**MEDICAL GENETICS ROTATION**

**STANFORD** **UNIVERSITY** **SCHOOL OF MEDICINE**

**DEPARTMENT OF PEDIATRICS**

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# Rotation Contacts and Scheduling Details

**Rotation Director:** Jon Bernstein, M.D., Ph.D.

  E-mail:              [jon.bernstein@stanford.edu](mailto:lhudgins@stanford.edu)

  Phone: 498-4937

  Pager: 23163

  Office Location: A097

**Educational Coordinator/Senior Genetic Counselor:**

Susan Schelley, MPH

Phone:  723-6858

Pager: 15460

Email:  [schelley@stanford.edu](mailto:schelley@leland.Stanford.EDU)

**Positions Available**:  1 spot each month, no training level preference,

              4 week rotation strongly encouraged; 2 week rotation possible

Contact Susan Schelley 3 to 4 months in advance of the rotation.  Please note that the rotation will try to accommodate individuals with less than 3 to 4 months notice; however, given the division’s obligations to other trainees in the medical school and genetic counseling program, this may not always be possible.

**Introduction**

The rotation provides a comprehensive introduction to the field of clinical genetics.  Residents have the opportunity to learn about a range of genetic disorders, genetic diagnostics and genetic counseling by participating in the evaluation of children in the General Genetics Clinic, Metabolic Genetics Clinic and Craniofacial Anomalies Clinic.  Residents are also encouraged to participate in the activities of the inpatient consult service.  By special arrangement residents may also observe in the Neurogenetics Clinic, Genetics of Skin Disorders Clinic, Down Syndrome Clinic or the Prenatal Genetics Clinic.  At the conclusion of the rotation, students will deliver a thirty minute presentation on a topic of their interest in genetic medicine.

The role the resident plays on the rotation may vary between that of an observer and active learner.  We encourage residents to take an active role, participate in all consults, and take the primary history and physical on several patients.  However, there will be situations in which the resident will not be the primary provider and will take an observer role.

**Weekly Schedule**

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| --- | --- | --- | --- | --- |
| Monday | Tuesday | Wednesday | Thursday | Friday |
| 8 – 8:30am  Morning Report | 8 – 8:30am  Morning Report | 8 – 8:30am  Morning Report | 8 – 8:30am  Morning Report | 8 – 9am  Grand Rounds |
| 9am – noon  General Genetics Clinic  (730) Welch) | 9 am – noon  General Genetics  Clinic  (730) Welch) | 8:30am – noon  Metabolic Clinic or Craniofacial Anomalies Clinic  (730 Welch) |  | 9:00am-10am  Medical Genetics Grand Rounds  LK101    10am  Neurogenetics Clinic  (1 clinic per month) |
| Noon Conference | Noon Conference | Noon Conference | 12:30-1:30pm  Prenatal Genetics Clinical Conference | 12pm-1pm  Perinatal Conference |
| 1:15pm-2pm  Metabolic Conference (A-051) |  | 1:30-2:30pm  NICU Genetics Teaching Rounds  (LPCH)  4-4:30pm  Molecular Genetics Journal Club LK120 |  |  |

# Rotation Specifics

# Orientation

Please contact Susan Schelley via email or voice mail prior to the first day of the rotation for instructions on where and when to begin the rotation.  Please email Jon Bernstein in advance of the rotation to arrange a meeting within the first 3 days of the rotation and then a follow-up meeting at the beginning of the 2nd week of the rotation. During these meetings, goals for the rotation will be reviewed.

# Rounds or Clinic Overview (or both if appropriate)

The Genetics rotation consists of both inpatient and outpatient experiences.  Typically, the consult service will receive 4-12 consults per week.  The resident may take the lead in gathering information and performing the history and physical as the consultant.  The resident will typically present the case to the fellow and then again present to the attending on rounds.  Since the service is small, rounds may be held at different times each day.  The resident should be available by pager and respond to pages if a consult is needed in the afternoons.

