Cambridge (CIE) IGCSE Biology

Your notes

Inheritance, Genes & Cell Division

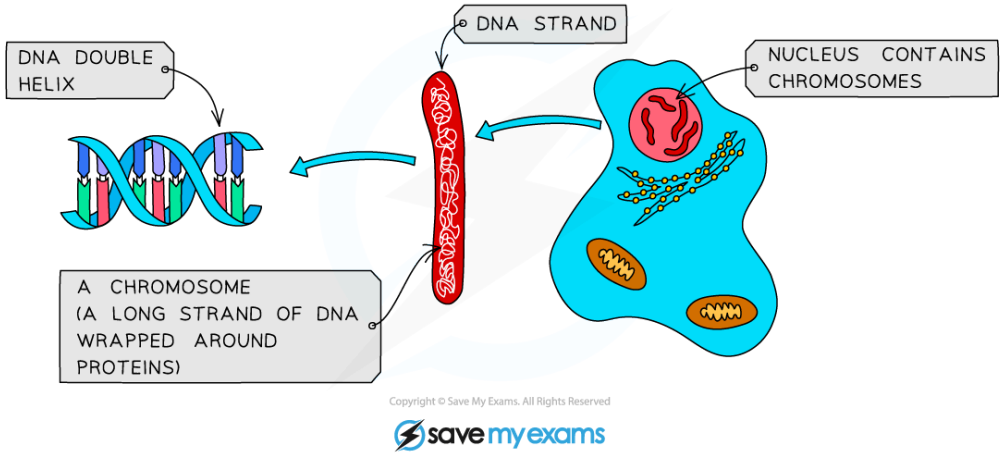
Contents

Your notes

The Structure of Genes

Inheritance of Chromosomes, Genes and DNA

- **Inheritance** is the **transmission of genetic information from generation to generation**
- **Chromosomes** are located in the **nucleus** of cells
- They are **thread-like structures of DNA, carrying genetic information in the form of genes**
- A **gene** is a short length of **DNA** found on a chromosome that **codes for a specific protein**
- This could be a structural protein such as collagen found in skin cells, an enzyme or a hormone
- Genes control our characteristics as they code for proteins that play important roles in what our cells do
- Think about zooming into the nucleus of a cell, as shown in the diagram below
  - On the right is the zoomed-out view
  - Which zooms in as we read across the diagram right-to-left



Genes are short lengths of DNA that code for a protein. They are found on chromosomes

- Alleles are **variations** of the same gene
  - As we have two copies of each chromosome (one from each parent), 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 do not code for the same thing
- 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**

Your notes

Examiner Tips and Tricks

The Zoom model is always useful when trying to viualise where you are in a cell, and what molecules are doing what. Imagine a zoom lens that has the power to zoom right out to look at a whole organism, but also to zoom right into individual molecules. This helps in recognising the structures in cell nuclei visible as chromosomes, then zooming in to picture the individual genes and then, the bases that make up the DNA

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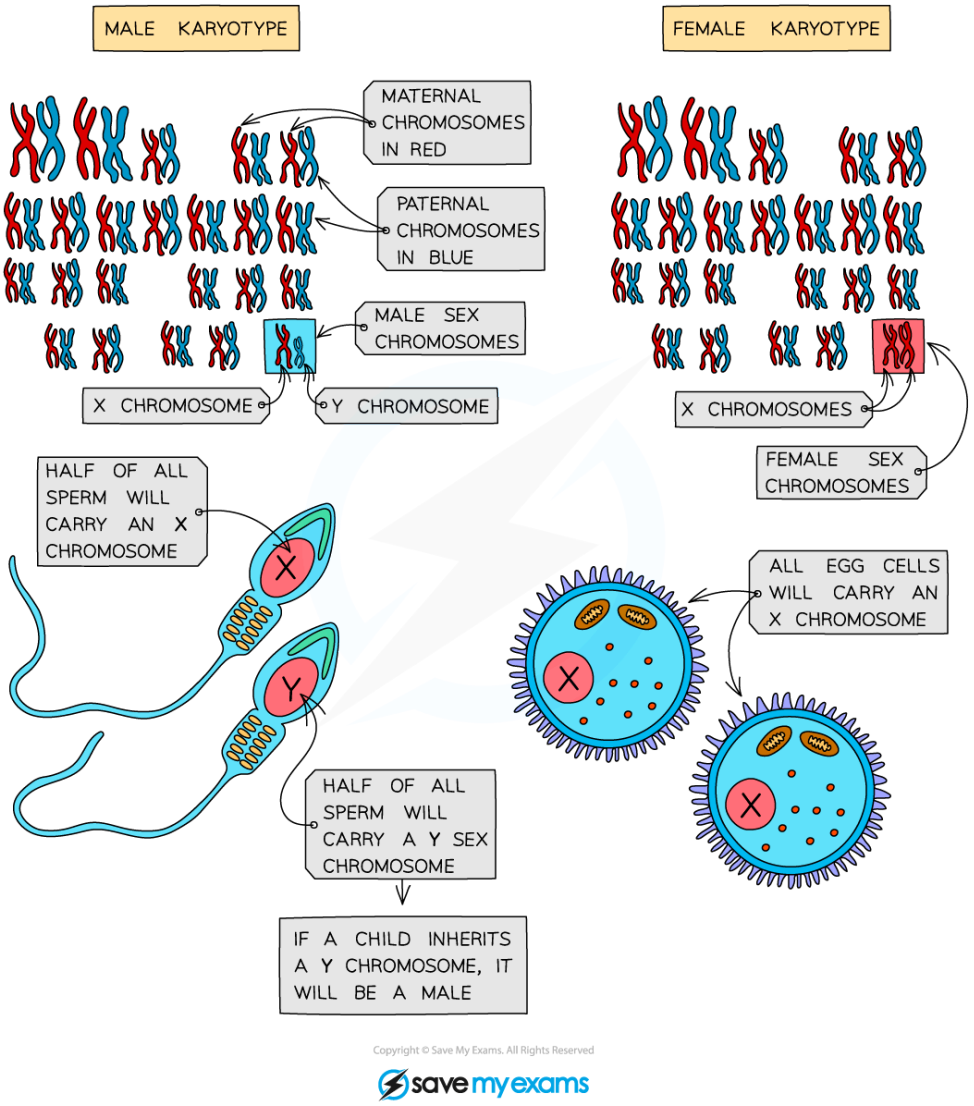
XX & XY Chromosomes

- Sex is determined by an **entire chromosome pair** (as opposed to most other characteristics that are just determined by one or a number of genes)
- **Females** have the sex chromosomes **XX**
- **Males** have the sex chromosomes **XY**
- As only a father can pass on a Y chromosome, he is **responsible for determining the sex of the child**
- He does this because
  - He produces (ejaculates) around 250 million sperm cells during sexual intercourse
  - Of those, half (125 million sperm) will be carrying his X chromosome
    - If one of these sperm fertilises the egg, the fetus will be female
  - The other 125 million of his sperm will be carrying his Y chromosome
    - Which will result in a male fetus if one of these fertilises the egg



Sperm cells determine the sex of offspring

- The inheritance of sex can be shown using a **genetic diagram** (known as a **Punnett square**), with the X and Y chromosomes taking the place of the alleles usually written in the boxes

