENETIC AETIOLOGY DETERMINES THE STATISTICAL APPROACH

Monogenic disorders



**A single variant with a large effect
is often sufficient to cause disease**
(high penetrance, clear inheritance patterns)

→ Family-based analysis / sequencing

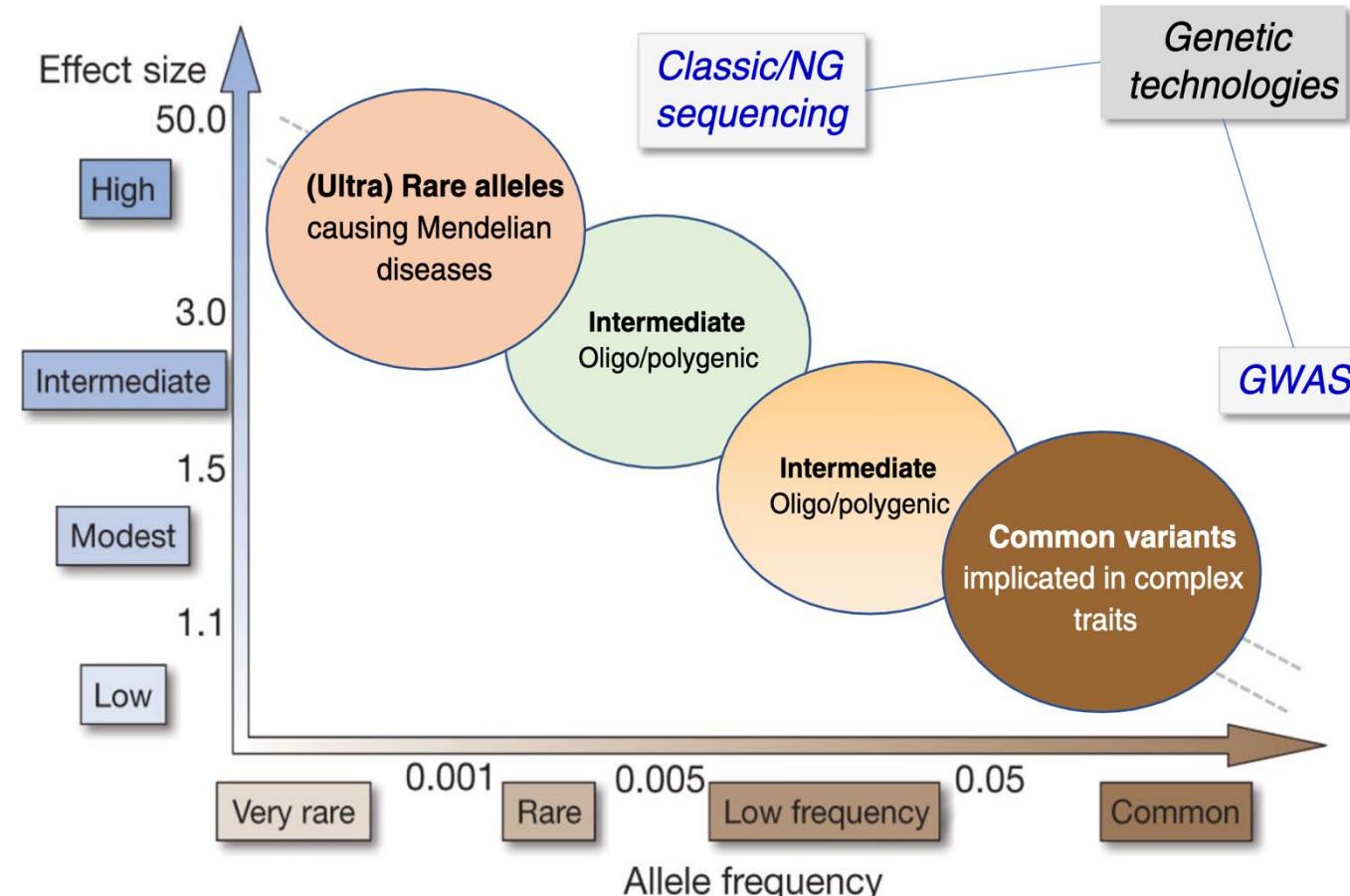
Common complex diseases



**Many variants, each with small effects
contribute to disease risk together with
