



## **PROGRAM**

**AND**

## **SIG MEETING SCHEDULE**

**Sheraton Sand Key Resort**

**CLEARWATER BEACH, FL**



## AGENDA

### Wednesday, May 01

7:00 am – 5:00 pm <i>Lobby II</i>	<b>APHMG Registration Desk Open</b> Staff: Sheilah Jewart
7:00 am – 8:00 am <i>Lobby II</i>	Continental Breakfast
8:00 am – 4:00 pm <i>Beach Room</i>	<b>Medical Genetics Residency Program Directors SIG Meeting</b>
11:30 am – 1:00 pm <i>Tent on Beach</i>	Lunch – Included
12:30 pm – 5:30 pm <i>Sand Key Room</i>	<b>Medical School Genetics Course Directors SIG Meeting</b>
1:15 pm – 4:30 pm <i>Gulf Room</i>	<b>Clinical Laboratory Training Program Directors SIG Meeting</b>
5:00 pm – 6:00 pm <i>Coquina – 3<sup>rd</sup> Fl</i>	<b>APHMG Council Meeting</b> Closed Meeting
6:00 pm – 7:00 pm <i>Beach/Gulf Room</i>	<b>APHMG Welcome Reception</b> <i>Visit Posters</i>
7:00 pm – 8:00 pm <i>Tent on Beach</i>	<b>APHMG Welcome Dinner</b>
8:00 pm – 9:30 pm <i>Beach/Gulf Room</i>	<b>APHMG Business Meeting</b> Dessert

## **Thursday, May 02**

7:00 am – 2:00 pm  
*Lobby II*

**APHMG Registration Desk Open**  
Staff: Sheilah Jewart

7:00 am – 8:00 am  
*Lobby II*

Continental Breakfast

8:00 am – 9:30 am  
*Beach/Gulf Room*

**Plenary Session I**  
**Examining Race, Ethnicity and Ancestry  
in Clinical Care**  
*Moderator: Shoumita Dasgupta*

**Workshop**  
**Healthcare Provider's Use of Race and  
Ethnicity: Where do we go from here?**  
•Vence Bonham, JD  
*National Human Genome Research Institute*  
•Alice Popejoy, PhD  
*Stanford University*

10:00 am – 10:30 am

Break

10:00 am – 11:30 am

**Plenary Session II**  
*Moderator: Shoumita Dasgupta*

*Beach/Gulf Room*

**Summary of NBME Visit**  
•Darrel Waggoner, MD  
*University of Chicago School of Medicine*

11:30 am – 12:30 pm  
*Tent on Beach*

Lunch – Included

12:30 pm – 1:45 pm  
*Beach/Gulf Room*

**Plenary Session III**  
*Moderator: Cindy Powell*

**Utilization of Other Healthcare Providers  
In Clinical Genetics**  
•Donald G. Basel, MD  
*Children's Hospital of Wisconsin*

