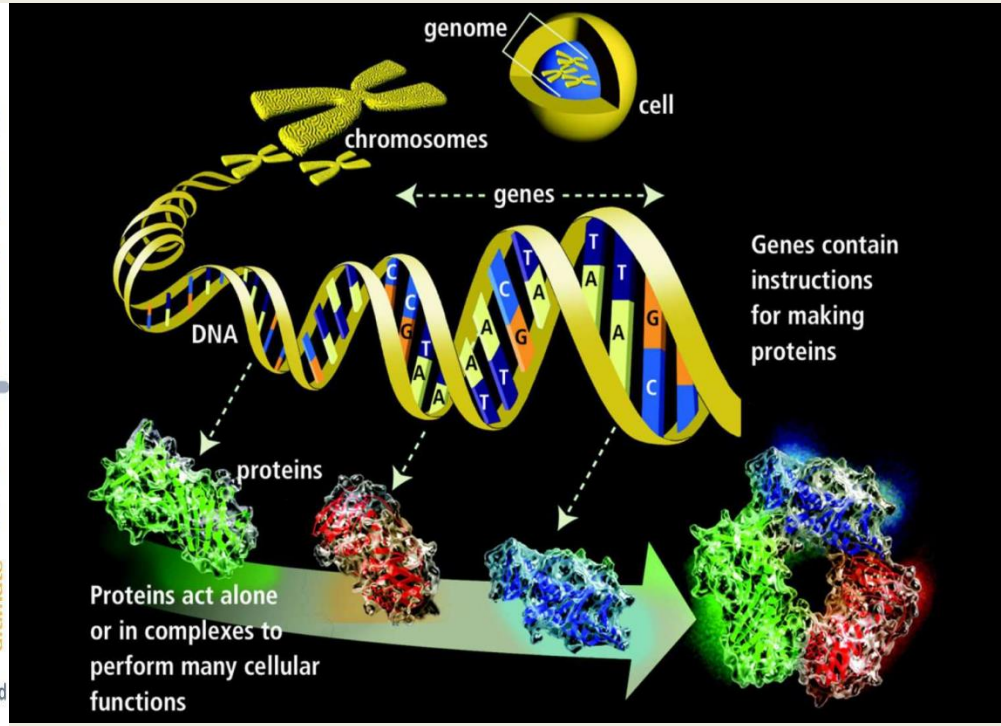
Biology Lesson Note For 11th Graders



Unit 4 - Genetics

4.4 Protein synthesis

4.5 Mendelian inheritance

4.6 Sex determination

4.7 Non-Mendelian inheritance

Part - II

By: Getaneh T.

4.4 protein Synthesis

- **Protein synthesis** is the stepwise process of the production of different types of proteins from amino acids.
- Protein synthesis involves **DNA**, **RNA** (mRNA, tRNA and rRNA), **amino acids,** various **enzymes** and **ribosome.**

- **DNA** stores genetic information used to produce different proteins.
- **Messenger RNA (mRNA)** transcribes genetic information from DNA in the nucleus with the help of enzyme **RNA polymerase**.
- **Transfer RNA** (tRNA) brings amino acids from the cytoplasm to the ribosome and it translates the message within the nucleotide sequence of mRNA to a specific amino acid sequence.
- **Ribosomal RNA (rRNA)** is a molecule in cells that forms part of the ribosome that help translate the information in mRNA into protein.
- **Ribosomes** are cytoplasmic organelles that translates the mRNA template into a polypeptide chain. – site of translation.
- **Amino acids** are the building blocks of proteins. There are 20 naturally occurring amino acids for protein synthesis.

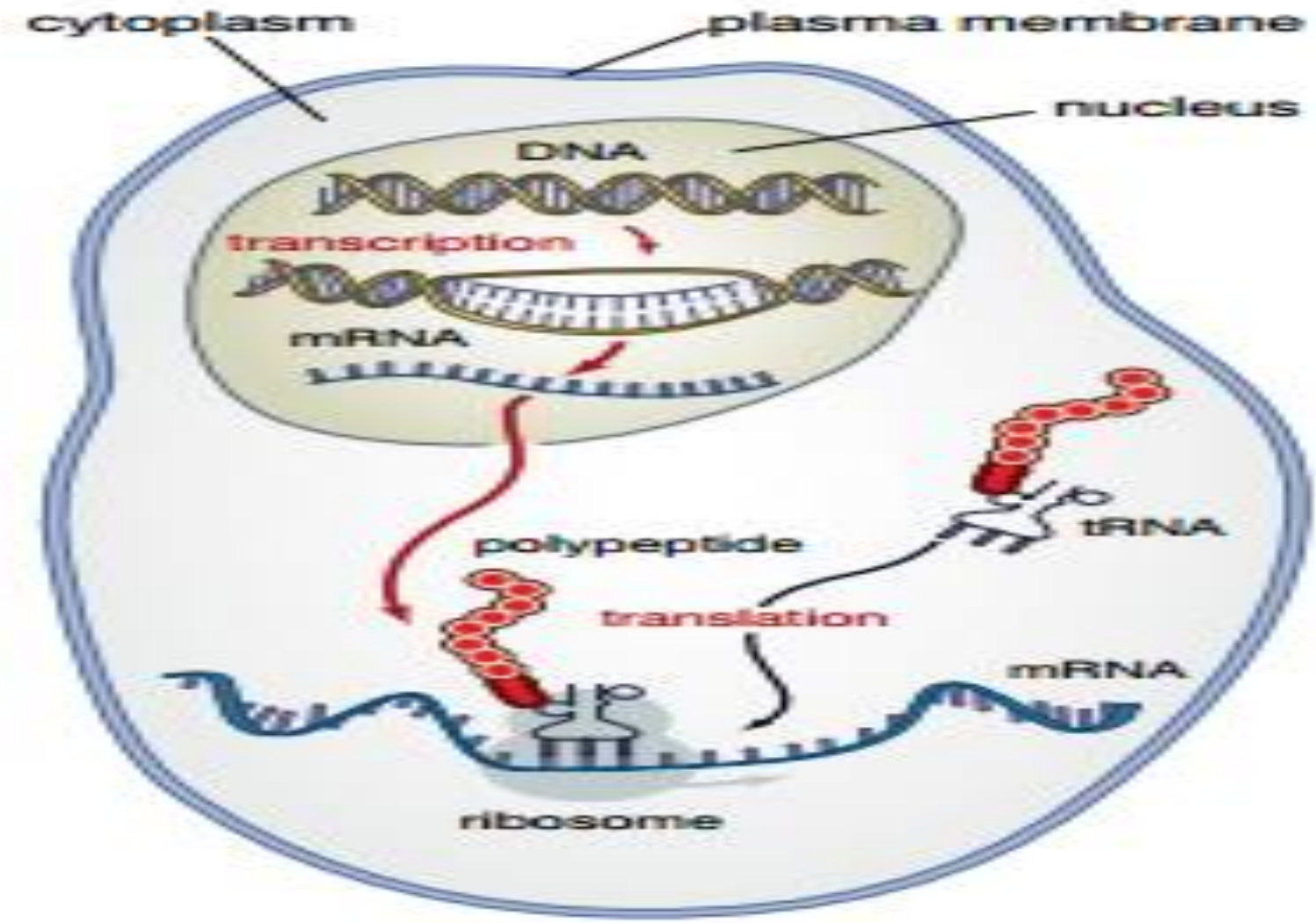


Figure 3.45 An overview of protein synthesis

Initiation → Elongation → Termination

How does a cell 'know' to make a protein?

- The code for a protein is specified by DNA (the genetic code).
- The DNA molecule contains all your hereditary information in the form of genes.
- A **gene** is a coded section of DNA; it tells our cells how to build specific proteins.
- Genes code for **EVERYTHING** our body needs and does.

✍ The following events occur during protein synthesis:

1. **Transcription -** rewriting the code of DNA into **messenger RNA** (mRNA).
   - mRNA is made from the template/antisense/non-coding strand of a DNA and has the same base sequence as the coding/sense strand of DNA.
2. **RNA splicing or processing** (post-transcriptional modification of mRNA)
   - removing **introns** (non-coding genes), and mRNA contains only **exons** (coding genes).
3. The mRNA travels from the nucleus through pores in the nuclear envelope to the ribosomes.
4. **Transfer RNA** (tRNA) - carry free amino acids from the cytoplasm to the ribosomes.
5. **Translation -** the ribosome reads the mRNA codons and assembles the amino acids carried by tRNA into a polypeptide (protein).

Transcription and translation

- The process of protein synthesis involves the conversation of instructions in DNA into a functional product (proteins) through **transcription** and **translation**.
- The process of biological information flow is called **central dogma**.

DNA → RNA → Protein

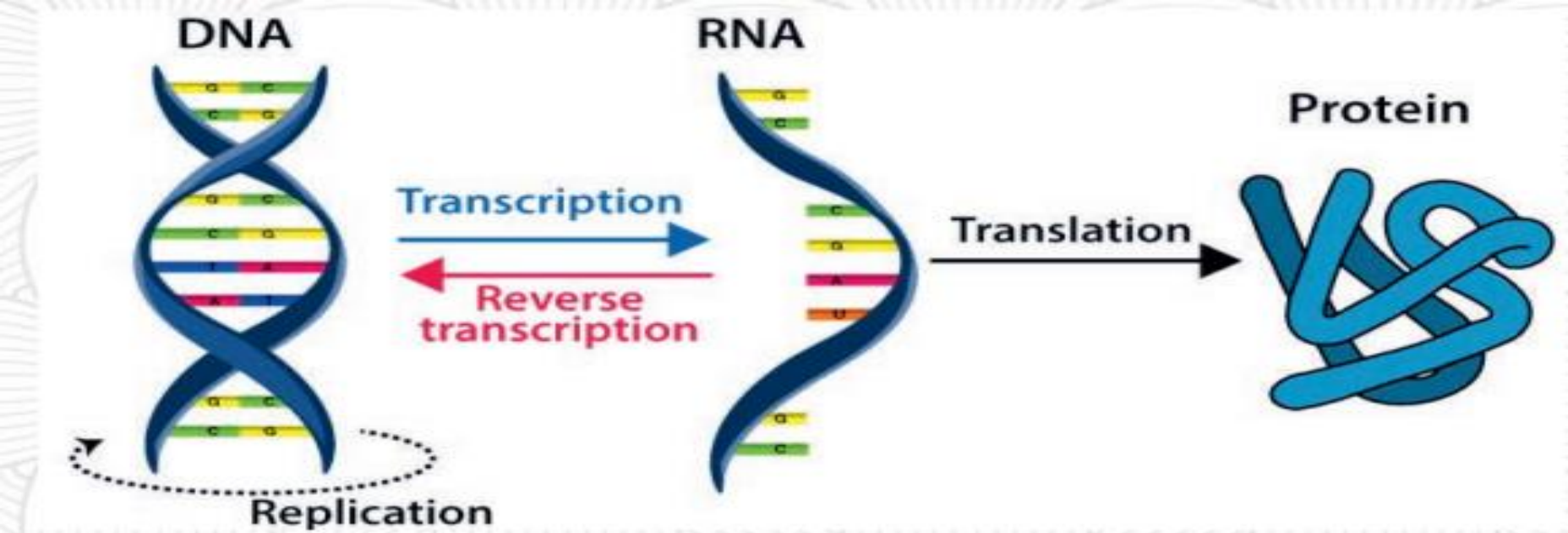
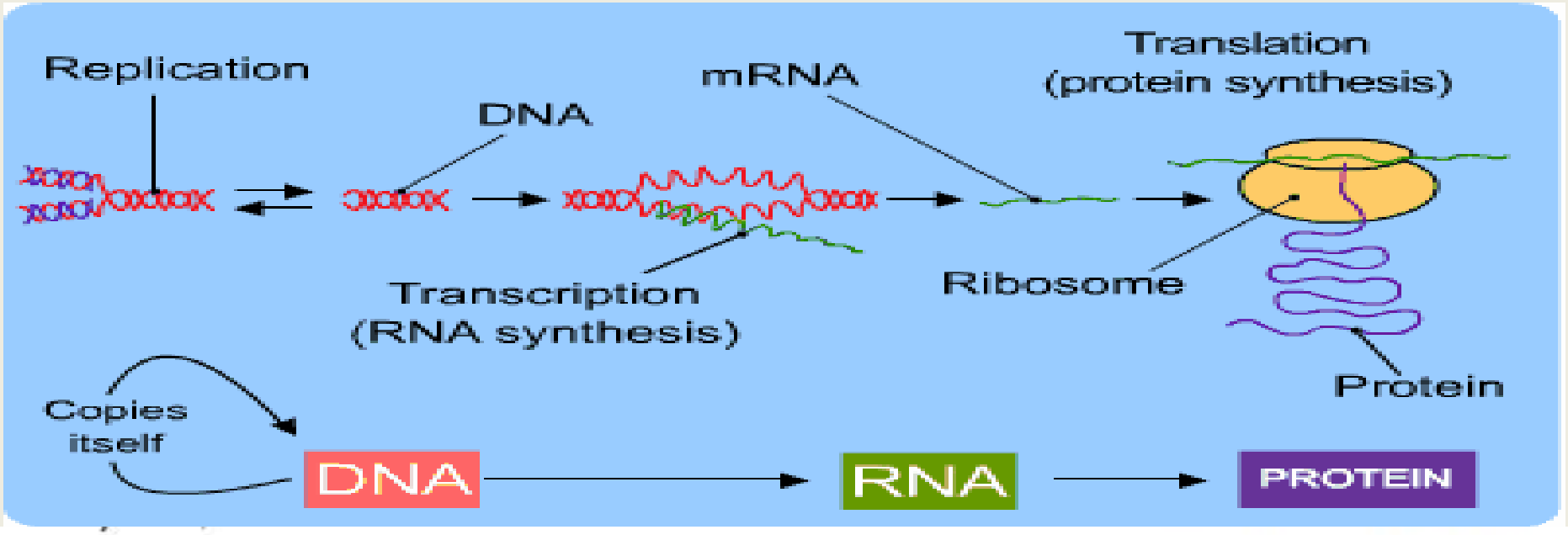


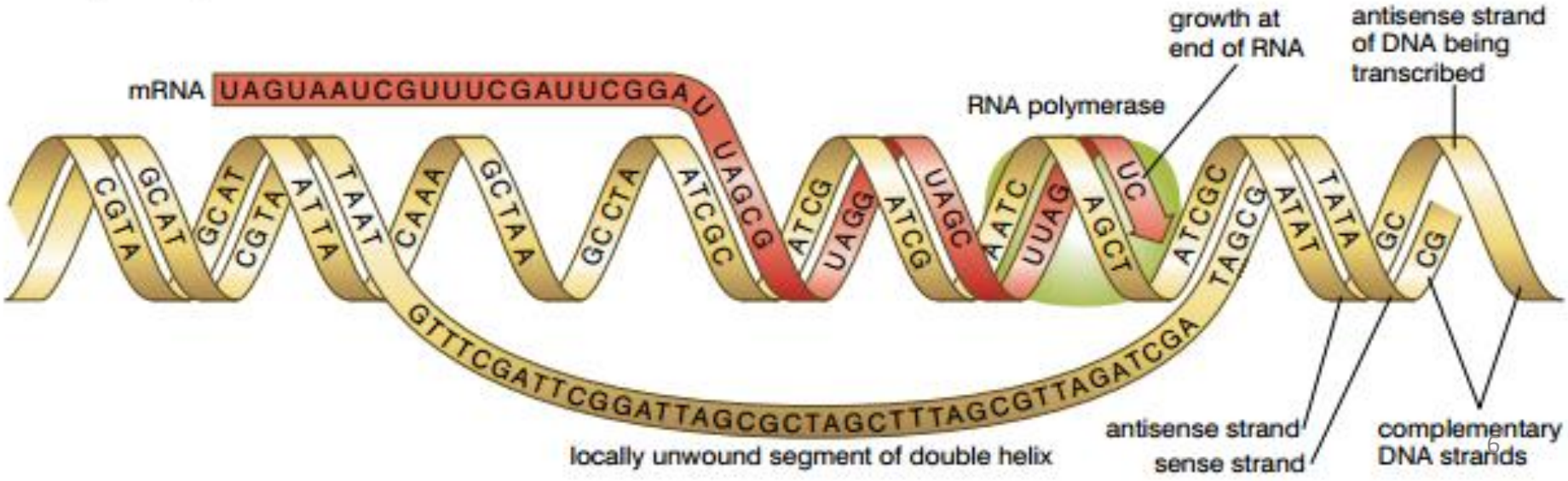
Figure 4.22 The central dogma of life

N.B: Reverse transcription is a process in which a DNA molecule is synthesized from an RNA template, which occurs in retroviruses. **RNA → DNA**

The "central dogma" of life

- states that information flows from DNA to RNA to Proteins. **DNA → RNA → Protein**



