

660F Clinical Genetics and Birth Defects

This rotation is not accepting international students

Course Name Clinical Genetics and Birth Defects

Course Director Maureen Bocian, MD

Academic Year 2020-2021

1. Course Director, Coordinator and General Administrative Information

FACULTY AND STAFF

Name	Office Location	Phone	Email
Director: Maureen Bocian, MD	City Tower, St. 800	714-456-5650	mebocian@uci.edu
Director: Neda Zadeh, MD	211 S. Main St.	714-288-3500	nzadeh@gmail.com
Coordinator: Frank Cruz	505 S. Main St., Ste. 525	714-456-5650	fcruz@hs.uci.edu

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

PREREQUISITES

This course is intended for 3rd and 4th-year students enrolled in the undergraduate medical education program at University of California, Irvine School of Medicine (UCISOM).

It is advantageous, but not required, for all students to have completed the Ob/Gyn, Medicine, and Neurology clerkships.

RESTRICTIONS

This course is intended for 3rd and 4th-year students enrolled in the undergraduate medical education program at University of California, Irvine School of Medicine (UCISOM).

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CHOC COURSE DIRECTOR:

Neda Zadeh M.D. is a medical geneticist in the Division of Medical Genetics at CHOC Children's Hospital. She specializes in the diagnosis and management of genetic syndromes and birth defects. Dr. Zadeh completed her residency training in pediatrics at CHOC, as well as fellowships in Clinical Genetics at Stanford University, and Clinical Molecular Genetics at UCLA. Dr. Zadeh is board certified in Clinical Genetics, Clinical Molecular Genetics and in Pediatrics.

Frank Cruz is the course coordinator for 660F course. Frank Cruz has been working as Student Coordinator for the UCISOM for 10+ Years. In addition to his Student Coordinator duties he also works as an assistant coordinator for the UCI-CHOC Pediatric Residency.

INFORMATION FOR THE FIRST DAY

Who/Where to Report on the First Day:

UCI Rotation: 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

CHOC Rotation: please contact Dr. Zadeh at nzadeh@gmail.com prior to rotation or call (714)288-3500, option 1.

Frank Cruz Student Coordinator will contact with further details. fcruz@uci.edu (714)456-5631.

SITE:

FOR MCWH/UCI BASED ROTATION: Patients are seen at UCIMC, UCI Gottschalk Clinic, Miller Children's, and CHOC

FOR CHOC BASED ROTATION: CHOC Only

DURATION: 4 weeks minimum

Scheduling Coordinator: UC Irvine students please call (714) 456-8462 to make a scheduling appointment.

Periods Available: The time of the course must be pre-approved by the elective director at least 3 months prior to the start of the course. No exceptions.

NUMBER OF STUDENTS ALLOWED: 2 per rotation

WHAT STUDENTS SHOULD DO TO PREPARE FOR THE COURSE

Review notes from the first year medical genetics course.

COMMUNICATION WITH FACULTY

Questions about logistics should be directed to the Course Coordinator. Direct questions, comments, or concerns about the course can be directed to the Course Director. Contact information and office location are at the beginning of this document.

The Course Director is also available to meet in person. Please email fcruz@hs.uci.edu to arrange an appointment. To ensure that your email will not be lost in the large volume of email received, please use the following convention for the subject line:

SUBJECT: COURSE NAME, your last name, your issue (e.g. XXX, Smith, Request for appointment)

2. Course Objectives and Program Objective Mapping

The following are the learning objectives for the 660F course. Students are expected to demonstrate proficiency in these areas in order to satisfactorily complete the course. In addition, the extent of a student's mastery of these objectives will help guide the course evaluation and grade.

Course Objective	Mapped UCI School of Medicine Program Objective	Sub Competency	Core Competency
Explain basic concepts regarding single-gene, chromosomal, multifactorial/polygenic, mitochondrial, and non-traditional patterns of inheritance in a manner easily understood by patients	A-2. Knowledge of the pathogenesis of diseases, interventions for effective treatment, and mechanisms of health maintenance to prevent disease	Disease Pathogen Treatment	Knowledgeable
	B-1. The ability to competently conduct a medical interview and counseling to take into account patient health beliefs, patient agenda and the need for comprehensive medical and psychosocial assessment	Medical Interview	Skillful

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<p>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</p>	<p>A-3. Knowledge of basic clinical skills required to meet the skills objectives, including interviewing, physical diagnosis, communication and clinical reasoning processes</p> <p>B-1. The ability to competently conduct a medical interview and counseling to take into account patient health beliefs, patient agenda and the need for comprehensive medical and psychosocial assessment</p>	<p>Basic Clinical Skill</p> <p>Medical Interview</p>	<p>Knowledgeable</p> <p>Skillful</p>
<p>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</p>	<p>B-3. The ability to articulate a cogent, accurate assessment and plan, and problem list, using diagnostic clinical reasoning skills in all the major disciplines</p>	<p>Patient Management</p>	<p>Skillful</p>
<p>Perform a basic dysmorphism examination; recognize</p>	<p>B-2. The ability to competently perform a complete and organ-</p>	<p>Physical Exam</p>	<p>Skillful</p>

