Genetics & Inheritance Scope

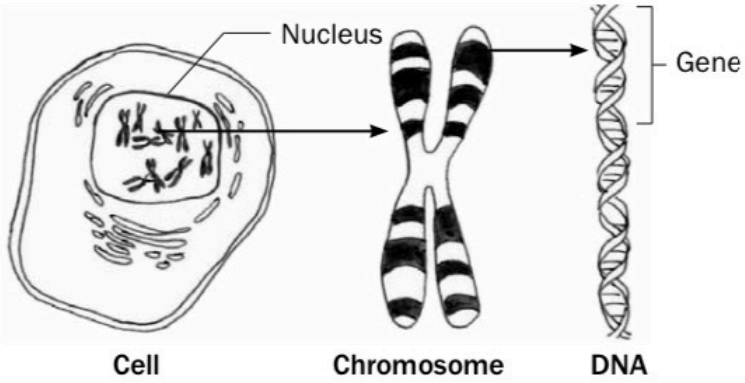
TOPIC	SUBTOPIC	KEY INFORMATION
GENETICS AND INHERITANCE	Genetic terms	• ALL GENETIC TERMS must be known well
	Complete dominance Incomplete dominance Co-dominance	• As stated in Exam Guidelines • State the Law of Dominance (see Examination Guidelines)
	Monohybrid crossing	• Understand and use the template • Position of meiosis and fertilization as well as P_1 and F_1 on the template • Answer the question at the end (usually for a compulsory mark) • Proportion and ratio of genotypes and phenotypes • State the Principle of Segregation (see Examination Guidelines)
	Inheritance of sex	• Differentiate between sex chromosomes (gonosomes) and autosomes (body cells) in the karyotypes of human males and females
	Sex-linked characteristics & disorders	• The correct way of writing it • Also in pedigree diagrams
	Blood groups	• Difference between genotype and phenotype of each blood group • Correct way of writing genotypes: e.g. I^Ai, I^Bi or ii or I^AI^B
	Dihybrid crossing	• Correct way of writing: Genotype: e.g. GGRR or GgRr Gametes: e.g. GR, Gr, gR, gr • Distinguish between dominant phenotype and dominant allele • Mendel's Principle of Independent Assortment (See Examination Guidelines)
	Pedigree diagrams	• Interpretation of pedigree diagrams • Give the phenotype, genotype • State which allele is inherited from each parent • The examiner DOES NOT have to provide a key
	Mutations	• Different types of mutations: Gene and Chromosome mutations • Different effects of mutations: harmful, useful and harmless • Refer to Examination Guidelines
	Genetic engineering: Stem cells, genetic modification and cloning	• Sources and uses • Benefits (Advantages and disadvantages) • Brief outline of processes
	Paternity testing and DNA profiling	• Describe how paternity testing is done not only identifying the father in a diagram but describe how the mother, father and child's DNA bars plays a role

Adapted from DBE revision guidelines

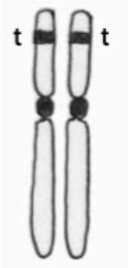
Genetics & Inheritance Notes

Key concepts

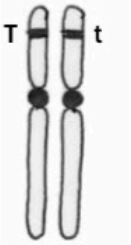
From mind the gap textbook

Term	Explanation	Diagram/Additional notes
Gene	A small portion of DNA coding for a particular characteristic.	
Alleles	Different forms of a gene which occur at the same locus (position) on homologous chromosomes.	**Dominant allele (T) – tall plant** **Recessive allele (t) – short plant**
Genotype	Genetic composition (make-up) of an organism.	
Phenotype	The physical appearance of an organism determined by the genotype, e.g. tall, short.	
Dominant allele	An allele that is expressed (shown) in the phenotype when found in the heterozygous (Tt) and homozygous (TT) condition.	
Recessive allele	An allele that is masked (not shown) in the phenotype when found in the heterozygous (Tt) condition. It is only expressed in the homozygous (tt) condition.	
Heterozygous	Two different alleles for a particular characteristic, e.g. Tt.	
Homozygous	Two identical alleles for a particular characteristic, e.g. TT or tt.	

