Learning objectives in Genetics
II Semester, Module N4 „Early Embryonic Development“

1. Cytogenetics, techniques for studying human chromosomes
2. Cytogenetic method of investigation in human genetics
3. Chromosome behavior during various stages of cell cycle and meiosis
4. Types of chromosomes
5. Molecular cytogenetics
6. Clinical indications for chromosome and genome analysis and spectrum of resolution
7. Karyotype
8. FISH - fluorescence in situ hybridization
9. Genome analysis using microarrays
10. Whole exome sequencing and whole genome sequencing
11. Cytogenetics. Chromosome abnormalities
12. Gene dosage, balance and imbalance
13. Numerical abnormalities of chromosomes - triploidy and tetraploidy, aneuploidy
14. Structural abnormalities of chromosomes
15. Unbalanced rearrangements (deletions and duplications, marker and ring chromosomes, isochromosome, dicentric chromosome)
16. Balanced rearrangements (reciprocal and Robertsonian translocations, insertions) rearrangements
17. Mosaicism
18. Frequency of chromosomal abnormalities
19. Spontaneous abortions
20. Mechanism of chromosomal abnormalities
21. Abnormalities of autosomes
22. Down syndrome; Chromosomes in Down syndrome
23. Trisomy 18 (Edwards syndrome)
24. Trisomy 13 (Patau syndrome)
25. Genomic disorders;
26. Autosomal deletion and duplication syndromes
27. Cri-du-chat syndrome
28. Diseases associated with genomic imprinting; Prader-Willi and Angelman syndromes
29. Abnormalities of sex chromosomes
30. Klinefelter syndrome - 47, XXY
31. 47, XYY syndrome
32. 47, XXX syndrome
33. Turner syndrome - 45, X
34. Disorders of gonadal and sex development
35. Gonadal dysgenesis
36. Development of ovaries and its maintenance
37. The chromosomal basis of sex determination
38. Y chromosome, X chromosome, cytogenetic abnormalities of sex chromosomes
39. Disorders of sex development affecting phenotypic sex

40. Neurodevelopmental disorders and intellectual disability
41. Developmental genetics and congenital defects
42. Dysmorphology: malformations, deformations and disruptions
43. Genetic, genomic and environmental causes of birth defects
44. Pleitropy: syndrome and sequence
45. Main concepts of human embryological development
46. Genes and environment in development
47. Developmental genetics and birth defects
48. Stem cells
49. Metabolic pathway of the cell
50. Pattern formation and *HOX* gene system
51. Cellular and molecular mechanisms of development
52. Interaction of developmental pathways during embryogenesis

Example tests:

All of the following types of cells are said to be arrested in G_0 phase **EXCEPT**:

A. Red blood cells;
B. Nerve cells;
C. Skin epithelial cells;
D. Heart muscle cells.

Newborn was admitted to the neonatal intensive care unit shortly after the birth due to multiple congenital abnormalities: cleft lip and palate, polydactyly (extra fingers on hands and toes), congenital heart defect and cutis aplasia (absent skin on the skull). Doctors suspected Patau syndrome which is caused by extra chromosome 13. Which cellular mechanism causes Patau syndrome?

A. Defect in the cell cycle checkpoints;
B. Meiotic nondisjunction;
C. Defective methylation of chromosome 13;
D. Mitotic nondisjunction.

Diagnosis of DiGeorge syndrome usually requires which of the following methods?

A. Chromosomal microarray
B. Whole exome sequencing
C. Karyotype
D. Whole genome sequencing

In sperm, most meiotic recombination between X and Y chromosome occurs in the pseudoautosomal region. Rarely, aberrant recombination take place outside this region, translocation the SRY gene from the Y chromosome onto the X. If such a translocation

event takes place and the resulting sperm fertilizes a normally developed oocyte, which combination of karyotype and phenotype is most likely to be observed?

A. XX individuals with ambiguous genitalia
B. XY individuals with ambiguous genitalia
C. XX individuals who are phenotypically male, but infertile
D. XY individuals who are phenotypically male, but infertile

Most people have two major linear creases across the palm of their hands (take a look). Two percent of people in the general population have a single transverse palmar crease (a so-called “simian crease”). This finding is very frequent in patients with Down syndrome. The single transverse palmar crease would be an example of:

A. Association.
B. Sequence.
C. Syndrome.
D. Malformation.