2:00 pm  
*Depart from Lobby II*

**Optional Excursion**  
Salvador Dali Museum

6:30 pm – 9:30 pm  
*Tent on Beach*

**APHMG Beach Party**

## **Friday, May 03**

7:00 am – 3:00 pm  
*Lobby II*

### **APHMG Registration Desk Open**

Staff: Sheilah Jewart

7:00 am – 8:00 am  
*Lobby II*

Continental Breakfast

8:00 am – 9:30 am  
*Beach/Gulf Room*

### **Plenary Session IV**

*Moderator: Kathy Hyland*

### **Making it Count Twice: Transform Your Genetics Teaching into Publications**

•*Sebastian Uijtdehaage, PhD*

*Uniformed Services University of the Health Sciences*

9:30 am – 10:00 am

Break

10:00 am – 11:00 am  
*Beach/Gulf Room*

### **Plenary Session V**

*Moderator: Donald Basel*

### **Artificial Intelligence and Genomic Analysis**

•*Bret Bostwick, MD*

*Baylor College of Medicine*

11:00 am – 11:30 am

### **Closing Remarks**

•*Cindy Powell, MD*

11:30 am – 12:30 pm  
*Beach Tent*

Lunch – included

12:30 pm – 3:00 pm  
*Beach/Gulf Room*

### **The TRIG Model for Teaching Genomic Medicine: A Train-the-Trainer Workshop**

•*Richard Haspel, MD, PhD*

*Chair, TRIG Working Group*

*Beth Israel Deaconess Medical Center*

•*Leah Burke, MD*

*Professor of Pediatric Geneticist*

*University of Vermont Medical Center*

# Medical Genetics Residency Program Directors SIG Meeting

## AGENDA

### Wednesday, May 01

8:00 am – 8:10 am	<b>Welcome &amp; Introductions</b> <i>•Hope Northrup, SIG Chair</i>
8:10 am – 8:30 am	<b>Report on the 2018 Match</b>
8:30 am – 9:00 am	<b>Update on the in-service Exam</b> <b>Report from ABMGG, including LGG Update</b> <i>•Mimi Blitzer, CEO ABMGG</i> <i>University Maryland School of Medicine</i>
9:00 am – 9:40 am	<b>Reports from the RRC/ACGME, Milestones Update</b> <i>•Laurie Demmer, Levine Children's Hospital</i> <i>•Kate Hatlak, ACGME</i>
9:40 am – 10:00 am	<b>Q&amp;A for ABMCG / ACGME</b>
10:00 am – 10:30 am	<b>Break</b>
10:30 am – 12:00 pm	<b>Plenary Session I</b> <b>Electronic Resources for Genomics Education</b>
12:00 pm – 1:00 pm <i>Tent on Beach</i>	<b>Lunch</b>
1:15 pm – 2:45 pm	<b>Plenary Session II</b> <b>Interactive Joint Session with Laboratory Directors</b> <b>Meeting to discuss Laboratory Rotations for Trainees</b>
2:45 pm – 3:45 pm	<b>Workshop for additional discussion about required Laboratory Rotations</b>