Each week, the upcoming clinic case load is reviewed and residents and fellows are assigned cases.  Residents are expected to create a differential diagnosis and review appropriate readings in advance.  For the first week of the rotation, residents are encouraged to contact Susan Schelley or Jon Bernstein to get patient assignments for the clinic.  Similar to the inpatient role, residents will often be asked to take a history and physical; dictations will be performed by the Fellow or Attending.    Follow-up labs will be tracked by the Fellow or Attending but residents are encouraged to follow-up on these out of clinical interest.

**Call Schedule**

There are no call responsibilities associated with the Genetics Selective.  If residents are taking call while on this rotation, they should provide Susan Schelley with call schedule and anticipated post-call absences at the start of the rotation.

**Weekend/Evenings**

There are no clinical duties for residents on the weekend or evenings

# Resident Roles, Responsibilities and Expectations

1.      Attend all outpatient genetics clinics

2.      The resident will perform a medical history, family history and dysmorphology examination of at least one patient per outpatient clinic session. The resident will formulate a differential diagnosis and diagnostic plan for the patient(s), and (under the supervision of the faculty preceptor) the resident will participate in the genetic counseling and treatment planning for the child (children).

3.      The resident will participate in performing inpatient consultations with the on-call attending and medical genetics resident

4.      The resident will attend all teaching conferences of the Division

5.      The resident will give a one 30 minute presentation on a topic of his/her choice at Medical Genetics Grand Rounds.  The topic for this talk should be reviewed with Dr. Bernstein in advance of preparation.

6.      Attend clinics, teaching and clinical conferences regularly and in a timely manner.

7.      Be available to participate in inpatient consultations.

8.      Communicate effectively with patients, families, and colleagues.

**Evaluation and Feedback**

Methods for evaluations will consist of:

        Resident performance with be evaluated using rating scales and narrative comments provided though the MedHub evaluation system.  Specifically, Susan Schelley will submit a Group Evaluation of the Resident after obtaining collective feedback from Genetics Faculty.

        Residents will also have opportunity to evaluate the selective and selective faculty using the MedHub evaluation system.

