

## I. Introduction

Welcome to the Medical Genetics and Genomics Residency in the Department of Human Genetics/Division of Medical Genetics. The ACGME accredited residency-training program in Medical Genetics and Genomics at Emory University School of Medicine is under the direction of Dr. Michael J. Gambello, Program Director. Liz McKenna ([Elizabeth.mckenna@emory.edu](mailto:Elizabeth.mckenna@emory.edu)) is the Program Coordinator and assists with orientation. The residency offers broad training in pediatric and adult medical genetics and genomics. Outpatient experiences are mainly in The Emory Clinic. Inpatient experiences are primarily at Emory University Hospital and Children's Healthcare of Atlanta (CHOA) at Egleston and Scottish Rite. The residency also includes laboratory training. For comprehensive information provided by the ACGME please see <http://www.acgme.org/Specialties/Overview/pfcatid/9>. You should review the ACGME Program Requirements. We will provide you with a copy of this document.

## II. Duration

The resident will spend two years training and participating in eighteen months of broad-based, clinically oriented medical genetics activities, 6 weeks of clinical laboratory training, and 3 weeks a year of vacation. Three months will be reserved for self-directed learning, research, further study of a specific field of interest, poster and manuscript preparation.

## III. Description

This orientation document contains important information about the two year training program. Residents will be an integral part of the Division of Medical Genetics during their two years of training. We welcome input at any time to improve the educational goals and objectives.

## IV. Rotations:

### 1. General Genetics (Pediatric, Adult) and Metabolism with Call

These rotations will be the main source of patient encounters during training. Residents will have approximately 12 months of this rotation spread across 24 months, typically in 1-2 month blocks. Outpatient clinics are at the Emory Clinic at 1365 Clifton Road, 2<sup>nd</sup> floor. About 85% of the patients are children, 15% adults. Residents will see both children and adults (see logbook requirements on the ABMGG website <http://www.abmg.org/>). Residents will see at least two patients per full day clinic at first. Initially Dr. Gambello will assign the resident patients. After several months, *it will be the resident's responsibility to review the clinic list the week before and coordinate with other residents and medical biochemical fellow. In general, the biochemical fellow has priority for metabolism patients to satisfy logbook requirements. Then senior residents have priority of junior residents. Once you have assigned patients, speak with Charlotte Peinhardt, CGC so she can make the schedule. Charlotte will show trainees how to access the patient list and referral documents on the Clinic Drive.* The evaluation of a patient will include history, pedigree, physical examination, documentation and follow-up. Residents should keep a running list of all the patients they see and labs to follow. **The ACGME requires that ALL cases be logged in as one of 5 categories. It is best to do this on a monthly basis and not wait until the end of training.** Metabolism clinics are mainly Mondays, Tuesdays and Wednesdays.

Whenever possible residents will also be scheduled in skeletal disorders clinic, craniofacial clinic (CFC), Disorder of Sexual Differentiation (DSD) clinic and vascular anomalies clinic (VAC). The skeletal disorders clinic is staffed by Dr. William Wilcox and Charlotte Peinhardt and held at the Emory Clinic. The CFC, DSD Clinics and VAC are at the at the CAP (Center for Advanced Pediatrics 1400 Tullie Rd NE, Atlanta, GA 30329). CFC is staffed by either Dr. Sanchez or Dr. Neira and one of the GCs. DSD clinic (every second Friday) are staffed by Dr. Li and Eileen Barr, CGC. VAC is staffed by Dr. Sanchez.

Due to the SARS-CoV-2 pandemic, we have embraced telemedicine to deliver care for rare diseases. We anticipate that we will continue to have telemedicine as part of our health care delivery system, regardless of the pandemic. The resident will learn how to evaluate and care for patients via telemedicine. The resident will work closely with the attending for all telemedicine visits.

During General Genetics and Metabolism rotations, residents will take In-House Call for one week each month during which time there will be no outpatient responsibilities. We will ensure that residents have call with all attending geneticists. Call will include home call in the evenings and on weekends, though residents may need to round on the weekends depending on the patient load. ***For the first 6 months ALL evening and nighttime calls should be discussed thoroughly with the attending on call.*** Each week residents will have one administrative day with no clinic patients. The day may be changed from semester to semester.

### **Goals and Objectives for General Genetics and Metabolism**

1. **Patient Care** - Provide family centered patient care that is developmentally and age appropriate, compassionate, and effective for the treatment of genetic disease and the promotion of health.
  - a. Determine whether a medical condition has a genetic etiology.
    - i. Obtain and document a medical history that includes a detailed prenatal history and a detailed family history (pedigree).
    - ii. Perform and document a thorough physical exam on a child or adult suspected of having a genetic disorder, identifying major and minor congenital anomalies, including measurements to determine normal v. abnormal.
    - iii. Develop a management plan for commonly encountered genetic disorders including choosing the appropriate tests (e.g. Know when to order a Next Generation Gene Panel vs. a Whole Exome Sequence).
    - iv. Identify resources in the community for diagnosis, genetic counseling, therapy and psychosocial support of children and adults with genetic diseases and congenital anomalies.

