660F Clinical Genetics and Genomics

This rotation is not accepting international students. CHOC Approval required

Elective at a Glance

Available: [X] UCI MS3 students [X] UCI MS4 students [] Extramural Students
Duration: **UCIMC**: 4 weeks, **CHOC:** 2 or 4 weeks
Number of Students: 2
Grading: H / P / F
Periods Available: Throughout the year

1. Course Director, Coordinator and General Administrative Information

FACULTY AND STAFF

Name	Office Location	Phone	Email
UCIMC: Natalie Gallant, MD, FACMG	3800 W. Chapman Ave, Suite 2200		ngallant@hs.uci.edu
CHOC: Neda Zadeh, MD	1201 W. La Veta Ave. Orange, CA 92868		nzadeh@gmail.com
Course Coordinator: Frank Cruz	505 S. Main St., Ste. 525	714-456-5650	fcruz@hs.uci.edu

DESCRIPTION

This course is designed to introduce students to the specialty of Medical (Clinical) Genetics and Genomics, including a variety of genetic disorders and congenital anomaly syndromes, genetic counseling, and genetic testing, by participating in the evaluation of children and adults in various clinical settings. Students should learn indications for referral for genetics consultation and the elements of a clinical genetics evaluation, including how to obtain a comprehensive family history and construct an accurate 3-generation pedigree, how to perform a dysmorphology examination, and how to approach the diagnosis and management of genetic disorders.

This elective consists of outpatient clinics and inpatient consultations at UCIMC and CHOC. Students must attend (virtual) weekly UCI teaching/patient management conferences and may be asked to present cases. There also are various weekly clinical genetics lectures at UCIMC during fall, winter, and spring quarters that students should attend if not engaged in clinical activities or consultations. Students have multiple clinical opportunities from which to choose each week, including outpatient consultation clinics at UCIMC and CHOC and may choose from various opportunities to work with different faculty in multiple half-day clinics per week—pediatric and adult genetics, prenatal genetics, cancer genetics, metabolic genetics—

and specialty clinics (cystic fibrosis, Prader-Willi syndrome, lysosomal storage disorders) .

PREREQUISITES
This course is intended for third- and fourth-year students enrolled in the undergraduate medical education program at the UCI School of Medicine. Ideally, medical students should have completed the Pediatrics, Ob/Gyn and Medicine clerkships, but these are not required.

RESTRICTIONS

- This course is intended for third- and fourth-year students enrolled in the undergraduate medical education program at UCI School of Medicine.
- UCIMC: Students must have the elective director's approval prior to UCIMC enrollment
- CHOC: All students must get prior approval via clinical scheduling coordinator (comsched@hs.uci.edu) for all CHOC-based rotations.
- This rotation does not accept international students.
- The time of the course must be pre-approved by the elective director at least three months prior to the start of the course. Rare exceptions may be granted.

COURSE DIRECTOR
Dr. Gallant is the course director for 660F course at UCIMC.
Dr. Zadeh is the course director for the 660F course at CHOC.

Frank Cruz is the course coordinator for the 660F course. He has been working as Student Coordinator for the UCI School of Medicine for 10+ Years, and he also works as an assistant coordinator for the UCI-CHOC Pediatric Residency. Mr. Cruz will contact you with further details. fcruz@hs.uci.edu (714)456-5631.

INFORMATION FOR THE FIRST DAY
Who/Time to Report to on First Day: The week usually begins with our Monday morning clinical case conference from 8:00 – 10:30 AM. This conference is held only by Zoom until further notice. Please email the course director one week before the elective starts for schedule updates and conference link.

Location to Report on First Day: Students will meet (virtually) with the course director after the Monday morning clinical conference for orientation to the rotation and clinic assignments. The first half-day clinic will likely be that afternoon.

SITE: UCIMC and CHOC

DURATION: **UCIMC:** 4 weeks, **CHOC:** 2 or 4 weeks

Scheduling Coordinator: UC Irvine students please email comsched@hs.uci.edu to make a scheduling appointment.

Periods Available: The time of the course must be pre-approved by the elective director at least three months prior to the start of the course. (Rare exceptions may be granted.)

NUMBER OF STUDENTS ALLOWED: Total of 2 trainees of any type—medical student, resident, and/or fellow.

WHAT STUDENTS SHOULD DO TO PREPARE FOR THE COURSE
Review notes from the first-year medical genetics course. See suggested reading (below).

