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General Biology

Lecture (3): Genetics

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Biology Department

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1

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1. Introduction into Genetics

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Overview: Variations on a Theme

- Living organisms are distinguished by their ability to reproduce their own kind.
- **Genetics** is the scientific study of heredity and variation.
- **Heredity** is the transmission of traits from one generation to the next.
- **Variation** is demonstrated by the differences in appearance that offspring show from parents and siblings.

3

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2.The Molecular Basis of Inheritance

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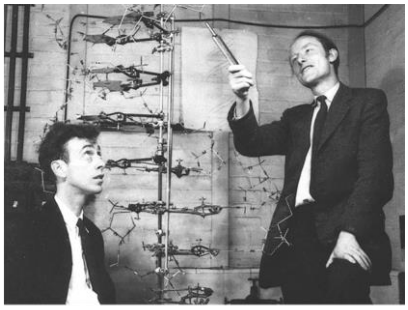
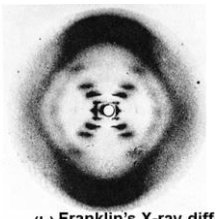
Overview: Life's Operating Instructions

- In 1953, James Watson and Francis Crick shook the world With an elegant double-helical model for the structure of deoxyribonucleic acid, or DNA.
- Watson and Crick deduced that DNA was a double helix, through observations of the X-ray crystallographic images of DNA
- The role of DNA in heredity worked out by studying bacteria and the viruses that infect them.
- DNA, the substance of inheritance and the most celebrated molecule of our time.



(a) Rosalind Franklin

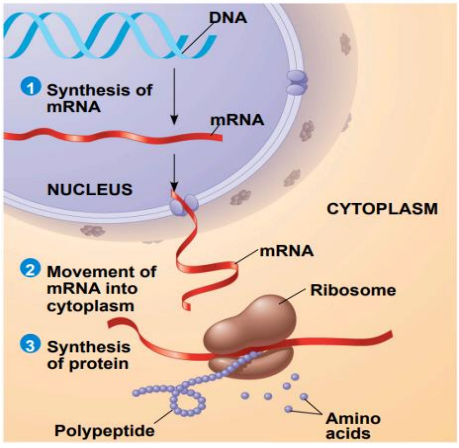


(b) Franklin's X-ray diff Photograph of DNA

5

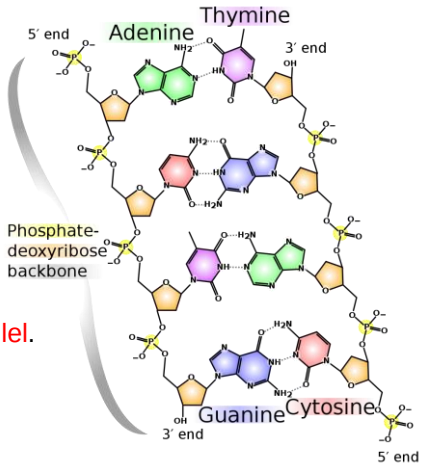
Nucleic acids store, transmit, and express hereditary information

- The amino acid sequence of a polypeptide is programmed by a unit of inheritance called a gene.
- Genes are made of DNA, a nucleic acid made of monomers called nucleotides .
- There are two types of nucleic acids:
  - Deoxyribonucleic acid (DNA)
  - Ribonucleic acid (RNA)
- DNA provides directions for its own replication.
- DNA directs synthesis of messenger RNA (mRNA) and, through mRNA, controls protein synthesis.
- Protein synthesis occurs on ribosomes.

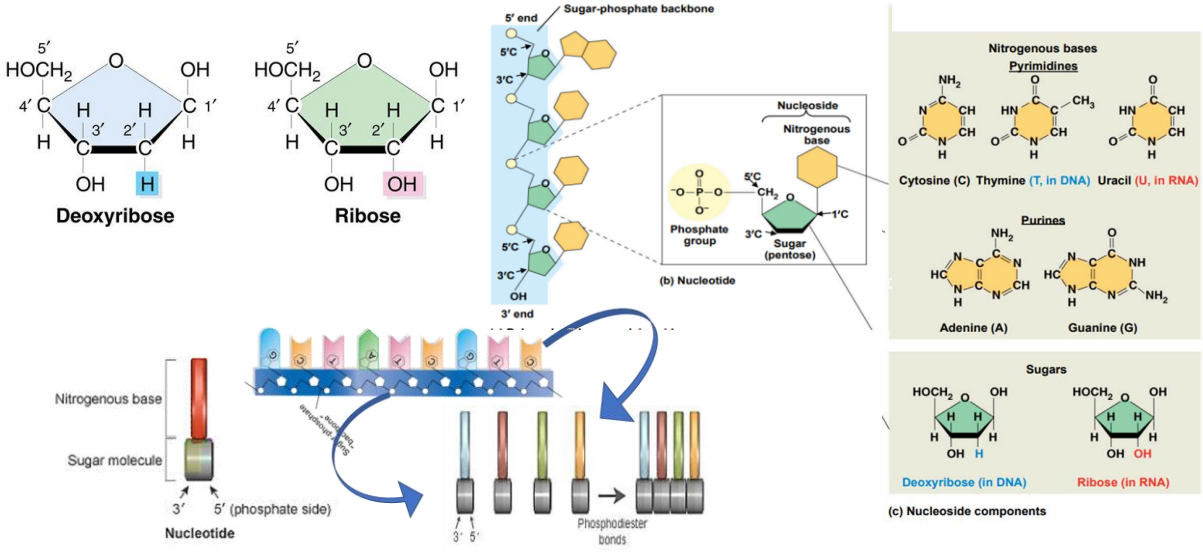


The Components of Nucleic Acids

- Nucleic acids are polymers called polynucleotides.
- Each polynucleotide is made of monomers called nucleotides.
- Each nucleotide consists of a nitrogenous base, a pentose sugar, and one or more phosphate groups.
- There are two families of nitrogenous bases
  - Pyrimidines (cytosine, thymine, and uracil)
  - Purines (adenine and guanine).
- In the DNA double helix, the two backbones run in opposite 5′→ 3′ directions from each other, an arrangement referred to as antiparallel.
- The nitrogenous bases in DNA pair up and form hydrogen bonds: adenine (A) always with thymine (T), and guanine (G) always with cytosine (C). Called complementary base pairing.



