**DNA: CODE OF LIFE**

**Nucleic acids**

Two types: DNA (Deoxyribonucleic acid) and RNA (Ribonucleic acid)

- Both DNA and RNA made up of nucleotides

Structure of nucleotide:

- Nitrogenous base: Purines and pyrimidines
  - Purines: Adenine (A) and Guanine (G)
  - Pyrimidines: Cytosine (C), Thymine (T) and Uracil (U)
  - Purines pairs with pyrimidines: Complementary base pair i.e. C-G, A-T in DNA but A-U in RNA
- Sugar portion (S): Deoxyribose in DNA; Ribose in RNA
- Phosphate portion (P): Same in DNA AND RNA

**DNA**

Location:

- In the nucleus: Nuclear DNA
  - Make up most of the DNA
  - Works with proteins to form chromatin network
- Outside the nucleus (Extra – chromosomal DNA)
  - In little quantities
  - Chloroplastic DNA and Mitochondrial DNA

Functions of DNA

- Forms proteins
- Heredity: parents to offspring

**RNA**

Three types:

- Ribosomal RNA (rRNA): Ribosomes in cytoplasm
- Messenger RNA (mRNA): nucleus, later attaches to ribosome
- Transfer RNA (tRNA): Cytoplasm

Functions of RNA

- Play a role in formation of proteins (Protein synthesis)

**Similarities between DNA and RNA**

- Sugar alternates with phosphate
- Nitrogenous bases: Adenine, Guanine and Cytosine
- Play a role in protein synthesis

**Differences between DNA and RNA**

| DNA | RNA |
|---|---|
| Double strand i.e. double helix | Single strand |
| Found in nucleus, chloroplast and mitochondrion | Found in nucleus, ribose and cytoplasm |
| Deoxyribose sugar | Ribose sugar |
| Adenine pairs with Thymine | Adenine pairs with Uracil |

**DNA replication**

- DNA makes an identical copy of itself
- It occurs before cell division (mitosis and meiosis) i.e. during Interphase
- It ensures equal number of chromosomes in daughter cells and also with the original

Process of DNA replication

- Double helix unwinds
- Weak hydrogen bonds between the nitrogenous bases break
- And the two DNA strands unzip
- Each original DNA strand act as a template to form a new strand
- By attaching free nucleotides from the nucleoplasm
- To form complementary (matching) strands i.e. A-T and C-G
- Each DNA molecule now consists of 1 original strand and 1 new strand
- The result is two genetically identical DNA molecules
- The entire process is controlled by enzymes

**DNA profile**

- Black bars left on x-ray film when extract of DNA is put through a special biotechnical process
- DNA profiling: method of identifying an individual by comparing his/her DNA profile with another known DNA profile

Uses of DNA profile

- Diagnosis of inherited disorders e.g. Haemophilia
- Identification of criminals

- Identification of brothers and sisters separate at birth
- Identification of a child's father: Paternity test
- Identification of unrecognized dead bodies

Debates against DNA profiling

- It is expensive
- It may reveal personal information such as HIV/AIDS that can be used against the person
- Human error in laboratory can lead to false results

**PROTEIN SYNTHESIS**

Occur in three stages: Transcription, movement of mRNA out of nucleus and Translation.

Transcription- formation of mRNA

- DNA double helix unwinds
- Weak hydrogen bonds of DNA break
- Forming two single strands of DNA
- One strand acts as a template (original)
- To form complementary strand which is mRNA
- Using free nucleotides from the nucleoplasm
- This process is called transcription
- Three adjacent bases on mRNA make up a codon
- The codon codes for an amino acid

Movement of mRNA out of the nucleus

- mRNA moves out of the nucleus
- through the nuclear pore
- into the cytoplasm
- where it attaches to the ribosome

Translation – using information from mRNA to form protein

- codons of mRNA matches with anticodons of tRNA
- to bring the required amino acids to the ribosome
- the amino acids link together by peptide bonds
- to form the required protein
- The process is called translation.

**MEIOSIS**

- It occurs after interphase
- It has four stages:
  - Prophase – first stage
  - Metaphase - middle stage
  - Anaphase – separation stage
  - Telophase – last (terminal) stage
- It has two parts: Meiosis I and Meiosis II

Differences between Meiosis I and Meiosis II

| Meiosis I | Meiosis II |
|---|---|
| Chromosomes double stranded | Chromosomes single stranded |
| Crossing-over takes place in Prophase I | No crossing-over |
| Chromosomes in homologous pairs at equator in Homologous pairs: Metaphase 1 | Chromosomes in individuals at equator: Metaphase 2 |
| Whole chromosomes are pulled to opposite poles in Anaphase 1 | Chromatids are pulled to opposite poles in Anaphase 2 |
| Chromosome number is halved during meiosis I | Chromosome number does not change during meiosis II |
| Results in two cells | Results in four cells |

The importance of meiosis:

- Forms haploid gametes or spores in organisms
- It maintains constant number of chromosomes from one generation to the next
- Introduces genetic variation: Prophase I (crossing over) and Metaphase I and II (random arrangement of chromosomes)

Similarities between mitosis and meiosis

- DNA replication takes place
- The nucleus divides
- The cytoplasm divides
- New cells are formed

Differences between Mitosis and Meiosis

| Mitosis | Meiosis |
|---|---|
| Forms somatic cells | Forms sex cells |
| One nuclear division | Two nuclear division |
| Two cells formed with same number of chromosomes as parent | Four cells formed with half the number of chromosomes as the parent |
| Two cells genetically identical to each other and to the parent | Four cells genetically different from each other and to the parents |
| During prophase, the chromosomes are not in pairs | During prophase I, the chromosomes come together in pairs |

| | |
|---|---|
| No crossing over | Crossing-over takes place during prophase I |
| Chromosomes splits into chromatids that are pulled in opposite poles | Whole chromosomes are pulled in opposite poles |

**Abnormal meiosis**

The following abnormalities may occur during meiosis:

- In anaphase I: one or more homologous pairs of chromosomes may not separate
- In Anaphase II: sister chromatids of one or more chromosomes may not separate
- The above abnormalities are together called non-disjunction and may lead to aneuploidy or polyploidy
    - Aneuploidy: gametes have one less (Monosomic) or one more (Trisomic) chromosome
    - Polyploidy: gametes have one extra set of chromosomes (3n) or two extra sets of chromosomes (4n)
- If the abnormal gamete fuse with a normal gamete or another abnormal gamete, they result in different genetic disorders e.g. Down syndrome

Down syndrome:

- It is an example of aneuploidy
- Extra chromosome at number 21 (trisomy 21)
- Occurs when a gamete with two copies of chromosome number 21 fuse with a gamete having one copy of chromosome number 21
- The result is a zygote with three copies of chromosome number 21 resulting in Down syndrome
- The individual with Down syndrome has 47 chromosomes instead of 46 chromosomes

Symptoms of Down syndrome

- Mental retardation
- Hearing loss
- Heart defects
- Decreased muscle tones
- Upwardly slanting eyes
- Small mouth and nose
- Abnormal ear shape
- Depressed nasal shape

- No cure for Down syndrome. Symptoms are treated.
- Down syndrome can be detected in unborn babies by **amniocentesis**. Amniocentesis – removal of foetal cells from mother's uterus to detect disorders in unborn babies.

Polyploidy

- It occurs when:
  - 2n gamete (abnormal) + n gamete (normal) = 3n (Triploid) zygote OR
  - 2n gamete (abnormal) + 2n gamete (abnormal) = 4n (Tetraploid) zygote
- It is common in plants especially angiosperms
- Rare in animals but found in some fishes, insects, amphibians and reptiles
- The only polyploid mammal known is a rat found in Argentina in 1999
- Scientists use polyploidy to produce new species
  Advantages of polyploidy
  - Larger fruits e.g. watermelons
  - Plants with large storage organs
- Polyploid individuals are created in laboratories by treating diving cells with colchicine. This drug prevents formation of spindle during mitosis. As a result, the duplicated chromosomes fail to separate.