environment and lifestyle**

→ Population-based studies

THE RELATIONSHIP BETWEEN FREQUENCY AND EFFECT SIZE

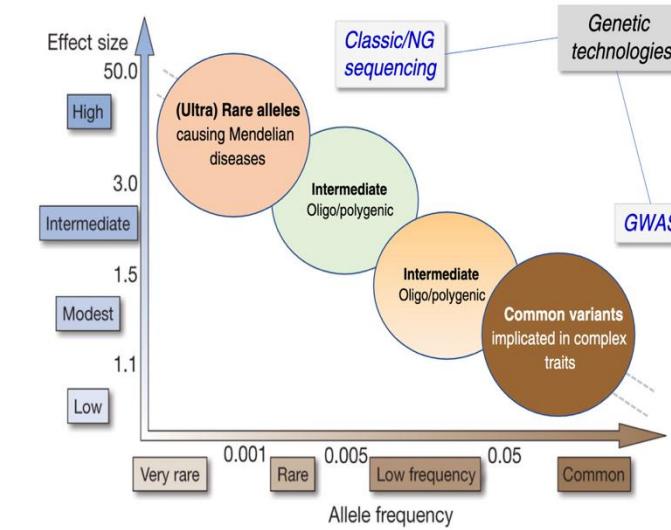
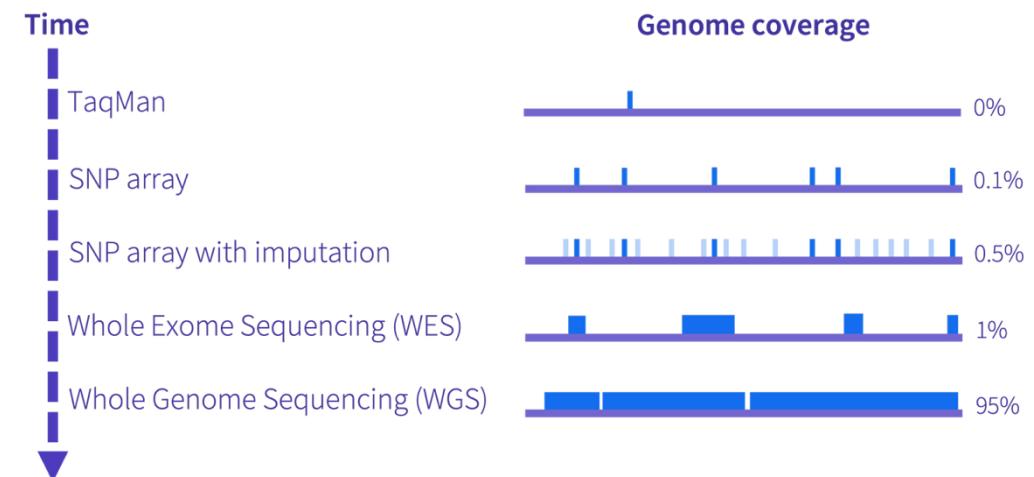


WHICH TECHNOLOGY?

The choice of technology to detect single nucleotide polymorphisms (SNPs) depends upon the application.