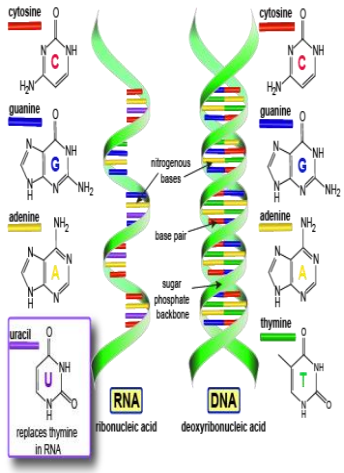
The Structures of DNA and RNA Molecules



The Structures of DNA and RNA Molecules

- RNA is slightly different from DNA in the following aspects

	DNA (Deoxyribonucleic acid)	RNA (Ribonucleic acid)
Location	Nucleus and mitochondria, chloroplast.	Nucleus and cytoplasm,
Structure	Double strand helix.	Single-strand helix.
Sugar	It contains Deoxyribose.	It contains Ribose.
Nitrogenous Bases	AT (adenine-thymine). GC (guanine-cytosine).	AU (adenine-uracil). GC (guanine-cytosine).
Length	Long with high molecular mass.	Short with low molecular mass.
Propagation	DNA is self-replicating.	RNA is synthesized from DNA.
Function	storage and transmission of genetic information.	Transfers genetic code for protein synthesis.
Type	_____	(tRNA), (rRNA), (mRNA),(snRNA)



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3. The Chromosomal Basis of Inheritance

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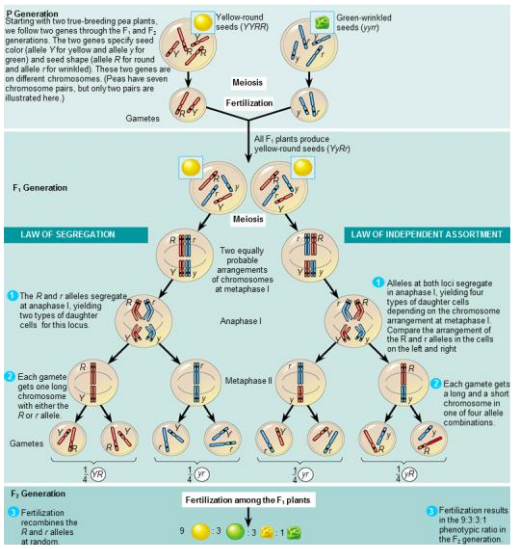
Chromosome Theory

- The chromosome theory of inheritance states that:
  - Mendelian genes have specific loci on chromosomes.
  - Chromosomes undergo segregation and independent assortment.
  - Thomas Hunt Morgan.
  - Provided convincing evidence that chromosomes are the location of Mendel's heritable factors.

The chromosomal basis of Mendel's laws



11

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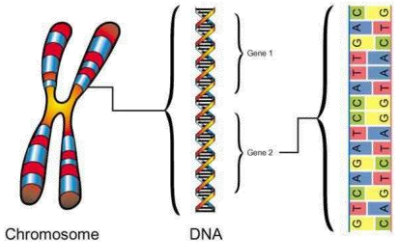
Inheritance of Genes

- In a literal sense, children do not inherit particular physical traits from their parents.
- It is genes that are actually inherited.
- **Genes** are the units of heredity, and are made up of segments of DNA.
- Genes are passed to the next generation through reproductive cells called **gametes** (sperm and eggs).
- Each gene has a specific location called a **locus** on a certain chromosome.
- Most DNA is packaged into chromosomes.
- One set of chromosomes is inherited from each parent.

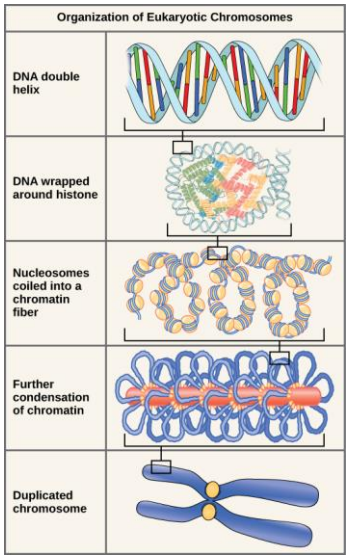


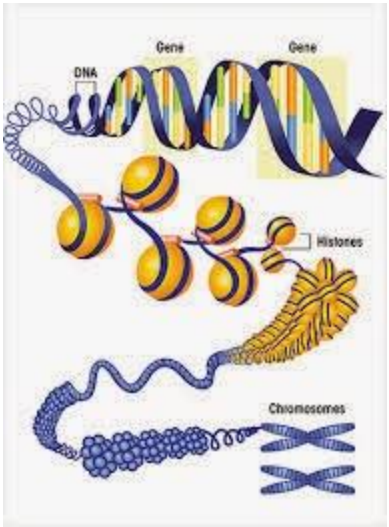
Genes

Offspring acquire genes from parents by inheriting chromosomes

12

DNA Packaging and Organization into Chromosomes





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4. The Cell Cycle.

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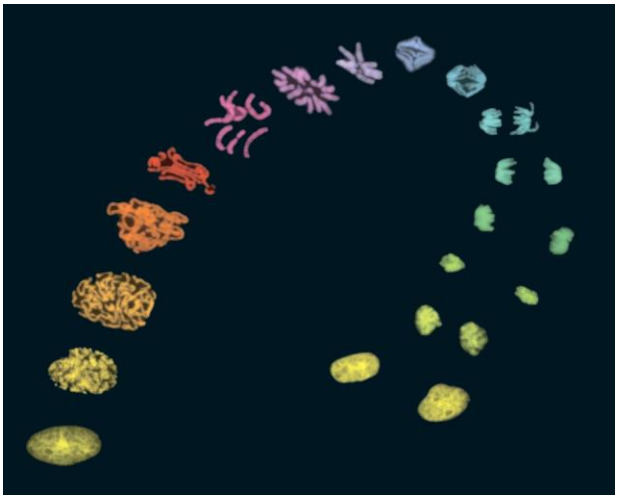
The Cell Cycle

- The ability of organisms to reproduce best distinguishes living things from non living matter.
- The continuity of life is based on the reproduction of cells, or cell division.
- The **frequency** of cell division varies with the type of cell.
- A **life cycle** is the generation-to-generation sequence of stages in the reproductive history of an organism.



15

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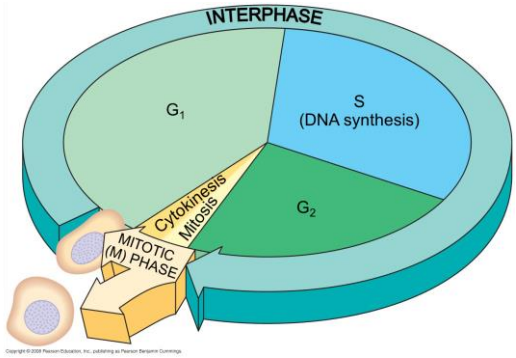
Phases of the Cell Cycle

- **The cell cycle consists of :**
  - Interphase (cell growth and copying of chromosomes in preparation for cell division).
  - Mitotic (M) phase (mitosis and cytokinesis).
  - Interphase (about 90% of the cell cycle) can be divided into subphases:
  - G1 phase ("first gap")
  - S phase ("synthesis")
  - G2 phase ("second gap")
  - The cell grows during all three phases, but chromosomes are duplicated only during the S phase.



