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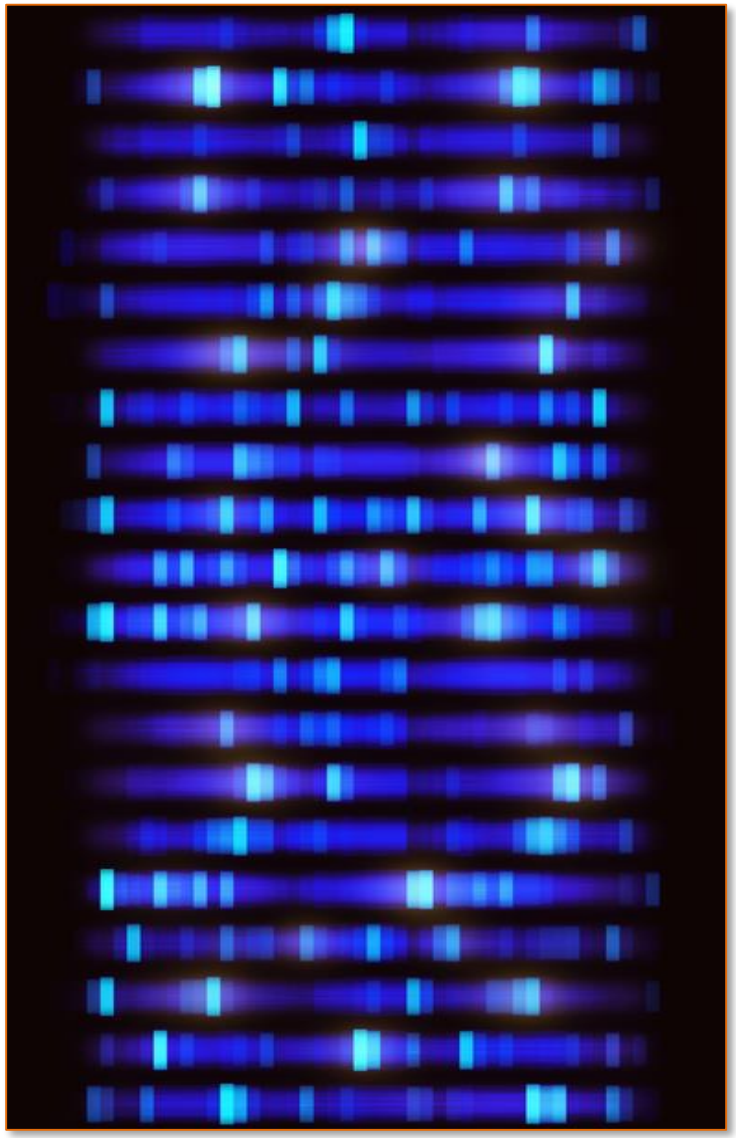
GCSE Biology

Edexcel Topic 3 – Genetics

Genetics - Overview

- Advantages and disadvantages of sexual & asexual reproduction (biology)
- Meiosis
- DNA structure
- DNA and the genome
- Protein synthesis (biology HT)
- The work of Mendel (biology)
- Genetic inheritance
- Sex determination
- ABO blood group inheritance
- Inherited disorders (biology HT)
- Variation
- Human Genome Project
- Mutations



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Genetics

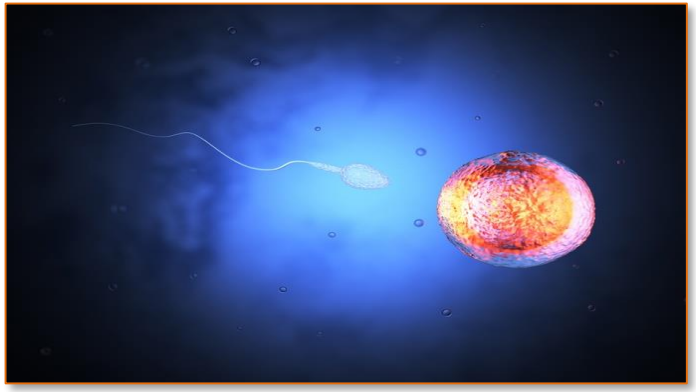
Part 1

- Advantages of sexual and asexual reproduction (biology)
- Meiosis

Genetics part 1 – Sexual and asexual reproduction

Sexual reproduction involves the joining of **male** and **female** gametes.
A **gamete** is the scientific term for a sex cell.
In animals, the gametes are the **sperm** and the **egg** cells.
In flowering plants, the gametes are the **pollen** and the **egg** cells.

In sexual reproduction, **mixing** of **genetic information** occurs which leads to **variety** in the offspring.
Every new **offspring** formed is **unique**.
The gametes are produced by **meiosis**.



Asexual reproduction involves only **one parent**. There is **no fusion** of gametes. **No mixing** of genetic information occurs. **All** offspring are genetically **identical** (called **clones**). Only **mitosis** is involved.

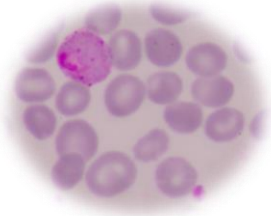
Genetics part 1 – Sexual and asexual reproduction (biology only)

There are **advantages** and **disadvantages** to both **sexual** and **asexual** reproduction.

Sexual reproduction	Asexual reproduction
Variation occurs in the offspring and population **but** energy must be put into **finding** and courting a suitable **mate**.	Only one parent is required so it is a **more efficient** use of time and energy as **do not** need to find a **mate.**
If the environment changes, variation will give a **survival advantage by natural selection**. There is a better chance of at least some organisms surviving.	**Many identical offspring** can be produced if conditions are favourable **however no variation** in the population occurs making it **susceptible** to **pathogens** or environmental change.
Natural selection can be **manipulated** and sped up by **humans, using selective breeding** to increase food production.	Occurs **faster** than sexual reproduction.

Genetics part 1 Sexual and asexual reproduction (biology only)

There are advantages to sexual and asexual reproduction, so some organisms **reproduce using both methods** depending on the circumstances.

Malarial parasites		Reproduce **asexually** in the **human** host, but **sexually** inside a **mosquito**. Video
Fungi		Reproduce **asexually** by **spores**, and **sexually** to give **variation**.

When **daffodils** flower they produce **seeds**. This is **sexual** reproduction.

Daffodils also reproduce **asexually** by **bulb division**.

Strawberry plants produce **seeds sexually** and reproduce **asexually** using **runners**. A genetically identical plant forms at the end of the runner.

Genetics part 1 – Meiosis

Meiosis leads to **non identical** cells being formed.
Mitosis leads to **identical** cells (clones) being formed.

Cells in the **reproductive organs** divide by **meiosis** to form **gametes**. In animals, the reproductive organs are the **ovaries** and **testes**.

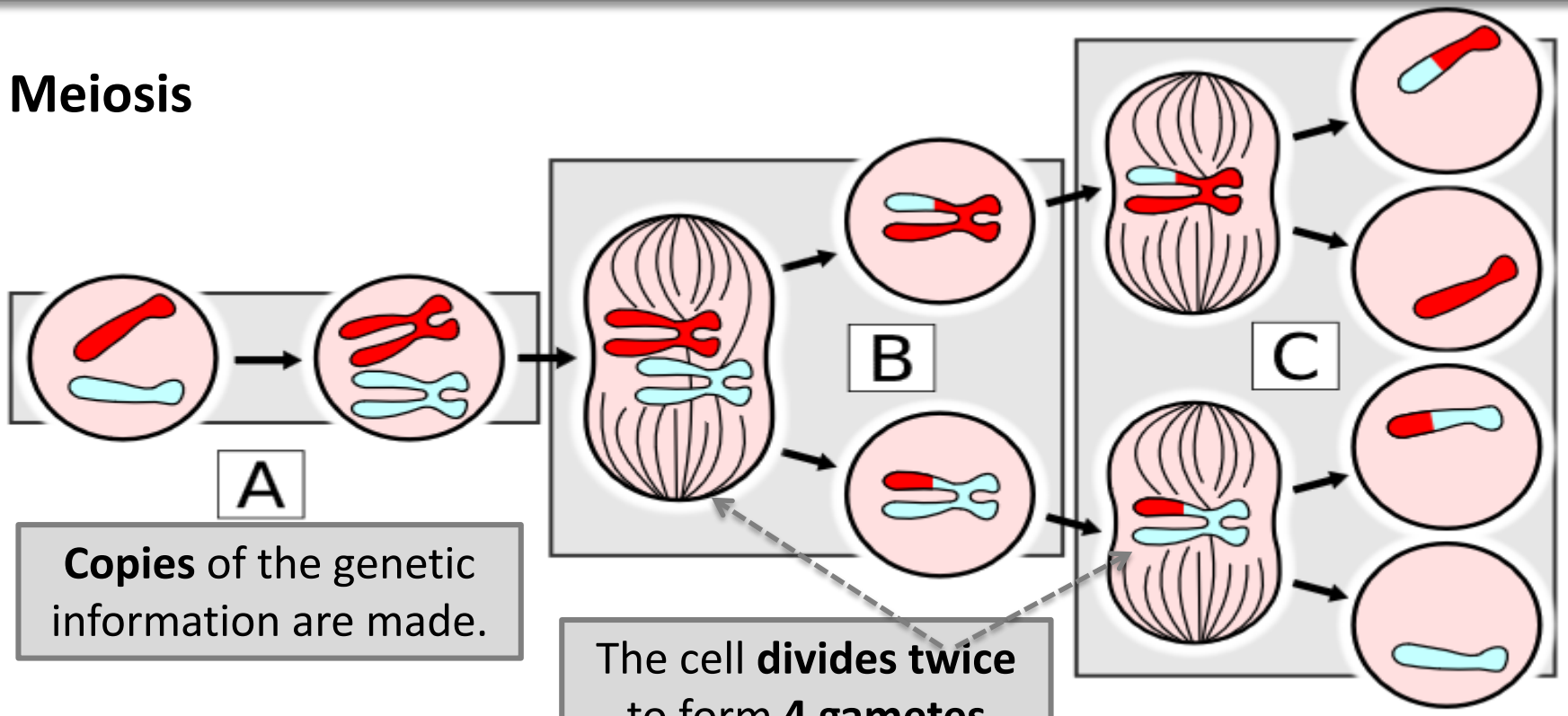
Meiosis is sometimes called **reduction division** because it **halves the number of chromosomes** in the gametes. When male and female gametes fuse during **fertilisation,** the number of chromosomes are restored.

This brother and sister have the same parents, but they look different. They show **variation** because of meiosis.