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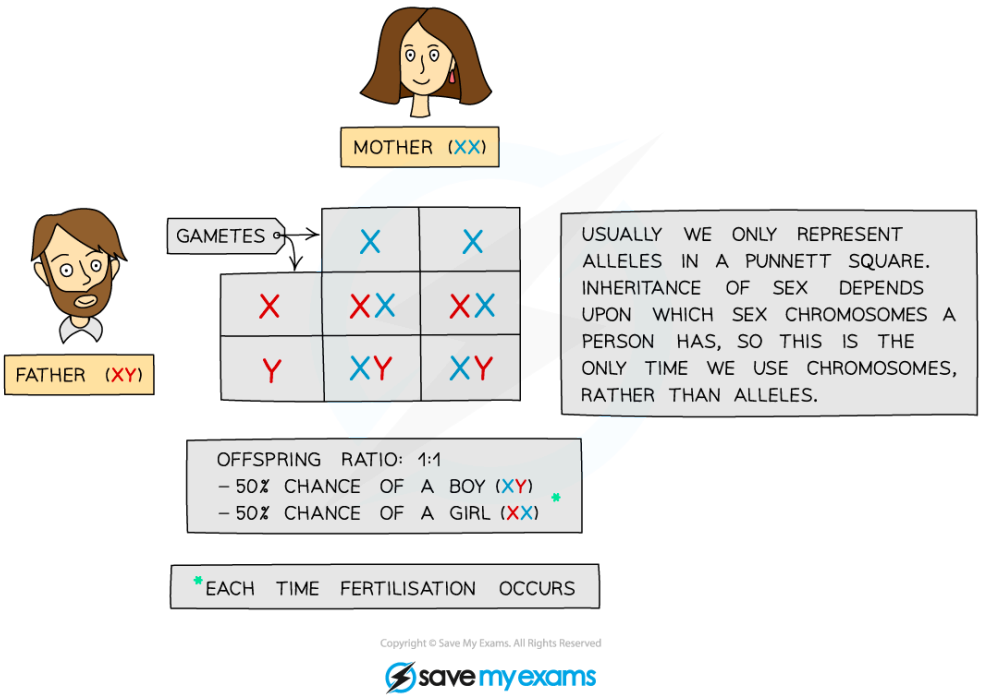

Punnett square showing the inheritance of sex

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DNA Base Sequence To Amino Acid Sequence: Extended

The DNA Base Sequence Determines the Amino Acid Sequence in Protein

- The DNA code (a series of **bases**) is converted into proteins (a series of **amino acids**)
- The process of protein synthesis has two stages
  - **Transcription** (rewriting the base code of DNA into bases of RNA)
  - **Translation** (using RNA base sequence to build amino acids into sequence in a protein)
- Therefore, the sequence of bases in a gene determines the sequence of amino acids that make a specific protein
- Different sequences of amino acids give different shapes and functions to protein molecules

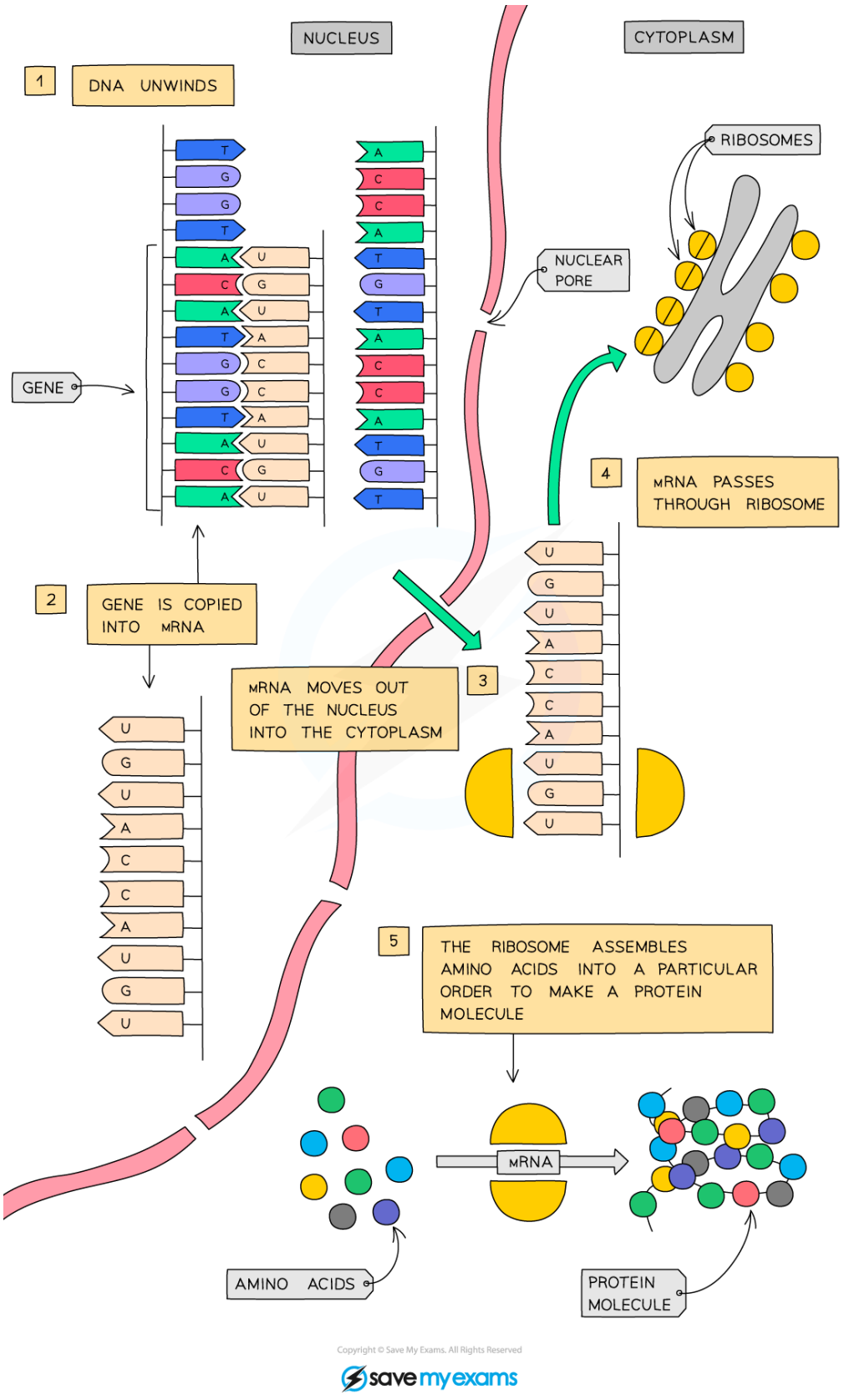
Examiner Tips and Tricks

Students often confuse sequences of **bases** (in DNA/RNA) with sequences of **amino acids** (in proteins).

Make sure you're clear which large molecule you're referring to and therefore, which monomers make up the larger molecule.

Your notes

Transcription & Translation: Extended



Protein synthesis

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- Proteins are made by **ribosomes** with the sequence of amino acids controlled by the **sequence of bases** contained within DNA
- DNA **cannot travel out of the nucleus** to the ribosomes (it is far too big to pass through a nuclear pore) so the base code of each gene is transcribed onto an RNA molecule called **messenger RNA** (mRNA).
- mRNA then **moves out of the nucleus** and attaches to a ribosome
- The ribosome 'reads' the code on the mRNA in groups of three
- Each triplet of bases **codes for a specific amino acid**
- In this way the ribosome **translates** the sequence of bases into a **sequence of amino acids** that make up a protein
- Once the amino acid chain has been assembled, it is released from the ribosome so it can fold and form the final structure of the protein

