# CHAPTER 7

# MUTATION

( Hours: 2L + 6T )

# CHAPTER 7: MUTATION

7.1 Overview the classification and types of mutation

7.2 Gene Mutation

7.3 Chromosomal Mutation

| UPS 3 | PSPM 1 |
|---|---|
| 6 MCQ | 11 marks |



# MUTATION

## 7.1 Overview the classification & types of mutation

- Definition of Mutation
- Classification of Mutation:
  i. Spontaneous Mutation
  ii. Induced Mutation
- Types of mutation:
  i. Gene/point Mutation
  ii. Chromosomal Mutation

## 7.2 Gene Mutation

- Base Substitution
- Base Insertion
- Base Deletion

## 7.3 Chromosomal Mutation

- Chromosomal Aberration
- Change in chromosomal number

# LEARNING OUTCOMES

**7.1 Overview the classification and types of mutation**

a) Define mutation. (C1 - Lecture)

b) Classify mutation into: (C3 - Lecture)

   i. Spontaneous mutation; and

   ii. Induced Mutation (e.g. exposure to mutagens)

c) State two types of mutation: (C1 - Lecture)

   i. Gene / point mutation

   ii. Chromosomal mutation

# WHAT IS MUTATION?

**Definition:**

A mutation is a **permanent change** in the **amount, arrangement** or **structure** of the DNA of an organism.

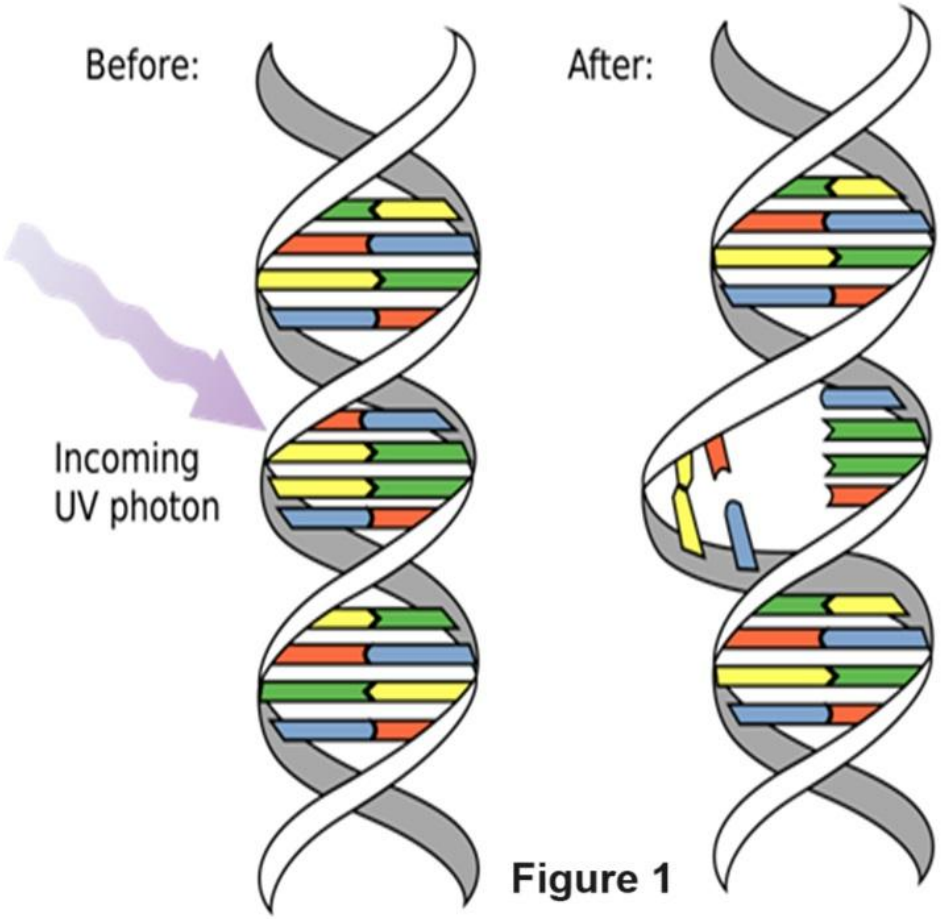
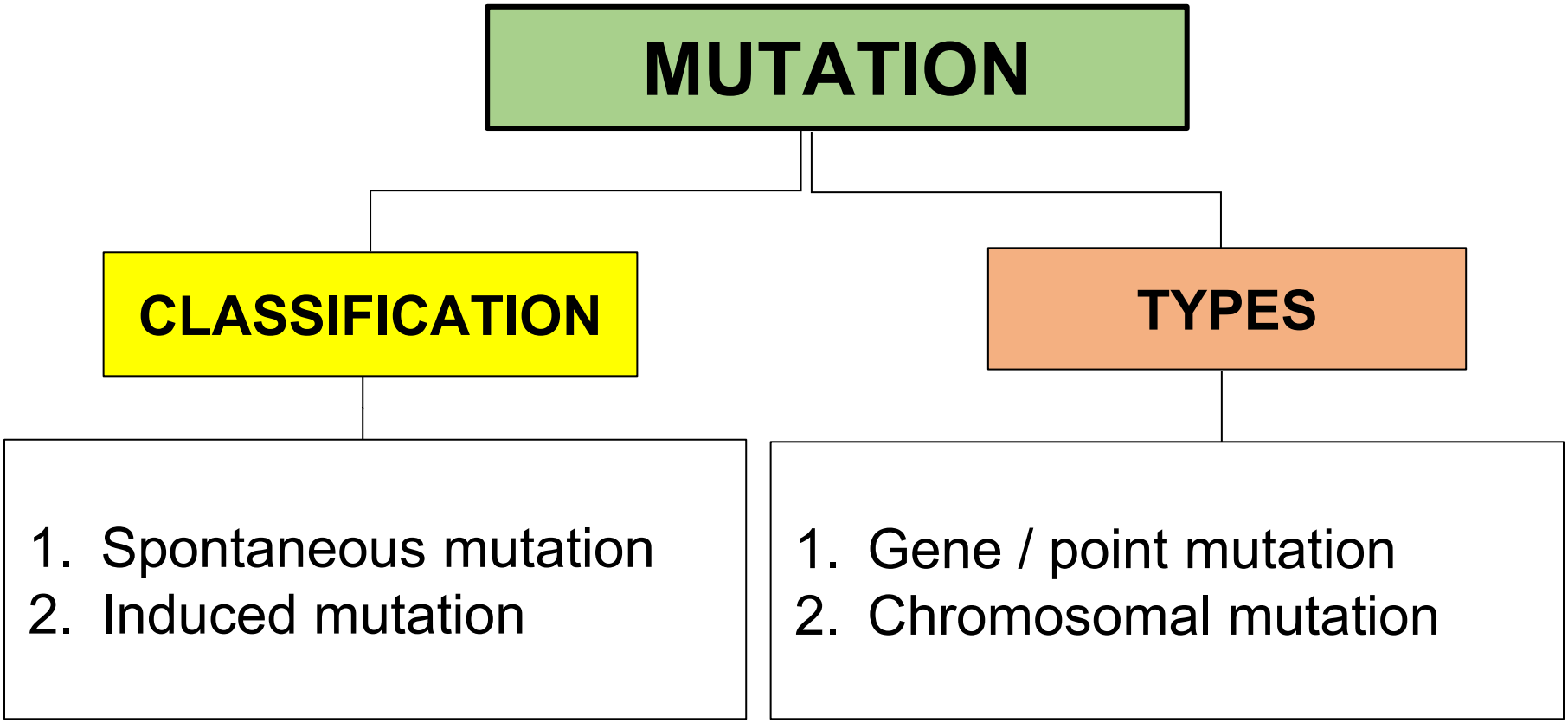



Figure 1

# CLASSIFICATION & TYPES OF MUTATION



# Classification of Mutation

## Spontaneous mutation

- Occur **naturally**/without human intervention
- Caused by:
  a) Mistakes in base pairing during **DNA replication**
  b) N**ondisjunction** during meiosis

## Induced mutation

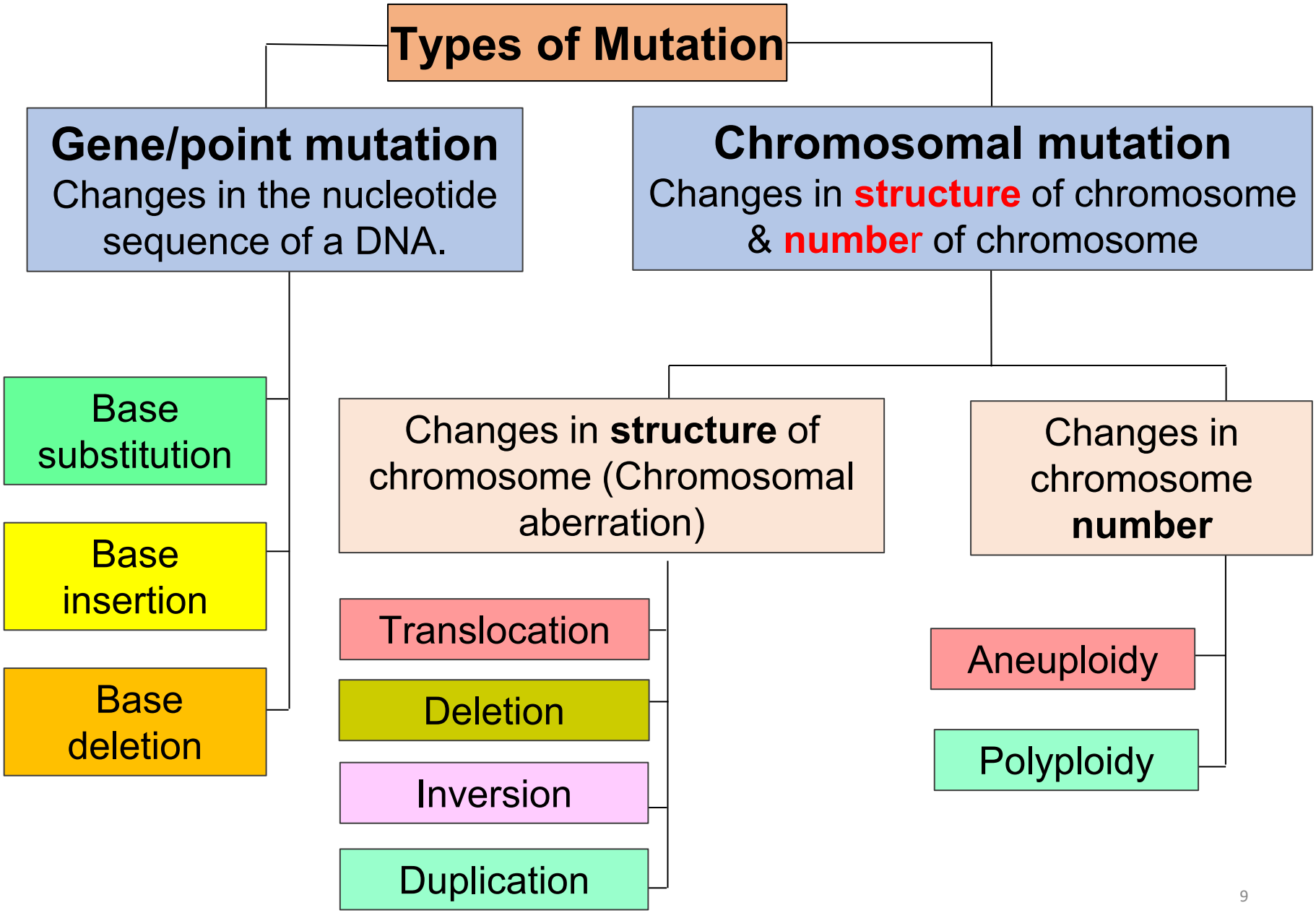
- Involve human intervention
- Occur when organism **exposed to mutagens** (**physical** mutagen & **chemical** mutagen)

# WHAT IS MUTAGEN?

A **chemical** or **physical agent** that interacts with DNA and can cause a mutation.

**Physical mutagen:**
Ultraviolet ray (UV ray)
Ionizing radiation (X-ray, gamma ray, alpha particles, neutron and electron)

**Chemical mutagen:**
Mustard gas, nitrous acid, base analogue etc.
Colchicine, ethidium bromide


Types of Mutation
Gene/point mutation
Changes in the nucleotide sequence of a DNA.
Chromosomal mutation
Changes in structure of chromosome & number of chromosome
Base substitution
Base insertion
Base deletion
Changes in structure of chromosome (Chromosomal aberration)
Changes in chromosome number
Translocation
Deletion
Inversion
Duplication
Aneuploidy
Polyploidy

# LEARNING OUTCOMES

## 7.2 Gene Mutation

a) Define gene mutation (C1 - Lecture)

b) State the three types of gene mutation (C1 - Lecture)

c) Explain three types of gene mutation: (C3 - Tutorial)

   i. Base substitution;

   ii. Base insertion; and

   iii. Base deletion

d) Explain base substitution (e.g.: Sickle cell anaemia as missense mutation) (C3 - Tutorial)

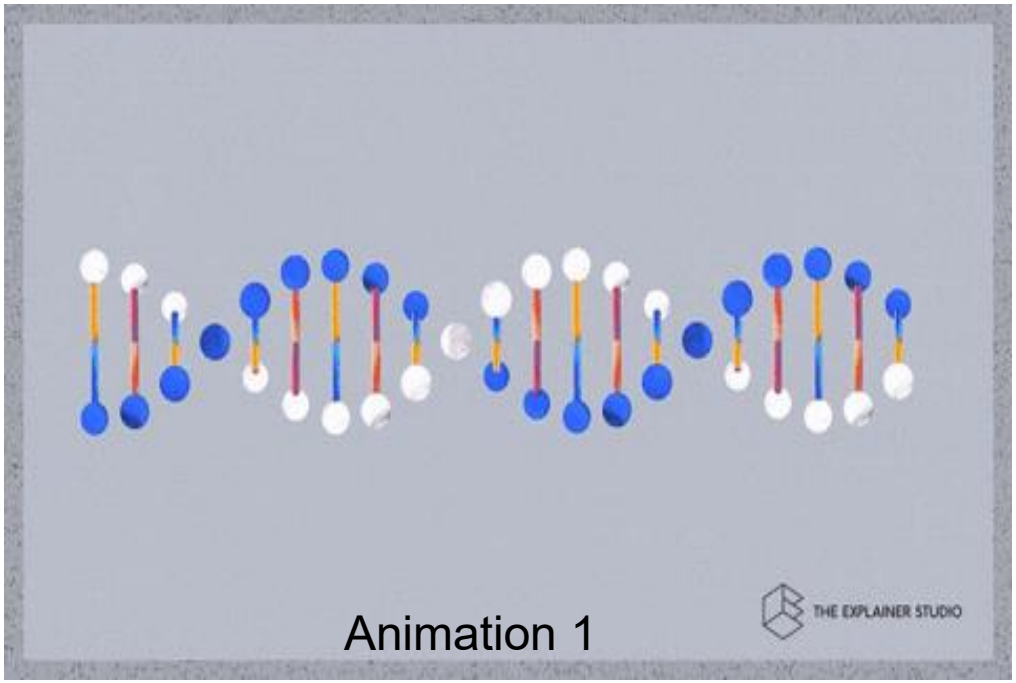
e) State the effect of base substitution (missense, nonsense and silent mutation) and base insertion and base deletion (frameshift mutation) (C1 - Lecture)

f) Explain base insertion and base deletion as a frameshift mutation. (C3 - Tutorial)

# GENE MUTATION

**Definition**:

Change in the **nucleotide base pair sequence of DNA** at a single locus in a particular region of chromosome that represent a gene.




Animation 1

# GENE MUTATION

- Also known as **point mutation**.
- Arise due to **mistake in base pairing** that occur during **DNA replication.**
- Result to **change in amino acid sequence** from original sequence.
- Thus **changes the protein**
- **Different protein produced** as the effect of mutation may **not function as normal.**