**DIVERSITY OF REPRODUCTIVE STRATEGIES IN VERTEBRATES**

Two types of fertilisation methods during reproduction of vertebrates: External and internal fertilisation

| External fertilisation | Internal fertilisation |
|---|---|
| Sperm fertilises the ovum outside female's body | Sperm fertilises the ovum inside female's body |
| No physical contact required between parents | There is a physical contact between parents |
| Water required for reproduction:<br>- Prevents drying out of eggs<br>- Allows sperm to swim towards the egg | Water not required:<br>- Adaptation of terrestrial animals to reproduce in dry areas |
| Large quantities of eggs and sperms produced | Little quantities of eggs and sperms produced |
| e.g. Frogs | e.g. Humans |

Internal fertilisation is divided into three types

- Oviparous
- Viviparous
- Ovoviviparous

Oviparous

- Egg layers
- Eggs fertilised internally before being laid or externally after being laid
- The eggs develop outside the mother's body
- Eggs hatches into a young animal e.g. frogs, birds etc.
  Disadvantage: Eggs exposed to predators
  Parents guard the eggs to increase chance of survival

Viviparous

- Young develop inside the mother's uterus after internal fertilisation
- Young get nutrients from the mother's blood through the placenta
- Young ones are born active e.g. Humans

Ovoviviparous

- Young develops from the eggs fertilised internally
- The eggs are retained within the mother's body after fertilisation
- The developing young obtains nutrients from the egg yolk and not from the mother
- The young then hatch inside the mother's reproductive system and born soon afterwards
- E.g. Reptiles and Fish.
  Advantages of vivipary and ovovivipary:
  - Young ones born active
  - Parental care
  - Internal fertilisation increases chances of offspring being produced

**Amniotic egg**

- It is found in reptiles and birds
- The amniotic egg is covered by a shell
- It has four membranes: Amnion, Chorion, Allantois and Yolk Sac

Amnion

- It encloses amniotic cavity with amniotic fluid
- It protects the embryo against mechanical shock

Chorion

- Allows gaseous exchange

Allantois

- Stores waste from the embryo
- Functions in gaseous exchange like chorion

Yolk sac

- Provides food to the embryo

Egg shells reduce dehydration. This allows birds and reptiles to occupy a wide range of habitats than amphibians

Mammals do not have an egg shell. Embryos are adapted to avoid dehydration by developing within their mother

Precocial and Altricial development

- In young birds (chicks) depending on availability or absence of predators

| Precocial development | Altricial development |
|---|---|
| Many predators | No predators |
| Lots of food available | Little food available |
| Eggs produced with lot of energy | Eggs produced with less energy |
| Well developed when the hatch:<br>- Eyes open<br>- Soft feathers called Down's feathers<br>- Can move around soon after hatching<br>- Able to feed themselves<br>- Not dependant on their parent | Poorly developed when they hatch:<br>- Eyes closed<br>- No Down's feathers<br>- Unable to move around<br>- Unable to feed themselves<br>Dependant on their parent |
| e.g. Turkeys and quails | In 60% of birds e.g. finches, crows etc. |
| Some species show both characteristics of Precocial and Altricial development e.g. hawks and owls | |

Parental care is shown in the following ways:

- Building of nests and burrows
- Care of eggs
- Provision of food for unborn/un-hatched young
- Provision of food for born/hatched young
- Protection of the young
- Providing social assistance to mature offspring

**HUMAN REPRODUCTION**

Male reproductive structure:

- Pair of testes that produce sperms
- Tubes related to carrying spermatozoa to the outside
  - Epididymis: Stores sperms temporarily
    Transfer sperms into the vas deferens
  - Vas Deferens/Sperm duct: Carries sperms from epididymis into the ejaculatory duct
  - Ejaculatory duct: Forces semen through the urethra
  - Urethra: Passage of semen and urine
- Accessory glands (seminal vesicle, prostate gland and Cowper's gland): Secrete fluid that promotes movement of semen. The fluid supply the spermatozoa with nutrients
- Penis: Transfers spermatozoa from male to female

Female reproductive structure:

- Pair of ovaries: produce eggs (ova)
- Pair of fallopian tubes (oviducts): Transfer ova from ovaries to uterus
- Uterus (Womb): Development of embryo
- Cervix: Neck of uterus extending to vagina
- Vagina: Receives spermatozoa from the male
- Vulva: External opening of the vagina

Gametogenesis: formation of gametes (sex cells)

Divided into two types: Spermatogenesis and oogenesis

Spermatogenesis:

- Forms spermatozoa from germinal epithelium of the testes
  - Cells of germinal epithelium undergo meiosis
  - Each cell that undergoes meiosis produces four haploid spermatids
  - The spermatids develop into a spermatozoa

Spermatozoa is divided into head, middle piece and tail

- Head
  - Made mainly of nucleus
  - 22 autosomes and Y-chromosome with nuclear DNA
  - Acrosome at the tip: enzyme that penetrates the ovum
- Middle piece
  - Made of numerous mitochondria (**Play a role in cellular respiration**)
  - Mitochondria provide the sperm with energy for locomotion
  - Mitochondrial DNA (**Mitochondrial Adam in evolution**)
- Tail
  - Enables the spermatozoa to swim very fast

Oogenesis

- Forms ova from germinal epithelium of ovaries
  - Germinal epithelium in ovary undergoes mitosis
  - Numerous follicles are produced
  - Each follicle has four cells inside
  - One out of four cells inside the follicle enlarges and undergoes meiosis
  - The enlarged cell survives and produce mature ovum

**Menstrual cycle** – changes that occur in the ovary and uterus of a female over a period of about 28 days

It is divided into ovarian cycle and uterine cycle

Ovarian cycle and ovulation:

- Controlled by FSH secreted by Pituitary gland
- Occurs inside the ovary
- Inside the ovary there are number of Graafian follicles
- Each Graafian follicle contains an ovum at different stage of development
- The more mature ova are in larger Graafian follicles
- The Graafian follicles releases **Oestrogen**
- Oestrogen prepares the uterus for attachment of fertilised ovum (implantation)
- Graafian follicle ruptures every 4 weeks to release an ovum: Ovulation
- The ovum is collected by funnels of the fallopian tubes
- LH converts ruptured follicle into a corpus luteum
- Corpus luteum releases **Progesterone**
- Progesterone maintains pregnancy if fertilisation has occurred
- Corpus luteum degenerates and drops progesterone levels if fertilisation did not occur
- The unfertilised ovum passes down the fallopian tube into the body and leaves the body by means of menstruation

Uterine cycle and menstruation

- Graafian follicle secretes oestrogen
  - Prepares the uterus for pregnancy
  - It makes the endometrium thicker, more vascular and more glandular
- After ovulation, corpus luteum secretes progesterone
  - Prepares uterus for implantation if fertilisation occur
  - Corpus luteum continues secreting progesterone if the ovum is fertilised
  - Corpus luteum becomes destroyed if the ovum is not fertilised
  - The progesterone levels decreases
  - The destroyed corpus luteum and egg results in menstruation

Menstruation

- Endometrium lining of the uterus comes off as menstrual period (bleeding)
- Menstruation lasts for about 5 days
- Menstruation occur every 14 days after ovulation

When a female stops releasing ova, she stops menstruating: Menopause (Between 45 and 55 years)

**Fertilisation and development of zygote into a blastocyst**

- Spermatozoa are released into the vagina during sexual intercourse
- The spermatozoa swim up the uterus into the fallopian tubes
- If an ovum is present at that time, one spermatozoon may penetrate and fertilise the ovum resulting in a diploid zygote i.e. sperm cell (n) + egg cell (n) = zygote (2n)
- The zygote contains genetic material from both male and female parent
- As the zygote passes down the fallopian tube into the uterus, it divides by mitosis to form a hollow ball of cells called the blastocyst