Genetics part 1 – Meiosis

This is the process a cell goes through to produce gametes:

Meiosis



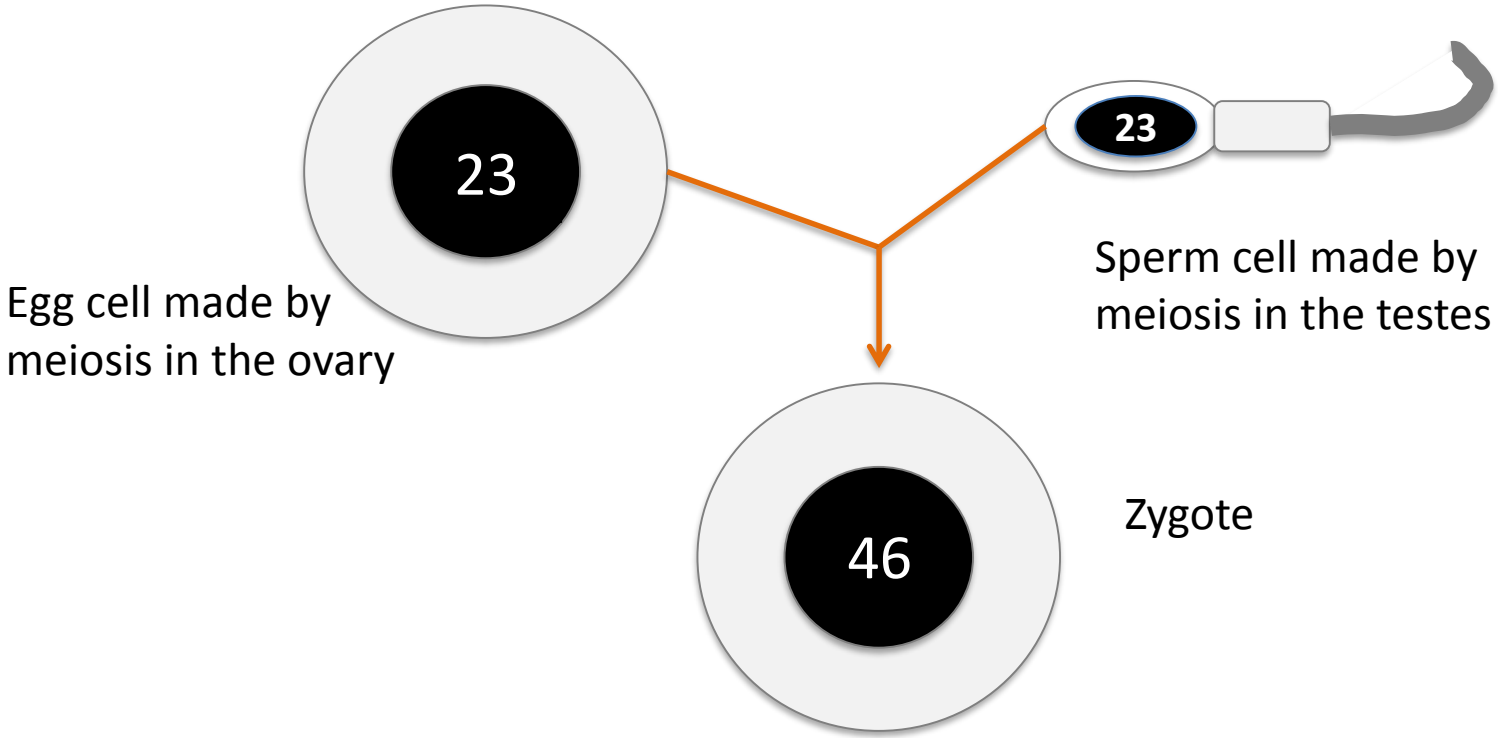
Copies of the genetic information are made.

The cell **divides twice** to form **4 gametes** each with a **single set** of **chromosomes**. This is **half** the number of the **original** cell.

All **4 haploid gametes** are **genetically different** from each other and the parent.

Genetics part 1 –Meiosis

A cell in the testes has **46** chromosomes. When this cell undergoes meiosis it produces 4 gametes each with **23** chromosomes. The same process occurs in the ovary to produce egg cells.
The male and female gametes **fuse** during **fertilisation** to restore the normal number of chromosomes.
The **fertilised** egg cell now contains **46** chromosomes. It is called a **zygote**.



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QuestionIT!

Genetics
Part 1

- Advantages and disadvantages of sexual and asexual reproduction (biology)
- Meiosis

Genetics part 1– QuestionIT

1. What is a gamete?
2. Name the male and female gametes in a) a human b) a daisy plant
3. For each row, tick **one** box to show which method of cell division is correct.

	Meiosis	Mitosis
Genetic mixing happens		
Gamete production occurs		
New cells show variation		
New cells have same number of chromosomes as parent		
Happens in skin cells		
Two divisions occur		
Two new cells are formed as a result		
Identical cells are formed		

4. What occurs at the point of fertilisation in humans?

5. How many chromosomes are found in the nucleus of a human:
   a) ovary cell ?
   b) egg cell ?
   c) embryo cell ?
   d) a cell just after fertilisation has occurred?
   e) a testes cell ?

6. What happens to the DNA just before the first meiotic division?

7. What does the term ‘haploid’ mean?

Biology ONLY

8. What are three advantages of sexual reproduction?

9. What are three advantages of asexual reproduction?

10. List three organisms which use both sexual and asexual reproduction methods.

11. Describe a disadvantage to an organism of i) using sexual reproduction and ii) asexual reproduction.

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AnswerIT!

Genetics
Part 1

- Advantages and disadvantages of sexual and asexual reproduction (biology only)
- Meiosis

Genetics part 1 – AnswerIT

1. What is a gamete?

   A cell with half the number of chromosomes of the parent cell.

2. Name the male and female gametes in a) a human b) a daisy plant

   a) Male = sperm Female = egg

   b) Male = pollen Female = egg

3. For each row, tick **one** box to show which method of cell division is correct.

	Meiosis	Mitosis
Genetic mixing happens	✓	
Gamete production occurs	✓	
New cells show variation	✓	
New cells have same no. of chromosomes as parent		✓
Happens in skin cells		✓
Two divisions occur	✓	
Two new cells are formed as a result		✓
Identical cells are formed		✓

4. What occurs at the point of fertilisation?

The nucleus of the sperm and egg cell fuse.

5. How many chromosomes are found in the nucleus of a human:

a) ovary cell ? **46**

b) egg cell ? **23**

c) embryo cell ? **46**

d) a cell just after fertilisation has occurred? **46**

e) a testes cell ? **46**

6. What happens to the DNA just before the first meiotic division?

The DNA is copied.

7. What does the term 'haploid' mean?

A single set of unpaired chromosomes.

Biology ONLY

8. What are three advantages of sexual reproduction?

Produce variation in the offspring
If environment changes can give a survival advantage meaning some organisms are likely to survive
Humans can speed up natural selection by selective breeding to increase food production

9. What are three advantages of asexual reproduction?

Only one parent needed
More time and energy efficient as do not need to find a mate
Faster than sexual reproduction
Many identical offspring can be produced when conditions are favourable

10. List three organisms which use both sexual and asexual reproduction methods.

Malarial parasites
Fungi
Strawberry plants
Daffodils

11. Describe a disadvantage to an organism of i) using sexual reproduction and ii) asexual reproduction.

i) Energy must be put into finding and courting a suitable mate.
ii) No variation in the population occurs making it susceptible to pathogens

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LearnIT!
KnowIT!

Genetics
Part 2

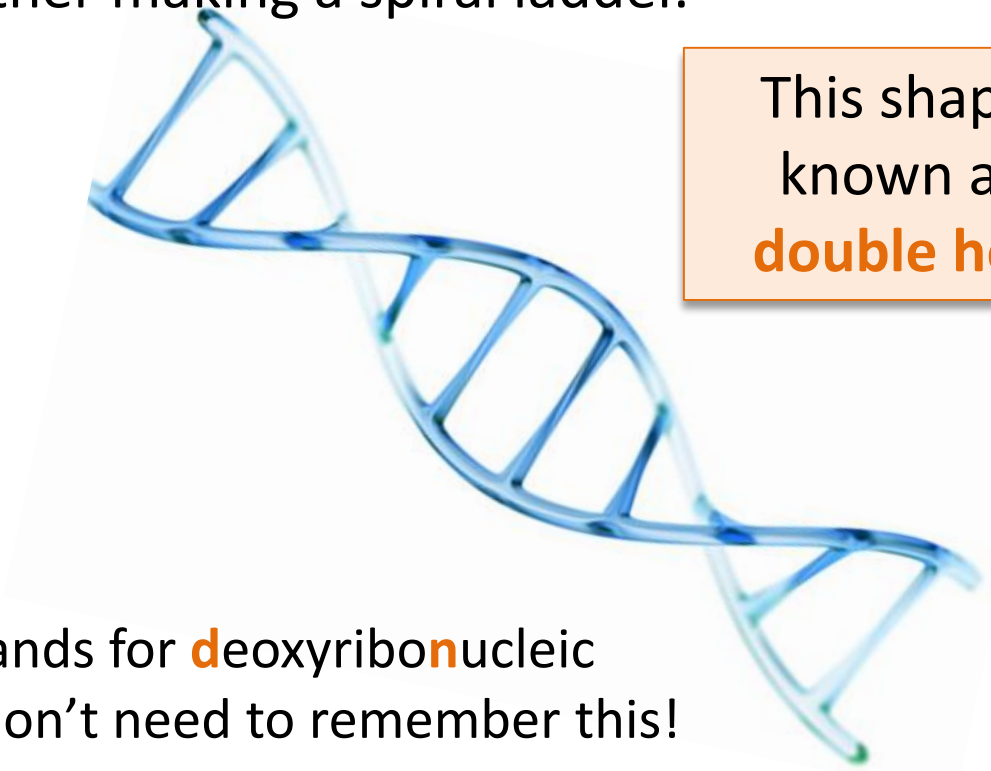
- DNA structure
- DNA and the genome
- Protein synthesis (biology HT)

Genetics part 2 – DNA structure

The **genetic material** in the nucleus of most cells is made from a chemical called **DNA**.

DNA is a **polymer** made from two strands which coil around each other making a spiral ladder.

A **polymer** is a large molecule made from many **smaller** molecules called **monomers**.



This shape is known as a **double helix**.

DNA stands for **d**eoxyribo**n**ucleic **a**cid. You don't need to remember this!

Genetics part 2 – DNA structure

DNA is made from repeating monomers called **nucleotides**. There are **4 different nucleotides**. The long strands of DNA consist of alternating sugar and phosphate sections.

Each nucleotide is made from three components:

- **A phosphate group**
- **A simple pentose sugar (this means it contains 5 carbon atoms)**
- **A base**

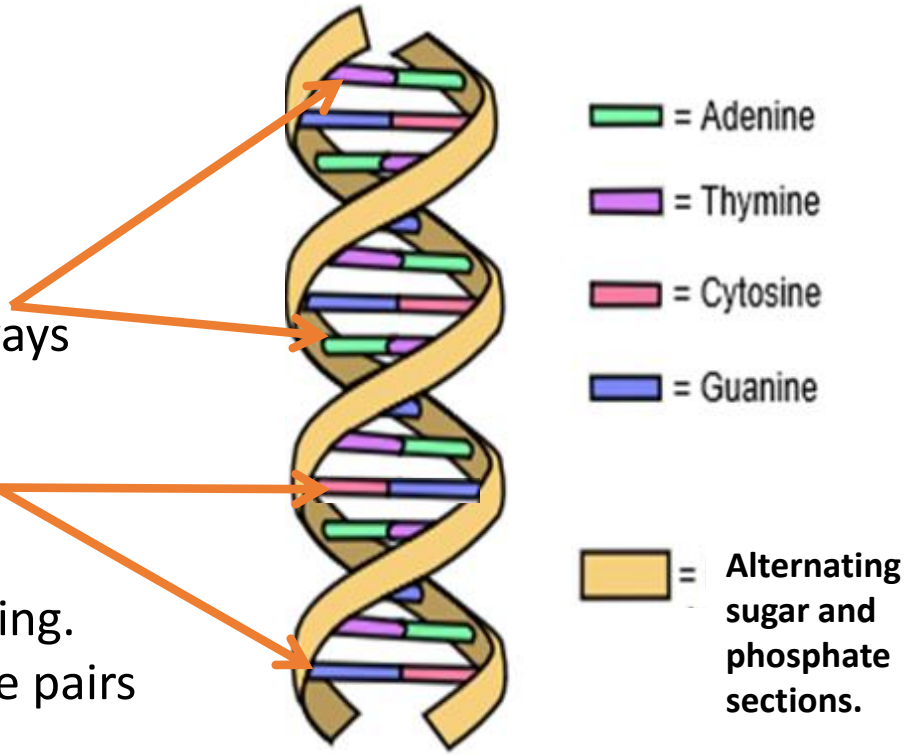
There are **4 bases in DNA**.
Adenine (A), thymine (T)
guanine (G) and cytosine (C)

Notice in the diagram that **adenine** always pairs up with **thymine** or vice versa.

Cytosine always pairs up with **guanine** or vice versa.