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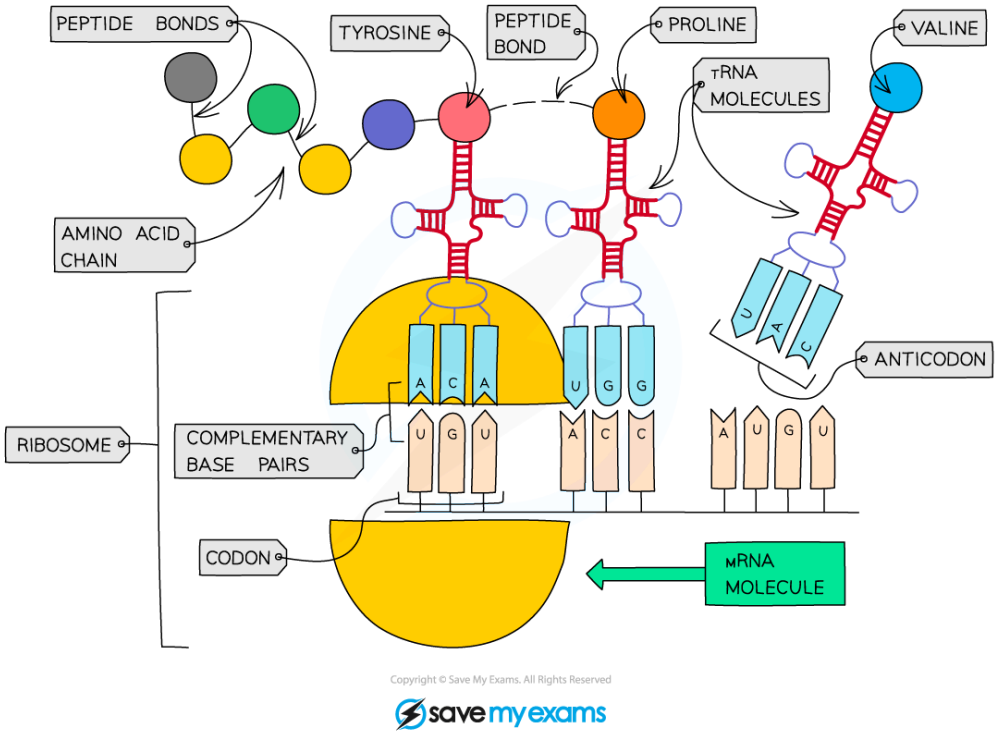
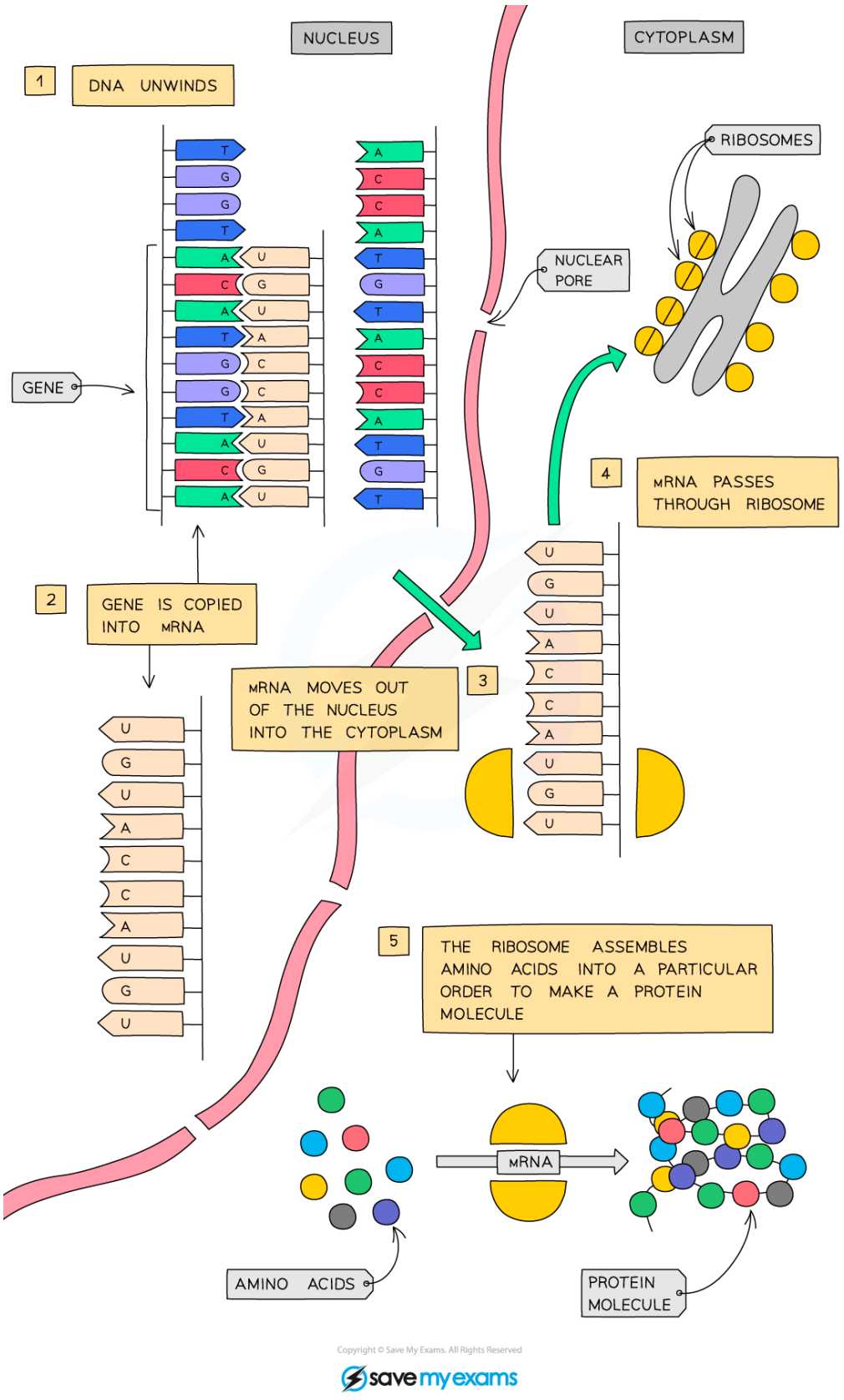


The triplet code of DNA (carried by mRNA) is read by the ribosome and amino acids are attached together in a specific sequence to form the protein

Control of Gene Expression: Extended

Your notes



Protein synthesis

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- **Expression** of a gene means whether that gene is transcribed and translated in a particular cell or not
- Most genes are not expressed in a particular cell
  - They are 'switched off'
  - Because that would be a waste of energy and other resources in the cell
- Only the genes whose proteins are vital to that cell's function are expressed ('switched on')

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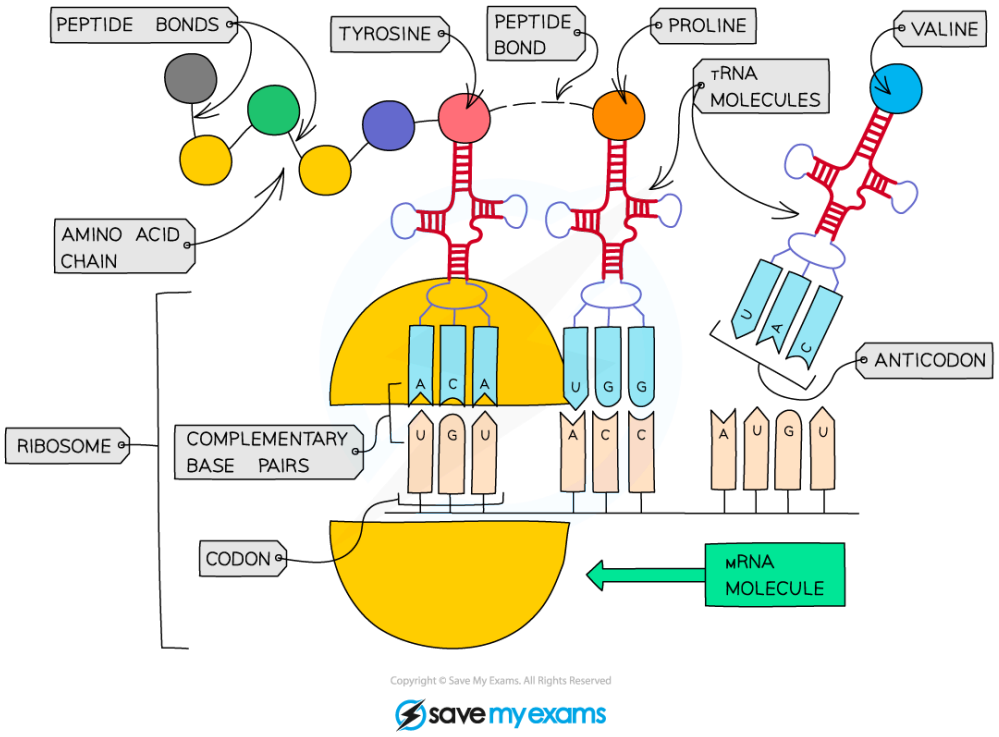