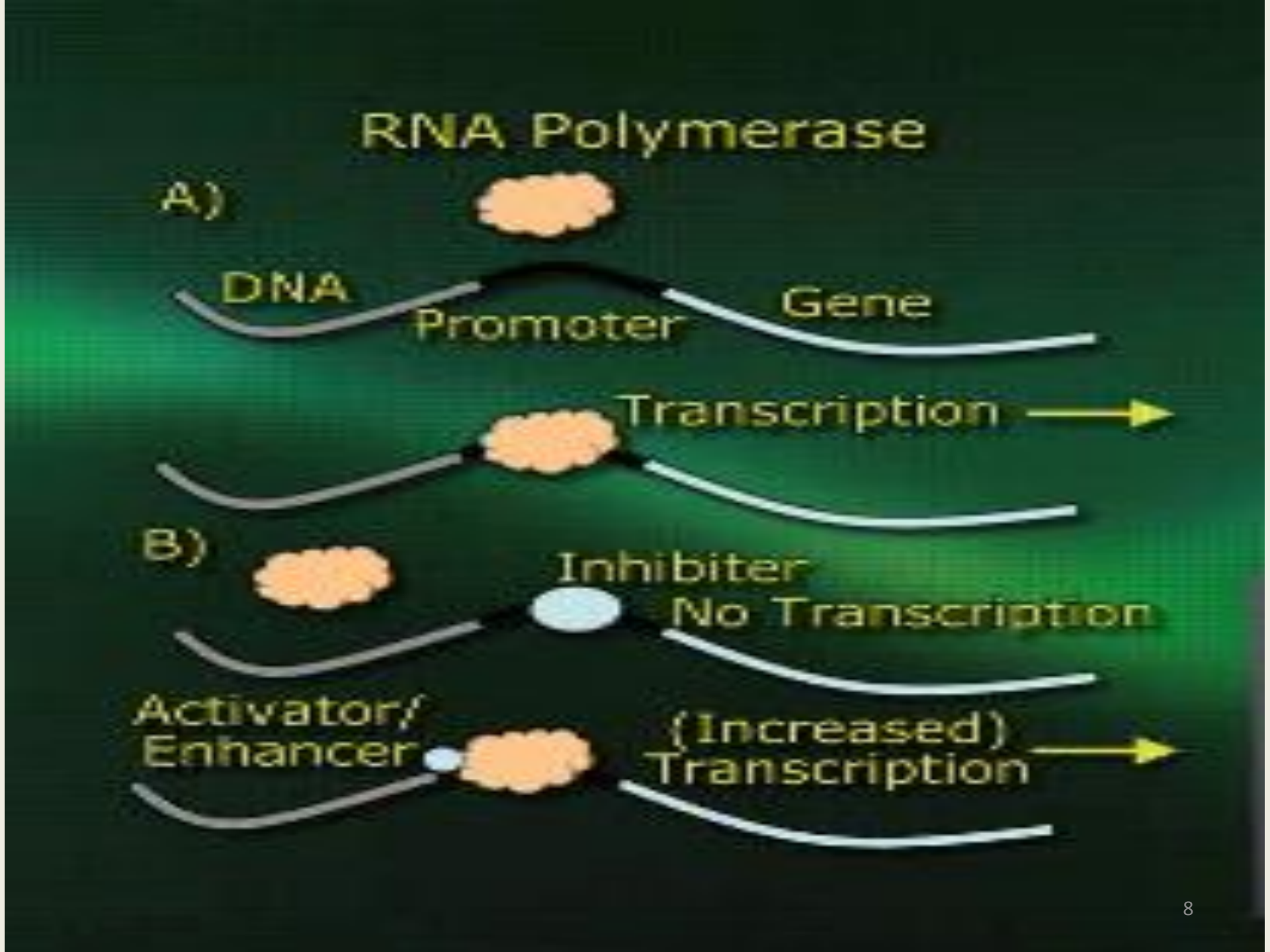


Transcription (DNA → mRNA)

- is the synthesis of **mRNA** molecules within the cell **nucleus** with the code for a protein copied from the genetic information contained in the DNA.
- Transcription produces an exact copy of a section of DNA known as messenger RNA (mRNA).
- **mRNA** carries complementary **genetic code** copied from DNA during transcription, in the form of **triplets of nucleotides** called **codons**.
- **A codon** is a sequence of three nucleotides (a trinucleotide) on an mRNA derived from the DNA that encodes a specific amino acid or stop signal.
- Each codon specifies a particular amino acid.
- There are only 20 naturally existing amino acids but the number of possible amino acids combination is = **64 triplets**.
- For 20 amino acids used for protein synthesis and 4 bases in DNA/mRNA there are **64 triplet codons** with the possible combination 4^3 = 64.
- Out of the 64 codons, **61** codes for amino acids and **3** are stop/termination codons.

DNA needs RNA!

Initiation → Elongation → Termination → Splicing


RNA Polymerase
A)
DNA
Promoter
Gene
Transcription
B)
Inhibiter
No Transcription
Activator/
Enhancer
(Increased)
Transcription

"Start" and "Stop" Signals

- **UAA, UAG** and **UGA** are **stop codons** (do not code for amino acids).
- **Stop codons** signify the end of a coding sequence/amino acid sequence or stop/terminate the process of protein synthesis.
- The codon **AUG**, which encodes the amino acid **methionine** is the **start codon** that initiates protein synthesis.

⇒ During Transcription:

1. An enzyme RNA polymerase binds to the promoter region of DNA (the starting point of transcription).
2. **Transcription factors** (proteins that control the rate of transcription) also bind to the promoter sequences with RNA polymerase.
3. RNA polymerase **unwinds** a portion of the DNA double helix, exposing the bases on each of the two DNA strands.
4. Free RNA nucleotides attach to appropriate base pairs on the DNA template and mRNA is formed with code (triplets of nucleotides called **codons**) for protein synthesis.
5. mRNA detaches from the DNA.and post transcriptional modification occurs.
6. mRNA leaves the nucleus to go out into the cytoplasm and binds to ribosomes.

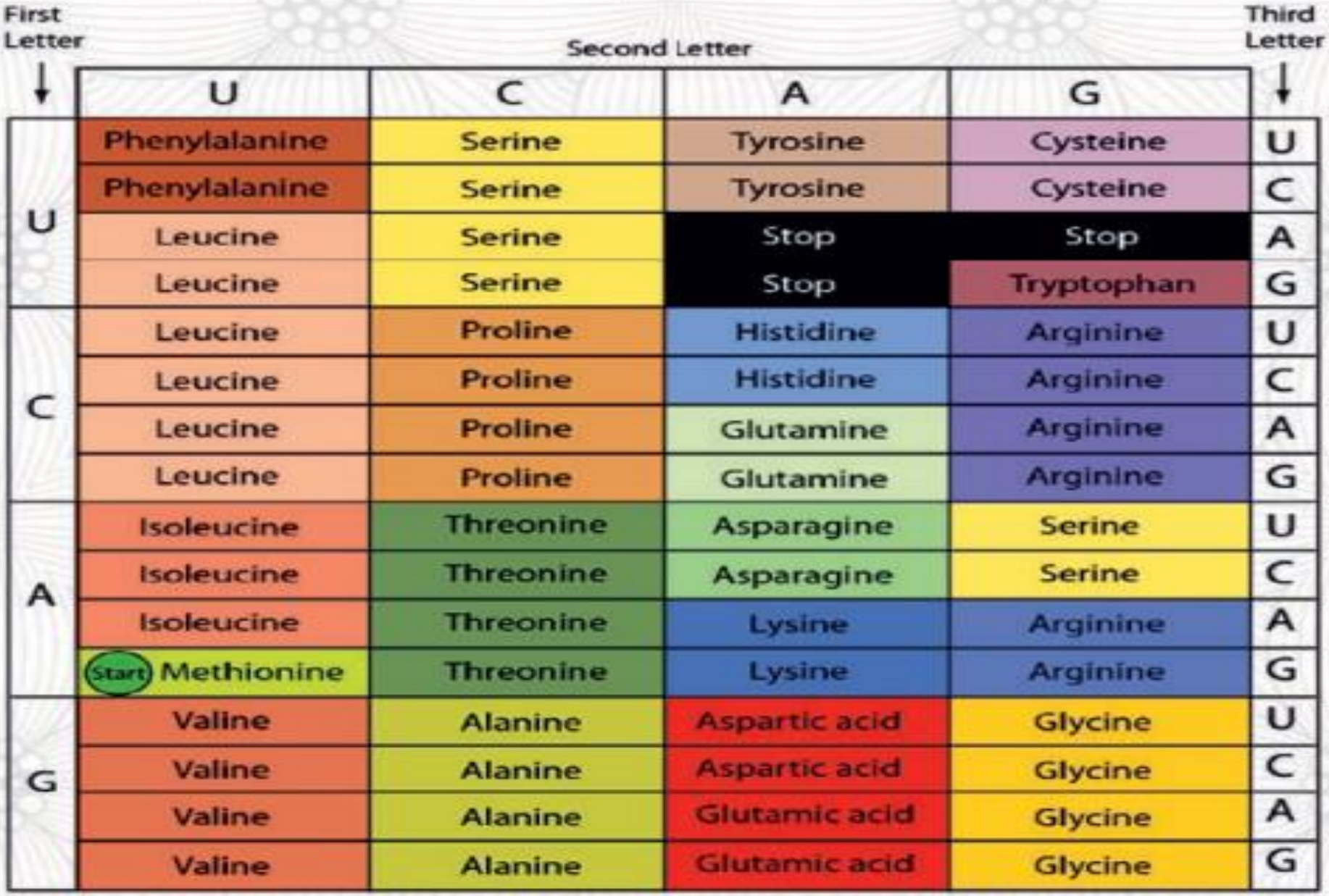


Figure 4.23 The genetic code

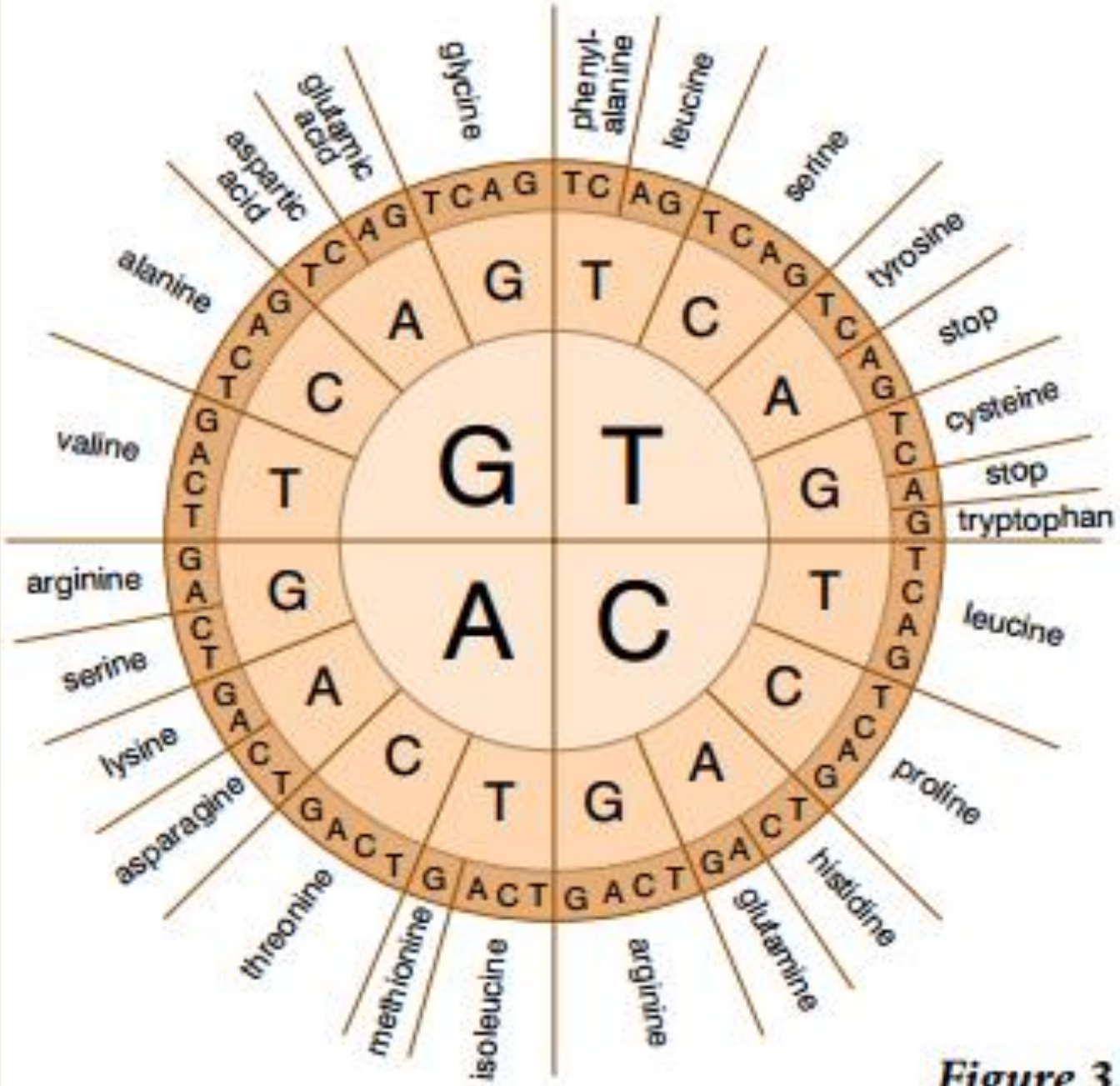


***Figure 3.47A** The genetic code*

Characteristics of the genetic code

1. **A triplet code** – sequence of three bases
2. **A degenerative code** – all amino acids except methionine (**AUG**) and tryptophan (**UGG**) are coded by several codons:
3. **A non-overlapping code** – each triplet is distinct from the next (5' – 3').
4. **A universal code** – the same in all (most) living organisms.
5. **A commaless code** – no punctuation between codons.
6. **Non-ambiguous code** – not having a double meaning.

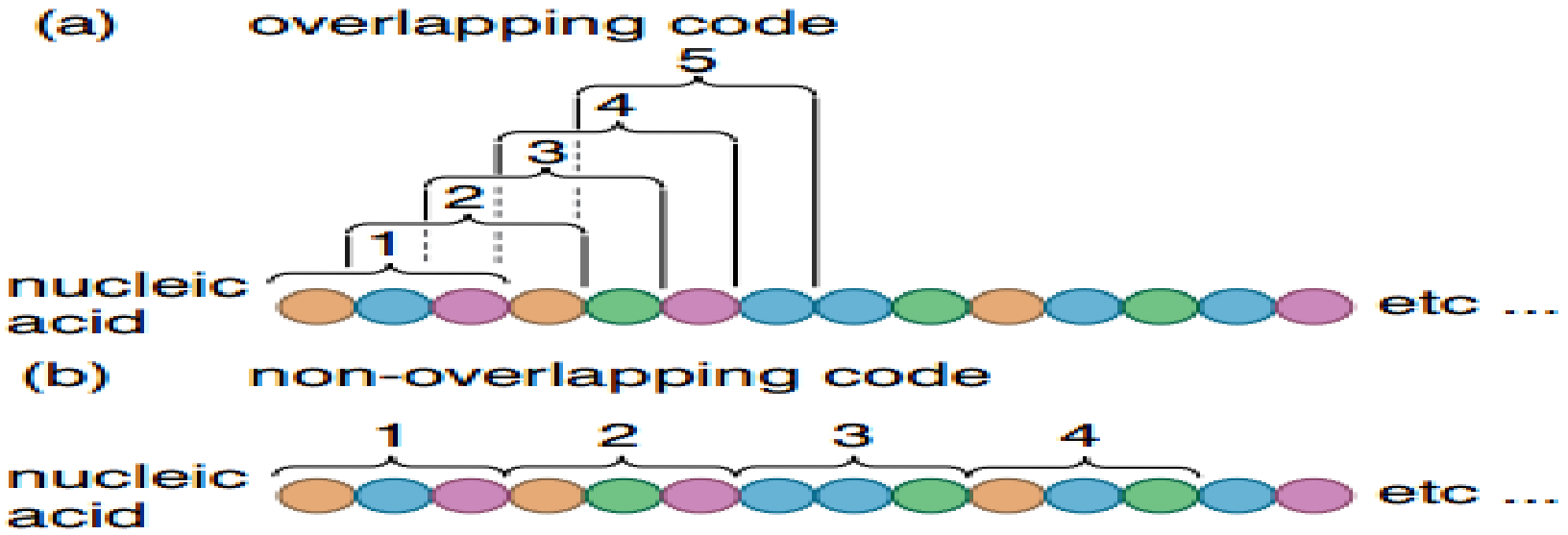
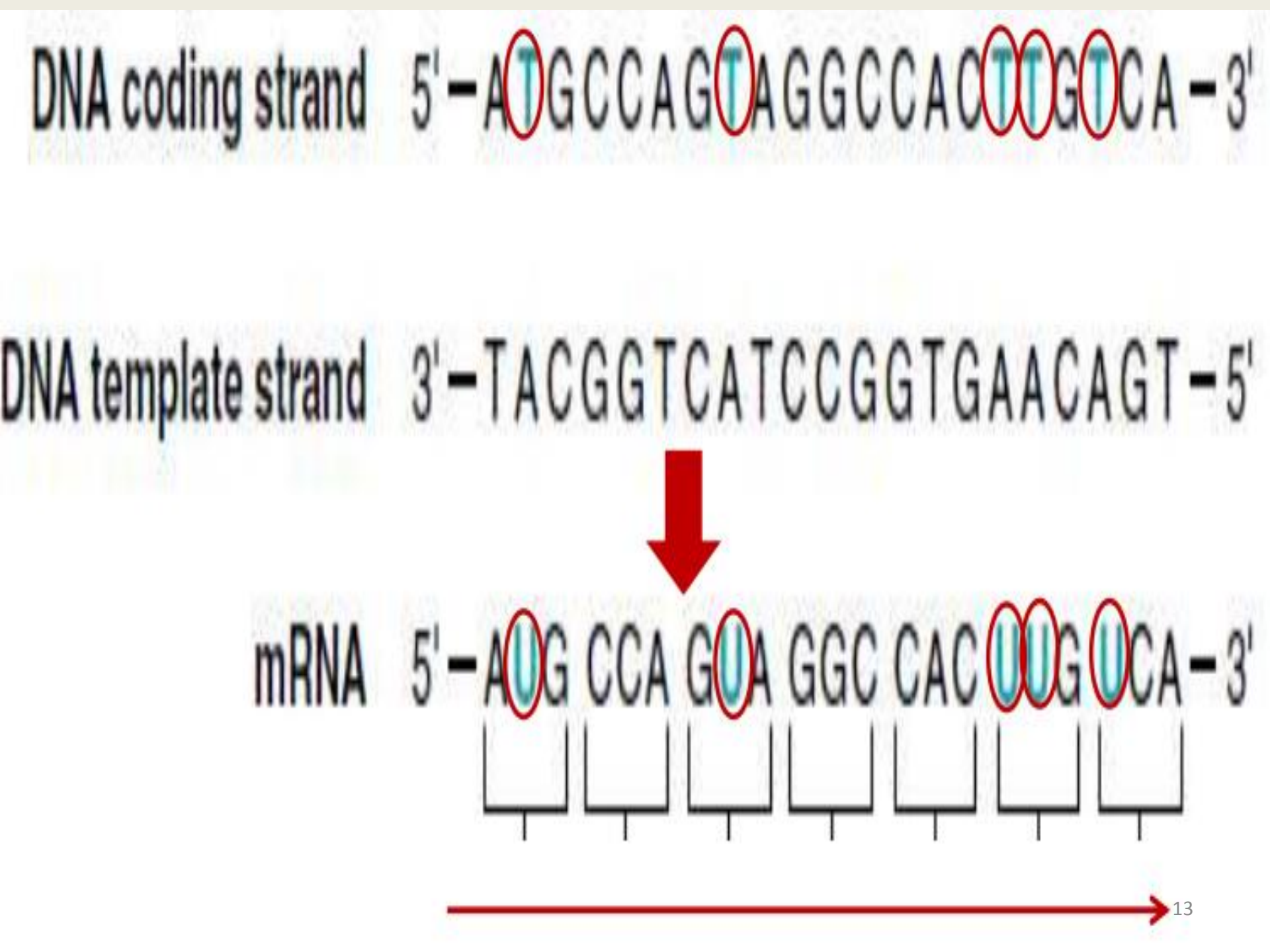


