660F CLINICAL GENETICS AND BIRTH DEFECTS

This rotation is not accepting international students

Course Description: The genetic rotation at different sites is designed to teach students about a range of genetic disorders, genetic diagnostic testing as well as genetic counseling by participating in the evaluation of children and adults in different clinical settings. The students will also learn how to perform a basic dysmorphology examination and elicit a comprehensive family history and construct a 3-generation medical pedigree

Site: CHOC Children's Hospital of Orange County
Site: Miller's Childrens Hospital

Site: UC Irvine/ Millers/CHOC

Course Director UCI: Maureen Bocian MD Email: mebocian@uci.edu

Course Description: Inpatient consultations. Outpatient clinics in pediatric genetic disorders and congenital anomalies, metabolic genetics, prenatal genetics, adult genetics, and cancer genetics. Weekly patient management/teaching conferences and formal lectures. Learn to obtain history and pedigree, analyze medical records, perform dysmorphology examinations, establish diagnosis and mode of inheritance, provide information about diagnosis, prognosis, and recurrence risk to patient/family. Understand genetic testing and informed consent; treatment and preventionoptions; genetic counseling; psycho-social and ethical issues.

Department: Pediatrics, UCI Division of Genetic and Genomic Medicine

Prerequisite: UC Irvine Students must have successfully completed their basic science course work and the Pediatrics Clerkship. Extramural students must be in their final year of undergraduate medical education and have completed the Pediatrics Clerkship. It is advantageous, but not required, for all students to have completed the Ob/Gyn, Medicine, and Neurology clerkships.

Instructing Faculty: Maureen Bocian, MD; Elizabeth Chao, MD; Pamela Flodman, MSc, MS; Natalie Gallant, MD; J. Jay Gargus, MD, PhD; June-Anne Gold, MD; Katherine Hall, MS; Meredith Jones, MS; Virginia Kimonis, MD; Deepika Nathan, MS; Kathryn Singh, MPH, MS; Moyra Smith, MD, PhD, MFA; Kathryn Steinhaus-French, MS; Maya Thangavelu, PhD; Valerie Watiker, MD; Michael V. Zaragoza, MD, PhD.

Course Website: None

Who/Where to Report on the First Day: UCI Pediatrics academic offices, Suite 800, The City Tower, 333 City Blvd. West, Orange, CA 92868; Tel: 714-456-7570. Report to Dr. Bocian at 8:00 AM. Please email the week before the elective starts for information updates: mebocian@uci.edu

Site UCI rotation: Patients are seen at UCIMC, UCI Gottschalk Clinic, Miller Children's, and CHOC

Site Coordinator: Frank Cruz fcruz@uci.edu (714)456-5650
Periods Available: Throughout the year except the last two weeks of December and during major Genetics conferences (please contact us to check before signing up - these change every year - they are usually in mid-March and October, but dates vary)

Duration: Minimum of 4 weeks

Number of Students: 1 or 2

Scheduling Coordinator:

- UC Irvine students please email comsched@uci.edu or call (714) 456-8462 to make a scheduling appointment. UCI genetics also requires prior approval please contact Dr. Bocian before scheduling.
- Non-UCI students—Please read the following information carefully:
  - Any student enrolled at a U.S. LCME medical school will use VSAS to apply. To apply, please refer to this website: http://www.aamc.org/programs/vsas/
  - Students from international schools which have an existing agreement with UC Irvine

Course Objectives: At the end of this rotation the student will be able to:

- Explain basic concepts regarding single-gene, chromosomal, multifactorial/polygenic, mitochondrial, and non-traditional patterns of inheritance in a manner easily understood by patients
- Elicit a comprehensive, multi-generational family medical history, construct an appropriate 3-generation pedigree medical pedigree, and recognize patterns of inheritance and other signs suggestive of genetic disease in the family
- Recognize features in a patient's medical history, physical examination, or laboratory results that suggest the presence of genetic disease; identify patients with strong inherited predispositions to common diseases and facilitate appropriate assessment of other at-risk family members; identify individuals and families who would benefit from clinical genetics services, including clinical genetic evaluation, genetic counseling, genetic testing prenatal genetic evaluation, and genetic screening
- Perform a basic dysmorphology examination; recognize and classify common congenital anomalies and patterns of anomalies
- Recognize when to initiate the evaluation of patients with possible inborn errors of metabolism
- Understand the results of common cytogenetic, molecular cytogenetic, molecular genetic, and biochemical genetic diagnostic
- Estimate recurrence risks for Mendelian, multifactorial, and mitochondrial disorders in affected families
- Describe approaches to providing genetic counseling for commonly-encountered genetic disorders; communicate information in a clear and non-directive manner that is suitable for individuals of different educational, socio-economic, ethnic and cultural backgrounds
- Understand how to provide patients with access to diagnostic and predictive tests that are appropriate for the condition in their family and know how to advise patients of the benefits, limitations, and risks of such tests; work with a medical genetics specialist to develop a comprehensive plan for the evaluation and management of patients with, or at- risk for, genetic disease
- Safeguard privacy and confidentiality of genetic information of clients and families
- Utilize community support services and agencies and support groups for genetic diseases appropriately
- Identify sources of credible, current information about genetics, including medical genetic textbooks, specific computerized databases, Pub Med searches, genetics journals, and web-based information; use new information technologies effectively to obtain current information about genetics; understand and evaluate the quality of genetic information on the internet

