



**22nd Annual Workshop &
Special Interest Groups Meetings**
May 04 – 06, 2016 • Tucson, Arizona



PROGRAM

AND

MEETING SCHEDULE

**WESTWARD LOOK RESORT
TUCSON, AZ**



**22nd Annual Workshop &
Special Interest Groups Meetings**
May 04 – 06, 2016 • Tucson, Arizona



**A Special Thank You to the
following Sponsors**



ELSEVIER



AGENDA

Wednesday, May 04

6:30 am – 5:00 pm
Sonoran Terrace

APHMG Registration Desk Open
Staff: Sheilah Jewart

7:00 am – 8:00 am
Sonoran Terrace

Breakfast – Included

8:00 am – 3:30 pm
Sonoran Ballroom

**Medical Genetics Residency Program Directors
SIG Meeting**

12:00 pm – 1:30 pm
Sonoran Terrace

Lunch – Included

1:00 pm – 5:00 pm
Sonoran Ballroom

**Clinical Laboratory Training Program Directors
SIG Meeting**

5:00 pm – 6:00 pm

APHMG Council Meeting
Closed Meeting

6:00 pm – 7:00 pm
Sonoran Rooftop Patio

APHMG Welcome Reception

7:00 pm – 8:00 pm
Sonoran Rooftop Patio

APHMG Welcome Dinner

8:00 pm – 9:30 pm
Sonoran Ballroom

APHMG Business Meeting
Dessert / Coffee

Thursday, May 05

6:30 am – 2:00 pm
Sonoran Terrace

APHMG Registration Desk Open
Staff: Sheilah Jewart

7:00 am – 8:00 am
Sonoran Terrace

Breakfast – Included

8:00 am – 10:00 am
Sonoran Ballroom

Plenary Session I
**Building an Evidence Base for Genomic
Medicine: Use of ClinGen in Clinical Care and
Training of Providers**
•*Jonathan Berg, MD, PhD, University North Carolina*
•*Andrew Faucett, MS, LGC, Geisinger Health System*

10:00 am – 10:30 am

Break

10:30 am – 12:00 pm
Sonoran Ballroom

Plenary Session II
Genetics in Health Disparities
•*Maximillian Muenke, MD, National Human Genome
Research Institute*
•*Melissa Davis, PhD, University GA Franklin College*
•*Rick Kittles, PhD, University of Arizona*

12:00 pm – 12:45 pm
Sonoran Terrace

Lunch – Included

12:45 pm – 2:00 pm
Sonoran Ballroom

Plenary Session III
ABMGG: New Pilot for Continuous Certification
•*Mimi Blitzler, PhD, University of Maryland / ABMGG*
•*Darrel Waggoner, MD, University of Chicago*

2:15 pm
Depart from Lobby

Optional Excursion
Hike or Tram Sabino Canyon

7:30 pm – 10:00 pm
Trails End

APHMG Western BBQ & Square Dancing

Friday, May 06

6:30 am – 6:00 pm
Sonoran Terrace

APHMG Registration Desk Open
Staff: Sheilah Jewart

7:00 am – 8:00 am
Sonoran Terrace

Breakfast – Included

8:00 am – 10:00 am
Sonoran Ballroom

Plenary Session IV
Newborn Screening in the Genomic Era

**Next Generation Sequencing in Newborn
Screening: Cystic Fibrosis and Beyond**

• *Mei Baker, MD, FACMG, University of Wisconsin*

Screening for SCID in the Genomic Era

• *Jennifer Puck, MD, University of California, SF*

10:00 am – 10:15 am

Break

10:15 am – 11:15 am
Sonoran Ballroom

Plenary Session V
**Our Experience with a Coursera Course in
Medical Genomics**

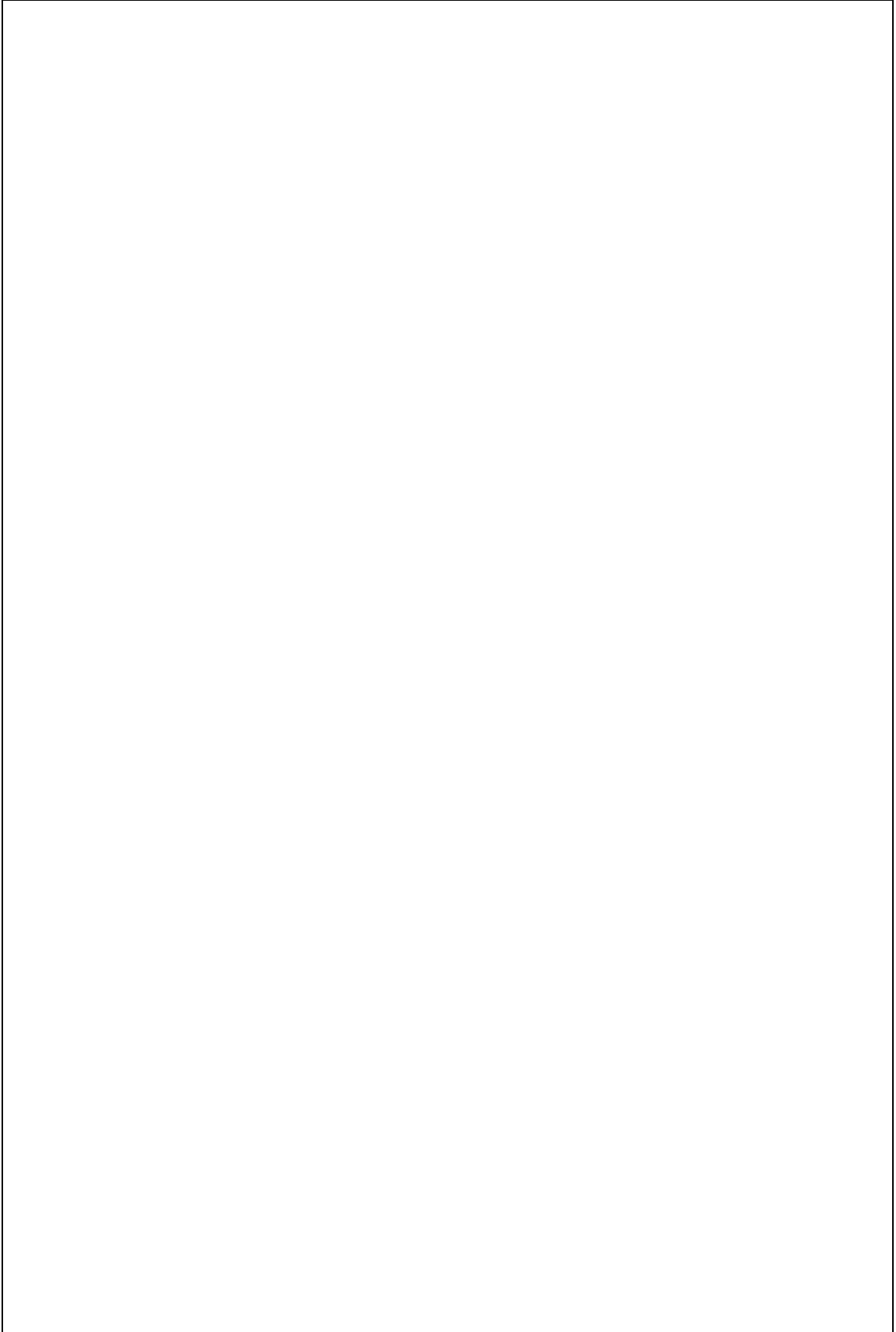
• *Robert Nussbaum, MD, University of California, SF
Invitae*