16

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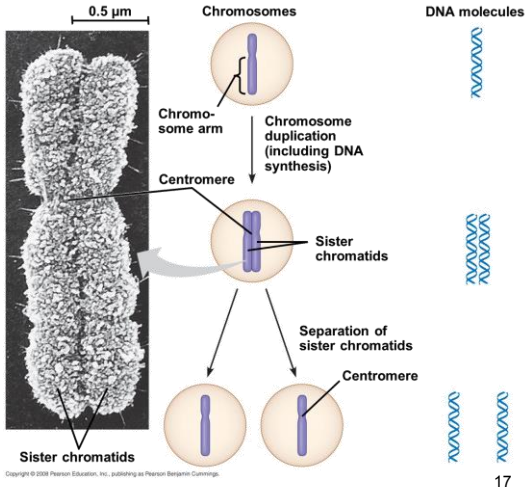
Interphase Stage

- 1st growth stage of cell division.
- Cell carries on its normal metabolic activities.
- Cells mature by making more cytoplasm & organelles, proteins and RNA .
- DNA Synthesis stage Occurs prior to division.
- DNA is copied or replicated. Because of duplication, each condensed chromosome consists of 2 identical chromatids joined by a centromere, called sister chromatids.
- Each duplicated chromosome contains 2 identical DNA molecules.



17

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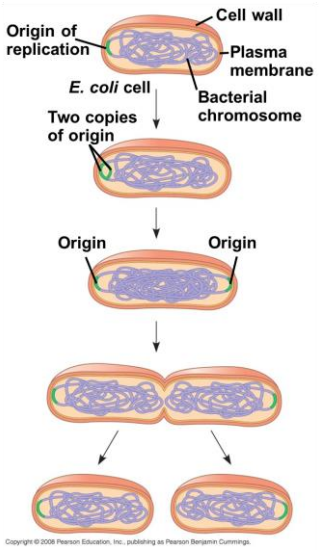
Cell division

- In **unicellular** organisms like in bacteria, division of one cell reproduces the entire organism called by Binary fission.
- **Eukaryotic** cell division consists of:
- Mitosis, the division of the nucleus.
- Cytokinesis, the division of the cytoplasm.
- Most cell division results in daughter cells with identical genetic information, DNA.
- A special type of division produces nonidentical daughter cells (gametes, or sperm and egg cells).



18

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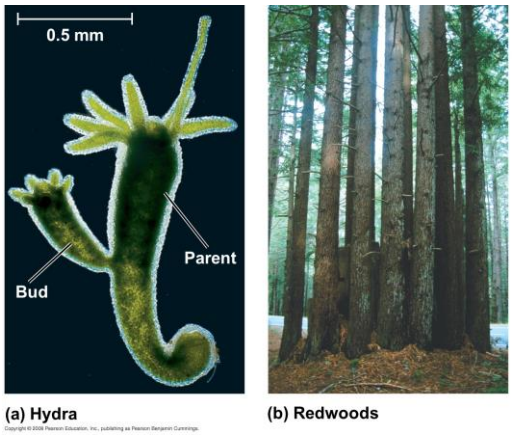
Comparison of Asexual and Sexual Reproduction

- In **asexual reproduction**, one parent produces genetically identical offspring by mitosis.
- A **clone** is a group of genetically identical individuals from the same parent.
- In **sexual reproduction**, two parents give rise to offspring that have unique combinations of genes inherited from the two parents.



19

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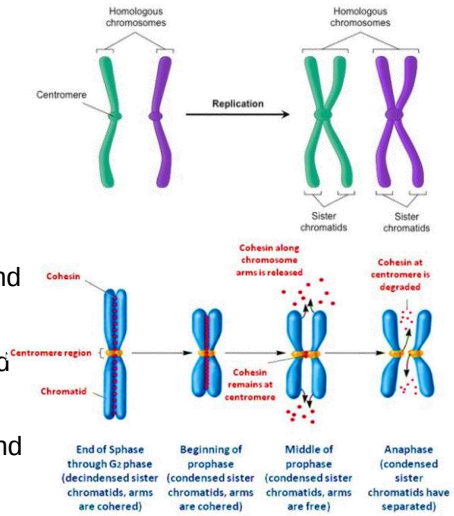
Mitosis

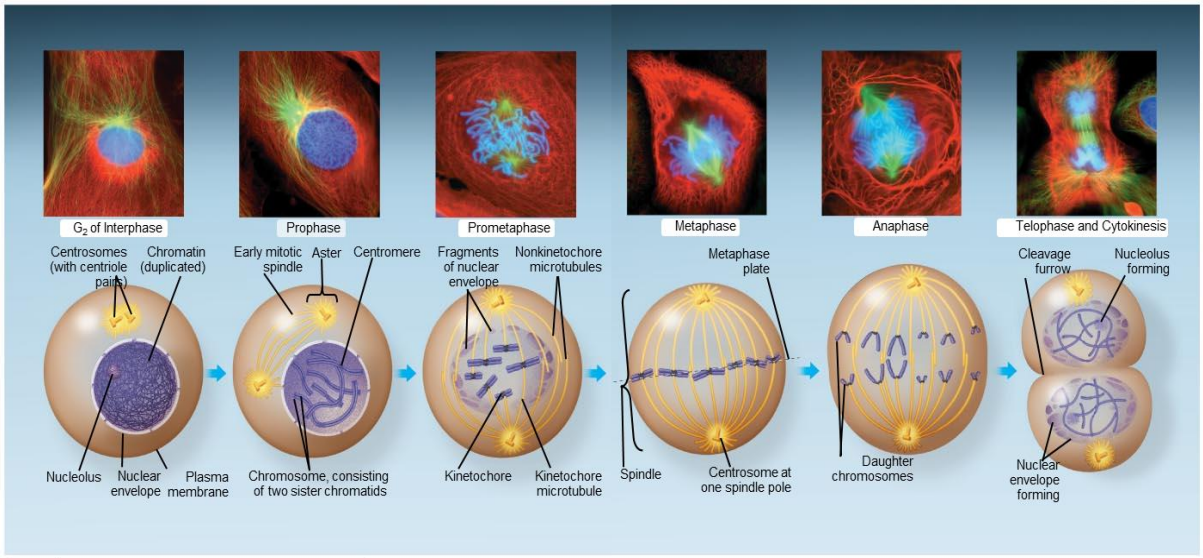
➢ Mitosis is divided into five phases:

- **Prophase**: The chromatin fibers become more tightly coiled, condensing into discrete chromosomes .
- **Prometaphase**: The two pairs of centrioles move to opposite poles, and mitotic spindle begins to capture and organize the chromosomes.
- **Metaphase:** the spindle has captured all the chromosomes and lined them up at the middle of the cell, ready to divide.
- **Anaphase:** the sister chromatids separate from each other and are pulled towards opposite ends of the cell.
- **Telophase:** Chromosomes cluster at opposite spindle poles and their identity is lost as discrete elements.
- **Cytokinesis:** the division of the cytoplasm to form two new cells.