The triplet code of DNA (carried by mRNA) is read by the ribosome and amino acids are attached together in a specific sequence to form the protein

- In this way, DNA **controls cell function** by **controlling the production of proteins**
- The proteins may be **enzymes, antibodies,** or **receptors** for **neurotransmitters**
- Although all body cells in an organism contain the same genes, **many genes in a particular cell are not expressed** because **the cell only makes the specific proteins it needs**

Haploid and Diploid Cells

- All humans have **23 different chromosomes** in each cell
- In most body cells, not including the gametes (sex cells), we have 2 copies of each chromosome, leading to a total of **46 chromosomes**

- Nuclei with two sets of chromosomes are known as **diploid nuclei**
- The **gametes** (egg and sperm cells) only have one copy of each chromosome, meaning they have a **total of 23 chromosomes** in each cell
- Nuclei with one set of unpaired chromosomes are known as **haploid nuclei**

Examiner Tips and Tricks

An easy way to remember the difference between haploid and diploid is to remember:

Haploid = **Ha**lf the normal number of chromosomes

It's worth noting that the human diploid chromosome number is 46. In an exam, you may be given information about a different species, with a different number of chromosomes. Make sure you read exam questions carefully.

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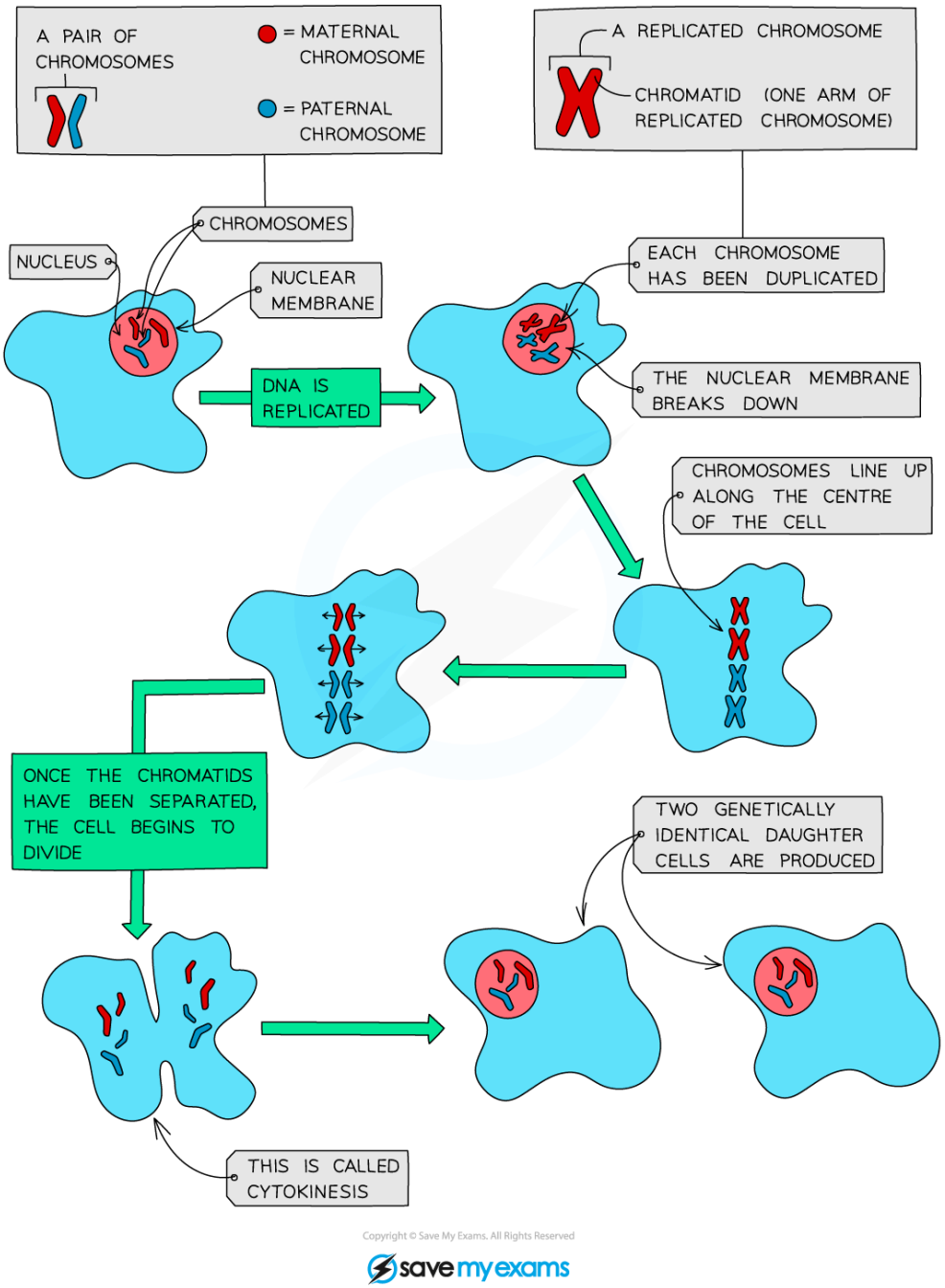
New Cells are Made by Mitosis: Extended

Extended Tier Only

Mitosis – Basics

- Most body cells have two copies of each chromosome
- We describe these cells as **diploid**
- When cells divide their chromosomes double beforehand
- This ensures that when the cell splits in two, each new cell still has two copies of each chromosome (is still diploid)
- This type of cell division is used for **growth, repair of damaged tissues, replacement of cells** and **asexual reproduction** and is known as **mitosis**
- Mitosis is defined as **nuclear division giving rise to genetically identical cells**



The process of cell division by mitosis

Process:

- Just before mitosis, each chromosome in the nucleus copies itself exactly (forms x - shaped chromosomes)
- Chromosomes line up along the centre of the cell where cell fibers pull them apart
- The cell divides into two; each new cell has a copy of each of the chromosomes

Importance:

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- All cells in the body (excluding gametes) are produced by mitosis of the zygote
- Mitosis is important for replacing cells e.g, skin cells, red blood cells and for allowing growth (production of new cells e.g. when a zygote divides to form an embryo)

Occurs in:

- *Growth*: mitosis produces new cells
- *Repair*: to replace damaged or dead cells
- *Asexual reproduction*: mitosis produces offspring that are genetically identical to the parent

