English Division I year
Medical Biology 2014/2015
Part II: Medical Genetics

Topic 8. Medical Genetics I - Dominant/recessive inheritance in humans. Blood groups in man

1. Structure of DNA and chromosomes.
2. Mitosis and meiosis, crossing over.
3. Mendelian inheritance.
4. Penetration, expression of genes.
5. Selected diseases inherited in the monogenic pattern in humans:
    a. hereditary spherocytosis -slides
    b. sickle cell anaemia
    - description of the conditions, differences in the blood smear of patients (differences between the appearance of red blood cells), diagnostics and treatment
    c. phenylketonuria
    - urinary test – experiment, report
    d. alkaptonuria
    - method of detection – experiment, report
    e. albinism - slides
6. Detection of antigens of ABO system:
    - in saliva – experiment, report
    - on surface of human erythrocytes:
        - with horse serum – experiment, report
        - with fitoagglutinin from *Dolichos biflorus* - experiment, report
7. Genetic tasks

Knowledge required before attending laboratory:

- the structure and the role of nucleic acids and chromosomes
- genotype, phenotype
- transmission of hereditary information
- mitosis, meiosis and crossing over, cell cycle
- The Mendel Laws – Mendelian genetics and inheritance
- genetic crosses
- basic of inheritance of traits and genetic disorders
- basic terms: antigen, antibody, agglutination, universal donor and recipient, lectins
- characteristic of AB0 and Rh systems (inheritance, structure of A and B antigens)

Recommended literature:

1. Syllabus
2. Campbell, J.B. Reece: Biology. Pearson, Benjamin Cummings, Seventh Edition 2005
3. Jorde L.B.; Carey J.C.; Bamshad M.J.; White R.L.: Medical Genetics, Third Edition 2006

Topic 9. Medical Genetics II – Pedigree analyses. Sex as a genetic trait

1. Pedigree analyses
   - pedigrees of selected features in humans
   - determination of genotypes
2. Sex of *Drosophilla sp.*
   a. female - microscopic slide fixed in ethanol, unstained, magnification 100x - demonstration
   b. male - microscopic slide fixed in ethanol, unstained, magnification 100x - demonstration
3. Sex chromatin in humans
   a. "drumstick" in neutrophilic leukocytes - microscopic slide fixed and stained by Pappenheim method, magnification 1000x - execution and description of the method, figure
   b. Barr body - photography – demonstration
   c. "Y body" in lymphocytes - photography - demonstration
4. Sex - linked traits
   a. Sex linkage - examples of tasks – description
   b. tests for color blindness by *Ishihara charts* - description
5. Genetic tasks

Knowledge required before attending laboratory:

- sex determination
- sex-linked traits
- sex-linked inheritance
- pedigree analysis

Recommended literature:

1. Syllabus
2. Campbell, J.B. Reece: Biology. Pearson, Benjamin Cummings, Seventh Edition 2005
3. Meder S., Windelspecht M.: Human Biology. McGraw-Hill Science/Engineering/Math, Twelfth Edition 2011

Topic 10. Medical Genetics III - Aneuploidy of autosomes and sex chromosomes

1. Chromosomes:
    a. in fruit fly *(Drosophila melanogaster)* larvae salivary glands - microscopic slide fixed in 70% ethanol, magnification 100x - figure
    b. from mouse *(Mus sp.)* bone marrow cells - microscopic slide, Giemsa stained, magnification 1000x-figure
    c. from human lymphocytes culture - microscopic slide, Giemsa stained, magnification 1000x-figure
2. Genome mutations - aneuploidy of sex chromosomes:
    a. monosomy 45,X - Turner syndrome
    - lymphocyte metaphase chromosomes, karyotype - photography
    - phenotypic traits of the syndrome - photography
    - morphogrammе - photography
    b. trisomy 47,XXY - Klinefelter syndrome
    - lymphocyte metaphase chromosomes, karyotype - photography
    - phenotypic traits of the syndrome - photography
    c. other trisomies
    - 47,XXX - karyotype - photography
    - 47,XYY - karyotype - photography
3. Morphogram - making of the own morphogram, report
4. Chromosomal abnormalities
    a. trisomy 21 (Down syndrome)
    - metaphasal chromosomes from lymphocytes, figure - demonstration
    - slides, demonstration
    b. trisomy 18 (Edwards syndrome)
    - methaphasal chromosomes from lymphocytes, figure - demonstration
    - slides, demonstration
    c. trisomy 13 (Patau syndrome)
    - metaphasal chromosomes from lymphocytes, figure - demonstration
    -slides, demonstration
5. Cancer chromosomal abnormalities
    a. He-La cells (Endometrial Uterus Cancer)- microscopic slide fixed in methanol, stained by Giemsa method, magnification 1000x - demonstration
    b. NK/Ly cells (Nemeth-Kellner Lymphoma) - microscopic slide fixed in methanol, stained by Giemsa method, magnification 1000x - demonstration
6. Structural chromosomal abnormalities
    a. "cri du chat" syndrome (5p deletion)
    - metaphasal chromosomes from lymphocytes, figure - demonstration
    - slides, demonstration
8. Genetic tasks