- Homozygous dominant (both alleles are dominant)
- Genotype TT
- Phenotype – tall



- Homozygous recessive (both alleles are recessive)
- Genotype tt
- Phenotype – short

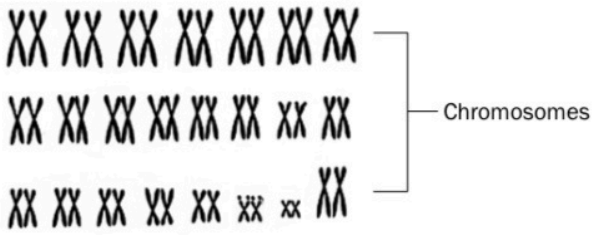


- Heterozygous (one dominant and one recessive allele)
- Genotype Tt
- Phenotype – tall

Genetics & Inheritance Notes

Key concepts

From mind the gap textbook

Term	Explanation	Diagram/Additional notes	
Monohybrid cross	Only one characteristic or trait is being shown in the genetic cross.	*Example*: Flower colour only, e.g. yellow flower or white flower **OR** shape of seeds only, e.g. round seeds or wrinkled seeds.	
Complete dominance	A genetic cross where the dominant allele masks (blocks) the expression of a recessive allele in the heterozygous condition.	In this type of cross the allele for tall (T) is dominant over the allele for short (t). The offspring will therefore **be tall** because the dominant allele (T) masks the expression of the recessive allele (t).	Tall (TT) × short (tt) ⟨ Tall (Tt)
Incomplete dominance	A genetic cross between two phenotypically different parents produces offspring different from both parents but with an intermediate phenotype.	*Example:* If a red-flowered plant is crossed with a white-flowered plant and there is incomplete dominance – the offspring will have **pink flowers (intermediate colour).**	Red flower – White flower ⟨ Pink flowers
Co-dominance	A genetic cross in which both alleles are expressed equally in the phenotype.	*Example:* If a red-flowered plant is crossed with a white-flowered plant and there is co-dominance the offspring has **flowers with red and white patches.**	Red flower × White flower ⟨ Flowers with red and white patches
Multiple alleles	More than two alternative forms of a gene at the same locus.	*Example:* Blood groups are controlled by three alleles, namely I^A, I^B and i.	
Sex-linked characteristics	Characteristics or traits that are carried on the sex chromosomes.	*Examples:* Haemophilia and colour-blindness The alleles for haemophilia (or colour-blindness) are indicated as superscripts on the sex chromosomes, e.g. X^HX^H (normal female), X^HX^h (normal female), X^hX^h (female with haemophilia), X^HY (normal male), X^hY (male with haemophilia).	
Karyotype	The number, shape and arrangement of all the chromosomes in the nucleus of a somatic cell.		
Cloning	Process by which genetically identical organisms are formed using biotechnology.	*Example:* Dolly the sheep was cloned using a diploid cell from one parent; therefore it had the identical genetic material of that parent.	
Genetic modification	The manipulation of the genetic material of an organism to get desired changes.	*Example:* The insertion of human insulin gene in plasmid of bacteria so that the bacteria produce human insulin.	
Human genome	The mapping of the exact position of all the genes in all the chromosomes of a human.	*Example:* Gene number 3 on chromosome number 4 is responsible for a particular characteristic.	

Genetics & Inheritance
Notes

Gregor Mendel

The father of genetics,.

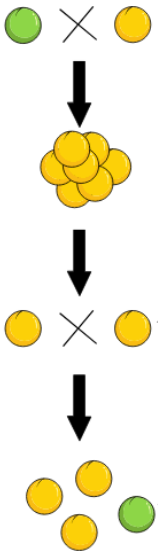
Mendelian experiment

Green x Yellow
gg x GG

All Yellow
Gg

Yellow x Yellow
Gg x Gg

Yello + Yellow + Yellow + Green
GG + Gg + Gg + gg



from his experiments with pea plants. The following laws were formulated:

Law(principle) of segregation

An organism possesses two 'factors' which separate or segregate so that each gamete contains only one of these 'factors',

Law of dominance

When two homozygous organisms with contrasting characteristics are crossed, all the individuals of the F1 generation will display the dominant trait
An individual that is heterozygous for a particular characteristic will have the dominant trait as the phenotype.