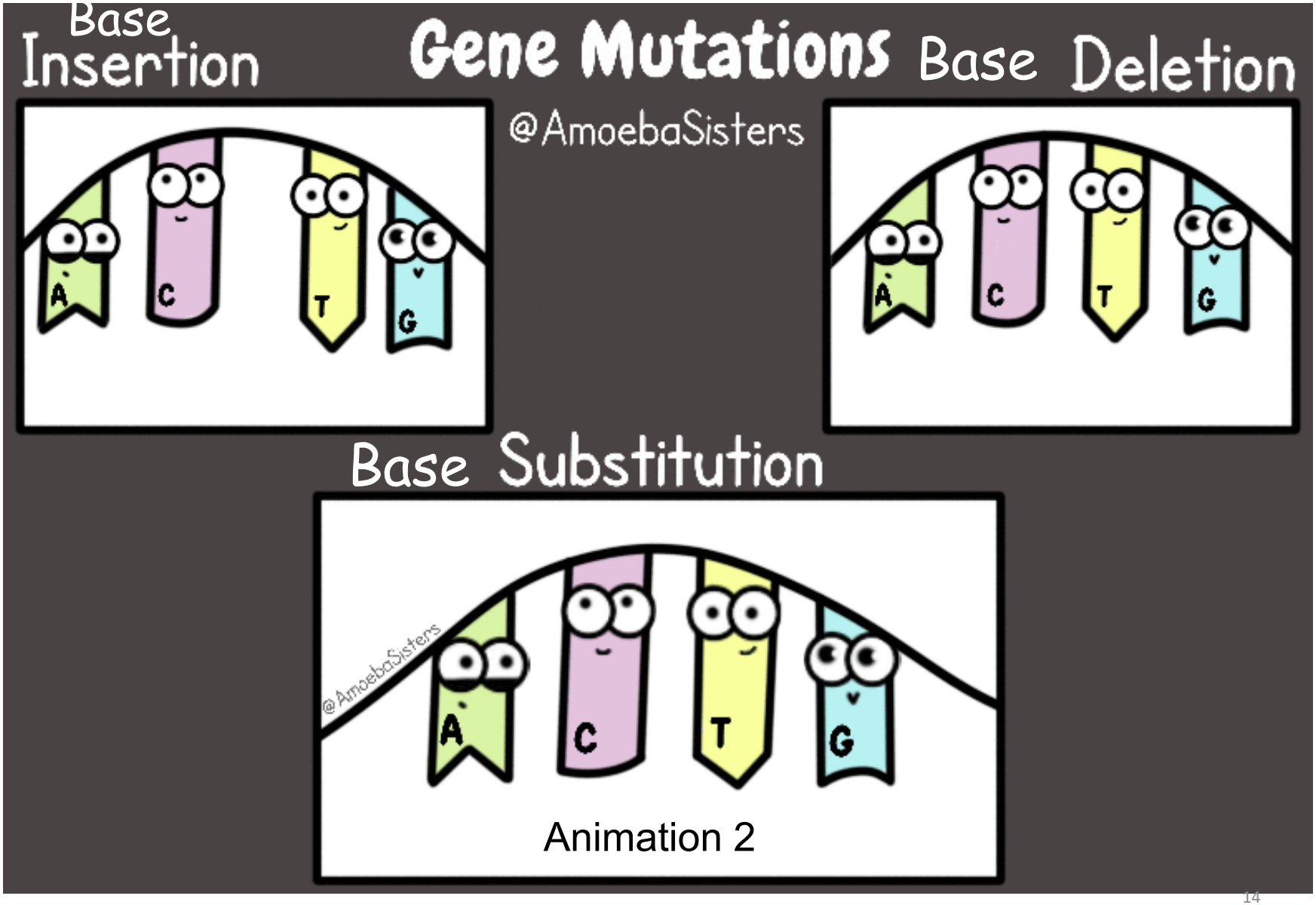
# TYPES OF GENE MUTATION

- **Base** Substitution
- **Base** Insertion
- **Base** Deletion

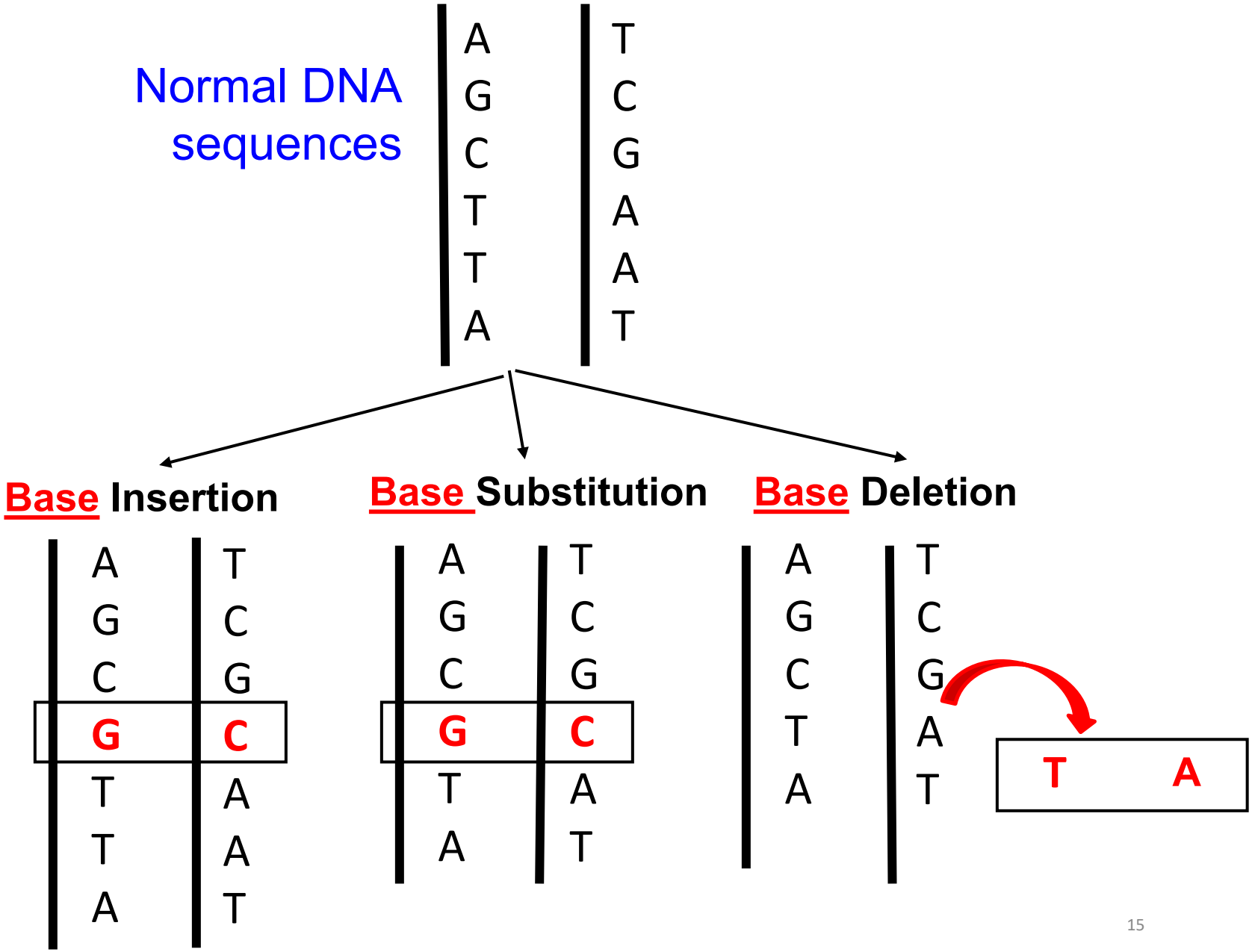
Tips from LKC

- Make sure use the term 'base' for each type of gene mutation

**Learning Outcome:**
**7.2 b) State the three types of gene mutation. (Lecture)**



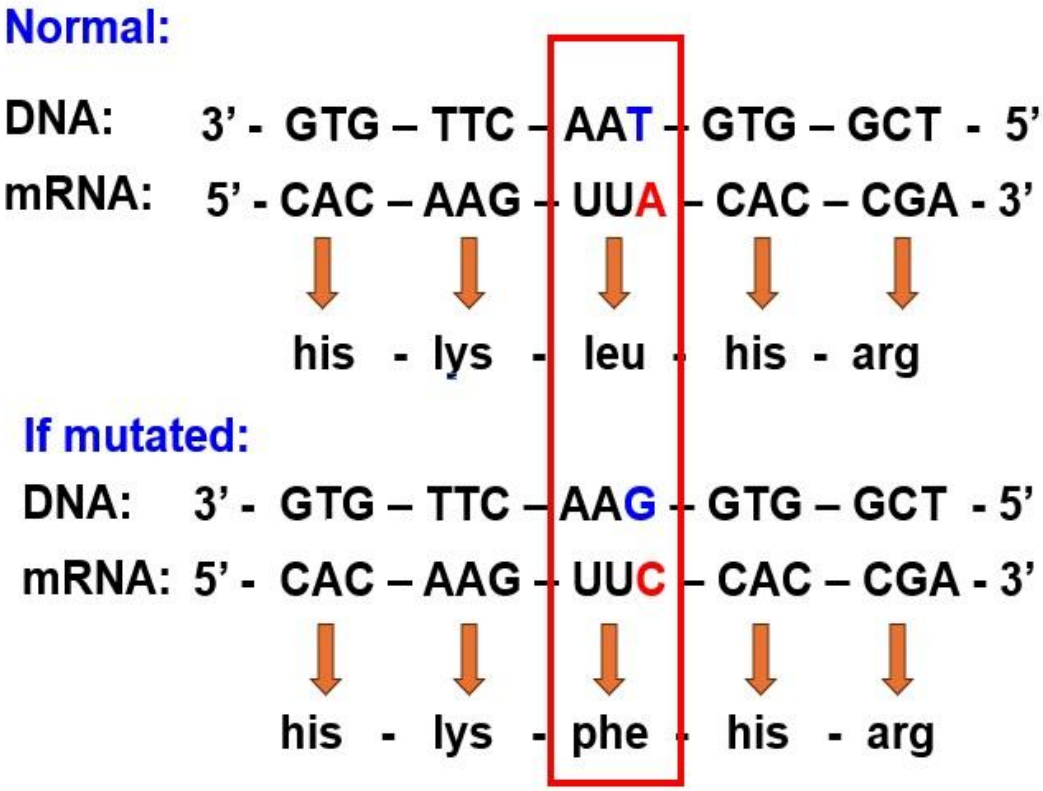

Animation 2


Normal DNA sequences
A G C T T A
T C G A A T
Base Insertion
A G C G T T A
T C G C A A T
Base Substitution
A G C G T A
T C G C A T
Base Deletion
A G C T A
T C G A T
T A

# i. Base Substitution

- One **nucleotide base pair is <u>replaced</u>** by other nucleotide pair in the DNA sequence.
- May cause changes in nucleotide base pair sequences that results in **changes of codon** in mRNA**.**
- This may lead to **changes of amino acids** during translation.
- Base substitution is a mutation that can cause:
  a) Missense mutation
     e.g. : Sickle cell anaemia as missense mutation
  b) Silent mutation
  c) Nonsense mutation

# i. Base Substitution




- In DNA, triplet base **3'-AAT-5'** is replaced with **3'-AAG-5'.**
- Base **T** of DNA is replaced with base **G.**
- Results in **changes of mRNA codon 5'-UUA-3' to 5'-UUC-3'.**
- Leads to **changes of amino acids (leu to phe).**

# ii. Base Insertion

**Definition:**

- **Addition of one or a few nucleotide base pair in** the DNA sequence.

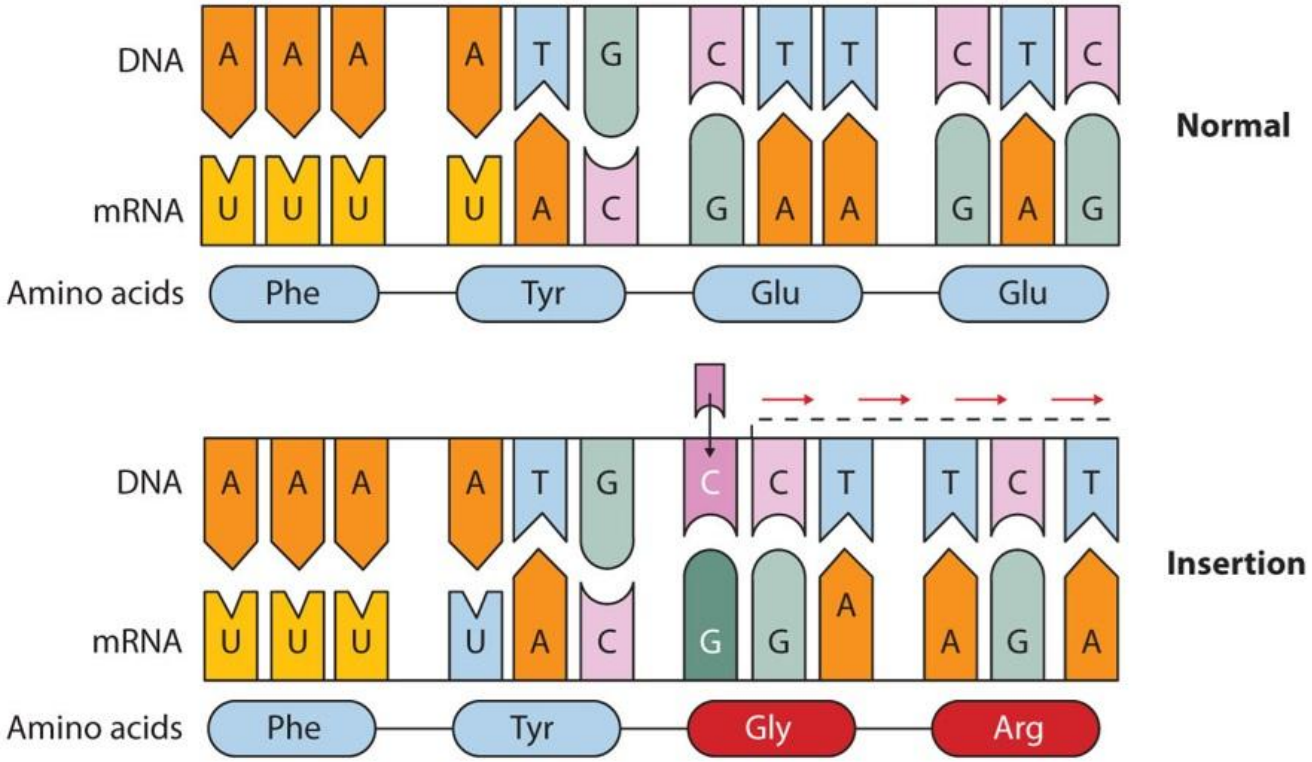



Figure 2

# iii. Base Deletion

**Definition:**

- **Loss/deletion of one or a few nucleotide base pairs** in DNA sequence.

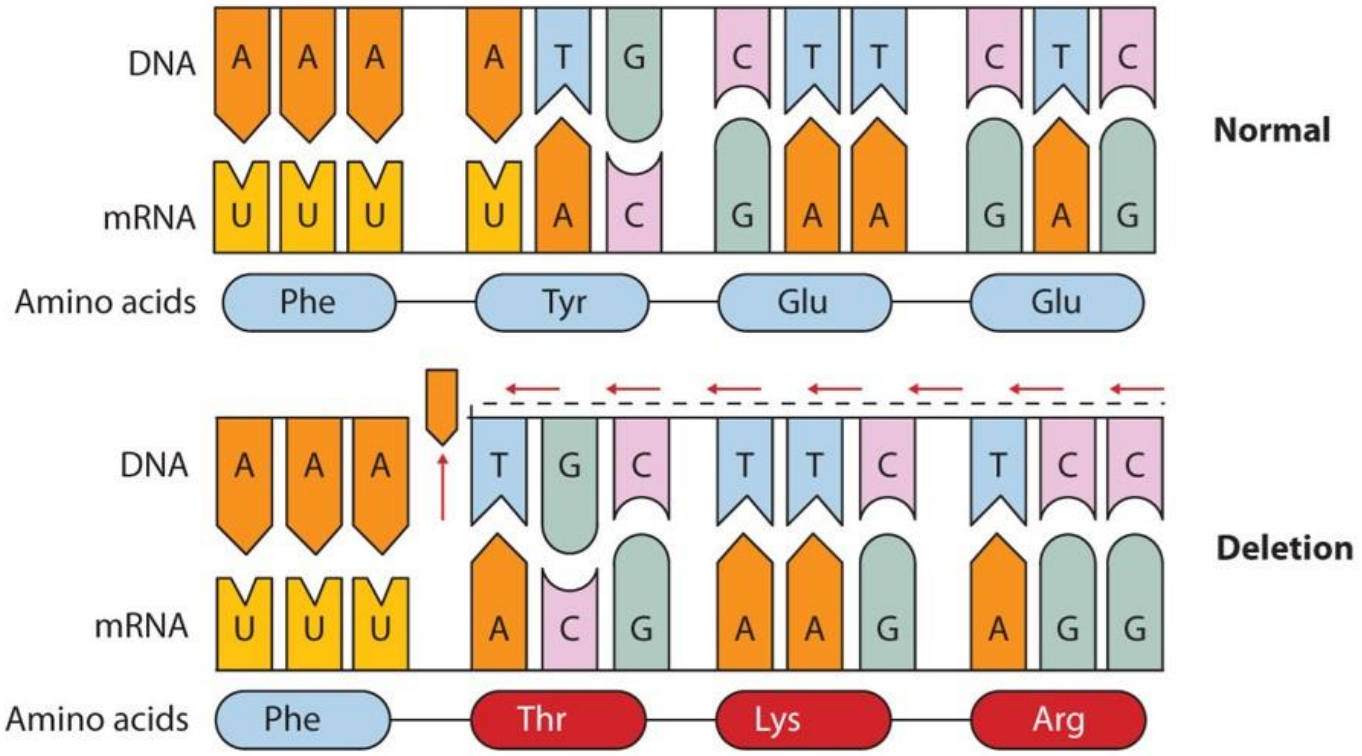
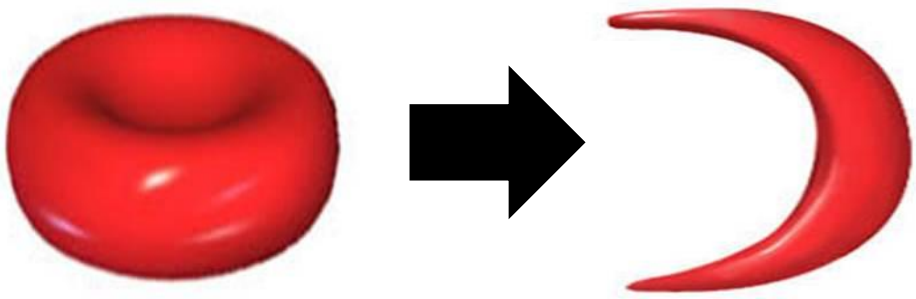



Figure 3

# Base Substitution: Missense Mutation (Sickle Cell Anaemia)

- Base substitution that **changes original codon to another codon** which codes for **different amino acid**
- As a result, less active protein is produced
- E.g: **Sickle cell anaemia** is genetic disorder caused by base substitution (contain abnormal hemoglobin)



Normal shape RBC  Sickle shape RBC

# i) Base Substitution: Missense Mutation (Sickle Cell Anaemia)

- In DNA, **triplet base** on DNA **3'- CTC- 5'** is **replaced** with **3'- CAC – 5'.**
- **Single** Base **T** of DNA is replaced with base **A**
- Causes changed the mRNA it encodes, leads to change occur at codon no. 6 of β-chain of hemoglobin.
- **Codon 5'-GAG- 3'** is replaced with codon **5'-GUG-3'.**
- **Glutamic acid** is replaced with **Valine** in both β-chain of hemoglobin.
- Abnormal **Hemoglobin/ Hb-S/ $Hb^S$** is formed.
- Causes abnormal sickle shaped red blood cell.

Tips from LKC
- If state the **sequence**, must use term **triplet base**
- If state the **base** only, must use term **single base**

Tips from LKC
- abnormal haemoglobin S ✗
- Hb-S/ $Hb^S$ / Abnormal haemoglobin / Haemoglobin S √

**Learning Outcome:**
**7.2 d) Explain base substitution (e.g : sickle cell anaemia as missense mutation) . (Tutorial)**

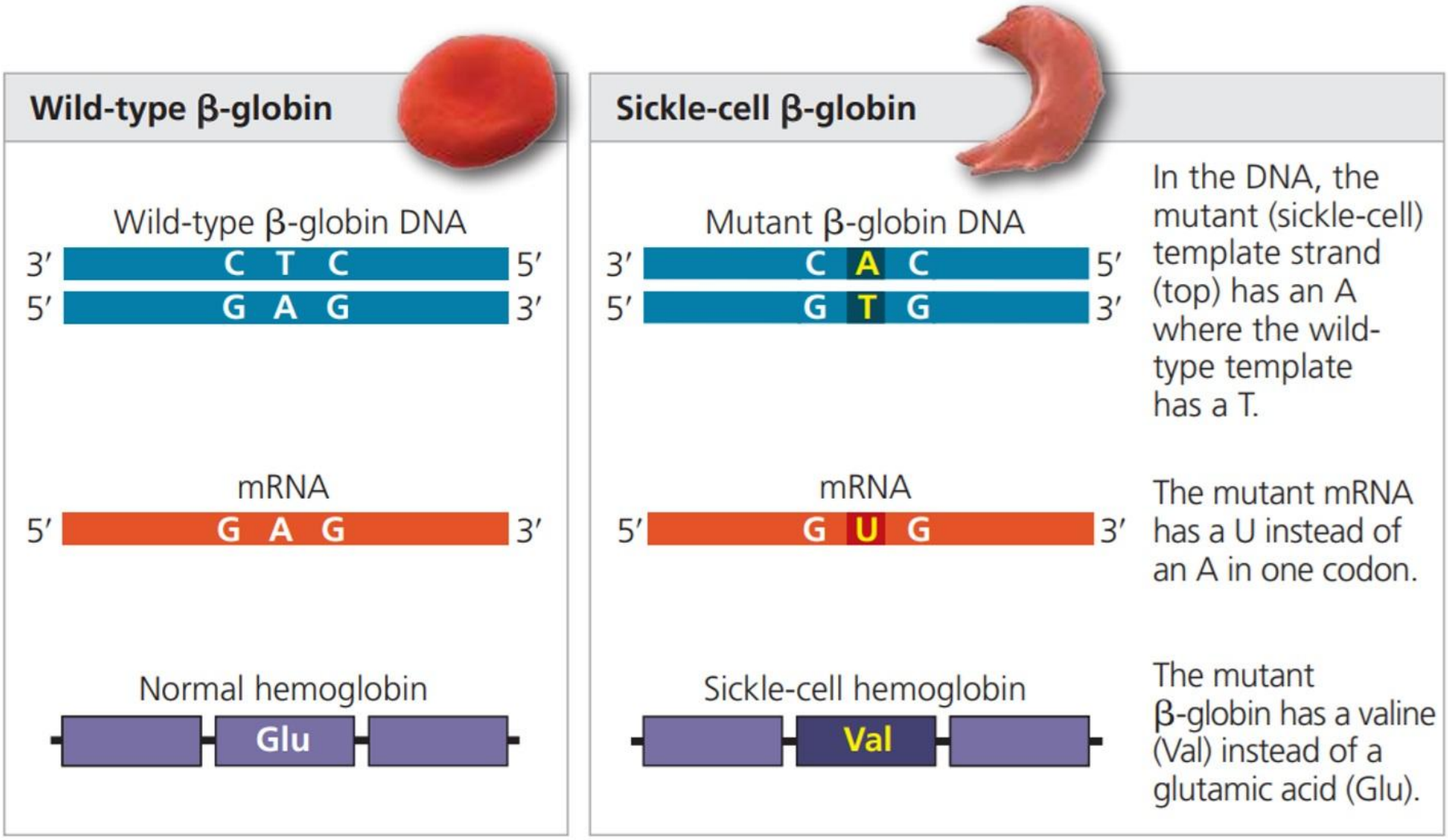



**Figure 4**

# i) Base Substitution: Missense Mutation (Sickle Cell Anaemia)

- These **sickled red blood cells** stuck in the blood capillaries, blocking the blood flow.
- Hemoglobin no longer efficient in transporting oxygen.
- Sickle-cell anaemia patients usually suffer from **heart problems, kidney failure, abdominal pains, paralysis and premature death.**

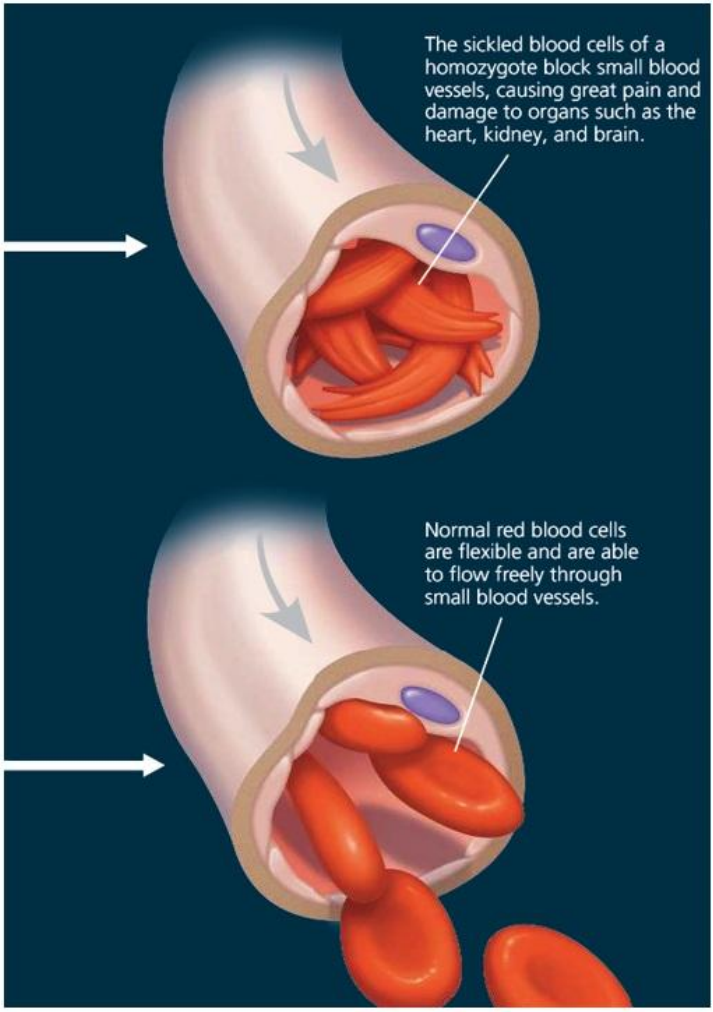



Figure 5

**Learning Outcome:**
**7.2 e) State the effect of base substitution (missense, nonsense and silent mutation) and base insertion and base deletion (frameshift mutation). (Lecture)**

# Effect of base substitution

## Missense mutation

- Substitution of base that change the codon into **different codon.**
- The altered codon codes for **different amino acid.**
- May have **little effect on protein** or cause **major change in a protein**.

## Silent mutation

- Substitution of base that change the codon
- But altered codon is code for the **same amino acid.**
- As a results, **same** protein is produced.