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and classify common congenital anomalies and patterns of anomalies	system-specific examination including a mental health status examination		
Recognize when to initiate the evaluation of patients with possible inborn errors of metabolism	B-3. The ability to articulate a cogent, accurate assessment and plan, and problem list, using diagnostic clinical reasoning skills in all the major disciplines	Patient Management	Skillful
Understand the results of common cytogenetic, molecular cytogenetic, molecular genetic, and biochemical genetic diagnostic	A-2. Knowledge of the pathogenesis of diseases, interventions for effective treatment, and mechanisms of health maintenance to prevent disease	Disease Pathogen Treatment	Knowledgeable
	B-3. The ability to articulate a cogent, accurate assessment and plan, and problem list, using diagnostic clinical reasoning skills in all the major disciplines	Patient Management	Skillful
Estimate recurrence risks for Mendelian, multifactorial, and mitochondrial disorders in affected families	A-2. Knowledge of the pathogenesis of diseases, interventions for effective treatment, and mechanisms of health maintenance to prevent disease	Disease Pathogen Treatment	Knowledgeable
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	B-1. The ability to competently conduct a medical interview and counseling to take into account patient health beliefs, patient agenda and the need for comprehensive medical and psychosocial assessment	Medical Interview	Skillful

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cultural backgrounds	<p>C-2. Professional behaviors reflecting compassion and respect for patient privacy, altruism and a commitment to comprehensive, holistic medical care</p> <p>C-3. Sensitivity and awareness of diverse cultures, health beliefs and social factors impacting patient health and illness</p>	<p>Compassion</p> <p>Cultural and Social Awareness</p>	<p>Altruistic</p> <p>Altruistic</p>
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	B-3. The ability to articulate a cogent, accurate assessment and plan, and problem list, using diagnostic clinical reasoning skills in all the major disciplines	Patient Management	Skillful
Safeguard privacy and confidentiality of genetic information of clients and families	C-2. Professional behaviors reflecting compassion and respect for patient privacy, altruism and a commitment to comprehensive, holistic medical care	Compassion	Altruistic
Utilize community support services and agencies and support groups for genetic	A-5. Knowledge of medical practice, including healthcare	Medical Practice	Knowledgeable

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diseases appropriately	<p>economics and health systems impacting delivery and quality of patient care</p> <p>B-5. The ability to practice effective preventive medicine by identifying, addressing and advocating for strategies to maintain health and well-being, to identify and treat disease early where appropriate and to advise on lifestyle practices</p>	Patient Management	Skillful
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	B-4. The ability to search the medical literature, including electronic databases, and to locate and interpret up-to-date evidence to optimize patient care	Evidence-Based Medicine	Skillful

3. Course Resources

TEXTS AND READINGS: SUGGESTED

- 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

TEXTS AND READINGS: SUPPORTING AND REVIEW

- 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

4. Major Exams, Assignments and Grading

MANDATORY SESSIONS

Students are given a copy of the weekly schedule on the first day of the rotation as well as locations of various clinics and mandatory to attend all clinical and teaching sessions as outlined. Students are also expected to attend morning report and noon conferences at CHOC with the pediatric residents.

MAJOR ASSIGNMENTS AND EXAMS

There are no assigned projects or exams. Students can speak with the course director individually if they are interested in preparing a presentation on a particular genetic condition during the rotation, although this is not mandatory.

GRADING

Medical Students are graded using the following scale: Honors (H), Pass (P), Fail (F), and Incomplete (I). For further information, please review the Grading Policy.

You have 30 days from the date of the grade to appeal any aspect of this grade. Please contact your Clerkship/course Director should you have any questions

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.

Requirements for "Pass": To receive a grade of Pass, students must demonstrate successful performance in all the following areas:

- Knowledge

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- Patient Care
- Practice-Based Learning
- Interpersonal & Communication Skills
- Professionalism
- Systems-Based Practice

Requirements for “Honors”: To receive a grade of Honors, students must demonstrate exceptional performance all the following areas:

- Knowledge
- Patient Care
- Practice-Based Learning
- Interpersonal & Communication Skills
- Professionalism
- Systems-Based Practice

Grounds for “Incomplete”: You will not be issued a grade until all elements of the course have been completed.

REMEDIATION

Remediation, if needed will be designed by the Course Director to suit the issue at hand.

Grounds for “Fail”: You will receive a grade of "Fail" if the requirements for passing the course have not been met. Please refer to the [Grading Policy](#) for the impact of the "Fail" grade to the transcript.