Knowledge required before attending laboratory:

- alterations of chromosome number or structure and genetic disorders they cause
- karyotype
- chromosome inheritance

Recommended literature:

1. Syllabus
2. Campbell, J.B. Reece: Biology. Pearson, Benjamin Cummings, Seventh Edition 2005
3. Meder S., Windelspecht M.: Human Biology. McGraw-Hill Science/Engineering/Math, Twelfth Edition 2011

Topic 11. Medical Genetics IV - Quantitative inheritance in humans

1. Distribution of random events frequency - balls distribution curve in the Galton apparatus – experiment, report
2. Examination of multifactor quantitative property in human population
    a. measurement of the head length and width – experiment, report
    b. calculation of the head length-width index – experiment, report
    c. measurement of the skull capacity by Broca method – experiment, report
    d. own skull capacity measurement by Lee Pearson method – experiment, report
3. Calculation of own body surface area.
4. Calculation of Rohrer index.
5. Genetic tasks

Knowledge required before attending laboratory:

- polygenic quantitative traits
- polygenic inheritance
- multifactor quantitative property in human population

Recommended literature:

1. Syllabus
2. Campbell, J.B. Reece: Biology. Pearson, Benjamin Cummings, Seventh Edition 2005
3. Jorde L.B.; Carey J.C.; Bamshad M.J.; White R.L.: Medical Genetics, Third Edition 2006

Topic 12. Medical Genetics V - Quantitative inheritance in humans. Genetic basis of transplantation.

1. Examination of multifactorial quantitative characteristics in human population
    a. Wechsler's intelligence scale
    b. Self-evaluation of IQ using MENSA test example
2. Human dermatoglyphics in genetics
    a. Types of human fingerprints patterns - diagrams - presentation with description
    b. Performing own dactylogram - experiment, report
    c. Examples of dermatoglyphic disturbances in various genetic disorders - diagrams, presentation with description
3. Genetic aspects of transplantation:
    a. Inheritance of HLA haplotypes – diagram - presentation
    b. Examples of HLA alleles with striking disease associations – table - presentation.
4. Diseases with complex inheritance
    a. Pedigree of a family with diabetes mellitus type l – diagram - presentation with description
5. Genetic tasks

Knowledge required before attending laboratory:

- polygenic traits – intelligence, dermatoglyphic patterns
- role of MHC
- basic of blood, organ and tissue transplantation

Recommended literature:

1. Syllabus
2. Campbell, J.B. Reece: Biology. Pearson, Benjamin Cummings, Seventh Edition 2005

Topic 13. Medical Genetics VI - Genetics of human population: Hardy-Weinberg equilibrium

1. Frequencies of some alleles in human population:
    a. those determining autosomal allele-morphological traits – phenotypes frequency table of students of the 1st course of Medical Faculty of Medical University of Lodz; calculation of gene and genotypic frequencies of one selected trait using Hardy-Weinberg rule
    b. multiple alleles – calculation of AB blood group frequency in South America - task
    c. recessive sex-linked alleles – calculation of frequency of disturbances in color vision in Caucasian and in African women
2. Recessive allele selection in population - task
3. Genetic drift – experiment, report

Knowledge required before attending laboratory:

- Hardy-Weinberg theorem/equilibrium
- preservation of allele frequencies
- condition for Hardy-Weinberg equilibrium
- population genetic and human health
- genetic drift, natural selection, sexual selection, gene flow, the bottleneck effect, the founder effect, gene variation, preservation of genetic variation

Recommended literature:

1. Syllabus
2. Campbell, J.B. Reece: Biology. Pearson, Benjamin Cummings, Seventh Edition 2005

Topic 14. Medical Genetics VII – Theoretical and practical assessment of Medical Genetics