aphmg
Association of Professors of Human and Medical Genetics

21st Annual APHMG Workshop and Special Interest Groups Meetings
May 6-8, 2015
Sheraton Sand Key Resort • Clearwater, Florida

PROGRAM
and
MEETING SCHEDULE

Useful information for APHMG Attendees

Sheraton Sand Key Resort	1160 Gulf Road, Clearwater Beach, FL; Tel: 727-595-1611
Registration, Badge and Program material pick-up	Lobby II – Wednesday 7:00 am – 6:30 pm Thursday 7:00 am – 2:00 pm Friday 7:00 am - noon
APHMG Staff Office	Conference Office
Medical Genetics Residency Program Directors SIG	Beach Room – see program for detailed agenda
Clinical Laboratory Training Program Directors SIG	Gulf Room – see program for detailed agenda
APHMG Workshop	Beach Room – see program for detailed agenda
APHMG Business Meeting and Dessert Reception	Beach Room
Medical School Genetics Course Directors SIG	Gulf Room – see program for detailed agenda
Breakfast/Lunch	Gulf Room, Friday lunch at Rusty's Bistro
Opening Reception	Pool side Grass (Gulf Room in the event of inclement weather)
Opening Dinner	Pool side Grass (Gulf Room in the event of inclement weather)
Thursday Afternoon Excursion	"Encounters with Dolphins", pre-registration required. Shuttle to Clearwater Marina starts at 3:00 pm, return by 5:30 pm.
Thursday Evening Dinner	Pool side Beach (Gulf Room in the event of inclement weather)
Internet Access	Complimentary WiFi in function space and guest rooms Code is **GENETICS**
Cameras, Cell Phones and Video Recording Devices	Attendees are asked to be respectful of their colleagues by turning off or putting mobile devices on vibrate before entering meeting rooms.
Guests (paid)	Guests are invited to participate in all meal functions but are not permitted to attend the meetings.
Restaurants/Bars	Poolside Café, Mainstay Tavern, Island Grille, Rusty's Bistro, Slo Joe's Turtle Bar
Concierge Desk	Mr. Charlie Creel
Activities	**Beachfront Pool:** Outdoor pool area, includes whirlpool and adjacent children's pool. **Outdoor Tennis Courts**: three courts overlooking the beach. Contact guest services to make reservations, complimentary rackets and balls. **Sheraton Fitness:** Life Fitness equipment and staff available. **Beach Rentals:** Umbrella and chairs, private cabanas, jet skis available for rent at the beach hut. **Golf:** Resort staff can book tee times and arrange transportation to nearby golf courses.

21st Annual APHMG Workshop and Special Interest Groups Meetings
May 6-8, 2015
FINAL AGENDA

Wednesday, May 6, 2015

7:00 am – 6:30 pm	**Registration Open**	*Lobby II*
7:00 am – 8:00 am	**Breakfast**	*Gulf Room*
8:00 am – 3:30 pm	**Medical Genetics Residency Program Directors' SIG Meeting –** refer to page 6	*Beach Room*
2:00 pm – 5:00 pm	**Clinical Laboratory Training Program Directors SIG Meeting –** refer to page 7	*Gulf Room*
5:00 pm – 6:00 pm	**APHMG Council Meeting** (Closed Meeting)	*Starfish Room, 7th floor*
6:00 pm – 7:00 pm	**Opening Reception**	*Poolside Grass*
7:00 pm – 8:00 pm	**Opening Dinner**	*Poolside Grass*
8:00 pm – 9:30 pm	**APHMG Business Meeting/Dessert Reception**	*Beach Room*

Thursday, May 7, 2015

7:00 am – 2:00 pm	**Registration Open**	*Lobby II*
7:00 am – 8:00 am	**Breakfast**	*Gulf Room*
8:00 am – 9:45 am	**Plenary Session I: Competencies: Where Did They Come From and How Can we Make Them Work for our Students, Residents and Lab Trainees** **Speaker:** Stanley Hamstra, PhD, ACGME	*Beach Room*
9:45 am – 10:15 am	**Break**	*Gulf Room*
10:15 am – 11:45 am	**Plenary Session II: Dare to Care: Demystifying the Approach to Problematic Medical Professionals** **Speaker**: Sandra Frazier MD, Assistant Dean of Professional Development, University of Alabama	*Beach Room*
11:45 pm – 12:30 pm	**Lunch**	*Gulf Room*