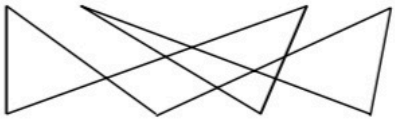
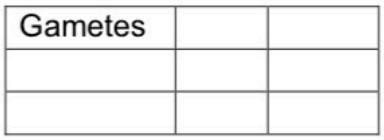
Law (principle) of independent assortment

The various 'factors' controlling the different characteristics are separate entities, not influencing each other in any way, and sorting themselves out independently during gamete formation.

Note: these laws cannot be paraphrased!

Genetics & Inheritance Notes

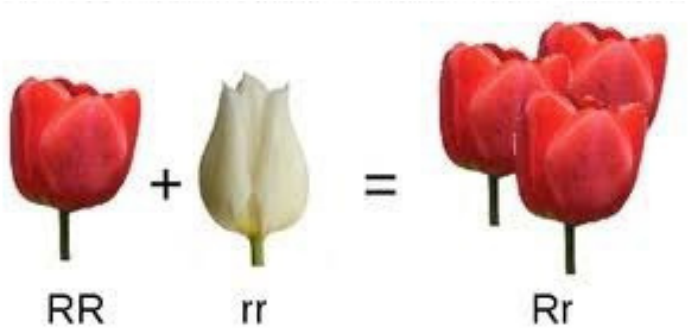
Genetic Diagrams

Layout of a genetic diagram	Explanation
P_1 Phenotype ..... x .....✓ Genotype ..... x .....✓ Meiosis Gametes ..... x ..... ✓ Fertilisation  **F_1** Genotype .....✓ Phenotype .....✓ P_1 and F_1 ✓ Meiosis and fertilisation✓	Visible trait is the **phenotype** e.g. tallness, shortness, etc. The genetic make-up of the individual is its **genotype** e.g. TT or Tt or tt The alleles segregate (or separate) during **meiosis** to form gametes. Each gamete has only one copy of each allele During **fertilisation** the individual gets one allele of the gene form each parent The matrix box used to determine the results of fertilisation is called a **Punnet square**
OR	
P_1 Phenotype ..... x .....✓ Genotype ..... x .....✓ Meiosis Gametes ..... x ..... ✓ Fertilisation  **F_1** Phenotype .....✓ Genotype .....✓ P_1 and F_1 ✓ Meiosis and fertilisation✓	

This is a template for a monohybrid cross.
It carries 2 free marks! Memorise!

