CHAPTER 12.1 – TYPES AND FACTORS OF VARIATION

Definition of variation

- Variation refers to the differences in characteristics found within the same population or species
- No two organisms are the same although they are from the same species including identical twins
- There will always be differences between them
- Normally, variation refers to physical characteristics observed in phenotypic differences caused by changes in structure, physiology and biochemistry
- Variation enables us to identify individuals within a population

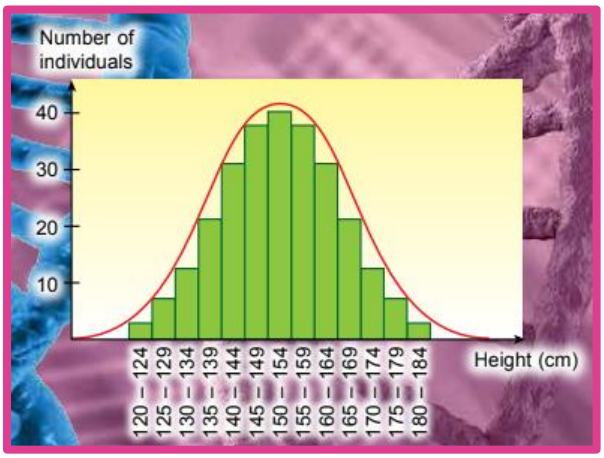
The necessity of variation for the survival of species

- Enables natural environment to continue selecting beneficial characteristics and remove the non-suitable ones
- Ensures survival of the species when the environment changes
- Allows cross breeding among species to form new species

Types of variation

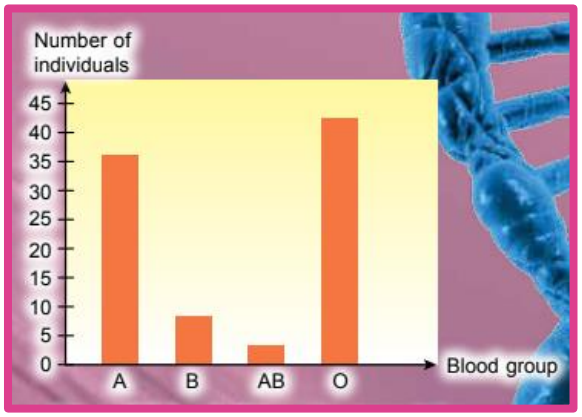
CONTINUOUS VARIATION

- Continuous variation is the variation in which the differences in the characteristic is not distinct
- Individuals show gradual differences in characteristic from one extreme to the other extreme
- A spectrum of phenotype is observed
- If data is obtained and plotted on a graph, a normal distribution or a bell-shaped curve will be obtained
- Most members of the population have intermediate phenotypes, which are characteristics found in between the two extremes
- Continuous variation is quantitative, it can be measured and graded from one extreme to the other extreme
- The characteristics are influenced by environmental factors
- Examples of characteristics which show continuous variation are height, body weight and skin colour



DISCONTINUOUS VARIATION

- ✓ Discontinuous variation shows distinct differences in characteristic
- ✓ If data is obtained and plotted on a graph, a discrete distribution or a bar chart with separate bars are obtained
- ✓ There are no intermediate characteristics
- ✓ The characteristic is qualitative, it cannot be measured or graded because the characteristic can only be determined by genetic factor
- ✓ The characteristic is not influenced by environmental factors
- ✓ Occurrence of discontinuous variation is due to genetic factors, therefore it can be inherited
- ✓ A characteristic is determined by a single gene with two or three alleles
- ✓ Therefore, the characteristic is easily seen
- ✓ Examples of discontinuous variations are the ability to roll tongue, eye colour and fingerprint pattern



Comparison between continuous and discontinuous variation

SIMILARITY

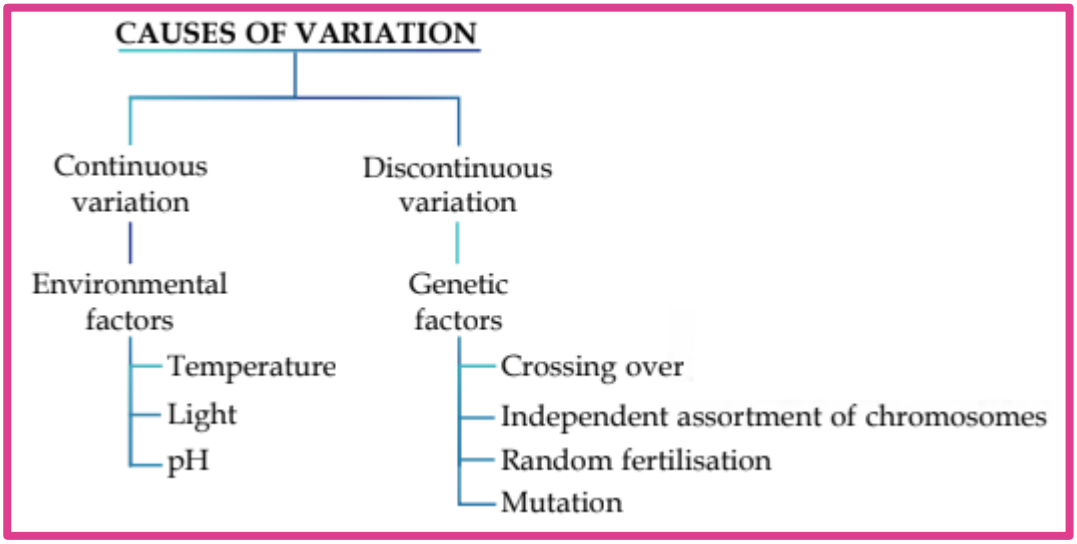
SIMILARITY
⌂ Shows differences in characteristics among individuals of the same species

