

## Chapter # 2

# Genes and Genomics

# Genes and Genomics

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## CELL BIOLOGY

- ***Cell* is the building block of human body and it is critical to understand its structure and function so that better treatment modalities can be achieved.**
- **There is similarity and difference of intracellular and extracellular aspects of cells of prokaryotic and eukaryotic origin.**

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## CELL AS A BUILDING BLOCK OF LIFE

- The cell is the structural and functional unit of all known living organisms.
- It is the smallest unit of a living organism, and is often called the building block of life.
- Humans have approximately 100 trillion or  $10^{14}$  cells, an example of a *multicellular* organism. On the other hand, a single-celled bacterium is called *unicellular*.
- A typical cell size is 10 micrometer while a typical cell mass is 1 nanogram.  
All animals and plants are made of cells and such concept was originally coined by Aristotle (384-322 BC).

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## **CELL AS A BUILDING BLOCK OF LIFE**

- All animals and plants are made of cells and such concept was originally coined by Aristotle (384-322 BC).
- In 1665, Robert Hooke observed for the first time the structure of a cell under a very primitive microscope.
- In 1674, Antonie van Leeuwenhoek discovered cells with structural organization within the cell.
- In 1824, H.J. Dutrochet, a French scientist, gave the idea of the cell theory

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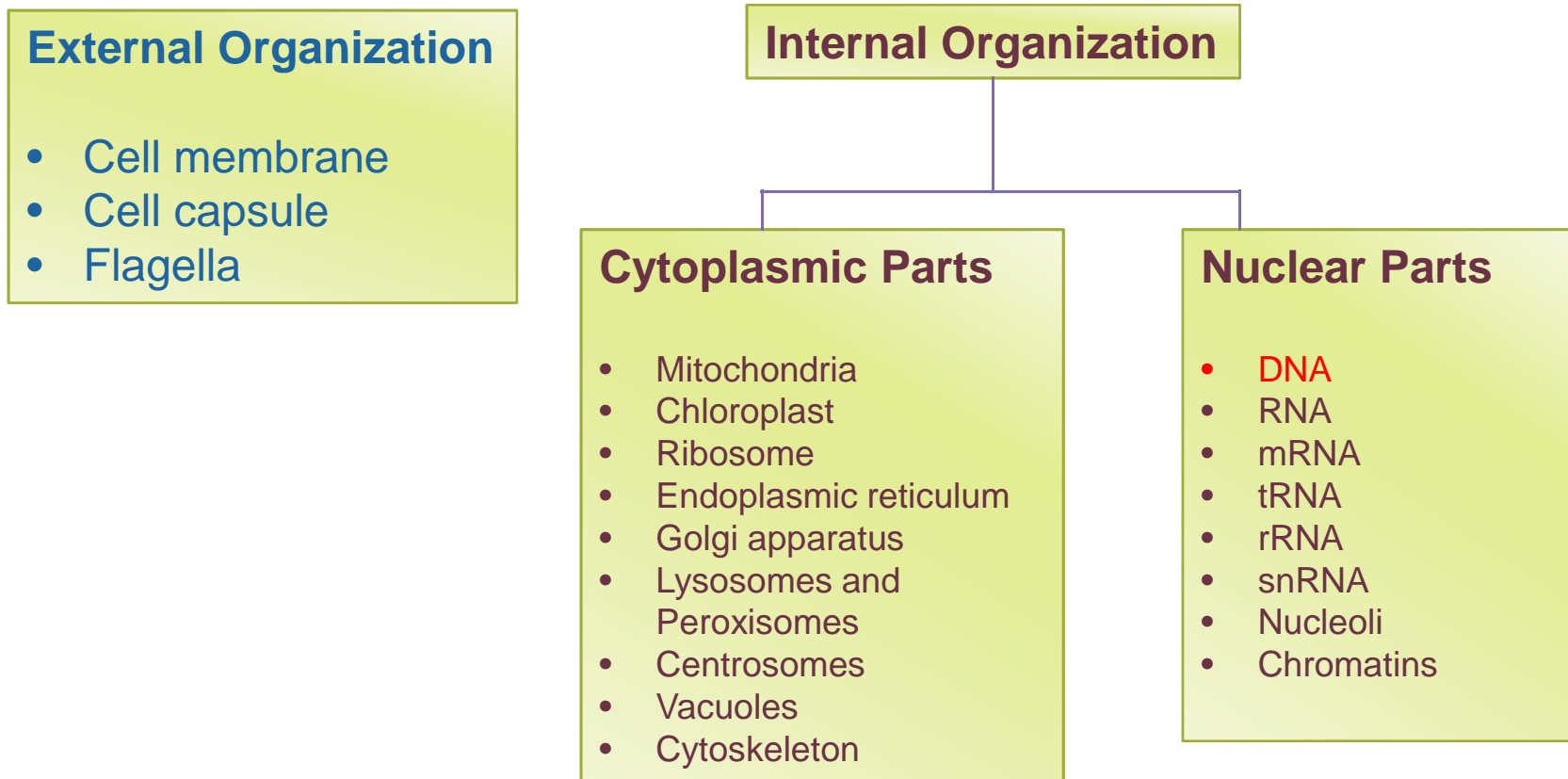
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## CLASSIFICATION OF CELL

- There are two different types of cells, the eukaryotic and prokaryotic cells.
- The **prokaryote cell** is simpler than a eukaryote cell, lacking a nucleus and most of the other organelles of eukaryotes.
- There are two kinds of prokaryotes, bacteria and archaea, These prokaryotes share a similar overall structure
- **Eukaryotic cells** are about 10 times the size of a typical prokaryote
- The major difference between prokaryotes and eukaryotes is that eukaryotic cells contain membrane-bound compartments in which specific metabolic activities take place

