GENETICS

Genetics is the scientific study of heredity and variation among organisms. It is the branch of science that accounts for the occurrence of similarities and differences among organisms of the same species; as well as explaining how traits are transmitted to off springs from their parents.

Inheritance refers to the process by which characters/traits are passed from parents to off springs.

The importance of genetics

- It is applied in genetic engineering to produce better breeds and varieties of plants and animals by altering their genetic constitution.
- It is important in courts of law to determine the paternity of the child.
- Genetics forms the basis of blood transfusion to determine compatible blood groups.
- Genetic counselling is important in preventing transmission of genetically determined diseases among married couples. This will help to relieve the families and community of the costs on treatment as well as the suffering of the sick and their families.
- It can be used in identification of criminals by use of finger prints and DNA profiling.
- It is used in molecular biology to manufacture artificial enzymes, hormones and vaccines by manipulating responsive genes from organisms.
- Forms the basis of cloning to increase the number of genetically important plants and animals.

Terms used in genetics.

1. **Chromosome**. These are thread-like structures bearing genes and located in the nucleus.
2. **Chromatid**. This is half of a chromosome split longitudinally.
3. **Bivalent**. This is a pair of homologous chromosomes.

4. **Gene**. This is a unit of the hereditable material found on the chromosome and responsible for controlling a particular trait/character.
5. **Allele**. This is the alternative form of the same gene. Most genes are made up of two alleles. Alleles of the same gene are represented by the same letter but the dominant allele is represented by a capital letter and the recessive allele by a small letter in the case of dominant-recessive characters
6. **Diploid**. This is a description of a cell, which has a whole set of chromosomes.
7. **Haploid**. This refers to a cell with half the set of chromosomes.
8. **Genotype**. This refers to the genetic composition of an organism.
9. **Phenotype**. This is the physical appearance or the outward expression of an individual.
10. **Dominant gene/dominant allele**. This is a description of a gene /allele whose effect is seen in the phenotype of the heterozygous individual. The effect of the dominant gene/allele is seen in the phenotype even in the presence of another gene/allele.
11. **Recessive**. This is a description of a gene whose effect is not phenotypically expressed in the heterozygous state. The effect of a recessive gene/allele is not seen in the presence of another (dominant) gene/allele.
12. **Homozygous**. This refers to a gene with two identical alleles for example if T represents the gene for height where tallness is dominant to shortness then the allele for tallness is T and that for shortness is t. an individual with TT is said to be homozygous tall and tt is said to be homozygous short.
13. **Homozygous dominant**. This is where both alleles of a gene determine a dominant character.
14. **Homozygous recessive**. This is where both alleles of a gene determine a recessive character.
15. **Heterozygous**. This refers to a gene with two different alleles for example if T represents the allele for tallness and t for shortness then Tt is the heterozygous state of this gene.
16. **Hybrid**. This is an offspring produced by parents of two different pure varieties.

17. **Pure breed**, is a breed which produces consistently same characteristics when bred with each other over generations.

18. **F1**, offspring produced by two pure breeds carrying contrasting characteristics.
19. **F2**, offsprings produced by selfing F1.

20. **Monohybrid inheritance**. This is a type of inheritance, which involves studying a single pair of contrasting characteristics.

21. **Dihybrid inheritance**. This is a type of inheritance, which involves studying two pairs of contrasting characteristics at ago

22. **Test cross ,** is a cross between an organism showing dominant character with that showing recessive character in order to establish whether the genotype of the organism showing the dominant character is homozygous or heterozygous

23. **Back cross**

This is the mating of an offspring with one of its parents.

MENDEL'S GENETIC EXPERIMENTS AND MONOHYBRID INHERITANCE

Monohybrid inheritance

Refers to the inheritance of a single pair of contrasting characteristics. Examples include, inheritance of height, blood groups, albinism, sickle cell anaemia, and sex linked characteristics etc.

This mechanism of inheritance was discovered by an Austrian monk and biologist Gregor Johann Mendel who carried out a number of genetic experiments using the garden pea plants *(Pisum sativum);* which he grew in the vegetable garden in his monastery. He later observed many sexually reproducing organisms and found out that they had variations among themselves despite being of the same species.

