GENETIC DISEASES.

These are inheritable complications suffered by particular individuals caused by defective genes.They include,

- Haemophilia.
- Red-green colour blindness.
- Sickle cell anaemia.
- Cystic fibrosis.
- Huntington's chorea.
- Phenylketonuria.

THE SICKLE CELL ANAEMIA.

Is a genetic disease which affects mainly the African, Americans, the people of Mediterranean countries and northern Africa.

CAUSES OF SICKLE CELL ANAEMIA (HOW SICKLE CELL ANAEMIA ARISE)

Sickle cell anaemia is caused by base substitution gene mutation. It occurs on one of the triplet nucleotide bases i.e Cytocine, thymine, cytocine in a DNA located on the 11th pair of chromosomes that carry genetic information for the production of beta polypeptide chain, determining formation of haemoglobin molecule.

During this base substitution gene mutation, the nucleotide base, Adenine replaces thymine in the DNA nucleotide triplet code i.e cytocine, thymine, cytocine (CTC), forming a wrong DNA triplet nucleotide base sequence i.e Cytocine, Adenine, Cytocine (CAC). During transcription, messenger RNA copies complementary DNA nucleotide base sequence which will include the codon Guanine, Uracil, Guanine (GUG) as a result of the base substitution, instead of the Guanine, Adenine, Guanine (GAG). This causes a mistake to occur on the 6th aminoacid in the beta polypeptide which is 146 amino acids long, where the amino acid valine replaces the aminoacid glutamic acid. The presence of valine instead of glutamic acid in the beta polypeptide leads to production of abnormal haemoglobin called haemoglobin S. This is because the messenger RNA codon GAG codes for aminoacid glutamic acid while GUG codes for amino acid valine.

Glutamic acid carries a negative charge and is polar where as valine is non-polar and hydrophobic. So, abnormal haemoglobin S is much less soluble than normal haemoglobin and it begins to crystallize when the oxygen concentration falls as it does in the capillaries of body tissues, resulting into the red blood cells normally biconcave disc shaped to assume the shape of a crescent or become sickle shaped, this reduces their surface area for absorption of oxygen and there fore, With their abnormal haemoglobins, the sickle shaped red blood cells are far less efficient at carrying oxygen in blood.

NOTE: Haemoglobins are found in red blood cells and are made of four polypeptide chains, two α-chains (Alpha chains) which is 141 aminoacids long and two B-chains (beta polypeptide chains) which is 146 amino acids long.. This mistake occurs at the 6th amino acid in the beta polypeptide chain where the amino acid glutamic acid for normal haemoglobin (HbA) is replaced by amino acid valine for the abnormal haemoglobin (HbS) called haemoglobin S.

SYMPTOMS/CONSEQUENCES/EFFECTS OF SICKLE CELL ANAEMIA DISEASE.

- Anaemia, low Red blood cells count.
- Oxygen deficiency.
- Poor blood circulation.
- Enlargement of the spleen.

(i) Anaemia and oxygen deficiency.

Anaemia and tendency of the red blood cells to change shape from Biconcave to sickle shape. Anaemia arises because the sickle shaped red blood cells are constantly destroyed in the spleen. The efficiency of the red blood cells to transport oxygen is reduced, oxygen deficiency occurs in the body. Deficiency of oxygen results into,

- Infections and frequent illness.
- Body weakness and fatique.
- Poor physical development.
- Dilation of the heart causing heart failure.

(ii) Poor blood circulation.

Poor blood circulation. This is because the sickle shaped red blood cells get jammed in the blood capillaries and small arteries. This will cause the following effects.

- Heart damage resulting into heart failures.
- Damage of the lungs causing pneumonia.
- Muscle and joint damage causing rheumatism and pain.
- Gut damage causing abdominal pain.
- Kidney damage causing kidney failure.
- Liver damage.

(iii) Enlargement of the spleen.