## Nonsense mutation

- Substitution that change a codon to **stop codon (5'-UAA-3' / 5'-UAG-3' / 5'-UGA-3').**
- Leading to a **shorter polypeptide and nonfunctional protein.**

# Effect of base substitution: Missense mutation

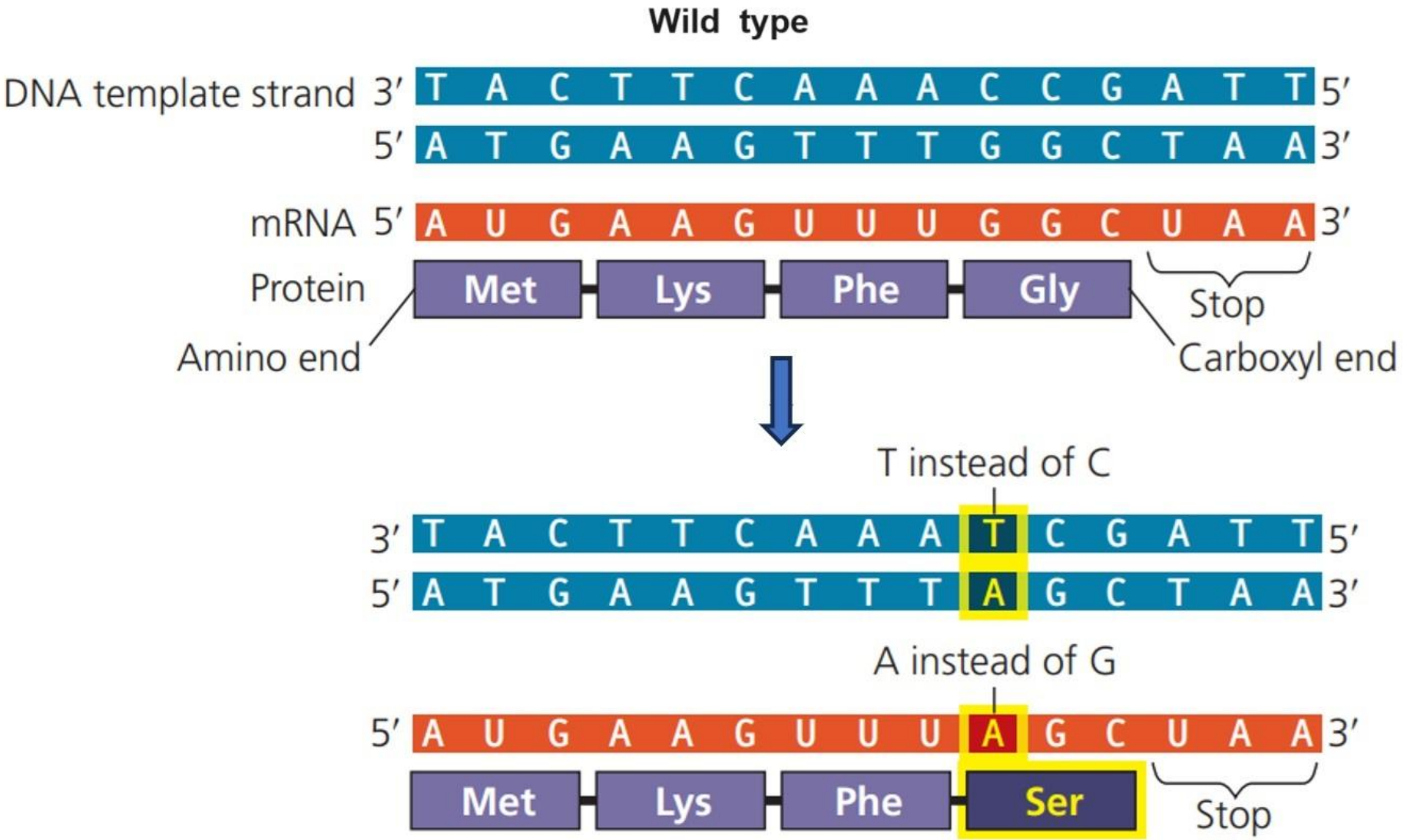





Figure 6

**Learning Outcome:**
**7.2 e) State the effect of base substitution (missense, nonsense and silent mutation) and base insertion and base deletion (frameshift mutation). (Lecture)**

# Effect of base substitution: Silent mutation

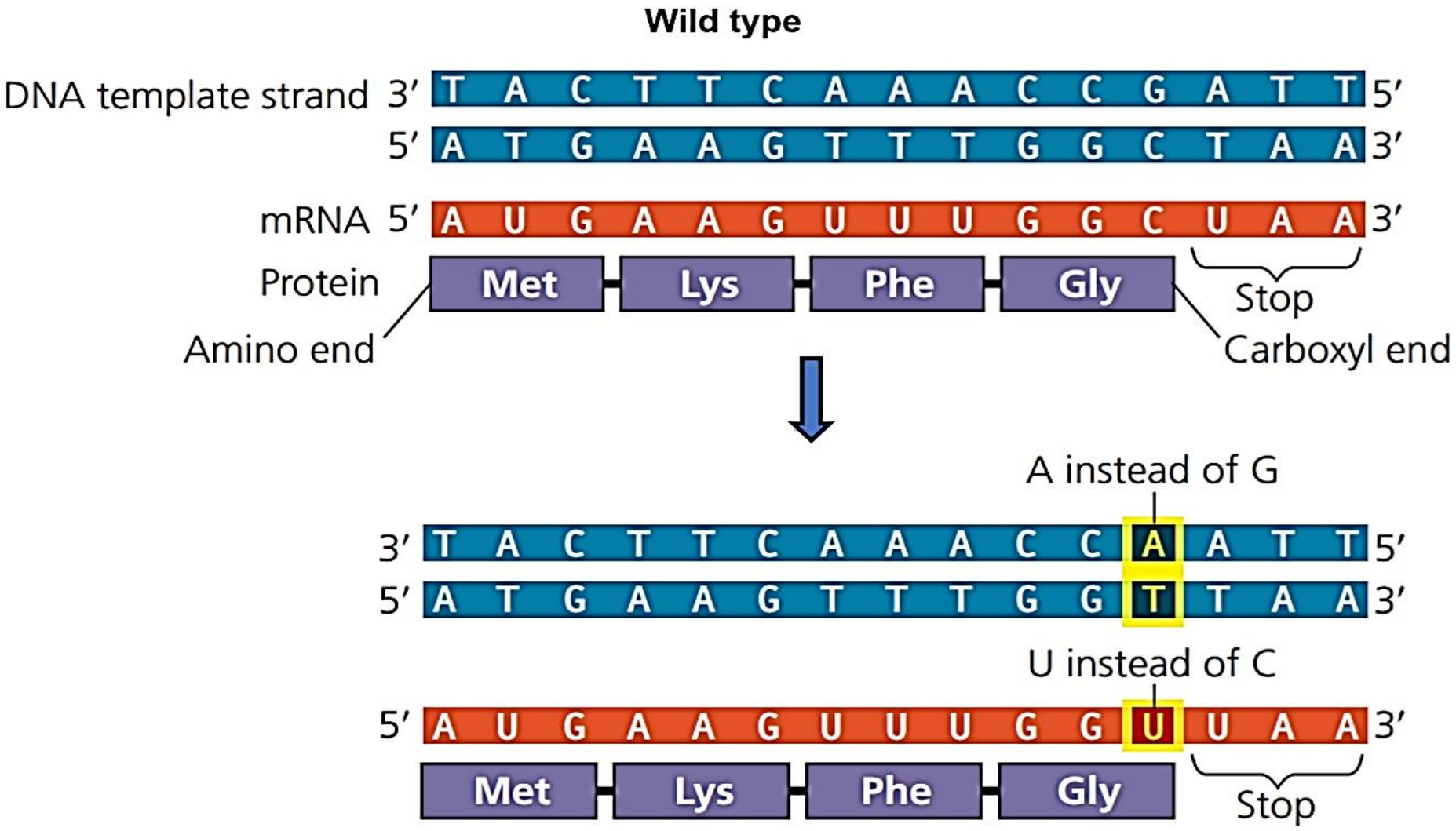



**Figure 7**

**Learning Outcome:**
**7.2 e) State the effect of base substitution (missense, nonsense and silent mutation) and base insertion and base deletion (frameshift mutation). (Lecture)**

# Effect of base substitution: Nonsense mutation

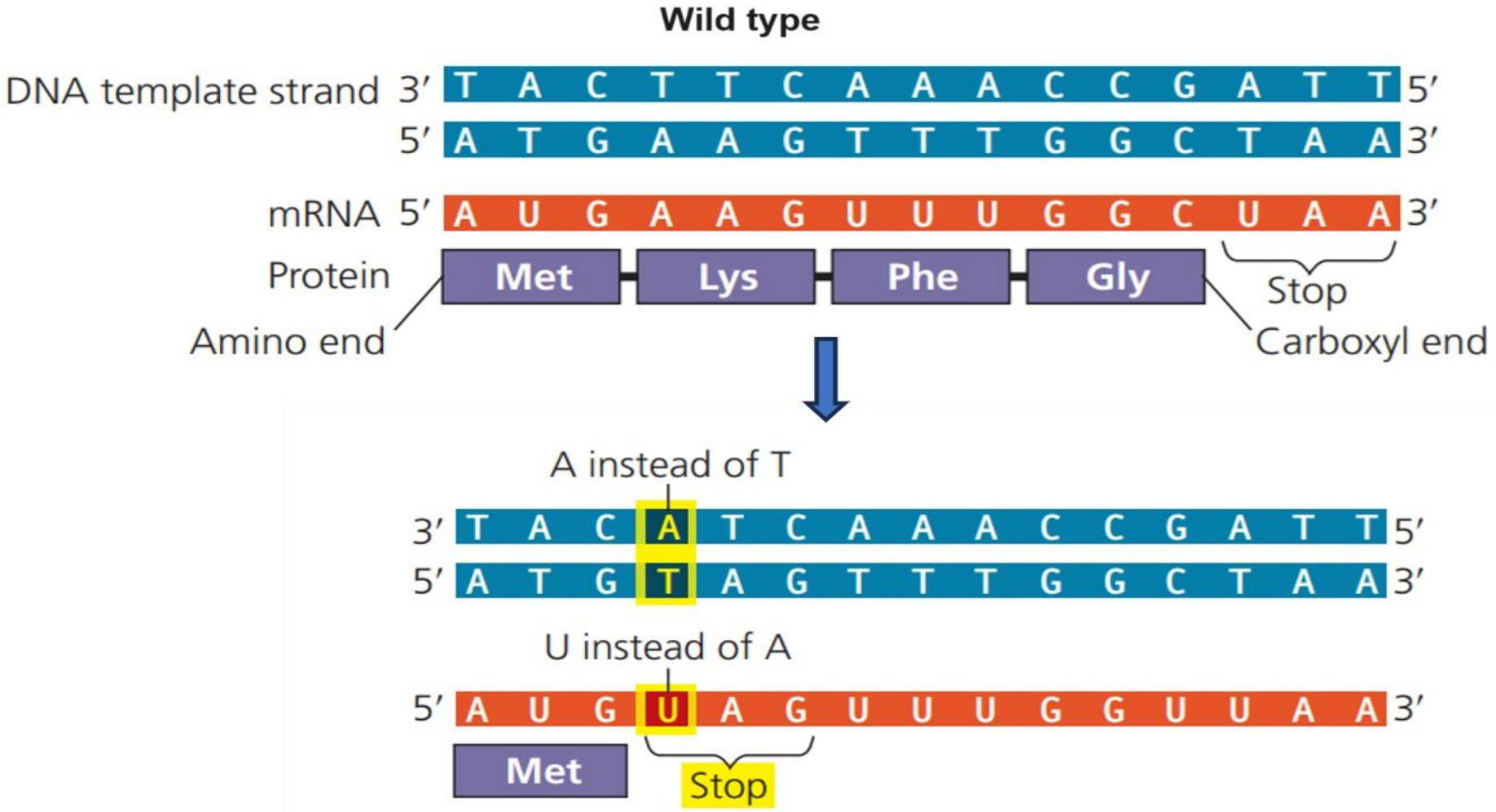



**Figure 8**

# Effect of Base Insertion and Base Deletion



**Frameshift Mutation**

# Frameshift Mutation

- Involve **insertion** or **deletion** of one or more nucleotide base pairs (but **not in a multiple of three**) into the DNA sequence of gene.
- Cause **alteration of reading frame of the DNA nucleotide base sequence after the point of mutation.**
- Shifting the reading frame of codon mRNA @ all the nucleotides after insertion/deletion point will be improperly grouped into codons.
- Results in an entirely **different sequence of amino acid of polypeptide chain** from the point of mutation.
- Causing **different protein** / **nonfunctional protein** to be produced.
- Effect : **Usually harmful / severe effect to organism**

# Base Insertion As a Frameshift Mutation

- One or more nucleotide base pairs is **inserted** into the DNA sequence of a gene.
- This causes the whole DNA base sequences (at and after the point of mutation) to be **changed/altered**
- Reading frame of **codons of mRNA is changed/altered**
- Resulting polypeptide consists of an entirely **different sequence of amino acids,** producing a **totally different proteins**
- The effect is very serious / harmful.

# Base Insertion As a Frameshift Mutation

Normal DNA : 3'- CTC CAA GGA TTT GGA -5'

Normal mRNA : 5'- GAG GUU CCU AAA CCU-3'

Polypeptide glu - val - pro - lys - pro

**Base insertion** (addition of nucleotide base C)

DNA : 3'-CTC CAA GGA CTT TGG A..-5'

mRNA : 5'-GAG GUU CCU GAA ACC U..-3'

Polypeptide glu - val - pro - glu - thr

# Base Insertion As a Frameshift Mutation

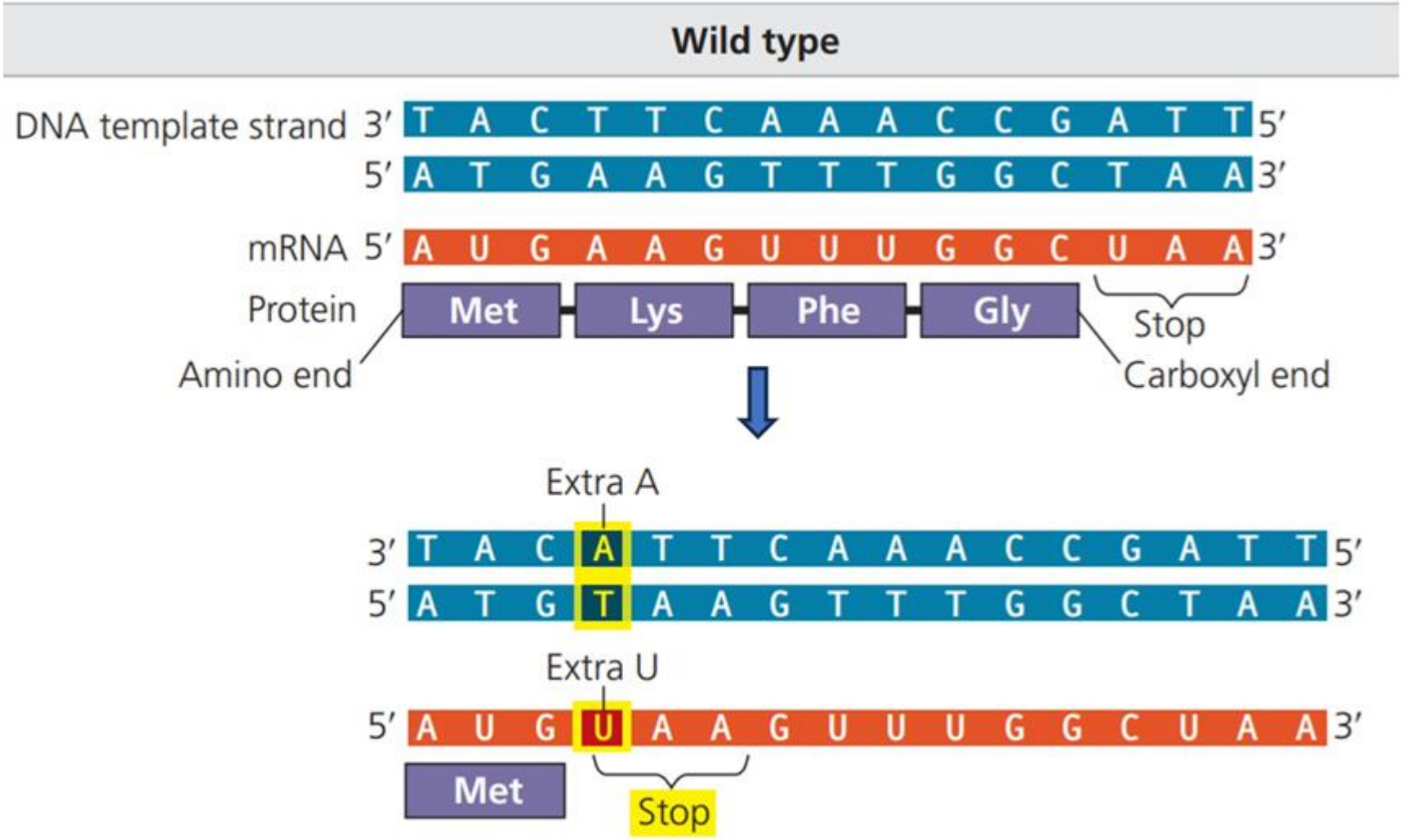



**Figure 9**

# Base Deletion As a Frameshift Mutation

- One or more nucleotide base pairs is **deleted** from the DNA sequence of a gene.
- This causes the whole DNA base sequences **at** and **after** the point of mutation to be **changed/altered**
- Reading frame of **codon of mRNA is changed/altered**
- Resulting polypeptide consists of an entirely **different sequence of amino acids,** producing a **totally different proteins**
- The effect is very serious / harmful.
- E.g.: **Thalassaemia**

# Base Deletion As a Frameshift Mutation

Normal DNA : 3'-CTC CAA GGA TTT GGA-5'

Normal mRNA : 5'-GAG GUU CCU AAA CCU-3'

Polypeptide glu - val - pro - lys - pro

<u>Base deletion</u> (deletion of nucleotide base C)

DNA : 3'-CTC AAG GAT TTG GA..-5'

mRNA : 5'-GAG UUC CUA AAC CU..-3'

Polypeptide glu - phe - leu - asn

# Base Deletion As a Frameshift Mutation

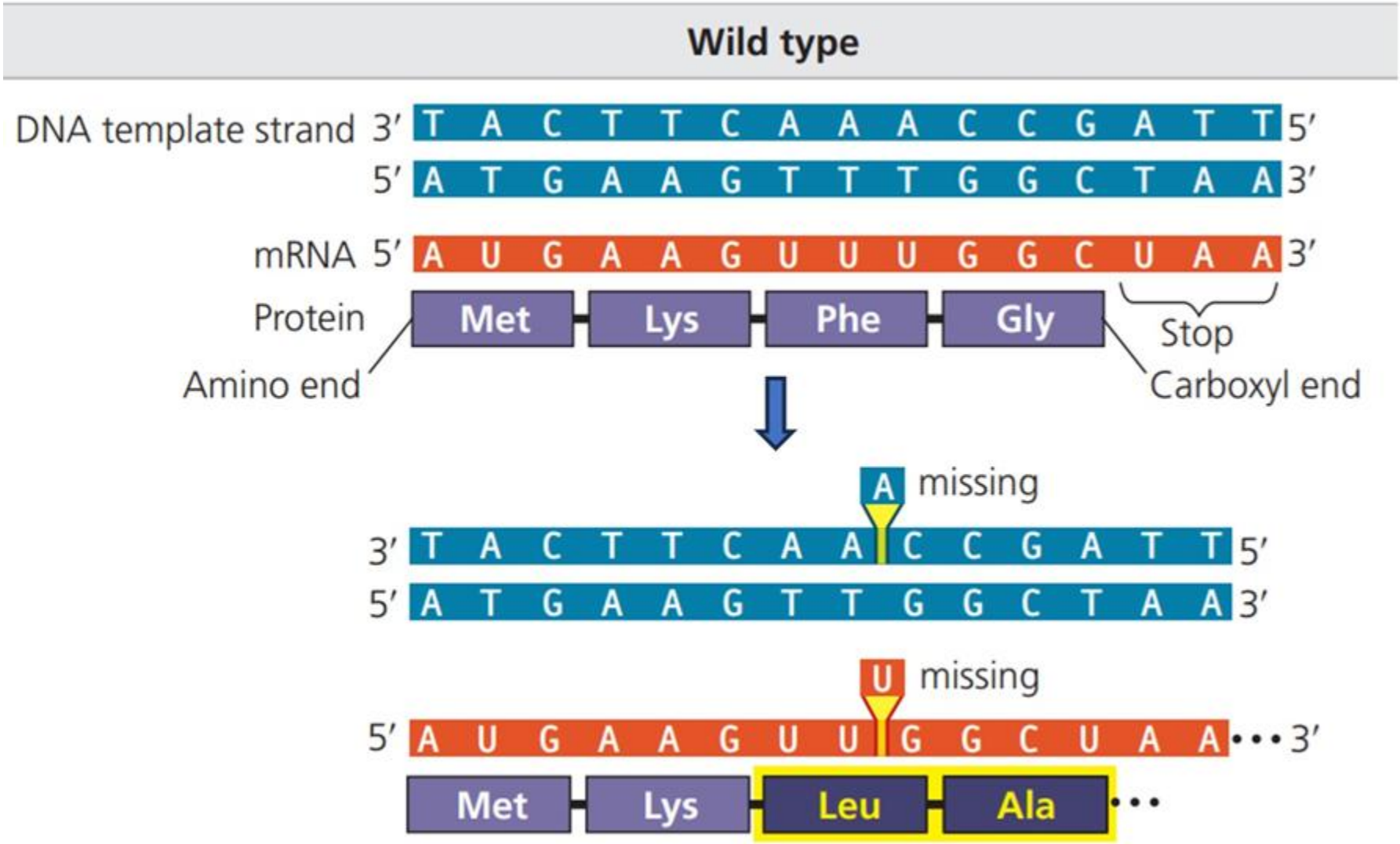



Figure 10

# LEARNING OUTCOMES

## 7.3 Chromosomal mutation

(a) Define chromosomal mutation (C1 - Lecture)

(b) State two types of chromosomal mutation (C1 - Lecture)

i. Changes in chromosomal structure/ chromosomal aberration

ii. Changes in chromosomal number

(c) Explain changes in chromosomal structure (C3 -Lecture)

(d) Explain types of chromosomal aberration (C3 - Tutorial)

# LEARNING OUTCOMES

## 7.3 Chromosomal mutation

e) Explain alterations of chromosome number (C3 -Tutorial)

f) State the types of the alterations: (C1 - Lecture)

i. Aneuploidy

ii. Polyploidy

g) Explain aneuploidy (C3 - Lecture)

h) State aneuploidy effect on autosomal chromosome and sex chromosome (C1 - Lecture)

