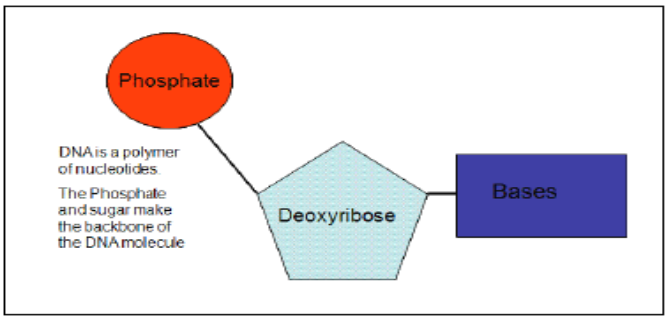
Week 3 Notes
NUCLEIC ACIDS

- Nucleic acids allow organisms to transfer genetic information from one generation to the next
- There are two types of nucleic acids

1. Deoxyribose Nucleic Acid (DNA)
2. Ribose Nucleic Acid (RNA)

- Both Nucleic acids are made up of monomers (or single units) called nucleotides.
- The nucleotide units themselves are made up of smaller types of components
- Each nucleotide contains a **phosphate**, a **sugar**, and a **nitrogen base**

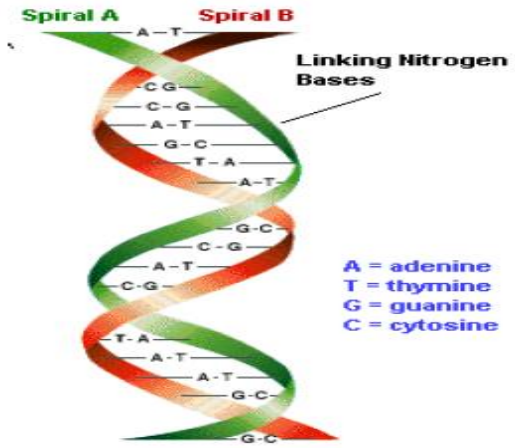
Structure of a Nucleotide



- Nucleotides are joined to form a long strand of nucleic acids
- When nucleotides are joined, the phosphate group of one nucleotide links is joined to the sugar of the next
- The phosphate – sugar link forms the backbone of the nucleic acids

1. DEOXYRIBOSE NUCLEIC ACID (DNA)

- DNA is made up of two strands of nucleic acids



- The sugar component of the nucleotide making DNA is known as deoxyribose sugar
- There are **four** different Nitrogen Bases that determines the nucleotide:

Adenine (A)
Guanine (G)
Cytosine (C)
Thymine (T)

- These nitrogen bases follows a base-pair rule where

  - ❖ Adenine always pair with Thymine **(A – T)**
  - ❖ Cytosine always pair wit Guanine **(C – G)**

- The two strands of DNA are held together by hydrogen bonds between adjacent complementary nucleotides
- The structure of DNA was not resolved until the early 1950's when James Watson and Francis Crick assembled all of the previously known information about DNA to construct a model of DNA
- The assembled strand of DNA takes on the structure of a double helix

2. RIBOSE NUCLEIC ACID (RNA)

- RNA is a single strand of nucleic acids
- The nucleotide making RNA is made up of the following components:

i) **Phosphate** group
ii) **Ribose sugar** (Note: the sugar in RNA is ribose sugar whereas the sugar in DNA is a deoxyribose sugar)
iii) **Nitrogen bases** (The four nitrogen bases of RNA are: Adenine (A), Guanine (G), Cytosine (C) and Uracil (U)

The complementary base pair rule in RNA is:

Adenine (A) pairs with Uracil (U)
Cytosine (C) pairs with Guanine (G)

- There are two major types of ribose nucleic acid:

i) **Messenger RNA (mRNA)**: function is to carry genetic information from the nucleus of the cell to the ribosome for protein synthesis