Enlargement of the spleen. This is because sickle celled red blood cells collect in the spleen for destruction so increase in the activities of the spleen leads to its enlargement.

OCCURRENCE AND DISTRIBUTION OF THE SICKLE CELL ANAEMIA IN A POPULATION.

The genes controlling sickle cell anaemia show codominance in some cases but sometimes described as recessive autosomal genes showing complete dominance. Homozygous recessive individuals possess both recessive allele(HbsHbs) and suffer from sickle cell anaemia and may die at an early age.

It shows codominance because heterozygous individuals possess both the dominant allele (HbA) and the recessive allele (Hbs) with a genotype (HbAHbs). The red blood cells of this individual contain about half of normal haemoglobin and about half of the abnormal haemoglobin S. In this case The Alleles HbA and Hbs are codominant but it shows complete dominance in some other cases this is because the heterozygous individuals (Hb^AHb^S) are described as showing sickle cell traits and the individuals do not suffer from the conditions of the sickle cell anaemia.

Heterozygous individuals are only affected at unusually very low oxygen concentrations like climbing at high altitudes. So some the haemoglobin can crystallize causing some few red blood cells to attain sickle shapes.

The sickle cell condition shows some advantages. It is widely distributed in a population and remain persistent. The frequency of the recessive alleles for abnormal haemoglobin S is high in malarial infected areas. This is because individuals carrying the recessive alleles do not suffer from malaria (are less susceptible to malarial infections) since the plasmodium parasites do not multiply in the red blood cells containing abnormal haemoglobins S, sickle celled red blood cells have reduced surface area and cannot absorb sufficient oxygen. Lack of enough oxygen in the red blood cells prevents adequate aerobic respirations to occur and many physiological processes can not take place in the plasmodium and may be destroyed inside the sickle shaped red blood cells, so its life cycle is not completed and many other red blood cells can not be infected. So, heterozygous individuals have a selective advantage over non-carriers and are more likely to survive and continue to pass the recessive alleles for abnormal haemoglobin in the next generations. The final frequency of the genes in the population is determined by the levels of malarial infections in the population. This is an example of balanced polymorphism.

CYSTIC FIBROSIS.

Cystic fibrosis is a recessive gene mutation very common in Europe. It is caused by deletion of three bases in agene(DNA) located on chromosome 7. The genes affected codes for normal formation of chloride channels in the plasma membrane, they are specific proteins on the plasma membrane known as Cystic fibrosis transmembrane regulator (CFTR). It allows diffusion of chloride ions into and out of epithelial cells.

In persons suffering from cystic fibrosis, the cystic fibrosis transmembrane regulators do not function and causing the defects of failure of chloride ions transport across the cell surface membrane of epithelial cells. The outward flow of chloride ions from the epithelial cells are prevented. Chloride ions are negatively charged, so in order to balance the negative charges which build up inside the cells more sodium ions enter into the epithelial cells, the high concentration of sodium ions in the cells in turn prevent water from leaving the cell, thus mucus lining the cell surfaces become thick and sticky. This affects mostly the lungs, pancreas and the liver where unusually thick mucus clogs lungs, liver and pancreas.

In the pancreas fibrous patches called cysts develop and in the lungs, when the thick mucus dries up, it causes blockage of air ways of the lungs, and branches of pancreatic ducts and the bile duct from liver into the gut. Repeated lung infections and digestive problems. Male and female infertility occurs, the individual secrete more saltier sweat.

Note: Cyst is a growth or swellings containing liquids that form on the body of a person.

HUNTINGTON'S CHOREA.

It is a genetic disease caused by an autosomal mutation which is dominant. The dominant mutant allele determines production of an enzyme known as huntingtin. The symptoms of the disease include,

- Deterioration of brain cells. This results into loss of intellectual ability.
- Involuntary muscular movements or loss of control of voluntary muscles by motor neurones. This results into uncontrollable shaking and dance (chorea) like movements.
- Hallucinations, mood and personality changes.