Key Topics:

- Genetic assessment and pedigree analysis
- Dysmorphology-principles and evaluation; evaluation of intellectual disabilities and developmental delay
- Mendelian inheritance: Clinical applications, representative disorders, and genetic counseling
- Nontraditional inheritance: Clinical applications, representative disorders, and genetic counseling
- Clinical cytogenetic and molecular cytogenetic analysis and abnormalities; common chromosomal disorders; genetic counseling
- Inborn errors of metabolism
- Prenatal genetic screening, diagnosis, and counseling
- Cancer genetic risk assessment and counseling
- Genetic screening; pre-symptomatic and predispositional genetic testing
- Genetic counseling principles
- Ethical/Social/Legal issues
- Databases and other on-line genetics resources

Competencies:

- Learn how to apply basic principles of human genetics to clinical medical practice
- Learn how to obtain a thorough genetic history and 3-generation pedigree and analyze medical records
- Learn how to perform a dysmorphology examination
- Learn when to suspect genetic and genetic-metabolic disorders
- Understand the importance of establishing the correct diagnosis and mode of inheritance
- Understand the importance of the way in which information about diagnosis, prognosis, and recurrence risk is imparted to patients and their families; apply appropriate techniques for conveying difficult medical information; recognize the importance of reiterating information to patients who are anxious or unfamiliar with the concepts being presented
- Understand basic clinical genetic testing methods and interpretation of results
- Understand basic concepts of prevention of genetic disease; understand the differences between screening, diagnostic, presymptomatic, predispositional, and association testing
- Know how to make appropriate referrals to genetics specialists and to genetics support groups, community groups, or other resources that can benefit the patient and family
- Be aware of major ethical, legal, and social issues in medical genetics
- Appreciate the sensitivity of genetic information and the need for privacy and confidentiality
- Recognize philosophical, theological, and ethical perspectives influencing utilization of genetic information and services. Respect patients' religious, cultural, social, and ethical beliefs, even if they differ from one's own beliefs; interpret one's own attitudes toward ethical, social, cultural, religious and ethnic issues and develop an ability to individualize each patient or family member; cope emotionally with patient responses and decisions. Appreciate the importance of sensitivity in tailoring information and services to clients' and families' culture, knowledge and language level
- Recognize the importance of delivering genetic education and counseling fairly, accurately, and without coercion; understand the concept of non-directive genetic counseling (respect the autonomy of all patients, but also provide guidance with decision-making when requested)

Educational Activities: Participation in various genetics clinics (pediatric genetic disorders and congenital anomalies, metabolic genetics, prenatal genetics, adult genetics, cancer genetics) and inpatient consultations under the direct supervision of a Genetics Division faculty member.

- Two patient case conferences per week (clinical and prenatal)
- Two or more didactic sessions per week (except summer quarter)
- An introduction to the use of medical genetics textbooks, specific computerized genetics databases, Pub Med searches, genetics journals, and web-based genetics information
- Observing genetic counseling regarding pre-symptomatic or predispositional genetic testing for cancer or neurogenetic disorders

What students should do to Prepare for the Rotation: Review notes from the first year medical genetics course. If available, be able to access one of the following paperback texts:

- Thompson & Thompson's Genetics in Medicine, Nussbaum, et al, 7th ed., W.B. Saunders
- Medical Genetics, Jorde et al, Mosby, 4th ed., 2009
- Emery and Rimoin's Essential Medical Genetics, ed. By Rimoin, Pyeritz, & Korf, 1st ed, June 2013

Clinical Responsibilities of the Student:

- Students should review the patient's genetics chart prior to a clinic visit and should look up pertinent information in the literature
- Because of the length and complexity of evaluations, student participation in outpatient clinics is largely observational. The student may be asked to obtain history and pedigree information from the patient or parent. Physical examination is done together with the faculty member.
- For inpatient consultations, the student will review the chart, obtain a history and construct a pedigree, and examine the patient together with the fellow and/or the faculty member on call, and formulate a diagnosis and plan together. The student may be asked to write a consultation report and give it to the faculty member to review, amend/append, approve, and sign.