DIFFERENCES

CONTINUOUS VARIATION	DISCONTINUOUS VARIATION
No obvious differences in characteristics	Obvious and distinct differences in characteristics
Presence of intermediate characteristics	No intermediate characteristics
Graph with normal distribution	Graph with discrete bars
Characteristic is controlled by many genes	Characteristic is controlled by one single gene
Influenced by environmental factors	Not influenced by environmental factors
Can be measured (quantitative)	Cannot be measured (qualitative)

Causes of variation

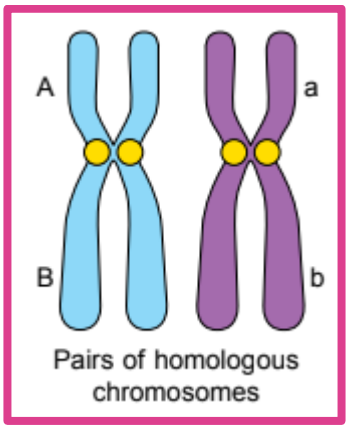
- Organisms of the same species differ in terms of morphology, physiology and genetics



Genetic factors

CROSSING OVER

- Crossing over occurs between non-sister chromatids of homologous chromosomes during prophase I of meiosis
- Recombination produces new combinations of genes
- Sister chromatids that separate during anaphase II of meiosis form gametes with different genetic materials at the end of meiosis



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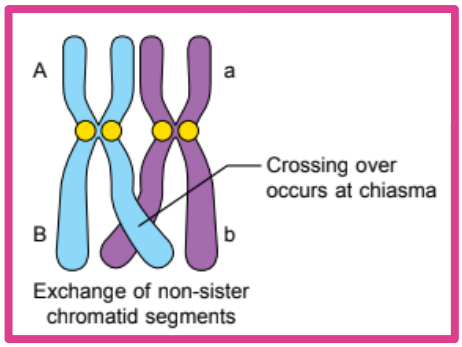

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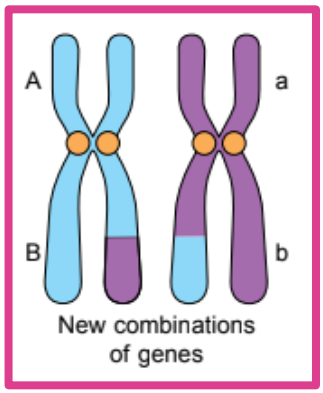
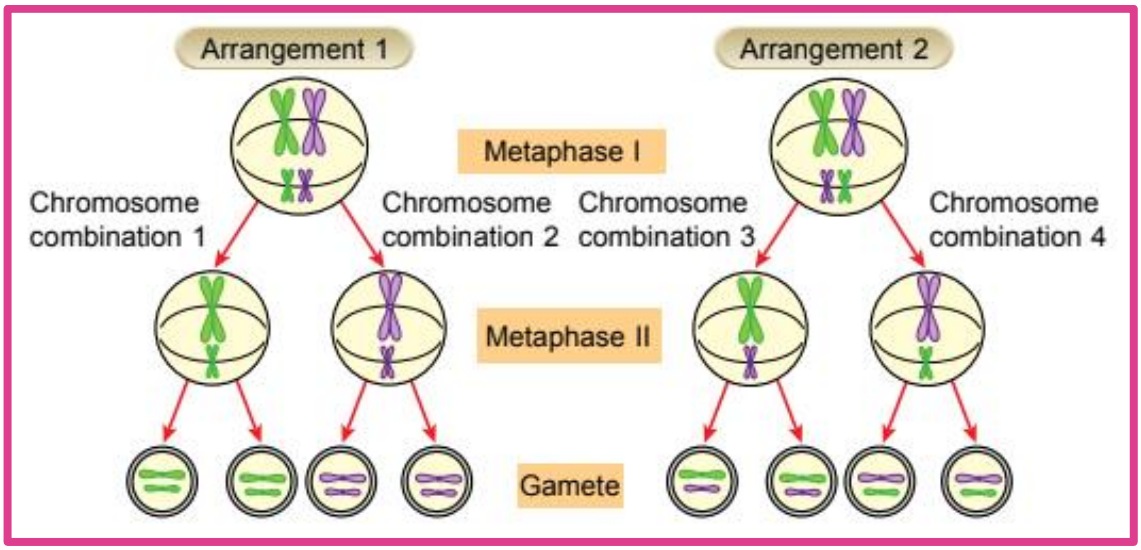


3

INDEPENDENT ASSORTMENT OF CHROMOSOMES

- During metaphase I of meiosis, a homologous chromosome pair (one maternal chromosome and one paternal chromosome) is arranged randomly on the equatorial plane of a cell
- There are two probabilities on the arrangement of homologous chromosomes on the equatorial plane for each diploid cell (2n=4)

- At the end of meiosis, different gametes will be produced with different combinations of paternal and maternal chromosomes, which results in the genetic contents of each gamete to be different from the others