Figure 3.46 Overlapping and non-overlapping codes


DNA coding strand 5'-ATGCCAGTAGGCCACTTGTCA-3'
DNA template strand 3'-TACGGTCATCCGGTGAACAGT-5'
mRNA 5'-AUG CCA GUA GGC CAC UUG UCA-3'

Transcription Practice?

Transcribe this non- coding DNA strand into a mRNA strand
*Remember: No Thymine in RNA. Instead use Uracil.   A=U

DNA

AAA TAG GAT ATC GGA TAC AGT

RNA

UUU AUC CUA UAG CCU AUG UCA

Translation (RNA → Protein)

- is the **synthesis of protein** from amino acids based on the genetic information instructed on mRNA with the help of rRNA, tRNA and enzymes.
- Transfer RNA (tRNA) carries a specific amino acid from cytoplasm to the ribosome.
- The tRNA contains an **anticodon** which is **complementary** to the **codon** on the mRNA.
- The anticodon on tRNA enables to recognize the codon of mRNA through complementary base pairing.
- **For example**, the genetic codon GUG (specifies an amino acid valine) binding with the complementary anticodon (CAC).
- The tRNA acts as an adapter, bringing the specific amino acid based on base complementarily.
- The ribosome guides the tRNA to bind to the mRNA if it is carrying an amino acid.

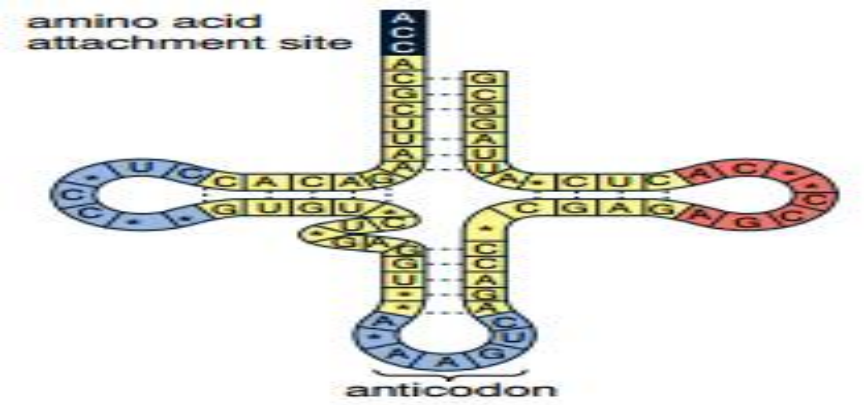


Figure 3.49 *The structure of tRNA*

✍ During translation:

1. mRNA carries the information from DNA align on the ribosome in the cytoplasm.
2. The ribosomes attach on to mRNA and let the tRNA loaded with specific amino acid to enter.
3. tRNA with anti-codon brings amino acids from the cytoplasm to the ribosomes.
4. The anti-codon of tRNA pairs with the codon of mRNA on the ribosome.
5. The information in messenger RNA (mRNA) translated into protein with the help of rRNA.
6. A polypeptide chain of amino acids will then form a protein.

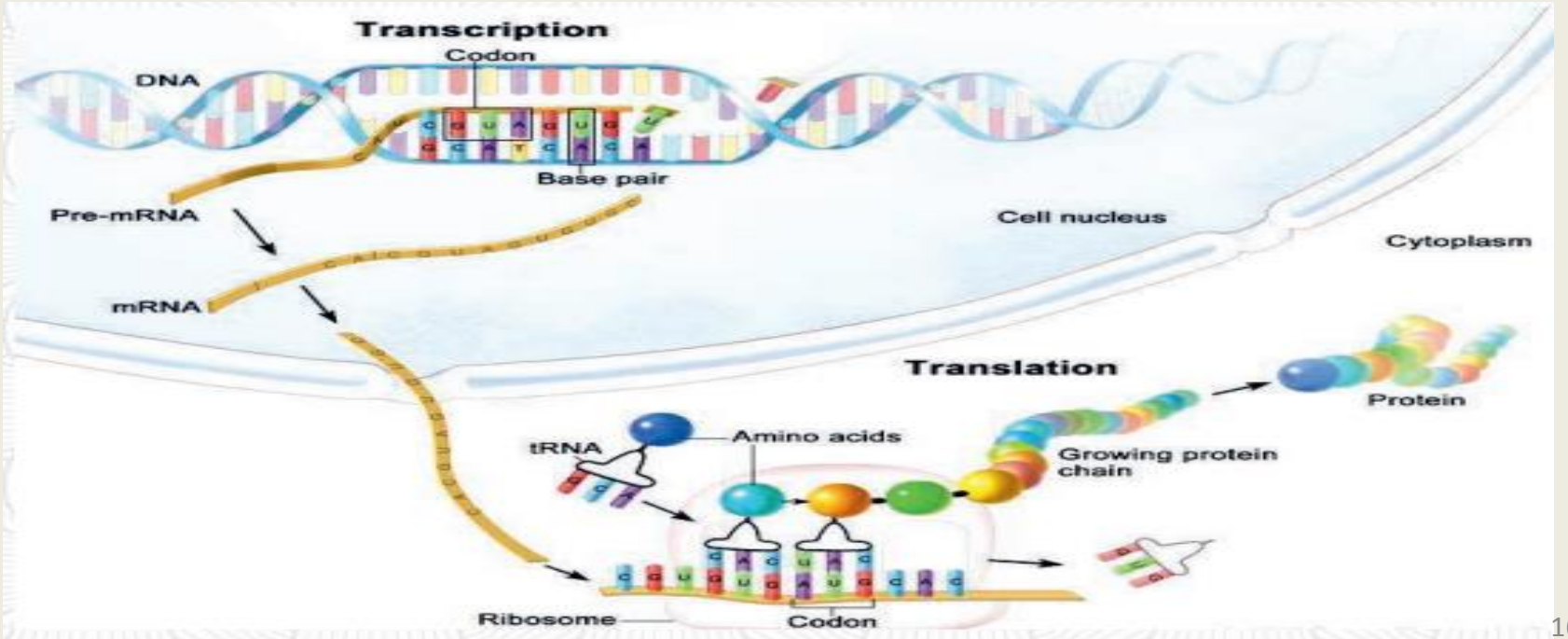


Figure 4.24 Transcription and translation in protein synthesis.

Exercise 1

I. Answer the following questions accordingly.

1. What process is involved in the synthesis of mRNA from DNA?
2. Where does transcription occur in eukaryotic cells?
3. What is produced during transcription?
4. What enzyme is responsible for transcribing DNA into RNA?
5. What do we call the region of DNA where transcription begins?
6. How many codons are there in the genetic code?
7. List the stop/termination codons that signals the end of a polypeptide chain.
8. Write the start codon. What type of amino acid is coded by start codon?
9. If a protein contains 100 amino acids, how many bases would there be in the mRNA that controls the production of this protein?
10. What molecule carries the genetic code from DNA to the ribosome?
11. What type of RNA carries amino acids to the ribosome?

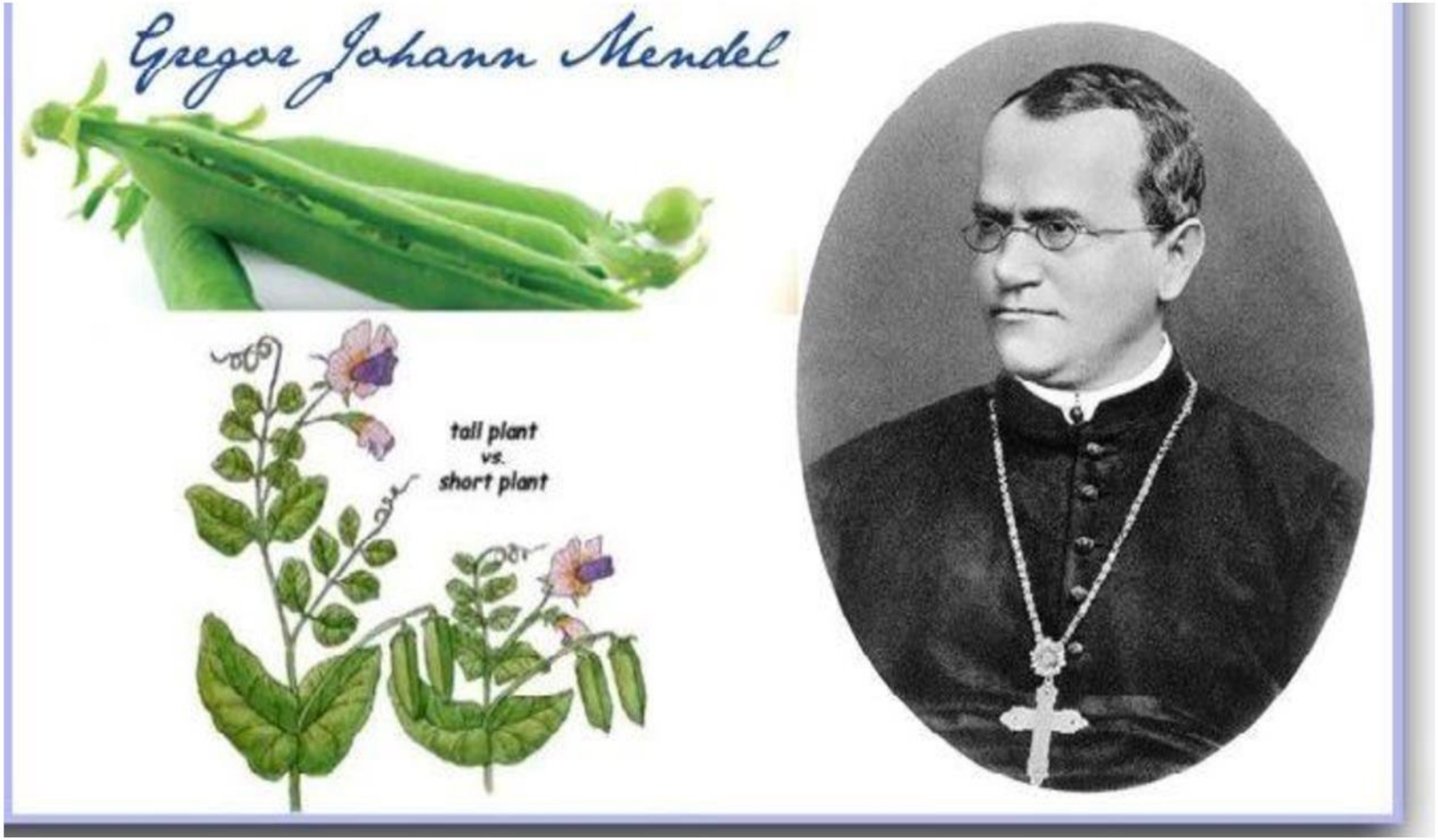
Exercise 2

I. Answer the following questions accordingly.

3’-CCG ATA CGC GGT ATC CCA GGG CTA-5’

1. If the above DNA strand is a template for the synthesis of mRNA, what would be the base sequences on mRNA?
2. Where does translation takes place?
3. In the genetic code dictionary, how many codons are used to code for all the 20 essential amino acids?
4. How many codons are needed to specify three amino acids?
4. Write the correct order of information flow or central dogma of modern biology?
5. How many nucleotides are needed to form condones for a protein that is 100 amino acids long?
6. What is the three letter sequences on mRNA molecule called?
7. What is the three letter sequences on tRNA molecule called?
8. Suppose the amino acid coding region in a mRNA is 1200 nucleotides long, how long is the protein in terms of amino acid number?
9. If the codon on mRNA reads **GUC**, what would be the anticodon on tRNA?
10. Describe the characteristics of the genetic code.

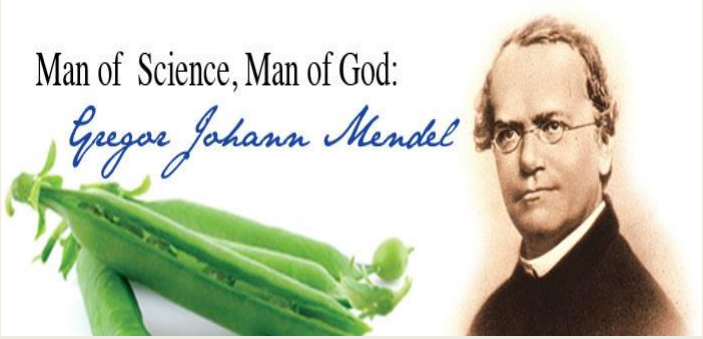
MENDELIAN GENETICS



Basic introductory questions

1. What is genetics?
2. What is heredity or biological inheritance?
3. Who 1st studied about the mechanism of inheritance?
4. What kind of organism did Mendel used for his investigations? Give reason.
5. How did Mendel conduct his experiment?
6. What is trait?
7. Gene Vs allele?
8. Dominant allele Vs Recessive allele?
9. Genotype Vs phenotype?
10. Homozygous Vs heterozygous?

4.5 Mendelian inheritance



Gregor Johann Mendel (1822 – 1884)

- was an Austrian monk (in Brno monastery).
- the "father" of genetics.
- discovered the **principles of inheritance** or **heredity**. 1864
- his work on heredity is the basis for the modern theory of genetics.
- he chose **garden peas (*Pisum sativum*)** for his work and investigation.
- Mendel chose **seven contrasting traits** of pea plants for his work.

- **Mendelian inheritance** refers to the patterns of inheritance of traits or transmission of traits, controlled by a single gene with two alternative alleles, from parent to offspring.

Why did Mendel chose garden pea (*Pisum sativum)* for his investigation?

⇒ Because **garden pea (*Pisum sativum*):**

- is easy to cultivate or grow.
- is a self pollinating plant.
- has many easily observable contrasting traits (7).
- the observable traits are controlled by single factors (genes).
- has short generation time or life span.

Terminologies related with Mendelian inheritance

1. **Gene -** *a coding section of DNA (chromosome) that determines a specific feature.*
2. **Allele -** *a version or form of a gene that determines a particular trait.*
3. **Locus** *- the position of a particular gene on a chromosome.*
4. **Homologous chromosome -** *a pair of chromosome with corresponding genes that together determine the same trait.*
5. **Trait** - a specific characteristic of an individual that can be determined by genes.
5. **Homozygous** – having identical or the same alleles for a gene on each of the chromosomes in the homologous pairs.

   Example: PP, pp, AA, aa, YY, yy …
6. **Heterozygous** – having different alleles for a gene on each of the chromosomes in the homologous pairs.

   Example: Pp, Aa, Rr, Yy …
7. **Genotype** – describes the pair of alleles for a particular gene possessed by an organism.

