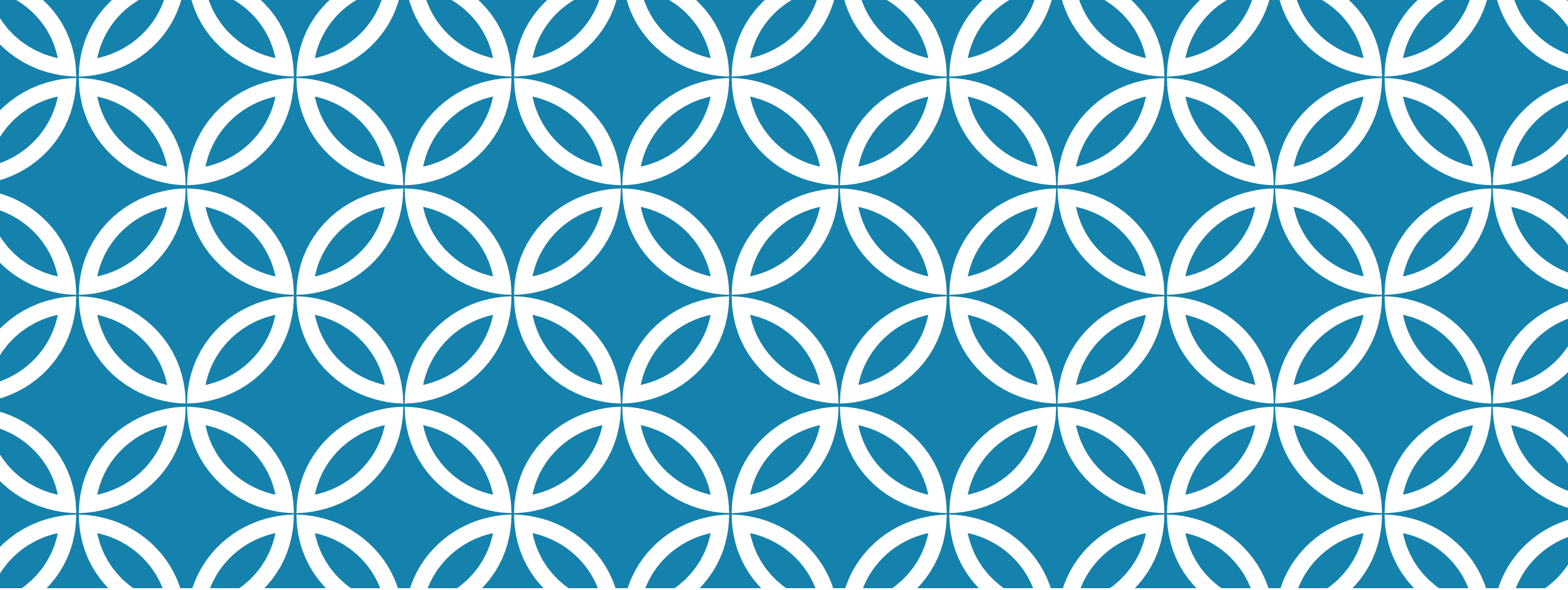


3.1 Genes

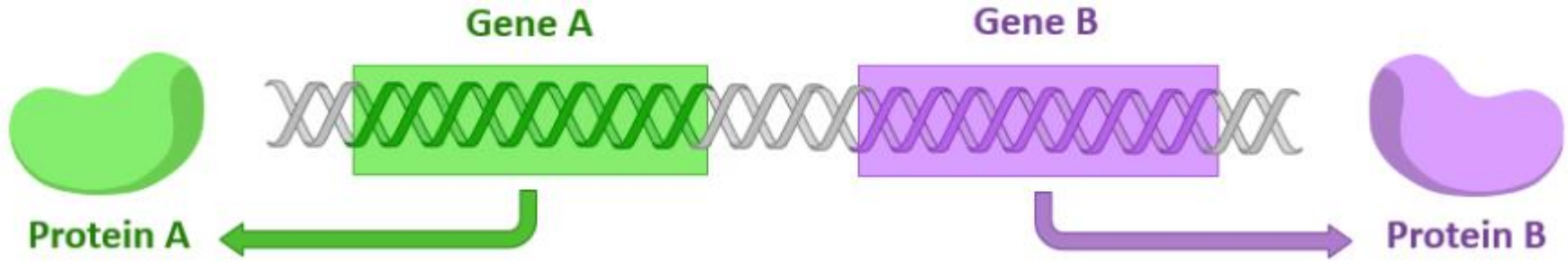
BASIC IDEA

DNA = genetic blueprint → codes for & determines an organism's characteristics

it encodes proteins, which carry out cellular processes

A specific length / sequence of DNA that encodes for a particular protein = **gene**

