DP IB Biology: HL

Your notes

3.3 Inheritance

Contents

3.3.1 Inheritance


Your notes

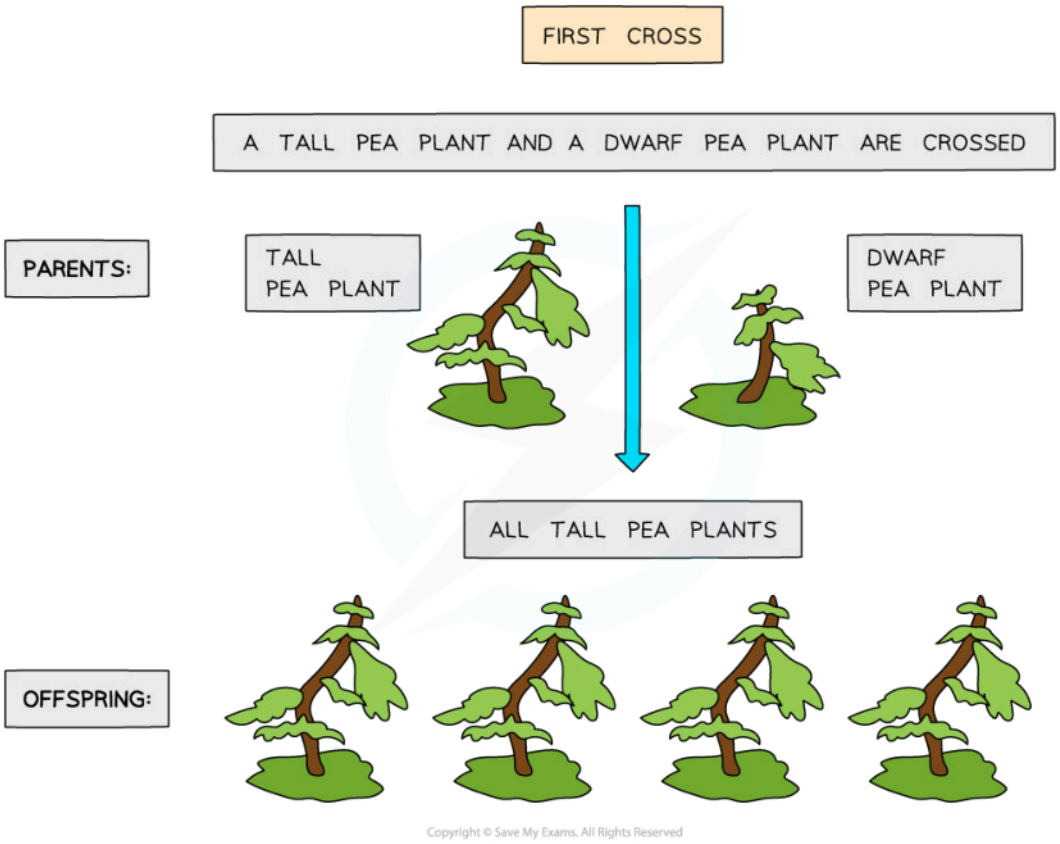
Mendel's Experiments

- **Gregor Mendel** was an Austrian monk
- He was trained in mathematics and natural history at the University of Vienna
- In the mid-19th century, Mendel carried out breeding experiments on large numbers of pea plants whilst looking after the monastery gardens
- He studied how **characteristics were passed on between generations of plants**
- Due to his extensive work on the understanding of inheritance, he is sometimes called the **father of genetics**

Mendel's groundbreaking work

- Mendel carefully **transferred pollen** from one pea plant to the reproductive parts of another
- This technique **eliminated any uncertainty** from his data since he knew which plants were fertilized by which pollen
- He **collected the pea seeds from these plants** and grew them in favourable conditions to find out their characteristics
- He also **cross-bred offspring peas** in order to find out which, if any characteristics would appear in future generations
- Mendel investigated the **height** of pea plants, the **colours of their flowers** and the **smoothness of their seed coat**

Your notes



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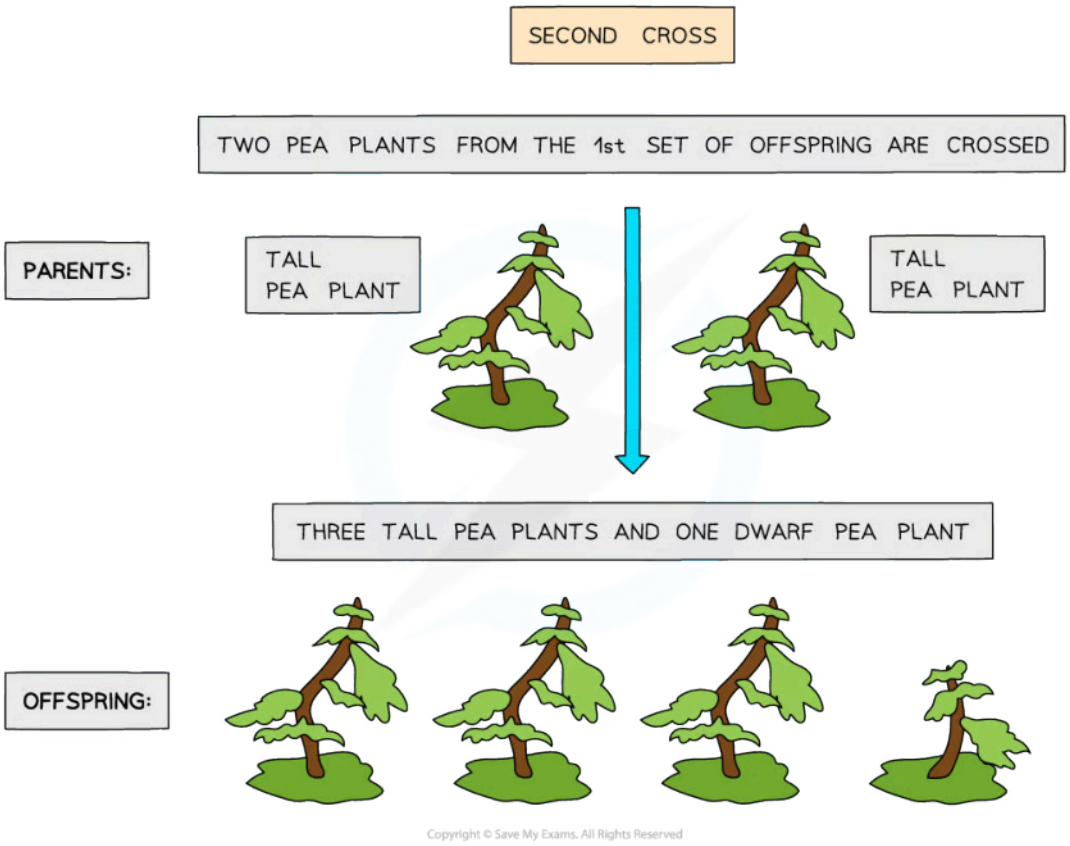


Mendel's pea plant crosses

Mendel's Pea Plant Results Table

Parental characteristics	Characteristics of first generation plants	Chraracteristics of second generation plants	Ratio of characteristics in second generation
Tall plant × dwarf plant	100% tall plants	868 tall plants and 277 dwarf plants	3.1 : 1
Round seed coat × wrinkled seed coat	100% Round seed coat	5474 round seed coat and 1850 wrinkled seed coat	3 : 1
Purple flowers × white flowers	100% Purple flowers	705 purple flowers and 224 white flowers	3.1 : 1

- Mendel found that characteristics were **inherited** in a **predictable pattern**
- **All** pea plants in the first generation had the same characteristic as **one** of the parental plants
- The offspring plants in the second generation had characteristics of **both** parent plants in a **3:1 ratio**
- Without knowing it, Mendel had discovered genes, he referred to them as **'units of inheritance'**
- He also discovered that some genes are **dominant** and some genes are **recessive**
- Different forms of the same gene are called **alleles**


Your notes

Mendel's Experimental Technique

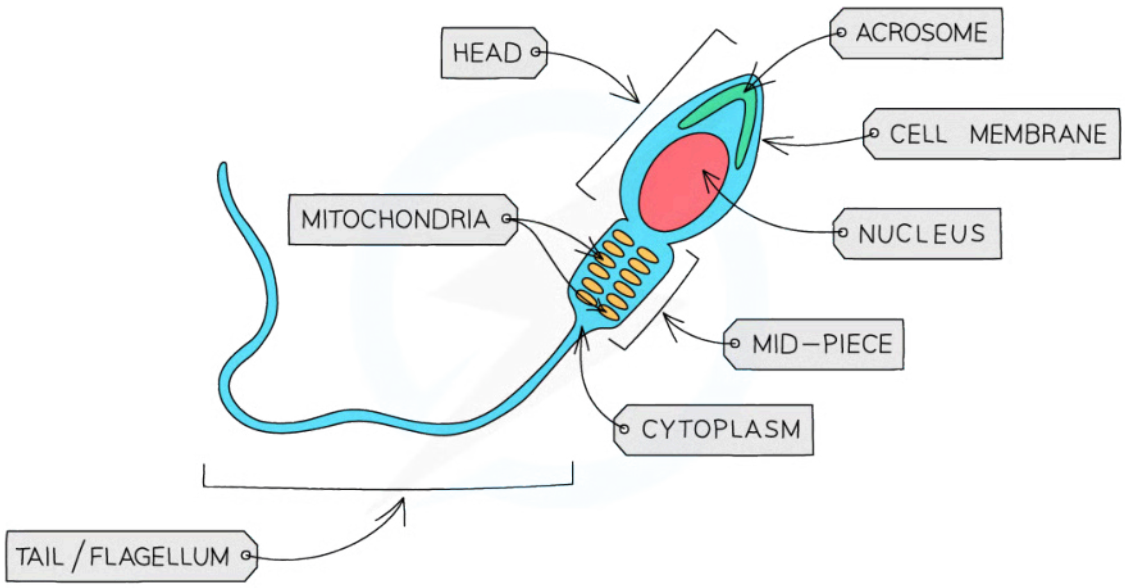
NOS: Making quantitative measurements with replicates to ensure reliability; Mendel's genetic crosses with pea plants generated numerical data

