

Pediatric Genetics - Jacksonville

PED E 38J | 4th Year Elective | Pediatrics | Clinical Science | Jacksonville

MDT 7400

Prerequisites

Completion of third year core rotation in Pediatrics.

Course Description

Students will attend assigned genetics clinics, participate in inpatient consultations, and attend genetics teaching conferences. Students will be provided a reading syllabus of original articles relating to the embryology of congenital anomalies and the approach to diagnosis of genetic disorders, which they are expected to read during the elective. Discussion will follow with faculty supervisor. The student will carry out an independent literature review of a genetics topic of his/her choice. The topic will be presented or to the faculty supervisor at the end of the rotation. The students will also participate in the department teaching activities including attending rounds, grand rounds, lectures, and teaching conferences.

Course Faculty and Staff

- [Pamela Trapane MD](#) (Director)
- [Dani Brown](#) (Course Staff)
- [Jennifer R. Hamilton BA](#) (Course Staff)
- [Ashley Volz](#) (Course Staff)
- [Frank J Genuardi MD, MPH](#) (Other Faculty)

Meeting Place and Time

8 AM: Dr. Trapane's office, 841 Prudential Dr, Ste. 900

Schedule: M-F 8-5

Genetic Clinics (841 Prudential Drive, Suite 1900)

Inpatient consultation: Wolfson Children's Hospital (800 Prudential Dr.) & UF Health Jacksonville (655 W 8th St.)

Course Materials

Elements of Morphology: Am J Med Genet Part A 149A:2-92

Smith's Recognizable Patterns of Human Malformation (Jones)

Introduction Dysmorphology Approach and Classification

Chapter 2 Morphogenesis and Dysmorphogenesis

Chapter 3 Genetics, Genetic Counseling, and Prevention

Chapter 4 Minor Anomalies: Clues to more serious problems and to the recognition of malformation syndromes

Emery and Rimoin's Principles and Practice of Medical Genetics

Section I – Basic Principles

Chapter 18 Human Developmental Genetics

Chapter 19 Human Malformation

Section III – Approaches to Clinical Problems

Chapter 43 A Clinical Approach to the Dysmorphic Child (Jones)

Chapter 44 Clinical Teratology

Chapter 45 Abnormal Mental Development

Chapter 46 Abnormal Body Size and Proportion

Metabolic and Molecular Basis of Inherited Disease
Chapter 66 Clinical Phenotypes: Diagnosis / Algorithms

Web Based Resources:

OMIM (www.omim.org/)

Gene Reviews (www.ncbi.nlm.nih.gov/books/NBK1116/)

Unique Rare Chromosome (www.rarechromo.org/)

Face 2 Gene (www.face2gene.com/)

Decipher (decipher.sanger.ac.uk/)

ClinVar (www.ncbi.nlm.nih.gov/clinvar/)

Additional Information

Classes Offered:

Pediatric Grand Rounds – every Wednesday 8-9

Clinical Conference – every Friday 830-10

Journal Club – 4th Friday of the month 10-11

Classes Offered

Period	Length	Credits	(Avail / Max) Slots
Period 1	4 Weeks (May 8 - Jun 4)	4	(1 / 1)
Period 2	4 Weeks (Jun 5 - Jul 2)	4	(1 / 1)
Period 3	4 Weeks (Jul 3 - Jul 30)	4	(1 / 1)
Period 4	4 Weeks (Jul 31 - Aug 27)	4	(1 / 1)
Period 5	4 Weeks (Aug 28 - Sep 24)	4	(1 / 1)
Period 6	4 Weeks (Sep 25 - Oct 22)	4	(1 / 1)
Period 7	4 Weeks (Oct 23 - Nov 19)	4	(1 / 1)
Period 8	4 Weeks (Nov 20 - Dec 17)	4	(1 / 1)
Period 9	4 Weeks (Jan 1 - Jan 28)	4	(1 / 1)
Period 10	4 Weeks (Jan 29 - Feb 25)	4	(1 / 1)
Period 11	4 Weeks (Feb 26 - Mar 24)	4	(1 / 1)
Period 12	4 Weeks (Mar 25 - Apr 21)	4	(1 / 1)
Period 13	4 Weeks (Apr 22 - May 19)	4	(1 / 1)