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/virtually. 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. 660F, 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 nontraditional 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 B-1. The ability to competently conduct a	Disease Pathogen Treatment Medical Interview	Knowledgeable Skillful

	medical interview and counseling to take into account patient health beliefs, patient agenda and the need for comprehensive medical and psychosocial assessment		
Elicit a comprehensive, patient and multigenerational family medical history, construct an appropriate three- generation pedigree, and recognize patterns of inheritance and other signs suggestive of genetic disease in the family.	A-3. Knowledge of basic clinical skills required to meet the skills objectives, including interviewing, physical diagnosis, communication and clinical reasoning processes 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	Basic Clinical Skill Medical Interview	Knowledgeable Skillful
Recognize features in a patient's medical history, physical examination, and/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	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

testing prenatal genetic evaluation, and genetic screening.			
Perform a basic dysmorphology examination; recognize and classify common congenital anomalies and patterns of anomalies.	B-2. The ability to competently perform a complete and organsystem-specific examination including a mental health status examination	Physical Exam	Skillful
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 tests.	A-2. Knowledge of the pathogenesis of diseases, interventions for effective treatment, and mechanisms of health maintenance to prevent 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	Disease Pathogen Treatment Patient Management	Knowledgeable Skillful
Estimate recurrence risks for Mendelian, chromosomal, 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 cultural backgrounds.	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 C-2. Professional behaviors reflecting compassion and respect for patient privacy, altruism and a commitment to comprehensive, holistic medical care C-3. Sensitivity and awareness of diverse cultures, health beliefs and social factors impacting patient health and illness	Medical Interview Compassion Cultural and Social Awareness	Skillful Altruistic Altruistic
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 diseases appropriately.	A-5. Knowledge of medical practice, including healthcare economics and health systems impacting delivery and quality of patient care 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	Medical Practice Patient Management	Knowledgeable 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, 8th ed., 2015, W.B. Saunders;
- Medical Genetics 6th Edition Jorde LB, et al. 2019, Elsevier.

- Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics: Foundations 7th Edition edited by Reed E. Pyeritz, et al., 2018, Academic Press

TEXTS AND READINGS: SUPPORTING AND REVIEW:

- Smith's Recognizable Patterns of Human Malformation: Expert Consult - Online and Print 8th Edition by Kenneth Lyons Jones MD, et al, 2021, Elsevier,
- Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics: Metabolic Disorders 7th Edition, edited by Reed E. Pyeritz, et al., 2020, Academic press
- Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics: Hematologic, Renal, and Immunologic Disorders 7th Edition, edited by Reed E. Pyeritz, et al., 2022, Academic Press
- Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics: Perinatal and Reproductive Genetics 7th Edition, edited by Reed E. Pyeritz, et al., 2021, Academic Press
- Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics: Cardiovascular, Respiratory, and Gastrointestinal Disorders 7th Edition, edited by Reed E. Pyeritz, et al., 2019, Academic Press
- Atlas of Inherited Metabolic Diseases, 4th ed., Nyhan, Lippincott, 2020
- Handbook of Physical Measurements, Gripp, Slavotinek, Hall, Allanson,; 3rd ed, Oxford, 2013;
- Cassidy and Allanson's Management of Genetic Syndromes, 4th Edition, edited by John C. Carey, et al, Wiley-Blackwell, 2020
- Gorlin's Syndromes of the Head and Neck, Hennekam, Krantz, & Allanson, 5th ed, Oxford U Press, 2010
- The Practical Guide to the Genetic Family History, Bennett, Wiley-Liss; 2nd edition, 2010.

WEBSITES:

- Concert Genetics: https://www.concertgenetics.com/clinicians/ · Concert's Genetic Test Finder allows clinicians to search a comprehensive catalog of all genetic tests on the market, comparing across key test details to determine which test makes the most sense for each patient.
- Genetic Testing Registry: https://www.ncbi.nlm.nih.gov/gtr/ (Concert Genetics is better)
- GeneReviews: http://www.ncbi.nlm.nih.gov/books/NBK1116/ Clinical genetic summaries for many disorders including diagnosis and testing; current and easy to read.
- OMIM: http://www.ncbi.nlm.nih.gov/omim Online Mendelian Inheritance in Man, searchable database that includes thousands of genetic disorders, useful clinical synopsis and references

- Elements of Morphology: http://elementsofmorphology.nih.gov/ Human Malformation Terminology – definitions of proper descriptive terms; photo illustrations
- MedGen: https://www.ncbi.nlm.nih.gov/medgen/ NCBI page with links to many Medical Genetics databases and tools:

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. It is mandatory to attend all clinical and teaching sessions as outlined. Students are also expected to attend clinical conferences and selected lectures.

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. Factors that will be taken into consideration for the final rotation grade (relative to level of training):

- Attendance and punctuality
- Enthusiasm / proactive learning
- Fund of knowledge
- Professionalism
- Cultural sensitivity
- Ability to formulate an initial assessment and plan of care for patients, including evidence of reviewing literature and applying the information to patient care
- Taking “ownership” in patient care, i.e., actively following up on pending labs/imaging, writing follow up notes on patients seen previously, and communicating/collaborating with appropriate team members

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 UCI 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. Midcourse 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
- 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.