MEDICAL GENETICS CLINICAL CARE ROTATION

Overview: The Medical Genetics Clinical Care Rotation (MGCCR) is the backbone of the clinical genetics experience for the Medical Genetics Residents/Fellows (hereafter referred to as the MGF). The MGCCR includes twelve of the eighteen months of clinical rotations required in the program. Every effort will be made for six months of the rotation to occur during the first year of training and the other six months to occur during the second year of training. In the first six months, the MGF will be closely supervised and learn the basic principals of Medical Genetics. The first six months on the MGCCR will have flexibility as the MGF will be taking their required didactic coursework during the first year of the program. Therefore, the MGF will be allowed time for class attendance, studying etc. while on the service. In contrast, during the second six months of training, the MGF will acquire more complex skills and have increasing responsibility for supervising the Medical Genetics consultative service including instruction of other trainees. Other trainees on the Genetics service include genetic counseling masters students, medical students, pediatric residents and fellows from other services (maternal-fetal medicine, pediatric endocrinology and molecular genetic pathology fellows). The MGF will not be involved in coursework etc, during the latter six months on the rotation. The MGCCR is a combination inpatient and outpatient rotation. The inpatient consultative service includes three hospitals: Memorial Hermann Children's Hospital, Lyndon Baines Johnson General Hospital (LBJGH), and the Shriners Hospital for Children. The outpatient sites include the Pediatric Medical Genetics Clinic at the Hermann Professional Building, the Pediatric Medical Genetics Clinic at LBJGH and Shriners Hospital Genetics Clinic.

During the first six months of the rotation, the MGF will be closely supervised by the Genetics Attending. The MGF will be instructed on how to approach the differential diagnosis for a referred patient including use of the many genetics diagnostic tools available such as software programs (LDDB and POSSUM) and medical literature searches. The MGF will provide the initial assessment of assigned patients followed by review with the attending. The MGF will acquire skills to include writing clinic notes as well as follow-up counseling letters to the patients/families. The MGF will observe counseling sessions ranging from straightforward scenarios such as chromosome disorders like Down syndrome to more complex situations such as mitochondrial disorders. During the second six months of the rotation, the MGF will progress to provision of genetic counseling under the supervision of the attending. By the completion of the 12 months on the MGCCR, the MGF should be confident in provision of genetic care from diagnosis through counseling and follow-up. Adherence to the 80-hour work week is mandated. Residents are supervised by faculty members who are boarded in Clinical Medical Genetics (either M.D., Ph.D. or M.D./Ph.D.).

Legend for Learning Activities		
AR - Attending Rounds	WH - Written Homework	FS – Faculty Supervision
ASR - Assigned Reading	DPC - Direct Patient Care	GJC-Genetics Journal Club
GCC-Genetics Clinical Conference	M/DO - Modeling/Direct Observation	GSOC-Genetics Sign-Out Conference

Legend for Evaluation Methods for Residents	
AE - Attending Evaluation	CR - Chart Review
DSP- Directly Supervised Procedures	DO - Direct Observation
360° - Global Evaluation	RWH - Review of Written Homework

Principal Educational Goals and Objectives by Relevant Competency

The principal educational goals for residents on this rotation are indicated for the relevant ACGME competencies. The tables below each goal list the corresponding educational objectives, the relevant learning activities, and the evaluation methods for each objective. The educational goals and objectives are applicable to PGY-4 and PGY-5 Medical Genetics Residents/Fellows. The expected competency level demonstrated by the residents should reflect their respective level of experience.

Competency 1 – Patient Care. Provide clinical care in the area of Medical Genetics to patients/families who are either affected or potentially affected by a condition that has a genetic component.

GOAL: Determine whether a medical condition has a genetic etiology.