Genetics & Inheritance

Types of dominance

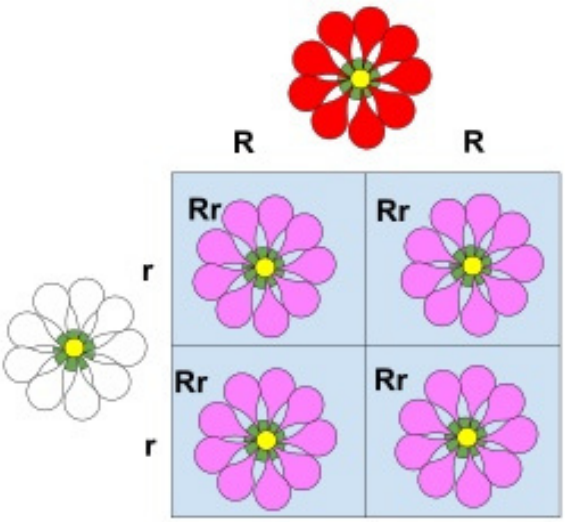


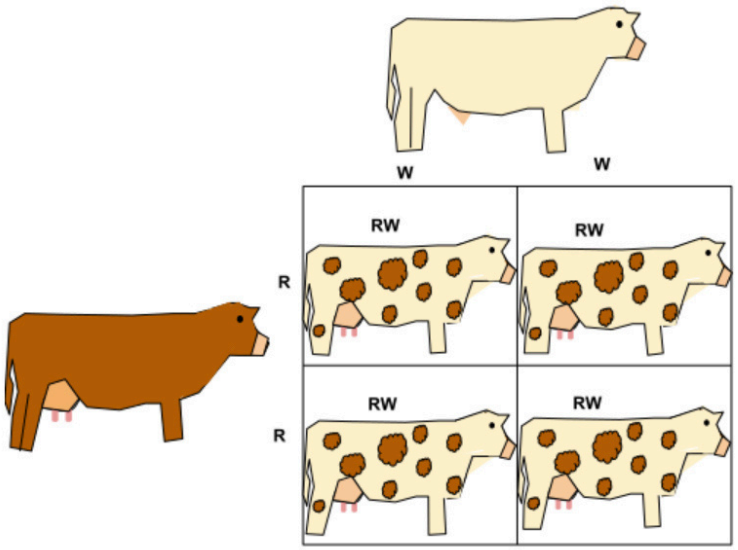
Complete dominance

In complete dominance, the dominant allele masks or blocks the expression of the recessive allele in the heterozygous condition.

Incomplete dominance

Incomplete dominance is a cross between two phenotypically different parents where no allele of the gene is either dominant or recessive. The offspring is different to both parents and the alleles blend to form a new or intermediate phenotype.





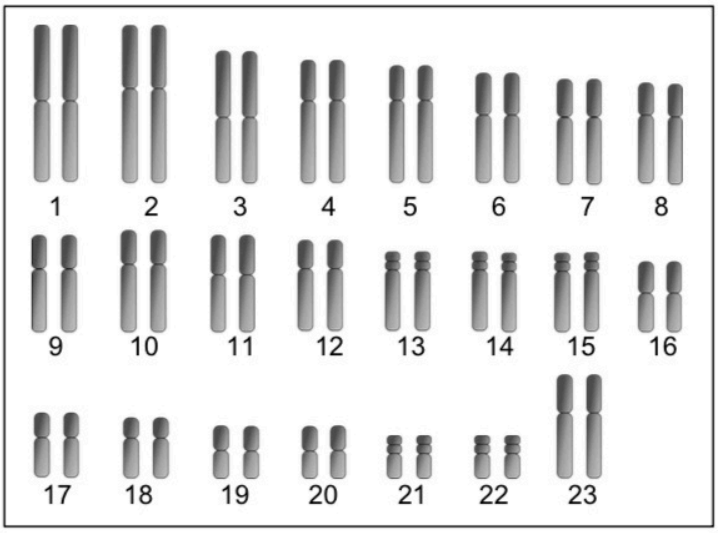
Co-dominance

In co-dominance both alleles of the gene are equally dominant so both will be expressed equally in the phenotype of the offspring.

Genetics & Inheritance

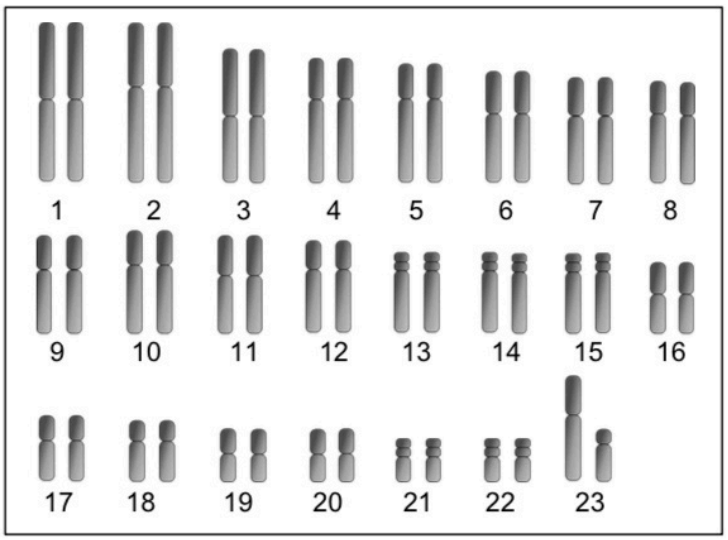
Sex determination

a genetic cross which shows inheritance of sex



Female karyotype

Gonosome of a female - XX



Male karyotype

Gonosome of a male - XY

P_1	Phenotype	**Male**	x	**Female**	
	Genotype	**XY**	x	**XX**	
Meiosis					
	Gametes	**X**	**Y** x	**X**	**X**

Fertilisation

Gametes	X	Y
X	XX	XY
X	XX	XY

F_1	Genotype	**XX**	**XX**	**XY**	**XY**
		2 (XX)		**2(XY)**	
	Phenotype	**Female (50%)**		**Male (50%)**	
	Phenotypic ratio	**1:1**			

The genetic cross shows that the percentage chance of having a boy or a girl is 50%.

