

The Goals and Objectives for the UAB Medical Genetics Residency Program
[Descriptions of the each rotation, including expectations, requirements, and
evaluation methods]

Overview

The Goals and Objectives for each educational activity of the UAB Medical Genetics Residency Program are presented below. The goal and objectives are presented in a manner to correlate with the 6 ACGME core competencies for each activity, along with the activity in which the competency will be mastered, and the evaluation method for each competency.

Graduated expectations

As their level of proficiency for a given skill or competency increases during the course of their training, we will have increased expectations for a resident's performance and involvement in a given case or activity. We expect to see a higher level of functioning as resident's gain experience, which leads to an increasing independence. Residents are expected to gain increasing competency in the various activities associated with the clinical practice of medical genetics, as outlined in the ABMG Milestones (Appendix 2)

Level 1: The resident demonstrates milestones expected of an incoming resident.

Level 2: The resident is advancing and demonstrates additional milestones, but is not yet performing at a mid-residency level.

Level 3: The resident continues to advance and demonstrate additional milestones, consistently including the majority of milestones targeted for residency.

Level 4: The resident has advanced and substantially demonstrates the milestones targeted for residency. This level is designed as the graduation target.

Level 5: The resident has advanced beyond performance targets set for residency and is demonstrating "aspirational" goals which might describe the performance of someone who has been in practice for several years. It is expected that only a few exceptional residents will reach this level.

It is expected that the progress towards independence will occur during both each rotation, and the entire residency training period. This expectation is also reflected in their **evaluations**, as each resident is judged not on an absolute scale, but with respect to their level of training and experience.

NOTES:

1. The clinic schedule is not listed here, as it is constantly changing. Please contact Program Coordinator Sandra Pilkinton for the current schedule.
2. While it is referenced in the rotation requirements, we do not list the required reading as it is updated regularly. Please see the rotation supervisor for the updated list.

GENERAL GENETICS (including general and specialty clinics and inpatient consultation) (NH Robin)

The goals and objectives listed in this section include the overarching core competencies involved in clinical in-patient and outpatient clinical patient care in medical genetics. These represent core competencies, as the skills are also common to other areas of the clinical practice of medical genetics, such as Prenatal Genetics and Cancer Genetics rotations.

Description and Requirements

The **General Genetics Outpatient** rotation consists of 3 one-month blocks during which the resident will focus on outpatient clinical activities, including both general clinics as well as specialty clinics. This includes increasing the number of resident continuity clinic from one half-day clinic twice per month to 1 half-day clinic per week.

The current clinic schedule should be reviewed and the resident should notify the faculty (physician and/or genetic counselor) associated with the clinic that they will be attending the specific clinics that month.

Expectations for Outpatient clinic rotation

- A. It is expected that the resident will attend *at least* 5 clinics per week, including 1) faculty genetics clinic (general and specialty); 2) their own continuity clinic (one per week). The resident should take primary responsibility for at least 2 patients per clinic. This includes a) performing a complete intake, including personal and family history with a 3-generation; b) performing a complete physical examination; and c) generating an impression/differential diagnosis and plan to be presented to the attending. Note: the expected level of competency will vary depending on the timing of training (eg, a new resident will not be expected to generate the same differential diagnosis and plan as a 2nd year resident)
- B. The resident should prepare 1-2 patient to be presented at clinic conference
- C. Complete recommended reading

The genetics faculty and staff participate in several **Multidisciplinary and Specialty clinics**. The resident must attend at least a specified number of these clinics during their training (to be documented in the Learning Portfolio).

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| - Biochemical Genetics (12) | - Tuberous Sclerosis (2) |
| - Cleft Clinic (6) | - Neurogenetics (2) |
| - Craniofacial Clinic (3) | - Skeletal dysplasia/connective tissue (2) |
| - Neurofibromatosis (3) | - Cystic Fibrosis (1) |
| - Marfan clinic (3) | - Medical Autism (1) |
| - Dental Genetics (2) | - Huntington (1) |
| - Adult Down syndrome (2) | - Muscular dystrophy (1) |

Consultation

There are 6 one-month **Inpatient Consultation** months. During these months the resident is responsible for staffing all requests for inpatient consultation, including both clinical genetics and metabolism services. An attending physician will always accompany the resident to consultations, but it is expected that the resident will progress in their level of independence. During these months, if time permits the resident should attend the various specialty clinics. However, the responsibilities of the in-patient service **always** take precedence over attending these clinics.

