Pediatric Medical Genetics Syllabus

Credit Hours: 2-4

Contact Information

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Course Information

Brief Description of Course

Medical Genetics involves any application of genetics to medical practice. It includes the studies of the inheritance of diseases in families, analyses of the molecular mechanisms through which genes cause disease, and the diagnosis and treatment of genetic disease. Medical genetics also includes genetic counseling, in which information regarding risks, prognoses, and treatments is communicated to patients, their families, and their primary care providers. As a result of seeing new and established patients in our Pediatric Medical Genetics Clinics, the medical student will develop an approach to establishing diagnoses in those families with a syndrome, both clinically and at the molecular level. The medical student will understand the basics of genetic counseling and provision of anticipatory guidance and management of common genetic conditions. The medical student will appreciate the collaboration required between the primary care provider and pediatric medical genetics subspecialist in providing optimal care to families with genetic disorders.

Course Goals

Learning Objectives (see as many patients as possible in time available):
- Understand basic genetic principles
- Obtain pedigree information from consult and
- Interpret inheritance patterns from pedigree information
- Identify key components of a genetics evaluation
- Distinguish autosomal dominant, autosomal recessive, X-linked recessive, X-linked dominant, mitochondrial, and multifactorial inheritance patterns
- Determine distinction between inborn errors of metabolism and syndromal etiologies of developmental delay
- Demonstrate understanding of indications for genomic testing
- Demonstrate understanding of indications for biochemical testing
- Interpret test results and understand implication of informing interview
- Demonstrate proficiency in accessing the genome browser
- Demonstrate ability to utilize databases to identify Mendelian disorders from cytogenomic SNP microarray results with genomic imbalances and/or segments of homozygosity
- Recognize value of collaboration between primary care providers, medical geneticists, genetic counselors, nurse practitioners, and dieticians in providing care to families with genetic conditions
- Navigate ethical issues related to genetic issues
  Learning guide to the 6 competencies for genetics trainees as outlined by the American Board of Medical Genetics and Genomics (ABMGG) http://abmgg.org/pages/program_learning.shtml

Course Format & Sample Schedule

Outpatient Clinics (Station 5, Third Floor PC Eccles Outpatient Facility 662-7511):

Mon – Biochemical Genetics (Longo, Botto, Underhill, Bentley, Shayota, Andrews)
Clinical Genetics (Flores, Goeke)

Tues – Clinical Genetics (Dent, Flores, Shayota, Viskochil)

Wed – Clinical Genetics (Flores, Goeke, Hagedorn, Viskochil)

Thurs – Clinical Genetics (Flores, Goeke, Hagedorn, Shayota, Elias)

Fri – Clinical Genetics (Flores, Viskochil, Elias)

General Genetics Providers
Audrey Rutz (GC), audrey.rutz@hsc.utah.edu
Brian Shayota (MD), brian.shayota@hsc.utah.edu
Caroline Hagedorn (NP), caroline.hagedorn@hsc.utah.edu
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Karin Dent (GC), karin.dent@hsc.utah.edu

Metabolic/Biochemical Genetics Providers
Abbey Bentley (NP), abbey.bentley@hsc.utah.edu
Ashley Andrews (NP), Ashley.andrews@hsc.utah.edu
Hunter Underhill (MD), hunter.underhill@hsc.utah.edu
Lorenzo Botto (MD), Lorenzo.botto@hsc.utah.edu
Nicola Longo (MD, PhD), Nicola.longo@hsc.utah.edu

Inpatient Rounds

Fridays, 1:15 to 2:30
On-call physician from previous week oversees patient presentations
Review of patients followed by walk rounds to selected patients

Biochemical Rounds

Wednesday morning 1:00-2:00 to review upcoming clinic following Monday
Friday afternoon 15:00-16:00 to review results from previous Monday Clinic

Case Management Conference

Wednesday at 2 to 3pm (3-minute discussion of selected patients)
Wednesday from 3-4pm formal presentation of various genetics topics

Other Clinical Genetics Experiences

Prenatal Diagnosis Conference (3rd Friday of every month; 7:30am; 2B329)
Prader-Willi Syndrome Clinic (2nd Thursday morning every other month)
Hearing Loss Clinic (1st Tuesday every month)
Tuberous Sclerosis Complex Clinic (4th Friday, every month)
Skeletal Dysplasia Clinic (variable)
Osteogenesis Imperfecta Clinic (2nd Wednesday every 3 months)
Neurofibromatosis Clinic (3rd Thursday every month – Riverton)

Processes in the medical genetics clinic setting

Orientation – On the first day of rotation go to the Eccles Outpatient Services building station 5 on the 3rd floor and touch base with one of the core faculty for the medical genetics training program (David Viskochil, Nicola Longo, Josue Flores). Patients will be assigned at that time.