RANDOM FERTILISATION

- Fertilisation between sperm and secondary oocyte is random
- Genetic recombination which occurs during crossing over and random arrangement of homologous chromosomes in meiosis, produces gametes with different genetic contents from their parents
- Therefore, a diploid zygote which is produced after fertilisation will have a new genetic combination

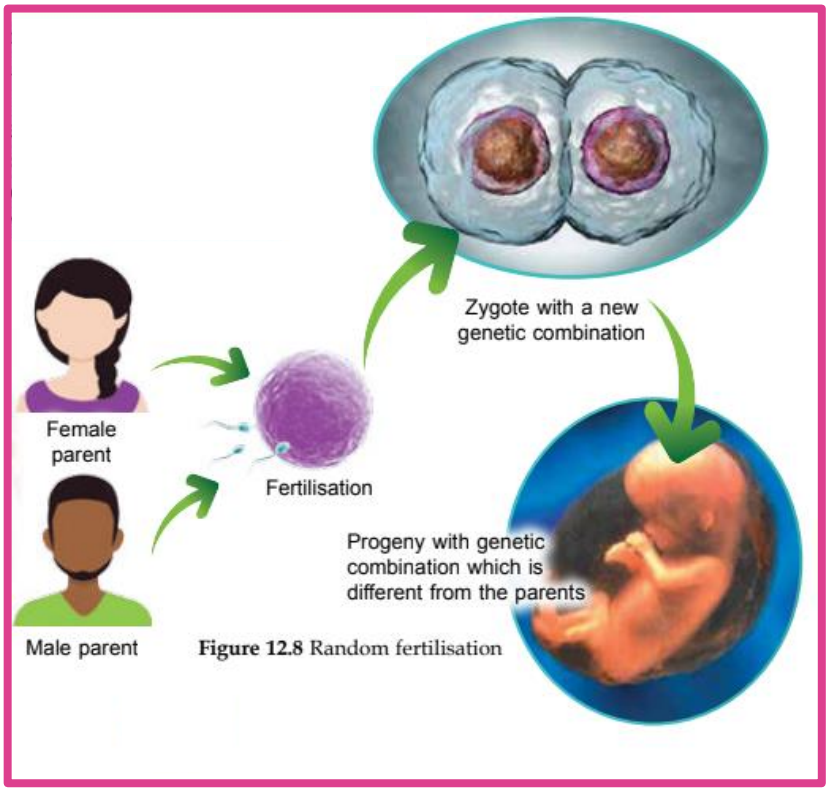
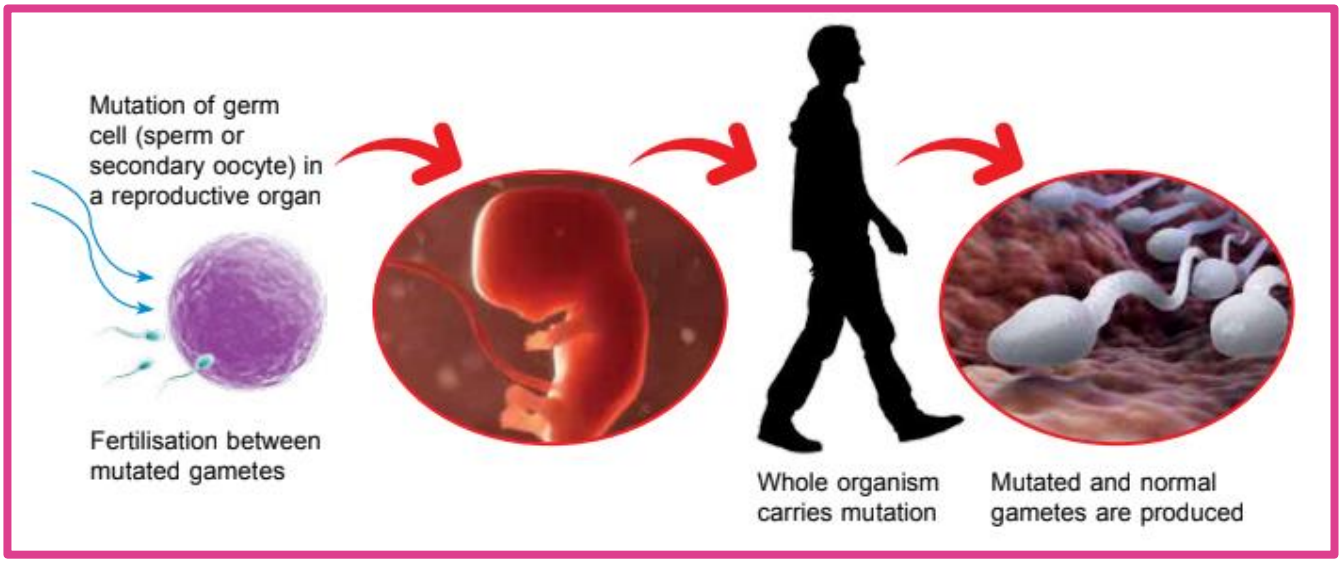


Figure 12.8 Random fertilisation

MUTATION

- Mutation is a permanent change which occurs spontaneously on genes or chromosomes
- Mutation creates new genotypes
- If mutation occurs in the gamete (mutation of germ cell), the characteristics determined by mutated genetic materials can be inherited
- Mutation of the somatic cell can cause variation but the characteristics cannot be inherited by the next generations



Environmental factors

- Variation caused by environmental factors is known as environmental variation
- Environmental factors that cause variation include abiosis factors such as temperature, light and pH
- In contrast to variation caused by genetics, the effect of environment on variation is small since it only involves phenotypic differences and not genotypic differences
- Environment can change allele frequency and genotype frequency in a population but cannot change the genotype
- Therefore, environmental variation cannot be inherited from one generation to the next

SOIL pH

- Hydrangea sp. plant produces blue flowers in acidic soil (pH less than 5.5) and pink flowers in alkaline soil

Hydrangea sp.