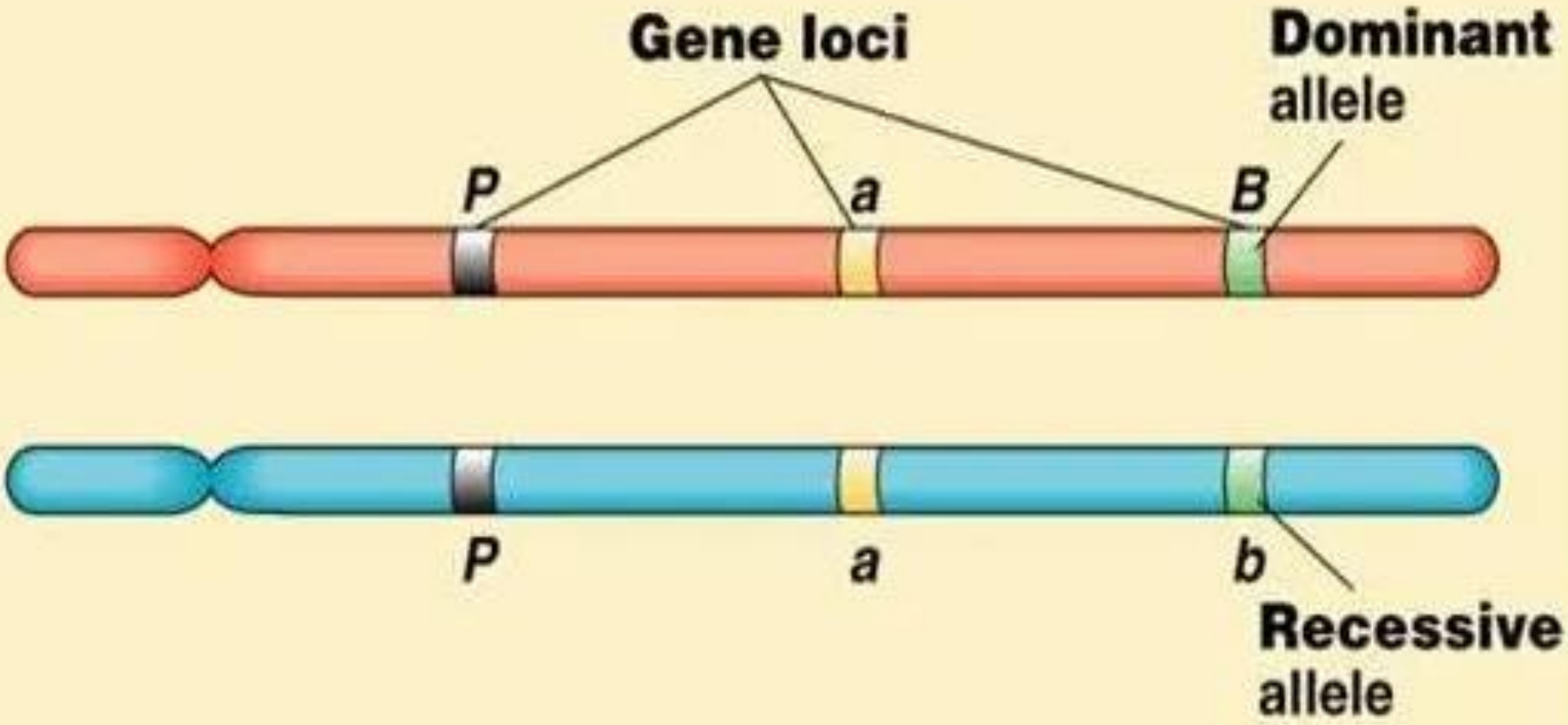
   - the genetic make up of an organism relating to a particular trait.

   Example: PP, Pp, pp, AA, Aa, aa, YY…
8. **Phenotype** - describes the trait or traits determined by a particular genotype.

   - the physical/external appearance or morphology of an organism relating to a particular trait.

   Example: Purple, short, round, wrinkled, green, yellow…

9. **Dominant allele** – an allele where the characteristics is expressed in the phenotype (external appearance) even if only one copy of the allele is present.
   - masks or hides the expression of recessive alleles.
   - written by upper case or capital letters.
10. **Recessive allele** – an allele where the characteristics is expressed in the phenotype (physical appearance) only if two copies of the alleles are present.
   - masked or hidden by dominant alleles.
   - written by lower case or small letters.
11. **Punnet square** - a chart used to determine the expected ratios of the possible genotypes and phenotypes in the offspring of two parents (probabilities).
12. **Genome** - is the total genetic composition of an organism.
13. **Monohybrid cross** - a cross between the two parents that are differing in one pair of alternating (contrasting) character (a single trait).
14. **Dihybrid cross** - a cross between the two parents that are differing in two pairs of alternating characters (two distinct traits).
15. **Test cross** - an experimental cross of an individual organism of dominant phenotype but unknown genotype and an organism with a homozygous recessive genotype (and phenotype).



Genotype: *PP* *aa* *Bb*

Homozygous for the dominant allele

Homozygous for the recessive allele

Heterozygous

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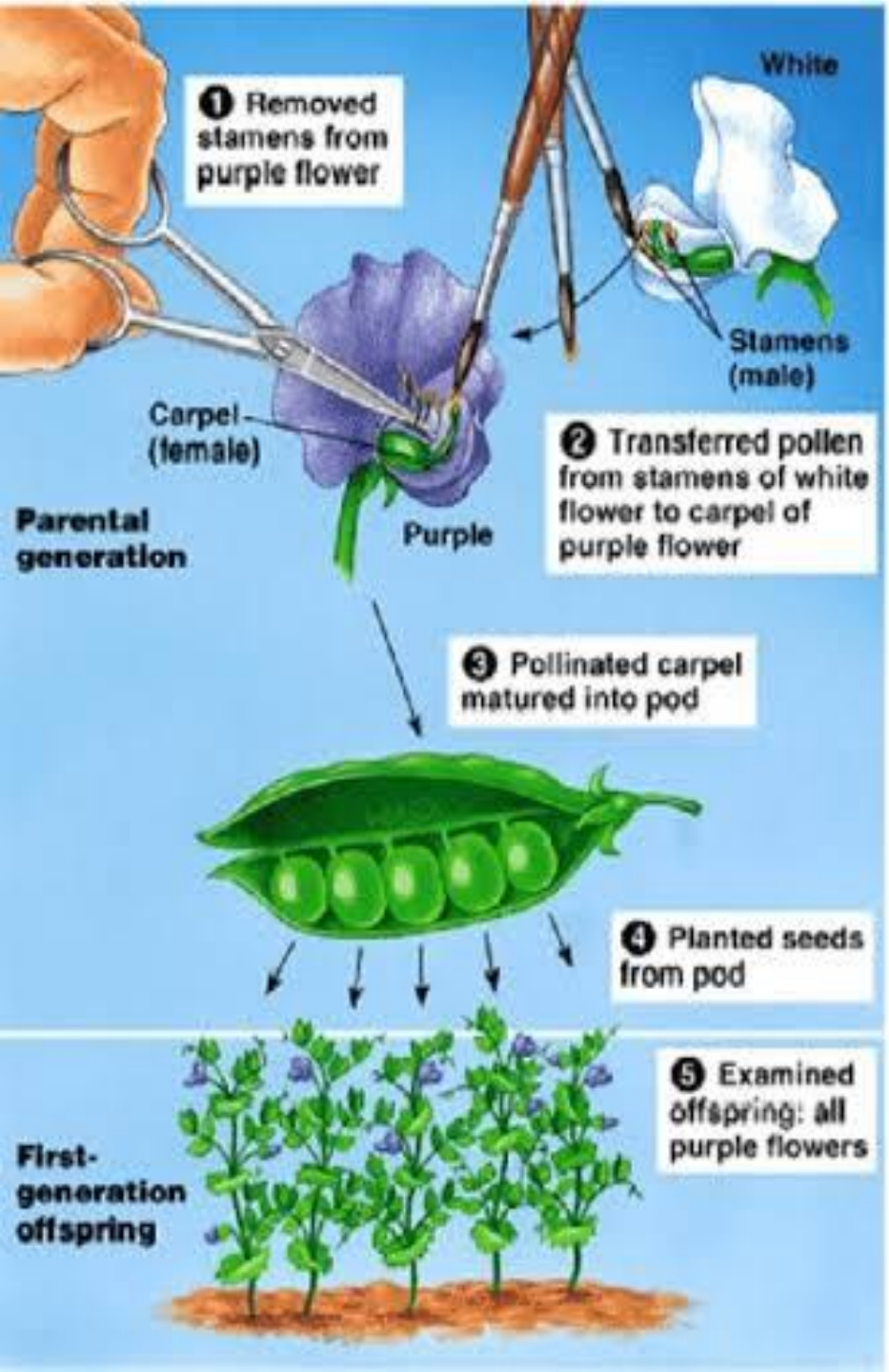

1 Removed stamens from purple flower
White
Stamens (male)
Carpel (female)
Purple
Parental generation
2 Transferred pollen from stamens of white flower to carpel of purple flower
3 Pollinated carpel matured into pod
4 Planted seeds from pod
5 Examined offspring: all purple flowers
First-generation offspring

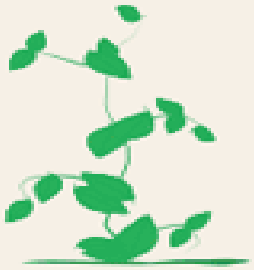
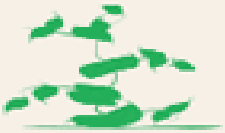
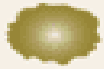
4.5.1 Mendelian Crosses

- Mendel conducted his experiments on pea plants (Pisum sativum) by cultivating them, crossing them with each other, and observing the pattern of inheritance in different stages of generation.
- Mendel studied seven pairs of different characters of pea plants with contrasting traits that exist in two forms.
- **The seven pairs contrasting traits of garden pea with dominant and recessive forms.**

S. No.	Character	Dominant	Recessive
1	Stem height	Tall	Dwarf
2	Flower colour	Violet	White
3	Flower position	Axial	Terminal
4	Pod shape	Inflated	Constricted
5	Pod colour	Green	Yellow
6	Seed shape	Round	Wrinkled
7	Seed colour	Yellow	Green

- **In order to determine the mechanism of inheritance of these seven pairs of contrasting traits from parents to offspring, Mendel conducted different types of crosses such as monohybrid crosses, dihybrid crosses and test crosses.**

The 7 contrasting traits of pea plants studied by Mendel

Characteristic studied	Dominant character	Recessive character
Stem length	Long 	Short 
Flower Position	Axillary	Terminal
Flower colour	Blue	White
Pod shape	Inflated	Constricted
Pod colour	Green	Yellow
Seed shape	Round	Wrinkled 
Seed colour	Yellow	Green

Genotype vs Phenotype

GENOTYPE

The genotype is an organism's genetic information.

BB
homozygous dominant

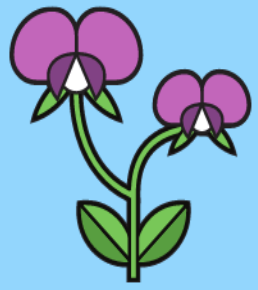
Bb
heterozygous

bb
homozygous recessive

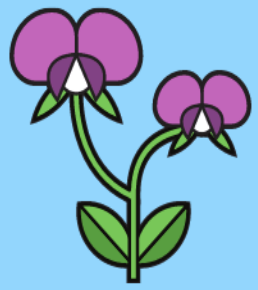
PHENOTYPE

The phenotype is the set of observable physical traits.

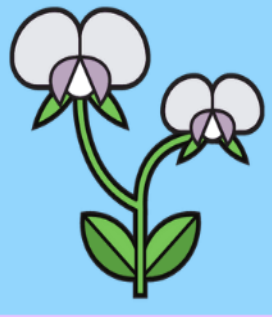
purple



purple



white



5.4.2 Monohybrid Cross

- **A monohybrid cross** is a cross between two parents to study the **inheritance of a single character** from each parent.
- Mendel performed seven types of monohybrid crosses, each involving contrasting traits for different characteristics.

⇒ **Mendel conducted his experiment as follows:**

1. He conducted a self-cross to checked the breeding line (he used **pure/true-breeding** lines for each trait) of all the traits (14 traits).
2. He **removed stamens** from the dominant traits to **prevent self-pollination**.
3. He transferred pollen grain using a brush from the dominant traits to the carpel of the recessive traits. (He made **artificial cross-pollination**).
4. He called the resultant offspring **F1 generation**. In the F1 generation, one of the traits was always seen in the offspring but not the other one. Why?
5. The F1 generations were then self-pollinated which gave rise to the **F2 generation**. In the F2 generation, both traits were observed.
6. Mendel counted the number of second-generation (F2) progeny with dominant or recessive traits and found a **3:1** ratio of dominant to recessive traits.
7. He called the characters that appear in the F1 generation **dominant** traits and those that appear for the first time in the F2 generation **recessive** traits.

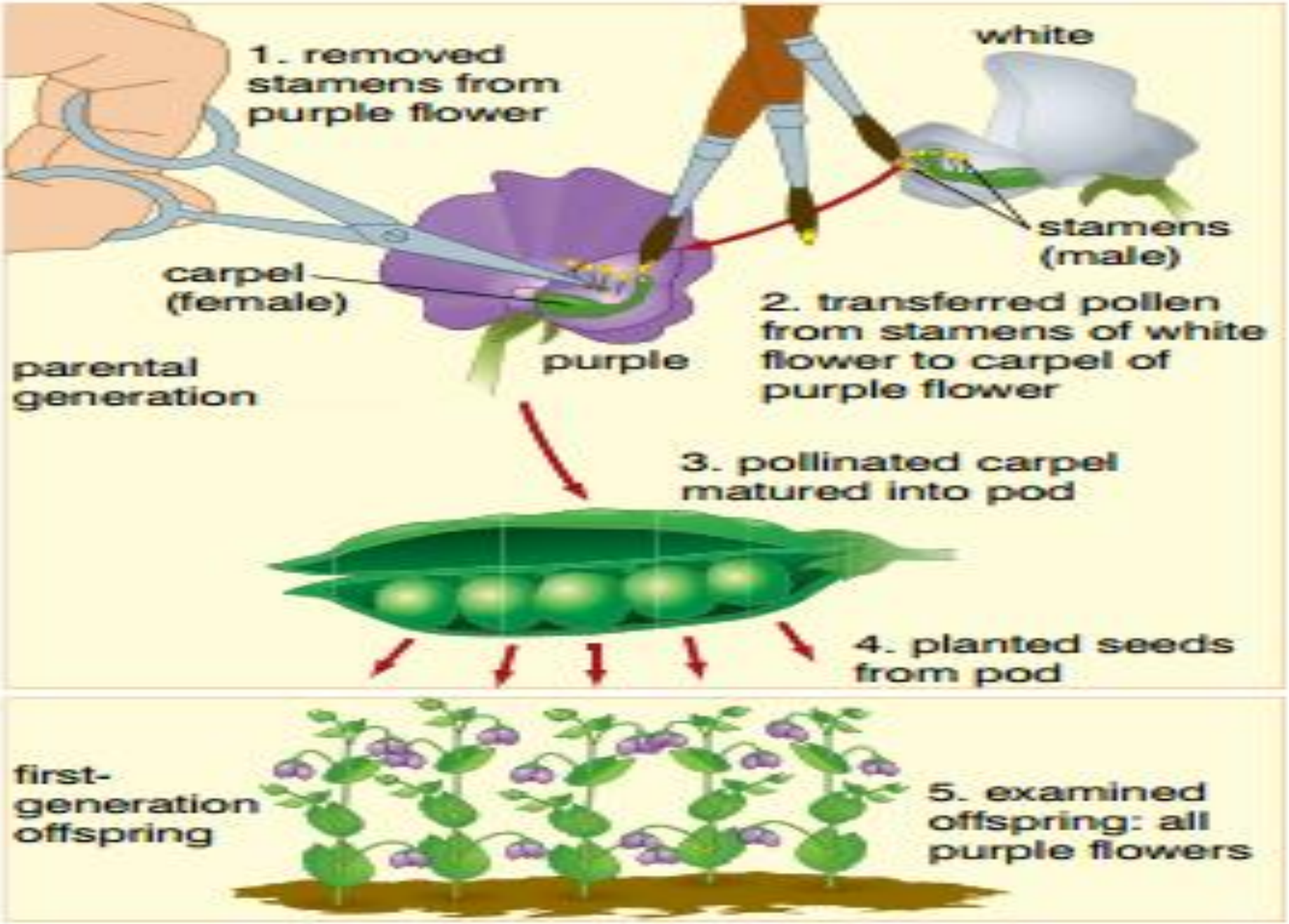


Figure 3.5 Mendel's technique

- **Example:** A monohybrid cross for tall plants with homozygous dominant genotype (TT) and dwarf/short plants with homozygous recessive genotype (tt) using the Punnett square in the F1 and F2.