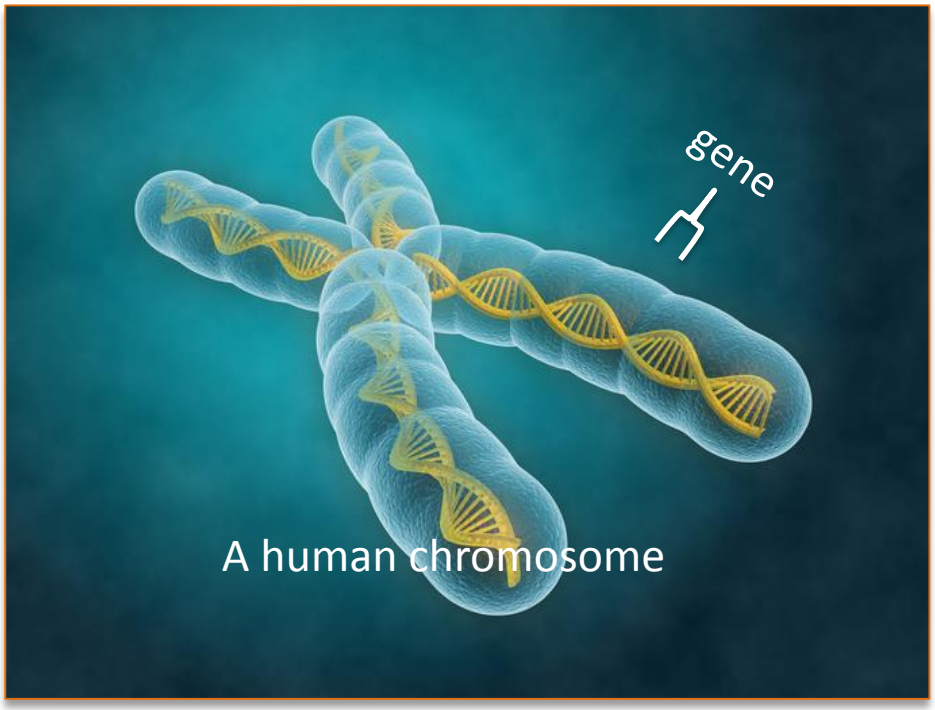
This is called **complementary** base pairing.

Weak hydrogen bonding holds the base pairs and the two strands together.



Genetics part 2 – DNA and the genome

DNA is arranged in structures called **chromosomes** inside a cell's nucleus.
The genome is the entire DNA of an organism.



A **gene** is a small section on a chromosome. Each gene **codes** for a particular sequence of **amino acids**, to make a specific **protein**.
A human has approximately 24 000 genes in total. Each single chromosome is made up of about 2000 genes.

In human body cells the **chromosomes** are normally found in **pairs**. Each cell has **23 pairs** of chromosomes.

Genetics part 2 – DNA and the genome

Extraction of DNA from fruit Video

Genetics part 2 – Protein synthesis (biology HT only)

Proteins have many different vital functions in our body.

- **Enzymes** – all made from protein.
- **Hormones** – all made from protein
- **Antibodies** – all made from protein
- **Structural** components such as muscle, hair and nail tissue.

Genes provide the **instructions** to make the **required protein** from **amino acids**.

It is estimated there are over 2 million proteins in a human body and all are made from combinations of just 20 different amino acids.

A sequence of **three bases** is the **code** for a particular **amino acid**. This is called a **codon**. The **order** of the **bases** controls the **order** in which **amino acids** are **assembled** to produce an individual protein.

Genetics part 2 – DNA structure (biology HT only)

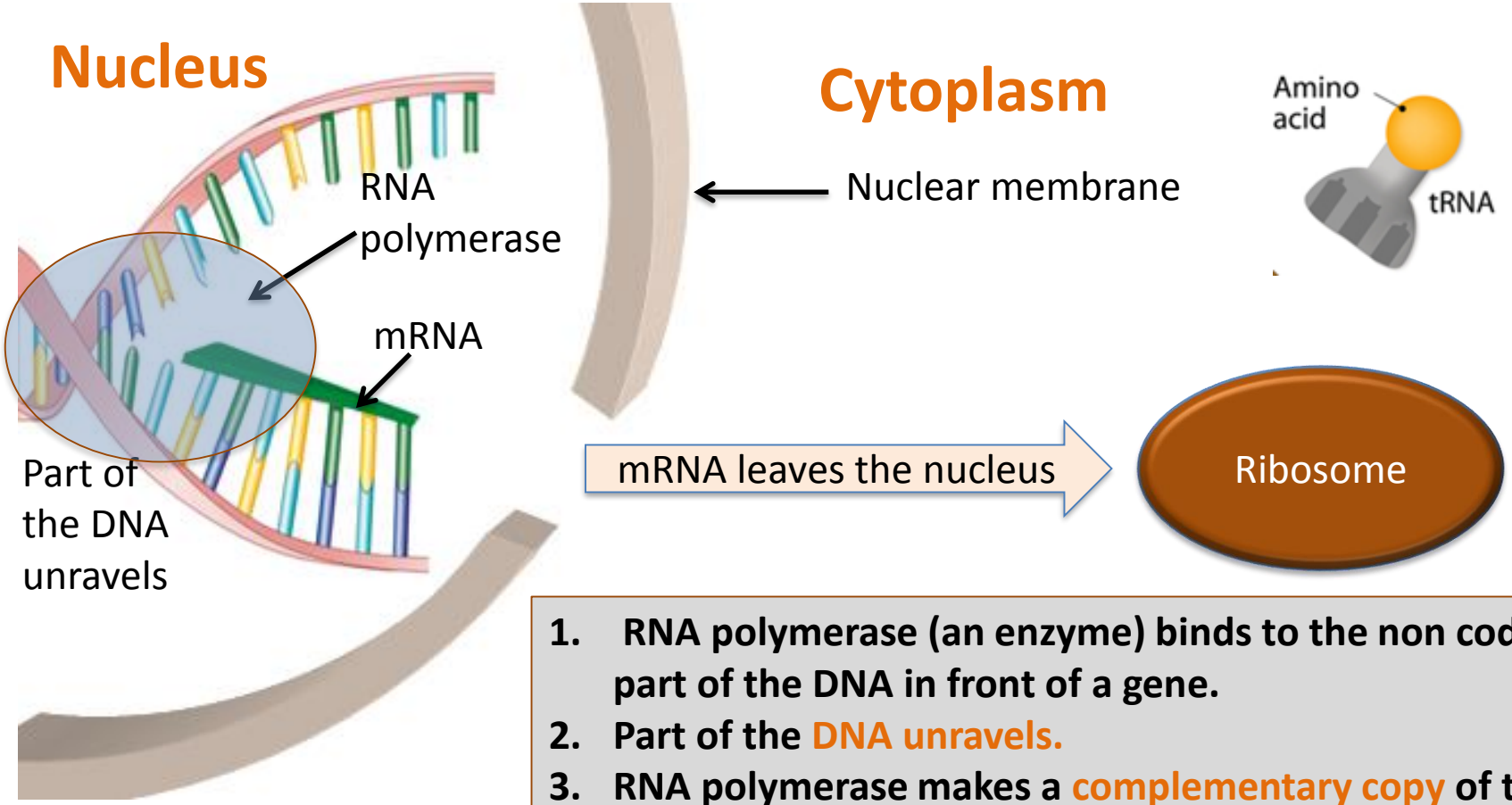
Every cell in an organism (apart from a gamete) with a nucleus will have a **complete set** of **chromosomes**. This means cells possess <u>every</u> gene that is needed to make every protein the organism ever needs to produce.

Most cells in the body will only **use** a very **small amount** of the **genes** available. If a **gene is activated** to convert the DNA code into a certain protein then we say the **gene** has been **expressed**. A muscle cell will never be required to make the protein used in the iris of the eye. This gene will not be expressed in the muscle cell.

Most of the **DNA does not** actually **code for proteins**. Research is still ongoing to find out what the function of the **non coding DNA** is. Non coding parts can **switch genes on or off** in a cell. **Mutations** in the **non coding DNA** may **affect** how **genes** are **expressed**.

Genetics part 2 – Protein synthesis (biology HT only)

Making new proteins: Transcription



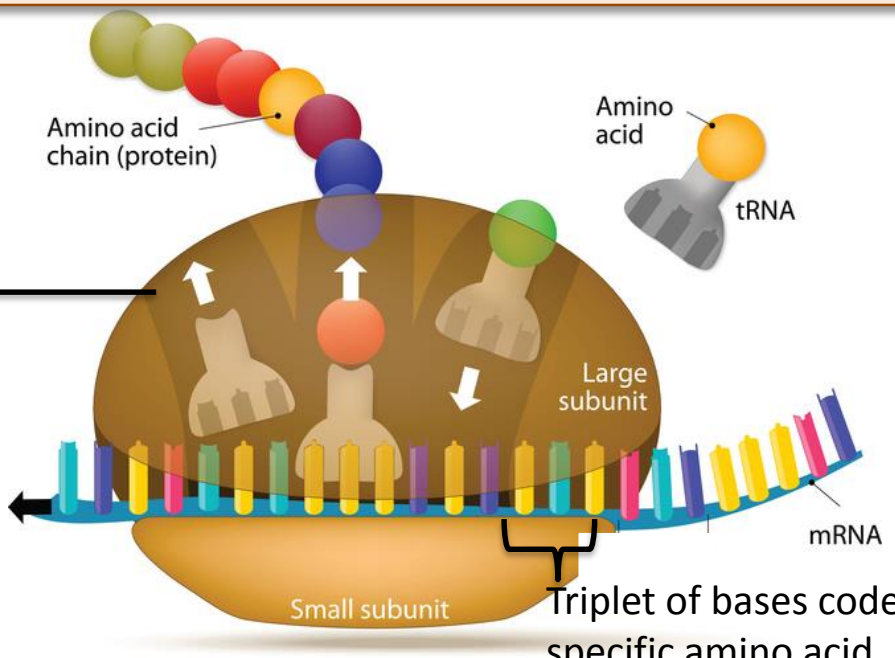
1. **RNA polymerase (an enzyme) binds to the non coding part of the DNA in front of a gene.**
2. **Part of the DNA unravels.**
3. **RNA polymerase makes a complementary copy of the coding DNA strand. This copy is called mRNA.**
4. **mRNA moves out of the nucleus into the cytoplasm. DNA is too large to leave the nucleus.**

Genetics part 2 – Protein synthesis(biology HT only)

Making new proteins: Translation

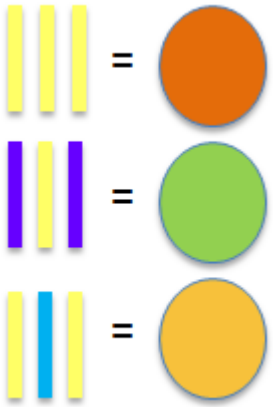
4. The mRNA travels to the ribosome in the cytoplasm and attaches to it.
5. Ribosomes translate each set of triplet bases into amino acids according to the mRNA template.
6. Amino acids are found in the cytoplasm. The correct amino acid for each triplet is brought to the ribosome by a carrier molecule called tRNA.
7. A long chain of amino acids form and link to form polypeptides. Their specific order forms a specific protein.