TEMPERATURE

→ Siamese cat inherited the gene which produces dark pigment enzyme for fur colour

→ This enzyme only functions at temperature less than the body temperature

→ Parts of the body with lower temperature are the ears, face, tail and paws

→ Therefore, these body parts are dark in colour

Siamese cat

LIGHT

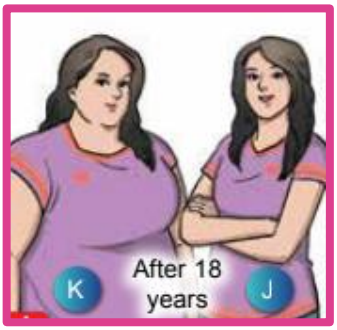
↘ Sunlight can alter skin colour
↘ People who are under the sun for a long period of time will have tanned skin
↘ For example, a construction worker Ultraviolet rays in sunlight can destroy melanin pigment of exposed skin
↘ Therefore, more melanin pigment will be produced, which cause the skin to be darker

Interaction between genetic and environmental factors

- Environmental factors can interact with genetic factors to cause variation, in which the environmental factors determine the phenotypes
- Characteristics inherited from parents such as height, intelligence and skin colour are greatly influenced by environmental factors
- An example which supports the effect of environmental factors on genetic factors can be observed in identical twins

INTERACTION OF ENVIRONMENTAL VARIATIONS IN DETERMINING PHENOTYPES OF IDENTICAL TWINS

» J and K are identical twins
» Both have similar genotype for obesity since they originated from the same embryo
» J and K were separated since childhood
» J likes low fat diet and enjoys physical activities whereas K likes high fat diet and does not like physical activities
» K is obese whereas J has an ideal body weight
» Conclusion: Differences in eating habits and environment while growing up produce different phenotypes



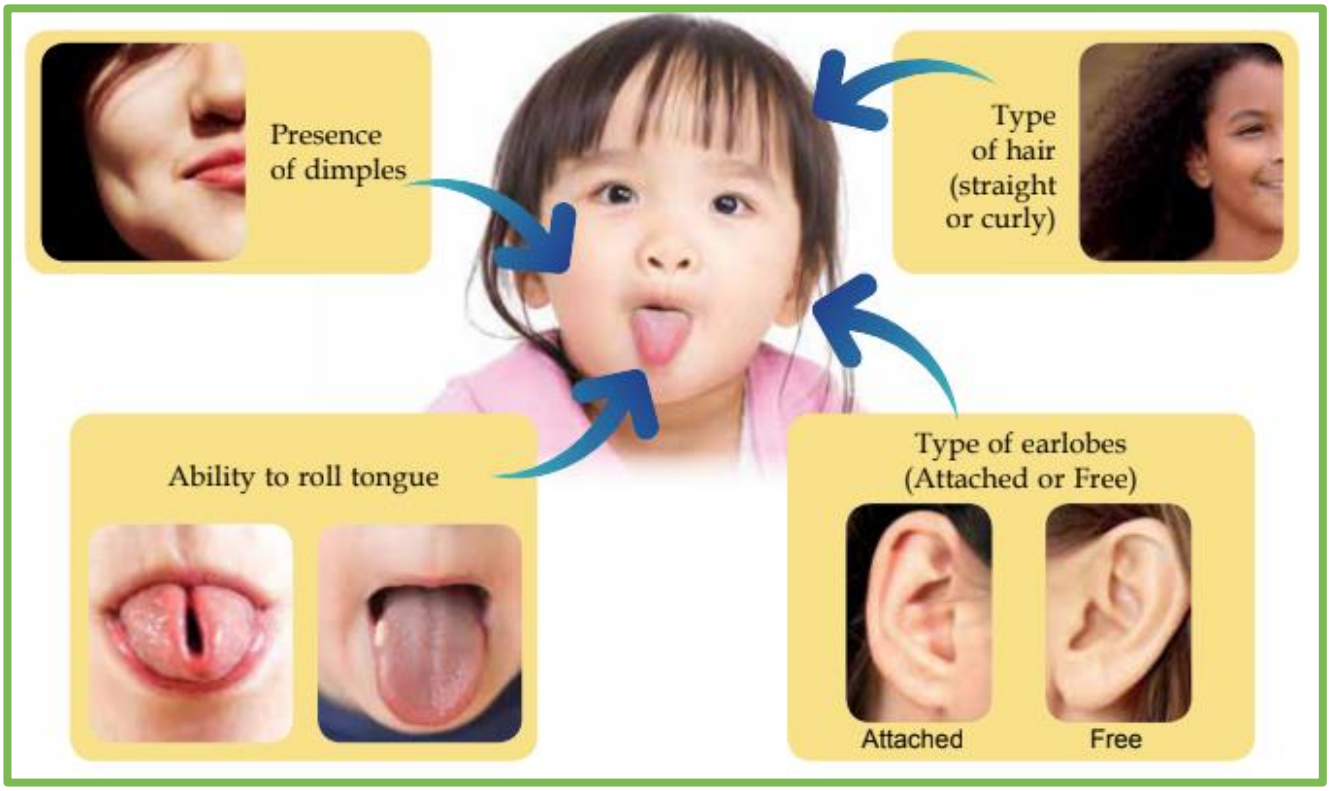
CHAPTER 12.2 – VARIATION IN HUMANS

Relation of variation to human inheritance

- Δ Human trait is controlled by a pair of alleles, in which an allele can be dominant or recessive
- Δ Dominant trait is observed when both dominant alleles are present or when one dominant allele is paired with a recessive allele whereas recessive trait is only shown when both recessive alleles are present
- Δ These genetic information causes variation in humans