	Principal Educational Objectives	Learning Activities	Evaluation Methods
1.	Obtain and document a medical history that includes a detailed prenatal history and a detailed family history.	DPC, AR, FS, GSOC	AE, CR, DO
2.	Perform and document a thorough physical exam that includes measurements to determine normal v. abnormal (i.e. inner canthal length, outer canthal length, palpebral fissure length, ear length, arm span, upper/lower segment ratio, total hand length, middle finger length, etc.) as indicated on patients referred for a Medical Genetics evaluation.	DPC, AR, FS, GSOC	AE, CR, DO
3.	Perform and document a thorough physical examination on a child suspected of a specific genetic disorder, identifying major and minor congenital anomalies that could be signs of an underlying genetic syndrome.	DPC, AR, FS, GSOC	AE, CR, DO
4.	Develop a management plan for commonly encountered genetic disorders, identifying principles of long-term management, including use of disorder-specific growth charts and practice guidelines.	DPC, M/DO, ASR, GSOC	AE, CR, DO
5.	Identify resources in your community for diagnosis, genetic counseling, therapy and psychosocial support of children with genetic defects and congenital anomalies.	ASR, AR	AE, CR, DO

GOAL: Conditions requiring urgent referral (Genetics and Inborn Errors of Metabolism). Recognize and respond to urgent and/or severe conditions related to genetics and inherited metabolic disorders.

	Principal Educational Objectives	Learning Activities	Evaluation Methods
1.	Identify, explain, provide initial management and support, and seek urgent referral for the following genetic and/or metabolic conditions: - Infants presenting with symptoms indicating the possibility of a severe inborn error of metabolism (e.g., metabolic acidosis, hyperammonemia, unexplained seizures, ketosis or hypoketosis, profound hypoglycemia). - Dysmorphic features found in chromosomal abnormalities and lethal skeletal dysplasias that require prompt diagnosis in the perinatal period (e.g., Trisomy 13, 18, 21, thanatophoric dysplasia).	DPC, FS, M/DO, ASR	AE, CR, DO, 360°, DSP

1.	- Unexplained critical illness or death suggestive of metabolic disorder, requiring collection of tissue samples before or at time of death. - Developmental delay with signs or symptoms suggesting an underlying metabolic or genetic disorder. - Physiologic changes or regression of milestones that suggest a possible metabolic etiology (e.g., urea cycle disorders, mitochondrial disorders, lysosomal storage diseases, abnormalities or organic/amino metabolism).		

<u>Competency 2 - Medical Knowledge</u>. Understand the scope of established and evolving biomedical, clinical, epidemiological and social-behavioral knowledge needed by a Medical Geneticist; demonstrate the ability to acquire, critically interpret and apply this knowledge in patient care.

GOAL: Recognize presenting symptoms, diagnose, describe the pathophysiology, and manage common presentations of the following genetic conditions.

	Principal Education Objectives	**Learning Activities**	**Evaluation Methods**
1.	Describe findings of chromosome abnormalities including: Common Trisomies (13, 18, 21), Turner syndrome (45X), Deletion syndromes (22q.11, Wolf-Hirschhorn, Smith-Magenis, etc.), and Sex chromosome aneuploidies (Klinefelter syndrome (47, XXY) and 47, XYY syndrome).	ASR, WH, GSCO, GCC, DPC	AE, DO, CR
2.	Describe findings of common single gene disorders including: cystic fibrosis, sickle cell anemia, neurofibromatosis, tuberous sclerosis, Marfan syndrome, achondroplasia, PKU and galactosemia.	ASR, WH, GSCO, GCC, DPC	AE, DO, CR
3.	Describe the evaluation of a patient with a potential metabolic emergency.	ASR, WH, GSCO, GCC, DPC	AE, DO, CR

GOAL: Differentiate disorders in patients associated with genetic predisposition or genetic disease from normal states or acquired disorders.

