# Variation and Mutation

## Learning Outcomes

- *18.1.1* – **Describe** variation as differences between individuals of the same species
- *18.1.4* – **State** that discontinuous variation is usually caused by genes only and continuous variation is caused by both genes and the environment
- *18.1.5* – **Investigate** and describe examples of continuous and discontinuous variation
- *18.1.6* – **Describe** mutation as genetic change
- *18.1.7* – **State** that mutation is the way in which new alleles are formed
- *18.1.8* – **State** that ionising radiation and some chemicals increase the rate of mutation
- *18.1.9* – **Describe** gene mutation as a random change in the base sequence of DNA
- *18.1.10* – **State** that mutation, meiosis, random mating and random fertilisation are sources of genetic variation in populations

# Variation

**Variation** · the difference between individuals of the same **species**

↳ group of organisms that can interbreed to produce fertile offspring

Even though all humans belong to the same species, *Homo sapiens*, they are not all the same and have many differences between each other

↳ Variation can either be due to <u>genetics</u> or <u>environmental factors</u> or both

→ **Genotypic variation**: variation caused by differences in genes (i.e. alleles) carried on chromosomes.

This variation is inherited from parents and you are born with this - it does not change in your lifetime

These are inherited characteristics

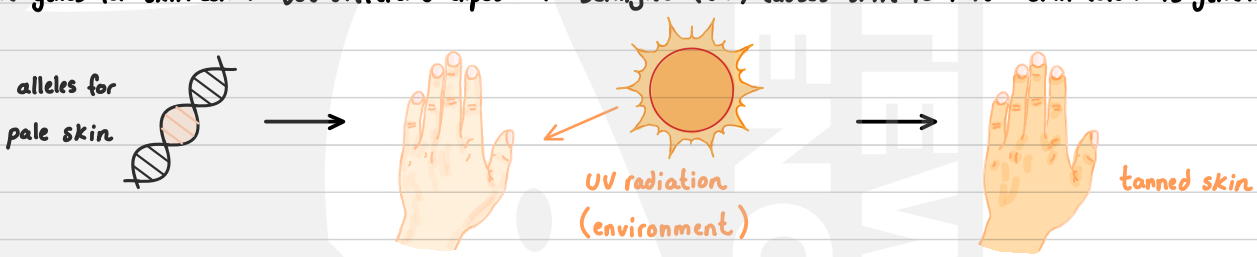
<u>ex</u>: all individuals have genes responsible for coding eye colour. Individual A has alleles for blue whereas B has alleles for green.


→ **Phenotypic variation**: variation caused by both differences in genes but also in the environment

This variation is not fully inherited and is impacted by the environment in which an organism lives (sunlight, temperature, diet, humidity, oxygen...)

These are acquired characteristics

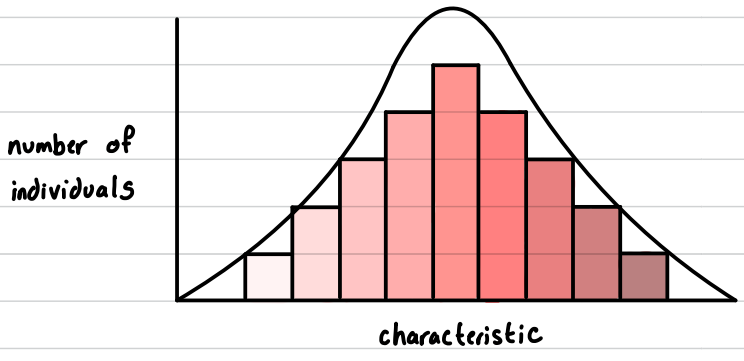
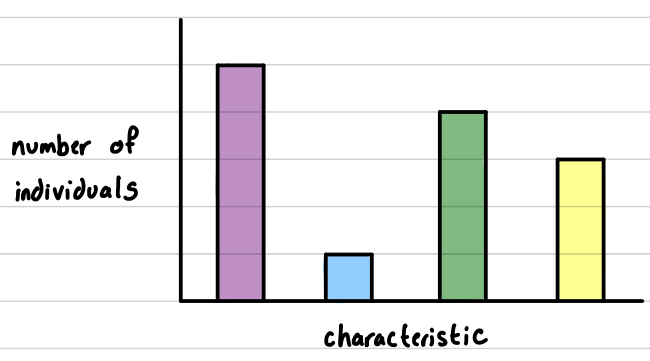
<u>ex:</u> we all have genes for skin colour but different exposure to sunlight (UV) causes skin to tan - skin colour is genetic, tan is acquired