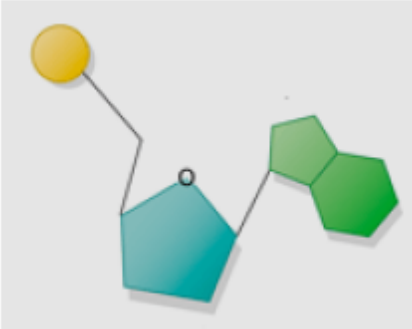
ii) **Transfer RNA (tRNA)**: role is to carry free amino acid to the ribosome to synthesise protein

Major differences between DNA and RNA

	DNA	RNA
1, Sugar	• deoxyribose sugar	• ribose sugar
2. Nitrogen Base	• has Thymine in stead of Uracil	• has Uracil instead of Thymine
3. Strand	• Double stranded	• Single strand

Self-check #3: Nucleic Acids

1. What does DNA stand for?
2. What are the monomers that make up DNA called?
3. Name the three main parts of the monomer identified in Question 2 – then label them on the picture



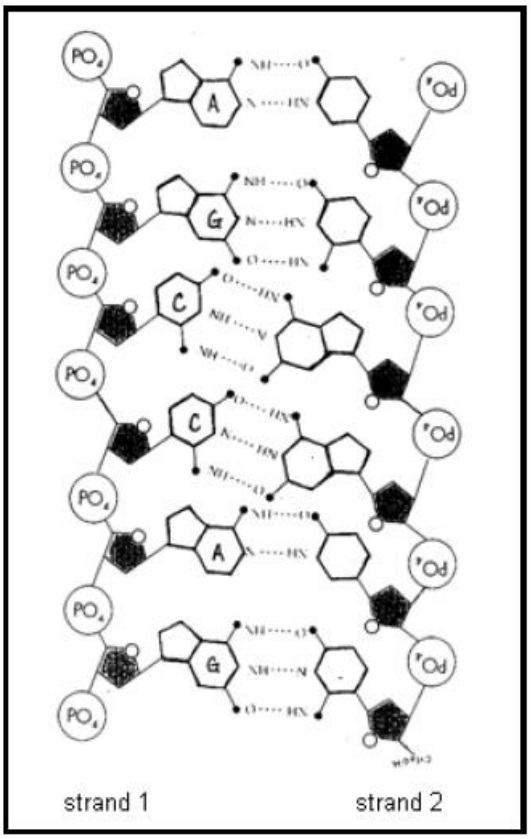
4. Name the four nitrogen bases that make up DNA molecules
5. Which nucleotides (A, C, T, G) base pair (match-up) with each other? What holds them together?
6. Name 2 differences between the structure of DNA molecule and RNA molecule
7. The following is a coding strand of DNA

5'-ATCGTGCGTGATATTTCCGAGT-3'

a) Write the complementary strand (template strand)
b) What is three-nitrogen base that code for a particular protein called?
c) How many triplets can be obtained from the DNA strand shown above?

8. Study the piece of DNA shown below and then answer the following questions

a) How many nucleotides are shown in the diagram?
b) Fill in the nitrogen bases of strand 2 using the complementary base-pair rule
c) Circle on the diagram a nucleotide
d) What are the backbones (sides) of the DNA ladder made of?
e) What are the "rungs" of the DNA made of?
f) Why is DNA referred to as the "blueprint" of life"?



9. What does RNA stand for?
10. What are the two types of RNA? Give their functions.

DNA REPLICATION

- DNA is able to copy (replicate) itself
- This is necessary because every cell must contain a complete set of all the genetic material needed for the development of any aspects of the organism
- It means that each cell must have a vast amount of coded message in the form of DNA, stored in the nucleus.
- This process of replication must take place before the chromosomes separate in cell division

Steps taken during the replication process

1. **Unwind**: the double helix form of the DNA unwind making the DNA looks like a ladder

2. **Unzip**: the bonds between the nucleotide on each strand break apart – rather like a zip being opened. The action takes place under the control of a special enzyme
3. **Copy**: as the strands separate, the bases are exposed. Nucleotides, which are present in large numbers in the surrounding nuclear material, are attracted to the matching bases on each strand