- b. 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.
    - i. Identify, explain, provide initial management and support, and seek urgent referral for the following genetic and/or metabolic conditions:  
Metabolic acidosis, hyperammonemia, unexplained seizures, ketosis or hypoketosis, hypoglycemia, and acute liver failure.
    - ii. Dysmorphic features found in chromosomal abnormalities that require prompt diagnosis in the perinatal period (e.g., Trisomy 13, 18, 21).
    - iii. Developmental delay or regression of milestones suggesting an underlying metabolic or genetic disorder.
2. **Medical Knowledge.** Understand the scope of established and evolving biomedical, clinical, epidemiological and social-behavioral knowledge in the area of medical genetics and genomics; demonstrate the ability to acquire, critically interpret and apply this knowledge in patient care.
- a. Recognize presenting symptoms, diagnose, describe the pathophysiology, and manage common presentations of genetic conditions.
    - i. Describe findings of common chromosome abnormalities including: Trisomy 21, Trisomy 18, Trisomy 13, Turner syndrome (45X), Klinefelter syndrome (47,XXY) and 47,XYY syndrome.
    - ii. Describe common deletion and duplication syndromes, and know how CNVs in general can affect phenotype.
    - iii. Describe findings of common single gene disorders such as neurofibromatosis, Marfan syndrome, achondroplasia, tuberous sclerosis complex, etc.
    - iv. Learn how to recognize disorders of the skeletal system through physical examination and X-ray analysis.
    - v. Learn disorders associated with orofacial clefting.
    - vi. Learn to diagnose and manage disorders of amino acid (e.g. PKU, branched chain amino acids – MSUD, urea cycle disorders), fatty acid (VLCADD, LCHADD, MCADD), carbohydrate metabolism, and other molecular diseases (Lysosomal storage disease, congenital disorder of glycosylation and peroxisomal biogenesis defect)
    - vii. Describe the findings and evaluation of a patient with a potential metabolic emergency.
    - viii. Know how to evaluate a child with a positive newborn screen.

- ix. Describe common patterns of Mendelian vs. non-Mendelian inheritance (autosomal dominant and recessive, X-linked, multifactorial, and the effect of maternal and paternal age).
  - x. Discuss unusual patterns of inheritance (mitochondrial defects, triplet repeat, imprinting).
- b. Differentiate disorders in patients associated with genetic predisposition or genetic disease from normal states or acquired disorders.
- i. Explain the findings on clinical history and examination that suggest a known or potential genetic disorder or inborn error of metabolism.
  - ii. Describe how well childcare differs in a child with a genetic condition, e.g., use of specific growth charts for specific conditions and physical findings.
  - iii. 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.
- c. Undifferentiated signs and symptoms. Evaluate, treat, and/or refer patients with the presenting signs and symptoms that suggest a genetic disease process.
- d. Laboratory Testing
- i. Biochemical Genetics – Learn the principles behind metabolite analyses including sample preparation and interpretation.
  - ii. Molecular Genetics and Cytogenetic - Learn the principles behind DNA and chromosome analysis including sample preparation, workflow and interpretation. Know how to navigate important databases for the interpretation of DNA sequence variants such as HGMD, ExAC, UCSD browser, ClinVar, ClinGen and OMIM.

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

- a. To participate in provision of genetic counseling to patients and or the patients' parents including: diagnosis, prognosis and recurrence risk (for the parents as well as for the child when he/she reproduces).
  - i. Participate in genetic counseling sessions.
  - ii. Talk to family members about sensitive issues that relate to a patient's genetic condition, e.g., coping with the child's altered needs in his/her home setting.
  - iii. Communicate effectively with physicians, other health professionals, and health related agencies to create and sustain information exchange and team work for patient care.
  - iv. Maintain accurate, legible, timely and legally appropriate medical records for patients in the outpatient and inpatient settings.

