Guide for preparation for 2nd concluding test

1. Define the terms.

	Characteristic	Example
Monogenic trait		
Poligenic trait		
Monofactoriale trait		
Multifactorial trait		
Normal trait		
Pathologic trait		
Dominant trait		
Recessive trait		
Intermediate trait		
Autosomal trait		
Gonosomal trait		
Holandric trait		
Monogenic determination		
Monoallelic determination		
Pleiotropy		
Polygeny		
Penetrance		
Uniparental dissomy		
Allele heterogeneity		
Locus heterogeneity		
Expresivity		
Epistasy		
Genomic imprinting		
Anticipation		
Genocopy		
Phenocopy		
Norm of reaction		
Genomic mutation		
Chromosomal mutation		
Gene mutation		

Induced mutation		
Spontaneous mutation		
Somatic mutation		
Generative mutation		
Lethal mutation		
Semilethal mutation		
Amorphic mutation		
Neomorphic mutation		
Isomorphic mutation		
Hipermorphic mutation		
Samesense mutation		
Missense mutation		
Nonsense mutation		
Sense mutation		
Point mutation		
Frame-shiht mutation		
Dynamic mutation		
Full aneuploidy		
Partial mutation		
Homogeneous aneuploidy		
Aneuploidy in mosaic		
Mutagen factor		
Terratogen factor		
Cancerigen factor		
HDNB		
Hereditary coefficient		

2. Fill in the table

Type of mutations	Genetic modification	Phenotype	Example
Samesense mutation			
Missense mutation			
Nonsense mutation			
Sense mutation			

Transition			
Transversion			
Amorphic mutation			
Hypermorphic mutation			
Neomorphic mutation			
Hipermorphic mutation			
Isomorphic mutation			
Exonic mutation			
Intronic mutation			
Reciprocal translocation			
Ring chromosome			
Inversion			
Robertsonian translocation			
Isochromosome p			
Isochromosome q			
Homogeneous monosomy			
Monosomy in mosaic			
Full mosaic			
Partial monosomy			
Homogenous trisomy			
Trisomy in mosaic			
Full mosaic			
Partial trisomy			
Free trisomy			
Trisomy in translocation			
Triploidy			

3. Identify the type of chromosomal error and their consequences on phenotype.

Karyotype	Type of error			Mechanism of production	Phenotype	Method of diagnostic
	Error	Additional / absent GM	Balanced / unbalanced			
47,XX,+21						
47,XX,+21/46,XX						
46,XY,i(21q)						

46,XX,21q+						
46,XY,rob21/21						
45,XY,rob 21/21						
45,XX,-21						
45,X						
45,X / 46,XX						
45,X / 47,XXX						
46,X,i(Xp)						
46,X,i(Xq)						
46,X,Xp-						
46,X,Xq-						
46,XX,5p-						
47,XXY						
48,XXXY						
49,XXXXY						
46,XY/47,XXY						

4. Identify the origin of aneuploidy:

Mother	Father	Child	Origin of aneuploidy
46,XX (Xg-)	46,XY (Xg+)	47,XXY (Xg-)	
		47,XXY (Xg+)	
		45,X (Xg-)	
46,XX (Xg+)	46,XY (Xg-)	45,X (Xg-)	
		47,XXX (Xg-)	
46,XX (Xg+)	46,XY (Xg-)	47,XXX (Xg+)	
		47,XXY (Xg-)	
		45,X (Xg+)	

5. Calculate the risk of HDNB in all couples. Motivate the answers.

Parents				Family history	Children		Risk for HDNB
Mother		Father			Genotype	Phenotype	
Genotype	Phenotype	Genotype	Phenotype				
	Rh+		Rh-	Father's sister had HDNB			
			Rh-	Older mother's brother had HDNB, bu she hadn't			

			Rh+	Older mother's sister had HDNB, bu she hadn't			
	Rh-		Rh+	Father's sister had HDNB			
	Rh+		Rh-	Mother's father had HDNB			
	HDNB		Rh+	Father is homozygote			
	Rh-			Father had HDNB			
	Rh+			Father - unknown			

6. Explain the following pathologies.

Pathology	Possible causes	Genealogic inheritance	Genetic diagnsotic
Monogenic disease			
Polygenic disease			
Monogenic syndrome			
Polygenic syndrome			
Down sdr.			
Patau sdr.			
Edwards sdr.			
„Cri-du-chat" sdr.			
Turner sdr.			
Klinefelter sdr.			
X-fra (FMR2 sdr.)			
Marfan sdr.			
Angelman sdr.			
Prader-Willi sdr.			
Phenilketonuria (PKU)			
DMD			
Hemophilia A			
Hemophilia B			
Achondrodisplasia			
ADPKD			
Cystic fibrosis			
Diabetes			
Schizophrenia			

7. Find several criteria for classification:
- hereditary traits
- variability
- mutations
- gene mutations
- chromosomal aberrations

8. Exercises with blood groups

9. Chromosomal syndromes (Down; Turner; Klinefelter; Patau; Edwards):
a. The cause of syndrome;
b. Associated phenotypes;
c. Mechanism of production;
d. Genetic diagnostic.
e. Prophylaxis.

10. Monogenic diseases (PKU, Marfan s., Hemophilia A, etc....;):
a. Cause

b. Manifestation:
    i. At molecular level
    ii. At cellular level
    iii. At level of organism
c. Characteristic of mutations;
d. Identification of genetic phenomena:
    iv. Expressivity
    v. Penetration
    vi. Pleiotropy
    vii. Genetic heterogeneity
e. Genealogic inheritance
    viii. Type of inheritance
    ix. Genotype of affected persons
f. Genetic diagnostic
g. Prophylaxis