aphmg
Association of Professors of Human and Medical Genetics

annual meeting

MAY 2-5, 2023

Kiawah Island Resort

Kiawah Island, SC

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PRESIDENT'S WELCOME

Dear Colleagues and Friends,

Welcome to the Annual Meeting of the Association of Professors of Human & Medical Genetics in Kiawah Island, South Carolina!

As we gather on beautiful Kiawah Island, we honor the Kiawah people, a subtribe of the *Cusabo* people, who were the first custodians of this land. We are thrilled to reunite here with dear colleagues and friends and engage with new colleagues.

Our APHMG leaders and members have been very active this year in forging collaborations with other professional organizations, continuing to improve inclusivity in our classrooms and clinics, disseminating our genetics curriculum competencies, and addressing genetics workforce challenges. As we prepare our students and trainees to practice genomic medicine in an inclusive and effective manner, we are aware of the current political climate and how laws restricting access to safe abortion and gender-affirming care impact clinical practice as well as genetics education. Plenary sessions on these topics will offer us as a genetics education community a forum to discuss ways to stand together and address these issues. We hope the opportunity to network and share ideas with colleagues will energize APHMG members and guests for future collaborations among clinicians, scientists, and educators.

Each of the APHMG Special Interest Groups (SIG) (Residency Program Directors, Medical School Course Directors, and Laboratory Fellowship Program Directors) have much to share and have individual meetings on Wednesday designed for discussion, problem solving, and the development and sharing of common resources. I am very excited about the work of our SIGs and committees this year, especially focusing on genetics competencies; the genetics workforce; diversity, equity, and inclusion in teaching and in clinic; and recruitment of outstanding trainees. I look forward to hearing the reports from our SIGs, committees and partner organizations at our business meeting, and welcome all of you join us for these updates and a bit of fun too!

We hope if you are not a member that you will take this opportunity to officially join our APHMG family, and we are delighted to have everyone here with us this week.

Welcome to Kiawah Island!

Kathy

Katherine M. Hyland, PhD
President, APHMG

CONFERENCE AGENDA

TUESDAY, MAY 2

All Day	*Main Arrivals / Explore Kiawah Island Resort*	*on your own*

WEDNESDAY, MAY 3

Note: All APHMG sessions take place in the West Beach Conference Center

7:00 am – 5:00 pm	APHMG Registration Desk Open	WBCC Lobby
9:00 am – 10:00 am	Continental Breakfast (*all attendees*)	WBCC Lobby
10:00 am – 5:00 pm	**Medical Genetics Residency PD SIG Meeting**	Azalea 4/5
12:00 pm – 1:00 pm	Group Lunch (everyone)	Carolina 5
1:00 pm – 5:00 pm	**Clinical Laboratory Training PD SIG Meeting**	Carolina 3
2:30 pm – 3:00 pm	COFFEE BREAK	WBCC Lobby
3:00 pm – 5:00 pm	**Medical School Genetics CD SIG Meeting – Part I**	Carolina 4
5:00 pm – 6:00 pm	APHMG Council Meeting (*Closed Meeting*)	Sweetgrass
6:00 pm – 7:00 pm	**Joint Welcome Reception with ABE**	West Beach Pool
7:00 pm – 8:00 pm	**Welcome Reception continued** (*APHMG only*)	West Beach Pool
7:00 pm – 8:00 pm	OPEN POSTER VIEWING	Carolina 5
8:00 pm – 9:30 pm	**APHMG Business Meeting** (*Dessert & Coffee*)	Carolina 3/4

Share your photos with us!
Scan to view & upload to the APHMG 2023 Meeting album

CONFERENCE AGENDA

THURSDAY, MAY 4

7:00 am -1:00 pm	APHMG Registration Desk Open	WBCC Lobby
7:00 am – 8:00 am	Continental Breakfast	WB Lobby / Carolina 5
8:00 am – 10:00 am	**PLENARY SESSION I:**	Carolina 3/4

Competency-Based Medical Education: Is it Time for Genetics?
Greg Ogrinc, MD, Senior VP, Certification Standards and Programs at ABMS
Miriam (Mimi) Blitzer, PhD, CEO, ABMGG; Professor, Univ. of Maryland SOM
Laura Edgar, EdD, Vice President, Milestones Development, ACGME
Kate Hatlak, EdD, Director, Faculty Development and Special Projects, ACGME