12:30 pm – 2:00 pm	**<u>Plenary Session III:</u> Incorporation of Clinical Trials and Therapeutics in Clinical Genetics and Medical Genetics Training Programs** **Speakers:** Bruce Korf MD, PhD, University of Alabama Susan Sparks MD, PhD, Genzyme	*Beach Room*
3:00 pm – 5:30 pm	**Thursday Afternoon Excursion:** "Encounters With Dolphins" - A 1.5 hour boat excursion that will include a sightseeing tour with Trained Guides pointing out the abundant wildlife and wild dolphins of Clearwater Tampa Bay. The boat will stop at a small deserted island for 30 minutes to collect shells, take a dip or go snorkeling. Shuttle bus will be provided to and from the Sheraton Sand Key Resort.	*Offsite*
6:00 pm	**Thursday Dinner - Beach BBQ:** Enjoy an evening of dining and fun with colleagues on the Sheraton Sand Key Resorts' 10 acre private beach. Activities will include glow in the dark corn hole and badminton or just bury your toes in the soft white sand and admire the spectacular sunset.	*Poolside Beach*

Friday, May 8, 2015

7:00 am – noon	**Registration Open**	*Lobby II*
7:00 am – 8:00 am	**Breakfast**	*Gulf Room*
8:00 am – 10:00 am	**<u>Plenary Session IV:</u> Economics of Clinical and Lab Genetics Services in the Current Health Care Climate** **Speakers:** Marc Williams MD, Director, Genomic Medicine Institute, Geisinger Health System David Flannery MD, Medical Director, ACMG ***The United States' Healthcare System: Evolution vs. Intelligent Design? Implications for Genetic Services*** Marc Williams MD, Director, Genomic Medicine Institute, Geisinger Health System ***You Say You Want A (Genomics) Revolution, But Who's Going To Pay (and How?)*** David Flannery MD, Medical Director, ACMG	*Beach Room*
10:00 am – 10:15 am	**Break**	*Gulf Room*
10:15 am – 11:15 am	**<u>Plenary Session V:</u> Genomic Medicine Training Opportunities in Medical Education**	*Beach Room*

	Speaker: William Scott, PhD, Professor of Human Genetics and Vice-Chair for Education, University of Miami	
11:15 am – 12:15 pm	**Lunch**	*Rusty's Bistro*
11:45 am – 5:30 pm	**Medical School Genetics Course Directors SIG Meeting** refer to page 8	*Gulf Room*

Posters: Submitted posters for the Medical School Genetics Course Directors SIG will be displayed during the meeting. Posters will be discussed on Friday, May 8th.

Medical Genetics Residency Program Directors SIG Meeting
Wednesday, May 6, 2015, 8:00 am – 3:30 pm – Beach Room

AGENDA

7:00 am – 8:00 am	**Breakfast** **Gulf Room**
8:00 am – 8:15 am	**Welcome:** Nathaniel Robin, MD
8:15 am – 8:30 am	**Report on the 2014 Match - Future Issues:** V Reid Sutton, MD
8:30 am – 8:45 am	**Update on the In-service Exam:** Mimi Blitzer, PhD
8:45 am – 9:15 am	**Report from ABMGG:** Mimi Blitzer, PhD
9:15 am – 10:00 am	**Reports from the RRC/ACGME:** V Reid Sutton, MD and Laura Edgar, EdD, CAE
10:00 am – 10:20 am	**Break**
10:20 am – 11:00 am	**Q&A for ABMGG/ACGME**
11:00 am – 12:00 pm	**Plenary Session (1): Challenges in Genetics Residency Education**
	Introduction: Nathaniel Robin, MD
	Is Dysmorphology Still Important?: Nathaniel Robin, MD
	Non Invasive Prenatal Screening: Susan Klugman, MD
	How Best to Use Their Time in the Clinical Laboratory Rotations: (TBD)
12:00 pm – 1:15 pm	**Lunch** **Gulf Room**
1:15 pm – 2:15 pm	**Plenary Session (2): Challenges in Genetics Residency Education**
	Expectations and Requirements in Cancer Genetics: Nathaniel Robin, MD
	Biochemical Genetics: V Reid Sutton, MD
	Integrating Research into Residency Education: TBD
2:15 pm – 3:30 pm	**Moving Forward: Best Practices to Make Our Training Better** Speakers and Topics: TBD