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## ORGANIZATION OF A CELL



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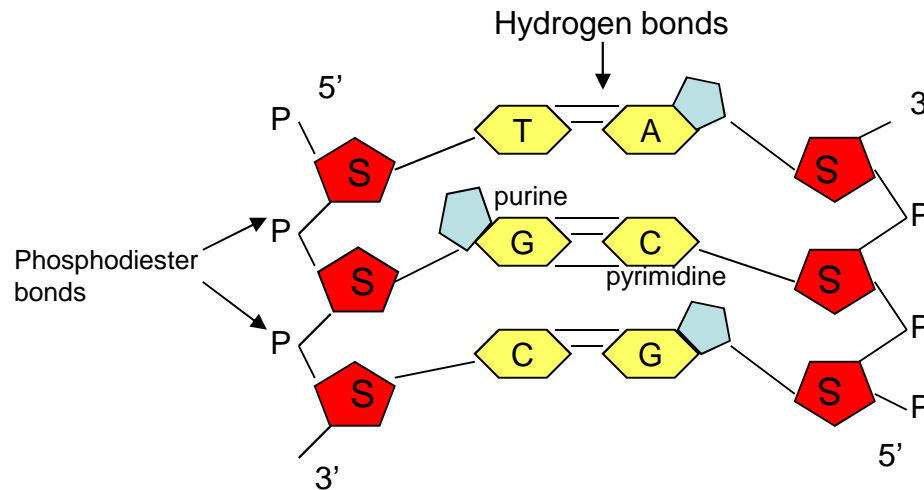
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## MACRO-MOLECULES OF BODY

- Living cells are primarily made up of water, a number of other molecules are also abundant within a cell such as *macromolecules*.
- Macromolecules provide structural support, store fuel, store and retrieve genetic information, and speed up biochemical reactions.
- There are four major types of macromolecules that play important functions in the life a cell:
  - **PROTEINS,**
  - **CARBOHYDRATES,**
  - **NUCLEIC ACID, AND**
  - **LIPIDS**

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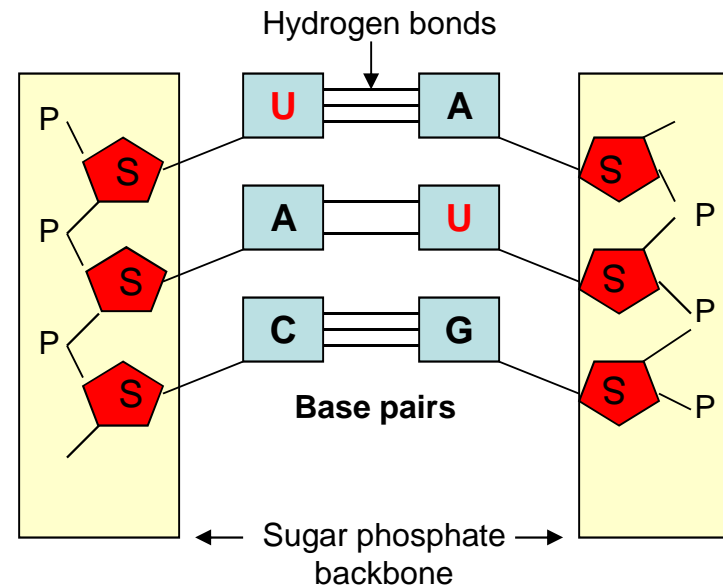
## DNA STRUCTURE



In most living organisms (except for viruses), genetic information is stored in DNA, which resides in the nucleus of living cells. It gets its name from the sugar molecule contained in its backbone (deoxyribose). However, it gets its significance from its unique structure. Four different nucleotide bases occur in DNA: adenine (A), cytosine (C), guanine (G), and thymine (T).