→ heritable factor that influences a specific characteristic



COMPARING NUMBERS OF GENES

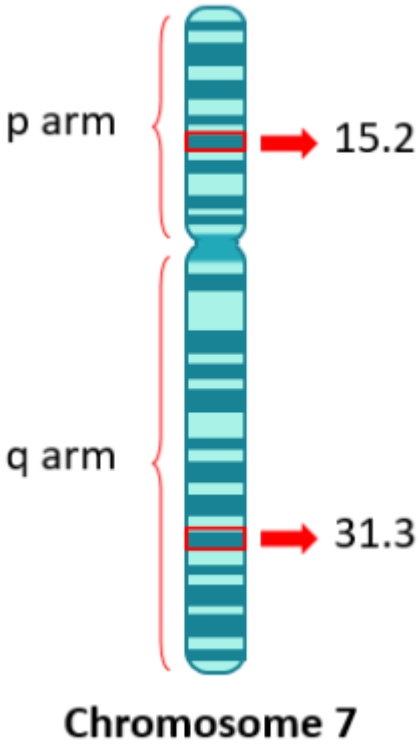
IB companion: p.142 orange box

GENE LOCI

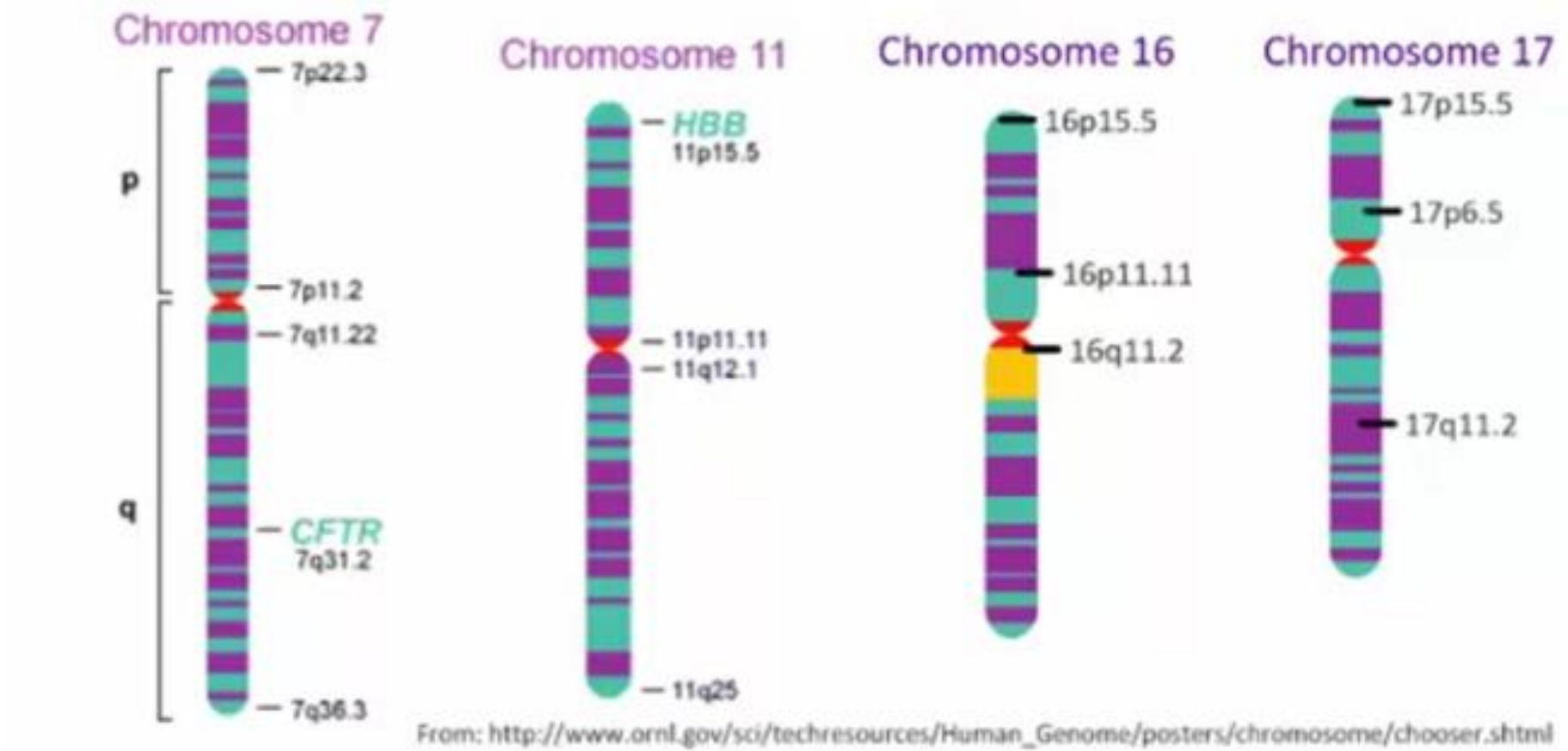
Position of a gene on a particular chromosome = **locus**
(plural = loci)

Eukaryotic gene loci identified to following features:

- chromosome number
- arm (p = short ; q = long)
- region and band

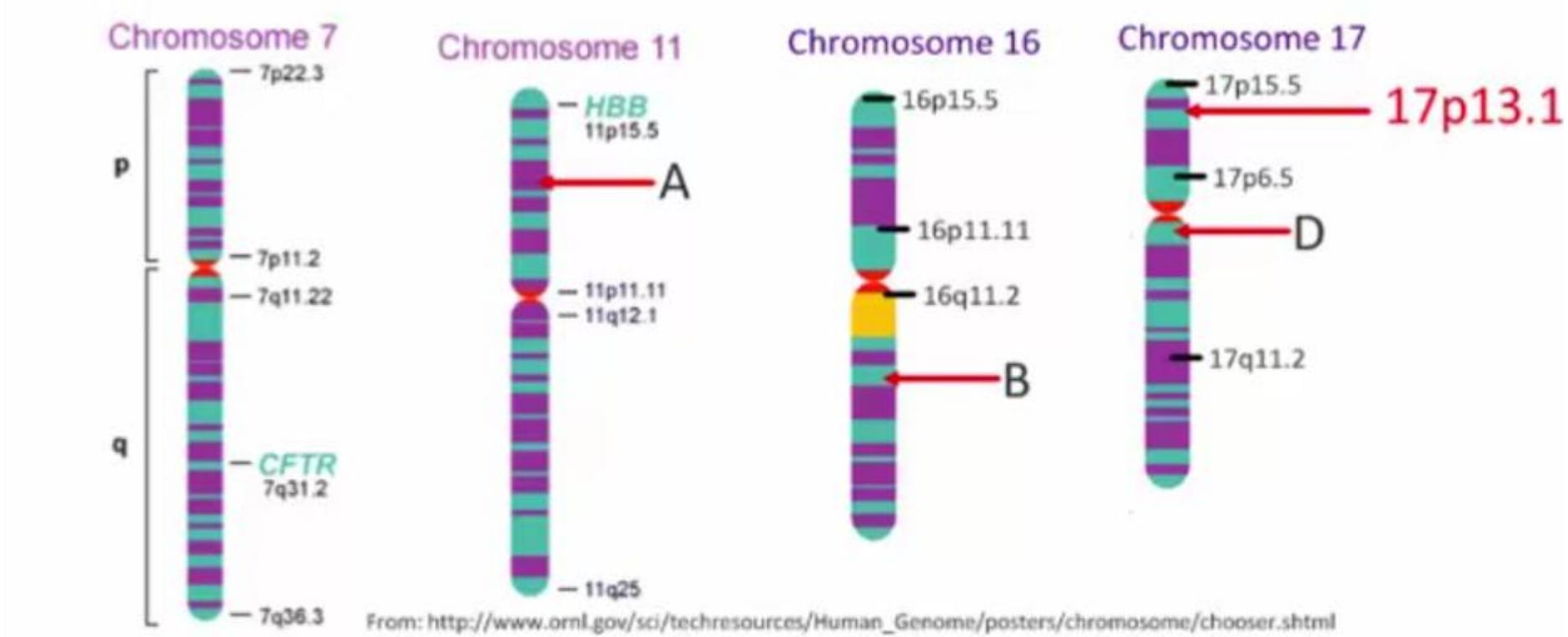


GENE LOCI: EXAMPLES



The gene TP53 (prevents tumours) is found at 17p13.1.

