LEARNING OBJECTIVES: <u>EL GENETICS MEDICAL GENETICS ELECTIVE</u>

Overarching Goals of Curriculum	Elective objectives: By the end of the rotation, students will be expected to:	Where/how taught *(location or learning activity)*	Taught by *(attending, fellows, etc.)*	How student's achievement of objective is assessed *(assessment method)*	How feedback is given *(feedback method)*	Quantity target *(target number of patients/ events during rotation)*
1, 2, 3, 4, 5, 6	1. **History skills:** Gather the important information that is needed to obtain a useful genetic history that includes: • Recognition of single gene disorders causing specific phenotypes. • Recognition of Congenital malformations and mental retardation. • Recognition of disorders of intermediary metabolism involving cardinal symptoms of inborn errors.	Outpatient clinics, inpatient consults	Attending and fellows	Direct Observation and Case Presentation	Verbal	2- 4 inpatients per week, 4 clinics per week
1, 2, 3, 4, 5	2. **Physical examination skills:** Complete a physical exam resulting in the: • Recognition of single gene disorders causing specific phenotypes. • Recognition of Congenital malformations and mental retardation. • Recognition of disorders of intermediary metabolism involving cardinal signs of inborn errors.	Outpatient clinics, inpatient consults	Attending and fellows	Direct Observation and Case Presentation	Verbal	2- 4 inpatients per week, 4 clinics per week
1, 2, 3, 4, 5, 6, 7, 8	3. **Knowledge/diagnostic and treatment skills:** Demonstrate the following skills: **Congenital malformations and mental retardation** • Recognition and assessment of multiple congenital anomalies, with or without mental retardation. • Effects of major teratogens and mechanisms of teratogenesis. • Use of cytogenetic& molecular genetic tests to diagnosis congenital malformations and mental retardation. • Use of literature to consider single gene disorders. **Predictive screening for genetic disease** • Principles of newborn screening; principles of follow up for abnormal tests.	Outpatient clinics, inpatient consults, laboratory	Attending and fellows	Direct Observation and Case Presentation	Verbal	2- 4 inpatients per week, 4 clinics per week

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	• Principles of carrier screening for single gene disorders based on ethnic background. • Principles of screening for disease pre-disposition. • Issues in advising patients and parents about the risks and benefits of screening. **Recognition of single gene disorders causing specific phenotypes** • Skeletal dysplasias. Distinctive dysmorphology or physical findings (e.g. NF, facial dysmorphic syndromes). Major organ abnormalities (renal, cardiac, gi, pulmonary, cns). **Disorders of intermediary metabolism** • Principles of acute management of metabolic emergencies. • Formulation of a differential diagnosis. • Principles of long term management of inborn errors. **Genes and disease: mechanisms of inheritance** • Genes and cancer. • Unusual mechanisms of gene action (trinucleotide repeats, imprinting). • Common disorders, e.g. diabetes. **Use of genetic testing** • Special issues in genetic testing in children. • Principles of selecting appropriate genetic tests. • Recognition of pitfalls in genetic testing.					
4	4. **Procedural skills:** Demonstrate the following skills in management of genetic disease: **Predictive screening for genetic disease** • Principles of newborn screening; principles of follow	Outpatient clinics, inpatient consults, laboratory	Attending and fellows	Direct Observation and Case Presentation	Verbal	2- 4 inpatients per week, 4 clinics per week

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	up for abnormal tests. • Principles of carrier screening for single gene disorders based on ethnic background. • Principles of screening for disease pre-disposition. • Issues in advising patients and parents about the risks and benefits of screening. **Treatment of genetic disease** • Environmental management. • Replacement of missing protein or gene product. • Gene therapy. **Skills in use of genetics resources** • OMIM (online mendelian inheritance in man) • Metabolic and Molecular Basis of Inherited Disease (OMMBID) (online textbook via YMS library. • POSSUM (syndrome diagnosis). • NCBI clinical genetics resources such as OMIM, GeneTests.					
4, 5, 6	5. **Attitude:** Demonstrate professional responsibility in working as a team member with other members of the care team, patients and families. **The student should exhibit sensitivity to the use of genetic information**: • How novel scientific discoveries can be evaluated in a clinical context. • Principles of communicating genetic information to parents and families. • Cultural and social issues in genetic information. • Problems in confidentiality. • Delivery of bad news. • Use of genetic consultants.	Outpatient clinics, inpatient consults, laboratory	Attending and fellows	Direct Observation and Case Presentation	Verbal	2- 4 inpatients per week, 4 clinics per week

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5, 6, 8	6. **Career/context**: Know the training/career pathway for Medical Genetics.	Outpatient clinics, inpatient consults, laboratory	Attending and fellows	Direct Observation and Case Presentation	Verbal	2- 4 inpatients per week, 4 clinics per week