CHAPTER 6

MUTATION

BIOLOGY UNIT

KMKT 2021/2022

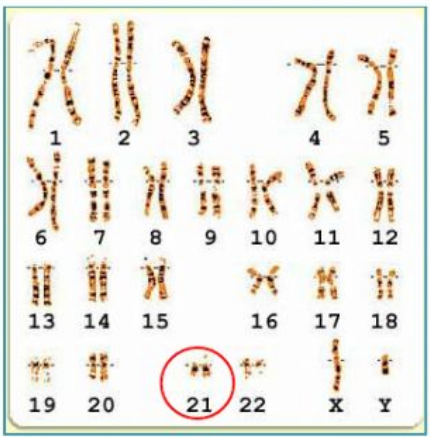
6.1 MUTATION CLASSIFICATION & TYPES

Learning outcome
At the end of the lessons, students should be able to:

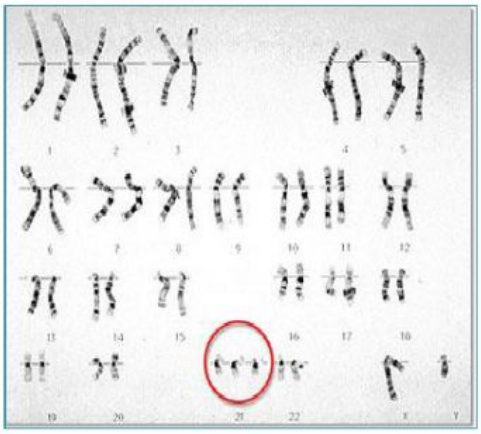
a. Define mutation
b. State two types of mutation:
   i. Spontaneous mutation
   ii. Induced mutation (e.g. exposure to mutagens).
c. Classify two classes of mutation to:
   i. Gene/point mutation
   ii. Chromosomal mutation
d. Define mutagen.
e. State types of mutagen:
   i. Physical (e.g. UV rays and gamma rays).
   ii. Chemical (e.g. colchicine and ethidium bromide).

Definition of Mutation

"A mutation is a _____ in the amount, arrangement or structure of the DNA of an organism".

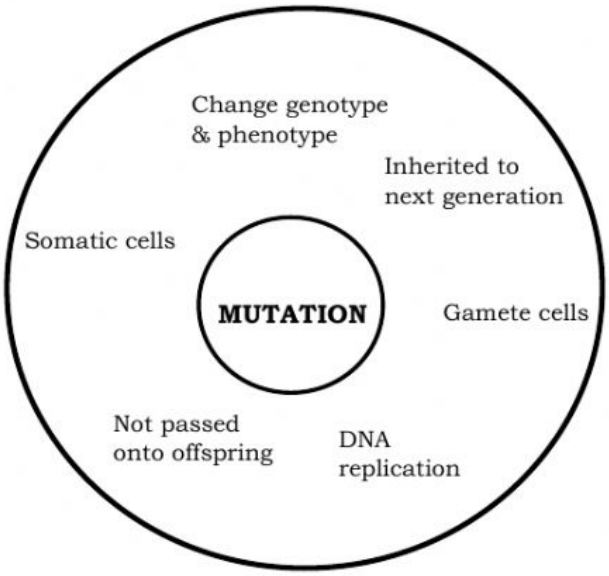


Normal karyotype



Abnormal karyotype (changes in number of chromosomes 21)

Figure 6.1: Example of chromosomal mutation.



Exercise 6.1 (a): Use all the ideas/words given to briefly describe about mutation.You can add another suitable word.

Mutation can occur during the _____ . Mutation result in a change of protein, thus it cannot function as it should be.

Mutation occur in _____ or _____ . Mutation in gamete cells will be _____ while in somatic cells it only can be inherited by daughter cells produced by mitosis. Mutation in somatic cells are _____.

Hence, mutation may lead to _____ that lead to the production of new traits

Classification of Mutation

Mutation is classified into:

a. _____ Mutation
b. _____ Mutation

Types of Mutation

a. _____ mutation. E.g: Non-disjunction
b. _____ mutation. E.g: Exposure to mutagen

Definition of Mutagen

"Biological, chemical, or physical agent, process, or substance that causes permanent genetic change (mutation) in a cell (other than which occurs during normal cell growth) due to genetic alterations or loss of genes or chromosomes".