Clinical Laboratory Training Program Directors SIG Meeting
Wednesday, May 6, 2015, 2:00 pm – 5:00 pm – Gulf Room

AGENDA

2:00 pm – 2:10 pm	**Welcome and Introductions** Linda Jeng, MD, PhD
2:10 pm – 3:30 pm	**Finalization and Implementation of Milestones** Linda Jeng, MD, PhD
3:30 pm – 3:40 pm	**Break**
3:40 pm – 4:30 pm	**ABMGG update (site visits, In-service exam, other)** Mimi Blitzer, PhD Laurie Demmer, MD
4:30 pm – 5:00 pm	**Brainstorming on Future Projects** Linda Jeng, MD, PhD

Medical School Genetics Course Directors SIG Meeting
Friday, May 8, 2015, 11:45 am – 5:30 pm – Gulf Room

AGENDA

11:45 am – 12:15 pm	**Announcements and Updates**
12:15 pm – 12:30 pm	**Report Back on Applications of Earlier APHMG Initiatives/Ideas**
12:30 pm – 1:30 pm	**Reflections on Curriculum Transitions Through a Genetics Lens**
	William Scott, PhD, Professor of Human Genetics and Vice Chair for Education, University of Miami
	Joann Bodurtha, MD, MPH Professor of Pediatrics and Oncology, Johns Hopkins University
1:30 pm – 3:00 pm	**Teachers Mentoring Teachers: The Gift of Peer Feedback Workshop**
	Katherine Hyland, PhD, Professor of Biochemistry and Biophysics, UCSF
3:00 pm – 3:30 pm	**Curricular Innovations Short Talks** *Active Learning Strategies Integrating Genetics During the Internal Medicine Clerkship* Tracy Weiler, Assistant Professor of Human and Molecular Genetics, Florida International University
	The Implementation of APHMG Genetics Competencies and Osteopathic Core Competencies in Medical Genetics at William Carey University College of Osteopathic Medicine Everett Roark, Assistant Professor of Microbiology and Genetics, William Carey University
3:30 pm – 3:45 pm	**Break/Poster Viewing**
3:45 pm – 4:15 pm	**Setting the Stage for Case Development**
	Videos in Medical Education: An Overview on Tools and Best Practices Jon Bernstein, Assistant Professor of Pediatrics, Stanford University
	Blended Learning: Linking Videos to Active Learning Exercises in the Classroom. Shoumita Dasgupta, PhD, Associate Professor of Medicine, Boston University School of Medicine
4:15 pm – 5:15 pm	**Flip and Elevate, Part 3: Developing Cases in Genomic Medicine**
5:15 pm – 5:30 pm	**Report Back on Case Development and Session Closing**

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POSTER ABSTRACTS FOR THE MEDICAL SCHOOL COURSE DIRECTORS SIG

POSTER #1

Mimi Blitzer, PhD
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Embedding Pharmacogenomics and Personalized Medicine Education into the Medical School Curriculum.

Mimi Blitzer, University of Maryland

As the cost of high-throughput DNA sequencing declines, the push to incorporate genomic data into various areas of clinical practice is steadily increasing. Recognizing this, organizations such as the AAMC have charged medical schools with training new physicians to be competent in their knowledge of genomic medical concepts. In response to this, University of Maryland School of Medicine (UMSOM) is incorporating analysis of pharmacogenomic data into the curriculum. All second-year medical students received a pharmacogenomic dataset generated from the DMET (Drug Metabolizing Enzymes and Transporters) molecular test, which consists of 1936 single nucleotide polymorphisms (SNPs) in 231 genes that have previously been shown to affect drug metabolism. Students were given the choice either to provide their own DNA sample and receive their personal data, or receive data from a de-identified sample. Through supplemental material provided with the results and reinforced in appropriate lectures, students were introduced to bioinformatics and the process of pharmacogenomic data analysis and interpretation. The clinical application of these data to inform clinical decision-making and drug management for the individual patient was emphasized during small group discussions. Anonymous surveys were administered before and after dataset distribution to determine if students thought the activity was educationally useful and whether the subject material was better understood and internalized when applied to their personal data. The surveys also assessed whether students perceived their approach the remainder of their medical school training would be different as a result of this activity. Approximately 88% of students chose to use their own sample; preliminary data indicate the activity was well received and valued. We anticipate students will use the DMET dataset throughout the remainder of their medical school education as a learning tool to better understand the impact and limitations of pharmacogenomic information in clinical care in the era of personalized medicine.