Genetics & Inheritance

Sex linked inheritance

Inherited characeristics carried on sex chromosomes

Colour-blindness

• A person is colour-blind is unable to tell different colours apart. For example, red-green colour-blindness is caused by an absence of the proteins that make up the red or green cones (photoreceptors) in the retina of the eye resulting in the person not being able to tell the difference between red and green.

Genotype	Phenotype
X^HX^H	Normal female
X^HX^h	Normal female (carrier)
X^hX^h	Haemophiliac female
X^HY	Normal male
X^hY	Haemophiliac male

Genotype	Phenotype
X^BX^B	Female with Normal vision
X^BX^b	Normal Female (carrier)
X^bX^b	Colour-blind female
X^BY	Normal male
X^bY	Colour-blind male

Haemophilia

• Haemophilia is the inability of the blood to clot due to lack of a blood clotting factor. If the sufferer were to cut themselves, the wound would continue to bleed until a clotting factor is transfused in hospital.

 Colour-blindness and haemophilia is caused by the recessive allele on the X- chromosome normally shown as (Xb) for colour-blindness and (Xh) for haemophilia

The Y chromosome does not carry any sex-linked disorder

Genetics & Inheritance

Multiple alleles

Characteristics controlled by more than two alleles

Blood type

There are four blood types in humans: A, B, AB or O. These phenotypes are controlled by three alleles but each person still inherits two alleles.

Genotype	Blood group
I^AI^A	A
I^Ai	A
I^BI^B	B
I^Bi	B
I^AI^B	AB
ii	O

IA is co-dominant to **IB** whereas **i** is recessive to both.

Blood type in paternity testing

The blood groups of the mother, possible father and child must be compared. If the blood groups of the adults do not correspond to or match the child's blood group then this man is not the father. If the blood groups of the adults correspond to or match the child's blood group, then there is a possibility that the man is the father and other tests need to be done as other men may have the same blood group.

Only DNA profiling can be conclusive as it looks at the similarities between the nucleotides in the DNA of the father and the child.

Genetics & Inheritance

DiHybrid cross

Dihybrid crosses involve two pairs of alleles representing two different characteristics, e.g.: the height of a plant and the colour of its seeds.

Apply the principle of Independent assortment for the formation of gametes

P_1	Phenotype	**Tall, purple**	x	**Tall, purple**
	Genotype	**TtPp**	x	**TtPp**
Meiosis				
	Gametes	**TP Tp tP tp**	x	**TP Tp tP tp**

Fertilisation

Gametes	**TP**	**Tp**	**tP**	**tp**
TP	TTPP	TTPp	TtPP	TtPp
Tp	TTPp	TTpp	TtPp	Ttpp
tP	TtPP	TtPp	ttPP	ttPp
tp	TtPp	Ttpp	ttPp	ttpp

F_1	Genotype	**9 different genotypes, as in the table above**
	Phenotype	**9 tall, purple flowered plants**
		3 short, purple flowered plants
		3 tall, white flowered plants
		1 short, white flowered plant

Lets take offspring with genotype -TtPp & TtPP and form their possible gametes

Genotype: TtPp X Genotype: TtPP

Gametes: TP Tp tP tp X Gametes: TP TP tP tP

Mendel says factors controlling characteristics are separate entities and will separate independently during gamete formation. Take each gamete as a sperm cell. There's 4 sperm cells each time and each will have those alleles