Clinic Encounters – Patients are assigned weekly from a master list based on trainee's schedule. Administrative personnel in the Medical Genetics office assemble the upcoming clinic encounter list and email to clinic personnel, including trainees. There is flexibility on how many patients will be evaluated and how clinic notes are generated. Each attending has individual preferences and it is the trainee's responsibility to contract their respective role with each outpatient clinic encounter. This could include observer/shadower only, history acquisition, perform physical examination, counsel family, and/or compose clinic note. Inpatient consults require working closely with the weekly attending to determine level of independence and expectations for provision of genetics services.

Overall expectations – Genetics encounters are team consultations, which means integrating your contribution with genetic counselors, nurse practitioners, dieticians, laboratory fellows, medical genetics fellows, and genetics medical assistants. Contracting with members of the team prior to individual encounters is essential to determine the role each member will play in the consultation.

Unique genetics experiences – Pedigree acquisition and integration of family history into a genetics consultation is a paramount expectation in this rotation. Trainees will be expected to be able to *procure a 3-generation pedigree* by the end of the rotation. Understanding the *clinical presentation* of known conditions is another key aspect of this rotation, which means spending time with family member on how the diagnosis was initially established for return patients. The generation of a *differential diagnosis* for new patient encounters is an art in medicine, especially so in genetics encounters as families begin their potential diagnostic odyssey. Once a genetic diagnosis is established, *intervention* with treatment protocols for known metabolic disorders and/or accession to clinical trials for rare syndromes are key components in the practice of medical genetics. Finally, sensitivity to the delivery of informing news with respect to a diagnosis possibly affecting additional family members (*genetic counseling*) is an essential competency in medical genetics that should be recognized during this rotation.

The competencies are adjusted to each trainee: Observer without medical training, shadower, medical student, pediatric resident, genetic counselor trainee, non-medical genetics fellow, and ABMGG genetics laboratory trainee.

Activities: **Pre-test at start of rotation; Attend clinics, consults, genetics conferences.**
Post-test at end of rotation
Evaluation: **e-value assessments based on attendance, consults, team participation**

Counseling issues in Genetics
Pedigree analysis
Preconception testing
Newborn screening
Informing interview
At risk family members
Incidental findings
Non-paternity
Consanguinity

Key Genetic Conditions:
Biochemical Genetics

       Urine organic acidopathies
           Glutaric Acidemia type I
           Methylmalonic Acidemia
           Propionic Acidemia
       Serum amino acidopathies
           Hyperglycinemia
           Phenylketonuria (PKU)
           Urea cycle defects
       Fatty Acid Oxidation Defects
           MCAD
           VLCHAD
       Galactosemia
Hyperammonemia
       Lactic acidosis
       Mitochondrial disorders

     <u>**Clinical Genetics**</u>
       Chromosome anomalies
           Down syndrome (trisomy 21)
           Edwards syndrome (trisomy 18)
           Patau syndrome (trisomy 13)
           Turner syndrome (45,X)
           Klinefelter syndrome (47,XXY)
       Microdeletion syndromes
           Deletion 22q11.2
           Williams syndrome
           Smith-Magenis syndrome
       Imprinting disorders
           Prader-Willi syndrome
           Angelman syndrome
           Beckwith-Wiedemann syndrome
       Trinucleotide expansion disorders
           Fragile X syndrome
           Huntington disease
           Myotonic dystrophy
       Skeletal dysplasias
           Achondroplasia
           Thanatophoric dysplasia
           Osteogenesis imperfecta
       Neurocutaneous disorders
           Neurofibromatosis type 1
           Tuberous sclerosis
           Von-Hippel Lindau syndrome
       Lysosomal storage disorders
           Mucopolysaccharidoses
           Pompe syndrome
           Gaucher disease
           Fabry disease
       Peroxisomal disorders
           X-Linked adrenoleukodystrophy
Zellweger syndrome
       Alexander disease
       Mosaicism
       Epigenetics
       Multifactorial disorders
Handouts: (https://uofu.box.com/s/gtu0mv39h0fkleu8oc8gcsu1nmontpmj)
     Pedigree Nomenclature
     PEDIATRICS Volume 132, Supplement 3, December 2013
     Principles of Anticipatory Guidance (https://pubmed.ncbi.nlm.nih.gov/1531255/)
     Competencies in Medical Genetics