20

Mitosis



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5. Meiosis and Sexual Life Cycles

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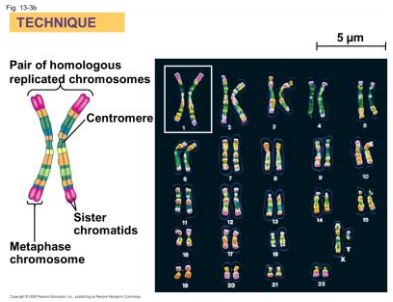
Meiosis and Sexual life Cycle

- Human **somatic cells** (any cell other than a gamete) have 23 pairs of chromosomes. one chromosome from each parent.
- Each set of 23 consists of 22 autosomes and a single sex chromosome.
- In an egg (ovum), the sex chromosome is XX ,while In a sperm cell, the sex chromosome may be either X or Y.
- The two chromosomes in each pair are called **homologous chromosomes**, are same in length.
- A **diploid cell** (2*n*) has two sets of chromosomes, For humans, the diploid number is 46 (2*n* = 46).
- A **haploid cell** (n) contains a single set of chromosomes, A gamete (sperm or egg) For humans, the haploid number is 23 (n = 23).



23

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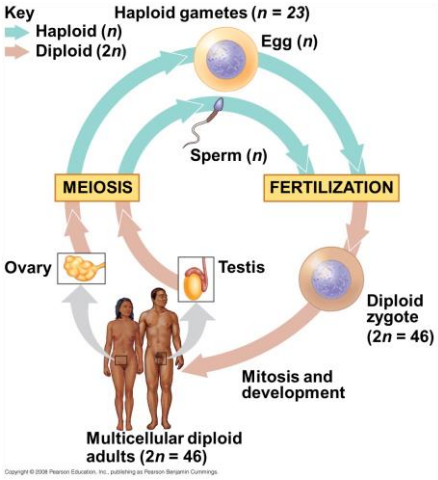
Behavior of Chromosome Sets in the Human Life Cycle

- **Fertilization** is the union of gametes (the sperm and the egg)
- The **fertilized egg** is called a **zygote** and has one set of chromosomes from each parent.
- The **zygote** produces somatic cells by mitosis and develops into an adult.
- At sexual maturity, the ovaries and testes produce haploid gametes.
- Gametes are the only types of human cells produced by **meiosis**, rather than **mitosis**.
- Meiosis results in one set of chromosomes in each gamete.
- **Fertilization and meiosis** alternate in sexual life cycles to maintain chromosome number.



24

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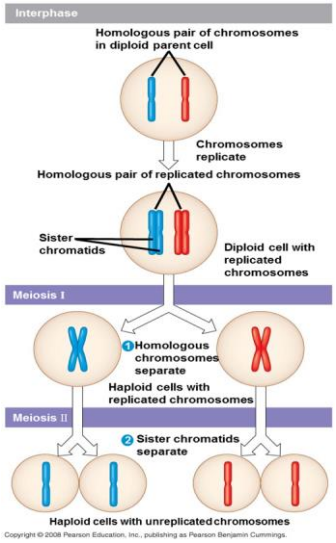
Meiosis reduces the number of chromosome sets

- Like mitosis, meiosis is preceded by the replication of chromosomes.
- Meiosis takes place in two sets of cell divisions, called **meiosis I** and **meiosis II.**
- The two cell divisions result in four daughter cells, rather than the two daughter cells in mitosis.
- Each daughter cell has only half as many chromosomes as the parent cell.



25

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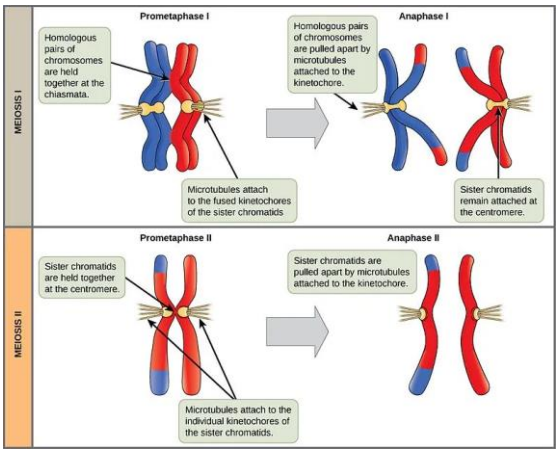
Meiosis reduces the number of chromosome sets

- In the first cell division (meiosis I), homologous chromosomes separate.
- Meiosis I results in two haploid daughter cells with replicated chromosomes; it is called the reductional division.
- In the second cell division (meiosis II), sister chromatids separate.
- Meiosis II results in four haploid daughter cells with unreplicated chromosomes; it is called the equational division.



26

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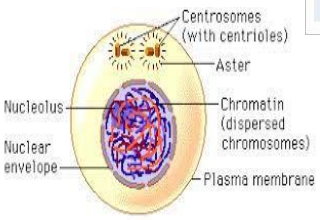
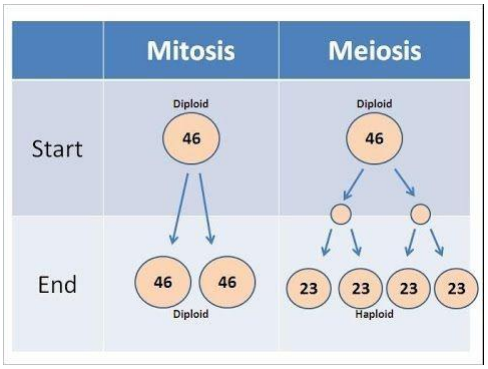
Meiosis I

- **Meiosis I** is preceded by interphase, in which chromosomes are replicated to form sister chromatids.
- The sister chromatids are genetically identical and joined at the centromere.
- The single centrosome replicates, forming two centrosomes.





27

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Meiosis I vs Meiosis II

Comparison	Meiosis I	Meiosis II
	Results two Daughter cells.	Results Four Daughter cells.
	Consist four stages Prophase I , Metaphase I, Anaphase I, Telophase I.	Consist four stages Prophase I I , Metaphase II, Anaphase II, Telophase II
	Homologous chromosome are present at the beginning.	Individual, bivalent chromosome are present at the beginning.
	Reduce the chromosome number in the daughter cell.	Equalizes the chromosome number for both parent and daughter cell.
Interphase	Interphase is followed by meiosis 1, DNA replication.	No interphase takes place prior to the meiosis II.

- Meiosis II is very similar to mitosis.