**Development into an embryo**

- The blastocyst develops into an embryo by the time it reaches the uterus
- The embryo becomes attached to the endometrium: Implantation
- Oestrogen has already prepared the uterus for implantation
  - It made the endometrium thick
  - Richly supplied with blood vessels
  - More glandular
- Since fertilisation took place, the corpus luteum continue secreting progesterone
  - Progesterone keeps the embryo attached to the endometrium

**Implantation and development**

- Soon after implantation, the blastocyst develops two extra-embryonic membranes around itself: Amnion and Chorion

Amnion:

- On the inside
- It has amniotic cavity filled with amniotic fluid. Functions of amniotic fluid are
  - Protection of embryo from mechanical injury
  - Prevents embryo from dehydration
  - Keeps the embryo within a small temperature range
  - Allows free foetal movement for growth and development

Chorion: on the outside and forms chorionic villi

- The chorionic villi together with the uterine tissue in which the villi are inserted makes up the placenta. Functions of placenta:
  - Attaches embryo to the mother
  - Allows diffusion of dissolved food from mother to the foetus
  - Allows gaseous exchange between the mother and the foetus
  - Allows diffusion of nitrogenous excretory waste from foetus to the mother
- The placenta is attached to the foetus by umbilical cord. The umbilical cord has umbilical artery and umbilical vein
  - Umbilical artery: Carries deoxygenated blood with nitrogenous waste from the foetus to the placenta

- Umbilical vein: Carries oxygenated blood with dissolved food from placenta to the embryo

**Gestation period (pregnancy)**

- The period during which the embryo develops inside the mother's uterus until the time the baby is born
- During this period, the embryo increases in shape and size as it grows

**Contraception (Contra**ry **to** Con**ception)**

- Contraceptives are used by humans to prevent pregnancy
- There are different methods of contraceptives

| Method | Effect |
|---|---|
| Condom | Stops sperms from getting into the vagina |
| Female condom (Femidom) | Stops sperms from getting into the uterus |
| Contraceptive pill | Contains artificial hormones that stops production of ova |
| Diaphragm | Prevents sperms from entering the uterus |
| Spermicides | Kills sperms; prevents sperms from entering the uterus |
| Male sterilisation (Vasectomy) | Sperm ducts are cut and tied. Semen without sperm is produced |
| Female sterilisation (Tubal litigation) | The fallopian tubes are cut and tied to prevent fusion of sperm and egg |
| Rhythm | Sexual intercourse is avoided three to four days before and after ovulation<br>This is between days 10 and 18 of the menstrual cycle |

**GENETICS AND INHERITANCE**

Genetics: Study of heredity

Heredity: characteristics passed from one generation to the other through the information coded in DNA

Gene: portion of DNA that carries genetic code

Allele: two genes occupying identical position and controlling the same characteristic

- Dominant allele: expressed in both genotype and phenotype
- Recessive allele: expressed only in genotype i.e. it is hidden

Homozygous organism (Pure-breed): it has two copies of the same allele for a particular characteristic

Heterozygous organism (Mixed-breed/hybrid): it has two copies of different alleles for a particular characteristic

Incomplete dominance: No allele is dominant over the other. The result in third phenotype which is in between the two phenotypes

Codominance: when both alleles are equally dominant. They produce third phenotype which is combination of the two phenotypes

Monohybrid cross: cross (reproduction) between two individuals involving the inheritance of one characteristic

Dihybrid cross: cross (reproduction) between two individuals involving the inheritance of two characteristics

Monohybrid crosses for complete dominance:

- To represent alleles
  - A letter is chosen according to the dominant allele
  - The upper case of the letter will represent dominant allele
  - The lower case of the letter will represent recessive allele
  - Genotype is expressed with letters
  - Phenotype is expressed with words

Consider the following genetic problem:

The allele for black fur is dominant over the allele for white fur in rabbits. A homozygous black rabbit is crossed with a homozygous white rabbit. Determine the possible phenotypes and genotypes of the offspring.

| | | | | |
|---|---|---|---|---|
| Key | Colour: B – Black | | b – white | |
| $P_1$ | Phenotype: | Black | x | White |
| | Genotype: | BB | x | bb |

Meiosis

| | | | |
|---|---|---|---|
| Gametes: | B ; B | x | b ; b |

Fusion/Fertilisation

| | | | | | |
|---|---|---|---|---|---|
| $F_1$ offspring | Genotype: | Bb | Bb | Bb | Bb |
| | Phenotype: | Black | Black | Black | Black |

Now consider the following problem:

A heterozygous black rabbit is crossed with a homozygous white rabbit. Determine the possible phenotypes and genotypes of the offspring

Key Colour: B – Black b – white

| | | | | |
|---|---|---|---|---|
| $P_1$ | Phenotype: | Black | x | White |
| | Genotype: | Bb | x | bb |

Meiosis

| | | | |
|---|---|---|---|
| Gametes: | B ; b | x | b ; b |

Fusion/Fertilisation

| | | | | | |
|---|---|---|---|---|---|
| $F_1$ offspring | Genotype: | Bb | Bb | bb | bb |
| | Phenotype: | Black | Black | White | White |

Monohybrid crosses for incomplete dominance:

In an incomplete dominance cross, a homozygous black rabbit is crossed with a homozygous white rabbit. Determine the possible phenotypes and genotypes of the offspring.

Key Colour: B – Black W – white

| | | | | |
|---|---|---|---|---|
| $P_1$ | Phenotype: | Black | x | White |
| | Genotype: | BB | x | WW |

Meiosis

| | | | |
|---|---|---|---|
| Gametes: | B ; B | x | W ; W |

Fusion/Fertilisation

| | | | | | |
|---|---|---|---|---|---|
| $F_1$ offspring | Genotype: | BW | BW | BW | BW |
| | Phenotype: | Grey | Grey | Grey | Grey |

Monohybrid crosses for codominance:

In a codominance cross, a homozygous black rabbit is crossed with a homozygous white rabbit. Determine the possible phenotypes and genotypes of the offspring.

Key Colour: B – Black W – white

$P_1$ Phenotype: Black x White

Genotype: BB x WW

Meiosis

Gametes: B ; B x W ; W

Fusion/Fertilisation

$F_1$ offspring Genotype: BW BW BW BW

Phenotype: Black & white Black & white Black & White Black & white

**The inheritance of blood groups in humans**

- Four blood types: **A**, **B**, **AB** and **O**
- There are three alleles controlling the four blood types i.e. $\mathbf{I^A}$, $\mathbf{I^B}$ and **i**
- An individual can have only two of the three alleles making up his or her genotype
- The **i** allele is recessive to $\mathbf{I^A}$ and $\mathbf{I^B}$ alleles. The $\mathbf{I^A}$ and $\mathbf{I^B}$ are co-dominant to each other

Consider the following genetics problem:

A man homozygous for blood type A married a woman heterozygous for blood type B. Determine the possible genotypes and the phenotypes of the offspring.

$P_1$ Phenotype: A x A

Genotype: $\mathbf{I^AI^A}$ x $\mathbf{I^BI^i}$

Meiosis

Gametes: $\mathbf{I^A ; I^A}$ x $\mathbf{I^B ; I^i}$

Fusion/Fertilisation

$F_1$ offspring Genotype: $\mathbf{I^AI^B}$ $\mathbf{I^AI^i}$ $\mathbf{I^AI^B}$ $\mathbf{I^AI^i}$

Phenotype: **AB** **A** **AB** **A**

**The inheritance of sex cells in humans**

- Every human body cell has 23 pairs of chromosomes. The 22 pairs are autosomes and 1 pair is sex cells (gonosomes)
- The are two types of sex cells: XX in females and XY in males
- There is a 50-50 chance of an offspring being a male/female. Consider the following cross:

Key Male: XY Female: XX

$P_1$ Phenotype: Male x Female

Genotype: XY x XX

Meiosis

Gametes: X ; Y x X ; X

Fusion/Fertilisation

$F_1$ offspring Genotype: XX XX XY XY

Phenotype: Female Female Male Male

**The inheritance of sex-linked characteristics**

- Genes for most bodily characteristics are located on 22 pairs of autosomes
- The 23rd pair contains sex cells. The sex cells also contain genes for certain bodily characteristics: sex-linked characteristics
- The X and Y chromosomes are different. In some cases, if an allele for a particular trait is on the X chromosome, there is no allele for the same trait on the Y chromosome because Y chromosome is smaller in size
- If the allele on the X chromosome in a male is a recessive one, then the recessive characteristic will automatically show up in an individual because there is no dominant allele on the Y chromosome to hide the effect of the recessive allele.
- Haemophilia and colour blindness are two examples of sex-linked genetic disorders.