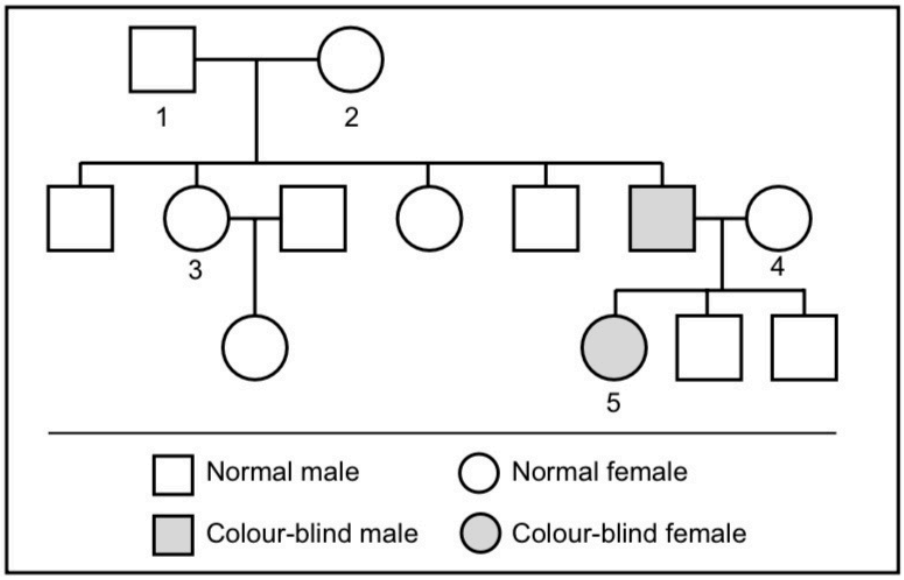
We apply the same rule for Ova formed. Only difference is only one ovum is released during ovulation. So one of the 4 gametes

Genetics & Inheritance

Genetic lineage

A **pedigree diagram** (also called a genetic lineage) is used to study the inheritance of characteristics in a family over a number of generations.

A genetic lineage showing inheritance of colourblindness



Key features

- There are 7 males and 6 females
- There are 3 generations in the family
- Only 1 male and 1 female is colourblind
- Parent 2 is heterozygous for colourblindness because one of their children is colourblind whereas some are not.

Colourblindness is sex-linked hence only parent 2 is mentioned ⬆️ (males only have one X.

- Person 4 is also heterozygous since they have a child with colourblindness.

	Genotype	Phenotype
Person 3 / Person 2	X^BX^B	Female with Normal vision
Person 3 / Person 2	X^BX^b	Normal Female (carrier)
Person 5	X^bX^b	Colour-blind female
Person 1	X^BY	Normal male
Husband of person 4	X^bY	Colour-blind male

Genetics & Inheritance

Mutations

A **mutation** is caused by a permanent change to the DNA of a cell.
Mutations can be **harmless**, **harmful** or **useful**.

Harmless mutations

- Changes the non-coding DNA
- This DNA is not involved in making proteins
- The structure or functioning of the cell/organism is not affected .

EG: freckles, blonde hair, baldness.

Useful mutations

- Changed the DNA responsible for the production of a specific protein.
- If the protein made increases the organism's chance of survival, it would be seen as a useful mutation.
- If the gene is passed on, it will lead to genetic variation that is advantageous to the individual.

Gene mutations

A change in nitrogenous base sequence of a DNA molecule.

Leads to:

- Haemophilia
- Colourblindness
- Sicklecell anemia
- Albinism

Harmful mutations

- Changes the coding DNA
- This DNA is responsible for the production of a specific protein.
- This changes the organism's physical appearance or functioning due to an incorrect / defective protein being made.

EG: may cause a genetic disorder. (Haemophilia, colourblindness, sickle cell anemia, albinism & Down syndrome)

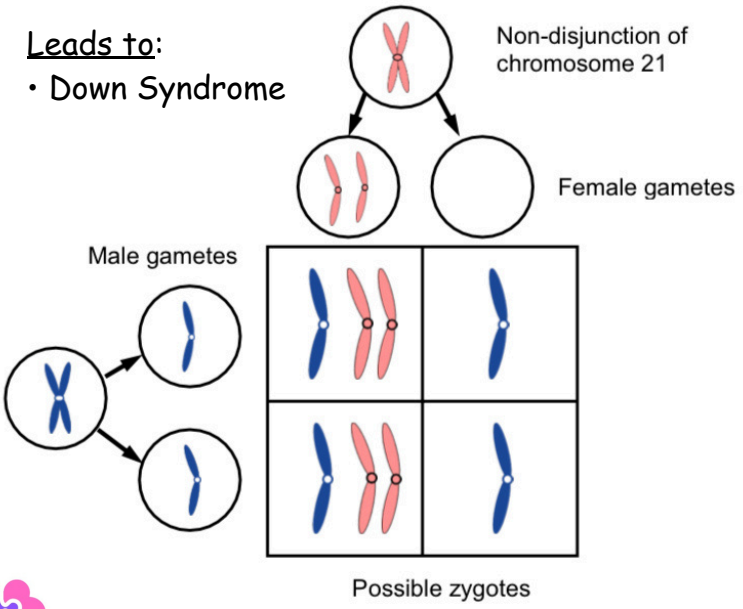
Mutations can occur in genes or chromosomes.