EXAMPLES OF CHARACTERISTICS AND TRAITS IN HUMANS

CHARACTERISTICS	TRAITS IN HUMAN	
	DOMINANT	RECESSIVE
Height	Tall	Short
Type of hair	Curly hair	Straight hair
Tendency to use hands	Right-handed	Left-handed
Presence of dimples	Presence	Absence
Ability to roll tongue	Can roll tongue	Cannot roll tongue
Type of earlobes	Free earlobes	Attached earlobes



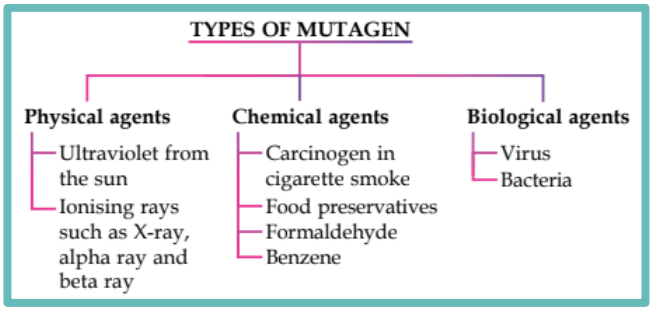
CHAPTER 12.3 – MUTATION

Mutation

- Mutation is a spontaneous and random change of genetic material, namely DNA of the cell in an organism
- A substance which causes mutation or increases the mutation rate to a dangerous level is called a mutagen
- Mutation occurs spontaneously in natural conditions
- New genetic material produced by mutation is called a mutant
- A mutant can exist as
    1) Mutant gene
    2) Mutant cell
    3) Mutant organelle
    4) Mutant individual

Types of mutagens

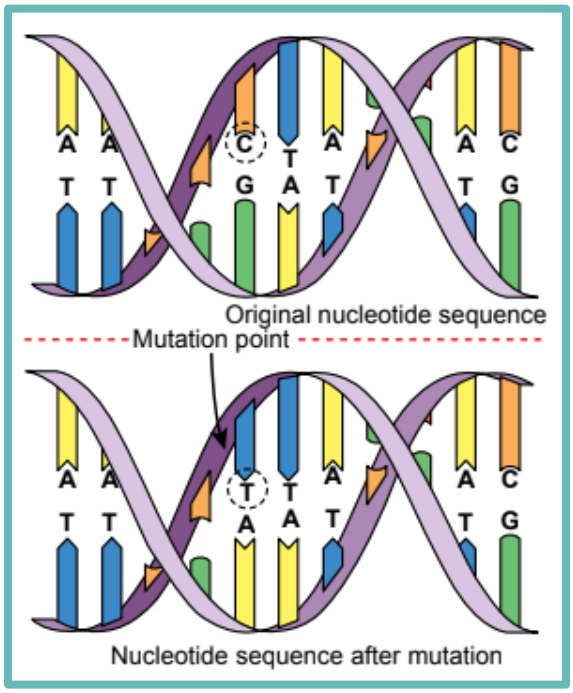
- Mutagen is divided into three types of agents
    1) Physical agent
    2) Chemical agent
    3) Biological agent