# LEARNING OUTCOMES

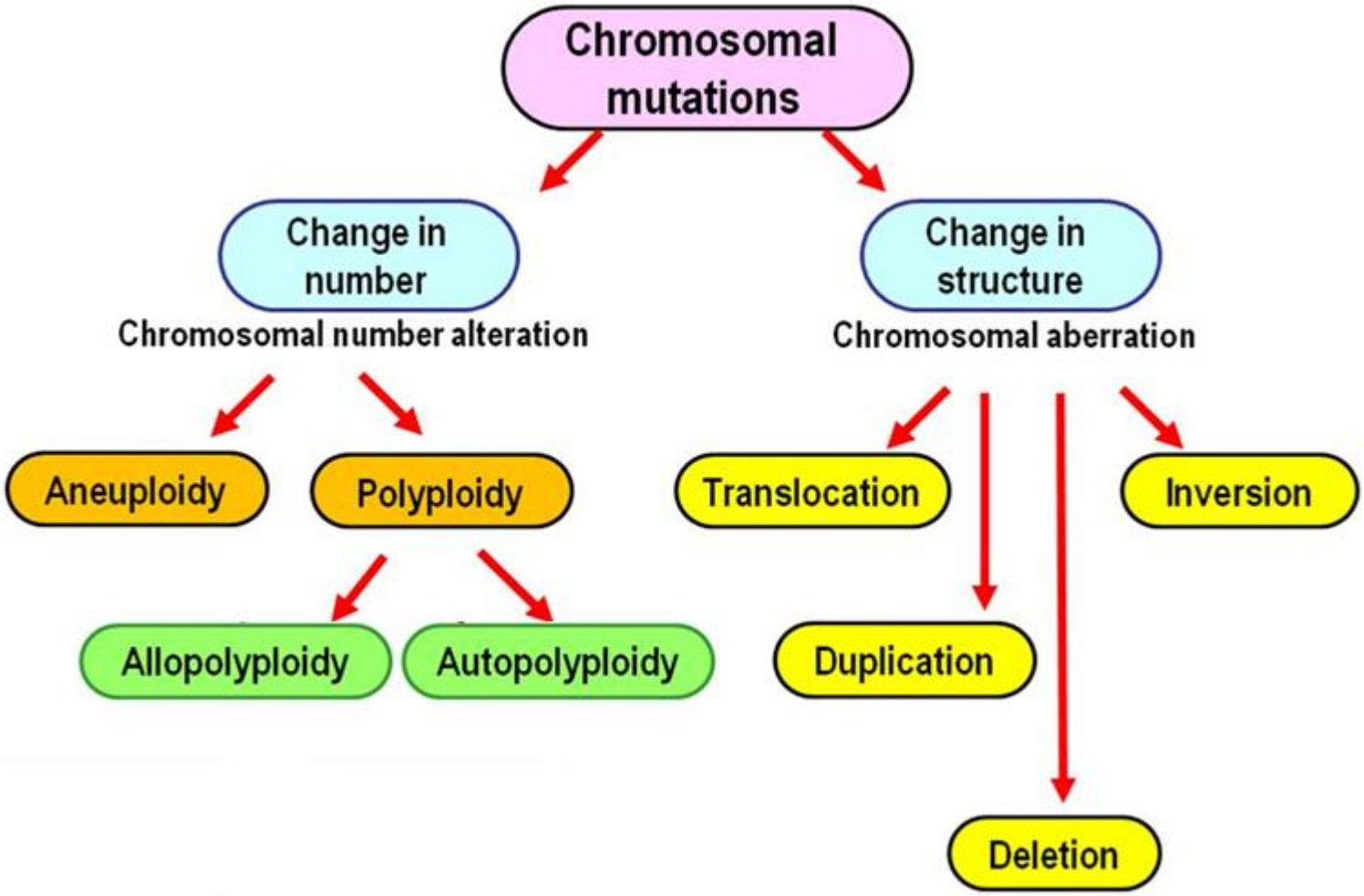
## 7.3 Chromosomal mutation

i) Explain autosomal abnormalities & their effects: (C3 - Tutorial)
   i. Monosomy (monosomy 21)
   ii. Trisomy (Down syndrome/ trisomy 21)

j) Explain sex chromosomal abnormalities. (C3 - Tutorial)
   i. Klinefelter syndrome (47, XXY)
   ii. Turner syndrome (45, XO)

k) Explain polyploidy. (C3 - Tutorial)
   i. Autopolyploidy
   ii. Allopolyploidy

# CHROMOSOMAL MUTATION



Learning Outcome:
7.3 a) Define chromosomal mutation (Lecture)
7.3 b) State two types of chromosomal mutation (Lecture)

# CHROMOSOMAL MUTATION

**Definition**:
Mutations that cause abnormalities in **chromosomal structure** or changes in **chromosome number**

2 types:

i. Chromosomal aberration
ii. Chromosomal number alteration

# CHROMOSOMAL ABERRATION

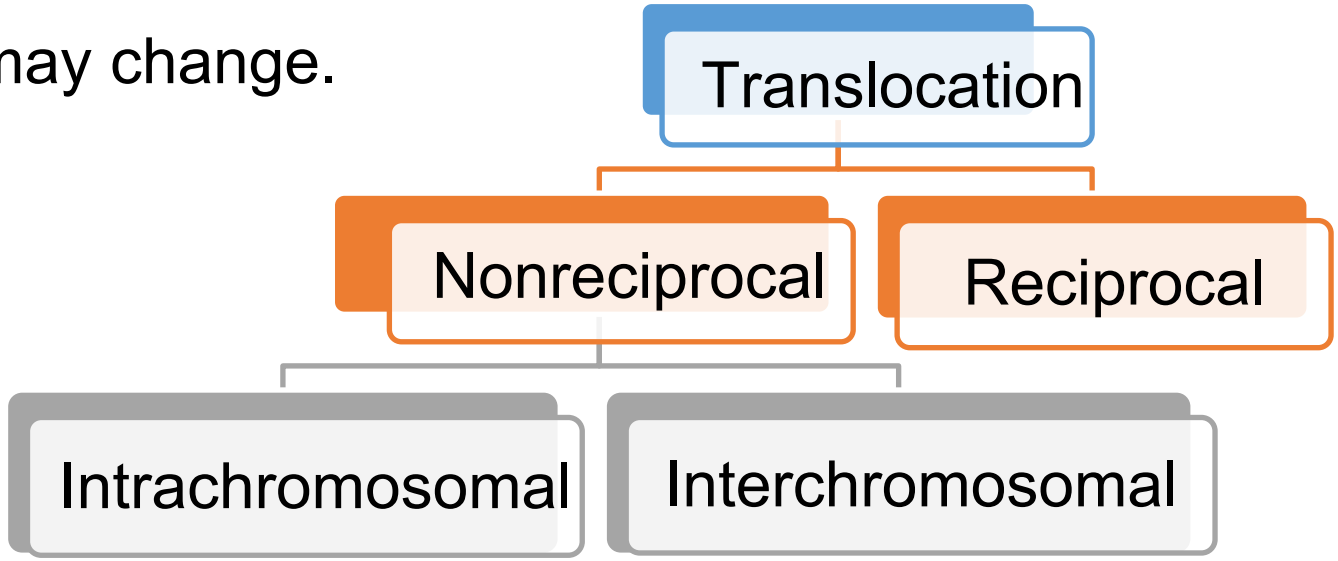
- **Changes to chromosomal structure** which result in abnormalities in the structure of chromosome.

- Four types of chromosomal aberration:

1) Translocation
2) Deletion
3) Inversion
4) Duplication

# 1. TRANSLOCATION

- Involves a **segment of a chromosome breaking off and reattached to another part of the same chromosome or other nonhomologous chromosome**.
- Changes in the position of the genes involved
- There is no gain or loss of genetic materials; no change in genotype.
- Phenotype may change.




**Tips from LKC**
Make sure **correct definition** and state the **types** of translocation

# i. Nonreciprocal translocation

- Involves only **one chromosome translocate a segment** to another region of the **same chromosome or another nonhomologous chromosome**.

## Intrachromosomal translocation

Segment of one chromosome **translocate to another region of the same chromosome**

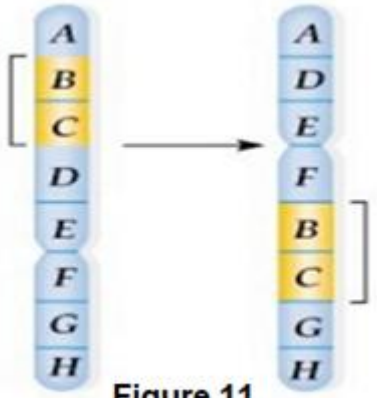



Figure 11

## Interchromosomal translocation

Segment of one chromosome **translocate to another nonhomologous chromosome**

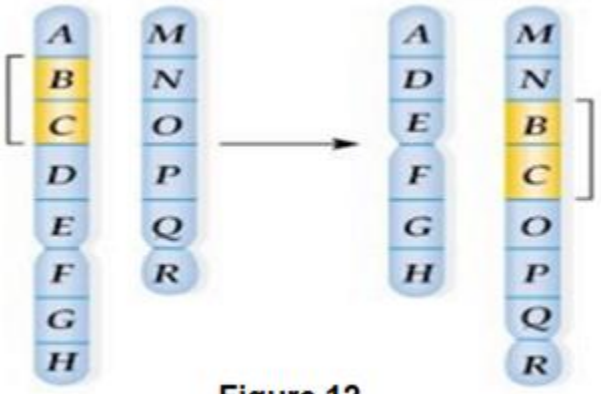



Figure 12

# ii. Reciprocal translocation

- Involves **exchanges of segments** between **two nonhomologous chromosomes.**
- Producing two new nonhomologous chromosome
- The most common type of translocation
- e.g: **Chronic Myelogenous Leukemia**

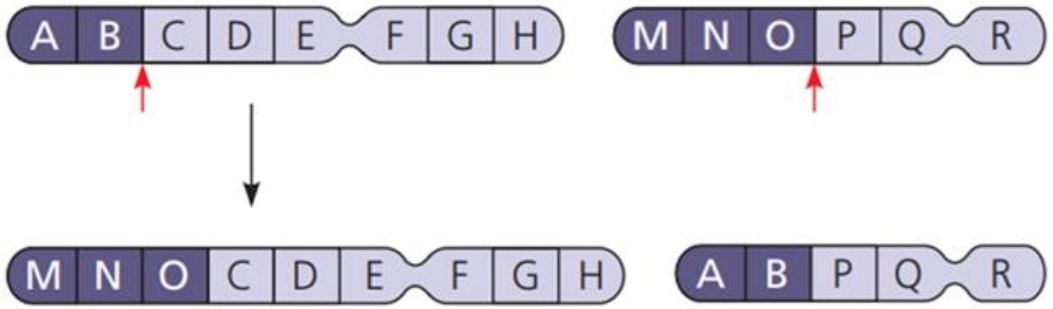



Figure 13

**Tip LKC :** Spelling must be correct
i.e. Chronic Myelogenous Leukemia//Chronic Myeloid Leukemia

# ii. Reciprocal translocation

## Chronic Myelogenous Leukemia

- Occurs when a reciprocal translocation happens during mitosis of cells that are precursors of white blood cells.
- **A large portion of chromosome 22** exchange with **a small fragment of chromosome 9** produces a **shortened chromosome 22** called **Philadelphia chromosome**

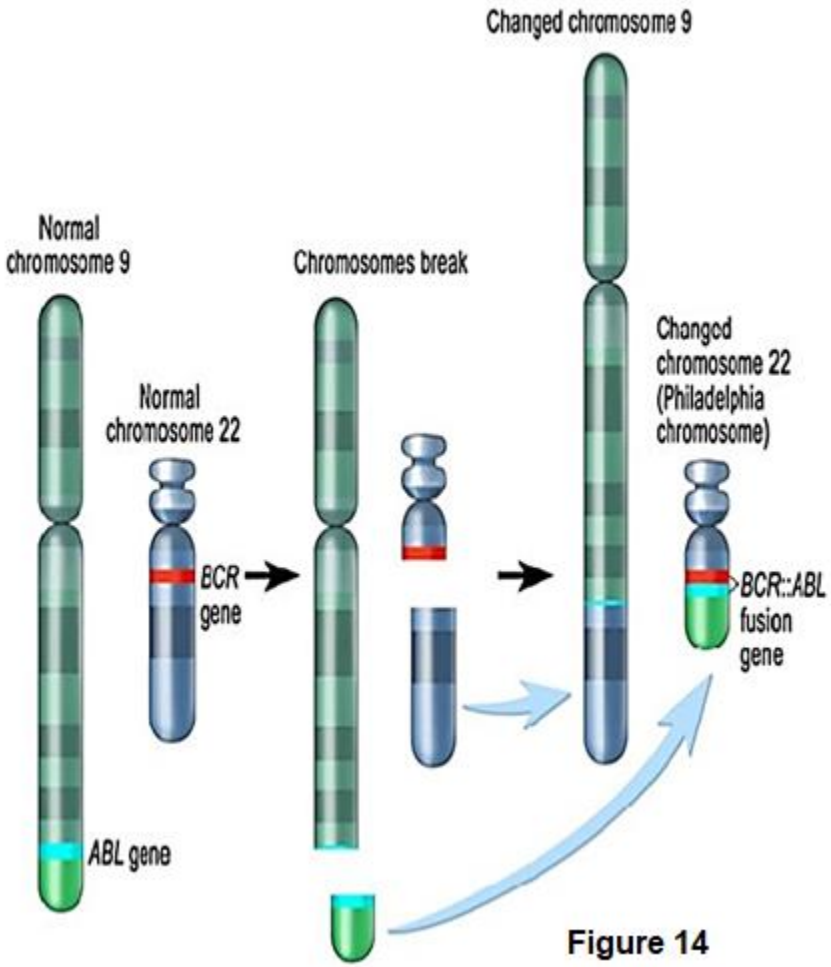



Figure 14

# ii. Reciprocal translocation

## Chronic Myelogenous Leukemia

- The exchange causes cancer by creating a new "fused" gene (oncogene) that leads to uncontrolled cell cycle progression
- Immature white blood cells (myeloblasts) accumulate in blood and bone marrow
- Overgrowth of myeloblasts impairs the development of other blood cells, leading to a shortage of red blood cells and platelets

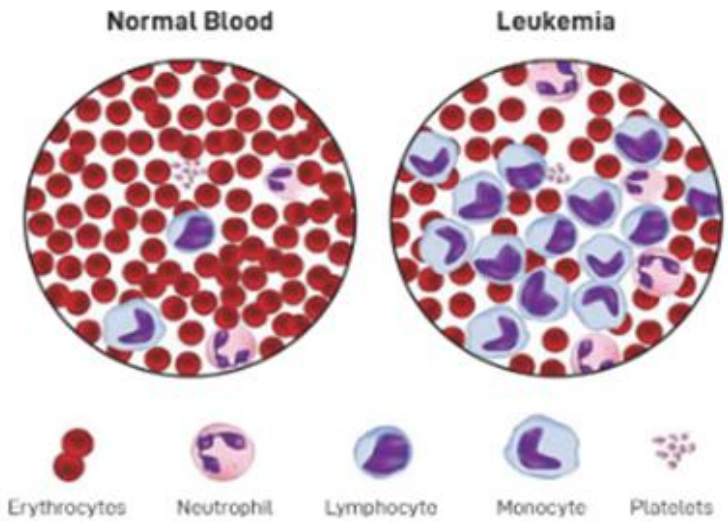



Figure 15

# ii. Reciprocal translocation

Symptoms of Chronic Myelogenous Leukemia:

- Excessive tiredness (fatigue)
- Fever
- Weight loss
- Enlarged spleen

# 2. DELETION

- **A segment of chromosome is <u>removed/lost</u>.**
- The affected chromosome missing certain genes and becomes shorter.
- If deletion affects the gene on both homologous chromosomes, the effect is harmful/ lethal.

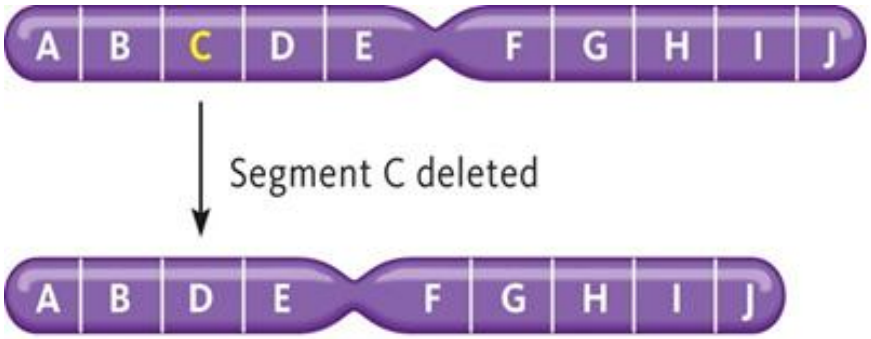
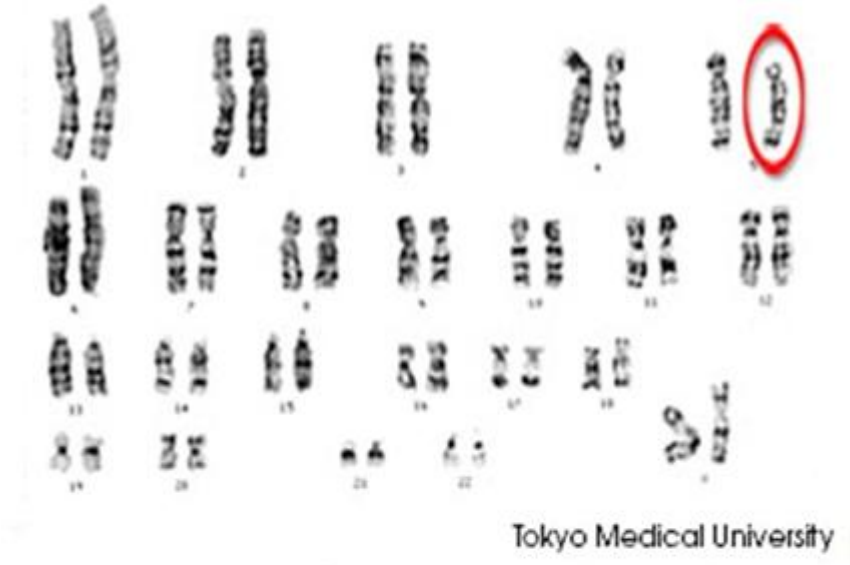



**Figure 16**

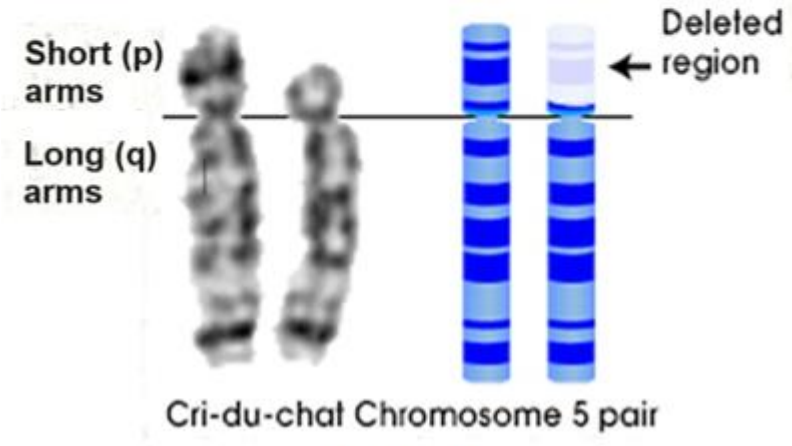
# 2. DELETION

e.g: ***cri du chat* syndrome**

- Caused by the **loss of a small part of the short arm of chromosome 5**.



Tokyo Medical University




Cri-du-chat Chromosome 5 pair

Figure 17

# 2. DELETION

***cri du chat*** syndrome individuals:

- Small head with unusual facial features
- Severe intellectual disabilities
- Cry sounds like the mewing of a distressed cat
- Usually die in infancy/ early childhood

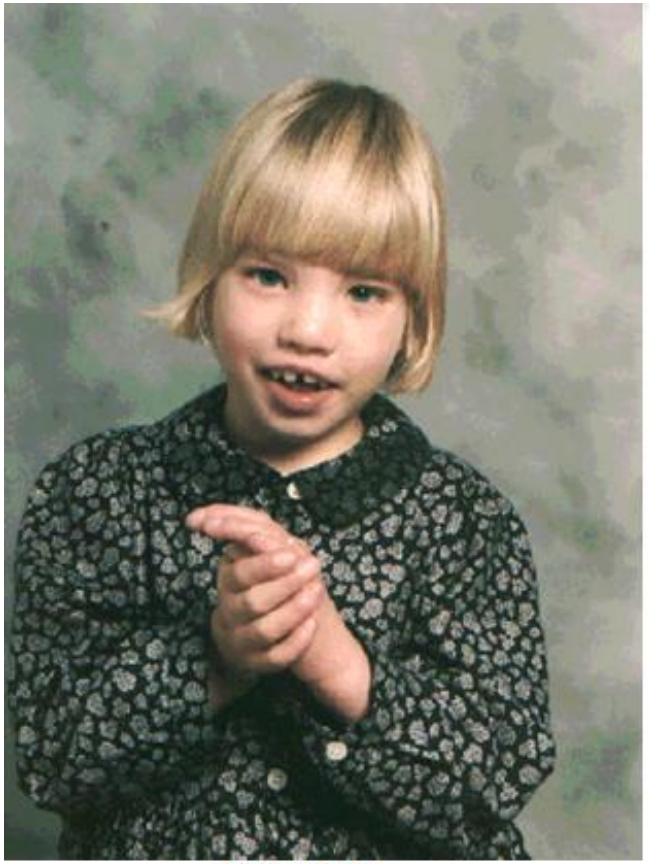


Figure 18

# 3. INVERSION

- The segment of a chromosome **breaks off and reattached in the opposite orientation** in the same chromosome, rearranging the linear gene sequence
- There is **no change in the genotype** but the **phenotype may change**.
- Two types:
  - Pericentric
  - Paracentric

