

**Name: M. Asif Khan**

**ID : 14710**

**Paper: Human Genetics**

**Instructor: Fazli sir**

**Exam : summer Final term**

**Date : 28/9/2020**

**QUESTION NO 1 ANSWER:**

- 1) one PCR cycle comprises of 3 steps (a) **Denturation** (b) Annealing (c) **extension**
- 2) **Resting phase** is often called the resting phase but cell is not at rest
- 3) The unwound form of the chromosome is called **Autosomes**
- 4) According to **Law of segregation** law of Medal, during the formation of gametes the paired alleles separate randomly so that each gametes receives one alleles or the other.
- 5) **Natural selection** Is the differential survival and reproduction of individuals due to differences in the phenotype

**QUESTION NO 2 ANSWER**

**1) Interphase:**

Composed of G1, S, G2, where cells spends most (90%) of its time often call the resting phase but cell is not at rest cell is not dividing but it is active.

Often called the “resting” phase but cell is not at rest. Cell may not be dividing but it is active.

**2) Check points in cell division:**

are control mechanisms in the eukaryotic cell cycle which ensure its proper progression where a cell check to make sure it is able to continue to the next phase Animation.

**G2**, The cell size must be large enough environment must be suitable.

**3) DNA synthesis phase:**

Cell makes a copy of its entire set of chromosomes (DNA)

Q: why is this necessary ?

Q: what would the chromosome look like now ?

the phase of the cell cycle in which DNA is replicated, occurring between G1 phase and G2 phase. Since accurate duplication of the genome is critical to successful cell division, the processes that occur during S-phase are tightly regulated and widely conserved.

#### 4) G1, G2, G<sub>0</sub> phases:

##### **Gap 1 (G1) first growth phase**

Period of rapid growth and new proteins and organelles are produced

Chromosomes are unwound (chromatin) preparing for DNA synthesis (s phase)

##### **Gap2 (G2), second growth phase,**

Cell grows larger in size in preparation for cell division. Produces organelles and structures needed for cell division. E.g. centrioles and nucleolus are duplicated.

Shortest part of interphase.

##### **Gap zero (G<sub>0</sub>)**

Cell leaves cell cycle can be temporary or permanent but not necessarily dead.

e.g. neurons.

#### **QUESTION NO 3 ANSWER:**

**Mutations:** In biology, a mutation is a change in the genetic material. This means changes to the DNA or to the chromosomes which carry the DNA. These changes are heritable (can be passed on to the next generation) unless they have lethal effects. Mutations can happen for several reasons. It can happen because of error when meiosis produces the gametes (eggs & sperms). Damage by radiation, or by certain chemical may cause mutations. Mutations occur at random. Also, by derivation, an individual carrying the mutation may be called a mutant or a mutation. So is the trait (character) most obviously affected by the mutation.

#### **Characteristics of mutation**

English farmer Seth Wright recorded case of mutation first time in 1791 in male lamb with unusual sort legs

The term mutation is coined by Hugo de Vries in 1900 by his observation in *Oenothera*. Systemic study of mutation was started in 1910 when Morgan genetically analyzed white eye mutant of *Drosophila*.

H. J. Muller induced mutation in *Drosophila* by using X-rays in 1927 ; He was awarded by Nobel prize in 1946.

### **Types of mutation:**

### **DNA mutations**

When DNA is copied mistakes are sometimes made – these are called mutations. There are four main types of mutations:

- Deletion, where one or more DNA bases are left out.
- Insertion, where one or more extra base is put in.
- Substitution, where one or more bases are changed for another base in the sequence.
- Duplication, where whole genes are copied.

### **Chromosome mutations**

These terms are explained in the third diagram.

- Deletion: a piece of chromosome is lost, together with any genes which may be on it.
- Duplication: part of a chromosome is repeated
- Inversion: part of a chromosome is reversed end to end
- Insertion: a smaller chromosome is added into a longer chromosome
- Translocation: part of a chromosome gets moved onto another chromosome.

### **QUESTION NO 4 ASNWER:**

#### **Mendal Genetics:**

Austrian monk

Born in 1822 near Brunn in Austria in a poor family.

Joined the St. Augustinian monastery.

Was send to university of Vienna.

Presenting his findings before national History society of Brunn in 1865.

But his finding were discarded due to the prevailing theories of Evaluation.