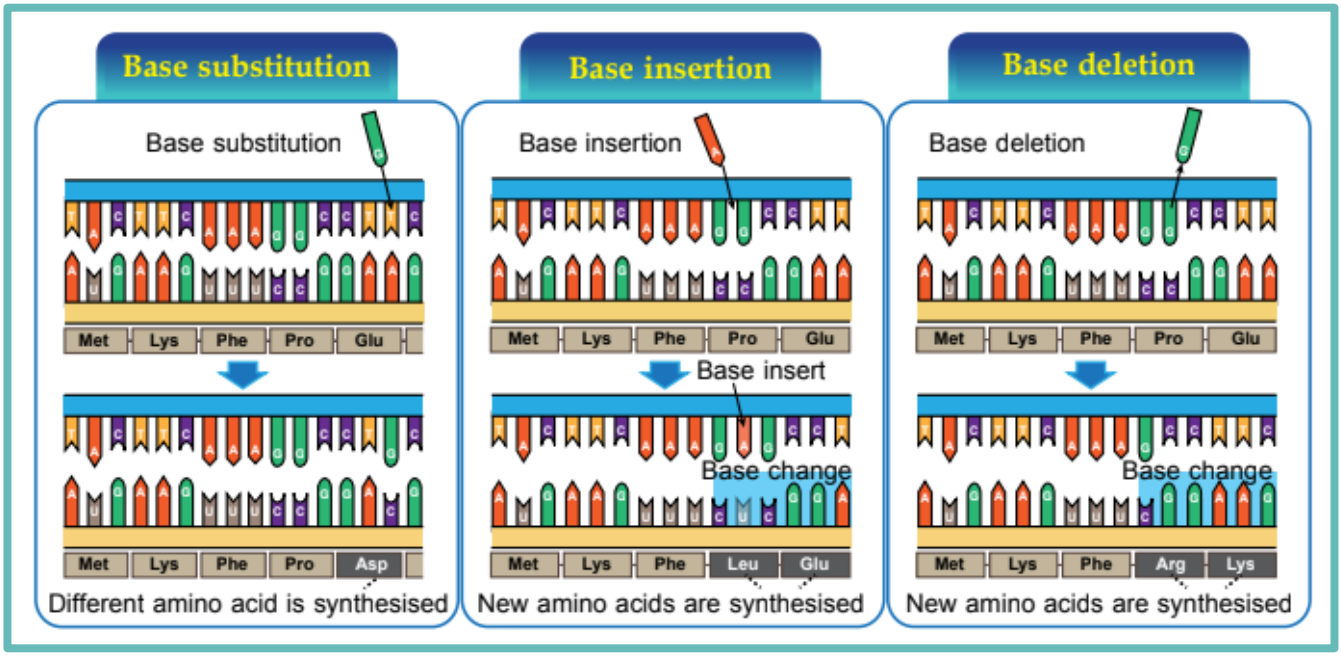


Types of mutations

Gene mutation

- Gene mutation occurs when there is a change in nucleotide base sequence of a gene
- Gene mutation is also known as point mutation
- The change alters the genetic code that is used to synthesise amino acid
- Therefore, there will be a change in protein structure and this new protein cannot function
- Gene mutation occurs by base substitution, base deletion and base insertion
- Gene mutation causes genetic diseases such as thalassemia, cystic fibrosis, sickle cell anaemia, albinism and haemophilia



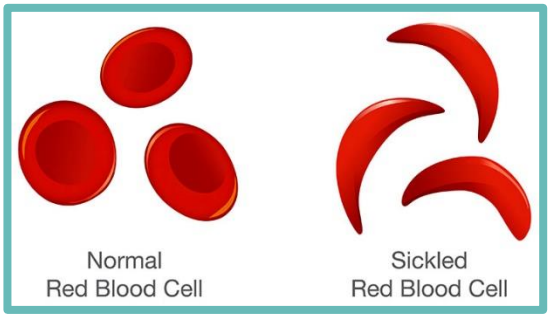


EXAMPLES OF GENETIC DISEASE

TYPES OF GENE MUTATION	EXAMPLES OF DISEASE
Base substitution	Sickle cell anaemia
Base insertion	Cystic fibrosis
Base deletion	Thalassemia

SICKLE CELL ANAEMIA

- ✓ Sickle cell anaemia is caused by a gene responsible for the synthesis of haemoglobin
- ✓ The red blood cells of a patient are in the shape of a crescent
- ✓ This is because the red blood cells are not properly formed
- ✓ Some red blood cells are normal whereas the rest are crescent shape



ALBINISM

- ⊕ An individual who experiences albinism is an albino
- ⊕ Albinism is due to the mutation of a gene which is responsible for producing pigments of skin, hair and eyes
- ⊕ Hence, the pigments are not produced in the albino

Chromosomal mutation

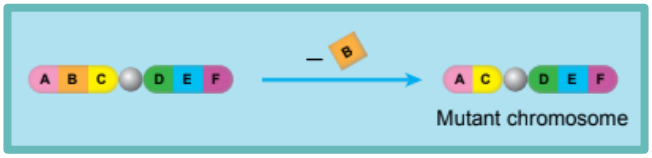
- ◊ Chromosomal mutation involves changes to chromosomal structure or changes to the chromosomal number
- ◊ Chromosomal mutation can change the characteristics of an organism

- ◊ Changes in chromosomal structure involve changes to the gene sequence in a chromosome
- ◊ The structural change causes abnormality to the chromosome, which is known as chromosomal aberration
- ◊ Types of chromosomal aberration
  - a) Deletion
  - b) Duplication
  - c) Inversion
  - d) Translocation

CHROMOSOMAL MUTATION

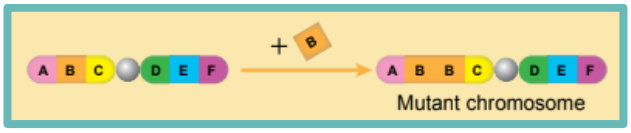
Deletion

- A part or a segment of chromosome is deleted or lost (loss of a few genes)



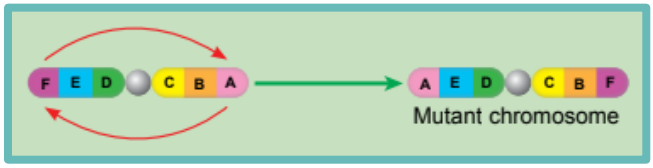
Duplication

- ➢ A part of a chromosome is copied which causes repetition of gene sequence



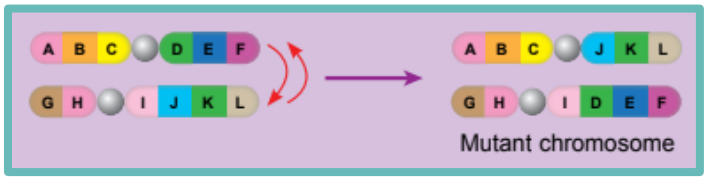
Inversion

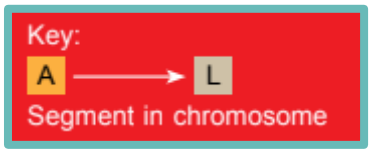
- ❖ A segment of chromosome is reversed end to end at 180o then re-join (change in DNA sequence of chromosome)