Chromosomal mutations

A change in the structure or number of chromosomes of a cell/organism.

Leads to:

- Down Syndrome



Genetics & Inheritance

Biotechnology

Biotechnology is the use of organisms (e.g. bacteria) or biological processes to improve the quality of human life.
examples in DNA profiling, genetic engineering, stem cell technology and cloning.

DNA Profiling

It is a form of biotechnology used for paternity testing, the identification of individuals, and for many other purposes.

Stem cells

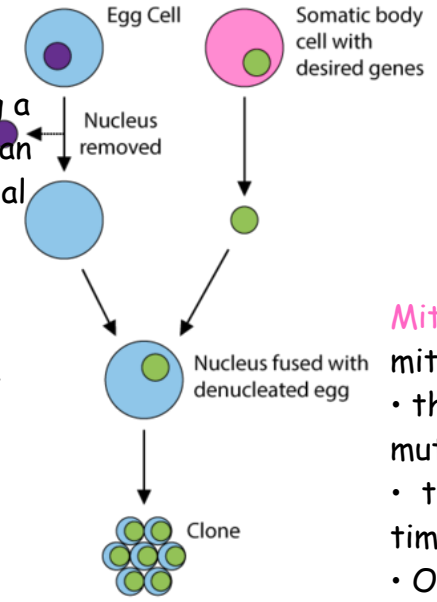
Stem cells are undifferentiated cells that have the ability to grow into any tissue in the body.

Two types:
- embryonic stem cells
- adult stem cells

Uses:
- replacing dead cells after a heart attack
- growing skin tissue to treat burn victims
- growing nerve cells to treat spinal cord injuries and Parkinson's disease

Cloning

Cloning is the natural or artificial process of creating a genetically identical copy of an organism or biological material (e.g. tissue).



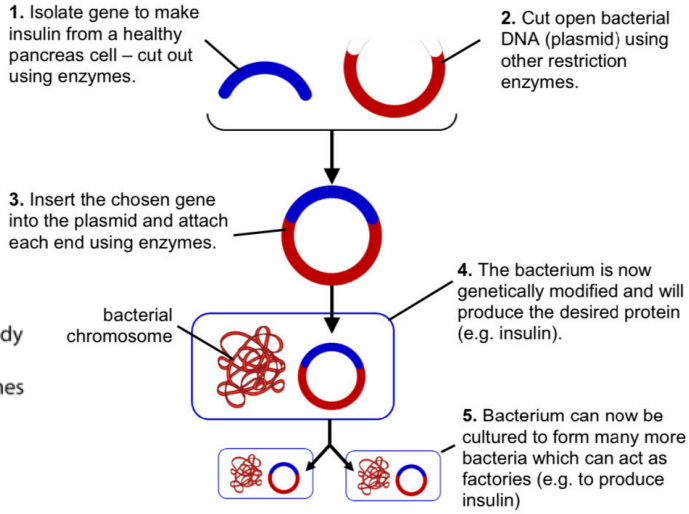
Advantages:
- replace damaged tissue e.g. skin, heart
- Genetic diseases could be prevented.
- improve food supply and quality.

Genetic engineering

Genetic engineering is used to alter the genome of a living cell for medical, industrial or agricultural purposes. This results in a genetically modified organism (GMO) or transgenic animal (animal with DNA from more than one species).

