HMX Fundamentals Genetics

Most conditions have some genetic basis, and a scientific revolution enabled by genomic technologies is changing the way that many diseases are diagnosed and treated as well as providing insight into health, risk factors, and disease prevention. A solid understanding of genetics has never been more important for the practice of medicine. In this course, participants will:

- Understand the fundamental concepts of genetics, including gene structure and genetic variation
- Learn about inheritance of disease, population-specific risk, and genetic testing
- See how increased knowledge of the human genome is applied in clinical settings, including through precision treatment of cancer and other diseases

Topics Covered

Course Overview
- Course introduction
- Meet the faculty
- Introduction to the human genome

The Central Dogma and Genetic Variation
- The central dogma and the relationship between genotype and phenotype
- Structure of a human gene and the effects of genetic variation

Mendelian Inheritance of Disease
- Meiotic segregation
- Modes of inheritance
- Pedigree analysis
- Penetrance and expressivity

Identifying Mendelian Disease Genes
- Haplotypes and linkage studies
- Determining causation of a variant
- Targeted genetic testing

Chromosomal Aberrations
- DNA segregation machinery
- Whole chromosome and structural aneuploidy
- Diagnostic techniques for chromosomal disorders

The Genetics of Cancer
- Germline and somatic mutations
- Tumor suppressors and oncogenes
- Two hit hypothesis
- Precision cancer treatments

Common Complex Traits
- Architecture of a complex trait
- Genome-wide association studies
- Heritability and missing heritability
- Understanding risk in common complex traits

Human Population Genetics
- Emergence and history of human traits
- Evolutionary forces and population dynamics
- Ancestry testing and population-specific risk

Beyond the Genome Sequence
- Mitochondrial inheritance
- Unstable repeats
- Epigenetic inheritance and imprinting
- Gene dosage and X-inactivation

Genetics and Precision Medicine
- Whole genome sequencing
- Pharmacogenomics
- Genome editing

Each HMX course is designed to give learners a solid foundation in the basic science principles that are relevant to human health and disease. Concepts are taught using whiteboard-style videos and animations and reinforced by interactive elements, true-to-life scenarios, and real patient cases to enhance learning.