Translocation

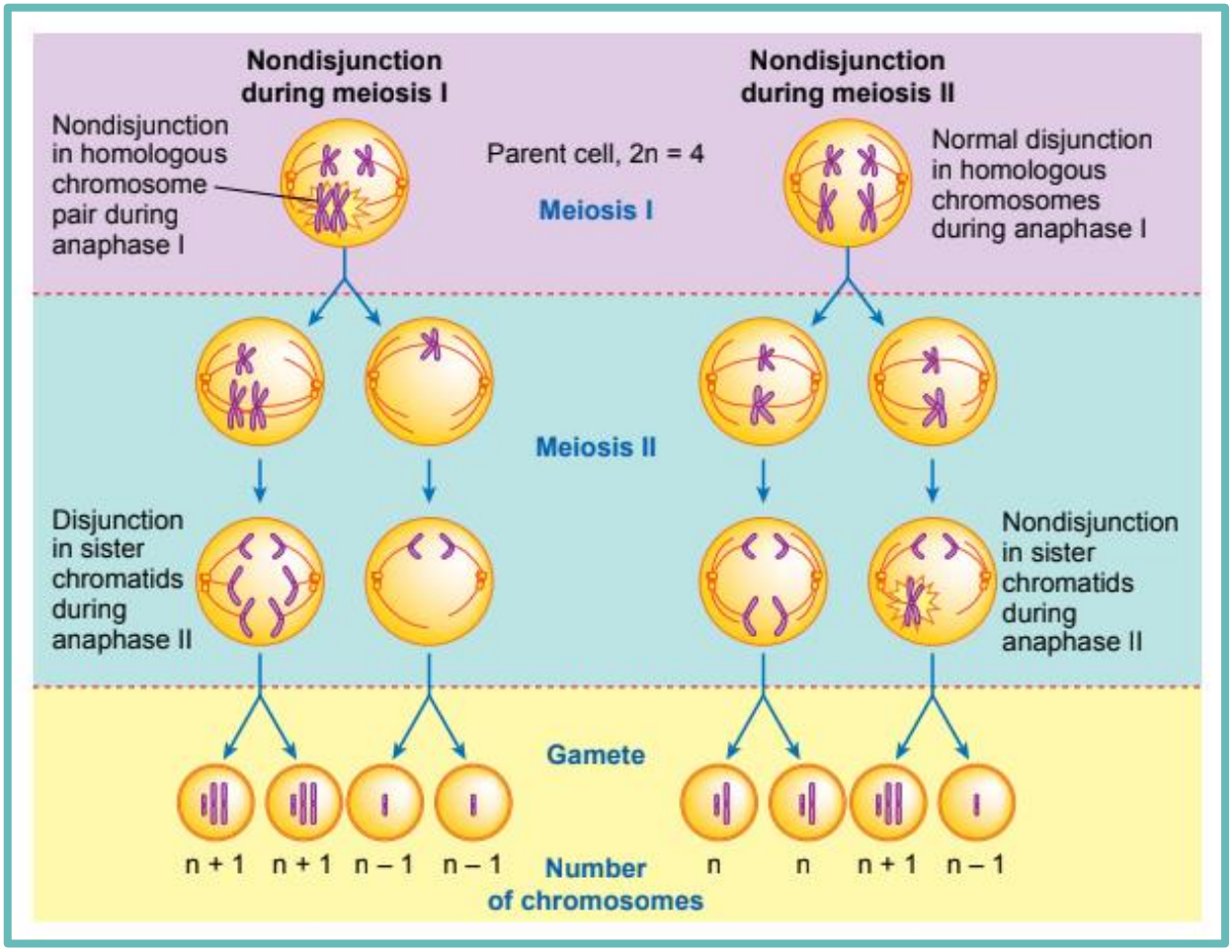
- → Part of the chromosome is cut, then join to another non-homologous chromosome