Haemophilia

- Condition where blood does not clot easily because of absence of clotting factors
- It mostly affects males
- It is a recessive condition since it is caused by a recessive allele found on X-chromosome
- If the dominant allele (normal) is shown by 'H' then the allele for haemophilia will be shown by 'h' since it is a recessive condition

Consider the following genetics problem:

A normal male marries a female with haemophilia. Determine the possible genotypes and phenotypes of their children.

Key Normal blood: H Haemophiliac: h

$P_1$ Phenotype: Normal Male x Haemophiliac Female

Genotype: $X^HY$ x $X^hX^h$

Meiosis

Gametes: $X^H$ ; Y x $X^h$ ; $X^h$

Fusion/Fertilisation

$F_1$ offspring Genotype: $X^HX^h$ $X^HX^h$ $X^hY$ $X^hY$

Phenotype: Normal Normal F Haemophilia M Haemophiliac M

**Colour-blindness**

- Various types exists
- The most common type is red-green colour-blindness, where a person cannot distinguish between red and green
- Red-green colour blindness is caused by a recessive allele
  - It is inherited in exactly the same way as haemophilia
  - Males more affected than females

**Dihybrid crossing**

- It involves inheritance of two characteristics e.g. height and shape, colour and shape etc.
- Gregor Mendel crossed pure breeding plants having seeds that are round and yellow with pure breeding plants with wrinkled and green seeds. Round was dominant over wrinkled and yellow dominant over green. His experiment for the dihybrid crosses can be represented as follows:

Key Shape: R – round r - wrinkled

Colour: Y – yellow y - green

$P_1$ Phenotype: Round and yellow x Wrinkled green

Genotype: RRYY x rryy

Meiosis

Gametes: RY ; RY ; RY ; RY x ry ; ry ; ry; ry

Fusion/Fertilisation

$F_1$ offspring Genotype: RrYy ; RrYy ; RrYy ; RrYy; #RrYy ; RrYy ; RrYy ; RrYy; # RrYy ; RrYy ; RrYy ; RrYy; # RrYy ; RrYy ; RrYy ; RrYy

Phenotype: All 16 offspring round and yellow

Mendel then crossed the $F_1$ generations

$P_2$ Phenotype: Round and yellow x Round and yellow

Genotype: RrYy x RrYy

Meiosis

Gametes: RY ; Ry ; rY ; ry x RY ; Ry ; rY; ry (Binomial&trinomial)

Fusion/Fertilisation

$F_2$ offspring Genotype: **RRYY**[1]; **RRYy**[2] ; **RrYY**[3]; **RrYy**[4] ; # **RRYy**[5] ; RRyy[1] ; **RrYy**[6] ; Rryy[2] ; # **RrYY**[7] ; **RrYy**[8] ; rrYY[1] ; rrYy[2]; # **RrYy**[9]; Rryy[3] ; rrYy[3] ; **rryy**[1]

Phenotype:

Round and yellow - 9

Round and green - 3

Wrinkled and yellow – 3

Wrinkled and green – 1

Phenotypic ratio 9:3:3:1

Based on the two above experiments, Gregor Mendel developed two laws

1. *Law of segregation*
   - For each characteristic, plants possess two 'factors' which separate so that each gamete contains one of each of these factors.
2. *Law of Independent Assortment*
   - The various 'factors' controlling different characteristics are separate entities , not influencing each other in any way, and sorting themselves out independently during gamete formation.

**Mutations**

- Sudden change to a structure of a gene
- They occur suddenly and randomly and may be caused by many environmental factors such as x-rays, UV rays, some chemicals etc.
- They are also called gene mutations
  - Gene mutations affect small parts as compared to larger chromosomal changes called chromosomal mutations or chromosomal aberrations

Gene mutations

- Involves individual genes
- Two types of gene mutations: Point mutations and Frame-shift mutations

Point mutation

- Also known as base substitution
- Change in a single base pair in DNA molecule at just one point
- It could lead to altered characteristics e.g. Sickle-cell anaemia

Frameshift mutation

- A single base pair may be inserted or deleted from a DNA molecule
- DNA will be different from point of insertion onwards
- It could lead to altered characteristics e.g. Albinism

Chromosomal mutations

- Changes in normal structure or number of chromosomes
- Chromosomes break and then rejoin incorrectly i.e. backwards or to the wrong chromosome
- Occur mostly in meiosis I when homologous pair of chromosomes fail to separate (Non-disjunction)

Effects of mutations

- They may be harmful or harmless to the organism

**Harmful (Lethal) mutations**: results in death so that harmful traits are not passed to the next generation

**Harmless mutations may be of two types:**

- Neutral mutations: no effect on the organism
- Advantageous mutations:
  - Passed on to the next generation
  - It may wipe all the other alleles controlling the same characteristics resulting in fixed mutation

**Genetic disorders caused by mutations**

- Down syndrome
  - Also called trisomy 21
  - Three copies of chromosome number 21 instead of two copies
- Sickle-cell anaemia
  - Mutant allele on chromosome 11
  - It causes red blood corpuscles to be sickle-shaped
  - It blocks small blood vessels
- Haemophilia
  - Failure of blood to clot due to absence of clotting agent
- Albinism
  - Lack of skin pigmentation called melanin

Mutations, Natural Selection and Evolution

- Mutations contribute to variations
- Mutations results in changed genotypes leading to new phenotypes
- Only organisms with advantageous mutations are able to survive
- This is called natural selection
- The organisms pass their favourable traits to the next generation
- Speciation occurs as a result of natural selection

**Genetic modification**

- It is deliberate change of the traits of an organism by manipulating its genotype using biotechnology
  - DNA of *E. coli* (bacterium) is recombined with a small DNA portion responsible for producing insulin, the bacterium is then tricked to produce the human insulin quickly
  - The above method can also be used to produce disease resistant plants or increase yield of some food types
  - The foods produced by genetic modification are called Genetically modified foods (GMF)

Arguments for genetic modification

- It allows production of medication or other resources cheaply
- It results in pest resistant and drought resistant crops
- Crop yield can be increased which will then increase food security
- It increases shelf life of fruits and vegetables which will then decrease food wastage

Arguments against genetic modification

- Initially it is an expensive process
- We are interfering with nature as we are trying to play God
- It may have potential health impacts
- We are unsure of long term effects of using GMOs

**Stem-cell research**

- Stem cells: undifferentiated cells that have the ability to differentiate and form any tissue or organ in the body
- Sources of stem cells
  - Embryo
  - Blood from umbilical cord and placenta (cord blood)
  - Stem cells from cord blood is becoming more popular than embryonic stem cells as they do not involve destruction of embryos to obtain the stem cells
- Uses of stem cells
  - Replacement of diseased or damaged organs e.g. islets of Langerhans
  - Treatment of diseases such as leukaemia, sickle cell anaemia etc.