GENE LOCI: EXAMPLES



Match the gene loci with the letters

16q13.2

11p13.1

17q1.1

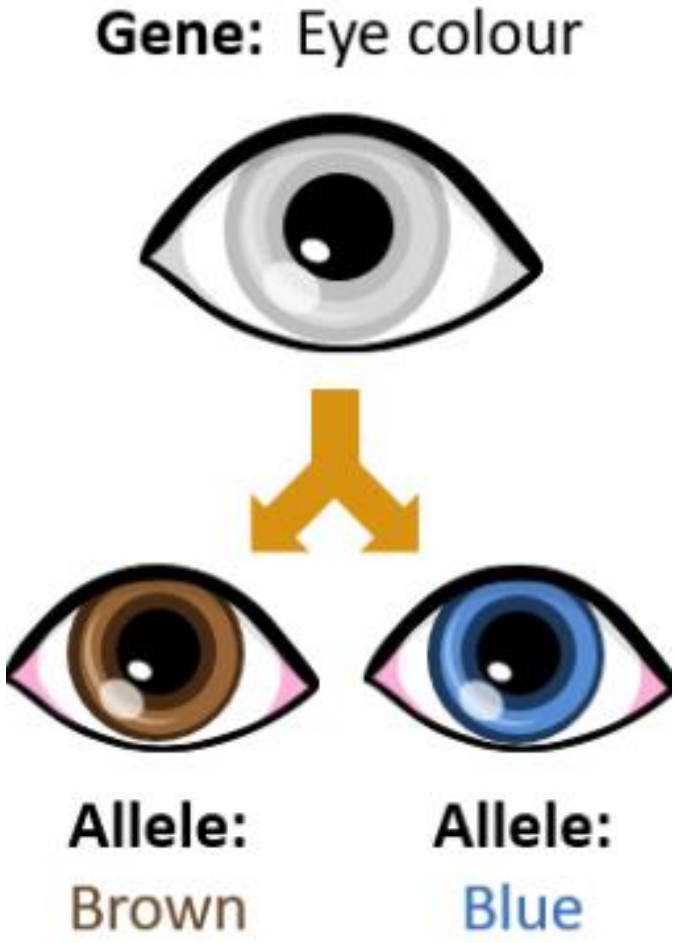
ALLELES

Alleles = different possible variations of a gene

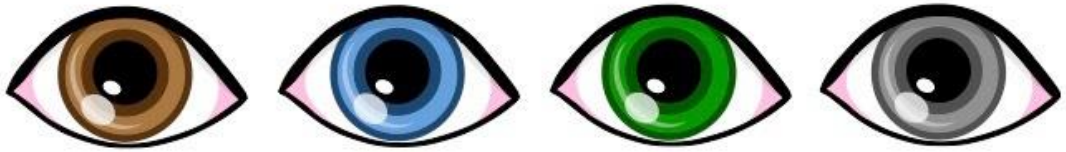
Genes encode a general characteristic: Eye color

Alleles encode for a specific alternative: Blue eyes

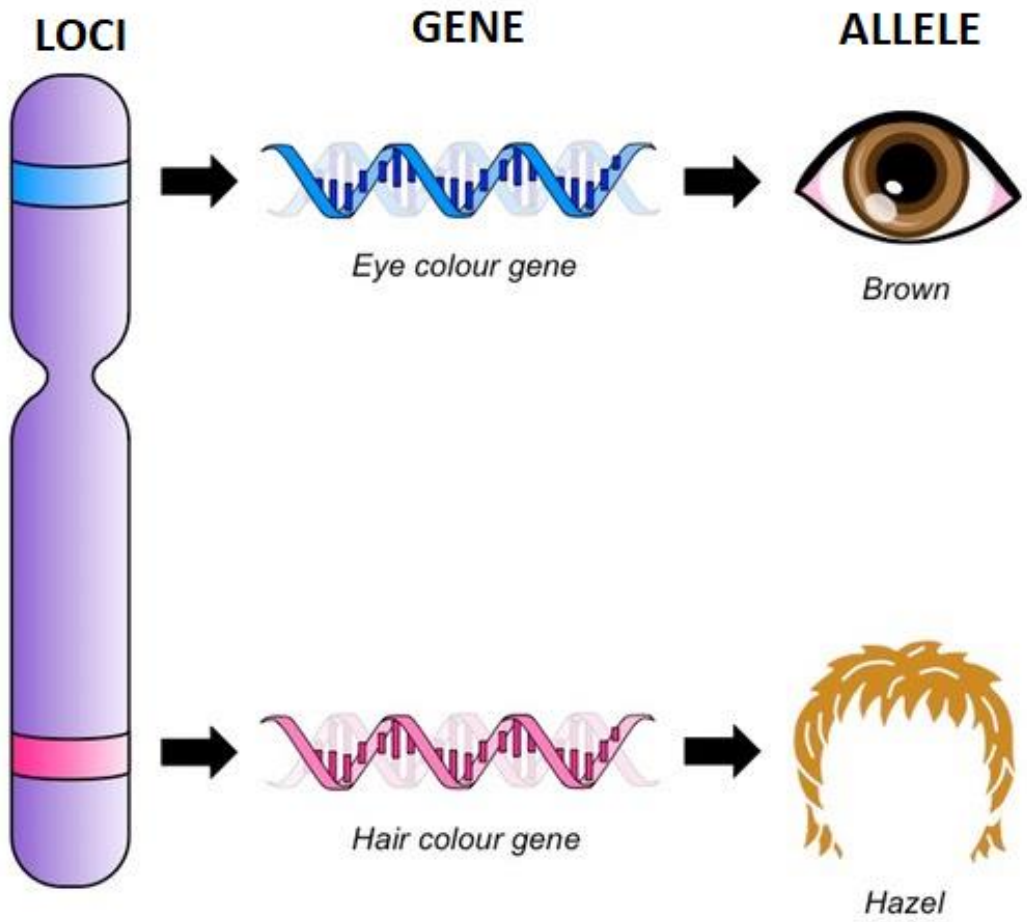
Alleles only differ from each other by one or few bases



ALLELES

Gene	Alternative Alleles
 Eye colour	 Brown Blue Emerald Grey
 Hair colour	 Blonde Red Brown Black

LOCUS VS GENE VS ALLELE



COX-2, SMOKING AND STOMACH CANCER

Data-based questions page 145

MUTATIONS

A **gene mutation** is a change in the nucleotide sequence of a section of DNA coding for a specific trait → may change a protein's function

Gene mutations can either be:

- **Somatic –** occur in body cells → not inherited
- **Germline –** occur in gametes → can be passed on to offspring

MUTATIONS

Gene mutations can be:

- Beneficial: new variations of a trait
- Detrimental: cannot carry out normal function
- Neutral: no effect on functioning

MUTAGEN

A mutagen is an agent that changes (mutates) the genetic material of an organism

Physical		Chemical			Biological	
						
UV *(from sun)*	X-rays *(medical)*	Carcinogen *(cigarettes)*	Processed foods	Cleaning products	Viruses *(HPV)*	Bacteria (*Helicobacter*)

TYPES OF MUTATIONS

Point mutations → modification of a single nucleotide within a sequence

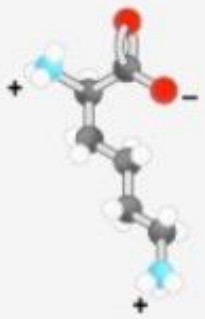
- **substitutions, insertions** or **deletions**

Substitution mutations can either cause:

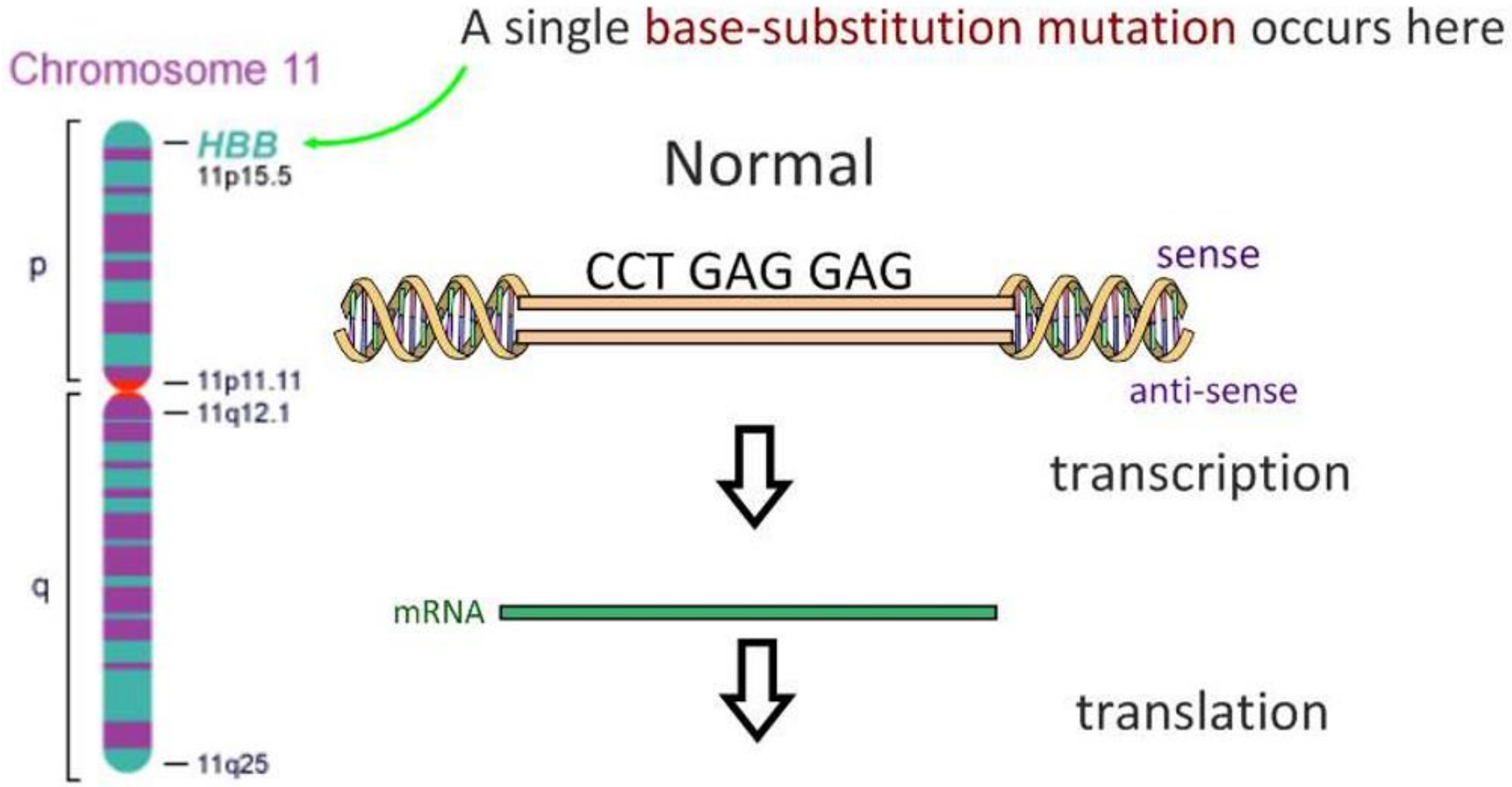
- *Silent mutations* – no change to sequence (*due to degeneracy*)
- *Missense mutations* – a signle amino acid is changed in the polypeptide
- *Nonsense mutations* – a stop codon is created, malfunction of polypeptide

Insertions and deletions cause frameshift mutations (*changes reading frame*)

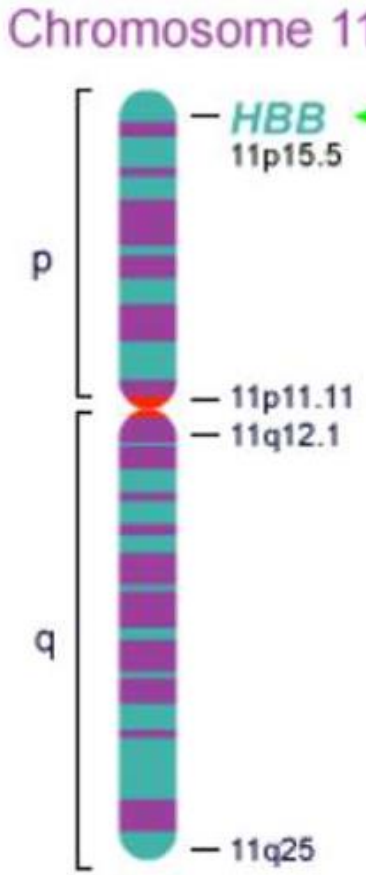
TYPES OF MUTATIONS:

	No mutation	Point mutations		
		Silent	Missense	Nonsense
DNA	TTC	TTT	TCC	ATC
mRNA	AAG	AAA	AGG	UAG
Protein	Lys	Lys	Arg	STOP
				