28

Meiosis I vs Meiosis II

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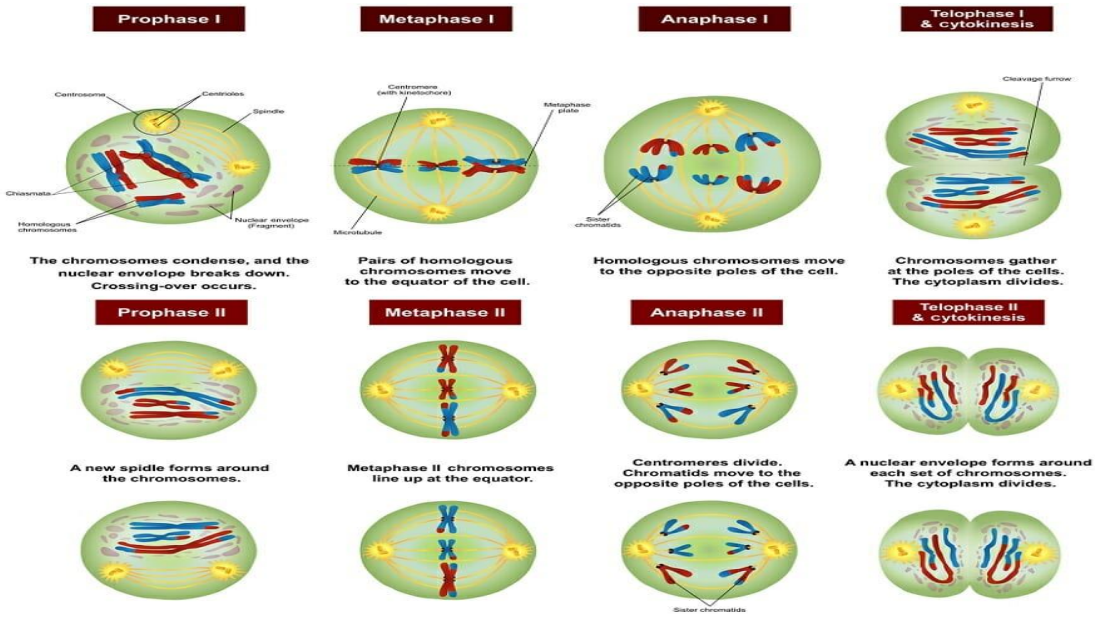
Comparison	Meiosis I	Meiosis II
Prophase	- The homologous chromosome Pairing (lines up closely together) and a **tetrad** is formed. Each tetrad is composed of **four chromatids**. - crossing over occurs by exchanging the genetic material between non-sister chromatids, leading to the genetic recombination.	- No chromosomal cross-over occurs.
Metaphase	Thethe homologous chromosomes(tetrads) arrange in an independent orientation of pairs of in the cell equator.	- The sister chromatids line up at the equator of the cell individually.
Anaphase	-The Homologous chromosomes are separated and pulled to the opposite poles.	- The sister chromatids are separated and pulled to the opposite poles.
Telophase	- The chromosomes arrive at opposite poles chromosomes begin decondensing and Nuclei form. - Haploid set of chromosomes; each chromosome still consists of two sister chromatids.	- The single sister chromatid are present in the daughter cells. - begin decondensing. Nuclei form.
Cytokinesis	- Usually occurs simultaneously, forming two haploid daughter cells.	

Meiosis II



Mitosis vs Meiosis

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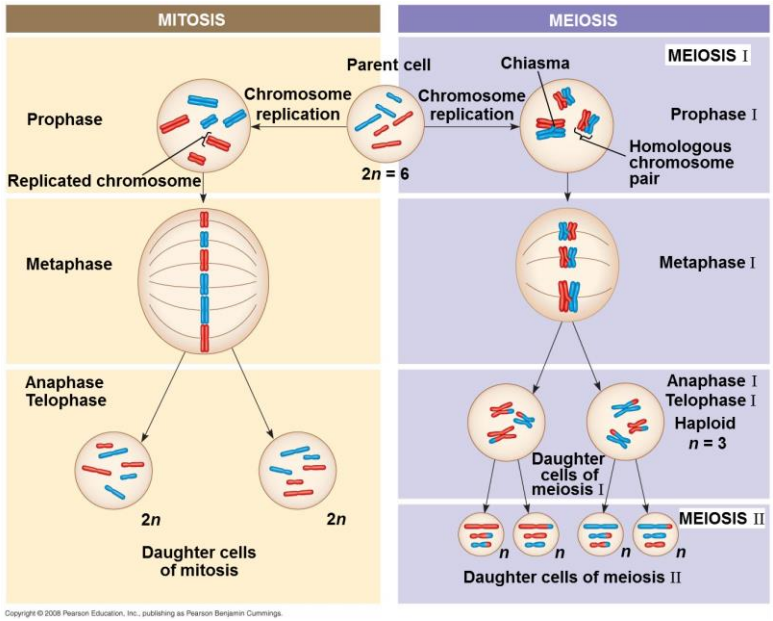

31

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Mitosis vs Meiosis

SUMMARY		
Property	**Mitosis**	**Meiosis**
DNA replication	Occurs during interphase before mitosis begins	Occurs during interphase before meiosis I begins
Number of divisions	One, including prophase, metaphase, anaphase, and telophase	Two, each including prophase, metaphase, anaphase, and telophase
Synapsis of homologous chromosomes	Does not occur	Occurs during prophase I along with crossing over between nonsister chromatids; resulting chiasmata hold pairs together due to sister chromatid cohesion
Number of daughter cells and genetic composition	Two, each diploid (2*n*) and genetically identical to the parent cell	Four, each haploid (*n*), containing half as many chromosomes as the parent cell; genetically different from the parent cell and from each other
Role in the animal body	Enables multicellular adult to arise from zygote; produces cells for growth, repair, and, in some species, asexual reproduction	Produces gametes; reduces number of chromosomes by half and introduces genetic variability among the gametes

32

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6. Mendel and the Gene Idea

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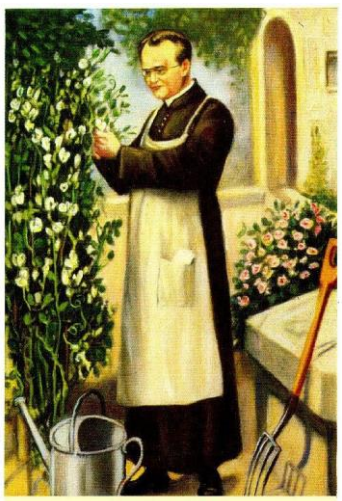
Mendel used the scientific approach to identify two laws of inheritance

- The "particulate" hypothesis is the idea that parents pass on discrete heritable units (genes)
- Mendel documented a particulate mechanism through his experiments with garden peas.
- Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments.



