VARIATION

Definition: Variation refers to the differences in the characteristics shown by organisms of the same species.

Organisms belonging to the same natural population **(same species)** would be expected to exhibit general similarity in their structural, physiological and biochemical characteristics but this is never the case; differences always occur even in identical twins and these are collectively referred to as **variation**.

TYPES OF VARIATION

There are two types/forms of variation, namely

- Continuous variation and Discontinuous variation

CONTINUOUS VARIATION

This is the occurrence of minor differences among organisms of the same species with no clear cut, hence producing intermediates.

Continuously varying characteristics are measurable and can be described in numerical forms; they can therefore be quantified and are described as quantitative characteristics/variations. They produced by a combined effect of very many genes at different loci. These genes are called polygenes and the system of inheritance is called polygenic inheritance.

Examples include height, weight, intelligence, size etc.

Polygenic inheritance

This refers to the inheritance of a single characteristic produced by a cumulative effect of several genes at different loci.

Most of the organisms' characteristics are actually produced by such genes; individually each of these genes may have little effect on the phenotype but their combined effect is significant. These genes always change positions due to reciprocal crossing over, producing an almost infinite variety of phenotypes which can greatly be modified by environmental factors. This forms the basis for continuous variation.

Given their quantitative (measurable) nature, the results of a large population represented on a frequency distribution curve usually produce a normal distribution curve as shown below.

Example

Mass/kg	50-52	52-54	54-56	56-58	58-60	60-62	62-64	64-66	66-68	68-70	70-72
Frequency	4	7	11	16	24	29	26	16	8	4	2

1. Represent the data on a histogram and frequency curve
2. Superimpose the frequency curve onto the histogram

The normal distribution curve shows a continuous gradation from one extreme to the other without any break. Most organisms show phenotypes close to the median, the number reduces towards the lower and upper extremes. E.g. Most human beings show intermediate heigh with only a few individuals being very tall or very short.

DISCONTINUOUS VARIATION

This is the occurrence of differences among organisms of the same species with clear cuts hence producing no intermediates.

Such characteristics are non-measurable and cannot be quantified and are therefore described as qualitative characters. They are controlled by one or two or only few genes usually with two allelic forms and are relatively unaffected by environmental factors.

Examples include blood groups in man, sex in plants and animals, wing length in drosophila, eye colour in drosophila, light and melanic forms of peppered moths etc.

Given their discontinuous variability, such characteristics can only be represented in form of bar graphs or even pie charts but not frequency curves.

SOURCES OF VARIATION

The ultimate (fundamental) factor that determines the phenotype is the genotype. However, the subsequent degree of expression is greatly influenced by environmental factors during growth and development of the organism.

Consider Mendel's tall pea plants which normally grow to a height of 6 ft, these can only do so if provided with adequate light, water and nutrients. Limited supply in any of these would inevitably prevent attainment of full height. Sources of variation are therefore both **environmental and genetic.**

Environmental sources of variation

This is when organisms of the same species show differences in their characteristics due to differences in their environment. Many of the differences in the phenotypic characteristics are as a result of exposure to different environmental conditions. These include temperature, light intensity, food supply, water supply social environment etc.

Genetic sources of variation

This is when organisms of the same species show differences in characteristics due to differences in their genetic makeup (Constitution)

Asexually reproducing organisms tend to have the same genetic composition but may show significant phenotypic differences which are as a result of environmental differences.

For sexually reproducing organisms, vast genetic changes occur at each generation during meiosis by **crossing over and independent assortment**, while others result from **random fusion** of gametes.

In his experiments, the Danish geneticist Johanssen demonstrated the influence of genetic and environmental influences on the phenotype of organisms. He made a series of experiments on the mean mass of seeds from a group of dwarf pea plants. He selected the heaviest and lightest seeds of each generation of self-pollinating and used these to produce the next generations. After several years, he observed that the mean mass of heavy seeds increased gradually while that of light seeds decreased progressively. The mean mass difference between the two lineages increased while the mass difference between any two seeds from the same lineages decreased progressively.

From these results, continuous variation can be defined as the cumulative effect of varying environmental factors acting on a variable genotype. The results also indicated that the extent of expression of a characteristic is primarily determined by genotype.

In this experiment, the plants must be self pollinating to conserve the original genotypes over the period of study.

Though both environment (nurture) and heredity (nature) interact to determine the final appearance of the character, environment can never increase the extent of a characteristic beyond that determined by the genotype.