11:15 am – 12:15 pm
Sonoran Terrace

Lunch – included

12:15 pm – 6:00 pm
Sonoran Ballroom

**Medical School Genetics Course Directors
SIG Meeting**



Medical Genetics Residency Program Directors SIG Meeting

AGENDA

Wednesday, May 04

8:00 am – 8:15 am Sonoran Ballroom	Welcome & Introductions • <i>Nathaniel Robin, MD, University of Alabama</i>
8:15 am – 8:30 am	Report on the 2015 Match • <i>Nathaniel Robin, MD</i>
8:30 am – 8:50 am	Update NRMP “All In” Policy • <i>Mona Signer, MPH, CEO & Executive Director</i> •, NRMP
8:50 am – 9:05 am	Update on the In-Service Exam • <i>Miriam Blitzer, PhD, University Maryland School of Medicine</i>
9:05 am – 9:30 am	Report from ABMGG • <i>Miriam Blitzer, PhD</i>
9:30 am – 9:45 am	Report from the ACGME • <i>Reid Sutton, MD, Chair 2016-17 ABMGG</i>
9:45 am – 10:00 am	Report from the Review / Recognition Committee • <i>Laura Edger, EdD, CAE, Executive Director</i> <i>Review Committee for Pathology, Radiation Oncology and</i> <i>Medical Genetics and Genomics</i>
10:00 am – 10:20 am	Break
10:20 am – 10:45 am	Q&A for NRMP, ABMGG, ACGME Panel
10:45 am – 11:30 am	Plenary Session Maximizing Resident-Genetic Counselor Education • <i>Dawn Allaine, MS, CGC, The Ohio State University</i> • <i>Nathaniel Robin, MD, University of Alabama</i>
11:30 am – 11:50 am	Small Group Discussions
11:50 am – 12:15 pm	Large Group Discussion

12:15 pm – 1:30 pm
Sonoran Terrace

Lunch – included

1:30 pm – 3:00 pm

**Improving Residents Experience in the Clinical Labs
Joint Session with LD SIG Meeting**

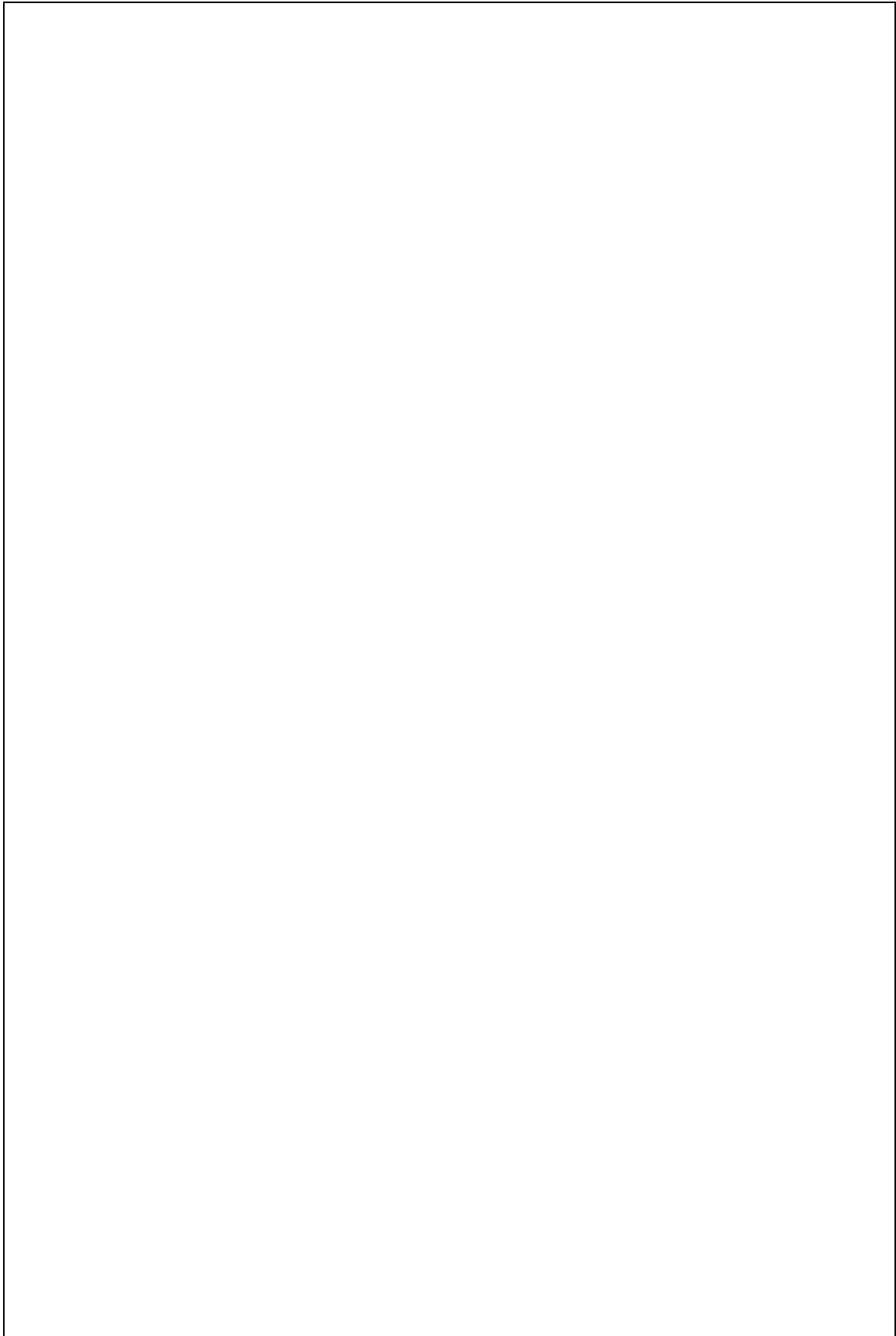
- Susan Klugman, MD*
- Linda Jeng, MD, PhD*