POSTER #2

Helen Jin
Boston University School of Medicine

Shoumita Dasgupta, PhD
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Cultural Competency in Medical Genetics Education: A Discussion Exploring LGBT Assisted Reproduction Technology

Shoumita Dasgupta and Helen Jin, Boston University School of Medicine

Social trends expanding the legalization of same-sex marriage have contributed to increasing numbers of same-sex couples raising children in the US. Same-sex families have the opportunity to raise biologically-related children through assisted reproduction technology (ART), including options such as sperm donation, in-vitro fertilization (IVF), or gestational surrogacy. Since these options differ from infertility services for heterosexual couples, developing culturally competent educational materials for medical students about the lesbian, gay, bisexual, transgender and queer (LGBTQ) patient population is a critical strategy for minimizing LGBTQ health care disparities in this area. At Boston University School of Medicine, we developed a case discussion for the Medical Genetics curriculum to highlight the applications of ancestry-based genetic testing, donor testing, and ART in same-sex family planning and a survey to assess student attitudes about use of ART with both heterosexual and same-sex couples. We found that students with a family member or close friend who identifies as LGBTQ were significantly more likely than other students to disagree with the concept that healthcare access is equivalent for LGBT individuals and other members of the population. Furthermore, before class significantly fewer students recommended IVF for an infertile heterosexual couple (63%) than for a healthy lesbian couple (80%), but those numbers began to converge (67% and 73%, respectively) after the class discussion. While it may appear that a declining number of students recommended ART for the lesbian case, in fact, more students recommended sperm donation to a lesbian couple with fertility concerns after the class (51%) than before the class (33%). These initial findings highlight the importance of both personal experience with the LGBTQ community and the impact of educational interventions in addressing LGBTQ healthcare disparities.

POSTER #3

Alice Hudder, PhD
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Genomics in Medicine: A Survey of Readiness Among Medical Residents at Millcreek Community Hospital in Erie, PA

Alice Hudder, Lake Erie College of Osteopathic Medicine

With the advent of the human genome project came the promise of personalized or precision medicine. Advances in technology have enabled the use of patient genomic information to inform medical diagnosis and treatment. This has created a knowledge gap for current and future physicians in the implementation of genomic information into clinical practice. This new capability in medicine has led to several important questions. For example, how will we integrate genomic information into clinical practice? How prepared are physicians to integrate genomics into patient care? And how will we train current and future physicians to make sense of this information? In large metropolitan areas there are a wide variety of specialists, including clinical geneticists, available for consultation. In smaller cities, like Erie, PA (population ~100,000), there is much more limited access to geneticists and this perhaps makes it more imperative for local physicians to achieve competency in this area. I conducted a survey of medical residents at Millcreek Community Hospital in Erie, PA to find out how they viewed genomics in medicine. Of those who responded, the majority felt that genomic information will improve patient care and that pharmacogenomics information in particular will affect patient treatment plans; however, the majority of medical residents did not feel competent to access and use human genomic information in their practice. In response to the results of this survey, a short curriculum in Genomics in Medicine has been proposed for all medical residents at Millcreek Community Hospital.