- v. Know how to provide pre and post test counseling for Gene Panels and Exome Sequencing, epigenetic abnormalities, dynamic mutations and mosaicism.
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.
- a. 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.
  - b. Identify the indicators that would lead the resident to seek a genetics consult.
  - c. Identify personal learning needs, systematically organize relevant information resources for future reference, and plan for continuing data acquisition if appropriate.
5. **Professionalism.** Demonstrate a commitment to carrying out professional responsibilities, adherence to ethical principles, and sensitivity to diversity.
- a. 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.
  - b. 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.
  - c. Demonstrate a commitment to carrying out professional responsibilities, adherence to ethical and legal principles, and sensitivity to diversity while providing care to children.
6. **Systems-Based Practice.** Understand how to practice quality health care and advocate for patients within the context of the health care system.
- a. Identify written and internet resources to aid in diagnosing a genetic or inborn error of metabolism, using physical findings along with laboratory examination.
  - b. Demonstrate sensitivity to the costs of clinical care in Medical Genetics and take steps to minimize costs without compromising quality.
  - c. Recognize the limits of one's knowledge and expertise and take steps to avoid medical errors.
  - d. Understand key aspects of health care systems as they apply to care of patients and their families, including cost control, billing and reimbursement.

- e. 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.

## **2. Cancer Genetics Rotation**

This is a one-month rotation directed by Dr. Vengoechea and Christine Stanislaw, CGC at the Winship Cancer Center and The Emory Genetics Clinic. While residents will have the opportunity to see cancer genetics cases throughout the resident's two years of training, this rotation will focus on cancer genetics. There will be no call during this rotation.

Monday – Clinical Case Conference 10:30 – 11:30; Plan schedule for the week with Dr. Vengoechea, Christine Stanislaw, CGC and Christine Tallo, CGC

Emory Clinic 1365 Clifton Road

Tuesday – Emory Clinic 1365 Clifton Rd

Wednesday – Emory Clinic 1365 Clifton Rd, Emory St. Joseph's or Emory Midtown

Thursday - Emory Clinic 1365 Clifton, Emory St. Joseph's or Emory Midtown

Friday - Emory Clinic 1365 Clifton

### **Goals and Objectives for Cancer Rotation**

**Patient Care** - Provide clinical care in genomics to patients/families who have or are at risk for a hereditary cancer syndrome.

1. Obtain and document a medical history that includes a detailed family history with particular attention to cancer or cancer-related findings in the family.
2. Provide genetic counseling for the most common hereditary cancer syndromes (e.g. HBOC, Lynch Syndrome) and for mid-penetrance cancer risk genes.
3. Work effectively with the multidisciplinary team to provide genetic services to the patient/family affected with cancer.

**Medical Knowledge** - Understand the scope of established and evolving biomedical, clinical, epidemiological and social-behavioral knowledge needed by a Medical Geneticist in the area of cancer genetics; demonstrate the ability to acquire, critically interpret and apply this knowledge in patient care.

1. Describe findings of common hereditary cancer syndromes including HBOC, Lynch Syndrome, FAP/AFAP and other syndromes that cause increased risk for breast, colon, and other cancer types.
2. Describe findings or more rare cancer and cancer-related conditions including Gorlin syndrome (basal cell nevus syndrome), Cowden syndrome, and Li-Fraumeni syndrome, etc.

3. Discuss current knowledge regarding the molecular basis of common hereditary cancer syndromes and more rare cancers and cancer-related conditions
4. Identify the current and future uses of DNA testing for genetic risk assessment of common hereditary cancer syndromes and more rare cancer and cancer-related conditions, including the distinction between high penetrance and mid penetrance risk genes.

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

1. Participate in/provide cancer genetic counseling sessions or common hereditary cancer syndromes and more rare cancer and cancer-related conditions.
2. Talk to the family members about sensitive issues that relate to the patient's/family's occurrence of cancer, e.g., coping with patient's/family's psychosocial needs relating to the cancer diagnosis and recurrence risk information
3. Communicate effectively with genetic counselors, physicians, and other health professionals and health related agencies to create and sustain information exchange and team work for patient care
4. Maintain accurate, legible, timely and legally appropriate medical records for the Cancer Genetics patients in the outpatient and inpatient setting

**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.

1. Develop strategies to learn about future advances in the understanding of the genetic basis of cancer, in order to incorporate into one's practice improved screening, identification, counseling and management of these disorders
2. Identify the indicators in a family affected by cancer that would lead the resident to seek a genetics consult
3. Identify personal learning needs, systematically organize relevant information resources for future reference, and plan for continuing data acquisition if appropriate

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

1. Discuss the ethical, legal, financial and social issues involved in genetic testing of patients/families at risk to develop cancer or a cancer-related condition, especially testing of children for status and providing medical care for patients with known fatal disorders.
2. Demonstrate personal accountability to the wellbeing of all patients, even when other physicians are primarily responsible for their care, e.g., by

following up on lab results, writing comprehensive notes, seeking answers to the difficult patient care questions, and communicating with primary care physicians

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

1. Identify written and internet resources to aid in counseling patients/families with cancer or risk to develop cancer including availability of research studies in which the patients/families might wish to participate
2. Demonstrate sensitivity to the costs of clinical care of Medical Genetics and take steps to minimize costs without compromising quality
3. Recognize the limits of one's knowledge and expertise and take steps to avoid medical errors
4. Understand key aspects of health care systems as they apply to care of patients and their families, including cost control, billing and reimbursement
5. Recognize and advocate for families who need assistance to deal with systems complexities, such as lack of insurance, long distance to the clinic, or need testing for relatives that live in other states.