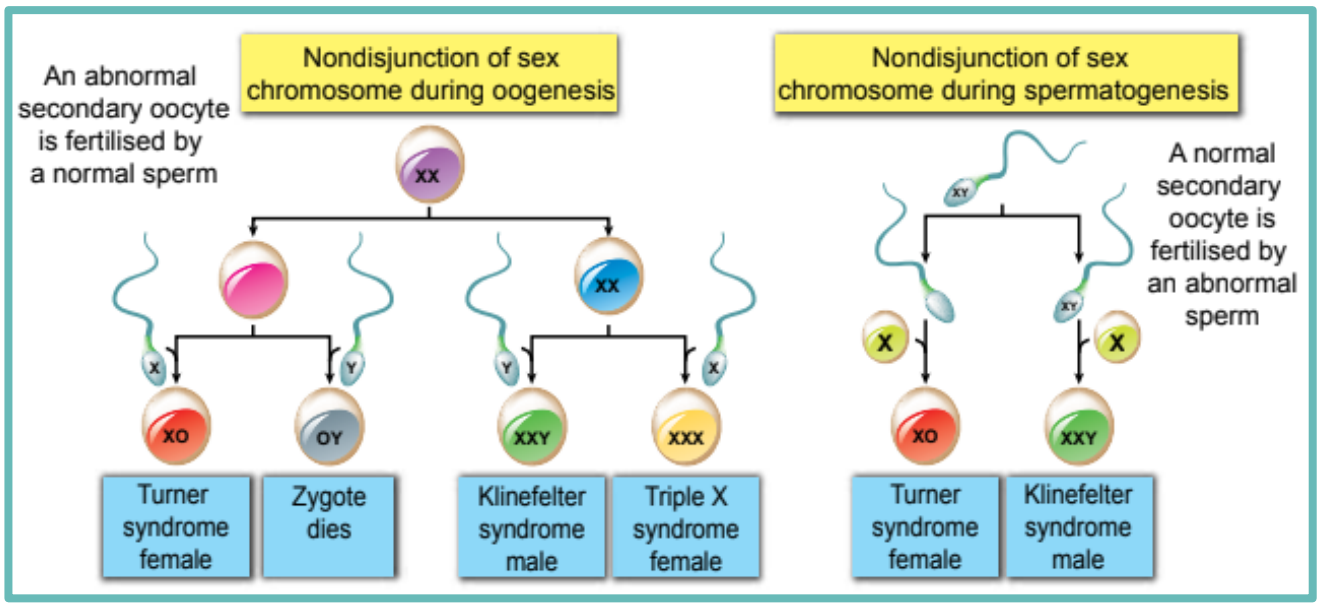
NONDISJUNCTION IN CHROMOSOMES DURING MEIOSIS

- ↘ A change in chromosomal number causes a diploid organism to lose one or more chromosomes, or gain one or more chromosomes
- ↘ The change occurs when homologous chromosomes fail to separate during anaphase I in meiosis or sister-chromatids fail to separate during anaphase II in meiosis
- ↘ This is probably because the normal spindle fibres fail to form during meiosis
- ↘ The phenomenon is known as nondisjunction and it causes abnormality in the number of chromosomes and in a gamete
- ↘ The gamete may lose one or more chromosomes or gain one or more chromosomes



DISEASES CAUSED BY MUTATION OF CHROMOSOMAL NUMBER

- Abnormality in the number of chromosomes can be due to nondisjunction during spermatogenesis or oogenesis
- Fertilisation that involves abnormal gametes will produce a zygote that develops into an individual with abnormal characteristics
- The individual experiences change in phenotype



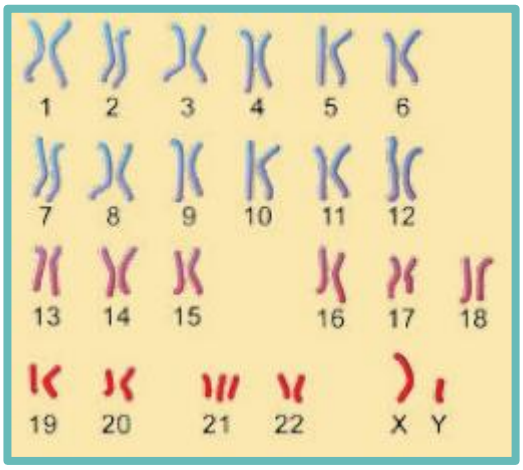
Characteristics of disease caused by chromosome mutation

Abnormalities in autosomes

DOWN SYNDROME

Chromosome number

- 2n + 1 = 47
- Has three number 21 chromosomes



Characteristics of the disease

- Δ An individual (male or female) has slant eyes, flat nose, protruding tongue, broad forehead and is usually mentally retarded

CRI DU CHAT SYNDROME

Chromosome number

- ⌂ The structure of chromosome changes due to a partial deletion on the short arm of chromosome number 5

Characteristics of the disease

- » The cry of affected infants is similar to that of a meowing kitten
- » Affected individual shows delayed development with mental and physical retardation
- » Most patients die during childhood

Abnormalities in sex chromosomes

KLINEFELTER SYNDROME

Chromosome number

- ✓ 2n + 1 = 47
- ✓ 44 + XXY

Characteristics of the disease

- ◊ A sterile man with small testes that fail to produce sperms
- ◊ Possesses voice and chest similar to those of a woman
- ◊ Has long legs and hands

JACOB SYNDROME

Chromosome number

- ⊕ 2n + 1 = 47
- ⊕ 44 + XYY

Characteristics of the disease

- Male who is taller than normal and has problem with pimples on his face
- Slow in acquiring speech and has learning disability
- Possesses weak muscles (hypotonia)

TURNER SYNDROME

Chromosome number

- (2n - 1) = 45
- 44 + XO

Characteristics of the disease

- ❑ Sterile female with lack of secondary female characteristics, undeveloped breasts and ovaries
- ❑ Weblike neck and low IQ

Mutation of somatic cells and gametes

DIFFERENCES BETWEEN MUTATIONS OF SOMATIC CELLS AND GAMETES

MUTATION IN SOMATIC CELLS	MUTATION IN GAMETES
Involves somatic cells such as skin cell and eye cell	Involves germ cell that produces gamete (Secondary oocyte or sperm)
Cannot be inherited by the next generation	Can be inherited by the next generation
Disease is present only in the individual with the mutation	Disease is present in the individual with the mutation and is also inherited by his/her descendants
Example: Disease related to nervous system	Example: All inherited diseases such as thalassemia