Later on his theories were again discovered simultaneously by;

Hugo de Vries in Holland

Carl Correns in Germany

Eric von Tschermak in Australia; in 1900.

**Mendel's Law of Segregation** states that a diploid organism passes a randomly selected allele for a trait to its offspring, such that the offspring receives one allele from each parent.

**Mendel's law of independent assortment** states that genes do not influence each other with regard to the sorting of alleles into gametes: every possible combination of alleles for every gene is equally likely to occur.

## QUESTION NO 5 ANSWER :

### Mitosis Definition

“Mitosis is that step in the cell cycle where the newly formed DNA is separated and two new cells are formed with the same number and kind of chromosomes as the parent nucleus.”

Mitosis is a process of asexual reproduction observed in unicellular organisms. Read on to explore what is mitosis, and the different stages of mitosis.

Table of Contents

Explanation

Features

Stages

Interphase

Prophase

Prometaphase

Metaphase

Anaphase

Telophase

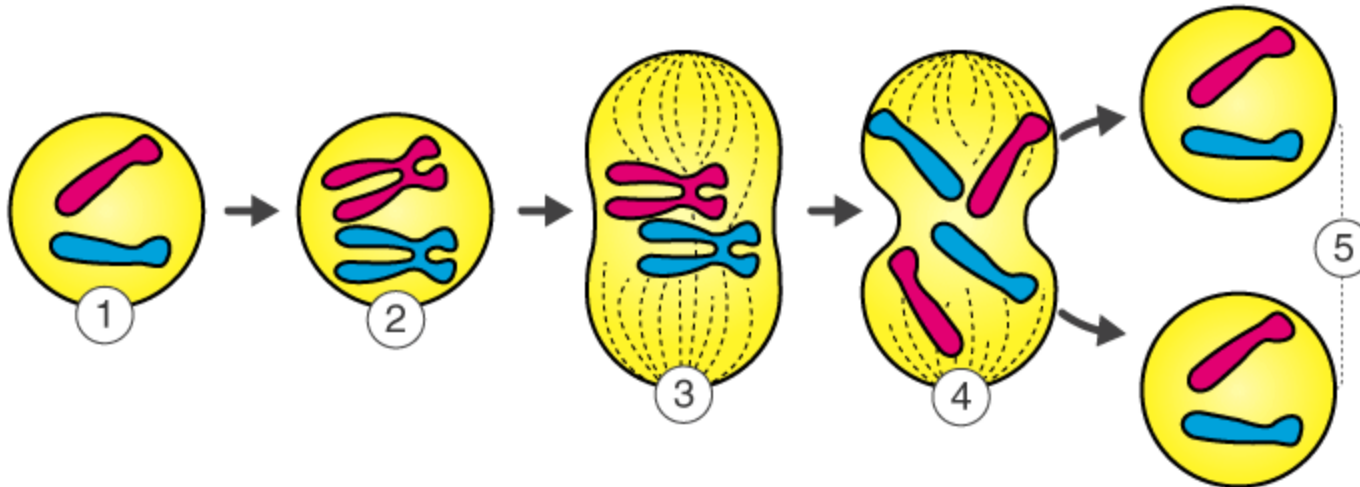
Function

Significance

**What is mitosis**

Cell division is the driving process of reproduction at the cellular level. Most eukaryotic cells divide in a manner where the ploidy or the number of chromosomes remains the same, except in the case of germ cells where the number of chromosomes is halved.

## MITOSIS : EQUATIONAL DIVISION



1 Interphase | 2 Prophase | 3 Metaphase | 4 Anaphase | 5 Telophase

Mitosis Diagram showing the different stages of mitosis

Mitosis is the phase of the cell cycle where the nucleus of a cell is divided into two nuclei with an equal amount of genetic material in both the daughter nuclei. It succeeds the G<sub>2</sub> phase and is succeeded by cytoplasmic division after the separation of the nucleus.

Mitosis is essential for the growth of the cells and the replacement of worn-out cells. Abnormalities during mitosis may alter the DNA, resulting in genetic disorders.

### Features of mitosis

In each cycle of cell division, two daughter cells are formed from the parent cell.

The cell is also known as equational cell division because the chromosome number in the parent cell and daughter cell is the same.