**Competency-based Goals and Objectives**

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| --- | --- | --- | --- |
| **Goal 1.  Understand the diagnostic approach to children with congenital anomalies.** | | | |
| Resident Objectives: | Instructional Strategies | Evaluation | ACGME Competency Goals |  |
| 1. Obtain and record a comprehensive birth history including pertinent details of maternal health, exposures and prenatal diagnostic studies. | Conduct interview and present to fellow.  Observe additional questions asked by Fellow and Attendings and reflect on what can be improved on future histories. | Verbal feedback from Attending | MK - Demonstrate an investigatory and analytic thinking approach to clinical  PC - Gather essential and accurate information about their patients  PC - Interview patients/families about the particulars of the medical condition for which they seek care, with specific attention to behavioral, psychosocial, environmental and family unit correlates of disease  P - Demonstrate sensitivity and responsiveness to patients’ culture, age, gender and disabilities  ICS - Use effective listening skills and elicit and provide information using effective nonverbal, explanatory, questioning, and writing skills  CS - Work effectively with others as a member or leader of a health care team or other professional group |  |
| 2. Perform multiple comprehensive physical examinations with attention to minor anomalies and congenital malformations | Perform exam independently then review findings with Fellows and Attendings in clinic and inpatient consultations.  Maintain list of all abnormal physical findings seen during the month and consider associated differential diagnoses. | Self-reflection  Attending feedback | ICS - Create and sustain a therapeutic and ethically sound relationship with patients  PC - Perform complete and accurate physical examinations |  |
| 3. Record a detailed family history in the form of a 3-generation pedigree. | Review pedigrees performed by Fellows and Attendings on consults  Review Pedigree Handout and article  Prenatal genetics conference | Review constructed Pedigree with Attending or Fellow | MK - Demonstrate an investigatory and analytic thinking approach to clinical situations |  |
| 4. Review pertinent medical literature in advance of patient visit.  Develop familiarity with electronic resources relevant to pediatric genetics. | OMIM  Gene Reviews  Clinical cases  Prenatal genetics conference  30 minute presentation at rotation conclusion | Attending feedback related to preparation for selected patients  Feedback from group on Grand Rounds presentation | MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatric  PC - Use information technology to support patient care decisions and patient education  PBLI Obtain and use information about their own population of patients and the larger population from which their patients are drawn  PBLI Locate, appraise, and assimilate evidence from scientific studies related to their patients’ health problems  PBLI Apply knowledge of study designs and statistical methods to the appraisal of clinical studies and other information on diagnostic and therapeutic effectiveness  PBLI Use information technology to manage information, access on-line medical information; and support their own education |  |
| **Goal 2.  Understand the diagnostic approach to children with inborn errors of metabolism.** | | | |
| Resident Objectives: | Instructional Strategies | Evaluation | ACGME Competency Goals |  |
| 1.      State initial management for patients with suspected metabolic disorder. | Study Enns article “Diagnosing inborn errors of metabolism”  Learning Module 2: IEM  Discussion with Attending/Fellows | Medhub  Self-assessment | MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics |  |
| 2.      Comprehend the hyperammonemia order set including the rationale for the diagnostic tests. | Review hyperammonemia order set.  Learning Module 2: IEM  Study Enns article “Diagnosing inborn errors of metabolism”  Clinical consult when available | Medhub  Self-assessment | MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics  PBLI Use information technology to manage information, access on-line medical information; and support their own education  · |  |
| 3.      Review the California Newborn Screen.  Recognize which diseases are and are not screened for.  State 10 diseases that are screened and 5 which are not. | Study Primer on Expanded Newborn Screening  Learning module 2: IEM  Review the California Newborn Screen website.  http://www.cdph.ca.gov/programs/NBS/Pages/default.aspx. | Fellow/Attending discussion | PBLI Use information technology to optimize learning\  MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics |  |
| 4.      Interperet Urine Organic Acids/Serum Organic Acids/Acylcarnitine profile. | Clinical consultations  Learning Module 2: IEM | Fellow/Attending discussion | MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics |  |
| **Goal 3.  Understand the principles of inheritance and genetic counseling.** | | | |
| Resident Objectives: | Instructional Strategies | Evaluation | ACGME Competency Goals |  |
| 1.      Define the modes of genetic inheritance. | Learning Module 3: Cleft lip and palate  Genetics prenatal conference | Fellow/Attending feedback  Self-assessment | MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics  · |  |
| 2.      Construct a 3 generation pedigree for a patient that you will see in clinic.  Predict the most likely mode of inheritance. | Genetics prenatal consult  Outpatient clinical encounter | Fellow/Attending feedback  Self-assessment | MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics  PC - Provide family-centered patient care that is culturally effective and developmentally and age-appropriate  PC - Interview patients/families about the particulars of the medical condition for which they seek care, with specific attention to behavioral, psychosocial, environmental and family unit correlates of disease  PBLI Use information technology to manage information, access on-line medical information; and support their own education  · |  |
| **Goal 4.  Be familiar with health supervision for children with common genetic disorders or minor anomalies, specifically:** | | | |
| Resident Objectives: | Instructional Strategies | Evaluation | ACGME Competency Goals |  |
