

# 2114 Pediatric Genetics/Metabolism

Course: Pediatric Genetics/Metabolism Course Number: PED 2114

Department: Pediatrics

Faculty Coordinator: Mary Carlin, MD

Assistant Faculty Coordinators: N/A

UTSW Education Coordinator Contact: Anthony Lee ([Anthony.Lee@UTSouthwestern.edu](mailto:Anthony.Lee@UTSouthwestern.edu))

Hospital: (Location of rotation) Children’s Medical Center  
 Bldg NB, 10th floor, Room 1022  
 Texas Scottish Rite Hospital  
 G6200 – UT Medical School, G Bldg – 6th floor  
 AC – Aston Center- 8th floor  
 MCD – McDermott Center, North Campus

Periods Offered: 1-12

Length: 4 weeks

Max # of Students: 1 (Post Clerkship Students)

First Day Contact: Mary Carlin, MD

First Contact Time: 7:30am

First Day Location: Children’s Medical Center –6<sup>th</sup> Floor Pavilion Genetics Clinic (Physician’s Touchdown F6-279)

Prerequisites: PED 1801 Pediatric Core Clerkship

## I. Course Description

Students will be integral members of a team participating in the provision of comprehensive consultations and for follow-up of patients admitted to the Genetics/Metabolism floor. Students will also participate in the outpatient Genetics/Metabolic clinic evaluating patients referred for the full scope of concerns related to pediatric genetics and metabolism.

| <u>Goals</u>   | <u>Objectives</u>  | <u>Assessment Methods</u>  |
|--|--|--|
| <b>Patient Care:</b><br>Students, together with supervising faculty, must be able to provide patient care that is compassionate, appropriate and effective for the treatment of health problems and the promotion of health. | <ul style="list-style-type: none"> <li>Students will demonstrate the knowledge, attitudes and skills necessary to perform appropriately focused and accurate histories and physical assessments and document the findings accurately in the health record.</li> <li>Students will assist in</li> </ul> | <ul style="list-style-type: none"> <li>Quality of Medical Records entries</li> <li>Skills evaluation from direct observation.</li> </ul> |

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|  | <p>development of evaluation and treatment plans, and take responsibility for implementation.</p> <ul style="list-style-type: none"> <li>• Students will develop procedural skills related to evaluating a child with a positive newborn screening result.</li> <li>• Construct a 3-generation pedigree from a patients' family history.</li> </ul>  |   |
| <p><b>Medical Knowledge:</b><br/>Students must demonstrate knowledge about established biomedical and clinical sciences and the application of this knowledge to patient care.</p>   | <ul style="list-style-type: none"> <li>• The Student will be able to explain how to use a pedigree to support a Mendelian mode of inheritance and/or designate obligate and/or possible carries of an inherited disease or trait.</li> <li>• The student will know how to assess and manage common complaints in one inborn error tested for by the current expanded newborn screening program and discuss how to confirm whether the infant is affected, what confirmatory test(s) are indicated and what treatment(s) might be initiated.</li> <li>• The student will know the pathophysiology of the usual cause of Down syndrome, the mechanism(s) involved and the recurrence risk for the parents, the siblings and the affected child.</li> <li>• Demonstrate how to examine an infant or a child for dysmorphic features and/or congenital anomalies.</li> <li>• Point out at least 5 features commonly seen in a child with Down syndrome.</li> <li>• List at least 6 prenatal exposures and/or maternal conditions that may be teratogenic to the fetus and identify at least one abnormality it may cause.</li> </ul> | <ul style="list-style-type: none"> <li>• Each student is required to create a case presentation and to present it to the faculty and fellows during the rotation. It is recommended that students present an interesting case they encountered during their rotation. However, they are also permitted to present on a topic of interest that relates to pediatric course topic.</li> </ul> |
| <p><b>Interpersonal and Communication Skills:</b><br/>Students must be able to demonstrate interpersonal and communication skills that result in effective information exchange with health care professionals and families.</p> | <ul style="list-style-type: none"> <li>• Use effective listening skills and elicit and provide information using effective nonverbal, explanatory, questioning, and writing skills. These include taking complete history from pediatric patient and their family and writing a careful evaluation in the chart.</li> </ul>  | <ul style="list-style-type: none"> <li>• Observations of faculty and staff.</li> </ul>  |

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|  | <ul style="list-style-type: none"> <li>• Work effectively with others as a member of a health care team.</li> </ul>   |   |
| <p><b>Practice-Based Learning and Improvement:</b><br/>Students must be able to assimilate scientific evidence and improve their patient care practices.</p>   | <ul style="list-style-type: none"> <li>• Locate and assimilate evidence from scientific studies related to their patients' health problems, including literature searches.</li> <li>• Use information technology to manage information, access on-line medical information; and support their own education.</li> </ul>   | <ul style="list-style-type: none"> <li>• Each student is required to create a case presentation and to present it to the faculty and fellows during the rotation. It is recommended that students present an interesting case they encountered during their rotation. However, they are also permitted to present on a topic of interest that relates to pediatric course topic.</li> <li>• Critical review of a relevant article.</li> </ul> |
| <p><b>Professionalism:</b><br/>Students must demonstrate a commitment to carrying out professional responsibilities, adherence to ethical principles, and sensitivity to a diverse patient population.</p> | <ul style="list-style-type: none"> <li>• Demonstrate respect, compassion, and integrity; a responsiveness to the needs of patients and their families; accountability to patients and their families and the profession; and a commitment to excellence and on-going professional development.</li> <li>• Demonstrate a commitment to ethical principles pertaining to provision or withholding of clinical care, and confidentiality of patient information.</li> <li>• Demonstrate sensitivity and responsiveness to patients' culture, age, gender, and disabilities.</li> </ul> | <ul style="list-style-type: none"> <li>• Observations of faculty and staff.</li> </ul>  |