10:00 am – 10:30 am	COFFEE BREAK & Poster Viewing	Lobby / Carolina 5
10:00 am – 10:10 am	Group Photo	West Beach Lawn
10:30 am – 12:00 pm	**PLENARY SESSION II:**	Carolina 3/4

The Clinical Genetics Workforce: Challenges and Opportunities
Kevin Glinton, MD, PhD, Baylor College of Medicine
Miriam (Mimi) Blitzer, PhD, CEO, ABMGG; Professor, Univ. of Maryland SOM
Cindy Powell, MD, Professor, Univ. of North Carolina

12:00 pm – 1:00 pm	Group Lunch	Carolina 5 / WB Lawn
1:30 pm – 4:30 pm	Optional Group Activity: **Bicycling on the Beach** **Reserve bikes though Kiawah Resort*	West Beach Bike Shop
6:00 pm – 9:00 pm	**APHMG Conference Dinner** *Included for all registered attendees.* *Meet at the WBCC Lobby starting at 5:45 pm for the Kiawah shuttle to Turtle Point Clubhouse*	Legends at Turtle Point *2nd floor*

CONFERENCE AGENDA

FRIDAY, MAY 5

7:00 am – 12:00 pm	APHMG Registration Desk Open	WBCC Lobby
7:00 am – 8:00 am	Continental Breakfast	WB Lobby / Carolina 5
8:00 am - 9:30 am	**PLENARY SESSION III:**	Carolina 3/4

Genetics Education in the Post-Dobbs Era
Laura Hercher, MS, Director of Student Research
Sarah Lawrence College Program in Human Genetics
Susan Klugman, MD, FACOG, FACMG, Professor,
Obstetrics and Gynecology and Women's Health
and Pediatrics, Albert Einstein College of Medicine
Montefiore Medical Center; President, ACMG

9:30 am - 10:00 am	COFFEE BREAK	WB Lobby
10:00 am - 12:00 pm	**PLENARY SESSION IV:**	Carolina 3/4

Leveraging Large-Scale Sequencing Projects for Students and Trainees
Sawana Biswas, MS, LCGC, Scientific and Clinical Lead
UCSF Health Center for Clinical Genetics and Genomics
Senior Genetic Counselor, UCSF
Rosanna Sanchez Russo, MD, FACMG, Asst. Professor,
Emory University (All of Us)
Anna Hurst, MD, MS, FACMG, Assoc. Prof., Dept. Genetics
University of Alabama at Birmingham
Katherine Hyland, PhD, Professor, Dept. Biochemistry,
Institute for Human Genetics, University of California San Francisco
Stephen Moore, MBS, PhD, FACMG, Assoc. Professor, Dept. Molecular and Medical Genetics, Oregon Health and Science University

12:00 pm - 12:15 pm	**Closing Remarks/Meeting Adjourns**	
12:30 pm – 2:30 pm	**Medical School Genetics CD SIG Meeting – Part II** (*with lunch*)	Carolina 3/4

aphmg annual meeting
MAY 2-5, 2023 KIAWAH ISLAND, SC

Safe travels home, everyone!

Don't forget to share your photos with us

Notes

MEDICAL GENETICS RESIDENCY PROGRAM DIRECTORS SIG MEETING AGENDA

WEDNESDAY, MAY 3

9:00 – 10:00	Continental Breakfast	WBCC Lobby
10:00-10:15	**Welcome & Introductions** Debra Regier, MD, PhD, Children's National Hospital	Carolina 3
10:15-10:30	**Report on the 2022 Match** Debra Regier, MD, PhD, Children's National Hospital	
10:30-11:00	**Report from ABMGG** Miriam (Mimi) Blitzer, PhD, CEO, ABMGG Professor, Univ. of Maryland SOM	
11:00-11:30	**Reports from the RRC/ACGME Update** Cindy Riyad, EdD, Director, Faculty Development and Special Projects, ACGME Melissa Meredith, MD, Chair RRC	
11:30-12:00	**Discussion**	
12:00-1:00	LUNCH BREAK	Carolina 5/WB Lawn
1:15-2:15	**Best Practices for At-Risk Trainees:** **Joint Session with Lab Director SIG** • Introduction to the topic • Panel discussion on implementation	
2:30 – 3:00	COFFEE BREAK	
3:00-4:00	**At-Risk Trainees: Case Scenarios and Conversations of Remediation Options** *(private tables for PD, Trainees, Coordinators)* *"Anonymized and combined" cases from membership – feel free to reach out to Dr. Regier if you have a case you would like anonymized and merged with others*	
4:00-5:00	**Citations: Prevention, Remediation and Best Practices** David Stevenson, MD Debra Regier, MD, PhD	
6:00-8:00	**APHMG Welcome Reception & Poster Viewing**	WB Pool / Carolina 5