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## RNA STRUCTURE

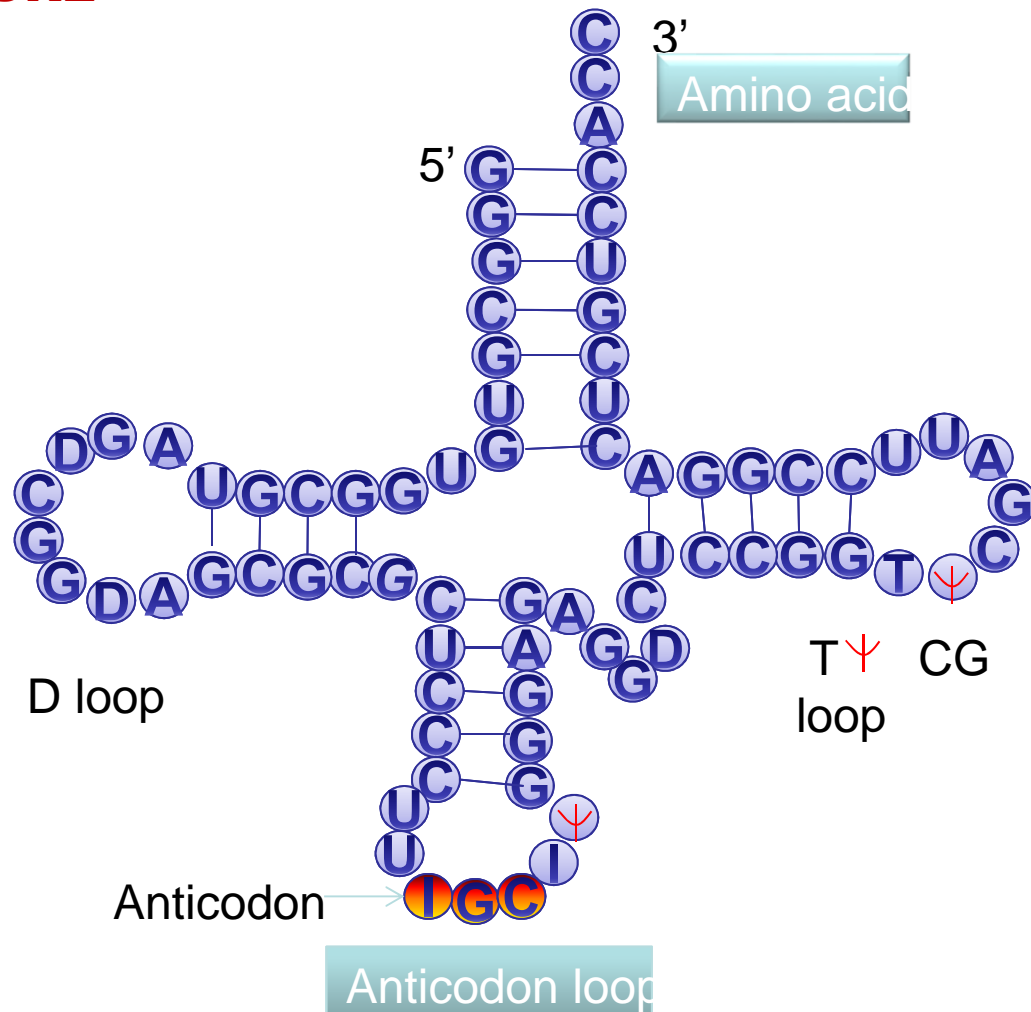


*Ribonucleic acid* (RNA) is a biologically important type of molecule that consists of a long chain of nucleotide units. RNA and DNA are both nucleic acids, but differ in three main ways. First, unlike DNA which is double-stranded, RNA is a single-stranded molecule in most of its biological roles and has a much shorter chain of nucleotides. Second, while DNA contains deoxyribose, RNA contains ribose, (there is no hydroxyl group attached to the pentose ring in the 2' position in DNA).

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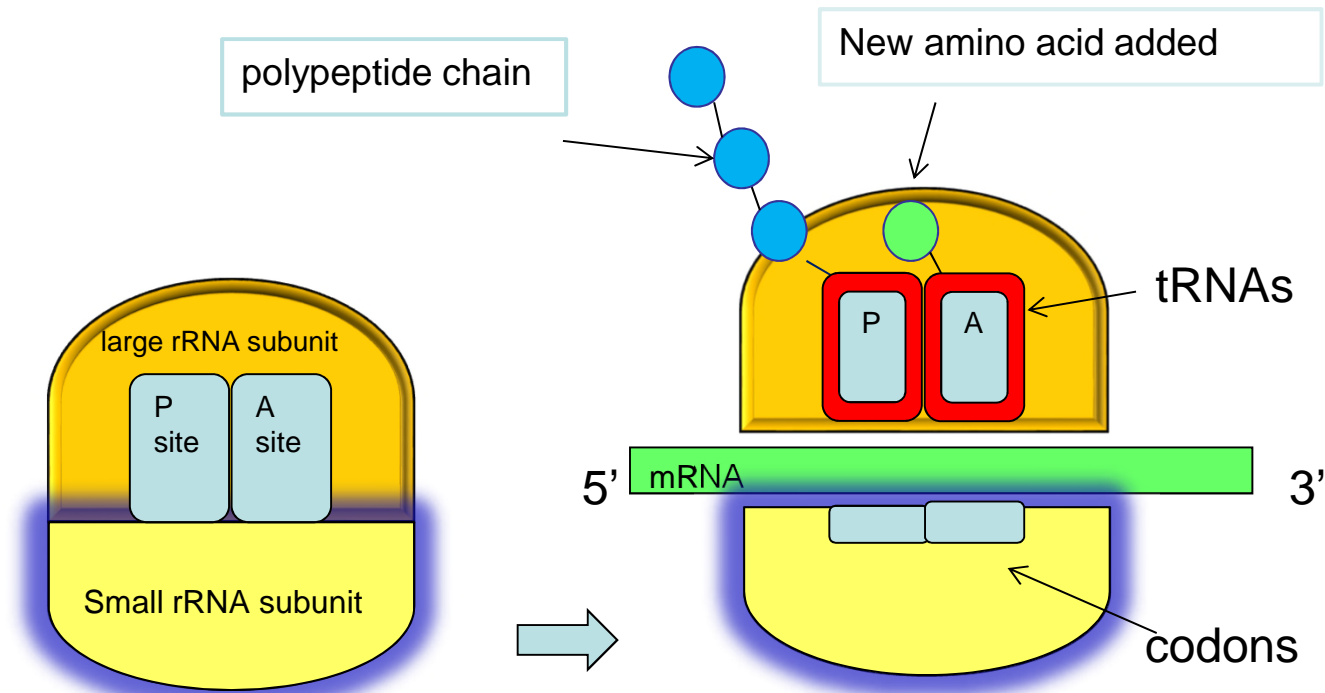
## TRANSFER RNA STRUCTURE

Transfer RNA (tRNA) is a small RNA molecule (usually about 74-95 nucleotides) that transfers a specific active amino acid to a growing polypeptide chain at the ribosomal site of protein synthesis during translation



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## RIBOSOMAL RNA STRUCTURE



*Ribosomal RNA* (rRNA) is the central component of the ribosome, the protein manufacturing machinery of all living cells. The function of the rRNA is to provide a mechanism for decoding mRNA into amino acids and to interact with the tRNAs during translation by providing peptidyl transferase activity. The tRNA then brings the necessary amino acids corresponding to the appropriate mRNA codon