Causes of genetic variation

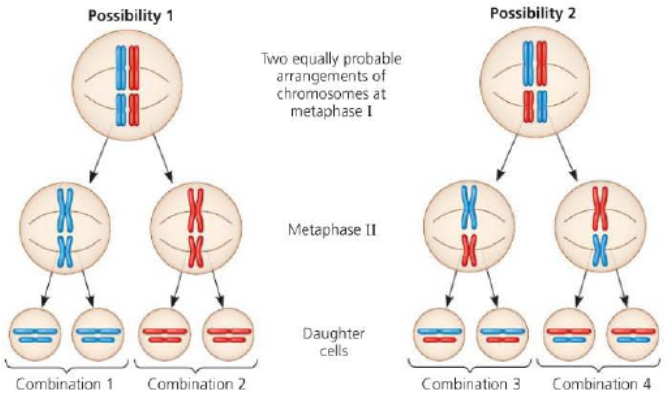
Crossing over

This refers to the reciprocal exchange of the genetic material between non-sister chromatids at the chiasmata during prophase I of meiosis. Crossing over leads to formation of new linkage groups called recombinant chromosomes ***(carry genes from two non-homologous chromosomes)*** which are genetically different from the original chromosomes. These pass into gametes which fuse to form a zygote that is genetically different from either parent.

Independent/random assortment of chromosomes

The random alignment of homologous pairs of chromosomes at the spindle equator during metaphase I and subsequent segregation (separation) anaphase I of meiosis

During metaphase I the orientation of chromosomes from one bivalent is totally independent of all other bivalents and determines which chromosomes pass into the same daughter cells. Subsequent segregation (separation) of homologous chromosomes at anaphase I produce a large variety of chromosome combinations, leading to genetic variation in the daughter cells. The orientation of chromatids at metaphase II is also random and determines which chromatids pass into the same daughter cells at anaphase II. Subsequent segregation (separation) produces more chromosomal combinations in the resultant gametes leading to more genetic variation.



Random fusion of gametes (fertilisation)

During fertilisation, the fusion of male and female gametes is random, with any male gamete potentially capable of fusing with any of the female gametes. This results into a combination of complementary haploid sets of maternal and paternal chromosomes leading to more genetic variation in the zygote.

The three sources of genetic variation above simply alter the arrangement of existing genes/alleles in DNA leading to a **change in combination of traits** in the next generation. Such sources are described as gene reshuffling (re-arrangement) and are responsible for the continuous type of variation in sexually reproducing organisms.

These changes however don't result into new alleles and the resultant gene combinations can easily be undone/reversed in subsequent generations before allowing for significant evolutionary change. They therefore don't generate major genetic changes which may be necessary for evolutionary change. Evolutionary significant changes are produced by mutation; the ultimate source if inheritable genetic changes (variations).

MUTATIONS

A mutation is a sudden/spontaneous change in the structure or/and amount of DNA of an organism. The information in mutated DNA can be expressed phenotypically by transcription into mRNA for translation into a protein leading to change in the characteristics of an organism. For multicellular organisms, the mutations affecting body cells (somatic mutations) can be passed to cells produced by mitosis of the mutant cell but not inherited to the next generation and most of these go unnoticed. It is only mutations affecting gamete producing cells and gametes that can be inherited into the next generations.

During mutation, some genetic material may be lost, doubled, inverted, translocated (moved), and mixed, resulting into mutants having different genetic constitution from the non-mutants. The mutants formed transmit these mutated genes to their offsprings through random fertilisation which makes the offsprings become different from the non-mutants.

CAUSES OF MUTATIONS

Mutations can happen spontaneously (randomly/by chance) during DNA replication or as errors during cell division. They can however be induced and accelerated by various environmental factors; which are called mutagens. Examples of mutagenic factor include:

- Chemicals like colchicines, formaldehyde and some food preservatives
- High energy particles like alpha and beater particles
- High energy radiations like X-rays, gamma radiations, ultra violet and cosmic radiations
- Viral infections like hapes
- Some pesticide like DDT
- Mustard gas

CHARACTERISTICS OF MUTATIONS

Some feature have been found to apply almost universally to all forms of mutations, these characteristic features include;

