Practical Part

1. Define the terms. Give examples:

- Gene imprinting
- Aneuploidy
- Balanced chromosomal aberration
- Unbalanced chromosomal aberration
- Reproduction disorder
- Anticipation
- HDNB
- Genetic disease
- Cancerogenesis
- Monofactoriale trait
- Monogenic trait
- Multifactorial trait
- Polygenic trait
- Karyotype
- Karyotyping
- Aneuploide clone
- Hereditary coefficient
- Barr body
- F body
- Chromatid
- Chromatin
- Chromosome
- Genomic fingerprinting
- Genetic disequilibrium
- Epistasy
- Heredity
- Expressivity
- Cancerigen
- Mutagen
- Terratogen
- Phenocopy
- Phenotype
- FISH
- Aneuploid gamete
- Gene
- Allele
- Amorphic gene
- Autosomal gene
- Feminization gene
- Hypomorphic gene
- Masculinization gene
- Non-allele gene
- Normomorphic gene
- Somatic gene
- Stabile gene
- Structural gene
- Tumor suppressor gene
- Single copy gene
- X-linked gene
- HOX gene
- Genocopy
- Genofond
- Genome
- Genotype
- Gonosome
- Linkage group
- Haplotype
- Allele heterogeneity
- Nonallele (locus) heterogeneity
- Genetic heterogeneity
- Heterozygote
- Homozygote
- Immunoglobulin
- Lyonisation
- Malformation
- Monosomy
- Chromosomal mosaic
- Chromosomal mutation
- Dynamic mutation
- Generative mutation
- Gene mutation
- Genomic mutation
- Induced mutation
- Lethal mutation
- Point mutation
- Semilethal mutation
- Somatic mutation
- Spontaneous mutation
- Norm of reaction
- Oncogene
- PCR
- Penetration
- Plasmotype
- Pleiotropy
- DNA polymorphism
- Clinical polymorphism
- Chromosomal polymorphism
- Phenotype polymorphism
- Proteinom
- Cellular immune response
- Humoral immune response
- DNA sequencing
- Chromosomal syndrome
- Molecular probe
- TcR
- Barr body test
- F body test
- Trisomy
- Variability
- Zygote

2. Steps and components required for methods of genetic analysis. Indications and limits for methods of genetic analysis:

a. Barr body test;
b. PCR;
c. Sequencing
d. Southern-blot
e. Karyotyping, differentiated painting;
f. FISH.

3. Questions based on lectures:

a. Genetics of development.
b. Genetics of sex development
c. Genetics of immune response
d. Cancer genetics.
e. Prophylaxis of human diseases.

4. Chromosomal syndromes (Down sdr.; Turner sdr.; Klinefelter sdr.; Patau sdr.; Edwards sdr.):

a. Cause of syndrome;
b. Associated karyotypes;
c. Mechanism of production in particular cases;

d. Diagnosis;
e. Prophylaxis.

5. **Monogenic diseases** (PKU, Marfan sdr., Hemophilia A, B , etc....;):
   a. Cause of disease
   b. Consequences of mutations at:
      i. molecular level
      ii. cellular level
      iii. level of organism
   c. Characteristics of mutations responsible for disease;
   d. Identification and explanation of genetic phenomena:
      i. Expressivity
      ii. Penetrance
      iii. Pleiotropy
      iv. Genetic heterogeneity
      v. Anticipation
   e. Genealogic inheritance
      i. Type of inheritance
      ii. Genotypes of affected persons
   f. Genetic diagnosis (methods of testing: required components, steps)
   g. Prophylaxis

6. **Analysis of family tree**:
   a. Type of inheritance
   b. Identification of genotypes
   c. Calculation of risk for disease
   d. Examples of diseases

7. **Normal genetic traits used for family analysis – blood groups etc.** (ABO; Rh, MN, Se/se; G/g; Hp; Xg).

8. Finding of arguments in favor of genetic determination of traits and listing of methods for diagnosis of pathology (Down sdr., Marfan sdr., PKU, hemophilia, etc....).

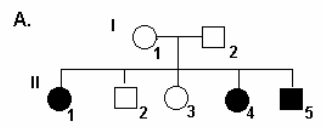
9. Calculation of hereditary and environment coefficients using twin method.

10. Calculation of frequency of genes and genotypes in population using Hardy-Weinberg principle.

11. Mutations and their consequences.

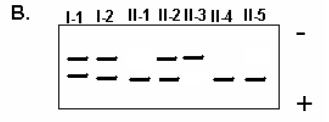
Mutation	Description of genetic changes	Type of mutation (gene, chromosomal, genomic)	Phenotypic consequences	Methods of identification
46,XX,5p-				
C2710T				
46,XY,rob21/21				
45,X/46,XX				
69,XXY				
46,X,r(X)				
A1867G				
1544-1547delAGGT				
46,XY,dup(16)(p11.2:p14.1)				
1125insG				
47,XX,+13				
48,XXXY				
46,X,i(Xq)				
46,XX,t 9/22				
45,XY,rob13/15				
4251delA				
46,XX/47,XX,+18				
46,XY,i(21p)				
$(CAG)_{60}$				
45,XX,rob(13/22)				
46,XX/47,XXY				

12.