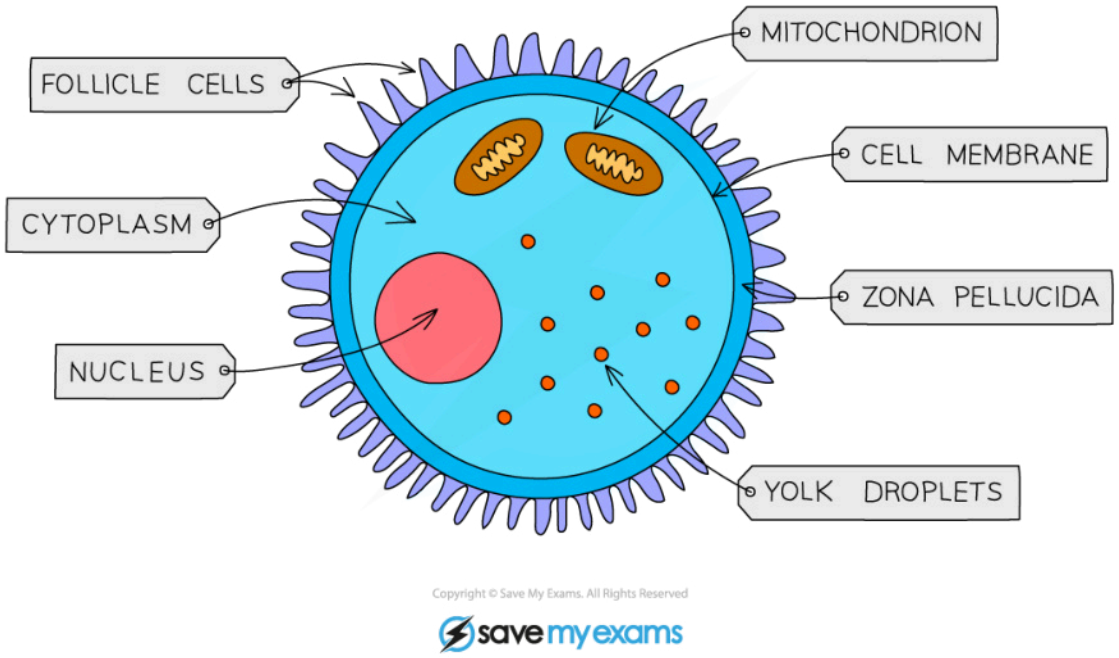
- Mendel was not the first to investigate inheritance using plants
- However, he was the first to generate strong numerical (**quantitative**) data, as opposed to observations only (**qualitative**)
- Mendel also used a **large number** of pea plants in his studies, for example in one investigation he recorded the characteristics for over 7,000 pea seeds
- By recording data for such a large number of seeds he ensured **reliability** in his conclusions
- **Many replicates** are important in a scientific investigation because they allow for:
  - Ensuring reliability of results
  - Identification of anomalous points
  - Further statistical analysis to establish a significant difference

Gametes

Your notes

- Gametes are the **sex cells** of an organism
- For example, the **sperm** and **egg** (ovum) cells in humans
- The **egg is larger than the sperm** as most of its space contains food to nourish a growing embryo
- The sperm cell contains **many mitochondria** to release energy for its motion
- Gametes **fuse** during fertilization to form a **zygote**
- These sex cells are formed during **meiosis** and only have **one** copy of each chromosome and so are **haploid** cells
  - For humans, that means the sperm and egg cells contain **23 single chromosomes** in their nucleus
  - As there is only **one chromosome from each homologous pair** there is only **one allele** of each gene present
    - This allele may be dominant, recessive or **co-dominant**





The structure of human gametes - the sperm and egg

Examiner Tip

You will not need to be able to quote all of Mendel's experiments and his results but you should be able to explain why there is always a 3:1 ratio in second generation organisms

Your notes

3.3.2 Inheriting Alleles


Your notes

Segregation of Alleles

- Meiosis is a form of **nuclear division** that results in the **production of haploid cells** from diploid cells
- During meiosis a diploid cell will **divide twice** to form **four haploid cells**
- It produces **gametes** in plants and animals that are used in **sexual reproduction**
- A diploid nucleus will contain **two** copies of each gene
- A haploid nucleus contains just **one** copy of each gene
- A diploid cell of genotype Yy will produce **two gametes carrying the Y allele** and **two carrying the y allele**
- The separation of alleles into different cells during meiosis is called **segregation**
- Segregation is important as it allows for **new allele combinations** in offspring

Diploid Zygotes

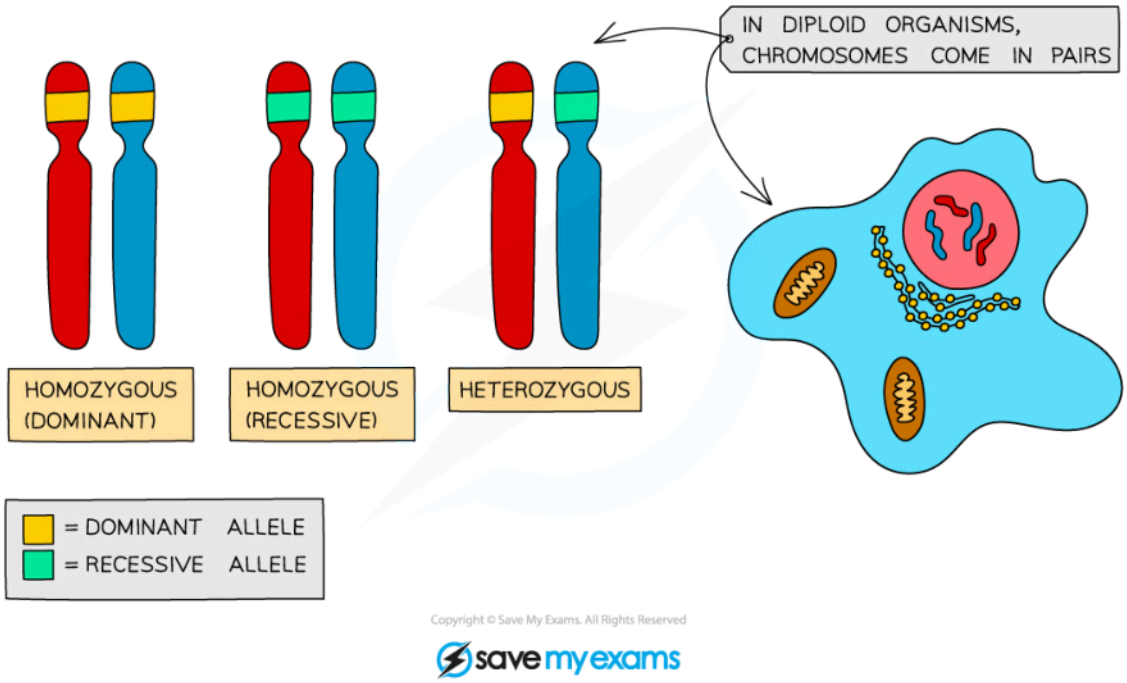
- Fusion of gametes results in diploid zygotes with two alleles of each gene that may be the same allele **or** different alleles
- Sexual reproduction is **a process involving the fusion of the nuclei of two gametes (sex cells) to form a zygote (fertilized egg cell) and the production of offspring that are genetically different from each other**
- Fertilization is defined as the **fusion of gamete nuclei**, and as each gamete comes from a different parent, there is **variation** in the offspring
- When a male and female gamete fuse their **chromosomes are combined**
- This means the resulting zygote is **diploid**
- The zygote contains two chromosomes of each type
- It will therefore also have **two alleles** of each gene
  - If the two alleles for a particular gene are the same then the **genotype** is described as **homozygous**
  - If the two alleles for a particular gene are different then the genotype is described as **heterozygous**

Dominant, Recessive & Co-Dominant Alleles


Your notes

- A **gene** is a short length of DNA found on a chromosome that codes for a particular **characteristic** (by coding for the production of a specific protein)
- **Alleles** are variations of the same gene
  - As we have two copies of each chromosome, we have two copies of each gene and therefore two alleles for each gene
  - One of the alleles is inherited **from the mother** and the other **from the father**
  - This means that the **alleles may not be the same**
  - For example, an individual has two copies of the gene for eye colour but **one allele could code for brown eyes and one allele could code for blue eyes**
- The **observable characteristics** of an organism (seen just by looking - like eye colour, or found – like blood type) is called the **phenotype**
- The **combination of alleles that control each characteristic** is called the **genotype**
- Alleles can be **dominant** or **recessive**
  - A dominant allele **only needs to be inherited from one parent** in order for the characteristic to be expressed in the phenotype
  - A recessive allele needs to be **inherited from both parents in order** for the characteristic to be expressed in the phenotype.
  - If there is only one recessive allele, it will **remain hidden** and the dominant characteristic will show
- If the two alleles of a gene are the same, we describe the individual as being **homozygous** (homo = same)
- An individual could be **homozygous dominant** (having two copies of the dominant allele), or **homozygous recessive** (having two copies of the recessive allele)
- If the two alleles of a gene are different, we describe the individual as being **heterozygous** (hetero = different)

Your notes



Alleles are different forms of the same gene. You can only inherit two alleles for each gene, and they can be the same (homozygous) or different (heterozygous). Alleles can be dominant or recessive.