Clinical Laboratory Training Directors SIG Meeting

AGENDA

Wednesday, May 04

12:00 pm – 1:00 pm Sonoran Terrace	Lunch – included
1:00 pm – 1:30 pm Sonoran Ballroom	Report from ABMGG • <i>Miriam Blitzer, PhD, University Maryland School of Medicine</i>
1:30 pm – 3:00 pm	Improving Residents Experience in the Clinical Labs Joint Session with PD SIG • <i>Susan Klugman, MD</i> • <i>Linda Jeng, MD, PhD</i>
1:30 pm – 1:40 pm	Biomedical Genetics Rotations • <i>Tina Cowan, PhD, Stanford School of Medicine</i>
1:40 pm – 1:50 pm	Cytogenetics Rotations • <i>Rhona Schreck, PhD, Ceders Sinai</i>
1:50 pm – 2:00 pm	Molecular Genetic Rotations • <i>Linda Jeng, MD, PhD, University Maryland School of Medicine</i>
2:00 pm – 2:30 pm	Goals for Clinical Fellows During Lab Rotations Breakout Groups
2:30 pm – 3:00 pm	Discussion Summary and Proposal for Competencies
3:00 pm – 3:15 pm	Break
3:15 pm – 3:25 pm Sonoran Ballroom	Welcome & Introductions • <i>Linda Jeng, MD, PhD, University of Maryland School of Medicine</i>
3:25 pm – 4:00 pm	Brainstorming Next Steps Follow-up from Joint LD and PD SIG Meeting
4:00 pm – 4:20 pm	Funding for Trainees / Fellows Survey Results
4:20 pm – 4:40 pm	Milestone Evaluation Tool • <i>Linda Jeng, MD, PhD</i>
4:40 pm – 5:00 pm	Request for Working Group Volunteers Generic Trainee Evaluation Form Trainee Funding