CLINICAL LABORATORY TRAINING PROGRAM DIRECTORS SIG MEETING AGENDA

WEDNESDAY, MAY 3

1:00-1:05	**Welcome & Introductions** Steve Moore, Chair and APHMG President Elect	Azalea 1/2
1:05-2:05	**Best Practices for At-Risk Trainees:** **Joint Session with Medical Genetics PD SIG**	
2:10-2:40	**Report from ABMGG** Mimi Blitzer, CEO, ABMGG	
2:40-3:10	COFFEE BREAK	
3:10-3:50	**Reports from ACGME** Cindy Riyad, ACGME Executive Director, Review Committees for Medical Genetics and Genomics, Pathology, and Preventive Medicine Melissa Meredith, NIH, ACGME Review Committee Chair MGG	
3:50-4:30	**LGG Program Directors Survey** Josh Deignan, UCLA	
4:30-4:50	**Fellow's Conference** Marco Leung, incoming Co-Chair LD SIG Nationwide Children's and The Ohio State University	
4:50-5:00	**Discussion and Closing** Steve Moore	

6:00-8:00	**APHMG Welcome Reception & Poster Viewing**	WB Pool / Carolina 5

MEDICAL SCHOOL GENETICS COURSE DIRECTORS SIG MEETING AGENDA

WEDNESDAY, MAY 3

2:30-3:00	COFFEE BREAK	WBCC Lobby
3:00 – 3:05	**Introduction and Welcome Remarks** Lauren Massingham, MD Chair CD-SIG Executive Committee	**Carolina 4**
3:05-4:05	**PLENARY SESSION AND WORKSHOP:** **GenZ and Me: Transforming Teaching Methods for the Current Generation of Medical Students** **Paul McDermott**, PhD, Professor of Medicine, Associate Dean for Faculty Affairs and Development, Director Academy of Medical Educators, Medical University of South Carolina	
4:05-4:15	**American College of Medical Genetics (ACMG) Student Special Interest Group (SIG) Program: Expanding the Knowledge of Medical Genetics and Genomics** Wesley Patterson, PhD, MSPA, PA-C, CAQ-Peds Greenwood Genetic Center	
4:15-5:00	**Selected Abstract Presentations** *Optimizing Onboarding for Medical Genetics and Genomics Residents* Annie Niehaus, Stanford University *Development of a Genetics-Specific Post-Graduate DEI Curriculum* Jessica Gold, Children's Hospital of Philadelphia *The UTHealth Houston Cardiovascular Genomics Certificate Program: Online On-Demand Genetic Training For Clinicians* Vinyana Murthy, George Washington University	
6:00-8:00	**APHMG Welcome Reception & Poster Viewing**	WB Pool / Carolina 5

FRIDAY, MAY 5

12:15	Grab & Go Lunch in foyer for CD-SIG attendees	
12:30-1:30	**NBME-Style Writing** Miriam Blitzer, PhD, CEO, ABMGG	**Carolina 3/4**
1:30-2:30	**Q-Bank Update** *Group interactive sessions to edit additional questions in NBME format*	

ORAL ABSTRACTS

O1

Optimizing Onboarding for Medical Genetics and Genomics Residents

Annie D. Niehaus, MD and David A. Stevenson, MD.
Division of Medical Genetics, Department of Pediatrics, Stanford University