- **Co-dominant alleles** have a combined effect on the phenotype
  - Certain **red-flowered plants** can be crossed with **white-flowered plants** of the same species, and the offspring's flowers have a **pink colour**
  - **Speckled chickens** show co-dominance between an **allele for white feathers** and an allele that causes the feathers to be **black**
  - The alleles are **both expressed to an equal extent** in the phenotype
- When completing genetic diagrams (punnet square diagrams), alleles are abbreviated to single letters
  - The dominant allele is given a **capital letter** and the recessive allele is given the same letter, but **lower case**
  - For example a tall (phenotype) pea plant can have the genotype **TT** or **Tt**; in this case 'T' represents the dominant tall allele and 't' represents the recessive dwarf allele

Inheritance of Blood Groups

Your notes

- Inheritance of blood group is an example of **co-dominance**
- This is of critical importance when deciding to give **blood transfusions** following injury or illness
- Use of the wrong blood group can cause an immune response that coagulates (solidifies) blood, leading to clots and serious illness/death
- There are **three** alleles of the gene controlling a person's blood group instead of the usual two
  - I represents the **gene** and the superscripts A, B and O represent the **alleles**
- Alleles I^A and I^B are **codominant**
- I^O is **recessive**
- I^A results in the production of **antigen A** on the surface of red blood cells
- I^B results in the production of **antigen B** on the surface of red blood cells
- I^O results in **no antigens** being produced on the surface of red blood cells
- These three possible alleles can give us the following genotypes and phenotypes

Blood Genotype & Phenotype Table

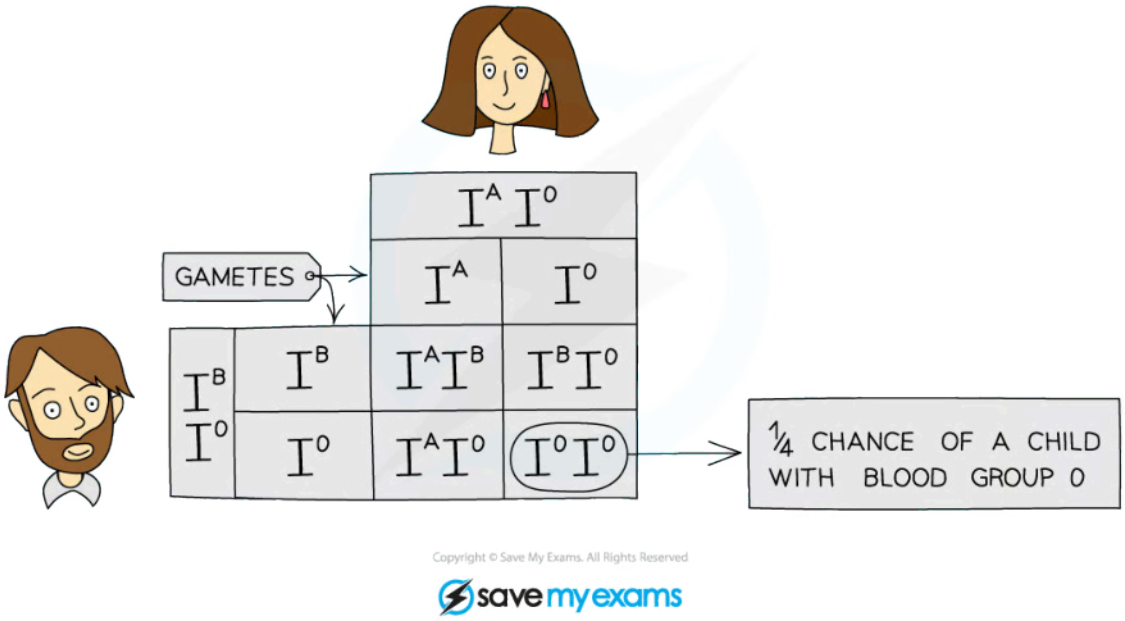
Genotype	Phenotype
I^AI^A or I^AI^O	A
I^BI^B or I^BI^O	B
I^AI^B	AB
I^OI^O	O

- We can use genetic diagrams to predict the outcome of crosses that involve the codominant alleles controlling blood groups

Worked example

Show how a parent with blood group A and a parent with blood group B can produce offspring with blood group O.

Your notes



Punnett square showing the inheritance of blood group

Examiner Tip

Take good care when hand-writing ABO blood group genotypes and alleles. The expected notation for ABO blood group alleles is that the letter I is always written in UPPERCASE and all the alleles must be in superscript. An example is $I^A I^O$

3.3.3 Skills: Inheritance

Your notes

Constructing Punnett Grids

- A **monohybrid trait** is one that is controlled by only **one gene**
- Generally, we consider that such a gene has **two alleles**
  - Either: one allele is dominant and the other is recessive
  - Or: the alleles are co-dominant

- A monohybrid cross starts with **pure-breeding parents** (homozygous), each displaying a different phenotype
- The **purpose of a Punnett grid** is to predict the probability of a certain offspring displaying a certain genotype or phenotype
  - In the case where multiple offspring are produced, Punnett grids can **predict the numbers of offspring** that will display a certain genotype or phenotype after a cross

Steps in constructing a Punnett Grid

1. Write down the **parental phenotypes** and **genotypes**
2. Write down all the **possible gamete genotypes** that each parent could produce for sexual reproduction
   - A useful convention is to write the gamete genotypes **inside a circle** to denote them as gametes (haploid cells)
3. Place each parental genotype **against one axis** of a Punnett grid (2 x 2 table)
4. In the boxes of the Punnett grid, combine the gametes into the possible genotypes of the offspring
   - This gives the offspring of the **F_1 generation** (1^{st} filial generation)
5. List the **phenotype** and **genotype ratios** for the offspring

Worked example

Sweet peas grow pods that are either green or yellow. The allele for green, **G**, is dominant to the allele for yellow, **g**.Construct a Punnett grid to predict the outcome when crossing green and yellow pure-bred plants to show the F_1 generation offspring.Using plants from the F_1 generation, construct a second Punnett grid to show the outcomes of the F_2 generation.

Step 1: Write down the parental phenotype and genotypes

Green coloured pods	Yellow coloured pods
GG	gg

Step 2: Write down all the possible gamete genotypes that each parent could produce

Your notes

G g

Step 3: Place each parental genotype against one axis of a Punnett grid (2 x 2 table)

		Green parent gametes	
		G	G
Yellow parent gametes	g		
	g		

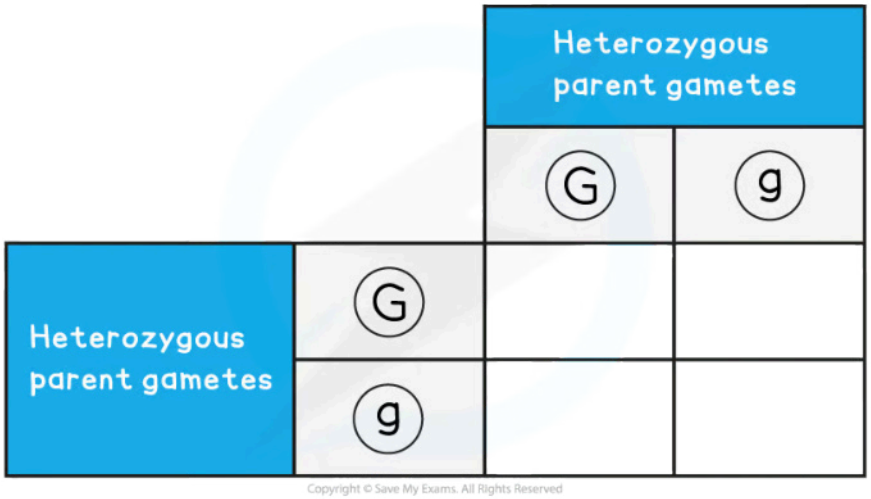
Step 4: Combine the gametes in each box of the Punnett grid

		Green parent gametes	
		G	G
Yellow parent gametes	g	Gg	Gg
	g	Gg	Gg

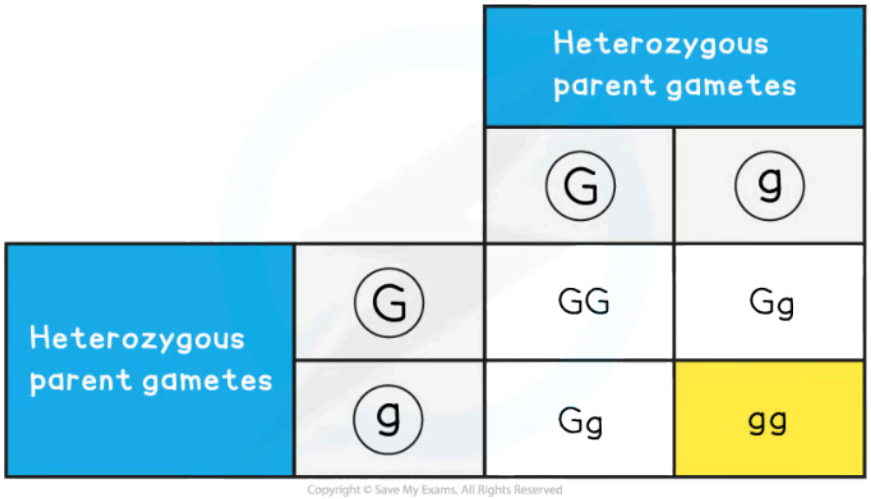
Genotypes of the F1 cross between homozygous green (GG) and homozygous yellow (gg) pea plants. All offspring (100%) have the genotype Gg and the phenotype is green.