4. **Rewind**: As these nucleotides join they form a further matching strand. When the process is complete, two identical double strands of DNA have been produced. One of these can then move into each new cell when the chromosomes separate. This means that each new cell has a complete set of all the genetic material of the original cell

UNIT 2: GENETICS

Learning Outcome

At the end of this unit, you should be able to:

- Discuss Mendel's experiment
- Define and give examples of phenotype, genotype, heterozygous, homozygous, recessive, dominant, allele, gene
- Construct monohybrid crosses to show the expected phenotype and genotype ratios in the offspring using punnet squares
- Describe the roles of X and Y chromosome in sex determination
- Define sex-linkage and give examples (colour blindness, haemophilia)
- Define mutation and karyotype
- State why karyotype is increasingly used
- Show through the use of karyotype that mutation occurs
- Give examples of the contemporary application of genetics (e.g. breeding of plants and animals)

What is heredity?

-It is when certain traits are passed from parent to children.

-Traits are characteristics such as eye colour, height and athletic ability.

-Heredity is passed through **genes** in the DNA molecule.

-In **biology** the study of **heredity** is **called genetics.**

Gregor Mendel is considered the father of science of genetics.

-Through experimentation he found that certain traits were inherited following specific patterns.

-He studied inheritance by experimenting with peas in his garden. Peas work as an excellent test subject as they can self-pollinate, cross fertilize and have several traits that only have two forms. This enables Mendel to easily control his experiments and reduced the possibility of the outcomes to something he could record and manage.

Mendel's Experiment

-He studies seven traits of the pea plant: seed color, seed shape, flower position, flower color, pod shape, pod color and the stem length.

-There were three major steps to Mendel's experiments;

1. He produced a parent generation of true-breeding plants. He made these by self-fertilizing the plants until he knew they bred true to the seven traits. For example, the purple flowering plants always produced seeds that made purple flowers. He called these plants the P generation (for parent).

2. He produced a second generation of plants (F1) by breeding two different true-breeding P plants.

3. He then produced a third generation of plants (F2) by self-pollinating two F1 generation plants that had the same traits.

Mendel's Results

-Mendel found some incredible results from his experiments.

F1 generation

-Mendel found that F1 generation all produced the same trait. Even though the two parents had different traits, the offspring always had the same trait.

For example, if he bred a P plant with purple flower with a P plant with a white flower, all of the offspring (F1) plants would have purple flowers. This is because the purple flower is the **dominant trait**. These results can be shown in a diagram, called **punnet square**. The **dominate gene** is shown with a **capital letter** and the **recessive gene** is shown with **lower case letter**.

Here the purple is the dominant genes shown with a “P” and the white is the recessive gene shown with a “w”.

	P	P
w	Pw	Pw
w	Pw	Pw

F2 Generation

-Mendel found that 75% of the flowers were purple and 25% were white. Even though both parents had purple flowers, 25% of the offspring had white flowers. This turned out to be because of a recessive gene or trait was present in both parents.

-Here is the punnet square showing that 25% of the offspring has two “w” genes causing them to have white flowers.

	P	w
P	PP	Pw
w	Pw	ww

Homozygous and Heterozygous

-When two of the genes are the same (like “PP” or “ww”) they are called **homozygous.**

-When they are different (like “Pw”) they are called **heterozygous.**

GENETIC TERMS

1. **Gene**: the unit of inheritance
2. **Allele**: an alternative form of a gene
3. **Inheritance**: refers to the way in which particular characteristics are passed from parents to their offspring
4. **Homologous chromosomes**: chromosomes of the same shape and size
5. **Genotype**: the genetic make-up of an organism. Usually expressed by the genotype
6. **Phenotype**: the physical appearance of an organism as determined by the genotype
7. **Dominant**: the gene that is always expressed. Usually represented by a capital letter. E.g. D or T
8. **Recessive**: the gene that usually hidden and only expressed when the dominant gene is absent. It is usually represented by a small letter. E.g. d or t
9. **Homozygous**: when both alleles are the same E.g. DD or dd
10. **Heterozygous**: when both alleles are different E.g. Dd or Tt
11. **Punnet Square**: a diagram used to predict the outcome of a particular cross
12. **Purebreed**: offspring that are the result of mating between genetically similar kinds of parents. Purebred is the same as true breeding