Purpose: The purpose of this project was to develop an Orientation framework for new Medical Genetics and Genomics (MGG) residents.
Background: Residents entering into MGG programs have diverse training backgrounds. While some may have completed full residencies (eg, pediatrics, internal medicine), others may have completed just a one-year internship. Adding to the complexity, training environments - ranging from smaller, community hospitals to large, academic, tertiary care centers - have the potential to differ starkly in terms of exposure to medical genetics. Additionally, many trainees have spent years in the workforce or completed additional degrees. Given diverse backgrounds, residents entering MGG programs potentially have vastly different baseline genetics and clinical knowledge. The American Board of Medical Genetics and Genomics (AMBGG) developed a list of core competencies to guide program directors during the development and evaluation of the educational content of training programs. However, the field is lacking in literature related to which of these competencies may have been mastered in prior training environments and/or those that are vital for new residents to learn within their first few days, weeks, or months. Greater characterization of the structure of an ideal "orientation" is needed.
Methods: A workgroup composed of residents and faculty reviewed our program's onboarding curriculum and coded topics based on the ABMGG Clinical Genetics and Genomics competencies (2019). The workgroup assessed which themes were represented during onboarding, and whether different training backgrounds influenced opinions on which topics should be prioritized in future years.
Results: Our current onboarding curriculum includes topics from all "six competences," with greater representation of topics related to patient care (10 hours) and systems-based practice (8.5 hours) compared to genetics knowledge (5 hours), interpersonal and communication skills (2 hours), practice-based learning and improvement (2 hours), and professionalism (2 hours). On a scale of one (not at all valuable) to five (incredibly valuable), residents ranked onboarding topics related to the core competencies of "patient care" and "systems-based practice" to have an average importance ranking of 4.4, whereas topics related to "genetics knowledge" received an average of 3.6. Additional themes that arose were the need to spend more time on the acute management of metabolic conditions and that residents from different training backgrounds prioritized the inclusion of similar topics during onboarding.
Conclusions: Results from this project will help guide the selection of onboarding topics at MGG residency programs. Future work can focus on ideal learning environments for these competencies.

O2

Development of a Genetics-Specific Post-Graduate DEI Curriculum

Jessica I Gold, Sanmati Cuddapah.
Division of Human Genetics, Department of Pediatrics, Children's Hospital of Philadelphia

Purpose: Clinical genetics, with its history of perpetuating racial pseudoscience, is increasingly recognizing race-based health inequities. Existing diversity, equity, and inclusion (DEI) efforts lack generalizability to this field. A targeted examination of medical genetics is necessary to reform structural racism.

Background: Modern clinical genetics is beset with structural racism. Dysmorphology atlases lack diversity, and patients with non-White ancestry are less likely to be diagnosed by this exam. Genetic screening favors variants common to patients of White European ancestry, leading to delayed diagnosis and increased morbidity in non-White patients. Underrepresented patients (of Black/Latinx ancestry) tend to have lower rates of genetic testing and a lower likelihood of receiving a diagnostic result. With increasing reliance on genetics for clinical management, disease surveillance, and risk stratification, it is critical to address race-based limitations and implement strategies for improvement.

Methods: We developed a curriculum for adult learners in the Divisions of Human Genetics at the Children's Hospital of Philadelphia (CHOP) and the University of Pennsylvania (Penn), which include clinical-based, laboratory-based, and research-based members. Our curriculum includes large-group expert lectures, small-group discussions, and book clubs. Curriculum assessment focused solely on post-graduate trainees of the medical genetics residency and advanced clinical and laboratory genetics fellowships, who completed a pre-test on the history of eugenics and dysmorphology, genomics, and genetic testing in diverse populations.

Results: Pre-test assessment demonstrated high perceived need for this curriculum among post-graduate trainees. Over 88% felt that addressing structural racism in medical genetics was "very important" with 100% indicating that formalizing learning would greatly impact their clinical practice. Average score on the knowledge assessment was 50.6±14.6%. Two of 4 scheduled expert lectures have occurred and were well-received with over 140 attendees each. Over 45 individuals joined our Genetics DEI book club, reading *Superior* by Angela Saini. All sectors of the CHOP/Penn Genetics were well represented, leading to a lively discussion on the intersection between expanded carrier screening and eugenics, challenges in consenting underrepresented minorities for genomic research, and methods to improve patient experience.

Discussion: Our curriculum has been embraced by CHOP/Penn Genetics. Active participants represent all sections of the division, enriching our examination into inequities at the personal, institutional, and global level. While this initial curriculum has skewed toward investigation and reflection, we anticipate a second phase focused on action, starting with a trainee-designed quality improvement project on delays in whole exome sequencing workflow for non-native English speakers.