Ribosome



Triplet of bases codes for a specific amino acid

3 bases = amino acid

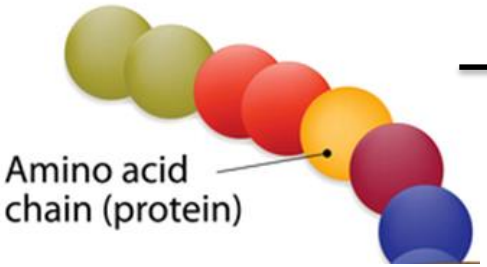


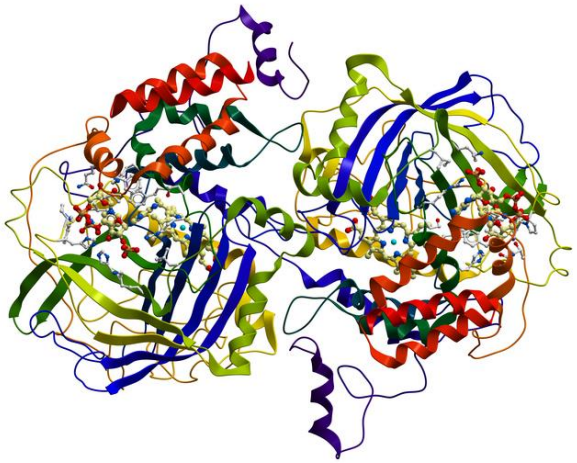
video

Genetics part 2 – Protein synthesis (biology HT only)

When the **protein chain** is complete, it **folds up** to form a **unique** shape.
This unique **shape** allows the protein to carry out its **role effectively**.

This could be as an enzyme, hormone or structures in the body such as collagen.





An enzyme

Genetics part 2 – Protein synthesis (biology HT only)

Mutations occur continuously producing genetic variants.

Prior to any **transcription** occurring, the **RNA polymerase** must **bind** to the **non-coding part** of the DNA in front of the gene.

If a **mutation** occurs in the **non-coding** part of the DNA then it may **affect** how well the **RNA polymerase** can **bind** to it.

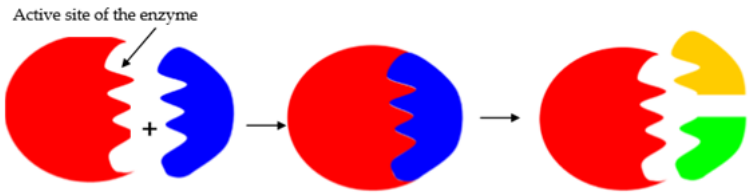
If **not all** the **mRNA** is **transcribed**, then this can **alter** the **quantity** of **protein** produced. This may **affect the function** or **phenotype** of the organism.

Genetic variations in the **non-coding DNA** can affect the **phenotype** of the organism even though this part of the DNA does not actually code for the protein.

Genetics part 2 – Protein synthesis (biology HT only)

Occasionally **a genetic variant in the coding DNA of a gene** results in an altered sequence of amino acids. This can produce an **altered protein** with a different shape which changes its activity.

Prior to a mutation



Active site binds with the substrate.

Following a mutation which results in an altered sequence of amino acids

Mutated active site has a different shape and no longer binds with the substrate.

The **active site** of an **enzyme** may have a **changed shape** and the enzyme will **not** be able to **bind** to the **substrate**. The enzyme will no longer function.

Structural proteins such as **keratin** or **collagen** may also **lose** their **strength** if their shape was altered.

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QuestionIT!

Genetics
Part 2

- DNA structure
- DNA and the genome
- Protein synthesis (biology HT)

Genetics part 2– QuestionIT

1. What is the definition of a polymer?
2. Name the shape which best describes the DNA polymer.
3. How is DNA organised inside the cell nucleus?
4. Which is larger, a chromosome or a gene?
5. What does a gene code for?
6. Where would you find a nucleotide?
7. What does a nucleotide consist of?
8. What are the symbols of the 4 bases found in DNA?
9. What does the code for an amino acid consist of?
10. a)Fill in the missing terms:

    The long strands of DNA are made of alternating _____ and _____ sections. Attached to each _____ is one of the four bases.
    The DNA polymer is made up of repeating _____ units. The strands of DNA are held together by _____ _____.
    b) What is the purpose of adding detergent to fruit when trying to extract its DNA?

Genetics part 2 – QuestionIT (biology only)

Higher Tier biology only

11. In the complementary strands of DNA – which base is T always linked to?
12. What type of molecule is RNA polymerase?
13. What is the role of RNA polymerase?
14. Which organelle in the cytoplasm carries out protein synthesis?
15. What do carrier molecules bring to this organelle from the cytoplasm?
16. What is this carrier molecule called?
17. Describe a codon.
18. What happens in protein synthesis once the protein chain is complete?
19. What happens if a mutation codes for a slightly altered enzyme protein with a different shape?
20. What can the non-coding parts of DNA do?

AnswerIT!

Genetics
Part 2

- DNA structure
- DNA and the genome
- Protein synthesis (biology HT)

Genetics part 2 – AnswerIT

1. What is the definition of a polymer?
   A polymer is a large molecule made from many smaller molecules called monomers
2. Name the shape which best describes the DNA polymer.
   Double helix
3. How is DNA organised inside the cell nucleus?
   Arranged in chromosomes
4. Which is larger, a chromosome or a gene? ***Chromosome***
5. What does a gene code for? ***A particular sequence of amino acids***
6. Where would you find a nucleotide? ***DNA***
7. What does a nucleotide consist of? ***Common sugar, phosphate group, base***
8. What are the symbols of the 4 bases found in DNA? ***A T C G***
9. What does the code for an amino acid consist of? ***A sequence of three bases***

10a. Fill in the missing terms:

The long strands of DNA are made of alternating ___*sugar*___ and ___*phosphate*____ sections. Attached to each ___*sugar*____ is one of the four bases.

The DNA polymer is made up of repeating _____*nucleotide*_____ units. The strands of DNA are held together by ___*hydrogen _bonds.*

10b.What is the purpose of adding detergent to fruit when trying to extract its DNA?

To break down cell membranes and release the DNA.

Higher Tier biology only

11. In the complementary strands of DNA – which base is T always linked to? *A*
12. What type of molecule is RNA polymerase?

    Enzyme or protein or polymer
13. What is the role of RNA polymerase?

    To bind to non-coding DNA in front of the gene and produce a complementary mRNA strand from the coding DNA of the gene.
14. Which organelle in the cytoplasm carries out protein synthesis? *Ribosome*
15. What do carrier molecules bring to this organelle from the cytoplasm? *Amino acids*
16. What is this carrier molecule called? *tRNA or transfer RNA*
17. Describe a codon

    A group of 3 nucleotides or bases which specifies the genetic code to synthesize a particular amino acid.
18. What happens in protein synthesis once the protein chain is complete?

    The protein folds up into a unique shape.
19. What happens if a mutation codes for a slightly altered enzyme protein with a different shape? *An enzyme may no longer fit the substrate binding site .*
20. What can non-coding parts of DNA do? *Switch genes on or off and affect how genes are expressed- the phenotype.*

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Genetics
Part 3

- The work of Mendel (biology)
- Genetic inheritance
- Sex determination
- ABO blood group inheritance

Genetics part 3 – The work of Mendel (biology)

Gregor **Mendel** was an Austrian Monk who was trained in mathematics and natural history. Whilst working in the monastery gardens, he studied the **inheritance** of **different characteristics** in **pea plants**.

Mendel observed that if he **cross bred red** flowering plants with **white** flowering plants, the majority of the **offspring** had red flowers but some were white.

He also cross bred **tall** plants with **short** plants, **green** and **yellow** peas, **wrinkled** and **smooth** peas and concluded there was a **predictable pattern of results**.

Gregor Mendel
(1822-1884)

"The father of modern genetics"

Genetics part 3 – The work of Mendel (biology)

Mendel kept meticulous **records** of the results of each cross and finally published them in **1866** along with his **hypothesis.**

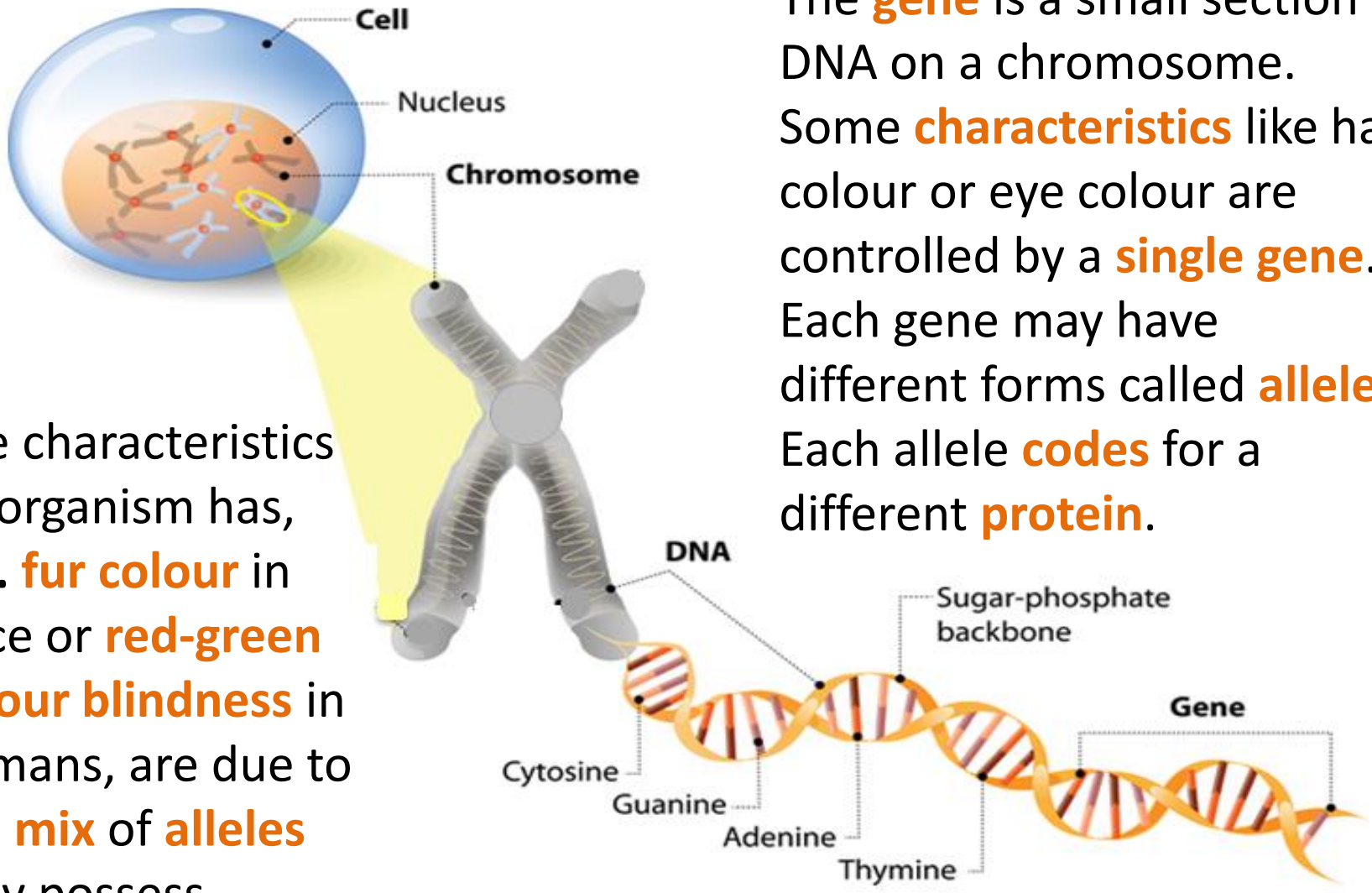
His hypothesis stated that:

- characteristics are held as **separate units of inheritance**
- the **units of inheritance** are **passed on** to offspring **unchanged**
- each unit of inheritance exists in **two forms** – **dominant** and **recessive.**

At the time scientists did **not know** about the existence of **chromosomes or DNA** and did not understand Mendel's work. It was almost twenty years after his death that scientists realised the **significance** of his findings. His work was repeated and **similar results** were achieved. **Chromosomes** were **seen** under the microscope and Mendel was given credit for his work.

The **units of inheritance** he proposed are now recognised as **genes**.

Genetics part 3 – Genetic Inheritance



The **gene** is a small section of DNA on a chromosome. Some **characteristics** like hair colour or eye colour are controlled by a **single gene**. Each gene may have different forms called **alleles**. Each allele **codes** for a different **protein**.

The characteristics an organism has, e.g. **fur colour** in mice or **red-green colour blindness** in humans, are due to the **mix** of **alleles** they possess.

