

## Genetics Elective Rotation Expectations and Procedures

For this rotation you will need to:

- Set up a meeting with Gustavo Maegawa (see contact information below) during the week ***prior*** to start of the rotation to complete learning contract and review expectations for the rotation.
- Schedule your genetics rotation for a consecutive 3-week block.
- Take no more than 1 work week off during your rotation.
- Attend at least 3 full-day clinics. This will constitute a minimum of **2** full-day clinics and **2** half-day clinics.
  - You are excused from Tuesday academic half-days as they conflict with clinic.
  - If your continuity clinic conflicts with clinic time, you must reschedule your continuity clinic if you are unable to meet minimum requirements for the rotation.
- Participate in Newborn Screening Program and meet with dietician on call as noted below.
- Be available in a timely manner for consults during rotation.
- Complete Extended Learning Project as noted below.
- Meet with one of our Attendings at the end of your rotation for a knowledge-based assessment as noted below.

### Resident Orientation

Genetics is an important elective rotation during a resident's training since patients with genetic conditions are seen by all specialists. It's important to have the resources and knowledge to work with these patients and to know when to refer for a genetics consultation. Each resident's goals for this rotation will differ based on where they are in their training. Prior to beginning your rotation, please contact Gustavo Maegawa at [gmaegawa@ufl.edu](mailto:gmaegawa@ufl.edu) to discuss rotation goals and expectations and to complete learning contract.

Residents will get out of this rotation what they put in. We are here as resources to help you navigate the complex world of genetics. Some of our basic objectives for our residents include:

- Be able to perform a clinical genetics evaluation and appropriate physical examination with attention to dysmorphism.
- Be able to take a detailed, targeted family history and recognize inheritance patterns in the pedigree.
- Be able to recognize common molecular, cytogenetic and biochemical syndromes.
- Become familiar with common cytogenetic, molecular and biochemical tests that are typically ordered to evaluate genetic disorders.
- Be able to recognize common medical and familial issues that warrant a referral to genetics for further evaluation.

Please review the following information to learn what to expect during this rotation.

## **Opportunities during Genetics Rotation:** outpatient clinics-

Gainesville and outreach, in-patient consults, newborn screening, resident projects

### **Outpatient Clinics**

You are expected to attend at least 3 full-day clinics within a **consecutive** 3 week block. This will constitute a minimum of **2** full-day clinics and **2** half-day clinics. Residents can, and should of course, attend more clinics than the minimum requirement.

### **Clinic Schedule**

- We have genetics clinics Tuesday from 8:00am until approximately 5:00pm at the Medical Plaza. You are expected to be at clinic for the full clinic day. If your continuity clinic conflicts with a general genetics clinic, you must reschedule your continuity clinic.
- We have genetics clinics the second Monday of the month and weekly on Wednesdays and Fridays from 8:00 am to approximately 5:00 pm at the Children's Medical Serviced building.
- We have craniofacial clinic Thursday afternoons (genetic counselor with multidisciplinary team. Please contact Heather Stalker directly to arrange attending this clinic).
- We have outreach and or Florida School for the Deaf and Blind clinics most months.

\*\*\*See posted schedule for more details.

### **Location**

- All Tuesday Gainesville clinics are located on the 2nd floor of the Medical Plaza in the Pediatric Specialty Clinic. Tuesday Workspace is in the back of clinic on the left, next to Cardiology's conference room
- All Monday, Wednesday, Friday clinics are at the Children's Medical Services building. Workspace is shared with endocrinology (room to right down your first hallway on right after entering CMS building).
- Craniofacial Workspace- conference room in back right corner
- Outreach clinics are in Daytona, the Panhandle (2-3 night trip), and St. Augustine at the Florida School for the Deaf and Blind (day trip).

### **Prior to a genetics clinic:**

- Email Genetic Counselor on schedule for clinic schedule and to set up clinic review.
- Clinic Review- typically 1-2 days before clinic with attending to discuss patients and plans

- Use resources attached and recommended literature to review conditions *prior* to clinic

### **Possible Roles in general genetics clinic**

As your comfort level with clinic increases, you will take on more roles with our patients. These possible roles include:

- Observe genetic counselor- medical intake, family history, genetic counseling (inheritance, recurrence risk, natural history, psychosocial counseling etc)
- Case Prep
- Review of Systems and Medical Intake
- Take Family History
- Perform Physical Exam
- Case Presentation to attending including discussion of differentials and potential testing

### **Consults**

#### **On call hours**

- Monday through Friday between 8:00am and 5:00pm unless medical emergency.