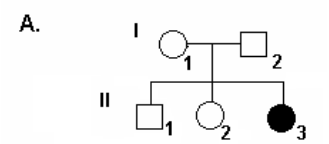


In the tree is shown a family where 3 children are affected by Gaucher disease. Using results of electrophoresis establish:

- Genotypes of all members of the family –

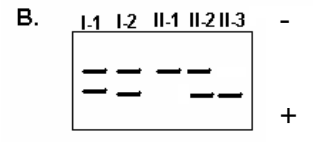


- Type of mutation

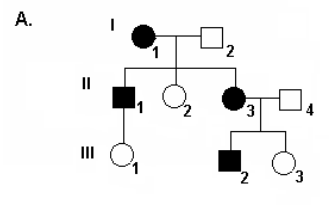


In the tree is shown a family where 1 child is affected by PKU. Using results of electrophoresis establish:

- Genotypes of all members of the family –

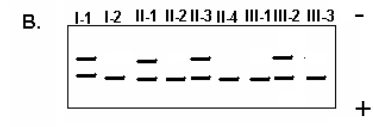


- Type of mutation

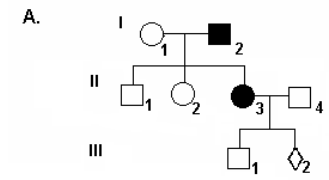


In the tree is shown a family where 4 children are affected by Marfan Syndrome. Using results of electrophoresis establish:

- Genotypes of all members of the family –

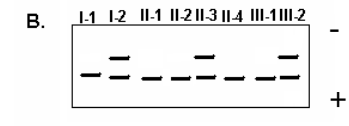


- Type of mutation

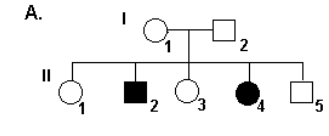


In the tree is shown a family where 2 members are affected by polydactily. Using results of electrophoresis establish:

- Genotypes of all members of the family –

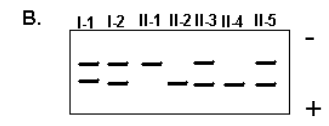


- Type of mutation



In the tree is shown a family where 2 children are affected by cystic fibrosis. Using results of electrophoresis establish:

- Genotypes of all members of the family –



- Type of mutation

13. Levels of expression of structural genes:

Gene	Level of expression		
	Primary	**Secondary**	**Tertiary**
DNA-polymerase			
RNA-polymerase			
Chatalase			
Spectrine			
Rh			
Melanin			
FBN1			
PAH			
Insulin			
ABL			
Ig			
Alcohol-dehydrogenase			
F8			
F9			
α globin			
β globin			
CFTR			
LDLR			
DMD			
HD			
PKD1			
HLA			
ABO			
SRY			

14. **Fill in the table. Show the origin of aneuploidy** (error of mitosis or meiosis)

Karyotype	**Chromosomal error**	**Mechanism of aneuploidy**	**When is produced**	**Phenotype**
45,X				
48,XXXY				
47,XXY				
47,XYY				
45,X/46,XX				
45,X/46,XY				
46,X,i(Xq)				
46,X,i(Yq)				
47,XX,+21				
47,XY,+21				
46,XX/47,XX,+18				
46,XY/47,XY,+8				
47,XX,+13				
47,XY,+21				
46,XX/47,XX,+21				
46,XY/47,XY,+21				

10. Analyze the karyotypes (For Barr-body and F-body tests indicate No and dimensions of bodies):

Karyotypes	Description of karyotype	Error	Phenotype	Test Barr	Test F
45,X					
47,XYY					
48,XXYY					
46,XY,16qh+					
46,XX,1qh-					
46, XY, del(21p)					
46,XY,inv (3)(q12.1q34.3)					
69,XXY					
45,XY,-7					
48,XXXX					
46,XY,dup(5p)					
46,XY, 14q+					
46,XY, 14q-					
46,XY, 14p-					
46,XX,9 ph+					
46,XY,i(21p)					
46,XY,i(21q)					
46,XY,i(8p)					
46,XY,i(8q)					
46,XY,del (14)(q32.2q34.1)					
46,X,r(X)					
46,X,i(Xp)					
46,X,i(Yp)					
46,X,i(Yq)					
47,X,iX(p),Y					
47,XX,i(Yq)					
46,XX/47,XX,+21					
45,X / 46,XX					
46,XY / 47,XXY					