POSTER #4

Katherine Hyland, PhD
University of California, San Francisco
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Creating Genomically-Literate Health Care Professionals: Incorporating Personal Pharmacogenomic Testing and Genetic Counseling into a Genomic Medicine Elective

K. Hyland[1,2], A Seol[3], C Boscardin[4], S. Baranzini,[2,5] B. Koenig[6], M Norton[2,7], R Nussbaum[2,8], M Sabbadini[9], J Shieh[2,9], K Weisiger[9], L Weiss[2,10]

(1) Department of Biochemistry and Biophysics, (2) Institute for Human Genetics, (3) School of Medicine, (4) Department of Medicine, (5) Department of Neurology, (6) Institute for Health and Aging, (7) Department of ObGyn, Division of Maternal Fetal Medicine, (8) Division of Genomic Medicine, Department of Medicine, (9) Division of Medical Genetics, Department of Pediatrics, (10) Department of Psychiatry

Purpose:

While advances in genomic technologies are impacting clinical practice across all disciplines of medicine, there is concern over lack of expertise among health care providers to interpret genomic test results and assess their clinical utility. Thus health care professional students require enhanced genomics training to better serve their future patients.

Methods:

To address this need, we developed a Genomic and Precision Medicine elective for medical and genetic counseling students. Since recent studies suggest participation in personal genomic testing has a positive impact on student educational experiences, we decided to offer an opportunity for personal genomic testing. Pharmacogenomic testing was chosen to avoid potential harm from whole genome analysis. Students are genotyped for pharmacogenomic variants in the *CYP2D6, CYP2C19, UGT1A1* and *IL28b* genes through the UCSF genetic testing laboratory. Prior to making the decision about participating in testing, students are introduced to applications, opportunities and challenges of genomic testing, and personally engage in the informed consent process with 2nd year genetic counseling students under the supervision of a licensed genetic counselor. Students who choose not to undergo testing are provided with example genotype reports. Using their own or example data, students learn how to interpret test results and assess clinical utility. In addition to analyzing pharmacogenomic data, case-based exercises using example WES and GWAS data are also incorporated.

Students enrolled in the course are asked to participate in a research study assessing knowledge and attitudes on genomic testing using a structured survey before and after the course. A control group not enrolled in the course was recruited from the 1st year medical school class. A follow up survey to assess knowledge retention will be delivered to the same study and control group 8 months later.

Results:

The course is launching spring 2015, data and analysis to follow.

POSTER #5

NJ Larsen
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J Grogan
Office of the Curriculum, Ross University School of Medicine

Genetics in a spiral, organ based curriculum

NJ Larsen and J. Grogan, Ross University School of Medicine

The Ross University School of Medicine (RUSM) preclinical curriculum follows a spiral organ-based framework during the first 4 semesters. Every student is required to complete two modules related to each of fundamentals plus 10 organ-systems. The first encounter with each module is in the first year, followed by a return to the same modules with greater clinical content in the second year. The primary pedagogy of instruction, outside of anatomy and clinical medicine learning, is large classroom instruction. All preclinical semesters include a clinical skills course with skills training labs, standardized patients, simulations, small group cognitive skills, community clinics, medical interviewing, hospital visits and service learning sessions which complement module-based learning.

A thematic review of topics in molecular biology and genetics learning was undertaken. The resulting map of genetics teaching indicates that semesters 1, 2, and 3 include more genetics learning than the semester 4 curriculum. The landscape of genetics learning is complex, with the fundamentals 1, reproductive 1 and reproductive 2 modules having the greatest weights of content. The results should assist the curriculum committee to better understand the genetics theme within the preclinical curriculum at RUSM. It is our hope that presenting at APHMG will allow us to discuss issues about integration of clinical learning with foundational genetic learning.

POSTER #6

Everett Roark, PhD
William Carey University College of Osteopathic Medicine
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The Implementation of APHMG Genetics Competencies and Osteopathic Core Competencies in Medical Genetics at William Carey University College of Osteopathic Medicine.

Everett Roark, William Carey University College of Osteopathic Medicine

Medical Genetics is of critical importance in the medical school curriculum. Physicians increasingly encounter a multitude of genetic disorders and diseases with genetic components. Also, physicians require the skills necessary to evaluate and apply knowledge from many different sources (drug companies, journals, etc.) in order to be effective lifelong learners. Therefore, having a comprehensive understanding (from basic science to clinical application) of the principles of Genetics and Genomics is vital in order for practicing physicians to develop skills necessary to make informed clinical decisions.