# Variation in Populations

**Population**: a group of organisms of the same species living in the same area

↳ When we examine populations, we can track how variable they are for a given characteristic.

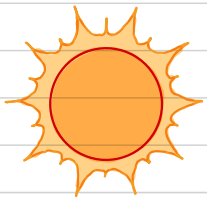
| Continuous Variation | Discontinuous Variation |
|---|---|
| No distinct categories; a spectrum of different varieties | Distinct categories; no intermediates |
| tends to be quantitative | tends to be qualitative |
| caused by both genes <u>and</u> the environment | caused (usually) by genes <u>only</u> environment minimal or no role |
| multiple genes responsible (polygenic) | few genes responsible (sometimes 1 only) |
| multiple alleles have an additive effect<br>ex: more alleles for pigmentation = darker | alleles do not have additive effect |
|  |  |
| examples: height, weight, skin colour, intelligence | examples: ABO blood groups, tongue rolling, eye colour |

# Sources of Genetic Variation

## Mutations

**mutation**: genetic change. Specifically, a random change in the nucleotide base sequence of DNA. Some factors can increase the rate of mutations.

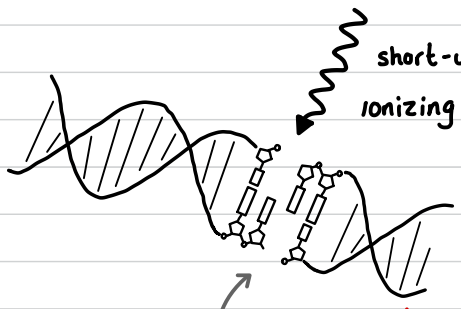
→ could be a base pair swap, insertion of 1 or more base pairs or deletion of 1 or more base pairs



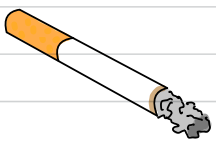
UV radiation

X-rays

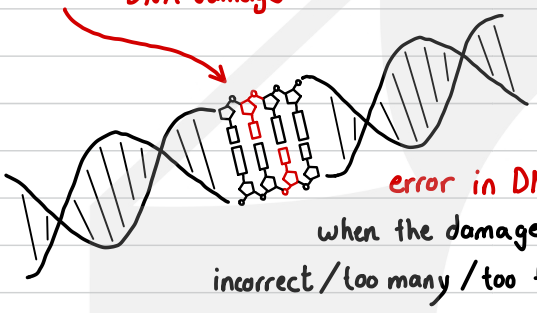
gamma radiation




Causes of Mutations

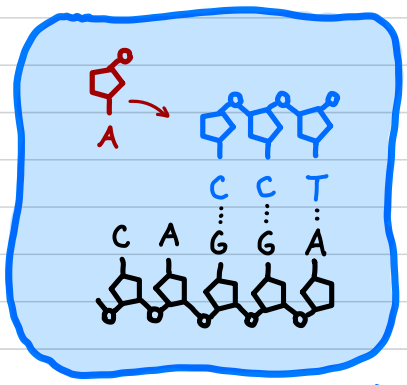


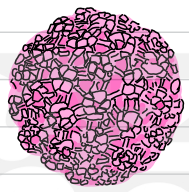
chemical mutagen
(ex: nicotine)