### II. Methods of Instruction

#### Didactic: (Schedule, Topic and Faculty)

| Day               | Time             | Conference                                | Venue                                  |
|-------------------|------------------|---|--|
| <b>Mondays</b>    | 8:00am – 9:00am  | Cancer Conference (Optional)              | North Campus                           |
|                   | 12:00pm – 1:00pm | Resident's Noon Conference                | Doctor's Dining Room (CMC Lobby Level) |
| <b>Tuesdays</b>   | 12:00pm – 1:00pm | Multidisciplinary Conference – Pediatrics | Moore Auditorium (CMC)                 |
| <b>Wednesdays</b> | 8:00am – 9:00am  | Pediatric Grand Rounds                    | Moore Auditorium (CMC)                 |
|                   | 12:00pm -1:00pm  | Resident's Noon Conference                | Doctor's Dining Room (CMC Lobby Level) |
| <b>Thursdays</b>  | 12:00pm – 1:00pm | Resident's Noon Conference                | Doctor's Dining Room (CMC Lobby Level) |
| <b>Fridays</b>    | 8:30am – 9:30am  | Friday Morning Report                     | Moore Auditorium (CMC)                 |
|                   | 9:30am – 10:30am | Genetics Conference                       | North Campus – McDermont Center        |

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|  | 12:00pm – 1:00pm                                  | Resident’s Noon Conference                                     | Doctor’s Dining Room (CMC Lobby Level) |
|  |   | Perinatal Case Conference (2 <sup>nd</sup> & 4 <sup>th</sup> ) | South Campus UTSW – G6200              |
|  | 3:00pm – 4:00pm<br>(2 <sup>nd</sup> Fridays only) | Journal Club   | TBD                                    |

### Clinical: (Schedule, Faculty Teaching and House-Staff Teaching)

| Day               | Time                      | Clinic  | Location   |
|-------------------|---------------------------|---|--|
| <b>Mondays</b>    | 8:30am – 12:00pm          | Genetics/Dysmorphology Clinic   | 6 <sup>th</sup> Floor Pavilion CMC                   |
|                   | 1:30pm – 4:00pm           | Genetics/Dysmorphology Clinic   | 6th Floor Pavilion CMC                               |
|                   | 4:00pm                    | Hospital Consultations (Check w/ Fellows and Attending)   | 6th Floor Pavilion CMC                               |
|                   | <b>Optional Clinic(s)</b> |   |  |
|                   | 12:30pm – 4:00pm          | Holoprosencephaly Clinic (Optional Clinic)  | Texas Scottish Rite Hospital                         |
| <b>Tuesdays</b>   | 8:30am – 12:00pm          | Metabolic Clinic  | 6th Floor Pavilion CMC                               |
|                   | 1:30pm – 4:00pm           | Metabolic Clinic  | 6th Floor Pavilion CMC                               |
|                   | 4:00pm -                  | Hospital Consultations (Check w/ Fellows and Attending)   | 6th Floor Pavilion CMC                               |
|                   | <b>Optional Clinic(s)</b> |   |  |
|                   | 12:30pm – 4:00pm          | Tuberous Sclerosis Clinic (1 <sup>st</sup> , 3 <sup>rd</sup> , 5 <sup>th</sup> Tuesdays only) (Optional clinic) | Texas Scottish Rite Hospital                         |
|                   | 1:30pm – 4:00pm           | Adult Genetics Clinic (Optional Clinic)   | Aston Center 8 <sup>th</sup> Floor                   |
| <b>Wednesdays</b> | 9:00am – 12:00pm          | Genetics Clinic   | 6th Floor Pavilion CMC                               |
|                   | 1:00pm – 3:00pm           | Metabolic Case Conference   | TBD – Check with Attending                           |
|                   | <b>Optional Clinic(s)</b> |   |  |
|                   | 9:00am – 12:00pm          | Rainbow Developmental Clinic (Optional Clinic)  | 6th Floor Pavilion CMC                               |
|                   | 1:00pm – 3:00pm           | Spina Bifida Clinic   | Texas Scottish Rite Hospital                         |
|                   | 12:30pm – 4:00pm          | Neurofibromatosis Clinic (weekly)   | Bldg NB, 10th floor, Room 1022                       |
| <b>Thursdays</b>  | 8:30am – 12:00pm          | Down Syndrome Clinic & Newborn Screening Follow up (every other week)   | 6th Floor Pavilion CMC                               |
|                   | 1:00pm – 4:00pm           | Genetics/Dysmorphology Clinic   | 6th Floor Pavilion CMC                               |
|                   | 4:00pm -                  | Hospital Consultations (Check w/ Fellows and Attending)   | 6th Floor Pavilion CMC                               |
|                   | 1:00pm – 4:00pm           | Molecular Genetics (One Thursday during rotation)   | McDermott Center, North Campus                       |
| <b>Fridays</b>    | 8:30am – 12:00pm          | Down Syndrome Clinic  | 6th Floor Pavilion CMC                               |
|                   | 2:00pm -                  | Hospital Consultations (Check w/ Fellows and Attending)   | 6th Floor Pavilion CMC                               |
| <b>Saturdays</b>  | 9:00am or 9:30am          | Metabolic Patient Rounds  | Main CMC Hospital (Check with Fellows and Attending) |

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## III. Overview of Student Responsibilities

- Evaluate new inpatients, present to attending and follow until discharge.
- In clinics, see new patients and present patient information to attending.

## IV. Method of Evaluation of Students and Requirements

Pass-fail grades; there are no examinations. Evaluations of the student by the faculty will be based on achievement of the stated objectives of the course. This will be assessed by the student's preparation, fund of knowledge, participation in clinic, and participation in didactic sessions.