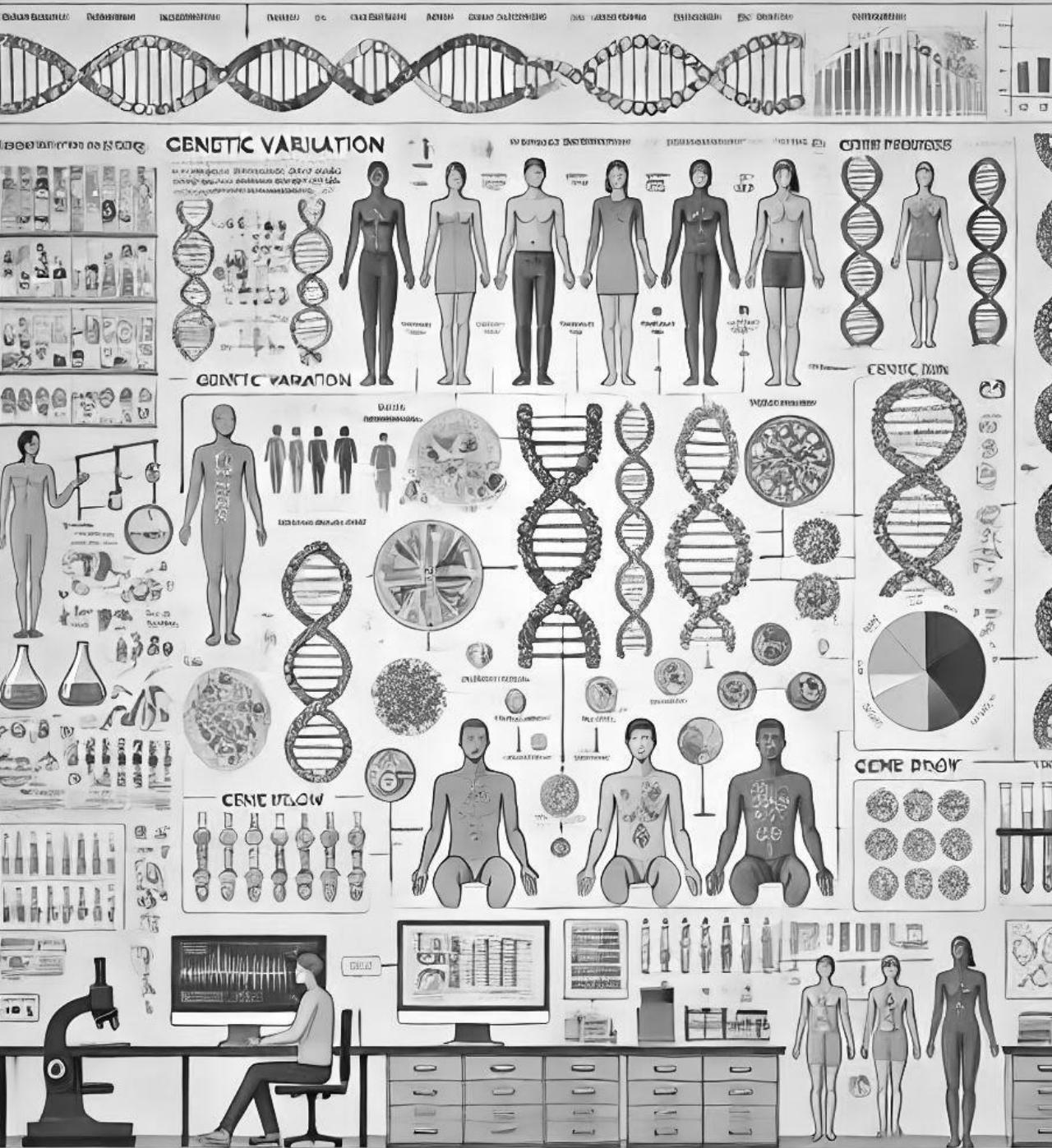
For Clinical utility **WES/WGS is preferred**

For GWAS and PGS **genotyping is preferred**



SUMMARY GENETIC VARIATION

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





BREAK

OUTLINE

12:30 – 12:45	Welcome (15 min)
12:45 – 13:10	Exercise Part I (25 min) [FIGURE RECAP]
13:10 – 13:35	Lecture - <i>What is precision medicine?</i> (25 min)
13:35 – 13:50	Break
13:50 – 14:20	Exercise Part II (30 min) [E1-E6]
14:20 – 14:35	Plenum (15 min) [SOLUTIONS]
14:35 – 15:00	Lecture - <i>Genetic variation</i> (25 min)
15:00 – 15:15	Break
15:15 – 15:45	Exercise Part III (30 min) [E7-E9 + R intro]
15:45 – 16:00	Plenum (15 min) [SOLUTIONS]
16.00 – 16:15	Evaluation + Crossword

SUMMARY

- 10 sessions - read the readme - prepare before class – **this is important**

- Precision 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

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16.00 – 16:15	Evaluation + Crossword

GENOMICS CROSSWORD

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

Hint: Use “-” to indicate a space