ORAL ABSTRACTS

O3

The UT Health Houston Cardiovascular Genomics Certificate Program: Online On-Demand Genetic Training for Clinicians

Vinyana Murthy, *University of Texas Health Science Center, Houston, TX*

Objectives: The primary goal of the UTHealth Cardiovascular Genetics Certificate program (CGC) is to fulfill an unmet need for adult cardiovascular genetic education by improving the recognition, assessment, and timely referral of patients with heritable cardiovascular diseases and by emphasizing practical skills to deliver personalized genomic medicine in daily clinical practice.

Methods: Content experts from five institutions and the National Institutes of Health collaborated to develop a free asynchronous online course in cardiovascular genetics for adult healthcare providers who do not have formal genetics training. The program consists of 24 modules in three tiers of increasing complexity that require the intensive application of genetic concepts to clinical scenarios. To unlock the modules, participants take a pre-course assessment of their genetics knowledge. In Tier 1, learners begin with short presentations on basic genetic concepts and terminology before progressing to apply these concepts to workshop-style activities in Tier 2 clinical scenarios. In Tier 3, learners navigate through prolonged cases which require them to contextualize genetic concepts in clinical practice. The clinical scenarios strongly emphasize collaborative assessment of patients and include simulated counseling sessions. The program is free and self-paced and provides CME or CNE credit to learners. Select pilot users in clinical programs at UTHealth Houston provided feedback via surveys implemented in REDCap before the course was published to outside learners on Canvas Catalog.

Results: Pilot learners strongly endorsed the course. They rated the modules as easy to navigate and wrote that the course improved competencies in the recognition of heritable diseases, interpretation of genetic information, and familiarity with cascade testing.

Conclusion: Provider training through the CGC is intended to benefit patients and save lives due to earlier detection of genetically triggered diseases and more frequent cascade testing. We will utilize longitudinal feedback to enhance the course experience for learners and to evaluate changes in clinical practice after exposure to CGC concepts.

POSTER ABSTRACTS

P1

Building Clinical Genetics and Genetic Testing Familiarity in Undergraduate Medical Education through Self-Directed, Small Group Case Studies

Mindy Hoang (OMS), Marie Discenza (OMS), and Michael B. Wells, PhD. *Idaho College of Osteopathic Medicine, Meridian, ID*

Purpose: Clinical Genetics is a growing medical education emphasis, as new technologies open possibilities for personalized patient treatments. This specialty interacts with one of the fastest growing areas of medical knowledge. Non-lecture activities can offer learners opportunities for small group-driven content exploration and application. Problem-based learning is an effective approach to add a clinical context to genetics training. We created an iterative series of worksheets, case studies, and role-playing scenarios tailored to improving student awareness and skills related to clinical genetics testing and analysis.

Background: Idaho College of Osteopathic Medicine is committed to training caring and competent physicians prepared to care for persons in our five-state region and beyond, with a focus on rural and medically underserved communities. According to various sources, there are less than 10 Clinical Geneticists in Idaho. More broadly, there is a growing need for physicians to be familiar with genetic testing, particularly in rural and underserved communities, as new tools are brought into practice and medical genetics knowledge continues to grow rapidly. Existing studies and toolkits introduce undergraduate medical students to this topic, but very few focus on exploring clinical decision making; and applying genetics skills within the context of detailed, dynamic patient scenarios.

Methods: Two iterations of instructional materials were created and utilized during subsequent years of a two-hour active learning session in the Reproductive System course. First, worksheets exploring online resources (UpToDate, OMIM) were created and utilized. Second, we created guided learning scenarios to provide a better understanding of medical genetics knowledge application within complex patient scenarios. Incorporating role playing into this session gave learners the opportunity to hone teamwork, peer teaching, effective communication, and empathy skillsets.

Results: Learners explored the scenarios in small groups, completed a series of related assessments, and provided anonymous, evaluative feedback about clinical genetics applicability in medical care before and after this session. Learners viewed worksheets as time-intensive. Case scenarios will be evaluated when this session occurs again, in early April 2023.

Discussion/Conclusions: Promoting familiarity with clinical genetics and testing as a skillset and a specialty supports growth of this cutting-edge field. Increased knowledge and resource usage support patient care and physician awareness of available treatment options and concerns.