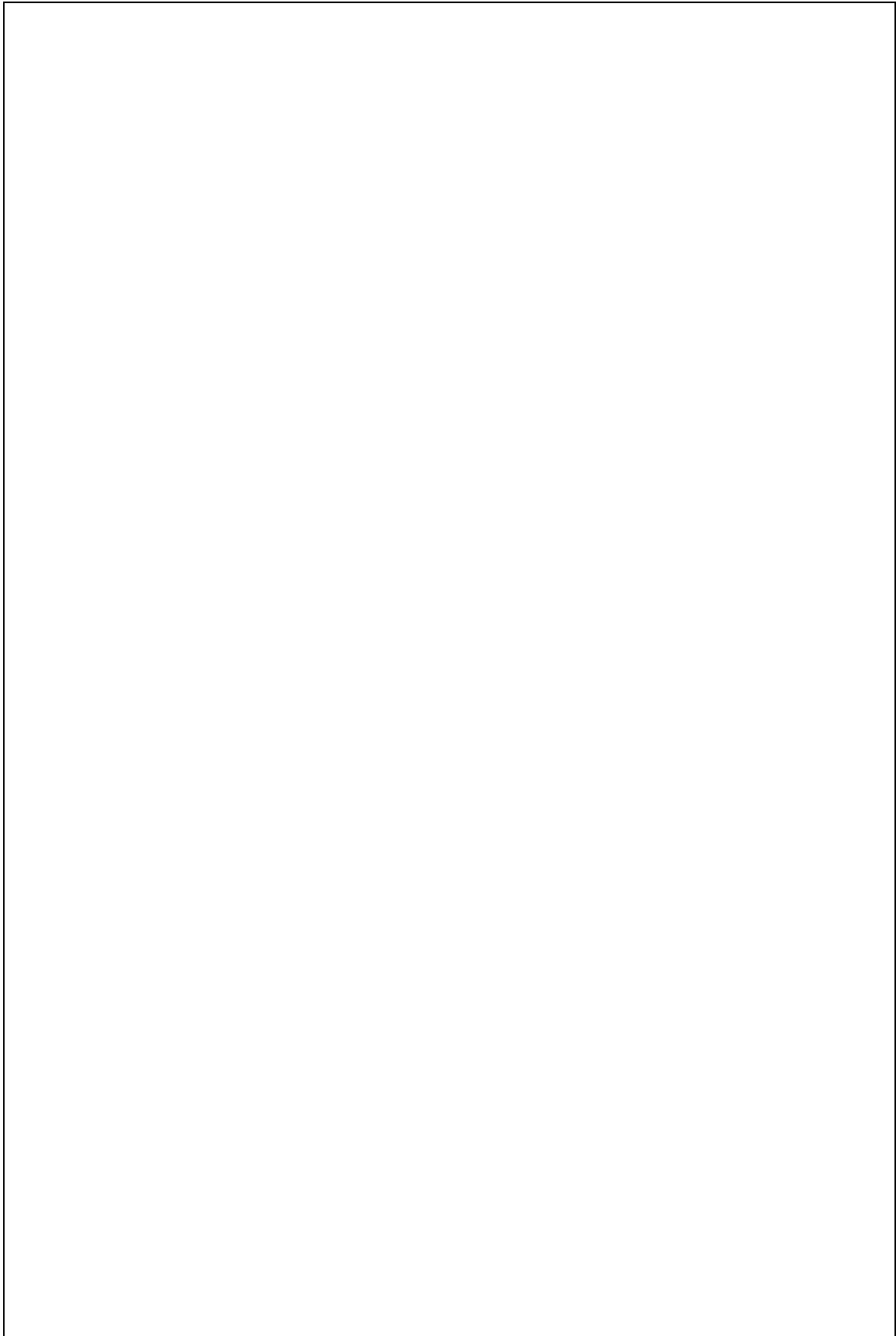




APHMG BUSINESS MEETING
Wednesday, May 05, 2016
8:00 pm - 9:30 pm - Sonoran Ballroom

AGENDA

Welcome	Darrel Waggoner
Updates:	
Meeting Planner	Darrel Waggoner
Training Residents in Genomics (TRIG) Working Group	
CD SIG survey: Clinical Cases	
Secretary-Treasurer Report	Cindy Powell
<u>SIG Updates</u>	
Residency Program Directors SIG	Nat Robin
Course Directors SIG	Shoumita Dasgupta
Lab Training Program Directors SIG	Linda Jeng
<u>Liaison Reports</u>	
ACMG	Mike Watson
ASHG	Mike Dougherty
ABMGG	Mimi Blitzer
<u>Other Business:</u>	
Nominations Committee Report	Demmer/Powell/Sutton
2017 Workshop: May 10-12, 2017 Sheraton Keys Clearwater	
Special Thanks	Darrel Waggoner



Medical School Genetics Course Directors SIG Meeting

AGENDA

Friday, May 06

12:15 pm – 12:30 pm Sonoran Ballroom	Announcements & Updates • <i>Shoumita Dasgupta, Boston University School of Medicine</i>
12:30 pm – 12:45 pm	Report back on applications of earlier APHMG Initiatives / Ideas
12:45 pm – 1:45 pm	From the Test Trenches: An Informal Discussion on UME Genetics / Genomics Curriculum • <i>Robert Nussbaum, MD, University of California, San Francisco & Invitae</i>
1:45 pm – 2:45 pm	Management of Genetics and Genomic Teaching Across the Medical School Curriculum: A Panel Discussion • <i>Sabrina Nunez, Washington University School of Medicine</i> • <i>Matt Velkey, Duke University Medical Center</i>
2:45 pm – 3:15 pm	Curricular Innovations Short Talks: The Anatomy to Genomics (ATG) Start Genetics Medical School Initiative: Exome Sequencing of Cadavers Used for Anatomy Instruction • <i>Glenn Gerhard, Temple University Katz School of Medicine</i> Improving Success in Medical School: The Michigan Prematriculation Curriculum • <i>Bev Yashar, University of Michigan Medical School</i>
3:15 pm – 3:35 pm	Poster Viewing / Break
3:35 pm – 4:00 pm	Discussion of Joint Meeting with Association of Biochemistry Educators (ABE) in 2017
4:00 pm – 4:30 pm	Turning Education Projects into Scholarly Work: A Primer • <i>Shoumita Dasgupta, Boston University School of Medicine</i> • <i>Kathy Hyland, University of California, San Francisco</i>
4:30 pm – 5:30 pm	Educational Scholarship Consult: Disseminating Your Creative Approaches to Medical Genetics Education • <i>Andrew Sobering, St. George's University</i>
5:30 pm – 6:00 pm	Report back on Consult Session and Closing Remarks

POSTER ABSTRACTS

Abstract #1: Common Threads: Reflective Practice Connecting Medical Genetics Concepts Across An Integrated Curriculum

Shoumita Dasgupta

Boston University School of Medicine, Boston, MA, 02118

Recent curricular surveys have identified increasing numbers of medical schools moving towards integrated pedagogical models. At the same time, AAMC data indicates that the average proportion of the pre-clerkship curriculum dedicated to medical genetics is approximately 2%¹. An integrated curriculum structure risks diluting the genetics concepts until the discipline identity is lost. To help preserve the identity of genetics within an integrated curriculum, a longitudinal reflective practice was introduced into the first year Principles Integrating Science and Medicine (PrISM) curriculum. This activity was tied to a series of patient visits held in four distinct modules of the PrISM curriculum. Following each patient session, the students were given an in-class reflective writing prompt. For the first two sessions, the anonymous writings focused on the stories' connections to their own lives and values, and subsequent sessions were paired with preceding patient visits. For example, the students' fall semester writings from meeting the parent of a child with trisomy 18 were returned to their peers for further comment after a spring semester session featuring a self-advocate with Down Syndrome. The writing guided students to contrast the similar genetic etiology and dramatically different prognoses of these conditions. Likewise, the students' fall semester reflections about women harboring hereditary breast and ovarian cancer-predisposing mutations were returned to their peers in the spring semester, after meeting a guest with multiple family members affected by Huntington's disease. While both are single gene disorders with genetic testing options, the ability to select risk-reducing procedures in one was juxtaposed with receiving a prediction for an incurable disease in the other. By structuring these reflective writings to highlight the common threads and contrasting outcomes between conditions, the students engaged with the clinical relevance of the genetics content and grappled with the ethical ramifications of genetic testing.