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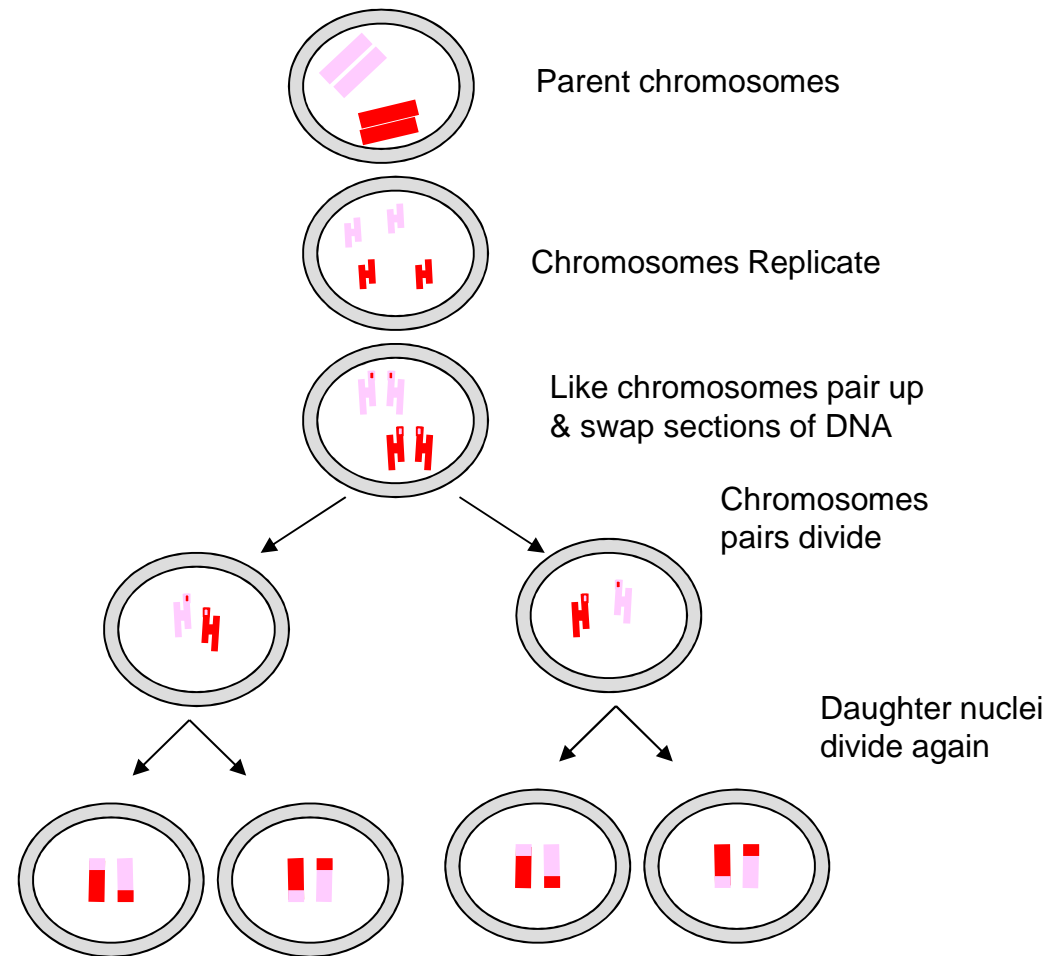
## CELL DIVISION

- All living cells undergo cell division in order to multiply and grow and there are two ways the cell can be divided such as **meiosis** and **mitosis**
- When cells are divided to yield two daughter cells, the genetic material must be divided equally so that each daughter cell contains identical DNA copies.

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## MEIOSIS

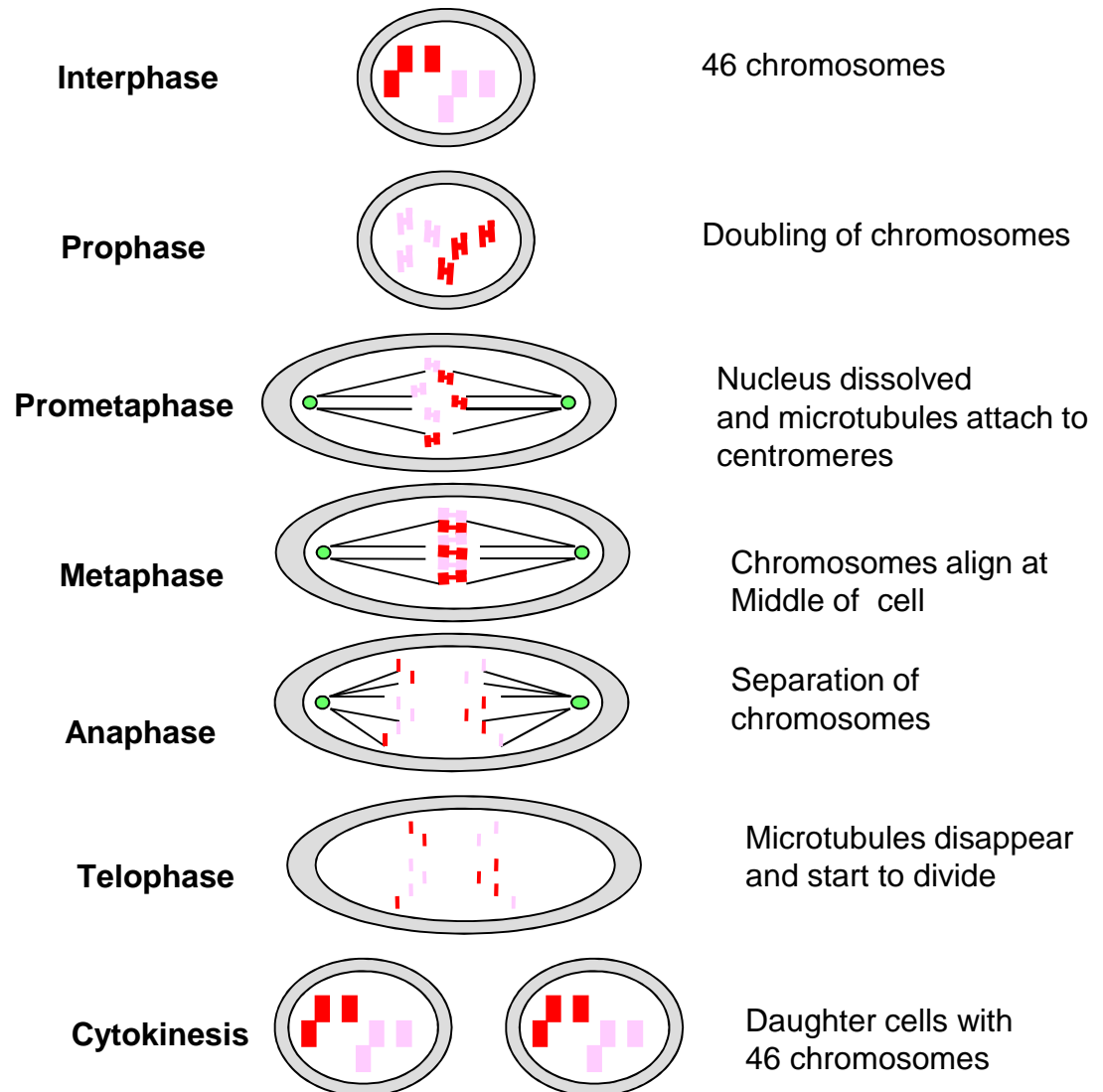
Meiosis is a process of reduction division in which the number of chromosomes per cell is reduced to half. In animals, meiosis always results in the formation of gametes, while in other organisms it can give rise to spores.