POSTER ABSTRACTS

P2

When You Hear Hoofbeats, Think Zebras: A Genetics Residency Curriculum Needs Assessment

Amanda Pritchard, *Div. of Pediatric Genetics, Metabolism, and Genomic Medicine, Department of Pediatrics, University of Michigan, Ann Arbor, MI*

Purpose: Geneticists are expected to have a broad fund of knowledge of many rare diseases. While there are lists of curriculum topics provided by the ACGME as well as ABMGG board review codes, there is not a standardized national course for trainees. This work aims to assess our program's current didactic curriculum in a way that could be recapitulated at other institutions.

Background: The didactic curriculum for our program has included auditing two graduate-level genetics courses in addition to supplemental Grand Rounds, Journal Clubs, Case Conferences, and Biochemical Genetics Didactics. However, as the some of the content these learning experiences may vary from year-to-year, this work focused on assessment of the genetic course curriculum through multiple methods.

Methods: A general needs assessment was first performed through curriculum mapping. The curriculum mapping charted the current graduate course lectures in relation to topics on the ABMGG Clinical Genetics and Genomics and General Exam content codes to identify gaps in coverage and areas covered in greater depth. In addition, five years of in-training exam missed question topics were collated to identify commonly missed topics. After completing these general needs assessments, a targeted needs assessment was initiated through ongoing semi-structured interviews to enable qualitative analysis.

Results: General needs assessment identified that the audited courses provide deep coverage of topics of chromosomal basis of inheritance, chromosome and gene structure and function, and Mendelian inheritance, among others. Certain clinical topics are sparsely covered. Pooled missed in-training exam topics revealed several commonly missed topics that may benefit from dedicated didactic sessions. Semi-structured interviews are ongoing; themes include noting the courses have more depth in some areas than may be needed for clinical geneticists, and several ways to improve the curriculum have been identified.

Discussion: Providing a strong foundation in genetics through a didactic curriculum requires ongoing curriculum assessment in the ever-changing landscape of the field. Periodic curriculum mapping and needs assessments can provide feedback to allow for improvements. Qualitative research methods may provide small training programs a way to identify themes and areas for development that may not be identified on standard evaluation forms or surveys.

P3

Improving Medical Student Comfort in the Delivery of Genetic Diagnoses

Gianna M. Arrizurieta[1], Juan P. Cordero[1], Elizabeth A. Roth[2], Jessica R. Vissicchio[2], Tracey Weiler[1], Andrew K. Sobering[2]
[1] *Florida International University (FIU) Herbert Wertheim College of Medicine, University Park, Florida;*
[2]*Augusta University/University of Georgia Medical Partnership (MP), Athens, Georgia*

Purpose To improve medical student comfort in the delivery of genetic diagnoses.

Background The delivery of difficult news is essential to medical practice and the development of these skills should begin early in medical training. Because genetics is often associated with difficult diagnoses, and it is introduced early in the preclinical medical curriculum, it is an excellent opportunity to practice learning of this skill.

Methods The genetics interest groups at AU/UGA Medical Partnership (MP) and Florida International University (FIU) Herbert Wertheim College of Medicine hosted independent lunch and learn sessions for first- and second-year medical students to learn about the delivery of difficult genetic diagnoses from a pediatrician or certified genetics counselor. First and second year medical students (n=48) at MP and FIU were given a pre-session survey to assess their comfort with delivering difficult news. Later, they were presented with the following case: A couple presents to their obstetrician after undergoing prenatal screening. The woman (G3P0) is 41-years-old and the prenatal amniocentesis is consistent with Down syndrome. The students broke into small groups to practice delivering the diagnosis to the family, while faculty moderators assisted students with questions and provided feedback on the practice scenarios. A combined total of 23 students in attendance completed a post-survey to gauge session response.

Results The pre-survey showed that the majority of students are uncomfortable with delivery of difficult genetic diagnoses. More than half of the 48 students felt very uncomfortable (21%) or somewhat uncomfortable (37%) in delivering genetic diagnoses prior to the session. The most cited factor influencing comfort level was lack of experience (89%). Other commonly selected factors included reaction of patient and family (60.9%), unfamiliarity with prognostic information (56.5%), and lack of information regarding the disease/disorder (50%). The post-session survey showed that practice helped to increase comfort level in delivery of genetic diagnoses. A majority of respondents agreed that formal training in the delivery of genetic diagnoses during preclinical curriculum was either important or very important (82.6%).