Why Mendel used garden peas

- They occurred in many varieties with distinct characters
- The plants were easy to cultivate
- All their offsprings were fertile
- They have a short life cycle that they reproduced so quickly
- The plants also had many contrasting characters with no intermediates
- Their reproductive structure were enclosed in petals which allowed for production of pure breeding plants due to self-pollination over many generations.

MENDEL'S EXPERIMENTS.

In one of his experiments, Mendel crossed tall pea plants with dwarf pea plants. In order to properly manage the cross, Mendel covered the stigma of all flowers of one group, and removed all the anthers from the flowers of another group of pea plants in order to prevent self-pollination, and transferred pollen using a brush. The resultant seeds were planted and he observed that all the F1off springs/**progenies** were tall.

He then selfed the F1 pea plants to get F2. This generation comprised of a mixture of tall and short pea plants in a ratio of 3 tall: 1short plants.

NB: The 3:1 ratio is known as Mendel's monohybrid ratio of the dominant and recessive characters respectively in the F2 generation.

Observation;

Mendel was able to observe that neither of the F1 nor F2 had intermediate phenotypes.

Conclusion;

He then concluded that inheritance is not the mixing/blending of features to produce intermediates but rather the process by which internal factors of the body may or may not express themselves in the phenotype.

From his conclusions, Mendel was able to formulate his first law of inheritance which is well known as the law of monohybrid inheritance/law of segregation/law of particulate inheritance

1st law, states that "The characteristics of an organism are controlled by internal factors which occur in pairs but only one can be carried in a single gamete".

Later with advancements in technology and microscopy, internal factors later came to be known as genes and Mendel's first law was modified. It can modernly be stated as follows. "***The characteristics of a diploid organism are controlled by alleles which occur in pairs but singly in gametes".***

A full genetic explanation of Mendel's first law and the 3:1 ratio

Let;

T represent the allele for tallness, t represents the allele for shortness

Parents : male x female

Phenotypes : Tall x short

Genotypes : TT x tt

Meiosis

Gametes : T T t t

Random fertilization :

F_1 genotypes : Tt Tt Tt Tt

Genotypic ratio: All Tt, Phenotypic ratio: All tall

To obtain F2 generation, F1 hybrids were selfed as shown below

Let;

T represent the allele for tallness, t represents the allele for shortness

Parents : male x female

Phenotypes : Tall x tall

Genotypes : Tt x Tt

Meiosis

Gametes : T, t, T, t

Random fertilization :

F_2 genotypes : TT, Tt, Tt, tt

Genotypic ratios: 1TT: 2Tt: 1tt; phenotypic ratios: 3tall: 1 Short

Note; for any genetic cross:

- Appropriate letters are 'let' to represent respective alleles involved
- Capital letters should be used to represent dominant alleles and small letters to represent recessive alleles.
- A cross**(X)** must be indicated to symbolize mating between the parents
- Directive words must be indicated to define each step of the cross

Mendel went on and carried out crosses on different contrasting characteristics of garden peas.

Table showing summary of results obtained by Mendel after investigating inheritance (monohybrid using various characteristics in pea plant (Garden Pea)

Contrasting characteristic of pea plant	**F1 generation phenotype**	**F2 phenotypic ratio**
Smooth seeded crossed(x) with wrinkled seeded plants	All smooth	3 smooth :1 wrinkled
Green pod X yellow poded plants	All Green	3 Green:1 yellow
Axil placed flowered X Terminal placed flowered plants	All Axil	3 Axil:1 Terminal flowered

Exercise

1. In a garden pea plant there are two forms of heights. When a pure breeding tall pea plant was crossed with a short pea plant all the offsprings obtained where tall when the offsprings were selfed a phenotype ration was obtained in F2.

a. Using suitable genetic symbols, workout the genotypes and phenotypes of the F2 generation

b. What are the phenotypic and genotypic ratios of the F2 generation

c. Suppose 700 pea plants where produced in the F2 generation

i. How many were tall?

ii. How many were short?

