MEDICAL GENETICS SPECIALTY CLINICS ROTATION

Overview: The Medical Genetics Specialty Clinics Rotation (MGSCR) is a one month required rotation for the Medical Genetics Residents/Fellows (hereafter referred to as the MGF). The MGSCR is designed to introduce the resident to utilization and value of multi-specialty clinics in the practice of Medical Genetics. Over the one month period of the rotation, the resident will attend the following multidisciplinary clinics:

1. Craniofacial/Cleft Lip and Palate (CLP)-third Monday all day at Suite 1400 in Hermann Professional Building (HPB)
2. Hemophilia/Thrombophilia-every Wednesday PM at 5th Floor Pediatric Clinic in HPB
3. CHOSEN [Chronic Health Oriented Services de Ninos (for Children)] Clinic-every Monday PM and every Wednesday AM at 5th Floor Pediatric Clinic in HPB
4. Shriners Hospital Metabolic (Rickets)-fourth Friday PM at Shriners Hospital Outpatient Clinic (SHOPC)
5. Shriners Hospital Cerebral Palsy (CP)-2-3 half days/week; day varies; SHOPC
6. Shriners Hospital Osteogenesis Imperfecta (OI)-fourth Friday PM at SHOPC
7. Shriners Hospital Spina Bifida Meningomyelocoele (SBMM)-third Friday all day at SHOPC
8. Shriners Hospital Arthrogryposis-fourth Monday all day at SHOPC
9. Shriners Hospital Screening (General)-1-2 times/month all day on Thursday at SHOPC

The clinics are designed around: single gene disorders (rickets, OI, hemophilias/thrombophilias); common, complex birth defects (spina bifida meningomyelocoele); a combination of single gene disorders and common birth defects (craniofacial/cleft lip and palate, arthrogryposis); or a mixture of genetic and non-genetic conditions (CHOSEN, CP, screening). There are subtle nuances between these clinics depending on the patient group. The MGF will learn the role of the Medical Geneticist in a multidisciplinary team that includes doctors from other specialties (pediatrics, pediatric intensive care, plastic surgery, hematology, pediatric endocrinology, orthopedic surgery, pediatric neurology, and pediatric neurosurgery) as well as nurses and other health care professionals (genetic nurses, genetic counselors, physical therapists, occupational therapists, nutritionists, speech pathology, dentists, etc.). For all of the clinics, the MGF will perform literature searches regarding conditions considered as diagnostic possibilities or observed in patients who are evaluated. The MGF will also learn to order specific genetic tests as appropriate for patients with suspected conditions versus patients with unknown diagnosis. Each clinic is described individually followed by a listing of Goals and Objectives for all of the clinics. Adherence to the 80-hour work week is mandated.

Craniofacial/Cleft Lip and Palate Clinic

The clinic meets once per month all day (third Monday) at the HPB (across the street from the Medical School and Hospital) in Suite 1400. The MGF will join a multidisciplinary team including plastic surgeons, dentist, orthodontic specialists, nutrition, speech pathology, and general pediatrician. Dr. Jacqueline Hecht is a regular participant in this clinic. She has performed research for many years on CLP searching for susceptibility genes. In addition to CL P, patients with a number of other genetic conditions are referred to this clinic [ie various craniosynostoses (Apert syndrome, Pfeiffer syndrome, Crouzon syndrome, Saethre-Chotzen syndrome), Goldenhar syndrome, Stickler syndrome, etc.]

Hemophilia/Thrombophilia Clinic

The clinic meets weekly on Wednesday afternoons at the HPB. The MGF will join a multidisciplinary team including hematologists, nurses, physical therapists, physician assistants, and a social worker. Aimee Williams, MS, CGC, has been involved with the clinic since 1997 and is an expert concerning these disorders. Patients referred to the clinic include those affected by hemophilia A and B and von Willebrand disease (VWD) as well as many rarer hemophilias/thrombophilias.

