**Name Fahad Ali**

**ID# 13708**

**Program BS MLT**

**Department Allied Health Science**

**Instructor name sir zahir mian**

**Subject human genetic**

**Date 28-09-2020**

Answer no 1 fill in the blanks

1. a-Denaturation

b- Annealing

c-extension

Question no 2

Answers

1. Interphase is the longest stage in the [eukaryote](https://biologydictionary.net/eukaryote/) [*cell cycle*](https://biologydictionary.net/cell-cycle/). During interphase, the [cell](https://biologydictionary.net/cell/) acquires nutrients, creates and uses proteins and other molecules, and starts the process of [cell division](https://biologydictionary.net/cell-division/) by replicating the DNA. Interphase is divided into three distinct stages, Gap 1, Synthesis, and Gap 2, which are discussed below. The purpose of interphase in all cell types is to prepare for cell division, which happens in a different stage of the cell cycle.
2. **Cell cycle checkpoints**  are control mechanisms in the [eukaryotic](https://en.wikipedia.org/wiki/Eukaryote) [cell cycle](https://en.wikipedia.org/wiki/Cell_cycle) which ensure its proper progression. Each checkpoint serves as a potential termination point along the [cell cycle](https://en.wikipedia.org/wiki/Cell_cycle), during which the conditions of the cell are assessed, with progression through the various phases of the cell cycle occurring only when favorable conditions are met. There are many checkpoints in the cell cycle, but the three major ones are: the G1 checkpoint, also known as the Start or [restriction checkpoint](https://en.wikipedia.org/wiki/Restriction_point) or Major Checkpoint; the [G2/M checkpoint](https://en.wikipedia.org/wiki/G2/M_checkpoint); and the metaphase-to-anaphase transition, also known as the [spindle checkpoint](https://en.wikipedia.org/wiki/Spindle_checkpoint). Progression through these checkpoints is largely determined by the activation of [cyclin-dependent kinases](https://en.wikipedia.org/wiki/Cyclin-dependent_kinase%22%20%5Co%20%22Cyclin-dependent%20kinase) by regulatory [protein subunits](https://en.wikipedia.org/wiki/Protein_subunit) called [cyclins](https://en.wikipedia.org/wiki/Cyclin%22%20%5Co%20%22Cyclin), different forms of which are produced at each stage of the cell cycle to control the specific events that occur therein.
3. **DNA Synthesis phase**

is the phase of the [cell cycle](https://en.wikipedia.org/wiki/Cell_cycle) in which [DNA](https://en.wikipedia.org/wiki/DNA) is [replicated](https://en.wikipedia.org/wiki/DNA_replication), occurring between [G1 phase](https://en.wikipedia.org/wiki/G1_phase) and [G2 phase](https://en.wikipedia.org/wiki/G2_phase).[[1]](https://en.wikipedia.org/wiki/S_phase#cite_note-:5-1) 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 phase

The first phase of interphase is G1 phase, from the end of the previous Mitosis phase until the beginning of DNA replication is called G1 (G indicating gap). It is also called the growth phase. During this phase the biosynthetic activities of the cell, which had been considerably slowed down during M phase, resume at a high rate. This phase is marked by synthesis of various enzymes that are required in S phase, mainly those needed for DNA replication. Duration of G1 is highly variable, even among different cells of the same species.

### G2 phase

After S phase or replication cell then enters the G2 phase, which lasts until the cell enters mitosis. Again, significant biosynthesis occurs during this phase, mainly involving the production of microtubules, which are required during the process of mitosis. Inhibition of protein synthesis during G2 phase prevents the cell from undergoing mitosis.

### G0 phase

Cells that have temporarily stopped dividing are said to have entered a state of quienscence,called **G0 PHASE**. G0 phase is viewed as either an extended G1 phase, where the cell is neither dividing nor preparing to divide, or a distinct quiescent stage that occurs outside of the cell cycle. G0 is sometimes referred to as a "post-mitotic" state, since cells in G0 are in a non-dividing phase outside of the cell cycle. Some types of cells, such as nerve and heart muscle cells, become post-mitotic when they reach maturity (i.e., when they are terminally differentiated) but continue to perform their main functions for the rest of the organism's life. Multinucleated muscle cells that do not undergo cytokinesis are also often considered to be in the G0 stage. On occasion, a distinction in terms is made between a G0 cell and a 'post-mitotic' cell (e.g., heart muscle cells and neurons), which will never enter the G1 phase, whereas other G0 cells may.

**Answer No 3**

**Mutation**

 Mutation is a change that occurs in our DNA sequence, either due to mistakes when the DNA is copied or as the result of environmental factors such as UV light and cigarette smoke.