### **3. Neurogenetics Rotation**

This is a one month, call free rotation. Residents will see patients in the TSC and NF clinics with Dr. Wolf, Neuromuscular Clinics with Dr. Verma, Huntington clinic with Amy Rosen, CGC, and dystonia/movement disorder clinic with Dr. Jinnah. Pediatric clinics are at the Center for Advanced Pediatrics. The Huntington and dystonia clinics are at The Emory Clinic. As soon as a month for this rotation is scheduled, individual times will be provided.

#### **Goals and Objectives for Neurogenetics**

**Patient Care** - Provide family centered patient care that is developmentally and age appropriate, compassionate, and effective for the treatment of neurogenetic disease and the promotion of health.

- a. Perform and document a thorough history and physical exam on a child or adult with a known or unknown neurogenetic disorder.
- b. Develop a management plan for commonly encountered neurogenetic disorders including choosing the appropriate tests (e.g. Know when to order a Next Generation Gene Panel vs. a Whole Exome Sequence).
- c. Identify resources in the resident's community for diagnosis, genetic counseling, therapy and psychosocial support of children and adults with neurogenetic diseases.

**Medical Knowledge** - Understand the scope of established and evolving biomedical, clinical, epidemiological and social-behavioral knowledge in the area of medical genetics and genomics; demonstrate the ability to acquire, critically interpret and apply this knowledge in patient care.



- a. Recognize presenting symptoms, know the differential diagnosis, describe the pathophysiology, and manage common presentations the following neurogenetic conditions.

Neuromuscular disease

Tuberous sclerosis complex

NF1 and associated Rasopathies

Dystonias and movement disorders

Huntington disease and other repeat associated neurogenetic disease

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

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

- i. Participate in genetic counseling sessions.
- ii. Talk to family members about sensitive issues that relate to a patient's neurogenetic condition, e.g., coping with the child's altered needs in his/her home setting. Palliative care.
- iii. Communicate effectively with physicians, other health professionals, and health related agencies to create and sustain information exchange and team work for patient care.
- iv. Maintain accurate, legible, timely and legally appropriate medical records for patients in the outpatient and inpatient settings.
- v. Know how to provide pre and post test counseling for Gene Panels and Whole Exome Sequencing.

**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.

- f. Develop strategies to learn about future advances in the understanding of neurogenetic disorders, in order to incorporate into one's practice improved screening, identification, counseling and management of such disorders.
- g. Identify the indicators that would lead the resident to seek a referral to one of the specialty neurogenetics clinics.
- h. Identify personal learning needs, systematically organize relevant information resources for future reference, and plan for continuing data acquisition if appropriate.

7. **Professionalism.** Demonstrate a commitment to carrying out professional responsibilities, adherence to ethical principles, and sensitivity to diversity.
- a. Discuss the ethical, legal, financial and social issues involved in genetic testing of children and adults for neurogenetic disorders that may present in adulthood, testing children for carrier status, and providing medical care for patients with known fatal disorders.
  - b. 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.
  - c. Demonstrate a commitment to carrying out professional responsibilities, adherence to ethical and legal principles, and sensitivity to diversity while providing care to children.

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

- d. Identify written and internet resources to aid in diagnosing a neurogenetic disorder using physical findings along with laboratory examination.
- e. Demonstrate sensitivity to the costs of clinical care and take steps to minimize costs without compromising quality.
- f. Recognize the limits of one's knowledge and expertise and take steps to avoid medical errors.
- g. Understand key aspects of health care systems as they apply to care of patients and their families, including cost control, billing and reimbursement.
- h. 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.