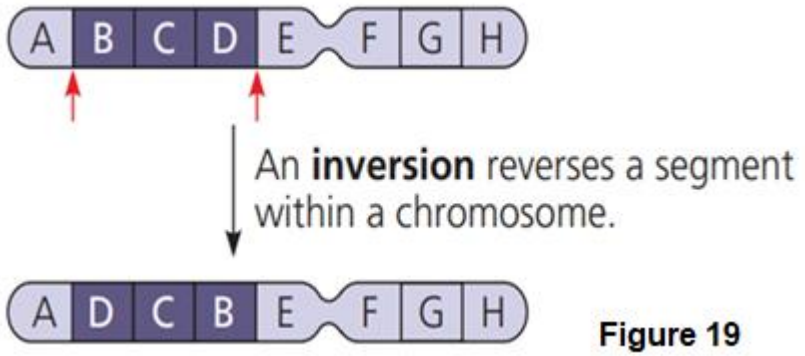



Figure 19

# 3. INVERSION

**Pericentric inversion**
Chromosome rearrangement in which a segment **include the centromere**.

**Paracentric inversion**
Chromosome rearrangement in which a segment **does not include the centromere**.

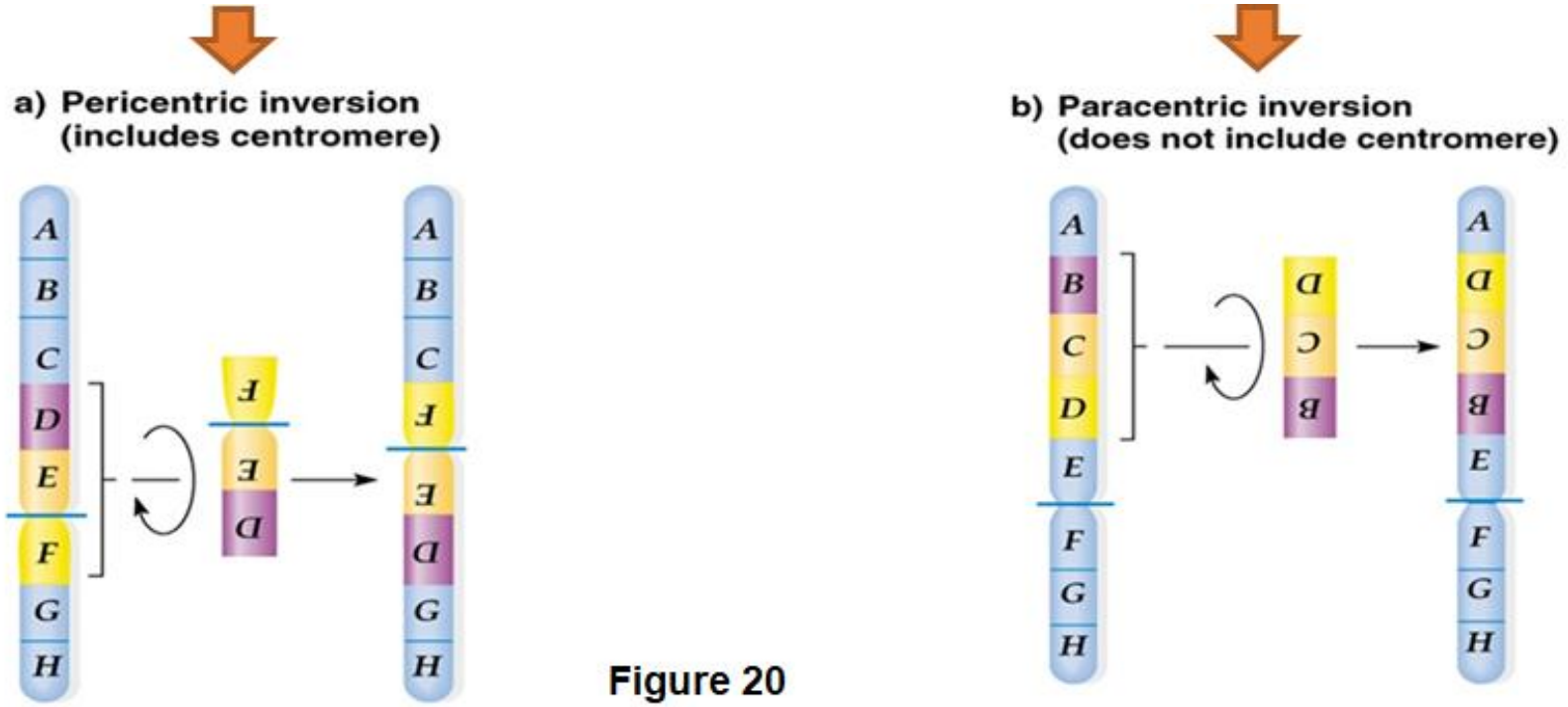



Figure 20

# 4. DUPLICATION

- **Repeated region/segment of a chromosome.**
- Results in, the presence of repetitive genes in the same chromosome.
- **Broken fragments of a chromosome become reattached as an extra segment to a sister or non-sister chromatid**
- May alter the function of the resulting protein.

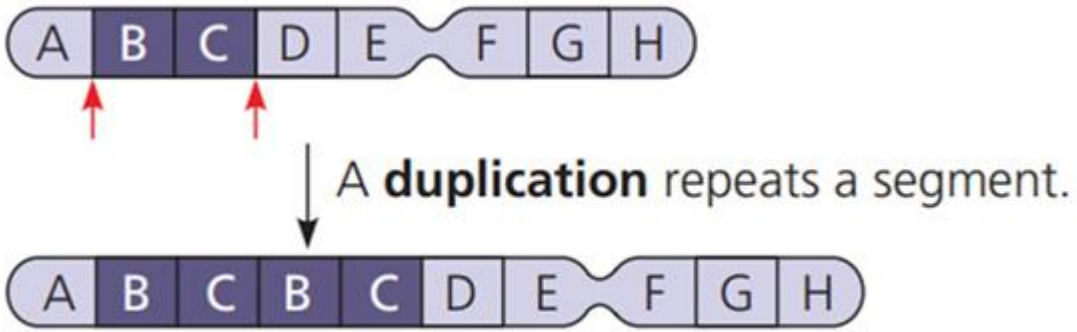
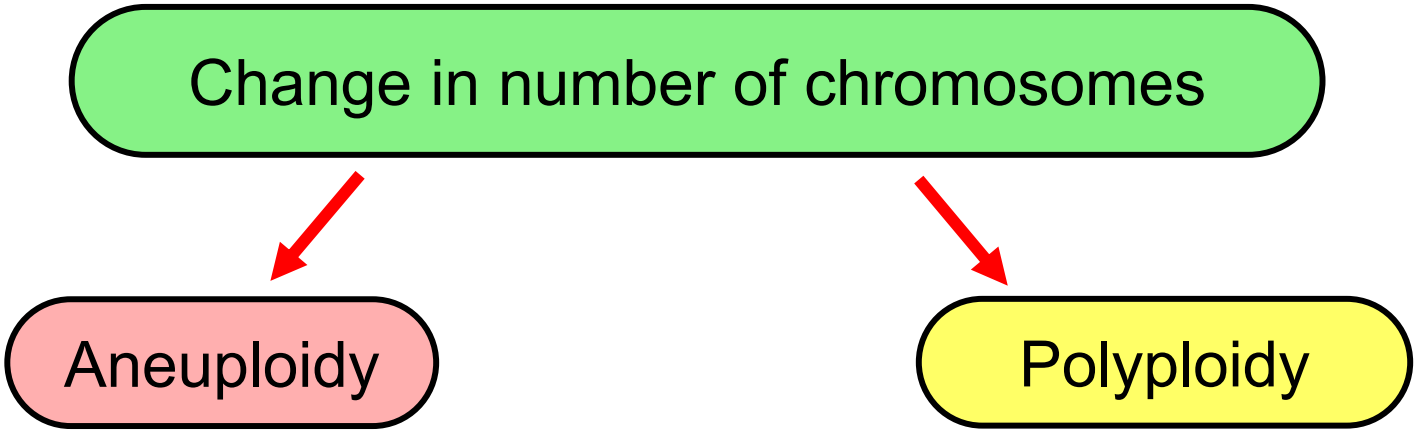



Figure 21

# ALTERATION OF CHROMOSOME NUMBER

- **Change in number** for **individual chromosomes** or the **whole set of chromosomes.**
- Usually the result of errors that occur during **meiosis** caused by **nondisjunction** during **meiosis.**
- Produce gamete with an abnormal number of chromosomes.

# ALTERATION OF CHROMOSOME NUMBER



# ANEUPLOIDY

- A condition in which the **addition or loss of one or more chromosomes** from the complete set of normal chromosome numbers in a diploid cell or individual.
- Can caused by **nondisjunction** during spermatogenesis/ oogenesis during meiosis I or meiosis II
- Can occur in **autosome** and **sex-chromosome**

# NONDISJUNCTION

- **The failure of homologous chromosomes to separate and move to opposite poles during anaphase I of meiosis I**

**OR**

**The failure of sister chromatids to separate and move to opposite poles during anaphase II of meiosis II**

- Caused by the failure of spindle fibres to form.
- Results in the production of gametes with an abnormal number of chromosome.

**Tip LKC**
**Must state :**
Failure in separation of **homologous chromosome** – during anaphase I
Failure in separation of **sister chromatids** – during anaphase II

# NONDISJUNCTION

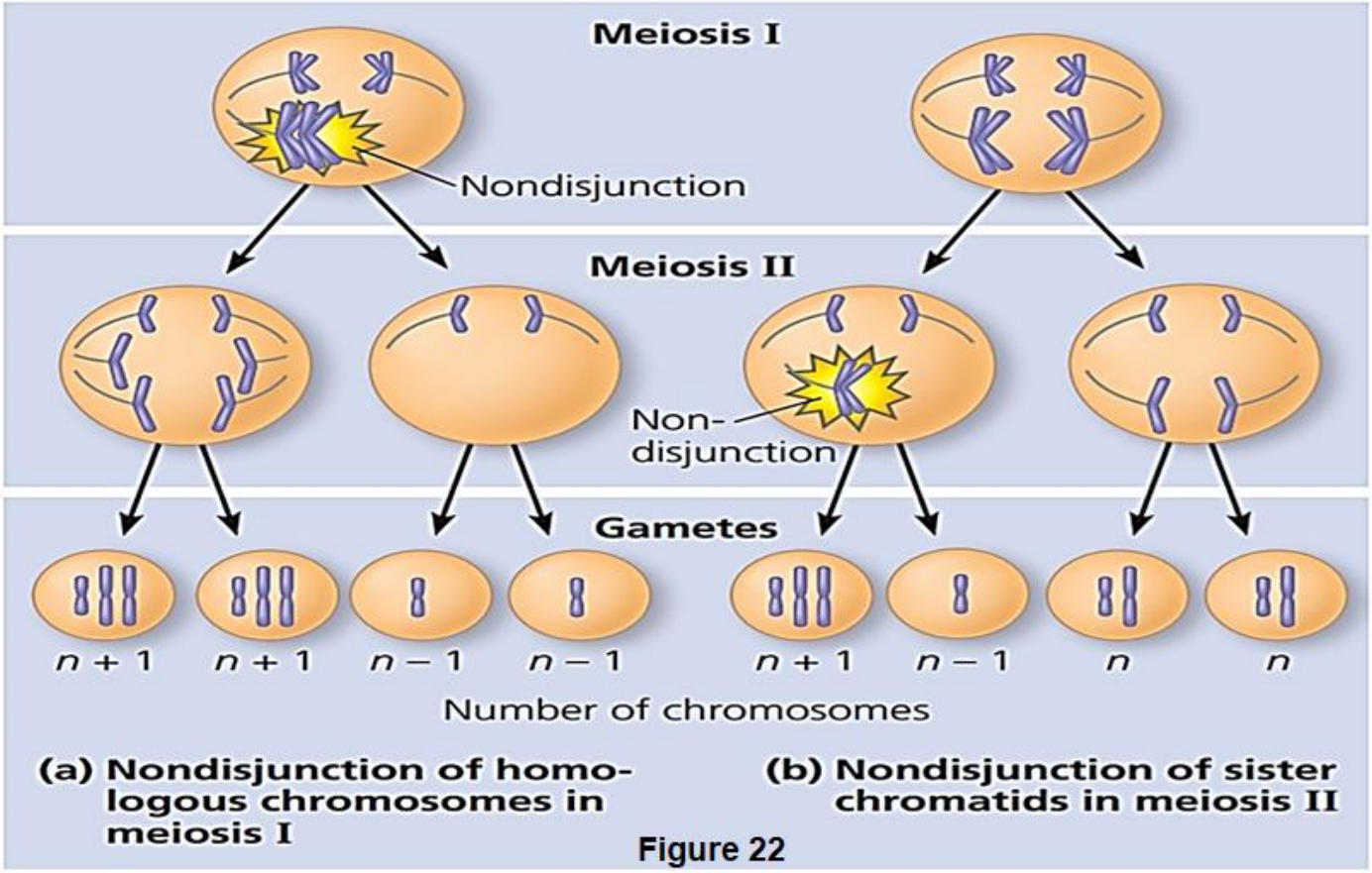



Figure 22

**Tip LKC :**

**Draw correct number and correct size of chromosome within cell**

# NONDISJUNCTION

- If fertilization occurs between normal gamete (n) and abnormal gamete (n -1)
  - ➢ produce abnormal zygote (2n–1)
  - ➢ Monosomy
- If fertilization occurs between normal gamete (n) and abnormal gamete (n + 1)
  - ➢ produce abnormal zygote (2n + 1)
  - ➢ Trisomy

# Nondisjunction of homologous chromosome (Anaphase I)

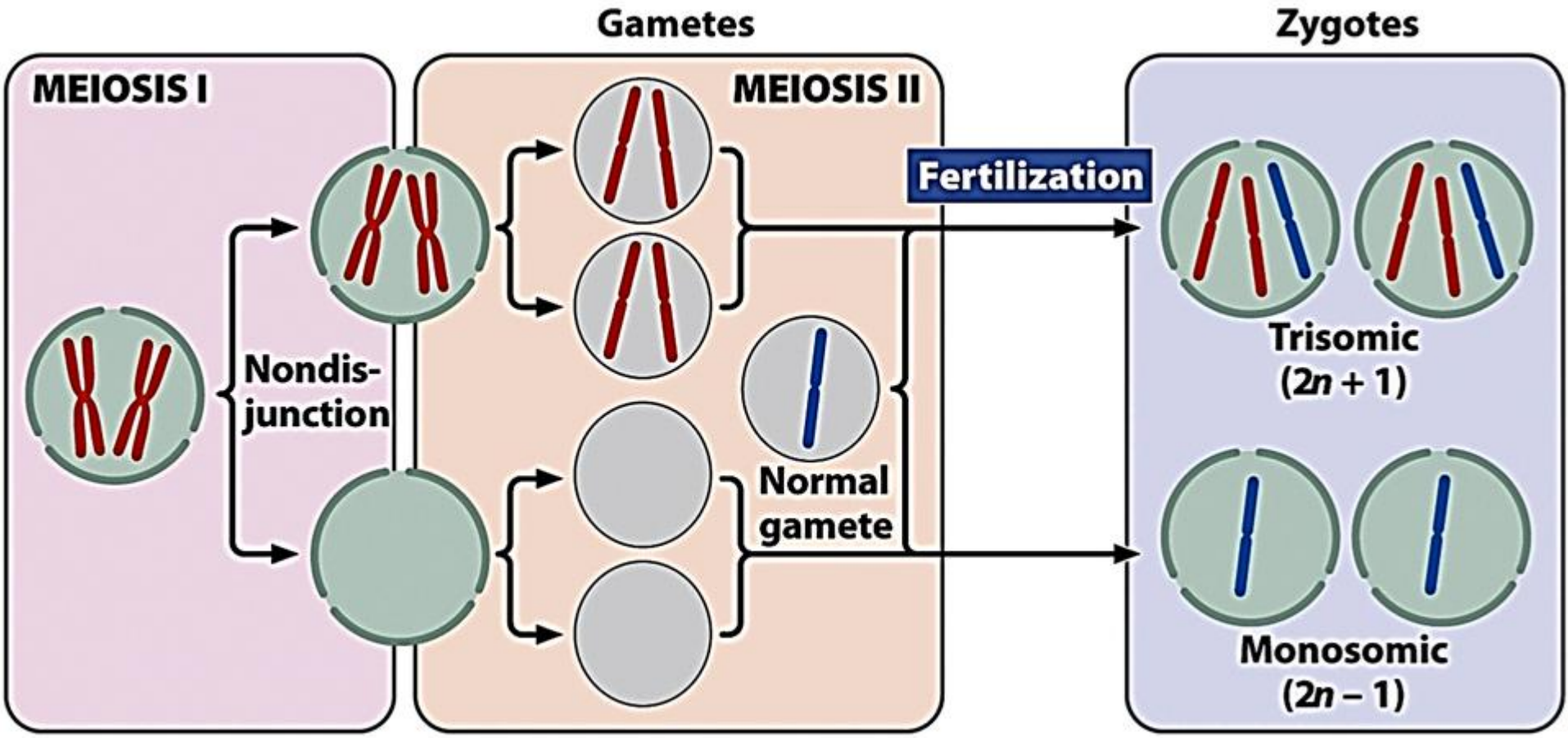



Figure 23

# Nondisjunction of sister chromatids (Anaphase II)

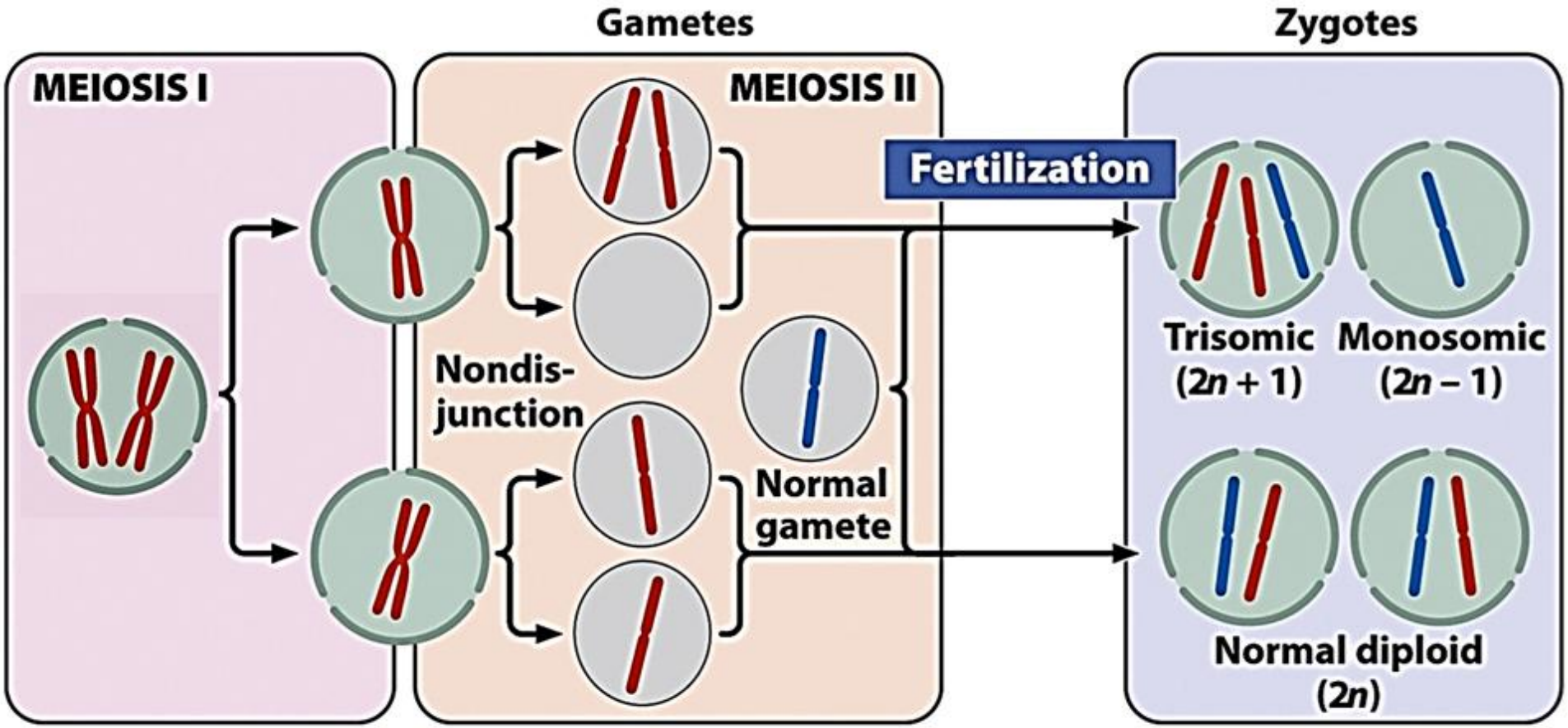



Figure 24

Learning Outcomes:
7.3 (g) Explain aneuploidy (Lecture)

