

## **PEDIATRICS RESIDENCY CLINICAL GENETICS ELECTIVE**

Elective rotation available for residents of all training levels

Residents participate in outpatient referral clinics and provide inpatient consultation services for children and adults who present with a broad spectrum of developmental abnormalities, multiple congenital anomalies, mental retardation, a wide range of clinical syndromes and single gene disorders. The clinical experience can be adapted to the level of experience of the resident. This Clinical Genetics Service provides genetic diagnostic and counseling services to the whole medical community of the Penn State Hershey Medical Center which includes children and adult services.

### **Value of the Rotation:**

- This elective exposes the resident to the full range of clinical genetics challenges and seeks to train the resident in the diagnosis, management and genetic counseling for the full range of genetic disorders including prenatal genetic counseling, newborn with congenital malformations, chromosome abnormalities, neuromuscular defects, late onset adult disorders and metabolic disorders.

### **Broad Goals:**

- The central goal is to expose the resident to a systematic approach to evaluate patients with a known or suspected genetic condition leading to accurate diagnoses, awareness of appropriate genetic testing, providing appropriate genetic counseling and organization of ongoing healthcare management.

### **Objectives:**

- The resident will gain an awareness of the importance of genetic factors in the evolution of human health and disease.
- The resident will learn to integrate the practice of clinical genetics in expanding basic knowledge of developmental embryology, dysmorphology and teratology.
- The resident is expected to be able to take a comprehensive history and perform a complete physical examination with accurate documentation of dysmorphic features.
- The resident will be able to obtain detailed family histories and construct multi-generation pedigree and be able to analyze the pedigree to determine mode of inheritance.
- The resident will become knowledgeable about the natural history, etiology, inheritance and management of the common disorders referred to clinical genetics.
- Through specific case studies, the resident will gain familiarity with the various modes of inheritance including Mendelian inheritance, mitochondrial inheritance, recurrence risk for multifactorial conditions and risk associated with familial chromosomal abnormalities.
- The resident will be provided with extensive online resources to become knowledgeable about informational resources about genetic disorders.
- The resident will be exposed to newborn screening and gain knowledge about the range of testing available for the newborn.
- The resident will be instructed in current genetic and genomic testing technologies and the specific clinical settings for their application.
- The resident will develop an understanding of the range of clinical indications for referring patients to genetics services.

### **Rotation Description:**

- The resident will participate in each clinic visit under the supervision of the attending clinical geneticist who usually will conduct the clinic session including obtaining complete medical history, family history and carrying out a complete physical examination pointing out specific physical findings. Following this intake, the resident will participate in a discussion of the differential diagnosis and recommended genetic testing. This is then followed with a comprehensive discussion with the patient and family summarizing the clinical findings and providing recommendations for clinical and genetic testing. Each clinic session will vary in time depending upon the complexity of the clinical presentation and extent of discussion with the patient and family.

**Resident Expectations:**

- The resident will be expected to attend every clinic session, review medical records prior to the clinic visit and develop pre-clinic differential diagnoses.
- Residents are expected to be available via page for consults and clinic from 8am-5pm, Monday - Friday.
- There are no night or weekend responsibilities directly related to this elective, however residents may be needed to staff cross-covers on the weekends and occasionally during the week.
- The only general exceptions are Thursday afternoons, when time is protected for didactics, and their assigned continuity clinic afternoon. Other exceptions, will be listed on the block docs that are sent out prior to the start of the rotation.
- Residents are expected to communicate with the attending early in the rotation if other time is going to be missed, such as for a doctor's appointment.

**Reading/Resources:**

- The resident will be provided with a list of electronic resources and reprints related to each of the clinical diagnosis.

**Individualized Curriculum:**

- The rotation may be adapted to interests of the resident and may include participation in the prenatal genetic counseling program in the Department of Obstetrics/Gynecology and participation in the genetic counseling program of the Cancer Institute.
- If 4 weeks of this elective are done during residency, this will count as a key elective.

**Feedback and Evaluation:**

- The resident will receive ongoing assessments of performance through discussions involving each clinical case. Each clinical session represents an opportunity to review basic genetic principles revolving around the clinical diagnosis, differential diagnoses and development of a management plan. Each clinic session begins with an overview of anticipated challenges and ends with a gathering of all participants to discuss individual perceptions of the experience and a critique of each counseling session.

**Logistics:**

**First day/orientation:** Residents are encouraged to contact the administrative office of the Division of Human Genetics several days before the rotation is to begin in order to receive details of the clinic schedules and to receive instruction about the availability of pre-clinical charts and recommended reading. Residents are to contact Susan Sell, MS, CGC, at C7520 extension 8414.

**What is the schedule/who will you work with:** Clinics are scheduled Monday 8:30–noon (Ladda), alternating Tuesday 8:30–noon (Gordon), Tuesday 1–5 p.m. (Ladda), Wednesday 8:30–5 PM Ladda), alternating Thursday 8:30–noon (Gordon). Appointments are also arranged outside of these time schedules to meet the specific need of an individual patient. Resident will be notified of inpatient consults and will be responsible for comprehensive reports to be reviewed and approved by the attending geneticist. Residents will participate in every clinic session.

The Clinical Genetics rotation also serves as an elective for fourth year medical students and for fellows in Reproductive Medicine. Other participants in Genetics Clinic sessions may include genetic counselors in training and M.D./PhD students with specific orientation for a career in genetics.

**Questions & Concerns:**

- o Please contact \_\_\_\_\_? with any questions, concerns or clarifications regarding this elective.