- Mutations are spontaneous; they can occur suddenly at any time.
- Mutations are recessive; the mutant alleles are recessive to the normal forms of the genes
- They are usually disadvantageous; most mutations interfere with normal body function hence exerting negative effects onto the organism. Some mutations are lethal and lead to death of the organisms. Some few mutations can be advantageous though less frequently.
- They are permanent/persistent. A mutation once occurred may stay in the population for very many generations without change.
- Mutations are rare. Experiments suggest that an average of about one base-pair changes spontaneously per 10^6 base-pair replications. If proteins are, on average, encoded by about 10^3 base-pairs then it would take about $l0^6$ cell generations before the protein contained a mutation

TYPES OF MUTATIONS

- Gene mutations
- Chromosome mutations

GENE MUTATIONS (Point mutations)

This refers to change in the base sequence of the DNA portion equivalent to one gene (cistron). Such mutations occur at a single locus of the chromosome and cause no structural change on DNA; they are therefore referred to as point mutations.

Types of gene/point mutations

- Deletion; this is where a nucleotide is completely lost from DNA
- Substitution; the replacement of one nucleotide with a different one
- Insertion (addition); the addition of one nucleotide into the DNA strand
- Duplication; this is where a nucleotide is repeated in a cistron
- Inversion; the reversal of two or more nucleotides within a cistron

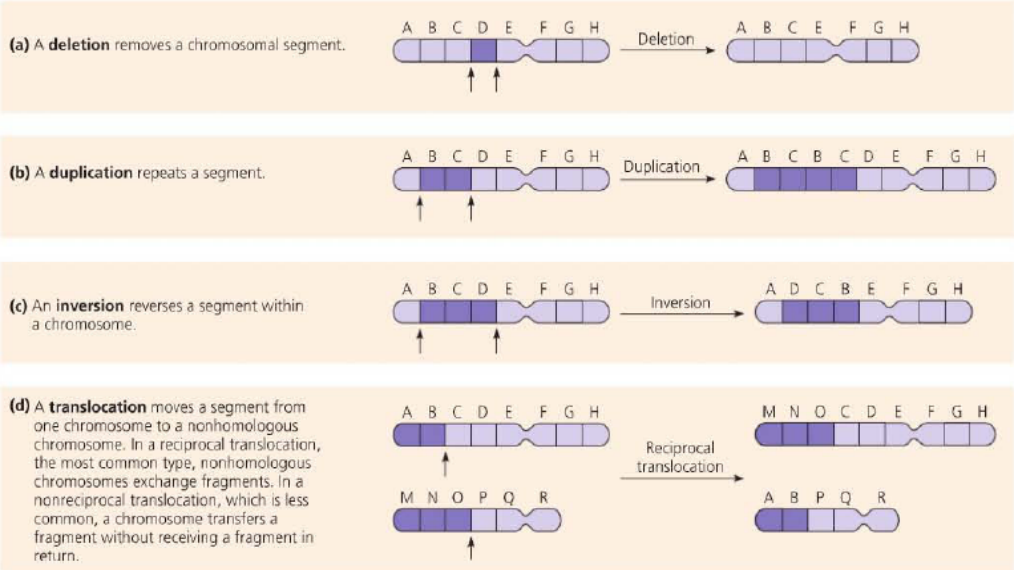
Chromosome mutations (chromosome abberations)

These involve change in number or structure of chromosomes

Structural chromosomal mutations

- Deletion; this involves total loss of a portion(s) from a chromosome. Such a mutation will lead to loss of several genes and shortening of chromosomes. It is the simplest form of chromosomal mutation that makes the chromosomes short of some genes. If such a mutation affects one of the homologous chromosomes, the alleles on the other chromosome will be expressed even if recessive. In case the same locus of both homologous chromosomes is affected, such mutations are usually lethal as the organism can no longer synthesise some proteins that may be vary paramount for survival.
- Substitution; involves replacement of chromosome portions with different ones
- Insertion; the addition of portions into the chromosomal strand Though this does not change the genotype, the phenotype changes due to change in the DNA base sequence. This implies that the order of gene loci is important; a phenomenon known as the position effect.
- Duplication; is where the gene sequence of a chromosomal portion is repeated
- Inversion; a portion of a chromosome breaks and joins the same chromosome after rotation through 180^0
- Translocation; a chromosomal portion breaks and joins either the other end of the same chromosome or another non homologous chromosome.