Goals and Objectives for the General Genetics and Consultation rotations

Competency 1 - Patient Care. Provide effective and compassionate clinical care patients and their families who have a genetic disorder, or at risk for having a genetic disorder.

GOAL: To be able to evaluate, diagnose, and manage with known or suspected genetic disease in children and adults in both the in-patient and out-patient setting.

Educational Objectives

1. Obtain and document a detailed medical, prenatal, and family history, and perform a complete physical/ dysmorphic exam
2. Identify the most important and relevant information
3. Order appropriate laboratory tests
4. Generate an appropriate differential diagnosis and plan
5. Provide appropriate counseling and education for patients/families, including identifying other at-risk family members
6. Communicate effectively with referring physicians and other healthcare

Competency 2 - Medical Knowledge. Recognize the extent of established and evolving biomedical, clinical, and epidemiological and social-behavioral sciences, and apply this knowledge to patient care.

GOAL: To be able to research, learn, and apply biomedical information to patient care.

Educational Objectives

1. Apply knowledge of anatomy, development, pathophysiology, natural history, clinical history, and inheritance to provide counseling, anticipatory guidance, and longitudinal management to patients with multisystem genetic disorders
2. Learn the common presenting signs for the various forms of metabolic disorders, including disorders of amino acids, organic acids, energy metabolism, and storage diseases. This should occur not only through personal interaction, but in phone consultation with known and potentially new patients
3. Able to identify appropriate therapeutic interventions, in both acute situations as well as long-term dietary management issues
4. Identify recent advances in genetic discovery (eg, new tests, treatments), and utilize these in patient care
5. Identify research-based studies or tests that may benefit the patient and/or family

Competency 3 – Practice-based Learning and Improvement. Demonstrate the ability to continually enhance their personal fund of clinical knowledge and clinical skills through ongoing learning as well as integrating new discoveries and advances, and to be able to critically review and analyze these discoveries and advances.

GOALS: 1) To demonstrate the ability to continue to learn through critical review and independent study; 2) to continue to enhance clinical skills and judgment through critiques of mentors and self-assessment; 3) to use new information and improved clinical skills in patient care.

Educational Objectives

1. Identify strengths, deficiencies, and limits in one's knowledge and expertise
2. Identify and perform appropriate learning activities
3. Demonstrate a willingness to seek out assistance when appropriate
4. Critically read and interpret scientific literature

Competency 4 – Interpersonal and Communications Skills. Display interpersonal and communication skills that result in information exchange, and partner with patients, their families and other healthcare professionals.

GOALS: 1) To provide compassionate, accurate, and informative genetic counseling to a patient and/or family, educating them on the diagnosis, prognosis and recurrence risks; 2) to communicate effectively with other members of the healthcare team.

Educational Objectives

1. Provide effective and informative genetic counseling in all situations
2. Communicate effectively with all members of the healthcare team
3. Generate informative clinical documentation

Competency 5 – Professionalism. Exhibit a commitment to carrying out professional responsibilities, adherence to ethical principles, and sensitivity to patients of diverse backgrounds

GOALS: 1) To conduct all activities in a professional manner to both patients, professional staff, and colleagues; 2) to act always in accordance with ethical principles; and 3) to demonstrate respect to patients of diverse cultures, socio-economic backgrounds, and religions.

Educational Objectives

1. Exhibit professional behavior, including promptly responding to messages, avoiding unexcused absences, being available when on-call or at assigned clinics
2. Be aware of and follow the ethical, legal, and social issues concerning genetic testing for a) children (eg, testing children for adult onset diseases and for carrier status) ; presymptomatic testing for cancer and neurodegenerative diseases in all ages
3. Respects patient/family's emotional state, cultural and/or educational background
4. Respond in a positive manner to both positive and negative feedback
5. Respect the barriers between research and clinical care

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

GOAL: Identify and seek to lessen the challenges a patient may face in dealing with the healthcare system

Educational Objectives

1. Identify written and internet resources for patients/families
2. Make appropriate use of referrals to other physician, health care professionals, and other resources (eg, social work, educational resources and mental health professionals)

3. Recognize and try to minimize the financial and personal burden of a genetics evaluation
4. Advocate for families who need assistance to deal with the healthcare system