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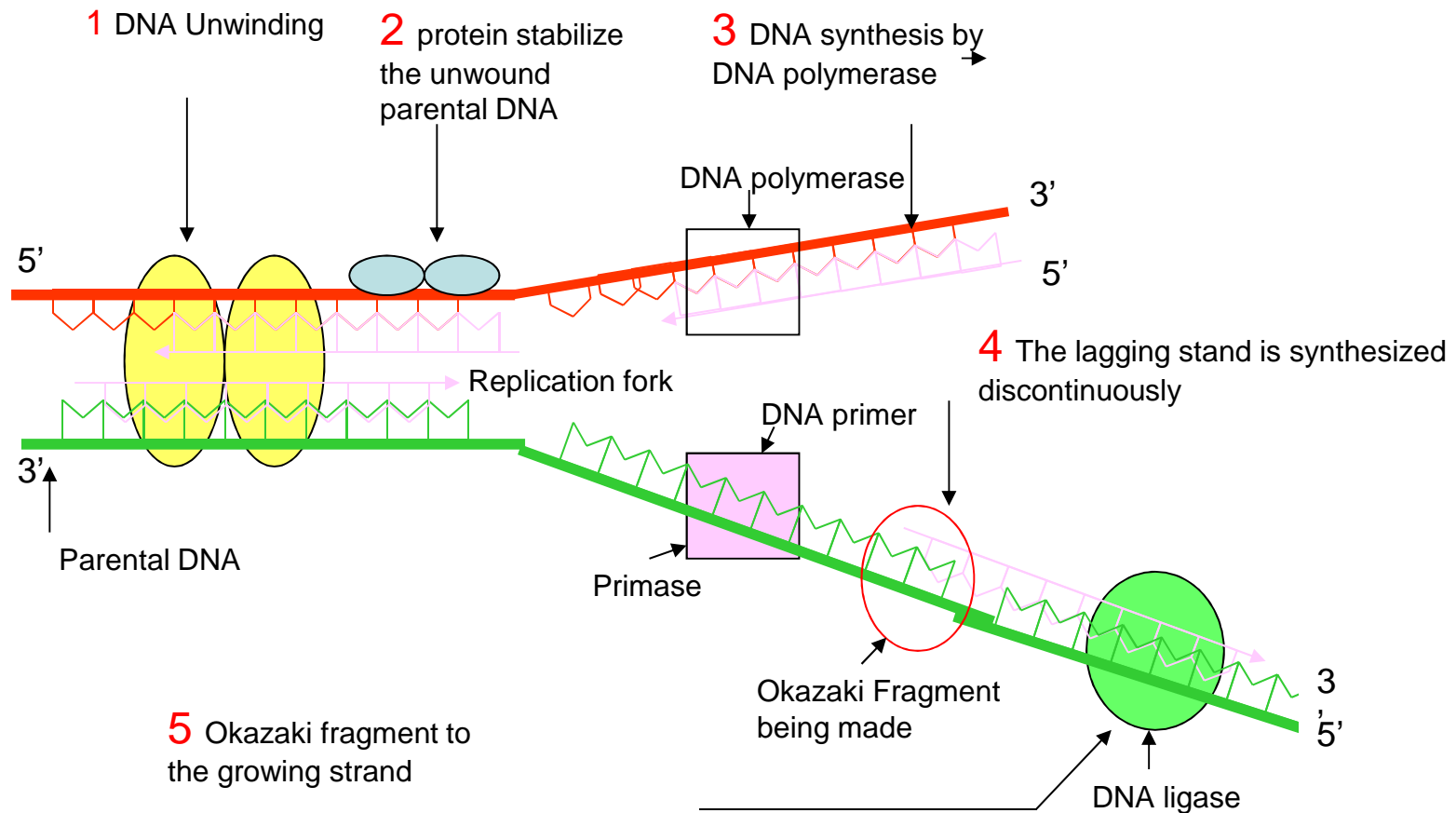
## MITOSIS

*Mitosis* is the process in which a eukaryotic cell separates the chromosomes in its cell nucleus into two identical sets in two daughter nuclei. It is generally followed immediately by cytokinesis, which divides the nuclei, cytoplasm, organelles and cell membrane into two daughter cells containing roughly equal shares of these cellular components