Genetics part 3 – Genetic Inheritance

Each gene has different forms of alleles. The alleles which are present are known as the **genotype**. These are often represented as letters such as **BB**. The genotype operates at a molecular level to develop the actual characteristics seen or the **phenotype**.

Most genes have two possible allele variations which are known as **dominant** or **recessive.**

Dominant alleles are represented by a **capital letter** e.g. B

Recessive alleles are represented by a **lower case letter** e.g. b

There are **3** possible combinations of alleles for each gene:

Two dominant alleles	**BB**
Two recessive alleles	**bb**
One dominant and one recessive allele	**Bb**

(always place the dominant allele first and do not use bB)

Genetics part 3 – Genetic Inheritance

Homozygous

Homo means the same.
Two of the same alleles.
BB means homozygous dominant.
bb means homozygous recessive.

Genotype

This word describes the alleles which are present for a particular feature e.g. Bb.

You need to be able to use and explain these terms.

Phenotype

This word describes what can be physically seen - black fur, blonde hair, blue eyes.

Heterozygous

Hetero means different.
Two different alleles are present.
Bb means heterozygous.

Genetics part 3 – Genetic Inheritance

In a particular species of mouse, the **dominant allele** operates at a molecular level to produce proteins that make the fur black. The **recessive allele** codes for white fur.

Phenotype = black fur
Genotype = ?
At least **one dominant** allele (B) is present because the mouse has black fur.
The mouse could be **genotype** BB **or** Bb.

KEY

Use B and b to represent the dominant and recessive alleles.
B = allele for black fur
b = allele for white fur

Phenotype = white fur
Genotype = bb
We know there are **no dominant** alleles present because the fur is white.

A recessive allele is only expressed if two copies are present and therefore no dominant allele present.

Genetics part 3 – Genetic Inheritance

Genetic cross

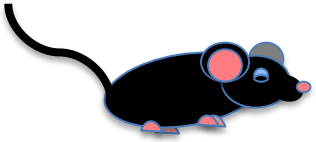
A **genetic cross** is a way of **modelling** the **potential outcome** from mating two parents where the **phenotype and genotype** are **usually known**. We use characteristics which are controlled by **a single gene** as it is easy to see the effect in the next generation and beyond.

A typical exam question might ask:

A female mouse which was **homozygous dominant** for black fur was mated with a male mouse which was **homozygous recessive** for white fur.

What are the possible outcomes for fur colour for their offspring?

What do we know?



Parent phenotype:	Black fur	White fur
Parent genotype:	BB	bb

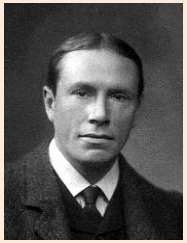
What gametes will be present?

in each egg	in each sperm
B B	b b

Genetics part 3 – Genetic Inheritance

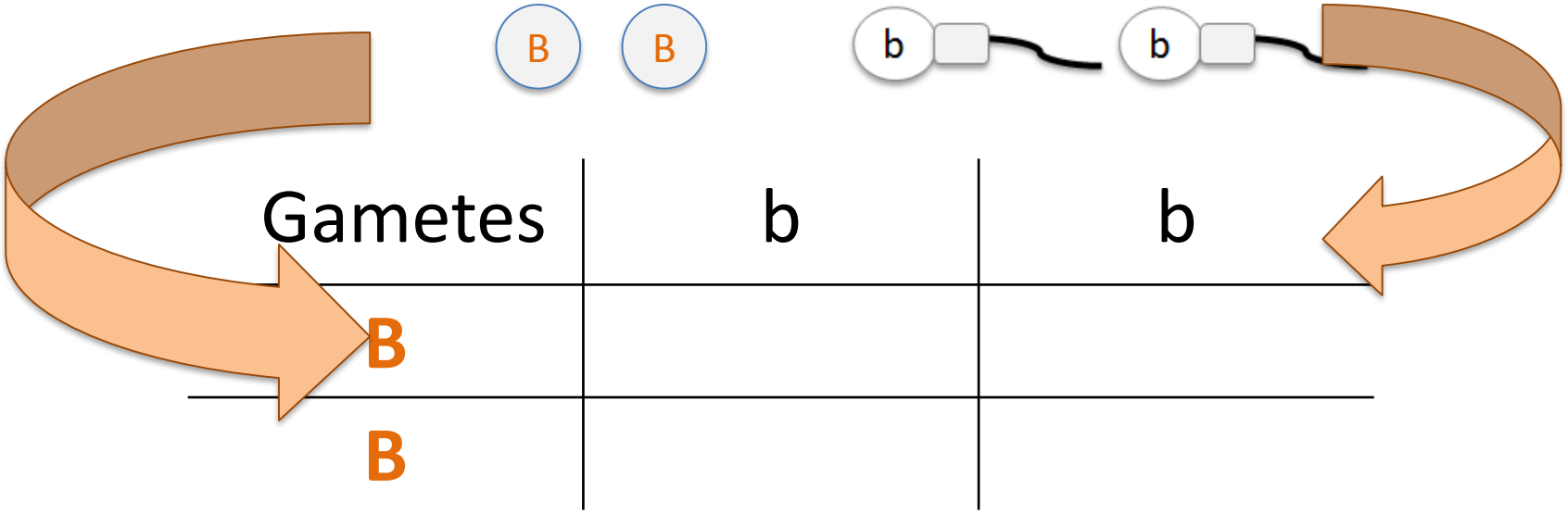
The Punnett Square

Reginald Punnett was a British geneticist who is most famous for creating the Punnett square diagram as a tool to predict the probability of genotypes in future offspring.



Parent phenotype: Black fur White fur

Parent genotype: BB bb

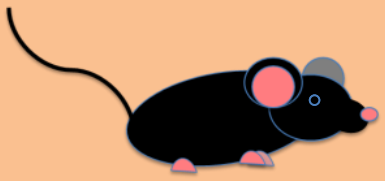


Genetics part 3 – Genetic Inheritance

If an egg containing a dominant allele (B) is fertilised by a sperm with a recessive allele (b) then the result is an offspring with genotype Bb.

Gametes	b	b
B	Bb	Bb
B	Bb	Bb

The possible offspring all have the **genotype Bb**.
This is described as **heterozygous**.



The **phenotype** of all offspring from these parents will be **black fur** because one dominant allele is always present.

Genetics part 3 – Genetic Inheritance

The **characteristic** of being a **tall** plant **or** a **short** pea plant is controlled by a **single gene**.
We will use the letter T to represent the gene for the purposes of a genetic diagram.
The allele which produces tall plants is **dominant** so we use **T**.
The allele for short plants is **recessive** so we use **t**.

Explain what would occur :

a) If two homozygous dominant plants were crossed.

The genotype for a homozygous tall plant is TT

Punnett square

Gamete	T	T
T	TT	TT
T	TT	TT

The genotype of **all** the future offspring will be TT and their phenotype will be **tall**. These parent plants will never produce short plants when crossed.

b) If two heterozygous plants were crossed.

The genotype for a heterozygous plant is Tt

Punnett square

Gamete	T	t
T	TT	Tt
t	Tt	tt

The **ratio** of **tall** plants to **short** plants likely to be produced is **3:1.**
In any four offspring, one would expect 1 homozygous dominant, 2 heterozygous and 1 homozygous recessive.

Genetics part 3 – Genetic Inheritance

Explain what would occur :

c) If a homozygous dominant plant and a homozygous recessive plant were crossed.

The **genotype** for a homozygous dominant plant is TT.
The **genotype** for a homozygous recessive plant is tt.

Punnett square

Gamete	t	t
T	Tt	Tt
T	Tt	Tt

The genotype of **all** the future offspring will be Tt and their phenotype will be **tall**. All offspring will be heterozygous. The probability of getting a tall plant is described as 1 or 100%.

b) If two homozygous recessive plants were crossed.

The **genotype** for a homozygous recessive plant is tt.

Punnett square

Gamete	t	t
t	tt	tt
t	tt	tt

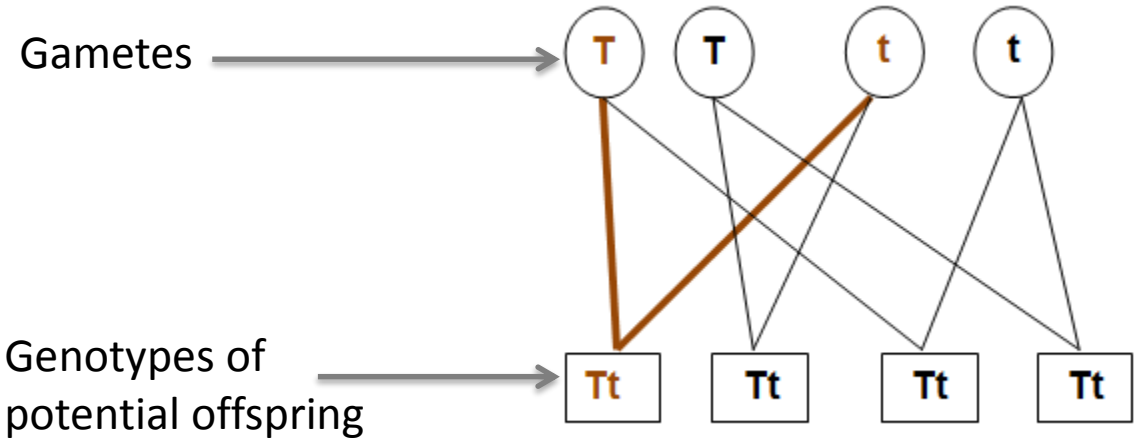
All future offspring will be **homozygous recessive**. Their **phenotype** will be **short.** These parents will **never** produce tall plants.

video

Genetics part 3 – Genetic Inheritance

Not all exam questions use the Punnett square layout to find out possible genotypes. The basic principle is always the same.

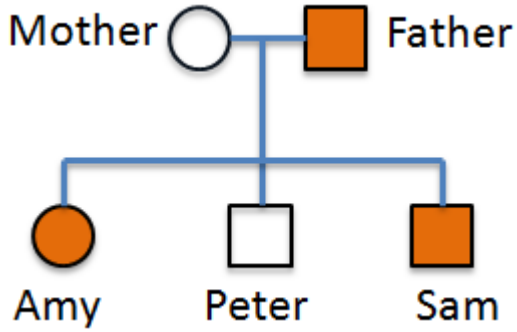
A genetic diagram could also look like this:



Genetics part 3 – Genetic Inheritance

A **family tree** can help to show how **genetic disorders** are inherited in a family. They can be used to work out the **probability** that a member of the family will **inherit** a disorder.
Read the **key** carefully to help you interpret a family tree accurately.

The family tree below shows the inheritance of a disorder caused by a **dominant** allele.



Key

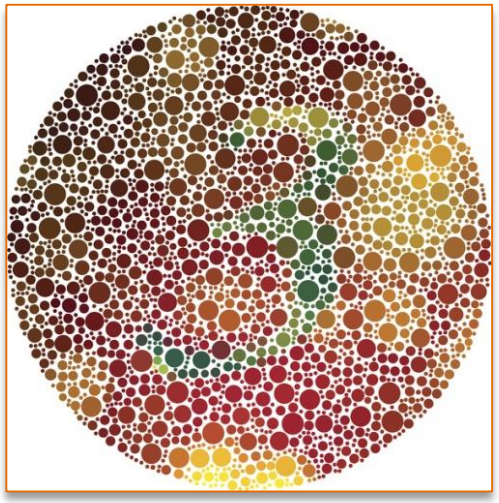
- Female without disorder
- Female with disorder
- Male without disorder
- Male with disorder

What is the genotype of the mother? **Homozygous recessive**
What is the genotype of the father? **Heterozygous**

If the father was **homozygous dominant** then **all** the **offspring** would have the disorder. Peter does not have the disorder and is **homozygous recessive.**

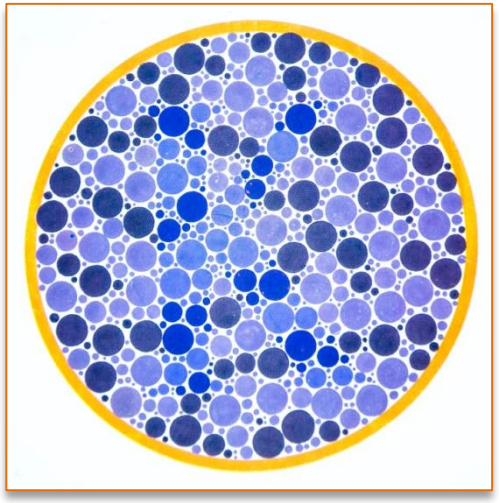
Genetics part 3 – Sex-Linked genetic disorder (biology HT)

Colour blindness is a **sex-linked genetic disorder** that a person is born with. It is caused by a faulty allele carried on the **X chromosome**.