Genetic counseling and gene cloning techniques can be applied to diagnose the disease and prevent frequency of its occurrences.

Dominant gene mutations are rare but the few known include Huntington's chorea. The heterozygotes are normal and can survive but it is lethal in the homozygous state.

PHENYLKETONURIA.

This is a genetic disease caused due to inability to convert the amino acid phenylalanine to another amino acid tyrosine, caused by a recessive autosomal condition. The new recessive allele does not code for the production of the enzyme that catalyses conversion of phenylalanine to tyrosine, phenylalanine accumulates in the body, the excess is converted to toxins. This will cause, severe mental retardation in infants, low life expectancy, Awkward posture during walking , dry, rough skin (eczyma).

VARIATION

Variation is described as difference in characteristics that exist between organisms of the same species living within the same natural population. Two forms of variations occur. These are :-

(i) Discontinuous variation
(ii) continuous variation

DISCONTINOUS VARIATION

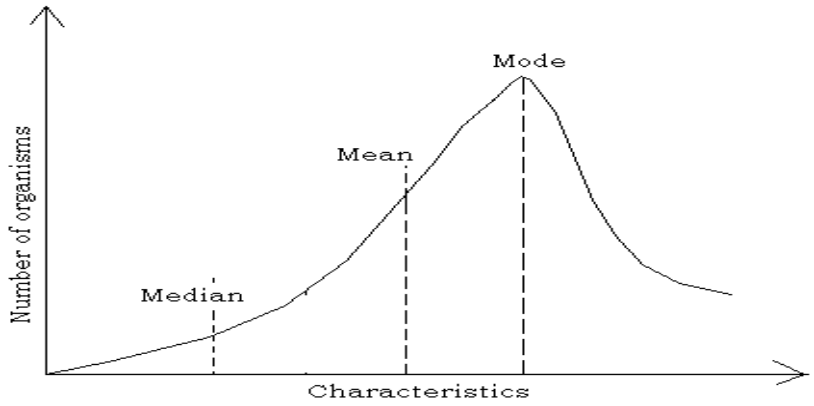
Is where certain characteristics in organisms are controlled by different alleles of a single gene. They show no intermediate characteristics within the population. Their phenotypic expressions are not affected by environmental factors.

Examples of characteristic which show discontinuous variations include: blood group in human, wing length in drosophila, melanic and light forms of the peppered moth (Biston betularia), and sex in animals and plants.

A histogram or a graph is used to represent difference frequencies Distribution of the variations in the characteristics within a population. In case of discontinuous variation the histogram represents or shows a skewed distribution in which the mean, mode and media all have different values.

Below is a histogram showing discontinuous variation in frequent distribution.

SKEWED DISTRIBUTION CURVE.



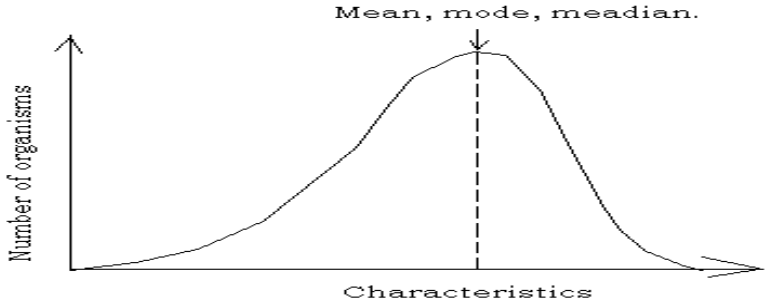
CONTINOUS VARIATION

Is where characteristics in organisms is controlled by many genes (polygenes). They show intermediate characteristics within the population. The phenotypic expressions of such characteristics are greatly influenced by environmental factors.

Examples of the characteristics showing continuous variations include; height, weight (mass), shape and sometimes colour of organisms.