CHOSEN (Chronic Health Oriented Services de Ninos (for Children)

The clinic meets two half days per week on Monday PM and Wednesday AM at the 5th Floor Pediatric Clinic in HPB. The MGF will join a nurse coordinator (Gayla Roberson, RN), a pediatric intensive care specialist (Dr. John Cochran), a social worker (Enrique Matallana, MSW) and a clinic coordinator. The CHOSEN Clinic is designed to provide primary care to patients who have multiple medical problems and often also have developmental delays/mental retardation. A significant number of Genetics patients who have multiple problems are referred for primary care to the CHOSEN Clinic as most pediatricians in private practice do not have the time and resources to help these families. Many of these families do not have financial resources to obtain services they need. Through the CHOSEN Clinic, these special patients can have much better care coordination and families get much needed help in obtaining resources available through federal and state agencies. It is important for the MGF to learn about care coordination of multiply handicapped children both physically and mentally. Many patients with different genetic conditions including chromosomal, single gene and multifactorial are followed through CHOSEN Clinic making it an ideal location to learn about utilization of healthcare resources and working within the medical system to help patients.

Shriners Hospital Metabolic (Rickets) Clinic

The clinic meets once per month on the fourth Friday afternoon at SHOPC. The MGF will join a multidisciplinary team that includes Barbara Dominguez, BSN, Genetics Nurse Specialist, Dr. Patrick Brosnan, pediatric endocrinologist, and Dr. Byron York, pediatric orthopedist. The Rickets Clinic is one of the largest clinics in the nation focusing on rickets. The most common type of rickets is the X-linked hypophosphatemic variety providing an excellent clinical example of an X-linked dominant condition with variable phenotype.

Shriners Hospital Cerebral Palsy (CP) Clinic

The clinic meets 2-3 half days/week at SHOPC. The schedule is published on a monthly basis and will be provided to the MGF for planning their attendance. The CP Clinic is staffed by a variety of orthopedic surgeons at Shriners Hospital as well as support staff including nurses, physical therapists, social workers, etc. The CP Clinic, like CHOSEN Clinic described above, includes many patients who have underlying genetic diagnoses. It provides the MGF with an opportunity to learn to distinguish multiply handicapped patients with genetic versus non-genetic etiologies for their problems. It also provides an opportunity to learn about obtaining resources for patients and working within a system.

Shriners Hospital Osteogenesis Imperfecta (OI) Clinic

The clinic meets once per month on the fourth Friday afternoon at SHOPC. The MGF will join a team of orthopedic surgeons and support staff. The OI patients will provide the opportunity to learn about a single gene disorder with extreme variability in phenotype. With the recent innovation of therapy for OI, the MGF will also have the opportunity to learn about how some genetic diseases can be effectively treated.

Shriners Hospital Spina Bifida Meningomyelocoele (SBMM) Clinic

The clinic meets once per month all day on the third Friday at SHOPC. The MGF will join a team including orthopedic surgeons, pediatric urology, pediatric neurosurgery and support staff. The MGF will learn about the multitude of problems faced by individuals affected with one of the most common complex birth defects. As we have learned through the increased longevity of SBMM patients after the advent of surgical intervention in the 1960s, the problems of SBMM extend beyond the typically associated physical handicaps of ambulation and bowel/bladder control to include learning deficits hampering the ability to integrate into society. Working with specialists from diverse specialties such as neurosurgery and urology reinforce the concepts of teamwork for approaching difficult care issues and utilization of resources.

Shriners Hospital Arthrogryposis Clinic

The clinic is held all day on the fourth Monday of each month at SHOPC. The MGF will join a team of orthopedic surgeons and support staff. The clinic includes all patients with a non-specific diagnosis of “arthrogryposis”. Examples of various diagnoses of patients referred to the clinic include arthrogryposis multiplex congenita (AMC), Escobar syndrome and multiple pterygium syndrome. The MGF will learn about the different diagnoses include in this broad descriptive category and how to distinguish genetic versus non-genetic diagnoses.