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## DNA REPLICATION PROCESS

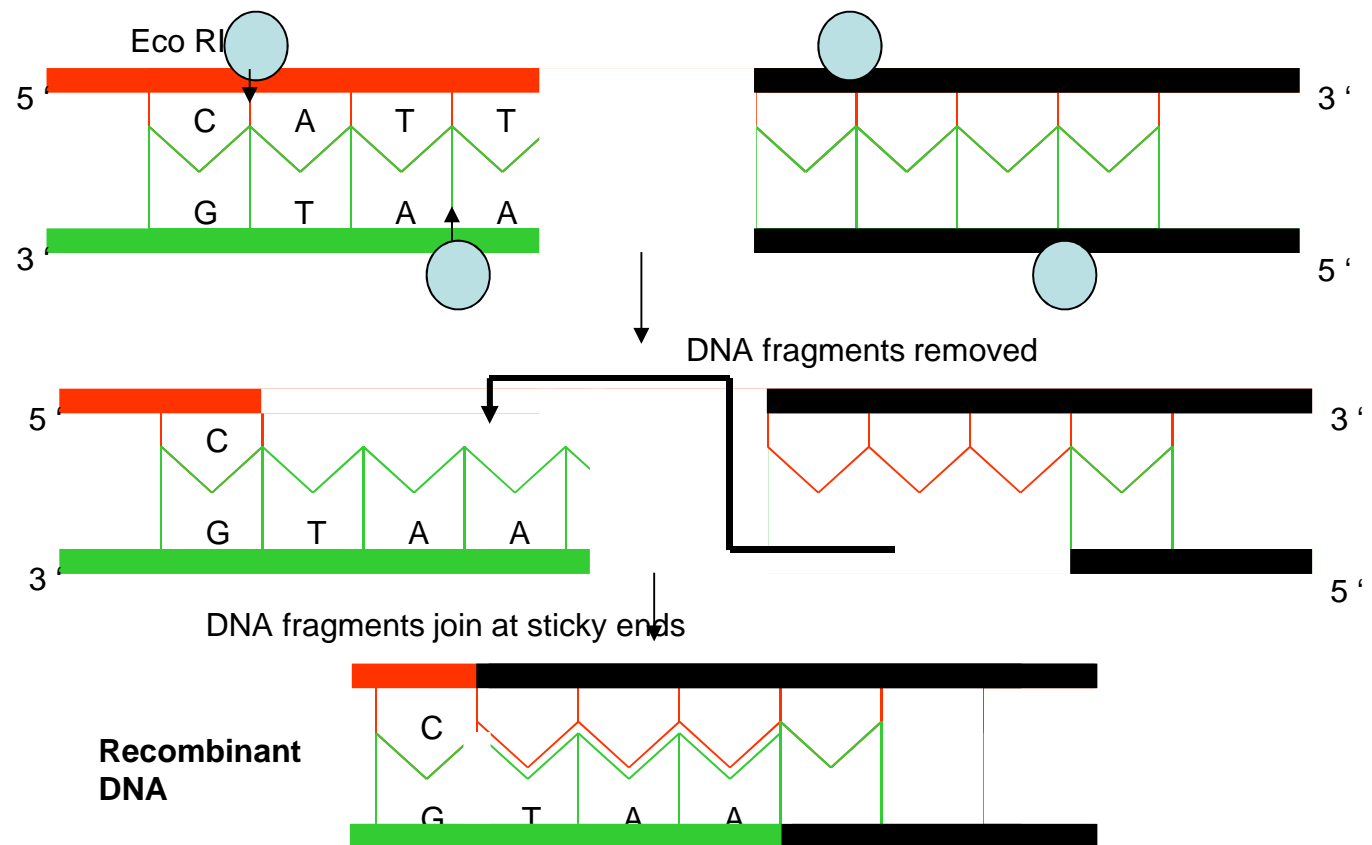


In a cell, DNA replication begins at specific locations in the genome, called "origins". Unwinding of DNA at the origin, and synthesis of new strands, forms a replication fork

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## ROLE OF RESTRICTION ENZYME

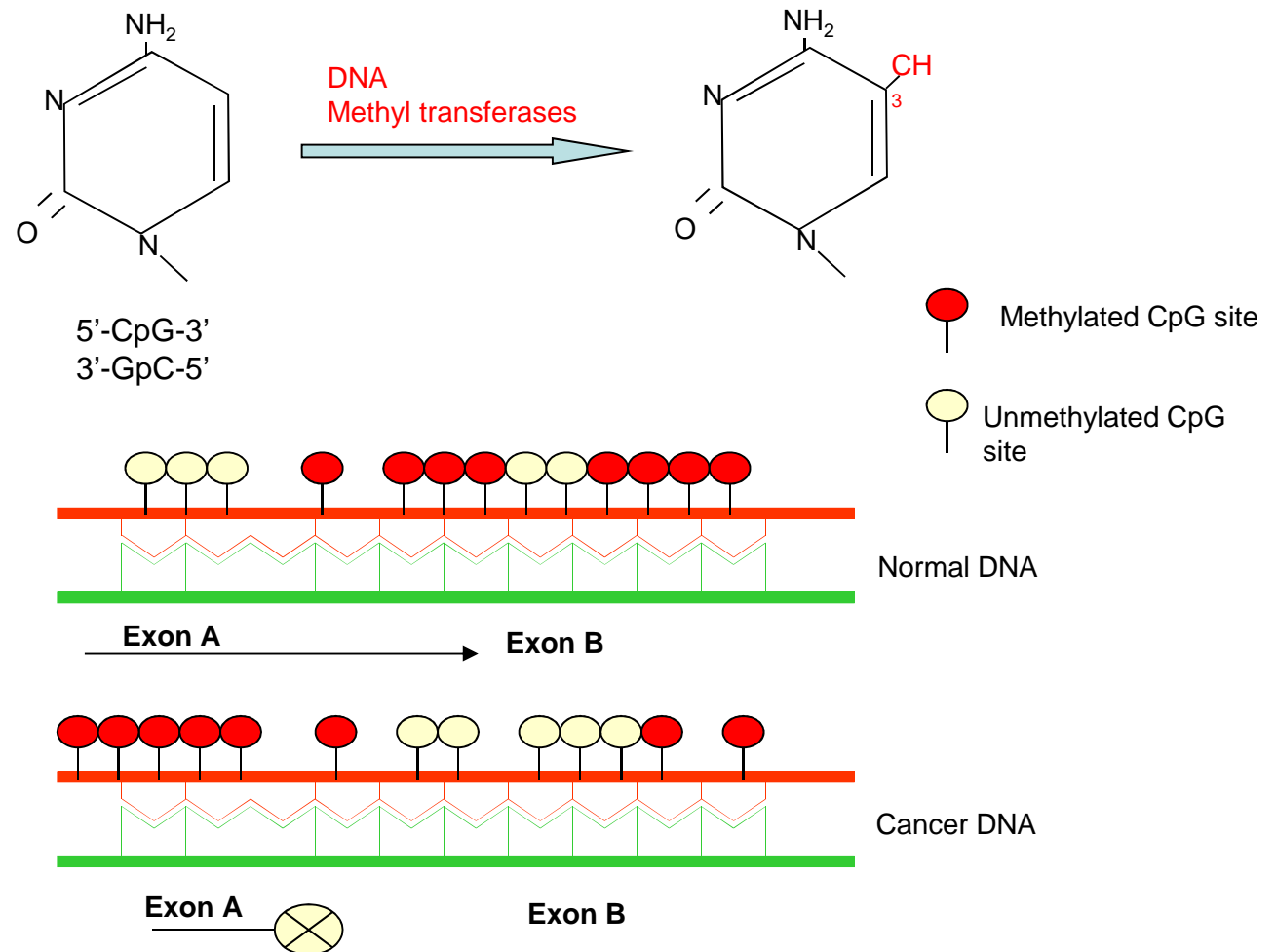
A **restriction enzyme** is an enzyme that cuts DNA at specific recognition nucleotide sequences (with Type II restriction enzymes cutting double-stranded DNA) known as restriction sites



