

## MEDICAL GENETICS CYTOGENETICS LABORATORY ROTATION

**Overview:** The Medical Genetics Cytogenetics Laboratory Rotation (MGCLR) is a required rotation for the Medical Genetics Residents/Fellows (hereafter referred to as the MGF). The MGCLR is a two week rotation that occurs in the Dynagene/Labcorp Cytogenetics Laboratory. The MGF will be supervised by Dr. Janice L. Smith. The MGF will observe laboratory methodologies for high resolution chromosome analysis from multiple different tissue types (white blood cells, skin fibroblasts, amniotic fluid, chorionic villi, bone marrows, and solid tissues). Additionally, the MGF will observe the fluorescence in-situ hybridization (FISH) technique as applied to analysis of prenatal samples, cancer samples as well as detection of constitutional deletions and rearrangements will be observed. Please see Section 7. Laboratory Data, A. Cytogenetics for a listing of the tests performed in the laboratory.

### **Description:**

The MGF will observe how samples are received in the laboratory and prepared for analysis. Observing the specimen receipt will provide the opportunity for the MGF to learn about quality assurance including safe guards to prevent sample mix-up or loss.

The MGF will observe the following methods/techniques from start to finish:

1. Tissue culture for samples from blood, skin biopsy, amniotic fluid samples, chorionic villi, bone marrows and solid tissues.
2. Cell harvest, slide preparation and staining.
3. Microscope analysis.
4. FISH
5. Computer digitizing for karyotyping.

### **Conferences:**

The MGF will participate in interpretation of results/diagnosis with the laboratory director, Dr. Smith. The MGF will participate in conferences and telephone consultations with clinicians involved in patient care. The MGF will participate in the Cytogenetics Case Review Conference on Mondays from 12-1PM. At the Cytogenetics Case Review, constitutional abnormalities are discussed. The MGF will also join the Director for Cancer Case Review Sessions as determined by the Director.

#### **Legend for Learning Activities**

AR - Attending Rounds	M/DO - Modeling/Direct Observation	E/C–Ethics/Communication Conferences
FS – Faculty Supervision	ASR - Assigned Reading	JC - Journal Club
CSOC-Case Sign-Out Conference	WH - Written Homework	RC - Research Conference

#### **Legend for Evaluation Methods for Residents**

AE - Attending Evaluation	DO - Direct Observation
DSP- Directly Supervised Procedures	RWH - Review of Written Homework
CR - Chart Review	CSR - Chart Stimulated Review
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 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: Observe laboratory techniques utilized in cytogenetic testing obtained on patients who are under assessment as possibly affected by a genetic disease.**

	Principal Educational Objectives	Learning Activities	Evaluation Methods
1.	Observe the following laboratory techniques: <ol style="list-style-type: none"> <li>1. Tissue culture for samples from blood, skin biopsy, amniotic fluid samples, chorionic villi, bone marrows and solid tissues.</li> <li>2. Cell harvest, slide preparation and staining.</li> <li>3. Microscope analysis.</li> <li>4. FISH,</li> <li>5. Computer digitizing.</li> </ol>	M/DO, FS	AE, DO

**Competency 2 - Medical Knowledge.** Understanding 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: Demonstrate knowledge regarding the results generated through testing in the Cytogenetics Laboratory.**

	Principal Education Objectives	Learning Activities	Evaluation Methods
1.	Participate in interpretation of laboratory data by the Cytogenetics Laboratory Director to diagnose chromosome abnormalities including aneuploidies, translocations, autosomal deletion syndromes, as well as distinguishing polymorphisms from abnormalities.	M/DO, FS, CSOC	AE, DO, CR

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

**GOAL: Participate in provision of results from the Cytogenetics Laboratory to referring genetic counselors/physicians or other appropriate professionals.**

	<b>Principal Educational Objectives</b>	<b>Learning Activities</b>	<b>Evaluation Methods</b>
1.	Participate in/provide results of cytogenetic testing to genetic counselors/physicians/other appropriate professionals.	WH, FS, M/DO	AE, DO
2.	Communicate effectively with genetic counselors, physicians, other health professionals, and health related agencies to create and sustain information exchange and team work for patient care.	FS, M/DO	AE, DO

**Competency 4 – Practice-based Learning and Improvement. 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.**

	<b>Principal Educational Objectives</b>	<b>Learning Activities</b>	<b>Evaluation Methods</b>
1.	Develop strategies to learn about future advances in the understanding of the cytogenetic basis of genetic disease, in order to incorporate into one’s practice improved screening, identification, counseling and management of these disorders.	ASR, M/DO, CSOC	DO, AE
2.	Identify personal learning needs, systematically organize relevant information resources for future reference, and plan for continuing data acquisition if appropriate.	ASR, M/DO	DO, AE,

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

	<b>Principal Educational Objectives</b>	<b>Learning Activities</b>	<b>Evaluation Methods</b>
1.	Discuss the ethical, legal, financial and social issues involved in genetic testing of patients/families, especially testing of children for disease status, and providing medical care for patients with known fatal disorders.	CSOC, E/C, M/DO	AE, DO
2.	Demonstrate personal accountability to the well being of all patients, even when other physicians are primarily responsible for their care, for example, by interpreting laboratory results and following with referring genetic counselors/physicians as appropriate.	M/DO, FS,	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 with genetic disease.	M/DO, FS	AE, DO

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

	<b>Principal Educational Objectives</b>	<b>Learning Activities</b>	<b>Evaluation Methods</b>
1.	Identify written and internet resources to aid in counseling patients/families with cytogenetic diseases including availability of research studies in which the patients/families might wish to participate.	ASR, WH, CSOC	AE, DO
2.	Demonstrate sensitivity to the costs of clinical care in Medical Genetics and take steps to minimize costs without compromising quality.	M/DO, FS	AE, DO

3.	Recognize the limits of one's knowledge and expertise and take steps to avoid laboratory errors.	M/DO, FS	AE, DO
4.	Understand key aspects of health care systems as they apply to care of patients and their families, including cost control, billing and reimbursement.	M/DO, FS	AE, DO
5.	Recognize and advocate for families who need assistance to deal with systems complexities, such as lack of insurance.	M/DO, FS	AE. DO