Step 5: Take two heterozygous offspring from the F_1 generation and cross them

Your notes



Step 6: Combine the gametes in each box of the Punnett grid



Punnett grid showing the results of the F2 generation

Phenotype ratio is 3:1 green:yellow, Genotype ratio is 1 GG: 2 Gg: 1 gg

Analysis of Genetic Crosses


Your notes

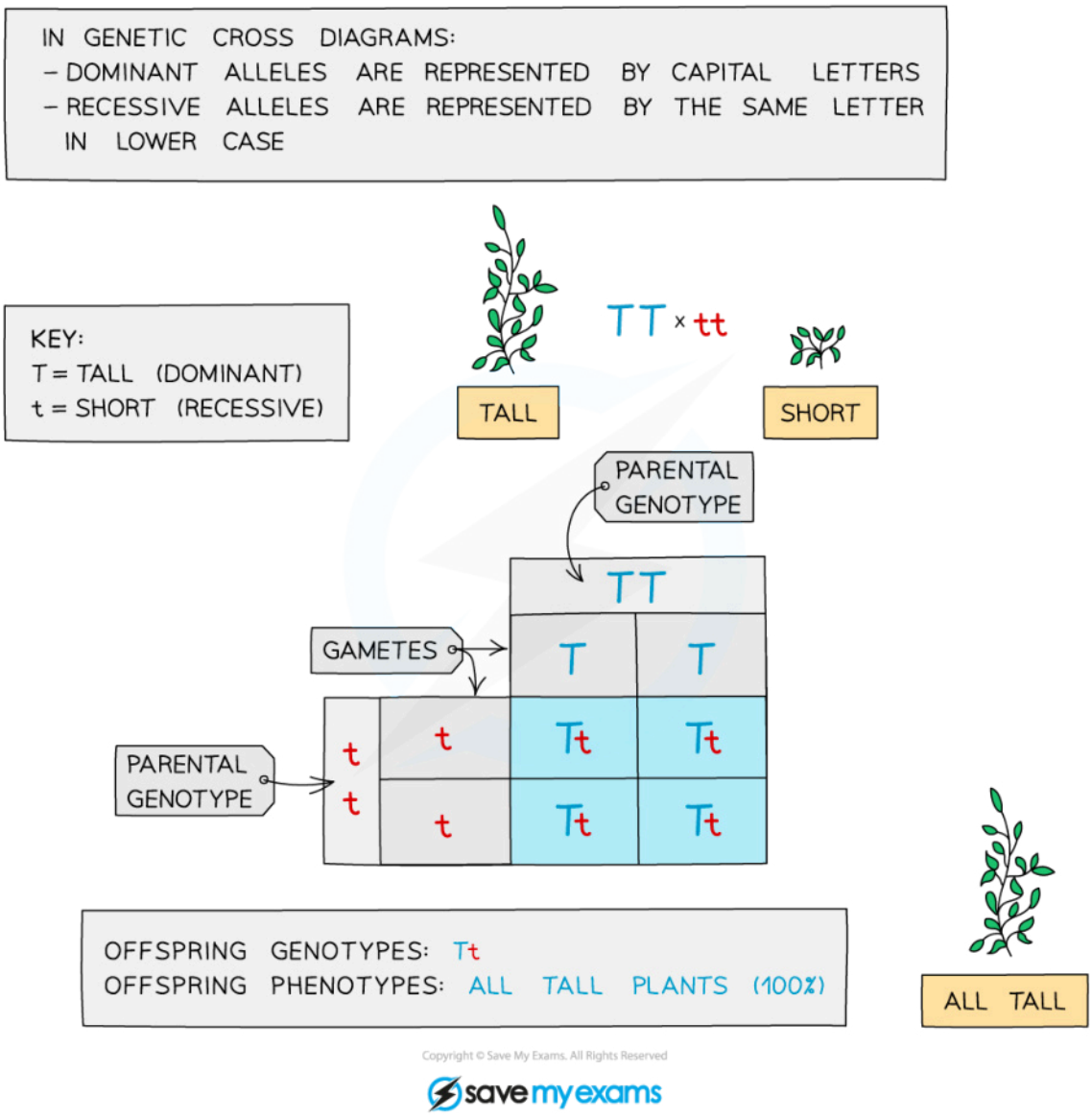
Comparison of predicted and actual outcomes of genetic crosses using real data

- A Punnett grid diagram shows the **possible combinations of alleles** that could be produced in the offspring of a certain genetic cross
- From this, we can deduce the **expected ratio** of these combinations
- The actual outcome achieved from the cross often presents **ratios different** to those deduced in the punnet grid
- This is because there is an element of **chance** involved with inheritance
- The **chi-squared test** is a statistical test that can be used to determine whether the differences between the observed ratios and the expected ratios are **significant or due to chance**

Using pea plants to demonstrate the predicted outcomes of genetic crosses

- The height of pea plants is controlled by a single gene that has two alleles: tall and short
- The tall allele is dominant and is shown as **T**
- The small allele is recessive and is shown as **t**
- A pure breeding short plant is bred with a pure breeding tall plant
- The term '**pure breeding**' indicates that the individual is homozygous for that characteristic

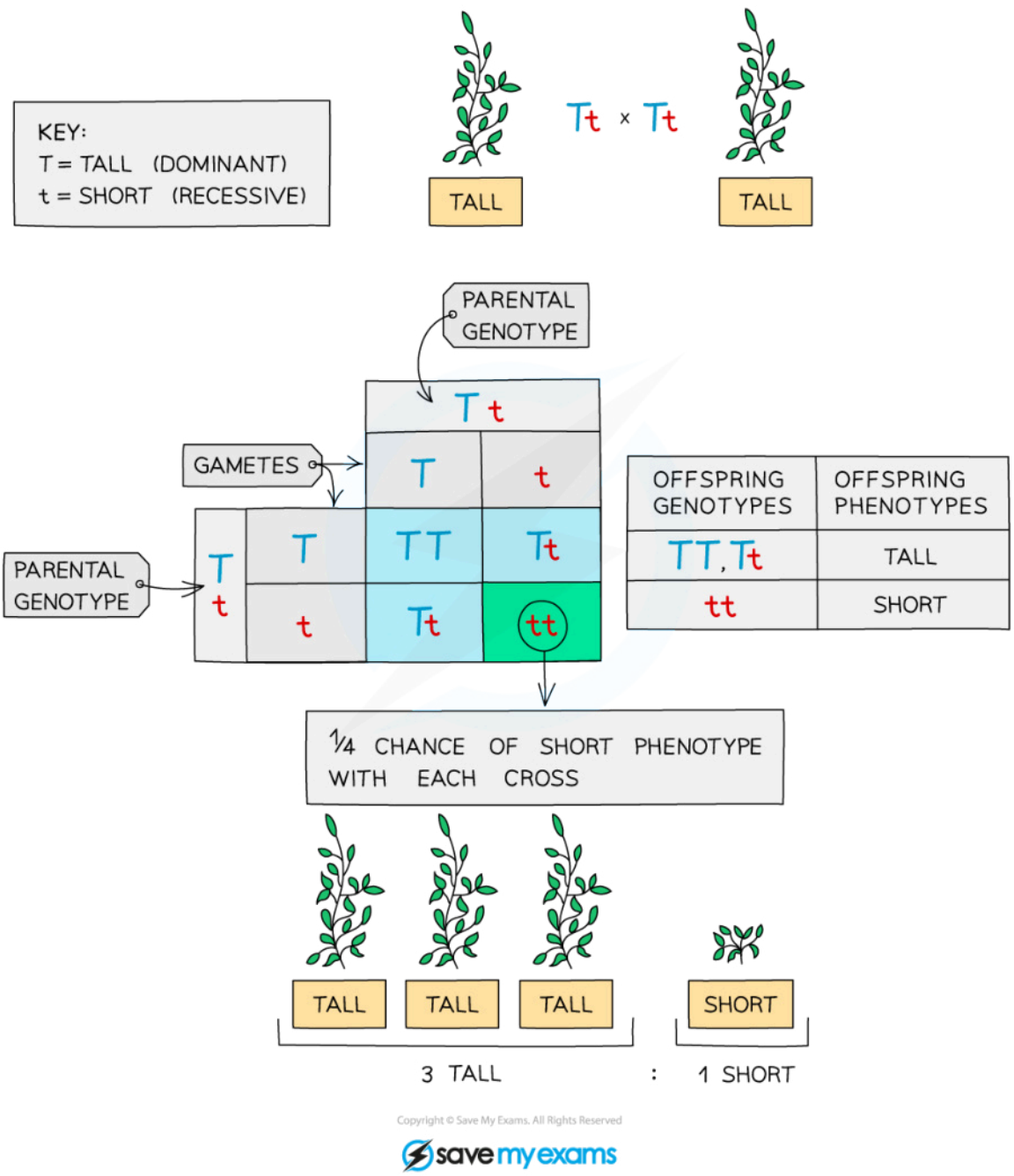
Your notes



A pure-breeding genetic cross in pea plants. It shows that all offspring will be have the tall phenotype.