Shriners Hospital Screening (General) Clinic

The clinic meets 1-2 times each month all day on Thursday at SHOPC. At this clinic triaging occurs for new patients in the Shriners Hospital system. Individuals from all the different clinics including Genetics Clinic participate. Patients are assigned all of their appointments to the different specialty clinics at Shriners Hospital. Attendance at this clinic should aid the MGF in honing skills to determine whether a patients’ constellation of symptoms merits genetic referral and work-up.

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		
RWH - Review of Written Homework	DO - Direct Observation	CR - Chart Review

AE - Attending Evaluation 360° - Global Evaluation

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.

<u>Competency 1 – Patient Care</u>. As part of a multi-specialty team, 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: Aid the team in determining whether a medical condition has a genetic etiology.

	Principal Educational Objectives	Learning Activities	Evaluation Methods
1.	Contribute to the work of the multidisciplinary specialty team by obtaining information from the patient/family that will aid in determining whether the presenting symptoms and/or condition have a genetic etiology (ie obtain a detailed prenatal history and a detailed family history).	DPC, AR, FS	AE, CR, DO
2.	Contribute to the work of the multidisciplinary specialty team by performing and documenting 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.) to aid in determining whether the presenting symptoms and/or condition have a genetic etiology.	DPC, AR, FS	AE, CR, DO
3.	When appropriate, 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	AE, CR, DO
4.	Contribute to the development a management plan for patients evaluated/cared for through the multidisciplinary specialty clinic identifying principles of long-term management, including use of disorder-specific growth charts and practice guidelines.	DPC, M/DO, ASR	AE, CR, DO
5.	Work with the multidisciplinary specialty team that includes other professionals such as physical therapists, nutritionists, social workers, etc. to identify resources in your community to meet the medical and intellectual needs of the patients.	ASR, AR	AE, CR, DO

GOAL: Recognize specific conditions encountered in the various multi-specialty clinics and learn how to diagnose, treat and manage these conditions.

	Principal Educational Objectives	**Learning Activities**	**Evaluation Methods**
1.	In the CL&P Clinic, participate in the diagnosis, management and counseling of patients/families with non-syndromic CL&P, syndromic CL&P and other craniofacial anomalies such as the various craniosynostoses syndromes, Goldenhar syndrome, hemifacial microsomia and Stickler syndrome.	DPC, FS, M/DO, ASR, GSOC	AE, CR, DO, 360°
2.	In the Hemophilia/Thrombophilia Clinic, participate in the diagnosis, management and counseling of patients/families with the common hemophilias/thrombophilias (ie Hemophilia A and B, von Willebrand's disease, Factor V Leiden,etc.)	DPC, FS, M/DO, ASR	AE, CR, DO, 360°
3.	In the CHOSEN Clinic, participate in the diagnosis, management and counseling of patients/families with lethal trisomies and rare chromosome disorders, single gene conditions (ie spinal muscular atrophy, various muscular dystrophies, Cornelia de Lange syndrome, etc.), teratogenic exposures (drugs and/or alcohol) and multifactorial conditions.	DPC, FS, M/DO, ASR	AE, CR, DO, 360°
4.	In the Shriners Hospital Metabolic (Rickets) Clinic, participate in the diagnosis, management and counseling of patients/families with X-linked hypophosphatemic, vitamin D-dependent rickets and autosomal dominant rickets.	DPC, FS, M/DO, ASR	AE, CR, DO, 360°
5.	In the Shriners Hospital Cerebral Palsy Clinic, aid the multidisciplinary specialty team in distinguishing between CP and an underlying genetic disease that is masquerading as CP.	DPC, FS, M/DO, ASR	AE, CR, DO, 360°
6.	In the Shriners Hospital Osteogenesis Imperfecta (OI) Clinic, participate in the diagnosis, management and counseling of patients/families with OI. Describe the different types of OI including underlying molecular bases and the different phenotypes that are observed. Describe the emerging treatment regimen for OI.	DPC, FS, M/DO, ASR	AE, CR, DO, 360°
7.	In the Shriners Hospital Spina Bifida Meningomyelocoele (SBMM) Clinic, participate in the diagnosis, management and counseling of patients/families with SBMM as well as other rarer types of neural tube defects (NTDs) followed through the SBMM Clinic. Describe the challenges faced by individuals with SBMM and other NTDs including not only physical problems (ie ambulation and bowel/bladder continence) but also learning problems that interfere with acquisition of everyday life skills (ie difficulties with numeracy).	DPC, FS, M/DO, ASR	AE, CR, DO, 360°