EXAMPLES OF MONOHYBRID INHERITANCE IN MAN

There are many genetically determined abnormalities and diseases that affect man (and other animals). Since these are genetic diseases, they can only be inherited from parents and their occurrence is determined by those genes inherited from parents during fertilization

Examples of such diseases include:

- ***Albinism***

NB: Research has showed that most of, though not all the genetic abnormalities are caused by recessive genes (alleles) and the genes responsible for normal conditions are dominant. This implies that for an individual to suffer from such diseases, they must have two copies of the responsive genes (homozygous recessive). The heterozygotes and the homozygous dominant individuals are normal. Though the former are phenotypically normal but their cells contain a copy of the recessive allele and are described as carriers.

INHERITANCE OF ALBINISM

Albinism is a recessive character which results into failure of formation of body pigments.

Albinos have the following characteristics as a result;

- Light-colored skin
- White hair
- Pink eyes

Example.

Man with normal skin marries an albino woman. Determine the genotypic and phenotypic ratio of their F1 offsprings.

Let **A** represent the allele for normal skin.

a represent the allele for albino color

Parents man x female

Phenotype normal x albino

Genotypes AA X aa

Gametes (A) (A) (a) (a)

Fertilization

	(A)	(A)
(a)	Aa	Aa
(a)	Aa	Aa

Genotypes -All Aa.

Phenotype - all normal but carriers.

Exercise.

A carrier man for albinism marries a carrier woman. Work out the F2 genotypic and phenotypic ratios.

EXCEPTIONS TO MENDELIAN INHERITANCE

The following do not conform to the process of inheritance as illustrated by Mendel.

1. Co-dominance
2. Incomplete dominance.
3. linkages
4. Multiple alleles.

CO-DOMINANCE

This is a condition where genes determining a particular character all show up in the phenotype. It mainly occurs in animals.

OR. It is where in the heterozygous state neither allele is completely dominant over the other i.e. the 2 alleles are co-dominant. This results in appearance of both the alleles in the phenotype.

Example.

Sickle cell Trait.

It is due to a mutation of a gene. A person suffering from sickle cell anaemia has a defective type of haemoglobin. ***It is caused by a recessive gene***

When the concentration of oxygen is low in blood, the red blood cells assume the shape of a sickle. Because of this, the red blood cells cannot absorb oxygen properly.

This is hereditary disease can be passed on to the children by the parents in their gametes. Sickle cell anaemia has a fatal effect on people who are homozygous for this mutated gene.

People who are heterozygous i.e. ***they have mutated and non-mutated genes, have normal red blood cells and abnormal red blood cells.***

Note. ***The gene for normal haemoglobin and abnormal haemoglobin are Co-dominant.***

In co-dominance, we represent alleles with different but all **capital letters** ***because none of the alleles is dominant over the other.***

Example:

A normal man married a woman with sickle cell anaemia. Determine the phenotype and genotype of the children.

Let **A** represent the allele for normal RBC

Let **S** represent the allele for sickle cell

Parents	Normal man	x	female carrier.
Genotypes	AA	x	SS
Meiosis	(A) (A)	x	(S) (S)

Fertilization

	A	A
S	AS	AS
S	AS	AS

Genotype of F1 offsprings - AS, AS, AS, AS.

Genotypic ratio - All AS

Phenotypes all normal but Carriers.

Note. *People with genotype* **AA** *have normal haemoglobin.*

People with **AS** *have both normal and abnormal haemoglobin but normal most, so are carriers and are said to have sickle cell trait.*

People with genotype **SS** *have sickle cell anemia.*

Example 2.

During the inheritance of fur/coat color in short-horned cattle, when red and white cattle are mated, the F1 hybrid has white fur thickly interspersed with red fur. This phenotype is referred to as roan.

So, Let ***R*** represent allele for red fur and ***W*** represent allele for white fur.

Parents Bull x cow

Phenotype Red x white

Genotypes RR x WW

Gametes (R) (R) (W) (W)

Fertilisation

	(R)	(R)
(W)	RW	RW
(W)	RW	RW

F1 genotype, all RW.

Phenotype, all roan.

F2

Parents Bull x Cow

Phenotypes roan x roan

Genotypes RW x RW

Gametes (R) (W) (R) (W)

Fertilisation

	(R)	(W)
(R)	RR	RW
(W)	RW	WW

F2 genotypic ratio 1 RR: 2 RW: 1 WW . Phenotypic ratio 1 red: 2 roan: 1 white.

