

Learning objectives in Genetics
II Semester, Module N3 „Cell and Tissue“

1. Molecular basis of inheritance and variability
2. Nucleic acids
3. DNA and RNA structure
4. DNA replication and its alternative models
5. Origins of replication and replication fork
6. Leading and lagging strands
7. Okazaki fragments
8. DNA replication enzymes
9. The human genome
10. Genome structure: nucleosome, chromatin, chromatid, chromosome
11. Histone and non-histone proteins
12. Heterochromatin and euchromatin
13. Nuclear and mitochondrial genomes
14. Extragenic DNA
15. Long and short interspersed nuclear elements
16. LINE and Alu sequences and their medical relevance
17. Satellite DNA
18. Gene organization and structure
19. Exons
20. Introns
21. Enhancers
22. Silencers
23. Insulators
24. Locus control regions
25. Protein-coding and regulatory genes
26. Genome expression
27. Transcription initiation, elongation, termination
28. RNA processing: capping; splicing; polyadenylation
29. Alternative splicing
30. RNA editing
31. micro-RNA
32. Genome expression: translation and genetic code
33. tRNA and aminoacyl t-RNA synthetize
34. Stages of translation: initiation, elongation, termination
35. tRNA
36. Posttranslational modifications and diversity of proteins
37. Regulation of gene expression
38. DNA methylation
39. Histone modification
40. Histone acetylation, methylation, phosphorylation

41. Chromatin remodeling
42. Changes in genome activity
43. Immunoglobulins and T-cell diversity
44. X inactivation
45. Imprinting
46. The variability of gene expression and its association with medicine.
47. Human genetic diversity. The concept of genetic polymorphism and mutation
48. Common genetic variation in humans; single nucleotide polymorphisms (SNP); insertion-deletion, microsatellite, mobile element insertion and inversion polymorphisms; copy number variants (CNVs)
49. ABO system and blood groups and their polymorphisms
50. Rh system
51. Gene mutation
52. Classification of gene mutations
53. Dynamic mutations
54. Variations in individual genomes
55. Effect of mutation on protein function
56. Replication errors
57. Diseases caused by DNA repair system errors
58. Single-gene inheritance, general overview and key concepts
59. Principles of pedigree construction
60. Its application in autosomal dominant and autosomal recessive disorders
61. Non-Mendelian types of inheritance
62. Pedigree construction and its application in X-linked dominant and recessive traits and Y-linked traits
63. Pseudoautosomal inheritance
64. Mosaicism; segmental and germline mosaicism
65. Genomic imprinting
66. main characteristics of mitochondrial inheritance
67. Unstable repeat expansions

Example tests:

During DNA replication what is the first process to occur?

- A. Sealing of the nicks between short DNA sections;
- B. Synthesis of the lagging strand;
- C. Unwinding of parental DNA;
- D. Synthesis of the leading strand.

Which of the following is true about histone proteins:

- A. They are positively charged;
- B. They contain methionine and valine;
- C. They bind to the base pairs of DNA;

D. There are about 20 histone types associated with DNA.

In which of the following regions of the gene does CGG expansion take place in Fragile X syndrome?

- A. Translated region;
- B. Noncoding region;
- C. Intron;
- D. Terminator.

An individual's ABO blood type is normally determined by:

- A. Genetic inheritance and environmental influences during life;
- B. Environmental influences alone;
- C. The inheritance of 1 of 3 possible alleles (A, B, or O) from each parent;
- D. Whether mother has been injected by RhIG during pregnancy or not

In familial hypercholesterolemia, individuals homozygous for the allele causing the disorder completely lack receptors on liver cells that take up cholesterol from the blood stream. Heterozygotes have one-half the number of receptors while individuals homozygous for the normal allele are phenotypically normal. This is an example of:

- A. Codominance;
- B. Incomplete dominance;
- C. Epistasis;
- D. Complete dominance.