MUTATION EXAMPLE: SICKLE CELL



A single base-substitution mutation occurs here



Normal

CCT GAG GAG

sense

GGA CTC CTC

anti-sense

transcription

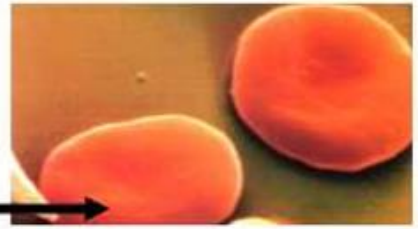
CCU GAG GAG

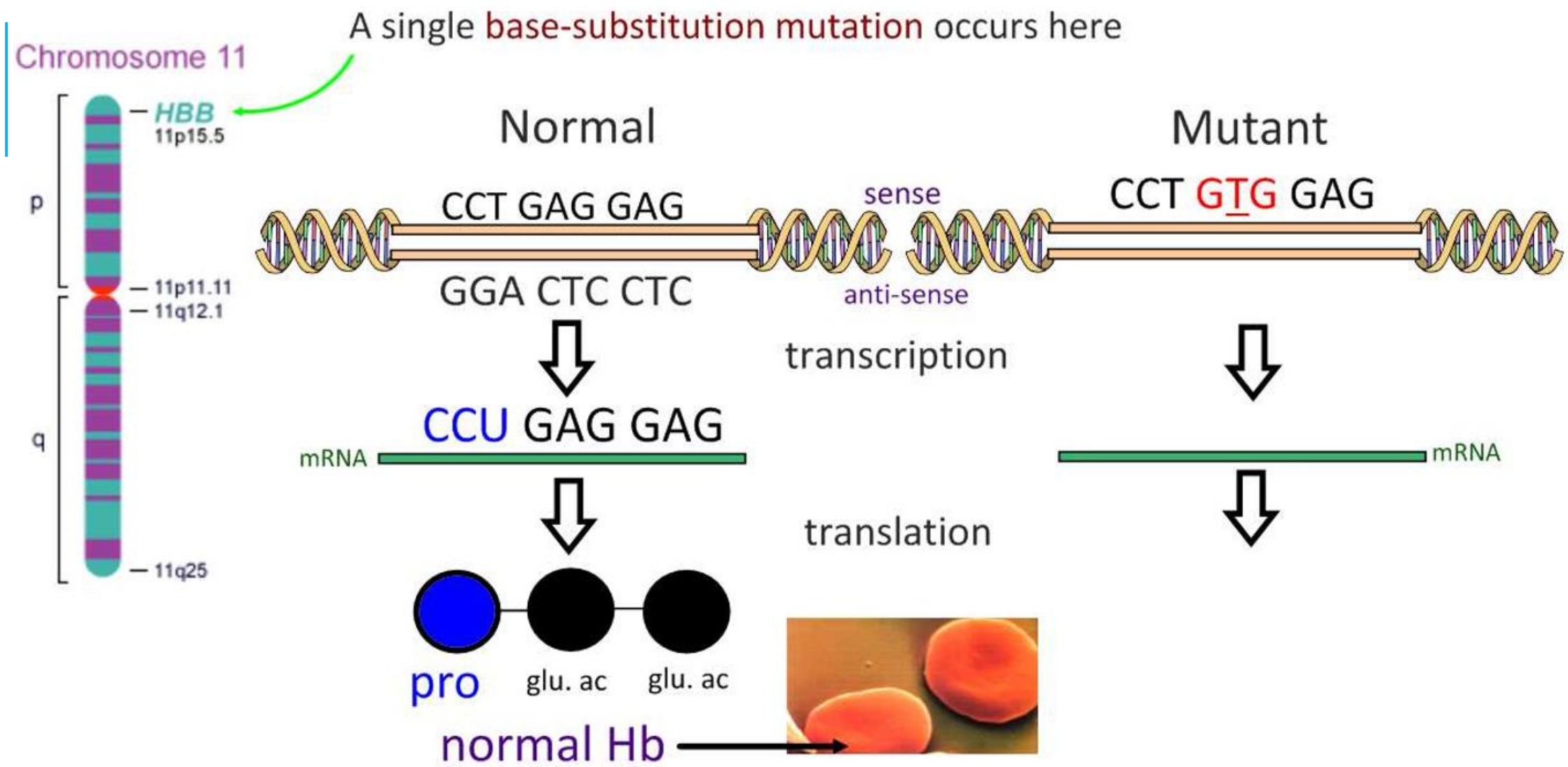
mRNA

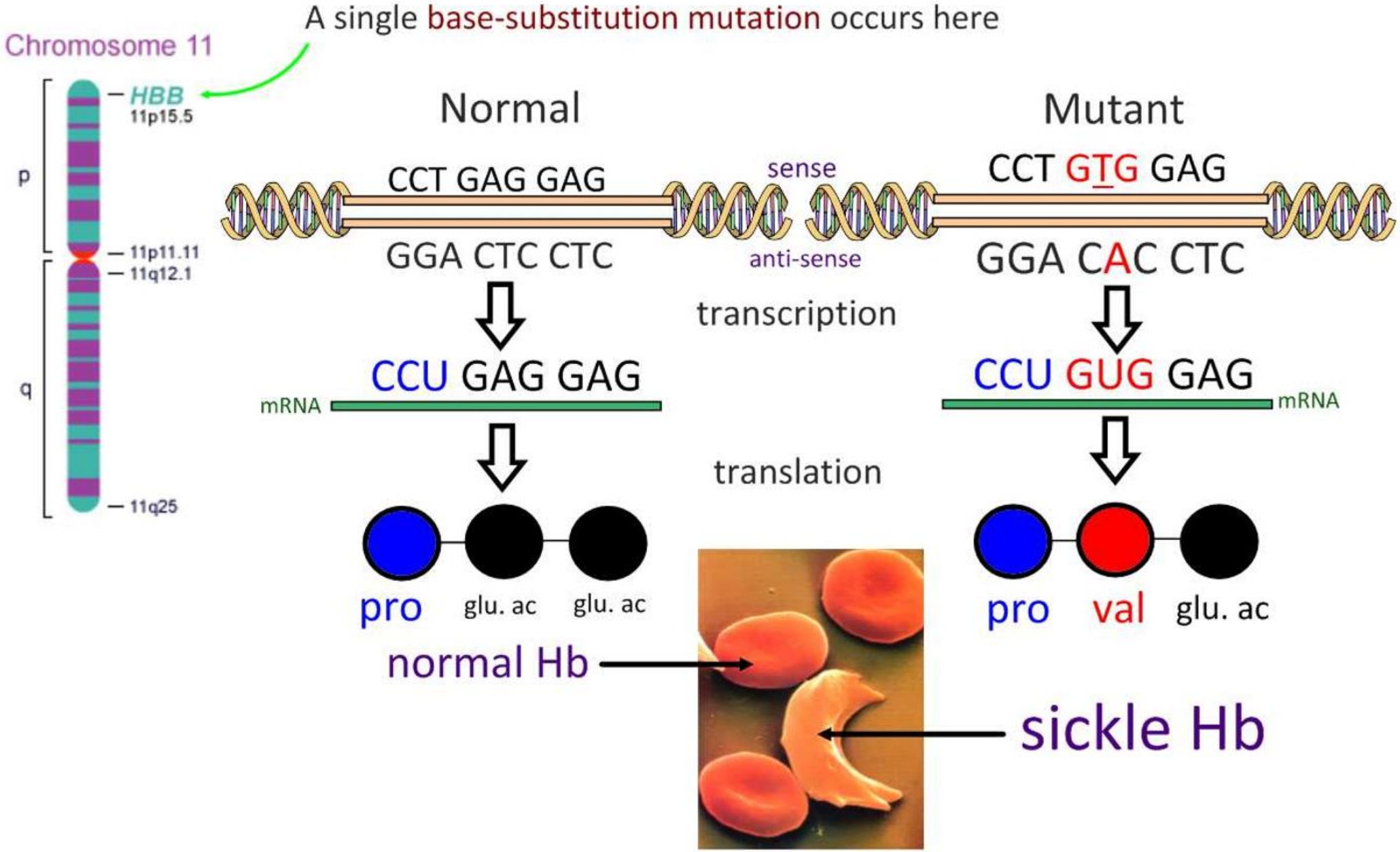
translation

pro glu. ac glu. ac

normal Hb




A single base-substitution mutation occurs here
Chromosome 11
HBB
11p15.5
p
11p11.11
11q12.1
q
11q25
Normal
Mutant
CCT GAG GAG
sense
CCT GTG GAG
GGA CTC CTC
anti-sense
transcription
CCU GAG GAG
mRNA
mRNA
translation
pro
glu. ac
glu. ac
normal Hb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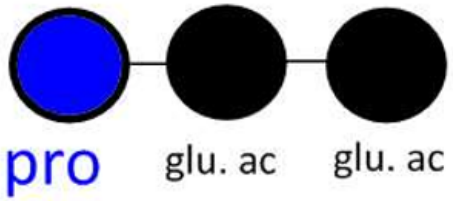
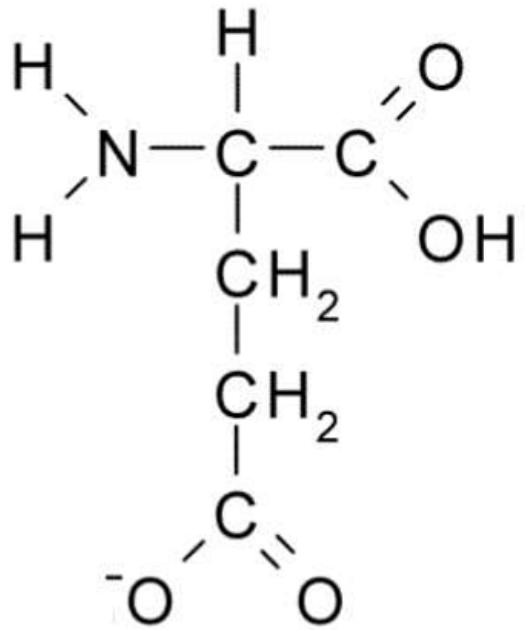
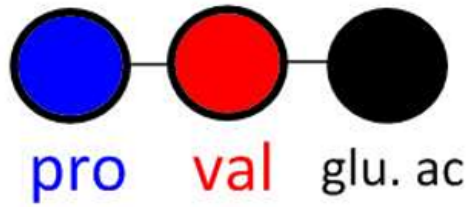
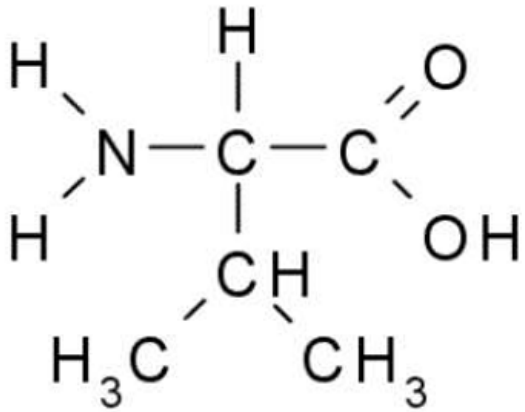
q
11q25
Normal
Mutant
CCT GAG GAG
sense
CCT GTG GAG
GGA CTC CTC
anti-sense
GGA CAC CTC
transcription
CCU GAG GAG
mRNA
CCU GUG GAG
mRNA
translation
pro
glu. ac
glu. ac
pro
val
glu. ac
normal Hb
sickle Hb