- Crossing the offspring from the first cross

Your notes



A genetic cross diagram (F2 generation). It shows a ratio of 3 tall : 1 short for any offspring.

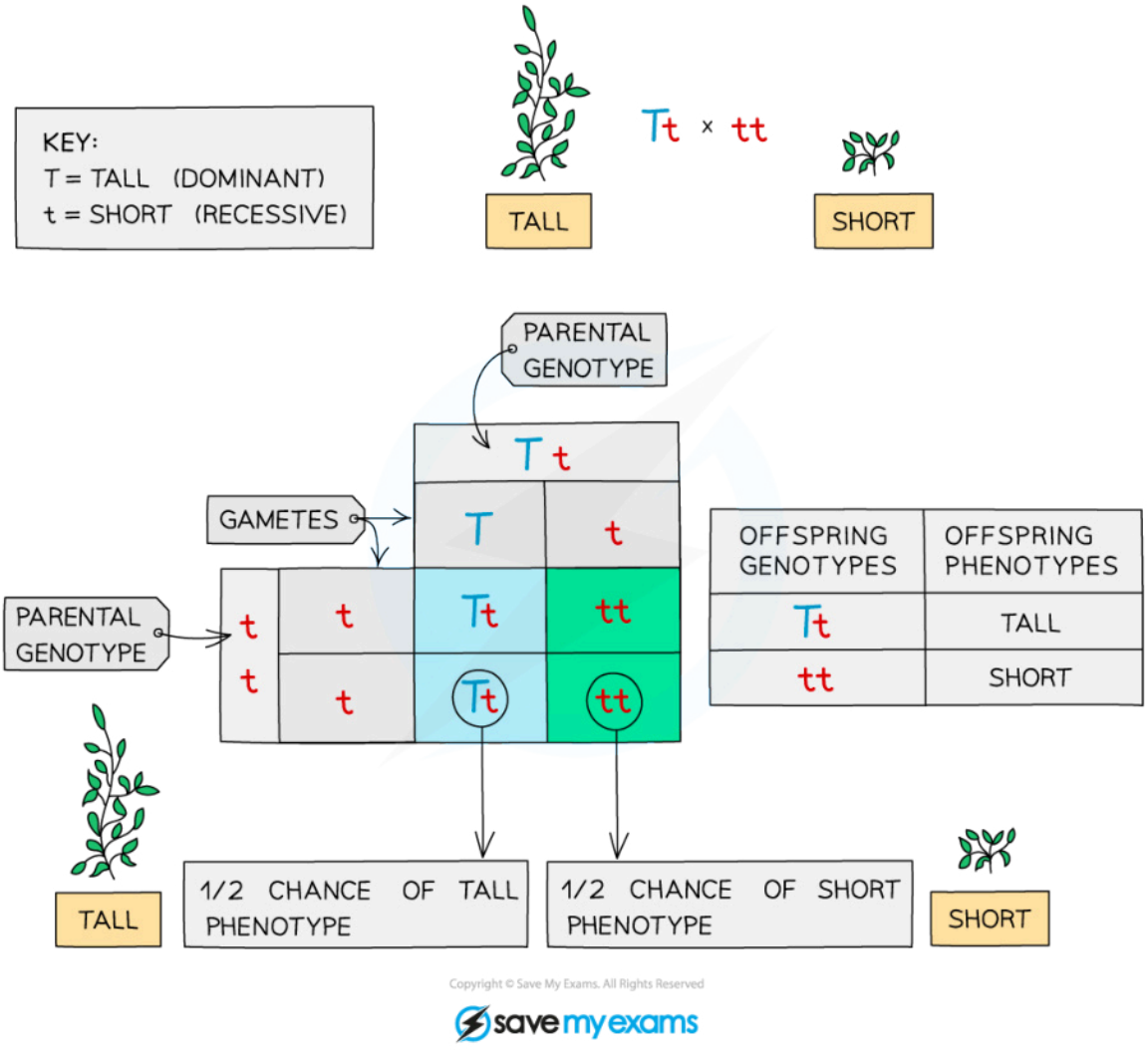
- All of the offspring of the first cross have the same genotype, **Tt** (heterozygous), so the possible combinations of offspring bred from these are: **TT** (tall), **Tt** (tall), **tt** (short)
- There is more variation in the second cross, with a **3:1** ratio of tall : short

- The F2 generation is produced when the offspring of the F1 generation (pure-breeding parents) are allowed to interbreed

Your notes

Crossing a heterozygous plant with a short plant

- The heterozygous plant will be tall with the genotype **Tt**
- The short plant is showing the recessive phenotype and so must be homozygous recessive – **tt**
- The results of this cross are as follows:



A cross between a heterozygous plant with a short plant

Examiner Tip

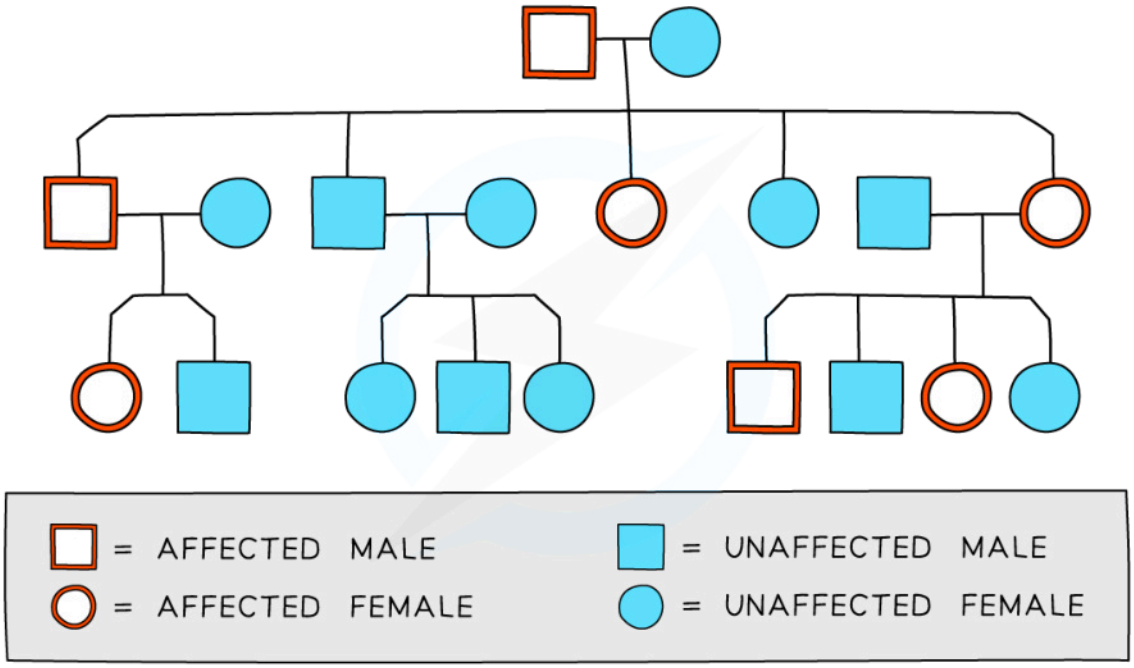
If you are asked to use your own letters to represent the alleles in a Punnett grid, try to choose a letter that is obviously different as a capital than the lower case so the examiner is not left in any doubt as to which is dominant and which is recessive.

Your notes

Pedigree Charts

Your notes

- Family pedigree diagrams are usually used to trace the **pattern of inheritance** of a specific characteristic (usually a disease) **through generations of a family**
- This can be used to work out the probability that someone in the family will inherit the **genetic disorder**



A family pedigree chart

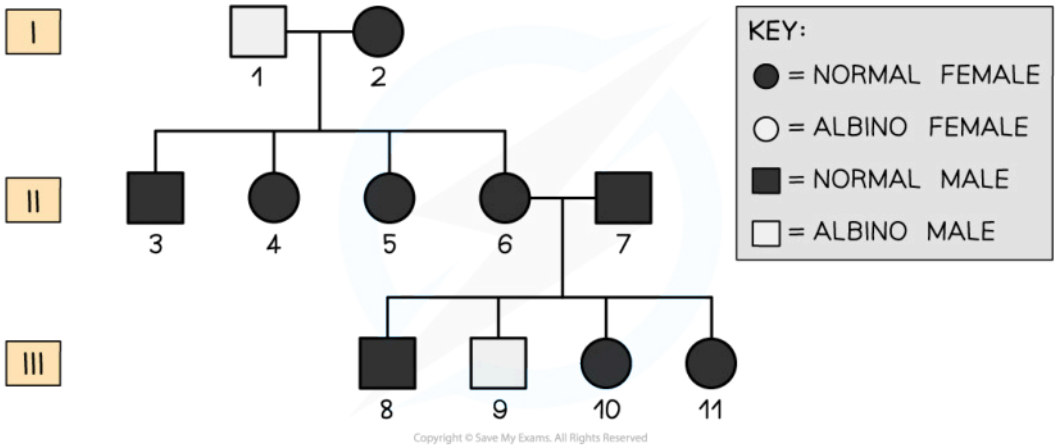
- Males are indicated by the **square shape** and females are represented by **circles**
- In this diagram, affected individuals are **red** and unaffected are **blue**
  - **Shading or cross-hatching** may also be used to show affected individuals
- **Horizontal lines** between males and females show that they have **produced children** (which are linked underneath each couple)
- Roman numerals may be used to indicate **generations**
- For each generation the eldest child is on the left and **each individual** is **numbered**
- The family pedigree above shows:
  - both males and females are affected
  - every generation has affected individuals
  - The eldest son (in the second generation) is affected
  - That there is one family group that has no affected parents or children
  - the other two families have one affected parent and affected children as well

Your notes

Worked example

Worked example: Pedigree charts

- Below is a pedigree chart which traces the inheritance of **albinism** across several generations. Albinism affects the production of the **pigment melanin** leading to lighter hair, skin and eyes.