This **gene** is **not found** on the much smaller **Y chromosome.**

Women need **two copies** of the faulty **recessive** allele to be colour blind, whilst **men** only need **one copy**.



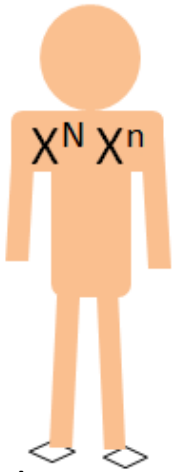
More men than women are affected by the disorder.

Genetics part 3 – Genetic Inheritance

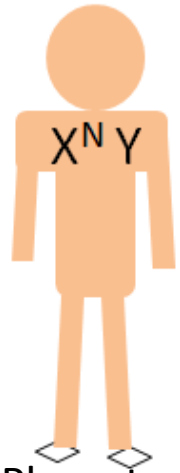
A **woman** with only **one copy** of the **recessive allele** is a **carrier** of colour blindness but is **not affected** herself.

The diagram shows a genetic cross between a **female carrier** and a **male** who has **normal** vision.

In a sex-linked genetic cross, we use the sex chromosomes (X or Y) with a super script symbol to represent the allele.



Phenotype : female normal vision (carrier)



Phenotype : male normal vision

Key

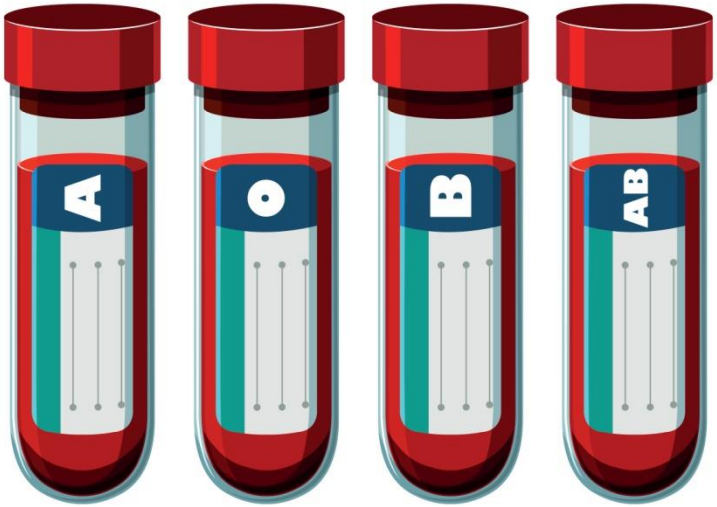
N= normal vision allele
n = colour blind allele

Punnett square

Gamete	X^N	X^n
X^N	$X^N X^N$	$X^N X^n$
Y	$X^N Y$	$X^n Y$

For this cross the **ratio** of **normal** vision **to colour blind** offspring is **3:1** .
There is a **1 in 4** chance (**25%**) of a child from these parents **being colour blind.**
This increases to **50%** if the child is a **male.**

Genetics part 4 – ABO inheritance (biology)



Human **blood group** is **inherited**.
There are **four** possible blood groups:
A, B, AB or O.

The gene for blood group has **three** different **alleles**: **I^A , I^B and I^O**

I^A and I^B alleles are **co-dominant**.
This means that if a person has the genotype I^A I^B then they are blood group AB. **Both alleles** are equally **dominant**.

I^O is **recessive** to both other alleles.

If a person has the **genotype** I^A I^O then the person has the blood group or **phenotype** A.

A person with **two recessive** alleles (I^O I^O) is blood group **O**.

Genetics part 4 – ABO inheritance (biology)

Genetic diagrams for co-dominant alleles can look complicated. A male with blood group B has a child with a woman with blood group A. What are the possible genotypes of the offspring?

	Male	Female
Phenotype:	Blood group B	Blood group A
Genotype:	I^BI^O	I^AI^O
Gametes:	I^B I^O	I^A I^O

Genetic Cross:

Gametes	**I^B**	**I^O**
I^A	**I^AI^B**	**I^AI^O**
I^O	**I^BI^O**	**I^OI^O**

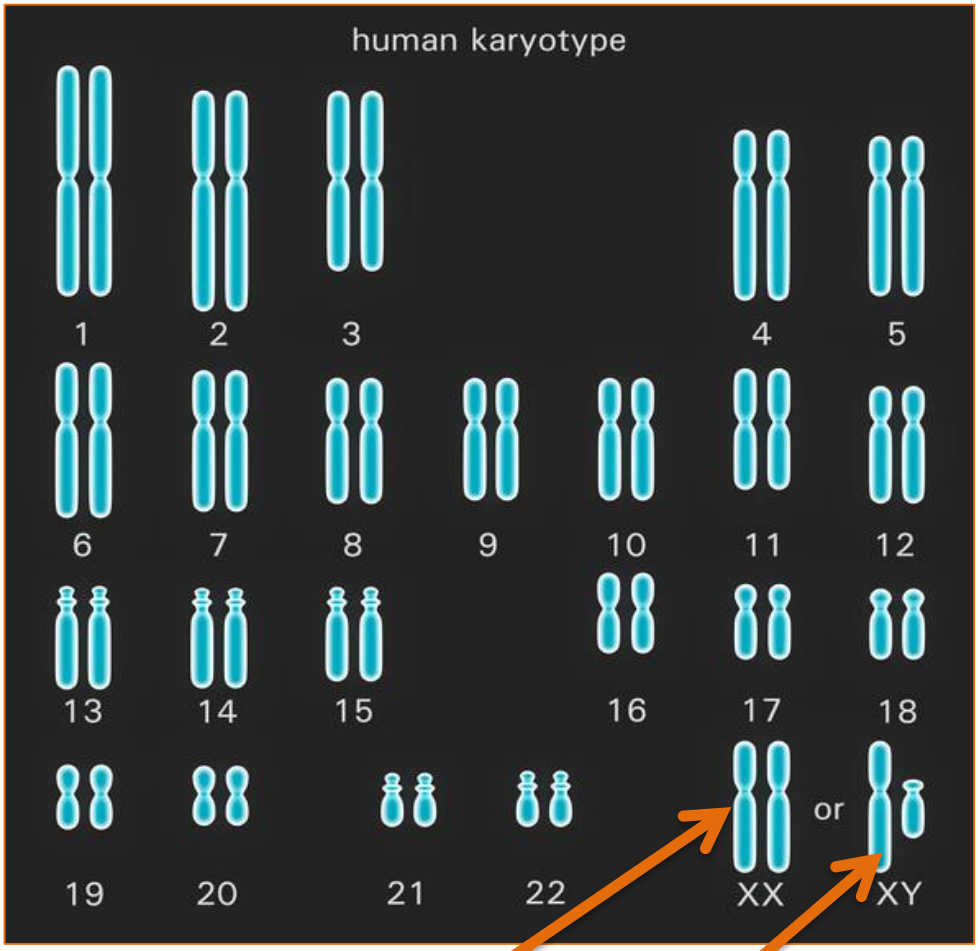
Offspring phenotypes: AB, A, B and O. From these parents, there is a **1 in 4 chance** or **25%** of the offspring being any of the four blood groups.

Genetics part 4 – Sex determination

An ordinary **human body cell** contains **23 pairs** of chromosomes. One chromosome of each pair comes from the egg and one from the sperm.

22 pairs of chromosomes control characteristics.

The **23rd pair** of chromosomes are called **sex chromosomes**. This pair carry the genes that determine sex. The sex chromosomes are **not identical** to each other and so are called **X** and **Y.** Sex is determined at **fertilisation.**



In **females** the sex chromosomes are the same **-XX**

In **males** the sex chromosomes are different-**XY**

Genetics part 4 – Sex determination

How is sex inherited?

The 23rd pair of chromosomes are responsible for determining the sex of a human.

The **Punnett square** is used to show the chances of an offspring being male or female.

A **woman** has the **genotype XX** and a **man** has the **genotype XY**.

Gametes	X	X
X	XX	XX
Y	XY	XY

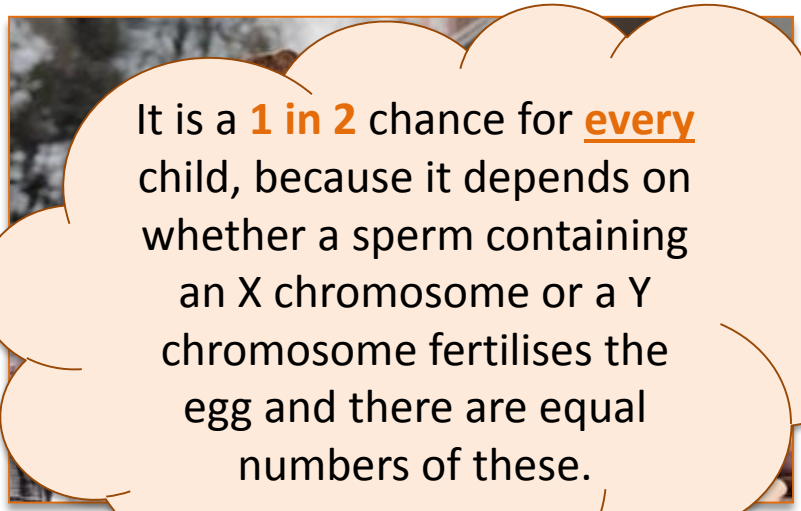
There is a **1 in 2** chance of the offspring being male or female.

This can also be described as a ratio of **1:1, 50:50** or **50%** or **0.5** of being male or female.

Genetics part 3 – Sex determination

A man and woman have three children – all boys. The woman is pregnant with their fourth child.

What is the chance of the next child being a girl?



It is a **1 in 2** chance for **every** child, because it depends on whether a sperm containing an X chromosome or a Y chromosome fertilises the egg and there are equal numbers of these.

I think it is more likely to be a girl as they already have 3 boys .

I think there is **more chance** of the new baby being a boy because they **have 3 boys already**.