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DNA methylation is one such post-synthesis modification. DNA methylation has been proven by research to be manifested in a number of biological processes such as regulation of imprinted genes, X chromosome inactivation, and tumor suppressor gene silencing in cancerous cells

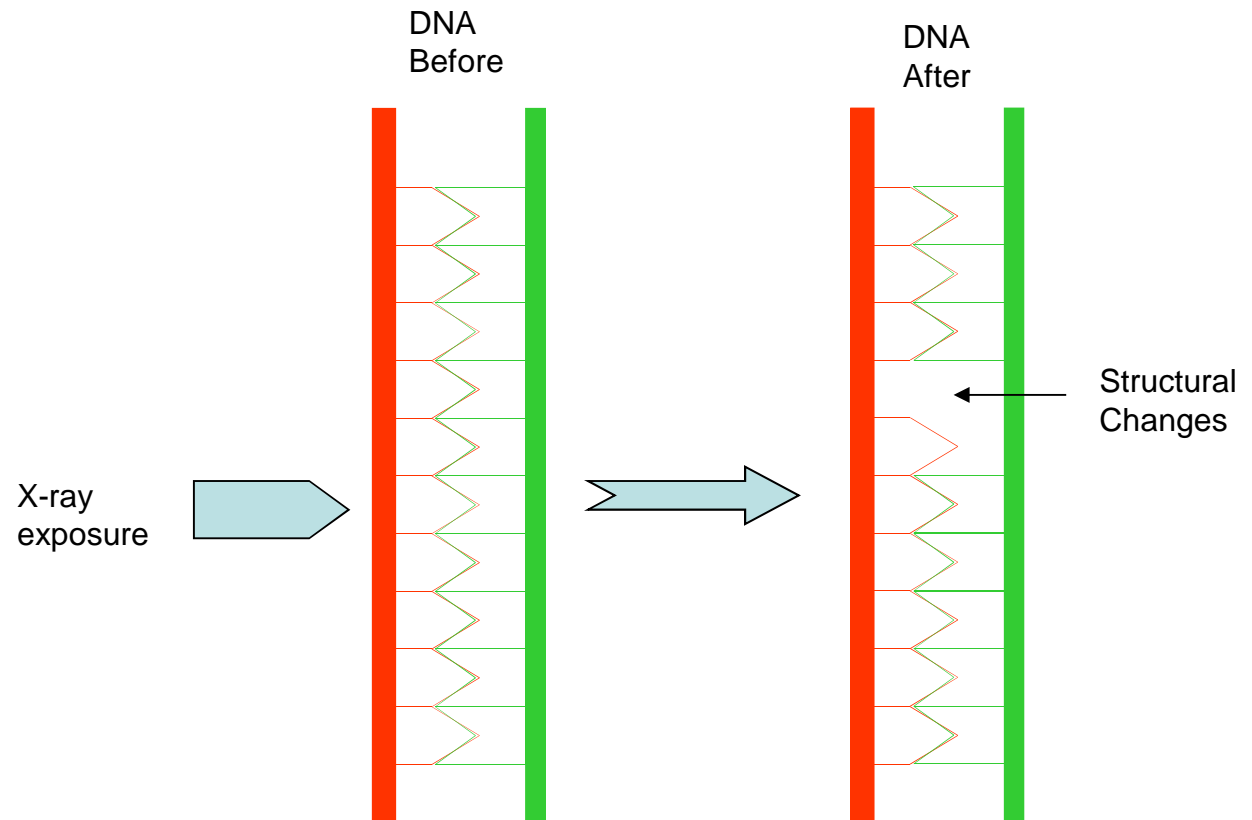
## DNA METHYLATION



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## DNA MUTATION PROCESS

*DNA mutation* is a change in the sequence of DNA. It can be caused by copying errors in the genetic material during cell division, by exposure to ultraviolet/ionizing radiation, chemical mutagens, or viruses, or by cellular processes such as hyper-mutation. It can also be induced by the organism itself.



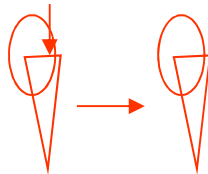
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## DNA AMPLIFICATION BY POLYMERASE CHAIN REACTION (PCR)