Read more: http://www.businessdictionary.com/definition/mutagen.html#ixzz4ACKPiECT

Types of Mutagen

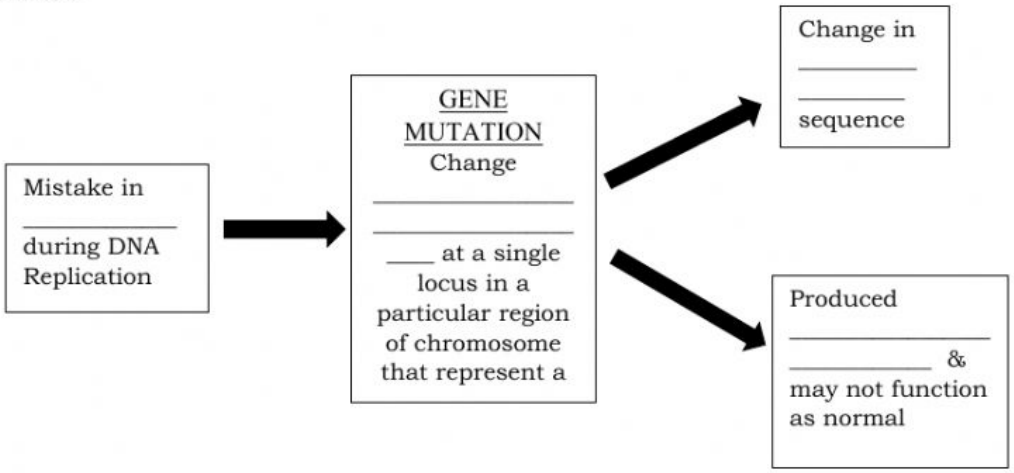
a. _____ mutagen. E.g: UV rays and Gamma rays.
b. _____ mutagen. E.g: colchcine and ethidium bromide.

6.2 GENE MUTATION

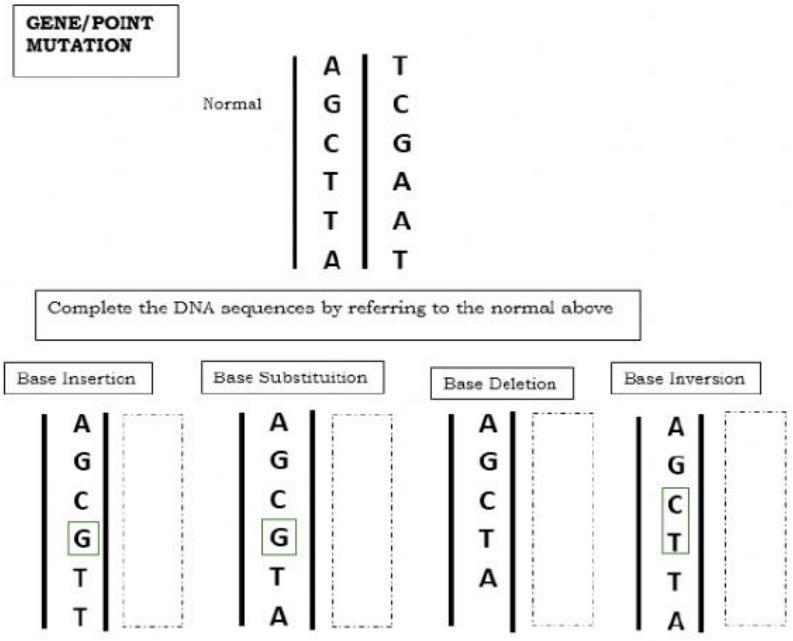
Learning outcome
At the end of the lessons, students should be able to:
a. Define gene mutation.
b. Define the four types of gene mutation:
   i. Base substitution
   ii. Base insertion
   iii. Base deletion
   iv. Base inversion
c. Describe the consequences of base substitution (e.g. Sickle cell anaemia)

Gene Mutation

Definition:



Classification of Gene Mutation



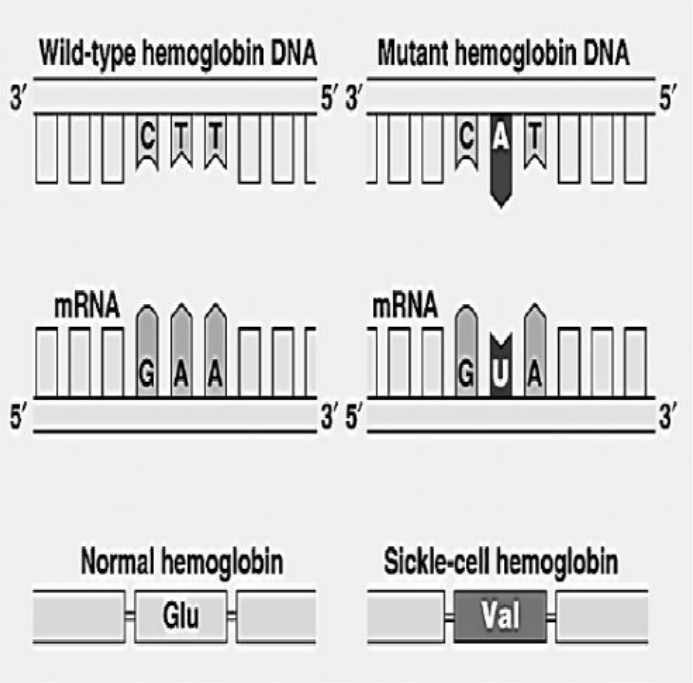
Base Substituition as Point Mutation

Definition:
One or a few base pairs in the nucleotides sequence in genes are replaced with other different base pairs.

BASE SUBSTITUTION

Changes in _____ _____ that result in changes in codon

Leads to changes of _____ during translation



About sickle cell anemia

-Abnormal in hemeglobin cause RBC becomes sickled chape.

