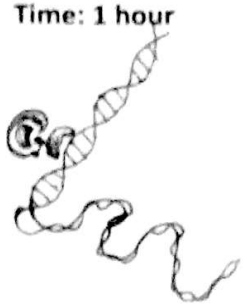
SBL 708 (Epigenetics in human health and disease): Minor I

August 28, 2016
Total: 25 marks
Time: 1 hour

Part I (MCQs / fill in the blanks): (1 x 11 = 11 marks):

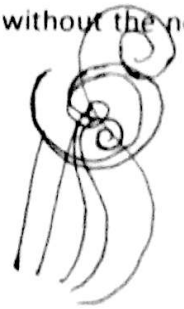


1) Which of the following is TRUE about miRNA?
   a) They inhibit RNA synthesis
   b) They directly inhibit translation
   c) They reduce the amount of RNA available for translation
   d) Their primary site of action is within the nucleus
   e) None of the above

2) Which of the following is TRUE?
   a) Histone acetyltransferases (HATs) are present only in the cytoplasm ✗
   b) HATs are present only in the nucleus ✗
   c) HATs influence transcription by increasing the basic charge on histone tails
   d) HATs are co-localize (are present) at the early replication foci
   e) None of the above

3) Which of the following histone epigenetic marks is NEVER associated with euchromatin?
   a) Histone acetylation
   b) Histone crotonylation
   c) Histone phosphorylation
   d) Histone sumoylation
   e) None of the above

4) Which of the following processes in the cell is completed without the need for epigenetically regulated mechanisms?
   a) Nucleotide excision repair
   b) Base excision repair
   c) Double-stranded DNA repair
   d) DNA replication
   e) None of the above



5) Which of the following is NOT an accurate representation of the extent of DNA methylation in embryonic stem cells?
   a) CpG > CpT
   b) CpC > CpA
   c) CpA > CpT
   d) CpG > CpA
   e) None of the above

6) Which of the following statements is TRUE about genomic imprinting?
   a) It occurs in all eukaryotes
   b) Paternally imprinted genes are more important than maternally imprinted genes for normal embryogenesis
   c) The presence of 150 imprinted genes in the human genome implies that 150 imprint control elements are present in the human genome
   d) Imprinted genes are highly conserved among closely related species
   e) None of the above

7) DNMT1 activity may be best documented in the
   a) G1 phase of the cell cycle
   b) G2 phase of the cell cycle
   c) S phase of the cell cycle
   d) M phase of the cell cycle
   e) None of the above

8) Which of the following statements is TRUE about imprinting?
   a) Imprinting-related epigenetic marks are modified immediately after fertilization
   b) Imprinting-related epigenetic marks are modified up to the blastocyst stage
   c) Imprinting-related epigenetic marks are never erased in any of the somatic cells throughout embryogenesis
   d) Parent-specific imprinting takes place before the placenta formation
   e) None of the above

9) In the absence of CAF-1
   a) Histone octamer assembly would be completely disrupted due to the non-availability H3 and H4 in the nucleus
   b) H3 and H4 would be available in the nucleus but octamer assembly will not occur
   c) Histone octamer assembly would be disrupted due to the non-availability H2A and H2B in the nucleus
   d) H2A, H2B, H3 and H4 would be available in the nucleus and octamer assembly will occur
   e) None of the above

10) UV-mediated damage in the cells is repaired by Nucleotide exci_____

11) _____ represents the most accurately copied epigenetic mark during cell division.

Part II: Short notes. Answer ANY TWO: (4+4 = 8 marks)

1) Early vs late replicating foci (4 marks)
2) Inheritance of histone epigenetic marks in cell division (4 marks)
3) Histone phosphorylation (4 marks)

Part III: Case study: (2+2+2 = 6 marks)

Ms. X inherited a faculty copy (mutated) of an imprinted gene (AB) on chromosome 8 from her father and Mr. Y inherited a faculty copy (mutated) of the imprinted gene (AB) on chromosome 8 from his mother. The imprinted gene AB is expressed only from the maternal allele. Expression of mutated protein AB or the complete lack of expression of protein AB is linked to disease.

1) Assuming that the genomes of Ms.X and Mr. Y do not have any other genetic or epigenetic abnormalities can you predict if Ms.X and Mr. Y will be diseased? Why? (2 mark)

2) Ms. X and Mr. Y get married and have 4 children (2 boys – Y1 and Y2 and 2 girls – X1 and X2). How many of these children do you expect NOT to have inherited the faculty gene AB? Why? (2 marks)

3) Now, in a hypothetical situation if Ms. X married Mr. Z (instead of Mr. Y) who does NOT have a faculty gene AB nor any other genetic or epigenetic abnormalities. If they had 4 children and 2 of them have a faculty copy of the gene AB. How many of the children born to Ms. X and Mr. Z will be diseased. Why? (2 marks)

Mom
X Y
AB AB

AA AB BA BB