**Types of mutation**

**Missense mutation**

This type of mutation is a [change in one DNA base pair](https://ghr.nlm.nih.gov/primer/illustrations/missense.jpg) that results in the substitution of one amino acid for another in the protein made by a gene.

**Nonsense mutation**

A [nonsense mutation](https://ghr.nlm.nih.gov/primer/illustrations/nonsense.jpg) is also a change in one DNA base pair. Instead of substituting one amino acid for another, however, the altered DNA sequence prematurely signals the cell to stop building a protein. This type of mutation results in a shortened protein that may function improperly or not at all.

**Insertion**

An [insertion](https://ghr.nlm.nih.gov/primer/illustrations/insertion.jpg) changes the number of DNA bases in a gene by adding a piece of DNA. As a result, the protein made by the gene may not function properly.

**Deletion**

A [deletion](https://ghr.nlm.nih.gov/primer/illustrations/deletion.jpg) changes the number of DNA bases by removing a piece of DNA. Small deletions may remove one or a few base pairs within a gene, while larger deletions can remove an entire gene or several neighboring genes. The deleted DNA may alter the function of the resulting protein(s).

**Duplication**

A [duplication](https://ghr.nlm.nih.gov/primer/illustrations/duplication.jpg) consists of a piece of DNA that is abnormally copied one or more times. This type of mutation may alter the function of the resulting protein.

**Frameshift mutation**

This type of mutation occurs when the addition or loss of DNA bases changes a gene's reading frame. A reading frame consists of groups of 3 bases that each code for one amino acid. A [frameshift mutation](https://ghr.nlm.nih.gov/primer/illustrations/frameshift.jpg%22%20%5Co%20%22Image%22%20%5Ct%20%22_blank) shifts the grouping of these bases and changes the code for amino acids. The resulting protein is usually nonfunctional. Insertions, deletions, and duplications can all be frameshift mutations.

**Repeat expansion**

Nucleotide repeats are short DNA sequences that are repeated a number of times in a row. For example, a trinucleotide repeat is made up of 3-base-pair sequences, and a tetranucleotide repeat is made up of 4-base-pair sequences. A [repeat expansion](https://ghr.nlm.nih.gov/primer/illustrations/repeatexpansion.jpg) is a mutation that increases the number of times that the short DNA sequence is repeated. This type of mutation can cause the resulting protein to function improperly.

**Answer No 4**

**Mendel law of segregation**

The principles that govern heredity were discovered by a monk named [Gregor Mendel](https://www.thoughtco.com/about-gregor-mendel-1224841) in the 1860s. One of these [principles](https://www.thoughtco.com/mendels-law-of-independent-assortment-373458), now called [Mendel's Law of Segregation](https://www.thoughtco.com/mendels-law-373515), states that [allele](https://www.thoughtco.com/allele-a-genetics-definition-373460) pairs separate or segregate during [gamete](https://www.thoughtco.com/gametes-373465) formation and randomly unite at [fertilization](https://www.thoughtco.com/sexual-reproduction-types-of-fertilization-373440).

## The Four Concepts

There are four main concepts related to this principle:

1. A [gene](https://www.thoughtco.com/genes-373456) can exist in more than one form or allele.
2. Organisms inherit two alleles for each trait.
3. When [sex cells](https://www.thoughtco.com/sex-cells-meaning-373386) are produced (by [meiosis](https://www.thoughtco.com/stages-of-meiosis-373512)), allele pairs separate leaving each [cell](https://www.thoughtco.com/facts-about-cells-373372) with a single allele for each trait.
4. When the two alleles of a pair are different, one is dominant and the other is recessive.

For example, the gene for seed color in pea plants exists in two forms. There is one form or allele for yellow seed color (Y) and another for green seed color (y). In this example, the allele for yellow seed color is dominant, and the allele for green seed color is recessive. When the alleles of a pair are different ([heterozygous](https://www.thoughtco.com/heterozygous-definition-373468)), the dominant allele trait is expressed, and the recessive allele trait is masked. Seeds with the [genotype](https://www.thoughtco.com/genotype-vs-phenotype-1224568) of (YY) or (Yy) are yellow, while seeds that are (yy) are green.

## Genetic Dominance

Mendel formulated the law of segregation as a result of performing [monohybrid cross](https://www.thoughtco.com/monohybrid-cross-a-genetics-definition-373473) experiments on plants. The specific traits that he studied exhibited complete [dominance](https://www.thoughtco.com/genetic-dominance-373443). In complete dominance, one [phenotype](https://www.thoughtco.com/phenotype-373475) is dominant, and the other is recessive. Not all types of genetic inheritance, however, show total dominance.