Arguments for usage of stem cells

- Embryos produced for in vitro fertilisation are destroyed anyway, it is acceptable for them to be used to help others
- Stem cells from cord blood do not damage the embryo

Arguments against usage stem cells

- Life begins as soon as the sperm fertilises the egg, so usage of human embryos for research is immoral
- Usage of stem cells from cord blood is unacceptable as scientist will be trying to play God

Cloning: production of an individual that is genetically identical to the organism from which it was produced using biotechnology

- When a haploid sperm fuses with a haploid egg, a diploid zygote is formed which then develops into an embryo eventually forming a new organism.

**Tracing genetic lineage using pedigree diagrams**

- A lineage or pedigree: line of descent that links existence of individuals or groups of individuals to their recent or distant ancestor
- When genetic information is used, it is called genetic lineage
- Pedigree diagrams: usually used to trace human ancestors
    - It represents parents, grandparents, great-grandparents and beyond

Uses of pedigree diagrams

- To learn how different traits are passed from parents to offspring
- To determine the probability of an offspring in a family having a particular genetic disorder
- To show dominant or recessive trait in a family and whether it affects males only or both males and females
- Remember the following steps when interpreting a pedigree diagrams
    - Study the key and opening statement(s) providing clues about phenotypes, dominant and recessive traits
    - Use the information from the key to write down phenotypes of all the individuals below each square or circle
    - Fill in the genotype of all the individuals with the recessive traits – it must have 2 recessive alleles (to lower case letter e.g. rr)
    - Fill in the first letter of the genotype in individuals that show dominant trait (capital letter e.g. R)
    - For every individual with two recessive alleles, it means that it has inherited one allele from each parent. Work forward and backwards to fill in the recessive allele for each parent and offspring
    - Any other individual showing the dominant characteristic will now be homozygous dominant (RR)or heterozygous dominant (Rr)

**HUMAN NERVOUS SYSTEM AND SENSE ORGANS**

- Humans use two systems to respond to the environment: the nervous system with its sense receptors and the endocrine system
- Nervous system detects changes in the environment and allow the body to react to these changes
- Nervous system also enables co-ordination of various activities of the body
- Nervous system is made up of nerves. Nerves are made up of nerve fibres. The nerve fibres have specialised nerve cells called neurons. There are three types of neurons:
    - Sensory neuron: Carry nerve impulses from receptors (e.g. nerve endings and sense organs) to central nervous system (brain and spinal cord)
    - Motor neurons: Carry impulses from the central nervous system to the effectors (muscles or glands)
    - Connector/interneuron: carry impulses within the central nervous system from sensory to motor neuron

- All neurons consist of a cell body with its dendrites and an axon. Nerve impulses always travel from the dendrites to the cell body to the axon.
    - The axon is covered by two membranes: myelin sheath and neurilemma
    - Myelin sheath: Provides electrical insulation and helps to speed up transmission of impulses
- Many neurons are involved in carrying nerve impulses
    - The neurons are lined up in such a way that axon terminals of one neuron lie to the dendrites of another
    - The neurons are not directly connected to each other. There is a microscopic gap (synapse) between the axon terminal of one neuron and the dendrites of another
    - Nerve impulses are carried along the neuron by electrical impulses but communication across the synapse is by means of chemicals called neurotransmitters.
    - The communication is called synaptic contact.

Parts of the nervous system

- Nervous system is divided into two parts
    - Central Nervous System (CNS): brain and spinal cord
    - Peripheral nervous system: all nerves outside the CNS

Central Nervous System

The Brain

- It has three parts: Cerebrum, cerebellum and brain stem

Cerebrum

- Largest part of the brain
- It has two hemispheres connected by bundles of nerve fibres, the largest of the bundles being corpus callosum
- Below corpus callosum is the thalamus
- Below thalamus is the hypothalamus

Functions of cerebrum

- It controls all voluntary actions
- It receives and interprets all sensation i.e. sight, hearing, smell, taste and touch
- It controls all the higher thought processes such as memory, judgement and reasoning

Hypothalamus controls:

- Body temperature
- Blood pressure
- Sleep
- Appetite
- Thirst
- Emotions

Functions of cerebellum

- Co-ordinates voluntary movements e.g. walking and running
- Maintains muscles tone, balance and equilibrium

The brain stem is made up of mid-brain, the pons Varolli and the medulla oblongata

Functions of medulla oblongata

- It contains reflex centres for involuntary actions such as breathing, regulation of heart beat, vasoconstriction and vasodilation etc.
- It conducts impulses from the spinal cord to the higher parts of the brain and vice versa
- It results in left side of the brain controlling the right side of the body and vice versa by allowing impulses to cross on the other side

The spinal cord

- It conducts impulses between the brain and the receptors and effectors
- It serves as a centre for reflex actions
- Reflex action: rapid, automatic response to a stimulus received by an organ or other receptor
- The path taken by an impulse in bringing about response to a stimulus during a reflex action is called reflex arc. Reflex arc is the functional unit of the nervous system
- Reflex arc involves
  - The spinal cord
  - The spinal nerves with three types of neurons
  - A receptor (located in sense organs) that picks up changes in the environment
  - An effector (muscle or gland) that responds to the changes
- Steps in a reflex action
  - Receptors receives the stimulus and convert it to nerve impulse
  - Sensory neuron conducts the nerve impulses through the dorsal root of the spinal nerve into the spinal cord
  - Connector neuron transmits impulses from sensory neuron to motor neuron
  - Motor neuron transmits impulses out of the spinal cord to the effector (muscle or gland)

**Peripheral nervous system**

- It is divided into somatic nervous system and autonomic nervous system

Somatic nervous system

- All parts of nervous system allowing the body to react to changes in the external environment
- It is made up of
  - Free nerve endings
  - Sense organs
  - Sensory nerves
  - Motor nerves

- Effector

Autonomic nervous system

- It is made up of
  - Centres in the brain and spinal cord, nerve cells in smooth muscles, glands and internal organs
  - Sympathetic and parasympathetic nerves running to the muscles, glands and internal organs. The sympathetic and parasympathetic nerves work antagonistically (in opposite directions) e.g. sympathetic nerves increases the heart rate while the parasympathetic nerves decrease the heart rate
- Its main function is to control sub-conscious activities of the body e.g. heartbeat, peristalsis, vasoconstriction etc. This helps to maintain homeostasis

**Disorders of the nervous system**

Alzheimer's disease

- It occurs when nerve tissues within the brain becomes damaged
- It usually occurs in very old people but can occur to people in their early 40s and 50s
- Memory loss and confusion are the symptoms of the disease
- No treatment as yet

Multiple sclerosis

- It occurs when the body's own immune system attacks and destroys the myelin sheath covering the neurons
- Patients have physical and mental disabilities
- The cause is unknown. Genetics, infections and environmental factors seem to play a role
- No treatment. Medical treatment available tries to slow the rate at which the disease progresses

**Injuries to the central nervous system**

- They may be caused by
  - injury to the brain or spinal cord
  - Stroke which reduces blood flow to one or more parts of the brain
- The effects of injuries depends on the part of the brain damaged
  - Medulla oblongata: breathing, swallowing and salivation will be affected
  - Back of cerebrum: poor vision
  - Cerebellum: Balance and equilibrium as well as co-ordination of involuntary movements will be affected
  - Spinal cord: impulses from the brain to and from different parts of the body will be affected

**Effects of drugs on central nervous system**

- Impulses are transmitted through a neuron and then across a synapse to another neuron by neurotransmitters
- The use of drugs may either stimulate or inhibit the action of the neurotransmitters
- As a result, drugs may have stimulant or depressant effects on the user
- Some common effects of drugs includes memory loss, paranoia, anxiety and confusion

**SENSE ORGANS AND SENSE RECEPTORS**

**Human eye**

3 layers ($S^o$,$C^m$,$R^i$): Sclera, Choroid and Retina

Sclera – Outer layer

Parts: Cornea and conjunctiva

Choroid - Middle layer

- Rich in blood vessels: Supply the eye with food

Parts: Ciliary body with ciliary muscles and suspensory ligament

Iris with 2 sets of muscles (circular and radial) and pupil in the middle

Retina – Inner layer

Parts: Lens, Optic nerve, yellow spot/fovea, blind spot

- 2 types of light receptors (Rods: Dim light and Cones: Colour vision & bright light)

