CHAPTER: 23

CHROMOSOMES AND DNA

Important points & Numerical values:

1. *Chromosomes were observed by German embryologist Walther Fleming in 1882, in salamander larva.*
2. *The term chromosome was proposed by Waldeyer, which literally means colored bodies.*
3. *Diploid: Species having two sets of chromosomes in their somatic cells are called diploid.*
4. *Polyploid: Species having more than two sets of chromosomes are called polyploid. Polyploid may be tetraploid, hexaploid.*
5. *Haploid: The number of chromosomes exactly the half of somatic number of chromosomes. Gametes and spores are usually haploid cells.*
6. *A haploid cell may be monoploid (one set), diploid (two sets), triploid (three sets) and etc.*
7. *Pencillium, a fungus has only one pair of chromosome while some ferns have more than 500 pairs.*
8. *All prokaryotes has single chromosome except V.cholera which has 2 chromosomes.*
9. *Different no. of chromosomes:*
   - *Mosquito = 6*
   - *Drosophila = 8*
   - *Pea = 14*
   - *Onion = 16*
   - *Maze = 20*
   - *Frog = 26*
   - *Honey bee = 32*
   - *Earthworm = 36*
   - *Rat = 40*
   - *Cow = 60*
   - *Donkey = 62*
   - *Mule = 63*
   - *Horse = 64*
   - *Wheat = 42*
   - *Rh monkey = 42*
   - *Tomato = 24*
   - *Female grasshopper = 24*
   - *Male grasshopper = 23*
   - *Radish = 18*
   - *Carrot = 18*
   - *Potato = 48*
   - *Tobacco = 48*
   - *Chimpanzee = 48*
   - *Dog = 78*
   - *Dove = 78*
   - *Hen = 78*
   - *Pigeon = 80*
   - *Sugar cane = 80*

23. H_1 is associated with small segment of DNA (linker DNA) b/w every two nucleosomes.
24. **Nucleosome:** Eight histone proteins forms octamere around which core DNA is found. Core DNA composed of 146 nucleotides.
25. Two nucleosomes are connected by linker DNA which is composed of 54 nucleotides.
26. Diameter of:
    - **DNA** is 2 nm
    - **Histone** is 2 nm
    - **Nucleosome** is 10 nm
    - **Solenoid** (chromatin fiber) is 30 nm
    - **Supper coil** is 200 nm
    - **Chromatid** is 700 nm
27. Chromosome is composed of chromatin of two types i.e euchromatin and heterochromatin.
28. **Euchromatin** mostly composed of coding region of DNA and is mostly used during transcription therefore it is less condense while,
29. **Heterochromatin** is composed of Junk DNA (non-coding) and is not used during transcription therefore it is more condense.
30. The word **gene** was first used by Johnson in 1909.
31. DNA have two types of genes:
    - <u>Structural gene:</u> Which give rise to mRNA and is composed of Exon (coding region) and intron (non-coding region).
    - <u>Regulatory gene:</u> The rate of transcription is controlled by promoter and terminator gene called the regulatory gene.
32. Promoter region is located at 5' end of coding strand of DNA.
33. Terminator region is located at the 3' end of coding strand of DNA.
34. The sequence of nucleotides on mRNA always match with the sequence of nucleotides of gene found on coding strand and for this purpose that mRNA must form in front of non coding strand.
35. Coding strand is also called sense strand and the opposite strand to the coding strand is called non coding strand. Non coding strand is also called non sense or antisense strand or template strand.
36. In eukaryotic genes, the functional sequence in structural region is called **exons** which is interrupted by a non-functional sequence called **introns**.
37. <u>Genes on chromosome no. 11:</u>
    - Insulin

10. Highest number of chromosomes found in a fern ophioglossum reticulatum i.e 1260 chromosomes while smallest number of chromosomes found in mermicia pallasola (ant) i.e sometimes single chromosome or sometimes a pair of chromosome.

11. At the terminal of at least one pair of chromosome has a nob like structure called **satellite**, composed of satellite DNA which contains VNTR (variable number of tendom repeat). These VNTRs are different in different members of the same species. These are used for the identification of a specie in DNA analysis or DNA finger printing or DNA test.

12. Tendom repeats are the consecutive repetition of nucleotides e.g AUGC, AUGC, AUGC. While non-tendom repeat is non-consecutive repetition e.g, AAGAACAACAA.

13. Nucleolar organizer region or secondary constriction gives rise to nucleoli during interphase.

14. The terminal ends of chromosomes are called telomeres which contains telomere DNA formed by telomerase enzyme.

15. Telomeres prevents the two chromosomes to attach with each other from their ends.

16. On the basis of position of centromere, a chromosome may be;
    - Metacentric → Centromere located at the center.
    - Submetacentric → Centromere located slightly away from the center.
    - Acrocentric → Centromere located near the end.
    - Telocentric → centromere located at the end.