Your notes

Mitosis & Stem Cells: Extended

Extended Tier Only

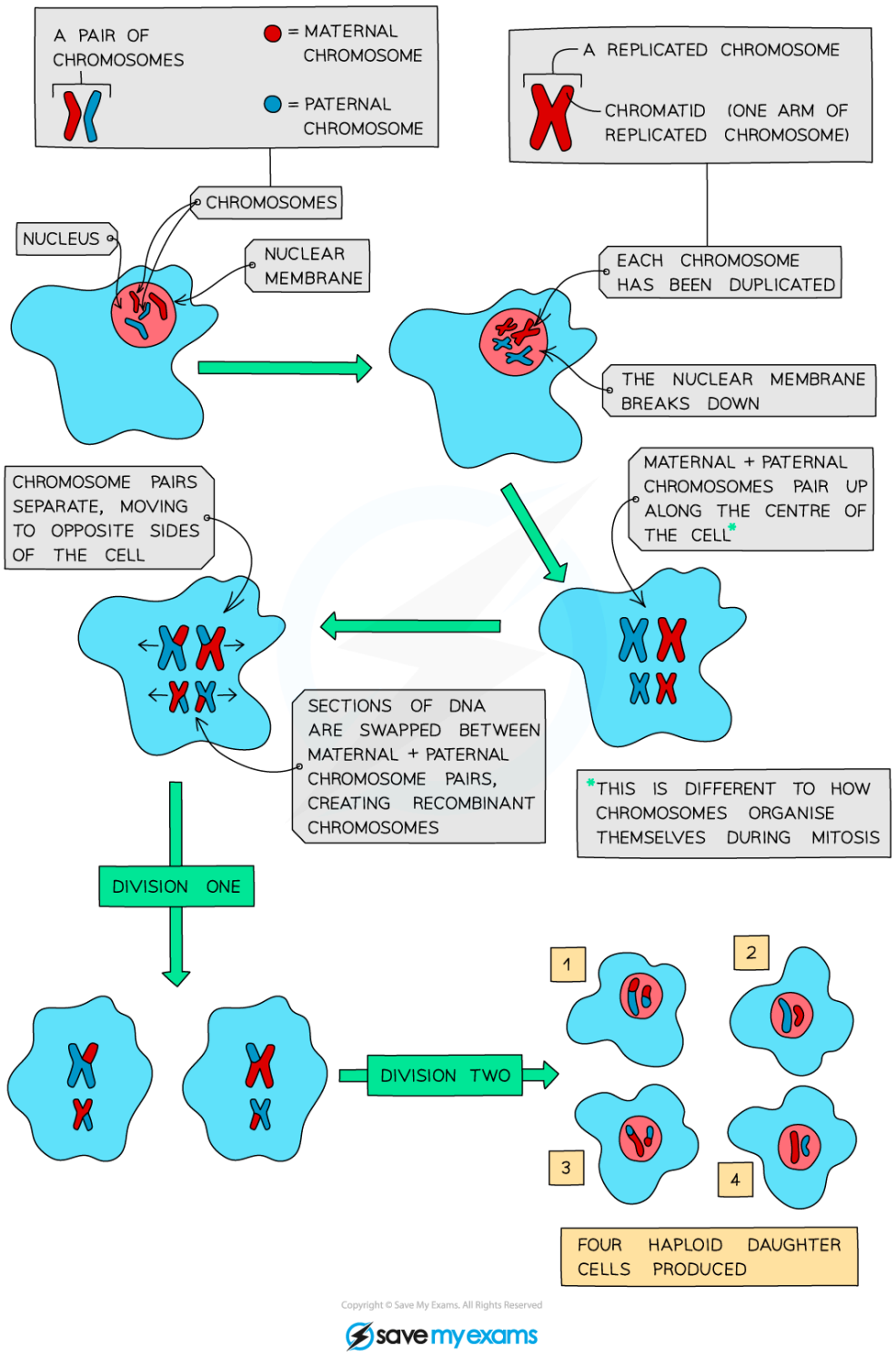
- Many tissues in the human body contain a small number of **unspecialised cells**
- These are called **stem cells** and their function is to divide by mitosis and **produce new daughter cells that can become specialised** within the tissue and be used for different functions
- The ultimate stem cell is the **zygote**
  - A zygote divides several times by mitosis to become a ball of unspecialised cells (around 200–300 cells)
    - These are **embryonic stem cells**
  - These cells are all the same and start differentiating as the fetus develops with recognisable features

Your notes

Meiosis: Extended

Extended Tier Only

- **Meiosis** is a type of nuclear division that **gives rise to cells that are genetically different**
- It is used to produce the **gametes** (sex cells)
- The number of chromosomes must be **halved** when the **gametes** (sex cells) are formed
- Otherwise there would be double the number of chromosomes after they join at fertilisation in the zygote (fertilized egg)
- This halving occurs during **meiosis,** and so it is described as a **reduction division** in which the **chromosome number is halved from diploid to haploid**, resulting in **genetically different cells**
- It starts with chromosomes doubling themselves as in mitosis and lining up in the centre of the cell
- After this has happened the cells divide twice so that only one copy of each chromosome passes to each gamete
- We describe gametes as being **haploid** - having half the normal number of chromosomes
- Because of this double division, meiosis produces **four** haploid cells



The process of cell division by meiosis to produce haploid gamete cells

Process:

- Each chromosome makes identical copies of itself (forming X-shaped chromosomes)

Your notes

- First division: chromosomes pair up along the centre of the cell, recombination occurs and then cell fibres will pull the pairs apart, each new cell will have one of each recombinant chromosome pair
- Second division: chromosomes will line up along the centre of the cell, cell fibres will pull them apart (as with mitosis)
- A total of four haploid daughter cells will be produced

Importance:

- Production of gametes e.g. sperm cells and egg cells, pollen grains and ovum
- Increases genetic variation of offspring
- Meiosis produces **variation** by forming **new combinations** of maternal and paternal **chromosomes** every time a gamete is made, meaning that when gametes fuse randomly at fertilisation, each offspring will be different from any others

Your notes

Differences between Mitosis & Meiosis

MITOSIS	MEIOSIS
TWO CELLS PRODUCED (KNOWN AS DAUGHTER CELLS)	FOUR CELLS PRODUCED (KNOWN AS DAUGHTER CELLS)
DAUGHTER CELLS ARE DIPLOID	DAUGHTER CELLS ARE HAPLOID
DAUGHTER CELLS ARE GENETICALLY IDENTICAL TO EACH OTHER AND TO THE PARENT CELL	DAUGHTER CELLS ARE GENETICALLY DIFFERENT FROM EACH OTHER AND THE PARENT CELL
ONE CELL DIVISION OCCURS	TWO CELL DIVISIONS OCCUR

Examiner Tips and Tricks

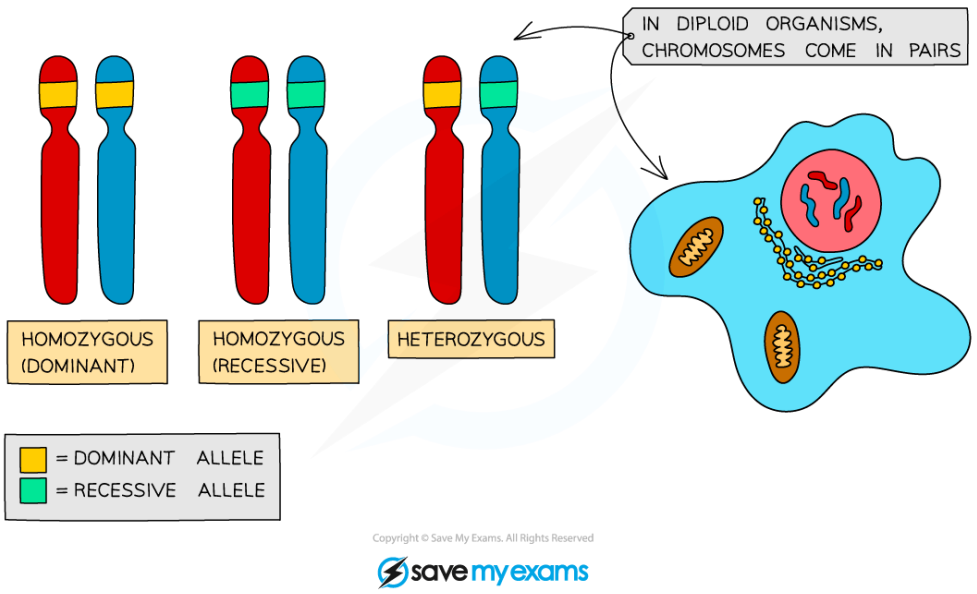
Questions on cell division often ask for differences between mitosis and meiosis. Learn two or three and remember to BE SPECIFIC when giving your answer.

You should also know the reasons for a specific type of cell division taking place and the types of cells where each happen.

Your notes

Inheritance: definitions

- **Inheritance** is the transmission of genetic information from one generation to the next generation
- A **gene** is a short length of DNA found on a chromosome that codes for a particular characteristic
- **Alleles** are versions, or forms, of a gene
  - Chromosomes exist in matching pairs, so individuals have two copies of each gene and therefore two alleles of each gene
    - One of the alleles is inherited from the mother and the other from the father
  - For example, an individual has two copies of the gene for eye colour; these alleles could be identical, or they could be different
- The observable characteristics of an organism 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 show up in the phenotype
  - A recessive allele needs to be **inherited from both parents** in order for the characteristic to show up 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)
- When completing genetic diagrams alleles are notated as single letters
  - The dominant allele is given a **capital letter** and the recessive allele is given the **lower case** version of the same letter



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Alleles of a gene can carry the same instructions or different instructions. You can only inherit two alleles for each gene, and they can be the same or different

- We **cannot always determine the genotype** of an individual just by looking at the phenotype
  - A phenotype associated with a dominant allele will be seen in both a dominant homozygous and a heterozygous genotype
- If two individuals who are both **identically homozygous** for a particular characteristic are bred together, they will **produce offspring with exactly the same genotype and phenotype as the parents**
  - We describe them as **pure breeding** as they will always produce offspring with the same characteristics
- A **heterozygous** individual can pass on **different alleles** for the same characteristic each time it breeds with any other individual and can therefore **produce offspring with a different genotype and phenotype than the parents**
  - heterozygous individuals are not pure breeding

Genetic diagrams

What is monohybrid inheritance?

