LS1201 Mid-Semester Examination

Name: Priyanshu Mahato

Roll No.: pm21ms002

Part - 1

Q1. A) Histone acetylation pattern.

Explanation: Epigenetic inheritance goes against the idea that inheritance happens only via DNA code that passes from parent to offspring. This means that a parent's experiences can be passed down to future generations without altering the primary structure of DNA in the form of epigenetic tags.

Since in options (B), (C), (D) we see that the primary structure of DNA is changed, they are clearly not the answers.

Q2. D) Nonsense mutation.

Explanation: A nonsense or a stop mutation, is a change in DNA that causes a protein to terminate its translation earlier than expected. As it is visible in the question, the CAG codon (which is a normal codon) is converted into TAG, which is a stop codon. Due to this, the DNA translation stops early and abruptly, and thus it can be called a nonsense mutation.

Q3. B) 16

Explanation: It is given in the question that the diploid chromosome number is 2n = 16. We know that after meiosis I the homologous chromosomes are separated but each set of homologous chromosomes has undergone 'DNA multiplication' which doubles the quantity of DNA in the cell giving rise to the sister chromatid of each and every chromosome. Thus, total no. of sister chromatids before the M phase is 32, whereas total no. of chromosomes before M phase is 16. Therefore, it doesn't change the number of chromosomes during the S phase of the cell cycle.

Q4. B) Autosomal Dominant

Explanation: *T*rait is not skipping generation and one copy of affected gene is enough to cause the disease. As there is no sexuality influence on the distribution of disease among children it won't be a sex linked.

Q5. A) Akash

Explanation: People with more than 35 CAG repeats are usually more susceptible to Huntington's Disease. In the given question, since only Akash has the number of CAG repeats more than 35 (39), he has the highest probability of developing Huntington's disease.

Q6. B) Frameshift Mutation

Explanation: Frameshift mutations are insertions or deletions in the genome that are not in multiples of three nucleotides. The divisibility by three is important as the cell reads a gene in groups of three bases. Each of these triplet codons corresponds to one of the 20 different types of Amino Acids used to build a protein. In the provided question, there are 23 nucleotides, which is clearly not divisible by three, so, we can be certain that it will give rise to a Frameshift Mutation which will be introduced into the protein.

<u>Part – 2</u>

Q1. As the cell acquires a mutation that disrupts the checkpoint after S-Phase, and we know that after the S-phase there exists a G_2 checkpoint which prevents a cell from entering mitosis if the DNA is damaged. It helps to maintain genomic stability. Thus, if the mutation occurs after S-Phase or in the G_2 checkpoint, then the daughter cells produced from this cell would have a higher probability of getting aneuploid.

The translation will start at the codon highlighted with the lighter shade of blue as it is a start codon which in this case also codes for methionine. The translation will stop at the codon highlighted with the darker shade of blue as it is a stop codon (one of UAA, UAG, UGA) that doesn't code for any amino acid.

Q3. Structural chromosomal abnormalities are those alterations in which, due to different genetic reasons or protein expression (due to a previous genetic mutation in the nucleotide sequence of a given gene), the structure of a chromosome is damaged. The integrity of the chromosome is lost and, depending on which and how many genes are involved, the consequences may be more or less serious.

- **Chromosomal deletions:** Chromosomal deletions are abnormalities in which a more or less large part of a chromosome is lost.
- ♣ Chromosome duplications: Chromosomal duplications are abnormalities in which a segment of a chromosome is repeated. Instead of having two copies of the same genetic segment, the person has three.
- ♣ Chromosomal inversions: Chromosomal inversions are abnormalities consisting of a change in direction of a genetic segment within a chromosome. The chromosome "breaks" in two places and the resulting DNA segment is reinserted but in the reverse direction, altering the way genes are transcribed into proteins.
- **Chromosomal Substitution:** A portion of the chromosome breaks and attaches to another chromosome.
- ♣ Balanced chromosomal translocations: Balanced chromosomal translocations are abnormalities that consist of a genetic segment of a chromosome moves and inserts into another chromosome without there being a loss or gain of total DNA. In the end, the genetic functionality is maintained, the genes are simply on another chromosome.
- ♣ Unbalanced chromosomal translocations: Unbalanced chromosomal translocations are anomalies that consist, again, in that a genetic segment of a chromosome moves and is inserted into another chromosome, although in this case, there is an alteration (due to loss or gain of DNA) in the said segment. Therefore, genetic functionality is in jeopardy.
- **← Chromosomal insertions:** Chromosomal insertions are abnormalities that consist of a segment of DNA from a chromosome has been transferred to an unusual position within the same chromosome or another. Again, if there is neither loss nor gain of DNA, the person will not suffer from any syndrome, just as with balanced translocations.

Q4. The central dogma represents the process DNA \rightarrow RNA \rightarrow PROTEINS.

The central dogma of life explains how bio-information is processed in living beings. This tells us how information stored in DNA is transformed into an mRNA and which finally results in the formation of proteins that are actually the worker molecules in a living organism and carry out all the physiological, morphological, anatomical processes. This explains how DNA, RNA, and proteins interact and function to carry out everything we are in this universe.

Q5.Trait: brown, allele: B
Trait: blue, allele: b
BROWN: Bb, BB

BLUE: bb

a) Since we have all progeny of F1 generation with the dominant trait, the parents have to be true-breeding (brown:BB, blue:bb)

	В	В
b	Bb	Bb
b	Bb	Bb

All the children have brown eyes(Bb)

b) If we have heterozygous parents for brown eye trait:

	В	b
В	ВВ	Bb
b	Bb	bb

Probability of children being with the blue eye is 1:3 or 25% chance.

c) The probability for the heterozygous brown parents getting a second child with the blue eye is 25% and the probability of getting second child as a boy is 50% the probability for the heterozygous brown parents getting a second boy child with the blue eye is 12.5%.