In plants, mitosis leads to the growth of vegetative parts of the plant like root tip, stem tip, etc.

Segregation and combination do not occur in this process.

The processes occurring during mitosis have been divided into different stages.

### Stages of mitosis

Right before prophase, the cell spends most of its life in the interphase, where preparations are made before the beginning of mitosis (the DNA is copied). However, since the actual process involves the division of the nucleus, prophase is technically the first stage of this process.

The different stages of mitosis occurring during **cell division** are given as follows-

### **Interphase**

Before entering mitosis, a cell spends a period of its growth under interphase. It undergoes the following phases when in interphase:

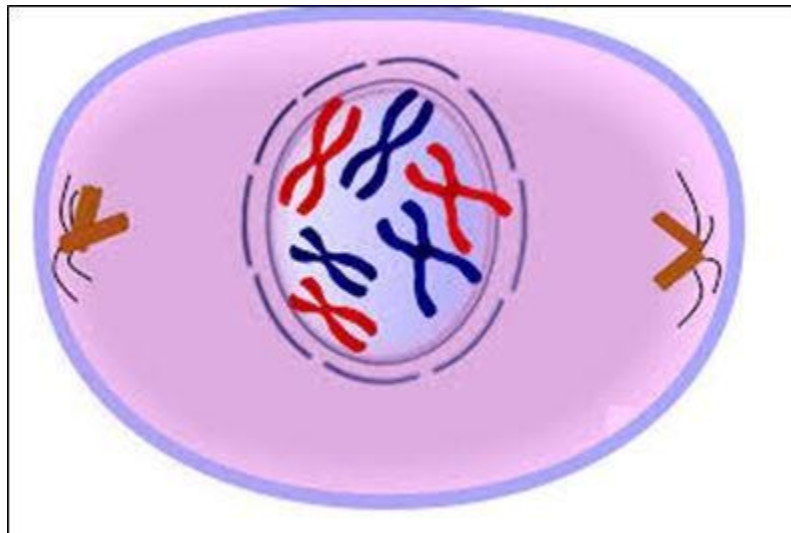
- G1 Phase: This is the period before the synthesis of DNA.
- S Phase: This is the phase during which DNA synthesis takes place.
- G2 Phase: This is the phase between the end of DNA synthesis and the beginning of prophase.

### **prophase**

Prophase immediately follows S and G2 phase of the cycle and is marked by condensation of the genetic material to form compact mitotic chromosomes composed of two chromatids attached at the centromere.

The completion of prophase is characterised by the initiation of the assembly of the mitotic spindle, the microtubules, and the proteinaceous components of cytoplasm that help in the process.

The nuclear envelope starts disintegrating.



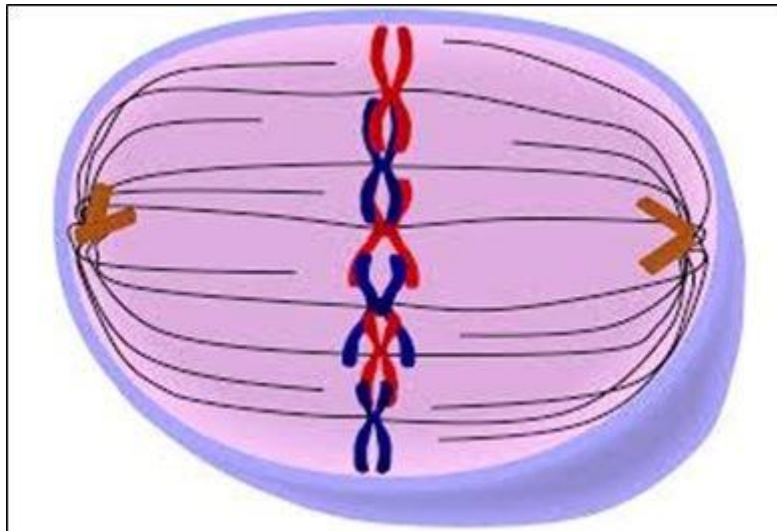
Prophase

### **Prometaphase**

In the prometaphase, the nuclear envelope disintegrates. Now the microtubules are allowed to extend from the centromere to the chromosome. The microtubules attach to the kinetochores which allow the cell to move the chromosome around.