#### **Consult Structure**

1. Consult is called into our division. Genetic Counselor preps case and may see patient prior to attending. Please be available when Genetics Office Staff calls/pages to help prep and see the case. If you do not respond or cannot come, we will call the Chief Resident's phone and notify them. Residents are expected to see the inpatient consults with the genetic counselor and follow-up accordingly with the attending physician. If the consult is to be seen the following day, you are expected to be there unless there are extenuating circumstances (i.e. clinic, pre-arranged vacation day, medical emergency). Failure to attend these consults may result in the loss of a sick/vacation day, as decided upon by the Chief Resident.

#### **Roles on Consults:**

For most consults you will be expected to:

- Collect all information on the patient.
- Examine the patient
- Develop differential diagnoses and possible recommendations for testing and screening
- Type a summary for attending's EPIC note
- Attend/Observe family meetings if set up for a known diagnosis

### **In-patient Admissions**

The timing and amount of in-patient admissions we have under our service varies throughout the year. Typically, these patients are our metabolic patients. We will keep you informed if we have a patient admitted under our service.

## **Newborn Screening**

### **Newborn Screening Timeline**

- During second week of your rotation, set up meeting with dietitian on call to discuss Newborn Screen Program.

### **Newborn Screening Overview**

- We receive positive metabolic newborn screens for all areas of Florida north of Orlando.
- Screens come in from the State Laboratory, Monday through Friday from 8:00am-5:00pm.
- For each screen we: review the screen results; contact the PCP to order confirmatory testing and/or bring family into genetics clinic to complete testing if local; contact the family to disclose screen results, follow results through the lab; and disclose final diagnosis based on confirmation testing.

### **Possible Roles for Newborn Screen:**

- Observe and participate in work-up of newborn screens

## **Genetics Meetings**

- We hold monthly genetics meetings for case presentations, discussions, and literature reviews.

## **Resident Extended Learning Projects**

Each rotating resident is expected to complete an Extended Learning Project (ELP). Please discuss with Resident Director at beginning of rotation. Possible ELPs include:

- Case Presentation
- Literature Review
- Update selected information on Genetics Website
- Compose learning questions for readings (see below)
- Write an RCPU newsletter
- Compile a patient resource packet on a specific condition

All Resident Extended Learning Projects require a PowerPoint presentation. The rough draft PowerPoint presentations will be due one week prior to the presentation date for review by the Resident Director.

### **Recommended Readings and Knowledge-Based Assessment**

Upon the completion of your rotation, you will meet with one of the Attendings for a recap of the rotation and to complete a knowledge-based assessment, which is a conversation on what you have learned throughout your rotation. You will be provided with a packet of required readings at the beginning of your rotation. You are expected to read each article and will be held accountable for the information in that packet during your knowledge assessment. Part of your evaluation will depend on the knowledge gained during your rotation. Please schedule this meeting on the last day of your rotation.

### **Recommended Reading:**

- Case Examples- cytogenetics and molecular genetics – includes overview of resources
- Copy Number and SNP arrays in clinical Diagnostics- review of microarrays
- Initial evaluation of Inborn Errors of Metabolism
- Patient Flow Example

### **Contact information**

Genetics Main Office 352-294-5050/5051  
Metabolic On-Call Phone 352-681-7040

### **Attendings**

Roberto Zori	<a href="mailto:zorirt@peds.ufl.edu">zorirt@peds.ufl.edu</a>	352-294-5036
Daniel Driscoll	<a href="mailto:driscdj@peds.ufl.edu">driscdj@peds.ufl.edu</a>	352-294-5446
Cheryl Garganta	<a href="mailto:cgarganta@ufl.edu">cgarganta@ufl.edu</a>	352-294-8525
Gustavo Maegawa	<a href="mailto:gmaegawa@ufl.edu">gmaegawa@ufl.edu</a>	352-294-5559
Sonja Rasmussen	<a href="mailto:Sonja.Rasmussen@peds.ufl.edu">Sonja.Rasmussen@peds.ufl.edu</a>	352-273-5934

### **Genetic Counselors**

Heather Stalker	<a href="mailto:stalkhj@peds.ufl.edu">stalkhj@peds.ufl.edu</a>	352-294-5042
Amy Jonasson	<a href="mailto:argladstone@ufl.edu">argladstone@ufl.edu</a>	352-294-5335
Sarah Nimrichter	<a href="mailto:snimrichter@ufl.edu">snimrichter@ufl.edu</a>	352-294-5039
Eric Traub	<a href="mailto:etraub@ufl.edu">etraub@ufl.edu</a>	352-294-5038

### **Dietitians**

Krista Mantay	<a href="mailto:mantak@shands.ufl.edu">mantak@shands.ufl.edu</a>	352-294-5037
Alexa Bledsoe	<a href="mailto:abledsoe@peds.ufl.edu">abledsoe@peds.ufl.edu</a>	352-297-9233
Anna McGuinness	<a href="mailto:anna.mcguinness@ufl.edu">anna.mcguinness@ufl.edu</a>	352-294-5040