# Clinical Laboratory Training Directors SIG Meeting

## AGENDA

### Wednesday, May 01

11:30 am – 1:00 pm  
*Tent on Beach*

Lunch – included

1:00 pm – 1:10 pm  
*Sand Key Room*

#### **Welcome**

•*Stephen Moore, PhD, SIG Chair*

1:15 pm – 2:45 pm  
*Gulf/Beach Room*

#### **Plenary Session I**

**Combined Residency Directors and Laboratory  
Directors to discuss Laboratory Rotations for Trainees**

3:00 pm – 3:45 pm  
*Sand Key Room*

#### **Report of the ABMGG**

•*Mimi Blitzer, University Maryland School of Medicine*

3:45 pm – 4:45 pm

#### **ACGME Discussion on Transition**

•*Miriam Blitzer, University Maryland School of Medicine*

•*Laura Edgar, ACGME*

•*Kate Hatlak, ACGME*

4:45 pm – 5:00 pm

Wrap-up

# Medical School Genetics Course Directors SIG Meeting

## AGENDA

### Wednesday, May 01

11:30 am – 12:30 pm  
*Tent on Beach*

**Lunch**

12:30 pm – 12:40 pm  
*Gulf Room*

**Announcements & Updates**

• *Jon Bernstein, SIG Chair*

12:40 pm – 1:55 pm

**Integration Genetics and Genomics in Clinical Years: Opportunities and Challenges**

• *Kim Dahlman, PhD, Vanderbilt University*

• *Kathy Hyland, PhD, University California, SF*

1:55 pm – 2:40 pm

**Teaching about race, ethnicity and ancestry: rationale and a practical approach**

• *Alice Popejoy, PhD, Stanford University*

2:40 pm – 2:50 pm

**Break / Poster Viewing**

2:50 pm – 3:35 pm

**Presentation of Selected Abstracts:**

Moderator: *Erin Strovel, PhD*

**Graded Discussion Posts and Their Role in Active Learning**

• *Tracey Weiler, PhD, Florida International University*

**Utilizing Small Group Sessions to Enhance Genetics Curriculum Understanding**

• *Lauren Massingham, MD, Warren Alpert Medical School of Brown University*

**Using Team-Based Learning to Introduce First Year Medical Students to Medical Genetics Databases**

• *Hana Anderson, PhD, University California, Davis*

3:35 pm – 5:05 pm

**Workshop on Curriculum Guidelines for Genetics in Medical Student Education**

• *Sabrina Nunez, PhD, Washington University, St. Louis*

• *Joe Bernstein, MD, PhD, Stanford University*

5:05 pm – 5:25 pm

**Poster Viewing**

5:25 pm – 5:30 pm

**Wrap Up / Reflections**

• *Jon Bernstein, SIG Chair*



**APHMG BUSINESS MEETING**  
**Wednesday, May 01, 2019**  
**8:00 pm – 9:30 pm – Beach/Gulf Room**

**AGENDA**

**Welcome** Cindy Powell  
**Secretary-Treasurer Report** Katherine Hyland

**SIG Updates**

Residency Program Directors SIG Hope Northrup  
Course Directors SIG Jon Bernstein  
Lab Training Program Directors SIG Stephen Moore

**Liaison Reports**

ACMG Mike Watson  
ASHG Mona Miller  
ABMGG Mimi Blitzer  
FASEB Capitol Hill Day Shoumita Dasgupta  
ISCC Shoumita Dasgupta  
Aquifer Sciences Project Kathy Hyland  
NHGRI Genomic Education for Cindy Powell  
Healthcare Providers

**Other Business:**

Nominations Committee Report Darrel Waggoner  
  
2020 Workshop Cindy Powell  
Open Discussion

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## **2019-20 OFFICERS AND COUNCIL**

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**Darrel Waggoner, MD**

University of Chicago



## ABSTRACTS

### **Promoting Interest in Medical Genetics through a Student-led Interest Group: An Interprofessional Initiative**

Zoë Mackay, BS<sup>1</sup>, Noreen Siddiqi<sup>1</sup>, MS, Reid McMurry<sup>1</sup>, MS, Shoumita Dasgupta, PhD<sup>2</sup>

<sup>1</sup>Boston University School of Medicine

<sup>2</sup> Boston University School of Medicine, Department of Medicine, Biomedical Genetics Section

**Background:** As genomic information is integrated into clinical practice, there is an increasing need for health care professionals to be well-versed in the approaches of genomic medicine. Currently there are 2 clinical geneticists per 1 million people in the United States. According to the American College of Medical Genetics and Genomics, only 50% of available clinical genetics training slots in the United States are routinely filled. The scarcity of clinical geneticists is a barrier to care for people with genetic conditions, and the shortage of patient-facing genetic counselors further amplifies this problem.

If the growing need for clinical geneticists is to be met, medical schools must facilitate interest in the field through enrichment and professional development opportunities. The Genetics Interest Group (GIG) was started in fall 2018 at Boston University School of Medicine (BUSM) to meet this need. The vision of this organization is to promote the equitable implementation of genetic testing and therapy in clinical practice by educating future providers.

**Methods:** The GIG hosted a podcast discussion and three guest-speakers to discuss Hemophilia, Huntington's disease, and the ethics of gene-editing, participated in a community health fair and Career Expo. The GIG also made genetics shadowing and research opportunities available to students. Both genetic counseling and medical students participated in this interprofessional group. At the end of the semester, all participants were surveyed.

**Results:** Survey results indicate that students were most interested in guest-speakers. When asked, the majority of students indicated their primary reason for participating was to learn from patients and advocates. Sessions featuring guest-speakers also had the highest attendance rates. Important outcomes reported by many students include increased interest in medical genetics and improved understanding of the role of genetic testing and therapy in clinical practice.