PowerPoint lectures:
>   **Clinical Genetics (HGEN 6470)**
>   **Clinical Biochemical Genetics (HGEN 7380)**
>   **Human Genetics (HGEN 6500)**

Educational and Instructional Modalities

As an example:

Modality	Percentage
Didactic	10%
Clinic Time	80%
Free Study	10%

Role of the Student in this Course

If you are sick or cannot come to clinic, please call Kairisti at (801) 585-7563 (kairisti.schut@hsc.utah.edu) to let the attending know.

Required Textbook(s)/Readings

Some required readings in the form of Journal Articles will be provided during the course of the rotation and will be accessible through Canvas. Textbooks below are available in offices of our faculty.

>   ***Medical Genetics: Jorde, Carey, Bamshad: 5th edition**
>   ***Management of Genetic Syndromes: 4th edition**
>   **Smith's Recognizable Patterns of Human Syndromes: 6th edition**
>   **Inborn Errors of Metabolism**
>   **Inborn Errors of Development**
>   **Syndromes of the Head and Neck**

Key Websites

>   **OMIM (Online Mendelian Inheritance of Man): www.omim.org**
>   **GeneReviews (www.ncbi.nlm.nih.gov/books/NBK1116/)**
>   **Genome browser (www.genome.ucsc.edu/)**

Assessment & Grading

Preceptor Evaluations

For Clinical Courses: This Clinical Course employs a preceptor evaluation which contributes to the student's overall course grade. Competencies are adjusted to level of training of the medical student.

Activities: Pre-test at start of rotation; Attend clinics, consults, genetics conferences; Post-test at end of rotation
Evaluation: e-value assessments based on attendance, consults, team participation

Assessments

Assessment Name	Weight toward Final Grade
Preceptor Evaluations	100%

Grading System

Students will receive a final letter grade of PASS (P), or FAIL (F) for this course.
Only elective courses with approved alternative grading schemes may use H/HP/P/F or H/P/F grading

>   **PASS:** A student who achieves the criteria, will be assigned a grade of PASS for the course.
>   **FAIL:** A student who fails to achieve the criteria for PASS, will be assigned a grade of FAIL for the course.

Student Feedback

Student feedback is an important aspect of curriculum quality improvement. Thus, students are expected to complete all assigned feedback surveys specific to a course by the due date.

Standard Policies

Please refer to the Student Handbook (on the Student Affair's website) for these policies:

Accommodations
Addressing Sexual Misconduct
Attendance policy
Dress Code
Examination and Grading Policies
Grade or Score Appeal
Professionalism, Roles & Responsibilities
Mistreatment

Alternate Name and/or Personal Pronoun

Class rosters are provided to the instructor with the student's legal name as well as 'Preferred' first name (if previously entered by you in the Student Profile section of your CIS account). While CIS refers to this as merely a preference, we will honor you by referring to you with the name and pronoun that feels best for you in class, on papers, exams, group projects, etc. Please advise us of any name or pronoun changes (and please update CIS) so we can help create a learning environment in which you, your name, and your pronoun will be respected.

Center for Disability & Access Services

The School of Medicine seeks to provide equal access to its programs, services and activities for all medical students. The Center for Disability and Access (CDA) provides accommodations and support for the educational development of medical students with disabilities. Medical students with a documented disability and students seeking to establish the existence of a disability and to request accommodation are required to meet with the CDA Director for recommended accommodations. The CDA will work closely with eligible students and the Academic Success Program to make arrangements for approved accommodations. The School of Medicine and CDA maintain a collegial, cooperative, and collaborative relationship to ensure compliance with federal and state regulations for students with disabilities.

Steven Baumann EdD, School of Medicine Senior Director of Academic Success Program, serves as the liaison between the School of Medicine and the CDA.

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