P generation: Phenotype = Tall X short

Genotype = TT X tt

Possible gametes = T T t t

F1

X	t	t
T	Tt	Tt
T	Tt	Tt

F1 Generation;

Genotype: All Tt

Phenotype: All tall (heterozygote or hybrid)

- The F1 generations self-crossed and produce F2 generations. F2 = F1 X F1

Phenotype = Tall X Tall

Genotype = Tt X Tt

Possible gametes = T t T t

F2

X	T	t
T	TT	Tt
t	Tt	tt

F2 Generation;

Genotype: 1TT : 2Tt : 1tt (1:2:1)

Phenotype: 3 tall : 1 short (3:1)

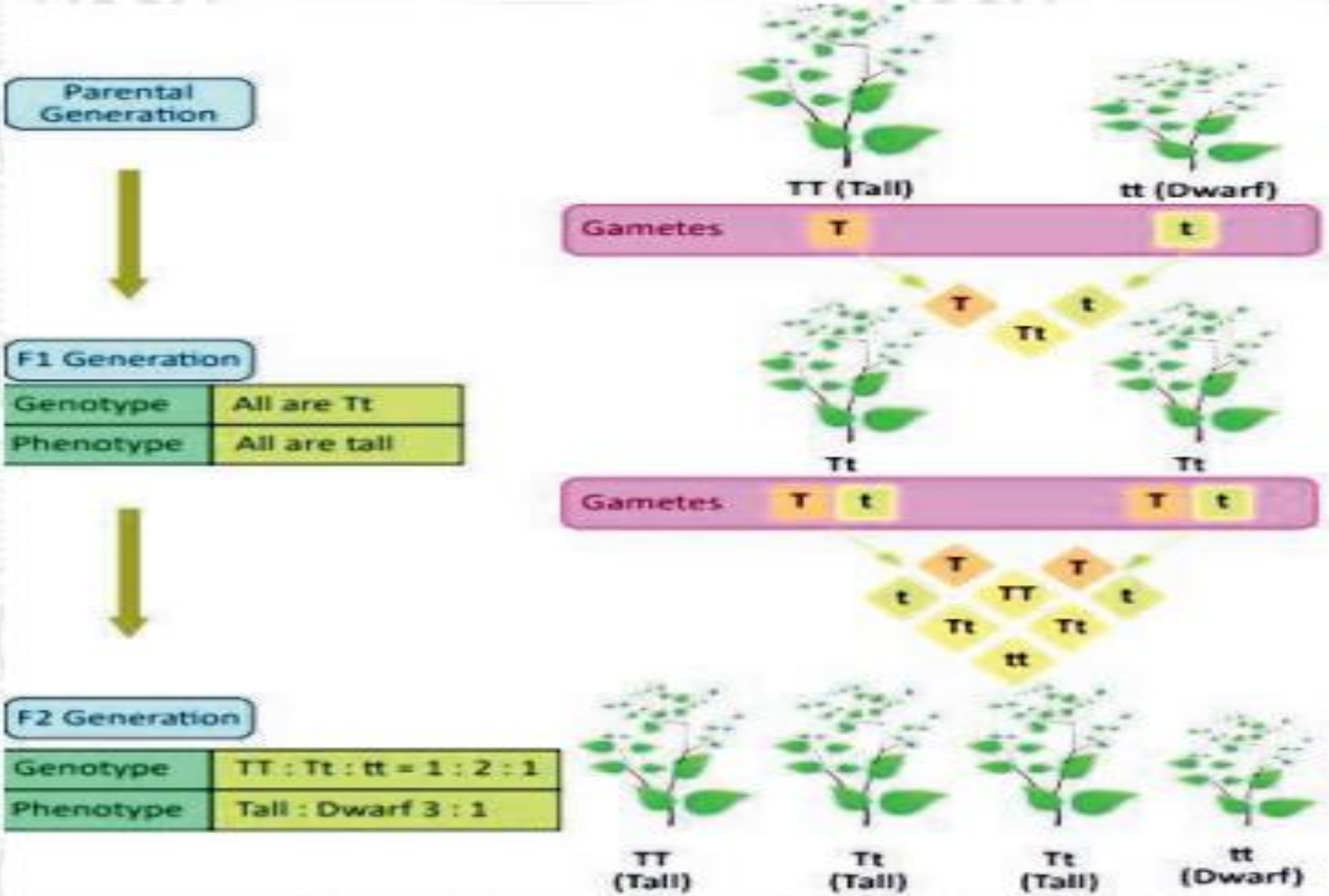


Figure 4.26. Monohybrid cross

Table 4.3 number of F2 generations and phenotypic ratios from Gregor Mendel

Characters	P1 crosses Dominant traits x recessive traits	F1	F2	Dominant : Recessive Ratio
Flower color	Purple x white	All purple	Dominant (705) : Recessive (224)	3.15:1
Flower position	Axial x terminal	All axial	Dominant (651) : Recessive (207)	3.14:1
Seed color	Yellow x green	All yellow	Dominant (6022) : Recessive (2001)	3.01:1
Seed shape	Round x wrinkled	All round	Dominant (5474) : Recessive (1850)	2.96:1
Pod shape	Smooth or inflated x constricted	All inflated	Dominant (882) : Recessive (299)	2.95:1
Pod color	Green x yellow	All green	Dominant (428) : Recessive (152)	2.82:1
Stem length	Tall x short	All tall	Dominant (787) : Recessive (277)	2.84:1

Table 3.1 Mendel's results

Character	Dominant trait	×	Recessive trait	F_2 generation dominant : recessive	Ratio
Flower colour	purple	×	white	705:224	3.15:1
Flower position	axial	×	terminal	651:207	3.14:1
Seed colour	yellow	×	green	6022:2001	3.01:1
Seed shape	round	×	wrinkled	5474:1850	2.96:1
Pod shape	constricted	×	inflated	882:299	2.95:1
Pod colour	green	×	yellow	428:152	2.82:1
Stem length	tall	×	dwarf	787:277	2.84:1

Mendel drew the following conclusions:

1. Every trait is controlled by two 'heritable factors'. One factor is **dominant** and one is **recessive** **(complete dominance).**
2. The only physical link between the generations is the gametes or sex cells. These must pass the factors from one generation to the next.
3. The heritable factors (alleles) separate when the gametes (sex cells) are formed; each gamete therefore contains only one allele controlling the trait.
4. Each offspring inherits one factor from each parent. If the dominant factor is present, it will be expressed even if the recessive factor is also present.
5. The recessive factor will be expressed if only recessive factors are present.
6. He also stated that the **gametes** (sex cells) **fuse randomly** at fertilization.

N.B: - Mendel studied each of the seven contrasting traits separately.

- He studied each trait in a controlled condition (controlled experiment).
- He used pure/homozygous/true-breeding plants for each traits.

Exercise 3

I. **Draw genetic diagrams to show the offspring that would result from the following crosses.**

1. Homozygous yellow-seed colored pea plant and homozygous green-seed colored pea plant *(Y-yellow, y-green*)
2. Heterozygous purple-flowered plant and white-flowered plant *(P-purple, p-white)*
3. Two heterozygous round-seeded pea plants *(R-round. r-wrinkled)*
4. A heterozygous yellow-seeded plant and a green-seeded plant

4.5.3 Dihybrid cross

- **Dihybrid cross** is a cross between two traits of individuals at a time.
- It means the inheritance of **two traits** controlled by **two distinct genes** at the same time.
- During dihybrid cross **4 possible gametes** are formed from each parent and **16 possible offspring's** are produced during fertilization.
- The phenotypic ratio of F2 generation in dihybrid cross is **9:3:3:1.**

Example: A cross between homozygous round, yellow seeded pea plant (RRYY) and homozygous wrinkled, green seeded pea plant (rryy). Work out in the F1 & F2.

P generation: Phenotype = Round, yellow X Wrinkled, green

Genotype = RRYY X rryy

Possible gametes (4) = **RY, RY, RY, RY** X **ry, ry, ry, ry**

F1

X	ry	ry	ry	ry
RY	ry	ry	ry	ry
RY	ry	ry	ry	ry
RY	ry	ry	ry	ry
RY	ry	ry	ry	ry

F1 Generation;

Genotype: All RrYy

Phenotype: All round, yellow

F1		Male gametes			
Generation	X	RY	Ry	rY	ry
Female gametes	RY	RYRY	RRYy	RrYY	RrYy
	Ry	RRYy	RRyy	RrYy	Rryy
	rY	RrYY	RrYy	YrYr	Yrry
	ry	Rr Yy	Rryy	rrYy	rryy

F2 generation

- Phenotype
  - Round Yellow = 9
  - Round green = 3
  - Wrinkled Yellow = 3
  - Wrinkled green = 1
  - 9:3:3:1phenotypic ration

Exercise: **What will be the phenotypic ratio if a heterozygote round, yellow pea plant crossed with wrinkled, green pea plant?**

Laws of Mendel

- From monohybrid and dihybrid crosses, Mendel discovered the two foundational laws of inheritance.

1. **Law of segregation** – **Mendel's 1st law.**

   - states that copies of genes randomly separate or segregate during meiosis (gamete formation) so that each gamete receives only one allele
   - the gametes (sex cells) fuse randomly at fertilization.

- The physical basis of Mendel's law of segregation is the first division of meiosis in which the homologous chromosomes with their different versions of each gene are segregated into daughter nuclei.

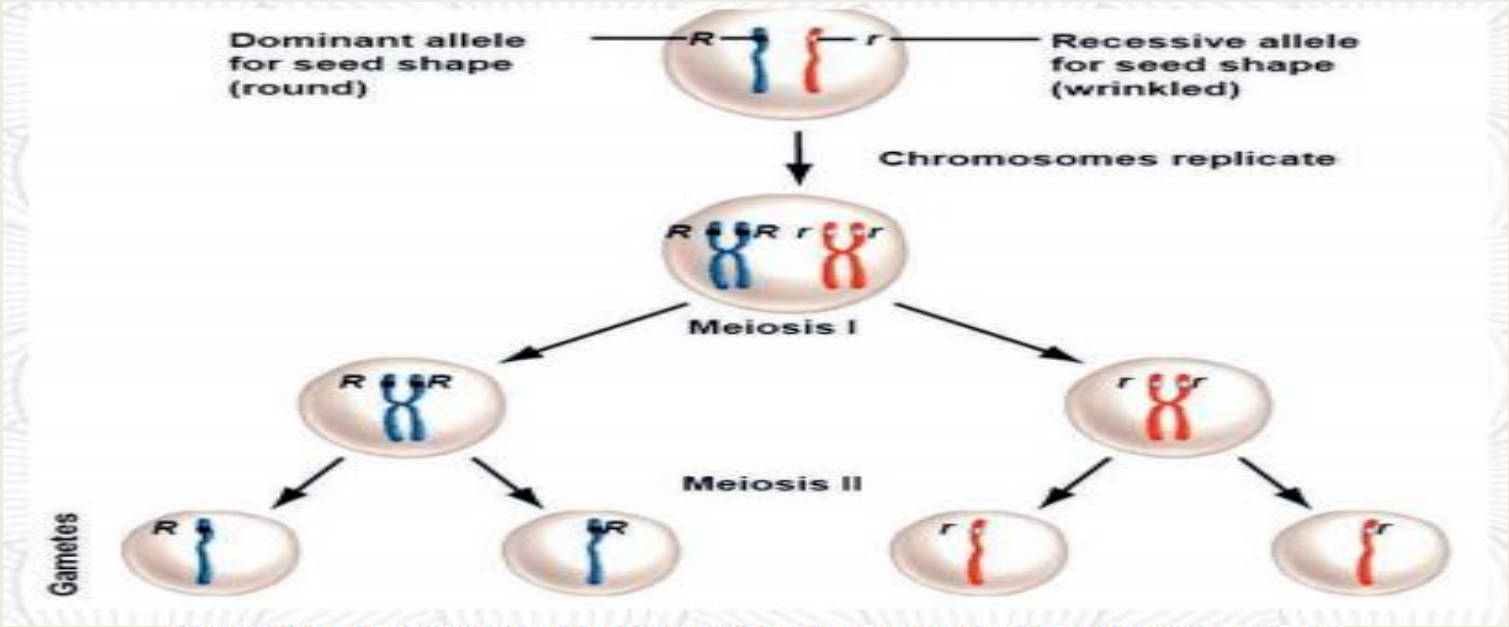


Figure 4.27 Segregation of maternal and paternal alleles into separate gametes

2. Law of independent assortment – Mendel's 2nd law.

- states that genes do not influence each other with regard to the sorting of alleles into gametes.
- states that the inheritance of one trait is independent of the inheritance of the other trait.
- the alleles of one pair segregate independently of the alleles of another pair controlling a different feature.

- The Law of independent assortment occurs in metaphase I of meiosis when pairs of chromosomes randomly align next at the center.

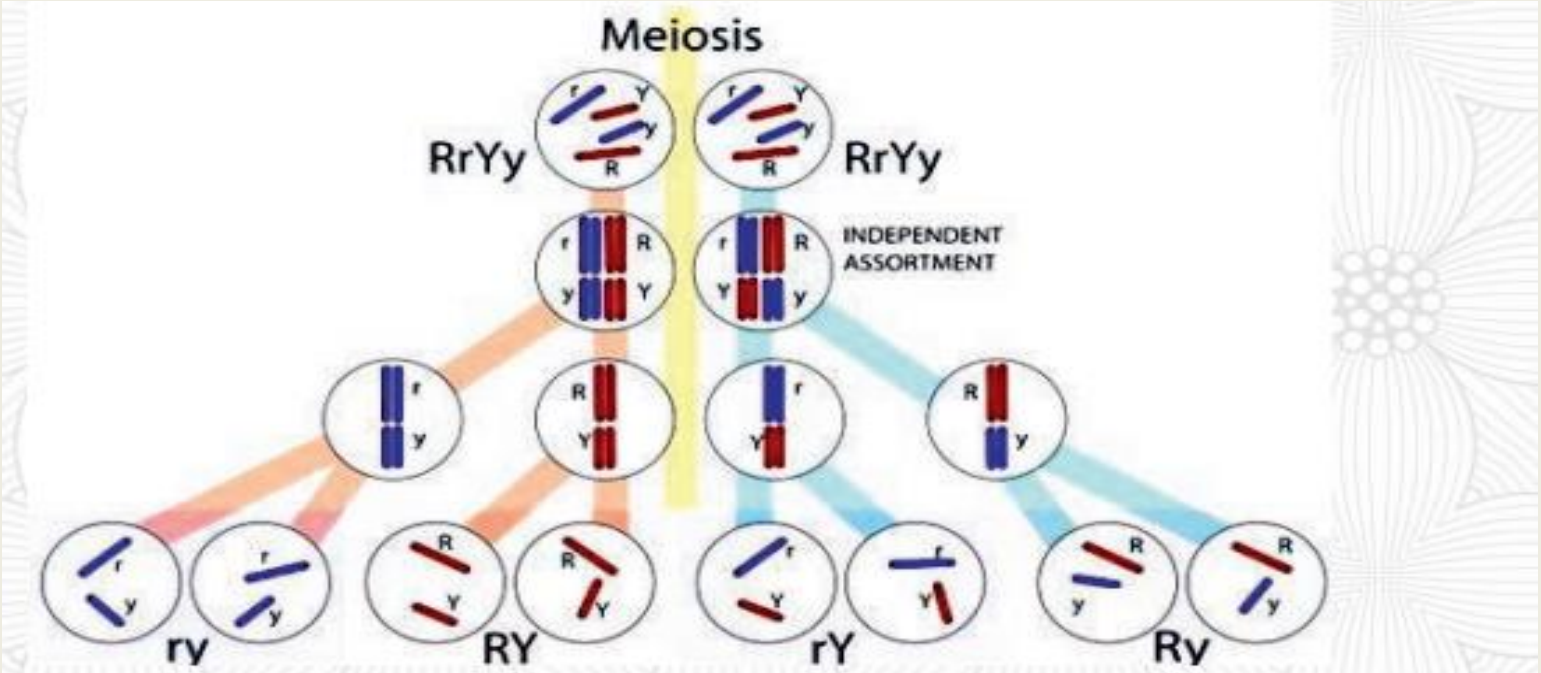


Figure 4.28 Independent assortment of alleles during gametes formation

4.5.4 Test Crosses

- The particular experimental cross to find out whether an organism is homozygous or heterozygous for a dominant trait is called the test cross.
- **Test cross** is used to **determine the unknown genotype** of an organism by crossing with a known **homozygous recessive** genotype.
- Individuals which are homozygous dominant or heterozygous will look exactly the same or have identical phenotypes.
- When we cross the unknown parent with homozygous recessive & **if the dominant trait is observed in all progenies**, the unknown genotype is **homozygous dominant**.
- When we cross the unknown parent with homozygous recessive & **if the recessive trait is manifested in any of their progenies**, the unknown genotype is **heterozygous dominant.**

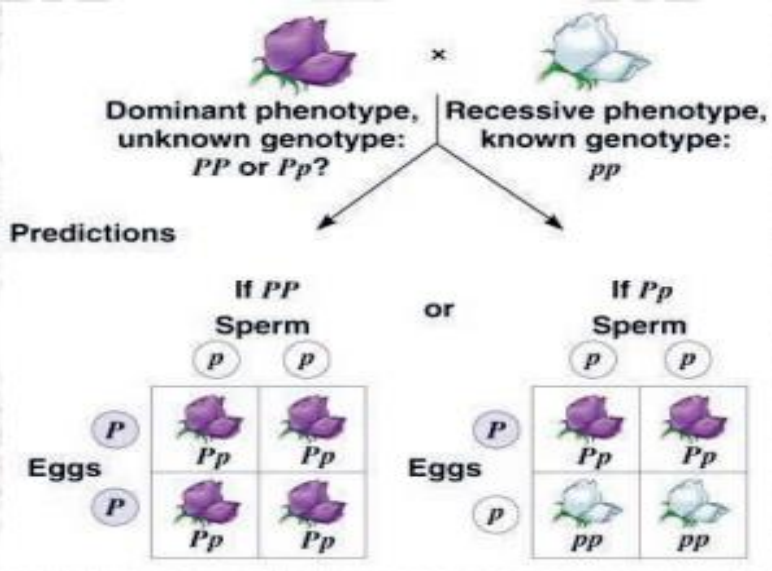


Figure 4.29 Test cross All purple 2 purple: 2 white = 1:1 phenotypic ratio

- **During dihybrid test cross, following breeding the dominant trait with a double recessive type (rryy), there are the following possible outcomes:**

1. If the progenies produced show **all the four possible phenotypes**, the original was **heterozygous for both features**.
2. If the progenies produced **all show the dominant phenotypes of both traits**, the original was **homozygous for both features**.
3. If the progenies produced **all show dominant phenotypes of one trait** but **some show both the dominant and recessive phenotypes of the other trait**, the original was **heterozygous for one trait** and **heterozygous for the other trait**.