GMO's are used:
- to breed more productive crops/animals to increase yield
- to produce drugs or hormones (e.g. insulin)
- to 'infect' cells to cure diseases (gene therapy) such as brain tumours and cystic fibrosis

Recombinant DNA technology



Mitochondrial DNA

Mitochondrial DNA (mtDNA) is found in mitochondria
- the only changes that occur are due to mutations.
- these mutations are used to work out a timeline of genetic ancestry.
- Only the mother's mtDNA is passed on to her offspring
- the scientists compare the mutations of different people to see how closely related they are.

Genetics & Inheritance

Terminology

Biological term	Description
Albinism	The condition that results from the absence of skin pigmentation
Alleles	Two alternative forms of a gene at the same locus
Artificial selection/selective breading	The breeding of organisms over many generations in order to achieve a desirable phenotype
Biotechnology	The use of biological processes, organisms or systems to improve the quality of human life
Clone	A copy of an organism that is genetically identical to the original organism
Cloning	The process by which genetically identical organisms are formed using biotechnology
Co-dominance	The type of inheritance where both alleles are equally dominant and both express themselves equally in the phenotype. E.g. A white cow crossed with a black bull will produce a calf with black and white patches
Complete dominance	The type of inheritance where the dominant allele masks the expression of the recessive allele in the heterozygous condition
Continuous variation	Type of variation within a population in which there is a range of intermediate phenotypes
Chromatin network	Visible as thread-like structures in the nucleus of an inactive cell
Chromosome	A structure made up of two chromatids joined by a centromere that carries the hereditary characteristics within the DNA
Dihybrid cross	A genetic cross involving two different characteristics e.g. shape and colour of seeds
Dominant allele	An allele that masks or suppresses the expression of the allele partner on the chromosome pair and the dominant characteristic is seen in the homozygous (e.g.: TT) and heterozygous state (e.g.: Tt) in the phenotype.
Gene	A segment of DNA/a chromosome that codes for a particular characteristic
Gene mutation	A change of one or more N- bases in the nuclear DNA of an organism.
Genetic variation	This includes a variety of different genes that may differ from maternal and paternal genes resulting in new genotypes and phenotypes.
Genotype	This is the total **genetic composition** of an organism. It is the information present in the gene alleles, for example BB, Bb or bb.
Genome	The complete set of chromosomes in the cell of an organism
Haemophilia	A sex-linked genetic disorder characterised by the absence of a blood-clotting factor
Heterozygous	An individual having two non-identical alleles for a characteristic

Genetics & Inheritance

Terminology

Biological term	Description
Homozygous	When two alleles that control a single trait (on the same locus) are **identical**.
Hypothesis	A tentative explanation of a phenomenon that can be tested and may be accepted or rejected
Incomplete dominance	The type of inheritance where both alleles express themselves in such a way that an intermediate phenotype is formed. E.g. A white flowering plant crossed with a red flowering plant will produce a pink flowering plant.
Locus	The exact position or location of a gene on a chromosome.
Mendel's Law of Dominance	When two individuals with contrasting homozygous alleles are crossed, the individuals of the first generation (F_1) will **ALL** resemble the parent with the dominant characteristic.
Mendel's Law of Independent Assortment	Alleles of a gene for one characteristic segregate independently of the alleles of a gene of another characteristic. The alleles for the two different genes will therefore come together randomly during gamete formation. This is also known as random assortment.
Mendel's Principle of Segregation	During gametogenesis the two alleles of a gene separate so that each gamete will receive one allele of a gene for a specific characteristic/trait.
Monohybrid cross	A genetic cross involving one characteristic e.g. colour of seeds
Mutation	A sudden change in the sequence/order of nitrogenous bases of a nucleic acid
Multiple alleles	When there are more than two possible alleles for one gene locus. e.g. blood groups
Phenotype	This is the external, **physical appearance** of an organism. The phenotype is determined by the genotype.
Pedigree diagram	A diagram showing the inheritance of genetic disorders over many generations
Recessive allele	An allele that is suppressed when the allele partner is dominant. The recessive trait will only be expressed/seen if both alleles for the trait are homozygous recessive e.g.: tt
Stem cells/meristematic cells	Undifferentiated cells that can develop into any cell type
Theory	Explanation of an observation that is supported by facts, models and laws