Discussion Our results indicate that a brief introduction to the delivery of sensitive genetic diagnoses followed by practice in breakout groups decreases discomfort and increases confidence in this skill. Participant feedback revealed that students would prefer more formal training with the skill integrated into curriculum. Every occasion to improve these skills is an opportunity to increase student knowledge and confidence as they prepare for their clinical careers.

POSTER ABSTRACTS

P4

Applications of Molecular Techniques in the Clinical Laboratory: An Immersive Educational Experience for Clinical Learners

Scott Turner, Guoyan Gao, Colleen Jackson-Cook, Elizabeth S. Barrie, Andrea Ferreira-Gonzalez. *Virginia Commonwealth University*

Purpose: The intensive two-week Molecular Pathology Practicum provides instruction and laboratory experience in the utilization of molecular techniques in the clinical laboratory.

Background: Opportunities for learners to get hands-on experience within a clinical genetics laboratory are limited. The practicum in Molecular Pathology at Virginia Commonwealth University was developed to provide an immersive 80-hour laboratory experience for a wide variety of clinical and non-clinical scientific professionals; including industry professionals, genetic counseling students, medical students, residents, and fellows from a variety of fields (Pathology, Genetics, Heme/Onc), as well as researchers and practicing physicians interested in continuing education in genetics and clinical laboratory medicine.

Methods: This course is the culmination of work over the past decade and has included over 100 participants from around the world and across various institutions and disciplines. Enrollment for the practicum is limited to no more than three participants/session. Through didactic presentations and diagnostic laboratory work, participants gained experience and an understanding of practice issues involved in molecular diagnostics. The effectiveness of the practicum for our clinical learners was assessed with a pre- and post-practicum quiz to assess knowledge gains.

Results: By following a strategy of limited enrollments, the practicum coursework has been tailored to each small group, based on their backgrounds and previous expertise. Upon completion of the practicum, participants can understand basic vocabulary and concepts of molecular diagnostics, and perform the following techniques: DNA extraction, RNA extraction, PCR, RT-PCR, quantitative PCR, fragment analysis, STR analysis, and sequencing technologies. They are also able to interpret the results of the tests performed, as well as identify and develop a plan for how molecular diagnostic techniques may be applied in their field or workplaces.

Conclusions: This practicum course has provided a unique educational opportunity for a variety of learners. This experience has supplemented education in genetics, molecular pathology, and laboratory medicine to rotating medical students for which these topics are underrepresented in the current curriculum allowing for more informed career decisions. It has allowed for a more in-depth understanding of methods and limitations of molecular techniques for genetic counselors, as well as specialist in infectious disease, oncology, and genetics, all of whom are responsible for clinical test ordering. This experience allows them to provide a more informed consultation with patients. Additionally, it has provided opportunities to bring knowledge and expertise to help establish molecular testing in clinical laboratories around the globe.

P5

Exploring the perceptions of osteopathic medical students on the importance of research in their medical education and residency placement

Neami Tervil, B.S, M.S., Zeynep Gromley Ph.D., Adam Gromley Ph.D. *Lincoln Memorial University-DeBusk College of Osteopathic Medicine Harrogate, TN*

Background: Currently, COMLEX-USA Level 1 and USMLE Step 1 licensure board exams reporting three-digit numeric scores are Pass/Fail. The National Resident Matching Program (NRMP) survey report in 2021 indicates that residency program directors rate the mean importance of involvement and interest in research in deciding whom to interview a 3.6/5. Rated on a scale of 1 (not all important) to 5 (very important). Due to these transitions and findings, we hypothesize that medical students will seek opportunities, such as research involvement, to be a competitive applicant for residency match.

Purpose: In this study we aim to assess first-year medical students' perceptions and views towards the importance of research experience in their medical education and for residency placement.

Methods: The first-year osteopathic medical students were recruited by email containing a link to an online Qualtrics questionnaire. The survey questions were aimed to explore whether the students were interested in gaining research experience and furthermore, we try to understand if the interest of residency specialty would be a factor for seeking research opportunities during their medical education. Participation was voluntary and data responses were collected anonymously.