¹ Plunkett-Rondeau J, Hyland K, Dasgupta S. Training future physicians in the era of genomic medicine: trends in undergraduate medical genetics education. *Genet Med*. 2015. doi:10.1038/gim.2014.208.

Abstract #2: Interprofessional Education Workshop To Enhance Genetic Communication Skills

Cecelia Bellcross and **Kathryn B. Garber**

Emory University School of Medicine, Atlanta, GA, 30307

Clinicians in many non-genetic specialties are often the first healthcare professional with whom patients discuss genetic screening and testing options and genetic test results. Although learning the precise language of genetics is critical preparation for these situations and to understand core genetic principles, use of this terminology may impede understanding by patients. Genetic counselors (GC) are trained to communicate concepts of risk, genetic information, and test results in a manner that is both understandable and sensitive to the patient's needs. To enhance such skills in first year medical and physician assistant (PA) students, we developed an inter-professional education workshop to foster good communication skills of genetic information. Senior GC students modeled a counseling session then worked in small groups with the medical and PA students, practicing the delivery of genetic information and receiving feedback from their GC mentors and peers.

**Abstract #3: The Anatomy To Genomics (ATG) Start
Genetics Medical School Initiative: Exome
Sequencing Of Cadavers Used For Anatomy
Instruction**

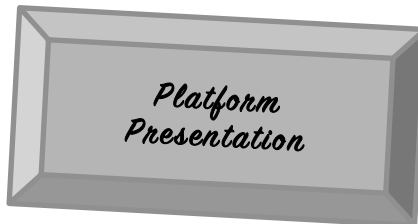
Glenn S. Gerhard, Qunyan Jin, Barbara V. Paynton and Steven N. Popoff
*Departments of Medical Genetics and Molecular Biochemistry and Anatomy and Cell Biology,
Temple University, Lewis Katz School of Medicine, Philadelphia, PA, 19140*

BACKGROUND: The increasing use of next generation DNA sequencing in clinical medicine is exposing the need for more genetics education in physician training. We piloted an initiative to determine the feasibility of incorporating exome sequencing data generated from DNA obtained from cadavers used for teaching Anatomy into a first year medical student integrated block-style course.

METHODS: We optimized the procedure to obtain DNA for exome sequencing by comparing the quality and quantity of DNA isolated from several tissues by two different extraction methods. DNA was sequenced using exome capture and analyzed using standard methods. Single nucleotide variants (SNVs) were selected for student teams to independently investigate and prepare presentations on their findings.

RESULTS: A total of seven cadaver DNAs were sequenced yielding high quality results. SNVs were identified that were associated, with known physical traits and disease susceptibility, as well as pharmacogenomic phenotypes. Students presented findings based on correlation with known clinical information about the cadavers' diseases and traits.

CONCLUSION: Exome sequencing of cadaver DNA is a useful tool to integrate Anatomy with Genetics and Biochemistry into a first year medical student core curriculum.



Abstract #4: Developing An Inquiry Habit Of Mind In Medical Education

Katherine Hyland^{1,2}, Michelle Hermiston^{3,4}, Scott A. Oakes⁵, Christopher Stewart³, Matt Trojnar⁶, Karen Hauer⁷, Christy Boscardin⁷, Sam Brondfield⁷, Gordon Strewler⁷ and H. Carrie Chen³

¹*Dept of Biochemistry and Biophysics*, ²*Institute for Human Genetics*, ³*Dept of Pediatrics*, ⁴*Dept of Hematology/Oncology*, ⁵*Dept of Pathology*, ⁶*Office of Medical Education*, ⁷*Dept of Medicine; University of California, San Francisco, CA, 94143*

PURPOSE: Despite rapid expansion in medical knowledge and scientific discovery, millions of Americans continue to suffer from health problems that lack effective therapies. There is a crucial need to address immense gaps in current knowledge and train physicians to solve these and future health problems. However, current emphasis on knowledge acquisition in medical training discourages curiosity and impedes development of skills needed for discovery. An inquiry-based curriculum, on the other hand, fosters a logical, critical, curious habit of mind, and guides learners to challenge current concepts and create new knowledge.

METHODS: We are developing an innovative medical curriculum that trains students to be curious, identify gaps in knowledge, ask the “right” questions, and develop a plan for discovery. We have worked collaboratively with a faculty team representing six “domains of science” to map and develop curricular elements, and have piloted integrated inquiry cases. In parallel, we piloted initial strategies for learner assessment, curriculum evaluation and faculty development. Using information from our pilots, we are refining and building teaching cases and methods for assessment, evaluation and faculty development.

RESULTS: The inquiry curriculum applies multiple domains of science (biomedical, clinical, educational, epidemiology/populations, social and behavioral, and systems), promotes spiral learning and unites students and faculty in the discovery process. It is composed of three elements:

1. **Core Inquiry Curriculum:** Weekly (during first two years) PBL cases, journal clubs and other exercises that integrate cutting-edge foundational sciences with clinical medicine; collaboratively approached in small groups of 8 students with a faculty facilitator.
2. **Inquiry Immersion:** Annual two-week blocks of scholarship skill-building experiences. Selective mini-courses dig deep into controversies in medicine.
3. **Deep Explore:** Required, in-depth scholarship experience, culminating in a capstone project that is completed incrementally during the four-year program.