Patient Care Responsibilities: There are no direct patient care responsibilities for students. Students should follow the progress of patients whom they see for inpatient consultation.

Call Schedule of the Student: No night or weekend call

Procedures to be Learned by the Student: None

Percentage of Time Student will Participate in Ambulatory Setting: 50%

Standardized Instructional Material: Each student is given a 2-hour introduction to the rotation, including the Division schedule and clinical venues, basic aspects of genetic history and pedigree analysis, dysmorphology examination, and an introduction to major textbooks and databases.

Conference/Lecture/Small Group Sessions: Weekly Division case management/teaching conferences are held on Mondays from 8:30 to 10:30 AM. Genetics didactic/small group sessions are held on Wednesdays and Fridays from 9:00 to 11:00 AM (except during the summer). Other lectures are variably available, depending on the quarter.

Course Hours Approximate Weekly Summary:

3-6	Clinic preparation
1-2	Computer Assisted Instruction
2	Conference

16	Outpatient clinics
2-8	Inpatient consultation
Var.	Laboratory
4-6	Lecture
12	Other: reading, writing
40-52	Total

Content Theme Integration: None

Official Grading Policy: Each student will be observed and evaluated by Genetics Division faculty and at times also by the Genetics fellow and Genetic Counseling graduate students. The standard UC Irvine elective evaluation form will be used to determine the final grade of a student. The students will be graded on a three-part system Honors/Pass/Fail. Mid-course feedback will be provided to the students by the course director. If the student fails the elective a grade of "F" will be permanently recorded on his/her transcript. The student can repeat the course for a second grade; however, the "F" will not be removed from the transcript.

Site: CHOC Children's Hospital of Orange County

Course Description: This rotation provides a comprehensive introduction in to the field of medical genetics. Students have the opportunity to learn about a range of genetic disorders, genetic testing and genetic counseling. Students are also encouraged to participate in the activities of the outpatient clinics and inpatient consultations.

Elective Director: Neda Zadeh, M.D. (714)288-3500, nzadeh@choc.org

Who to Report to on the First Day: Frank Cruz Student Coordinator will contact student with further details. fcruz@uci.edu (714)456-5650

Site Coordinator: Frank Cruz fcruz@uci.edu (714)456-5650

Scheduling Coordinator: UC Irvine students please email comsched@uci.edu or call (714) 456-8462 to make a scheduling appointment. Please read the following information carefully. Any student enrolled at a U.S. LCME medical school will use VSAS to apply. To apply please refer to this website http://www.aamc.org/programs/vsas/

Periods of available: throughout the year.

Duration: minimum of 2 weeks.

Number of Students: 1-2 per period.

Course Objectives:

- Learn about a range of genetic disorders, genetic diagnostic testing and genetic counseling by participating in the evaluation of children and adults in our different outpatient clinics.
    - Learn to perform a basic dysmorphology exam.
    - Learn to obtain and draw a three-generation pedigree.
    - Estimate recurrence risks for Mendelian disorders in affected patients and families.
    - Learn about common cytogenetic and molecular tests utilized in clinical practice.

    - Understand the difference between diagnostic testing and screening.
    - Understand the abilities and limitations of specific genetic testing.
    - Understand the interpretation of genetic testing and different technologies available.
    - Learn about currently available genomic databases and clinical databases utilized in the care of our patients.
    - Appreciate the importance of a sensitive bedside manner, providing compassion in discussing different genetic conditions with patients and their families, and providing genetic education and counseling in a non-directive fashion.
- Participate in the activities of the inpatient consult service.
    - Student will review patient's chart and look up pertinent information in the medical literature.
    - Student will obtain a past medical history, family history and the physical examination will be performed in conjunction with the attending.
    - Follow the progress of the patient during the hospitalization.
- Observe patient care and management in multidisciplinary genetics clinics (craniofacial clinic, NF clinic, muscular dystrophy clinic etc.).

Lectures:

One individualized or small group lecture will be given formally per week on the topic of the student's choice. Students are also expected to attend resident morning report and noon conferences. Further education will be provided during genetics rounds and outpatient clinics.

Call schedule: None.

Preparation for the Rotation:

During the rotation, several texts and journal articles will be available, some of which are listed below:

- Handbook of Normal Physical Measurements, Hall, 1st ed, Oxford, 1989
- Management of Genetic Syndromes, Cassidy and Allanson, First ed, 2001
- Principles & Practice of Medical Genetics, Emery & Rimoin, 4th ed, 2001
- Smith's Recognizable Patterns of Human Malformation,
- Syndromes of the Head and Neck, Gorlin, 4th ed, Oxford U Press, 2001
- Thompson & Thompson's Genetics in Medicine, Nussbaum, et al, 6th ed., W.B. Saunders, 2001
- Medical Genetics, Jorde et al, Mosby, 3rd ed., 2003