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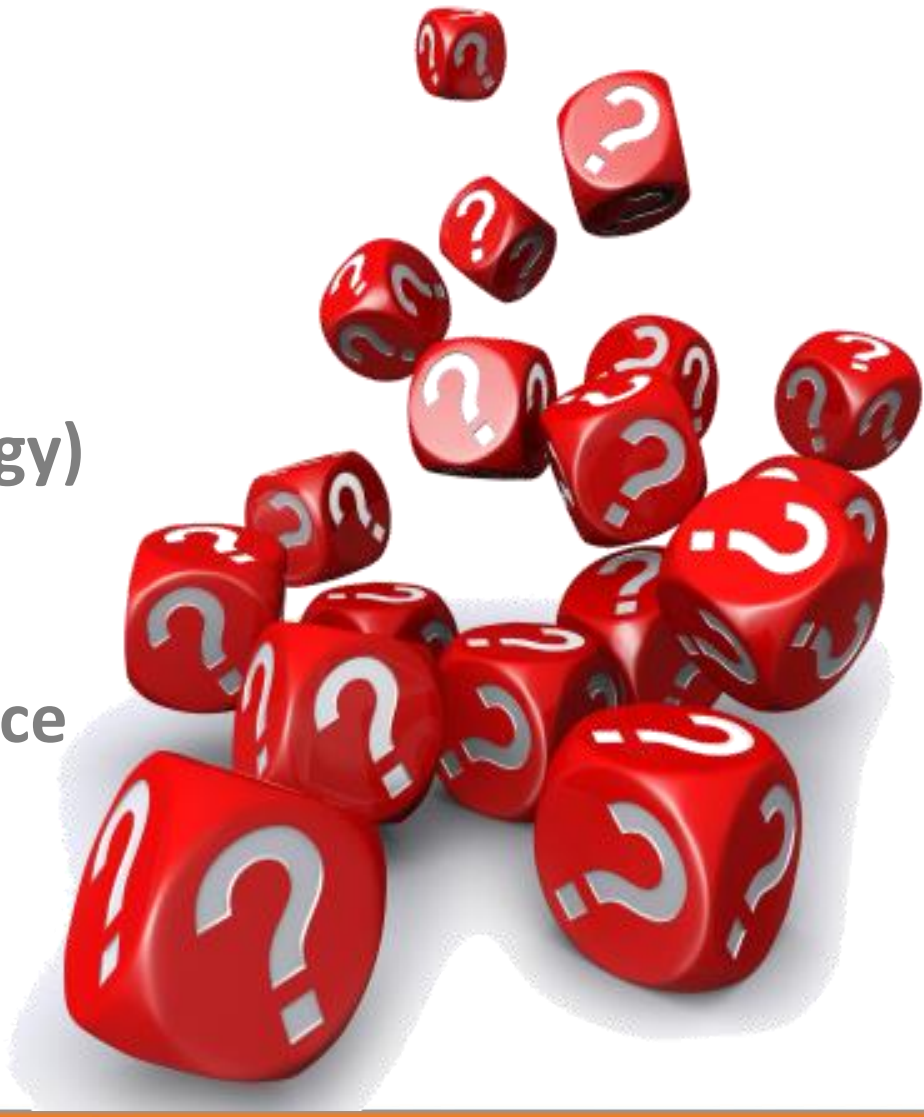
QuestionIT!

Genetics

Part 3

- The work of Mendel (biology)
- Genetic inheritance
- Sex determination
- ABO blood group inheritance



Genetics part 3 – QuestionIT

1. Complete the sentences below using one of the following words:

genotype *phenotype* *homozygous* *gamete* *chromosome*

The same alleles for a particular characteristic are described as .....
Mice fur can be described by its colour. This is known as the .....
The alleles for a particular characteristic determine its .....
The pollen of a rose plant can also be described as a male .....

2. What are the different forms of a gene called ?

3. Why is it easy to describe an organisms phenotype for a particular characteristic but very difficult to state the genotype?

4. List the alternative ways of describing the following outcome of a genetic cross:

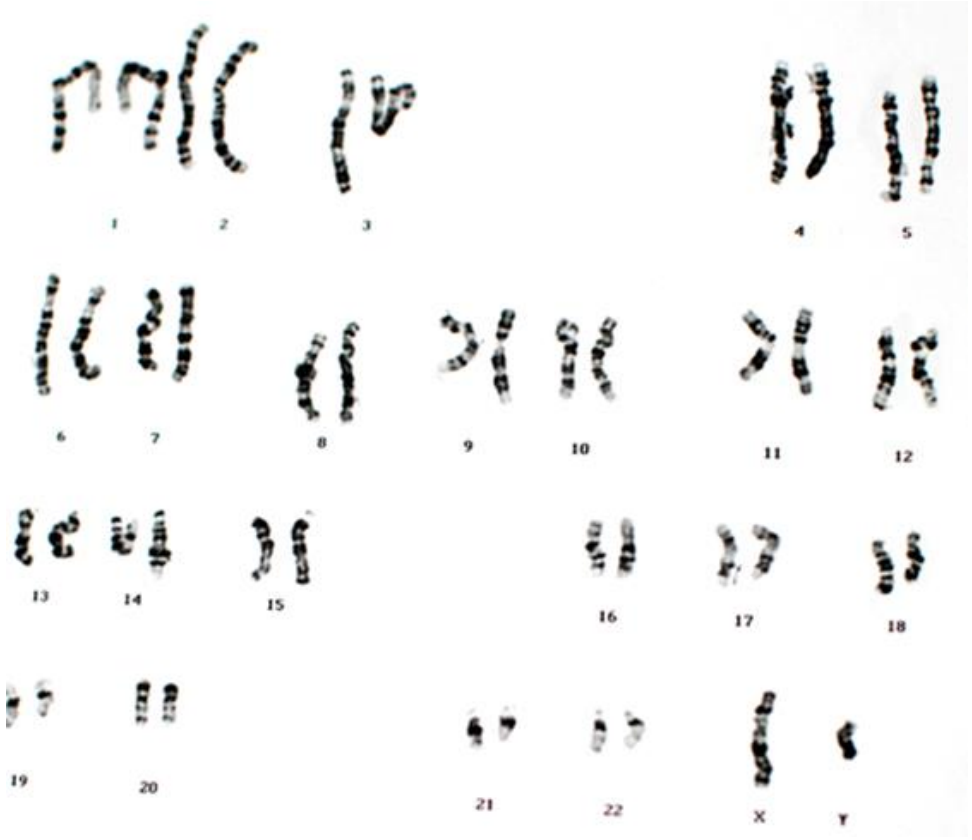
3 in every 4 crosses will have black fur.

5. If two heterozygous tall plants were crossed 75 times, what proportion of the offspring would you expect to be short?

Genetics part 3 – QuestionIT

Look at the following picture of a persons chromosomes.

6. How many chromosomes does this person have in a normal body cell?
7. What sex is the person and how do you know?



8. Black fur is dominant to brown fur in mice.
Use B to represent black fur and b to represent brown fur.

a) Complete the Punnett square diagram to show the outcome of a genetic cross between two heterozygous mice.

Gametes		

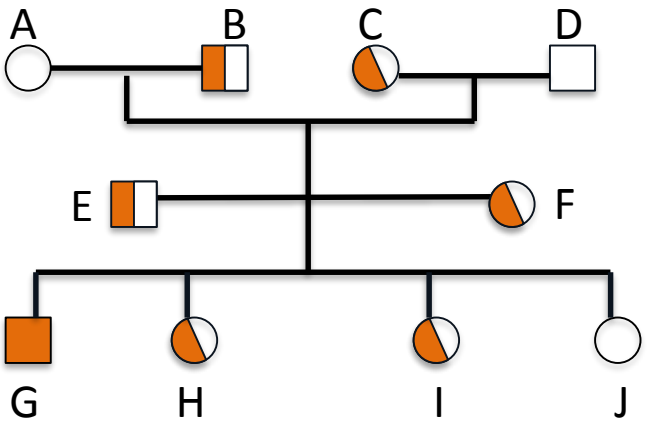
b) What is the phenotype of the parent mice?

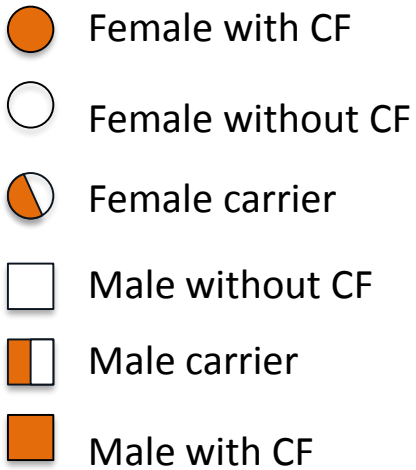
c) What are the chances of a homozygous recessive mouse being born to two heterozygous mice?

9. What is meant by the term zygote?

Genetics part 3 – QuestionIT

10. The family tree shows the inheritance of cystic fibrosis (CF). Cystic fibrosis is a condition caused by a recessive allele. Let F = healthy allele and f = CF allele





a) What is the genotype of a female carrier?
b) Explain why person G inherited cystic fibrosis.
c) Explain why person J did not inherit cystic fibrosis.
d) What is the probability of a fifth child having cystic fibrosis?
e) Many people feel opposed to the screening of embryos to identify the CF allele. Suggest **two** reasons why they may feel this way.

Genetics part 3 – QuestionIT

11. The flower colour for pea plants is controlled by a single gene. Red flowers are dominant to white flowers. A homozygous dominant plant and a heterozygous plant are crossed to produce offspring.

a) What is the phenotype of the homozygous dominant plant?

b) Draw a genetic diagram to show all the possible arrangements of alleles in the offspring.

c) What is the chance of producing homozygous dominant red flowering plants?

Biology only

12. What were the three conclusions Mendel proposed after his work on the inheritance in peas?

13. What made it hard for scientists at the time to understand Mendel's ideas?

14. How many alleles exist for inheritance of blood group?

15. What is meant by co-dominance?

16. If someone has a blood group of A, what alleles do they possess?

Genetics part 3 – QuestionIT (biology only)

17. A woman with the genotype $X^N X^N$ for colour blindness (where N = the allele for normal vision) can be described as:

Tick all word/s that apply .

Colour blind		**Normal vision**		**Carrier**	
Homozygous dominant		**Heterozygous**		**Homozygous recessive**	

AnswerIT!

Genetics

Part 3

- The work of Mendel (biology)
- Genetic inheritance
- Sex determination
- ABO blood group inheritance

Genetics part 3 – AnswerIT

1. Complete the sentences below using one of the following words:

   genotype *phenotype* *homozygous* *gamete* *chromosome*

   The same alleles for a particular characteristic are described as ***homozygous***
   Mice fur can be described by its colour. This is known as the ***phenotype***
   The alleles for a particular characteristic determine its ***genotype***
   The pollen of a rose plant can also be described as a male ***gamete***

2. What are the different forms of a gene called ?

   Alleles

3. Why is it easy to describe an organisms phenotype for a particular characteristic but very difficult to state the genotype?

   Most characteristics are a result of multiple genes interacting .

4. List the alternative ways of describing the following outcome of a genetic cross:

   3 in every 4 crosses will have black fur.

   75% 0.75 3/4

5. If two heterozygous tall plants were crossed 75 times, what proportion of the offspring would you expect to be short?

   25% or 19

Genetics part 3 – AnswerIT

Look at the following picture of a persons chromosomes.

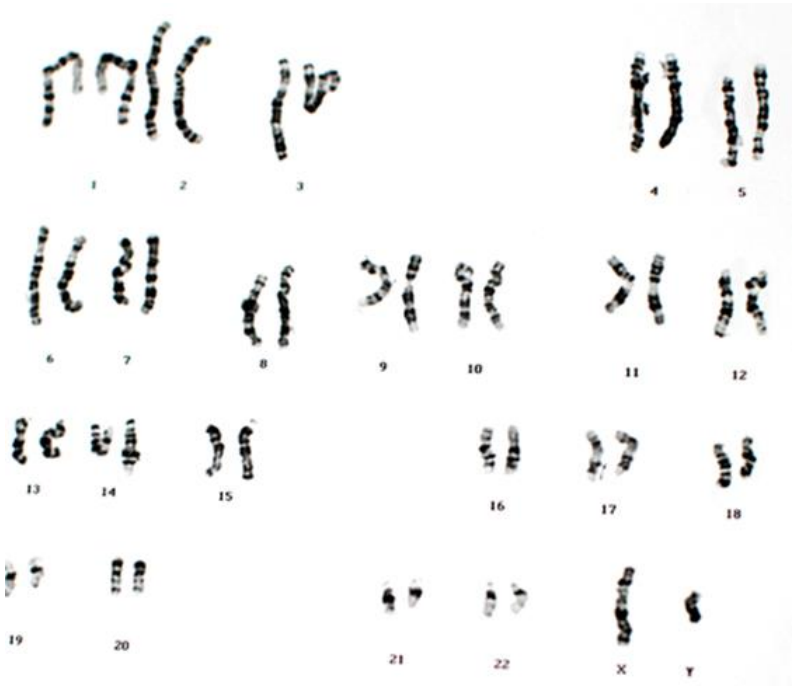
6. How many chromosomes does this person have in a normal body cell?