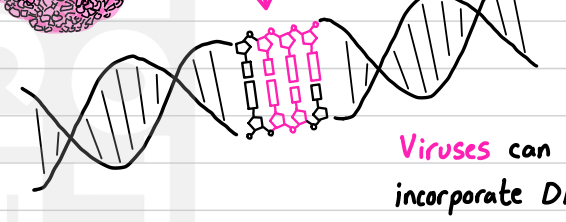
error in DNA repair
when the damage is fixed,
incorrect / too many / too few bases are used

**Error in DNA replication**
DNA polymerase adds the incorrect, non-complementary base during replication and this mistake is missed during proofreading



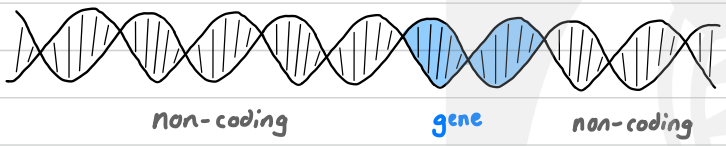
ex: HPV



Viruses can alter / incorporate DNA in infected host cells, changing it

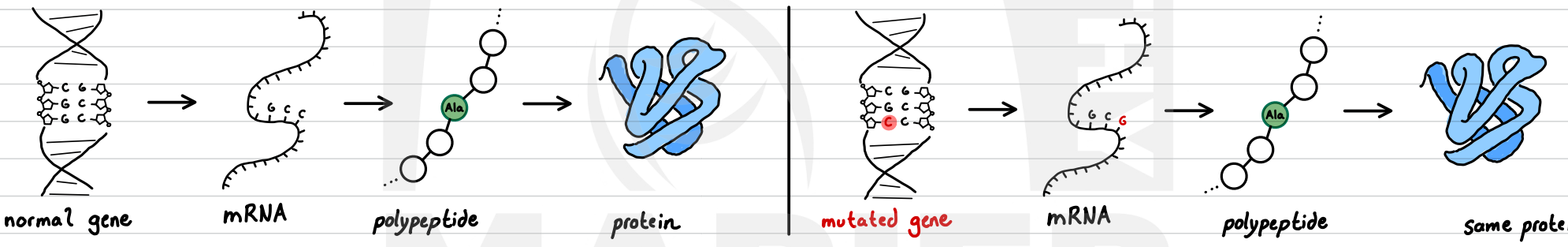
### Impact of mutations:

😐 neutral: if a mutation occurs in a non-coding section (DNA that is not a gene) proteins being synthesized unaffected

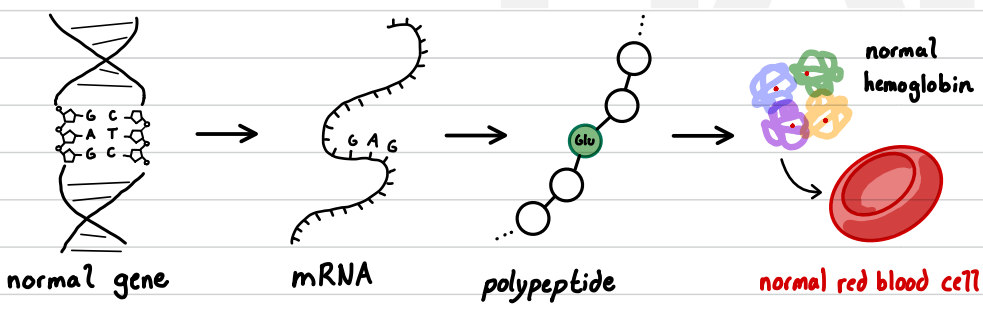



* More than 95% of human DNA is non-coding Even though they don't code for proteins they are important for gene regulation, structure, and more