Two substances filling the eye:

- Aqueous humour: Liquid between cornea and lens
- Vitreous humour: Jelly-like substance filling up the space behind the lens

Optic nerve: carries nerve impulses from the eye to the cerebrum for interpretation

Accommodation – change in lens shape to form clear images on retina for near and distant (far) objects.

| Near vision ( < 6m) | Distant vision ( > 6m) |
|---|---|
| Ciliary muscles contract | Ciliary muscles relax |
| Suspensory ligaments loosen | Suspensory ligaments stretches |
| Tension on lens capsule decreases | Tension on lens capsule increases |
| Lens more round | Lens less round (flattens) |
| Less light enter the eye | More light enter the eye |
| Clear image formed on the retina: near object | Clear image on the retina: distant object |
| | |

**Pupillary mechanism** – change in diameter of the pupil to control amount of light entering the eye.

| Dim light | Bright light |
|---|---|
| Radial muscles of the iris **contract** | Radial muscles of the iris **relax** |
| Circular muscles of the iris **relax** | Circular muscles of the iris **contract** |
| Pupil **big** (dilates) | Pupil **small** (constricts) |
| **More light** enters the eye | **Less light** enters the eye |
| | |

**Diseases and disorders of the eye**

Cataracts

Cause: cloudy, milky (opaque) lens

Treatment: Replacement of the cloudy lens with man-made (synthetic) lens by operation (surgery)

Astigmatism

Cause: Uneven surface of cornea

Symptoms: Blurred (unclear) images, headache and fatigue, eye irritation.

Treatment: Prescription lenses

| Long-sightedness (Hypermetropia) | Short-sightedness (Myopia) |
|---|---|
| Causes: - Eyeball too round | Cause: - Eyeball too long |
| - Lens unable to be more round (Convex) | - Lens unable to be flat enough (less convex) |
| Symptom: blurred images of near objects | Symptom: blurred images of distant objects |
| - Clear images of near objects falls behind the retina | - Clear images of distant objects falls in front of the retina |
| Treatment: Glasses with convex lenses | Treatment: Glasses with concave lenses |

**The Human ear**

3 parts: Outer ear, Middle ear and Inner ear

Outer ear

- Pinna
- Auditory canal with cerumen (wax) and hairs
- Cerumen (wax) and hairs prevent dust and small organisms from entering the ear.
    - Cerumen also prevents ear drum from drying out

Middle ear: air-filled cavity

- Tympanic membrane (ear drum): Separates outer and middle ear
- 3 ossicles ($H^oA^mS^i$):
  - Hammer (Malleus)
  - Anvil (Incus)
  - Stirrup (Stapes)
- Eustachian tube: maintains equal pressure on both sides of ear drum
- Round window: absorbs pressure set up in the middle ear
- Oval window: separates middle ear from inner ear

Inner ear: Fluid-filled cavity

Subdivided into Bony labyrinth and membranous labyrinth

Bony labyrinth filled with a fluid called <u>perilymph</u>

- 3 Semi-circular canals with ampulla at the base of each
- Vestibule (Sacculus and utriculus)
- Cochlea

Membranous labyrinth filled with a fluid called <u>endolymph</u>

- Membranous sacs and tubes floating in the perilymph

Auditory nerve: carries nerve impulses from the ear to the cerebrum for interpretation

<u>Pathway of hearing</u>

Pinna → Tympanic membrane (vibrates) → Vibration of ossicles (Hammer, Anvil and Stapes) →Oval window (vibrates) → Cochlea (fluid stimulate organ of Corti) → Organ of Corti convert the stimulus into a nerve impulse→ Nerve impulse carried to the cerebrum by auditory nerve for interpretation

Pressure in the inner ear eased out through the round window into the Eustachian tube.

Balance restoration

Sudden change in speed and direction

↓

endolymph in semi-circular canals moves

↓

movement of endolymph stimulates cristae (receptors) within ampullae

↓

cristae convert the stimulus into nerve impulse

↓

Auditory nerve (via vestibular branch) transmits the impulse to cerebellum

↓

Cerebellum sends message to the muscles to restore balance

Sudden change in head direction

↓

gravitational pull stimulates maculae (receptors) within sacculus and utriculus

↓

Maculae convert the stimulus into nerve impulse

↓

Auditory nerve (via vestibular branch) transmits the impulse to cerebellum

↓

Cerebellum sends message to the muscles to restore balance

**Diseases and disorders of the ear**

Middle ear infections:

Cause: Viruses and bacteria that result in accumulation of fluids that causes pressure and pain

Treatment:

- Grommets – use air to clear the middle ear until infection is removed
- Medication
- Drainage of the middle ear

Deafness:

Cause:

- Damaged ear drum
- Hardened wax
- Injury to parts of the ear, nerves or brain
- Ageing

Treatment:

- Hearing aid with 3 parts
  - Speaker
  - Amplifier
  - Speaker
- Cochlear implants by operation in severe cases
  - Stimulate any functioning auditory nerve inside the cochlea with an electric field

**HUMAN NDOCRINE SYSTEM**

- It works with nervous system and its sense receptors to respond to changes in the environment
- It is made up of endocrine glands that secretes hormones directly into the blood stream

| Gland | Hormone(s) and Functions |
|---|---|
| 1. Hypothalamus | 1.1.ADH – maintains water balance |
| 2. Pituitary gland/Hypophysis | 2.1 Growth Hormone (GH)<br>- Controls growth of skeletal muscles<br>- Controls growth of the skeleton<br>2.2 Prolactin – produces milk<br>2.3 LH – stimulates ovulation<br>- Convert Graafian follicle into corpus luteum<br>2.4 FSH – stimulate production of an ovum in the Graafian follicle in the ovary<br>2.5 TSH – Controls the levels of thyroxin produced by the thyroid gland |

| | |
|---|---|
| 3. Thyroid | 3.1 Thyroxin – regulates metabolic rate e.g. cellular respiration<br>- Affects growth and functioning of the heart and nervous tissue |
| 4. Pancreas | 4.1 Insulin: Lowers blood sugar level<br>4.2 Glucagon: Increases blood glucose level |
| 5. Ovaries | 5.1 Oestrogen – prepares the uterus for implantation<br>- Development of secondary characteristics in females<br>5.2 Progesterone – maintains the endometrium during pregnancy |
| 6. Adrenal | 6.1 Adrenalin "flight or fight hormone" – prepares the body for emergencies<br>6.2 Aldosterone – maintains salt balance<br>- Works with ADH to maintain water balance |
| 7. Testes | 5.1 Testosterone – Development of secondary sexual characteristics in male |

**Examples of negative feedback**

TSH (Thyroid Stimulating Hormone) and Thyroxin

| **Thyroxin levels low** | **Thyroxin levels high** |
|---|---|
| Pituitary gland stimulated to secrete **more** TSH | Pituitary gland stimulated to secrete **less** TSH |
| Thyroid gland secretes **more** thyroxin | Thyroid gland secretes **little or no** thyroxin |
| Thyroxin levels **raised** to normal limits | Thyroxin levels **reduced** to normal limits |

Insulin and Glucagon

| **Blood sugar level low** | **Blood sugar level high** |
|---|---|
| Glucagon secreted by pancreas | Insulin secreted by pancreas |
| Glucagon stimulates conversion of glycogen to glucose | Insulin stimulates conversion of glucose into glycogen and fat |
| Glucose is released into the bloodstream | Glycogen and fat are stored in liver and muscle |
| Blood sugar level raised to normal limits | Blood sugar level reduced to normal limits |

