

MEDICAL GENETICS RESIDENCY

CLINICAL GENETICS ROTATION—UNIVERSITY OF WASHINGTON CLINICS

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INTRODUCTION

Clinical geneticists are physicians who have completed a medical genetics residency, after completing two or more years of a primary specialty such as internal medicine, or pediatrics. The rapid pace of scientific discovery is improving the ability to diagnose, manage, treat and prevent genetic conditions, and this is reflected in an increasing demand for genetic services. New opportunities include genetic testing for cancer predisposition, expanded newborn screening, pharmacogenetic testing, and genomic analysis, including whole exome and whole genome sequencing.

Medical genetics encompasses essentially all of medicine, because all organ systems manifest genetic conditions. Relevant skills common to all of genetics include: history taking, risk assessment physical examination, communication skills, and interpretation of genetic testing. Special areas of expertise in the UW Genetic Medicine include the following: Connective tissue disorders, cancer genetics, neurogenetics, dermatologic genetics, autism genetics, and cardiac genetics.

GOAL

The goal of the Medical Genetics rotation at the University of Washington Genetic Medicine Clinics is to help residents to understand the clinical spectrum of genetic disorders that manifest in adults, to recognize when to refer patients for genetic consultation, to provide them with the tools necessary to make genetic diagnoses and to provide families with appropriate genetic counseling, referral and follow-up medical care, and to understand the ethical, privacy, social and psychological issues affecting genetic testing.

OBJECTIVES

The objectives are presented below, organized by the general competencies defined by the Accreditation Council for Graduate Medical Education (ACGME) as expected of all residents.

Patient Care

Residents will demonstrate ability to:

- Gather appropriate and accurate family history and clinical information and reduce these to a correct family pedigree
- Perform all the necessary elements of physical examination for the diagnosis and differential diagnosis of defined genetic disorders
- Explain the mode of inheritance, natural history, availability of prenatal diagnosis, availability and efficacy of interventions in the natural history of the disorder, and expected prognosis of specific genetic diagnoses in language that is appropriate to family background and education
- Determine the appropriate routine and specialized genetic testing for individuals at risk for, thought to have, or clearly affected with a genetic disorder
- Make appropriate referrals to medical, social, or peer group agencies
- Interpret laboratory test results within the clinical context
- Use clinical decision-making concepts and techniques in interpreting results
- Transmit information about the disorder to the family and referring physician in written and verbal form and, when done in person, assess the effect of the interaction and to assist in anticipating and dealing with any emotional consequences

Medical Knowledge

Residents will demonstrate:

- Knowledge of genetic conditions that manifest in adult life, their natural history, molecular basis when known, and current literature concerning recent advances in knowledge, prevention, and treatment
- Knowledge of mutational mechanisms
- Knowledge of the characteristics and limitations of major laboratory techniques used for genetic testing for these disorders
- Ability to use a variety of resources to investigate clinical genetics questions
- Knowledge and understanding of the concepts of Mendelian and “non-Mendelian” modes of inheritance, penetrance, expressivity, and genotype/phenotype correlation
- Development of a personal strategy to regularly maintain and update medical knowledge
- Knowledge of community resources available to patients and their families, including peer groups
- Knowledge of the principles of genetic counseling

Practice-Based Learning and Improvement

Residents will demonstrate:

- Ongoing identification and remediation of gaps in personal medical genetics knowledge

Interpersonal and Communication Skills

The resident will demonstrate:

- Ability to communicate clearly and effectively with patients and their families, with other clinicians, genetic counselors, and other medical personnel involved in the care of their patients and families
- Ability to use appropriate modes of communication (direct, telephone, e-mail, written)
- Ability to prepare and deliver effective presentations about families and about specific disorders

Professionalism

The resident will demonstrate:

- Knowledge and understanding of ethical, privacy, social and psychological issues affecting genetic testing
- Maintenance of confidentiality of patient information
- Respectful behavior towards all patients and medical personnel
- Dress professionally, including wearing an ID badge.
- Dictating a complete clinic note within 24 hours of the clinic visit to document the history, family history, physical exam, labs, assessment and plan as discussed with the attending for review/signature by the attending.
- Prompt and courteous response to all pager and telephone calls
- Regular, punctual attendance, and participation in conferences and meetings and rotation responsibilities

Systems-based practice

The resident will demonstrate:

- Understanding of the role of the laboratory in the health care system, and the importance of reliable, cost-effective and timely laboratory results in clinical decision-making
- Ability to work with clinicians, administrators and others to determine the role of the laboratory in specific situations to optimize patient outcomes

RESPONSIBILITIES

Orientation to Clinics

Residents should contact Dr. Hisama, or if not available, then Robin Bennett, M.S., C.G.C., the clinic co-director at robinb@u.washington.edu a few days before beginning the clinic rotation. The resident will receive a schedule and information about the clinic by email prior to starting the rotation. The initial orientation to the physical areas and key personnel will be at the first Clinic in which they are scheduled. This will be at 8am if the first clinic is scheduled on Tuesday in the Center for Human Development and Disabilities (CHDD, ROOM 246) and 8am if the first clinic is scheduled on Thursday, in the Medicine 8SE East Clinic of UWMC.