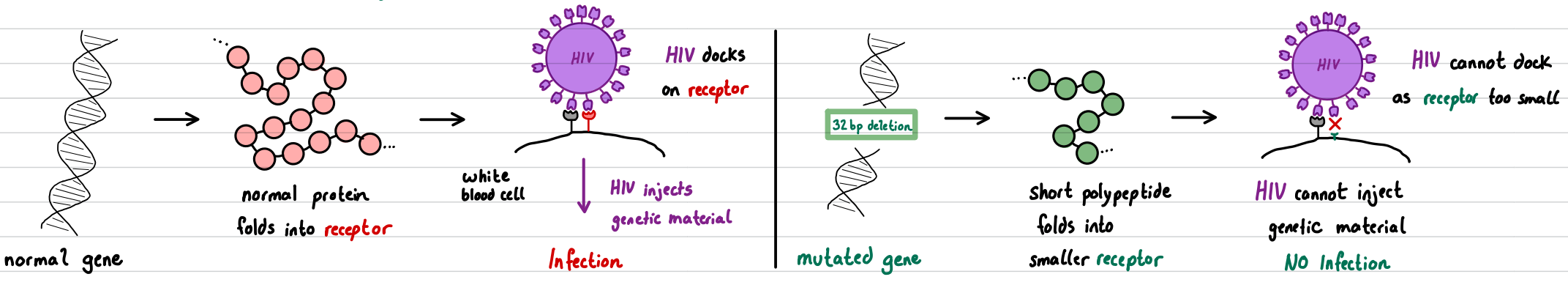
😐 neutral: if a mutation occurs in a gene, but the mutated codon codes for the same amino acid. No change as same protein being made.




☹ negative: if a mutation occurs in a gene, the mutated codon(s) codes for a different / altered protein. → New allele (could be disadvantageous, like disease)




☺ positive: if a mutation occurs in a gene, the mutated codon(s) codes for a different / altered protein. → New allele (could be advantageous, like resistance)



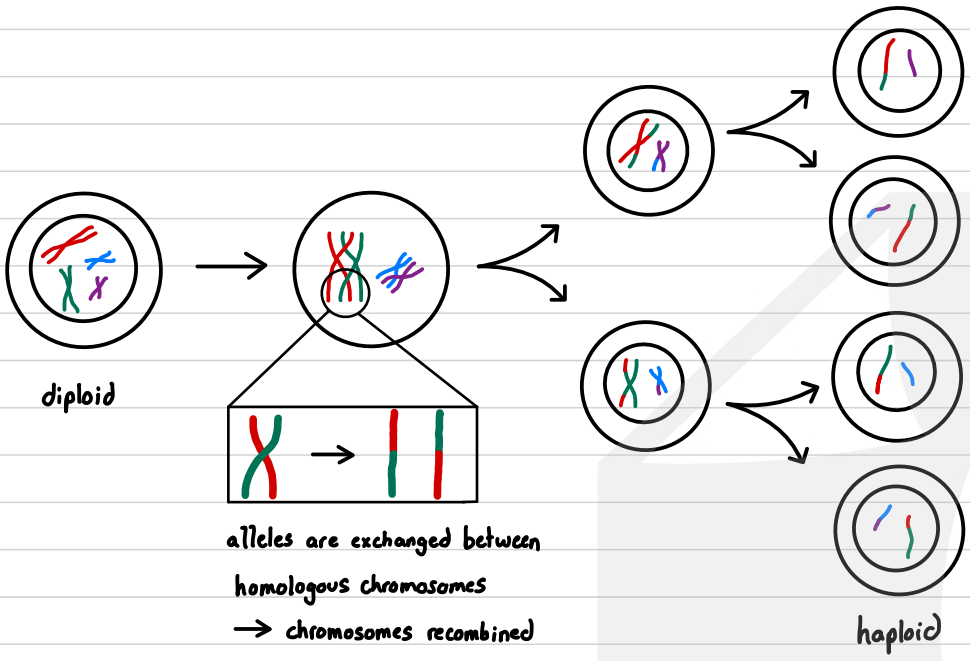
# Sources of Genetic Variation

Note: all organisms (unicellular or multicellular) are subject to mutations. Mutations are the source of new alleles and new characteristics. however, some organisms reproduce sexually and this further increases variation as it allows new combinations to occur

