

## **Program of General Pathology and Physiopathology**

CELLULAR RESPONSES TO STRESS AND TOXIC INSULTS: ADAPTATION, INJURY, AND DEATH

ACUTE INFLAMMATION

CHRONIC INFLAMMATION

TISSUE RENEWAL, REPAIR, AND REGENERATION

NEOPLASIA

INFLAMMATION AND CANCER

HORMONE-DEPENDENT CANCERS

BLOOD VESSELS

ATHEROSCLEROSIS

HEMODYNAMIC DISORDERS

THROMBOEMBOLIC DISEASES

THE HEART

SHOCK

DISEASES OF WHITE BLOOD CELLS, LYMPH NODES, SPLEEN, AND THYMUS

RED BLOOD CELL AND BLEEDING DISORDERS

LIVER AND BILIARY TRACT

THE ENDOCRINE SYSTEM

### **REFERENCE TEXTBOOKS:**

Robbins & Cotran- Pathologic Basis of Disease

**9th Edition** With STUDENT CONSULT Online Access

Robert A. Weinberg- The Biology of Cancer

**2nd Edition**- Garland Science- Taylor & Francis Group

### **ORAL EXAM**

#### **Program of Medical Genetics**

- THE HUMAN GENOME: GENES AND ORGANIZATION
- NEXT GENERATION SEQUENCING (NGS), THE EXOME
- PENETRANCE AND EXPRESSIVITY, ANTICIPATION
- HOMOZYGOSITY AND COMPOUND HETEROZYGOSITY
- HAPLOINSUFFICIENCY, CONGENITAL AND SYNDROMIC DISORDERS
- MECHANISM OF SPLICING AND ITS ALTERATIONS
- CLASSES OF POINT MUTATIONS, TRANSITION AND TRANSVERSION, CONSERVATIVE MISSENSE, NONSENSE, NONSTOP
- INSERTIONS, DELETIONS AND FRAME-SHIFT AND NON-DUPLICATION, GENE CONVERSION
- PATHOLOGICAL SIGNIFICANCE OF THE VARIOUS CLASSES OF DNA VARIATIONS
- INTERNATIONAL NOMENCLATURE OF GENETIC VARIATION AND SIGNIFICANCE OF REPORTING
- GENETIC COUNSELING: REPRODUCTIVE RISK
- PRENATAL DIAGNOSIS AND PRESYMPTOMATIC TESTING
- KARYOTYPE ANALYSIS, THE FISH
- MOLECULAR KARYOTYPING BY ARRAYCGH
- ANEUPLOIDY IN ABORTIONS AND RISK OF RECURRENCE

- TRIPLOIDY, TETRAPLOIDY
- THE X CHROMOSOME INACTIVATION AND PAR
- AUTOSOMAL TRISOMIES
- SEX CHROMOSOME TRISOMIES
- MONOSOMIES, TURNER SYNDROME
- CHROMOSOMAL DELETIONS, PARACENTRIC AND PERICENTRIC INVERSIONS
- BALANCED AND UNBALANCED TRANSLOCATIONS, ROBERTSONIAN, CHROMOSOMAL MARKERS
- SUBMICROSCOPIC DELETIONS AND DUPLICATIONS (WILLIAMS S., DIGEORGE, CRI DU CHAT, SMITH-MAGENIS)
- MONOALLELIC MENDELIAN DISORDERS WITH DE NOVO MUTATIONS (CRANIOSYNOSTOSIS, ACHONDROPLASIA)
- AUTOSOMAL DOMINANT MENDELIAN DISORDERS (NEUROFIBROMATOSIS, MARFAN, POLYCYSTIC KIDNEY DISEASE)
- X-LINKED DISORDERS (DUCHENNE AND BECKER MUSCULAR DYSTROPHY, HEMOPHILIA, X-LINKED MENTAL RETARDATION, RETT SYNDROME)
- AUTOSOMAL RECESSIVE DISORDER (CYSTIC FIBROSIS, ALPHA AND BETA THALASSEMIA, SPINAL MUSCULAR ATROPHY, HEMOCHROMATOSIS, GLYCOGEN STORAGE DISORDERS, LYSOSOMAL STORAGE DISORDERS)
- MENDELIAN DISORDERS WITH GENETIC HETEROGENEITY (LIMB-GIRDLE MUSCULAR DYSTROPHIES, RETINITIS PIGMENTOSA)
- DYNAMIC MUTATIONS IN NON-CODING REGIONS (FRAGILE X, MYOTONIC DYSTROPHY) AND CODING REGIONS (HUNTINGTON'S DISEASE, SPINOCEREBELLAR ATAXIAS)
- MUTATIONS IN CHROMOSOMAL REGIONS WITH IMPRINTING (PRADER-WILLI SYNDROME, ANGELMAN SYNDROME, BECKWITH-WIEDEMANN SYNDROME, SILVER-RUSSELL), UNIPARENTAL DISOMY
- MUTATIONS OF MITOCHONDRIAL DNA (MERFF, MELAS, LHON, KS, LEIGH SYNDROME)
- GENETIC PREDISPOSITION
- MULTIFACTORIAL TRAITS
- GWAS STUDIES
- MICRORNA FUNCTION AND ROLE IN GENETIC DISEASES
- GENERAL PRINCIPLES OF ADVANCED THERAPIES FOR GENETIC DISEASES

#### **REFERENCE TEXTBOOKS:**

- Tom Strachan, Andrew Read. Human Molecular Genetics, 4<sup>th</sup> edition. Garland Science
- Thompson and Thompson Genetics in Medicine. Saunders 7<sup>th</sup> edition

#### **ORAL EXAM**