Clinic Responsibilities

Residents will:

- Contact Dr. Hisama one to a few days prior to each clinic to determine which families they will see in the next clinic and to discuss known genetic issues with the genetic counselor that has contact with the family
- Review the written information assembled concerning each patient and family to be seen
- Discuss the diagnostic, genetic, psychosocial and counseling issues with the attending physician and determine if more information is needed
- Review current and other important literature about the condition anticipated or known to exist in the family prior to seeing the family
- See and evaluate the family or patient in the clinic— determine the reason for the visit, complete and check the family and social history provided, confirm the history and presentation as appropriate, perform the appropriate physical examination, and outline the issues to be discussed with the family
- Present the family to the attending physician and discuss the counseling to be provided or determine whether additional information needs to be collected
- Counsel the family with the attending physician, as decided during the presentation
- Determine what follow-up medical procedures or genetic testing need to be performed and obtain the appropriate samples with necessary consent
- Dictate the clinic note on the day of clinic (always required for documentation) and prepare a letter to the family (if needed) by the following day (later if agreed with the attending physician because of pending results)
- Edit and correct both through the electronic facilities available at the hospital within 24 hours of posting
- Prepare a concise presentation of the family and genetic condition for the clinic conference
- Communicate with referring physicians in a timely fashion regarding the results of the evaluation in clinic

Weekly Schedule and Location of Clinics, and Didactic Activities.

MONDAY: No scheduled activities. Preparation and reading for clinic patients that week.

TUESDAY: Clinic 8A-5P CHDD building. Meet in Room 246.

WEDNESDAY: Medical Genetics Journal Club. 8:30A-9:30A. K-250. Presentation of recently published articles.

Clinic Case Conference 9:30A-10:30A K-250. Weekly presentation of selected case(s). May also include update on pathogenetics of the disease.

THURSDAY: Clinic 8A-3P UWMC. 8SE. Take Cascade elevators to 8th floor, turn right, go to second desk and ask for physicians' work room.

FRIDAY: Medical Genetics Seminar 12:30P-1:30P K-069.

METHODS USED IN EVALUATING RESIDENT AND PROGRAM PERFORMANCE

At the end of the rotation, the resident is evaluated in writing and their performance reviewed with them verbally by every attending and fellow he or she has interacted with for a significant amount of time. The evaluator rates the resident on a nine-point scale in each component of clinical competence (i.e. patient care, medical knowledge, practice based learning improvement, interpersonal and communication skills, professionalism, system based learning, educational attitudes, leadership, overall clinical competence).

The resident is given the opportunity to evaluate in writing the quality of the curriculum and the extent to which the educational goals and objectives of the rotation have been met. The resident also evaluates the teaching competence of each attending, genetic counselor, and fellow with whom s/he has interacted for a significant amount of time.

REFERENCES FOR THE ROTATION

Bennett RL, French KS, Resta RG, Doyle DL. [Standardized human pedigree nomenclature: update and assessment of the recommendations of the National Society of Genetic Counselors.](#) J Genet Couns. 2008 Oct;17(5):424-33.

Riley BD, Culver JO, Skrzynia C, Senter LA, Peters JA, Costalas JW, Callif-Daley F, Grumet SC, Hunt KS, Nagy RS, McKinnon WC, Petrucelli NM, Bennett RL, Trepanier AM. Essential Elements of Genetic Cancer Risk Assessment, Counseling, and Testing: Updated Recommendations of the National Society of Genetic Counselors. [Genet Med.](#) 2007 Jun;9(6):385-9.

Pletcher BA, Toriello HV, Noblin SJ, Seaver LH, Driscoll DA, Bennett RL, Gross SJ. [Indications for genetic referral: a guide for healthcare providers.](#) Genet Med. 2007 Jun;9(6):385-9.

<http://uwresidents.com> Under Programs, select Medical Genetics

<http://www.genetests.org>

<http://ashg.org> The American Society of Human Genetics

<http://acmg.net> The American College of Medical Genetics