Inquiry elements comprise 25% of UCSF’s new medical curriculum, reflecting a strong commitment to developing an “Inquiry Habit of Mind”. The curriculum will launch in August 2016.

CONCLUSION: Developing an inquiry-based curriculum will equip future physicians with the skills to address gaps in medical knowledge and discover innovative ways to address health problems of the future.

Abstract #5: A Two-Pronged Approach To Genetics Curriculum

Thomas Pace², Robert Saul², Peggy Wagner¹, Anna Cass², Robert Best¹ and **Renee LeClair**¹
¹University of South Carolina School of Medicine, ²Greenville Health System, 701 Grove Rd., Greenville, SC, 29605

BACKGROUND: For students to achieve competency within a genetics curriculum they must be able to confidently integrate basic knowledge with nuances of clinical care, delivery, counseling and referral. The latter of these facets are more specialized to clinical genetics and pose an evolving educational challenge. We have met this challenge with a two-pronged approach of horizontal integration of basic and clinical science in the M1 year, and vertical integration to provide longitudinal continuity of genetics content across the M1 and M2 years. This approach was delivered across parallel basic and clinical science courses.

METHODS: Horizontal integration was achieved with a series of mini-portfolio based projects that incorporated aspects of behavioral, social or clinical science along with the basic science genetics content. These projects were delivered (and assessed) within the basic science course. One project tasked students with taking a detailed family history used to: 1) generate a detailed pedigree consistent with the information gathered, 2) predict carriers or at-risk individuals, and 3) identify appropriate testing to determine affected or carrier status. In each of these mini-projects the application of basic knowledge (Mendelian and non-Mendelian inheritance, risk calculations and basic genomic diagnostics) was integrated with the assessment of other core competencies (interpersonal and communication skills, patient care and practice-based learning and improvement) that are an essential part of a genetics curriculum. To provide longitudinal continuity of genetics content, we intentionally selected 5 cases (sickle cell, breast cancer, prostate cancer, Parkinson's disease and arthritis) that were revisited across the undergraduate years within the clinical module. The initial interaction with the cases provided basic context while the M2 year reinforced content and explored nuances of the presentation.

RESULTS and CONCLUSIONS: The two-pronged approach within our curricular structure helps reduce marginalization of genetics content while requiring minimal dedicated time within any given week. Additionally, this spiraled design provides opportunity for assessment of developing clinical skills essential to burgeoning physicians and a platform for vertical integration of clinical genetics and genomics content across all 4 years.

Abstract #6: Utilization Of Genetic Counselors As Educators In Medical Schools

Subhadra Ramanathan

Loma Linda University, Loma Linda, CA, 92354

AIM: A preliminary assessment of the utilization of genetic counselors as educators in graduate medical education, especially in medical schools.

METHODS: 15-question survey, created on surveymonkey.com, was posted to the listserv of the National Society of Genetic Counselors (NSGC). The survey was available to 792 members and was open for 9 days.

RESULTS: A total of 100 responses were collected, all of them from genetic counselors, for a response rate of 12.6%. Among them, 32% were teaching in a medical school course and 40% were teaching in other graduate school courses, including programs in genetic counseling, nursing and physician assistant training. Among those teaching in medical schools, the majority (60%) taught in the freshman class, but at least 12% were involved in teaching in the Senior year. The most common topics of instruction were prenatal diagnosis/screening/teratogens, inheritance patterns and principles of genetic counseling. Others taught in a specialty, such as cancer, neuro/cardiogenetics, laboratory testing methods, quantitative genetics, metabolic genetics and ethics. About 44% of genetic counselors who taught in medical schools spent only 1- 5 hours in the classroom every year, 25% spent 5-10 hours, 6% spent 10-15 hours and 6% taught for more than 15 hours (26 respondents). The majority (80%) felt underutilized as educators. Only 9 (28%) participated in Active Learning (AL) modules in the medical school. Frequent AL topics were inheritance patterns, principles of genetic counseling, laboratory testing methods and quantitative genetics. Only 8 (22%) genetic counselors who taught in other graduate programs reported being involved in AL modules.

DISCUSSION: In the 2014 professional status survey of 1,896 genetic counselors, 64% reported being involved in the education of medical students, residents, fellows and physicians, formally and informally. This smaller survey illustrates that genetic counselors are involved in formal teaching of a variety of genetics topics in medical schools, although only a minority are involved in AL modules. Genetic counselors perceive that they could be better utilized as educators in graduate medicine, as most of them spend 10 hours or less in the classroom each year. As medical schools develop innovative curricula to teach genetics and genomic medicine to their students, AL modules are increasingly being incorporated. Recruiting genetic counselors to serve as educators will harness the expertise of available but underutilized professionals who are particularly qualified to do so by virtue of their training and experience.

**Abstract #7: Rare Diseases Research Certificate Program: Equipping
The Next Generation In Rare Disease Research**

Debra Regier, Bryce Comstock, Jeff Sestokas, Raven Tolson, Rachel Sarnacki, Mary Ottolini and Marshall Summar

Division of Genetics and Metabolism, Children's National Medical Center, Washington, DC, 20010

BACKGROUND: Rare disease research is a unique field that requires particular skills not consistently fostered in the current academic research setting. Rare disease research spans multiple medical specialties and funding streams. Rare diseases are defined by the small size of the affected patient populations and multidisciplinary approaches in research. The demand for this research does not match the current supply of researchers prepared to partake in the field. This program is designed to prepare the next generation of researchers for the rare disease field.

METHODS: A cohort of early career researchers will be selected from a variety of specialties and sub-specialties. The program includes an in-person seminar at the beginning of the program and an in-person seminar at the end of the program, with semimonthly webinars and asynchronous learning for the months in between. Outcomes are measured by posters submitted for conferences and scored by a rubric, communications skills measured by presentation rubric, their publications post program, and their five-year occupational outcomes.