In 2010, the William Carey University College of Osteopathic Medicine took in its first class. Genetics was taught didactically as a subset of Human Embryology. This resulted in a condensed Genetics component that was heavily focused on developmental genetics. In 2013, the Genetics class was separated into an independent course and the APHMG Genetics Competencies were used as a framework for covering essential concepts in the Genetics class. The class focused on the impact of genetics and genetic technology on the lives of patients. In addition, the Genetics course included concepts that are compatible with four osteopathic core competencies: Osteopathic Philosophy, Medical Knowledge, Patient Care, and Practice-Based Learning and Improvement. We now deliver the curriculum using various methods, including: didactic lectures, group projects, student presentations, and clinical guest lectures. Since implementing these changes, we have seen gains in the course rating and student performance.

POSTER #7

Andrew K. Sobering, PhD
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Using *PowerPoint Show* as a Platform for Student Engagement

Andrew sobering, St. George's University

This poster describes an application of PowerPoint which engages students by creating interactive vignettes. Using an ordinary PowerPoint presentation, a scenario is created, questions are posed, and each answer provides a hyperlink explaining the outcome of that decision. The file is saved in a manner which forces students to navigate through the vignette using only the hyperlinks. To enable this feature, the following four steps must be followed when saving the file:

1. Select the **SLIDE SHOW** tab (top menu choices)
2. Click on the **Set Up Slide Show** button (fifth button from left)
3. Look at the top left panel of the pop up menu, and select **Browsed at a kiosk (full screen)**
4. Save the file as a **PowerPoint Show format** (PPSX)

When these four steps are followed, the PPSX file will open in display mode, the arrow keys are disabled, and the only way to navigate through the vignette is by clicking on the embedded hyperlinks. This poster describes the construction of this teaching tool, my experiences with it, and a live example set up on a lap top computer to demonstrate it. The idea for this poster came from the 2014 Course Directors SIG at the 2014 APHMG meeting in Napa California where we formed breakout groups to create case-based content to support a flipped-classroom model.

Poster #8

Tracey Weiler PhD
Herbert Wertheim College of Medicine, Florida International University
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Marta Cuellar MD
Herbert Wertheim College of Medicine, Florida International University

Amalia Landa-Galindez, MD
Herbert Wertheim College of Medicine, Florida International University

Active Learning Strategies Integrating Genetics during the Internal Medicine (IM) Clerkship

Tracey Weiler PhD, Marta Cuellar MD, Amalia Landa-Galindez, MD, Florida International University

Background

In this era of individualized medicine, medical schools have a responsibility to graduate students who can apply genetic competencies in clinical practice. Although the APHMG Medical School Core Curriculum in Genetics guides curriculum development in many institutions, strategies for integration and assessment in third year clerkships are needed.

Objective

At FIU-HWCOM we implemented an online module to enhance the following genetic competencies among 3rd year IM students: 1) collection of genetic information from patient interviews; 2) construction and interpretation of 3-generation pedigrees; 3) prioritization of individuals requiring genetic testing; 4) identification of appropriate genetic testing technologies.

Methods

We designed a two-part activity consisting of a self-learning module (SLM) followed by an innovative assessment tool requiring application of concepts in a simulated clinical setting.

The online SLM included a study guide summarizing key genetic concepts from years 1 and 2 enhanced by interactive family history exercises and case scenarios from the NCHPEG www.nchpeg.org/pa/ website.

The assessment required students to analyze a pre-recorded patient-practitioner encounter focused on an adult genetic disorder. Students were required to draw an accurate pedigree and provide detailed written responses to the specific learning objectives (LO) described in the table.

Results

114 students have completed the module. The table indicates the percentage of students that achieved a passing score for each learning objective.

LO1	LO2	LO3	LO4	LO5
draw pedigree reflective of vignette	interpret inheritance pattern	identify genetic red flags	identify appropriate individuals for testing	suggest appropriate genetic testing technology
88%	82%	96%	84%	64%

Conclusion

Assessment results demonstrated that the majority of students effectively applied genetic knowledge, however multiple areas still need reinforcement. In the future, we also intend to assess the students' ability to calculate recurrence risks and demonstrate an understanding of counseling strategies.

SAVE THE DATE

22ND Annual APHMG Workshop and Special Interest Group Meetings

May 4 – 6, 2016

Westward Look Wyndham Grand Resort & Spa, Tucson, Arizona

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