MONOHYBRID CROSS

- A type of cross that involves the studying of the inheritance of only **ONE** characteristic
- A punnet square is often used to show this cross and to find the possible genotype of the offspring and in what proportion

For example: colour is a characteristic of the human eye. The allele coding for brown eyes is dominant to the allele coding for blue eyes. Using the letter B to represent the dominant brown eye allele, the recessive blue eye allele is represented by b

The three possible genotypes and phenotypes for eye colour are:

Genotype	BB	Bb	bb
Genotype description	Homozygous dominant	Heterozygous	Homozygous recessive
Phenotype (appearance)	Brown eye 	Brown eye 	Blue eye 

Joseph and Mary have three children. John has blue eyes and Mary is homozygous for brown eyes.

QUESTIONS

1) What are the **genotype** of Joseph and Mary?

   Joseph: _____

   Mary: _____

2) Draw the Punnet Square to show the **possible genotype** of the offspring of Joseph and Mary?

3) What **proportion** of their children will have brown eyes?

4) What is the **probability** of their next child having blue eyes?

ROLES OF X AND Y CHROMOSOMES IN SEX DETERMINATION

Chromosomes – carriers of the code

- All of the many millions of cells in our bodies must carry some sort of set of instructions which can tell the cell how to grow and how and when to carry out all its functions.
- In fact, every cell must carry the complete set of instructions for the development and control of the whole body
- The different cells must be able to select and use only the parts of the code that apply to them at any time
- In an human's embryo, some of the developing cells must grow into kidney cells and carry out the functions of the kidney, other cells will grow into brain cells and so on
- As well as all this, the set of instructions must be able to be passed on unchanged into new cells and even into new individuals when they reproduce.

The human genome is organized into 23 pairs of chromosomes (22 pairs of autosomes and one pair of sex chromosomes), with each parent contributing one chromosome per pair.

The **X** and **Y chromosomes**, also known as the **sex chromosomes**, determine the biological sex of an individual:

-females inherit an X chromosome from the father for a XX genotype.
- males inherit a Y chromosome from the father for a XY genotype (mothers only pass on X chromosomes).

-The presence or absence of the Y chromosome is critical because it contains the genes necessary to override the biological default - female development - and cause the development of the male reproductive system.

Note: Men determine the sex of their baby depending on whether their sperm is carrying an X or Y chromosome. An X chromosome combines with the mother's X chromosome to make a baby girl (XX) and Y chromosome will combine with the mother's to make a boy (XY)

Sex-Linkage

-Sex linkage or sex-linked inheritance is the phenomenon of phenotypic expression of an allele which is linked to a sex chromosome. The expression of an allele is dependent on the sex of an individual.

Examples:

i)Color Blindness: It is a sex-inked disorder where the person cannot distinguish between red and green colour. It is caused by a recessive gene located on X chromosome. It is mostly expressed in males, rarely seen in females. The genotype of an individual who is color blind and the carrier will be:
Carrier female: XcX
Colour blind male: XcY
Colorblind female: XcXc.

ii)Haemophilia: It is a hereditary disorder in which blood does not clot. It is X-linked recessive disorder. It is also called as bleeder's disease. The gene for haemophilia is located on a non-homologous region of the X chromosome but their alleles are absent in Y chromosome. The genotype of an individual who is color blind and the carrier will be:
Haemophiliac female: XhXh
Haemophiliac male: XhY
Carrier female: XhX.