17. Chromosome is made up of 40% DNA and 60% protein.

18. Average sized human chromosome has approx. 5 cm long DNA which consist of about 140 million nucleotides.

19. Histone protein is positively charged basic protein due to the presence of basic amino acids lysine and arginine.

20. There are 5 types of histone proteins (H_1, H_2A, H_2B, H_3 and H_4) found in the chromosome.

21. The positive charge of histone protein and negative charge of DNA are strongly connected to each other and forms nucleosome.

22. About every 200 nucleotides of the duplex DNA wrap twice around the core of 8 histones (two of each H_2A, H_2B, H_3 and H_4) thus forming a complex known as nucleosome (10nm thick).

- Sickle cell anemia
- Leukemia
- Albinism

38. **Genes on chromosome no. 19:**
    - H – substance
    - Secretor gene
39. Blood group gene = chromosome no. 9
40. Phenyl ketanuria gene = chromosome no. 12
41. Rh factor gene = chromosome no. 1
42. **Semi-conservative model:** In this model, the parent strands are partially conserved in both daughter DNA molecules.
43. **Conservative model:** In this model, the parental DNA is fully conserved in the next generation.
44. **Dispersive model:** In this model, the daughter DNA molecules would be a mixture of old and new fragments.
45. The most acceptable model is semi-conservative model of DNA replication, experimentally proved by Meselson-Stahl experiment in 1906.
46. **Origin of replication:** A specific sequence of nucleotides along the length of DNA from where the process of replication begins.
47. In eukaryotic DNA, there is more than one origin of replication sites while in prokaryotic DNA, there is only one origin of replication.
48. DNA gyrase (or topoisomerase) opens the turns of DNA duplex so the DNA is converted from spiral ladder like form to straight ladder like form.
49. DNA helicase breaks the hydrogen bonding so the two strands becomes separated from each other.
50. Single strand binding proteins (SSB) prevents the single strands of DNA molecule to pair up again.
51. Hydrogen bond is always formed spontaneously, do not requires an enzyme.
52. Hydrogen bond is broken by,
    - Invivo (during DNA replication) → helicase enzyme
    - Invitro (in PCR) → Heat
53. Function of different DNA polymerases:
    - DNA Pol-I: Its functions are exonuclease activity + polymerization + termination. It replaces the RNA primer by DNA nucleotide.
    - DNA Pol-II: Its function is the repairing of DNA.

- *DNA Pol-III: It performs the real polymerization, adds nucleotides at the 3' end of primer so the direction of replication becomes 5' to 3'. It also perform proof reading from 3' to 5' direction.*

54. *DNA polymerase-III requires a primer for its polymerization process while RNA polymerase donot require any primer.*
55. *Primer is a short oligonucleotide strand of 15-20 RNA nucleotide which acts as a start site for the activity of DNA polymerase-III*
56. *DNA polymerase, RNA polymerase and ribosomes always moves from 5' to 3' end.*
57. *Transcription is the RNA formation and enzyme RNA polymerase adds RNA nucleotide which are rNTPs. RNA polymerase does not require any primer to initiate polymerization.*
58. *Promotor, a regulatory region of the gene provides a binding sites for RNA polymerase.*
59. *The two common promotor in prokaryotes are TATAAT (–10) sequence & TTGACA (–35).*
60. *In eukaryotes TATA (TATA Box) also called – 25 sequence and CAAT (CAAT Box) or –70 sequence.*
61. *Names of these sequences (-10, -35, -25, -70) refers the position of these sequences before the initiation site of structural region of the gene.*
62. *RNA polymerase consists of 4 subunits;*
    - *beta*
    - *beta'*
    - *sigma*

    } *core enzyme → require for polymerase activity.*

    - *sigma factor → required for RNA polymerase to bind to the promotor.*

53. *In prokaryotes only one type of RNA polymerase is formed, which synthesize all the three RNAs i.e rRNA, mRNA and tRNA.*
54. *In eukaryotes there are 3 types of RNA polymerase namely,*
    - *RNA polymerase-1 which synthesize rRNA*
    - *RNA polymerase-2 which synthesize mRNA*
    - *RNA polymerase-3 which synthesize tRNA.*
55. *One of the two strands of gene in transcription process is sense strand or coding strand and the other strand to which mRNA is complementary is antisense strand or non-coding strand or template.*
56. *Post transcriptional modification is a process in which a primary mRNA is converted into mature and functional RNA.*

57. This process only occurs in eukaryotic cells and absent in prokaryotic cells because of the absence of definite nucleus and introne.

58. Post transcriptional modification consists of two events,
    - Addition of cap and tail to protect mRNA from degradation by enzymes like phosphatases and nucleases.
    - RNA splicing to remove non-protein coding sequence.

59. **RNA splicing:** The removing of introns and reattachment of exons is called RNA splicing. RNA splicing is performed by splicosome (a complex of RNA + protein). RNA in splicosome acts as a ribozyme.

60. Ribozyme is RNA which performs enzymatic function.

61. A cap is in the form of 7-methyl GTP, which is linked from its 5' end to the 5' end of mRNA.

62. A small chain of 30 to 500 adenine nucleotide called poly-A tail which is attached to the 3' end of the mRNA.

63. **Genetic code** is a system of codon (a triplet of nucleotide) consisting of only four types of nucleotides Adenine (A), Guanine (G), Cytosine (C) and Thymine (T).