#### 4. Prenatal Genetics

This one month rotation is at Emory University Hospital Midtown 550 Peachtree St NE; 15<sup>th</sup> Floor. Martina Badell, MD ([mbadell@emory.edu](mailto:mbadell@emory.edu)) directs the rotation and is the resident's main contact. The resident will take no call during this month. If the resident can attend clinical conference and grand rounds remotely, they are encouraged to do so, but obtaining prenatal cases is the priority during this month. The schedule and goals and objectives follow:

<b>Monday</b>	Prenatal Clinic (PNC) 8am-5pm
<b>Tuesday</b>	PNC 8am-5pm
<b>Wednesday</b>	Grand Rounds 8-9am, PNC 9-12:00am, Didactic MFM conference 1-2pm
<b>Thursday</b>	PNC 8am-5pm
<b>Friday</b>	PNC 8am-5pm

## **Goals and Objectives for Prenatal Genetics**

### **1. Patient Care.**

- a. Provide genetic counseling, follow up care and management as appropriate to women and (when applicable) their partners. Develop empathy and understanding for pregnant women who have been determined to have a fetus with a genetic diagnosis and/or a congenital anomaly/anomalies
- b. Obtain an accurate history, construct and analyze a pedigree and recognize details relevant to genetic diagnoses
- c. Organize and implement appropriate laboratory testing and imaging studies for patients and fetuses with known or suspected genetic diagnoses
- d. Provide appropriate advice to care providers seeking consultation about work-up and management of genetic disorders and work effectively in a multidisciplinary setting
- e. Use prenatal diagnostic techniques appropriately, including amniocentesis and chorionic villus sampling (CVS).

### **1. Medical Knowledge:**

- a. Understand the scope of established and evolving biomedical, clinical, epidemiological and social-behavioral knowledge needed by the Medical Geneticists; demonstrate the ability to acquire, critically interpret and apply this knowledge in patient care:
- b. Demonstrate knowledge regarding the genetic conditions encountered in the prenatal setting:
- c. Describe Indications for and procedures for prenatal testing including screening and invasive testing
- d. Discuss the capabilities and limitations of prenatal testing, including prenatal array, quad screen, and NIPT.
- e. Discuss the definitions and implications of true vs. pseudomosacism detected by invasive testing
- f. Actively participate in the medical education of other learners, including medical students, residents & fellows from other disciplines and genetic counseling students

### **2. Interpersonal and Communications Skills.**

- a. Demonstrate Interpersonal and communication skills that result in information exchange and partnering with patients, their families and professional associates
- b. To participate in/provide prenatal genetic counseling to the pregnant woman and (when appropriate) her partner
- c. Participate in/provide prenatal genetic counseling sessions in commonly encountered parental genetic conditions such as AMA and positive screening tests for aneuploidy and NTD (NIPT, Quad Screen)
- d. Participate and provide counseling for more complex disorders such as Mendelian disorders and complex structural anomalies

- e. Talk to the patient and family (where appropriate) about sensitive issues that relate to potential/detected fetal anomalies/cancer risk and/or diagnosis, including family/psychosocial issues related to diagnosis and recurrence risk information
- f. 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
- g. Maintain accurate, legible, timely and legally appropriate medical records

**3. 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.

Develop strategies to learn about future advances in the understanding of prenatal genetic conditions in order to incorporate into one’s practice improved screening, identification, counseling and management of these disorders

Identify the indicators in a pregnancy that would indicate the need for a prenatal genetic consultation

Identify personal learning needs, systematically organize relevant information resources for future reference and plan for a continuing data acquisition if appropriate

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

Discuss the ethical, legal, financial and social issues involved in prenatal genetic testing including both screening and diagnostic testing

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

Demonstrate a commitment to carrying out professional responsibilities, adherence to ethical and legal principles and sensitivity to diversity while providing care to pregnant women and their partners who either have a fetus with an anomaly or genetic disease or who are at risk for having a fetus with an anomaly and/or genetic disease

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

Identify written and internet resources to aid in counseling pregnant women and their partners when appropriate who have or at risk for having a fetus with an anomaly or genetic disease including availability of research studies in which the patients/families may wish to participate

Demonstrate sensitivity to the costs of clinical care in prenatal medical genetics and take steps to minimize costs without compromising quality

Recognize the limits of one's knowledge and expertise and take steps to avoid medical errors

Understand key aspects of health care systems as they apply to care of pregnant women and their families, including cost control, billing and reimbursement

Recognize and advocate for pregnant woman who need assistance to deal with systems complexities, such as lack of insurance, transportation difficulties

## **5. Clinical Laboratory Rotations.**

Due to a major restructuring of the Emory Genetics Laboratory, rotations will be done at an outside laboratory. We are currently in negotiation with Greenwood Genetics Laboratory and hope the 6 weeks of laboratory training will occur there. The Emory Dept. of Human Genetics will pay for housing for this away rotation.

