Bio-informatics assignment-1 (DNA Replication**)**

Q1 Explain the 3 main steps in the process of DNA replication. Name the enzymes that goes with each step:

STEP 1: Initiation

Replication begins at a location on the double helix to which certain initiator proteins bind and trigger unwinding. Enzymes known as **helicases** unwind the double helix by breaking the hydrogen bonds between complementary base pairs, while other proteins keep the single strands from rejoining.

- **Helicases – unwinds DNA**

STEP 2: Elongation

With the primer as the starting point for the leading strand, a new DNA strand grows one base at a time. The existing strand is a template for the new strand. For example, if the next base on the existing strand is an A, the new strand receives a T. The enzyme DNA **polymerase** controls elongation, which can occur only in the leading direction. The lagging strand unwinds in small sections that DNA polymerase replicates in the leading direction.

- **polymerase – creates complementary strand**

STEP 3: Termination

Once both the continuous and discontinuous strands are formed, an enzyme called **exonuclease** removes all RNA primers from the original strands. These primers are then replaced with appropriate bases. Another exonuclease “proofreads” the newly formed DNA to check, remove and replace any errors. Another enzyme called **DNA ligase** joins Okazaki fragments together forming a single unified strand. The ends of the linear DNA present a problem as DNA polymerase can only add nucleotides in the 5’ to 3’ direction. The ends of the parent strands consist of repeated DNA sequences called telomeres. Telomeres act as protective caps at the end of chromosomes to prevent nearby chromosomes from fusing. A special type of DNA polymerase enzyme called **telomerase** catalyzes the synthesis of telomere sequences at the ends of the DNA. Once completed, the parent strand and its complementary DNA strand coils into the familiar double helix shape. In the end, replication produces two DNA molecules, each with one strand from the parent molecule and one new strand.

- **ligase – joins DNA fragments together**

Q2 In which direction are new nucleotides added during replication?

new nucleotides are added in the **5' to 3'** direction during replication

Q3 Below is a single strand of DNA. Below each letter write the complementary strand of DNA

3` A - T – G – C – G – C – G – A – T – T – A – T – C – G – C 5`

| | | | | | | | | | | | | | |

5` T – A – C – G – C – G – C – T – A – A – T – A – G – C – G 3`

Q4 From the double helix that you got in question 4, using 2 different colors, create 2 new strands from the original strand. Label which color represents the strand from original strand and which color represents the daughter strand.

Original strand:

3` A - T – G – C – G – C – G – A – T – T – A – T – C – G – C 5`

| | | | | | | | | | | | | | |

5` T – A – C – G – C – G – C – T – A – A – T – A – G – C – G 3`

New strands:

3` A - T – G – C – G – C – G – A – T – T – A – T – C – G – C 5`

| | | | | | | | | | | | | | |

5` T – A – C – G – C – G – C – T – A – A – T – A – G – C – G 3`

3` A - T – G – C – G – C – G – A – T – T – A – T – C – G – C 5`

| | | | | | | | | | | | | | |

5` T – A – C – G – C – G – C – T – A – A – T – A – G – C – G 3`

blue color represents strand from original strand and green color represents strand from daughter strand

Q5 A) single gene disorder.

Sickle cell anemia (SCA)

B) Write a brief description of the disorder.

Sickle cell anemia (SCA) is the most common and most severe form of sickle cell disease (SCD), a group of inherited blood disorders caused by a genetic mutation. This mutation affects the body's production of hemoglobin, the oxygen-carrying protein in red blood cells.

C) Explain the disorder in terms of gene sequence and how it affects the transcription [to mRNA] or translation [to protein] process.

SCA is caused by mutation in the gene that tells your body to make the iron-rich compound that makes blood red and enables RBC to carry oxygen from your lungs throughout your body. In SCA, the abnormal hemoglobin causes red blood cells to become rigid, sticky and misshapen.

This is an example of substitution type of mutation

● **Transcription :**

Normal Haemoglobin molecule :

DNA Sequence : 3’ - TAC - CAC - GTG - GAC - TGA - GGA – C **T** C - CTC - 5’

Mutated Haemoglobin Molecule :

DNA sequence: 3’ - TAC - CAC - GTG - GAC - TGA - GGA – C **A** C - CTC - 5’

● **Translation :**

Normal Haemoglobin molecule :

mRNA sequence: 5’ - AUG - GUG - CAC - CUG - UCU - CCU – G **A** G - GAG - 3’

Amino acid sequence : start - Val - His - Leu - Ser - Pro - Glu – Glu

Mutated Haemoglobin Molecule :

mRNA sequence: 5’ - AUG - GUG - CAC - CUG - UCU - CCU – G **U** G - GAG - 3’

Amino acid sequence: start - Val - His - Leu - Ser - Pro - Val - Glu.