Comparing glutamic acid and valine





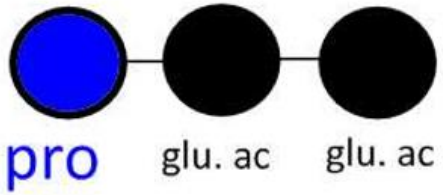


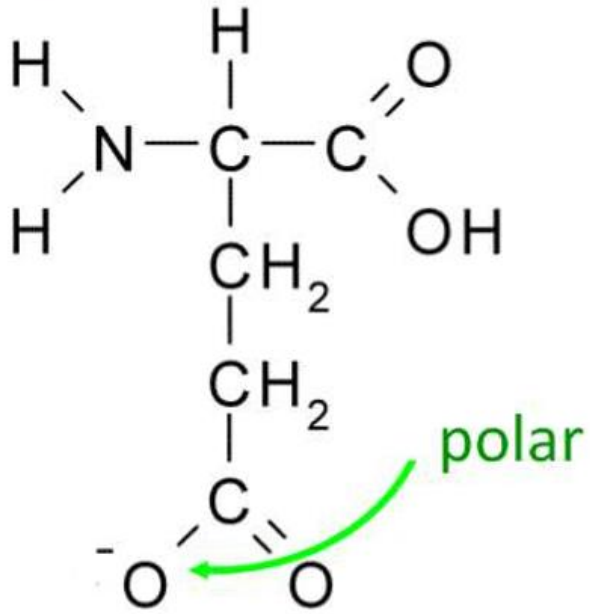


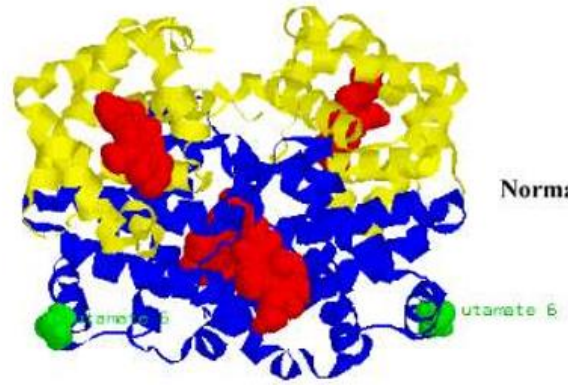
What differences in properties can we deduce from the structures?

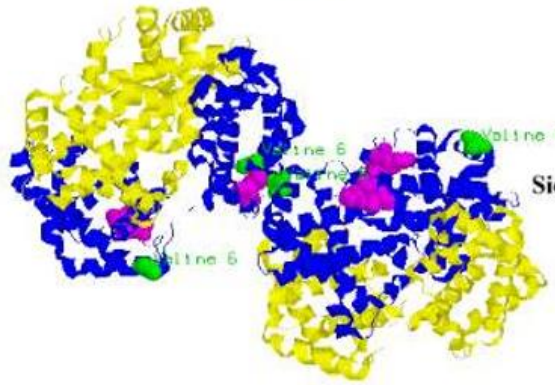
How might this change the quaternary protein structure?

Comparing glutamic acid and valine

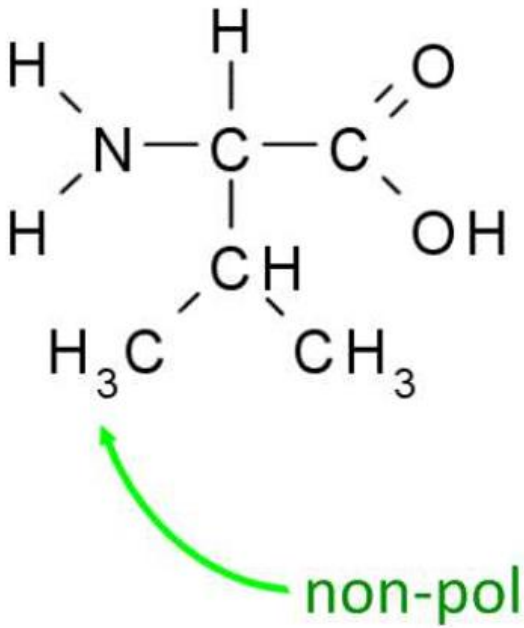








Note: The Sickle hemoglobin image is drawn at 50% of the size of the Normal hemoglobin



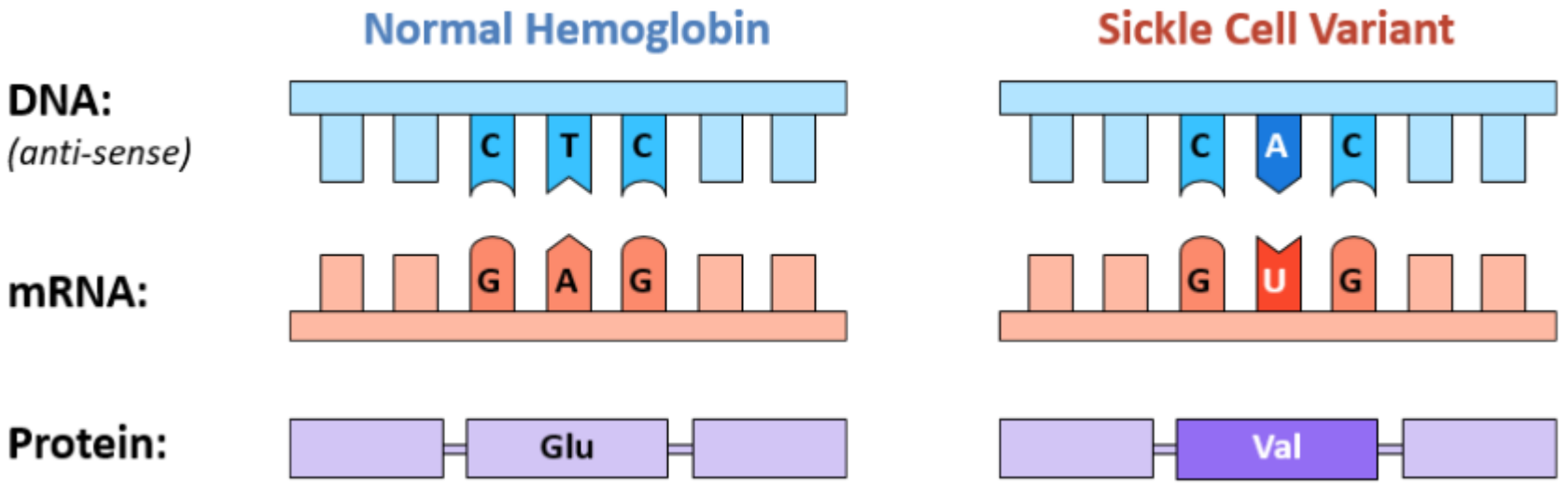
What differences in properties can we deduce from the structures?