64. There are 64 codons possible from an alphabet of 4 (letters or) nucleotides.

65. Codon consists of;
    - Coding codon → 61
    - Non-coding codon (stop codon) → 3 (UGA, UAG, UAA)

66. Some amino acids are coded by one codon i.e, methionine and tryptophan.

67. Amino acid leucine, serine and arginine are encoded by six codons.

68. Genetic code show redundancy but no ambiguity.
    - **Redundancy:** when one amino acid is coded by more than one codon. Eg: valine is coded by four codon.
    - **Ambiguity:** when one codon codes for more than one amino acid and there is no such codon which codes for more than one amino acid.

69. Genetic code is universal except mitochondrial DNA;

Codon	Nuclear DNA	Mitochondrial DNA
UGA	Stop codon	Tryptophan
AUA	Iso leucine	Methionine
AGA & AGG	Arginine	Stop codon

70. *In translation, the mRNA produced by transcription is decoded by ribosome to produce specific amino acid that will later fold into active protein.*

71. *Activation of amino acids refers to the binding of free amino acids dispersed in cytoplasm to the 3' end of particular tRNA molecules.*

72. *Amino acid always attach with 3' end of tRNA with the help of enzyme aminocyl tRNA synthetase.*

73. *First tRNA always attach with P-site and all other tRNA attach with A-site of ribosome.*

74. *Ribosomal subunits slightly move along mRNA from 5 to 3' direction so that a new codon is exposed to A site.*

75. *A peptide bond is formed between successive amino acid by an enzyme called peptidyl transferase or ribozyme.*

76. *Peptidyl transferase or ribozyme is present in large subunit of ribosome of both prokaryotes and eukaryotes.*

77. *All enzymes are proteinecious except ribozyme, which is RNA in nature and performs the enzymatic activity i.e peptide bond formation.*
Note: Peptide bond is broken by pepsin but formed by ribozyme.

78. ***Operon:** when a cluster of genes are controlled by one promotor and one terminator is called operon.*

79. ***Positive gene regulation:** When the expression of gene is quantitatively increased e.g lac operon.*

80. ***Negative gene regulation:** When the expression of gene is quantitatively decreased e.g trp operon*

81. *In lac operon:*
    - *Z-gene → beta galactosidase (convert lactose into glucose + galactose)*
    - *Y-gene → permease (have role in the transport of glucose across the bacterial membrane)*
    - *A-gene → trans acetylase (have NO role in the lactose metabolism)*

82. *In lac operon, repressor has role in negative gene regulation while lactose stops the repressor and acts as inducer.*

83. ***Mutation:** permanent change in DNA is called mutation. Any agent which causes mutation is called **mutagens.***

84. *1st discovered mutagen is mustard gas.*

85. ***Chromosomal aberration:** When change occurs in no. of chromosomes (Aneuploidy) or complete set of chromosome (Euploidy) is called chromosomal aberration.*

86. **Aneuploidy:** *when change occurs in number of chromosome*
e.g;
- *Monosomy (2n – 1) → Turner's syndrome*
- *Nullisomy (2n – 2)*
- *Trisomy (2n + 1) → Down's syndrome and klinfelter's syndrome.*
- *Tetrasomy (2n + 2)*

87. **Euploidy:** *when change occurs in complete set of chromosome. Eg: hexaploid (wheat=6n).*

88. *Sickle cell anemia also called Drepanocytosis is an autosomal recessive genetic disorder in which the 6 number amino acid of both beta-chains of hemoglobin is glutamate which is replaced by an abnormal amino acid valine.*

89. *Hb – A → adult Hb (normal Hb)*
Hb – S → sickle cell Hb (abnormal)
Hb – F → foetal Hb (this Hb found only in embryonic condition)

90. *PKU (phenyl ketanuria) is genetically transferred as well as enzyme deficiency disease.*

91. *PKU occurs due to the deficiency of enzyme called phenylalanine hydroxylase which converts phenylalanine into tyrosine. Tyrosine is converted into melanin by tyrosinase enzyme. (Both phenylalanine and tyrosine are aromatic amino acids).*

92. *Down syndrome is autosomal non-disjunction and occurs in chromosome number-21 and person have an extra 21 number chromosome.*

93. *Down's syndrome was 1st identified by Dr. Down in 1866 but its genetic base was studied by Dr. Jerome Lejeune in 1959.*

94. *Incidence of Down syndrome is estimated 1 per 800 births before the age of 30.*

95. *Women have risk of Down syndrome upto about 1 in 350 by age 35, by 40 the risk rises to about 1 in 100.*

96. *Average IQ of children with Down syndrome is around 50 compared to normal children with an IQ of 100.*

97. *In klinefilter's syndrome, males have an extra X-chromosome (XXY).*

98. *Klinefelter's syndrome affects 1 in 500 to 1000 males.*

99. *Most variants of Klinefelter's syndrome are much rare occurring in 1 in 50,000 or fewer male births.*

100. *Turner's syndrome is chromosomal disorder which is characterized by the missing of one X chromosome (44+X).*