Evaluated Competencies

#1 Professionalism

Educational Objectives: Student will be expected to show respect for colleagues and patients and compassion for patient and families in all interactions. Student will follow highest ethical standards ; follow all the rules and regulations and HIPAA standards. By the end of the rotation, the student will be able to: · Demonstrate reliability in the daily function. · Maintain integrity and honor in complex situations. · Seek to exceed expectations. · Maintain positive attitude amidst chaos. · Avoid shortcuts that omit components of patient care. · Accept responsibility for own actions. · Demonstrate accountability for actions of yourself and the health care team. · Advocate for best quality of care possible. · Identify cultural and personal issues of patients/families that affect patient care decisions. Recognize personal biases that may conflict with patients/families in decision-making and development of treatment plans. · Resolve conflicts in favor of the patient/family when possible. · Respect the input and importance of the family/patient and each member of the health care team. · Recognize and address ethical issues confronted daily. · Maintains and advances knowledge and skills independently. Accept assigned roles and responsibilities. · Effectively use time. · Maintain a safe, effective and responsible practice.

Method of Evaluation: Preceptor observation.

#2 Patient Care

Educational Objectives: Student will demonstrate ability to gather appropriate patient management data and to present this in oral and written form. By the end of the rotation, the student will be able to:

- Perform a thorough physical examination with emphasis on identifying major and minor congenital anomalies which may be signs of underlying genetic syndromes.
- Provide primary care and participate as a team member in medical and educational planning for patients with genetic disorders.
- Participate in the process of genetic counseling and then review the experience with an experienced genetic counselor.
- Demonstrate commitment to collect appropriate screening histories, participate in neonatal screening programs, provide initial counseling, and utilize resources for genetic counseling.
- Recognize, provide initial evaluation and counseling for, and identify resources for emergency consultation regarding:
 - infants presenting with symptoms that indicate the possibility of a severe inborn error of metabolism (e.g., metabolic acidosis, hyperammonemia, unexplained seizures).
 - chromosomal abnormalities that require prompt diagnosis in the perinatal period (e.g., Trisomy 13, 21).
 - unexplained critical illness or death suggestive of metabolic disorder, requiring collection of tissue samples before or at time of death.
 - developmental delay with signs or symptoms suggesting an underlying metabolic or genetic disorder.

Method of Evaluation: Preceptor observation. Review of write-ups.

#3 Medical Knowledge

Educational Objectives: Student will be expected to have the understanding and knowledge of pathophysiology, clinical features, differential diagnosis, diagnosis, treatment or therapy options, and outcome of more commonly encountered genetic disorders. Student will be required to attend educational conferences, lectures, department grand rounds, and other meetings pertaining to medical genetics. Student will be expected to research topics encountered in clinical care through the course materials to increase medical knowledge. By the end of this rotation the trainees will be able to: · Identify key concepts related to gene structure, molecular genetic techniques, mutations, and common patterns of inheritance (autosomal dominant and recessive, X-linked recessive, multifactorial, and related to maternal age). · Describe common methods of genetic diagnosis, including genetic screening tests available, and identify resources for up-to-date information on this topic. · Explain the concept of malformation etiologies: chromosomal, multifactorial, teratogenic, due to intrauterine factors. · Discuss key concepts related to testing for carrier states and genes predisposing to cancer and adult onset disorders (e.g., Alzheimer's). · For commonly encountered disorders (e.g., Trisomy 21, Turner Syndrome, Fragile X, neurofibromatosis, PKU) list presenting signs and symptoms and identify principles of long-term management. · Identify the disorders screened for in newborn screening in various states and specifically in Florida. Identify the method of testing for each of the disorders and the common problems encountered during the newborn screening process.