Sometimes the body cannot reduce the blood glucose level to normal limits. This results in kidneys releasing the excess glucose. The excess glucose will be found in urine resulting in diabetes mellitus. There are two types of diabetes mellitus i.e. Type 1 and Type 2

| Type 1 diabetes mellitus | Type 2 diabetes mellitus |
|---|---|
| Pancreas cannot produce insulin | Pancreas produces little insulin |
| In young people (under the age of 30) | In old people (Over the age of 40) |
| People are born with the condition | People get it because of poor diet habits leading to obesity |
| Inject insulin to reduce the blood sugar level | Healthy diet and exercise reduce blood glucose level |

Symptoms of diabetes mellitus:

- Glucose in urine
- Frequent urination
- Extreme thirst
- Fatigue
- Vomiting
- Weight loss
- Blurred vision
- Non-healing wounds

Treatment of diabetes:

- Exercise
- Diet suitable for diabetic person
- Using prescribed medication for management of diabetes mellitus



Thermoregulation (Temperature regulation)

Normal body temperature of humans is 37°C. The body works via negative feedback to maintain that temperature. Skin plays a major role in the process

| Hot day | Cold day |
|---|---|
| - Increase in diameter of blood vessels (Vasodilation) | - Decrease in diameter of blood vessels (Vasoconstriction) |
| - More sweat | - Less or no sweat |
| - Hair follicles of the skin: horizontal (lie down) | - Hair follicles of the skin: vertical (upright) |
| | |

**RESPONDING TO THE ENVIRONMENT (PLANTS)**

**Plant hormones**

Hormone – chemical substance produced by organisms to control activities in a cell

3 plant hormones: Auxins, Gibberellins and Abscisic acid

Auxins

- Promote elongation
- Stimulate cell division
- Stimulate root development
- Bring about apical dominance
- Stimulate fruit development
- Keep the cell wall elastic
- Coordinates the response of plants to light and gravity

Gibberellins

- Stimulate germination of seeds
- Stimulate development of flowers
- Break dormancy in buds
- Delay ageing of leaves and fruits
- Increases fruit size
- Promote elongation of the internodes of stems

Abscisic acid

- Slow or stop plant growth
- Help plant to survive harsh weather conditions
- Stop seeds from germinating too soon
- Cause stomata to close (reduce water loss)
- Inhibits opening of buds

Tropism – plant response towards stimulus

| **Phototropism** | **Geotropism** |
|---|---|
| Plant response towards light | Plant response towards gravity/earth |
| Stems are positively phototropic | Stems are negatively geotropic |
| Roots are negatively phototropic | Roots are positively geotropic |

**Plant defence mechanisms**

2 types: Chemical and physical defence

Chemical defence

- Production of milky sap which cause inflammation e.g. Pencil tree (Mohloko)
- Production of thick, sticky, pale yellow substance called resin
  - Resin gums up insects mouth
- Production of less digestible chemical e.g. Sisal plant
- Production of less nutritious chemicals e.g. Aloe
- Hairs on plants release irritating chemicals e.g. Stinging needle plant (Mochachamogope)
- Production of toxic chemicals e.g. Night shade (Thola)

Physical defence

- Produce thorns on stems and leaves
  - Protect them against herbivores

**EVOLUTION**

- It is a process by which things that exist today arose from things that existed in the past

Biological evolution – changes that living organisms have undergone over long periods of time to give rise to present life forms e.g. long necks of giraffes. Biological evolutions means:

- All present life forms have **descended** from and are related to those that lived in the past
- All present-day life forms may look different from those that they descended from because they became modified from one generation to the another

**Theory of evolution**

Theory (everyday language) – thoughts, ideas, opinions not supported by **facts** e.g. what is your theory of learners performing poorly in mathematics during examination as compared to test, you will give your own opinion that does not have to be supported by facts

Theory (life science – biology) – an explanation that can be supported by facts, laws and tested hypotheses, for something that has been observed in nature

**Theory of evolution** – all things we see today arose from things that existed in the past but they look different because things change over long periods of time

**Evidence of evolution**

- Fossil record
- Comparative anatomy
- Comparative embryology
- Biogeography
- Molecular biology and genetics

Fossil record

Fossils are traces or remains of organisms that lived in the past

Study of fossils is called palaeontology

The history of life on earth is based on fossils and the rock layers in which the fossils are found.

Where are fossils found?

- Layers of sedimentary rocks
- Layers of ice

How is fossil age determined?

By using Relative and radiometric dating

Relative dating: age worked out in relation to another fossil or a geological event such volcanic eruption. It is not specific

Radiometric dating: attempts to find out how many years ago was the fossil formed. It is specific

NB: The top layer of a rock or ice is the most recent with bottom layer being the oldest during fossil dating (**Foundation of a house**)

Both relative and radiometric dating methods are used together to determine the age of a fossil.

Evidence from comparative anatomy/modification by descent

Modification by descent: the way inherited characteristics change over time in related organisms

Example 1

Wings of butterflies, bats and birds

- They appear to be similar with similar functions but they have evolved in different ways (from different ancestors): analogous structures

Analogous structures are evidence of convergent evolution which is when organisms which are not closely related evolve similar characteristics independently

Example 2

Basic forelimb structures of all vertebrates

They appear different with different functions however they evolved from common ancestor; all the forelimbs of the vertebrates have phalanges: homologous structures

> Homologous structures are evidence of divergent evolution which is when organisms that evolved from the same ancestor evolve different structures with different functions

Comparative embryology

Vertebrates' embryos look similar in early developmental stages. They have the following in common:

- Nerve cord which becomes a spinal cord
- Supporting rod called notochord that develops into a vertebral column
- Gill slits, a fish-like heart and kidney, and a tail

As the embryos develop, they lose some of these features to develop into vertebrates of their group

Evidence from biogeography

Biogeography: study of past and present distribution of individual species

Example: two islands with similar habitats in different parts of the world are populated by two different species

> Each species is similar to the one nearest to the mainland: Darwin explained that these similar structures have evolved from similar species on the nearest mainland

Evidence from molecular biology and genetics

It shows organisms that are related to each other not the organisms that evolved from each other. The evidence comes from

- Identical DNA structure
- Identical protein synthesis and similar proteins
- Similar sequences of genes
- Similar portions of DNA with no function
- Similar respiratory pathways

**Origins of ideas**

Lamarckism

- Idea that an organism can pass on characteristics that it acquired in its lifetime to its offspring
- The theory was named after Jean-Baptiste de Lamarck (1744-1829)
  He used two laws to describe his theory:
- Law of use and disuse
- Law of inheritance of modified/acquired characteristics

<u>Law of use and disuse</u>

- Organs became modified/adapted according frequency of use
- Frequently used organs became bigger, stronger or changed so that it can work better
- Disused organs became smaller until they totally disappeared

<u>Law of inheritance of modified/acquired characteristics</u>

- Modifications from Law of use and disuse will be transmitted to the offspring

Lamarck's explanation is rejected by most biologists. The biologists say that:

- Organisms evolved not because not because they wanted as Lamarck indicated, they evolved randomly in response to the environment
- Lamarck's explanation has very little evidence to support it.