In [incomplete dominance](https://www.thoughtco.com/incomplete-dominance-a-genetics-definition-373471), neither allele is completely dominant over the other. In this type of intermediate inheritance, the resulting offspring exhibit a phenotype that is a mixture of both parent phenotypes. Incomplete dominance is seen in snapdragon plants. Pollination between a plant with red flowers and one with white flowers produces a plant with pink flowers.

In codominance relationships, both alleles for a trait are fully expressed. Codominance is exhibited in tulips. [Pollination](https://www.thoughtco.com/facts-about-pollen-373610) that occurs between red and white tulip plants can result in a plant with flowers that are both red and white. Some people get confused about the differences between incomplete dominance and codominance.

**Mendel law of independent assortment**

In the 1860s, a monk named Gregor Mendel discovered many of the principles that govern heredity. One of these principles, now known as [Mendel's law of independent assortment](https://www.thoughtco.com/independent-assortment-373514), states that [allele](https://www.thoughtco.com/allele-a-genetics-definition-373460) pairs separate during the formation of [gametes](https://www.thoughtco.com/gametes-373465). This means that traits are transmitted to offspring independently of one another.

**Key Takeaways**

* Due to the law of independent assortment, traits are transmitted from parents to offspring independently of one another.
* Mendel's law of segregation is closely related to and foundational to his law of independent assortment.
* Not all inheritance patterns conform to Mendelian segregation patterns.
* Incomplete dominance results in a third phenotype. This phenotype is an amalgam of the parent alleles.
* In co-dominance, both of the parental alleles are expressed fully. The result is a third phenotype that has characteristics of both alleles.

Mendel discovered this principle after performing [dihybrid crosses](https://www.thoughtco.com/dihybrid-cross-a-genetics-definition-373463) between plants that had two traits, such as seed color and pod color, that differed from one another. After these plants were allowed to self-pollinate, he noticed that the same ratio of 9:3:3:1 appeared among the offspring. Mendel concluded that traits were transmitted to offspring independently.

The image above shows a true-breeding plant with the dominant traits of green pod color (GG) and yellow seed color (YY) being cross-pollinated with a [true-breeding plant](https://www.thoughtco.com/true-breeding-plant-373476) with yellow pod color (gg)and green seed color (yy). The resulting offspring are all [heterozygous](https://www.thoughtco.com/heterozygous-definition-373468) for green pod color and yellow seed color (GgYy). If the offspring are allowed to self pollinate, a 9:3:3:1 ratio will be seen in the next generation. About nine plants will have green pods and yellow seeds, three will have green pods and green seeds, three will have yellow pods and yellow seeds, and one will have a yellow pod and green seeds. This distribution of traits of typical of dihybrid crosses.

**Answer No 5**

**Mitosis** is a process where a single cell divides into two identical daughter cells (cell division).

During mitosis **one** cell? divides once to form **two** identical cells.

The major purpose of mitosis is for growth and to replace worn out cells.

If not corrected in time, mistakes made during mitosis can result in changes in the DNA? that can potentially lead to genetic disorders?.

Mitosis is divided into five phases:

**1. Interphase:**

The DNA in the cell is copied in preparation for cell division, this results in two identical full sets of chromosomes?.

Outside of the nucleus? are two centrosomes, each containing a pair of centrioles, these structures are critical for the process of cell division.

During interphase, microtubules extend from these centrosomes.

**2. Prophase:**

The chromosomes condense into X-shaped structures that can be easily seen under a microscope.

Each chromosome is composed of two sister chromatids, containing identical genetic information.

The chromosomes pair up so that both copies of chromosome 1 are together, both copies of chromosome 2 are together, and so on.

At the end of prophase the membrane around the nucleus in the cell dissolves away releasing the chromosomes.

The mitotic spindle, consisting of the microtubules and other proteins, extends across the cell between the centrioles as they move to opposite poles of the cell.

**3. Metaphase:**

The chromosomes line up neatly end-to-end along the centre (equator) of the cell.

The centrioles are now at opposite poles of the cell with the mitotic spindle fibres extending from them.

The mitotic spindle fibres attach to each of the sister chromatids.

**4. Anaphase:**

The sister chromatids are then pulled apart by the mitotic spindle which pulls one chromatid to one pole and the other chromatid to the opposite pole.

**5. Telophase:**

At each pole of the cell a full set of chromosomes gather together.

A membrane forms around each set of chromosomes to create two new nuclei.

The single cell then pinches in the middle to form two separate daughter cells each containing a full set of chromosomes within a nucleus. This process is known as cytokinesis.

