ABMGG TRAINEE CORE LECTURES 2018-2020 Wednesday 11:00 am – 12:00 pm Room DD2205 MCN

Date	Speaker	Торіс
07/04/2018	Hamid	Genetic Databases
07/11/2018	Phillips	Patterns of inheritance, mosaicism, imprinting and anticipation
07/18/2018	Phillips/Owen	Newborn Screening
07/25/2018	Phillips	Recurrence risks and Hardy Weinberg
08/01/2018	Hannig	Bayesian risk calculations
08/08/2018	Duis	Fetal demise evaluation
08/15/2018	Dudak	Reproductive genetics
08/22/2018	Phillips	Fundamentals of Molecular Genetics
08/29/2018	Duis	Approach to a dysmorphic patient
09/05/2018	Reimschisel	Approach to inborn metabolic diseases
09/12/2018	Duis	Approach to an autistic patient
09/19/2018	Phillips	Approach to skeletal dysplasias
09/26/2018	Wheeler	Aneuploidies
10/03/2018	Wheeler	Chromosomal translocations and inversions
10/10/2018	Wheeler	Cytogenetics techniques

Table 2

10/17/2018	Wheeler	aCGH in clinical practice
10/24/2018	Hamid	Genomic sequencing in clinical practice-I
10/31/2018	Cogan	Genomic sequencing in clinical practice-II
11/07/2018	Weisner	Introduction to cancer genetics
11/14/2018	Pal	Hereditary cancers
11/21/2018	Phillips	Fatty acid oxidation disorders
11/28/2018	Hamid	Aminoacidopathies
12/05/2018	Morgan	Disorders of carbohydrate metabolism
12/12/2018	Morgan	Urea cycle diseases
12/19/2018	Morgan	Organic acidemias
07/03/2019	Owen/Wey	Nutritional management of metabolic disorders
07/10/2019	Reimschisel	Interpretation of plasma amino acids
07/17/2019	Reimschisel	Interpretation of organic acids and acylcarnitine profiles
07/24/2019	Duis	Epigenetic disorders
07/31/2019	Duis	Overgrowth syndromes
08/07/2019	Morgan	Lysosomal storage disorders
08/14/2019	Reimschisel	Primary mitochondrial diseases

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08/21/2019	Reimschisel	Neurogenetics
08/28/2019	Phillips	Triplet repeat disorders
09/04/2019	Phillips	Disorders of sexual differentiation
09/11/2019	Friedrich	Common genetic disorders in adults-I
09/18/2019	Friedrich	Common genetic disorders in adults-II
09/25/2019	Duis	Fetal demise evaluation
10/02/2019	Hannig	Genetics of hearing loss
10/09/2019	Fairbrother	Communication with patients and families (delivering diagnoses and bad news)
10/16/2019	Fairbrother	Genetic testing
10/23/2019	Fairbrother	Explaining difficult terms and concepts
10/30/2019	Hamid	Grant writing
11/06/2019	Van Driest	Pharmocogenomics
11/13/2019	Phillips	Ethical problems in genetics
10/20/2019	Phillips	Clinical trials in genetic disorders
10/27/2019	Faculty and Residents	Program Review

In addition to the core lectures above the residents will be take additional course work as outlined below

1. **Human Genetics 340 course (required)**. Designed to cover background and latest advances in human genetics. Topics will include an overview of mutational

mechanisms, cytogenetics (detection and description of chromosomal abnormalities), biochemical genetics (gene defects in biochemical pathways, inborn errors of metabolism), molecular genetics (gene structure, function, and expression). Topics are discussed with reference to specific human genetic diseases.

- 2. Human and Mammalian Genetics and Genomics Short Course (optional). the Human and Mammalian Genetics and Genomics Short Course topics include the following: Chromosome structure and function, 3D genome organization, Mendelian inheritance, Genotype to Phenotype, Transcriptional and post transcriptional regulation of gene expression, Next generation sequencing, Genome Editing, Molecular genetics, Epigenetics and methylation, Genetics of Autoimmunity, RNA sequencing, Exome sequencing, Genetics Association studies, Complex traits, Quantitative Systems Biology, Human Genomic Databases, Topics in Cancer Biology, Interpretation of Genetic Variants, Mouse Genome, Telomeres, Genetic Recombination, Copy Number Variations, HLA complex in Disease, Genetics of Infectious Diseases, Inborn Errors, Genetics and Environment, Malformation Syndromes, Precision Medicine, Pharmacogenomics, Metabolomics, Biomedical Ethics, Prenatal Screening, Treatment of Genetic Diseases and Stem cells.
- 3. Metabolic Academy (optional). This week-long course offers an immersive exposure to metabolic disorders. Topic covered in the course include the following: Amino Acid Disorders, Urea Cycle Disorders, Organic Acidemias, Disorders Of Vitamin Metabolism (Cobalamin, Folate, Biotin, Pyridoxine), Biochemical Genetics Laboratory (Amino Acids, GC/MS, Tandem MS, NBS), Introduction to Metabolic Nutrition, Hypoglycemia and Gluconeogenesis, Glycogen Storage Diseases, Inborn Errors of Fructose and Galactose Metabolism, Fatty Acid Oxidation Defects and Ketone Body Metabolism, Glucose Transporter Defects, Lactic Acidemia and Defects of Pyruvate Metabolism, Mitochondrial Energy Metabolism, Respiratory Chain Defects, Disorders of Metal Cofactors (Copper, Molybdenum), Defects of Neurotransmitter and Creatine Metabolism, Disorders of Phospholipid Synthesis, Disorders of Non-Mitochondrial Lipid Metabolism, Congenital Disorders of Glycosylation, Lysosomal Storage Disease Overview, Newborn Screening.
- 4. Option to take Human Genetics 341(optional)
- 5. <u>Option</u> to take additional more specific course work based on interests (program director will provide guidance)
- 6. Teach at least one lecture to Pediatric residents on Metabolic disease with the attending physician (required)
- 7. Teach at least one lecture to Pediatric residents on Dysmorphology with the attending physician (required)
- 8. Participate in monthly case-based didactics with a focus on ABMGG curriculum (required)