- Using the pedigree chart, deduce and explain the following:

1. What type of allele causes albinism
2. The genotype of individuals named **9** and **7**
3. The possible genotypes of **10** and **11**

1. Albinism is caused by a **recessive allele**
   - Explanation: We can tell this from the pedigree chart because expression of the disease skips generation II. Also, person number 9 is an affected individual despite his parents (6 and 7) being unaffected. 6 and 7 must both be carriers of the recessive allele and 9 has inherited one recessive allele from each parent.
   - It is unlikely to be a **sex-linked disease as both females and males** have the condition

2. The genotype of person 9 must be **homozygous recessive** (aa) and the genotype of 7 must be **heterozygous** (Aa)
   - Explanation: 9 is an affected individual with albinism (which is determined by the recessive allele). 7 must be heterozygous in order for him to pass on the recessive allele to person 9

3. The possible genotypes of 10 and 11 are **heterozygous** (Aa) or **homozygous dominant** (AA)
   - Explanation: This is because they are unaffected individuals so must possess at least one dominant allele (A), however, it is possible that they each inherited a dominant allele from each parent

Examiner Tip

When answering questions about pedigree charts for genetic diseases, it is always useful to remember which phenotype is caused by the recessive allele. You can write these genotypes onto your chart and it will give you a good starting point for working out the possible genotypes of the rest of the individuals in the chart.

Your notes

3.3.4 Inheritance of Genetic Diseases


Your notes

Causes of Genetic Diseases

- A gene can affect the phenotype of an organism
  - A gene codes for a single polypeptide
  - The polypeptide affects the phenotype through a particular mechanism

- The phenotype of an individual can also be affected by the **environment**
- A **genetic disease** is caused by a gene which results in an abnormal protein that **alters the phenotype** of the individual
- Most genetic diseases are caused by recessive alleles on autosomal chromosomes
  - This means that an individual would need **two** copies of the recessive allele in order to develop the disease
  - Individuals that are heterozygous do not suffer from the disease but are **carriers** and can pass the recessive allele on to the **next generation**
  - A disease determined by a recessive allele includes **cystic fibrosis**

- Some diseases are caused by dominant
  - This means that only **one** copy of the allele is required in order to develop the disease and this one copy can also be passed on to the next generation
  - Individuals that are homozygous dominant, **will suffer from the disease** and will also pass the allele on to the next generation with **100%** probability
  - A disease determined by a dominant allele includes **Huntington's disease**

- It is also possible, but rare, for a disease to be caused by codominant alleles
  - This means that in individuals with heterozygous genotype, **both alleles** are expressed in the phenotype
  - Therefore giving a **3rd phenotype** that is different to the homozygous phenotypes
  - A disease determined by codominant alleles includes **sickle cell anaemia**

- The genes which causes some genetic diseases are found on the **sex chromosomes**
  - This means they affect males and females **differently**
  - Examples of sex-linked diseases include **haemophilia** and **colour blindness**

Examiner Tip

You may be asked to predict the inheritance of diseases like the ones above. An example question would be:

Max and Jane are trying for a baby but they are concerned about the possibility of their child having haemophilia. Neither Max or Jane have haemophilia themselves but Jane's father had the condition. What are chances that their child could have haemophilia?

For questions like this, it is very important to gather early on whether the abnormal allele that causes the disease is dominant or recessive and if there is any sex linkage. In this example for haemophilia, the abnormal allele is recessive and the gene is sex-linked. Then the next step would be to work out the genotypes of the parents from the information given and use this to create a genetic diagram.


Your notes

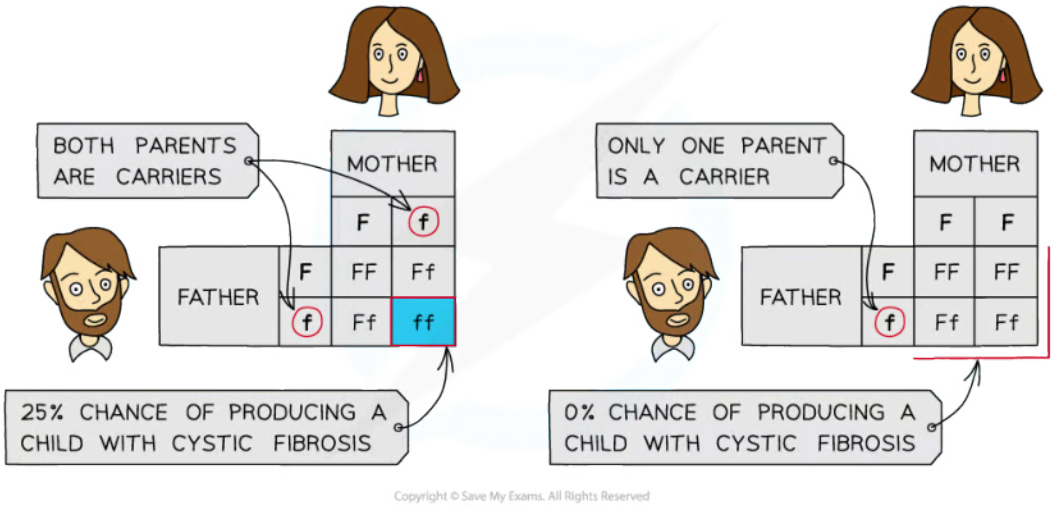
Examples of Genetic Diseases


Your notes

Cystic Fibrosis

- Cystic fibrosis is a genetic disorder of **cell membranes** caused by a **recessive** allele (**f**) of the CFTR gene located on chromosome 7
  - This gene codes for the production of chloride ion channels required for secretion of sweat, mucus and digestive juices
- A fault in the CFTR gene leads to production of **non-functional channels**
- This leads to reduced levels of **sodium chloride** in secretions which subsequently reduces the movement of **water by osmosis** into the secretions
- Ultimately resulting in the body producing large amounts of thick, sticky mucus in the air passages
- Over time, the mucus builds up in the **lungs** leading to infections and blocks narrow passage ways, such as the **pancreatic duct,** leading to reduced digestive efficiency as a result of less enzyme secretions
- Cystic fibrosis is determined by a recessive allele, this means:
  - People who are **heterozygous** (only carry one copy of the recessive allele) won't be affected by the disorder but are **'carriers'**
  - People must be **homozygous recessive** (carry two copies of the recessive allele) in order to have the disorder
  - If both parents are carriers, the chance of them producing a child with cystic fibrosis is 1 in 4, or 25%
  - If only one of the parents is a **carrier** (with the other parent being homozygous dominant), there is no chance of producing a child with cystic fibrosis

Your notes

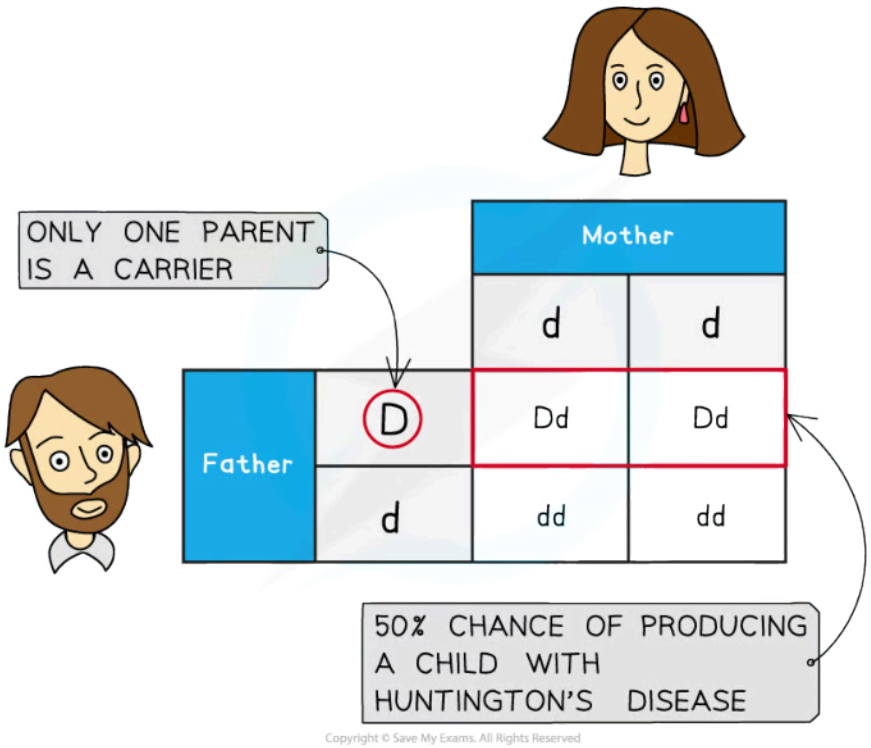


Inheritance of cystic fibrosis if both parents are carriers or if only one parent is a carrier