- Monohybrid inheritance is the inheritance of characteristics controlled by a **single gene** (mono = one)
- This can be determined using a genetic diagram known as a **Punnett square**
  - A Punnett square diagram shows the **possible combinations of alleles** that could be produced in the offspring

- From this the **ratio** of these combinations can be worked out

Your notes

Worked Example

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**

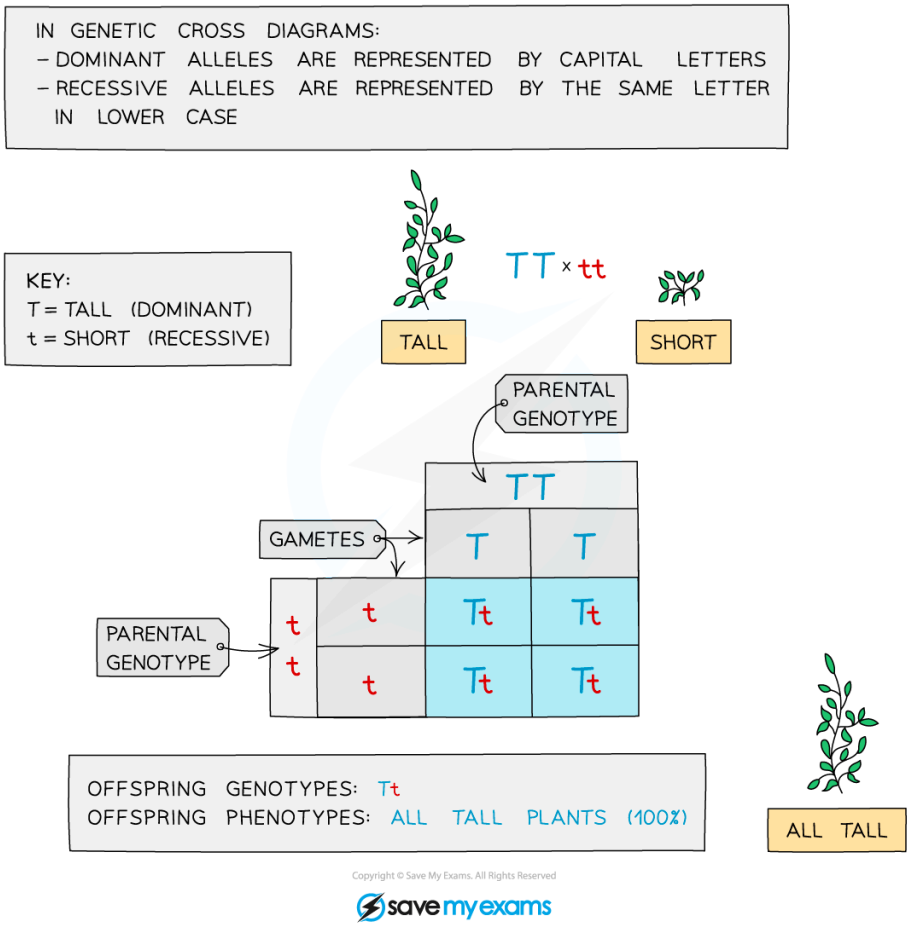
Show the possible allele combinations of the offspring produced when a homozygous short plant is bred with a homozygous tall plant. Determine the probability that any offspring will be tall

Step 1: construct a Punnett square

- The parents are homozygous, so:
  - tall = **TT**
  - short = **tt**
- The Punnet square should indicate:
  - parent gametes
  - offspring genotypes
  - an indication of which offspring are tall

Step 2: determine the probability of tall offspring

- All offspring are **Tt**
- The probability that they are tall = 100 %



Your notes

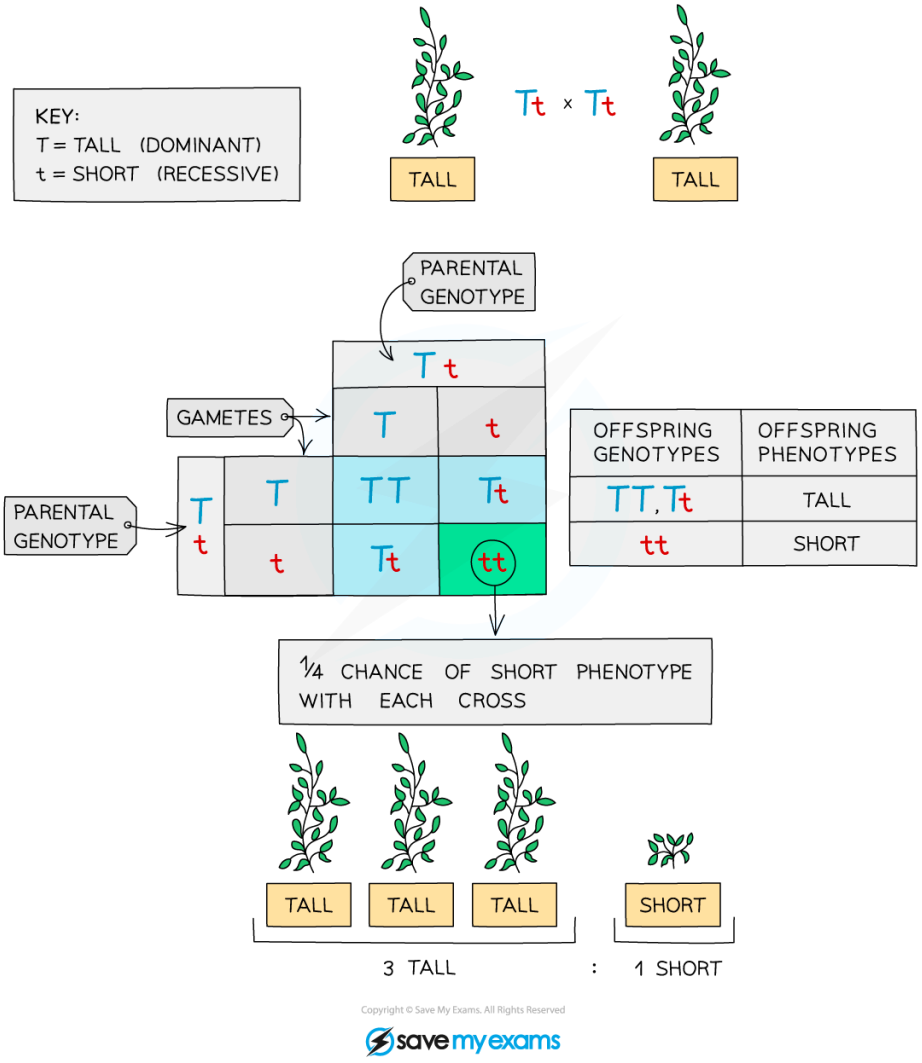
Show the possible allele combinations of the offspring produced when two of the offspring from the first cross are bred together. Determine the probability that any offspring will be short.

Step 1: construct a Punnett square

- The offspring from the first cross are all **Tt**
- The Punnet square should indicate:
  - parent gametes
  - offspring genotypes
  - an indication of which offspring are short

Step 2: determine the probability of short offspring

- Offspring genotypes = **TT**, **Tt**, **Tt**, **tt**
- Offspring phenotypes = 3 x tall, 1 x short
- The probability that they are tall = 25 %



Your notes

Worked Example

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**

Show the results of crossing a heterozygous plant with a short plant. Determine the probability of the offspring being short.