How might this change the quaternary protein structure?

SUMMARY: SICKLE CELLS

Sickle cell anemia is caused by a base substitution in hemoglobin

- 6th codon (*sense strand*): **GAG → GTG**
- Amino acid: **Glu → Val**



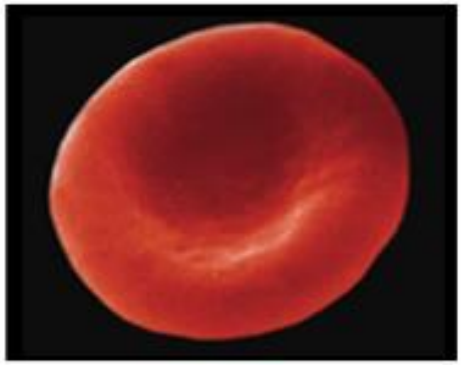
MUTATION CONSEQUENCE: ANAEMIA

The amino acid change alters structure → hemoglobin forms insoluble strands

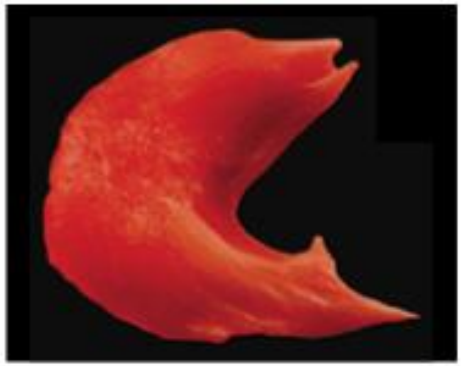
These insoluble strands change the shape (*sickle shape*)

The red blood cells cannot carry oxygen effectively and are destroyed at a higher rate – leading to **anemia**

Sickle cells are more likely to form clots

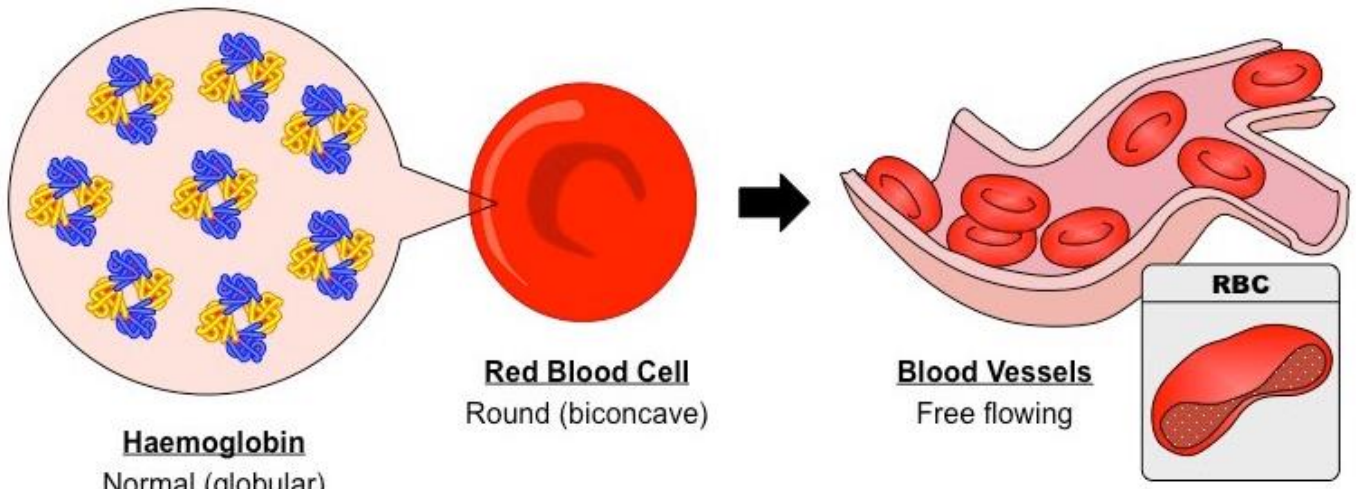


Normal Blood Cell



Sickle Blood Cell

Normal (Wild-Type) Haemoglobin

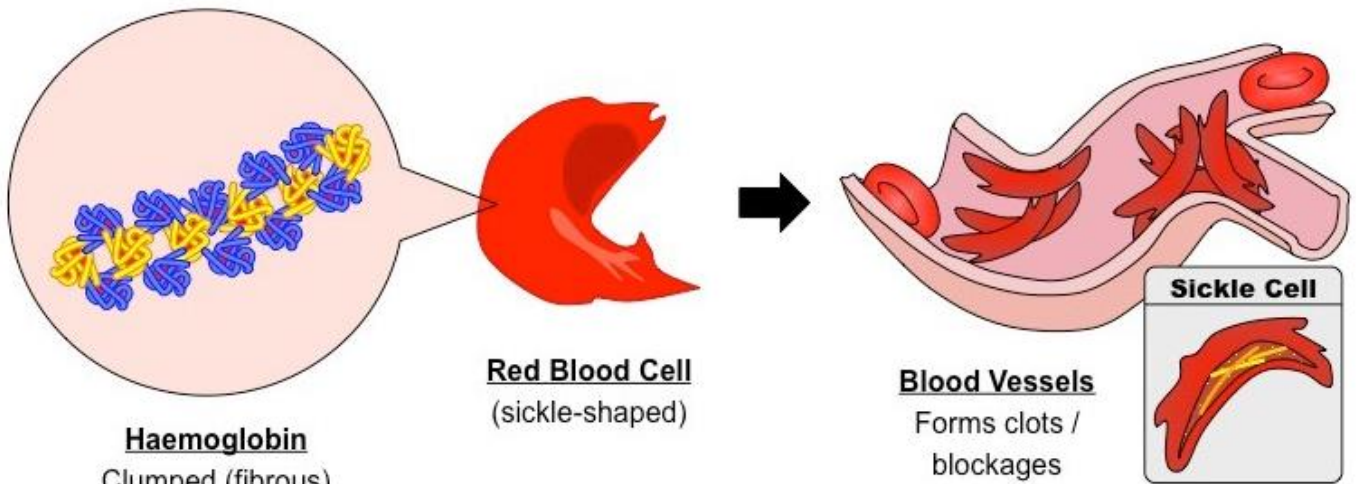


Haemoglobin
Normal (globular)

Red Blood Cell
Round (biconcave)