If a frequency distribution of continuous variation is plotted, a bell shaped graph called a normal distribution curve or Gaussi curve is obtained.In normal distribution curve, the means, mode and media have the same value.A histogram showing frequency distribution of continuous variation.

NORMAL DISTRIBUTION CURVE.



DIFFERENCES BETWEEN DISCONTINOUS AND CONTINOUS VARIATION.

DISCONTINOUS VARIATION	***CONTINOUS VARIATION.***
(i) Phenotype differences are distinct and separate.	(i) Phenotype differences are very slight.
(ii) The population shows no intermediate characteristics. They have distinct forms.	(ii) The population show intermediate characteristics.
(iii) The differences that exist in their characteristics are qualitative and can not be measured.	(iii) The differences in their characteristics are quantitative and can be measured.

(iv) Controlled by a single gene. (v) The phenotypes are not affected or influenced by the environmental conditions. (vi) Their frequency show skewed distribution curve.	(iv) Controlled by combined effect of many genes (polygenes). (v) The phenotypes can be influenced by the environmental factors. (vi) Their frequency show normal distribution curve.

SOURCES OF VARIATION

These include the following

(i). Gene reshuffling

This is the process of recombination of genes/alleles which occurs in sexual reproduction causing genetic variation between offsprings and their parents. The reshuffling of genes result into genetic variation and they occur in the following.

• A reciprocal crossing-over of genes between chromatids of homologous chromosomes during prophase I of meiosis. This separates linked genes and form combination of alleles and new linkage groups and is a source of genetic recombination of alleles.

• The random and independent assortment of homologous chromosome during metaphase I of meiosis. This determines the directions in which the pairs of chromosomes move during anaphase I of meiosis and subsequently gives rise to a large number of different chromosome combinations in gametes and form new combinations of alleles.

• A complete random fusion of male and female gametes containing complementary sets of haploid chromosomes producing a diploid zygote.

Gene reshuffling is the main basis of continuous variation however , it does not generate a major change in genotype and neither does it effectively and does not result into formation of new species.

(ii) Mutation

Is the change in the chemical structure or the amount of DNA in the chromosome of an organism. Only those mutations which occur during the formations of the gametes can be inherited, these mutations produce sudden and distinct difference in characteristics between individuals of the same species and therefore basis of continuous variations.

Gene mutation may lead to several alleles occupying a specific gene locus. This uses both the heterozygosity and size of the gene pool of the population and leads to increase in variation within the population.

NOTE:

Mutations which occur during meiotic cell division (gamete formation) also results into gene – reshuffling.

Gene reshuffling merely shuffles the genes to form new combinations of alleles, but the mutation is the ultimate source of the inherited sources of variation.

Mutations occur due to, errors in DNA replications, errors in cell division, or induced by the environmental factors.

(iii) Environmental effects

Organisms of identical genotypes when subjected to different environmental condition, the individuals tend to show differences in phenotypes. Implying that the final phenotypic expression (physical appearance) of an organism is the result of its genotype and the effect of the environment upon it.

Temperature, light intensity, e.t.c are largely responsible for continuous variation within a population.

JOHANSEN'S EXPERIMENT

Johanssen demonstrated the effect of the interactions of genotypes and environment on phenotypes.

In a series of experiments on the mass of dwarf bean seeds, he selected the heaviest and lightest seeds from each generation of self-pollinating dwarf bean plants and used these to produce the next generations. After repeating these experiments for several years, e found only small differences in the mean mass of seeds from the same selected line that is heavy or light but large differences in the mean mass of seeds from different selected lines that is heavy or light. This suggested that both heredity and environment were influencing the phenotypic appearance of the characteristics.

In the development of human characteristics such as personality, temperament and intelligence, there is evidence to suggest that both nature (hereditary factor) and nurture (environmental factors)interact to influence the final appearance of the characteristics.

Identical twins can also be helpful to demonstrate the effects of both genotypes and environment on the continuous variation. The very close resemblance between identical twins is genetical while any slight difference in their characteristics is environmental.