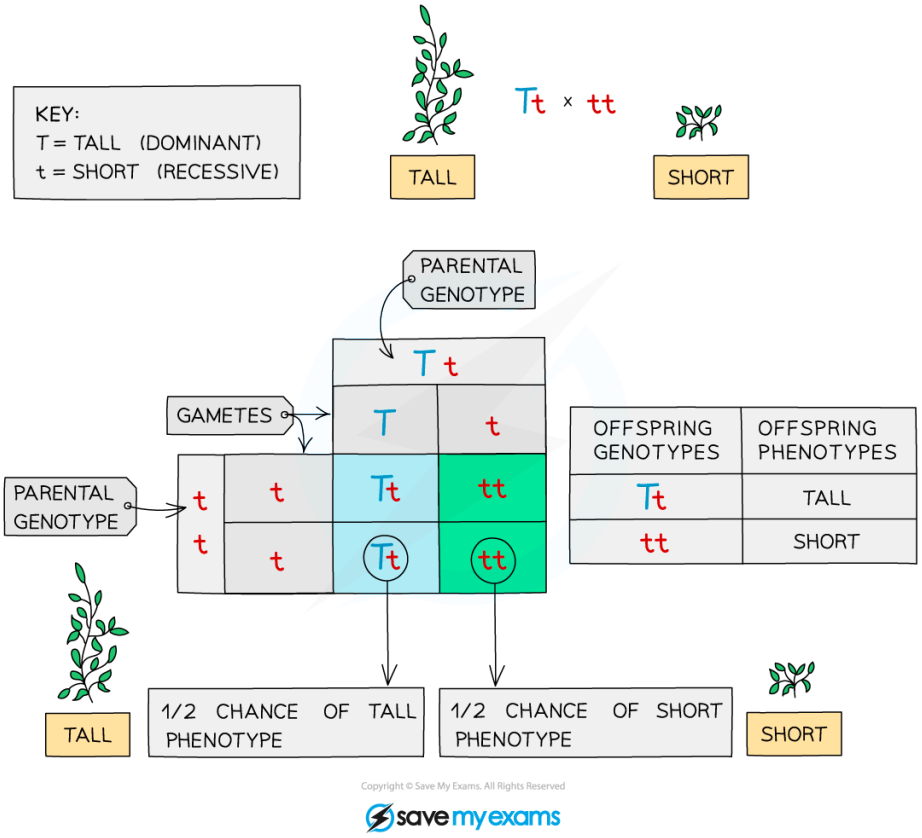
Step 1: construct a Punnett square

- The heterozygous parent has the genotype **Tt**
- The short parent has the genotype **tt**
- The Punnet square should indicate:
  - parent gametes
  - offspring genotypes

- an indication of which offspring are short

Step 2: determine the probability of short offspring

- Offspring genotypes = Tt, Tt, tt, tt
- Offspring phenotypes = 2 x tall, 2 x short
- The probability that they are tall = 50 %



Constructing Punnett squares

- You may be asked to determine the ratio of different allele combinations in the offspring, calculate a percentage chances of offspring showing a specific characteristic or just determine the phenotypes of the offspring; this can be done using a Punnett square as follows:

1. Determine the letter that will be used to notate each allele
    - Some exam Qs will provide this information
    - If not then you should select a relevant letter, e.g. E and e for **e**ye colour
2. Determine the parental genotypes
3. Determine the gametes produced by each parent and add them to the Punnett square headings
4. Fill in the middle four squares of the Punnett square to work out the possible genetic combinations in the offspring

Your notes

5. Indicate clearly on your Punnett square which individual will show each phenotype, e.g. with labels or colour coding

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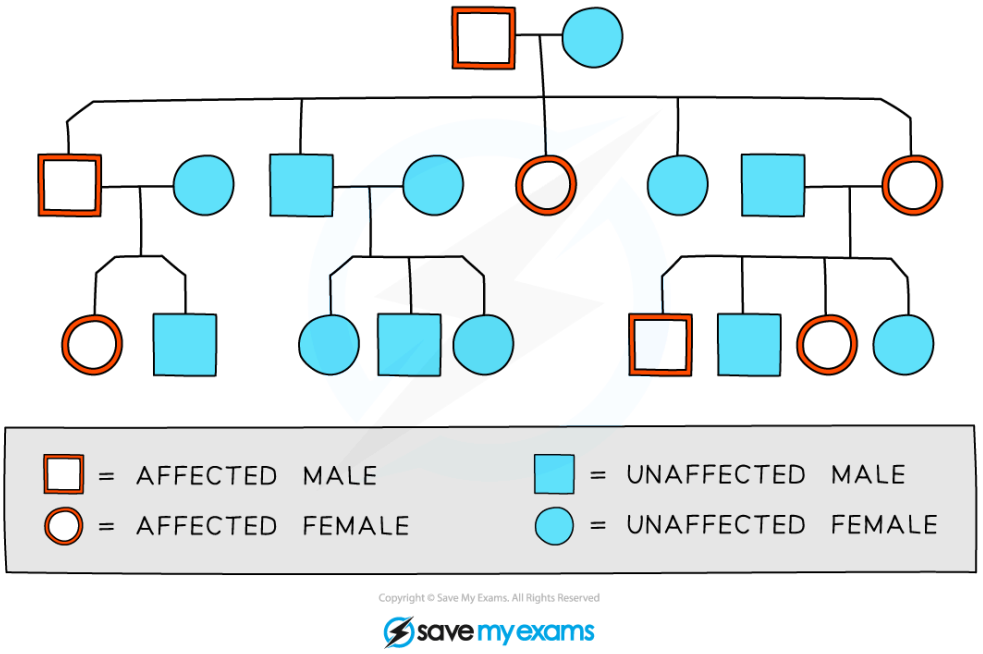
Examiner Tips and Tricks

If you are asked to use your own letters to represent the alleles in a Punnett square, 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.

For example, C and c are not very different from each other when written by hand, whereas A and a are!

Pedigree diagrams

- 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**
- Affected individuals are **red** and unaffected are **blue**
- Horizontal lines between males and females show that they have produced children (which are shown underneath each couple)
- The family pedigree above shows:

- both males and females are affected
- every generation has affected individuals
- 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

Identifying an unknown genotype: extended

- Breeders can use a **test cross** to find out the genotype of an organism showing the dominant phenotype
- This involves crossing the unknown individual with an individual showing the recessive phenotype - if the individual is showing the recessive phenotype, then its genotype must be homozygous recessive
- By looking at the **ratio of phenotypes in the offspring**, we can tell whether the unknown individual is homozygous dominant or heterozygous

