Basic Training in Reproductive Medical Genetics

Foundations in Genomics

Week 1 – Principles of Genetics

Foundational principles of molecular genetics, embryology, inheritance, genetic disease mechanisms, mosaicism, and introductory cytogenetics relevant to reproductive medicine.

Key Topics Covered:

- DNA structure and function, transcription, translation, and mRNA processing
- Genetic variants and molecular mechanisms of disease
- Foundations of embryology and early human development
- Mendelian inheritance patterns and phenotypic variability
- Mitochondrial inheritance and mitochondrial disease
- Germline and somatic mosaicism and their clinical implications
- Trinucleotide repeat disorders and anticipation
- Complex inheritance and multifactorial disease
- Mitotic errors in reproduction and genetic risks associated with advanced paternal age
- History and principles of cytogenetics, chromosome banding, and karyotyping
- Molecular cytogenetics and cytogenetic nomenclature

Workshops & Cases:

Genetics terminology and patient education workshops, pedigree construction and risk analysis, cytogenetics laboratory tour, and case-based learning focused on autosomal dominant, X-linked, and trinucleotide repeat inheritance. Supplemental activities reinforce cell cycle, meiosis, embryogenesis, DNA extraction, transcription, translation, and the history of cytogenetics.

Week 2 – Chromosomal Abnormalities & Laboratory Genetics

Chromosomal abnormalities, cytogenomic testing, dysmorphology, prenatal phenotype assessment, and laboratory methods for identifying genomic variants.

Key Topics Covered:

- Constitutional chromosomal abnormalities and their clinical consequences
- Autosomal and sex chromosome aneuploidies
- Structural chromosomal rearrangements
- Origins and clinical implications of chromosomal mosaicism
- Cytogenomic microarray applications in prenatal diagnosis
- Microdeletion and microduplication syndromes
- Dysmorphology and recognition of prenatal and postnatal phenotypes
- First trimester genetic sonography
- Interpretation of common karyotype and microarray findings
- Clinical utility and limitations of karyotype and microarray testing
- Products of conception (POC) testing and interpretation of common findings

• Introduction to molecular laboratory methods and detection of single nucleotide variants
• Genotyping, sequencing, and next-generation sequencing technologies

Workshops & Cases:
Chromosomal disease case series, cytogenomic and microarray clinical application workshops, 22q11.2 deletion syndrome phenotype evaluation, and interactive interpretation of PGT, FISH, karyotype, and microarray results. Case-based learning includes chromosomal rearrangements, abnormal microarray findings, and X-linked adrenoleukodystrophy (XALD).

Week 3 – Prenatal Screening & Diagnostic Testing

Prenatal screening and diagnosis, cell-free DNA technologies, carrier screening, newborn screening, invasive testing, and clinical interpretation of mosaic and atypical results.

Key Topics Covered:
• Maternal serum screening for fetal chromosomal abnormalities
• Basic principles and clinical application of cell-free DNA screening for common aneuploidies
• Expanded NIPT and screening for additional chromosomal conditions
• Noninvasive prenatal diagnosis for monogenic disorders
• Prenatal screening considerations in twin pregnancies
• Integration of cfDNA screening from laboratory testing to clinical practice
• Carrier screening in the genomic era and implications for reproductive counseling
• Newborn screening and early identification of actionable genetic conditions
• Amniocentesis and chorionic villus sampling (CVS)
• Invasive prenatal diagnosis in twin pregnancies
• Clinical implications of mosaicism in prenatal diagnostic testing
• Counseling and management of unusual, atypical, or discordant prenatal screening results

Workshops & Cases:
NIPT interpretation workshops, carrier screening counseling, delivery of atypical genetic results, and diagnostic prenatal testing sessions. Case-based learning addresses discordant fetal sex, single-gene NIPT findings, spinal muscular atrophy (SMA), and cystic fibrosis, with additional discussion of mosaicism and unusual cfDNA results.

Week 4 – Adult & Complex Disease

Ethical, legal, and social considerations in reproductive genetics alongside adult-onset disease, cancer genetics, cardiovascular genetics, neurogenetics, genomic privacy, and emerging precision medicine.

Key Topics Covered:
• Ethical and social issues in prenatal genetic testing
• Legal considerations surrounding prenatal genetic testing
• Genetic privacy and implications of prenatal genomic screening
• Direct-to-consumer genetic testing and patient counseling considerations

• Industry and academic collaboration in genomic innovation
• Precision medicine and treatable fetal findings
• Genetic basis of cancer and hereditary cancer syndromes
• Cancer genetics in women's health
• Cryopreservation and indications for genetics referral
• Neurogenetics and adult-onset disorders including Alzheimer disease, Huntington disease, and Parkinson disease
• Cardiovascular genetic disease across the lifespan
• Autism, intellectual disability, and mental health conditions in family pedigrees
• Recognition of common genetic conditions and inheritance patterns through pedigree analysis

Workshops & Cases:
Precision care and treatable fetal findings workshops, cancer genetics pedigree counseling, neurodevelopmental and mental health pedigree analysis, and evaluation of common conditions in family histories. Case-based learning focuses on ethical and legal issues, direct-to-consumer testing, hereditary cancer, neurogenetics, and cardiomyopathy.