23 pairs or 46

7. What sex is the person and how do you know?

A male

Because the sex chromosomes are different (XY)



Genetics part 3 –AnswerIT

8. Black fur is dominant to brown fur in mice.
Use B to represent black fur and b to represent brown fur.
a) Complete the Punnett square diagram to show the outcome of a genetic cross between two heterozygous mice.

Gametes	*B*	*b*
B	*BB*	*Bb*
b	*Bb*	*bb*

b) What is the phenotype of the parent mice?

Black fur

c) What are the chances of a homozygous recessive mouse being born to two heterozygous mice?

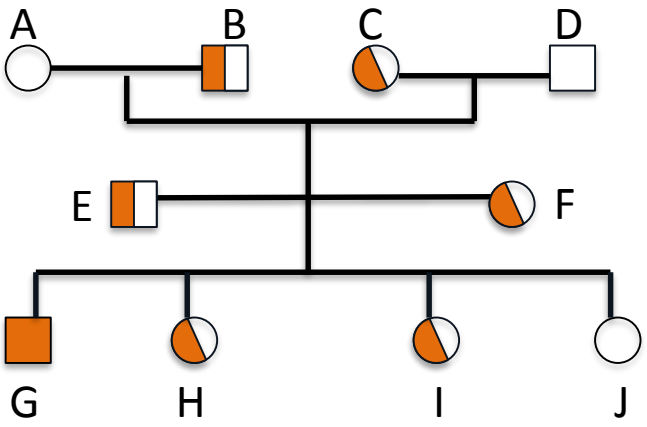
1 in 4 or 25% or 0.25

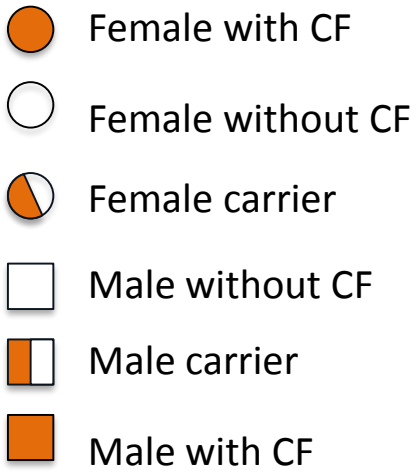
9. What is meant by the term zygote?

A zygote is the single cell formed when the gametes fuse at fertilisation.

Genetics part 3 – AnswerIT

10. The family tree shows the inheritance of cystic fibrosis (CF). Cystic fibrosis is a condition caused by a recessive allele. Let F = healthy allele and f = CF allele





a) What is the genotype of a female carrier? ***Ff***

b) Explain why person G inherited cystic fibrosis

Inherited a recessive allele from both E and F.

As no dominant allele present, the CF alleles code for faulty cell membranes

c) Explain why person J did not inherit cystic fibrosis.

Inherited two dominant alleles from E and F

d) What is the probability of E and F having a fifth child with cystic fibrosis?

1 in 4, 25%, 0.25, 1:3 or ¼

Genetics part 3 – AnswerIT

e) Many people feel opposed to the screening of embryos to identify the CF allele. Suggest **two** reasons why they may feel this way.

Screening procedure may cause miscarriage or damage embryo/fetus
The embryo has a right to life
Screening is expensive and could be used for other purposes
Have to make moral/ethical or religious decisions as a result such as whether to terminate.

Genetics part 3 – AnswerIT (HT only)

Higher tier question

11. The flower colour for pea plants is controlled by a single gene. Red flowers are dominant to white flowers.
A homozygous dominant plant and a heterozygous plant are crossed to produce offspring.
Let R represent the dominant allele.

a) What is the phenotype of the homozygous dominant plant?

Red flowers

b) Draw a genetic diagram to show all the possible arrangements of alleles in the offspring.

Gamete	*R*	*r*
R	*RR*	*Rr*
R	*RR*	*Rr*

c) What is the chance of producing homozygous dominant red flowering plants?

1 in 2 50:50 50% or ½ or 1:1

Genetics part 3 – AnswerIT

Biology only

12. What were the three conclusions Mendel proposed after his work on the inheritance in peas?

that characteristics are held as separate units of inheritance
the units of inheritance are passed on to offspring unchanged
each unit of inheritance exists in two forms – dominant and recessive.

13. What made it hard for scientists at the time to understand Mendel's ideas?

they did not know about the existence of chromosomes or DNA

14. How many alleles exist for inheritance of blood group?

Three different alleles: I^A , I^B and I^O

15. What is meant by co-dominance?

alleles are equally dominant.

16. If someone has a blood group of A, what alleles do they possess?

EITHER I^A I^A OR I^A I^O

17. A woman with the genotype X^N X^N for colour blindness (where N = the allele for normal vision) can be described as:
Tick all word/s that apply .

Colour blind		**Normal vision**	✓	**Carrier**	
Homozygous dominant	✓	**Heterozygous**		**Homozygous recessive**	

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LearnIT!
KnowIT!

Genetics
Part 4

- Variation
- Human Genome Project
- Mutations

Genetics part 4 – Variation

Variation means that individuals in a population show **differences** in characteristics. Population is the number of one species in a habitat. The **genome** and its **interaction** with **the environment** influence the development of a **phenotype** in an organism.

Variation within a **population** of a species is usually **extensive,** mostly arising from **mutations**.

Causes of variation may be:

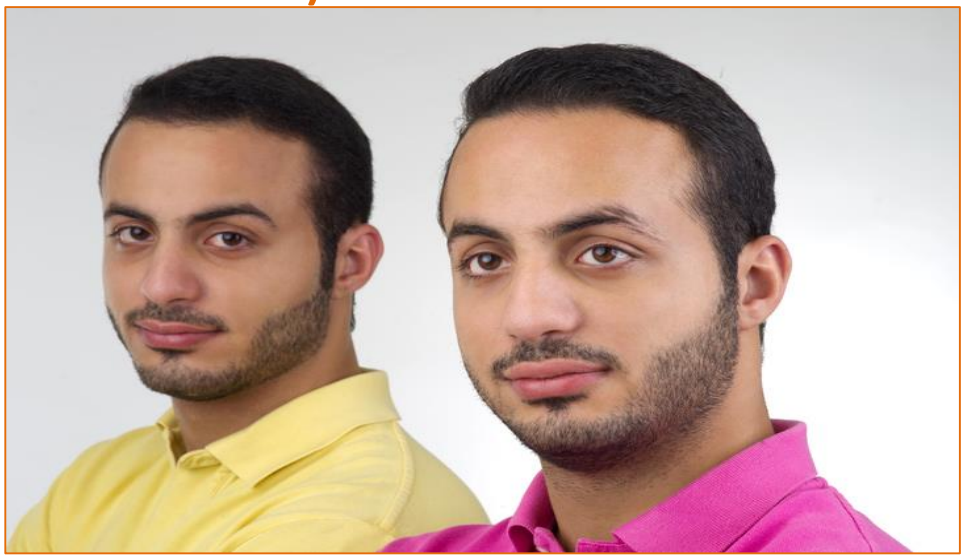
❑ **differences** in the **genes** individuals have inherited due to **mutation** and **sexual reproduction.**

❑ **differences** in the **environmental** conditions have led to variation **(acquired characteristics).**

❑ or a **combination** of both genetic and environmental causes.

Genetics part 4 – Variation

Identical twins are produced from the same egg and sperm. They show **very little** or no **genetic variation**. One twin has a scar and this is **environmental** variation (an **acquired characteristic)**



Factors which are **influenced** by **both genetic and environmental** variation are:
skin colour (can be tanned),
weight (can be affected by food availability) or being athletic.

Human phenotypes which are caused by **genetic variation** are:
Eye colour
Natural hair colour
Nose shape
Ear lobe shape
Blood group

Human phenotypes influenced by the **environment** are:
Hair length
Accents
Tattoos
Scars
Language spoken

Genetics part 4 – Variation

- **Most phenotypic features** are actually a result of **multiple genes** interacting, rather than single gene inheritance.

- We are able to describe a **phenotype** for these features **but cannot write down a genotype** because more than one gene is involved.

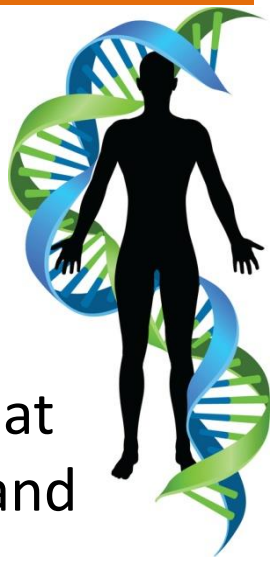
- Some characteristics in a species show **continuous variation**. There is a **range** of values and an individual can be anywhere in between the maximum and minimum. **Height** is an example.

- **Discontinuous variation** is when there is a limited number of values that an individual can possess. There is **no range**. **Blood group** and **gender** are examples of discontinuous variation.

Human eye colour is an example of when **multiple** genes affect the phenotype.

Genetics part 4 –DNA and the genome

The **genome** of an organism is defined as the **entire genetic material** of that organism.



The **Human Genome Project** (HGP) was an international scientific research project set up to **map all the genetic information** in a human being.

It began in 1990 and was completed in 2003. The whole **human genome has now been studied** and this will have great significance for **medicine** in the future. This work to understand the human genome is important for several reasons:

- ❑ To enable scientists to search for the **genes linked to different types of disease** to look for possible treatment or correction.
- ❑ To enable doctors to better **understand and treat inherited disorders** e.g. cystic fibrosis, sickle cell anaemia, thalassemia.
- ❑ To be able to trace historic **human migration patterns.** **video**

Genetics part 4 – Mutations

All **genetic variations** arise from **mutations**.

A mutation is a change in the DNA sequence of an organism. **Mutations** are occurring **continuously** during cell division. **Most** genetic mutations **do not alter** the **phenotype.** Some mutations will have a small effect on the phenotype, and rarely a single mutation will significantly affect the phenotype. If a **new phenotype** is **suited** to an environmental change, it can **lead** to a relatively **rapid change** in the **species**. An example is seen below:

The peppered moth is camouflaged against tree bark. Birds cannot see it easily. The peppered moth lives long enough to breed and pass on its genetic information. This phenotype is found mainly in the countryside now.

During the industrial revolution, tree bark and buildings in cities and towns became blackened. The peppered moth became easy prey for the birds. A mutation occurred which changed the colour of the moth to black. The black phenotype is now found in large numbers in cities.

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QuestionIT!

Genetics

Part 4

- Variation
- Human Genome Project
- Mutations

Genetics part 4 – AnswerIT

1. Write down the definition of the genome of a horse.

2. List **two** reasons why it is important to understand more about the human genome.

3. What can bring about genetic variation in a species?

4. What is meant by ‘an acquired characteristic’?

5. What is the chance of most genetic mutations having an effect on the phenotype? **High, Medium or Low**

6. What is meant by discontinuous variation?

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AnswerIT!

Genetics
Part 4

- Variation
- Human Genome Project
- Mutations

Genetics part 4 – AnswerIT

1. Write down the definition of the genome of a horse.
   All the genetic material of the horse.
2. List **two** reasons why it is important to understand more about the human genome.
   To be able to identify the genes which are linked to different diseases.
   To understand and be able to treat inherited disorders.
   Useful in tracing human migration patterns from the past.
3. What can bring about genetic variation in a species?
   Mutations
4. What is an acquired characteristic?
   A characteristic caused by the organisms environment e.g. a scar
5. What is the chance of most genetic mutations having an effect on the phenotype? **High, Medium or Low**
   Low – very few mutations have an effect on the phenotype
6. What is meant by discontinuous variation?
   A feature has a limited number of possible values. e.g. gender