Huntington's Disease

- Huntington's disease is a genetic condition that develops as a person ages
- Usually a person with the disease will not show symptoms until they are 30 plus years old
- An individual with the condition experiences **neurological degeneration**; they lose their ability to walk, talk and think
- The disease is ultimately fatal
- It has been found that individuals with Huntington's disease have **abnormal alleles of the *HTT* gene**
  - The *HTT* gene codes for the protein **huntingtin** which is involved in neuronal development
- The **abnormal allele is dominant** over the normal allele
  - If an individual has one abnormal allele present they will suffer from the disease
  - If only one parent is a carrier of the dominant allele, there is still a 1 in 2 or 50% chance of producing a child with the disease

Your notes



Huntington's is caused by a dominant allele

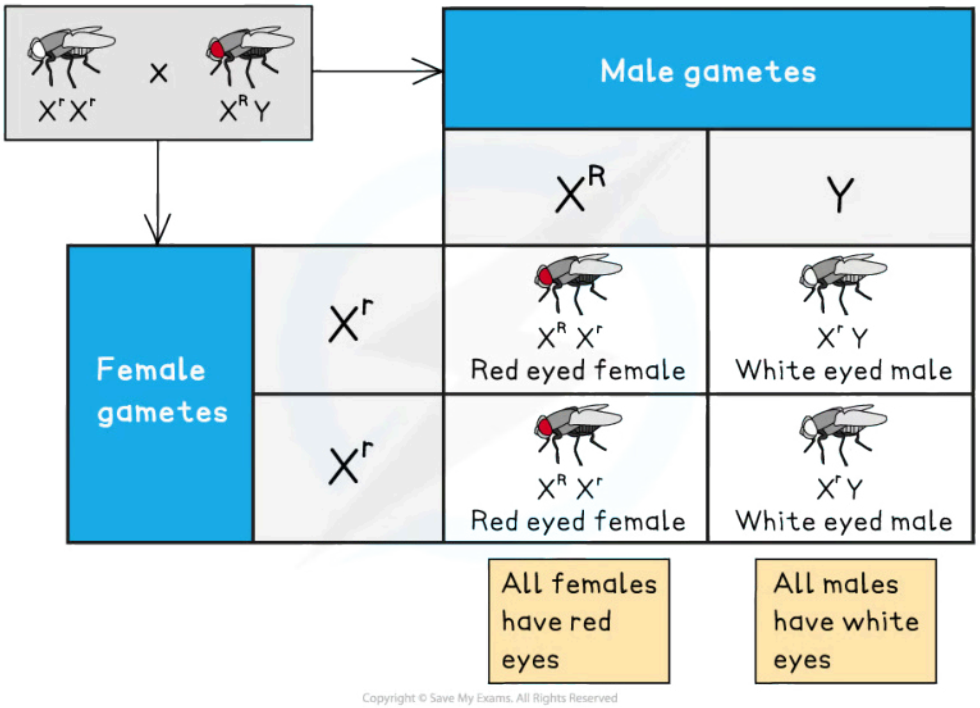
Sex-linked Genetic Diseases


Your notes

- Some genetic diseases in humans are sex-linked
- Inheritance of these diseases is different in males and females
  - Sex-linked genes are only **present on one sex chromosome and not the other**
  - This means the sex of an individual affects what alleles they pass on to their offspring through their gametes
- If the gene is on the X chromosome **males (XY) will only have one copy** of the gene, whereas females (XX) will have two
- There are **three phenotypes** for **females** - normal, carrier and has the disease, whereas **males** have only **two** phenotypes – normal or has the disease

Sex linkage in drosophila

- Thomas Morgan discovered sex linkage in *Drosophila* through studying the **ratios** of offspring produced from crosses of flies with different **eye colours**
- He found that when crossing white eyed females and red eyed males, the ratios achieved were different to those observed when crossing red eyed females with white eyed males
- For example, when crossing a red eyed male, X^RY, with a white eyed female, X^rX^r, Thomas Morgan found that all female offspring had red eyes and all male offspring had white eyes
  - If this was a gene found on an autosome, he would have expected to see that **all** of the first filial (F_1) generation would have red eyes because red is dominant
- Analysis of the results lead Thomas Morgan to deduce that the gene for eye colour was found on the **X chromosome** and therefore males possessed only **one** allele for that phenotypic characteristic



Eye colour in Drosophila is determined by a gene found on the sex chromosomes

Examiner Tip

The expected notation when writing about sex linked alleles is to use upper case 'X' and 'Y' for the chromosome, next to superscript letters to represent the allele. For example X^fX^f or X^fY.

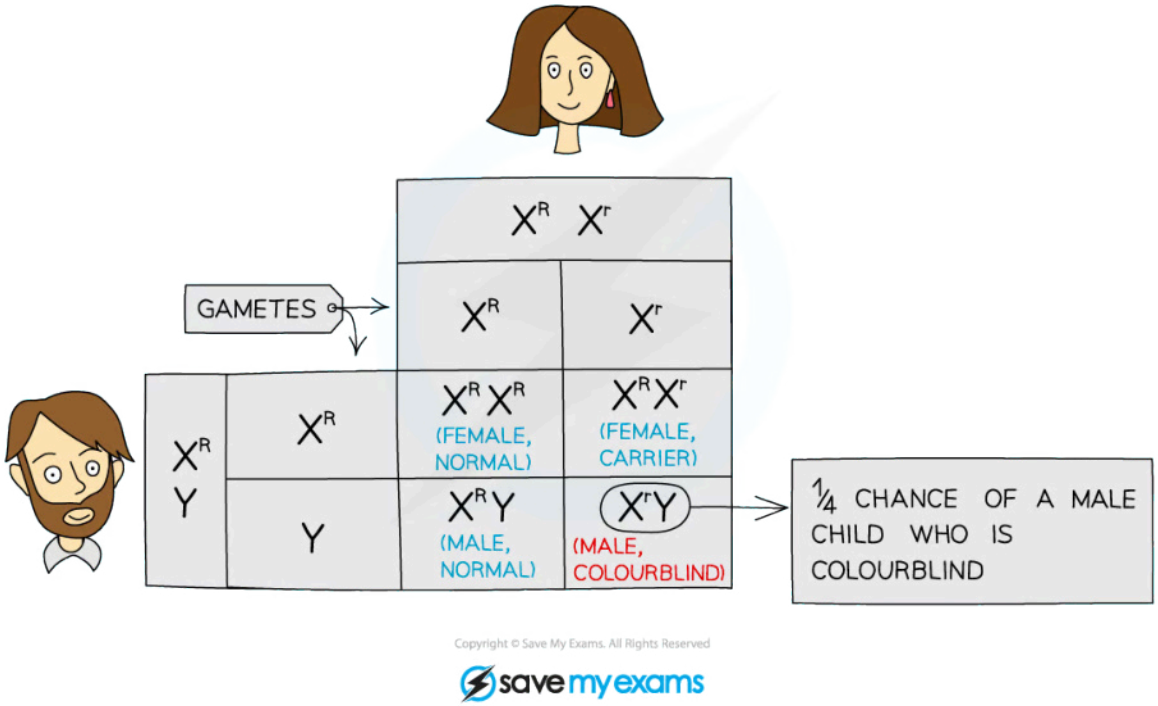
Your notes

Examples of Sex-Linked Genetic Diseases

Your notes

Red-green colour blindness

- The gene which is responsible for synthesizing the **photoreceptor proteins** of the eye, are found on the **X chromosome**
- The photoreceptor proteins are made in the cone cells of the eye and detect the specific **wavelengths** of light entering the eye
- Red-green colour blindness is caused by a **recessive allele** of this gene
- Males are more likely to be red-green colour blind as they only posses 1 allele for the gene, whereas females have 2 alleles and need to inherit 1 faulty allele from both parents in order to be colour blind



Punnett grid showing the inheritance of colourblindness, an X-linked condition

Haemophilia

- Haemophilia is a well known sex-linked disease
- There is a gene **found on the X chromosome** that codes for a protein called factor VIII. Factor VIII is needed to make blood clot
- There are two alleles for factor VIII, the dominant **F** allele which codes for normal factor VIII and the recessive **f** allele which results in a lack of factor VIII
- When a person possesses only the recessive allele **f**, they don't produce factor VIII and their blood can't clot normally
- If males have an abnormal allele they will **have the condition** as they have only one copy of the gene

- Females can be heterozygous for the faulty gene and not suffer from the condition but act as a **carrier**
- This means that haemophilia is a potentially fatal genetic disease which affects males more than females

Your notes

Worked example

Worked example: Haemophilia

- The genetic diagram below shows how two parents with normal factor VIII can have offspring with haemophilia

Parental phenotypes: carrier female x normal male

Parental genotypes: X^FX^f X^FY

Parental gametes: X^F or X^f X^F or Y

Monohybrid Punnett Square with Sex-linkage Table

		Male gametes	
		X^F	Y
Female gametes	X^F	X^FX^F / female with normal blood clotting	X^FY / male with normal blood clotting
	X^f	X^FX^f / carrier female	X^fY / male with haemophilia

Predicted ratio of phenotypes in offspring

1 female with normal blood clotting : 1 carrier female : 1 male with haemophilia : 1 male with normal blood clotting

Predicted ratio of genotypes in offspring: 1 X^FX^F : 1 X^FX^f : 1 X^FY : 1 X^fY

Examiner Tip

Make sure to include all of your working out when constructing genetic diagrams. It is not enough just to complete a punnett grid, you need to show that you have thought about the **possible gametes** that can be produced by each parent.Also, remember to state the **phenotype** as well as the genotype of the offspring that result from the cross. Read the questions carefully when answering sex-linked inheritance questions – is the question asking for a probability for **all** children or is it asking about a specific sex (males or females).