# NONDISJUNCTION

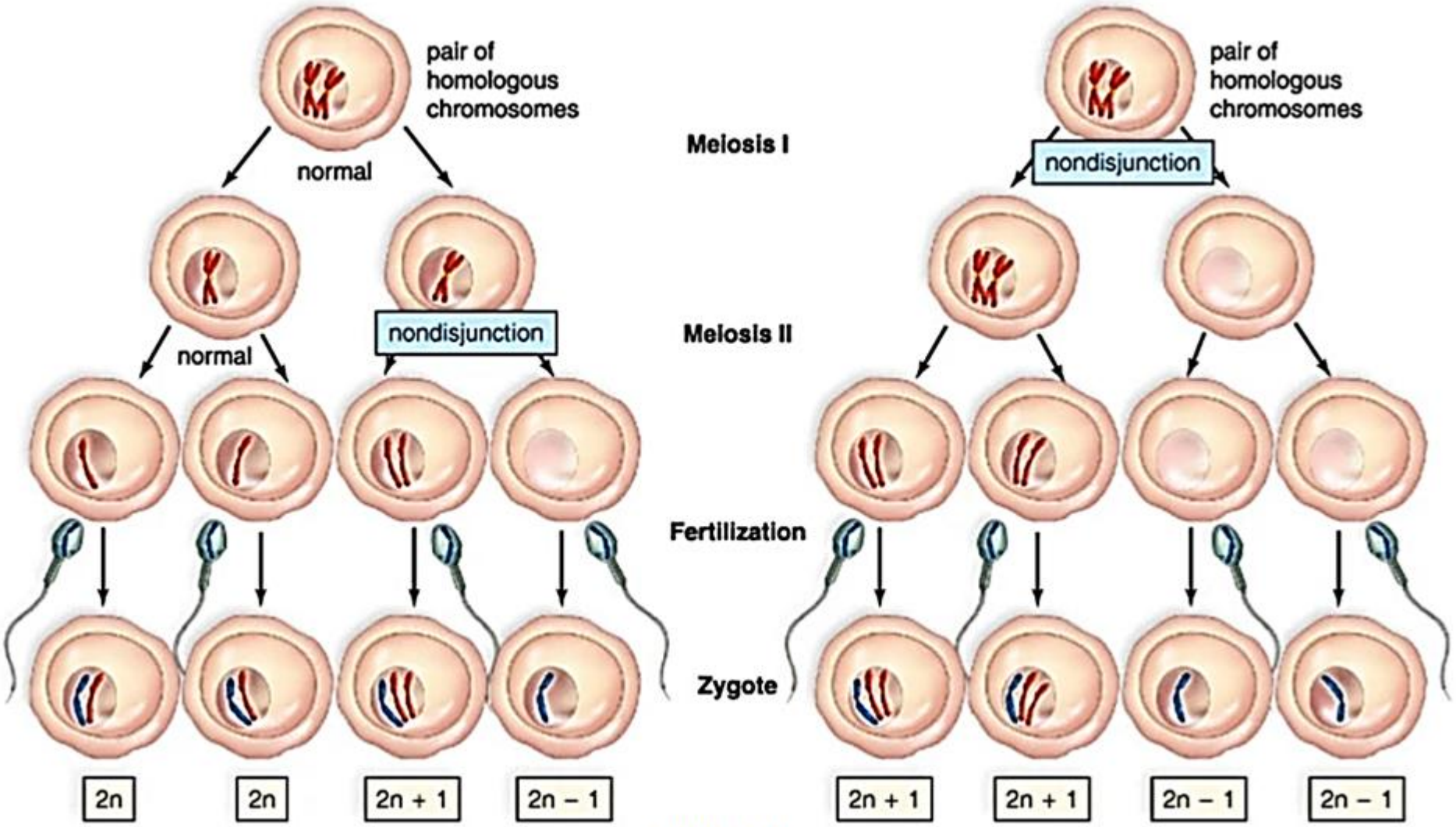
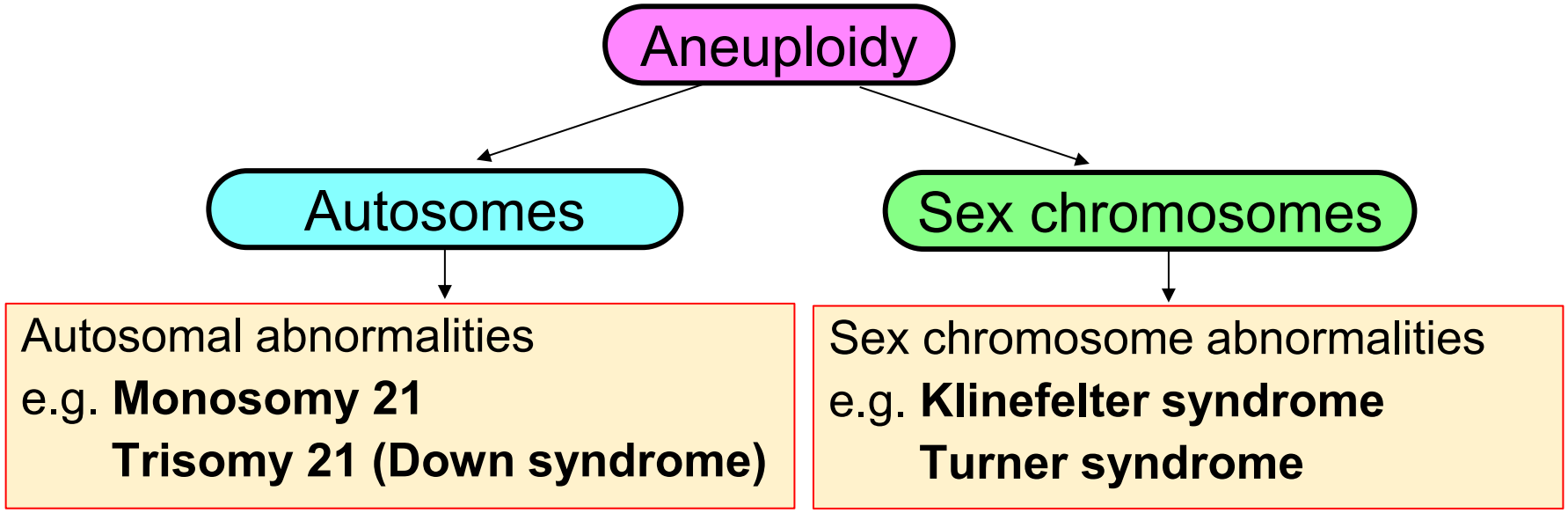



Figure 25

# ANEUPLOIDY

- Effect of Aneuploidy :



# AUTOSOMAL ABNORMALITIES

## Monosomy 21

- A diploid organism has only one chromosome 21;

  2n-1 (45 chromosomes)
- Fatal during early embryonic development.
- Rare disease




Figure 26

# AUTOSOMAL ABNORMALITIES

## How does monosomy 21 occurs?

- **Due to the nondisjunction of chromosome 21 during meiosis I or meiosis II**
- Failure of homologous chromosome to separate and move to opposite poles during anaphase I OR

  Failure of sister chromatids to separate and move to opposite poles during anaphase II.
- Produce abnormal gamete (n-1) with one **less** chromosome 21.
- Fertilization between normal gamete (n) and abnormal gamete (n -1) produces a zygote (2n-1) that has 45 chromosomes with only one chromosome 21.

Tips from LKC
- state the type of chromosome involved (example : chromosome 21)
- State which phase does the nondisjunction occur (example : anaphase I or anaphase II)

# AUTOSOMAL ABNORMALITIES

## Monosomy 21

Symptoms:

- Short distance between eyes
- Large ears
- Contracted muscle



Figure 27

# AUTOSOMAL ABNORMALITIES

## Trisomy 21 (Down syndrome)

- A diploid organism with extra one chromosome 21; 2n+1 (47 chromosomes)
- Also known as **Down syndrome**
- A woman's chance of giving birth to a Down syndrome child increases with age

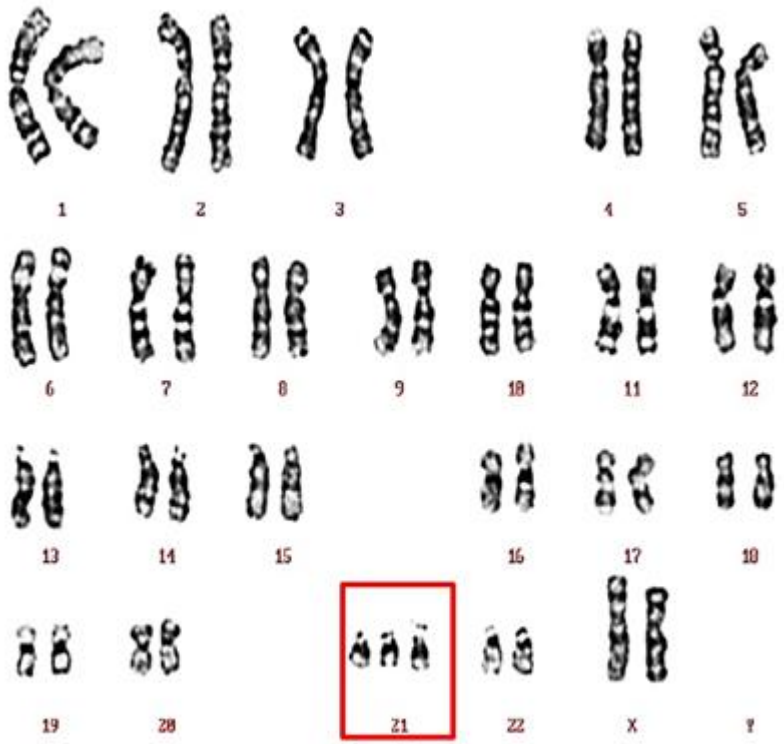



Figure 28

Tips from LKC
- Make sure Down syndrome is written with capital letter **D**

Learning Outcomes:
7.3 (i) Explain autosomal abnormalities and their effects (Tutorial)

# AUTOSOMAL ABNORMALITIES

## How does Trisomy 21 (Down syndrome) occurs?

- Due to the nondisjunction of chromosome 21 during meiosis I or meiosis II
- **Failure of homologous chromosome to separate and move to opposite poles during anaphase I OR**

  **Failure of sister chromatids to separate and move to opposite poles during anaphase II**
- Producing abnormal gametes (n+1) with **extra** one chromosome 21.
- When an abnormal gamete with an **extra** chromosome 21 (n+1) fertilizes with a normal gamete (n), produces a zygote (2n+1) that has 47 chromosomes with three copies of chromosome 21.

Tips from LKC
- state the type of chromosome involved (example : chromosome 21)
- Abnormal gamete with **extra** chromosome 21

# Nondisjunction of chromosome 21

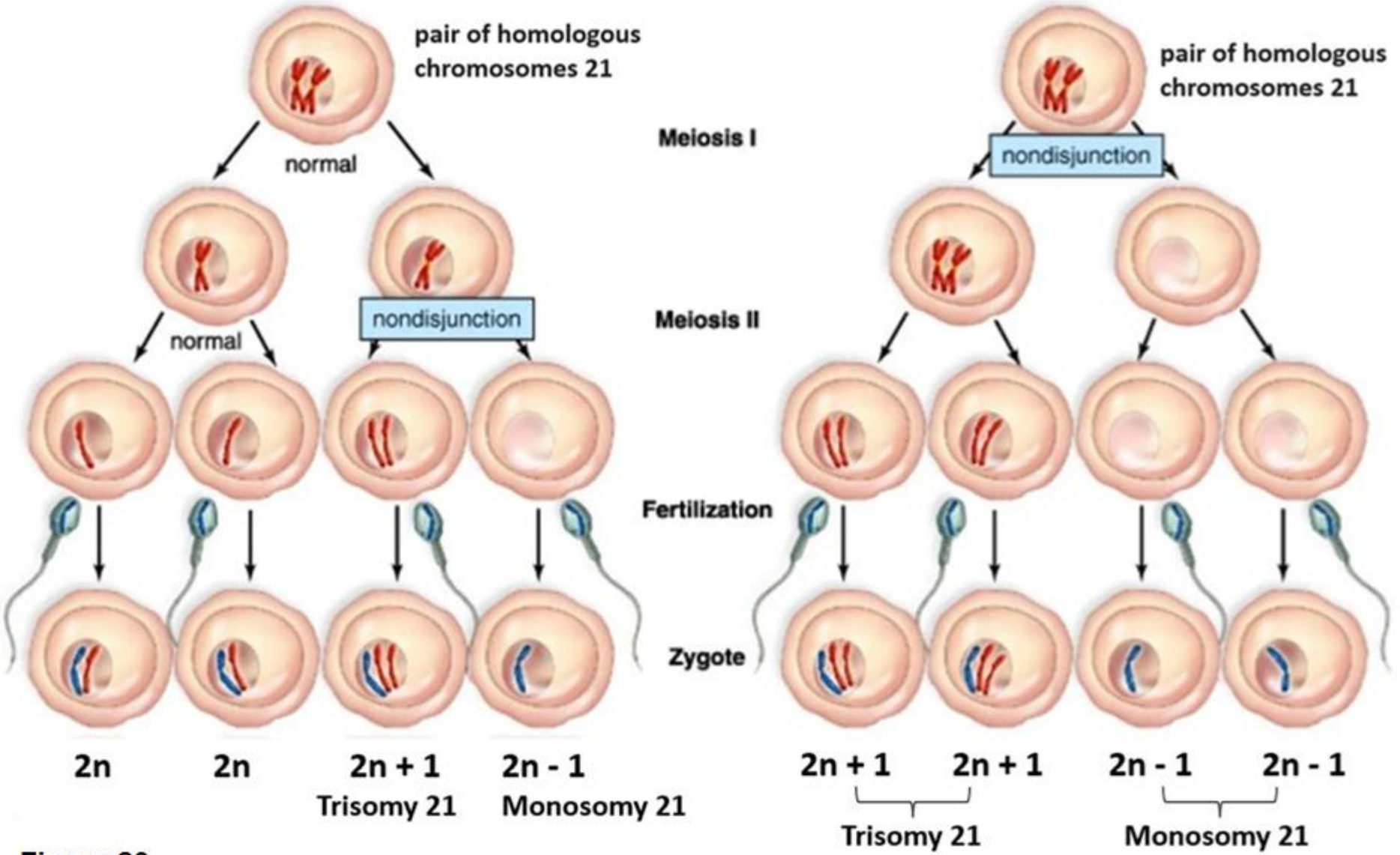



Figure 29

# AUTOSOMAL ABNORMALITIES

## Trisomy 21 (Down syndrome)

- Broad face and slanted eyes
- Short stature
- Protruding furrowed tongue
- Have sterile / have underdeveloped sexual organ
- Growth failure
- Mental retardation
- Congenital heart disease
- Short life span.




Figure 30

# SEX CHROMOSOMAL ABNORMALITIES

- Abnormalities in sex chromosome number caused by nondisjunction during:

  ➢ Spermatogenesis:
  ✓ producing sperm containing an abnormal number of sex chromosomes (O/ XY/ XX/ YY sperm)

  ➢ Oogenesis:
  ✓ producing ovum containing an abnormal number of sex chromosomes (O/ XX ovum)

Tips from LKC
- state the type of chromosome involved (example : sex chromosome)
- Must state correct process and cell involved
  (example : nondisjunction in **spermatogenesis** produce **sperm** containing abnormal number of sex chromosome

# SEX CHROMOSOMAL ABNORMALITIES

- If fertilization occurs between the normal gamete, n and the abnormal gamete, n+1 or n-1, the zygote will have abnormal numbers of sex chromosome.
- The possible abnormal zygotes formed are:
  - Trisomy: **Klinefelter syndrome (44+XXY** or **47, XXY)**
  - Monosomy: **Turner syndrome (44+XO** or **45, XO)**

# SEX CHROMOSOMAL ABNORMALITIES

## Klinefelter syndrome (47, XXY)

- A genetic condition that results when a boy is born with **an extra copy of the X chromosome**.

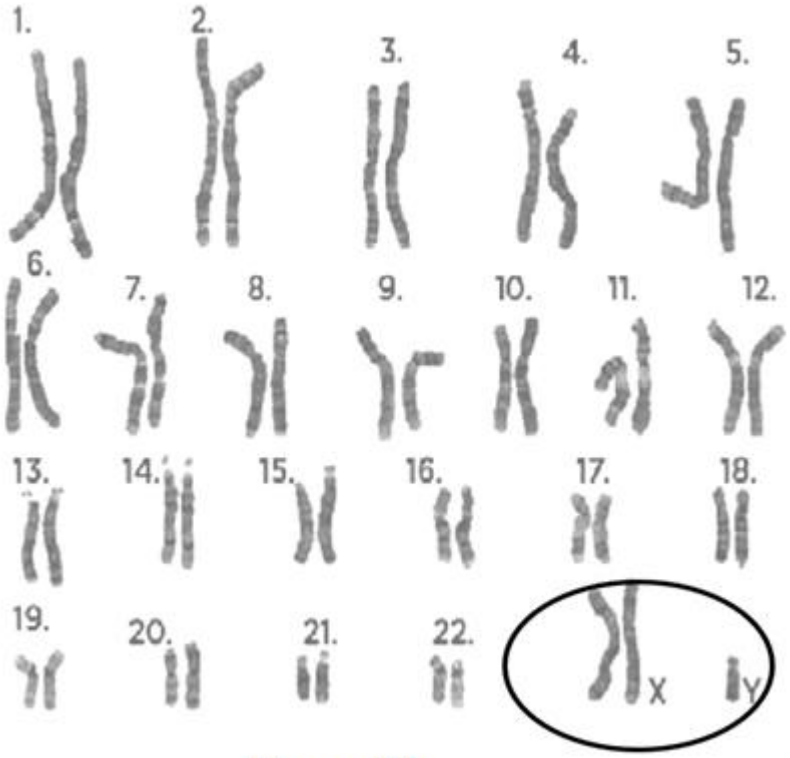



Figure 31

# SEX CHROMOSOMAL ABNORMALITIES

## Klinefelter syndrome (47, XXY)

How it occurs?

- Due to **nondisjunction of sex chromosome in anaphase I/meiosis I during spermatogenesis in male OR anaphase I/meiosis I or anaphase II/ meiosis II during oogenesis in female**.

- Produce **sperm** with an abnormal number of chromosomes, **22+XY OR**

  **ovum** with an abnormal number of chromosome **22+XX**

Tips from LKC
- state the type of gamete (sperm or ovum) involved

# SEX CHROMOSOMAL ABNORMALITIES

## Klinefelter syndrome (47, XXY)

- When an **abnormal sperm**, **22+XY** fertilizes with a **normal ovum**, **22+X** produces a zygote with an abnormal number of chromosomes with **44+XXY** or **47, XXY**

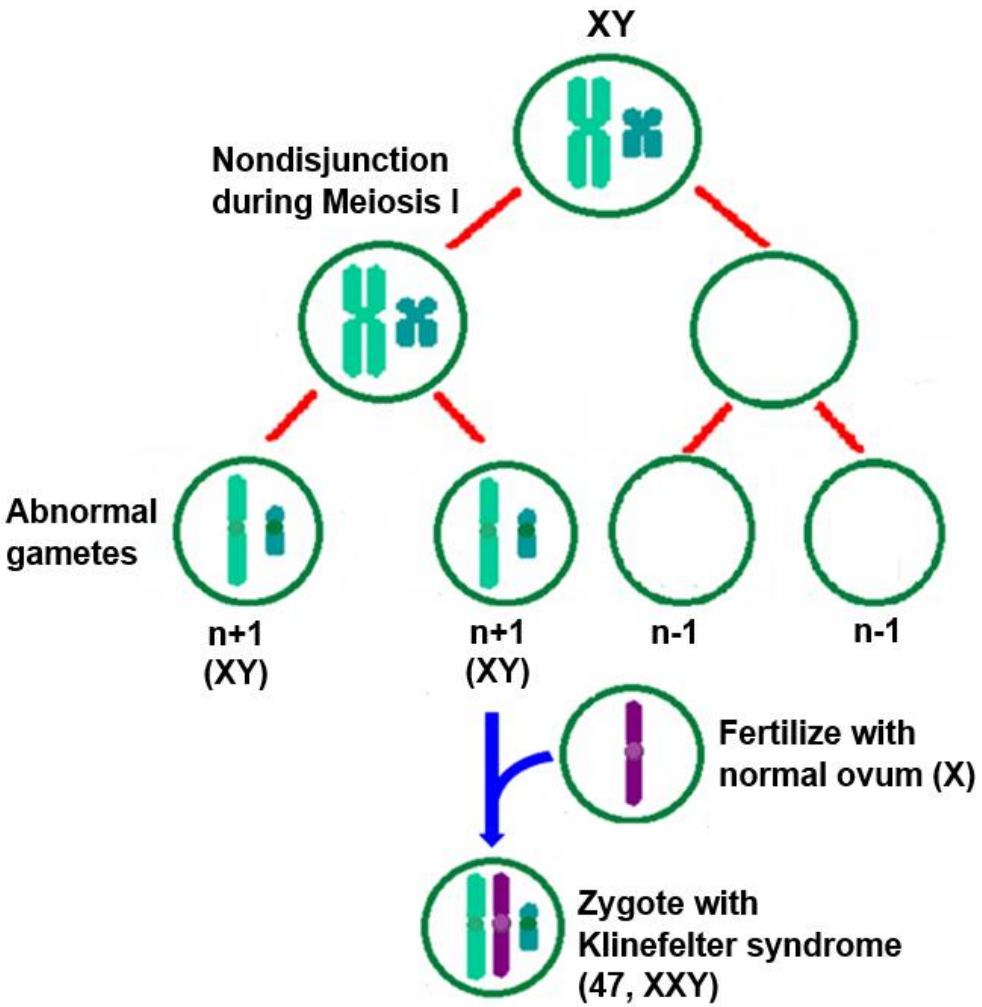
**OR**

- When an **abnormal ovum, 22+XX** fertilizes with **normal sperm, 22+Y** produce a zygote with an abnormal number of chromosomes with **44+XXY** or **47, XXY**

Tips from LKC
- state the genotype
- Example : 47, XXY

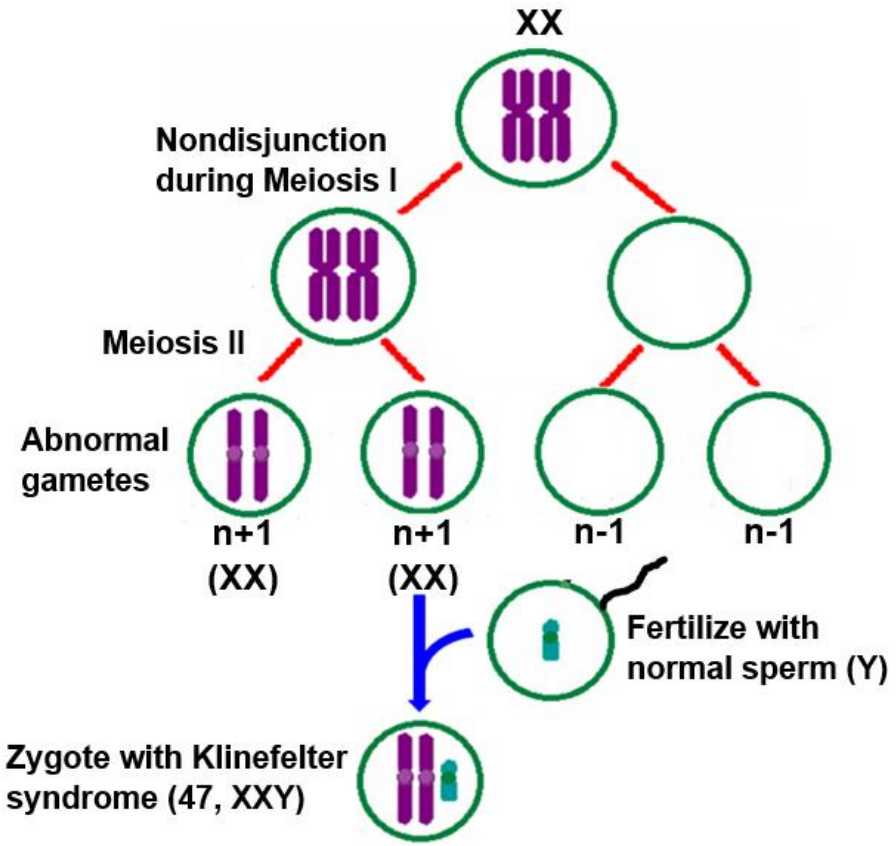
# Klinefelter syndrome (47, XXY)