NB; Crossing over involves reciprocal exchange of the genetic material between homologous chromosomes. This simply changes the allele sequence of chromosomes but no gene loci are lost; similarly does inversion and translocation

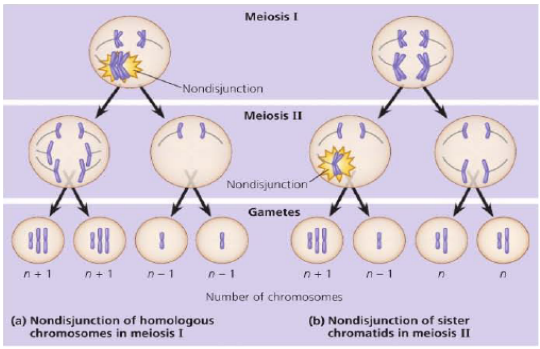


Change in number of chromosomes

These usually occur as a result of errors during meiosis and may involve change in number of complete sets or simply number of chromosomes; the two cases are termed as **polyploidy and aneuploidy** respectively

ANEUPLOIDY

This is when a cell/organism has one or more chromosomes more or less than the normal diploid number. This is as a result of failure of homologous chromosomes or sister chromatids to separate during anaphase I and II of meiosis respectively (Non disjunction).



Non disjunction is the failure of one or more pairs of homologous chromosomes or chromatids to separate at anaphase of meiosis, such that the daughter cells get less or more chromosomes than the normal haploid number.

The other chromosomes not affected by non-disjunction are usually distributed normally by meiosis into gametes formed.

Fusion of such gametes with the normal haploid gametes leads to different forms of polysomy (possession of excess chromosomes) in the zygotes as discussed below.

Polysomy	Description/cause	General representation
Monosomy	A gamete that is lacking one chromosome fuses with a normal haploid gamete	2n-1; The chromosome is represented singly in the zygote
Trisomy	A gamete with one extra chromosome fuses with a normal haploid gamete	2n+1; The chromosome is represented in triplicate in the zygote
Tetrasomy	A gamete with two extra chromosomes fuses with a normal haploid gamete	2n +2; Two chromosomes each represented in triplicate, making a total of 4 copies
Etc	Etc	etc

After fertilization, mitosis will subsequently transmit the anomaly to all embryonic cells. If the organism survives, it usually has a set of traits caused by the abnormal number of the genes associated with the extra or missing chromosome. Zygotes missing some chromosomes have much lower chances of surviving.

Non disjunction can also occur during mitosis. If such an error takes place early in embryonic development, then the aneuploid condition is passed along by mitosis to a large number of cells and is likely to have a substantial effect on the organism.

Down syndrome is an example of trisomy in humans, this and heterosomal non disjunction are discussed later in the chapter.

POLYPOIDY (EUPLOIDY)

This is when a cell/organism has more than two complete sets of chromosomes

Examples include triploidy (3n), tetraploidy (4n) etc

In plants, triploid cells are formed by fusion of a diploid egg cell with a haploid male nucleus forming a triploid endospermic nucleus; this degenerates in some seeds but persists as the endosperm in endospermic seeds. Tetraploidy may be a result of successful DNA replication but chromosomes fail to separate normally at anaphase; the nucleus therefore does not divide. This can be induced artificially by treating cells in early prophase with colchicine; a chemical that inhibits spindle fibre formation in living cells.

NB: A modified form of polyploidy occurs in some animal cells where chromosomes replicate and separate within an intact nucleus without cell division. This is called endomitosis and occurs in liver cells (4n), salivary glands of drosophila.

Polyploidy is more common in plants than animals, in fact most of the known angiosperm species are polyploidy. The lower occurrence in animals is due to the fact that the increased number of chromosomes makes the process of gamete formation by meiosis more prone to errors which

makes most polyploidy animals infertile. These die minus passing the condition to next generations

Plants on the other hand can reproduce vegetatively without use of gametes. The condition is associated with several advantages which include the following

- Hardness to harsh conditions
- Resistance to diseases
- Faster growth
- Increased fruit yield
- Increased fruit size etc;

Such offsprings are much better adapted to the environment, this is known as hybrid vigour. However, better adapted polyploids may fail to interbreed with diploid organisms, thereby forming a new species due to possession of extra sets of chromosomes beyond the diploid number. This is termed as **interspecific hybridisation.**

In fact most of the existing angiosperms are polyploids due to the above advantages, for example bananas are 3n and wheat is 6n.

There are two types of polyploidy, depending on the origin of the chromosome sets

AUTOPOLYPLOIDY

This is when all the extra complete sets of chromosomes are derived from the same species. Plant cells may undergo replication without division to produce 4n cells, the amount of cytoplasm increases to balance with that of the nucleus leading to a general increase in the size of plant organs.