Other examples of co-dominance include blood groups ABO.

Allele **A** and **B** are co-dominant, that is why we have blood group **AB,** however **O** is recessive for both **A** and **B.**

INCOMPLETE DOMINANCE

This is a condition in the heterozygous where neither of the alleles is dominant over the other and the phenotype of the offspring is an intermediate between that of the parents. ***It mainly occurs in plants.***

E.g. in plants, when a red flowered plant is crossed with a white flowered plant, the offspring produced pink and white flowers in a ratio of 1:2:1 respectively.

Example 1.

Consider petal colour in flowers.

Let the allele for red flowers be R.

Let the allele for white flowers be W

Phenotype phenotype Red petal x white petal

Genotypes RR x WW

Gametes (R) (R) (W) (W)

Fertilisation

	(R)	(R)
(W)	RW	RW
(W)	RW	RW

F1 genotypes, all RW. Phenotypes, all pink.

F2 (selfing F1)

Phenotypes pink x pink

Genotypes RW x RW

Gametes R W R W

Fertilisation

	R	W
R	RR	RW
W	RW	WW

F2 genotypic ratio 1 RR: 2 RW: 1 WW

Phenotypic ratio 1 red: 2 pink: 1 white.

MULTIPLE ALLELES

This is where one character is determined by more than two alleles. This implies that a single gene contains more than two alleles. An example is blood group inheritance.

Inheritance of blood groups

The gene controlling blood groups is made up of three different alleles (multiple alleles). These alleles are A, B and O. The inheritance of blood groups is also an example of co-dominance.

There are 4 blood groups that is group A, B, AB and O. the alleles for blood groups are represented as I^A, I^B and I^O.

I^O is recessive to I^A and I^B.

I^A and I^B are co-dominant. An individual inherits two of these alleles one from each parent.

The table below shows the possible blood groups that can arise from the different genotypes.

Genotype (alleles)	Blood group (phenotype)
I^AI^O	A
I^AI^A	A
I^BI^O	B
I^BI^B	B
I^AI^B	AB
I^OI^O	O

Example:

A man heterozygous for blood group A marries a woman who is heterozygous for blood group B. Work out the genotypes of possible offsprings.

Solution

Let;

A, B and o represent the alleles for blood groups A, B and O respectively

Parents	:	Father	x	Mother
Phenotypes	:	Blood group A	x	Blood group B
Genotypes	:	Ao	x	Bo
Meiosis				
Gametes		A, o		B, o
Random fertilization	:			
Offspring genotypes	:	AB, Ao, Bo, oo		

Offspring phenotypes: Blood groups AB, A, B and O

Exercise

Work out the possible blood groups of the offsprings produced if a man of blood group A marries a woman of blood group AB

SEX DETERMINATION

In man, there are 23 pairs of chromosomes; of these only one pair carries genes for sex determination. These are called sex chromosomes (heterosomes) designated X and Y, and the other 22 pairs are called autosomes. A genotype XX is described as homogametic and is female while XY is described as heterogametic and is a male. During meiosis, the two sex chromosome segregate such that each ovum carries one X chromosome, half of the sperms carry an X chromosome and the other half carry a Y chromosome.

If a sperm carrying an X chromosome fuses with the ovum, the zygote is female and if the sperm is carrying a Y chromosome, the zygote is male. Sex is therefore determined by the sex chromosome carried in the sperm as a matter of chance.

Example.

Parents : Father x Mother

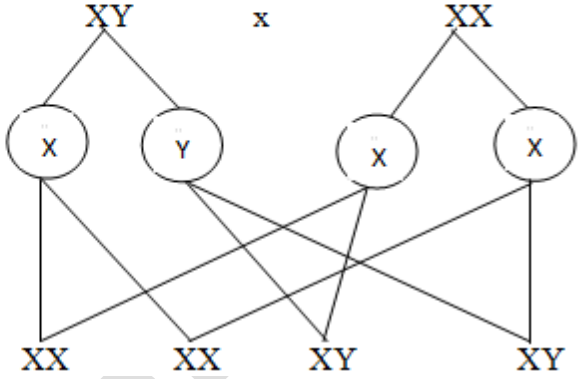
Phenotypes : Male x Female

Genotypes : XY x XX

Meiosis

Gametes

Random fertilization :



F_2 genotypes : XX XX XY XY

Genotypic ratios: 1XX: 1XY

Phenotypic ratios: 1female: 1male

This shows that there is a 50% chance of any child being a male or female.