### **Metaphase**

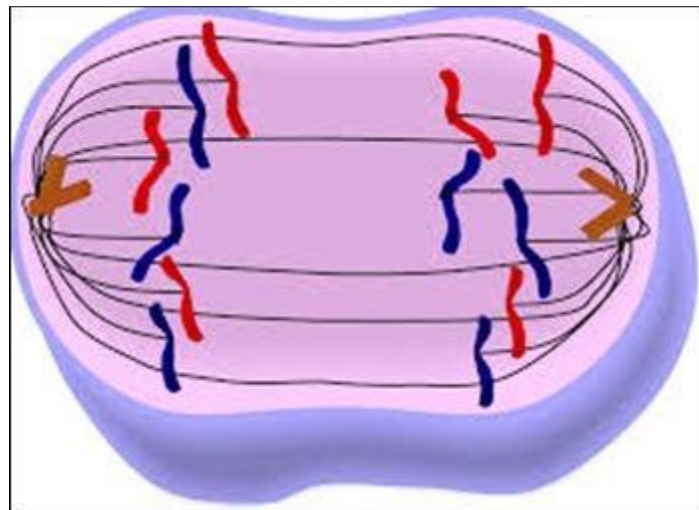
At this stage, the microtubules start pulling the chromosomes with equal force, and the chromosome ends up in the middle of the cell. This region is known as the metaphase plate. Thus, each cell gets an entire functioning genome.



Metaphase

### Anaphase

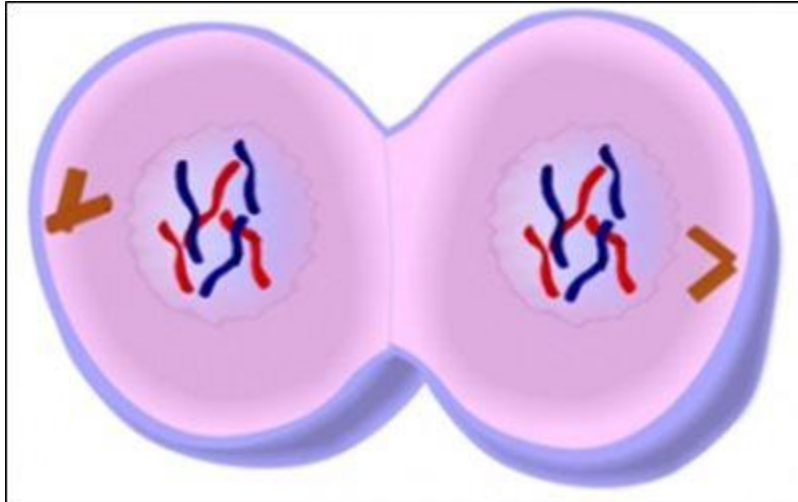
The splitting of the sister chromatids marks the onset of anaphase. These sister chromatids become the chromosome of the daughter nuclei. The chromosomes are then pulled towards the pole by the fibres attached to the kinetochores of each chromosome. The centromere of each chromosome leads at the edge while the arms trail behind it.



Anaphase

### Telophase

The chromosome that cluster at the two poles start coalescing into an undifferentiated mass, as the nuclear envelope starts forming around it. The nucleolus, Golgi bodies and ER complex, which had disappeared after prophase start to reappear.



Telophase

Telophase is followed by cytokinesis, which denotes the division of the cytoplasm to form two daughter cells. Thus, it marks the completion of cell division.

Also Read: cell cycle

### **Function of Mitosis**

Following are the two important functions of mitosis:

- Mitosis helps in the development of an organism. In single-celled organisms, mitosis is the process of asexual reproduction.
- Mitosis helps in the replacement of damaged tissues. The cells near the damaged cells begin mitosis when they do not sense the neighbouring cells. The dividing cells reach each other and cover the damaged cells.

### **Significance of Mitosis:**

- Mitosis is responsible for the development of the zygote into an adult.
- Equal distribution of chromosomes to each daughter cell.
- It is responsible for the growth and development of an individual.
- It maintains the constant number of chromosomes in all body cells of an organism.
- Mitosis is required for asexual reproduction, vegetative propagation in plants and also responsible for repair and regeneration of damaged tissues.
- Mitosis helps in maintaining purity of genome as there is no recombination or crossing over takes place.
- It is responsible for repair and regeneration of old and damaged cells in animals e.g. gut epithelium, blood cells, etc.