8.	In the Shriners Hospital Arthrogryposis Clinic, participate in the diagnosis, management and counseling of patients/families with arthrogryposis including sporadic (ie arthrogryposis multiplex congenital) forms as well as single gene disorders (ie Escobar syndrome, multiple pterygium syndrome, popliteal pterygium syndrome)	DPC, FS, M/DO, ASR	AE, CR, DO, 360°
9.	In the Shriners Hospital Screening Clinic, participate in triaging patients to various clinics including Genetic clinic.	DPC, FS, M/DO, ASR	AE, CR, DO, 360°

Competency 2 - Medical Knowledge. 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.	After attending the Craniofacial/CL&P Clinic, be able to list the common craniosynostoses and describe the phenotype and genotype of each.	ASR, WH, GSCO, GCC, DPC	AE, DO, CR
2.	After attending the Craniofacial/CL&P Clinic, describe the difference between non-syndromic and syndromic disorders that have CL&P as a feature.	ASR, WH, GSCO, GCC, DPC	AE, DO, CR
3.	After attending the Hemophilia/Thrombophilia Clinic, describe the common hemophilias/thrombophilias (ie Hemophilia A and B, von Willebrand's disease, Factor V Leiden,etc.).	ASR, WH, GSCO, GCC, DPC	AE, DO, CR
4.	After attending the CHOSEN Clinic, describe the clinical features of the lethal trisomies and rare chromosome disorders, single gene conditions (ie spinal muscular atrophy, various muscular dystrophies, Cornelia de Lange syndrome, etc.), teratogenic exposures (drugs and/or alcohol) and multifactorial conditions.	ASR, WH, GSCO, GCC, DPC	AE, DO, CR
5.	After attending the Shriners Hospital Metabolic (Rickets) Clinic, describe the clinical features of X-linked hypophosphatemic, vitamin D-dependent rickets and autosomal dominant rickets.	ASR, WH, GSCO, GCC, DPC	AE, DO, CR
6.	After attending the Shriners Hospital Osteogenesis Imperfecta (OI) Clinic, describe the clinical features including variability, genetic bases, testing modalities and treatment for all types of OI.	ASR, WH, GSCO, GCC, DPC	AE, DO, CR
7.	After attending the Shriners Hospital Spina Bifida Meningomyelocoele (SBMM) Clinic, describe SBMM as well as other NTDs including lethal forms such as anencephaly and craniorachischisis. Discuss the postulated genetic and environmental susceptibility factors. Discuss prevention by vitamin supplementation as well as prenatal testing available for detection.	ASR, WH, GSCO, GCC, DPC	AE, DO, CR
8.	After attending the Shriners Hospital Arthrogryposis Clinic, describe sporadic (ie AMC) as well a single gene (ie Escobar syndrome, multiple pterygium syndrome, popliteal pterygium syndrome) diagnoses encountered at the clinic.	ASR, WH, GSCO, GCC, DPC	AE, DO, CR

9.	After attending the Shriners Hospital Screening Clinic, describe a system for triaging patients encounterd in a pediatric orthopedic hospital to various clinics with focus on how patients are selected for referral to Genetics Clinic.	ASR, WH, GSCO, GCC, DPC	AE, DO, CR

<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.	Provide genetic counseling when appropriate as part of the genetics contribution to the care of the patient/family evaluated and cared for at the multidisciplinary specialty clinic.	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 supervision of the Attending Faculty member at the multi-disciplinary specialty clinic, 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 patients in the multidisciplinary specialty clinics.	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 evaluation in the various multidisciplinary specialty clinics.	GJC, AR, ASR, GCC, 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, GCC, 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.	Recognize the limits of one's knowledge and expertise and take steps to avoid medical errors.	DPC	AE
4.	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
5.	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