**Conclusions:** These findings inform strategies for engaging students in genomic medicine through student interest groups. Guest-speakers most successfully promote interest and engagement. This suggests that future providers are interested in learning from patients with genetic conditions and may incorporate a knowledge of genetics into

their clinical practice. Interprofessional interest groups also encourage critical partnership across the health professions beginning at the trainee stage.

Keywords: Medical education, Interest group, Interprofessional

## Using team-based learning to introduce first year medical students to medical genetics databases

Hana Anderson<sup>1,2</sup> and Amy Studer<sup>3</sup>

University of California, Davis, Department of Internal Medicine<sup>1</sup>, Department of Cell Biology and Human Anatomy<sup>2</sup>, Blaisdell Medical Library<sup>3</sup>

**Background:** The ability to identify reliable information for diagnosis and management of patients with genetic conditions is becoming an essential skill for clinicians. However, navigating online genetics resources can easily overwhelm first year medical students. Previously, we used didactic demonstration to introduce genetic database websites (e.g. OMIM) to students, but it was insufficient for student skill development. To address this, a team-based learning (TBL) session was designed and implemented to introduce first year medical students to well-established medical genetics databases and promote proficiency in information mining.

**Methods:** Prior to the TBL session, students completed both an IRAT covering medical genetics concepts from lecture and a vignette-based hands-on tutorial guiding them through OMIM to formulate a working diagnosis of Marfan syndrome. The tutorial included online answer submission (using Qualtrics survey software) enabling formative assessment of students' ability to effectively use OMIM. During the TBL session, 18 teams of 6-7 students engaged in active learning activities in a high-tech propeller room. First, the IRAT and pre-class tutorial were reviewed and important external links on OMIM (e.g., *GeneReviews*) highlighted. Then, using credible medical genetics resources, teams researched responses to questions about three cases (ADPKD, allopurinol hypersensitivity syndrome (AHS), and an optional pharmacogenetics case). To debrief, teams presented their findings with the larger group (ADPKD case) and submitted answers online for the remainder. Students were asked to complete a voluntary survey at the end of the session.

**Results:** The pre-class tutorial was highly effective in developing students' knowledge and skills in using medical genetics databases, with 109/112 students correctly identifying the working diagnosis using OMIM. Answer submission data for the AHS case confirmed the use of credible information sources [e.g., *Medical Genetics Summaries* (15 teams), OMIM (13), and Genetic and Rare Diseases Information Center - GARD (3)]. Eighty percent of student survey respondents reporting feeling more confident in using medical genetics databases at the end of the session (n=20).

**Conclusion:** There is demand for incorporating active learning and self-directed learning into pre-clinical curriculum. This structured TBL session provides students with an effective springboard for developing life-long learning skills in the rapidly changing field of human genetics.

Key words: team-based learning, medical genetics resources, self-directed learning

## **Making Epidemiology Relevant through Interdisciplinary Case-Based Teaching**

Cheryl McSweeney, MD, MPH<sup>1</sup>; Shoumita Dasgupta, PhD<sup>2</sup>; Sharon Phillips, MSc, MD<sup>3</sup>; Molly Cohen-Osher, MD, MMedEd<sup>1</sup>

<sup>1</sup>Boston University School of Medicine, Department of Family Medicine

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<sup>3</sup>Tufts Medical Center Community Care, Department of Family Medicine

**Background:** As educators, we are striving for methods that will make our teaching engaging and memorable for learners. The use of active learning methods has the potential to increase student retention and ability to apply learned concepts. Moreover, creation of interdisciplinary educational activities allows students to engage in application of material in the context of clinically-relevant situations.