	Principal Educational Objectives	**Learning Activities**	**Evaluation Methods**
1.	Describe common patterns of Mendelian vs. non-Mendelian inheritance (autosomal dominant and recessive, X-linked, multifactorial, and the effect of maternal and paternal age) and demonstrate the ability to construct a pedigree.	ASR, WH, GSCO, GCC, DPC	AE, DO, CR
2.	Discuss unusual patterns of inheritance (mitochondrial defects, anticipation and triplet repeat disorders, imprinting, uniparental disomy/isodisomy).	ASR, WH, GSCO, GCC, DPC	AE, DO, CR
3.	Identify common diseases with known inheritance patterns and describe the mode of inheritance, including: cystic fibrosis, sickle cell anemia, Marfan syndrome, tuberous sclerosis, neurofibromatosis, and PKU.	ASR, WH, GCC, DPC GSCO, AR	AE, DO, CR
4.	Identify common disorders with unusual inheritance patterns and describe the mode of inheritance, including: trinucleotide repeat	DPC, ASR, WH, GCC	AE, DO, CR

	disorders (Fragile X syndrome, myotonic dystrophy, Huntington disease, etc.) and maternally inherited mitochondrial disorders (MELAS).	GSCO, AR	
5.	Explain the findings on clinical history and examination that suggest a known or potential genetic disorder or inborn error of metabolism.	DPC, ASR, WH, GCC GSCO, AR	AE, DO, CR
6.	Describe how well child care differs in a child with a genetic condition, e.g., use of specific growth charts for specific conditions and physical findings.	DPC, ASR, WH, GCC GSCO, AR	AE, DO, CR
7.	Identify appropriate clinical and laboratory tests to help identify genetic diseases and inborn errors of metabolism. Explain the reason for the test to a family and interpret the results, with the assistance of a geneticist. The tests should include the following: - Chromosome analysis (both metaphase and prophase) and FISH testing for specific disorders. - Plasma and urine amino acids, urine organic acids, ammonia level, venous pH, lactate, pyruvate, and blood acylcarnitine profile. - Molecular testing for triplet repeat and imprinting disorders. - DNA mutational testing for selected disorders. - Newer and future technologies developed for detection of genetic disorders (e.g., CGH/microarray technology).	DPC, FS, M/DO	AE, DO, CR

GOAL: Undifferentiated signs and symptoms (Genetics and Inborn Errors of Metabolic). Evaluate, treat, and/or refer patients with the presenting signs and symptoms that suggest a genetic disease process.

	Principal Educational Objectives	**Learning Activities**	**Evaluation Methods**
1.	Create a strategy to determine if the following presenting signs and symptoms are caused by genetic disease or an inborn error of metabolism and determine if the patient needs treatment or referral. - Developmental delay - Dysmorphic features - Poor feeding - Vomiting - Failure to thrive - Seizures - Short stature - Unusual behavior - Short stature - Hearing loss - Cleft lip/palate - Respiratory disorders - Obesity - Skin lesions - Hypotonia	DPC, AR, M/DO, ASR, WH, GCC, GSOC	AE, CR, DO

GOAL: Molecular medicine. Recognize genetic factors in common diseases of childhood and adulthood.

	Principal Educational Objectives	**Learning Activities**	**Evaluation Methods**
1.	Discuss current knowledge regarding the molecular basis of common childhood and adult conditions.	ASR, DPC	AE, CR, DO
2.	Identify the current and future uses of DNA testing in the office setting, including diagnosis of infectious diseases using DNA, pharmacogenetic testing for inborn errors of metabolic pathways prior to prescribing, DNA chips to identify genetic etiologies for complex disorders (e.g., congenital heart disease, seizure disorders, etc.).	AR, ASR	AE, CR, DO

<u>Competency 3 – Interpersonal and Communications Skills</u>. Demonstrate interpersonal and communication skills that result in information exchange and partnering with patients, their families and professional associates.

GOAL: To participate in provision of genetic counseling to a patient and or the patient's parents including: diagnosis, prognosis and recurrence risk (for the parents as well as for the child when he/she reproduces).