Darwinism

- Named after Charles Darwin (1809-1882)
- His theory based on natural selection

Darwin based his theory on

- Variation: There is variation (difference) in appearance and abilities of organisms within every species
- Offspring: Species produce large number of offspring but they do not all survive
- Competition: Organisms compete for limited resources such as food
- Genetics: Organisms pass favourable characteristics to their offspring
- Natural selection: The organisms with useful (beneficial) characteristics are more likely to survive and reproduce

| Lamarckism | Darwinism |
|---|---|
| Variation in offspring caused by individuals in the population that wanted to change | Variation in offspring shown from moment of their production |
| Change: Individuals in the population that wanted to change | Change: Environmental factors acting randomly on the population. |
| Change: Individuals adapting to the environment | Change: nature choosing individuals best suited to survive to the environment |
| Individuals in a population change | Whole population changes |
| Changes inherited from parents to offspring | Characteristics passed from generation to generation |

Punctuated equilibrium vs Darwinism

| Darwinism (Grade 1, 2,3 etc.) | Punctuated equilibrium (Grade 1, 5, 9,11 etc.) |
|---|---|
| Evolution occur gradually over long periods of time | Evolution involves long periods of little or no change (equilibrium), they alternates with short rapid changes caused by natural selection |
| New species formed slowly over long period | New species formed quickly within short period compared to the long periods of little or no change |
| Supported by many transitional fossils which show gradual change over time | Supported by absence of transitional fossils indicating periods of artificial change |

**Speciation -** Occurs when a single species splits into two different species because of natural selection

- If a population of single species
- Becomes separated by a geographical barrier (sea, river or mountain)
- Then the population splits into two
- No gene flow occur between the two populations
- Because each population may be exposed to different environmental conditions
- Natural selection occurs independently in each of the two populations
- The individuals of the two populations become very different from each other
- Genotypically and phenotypically
- The populations will not be able to reproduce even if they were to mix again
- Because they have become different species

**Mechanisms of reproduction isolation**

- Prevents two species from producing viable, fertile offspring. This may occur in various ways:
- Breeding at different times of the year
- Chances of mating reduced
- Species-specific Courtship behaviour
- Results in courtship behaviour that do not attract individuals of other species even if they are closely related
- Adaptation to different pollinating agents
- Some closely related species of plants have different appearance that attracts different pollinating agents. This prevents cross-pollination between the different species
- Prevention of fertilisation
- Different copulatory organs in closely related species. The male organs do not fit into the female organs, the sperm cannot fertilise the female's egg.
- Infertile offspring
- Closely related species mate and produce infertile offspring. The offspring cannot produce more offspring of its kind therefore no new species is formed.

**Evolution in present times**

- It was sometimes thought that evolution took place long time ago and not taking place now
- It was also thought that evolution takes place only over thousands or millions of years
- There is proof of that evolution is still taking place even now
- The proof also shows that evolution can also take place within a short period of time.

<u>Development of resistance to insecticides in insects</u>

- Mosquitoes carry a parasitic protozoan called *Plasmodium* that causes malaria
- DDT was introduced to prevent malaria by spraying it in the walls of the house
- At first, the DDT was very effective in reducing malaria by killing the mosquitoes
- The mosquitoes eventually developed resistance to DDT and made it ineffective
- **The development of DDT resistant strain of mosquitoes can be explained as follows**:
- Mosquitoes produce large number of offspring with great variation
- At first most mosquitoes were not resistant to malaria, only a small number did
- DDT killed most of those mosquitoes that were not resistant to malaria
- The small number that was resistant to DDT survived due to natural selection
- The DDT-resistant mosquitoes which survived produced offspring that was also DDT-resistant
- The DDT-resistant mosquitoes increased in number in the next generation
- In that way, the mosquitoes made DDT ineffective by producing DDT-resistant mosquitoes over many generations
- That made the DDT ineffective in killing the mosquitoes

In similar way, each of the following can be explained through natural selection

- The variation in the bill and body size in the Galapagos finches
- Resistance to antibiotics in various bacteria e.g. in TB
- HIV resistance to anti-retrovirals

**Evidence of common ancestors in hominids including humans**

- Fossil evidence
- Cultural evidence: Tool making
- Genetic evidence

Fossil evidence

- This shows how characteristics of hominids changed over time
- The characteristics that changed are:
- Position of the foramen magnum
- Size of cranium
- Brow ridges
- Dentition and
- Palate shape

Cultural evidence: Tool making

- Studies show that other primates made tools and weapons just like the humans
- *Homo habilis* (Handy man) made the first tools about 2 million years ago
- The weapons were used for fishing, farming, hunting etc.

Genetics evidence: Mitochondrial DNA

- Humans are closely related to Chimpanzees than gorillas
- There is only 1,4% difference in the DNA nucleotide sequence of humans and chimpanzees
- Similarities in mutations present on mitochondrial DNA are also used to trace ancestors of all modern humans

Similarities between humans and the African apes

- Large brain compared to body mass
- Reduced sense of smell
- Parts of brain processing information from hands and eyes are enlarged
- Eyes in front
- Eyes with colour vision
- Arms rotate freely
- Long upper arms
- Elbow joints allow rotation of upper the forearm
- Flat nails instead of claws
- Opposable thumb that works in opposite direction to their fingers (money)
- Upright posture

| African Apes | Humans (*Homo sapiens*) |
|---|---|
| Small brain | Large brain |
| Brow ridges well developed | Brow ridges poorly developed |
| Less curved spine | More curved spine |
| Long, narrow pelvis | Short, wide pelvis |
| Large canines | Small canines |
| Long and rectangular mouth palate | Small and semi-circular mouth palate |
| Large jaws | Small jaws |
| Round chin (poorly developed) | Square chin (well developed) |
| More protruding jaws (more prognathous) | Less protruding jaws (less prognathous) |
| Cranial ridge across the top of the skull | No Cranial ridge |
| Foramen magnum in a backward position | Foramen magnum in a forward position |
| Brow ridge well developed | Brow ridge poorly developed |

Fossils related to human evolution

| Scientist | Fossil | Place | Year |
|---|---|---|---|
| Michael Brunet | Toumai (*Sahelanthropus tchadensis*) | Chad | 2001 |
| Donald Johanson, Yves Coppan and Tim White | Lucy (*Australopithecus afarensis*) | Ethiopia | 1974 |
| Raymond Dart | Taung baby (*Australopithecus africanus*) | South Africa | 1924 |
| Robert Broom | Mrs Ples (*Australopithecus africanus*) | South Africa | 1947 |
| Mary and Louis Leakey | Nutcracker man (*Paranthropus boisei*) | Tanzania | 1959 |
| Robert Broom | *Paranthropus robustus* | Kroomdraai, SA | 1938 |
| Mary Leakey | Laetoli footprints | Tanzania | |
| Stephen Motsumi and Nkwane Molefe | Little foot (Stw 573) (*Australopithecus africanus*) | Sterkfontein, SA | 1994 |
| Peter Nzube | Handy man (*Homo habilis*) | Olduvai Gorge, Tanzania | 1959 |
| Mathew Burger | Karabou (*Australopithecus sediba*) | Cradle of Humankind, SA | 2008 |
| Tim White | Ardipithecus ramidus | North-East Ethiopia | |
| Eugene Dubois | Homo erectus | Java in Indonesia and then Swartkrans | |
| Tim White | Homo sapiens | Ethiopia; Border Cave(KZN, Blombos Cave in W. Cape | |

The following information was observed in evolutionary trend of the fossils studied:

- Increase in brain size – processing more information
- Movement of foramen magnum to more forward position – Bipedalism
- Change from more prognathous to less prognathous face – smaller jaw
- Decrease in size of canines – cooked food

- Brow ridges became less developed – smaller jaws with little force

Advantages of Bipedalism

- Free hands to carry food, babies, tools etc.
- Clear view of surroundings for food and predators
- Efficient movement
- Faster cooling of the body in hot environments
- Display of the male sex organs as part of courtship display

**Out of africa hypothesis**

- Humans originated in Africa and then migrated out to the rest of the world
- Based on **fossil evidence** and **genetic evidence**

Fossil evidence

- Traced by the fossils found in South Africa and parts of Africa

Genetic evidence

- Traced with DNA from the Y-chromosome: Males
- Traced with DNA from the Mitochondrion (mtDNA): Females

**Alternatives to evolution**

Creationism

- All living organisms have been created by Supreme Being
- The organisms have not changed since their creation
- Different forms of organisms were created to function in particular setting

Intelligent Design (ID)

- Diversity of organisms on Earth came because each group of organisms were intelligently designed
- ID theorists prefer not to be called creationists because their evidence is purely scientific and not faith-based