Back cross

- is the mating of a hybrid organism (offspring of genetically unlike parents) with one of its parents or with an organism genetically similar to the parent.
- is a cross between the F1 hybrid individual and either of the two parents (either recessive/or dominant).
- Backcross is useful in genetics studies for isolating (separating out) certain characteristics in a related group of animals or plants.

Exercise 5

I. Answer the following questions accordingly.

1. Suppose in a monohybrid cross, 160 F2 plants were produced, what is the number of the plants that are expected to have the dominant and the recessive phenotypes?
2. A genetic cross between two F1 –hybrid pea plants having yellow seeds (dominant) will yield what percent of green-seeded (recessive) plants in the F2 generation?
3. Regarding seed characteristics, suppose a heterozygous round, yellow pea plant of RrYy genotype is selfed and produced 256 seeds, how many of the seeds are expected to be round, yellow?
4. In a cross between round, green pea of RRyy genotype and wrinkle, yellow pea of rrYY genotype, what is the expected genotype of the F1?
5. When the F1 hybrid of a monohybrid cross is back crossed with the homozygous recessive parent, what percentage of the offspring would be homozygous recessive?
6. Supposing that 1000 individuals were produced from a cross between Bb and bb, how many of them are expected to have the Bb genotype?

Mendelian genetics in humans

How can parents showing one version of a feature have children with the other version?

Exercise 6

I. Answer the following questions accordingly..

1. Suppose a man homozygous for free earlobes (E) and a woman homozygous for attached earlobes (e) have a child. Determine the possible genotypes and phenotypes of a child?
2. Suppose a man and a woman are heterozygous for free earlobes have a child. What will be the possible genotypes and phenotypes of a child?
3. Two parents of genotype Aa are cross-bred. The alleles show complete dominance. What proportion of the offspring will phenotypically look like their parents?
4. In human albinism is determined by a recessive autosomal gene. If two parents produced a legitimate child who is albino, the child has most likely inherited the defect from:
5. If two normally pigmented persons heterozygous for the same albinism allele married, what proportion of their children would you expect to be albino?

4.6 Sex determination in humans

- Map of chromosomes in the nucleus of a cell is called **karyotype**.
- Human karyotype shows 46 (23 Pairs) chromosomes.
  - 44 (22 pairs) – **autosomes** (non-sex chromosomes), and
  - 2 (1 pair) – **allosomes** (sex chromosomes).
- Normal human **males** - **XY** (**heterogametic** sex)
- Normal human **females** – **XX (homogametic** sex).
- In any mating in mammals, the predicted ratio of males to females is **1:1 (50% : 50%).**

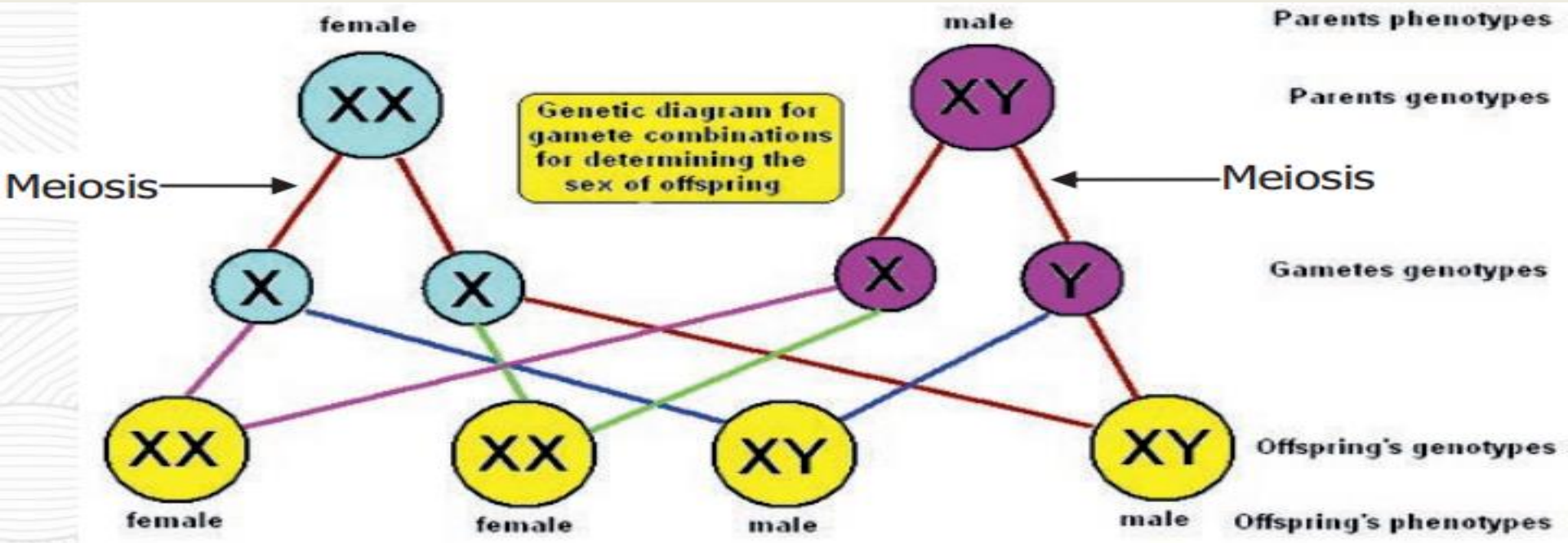
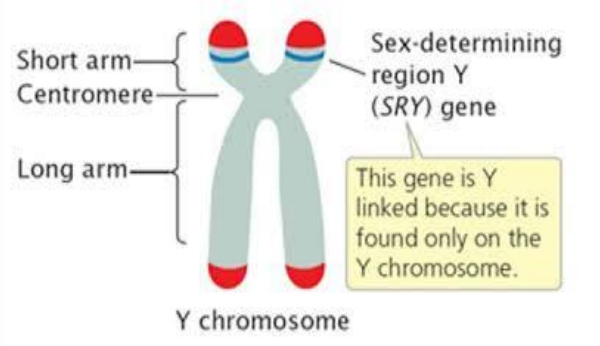


Figure 4.30 Sex determination

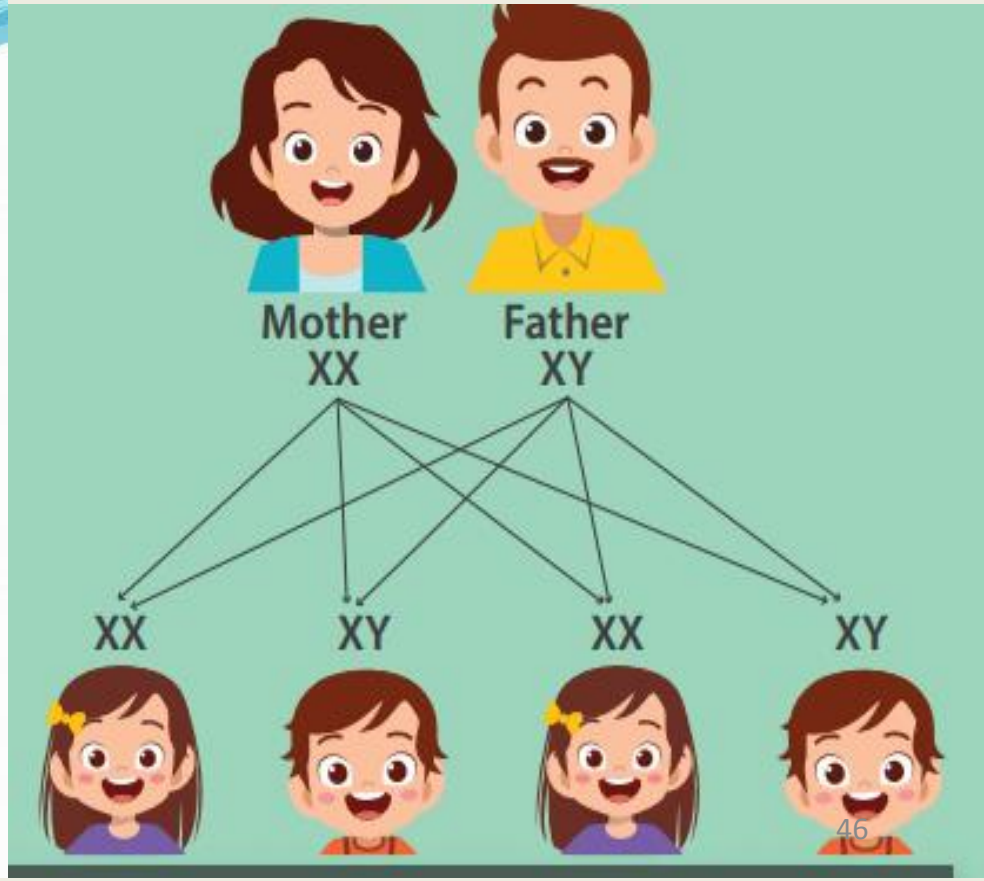
What exactly determines sex in humans?

- The **SRY gene** on Y chromosome determine a person's sex.
- **SRY gene** – determines the formation of testes.
- When the SRY gene is activated, the **bi-potential gonad** develops into a testis, and the embryo becomes male.
- If the SRY gene is absent, genes on the X chromosome and on the autosomes cause the bi-potential gonad to develop into an ovary, and the embryo becomes female.

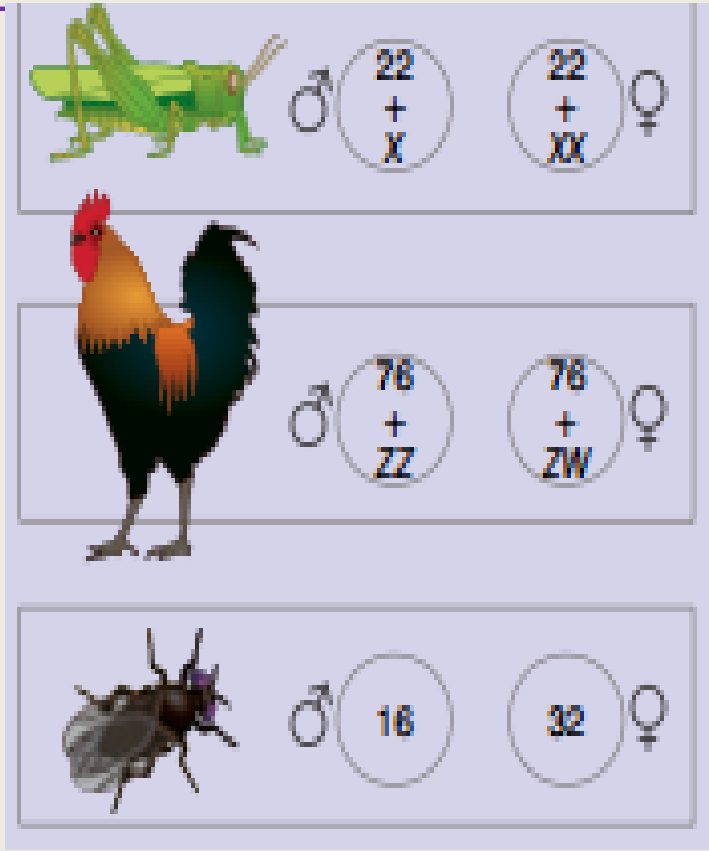
The SRY gene: The Male determining factor.



- SRY gene is located on the short arm of the Y chromosome
- This Gene causes male gonadal development.



Sex determination in other organisms



1. In birds such as chickens (**ZW and ZZ**)

- **females** are heterogametic sex (ZW) and
- **males** are homogametic sex (ZZ)

2. In some insects such as grasshopper (**XX and X**)

- **females** have two X chromosomes (XX)
- **males** have only one X chromosome (X)

3. In some insects such as honeybees (**haplodiploidy**)

- **Females** (queens and workers) are diploid (2n)
- **Males** (drones) are haploid (n)

4. In reptiles such as alligators (**temperature**)

- **females** – from eggs incubated at low temperature.
- **males** – from eggs incubated at high temperature.

5. Some species such as earthworm are **hermaphrodite** (have both male and female sex organs), have no sex-determination system (**hermaphroditism**).

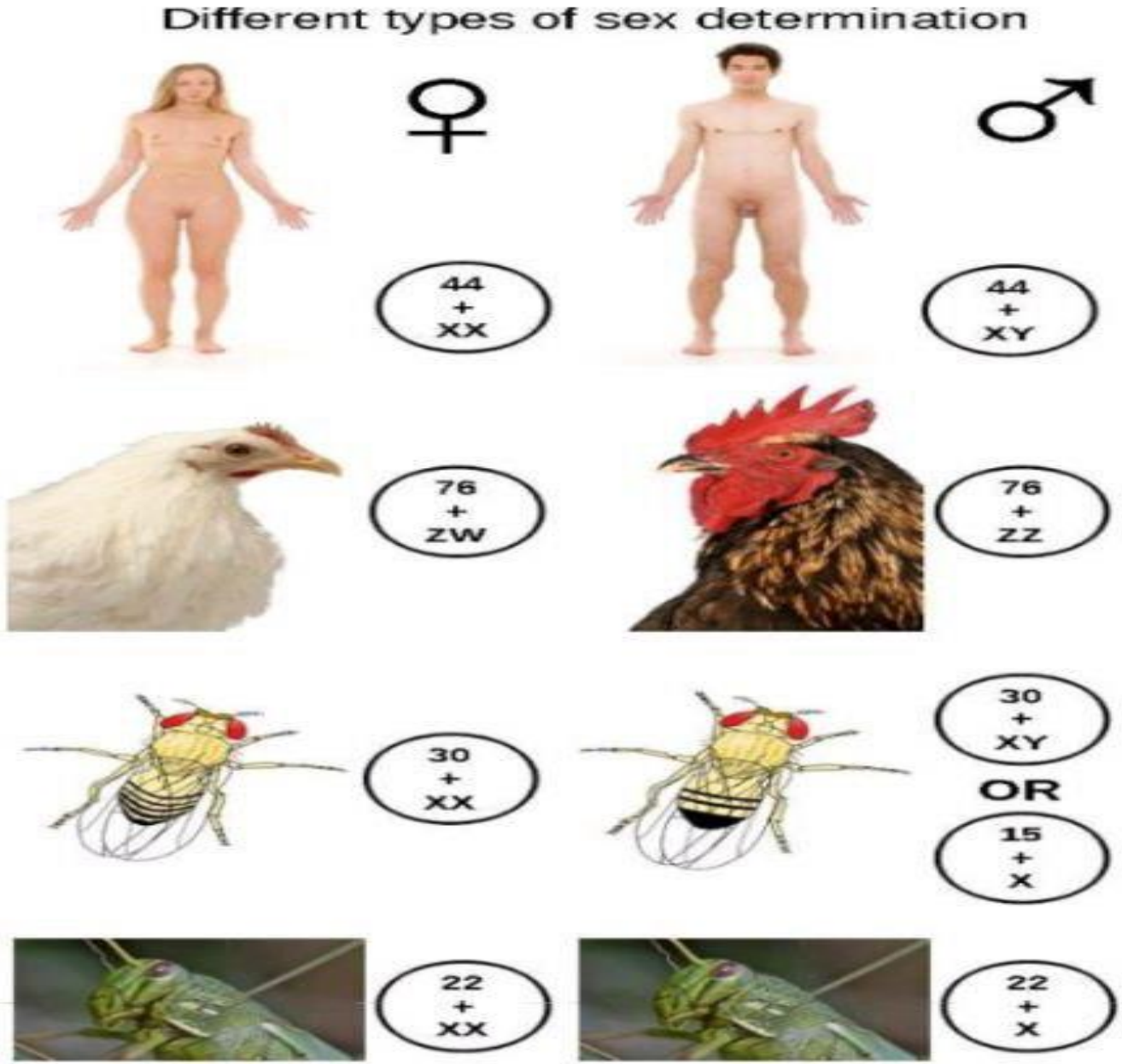
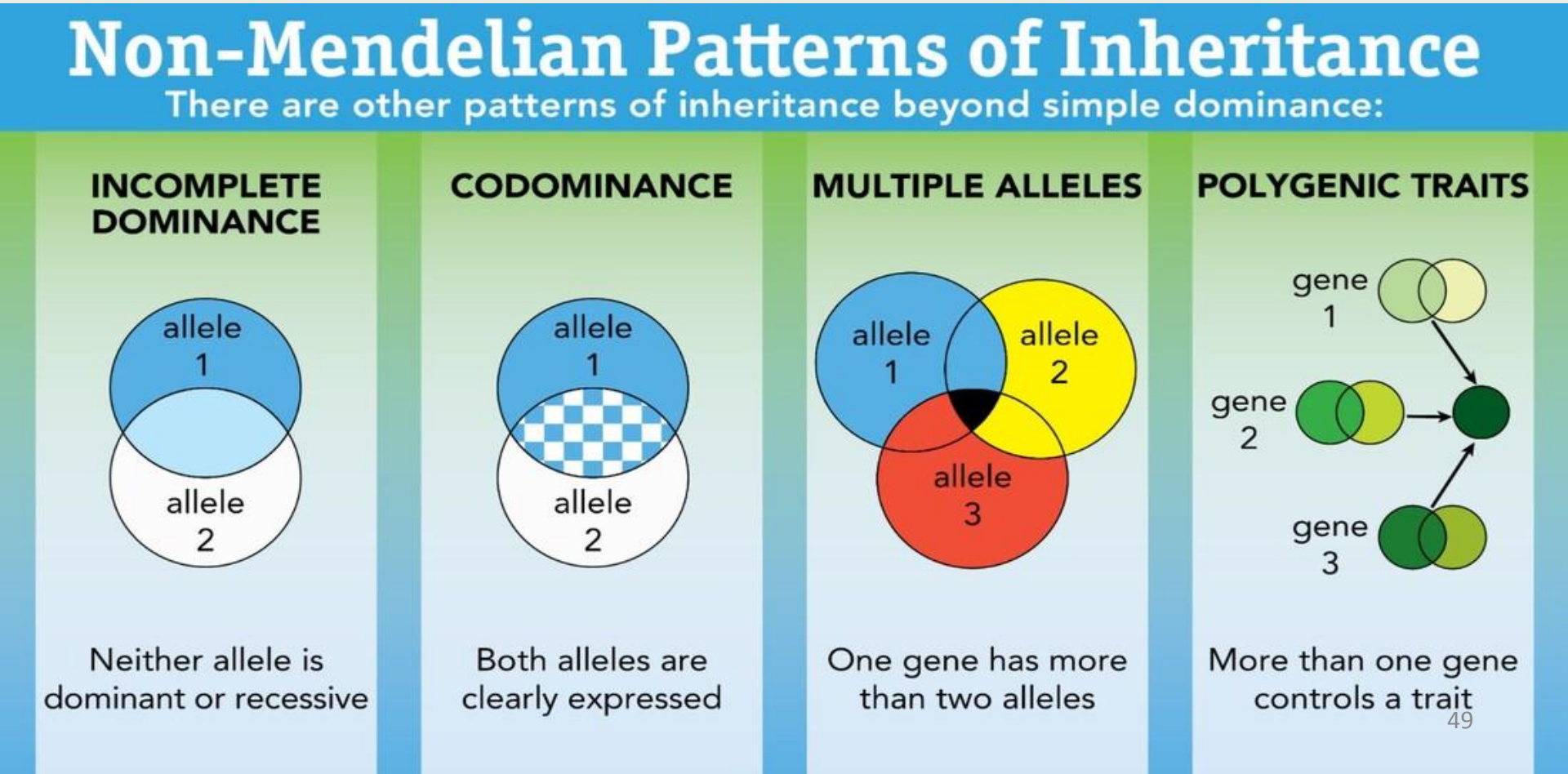