Mutation

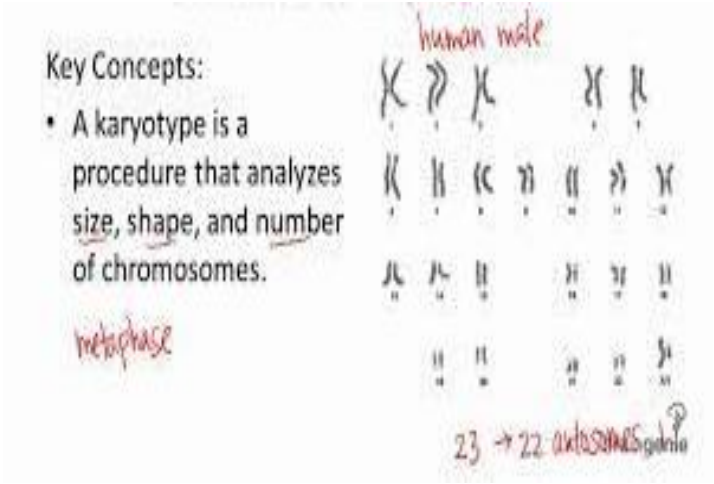
-It is a change in a DNA sequence. It can result from DNA copying mistakes made during cell division, exposure to ionizing radiation, exposure to chemicals called mutagens or infection by viruses.
-May **occur in somatic cells** (aren't passed to offspring)
-May **occur in gametes** and be passed to offspring

Gene Mutation

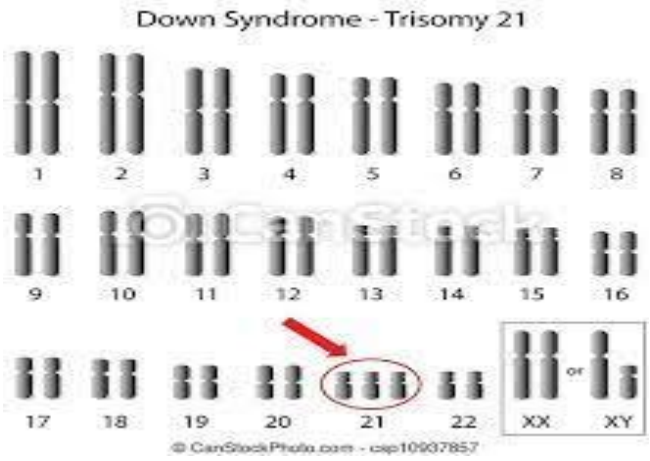
-A gene mutation is a change in one or more genes. Some mutations can lead to genetic disorders or illness.

Karyotype

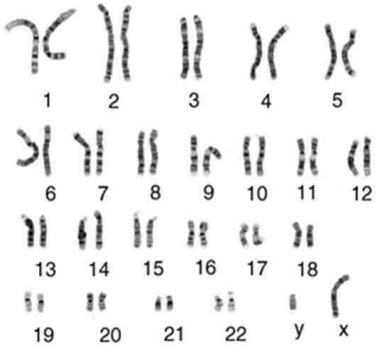
- Is an individual's collection of chromosomes. The karyotype is used to look for abnormal numbers or structures of chromosomes.
- Chromosomes usually extracted during **METAPHASE** of a cell



- The typical human karyotypes contain 22 pairs of autosomal chromosomes and one pair of sex chromosomes.
- Karyotypes can reveal changes in chromosome number, such as trisomy 21 (Down syndrome). Careful analysis of karyotypes can also reveal more structural changes, such as chromosomal deletions, duplications, translocations or inversions.



Mutations that produce changes in whole chromosomes are known as **chromosomal mutations**.



A karyotype is an organized profile of a person's chromosomes. In a karyotype, chromosomes are arranged and numbered by size, from largest to smallest. This helps scientists quickly identify chromosomal changes that may result in a genetic disorder.

Passing on the code

- We often hear such comments as – "doesn't she look like her mother' or 'he has got is father's eyes', or 'you can see that they are brothers'
- Although all living things differ, brothers and sisters tend to saw similar features
- These family similarities are inherited from their parents in **sexual reproduction**