-easy to break or stuck in blood capilaries

-not efficient in transporting oxygen

-patients usually suffer from heart problems, kidney failure, abdominal pains, paralysis and premature death.

Ref: https://en.wikipedia.org/wiki/Sickle-cell_disease

6.3 CHROMOSOMAL MUTATION

Learning outcome
At the end of the lessons, students should be able to:
a. Define chromosomal mutation
b. State two types of chromosomal mutation:
   i. Changes in chromosomal structure/chromosomal aberration (e.g: cri du chat)
   ii. Changes in chromosomal number (e.g: Down Syndrome)
c. Define the types of chromosomal aberration:
   i. Translocation
   ii. Deletion (segmental deletion) (e.g: *cri du chat*)
   iii. Inversion
   iv. Duplication
d. Define the types of the alteration of chromosomal number:
   i. Aneuploidy (e.g: Down Syndrome)
   ii. Euploidy/polyploidy

Chromosomal Mutation

Find the correct terms used in chromosomal mutation.

TERMS	EXPLAINATION
	1. A segment of a chromosome breaks, turned 180°C and reattaches in opposite orientation.
	2. The failure of a pair of homologous chromosomes to separate during meiosis I or sister chromatids to separate during meiosis II
Chromosomal Aberration	3. Chromosomal abnormalities that occur due to structural changes of the chromosome
	4. Addition or losing one or more individual chromosomes
Chromosomal number alteration	5. Change in number for individual chromosome or for the whole set of chromosomes
	6. The loss of one chromosomal segment containing one or more genes
	7. Alteration in the structure or number of the chromosome
Chromosomal duplication	8. A segment of a chromosome duplicated
	9. When chromosomal segment breaks and reattaches to another part of the same chromosome or other chromosome.
	10. Addition or losing the whole set of chromosomes

Non-Disjunction

Definition: The failure of a pair of homologous chromosomes to separate during meiosis I, or sister chromatids to separate during meiosis II

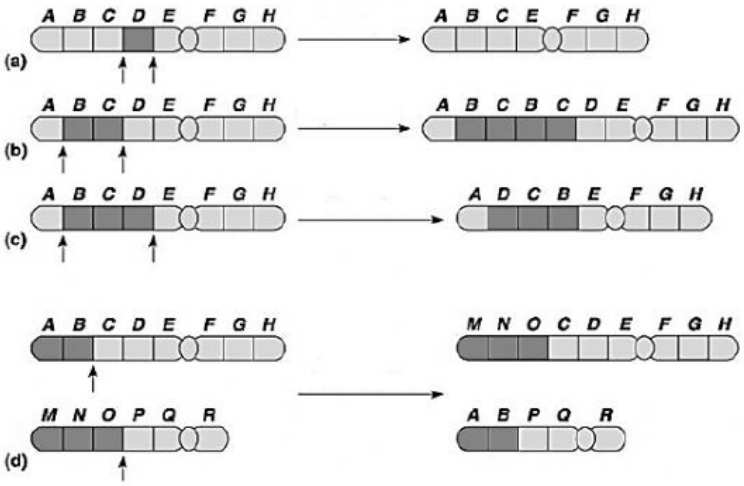
Why? →Because of spindle fibre CANNOT form

Effect? production of gametes with an abnormal number of sex-chromosomes or autosomes

Classification of Chromosomal Mutation

1. Chromosomal Aberration

Identify the type of Chromosomal Aberration based on the diagram below



A) _____

B) _____

C) _____

D) _____

2. Chromosomal Number Alteration

Definition = Change in number for individual chromosome or for the whole set of chromosomes

- Usually occurs due to _____ during meiosis I or meiosis II
- Producing gametes with abnormal number of chromosomes
- Change in the _____ of chromosomes lead to a chromosomal disorder

Complete all the information that can be associated with the abnormalities below:

	ABNORMALITIES	INFORMATION
1.	Trisomy 21 	• Also known as: • Symptoms: o growth failure o mental retardation o reduced resistance to disease o slanting eyes, broad flat face, short and o heart abnormalities • Number of chromosome : • Ploidy number: • Type of mutation: • Autosomal abnormality

	ABNORMALITIES	INFORMATION
2.	Cri Du Chat Syndrome 	• Number of chromosome: • Ploidy number:
		• Type of mutation:
		• Autosomal abnormality
		• number of chromosome: • Ploidy number:

Type of Alteration

1. **Aneuploidy**

- _____ one or more individual chromosomes
- Because of _____during _____
- Can occur in autosome and sex chromosome
- Eg : Monosomy, Triosomy

2. **Euploidy / Polyploidy**

- the condition of an organism having more than two sets of chromosomes (> 2n) →an increase in the whole set of chromosomes
- 2 types: _____ and _____
- common in plants / crops (e.g oats, cotton, potatoes, tobacco, wheat, grass)
- better qualities (e.g. more yields, more resistant to diseases / pests, grow faster, adaptation to a new environment)
- all chromosomes are not separated during meiosis (non-disjunction)