Your notes

Human Genetic Diseases

- There are **thousands** of genetic diseases that affect humans, although many are caused by recessive alleles and are therefore very **rare**
- Naturally numbers of people suffering from diseases caused by recessive allele remains very low due to the low probability of inheriting 2 recessive alleles. This is the recognised pattern of **Mendelian inheritance**
- Additionally, the **Human Genome Project** made it possible to **sequence** the entire human genome and opened up the opportunity for scientists to pinpoint which part of the genome is responsible for specific genetic diseases
- Now individuals can go through tests to sequence their own genome in order to establish the likelihood of inheriting an allele which could cause a genetic disease
- Parents may choose to go through this sequencing process before having children so that they are aware of the risks
  - They can make informed decisions based on the outcome of the test

3.3.5 Mutations & Disease

Your notes

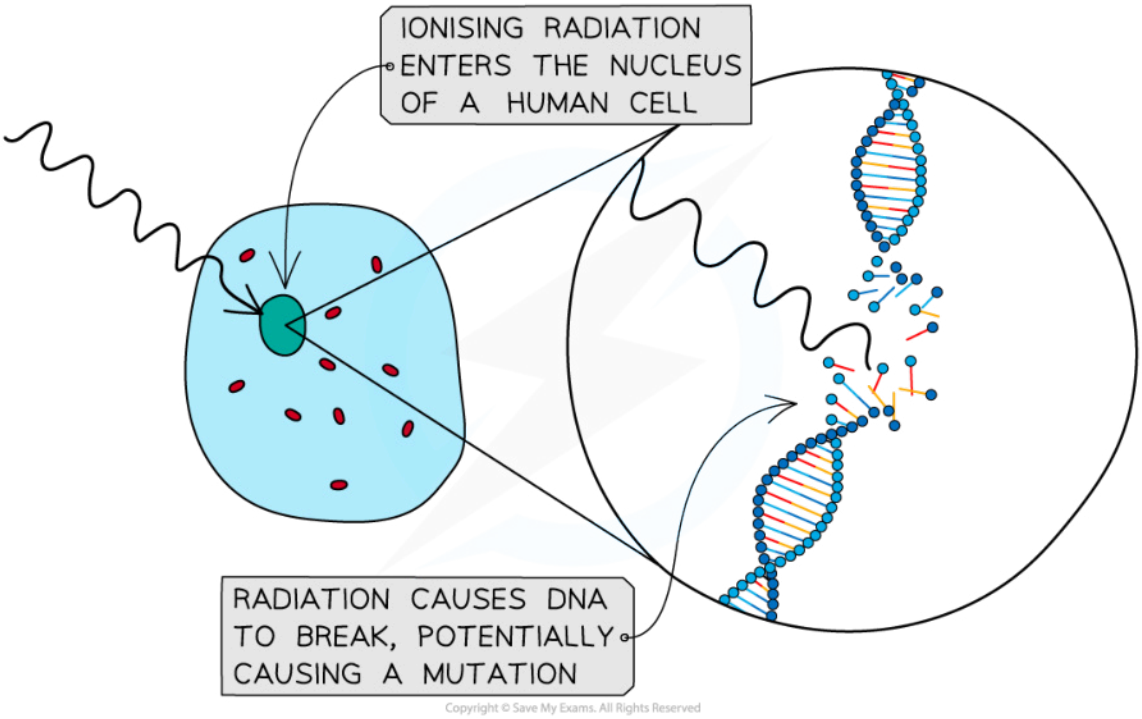
Causes of Mutations

- A **mutation** is a change in the sequence of base pairs in a DNA molecule
- They occur **randomly** and **continuously** to create new alleles of a gene
  - Often only with a very small number of differences in the base sequence
- As the DNA base sequence determines the sequence of amino acids that make up a polypeptide, **mutations in a gene can sometimes lead to a change in the polypeptide** that the gene codes for
  - If the change is significant, this could be harmful for the organism (mutations are rarely beneficial). It **may affect the ability of the protein to perform its function**
  - For example:
    - If the shape of the active site on an enzyme changes, the substrate may no longer be able to bind to the active site
    - A structural protein (like collagen) may lose its strength if its shape changes
- Most mutations are **neutral** because they **do not alter the polypeptide** or only alter it slightly so that its structure or function is not changed
  - This is because the genetic code is **degenerate**
- Mutations in body cells can lead to cancer. These mutations are often eradicated when the individuals dies
- Mutations of cells which are involved in gamete production can be **inherited** by the next generation

Mutagenic agents

- There are natural mechanisms that take place within cells to ensure the accuracy of **DNA replication**
  - These mechanisms involve proofreading and repairing damaged DNA
- When the mutation rate of a cell rises to above a normal (usually low) rate then these mechanisms have become ineffective
- Mutagenic agents are environmental factors that **increase the mutation rate of cells**
- **Radiation** can cause chemical changes in DNA, this includes:
  - **High-energy** radiation such as UV light
  - Ionising radiation such as X-rays, gamma rays and alpha particles
- **Chemical substances** can also caused changes to DNA, examples include
  - Benzo[a]pyrene and nitrosamines found in tobacco smoke
  - Mustard gas used as a chemical weapon in World War I

Your notes



Ionising radiation can cause mutations in DNA

Effects of Radiation

Your notes

Chernobyl Nuclear Disaster

- **Chernobyl** Nuclear Power Plant is in Ukraine
- In 1986 an incident at the plant caused an explosion and fire in the nuclear reactor core
- A large amount of radioactive material was released from the plant and went into the air
  - Radioactive isotopes of xenon, krypton, iodine, caesium, and tellurium were released as well as large amounts of small particles of uranium
- Hundreds of thousands of people were evacuated from the surrounding area to protect them from being contaminated
- An exclusion zone of around 2,600 square kilometres is still in place around the power plant
  - This is because the level of radiation in the area is still very high

The Chernobyl Disaster is probably the worst nuclear disaster in history

- The effects of the Chernobyl powerplant explosion were significant:
  - The total number of radiation related deaths to date has reached 4000
  - Large areas of pine forest **turned brown** and **died** in the weeks afterwards
  - **Agricultural animals** died due to **thyroid damage** caused by radioactive iodine and the **consumption** of contaminated meat e.g. lamb, was **banned**
  - **Milk** produced contained high levels of iodine in areas where waterways had been contaminated

Your notes

- **Bioaccumulation** of radioactive materials occurred in nearby waterways affecting fish in countries thousands of kilometres away and **contaminating drinking water** for many species
- Thousands of cases of **thyroid cancer** were recorded as a direct result of the radioactive iodine, including over 4000 in children and adolescents

- However, despite all this, there is **no significant evidence** of an increase in **solid cancers** or **leukaemia** even in the most affected population
- With no human habitation within the exclusion zone since the explosion, **other wild animals and many species of plants** have moved in and **colonised** the area
  - Despite the high radiation levels, the life expectancy of these organisms has not been shortened
- The long term effects on those who were exposed to **low levels** of radiation are yet unknown as studies continue

Nuclear bombing of Hiroshima and Nagasaki

- Two atomic bombs were dropped in Japan towards the end of World War II. One on Hiroshima and one on Nagasaki
- Between 150 000 and 200 000 people died as a direct result of the bombs
  - Half of these people died on the day the bombs were detonated and the rest died in the months immediately afterwards as a result of **burns, radiation sickness, injuries or through illnesses and malnutrition**
- Huge studies were carried out on other **survivors,** compared to a control group, to build a bigger picture of the longer term impact of high **exposure to radiation**
  - Incidence of cancer were much higher in the survivors studied compared to the control group
  - An **increase in leukaemia** cases was seen in both cities after a 2 year delay, which reached a peak around 6 years after the bombings
  - Those who were closer to ground zero seemed to be more seriously affected
  - There were also thousands of recorded **cancerous tumours** in the groups being monitored, although due to **confounding factors**, only around 800 could be formally attributed to the effects of radiation
- A large study was also carried out into the effects on **babies** pre and post birth
  - It was expected that there would be high numbers of mutations resulting in subsequent stillbirths or deformities, however, the numbers of incidence recorded were **not significant**
  - There was **no evidence** to suggest that babies conceived by survivors of the bombings were more likely to be born with birth defects
- There were many more **social impacts** associated with the bombs
  - The survivors were labelled 'Hibakusha', meaning 'the explosion affected people' and the associated stigma lead to widespread discrimination
    - There were **concerns** about whether the Hibakusha were **contagious** or whether the illnesses that they experienced were **heritable**. **Sterilisation** programs were even considered
    - As a result, many survivors struggled to find **employment or marry**