### **Goals and Objective for Laboratory Rotations**

#### **Patient Care**

- Identify key elements of laboratory testing (turn-around time, collection tubes, test ordering, pre-analytic, analytic, and post-analytic phases)
- Analyze laboratory data with assistance
- Describe clinical indications for testing
- Recognize the importance of clinical history in interpreting laboratory results
- Recognize role of CLIA in governing clinical laboratory testing, and distinguish between clinical and research testing.
- Describe situations in which laboratory reports may be amended (changes in variants status, etc.).
- Recognize variability in test results reporting among different laboratories.
- Propose additional testing based on test results and/or patient history

#### **Genetics Knowledge**

- Recognize major disease categories and testing approaches
- Describe the basic principles of laboratory testing relevant to the test being requested
- Differentiate methodologies utilized in the laboratory (including strengths and limitations; multiplex analysis vs. single metabolite/targeted testing)
- Recognize, interpret and apply basic elements of laboratory reports (i.e., metabolites and nomenclature)
- Describe ethical issues surrounding genetics testing

#### **Interpersonal and communication skills**

- Recognize the importance of effective communication between all members of the health care team (including laboratory)
- Use good communication skills when interacting with other members of the health care team

### **Practice-based Learning and Improvement**

- Describe basic elements of laboratory safety
- Recognize relevant aspects of laboratory quality control and quality assurance
- Employ available databases and other online resources

### **Professionalism**

- Assist other members of the health care team to optimize patient care
- Adhere to professional responsibilities
- Apply ethical practices

### **Systems-based Practice**

- Identify available professional guidelines and standards for laboratory practice
- Recognize test cost, utilization and effect on patient management (including tests not performed at the institution of care)

## **6. Scholarly Activity**

Each resident must demonstrate scholarship through submission of at least one scientific presentation, abstract, or publication. This is an ACGME requirement.

## **7. Self-Directed Learning**

Residents will have 3 months of self-directed learning that can be in any aspect of medical genetics. This is a good time to explore required scholarly activity in the form of a case report, case series, or even a project with one of the faculty. Some examples of self-directed learning include: A week at the Georgia NBS Laboratory, Manuscript/abstract/poster preparation, electives in pediatric neuroradiology, pediatric hepatology, pediatric pathology, and other clinical experiences.

## **V Supervision Policy**

The Department of Medical Genetics follows the ACGME requirements regarding resident supervision. These policies are distributed to all supervising attending physicians.

## **VI Outpatient Activities**

The Medical Genetics Resident will be under the supervision of an attending physician for all patients he/she evaluates and treats through the Medical Genetics and Genomics Training Program. The supervision in the outpatient clinic will always be **Indirect Supervision, with Direct Supervision immediately available**. During the first year of residency, the resident will be asked to complete a full history and physical examination for each patient he/she is assigned. The history/physical does not need to be completed under the direct observation of an attending physician, unless the attending physician deems it necessary after deficiencies have been found. The resident shall then present the findings to the attending physician and formulate a differential diagnosis and plan for testing. The attending physician will then do an independent physical examination of the patient in front of the resident. During the second year, the resident will be

given more independence and the opportunity to perform his/her own counseling with the patient, while under the direct observation of the attending physician. The resident is expected to provide medical documentation for each patient he/she evaluates in clinic. This documentation will be reviewed and signed by the attending physician. All laboratory/imaging study results from the patient encounter will also be reviewed by the attending physician and the treatment plan / plan for further diagnostic tests will be discussed with the resident prior to any further discussion between the resident and the patient. All notes must be completed within 24 hours.

## **VII Inpatient / Call Activities**

For inpatient consultations or on call telephone questions the supervision will be **Indirect Supervision with Direct Supervision available**. For nighttime home call, the resident discusses a plan with an MD or parent, then calls genetics attending and discusses and amends plan as necessary. All nighttime calls to resident on call are discussed with attending for both years of training. All notes must be completed with 24 hours.

## **VIII Duty Hours Policy**

It is the policy of the Medical Genetics Training Program to follow guidelines established by the ACGME (see <http://acgme.org>) regarding duty hours for residents in an accredited training program. As we are not an admitting service, but strictly an inpatient consultative service with primary outpatient clinical responsibilities, we have no in-house call. All after-hours call responsibilities are from home. Each resident takes call for an average of one week out of four.

- Residents must not work more than 80 hours per week averaged over a month,
- Residents must receive at least one day in seven away from clinical duties.

Each resident is required to keep a time card consisting of their work hours over a continuous 4 week period two times per year as part of the duty hours monitoring system.

The Program Director must provide information to residents, fellows, and faculty members regarding effects of loss of sleep and chronic fatigue. Currently, the GMEC recommends the using SAFER program available from the GME Office, the Dinges presentation found at the ACGME website, and asking faculty members of Emory University School of Medicine who have expertise in this area.

## **IX. Clinical and Research Operation Meetings**

The resident is required to attend a monthly one hour monthly clinic operations meeting, and a one hour monthly clinical research meeting. These will occur on alternate Mondays when there are no scheduled Grand Rounds. This is an excellent opportunity to take part in quality improvement, hear the resident's thoughts regarding operations, learn from the genetics team, and provide suggestions for quality and practice improvement.