## Nondisjunction during meiosis I of spermatogenesis

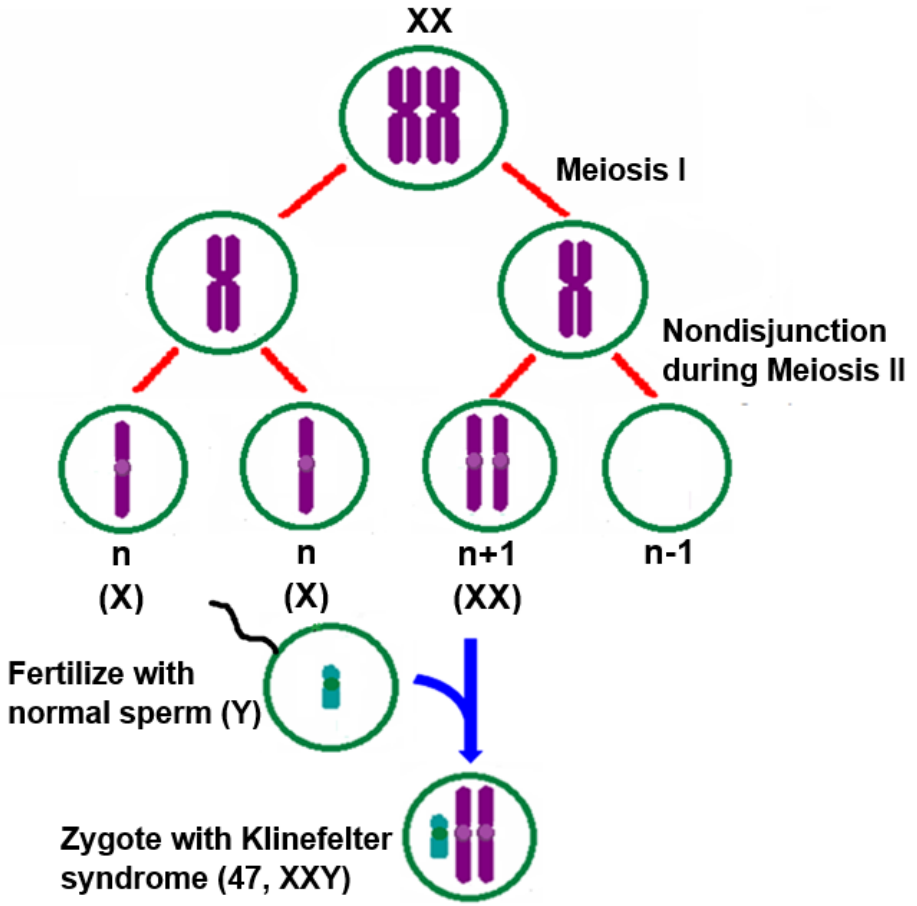


# Klinefelter syndrome (47, XXY)

## Nondisjunction during meiosis I of oogenesis




## Nondisjunction during meiosis II of oogenesis



# SEX CHROMOSOMAL ABNORMALITIES

## Klinefelter syndrome (47, XXY)

Symptoms:

- Small testes; fail to form sperm (sterile)
- Voice like a female
- Long legs and arms
- Has female secondary sexual characteristics (breast enlargement)
- May have subnormal intelligence

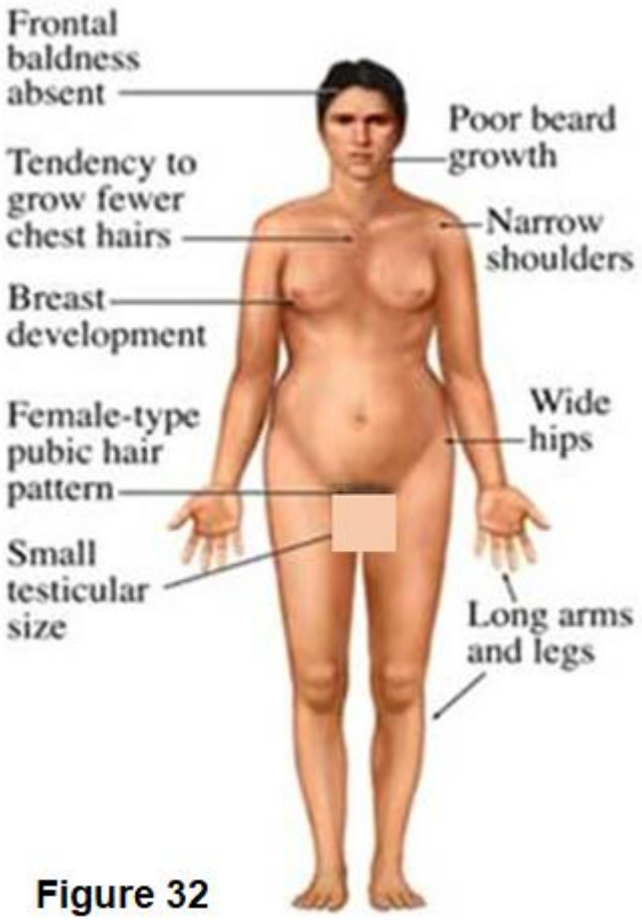



Figure 32

# SEX CHROMOSOMAL ABNORMALITIES

## Turner syndrome (45, XO)

- A genetic condition that affects only females, results when one of the X chromosomes is missing.
- May be diagnosed before birth (prenatally), during infancy or in early childhood

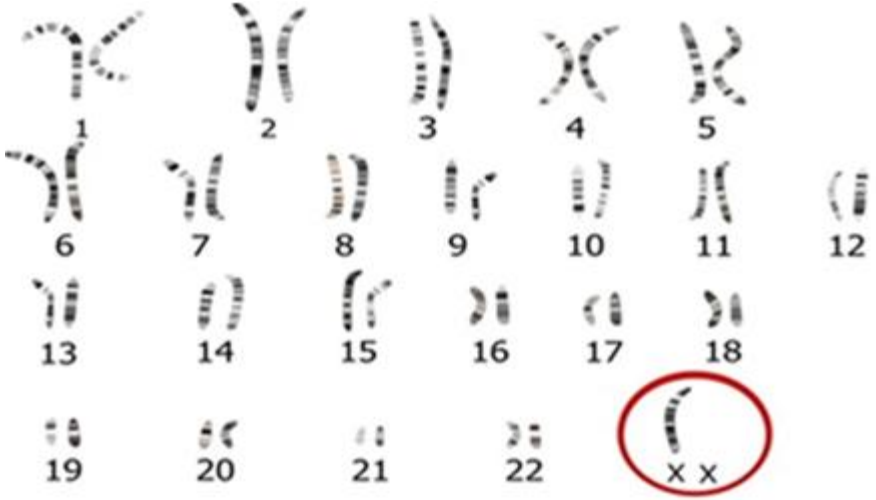



Figure 33

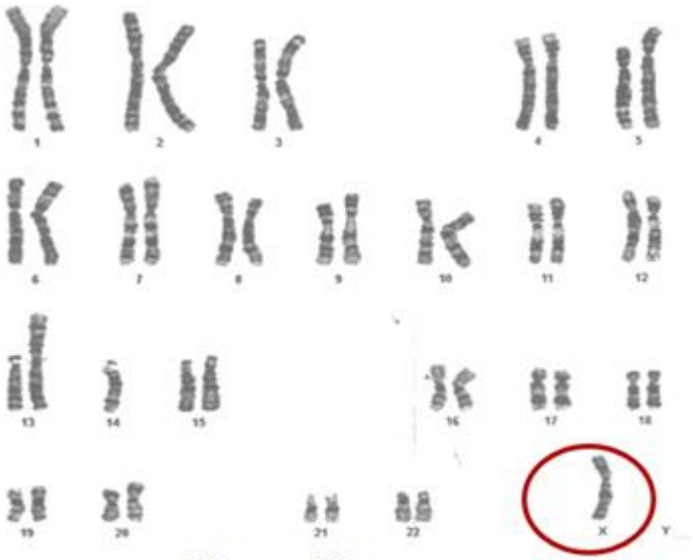


Figure 34

# SEX CHROMOSOMAL ABNORMALITIES

## Turner syndrome (45, XO)

How it occurs?

- Due to nondisjunction of sex chromosome in anaphase I/ meiosis I or anaphase II/ meiosis II during spermatogenesis in male **OR** oogenesis in female

- Produce sperm with an abnormal number of chromosomes, 22+O **OR** ovum with an abnormal number of chromosomes, 22+O.

# SEX CHROMOSOMAL ABNORMALITIES

## Turner syndrome (45, XO)

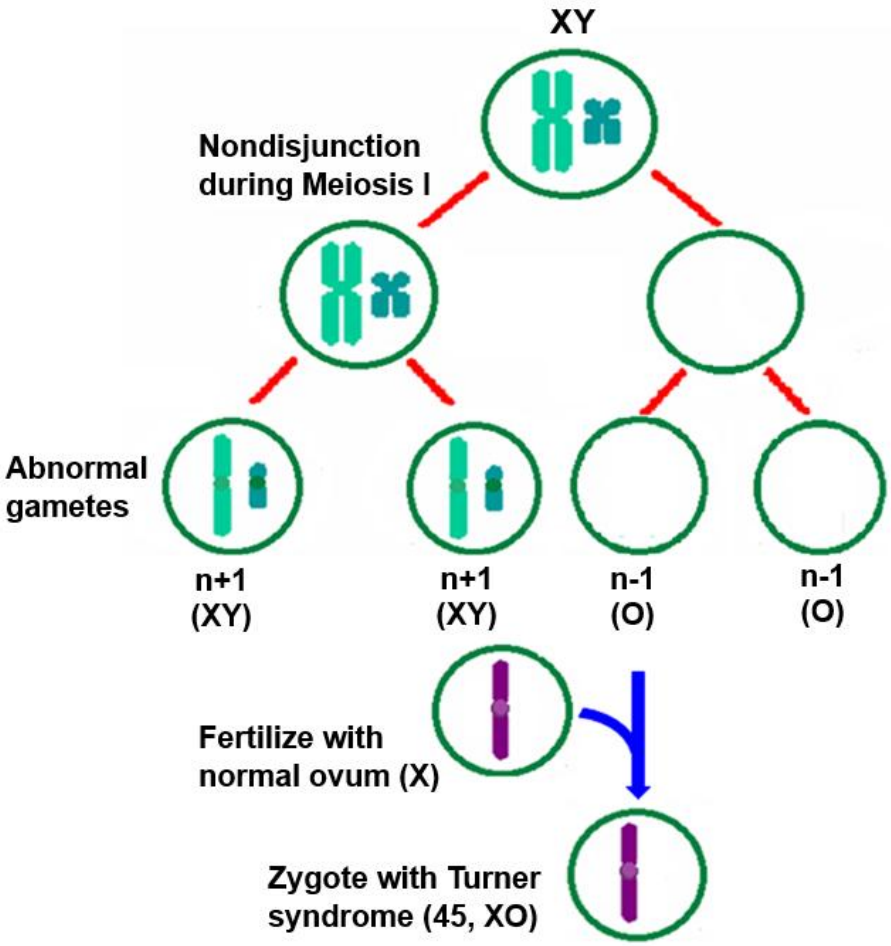
- When **abnormal sperm,** 22+O fertilizes with **normal ovum,** 22+X produce a zygote with an abnormal number of chromosomes with **44+XO** or **45, XO**

**OR**

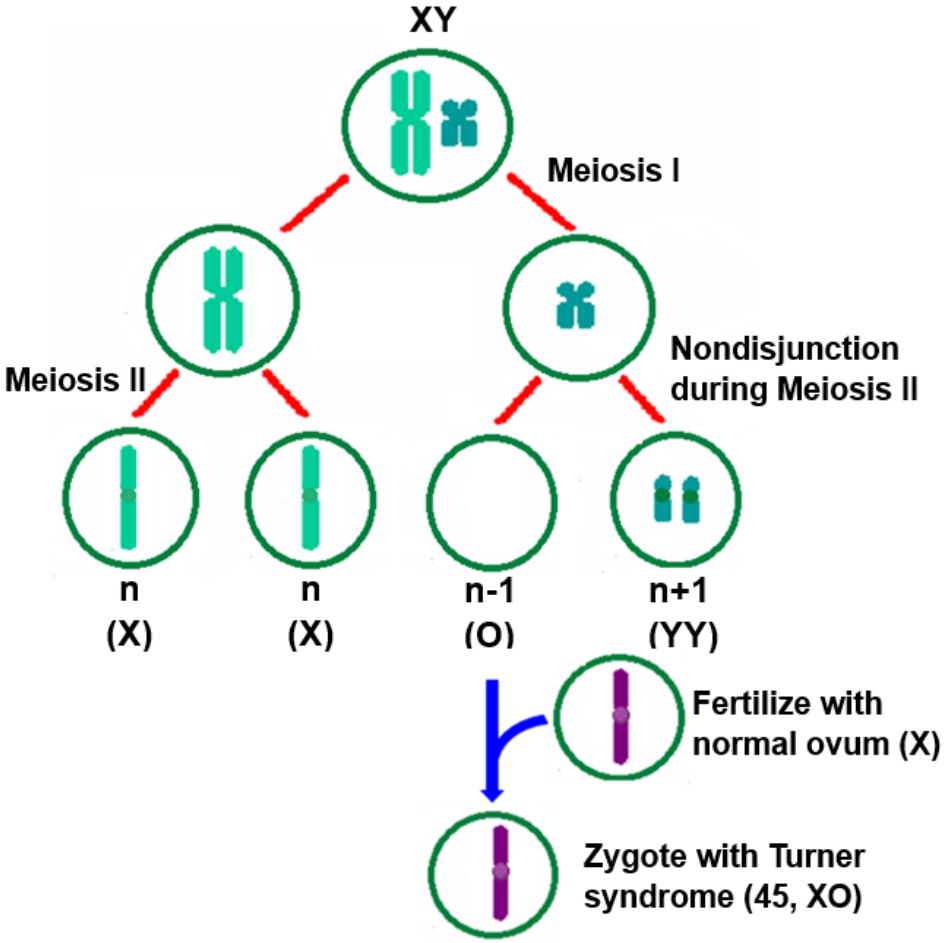
- When an **abnormal ovum,** 22+O fertilizes with **normal sperm, 22+X** produces a zygote with an abnormal number of chromosomes with **44+XO** or 45, XO

# Turner syndrome (45, XO)

## Nondisjunction during meiosis I of spermatogenesis

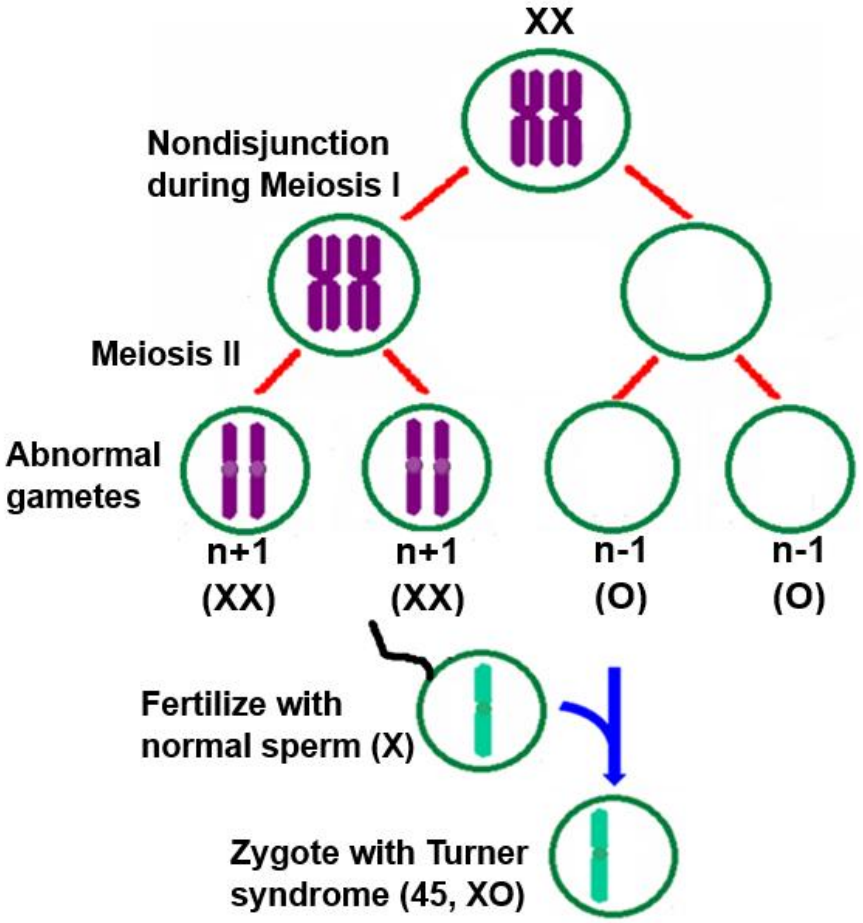



## Nondisjunction during meiosis II of spermatogenesis

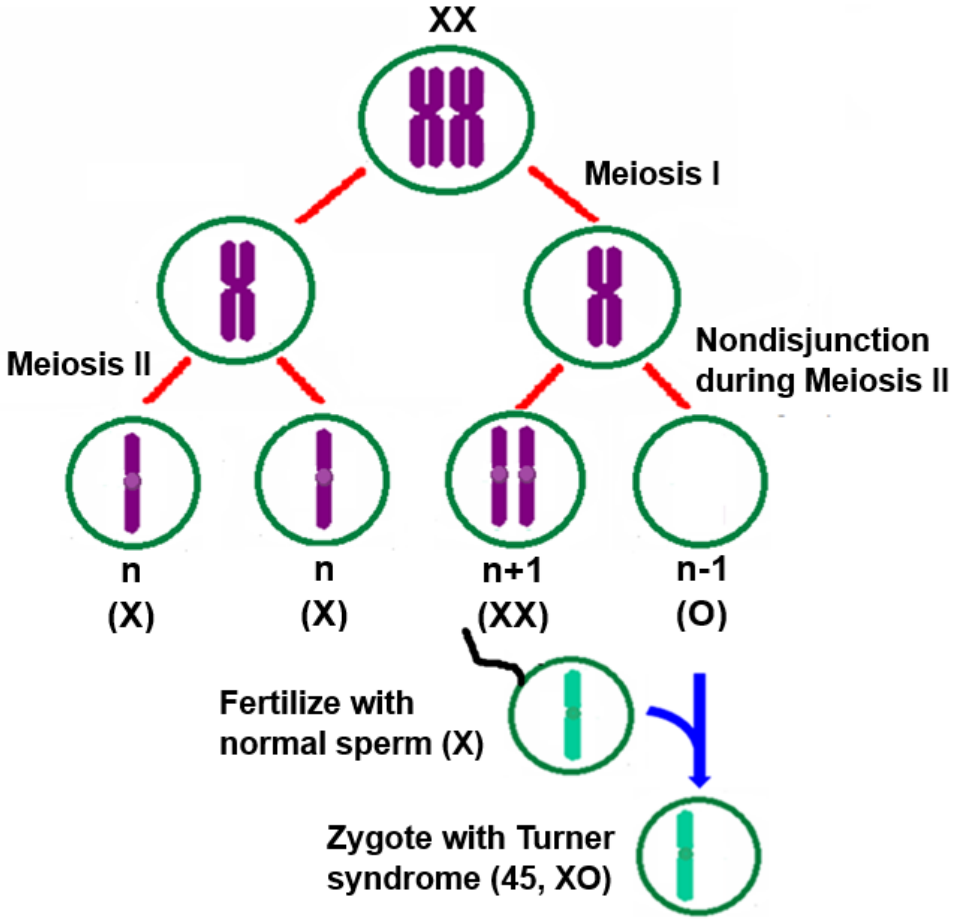


# Turner syndrome (45, XO)

## Nondisjunction during meiosis I of oogenesis




## Nondisjunction during meiosis II of oogenesis



# SEX CHROMOSOMAL ABNORMALITIES

## Turner syndrome (45, XO)

Symptoms:

- Dwarf / short stature
- Ovaries and breast do not develop
- Sterile
- Webbed neck
- Heart defects
- Most have normal intelligence

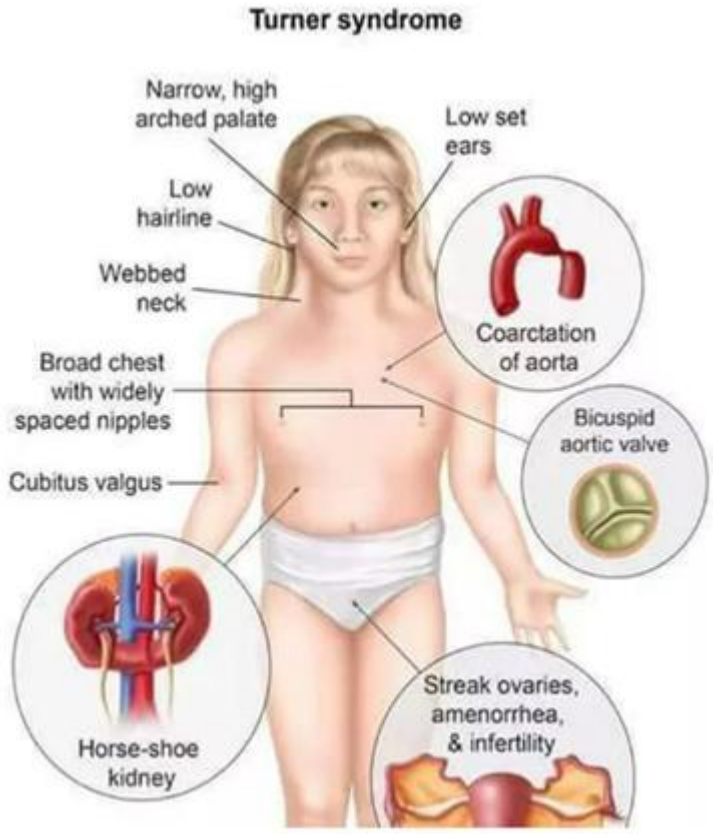



Figure 35

# POLYPLOIDY

- **A chromosomal alteration in organisms that possess more than two complete chromosome sets.**
- May occur naturally **OR** by induction (e.g. using chemicals such as colchicine)
- Spindle fiber formation is prevented, causing complete nondisjunction involving all chromosomes and resulting in the doubling of chromosome number.
- 2 types: **autopolyploidy** and **allopolyploidy**

# POLYPLOIDY

- Common in plants/crops
  e.g.: Bananas (triploid), wheat (hexaploid), strawberries (octoploid)
- Polyploid plants often have better qualities than their parents
  - ➢ Hybrid vigor e.g. increased size, resistance to disease and seedless fruits
- Uncommon in animals; usually has lethal effects during embryonic development