Genetically unique gametes created via meiosis × random pairing of mates × random fertilization of gametes = offspring with a unique combination of alleles (different from each other and parents)

## Meiosis

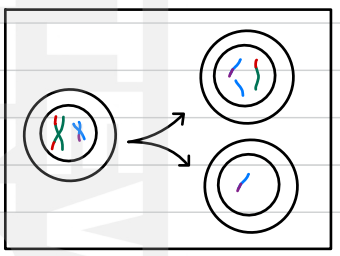
**Meiosis**: a reduction division in which the chromosome number is halved from diploid to haploid, resulting in genetically unique gametes




end result are 4 haploid gametes which are genetically unique

these differentiate and mature into sperm
(testes in mammals, anthers in flowering plants)

these differentiate and mature into ova
(ovaries in mammals, ovules in flowering plants)



an error in division could result in a gamete with too many or too few chromosomes. If these fertilize offspring will have genetic disorder
ex: Down Syndrome
(47 chromosomes instead of 46)

## Random Mating

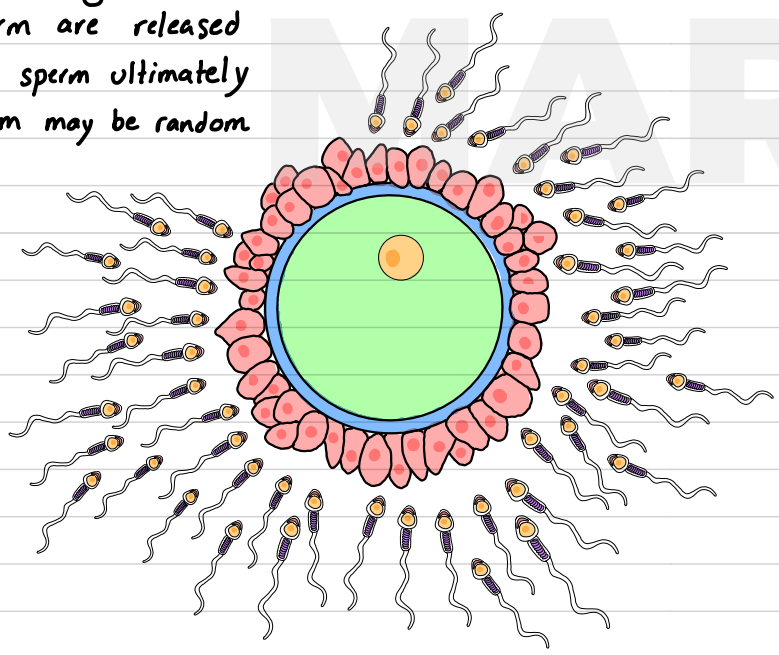
Even though most sexually-reproducing organisms select mates prior to reproduction, random mating still occurs

Ex: In plants, which plant pollinates another can be strongly determined by weather conditions, like wind or currents
∴ which alleles are combined is random

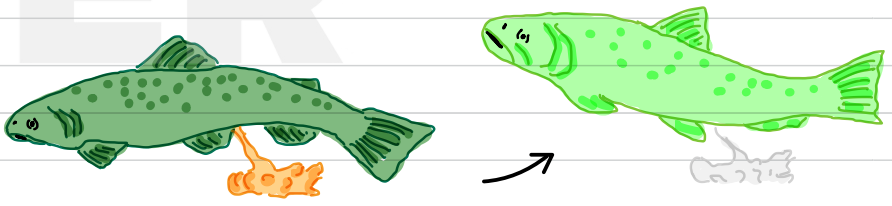
ex: In animals there is typically mate selection. But not all traits are selected for, such as blood type. So it is often random for certain traits which get combined and mixted

