JBL302: Human Disease Genetics

Organization of the human nuclear genome: general organization, protein-coding genes, RNA genes, repetitive DNA, mitochondrial genome, human genetic variability, including chromosome abnormalities and their consequences.

Genes in pedigrees and populations: monogenic versus polygenic threshold theories, monogenic patterns and their complications, rare genetic disorders, pathogenic variants and genotype-phenotype correlations, emerging paradigms in personalized medicine.

Mammalian development- a genetic perspective, cell specialization, pattern formation, morphogenesis, early human development, neural development, and conserved developmental pathways.

Model organisms, human pluripotent stem cells to study genetic disorders, organoid and assembloid technologies to investigate neurological disorders, gene editing, and genetic approaches to treating diseases.

Genome-wide association studies: fundamental concepts, why and how are these studies conducted, population stratification, interpretations of results, causality of the signals identified, family studies of complex diseases, literature readings on complex neurological disorders.

Suggested reading:

An Introduction to Genetic Analysis by Griffiths et. al. (WH Freeman and Company)
Human Molecular Genetics by Tom Strachan and Andrew Read (Garland Science)
Selected research and review articles will be provided.