Worked Example

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**

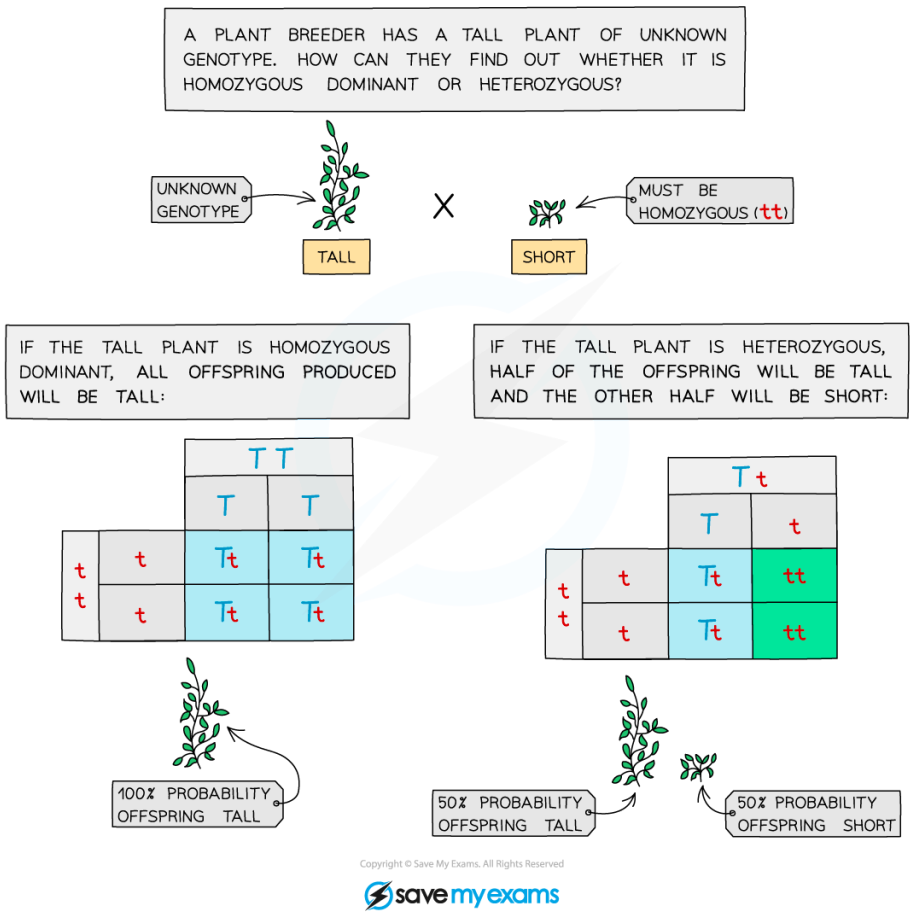
A plant breeder has a tall plant of unknown genotype. Show how they can find out whether it is homozygous dominant or heterozygous.

Step 1: construct Punnett squares

- The short plant is showing the recessive phenotype and so must be homozygous recessive **tt**
- The tall parent could be **either TT or Tt**, so we will need to produce two Punnett squares to show the different outcomes
- The Punnet squares should indicate:
  - parent gametes
  - offspring genotypes
  - which offspring are tall or short

Step 2: determine how the outcomes of the two crosses would differ

- Homozygous tall parent offspring = Tt = all tall
- Heterozygous tall parent offspring genotypes = Tt or tt = 50 % tall and 50 % short
- The presence of any short offspring indicate that the unknown parent has a heterozygous genotype



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Codominance: Extended

Extended Tier Only

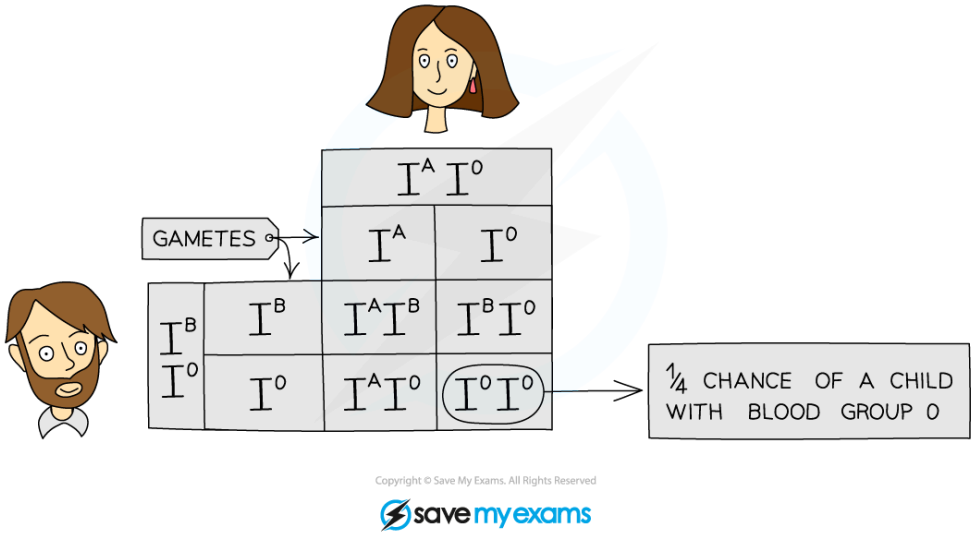
Codominance

- Codominance occurs when **both alleles in heterozygous organisms contribute** to the phenotype
- Inheritance of blood group is an example of codominance
- There are three alleles of the gene governing this instead of the usual two
- Alleles I^A and I^B are codominant, but both are dominant to I^O
- I represents the gene and the superscript A, B and O represent the alleles
- I^A results in the production of antigen A in the blood
- I^B results in the production of antigen B in the blood
- I^O results in no antigens being produced in the blood
- These three possible alleles can give us the following genotypes and phenotypes:

GENOTYPE	PHENOTYPE
$I^A I^A$ OR $I^A I^O$	A
$I^B I^B$ OR $I^B I^O$	B
$I^A I^B$	AB
$I^O I^O$	O

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

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



Punnett square showing the inheritance of Blood Group

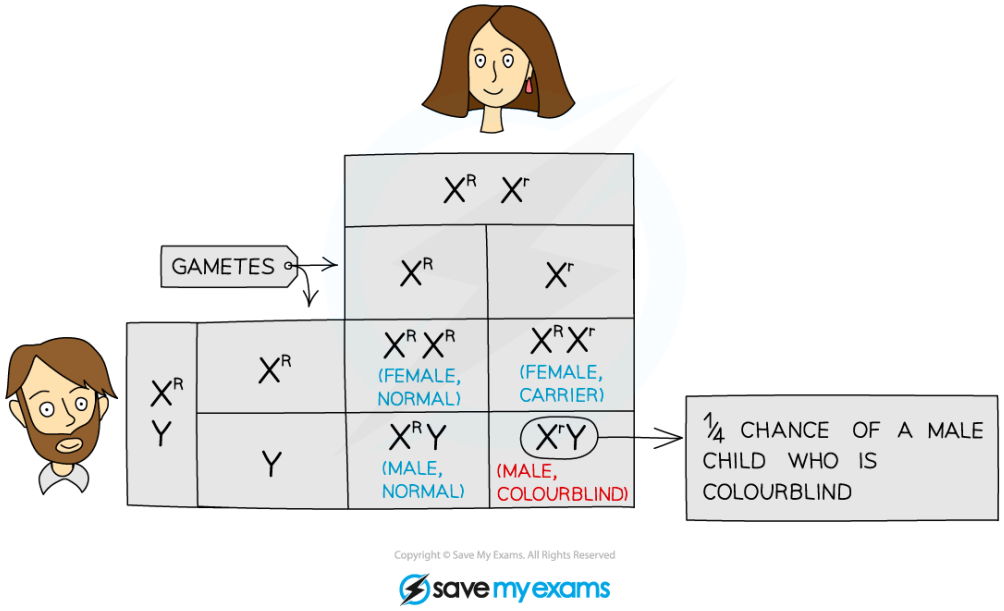
- The parent with blood group A has the genotype I^AI^O
- The parent with the blood group B has the genotype I^BI^O
- We know these are their genotypes (as opposed to both being homozygous) as they are able to produce a child with blood group O and so the child must have inherited an allele for group O from each parent
- Parents with these blood types have a 25% chance of producing a child with blood type O

Sex-Linked Characteristics: Extended

Extended Tier Only

- Alleles on the same chromosome are said to be **linked**
- When alleles that control a particular characteristic are found on the **sex chromosomes**, we describe the inheritance that results as **'sex linked'**
- In almost all cases, there are **only alleles on the X chromosome** as the **Y chromosome is much smaller**
- Because males **only have one X chromosome**, they are **much more likely to show sex-linked recessive conditions** (such as red-green colour blindness and haemophilia)
- Females, having two copies of the X chromosome, are likely to inherit one dominant allele that masks the effect of the recessive allele
- A female with one recessive allele masked in this way is known as a **carrier**; she doesn't have the disease, but she has a 50% chance of passing it on to her offspring
- If that offspring is a male, he will have the disease

- The results of a cross between a normal male and a female who is a carrier for colourblindness is as follows:



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

- In the cross above, there is a 25% chance of producing a male who is colourblind, a 25% chance of producing a female carrier, a 25% chance of producing a normal female and a 25% chance of producing a normal male

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