Forward Primer

Reverse Primer

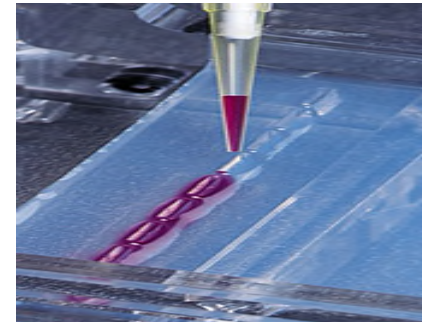
DNA sample



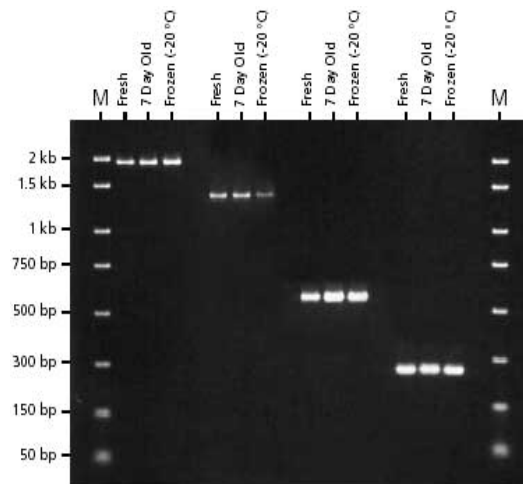
Sample preparation



PCR machine



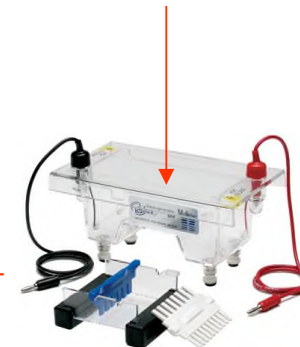
Sample loading on gel



Final PCR Product



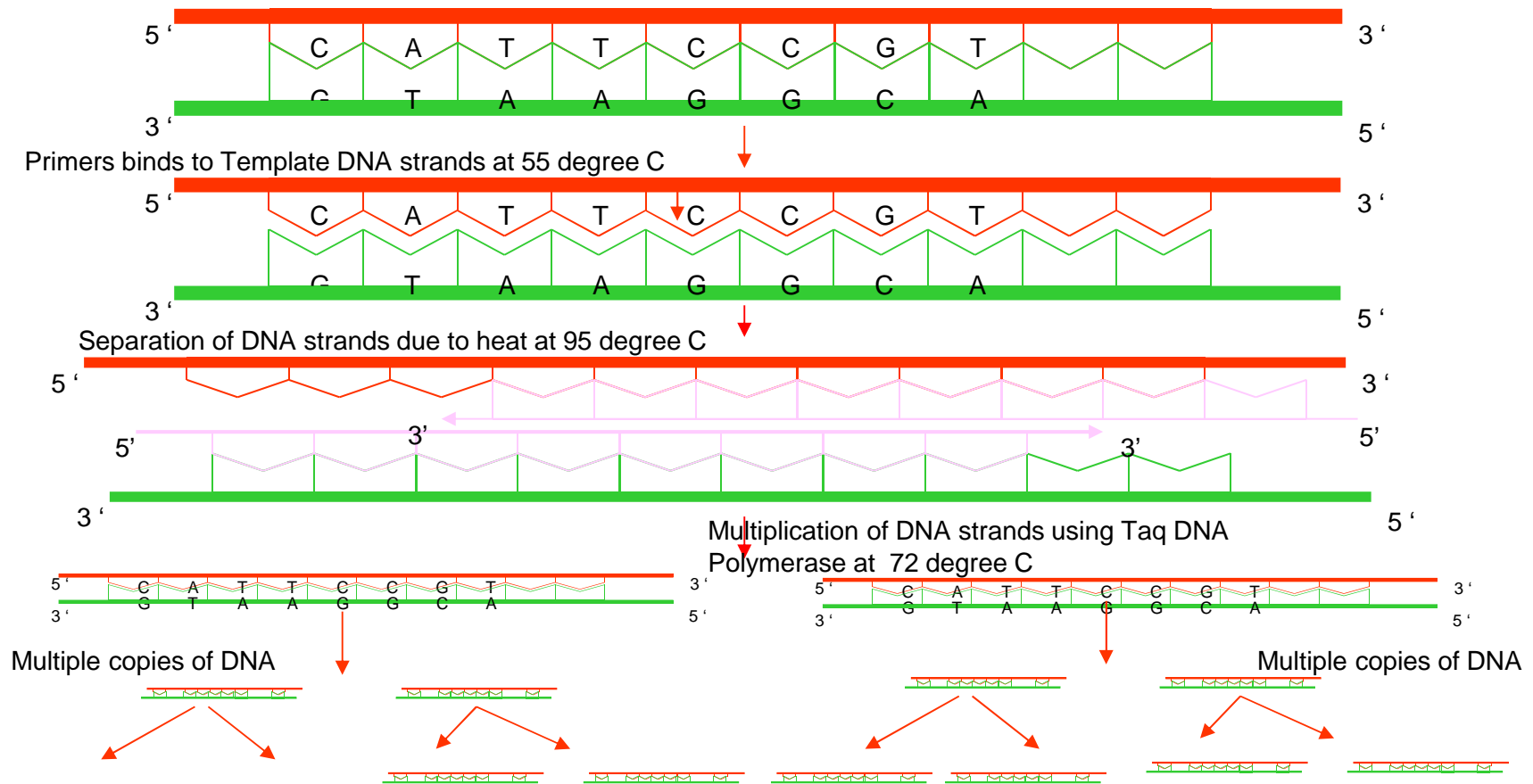
Gel documentation system



Gel electrophoresis

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## DNA MUTATION PROCESS



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## **FIND A SOLUTION FOR A PROBLEM**

- Q1. What is the cell theory?
- Q2. Describe the characteristics of a prokaryotic cell.
- Q3. Explain Mendelian's genetics.
- Q4. Explain supercoiling in DNA molecule.
- Q5. Describe the role of DNA polymerase in replication.
- Q6. What are topoisomerases and helicases?
- Q7. How does DNA methylation occur?
- Q8. What is a polymerase chain reaction?
- Q9. How does forensic DNA profiling done with PCR tool?

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## CRITICAL THINKING

Q1 In order to identify the real culprit among a group of crime suspects, what technique can be used to establish the identity of the culprit? Explain with suitable examples.

Q2 Is it possible to study the genetic information of an individual by working with mRNA only? Explain.

Q3 What would be the status of gene expression in case mRNA is not available?

Q4 What will happen if nuclear DNA is circular in shape and mitochondrial DNA is linear in shape?