GREGOR MENDEL

34

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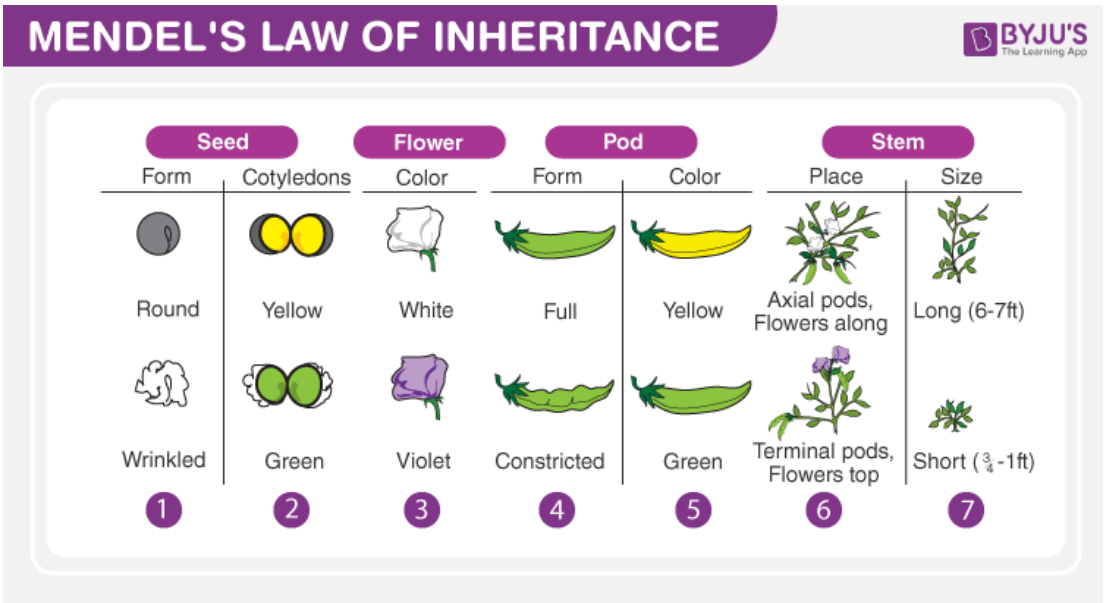
Why Mendel's choose pea plant for his Experiment ?

➢ Advantages of pea plants for genetic study:

- Mendel performed 28,000 of crosses, Seven pairs of contrasting characters were chosen for his study..
- Peas had many sharply defined inherited characters, **characters** (such as flower color); character variants (such as purple or white flowers) are called **traits.**
- Mating of plants can be controlled.
  - Each pea plant has sperm-producing organs (stamens) and egg-producing organs (carpels).
  - Cross-pollination (fertilization between different plants) can be achieved by dusting one plant with pollen from another.
- The self fertilization through many generations helps in easily obtaining the pure line with constant trait in pea plants.
- Pea plants has Short life time.

35



36

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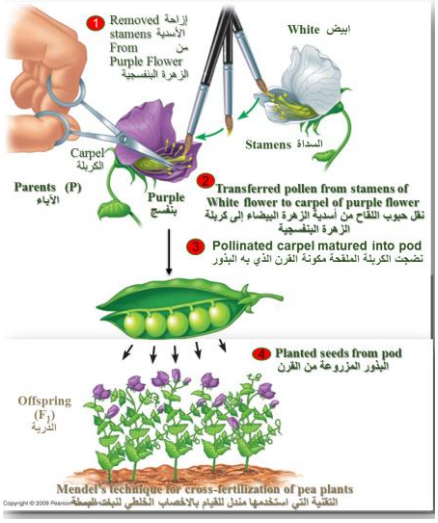
Mendel's Experimental Methods

- In a typical experiment, Mendel mated two contrasting, true-breeding varieties, a process called **hybridization.**
- The true-breeding parents are the **P generation.**
- The hybrid offspring of the P generation are called the **F_1 generation.**
- When F_1 individuals self-pollinate, the **F_2 generation** is produced.
- When Mendel crossed contrasting, true-breeding white and purple flowered pea plants, all of the F_1 hybrids were purple.



37

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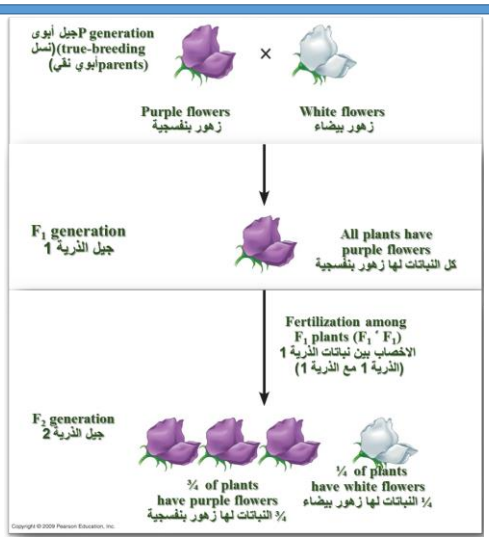
Mendel's Experimental Methods

- When Mendel crossed the F_1 hybrids, many of the F_2 plants had purple flowers, but some had white.
- Mendel discovered a ratio of about three to one, purple to white flowers, in the F_2 generation.
- Mendel developed a hypothesis to explain the 3:1 inheritance pattern he observed in F_2 offspring.
- Four related concepts make up this model.



38

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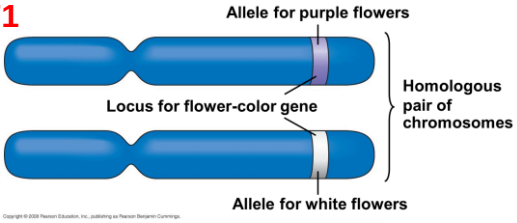
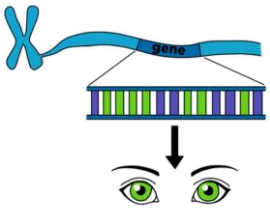
Mendel concepts: The Rule of Dominance

1- Mendel's **observed** in every case, the purple flower factor was affecting flower color in the F_1 hybrids, while the white flower seemed to **disappear** in the **F1** generation**,** only to **reappear** unchanged in **one-fourth** of the **F2** plants**.**

- Mendel called the purple flower color a dominant trait and the white flower color a recessive trait.



2- Mendel **concluded** that each organism has **two factors** that **control** each of its **traits called** "heritable factor" is what we now call a gene and that they are located on **chromosomes.**



39

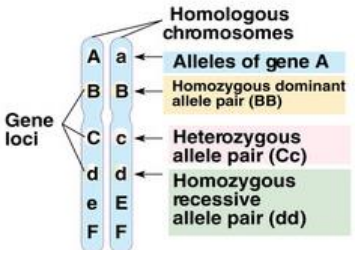
23/02/1444

Mendel concepts: The Rule of Dominance

3- Genes exist in alternative forms, Called alleles.

- An organism's has two alleles are located on different copies of a chromosome, one inherited from the female parent and one from the male parent.

- The two alleles at a locus on a chromosome may be identical as in the true-breeding plants of Mendel's P generation or differ, as in the F_1 hybrids.

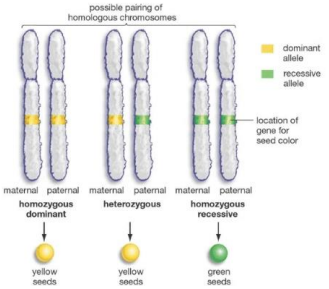


4- Mendel recording the results of crosses, he use the same letter for different alleles of the same gene.

- The An uppercase letter is used for the dominant allele. (e.g. PP).

- The lowercase letter for the recessive allele. (e.g. pp),

- The dominant allele is always written first. (e.g. Pp).