Figure 4.2. Sex determination

4.7 Non-Mendelian inheritance

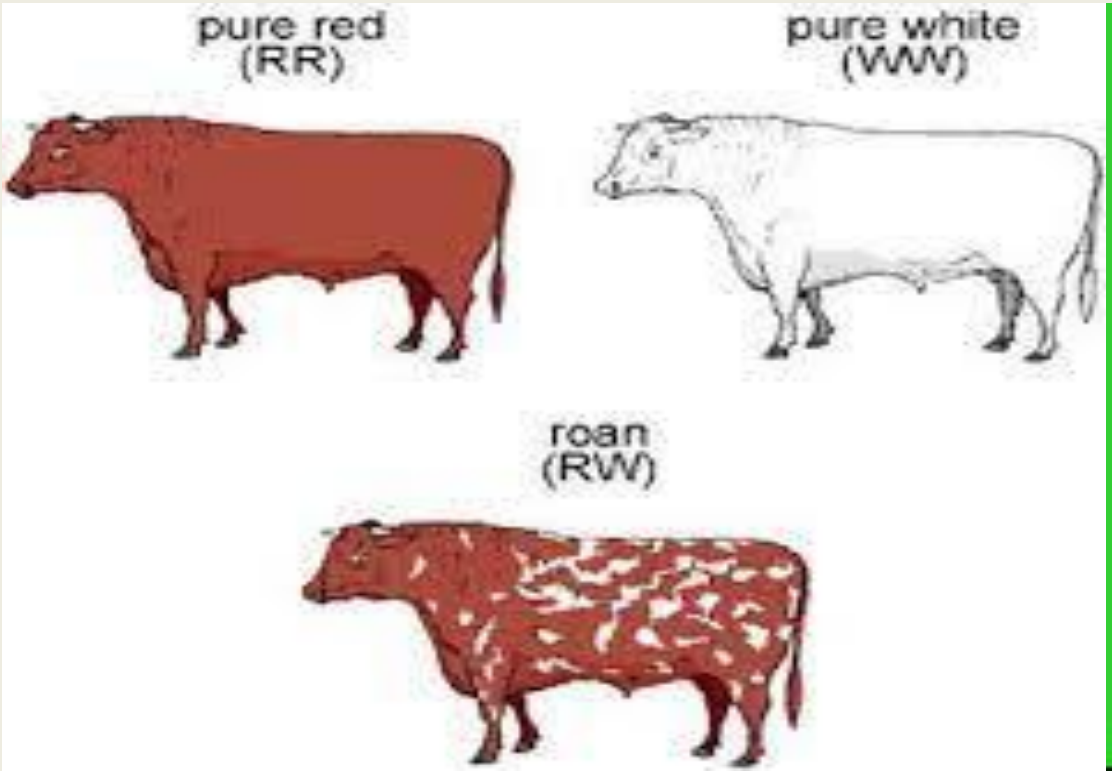
- is a form of genetic inheritance that is not in accordance with Mendel's law and don't follow one or more laws of the Mendelian genetics.
- **Examples of non-Mendelian inheritance includes;**
  - co-dominance
  - incomplete dominance
  - multiple allelism
  - gene linkage

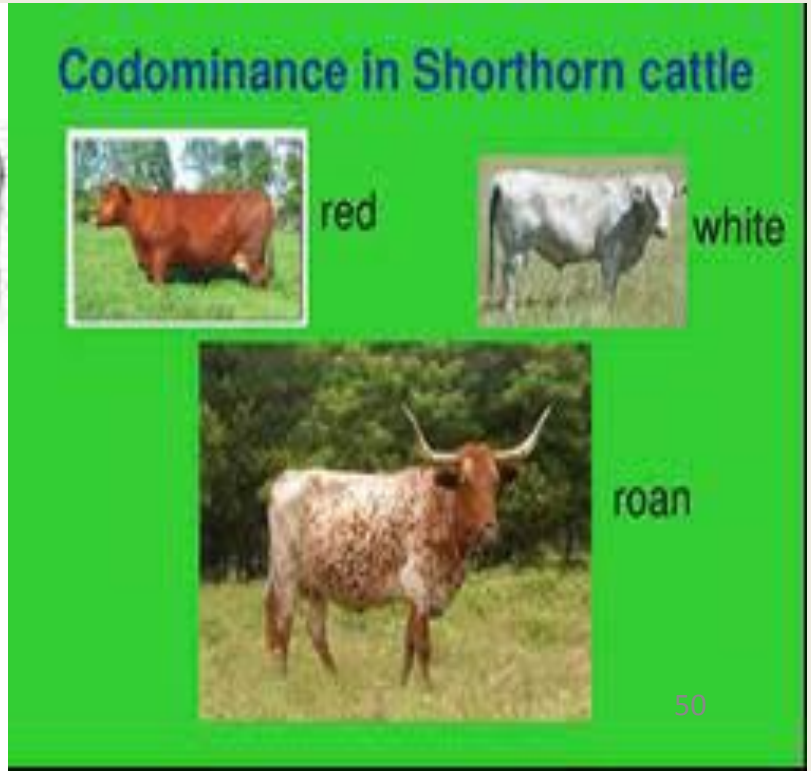


4.7.1 Co-dominance, Incomplete dominance and Multiple alleles

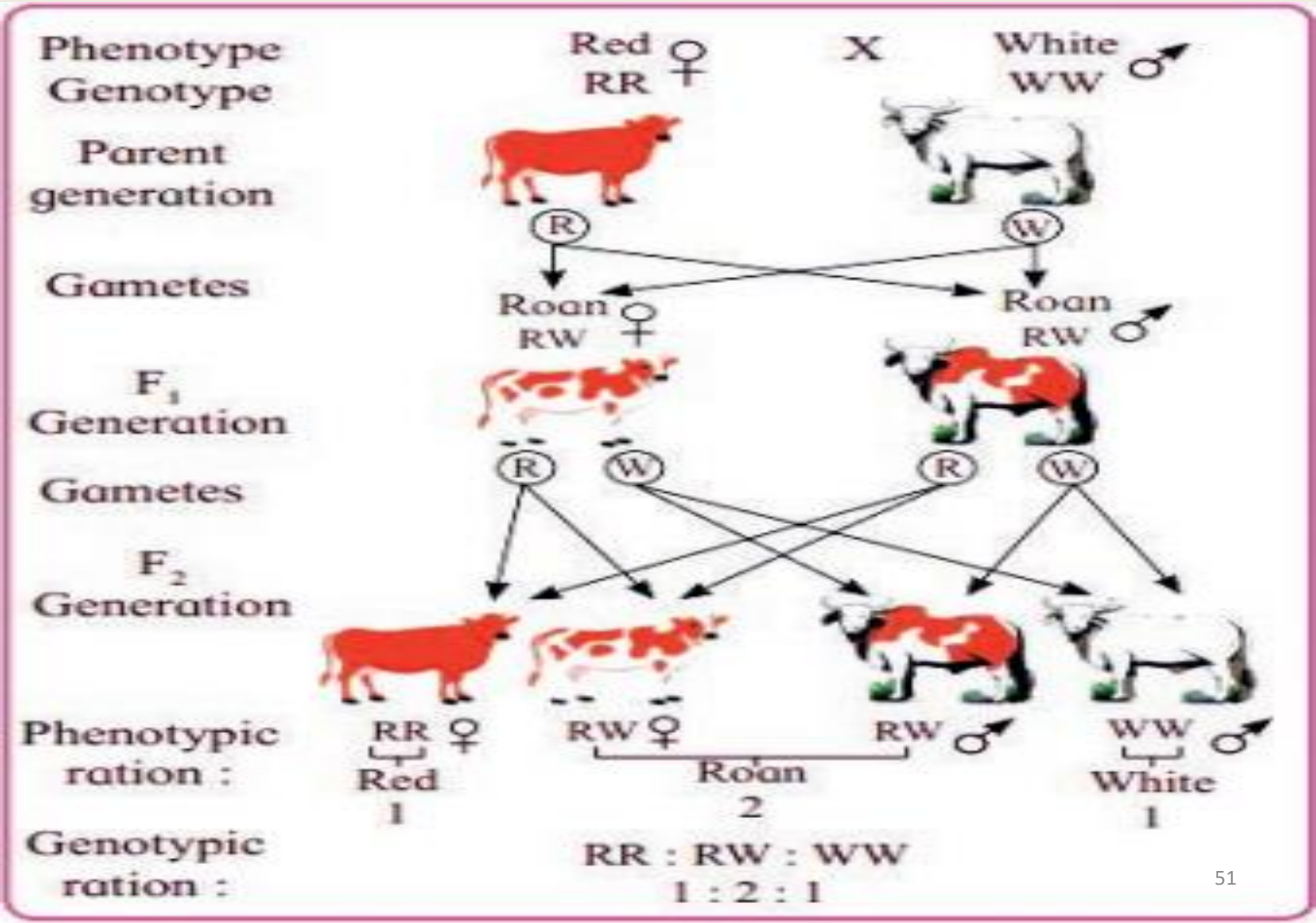
a. Co-dominance

- is a condition in which both alleles are expressed equally rather than a dominant allele taking complete control over a recessive allele.
- It means that when an organism has two different alleles (heterozygote), it will express both alleles at the same time.
- **Example:** In short-horned cattle, a cross between **red** and **white** cattle that produce ratio **1 red: 2 roans: 1 white** coat color because of co-dominance. Heterozygous individuals (RW) show roan coat color as both the alleles express themselves equally.





Co-dominance in the cross between red and white short-horned cattle

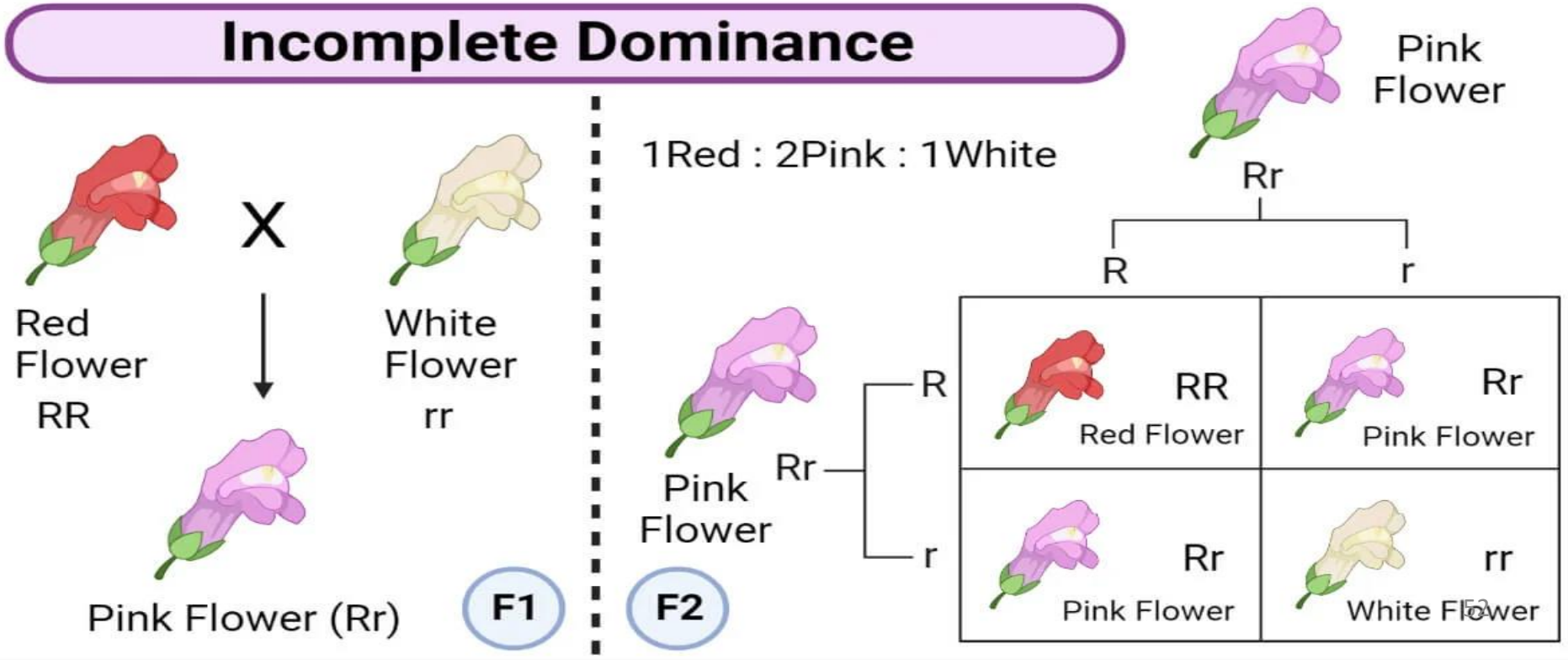


b. Incomplete dominance (partial dominance)

- **Incomplete dominance** is a form of genetic inheritance in which **both alleles of a gene are partially expressed**, often resulting in **an intermediate or different phenotype.**
- When the genotype is heterozygous, then the trait will be a blend of the two alleles.
- **Example:** the **white (RR)** and **red (rr)** varieties of rose are hybridized resulting **pink (Rr)** rose offspring.

P generation: **Phenotype** = white rose X red rose

Genotype = RR X rr



Incomplete Dominance

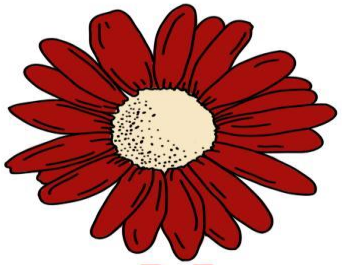
Neither allele is dominant

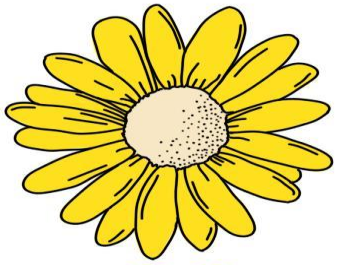
When an organism is heterozygous for a trait, it will show a third phenotype

The third phenotype is a blend of the other two

In this example, the letter A represents the gene

R and Y represent the different alleles





Black

B1B1

B2B2

White

B1B2

Spackeld

c. Multiple alleles (Multiple allele inheritance)

- **Multiple allele inheritance** means the inheritance of a trait controlled by a **single gene** containing **more than two alleles**.
- In multiple allele inheritance, an individual can only have two alleles of the gene. Each biological parent transmits one of their two alleles to their child.
- **An example** of a trait with multiple alleles is **ABO blood group system** in humans, because it exists in three allelic forms: **A**, **B**, and **O**.
- In the ABO blood grouping system, there are four blood groups, determined by the presence or absence of the **antigens A and B** on the surface of the red blood cells.
- **There are three alleles involved in the inheritance of the four blood groups:**
  - A, which determines the production of the A antigen
  - B, which determines the production of the B antigen
  - O, which determines that neither antigen is produced
- Alleles **A** and **B** are **co-dominant,** but **O** is recessive to both.