## Random Fertilization

In sexually reproducing individuals, millions of sperm are released at once! Which sperm ultimately will fertilize ovum may be random



In many fish, the female releases hundreds-thousands of eggs where the male then releases millions of sperm. Which gamete fuses with which is often random



females release many eggs

males release many sperm

# Assessment Tasks

Answer the following questions:

(1) The following DNA sequence codes for a protein:

TAC GGT CAG GCC ACC CTC GAA TTA AGA TTC ATT

determine the polypeptide chain it codes for

(2) The sequence got mutated when an extra base was inserted

TAC GGC TCA GGC CAC CCT CGA ATT AAG ATT CATT

↑

determine the polypeptide chain it codes for and justify whether the protein will be impacted

Second letter

| First letter | U | C | A | G | Third letter |
|---|---|---|---|---|---|
| U | UUU Phe<br>UUC Phe<br>UUA Leu<br>UUG Leu | UCU Ser<br>UCC Ser<br>UCA Ser<br>UCG Ser | UAU Tyr<br>UAC Tyr<br>**UAA Stop**<br>**UAG Stop** | UGU Cys<br>UGC Cys<br>**UGA Stop**<br>UGG Trp | U<br>C<br>A<br>G |
| C | CUU Leu<br>CUC Leu<br>CUA Leu<br>CUG Leu | CCU Pro<br>CCC Pro<br>CCA Pro<br>CCG Pro | CAU His<br>CAC His<br>CAA Gln<br>CAG Gln | CGU Arg<br>CGC Arg<br>CGA Arg<br>CGG Arg | U<br>C<br>A<br>G |
| A | AUU Ile<br>AUC Ile<br>AUA Ile<br>**AUG Met** | ACU Thr<br>ACC Thr<br>ACA Thr<br>ACG Thr | AAU Asn<br>AAC Asn<br>AAA Lys<br>AAG Lys | AGU Ser<br>AGC Ser<br>AGA Arg<br>AGG Arg | U<br>C<br>A<br>G |
| G | GUU Val<br>GUC Val<br>GUA Val<br>GUG Val | GCU Ala<br>GCC Ala<br>GCA Ala<br>GCG Ala | GAU Asp<br>GAC Asp<br>GAA Glu<br>GAG Glu | GGU Gly<br>GGC Gly<br>GGA Gly<br>GGG Gly | U<br>C<br>A<br>G |

(3) Cancer is a result of a mutation in your body cells where they grow and divide uncontrollably, resulting in a tumor. If a person's skin cells mutate and become cancerous and they have a child - will the child have cancer as well? Explain.

(4) Cigarettes and vape pens contain several mutagens. Assess the following statement: "smoking cigarettes will give you cancer"

(5) Over time, it is more common for mutations to occur in non-coding than coding sections of DNA. Explain why.

(6) In the past, identical twins were used (unethically) in studies to determine which characteristics were more determined by genetics or the environment. Explain how this works.

(7) <u>Investigation</u>

Aim: to determine the distribution of certain characteristics of a local population

Method: 1- Create a table like the one below:

| Participant | eye colour | blood group (A/B/AB/O) | tongue roller (yes/no) | height (cm) | shoe size | hand span (cm) |
|---|---|---|---|---|---|---|
| A | | | | | | |
| B | | | | | | |
| C | | | | | | |

* always ask permission and consent when gathering data. Keep names anonymous.

2- Now create a tally for <u>one</u> continuous and <u>one</u> discontinuous characteristic. Example below:

| height groups (cm) | tally |
|---|---|
| 155 - 159 | |
| 160 - 164 | |
| 165 - 169 | |
| 170 - 174 | |
| 175 - 179 | |
| 180 - 184 | |
| 185 - 189 | |
| 190 - 194 | |

create equal categories that fit your data range

| blood groups | tally |
|---|---|
| A | |
| B | |
| AB | |
| O | |

3- Graph both datasets

4- Analyze your data. Any patterns? Outliers?