RESULTS: We have successfully funded this novel program design and have begun the first year of this cohort including 21 participants from multiple specialties. The range of participants includes all types of specialty (pediatric, internal medicine, obstetrics and gynecology) and sub-specialty populations (pulmonary, hematology, oncology, nephrology, genetics, etc.). Cohort members expressed their satisfaction with didactics and one-on-one support of their research and trouble-shooting during the monthly series. Short-term evaluations of improved satisfaction with research and comfort with rare disease methods of research will be shown; however, long-term outcome data is pending.

CONCLUSIONS: Given the lack of prepared researchers participating in the rare disease field, this program has been a success by highlighting and identifying funding to support early career rare disease researchers. Specific curriculum to teach the nuances and best practices in rare disease research are essential to effectively equip and encourage early career researchers.

Abstract #8: Survey Of Primary Care Physician Opinions Of Most Important Genetic Disorders For Medical Students

Andrew K. Sobering¹, Shruthi Rethi¹, Emily Smith¹ and Tracey Weiler²

¹*St. George's University, St. George's, Grenada*, ²*Herbert Wertheim College of Medicine, Florida International University, Miami, Florida, 33199*

Professional genetics societies including the American College of Medical Genetics and Genomics (ACMG) and the Association of Professors of Human and Medical Genetics (APHMG) have identified potential gaps pertaining to genetics and genomics in the 4 year undergraduate medical curriculum¹. A long range plan of these organizations is to create case-based teaching tools to address these gaps². To best tailor these tools to the majority of medical students who entering primary care specialties, practicing physicians were surveyed regarding their opinions of the most important genetic diseases that should be included in the undergraduate medical curriculum.

An anonymous survey was developed including a set of demographic questions as well as a question asking practicing primary care physicians to provide a ranked list of genetic disorders that they feel should be included in the four years of the undergraduate medical school curriculum.

The first site to deploy the survey was at the St. George's University clinical faculty meeting, where 84 physicians participated: 55 of whom practice in the USA, 20 in the Caribbean, eight in the UK, and one in Europe. As expected, geographical location has a strong effect on the opinion of disorders of importance. As an example: ~75% of Caribbean physicians choose sickle cell disease (SCD) as an important genetic disorder for a teaching case (~55% as the most important), compared to ~47% of North American physicians (~10% as the most important). Additional analysis of the survey data stratifying by physician age and specialty will be presented, including the overall top 10 list of disorders considered most important by this cohort of surveyed physicians.

Additional sites will be surveyed in the near future, including Florida International University, Miami, Florida and University of Toronto, Canada. This data will inform the focus of case-based teaching materials for use in undergraduate medical education. Evidence from the first survey suggests that the list of disorders used for undergraduate medical education should be tailored to the region where most students will eventually practice.

¹ Plunkett-Rondeau et al *Genet Med.* 2015; 17:927-934

² American College of Medical Genetics and Genomics. American College of Medical Genetics and Genomics Strategic Plan. Accessed via the ACMG web site: https://www.acmg.net/docs/ACMG_FINAL_Strategic_Plan_071715_V2.pdf?hkey=54b2055f-09ae-402d-9e34-d7cb7cd2eec3.

Abstract #9: An Active Learning, Team-Based Approach For Cystic Fibrosis Cases

Susan Viselli and Nalini Chandar

Department of Biochemistry, Northwestern University, Downers Grove, IL, 60515

BACKGROUND: With goals of providing medical students practice calculating carrier frequencies, and describing genotypic variations in CF, we developed 2 cases. Students reviewed these individually and as members of their previously- formed teams. The first, an Instructional Case, included a healthy individual with a first-degree relative with CF, and had individuals from populations with different CF prevalence. The second, an Evaluation Case, included a male with congenital bilateral absence of vas deferens, as a consequence of compound heterozygosity for CFTR mutations.

METHODS: Monday, the week after 4 lectures on single-gene inheritance were given, the Instructional Case and 5 learning objectives were posted to Blackboard. Students were asked to review these prior to a faculty-guided 2-hour session on Thursday. This began by polling students using clickers and audience-response software. Faculty reviewed response statistics to multiple-choice questions and when needed, explained concepts. Scores were not recorded. Each team was next given the Evaluation Case and questions to answer over the next hour. A faculty-led discussion followed. Each team member received the same grade on the exercise. Lastly, students completed multiple-choice quizzes and individual scores were recorded.

RESULTS AND CONCLUSIONS: This use of active learning in a team-based setting effectively employed individual accountability along with team work to aid in student understanding of difficult concepts. Based on polling results, probability of a child affected with CF, given parental carrier risk, was initially answered correctly by only ~80% of 206 students. After additional examples, similar questions were answered correctly by close to 100% of students polled. Identifying carrier risk for a healthy person with a sibling with CF was answered correctly by < 50% of students initially but by >75% on the graded quiz. Team exercise grades ranged from 74-98% (mean 86.73 ± 5.11), with the most difficulty assessing carrier risk after negative results for a small number of CFTR mutations. The mean quiz score was $91.75 \pm 11.51\%$ (range 60-100), with 63% scoring 100%. The most commonly- missed question concerned carrier risk when a parent or sibling has CF; answered incorrectly by 64.17% of those missing 1 question and by 66.66% of those missing 2. Overall, students prepared for the exercise and remained engaged. We saw improvement in answers to questions related to the initially most difficult concepts.