| 1.  Downs Syndrome:  State the medical complications of Down Syndrome and the suggested screening labs/studies. | Attend Down’s Syndrome Clinic  Review AAP Guidelines: Health Supervision for Children with Down Syndrome | Self-assessment  Clinical feedback | PBLI Obtain and use information about their own population of patients and the larger population from which their patients are drawn  MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics  PC - Make informed decisions about diagnostic and therapeutic interventions based on patient information, preferences, up-to-date scientific evidence, and clinical judgment  PC - Provide effective health maintenance and anticipatory guidance |  |
| 2.  Neurofibromatosis:  Define the clinical findings and associated complications of neurofibromatosis. | Review AAP Guidelines: Health Supervision for Children with Neurofibromatosis  Clinical cases | Self-assessment  Clinical feedback | PBLI Obtain and use information about their own population of patients and the larger population from which their patients are drawn  MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics  PC - Make informed decisions about diagnostic and therapeutic interventions based on patient information, preferences, up-to-date scientific evidence, and clinical judgment  PC - Provide effective health maintenance and anticipatory guidance |  |
| 3.  Single Umbilical Artery | Study The Importance of Minor Anomalies in the Evaluation of the Neweborn (Hudgins, Neoreviews, 2003)  Prenatal Genetics Conference | Self-assessment  Clinical feedback | PBLI Obtain and use information about their own population of patients and the larger population from which their patients are drawn  MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics |  |
| 4.  Sacral Dimple: State how one would appropriately manage a sacral dimple found on a healthy newborn exam.  Specify the clinical significance of a sacral dimple, clarify which dimples are of concern, generate a differential diagnosis for sacral dimple. | Study The Importance of Minor Anomalies in the Evaluation of the Neweborn (Hudgins, Neoreviews, 2003)  Generate the differential diagnosis for sacral dimple. | Self-assessment  Clinical feedback | PBLI Obtain and use information about their own population of patients and the larger population from which their patients are drawn  MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics  PC - Make informed decisions about diagnostic and therapeutic interventions based on patient information, preferences, up-to-date scientific evidence, and clinical judgment  · |  |
| 5.  Ear pits or tags | Study The Importance of Minor Anomalies in the Evaluation of the Neweborn (Hudgins, Neoreviews, 2003) | Self-assessment  Clinical feedback | PBLI Obtain and use information about their own population of patients and the larger population from which their patients are drawn  MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics  PC - Make informed decisions about diagnostic and therapeutic interventions based on patient information, preferences, up-to-date scientific evidence, and clinical judgment  PC-Provide effective health maintenance and anticipatory guidance |  |
| 6.  Single palmar crease | Study The Importance of Minor Anomalies in the Evaluation of the Neweborn (Hudgins, Neoreviews, 2003) | Self-assessment  Clinical feedback | PBLI Obtain and use information about their own population of patients and the larger population from which their patients are drawn  MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics  PC - Make informed decisions about diagnostic and therapeutic interventions based on patient information, preferences, up-to-date scientific evidence, and clinical judgment |  |
| 7.Syndactyly/polydactyly | Study The Importance of Minor Anomalies in the Evaluation of the Neweborn (Hudgins, Neoreviews, 2003) | Self-assessment  Clinical feedback | MK - Demonstrate an investigatory and analytic thinking approach to clinical situations  MK - Demonstrate sufficient knowledge of the basic and clinically supportive sciences appropriate to pediatrics  PC - Make informed decisions about diagnostic and therapeutic interventions based on patient information, preferences, up-to-date scientific evidence, and clinical judgment  PC - Provide effective health maintenance and anticipatory guidance |  |
| **Goal 6.  Be familiar with community resources for children with birth defects and genetic disorders.** | | | |
| Resident Objectives: | Instructional Strategies | Evaluation | ACGME Competency Goals |  |
| 1.  State the services provided by Occupational Therapy versus Physical Therapy. | Create a list of appropriate referrals for services for one of the patients with a genetic disorder that you see in clinic.  Attend Craniofacial Clinic  Attend Down’s Clinic | Self-evaluation | SBP - Advocate for quality patient care and assist patients in dealing with system complexities  SBP - Work in inter-professional teams to enhance patient safety and improve patient care  · |  |
| 2.  Describe the state requirements regarding IEP, a patient/families rights/ and the role of the physician. | If available, discuss the IEP of one of your patients in clinic.  Review article “Pediatrician’s Role in Developing and Implementing IEP” | Attending and multidisciplinary team feedback. | SBP - Advocate for quality patient care and assist patients in dealing with system complexities  ICS - Communicate effectively with physicians, other health professionals, and health related agencies  · |  |
| 3.  Describe who qualifies for the Regional Center and Early Start.  Explain how you would refer one to these centers. | Review case of your selected patient.  Discuss with attending whether they qualify for Regional Center. | Attending and multidisciplinary team feedback. | SBP - Advocate for quality patient care and ssist patients in dealing with system complexities |  |
| 4. Locate family support groups in the Bay Area. | Generate a list of support groups and resources for a selected patient you saw in clinic. | Discuss your list of recommended resources with the care team. | PBLI Participate in the education of patients, families, students, residents and other health professionals |  |

PBLI = practice based learning and improvement

ICS = interpersonal and communication skills

P= professionalism

MK= medical knowledge

PC= patient care

SBP = systems based practice

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