## X. Newborn Screening

Emory Medical Genetics has the newborn screening (NBS) follow-up contract for the state of Georgia through the Georgia Department of Public Health (DPH). All positive screens are funneled through a group of nurses coordinated by Director Angela Wittenauer MSN, FNP-C, RN. Emory maintains a NBS database of all screened babies in Georgia. You will be granted access to this database. Once you have your Emory email, send an email to Angela ([alwitte@emory.edu](mailto:alwitte@emory.edu)) explaining who you are and she will facilitate your access to the database. Arrange, by email, to spend an hour or so with Angela who will help you navigate the NBS database. You should also submit a ticket at [help.emory.edu](http://help.emory.edu) and request that you be added to the metabolic distribution list: EUHG\_DOMG\_METAB. All positive screens related to inborn errors that require diagnostic testing are sent to the distribution list. When you are on call, you will receive many calls from care givers from around the state about follow-up. Your access to the database as well as inclusion in the EUHG\_DOMG\_METAB list will be indispensable. Often you can search you email for the child's name and you will find it in a EUHG\_DOMG\_METAB email. If not you can access the database. **AS ALWAYS, YOU SHOULD REVIEW ALL PLANS REGARDING NBS WITH THE ATTENDING ON CALL**, especially during the first 6 months. Not uncommonly, if you access the database, you will find the follow up recommended by the nurses. This can simply be reiterated to the caregiver. When you discuss a case with a caregiver, you should always document your conversation in the NBS database. Angela and the attendings will show you how.

## XI. IMPORTANT NUMBERS

- |                                                                                                                                                         |                                   |
|---------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------|
| a. Dr. Gambello                                                                                                                                         | 404-727-6483<br>Cell 713-516-0049 |
| b. Liz McKenna                                                                                                                                          | 404-727-3067                      |
| c. The Emory Clinic - Medical Genetics Clinic                                                                                                           | 404-778-8570                      |
| d. The Emory Clinic FAX                                                                                                                                 | 404-778-8562                      |
| e. Jessica Cruz for Special Appointment Requests                                                                                                        | 404-778-8524                      |
| f. If a PEDIATRICIAN/PEDIATRIC SPECIALIST wants the geneticist on call they should call the transfer center at CHOA (Children's Healthcare of Atlanta). | 404-785-7778                      |
| g. If a patient wants to page a geneticist<br>The PIC system is an EMORY based system                                                                   | 404-686-5500 PIC 50263            |
| h. Emory Genetics Laboratory                                                                                                                            | 404-778-8499                      |
| i. Emory Medical Labs (EUH)                                                                                                                             | 404-712-5227                      |
| j. Children's Egelston General number                                                                                                                   | 404-785-6000                      |



**PAGE CODE – Often the resident will be paged to a 5 digit number. This should help DECODE a 5 digit page.**

<b>2-1234</b>	<b>404-712-1234</b>	<b>Emory Univ.</b>
<b>3-1234</b>	<b>404-943-1234</b>	<b>PICU/CICU Phones</b>
<b>4-1234</b>	<b>404-565-1234</b>	<b>CHOA Voalte</b>
<b>5-1234</b>	<b>404-785-1234</b>	<b>CHOA</b>
<b>6-1234</b>	<b>404-686-1234</b>	<b>EUHM</b>
<b>7-1234</b>	<b>404-727-1234</b>	<b>Emory Univ.</b>
<b>8-1234</b>	<b>404-778-1234</b>	<b>TEC/EHC/Emory U</b>

**XII. Evaluations**

After each rotation, residents will be evaluated by attending(s), genetic counselor(s), dietitian(s) and nurses. They will use template evaluations appropriate for the specific rotation. For example, for the general genetics/metabolism rotation, attending physicians would fill out three forms, the Adult Genetic Evaluation, Pediatric Evaluation and the Metabolism evaluation. Dietitians would only fill out the metabolism evaluation. Residents are provided with all the evaluation forms.

The Clinical Competency Committee (CCC), is composed of:

1. Hong Li, MD, PhD
2. William Wilcox, MD, PhD
3. Rossana Sanchez, MD
4. Jaime Vengoechea, MD
5. Stephanie Wechsler, MD
6. Juanita Neira, MD
7. Emily Black, MD

This committee will evaluate each resident every six months using the ACGME Medical Genetics Milestones. The CCC will use rotation evaluations and any other means that help them identify progress and deficiencies. Residents are given the milestones.

The Program Evaluation Committee will include the above members but will also include residents. This group will evaluate the program yearly and identify areas for improvement.