Colchicine and related chemicals are used in plant breeding to induce autopolyploidy; as in tobacco and tomatoes. Autoploids are always fertile as diploids if they have an even number of chromosomes to allow for complete pairing of homologous chromosomes during meiosis. In fact polyploids have fewer abnormalities as compared to anueploids.

ALLOPLOIDY

This is the presence of complete sets of chromosomes from different species.

Such organisms are usually sterile unless the total number of chromosomes is an exact multiple of the original haploid number of chromosomes. E.g. 2(n1+n2), 3(n1+n2) etc as this will allow for complete chromosomal pairing during meiosis

Allopolyploidy does not occur in animals because there are fewer instances of cross breeding between species. Polyploidy does not add new genes to the gene pool but simply produces new gene combinations.

IMPLICATIONS OF MUTATIONS

The most immediate effect of mutation is genetic variation, which leads variation in phenotypes.

Mutations are almost always known to be disadvantageous because of their harmful effects on

mutants, some of which are even lethal. Although the frequency of aneuploid zygotes may be quite high in human, most of these chromosomal alterations are so disastrous to development that the embryos are spontaneously aborted long before birth. Some types of aneuploidy appear to upset the genetic; some individuals can survive but carry a set of traits (syndrome) characteristic of that type of chromosomal aberration

Down's syndrome (mongolism)

It is a congenital genetic disorder due to inheritance of three copies of chromosome 21. It is a trisomic form of polysomy due to non disjunction of an autosomal chromosome 21 (also called trisomy 21), or translocation of this chromosome usually onto chromosome 15 making this chromosome abnormally longer.

Mongolism usually leads to miscarriage in mothers and the chances of it to occur increases with age of the females; mothers are therefore advised to bear their children earlier in life. This form of syndrome results in individuals having the following characteristics;

- ❖ They are mentally retarded
- ❖ They have a short life span due to their low resistance to infections
- ❖ Frequent saliva flow from the mouth
- ❖ They have a short stature (natural standing height)
- ❖ They have slit eyed appearance.

HETEROSOMAL NON-DISFUNCTION

These are non-disjunctions of sex chromosomes. As a result some gametes have an extra sex chromosome or no sex chromosome at all. Fusion of such a gamete with a normal haploid gamete produces the following forms of aneuploidy.

Heterosomal non disjunctions tend to have less profound effects than autosomal ones. This is because the Y chromosomes carry very few genes and the extra X chromosomes in all somatic cells are inactivated into Barr bodies-whose genes are not transcribed. In fact for a normal somatic cell, only one X chromosome remains active and the other is condensed onto a very small inactive rod called the Barr body.

Klinefelter's syndrome (XXY)

This occurs when an ovum with two x chromosomes fuses with a normal haploid sperm or when a sperm with two sex chromosomes fuses with a normal haploid ovum.

The individuals are male with poorly developed secondary characteristics but tend to exhibit a number of feminine traits which include the following;

- ➢ Poorly developed testes with very small penises

- They have sparse pubic hair and little facial hair
- High pitched voices
- Some breast development occurs and wide hips
- They are infertile due to impaired spermatogenesis
- They have very subnormal intelligence
- They tend to be taller than the average height

Triple X syndrome (XXX)

Also called trisomy X and results from fusion of a normal haploid sperm carrying an X chromosome fusing with an ovum with two X chromosomes.

It occurs in females who are fertile, mentally and physically normal as the diploid females only that they tend to have a high sex libido.

XYY syndrome

They are males produced when the Y chromosomes fail to separate after replication leading to sperms with two Y chromosomes. These individuals don't exhibit any well developed syndrome but may have the following characteristics;

- They are usually very aggressive and therefore common in prisons and security forces
- They tend to be giants and taller than abnormal heights
- They are fertile and mentally normal

Turner's syndrome (XO)

It is a form of monosomy resulting from fusion of a normal sperm carrying an X chromosome with an ovum lacking a sex chromosome. It is the only viable form of monosomy known to occur in human beings and is also called monosomy X.

The individuals are females who lack most of the secondary female characteristics as follows

- They have poorly developed gonads and are infertile
- Do not show secondary sex characteristics like enlarged breast and menstrual cycles
- They are shorter than normal height
- They also have a skin fold around their neck

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