Abstract #10: Warfarin Pharmacogenomics Case-Based Discussion

Tracey Weiler, Helen Tempest, Georg Petroianu and Gagani Athauda

Herbert Wertheim College of Medicine, Florida International University, Miami, FL, 33199

Pharmacogenomics correlates individual genetic variation with drug responses and is an integral part of precision medicine. Increasingly, practitioners have to incorporate patient genotypes when considering therapy options, including drug choice and dosing. Pharmacologic therapies often target a specific protein, which can exhibit allelic heterogeneity, thus specific drugs may demonstrate patient-specific sensitivity. Furthermore, the cytochrome P450 system metabolizes xenobiotics, and may convert a drug to a more-or-less bioactive substance. CYP450 enzymes also exhibit variation, particularly with respect to the rate of metabolism. For drugs that exhibit significant morbidity in patients with a specific genotype, best practices suggest that the patient's genotype should be taken into account when prescribing the drug. We sought to expose first-year medical students to the practice of pharmacogenomics through a case-based discussion (CBD).

A warfarin pharmacogenomics CBD was delivered to medical students during their Pharmacology course. A short study guide was provided including defined learning objectives. During the session, a brief didactic lecture summarizing the study guide was followed by presentation of 3 cases with different indications for warfarin therapy. Students in small groups answered questions related to the patient's warfarin sensitivity, bleeding/clotting risk, modifications of warfarin dose and other patient counseling issues. After working through the cases, answers were elicited using voting cards, and students were asked to defend their answers. An anonymous survey was administered to determine student satisfaction as well as their understanding of pharmacogenomics concepts.

The session was well received, and informal polling of voting responses revealed that the majority of groups came to the correct answers during the CBD. We believe that the **combination** of pre-session study guide, brief summary didactic lecture followed by the student discussion of disparate cases of warfarin treatment were responsible for student comprehension. Results of the survey indicated that the session increased awareness of the role of genotypes in drug response and furthermore, it increased student's confidence to discuss how pharmacogenomics can be used in clinical care.

We do not have information about the success of the intervention with respect to an individual student's ability to understand and use pharmacogenomics in clinical settings. In future, an individualized pre/post-test of pharmacogenomics knowledge will be administered to elucidate the relative utility of the study guide alone vs. the addition of the didactic lecture and group discussion.

Abstract #11: A Post-Baccalaureate Masters In Medical Genomics: Does It Affect Career Choices?

Karen Weissbecker

Hayward Genetics Program, Tulane University School of Medicine, New Orleans, LA, 70112

For the past 17 years, Tulane Hayward Genetics Center has offered a 1-year post-baccalaureate Masters in Medical Genetics and Genomics. The program is multidisciplinary with classes in medical, clinical, biochemical, and molecular genetics as well as cytogenetics and genetic epidemiology. In addition to coursework, students observe our geneticists and genetic counselors in clinic and write a thesis-style paper.

Most students are pre-med and the advertised purpose of the program is to “give graduates an in-depth understanding of the rapidly advancing field of human genetics and to prepare individuals for careers in the health sciences.” Our secondary purpose was to hope that we could perhaps influence future doctors and increase recruitment of trainees in Medical genetics. The Bambury Summit reported that the number of physicians choosing to enter the field of medical genetics is small, and getting smaller (Korf, Feldman and Wiesner, *Genetics in Medicine*, 7:433, 2005). The number one reason given for the decline was “lack of awareness of genetics/insufficient exposure in medical school”.

In assessing the first 12 years of the program, we found that of 116 students, 71 went on to medical or osteopathic school. Of the 20 medical students who have been out long enough and for whom we know what specialty they chose, none chose Medical Genetics. However, three students went into Genetics Counseling and 9 went into Doctorial programs – almost all with a genetic focus. In addition, one of our alumni is a senior scientist at Courtagen. Therefore, while we did not achieve our goal of recruiting future Medical Geneticist (yet), the program has influenced students in other genetic -related careers. A follow-up survey of knowledge and attitudes of genomics and genetic testing is planned to assess whether our alumni are better prepared than their colleagues for the era of precision medicine.

Abstract #12: Improving Success in Medical School: The Michigan Prematriculation Curriculum

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Students entering The University of Michigan Medical School (UMMS) have highly diverse academic backgrounds, including varied academic foci and life experiences. This diversity can initially hamper learners as they transition to the high intensity medical school-learning environment. The new UMMS curriculum is focused on integrating foundational science with related clinical experiences. The starting point for this integration, the Foundations in Molecular Medicine (FMM) course is focused on genetics and biochemistry. In order to enhance student readiness for FMM, we developed a formal prematriculation program (PMP) so students can self-assess knowledge, identify gaps and correct as needed. This voluntary asynchronous, on-line, formative intervention, included specified learning objectives, multiple-choice exams, identification of areas for further study/review, and links to online resources. 95% of the entering 2015-M1 class participated in the PMP, 65% completed all 10 pre- and post-test assessments with a range of pre- and post-test score improvements between 0.34% (amino acids/proteins) – 10.85% (cell biology). Written feedback from participants highlighted positive aspects related to refreshing knowledge, assessing base line knowledge and weaknesses while some learners felt there was too much redundancy with sequence content and limitations in the depth and breadth of content. Analysis of results from prematriculation program assessments (PMPAs) in association with first-half and second-half cumulative outcomes for FMM showed PMPA-cumulation (calculated as cumulative PMPA score (n=114, 67% of M1-2015) for students who completed at least three of the five post-program assessments) had moderate, statistically-significant correlations with both FMM first-half cumulation ($R=0.401, p<0.001$), and FMM second-half cumulation ($R=0.393, p<0.001$). Multiple regression analysis found an impact of PMPA-cumulation in both settings even after controlling for GPA and MCAT-composite scores (FMM first-half cumulation ($p=0.002$) and FMM second-half cumulation ($p=0.003$). Future directions for the PMP include increasing the depth and breadth of content, understanding the impact on longterm individual performance and exploring opportunities for additional utilization in the UMMS curriculum.



*Platform
Presentation*

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