#4 Practice-Based Learning

Educational Objectives: Student will demonstrate ability to perform self-directed activities to increase knowledge. By the end of this rotation the student will be able to: · Participate in the process of genetic counseling and then review the experience with an experienced genetic counselor. · Demonstrate the ability to critically appraise the medical literature. · Incorporate evidence (when possible) into the decision-making and treatment plans of common genetic health care issues. · Locate patient information efficiently. · Analyze own practice experience and perform practice-based improvement using a systematic methodology. · Seek feedback on performance on a regular basis from attending and peers. · Provide effective feedback to students and other members of team. · Reflect on own performance and develop plan for improvement. · Ask questions of other members of team to guide thinking. · Participate in student orientation to delineate roles/responsibilities. · Use clinical encounter for teaching opportunities. · Address the need for balance of professional and personal activities.

Method of Evaluation: Student will be asked to give at least one oral presentation about a topic encountered in clinical practice and selected by the student in consultation with the faculty. Student also present findings from a literature review on a separate topic in genetics of his/her choice. Student will also be expected to perform literature searches for topics associated with the patients they encounter in clinical practice and apply the literature to clinical care decisions.

#5 Interpersonal and Communication Skills

Educational Objectives: Students will be expected to communicate history, physical exam findings and care plan verbally in a concise, complete, and organized manner to team members during and after rounds. Students will also document this information clearly in charting to optimize communication and care coordination. By the end of this rotation the student will be able to: · Explain indications for testing in the primary care setting for genetic or metabolic disorders (e.g., for findings such as short stature, developmental delay, minor congenital anomalies, failure to thrive, seizures, family history suggestive of certain inherited conditions). · Collect an appropriate family history for genetic disorders and identify risks when present, and triage families appropriately for genetic counseling. · List the disorders screened for in one's state neonatal screening program, provide initial counseling, and explain management for an infant with a positive neonatal screening test for each disorder. · Seek information from families and other health care professionals to assure patient-centered care that addresses patient/family needs. · Verify information from appropriate sources, filling in gaps to promote optimal care. · Demonstrate effective listening during interactions with patients, families and other members of the health care team. · Verify understanding of the patient, families and members of health care team. · Maintain approachable demeanor for families, nurses and other members of health care team. · Avoid words/actions that diminish willingness of others to provide information. · Respond to pages and questions in a timely manner. · Create and sustain a therapeutic and ethically sound relationship with patients and their families. · Provide information to families and other health care workers that is accurate, appropriate for their level of understanding and consistent with the overall treatment plan. · Recognize own personal biases that affect information provided. · Document patient care in the medical record following guidelines in a timely fashion. · Effectively use an interpreter. · Enter orders effectively and in a timely fashion. · Actively participate in work rounds, emphasizing facts. · Provide effective feedback to residents, peers and faculty. · Incorporate knowledge of medical literacy in presenting information to parents and patients. · Seek complete information needed to provide consultation requested by other services. · Understand the management of psychosocial problems that affect children with complex chronic disorders and their families.

Method of Evaluation: Preceptor observation. Review of clinical notes.

#6 Systems-Based Practice

Educational Objectives: Student will gain an understanding of the multidisciplinary approach to care of complex patients, including medical genetics, genetic counseling, and coordination of care with specialties providing therapeutics. By the end of this rotation the student will be able to: · Explain how to identify and use resources in one's community for diagnosis, genetic counseling, therapy, and psychosocial support of children with genetic defects and congenital anomalies. · Discuss public health strategies to reduce risk for genetic disorders and congenital anomalies (e.g., early identification and screening programs to detect disease and carrier states, prenatal care, genetic counseling). · Be familiar with when to seek consultation, when to refer to the appropriate sub-specialist and how to

Method of Evaluation: Preceptor observation. Attendance and participation in multidisciplinary meetings.