Blood Vessels
Free flowing

'Sickle Cell' Haemoglobin

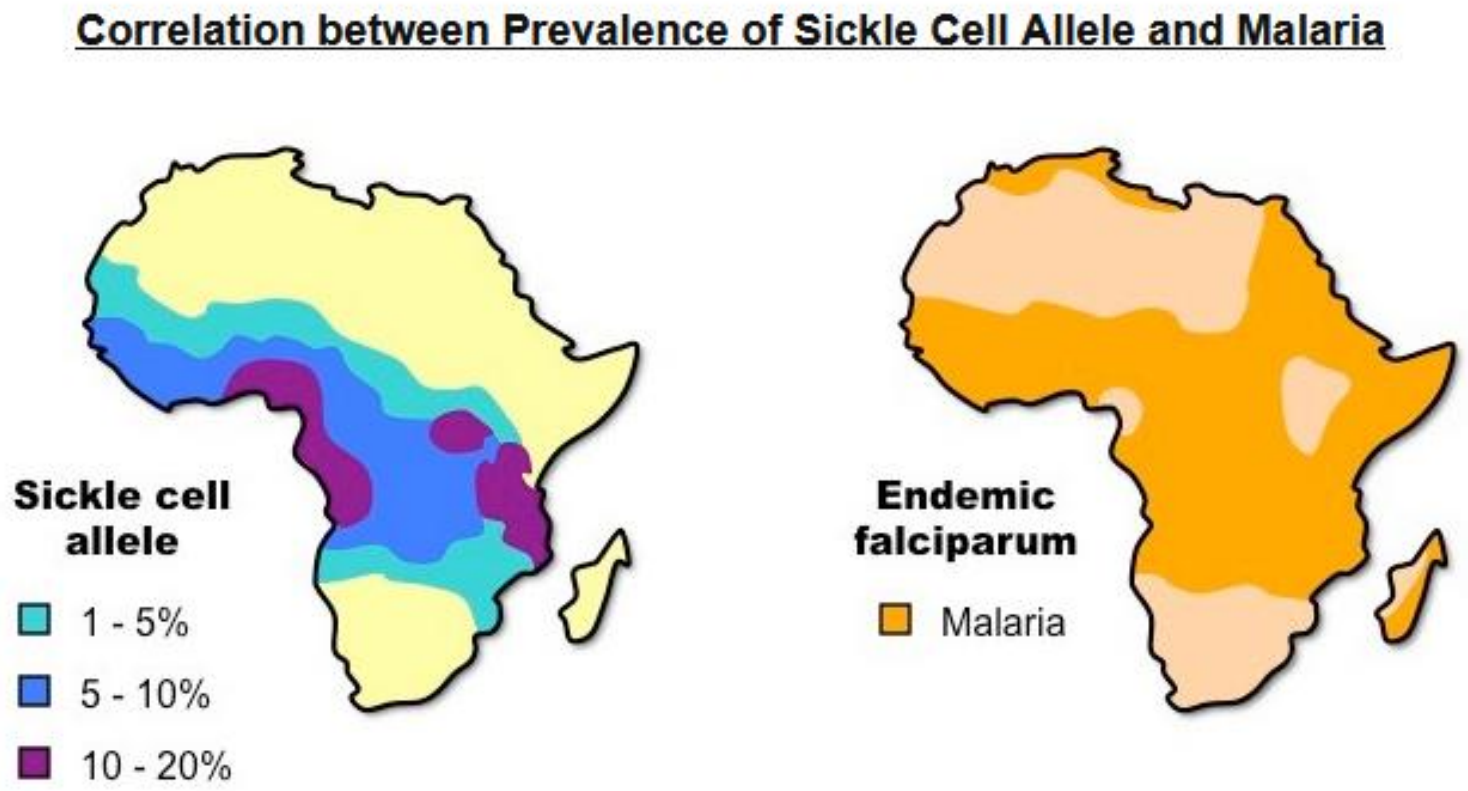


Haemoglobin
Clumped (fibrous)

Red Blood Cell
(sickle-shaped)

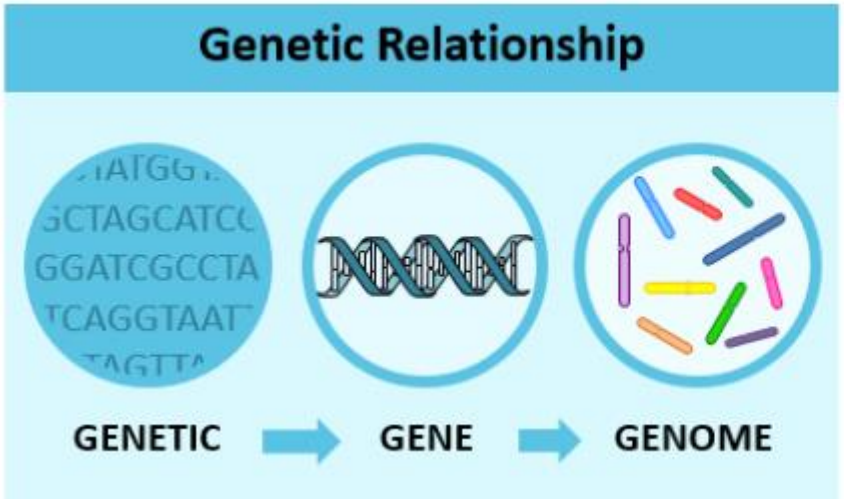
Blood Vessels
Forms clots / blockages

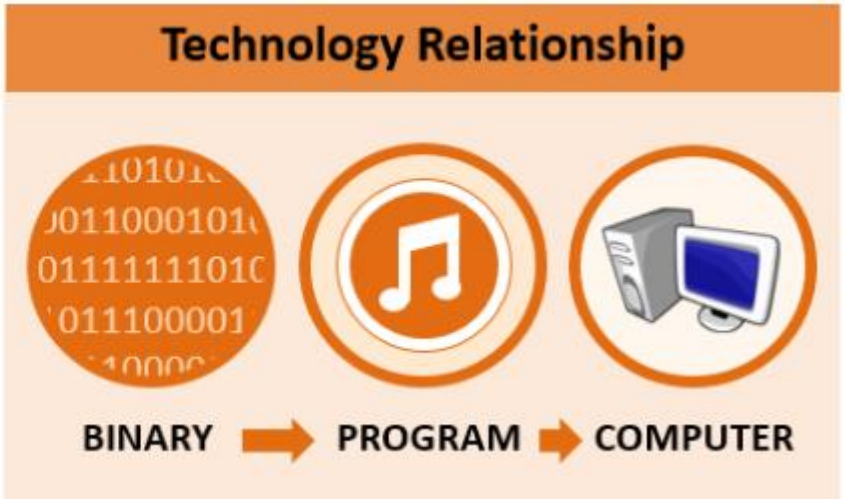
SICKLE CELL VERSUS MALARIA



GENOMES

Genome = totality of genetic information in a cell or organism (coding + non-coding sequences)





HUMAN GENOME

The Human Genome Project (HGP) → international cooperative venture → sequence entirety of human genome.

- 46 chromosomes
- ~3 billion base pairs
- ~21 000 genes

GENETICS COMPARISON

Species differ in genome size, number of chromosomes and number of genes

There is **no** clear correlation between these factors → but other trends

- viruses and prokaryotes tend to have a smaller genome + fewer genes
- eukaryotes have huge variation in genome size and gene numbers

Overall **no** consistent or reliable pattern.

GENOME SIZE

Species	*T2 phage*	*Escherichia coli*	*Drosophila melanogaster*	*Homo sapiens*	*Paris japonica*
Genome Size	170,000 bp	4.6 million bp	130 million bp	3.2 billion bp	150 billion bp
Common Name	 Virus	 Bacteria	 Fruit fly	 Human	 Canopy Plant

CHROMOSOME NUMBER

Species	*Parascaris equorum*	*Oryza sativa*	*Homo sapiens*	*Pan troglodytes*	*Canis familiaris*
Chromosomes	4	24	46	48	78
Common Name	 Roundworm	 Rice	 Human	 Chimpanzee	 Dog

GENE NUMBERS

Species	*Escherichia coli*	*Gallus gallus*	*Homo sapiens*	*Daphnia pulex*	*Oryza sativa*
Genome Size	~4,200	~17,000	~21,000	~31,000	~38,000
Common Name	 Bacteria	 Chicken	 Human	 Water flea	 Rice