**Methods:** In 2018 we developed an interdisciplinary case to blend the learning of epidemiology and biostatistics with medical genetics. Students were prompted to consider the value of offering genetic testing for hereditary breast and ovarian cancer, currently recommended for high risk individuals with a family history of the condition, to the general population, an extension of current screening recommendations recommended by some scientists and clinicians. The students use data about sensitivity and specificity to calculate the positive predictive value of this genetic test in different population settings. Students applied this information in small groups to a discussion of the ethics of genomic medicine and the implications of their calculations on health policy around the implementation of screening tests. Discussion was either conducted independently outside of class (2018) or was facilitated by a pair of experts in public health and genetics (2019). Student responses to the questions posed in the case were submitted and graded using a developed rubric to assess the quality of responses and the understanding and integration of the material. Students also completed an in-class survey about the small group experience.

**Results:** To assess the effectiveness of the case in increasing the retention of epidemiologic concepts, performance on a multiple choice exam given after the case was compared with the performance on the same exam from the year prior to implementation. Use of the case both independently and in facilitated groups improved performance on the exam.

**Conclusions:** The use of a case method that allows for the practice and then application of these concepts to relevant, real-life medical situations has the potential to increase the engagement of students in learning and their retention of the concepts. The methods we used can be applied to implement similar case-based approaches at other institutions integrating disparate topics into meaningful learning experiences.

**Keywords:** Interdisciplinary, Public Health, Cancer Predisposition Genetic Testing

### **Utilizing small group sessions to enhance genetics curriculum understanding**

Lauren Massingham MD, Chanika Phornphutkul MD, Thais Mather PhD, Kristina Monteiro PhD, Luba Dumenco, MD.

The Warren Alpert Medical School of Brown University

**Background:** Classically, lecturing has been the predominant mode of instruction in university settings, including medical schools. In recent years, active learning has become a more prominent teaching strategy. Freeman et al found that student performance on examinations and concept inventories was increased in the active learning group compared with traditional lecturing. Traditionally at our institution, the general impression of the medical students was that the genetics content is difficult to understand and overwhelming. A few years ago at APHMG, we presented newly developed short patient videos that focused on their genetic condition, medical treatments and the stresses this condition causes that were utilized in the first year core genetics curriculum. This was implemented to give the medical students some emotional or experiential context. In order to further enrich the curriculum, small group sessions designed to build upon the core curriculum and these video segments have been implemented. The goal of the small group sessions are to further enhance the preclinical learning and give “real world” examples for the students to connect with didactic material.

**Methods:** Two small group sessions were designed with three cases for each session. One case for each session was related to one of the videos. Each case was designed with a short case description and a few questions that related to the key concepts that we wanted to ensure the students understood. Feedback regarding the small group sessions was collected via survey at the end of the lecture series.

**Results:** The small group sessions were very well received. One of the cases discussed was phenylketonuria, which also included a taste testing session of some PKU formulas and foods that one student reported was a “visceral experience I won’t soon forget”. Another comment was that small groups were “helpful to clarify and explain concepts”.

**Conclusions:** Overall the feedback has been very encouraging that utilizing the affective (emotional) domain by incorporating videos and active learning via small group sessions enhances engagement in the material, with the goal of enhancing knowledge transfer and providing better patient care. To evaluate this active learning technique in a more objective manner we would like to implement this material multi-institutionally to enhance sample size and confer a more robust evaluation.

Key words: Active-learning, small groups, video

## **Genetics Primary Care Clinic Initiated by Combined Pediatric-Medical Genetics and Genomics Residents: A Potential National Model**

Kuntal Sen, MD<sup>1,2</sup> ; Shagun Kaur, MD<sup>1,2</sup> ; Yvonne Friday, MD<sup>1</sup> ; Michelle Cichon, CGC<sup>3</sup> ; Lynn Smitherman, MD<sup>1</sup> ; Gerald Feldman, MD PhD<sup>2,3,4</sup>