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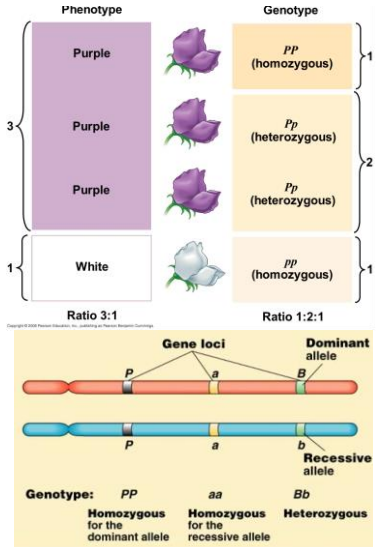
Mendel concepts: The Rule of Dominance

- The way an organism looks and behaves is called its phenotype. (e.g. purple, white).
- The allele combination an organism contains is known as its genotype. (e.g. PP, Pp, pp).
- An organism is homozygous for a trait if its two alleles for the trait are the same. (e.g. PP or pp); also called pure.
- An organism is heterozygous for a trait if its two alleles for the trait are different from each other. (e.g. Pp); also called hybrid.



41

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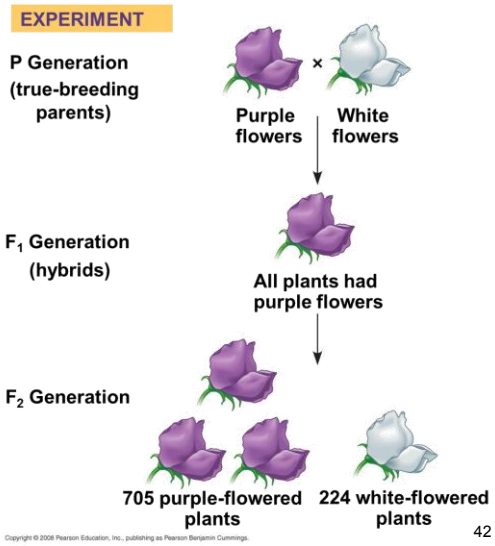
Mendel's law: Law of Dominance

- In a cross of parents that are pure for contrasting traits, only one form of the trait will appear in the next generation.
- The Offspring that are hybrid for a trait will have only the dominant trait in the phenotype.



42

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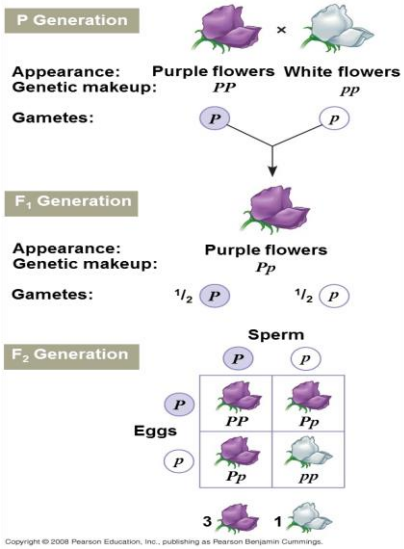
Mendel's law: Law of Segregations

- The **law of segregation**, states that the two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes.

- Thus, an egg or a sperm gets only one of the two alleles that are present in the somatic cells of an organism.

- This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes in meiosis.



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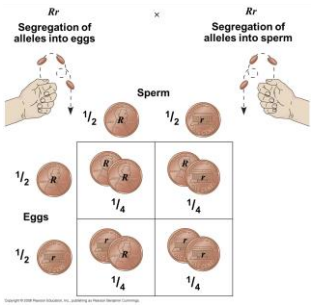
Mendel's law: Law of Independent Assortment

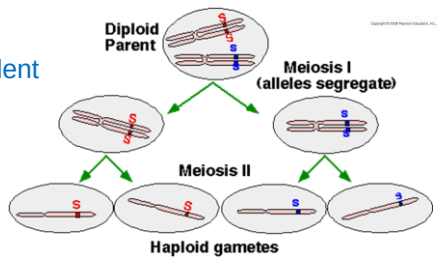
- Using a dihybrid cross, Mendel developed the law of independent assortment.

- **The law of independent assortment** states that each pair of alleles segregates independently of each other pair of alleles during gamete formation.

- this law applies only to genes on different, nonhomologous chromosomes.

- Mendel's laws of segregation and independent assortment reflect the rules of probability.





44

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7. From Gene to Protein

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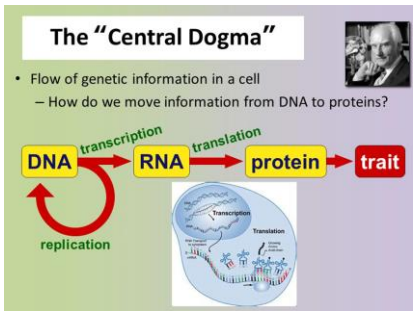
Overview: The Flow of Genetic Information

- **The information content of DNA** is in the form of specific sequences of nucleotides along the DNA strands.
- **The DNA inherited by an organism** leads to specific traits by dictating the synthesis of proteins.
- Cells are governed by a cellular chain of command
  DNA ⟶ **RNA** ⟶ **protein**
- The process by which DNA directs protein synthesis, called **gene expression** includes two stages, called **transcription and translation.**
- Protein are complex molecules, polypeptide are folded, and combined with other polypeptides to form a protein.



46

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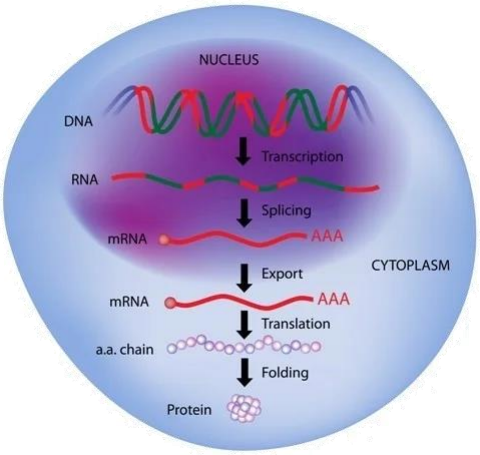
Basic Principles of Transcription and Translation

➢ Transcription:

- The first step in gene expression. It involves copying a gene's DNA sequence to make an RNA molecule.
- The main enzyme involved in transcription is **RNA polymerase**, which uses a single-stranded DNA template to synthesize a complementary strand of RNA.
- RNA polymerase builds an RNA strand in the 5' to 3' direction by adding each new nucleotide to the 3' end of the strand.