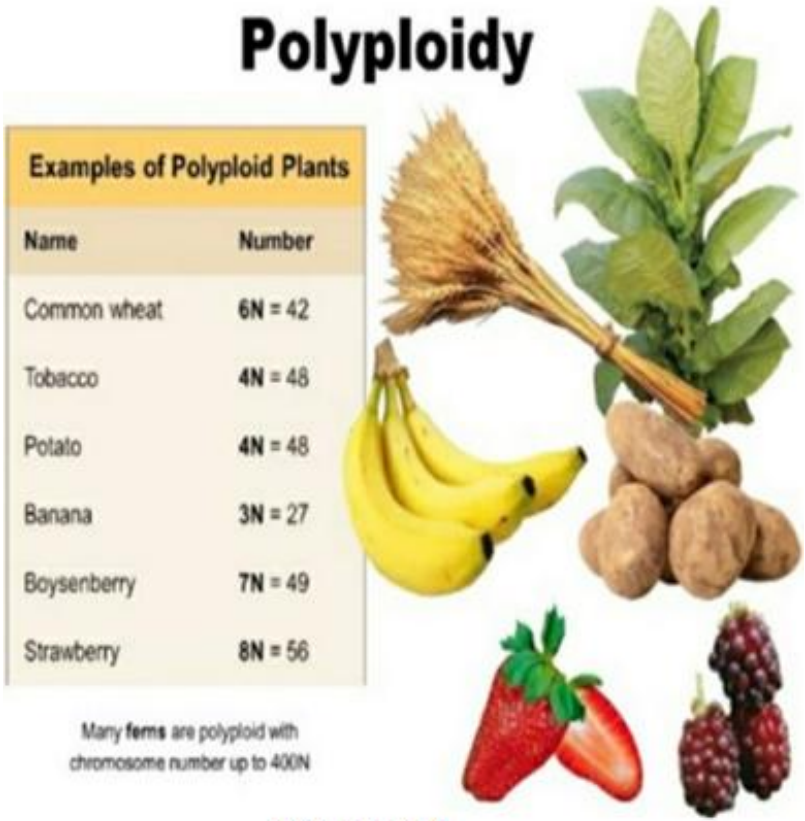



Figure 36

# POLYPLOIDY

- Method of naming polyploidy is based on the total sets of chromosomes present:
  - 3n (3 sets) - triploid
  - 4n (4 sets) - tetraploid
  - 5n (5 sets) - pentaploid
  - 6n (6 sets) - hexaploid
  - 8n (8 sets) - octaploid
  - 10n (10 sets) - decaploid

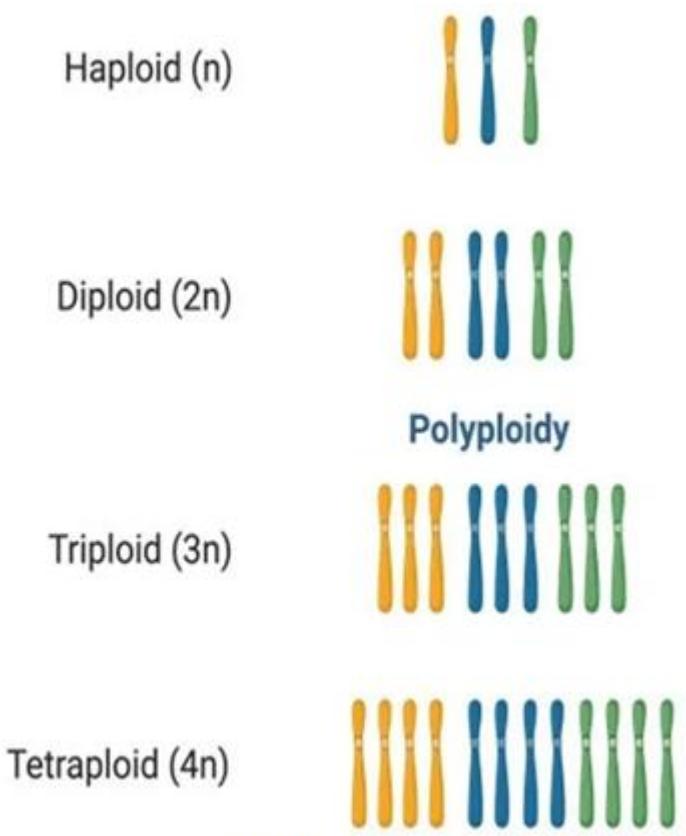



Figure 37

# POLYPLOIDY

## Autopolyploidy

- A condition in which an **individual has more than two chromosome sets that are all derived from the same species.**
- All of the chromosomes are identical
- E.g. Strawberry, sugarcane, potato

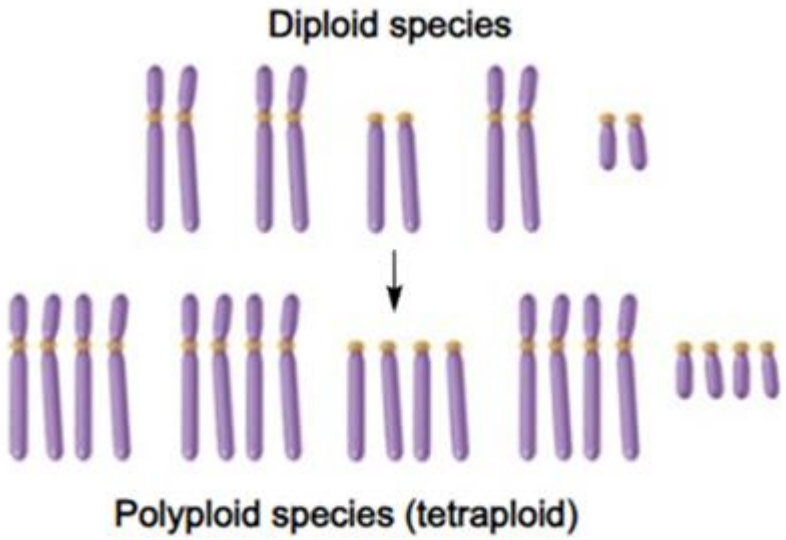



Figure 38

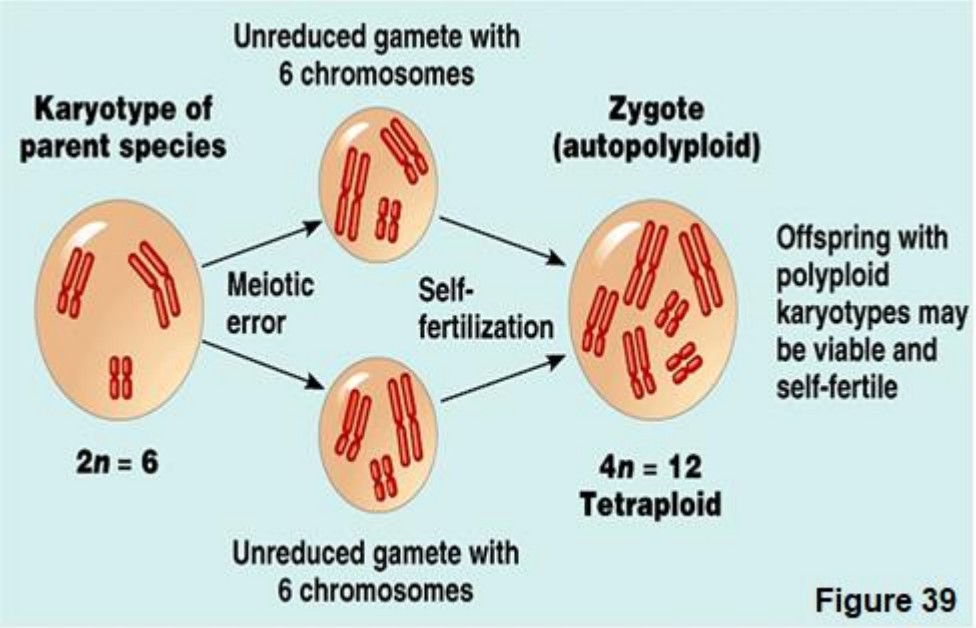
# POLYPLOIDY

## Autopolyploidy

Example:

- A failure of cell division doubles a cell's chromosome number from diploid 2n to tetraploid 4n

- Caused by **nondisjunction in meiosis** that causes the formation of **diploid gametes.**

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# POLYPLOIDY

## Autopolyploidy

Self fertilization between **two unreduced gametes** from a diploid cell

Parental genotype: **AA** **X** **AA**

Meiosis (nondisjunction)

Gamete: **AA** ○ ○ **AA**

Random fertilization:

Offspring genotype: **AAAA**
(fertile tetraploid)

# POLYPLOIDY

## Autopolyploidy

Self fertilization between **an unreduced gametes** from a diploid cell

Parental genotype: **AA** **X** **AA**

Meiosis (nondisjunction)

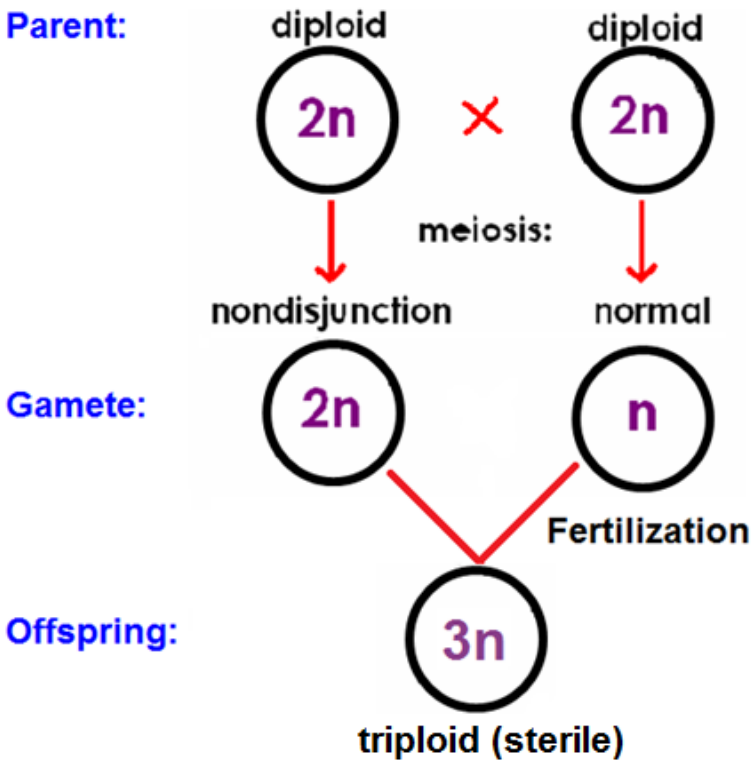
Gamete: **AA** ○ **A** **A**

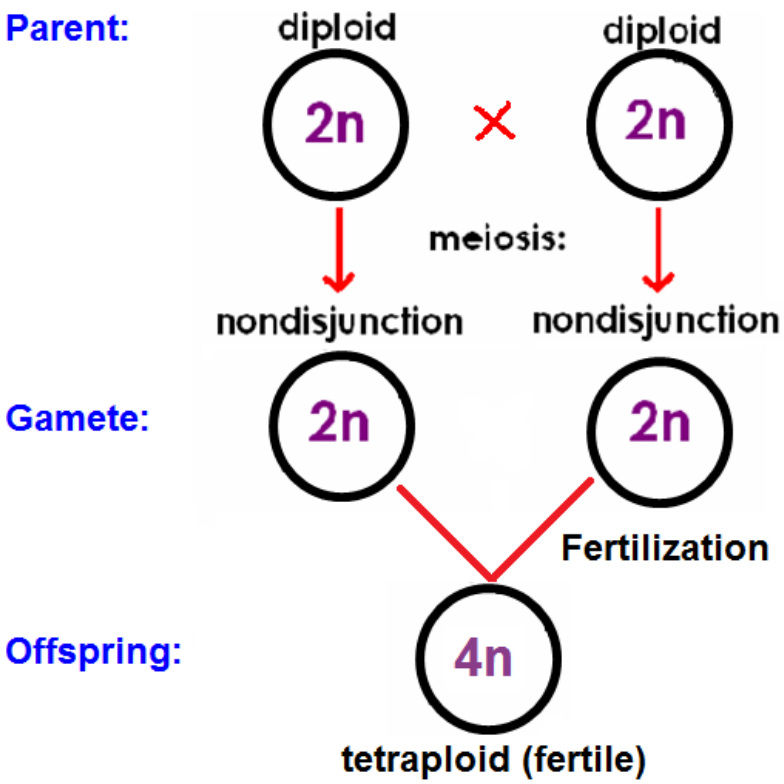
Random fertilization:

Offspring genotype: **AAA** (sterile triploid)

**Sterile autopolyploids** can be induced to undergo **chromosome doubling** to become **fertile** by use of chemicals (e.g.: Colchicine) which interferes with spindle formation (mitotic error)

# Autopolyploidy




- Autopolyploid individual is fertile if it has an even number of chromosome sets (4n, 6n, 8n etc.)

# POLYPLOIDY

## Allopolyploidy

- A condition in which an **individual has more than two chromosome sets as a result of two different but closely related species interbreeding and combining their chromosomes**
- Due to species hybridization.

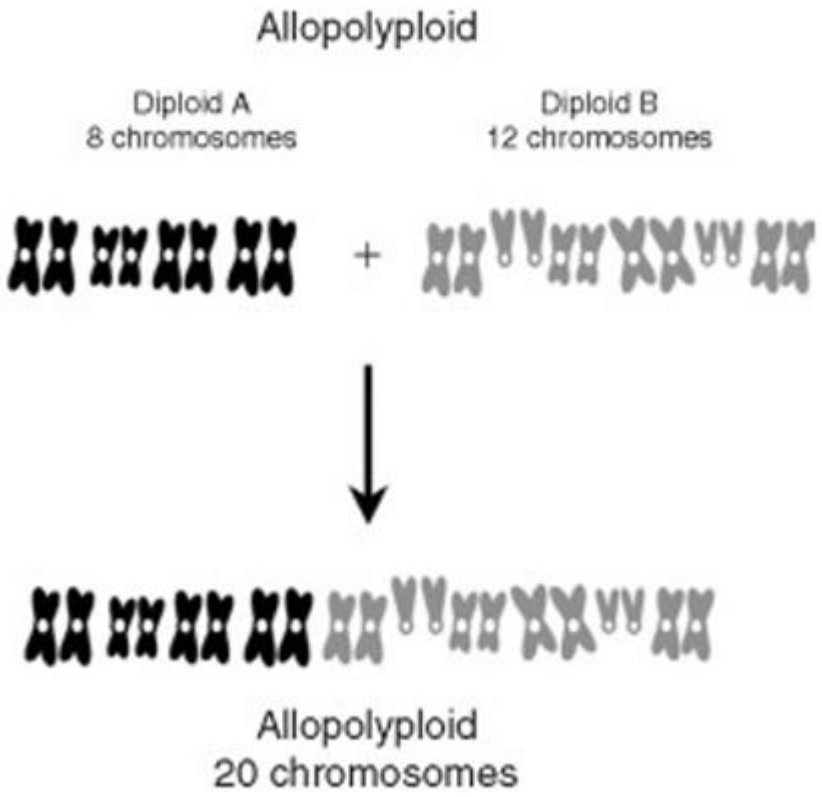



**Figure 40**

# POLYPLOIDY

## Allopolyploidy

- When 2 different species interbreed, they will produce hybrid offspring.
- **Hybrid is sterile** because the **chromosomes cannot pair during meiosis (not homologous).**
- Sterile hybrid can reproduce asexually by **vegetative propagation.**

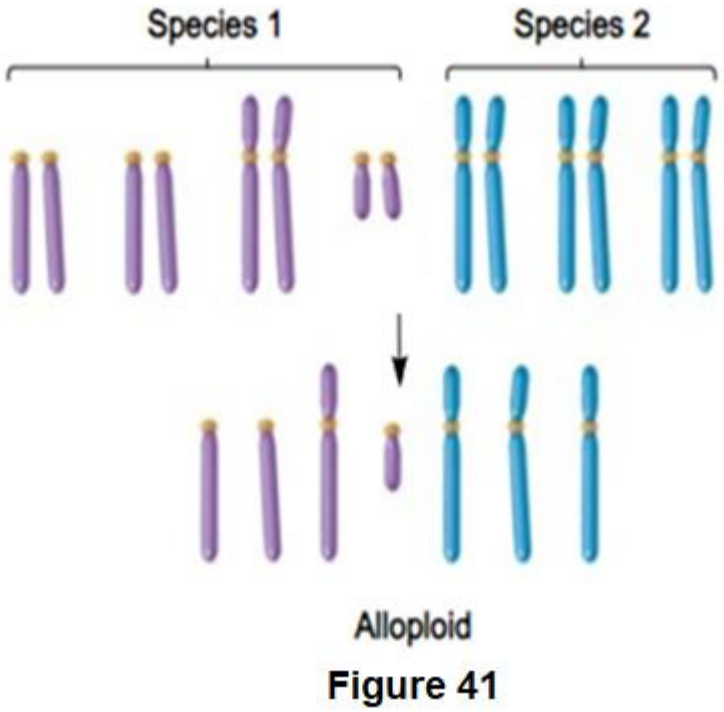



Figure 41

# POLYPLOIDY

## Allopolyploidy

- A sterile hybrid can be changed into a fertile hybrid (allopolyploid) by **chromosome doubling**.
- The homologous chromosomes are present and can undergo synapsis during meiosis to produce gametes.
- The hybrid can interbreed with each other, but they cannot interbreed with either parent species.

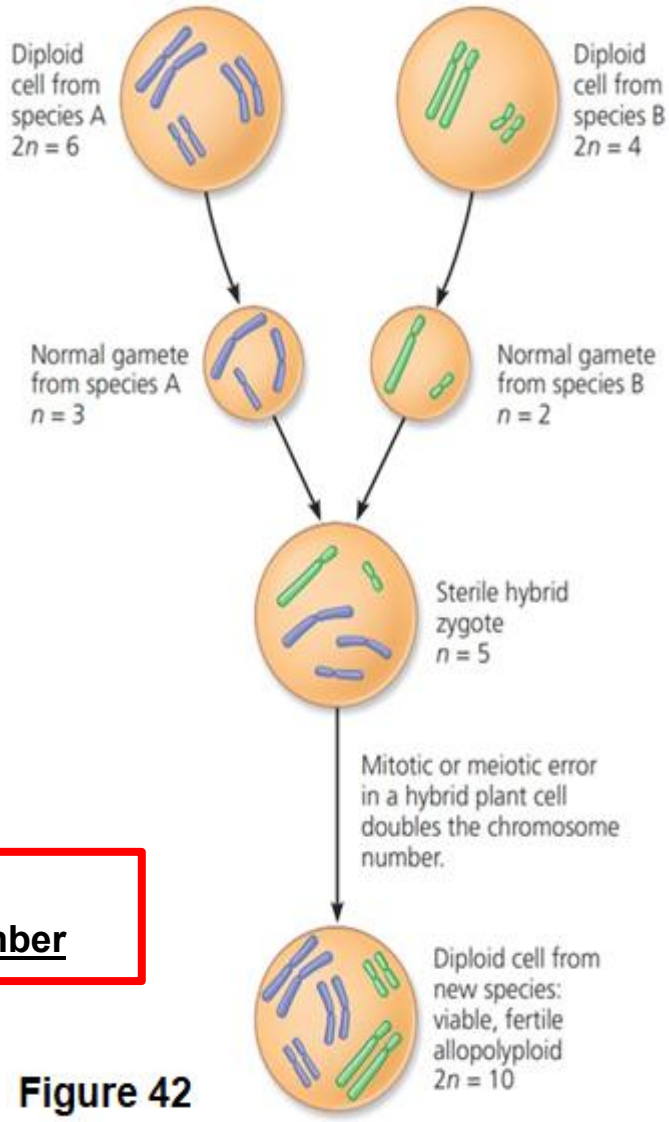



Figure 42

Tips from LKC
- Chromosome doubling occur/ Doubling the chromosome **number**

# POLYPLOIDY

## Allopolyploidy

Example:

- Bread wheat (*Triticum aestivum*) is an allohexaploid, containing two sets of chromosomes from each of three different species

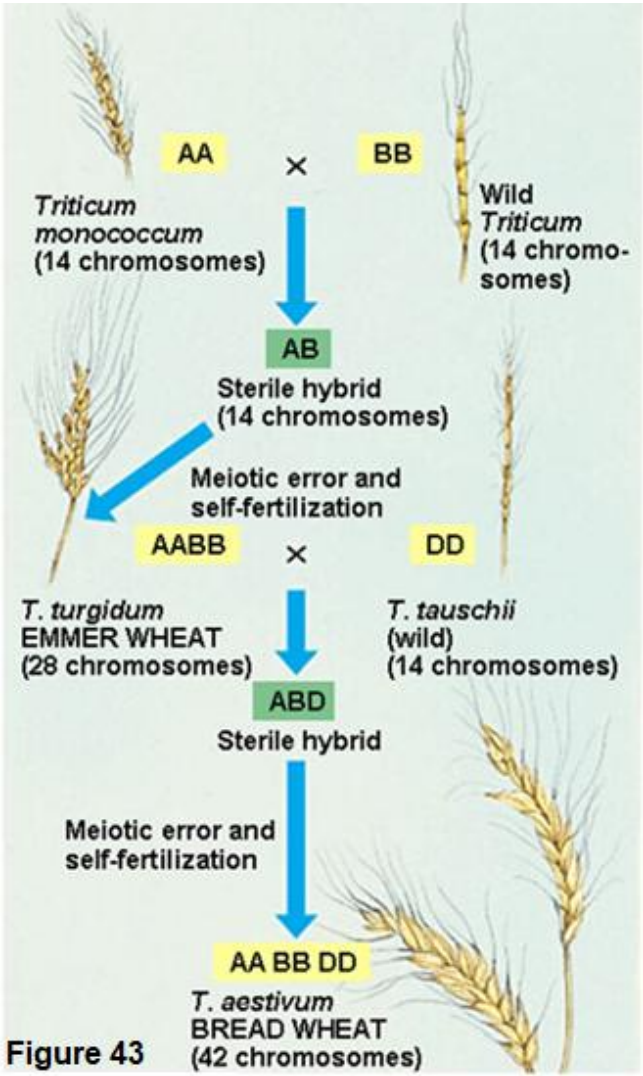



Figure 43

# ii) Allopolyploidy

Example 1:

***Spartina maritima*** x ***Spartina alterniflora***

*(2n=56)*
n=28

*(2n=70)*
n=35

Hybrid *Spartina townsendii*

n+n= 63

(**Sterile**)

**Chromosome doubling**

Why sterile?

- No homologous chromosome/ pair
- Cannot undergo meiosis
- Cannot produce gamete

*Spartina anglica*

*(fertile)*

AABB

2n=126 (tetraploid)

Why fertile?

- Has homologous chromosome/ pair
- Can undergo meiosis
- Can produce gamete

Tips from LKC

Why sterile?

- No pairing of homologous chromosome ✗
- No homologous chromosome for pairing ✓



# REFERENCES


- Campbell N.A & Reece, J.B., Biology, 12th ed. (2021), Pearson Education, Inc.
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