<sup>1</sup>Division of General Pediatrics and Adolescent Medicine, <sup>2</sup>Division of Genetics, Genomic and Metabolic Disorders, Children's Hospital of Michigan, <sup>3</sup>Center for Molecular Medicine and Genetics, <sup>4</sup>Departments of Pediatrics and Pathology, Wayne State University School of Medicine

**Background:** The four year combined pediatrics-medical genetics and genomics residency program currently requires a general pediatrics continuity clinic in the first two years of residency and a longitudinal genetics continuity clinic thereafter. As most genetics patients are seen less than or equal to once a year, the genetics clinic is limited in scope and patients. A primary care continuity clinic exclusively for patients with genetic disorders helps overcome this shortcoming, while providing an educational relevance for the combined trainees.

**Aim:** Our goal is to develop a primary care clinic for the care of individuals with confirmed genetic and metabolic disorders. The clinic is not meant to replace the regularly scheduled genetics clinic appointments with a clinical geneticist.

**Method:** Patient inclusion criterion is having a confirmed genetic condition. A list of potential patients currently seen at our institute using ICD-10 genetic disease codes was developed. The residents contacted the families to invite them to the clinic which is scheduled as a weekly ½ day clinic and staffed by a general pediatrician. New patients to the system are also referred using the same criterion. Patient/family surveys are provided to each family seen.

**Results:** Since Nov. 2018, 10 patients with chromosomal disorders, 8 with single gene disorders, 3 with inborn errors of metabolism, and 4 with other diagnoses have been scheduled. Nine patients were scheduled through direct contact, 2 from hospital referrals, 8 from pediatrics and 6 from genetic and metabolic clinics. Six patient surveys were completed showing positive response to the clinic.

**Conclusion:** The Genetics Primary Care Clinic represents a unique resident-initiated program that can set a new paradigm for the care of children with orphan diseases, while augmenting the educational experience for the trainees. We believe that this will satisfy training requirements and improve the care of such patients. To the best of our knowledge, such a model does not exist in the country currently.

Keywords – Primary care, Combined Pediatric-Medical Genetics and Genomics Residency

## **Graded Discussion Posts and Their Role in Active Learning**

Fabio Garrotte and Tracey Weiler

Herbert Wertheim College of Medicine, Florida International University

The Graduate Certificate in Molecular and Biomedical Sciences at FIU is a rigorous post-baccalaureate, pre-medical program including courses in Medical Molecular Biology and Medical Genetics. Graded discussion boards are a significant component of these courses. They are designed to improve critical thinking, develop active learning and improve communication skills.

**Part I** involves creating an original post regarding class content from the past two weeks. Students post an explanation of a topic in which they have gained a good understanding, after which they describe a topic area in which they do not feel confident about their understanding. Alternatively, they can post an analogy of something they have learned in those two weeks that would assist their peers' learning of the material.

**Part II** requires that students reply to **at least one** of their peers' posts, either explaining concepts or constructing NBME questions that can be used for practice testing. The grading is focused on critical thinking, effective writing and communication skills.

**Part I** allows students to demonstrate their level of comprehension of a topic. The act of writing serves to help students measure their depth of understanding. Furthermore, the task of describing areas in which they are unconfident pushes students to frame their areas of confusion in a way that their peers can help. **Part II** enhances knowledge, active learning competencies, communication skills, and engagement as the students explain difficult concepts, often by using multiple resources to review the material. Once the assignment is complete, faculty participate in the discussion reinforcing good posts and clarifying others.

The use of discussion posts has proven to be of immense value for both students and faculty. By completing all tasks, students are not only given the opportunity to reinforce their knowledge but also to learn from each other. Students felt like these assignments were beneficial, since their completion significantly reduced the study time of subjects reviewed in the discussion posts. Faculty gain an appreciation of topic areas that students understand well and those that need further clarification. In many cases, students clarify each other's posts, minimizing faculty time. Faculty explanations can be posted to the discussion board for the benefit of all students, rather than to one student through email or in person. As a result, discussion boards prove to be an excellent study guide for exams.