SYLLABUS FOR HUMAN GENETICS PH.D. ENTRANCE TEST (W.E.F 2026)

Structure and Functions of Cell Organelles

Mitochondria, Ribosome, Golgi Complex, Endoplasmic Reticulum, Peroxisomes and Lysosomes, Nucleus

Cell, Plasma Membrane and Cytoskeleton

Cell: Structure and Organization, Plasma Membrane, Structure of Plasma Membrane with special emphasis on various models, Functions of Plasma Membrane, Transport across membrane, Mechanisms of Endocytosis and Exocytosis, Cytoskeleton, Microfilaments: Structural organization, cell motility and cell shape, Microtubule: Structural and Functional organization, Intermediate filaments

Cell Cycle and Cell Signalling

Cell cycle and its regulation, Cell-Cell Interaction, Cell adhesion molecules, Cellular Junctions, Extracellular matrix, Signal transduction, Intracellular receptor and cell surface receptors, Signalling via G-protein linked receptors (PKA, PKC, CaM kinase), Enzyme linked receptor signaling (Growth factor receptor signaling; JACK-STAT pathway), Network and cross-talk between different signal mechanisms, Programmed cell death (Apoptosis)

Molecular Biology

DNA – Structure, types & functions, DNA Replication in Prokaryotes & Eukaryotes, RNA - Structure, types & functions, Transcription factors & Gene Expression, Post transcripted RNA processing, Translation: Protein formation, Cell -based DNA cloning, Principles of DNA cloning, Vector Systems for cloning different sizes of DNA fragments, Expression cloning, PCR based DNA cloning & DNA Analysis, Principles of PCR, Applications of PCR, Real Time PCR

Human Genome

Basic concepts of Human Genome, Human Gene Families, Homology, Parelogs & Orthologs, Mitochondrial Genome, Repetitive DNA and its types, Transposable elements in Eukaryotes**.**

Human Cytogenetics

Genetic basis of Cancers, Oncogenes & Proto-oncogenes, Tumor Suppressor Genes, Genetics of common Cancers, Human Congenital Anomalities; Neural Tube Defects, Anencephaly, Encephalocele, Hydrencephaly, Spina bifida including myelomeningocele and others, Cleft Lip/Cleft Palate, Uniparental Disomy; Prader-Willi Syndrome, Angelman Syndrome, Beckman Weidworth Syndrome, Human Heredity and Social Welfare; Color, Form and Distribution of hair on the head, Color and Vision of Eye and Vision defects, Shape and size of hands & Feet and their abnormalities, Abnormalities and inheritance patterns of Skin, Muscles & Bones, Eugenics: Definition, History, Positive and Negative Eugenics, Euphenics, Euthenics, Heredity of Twins.

Bioinformatics

Introduction: Historical overview and definition, Applications, Major databases in bioinformatics: Nucleic acid databases, Genome databases, Protein databases, Molecular biology and bioinformatics, Bioinformatics software, Information Search and Data Retrieval: The world wide web, Tools for web search, Data Retrieval tools

Metabolic disorders

Introduction to Carbohydrates Metabolism, Disorders of Carbohydrates Metabolism, Lactose Intolerance, Glycogen Storage Disease (G-6PD), Fructose Intolerance, Diabetes Mellitus, Galactosemia, Introduction to Proteins & Amino acids, Disorders of Amino acids metabolism, Phenylketonuria, Alkaptonuria, Tyrosinemia, Albinism, Metabolic Disorders of Purines and Pyrimidines, Hyperuricemia, Lesch-Nyhan Syndrome, Metabolic disorders of Porphyrin, Acute Intermittent Porphyrin, Erythropoietic Porphyria, Metabolic disorders of Glycosaminoglycans & Glycoproteins, Mucopolysaccharidosis, Mucolipidosis, Introduction to Lipids & Fatty acids & their metabolism, Disorders of Lipid Storage, Tay Sachs Disease, Krabbe Disease, Disorders of Fatty acid Metabolism, Hyperlipidemia, Hypercholesterolemia

Applied Medical Genetics

History and impact of genetics in medicine, Clinical aspects of medical genetics, Animal models for the study of human genetic diseases, Evolving molecular cytogenetic technologies, Comparative genomic hybridization, Spectral hybridization, Human mitochondrial DNA and related diseases

Human Genetic disorders

Principle and strategies in identifying human disease genes, Position independent strategies for identifying disease gene, Positional cloning, Confirming a candidate gene, Ways to identify disease genes, Rules for nomenclature of mutations and databases of mutation, Loss of function mutations, Gain of function mutations, Molecular pathology: from gene to disease, Molecular pathology: from disease to gene, Molecular pathology of chromosomal disorders, Inheritance pattern of genetic diseases; Autosomal disorders, X-linked dominant disorders, X- linked recessive disorders, Colour blindness, Hemoglobin and Hemoglobinopathies, Structure of hemoglobin, Genetic control of hemoglobin synthesis, Developmental control of globin gene, Gene mutation and related abnormalities of hemoglobin, Neuromuscular diseases, Central nervous system orders, Disorders of mental functions, Disorders of bone and connective tissues, Oral and craniofacial disorders, Deafness and renal diseases

Chromosomal abnormalities and genetic testing

Chromosomal abnormalities in human cancers, Chromosomal changes associated with leukemias, Chromosomal changes associated with solid tumors, Chromosomal associated with benign tumors, Autosomal dominant disorders with the predisposition to develop cancer, Association of HPV with human cervical carcinoma, Prenatal testing and reproductive genetics, Pre-implantation of genetic diagnosis, Detection of genetic diseases, Treatment of genetic diseases, Management of genetic diseases, Mechanism of Gene silencing

Genetic Counselling

Genetic counseling: an introduction, Genetic counseling in Mendelian disorders, Genetic counseling in common non- Mendelian disorders, Genetic counselors as educators, Risk assessment as a part of Genetic counseling, Risk communication a complex process, Genetic testing issues, Privacy and Confidentiality, Genetic Counselling registers, Genetic counselling clinics and its working, Objectives and Outcomes of Genetic Counselling, Genetic counseling strategies for working with families, Developmentally based approaches for counseling children and adolescents, Genetic counseling for women with intellectuals disabilities, Actively engaging with patients in decision making.

Clinical genetics

Principles and practice of clinical genetics; History , Nature and frequency of genetic diseases, Molecular and Biochemical basis of genetic diseases, Late onset genetic disorders- Alzheimer, Multifactorial diseases- Atherosclerosis, Diabetes mellitus, Nature and Nurture: Distangling the effects of genes and environment, Cytogenetic techniques in disease detection, Chromosome abnormalities and pregnancy loss, Ring chromosome and related genetic disorders, Chromosomal rearrangements and their impact on human health, Reprogenetics-Germinal Choice Technology