	Principal Educational Objectives	**Learning Activities**	**Evaluation Methods**
1.	During the first six months of the rotation, participate in genetic counseling sessions for chromosome disorders, single gene disorders multifactorial disorders, and mitochondrial disorders. In the latter six months of the rotation, provide genetic counseling for all of the categories of genetic conditions listed above under the supervision of the attending faculty member.	DPC, AR, FS, WH, M/DO	AE, CR, DO, 360°
2.	Write genetic counseling letters to patients/families after participation in/provision of genetic counseling sessions for chromosome disorders, single gene disorders, multifactorial disorders, mitochondrial disorders, sporadic conditions, and conditions with unknown etiology.	DPC, FS, WH, M/DO	RWH, AE
3.	Under direct supervision of the Genetics Faculty member, discuss sensitive issues that relate to a patient's genetic condition such as limitations for acquisition of life skills in mentally retarded patients, long-term plans for mentally retarded patients who most likely will outlive their parents, etc.	DPC, AR, FS	AE, CR, DO
4.	Communicate effectively with physicians, other health professionals, and health related agencies to create and sustain information exchange and team work for patient care.	DPC, AR, FS	AE, CR, DO
5.	Maintain accurate, legible, timely and legally appropriate medical records for Medical Genetics patients in the outpatient and inpatient setting.	DPC, AR, FS	AE, CR, DO

<u>Competency 4 – Practice-based Learning and Improvement</u>. Demonstrate knowledge, skills and attitudes needed for continuous self-assessment, using scientific methods and evidence to investigate, evaluate, and improve one's patient care practice.

	Principal Educational Objectives	**Learning Activities**	**Evaluation Methods**
1.	Develop strategies to learn about future advances in the understanding of genetic disorders, in order to incorporate into one's practice improved screening, identification, counseling and management of such disorders.	GJC, AR, ASR, GCC, GSOC	DO, AE, CR
2.	Identify the indicators that would lead you to seek a genetics consult.	GJC, AR, ASR, GSOC	DO, AE, CR
3.	Identify personal learning needs, systematically organize relevant information resources for future reference, and plan for continuing data acquisition if appropriate.	GJC, AR, ASR, GSOC	AE, CR, DO

<u>Competency 5 – Professionalism</u>. Demonstrate a commitment to carrying out professional responsibilities, adherence to ethical principles, and sensitivity to diversity.

	Principal Educational Objectives	Learning Activities	Evaluation Methods
1.	Discuss the ethical, legal, financial and social issues involved in genetic testing of children for genetic disorders that may present in adulthood, testing children for carrier status, and providing medical care for patients with known fatal disorders.	GSOC, AR, DPC, GJC, GCC	AE
2.	Demonstrate personal accountability to the well being of all patients, even when other physicians are primarily responsible for their care, for example, by following up on lab results, writing comprehensive notes, seeking answers to difficult patient care questions, and communicating with primary care physicians.	M/DO, GJC, AR, ASR, GCC, GSOC	AE, DO
3.	Demonstrate a commitment to carrying out professional responsibilities, adherence to ethical and legal principles, and sensitivity to diversity while providing care to patients/families affected by genetic conditions.	GJC, AR, ASR, GCC, GSOC	AE, DO

<u>Competency 6 - Systems-Based Practice</u>. Understand how to practice quality health care and advocate for patients within the context of the health care system.

	Principal Educational Objectives	Learning Activities	Evaluation Methods
1.	Identify written and internet resources to aid in diagnosing a genetic or inborn error of metabolism, using physical findings along with laboratory examination.	DPC, FS, ASR	AE, CR, RWH
2.	Demonstrate sensitivity to the costs of clinical care in Medical Genetics and take steps to minimize costs without compromising quality.	DPC, FS, ASR	AE, CR, RWH
3.	Identify research studies for patients' possible participation and facilitate enrollment of patients in research studies (ie consent patients, aid in obtaining and sending samples, etc)	DPC, FS, ASR	AE, CR, DO
4.	Recognize the limits of one's knowledge and expertise and take steps to avoid medical errors.	DPC	AE
5	Understand key aspects of health care systems as they apply to care of patients and their families, including cost control, billing and reimbursement.	DPC	AE
6.	Recognize and advocate for families who need assistance to deal with systems complexities, such as lack of insurance, multiple medication refills, multiple appointments with long transport times, or inconvenient hours of service.	DPC	AE