SEX LINKAGE.

In humans, there are several thousands of characteristics each genetically controlled. With only 23 pairs of chromosomes, each chromosome must therefore carry many genes; a phenomenon that does not exclude sex chromosomes. These in addition to genes responsible for sex differences may carry genes determining some other features in the body.

Sex-linked genes are genes carried on sex chromosomes and inherited together with those determining sex. Sex linked traits (characters) are traits determined by genes carried on sex chromosomes and inherited together with those determining sex.

Note: The Y chromosomes don't carry genes, sex linked genes are specifically carried on the X sex chromosomes but not on the Y chromosome.

Examples of sex linked characters in man include the following

- Haemophilia
- Colour blindness

Most of these characters are caused by recessive alleles and in a genetic cross, these must be represented as <u>superscripts</u> on the X sex chromosome.

HAEMOPHILIA (BLEEDERS' DISEASE)

Haemophilia is a recessive sex-linked blood disorder that leads to absence of one or more blood clotting factors, leading to prolonged bleeding even from minor cuts.

Just like other sex-linked traits, haemophilia is carried on the X chromosome and the responsive allele is recessive to the normal allele. The condition interferes with formation of blood clotting factors; whose absence greatly delays the blood clotting process. This results into prolonged bleeding and excess blood loss even from minor cuts which may lead to death.

The allele being recessive, haemophiliac females must inherit two copies of the defective allele while males inherit one copy. The heterozygous females show normal blood clotting and are described as carriers.

This is because the other X chromosome carries a dominant allele needed for normal blood clotting which suppresses the recessive allele for haemophilia. The males lack the alternative allele and the recessive allele is automatically expressed phenotypically.

Example: When a carrier woman is married to a normal man

Let;

H represent the allele for normal blood clotting, h represents the allele for haemophilia

Parents : Female x Male

Phenotypes : Normal x Normal

Genotypes : X^HX^h x X^HY

Meiosis

Gametes : X^H X^h X^H Y

Random fertilization :

F_1 genotypes : X^HX^H X^HY X^HX^h X^hY

It can be noted that there is a 50% chance of a daughter being a carrier and a 50% chance of a son being haemophiliac. Sons can only inherit haemophilia (and other sex linked traits) from their mothers but not fathers as they only inherit the father's Y chromosome and not the X chromosome that carries sex linked genes. Girls can inherit from both parents.

Today, people with hemophilia are treated as needed with intravenous injections of the missing protein.

COLOUR BLINDNESS

It is a recessive sex linked character that leads to inability of the individual to distinguish between colours.

It is caused by a recessive allele, carried on the X chromosome and inherited in the same way as haemophilia. Colour vision is due to presence in the retina of red, blue and green cones needed for seeing the respective colours. The recessive alleles result into absence of some of these cones which renders inability to identify such colours from other related colours. This is called colour blindness; the commonest being red-green colour blindness where individuals lack red and green cones in their eyes.

EXERCISE

Green colour blindness is sex linked in man. A normal man married a colour blind woman. Using suitable genetic symbols workout the genotypes and phenotypes of their children.

PRACTICAL APPLICATION OF PRINCIPLES OF HEREDITY IN PLANT AND ANIMAL BREEDING.

- **Hybridization**

It is the process of producing an offspring by crossing two organisms (parents) of different varieties, species, or genera. The resulting offspring is called **Hybrid**. Hybridization is useful in Agriculture to produce hybrid crops and some hybrid Animals of better qualities.

The parents involved are usually carrying specific dominant genes e.g. In maize crossing, two parents one having a characteristic of producing large sized maize cobs and other that grow faster produce a Hybrid Offspring with both characteristics.

In livestock, Hybridization is useful in production of:

-Goats with high milk yields, Fast growing chicken, Cattle with high milk yields.

IMPORTANCE OF HYBIRDIZATION

- It leads to production of climate/weather tolerating varieties of crops and animals.
- It leads to development of high yielding varieties of crops and livestock hence an increased Agricultural output.
- It leads to production of diseases/pest resistant varieties of crops and animals.
- Produce faster growing varieties of crops and livestock that mature in a short period of time.