47

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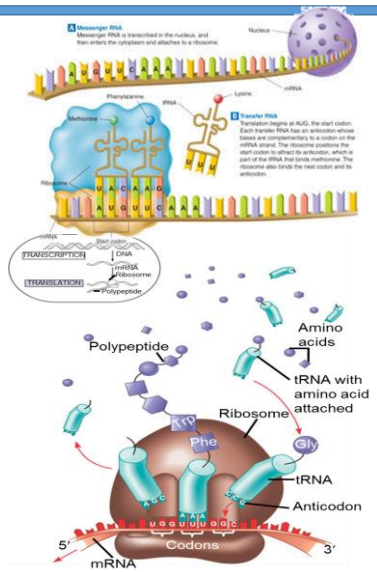
Basic Principles of Transcription and Translation

- ➢ Once the DNA has been transcribed to mRNA, the codons must be translated to the amino acid sequence of the protein.
- ➢ Protein is constructed in the cytoplasm.
- ➢ Three steps of translation:
  - Initiation: mRNA binding is binding with subunit of ribosome and aminoacyl-tRNA.
  - Elongation: Ribosome moves along mRNA, making polypeptide chain and extending protein.
  - Termination: Polypeptide chain is released from tRNA, and ribosome dissociates from mRNA.



48

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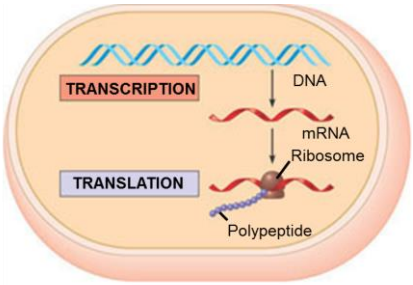
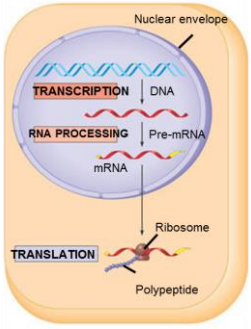
Transcription and Translation in Prokaryotes and Eukaryotes

- **In prokaryotes**
  - Transcription and translation occur together.



(a) Prokaryotic cell. In a cell lacking a nucleus, mRNA produced by transcription is immediately translated without additional processing.

- **In eukaryotes:**
  - RNA transcripts are modified before becoming true mRNA



(b) Eukaryotic cell. The nucleus provides a separate compartment for transcription. The original RNA transcript, called pre-mRNA, is processed in various ways before leaving the nucleus as mRNA.

49

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8. Regulation of Gene Expression

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Gene Regulation in Prokaryotes

- **Prokaryotes** and **eukaryotes** alter gene expression in response to their changing environment.
- **In Prokaryotic:** Gene expression is controlled by the operon model.
- An **operon** is the entire stretch of DNA that includes the operator, the promoter, and the genes that they control.
- A cluster of functionally related genes can be under coordinated control by a single on-off "switch".
- The **regulatory** "switch" is a segment of DNA called an **operator** usually positioned within the promoter.
- The operon can be switched off by a protein **repressor ,** The repressor prevents gene transcription by binding to the operator and blocking RNA polymerase.

51

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Gene Regulation in Prokaryotes

- The **repressor** can be in an active or inactive form, depending on the presence of other molecules.
- A **corepressor** is a molecule that cooperates with a repressor protein to switch an operon off.
- For example, *E. coli* can synthesize the amino acid tryptophan.
- By default the *trp* operon is on and the genes for tryptophan synthesis are **transcribed.**
- When **tryptophan** is present, it binds to the *trp* repressor protein, which turns the operon off .
- The repressor is active only in the presence of its corepressor tryptophan; thus the *trp* operon is turned off (repressed) if tryptophan levels are high.

52

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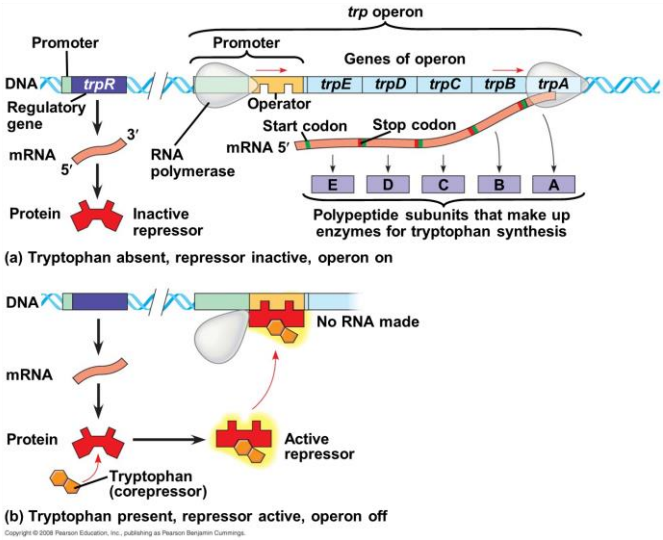
Gene Regulation in Prokaryotes



53

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Gene Regulation in Eukaryotes

- In multicellular eukaryotes, gene expression regulates development and is responsible for differences in cell types.
- **RNA** molecules play many roles in regulating gene expression in eukaryotes.
- Almost all the cells in an organism are genetically identical.
- Differences between cell types result from **differential gene expression**, the expression of different genes by cells with the same genome.
- **Errors** in gene expression can lead to diseases including cancer.
- Gene expression is regulated at many stages.

54

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9. Biotechnology

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The DNA Toolbox

- **Biotechnology** is the manipulation of organisms or their genetic components to make useful products.
- **Recombinant DNA**, nucleotide sequences from two different sources, often two species, are combined *in vitro* into the same DNA molecule.
- **Methods** for making recombinant DNA are central to **genetic engineering**, the direct manipulation of genes for practical purposes.
- **Microarray is a** DNA technology to measurement of gene expression of thousands of different genes.
- **Gene cloning** involves using bacteria to make multiple copies of a particular gene to producing **a protein product.**

56

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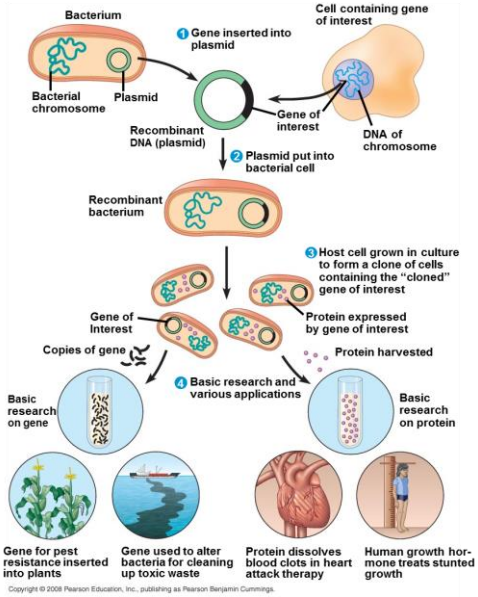
DNA Cloning and Its Applications



57

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- a A. Urry, Micheal L.Cain, Steven A. Wasserman, Peter V. Minorsky, Rebecca B. Orr, Neil A. Campbell, Publisher: Pearson, Year: 2020, ISBN: 9780135988046; 0135988047.
- 1- Mitosis https://www.youtube.com/watch?v=f-ldPgEfAHI.
- 2- Meiosis https://www.youtube.com/watch?v=VzDMG7ke69g.
- 3- DNA transcript and translate https://www.youtube.com/watch?v=8_f-8ISZ164&t=330s

58