1. Hong Li, MD, PhD
2. William Wilcox, MD, PhD
3. Rossana Sanchez, MD
4. Jaime Vengoechea, MD
5. Charlotte Peinhardt, CGC
6. Emily Black, MD
7. Resident(s)

### **XIII. OTHER REQUIREMENTS:**

1. **Clinical Conference.** Occurs Tuesday mornings at 8:00 AM except during summer months (June, July, August) in WH300. Residents are expected to present at this conference once a semester. Drs. Li or Vengoechea will assign the resident times to present. The presentation will be 25-30 minutes, and include a succinct history and pertinent physical examination. Occasionally, photographs of the patient are shown (with permission). Then a brief differential diagnosis, followed by a diagnosis (if made), and a discussion of the disorder. There will be a faculty mentor assigned to the resident's clinical conference time. The resident must review the presentation with the faculty mentor as well as generate learning objectives. This **MUST** be done a week in advance of the actual conference in order for the conference to qualify for CME.
2. **Medical Genetics Grand Rounds:** Mondays during the academic year from 8:00-9:00 in the WH300. Residents will attend all grand rounds. If the resident is at a remote location, they can log into webex/Zoom. Residents will be required to present one grand rounds presentation a year at the end of the academic year. Topics should be discussed with Dr. Gambello or one of the other attendings involved in the case.
3. **Metabolism Rounds:** Occurs 9:00-10:00 WH300. The resident should prepare to see patients by reviewing the biochemical defect. There is a textbook called Atlas on Inherited Metabolic Diseases. Read the pertinent chapters in that book. The following weeks the resident will also present follow-up on the patients she saw in clinic the week before.
4. **Emory Genetics Laboratory Rounds.** Excellent educational session on Thursdays 12:30 – 1:30. These are at EGL in Tucker and currently the residents will only go to these during their laboratory rotations. The Department of Human Genetics is working on having these sessions teleconferenced.
5. **Journal Club** - This occurs on the last Tuesday of every month from 8:00-9:00 instead of clinical conference. Drs. Li and Gambello will give the resident details.

## **XIV. COURSE WORK**

There are several required courses during the first year of training.

HGC 715 Human Genetics

Fall Semester Monday and Wednesday 2:15 – 3:45

Course Director – Kate Garber, PhD

HGC 745 Medical Genetics      Fall and Spring Semesters

Fall Tuesday 3-5 PM Spring TBD

Course Director – Stephanie Wechsler,

The syllabus for each class will be provided by the instructors. These courses take priority over everything else except a critically ill patient.

## **XIV. Books - Internet Resources**

**Gene Reviews - Usually the most up to date resource**

**OMIM**

**UCSC Browser**

**Smith's Recognizable Patterns of Human Malformation**

**Management of Genetics Syndromes 2nd edition**

**Emery and Rimoin's Principles and Practice of Medical Genetics 6th Edition - There is an electronic copy in the Books folder on the clinical drive**

**Atlas of Metabolic Diseases      William L. Nyhan, Bruce , Bruce A. Barshop, Aida I Al-Aqeel**

**Inborn Metabolic Diseases 5th Edition Jean-Marie Saudubray, Georges van den Berghe, John H. Walter - There is an electronic copy (4th edition) in the Books folder on the clinical drive**

## **XV. MEETINGS/National Course**

The DOMG is committed to each resident's professional development. Financial support is provided for the following

1. The SIMD North American Metabolic Academy. This course is usually given in October and pre-registration is a must. Submit an application in JULY of the first year (second year is also acceptable). See the following website: <http://simdnama.org/>
2. The American College of Medical Genetics has its annual meeting in March or April. We expect the resident to submit an abstract.

3. The American Society of Human Genetics has its annual meeting in October.

The DOMG does not provide funding for Board Exams.

#### **XVI. ACGME and ABMGG Logbook/Patient Tracking**

It is imperative the resident enter the number of cases in several categories into the ACGME Resident Case Logs System. The data provided to ACGME is used for accreditation, maintained confidentially, and is not distributed for commercial use. Liz will assist in gaining the resident access to the database. The resident should maintain a separate log of ALL cases. They should be aware of the types of cases that are required for the ABMGG logbook so they can schedule patients accordingly.

#### **XVII. Encouraging a Healthy Lifestyle**

The Division of Medical Genetics and the MGG training program encourages a healthy lifestyle for all residents, faculty and support staff. The resident is encouraged to explore the Emory School of Medicine Resident Wellness website (<https://med.emory.edu/education/gme/wellness/index.html>). There are a variety of modules and workshops that address time management, Sleep hygiene, and other areas of wellness. The program understands that life can present many unexpected stressors during training. We are committed to working with our residents so that they can maintain a level of training while providing the flexibility of accommodating life's curve balls. Resident are encouraged to discuss any issues with Dr. Gambello who is available at any time (713-516-0049).