MUTATION

This is a sudden change in genetic makeup of an organism.

Types of mutation

1. Chromosome mutation: this is a sudden change in the structure of a chromosome.
2. Gene mutation, this may involve deletion, duplication of a gene at a locus.

Causes of mutations.

1. High temperatures.
2. Chemicals such as mustard gas, Colchicines etc.
3. High-energy particles such as alpha and beta particles.
4. High-energy radiations such as x-rays, gamma rays and ultra violet radiations.

Note*; most mutations are disadvantageous and recessive. They are rare but persistent in the population.*

- *Substances that cause mutation are also called mutagens.*

VARIATION.

It refers to the differences in characteristics between organisms of the same species.

TYPES OF VARIATION

1. Environmental (non-genetical / non-inheritable).

Is variation due to some environmental factors / food, water / changing the phenotypes of an organism. Most of these occur in somatic cells (but not germinal cells and hence they are not inherited and not a value in evolution).

2. Genetic (inheritable) variations.

Is a variation due to changes occurring in the genotype of an organism. Such variations are inherited and result in variation of new species.

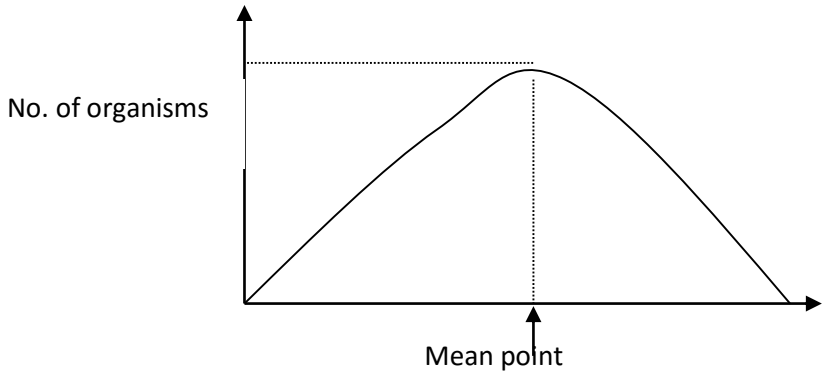
Types of genetic variation

a. ***-continuous variation***
b. ***-discontinuous / discreet variation***

A. ***Continuous variation****:*

Is when there is no clear cut difference in characteristics between organisms of the same species. The characteristics vary marginally between two extreme ends and most organisms are in the middle.

When plotted, such characteristics give a smooth bell shaped curve known as the normal distribution / Gaussian curve.



Examples of variations in mass / linear dimensions / shapes, colour, intelligence / ear lobe length, yields of milk, number of grains in the maize cob etc.

Such characteristics are controlled by the combined effect by a number of genes called the polygenes (i.e. polygenic character) and environmental factors. These characteristics are measurable i.e. (quantitative).

b. ***Discontinuous / discreet variations***

Is when characteristics within a population vary between individuals by clear cut differences (with no intermediate?)

Plotting such characters **don't** give a normal distribution curve since they don't show a smooth gradation between the two extremes.

Examples:- sex phenotype, blood groups, wing length, tongue rolling, eye colour in drosophila, thumb prints albinism sickle cell, haemophilia, polydactyl (extra finger inheritance).

These characteristics are controlled by one /two major genes which may have two or more allelic forms in a population.

CAUSES OF GENETIC VARIATION

- Crossing over of homologous chromatids during prophase 1 of meiosis which produce gene recombination in gametes and therefore offsprings.
- Random assortment of chromosomes during anaphase 1 of meiosis, this leads to varied offsprings.
- Mutation, the sudden change in the genetic composition of an organism also bring about variation.

Role of mutation in the evolution of new species

Mutation produces genetic variations which can be passed onto successive generations.

The advantageous ones enable organisms to adapt to their environment. Isolated groups of a species in different environments adapt differently and eventually form new species e.g. resistance of malarial parasites to anti- malarial drugs.

When drugs are administered few individuals of mutant strain that are resistant survive. These soon reproduce and become the majority of the population.

Congratulations, you have come to the end of genetics!