Table 4.4 The genotype and phenotype of ABO blood groups

ABO Blood Group	
Genotype	Phenotype (Blood type)
AA	A
AO	A
BB	B
BO	B
OO	O
AB	AB

Table 4.5 Antigens and antibodies in ABO blood types

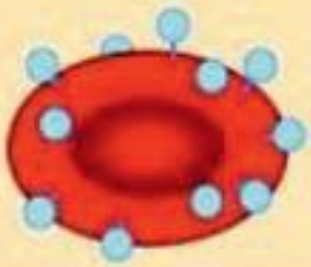
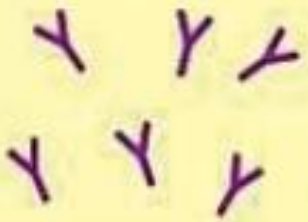
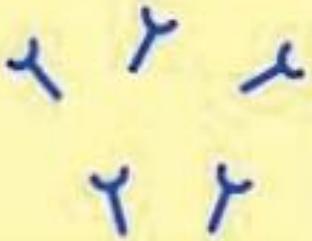
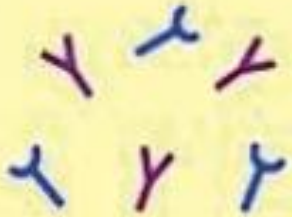
ABO Blood Groups				
Antigen (on RBC)	Antigen A 	Antigen B	Antigens A + B	Neither A or B
Antibody (in plasma)	Anti-B Antibody 	Anti-A Antibody 	Neither Antibody	Both Antibodies 
Blood Type	**Type A** Cannot have B or AB blood Can have A or O blood	**Type B** Cannot have A or AB blood Can have B or O blood	**Type AB** Can have any type of blood Is the universal recipient	**Type O** Can only have O blood Is the universal donor

Table 4.3. Possible ABO blood types of children from parents of various blood groups

No	Parents		Children	
	Phenotype	Genotype	Phenotype	Genotype
1	O x O	ii x ii	O	Ii
2	O x A	ii x $I^A I^A$ or $I^A i^A$	O, A	ii, $I^A i$
3	O x B	ii x $I^B I^B$ or $I^B i$	O,B	ii, $I^B i$
4	O x AB	ii x $I^A I^B$	A, B	$I^A i$, $I^B i$
5	A x A	$I^A I^A$ or $I^A i$ x $I^A I^A$ or $I^A i$	A,O	$I^A I^A$, $I^A I^i$, ii
6	A x B	$I^A I^A$ or $I^A i$ x $I^B I^B$ or $I^B i$	A, AB, O, B	$I^A i$, $I^A I^B$, ii, $I^B i$
7	A x AB	$I^A I^A$ or $I^A i$ x $I^A I^B$	A, AB, B	$I^A I^A$, $I^A I^i$, $I^B i$, $I^A I^B$
8	B x B	$I^B I^B$ or $I^B i$ x $I^B I^B$ or $I^B i$	B, O	$I^B I^B$, $I^B I^i$, ii
9	B x AB	$I^B I^B$ or $I^B i$ x $I^A I^B$	A, B, AB	$I^A i$, $I^B I^B$, $I^B i$, $I^A I^B$
10	AB x AB	$I^A I^B$ X $I^A I^B$	A, B, AB	$I^A I^A$, $I^B I^B$, $I^A I^B$

Exercise 7

I. Perform the following genetic crosses.

1. If, due to incomplete dominance, the F1 plants from a cross of red flowered X white flowered in the F2 generation?
2. If the father is blood group O and the mother is blood group AB, then show the blood group of their children with phenotypic and genotypic ratio.
3. Suppose a lost found baby is claimed by the four families whose blood genotypes are shown. If the baby has blood type O, what would be the possible of genotypes of the parent?

4.7.2 Rh factor inheritance in humans and its medical importance

- Rh factor (D antigen) is a substance appearing on the surface of erythrocytes (RBCs) in 85% of people.
- The Rh factor is inherited independently of the ABO blood types.
- There are **two different alleles** for the Rh factor known as **Rh+** with **dominant Rhesus D antigen** and **Rh-** , without antigen.
- People who are **Rh+/Rh+** or **Rh+/Rh-** genotype possess the Rh (D) antigen and are called **Rh positive**.
- People who are Rh-/Rh- do not possess the antigen are called **Rh negative**.

Table 4.6 Rh blood group type

Genotype	Phenotype	Blood Proteins(Antigen)	Blood transfusion	
			Donates to	Receives from
Rh+/ Rh +	Rh+	Rh(D) proteins	Rh+	Rh+, Rh-
Rh +/ Rh -	Rh+	Rh(D) proteins	Rh+	Rh+, Rh-
Rh-/ Rh-	Rh-	None	Rh-	Rh-

- A parent who is Rh negative can only pass an Rh- allele to her/his son or daughter.
- A parent who is Rh positive can pass either an Rh+ or Rh- allele to his/her son or daughter.
- A man who is heterozygous with the Rh+ blood group and a woman with the Rh- blood group may have a child with Rh+ or Rh- blood groups.
- If the Rh factor is incompatible between the mother and her unborn child, i.e. when the Rh- mother's child, inherits the Rh+ from the father, it results in a **serological conflict** (the mother's antibodies attack the baby).
- However, it is possible to prevent Rh sensitization complications by taking **anti-D vaccine.**

Table 3: Illustration of Rhesus Compatibility Match

Father	Mother	Compatibility
Rh – Positive	Rh – Positive	Compatible
Rh- Negative	Rh- Negative	Compatible
Rh – Positive	***Rh – Negative***	***Incompatible***
Rh – Negative	Rh- Positive	Compatible

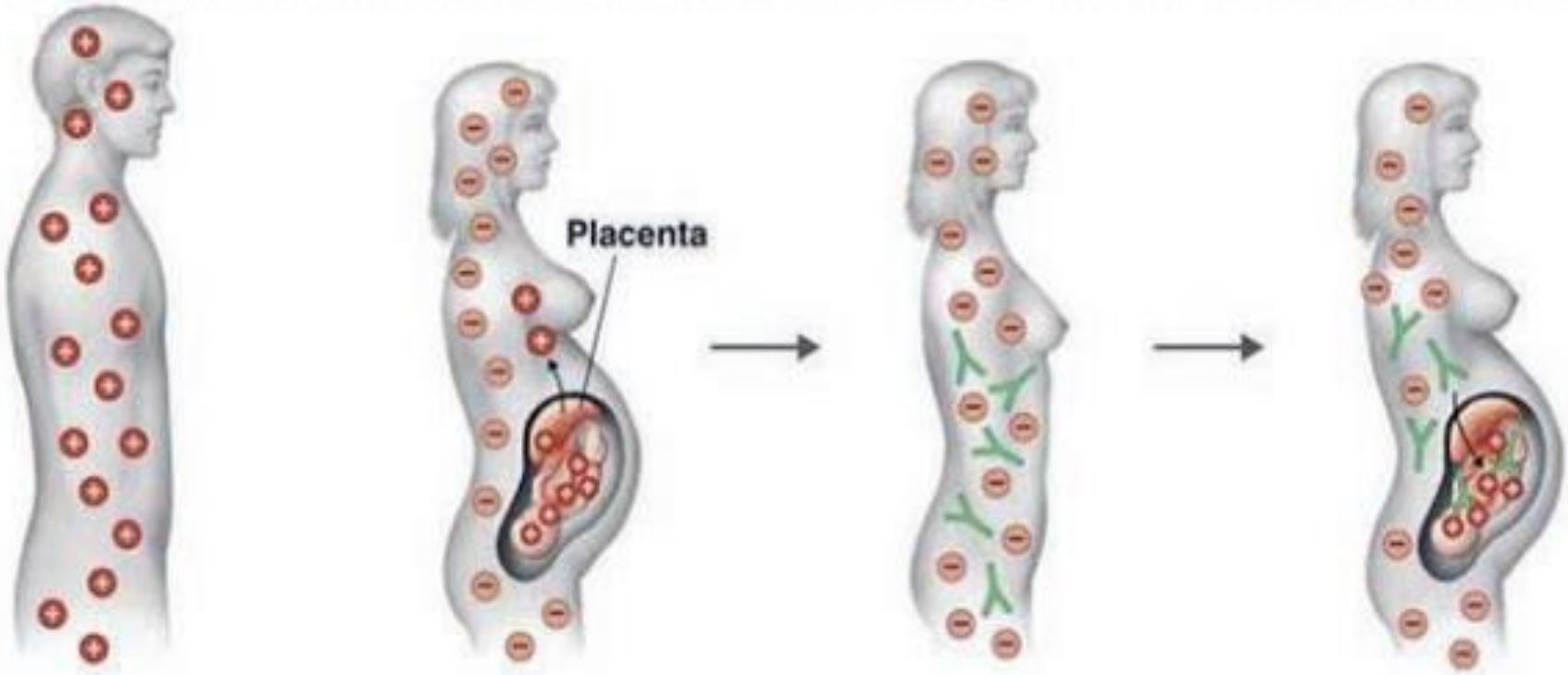


Figure 4.32 Rh Sensitization during pregnancy

Table 4.4 possible Rh blood type of children from parents of various blood groups

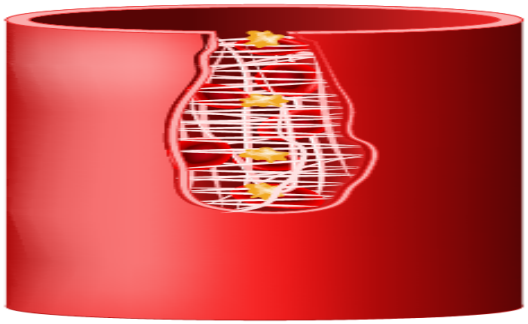
	Parents		Children	
1	Phenotype	Genotype	Phenotype	Genotype
2	$Rh^+ \times Rh^+$	+/+	Rh^+	+/+
3	$Rh^+ \times Rh^-$	+/-	Rh^+, Rh^-	+/-
4	$Rh^- \times Rh^-$	-/-	Rh^-	-/-

4.7.3 Sex-linked inheritance in humans

- **Sex-linked inheritance** refers to the inheritance of traits that are influenced by genes carried on the **sex chromosomes** (X or Y).
- **Hemophilia** and **red-green color blindness** are common examples of a sex-linked (X-linked) genetic diseases in humans.
- The genes that control hemophilia and color blindness are carried on the **X -chromosome.**
- Both hemophilia and color blindness are determined by **recessive alleles.**
- A man needs to inherit **only one X chromosome** with a recessive hemophilic or color blindness allele to suffer from the condition, because males have only one X chromosome.
- A woman needs to carry **both the recessive alleles** for hemophilia and color blindness to suffer from the condition, because females have two X chromosomes.

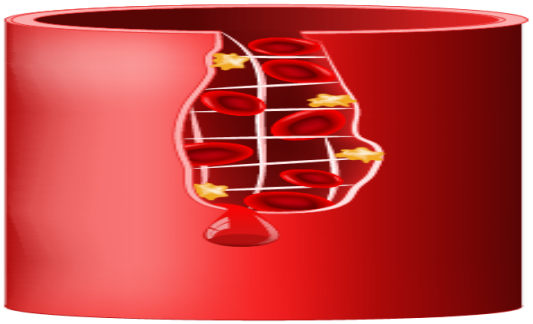
Hemophilia

Healthy person



Blood clots stop the bleeding after an injury

Hemophilia patient



The body lacks the required factor to produce blood clots, thereby causing excessive bleeding

- **Characteristics of sex-linked traits determined by recessive alleles on the X chromosome includes;**
    - they are much more common among males (males are hemizygous)
    - affected males inherit the allele from their mother only
    - affected females inherit one allele from each parent
    - females who are heterozygous for the condition are called carriers
    - they may 'skip' a generation and then appear in the males only
- **Genotypes of sex-linked traits** include the appropriate **sex chromosomes** as well as the **alleles**.
- For example, for hemophilia, H represents the allele for normal clotting and h represents the allele for hemophilic.
- **The possible genotypes and phenotypes of hemophilia in male and female are;**

Genotype	Phenotype
$X^H X^H$	Normal female
$X^H X^h$	Normal female (carrier)
$X^h X^h$	Hemophiliac female (homozygous)
$X^H Y$	Normal male
$X^h Y$	Hemophiliac male

Example: 1. If a father is normal clotting and a mother is normal but carrier.

Parents	Male (XY)	
	Gametes X	Y
Female (X^HX^h)	X^H	X^HX^H	X^HY
	X^h	X^HX^h	X^hY

- Daughters – 100% normal (50% carrier)
- Sons – 50% normal and 50% hemophiliac

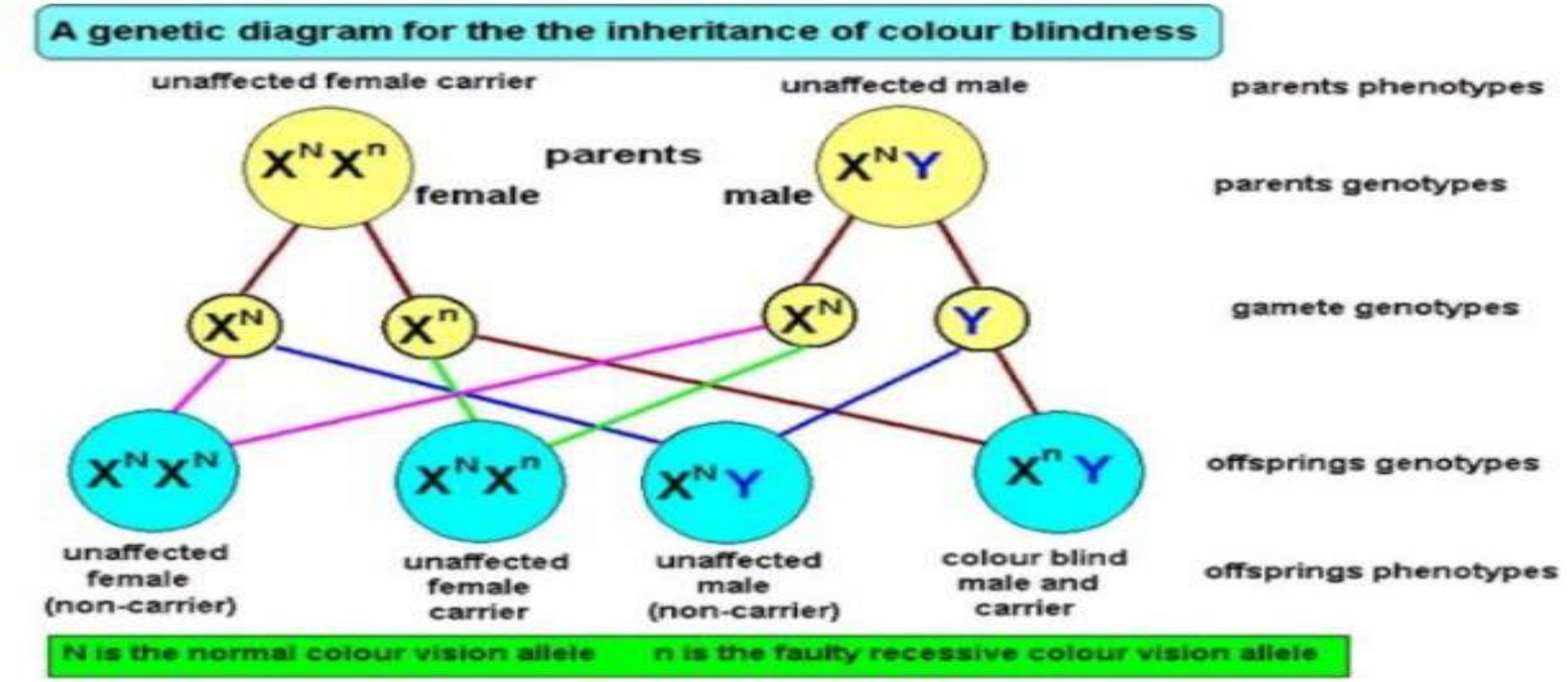


Figure 4.4. Inheritance of color blindness

Exercise 8

1. A normal visioned man whose father was color-blind, marries a woman whose father was also color-blind.
    a. What are the chances that their sons to be color-blind?
    b. What are the chances that their daughters to be color -blind?
2. A woman with two genes for hemophilia and one gene for color blindness on one of the 'X' chromosomes marries a normal man.
    a. What is the chance of their son to be hemophilic?
    b. What is the chance of their daughter to be hemophilic?

4.7.4 Environmental effects on phenotype

- The environment can affect the phenotype of organisms that live in different environmental conditions.
- **A phenocopy** is an environmentally induced phenotype of an individual, which is identical to the phenotype of another individual determined by genotype.
- The phenocopy induced by the environmental conditions mimics the phenotype produced by a gene.
- Most of the time phenocopies can result from exposure to radiation, chemicals poisons, temperature shocks etc.
- An example of phenocopy due to temperature variation is observed in Himalayan rabbits.

Himalayan rabbits have a white colored coat along with a black tail, nose, and ears when raised in moderate temperatures. However, they also show black coloration of their coats when raised in cold temperatures, resembling the genetically black rabbits (Figure 4.33).

Figure 4.33 White and Black(phenocopy) Himalayan rabbit

“Believe you can and you are hallway there!!”