MEDICAL GENETICS COURSE BEMC 2021-2022

Prof. Cristiano Simone

- **Gene, Epigenetic and Epigenomic Aspects of Gene Expression**: Gene Structure, Gene Organizzation, Gene Expression, Chromatin Architecture and Function, Allelic Imbalance, Genomic Imprinting, X Chromosome Inactivation, Non Coding RNA Genes, Gene Expression as the Integration of Genomic And Epigenomic Signals, Variation in Gene Expression and its Relevance to Medicine.

- **Human Genetic Diversity:** Mutation and Polymorphism, Types of Mutations and their Consequences, The Origin and Frequency of Different Types of Mutations, Clinical sequencing studies, The Nature of Genetic Variation, Inherited Variation and Polymorphism in DNA, Variation in Individual Genomes.

- **Genetic Inheritance Patterns:** Genotype and phenotype**,** Dominant and recessive traits in Autosomal Patterns of Mendelian Inheritance, Patterns of Single-Gene Inheritance, X-Linked Inheritance, Mosaicism, Dynamic Mutations: Unstable Repeat Expansions, Maternal Inheritance of Disorders Caused by Mutations in the Mitochondrial Genome.

- **Principles of Clinical Cytogenetics and Genome Analysis:** Chromosome and kariotype, Medical Relevance of Mitosis and Meiosis, Human Gametogenesis and Fertilization, Chromosome Abnormalities and Relative Mechanisms, Disorders associated with Genomic Imprinting, Chromosome and Genome Analysis in Cancer, Aneuploidy, Prenatal Diagnosis and Indications for Prenatal Testing.

- **Risk Assessment and Genetic Counseling:** Family History in Risk Assessment, Genetic Counseling in Clinical Practice**,** Determining Recurrence Risks**,** Empirical Recurrence Risks**,** Molecular and Genome-Based Diagnostics.

- **Hemoglobinopathies:** The Hemoglobins and the Effect of Mutation On Globin Function, α And β-Thalassemias, Hereditary Persistence of Fetal Hemoglobin, Sickle Cell Disease.

- **Neurodegenerative disorders:** Alzheimer Disease (Cerebral Neuronal Dysfunction and Death, MIM 104300), Disorders of Mitochondrial DNA, Leber Hereditary Optic Neuropathy, Disease due to the Expansion of Unstable Repeat Sequences.

- **Hearing Loss:** Infant Deafness, Syndromic Deafness with Autosomal Transmission, Syndromes with X-Linked Transmission, Mitochondrial Hearing Loss Syndromes, Non Syndromic Hearing Loss with Autosomal Transmission, Mitochondrial Non-Syndromic Hearing Loss.

10. Oncogenetics (Part I): Neoplasia, Genetic Basis of Cancer, Familial Occurrence of Cancer, Cancer Gene Discovery in Families**,** Cancer and the Environment.

11. Oncogenetics (Part II): Sporadic Cancer**,** Activation of oncogene**,** Loss of tumor suppressor gene**,** Cytogenetic Changes in Cancer, Gene amplification in cancer, Applying Genomics to Individualize Cancer Therapy, Application of gene signatures, Gene expression profiling in cancer prognosis, Targeted cancer therapy, Cancer pathway, Cancer therapy and heterogeneity leading to the drug resistance.

12. Treatment of Genetic Diseases: Treatment by the Manipulation of Metabolism, Treatment to Increase the Function of the Affected Gene or Protein, Modulation of gene expression, Modification of the somatic genome transplantation, Precision medicine: the Present and Future of the Treatment of Mendelian Disease, The Current State of Treatment of Genetic Disease, Special Considerations in Treating Genetic Disease, Gene Therapy**.**