Results: Our survey results revealed about 80% of students who took the survey reported that research experience is important factor for residency placement. These respondents plan on seeking research during the summer between first and second year of medical education. Interestingly, respondents who disagreed with the statement were among the group who reported an interest in primary care fields of family medicine, internal medicine, pediatrics and OBGYN.

Conclusion: Our data suggest that students who would like to be a competitive applicant for residency placement seek research opportunities. Our results suggest that students believe that involvement or understanding of research is important for residency placement and majority plan on seeking research during medical education. Further research should be conducted to determine if medical students' perspectives will change in third and fourth year for the residency application cycle.

POSTER ABSTRACTS

P6

Cytogenetics and Molecular Genetics in the Era of Cancer Genomics: Development of a Course for LGG Trainees

Suzanne Hart, PhD, *Director of Education, NHGRI Medical Genetics and Genomic Medicine Residency and Fellowship Training Programs*

Purpose: To develop tools to support laboratory cancer training for Laboratory Genetics & Genomics (LGG) trainees at NHGRI/NIH.

Background: Feedback from NHGRI/NIH trainees who took the 2021 ABMGG certifying exam in LGG indicated that additional education in cancer genetics would have been desirable, especially additional practice with cytogenetics nomenclature**.**

Methods: Using the ACMG short course as a guide, a 14-week course was developed, evenly split between cytogenetics and molecular genetics. Each week consisted of 2 hours of presentations that were live-streamed over zoom as well as recorded for review at a later time. Presenters were all experts in the field. Both core lectures and case studies were presented. Presenters were encouraged to use zoom polling questions to gauge student learning in real time. Student knowledge was formally assessed based on performance on a midterm and final exam, both administered online.

Results: The course was offered through the Foundation for the Advanced Education in the Sciences (www.faes.org) for the first time in the spring of 2022 under course code GENE 527. There were 20 students enrolled, including 12 LGG fellows from our training program at NHGRI/NIH. The remaining 8 students were NHGRI clinical trainees (n=2), a a graduate of the NHGRI Molecular Fellowship (n=1), fellows/employees at Johns Hopkins University, University of Chicago and University of Pittsburgh (n=5). Only one student (employee at a university) failed to complete the course.

Conclusions: Based upon the course feedback, we anticipate continuing to offer this course biannually (in the spring of even years). Since this is a virtual course, enrollment is open to all.

P7

Exploring the Graduate Medical Education Framework for Training in Clinical Gene Therapy

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Background: Clinical gene therapy is an emerging field in medicine that aims to cure or alleviate genetic disorders. The complexity of gene therapy and ongoing efforts to develop novel gene therapy products prompted us to explore approaches to clinical training focusing on emerging gene therapy drugs.

Objectives: To explore the feasibility, advantages and disadvantages of approaches to training residents in GME environments with variable resources.

Methods: We conducted a literature review to identify published opinions on gene therapy training. Expert opinions were sought from professionals with expertise in the ACGME and ABMGG requirements, in the field of clinical research, regulatory medicine, and gene therapy development.

Results: We identified several potential strategies that medical genetics and genomics programs can use to advance clinical training in gene therapy: short term rotations, a clinical research non-accredited year, and non-standard training program. (1) Short term rotations can allow residents to gain introductory experiences in selection of appropriate candidates for drug administration, management of adverse events and long-term follow-up. This can be achieved by rotating through established multidisciplinary gene therapy clinics for a short period of time. (2) A non-accredited clinical research year can provide an opportunity for residents to pursue advanced specialized training or research opportunities in gene therapy that are not covered by the standard curriculum. This can include attending specialized courses and pursuing focused clinical experiences in a specific area of gene therapy conducted by individual labs. This non-accredited year can help residents tailor their training to their individual interests and career goals. (3) Non-standard training programs can create opportunities for residents to gain experience with the gene therapy technologies and techniques in a structured environment utilizing the ACGME Milestones 2.0 framework.

Conclusions: Medical genetics and genomics programs can play critical role in preparing residents for emerging gene therapies by providing a strong foundation in genetics and genomics, training in genetic counseling, exposure to the latest gene therapy technologies and techniques. Surveys, focus groups, and expert opinions can be unitized to develop topics for curriculum and proficiency expectations using the ACGME Milestone 2.0 framework.

P8

Sleep Deprivation: The Effect of Home-based On-Call in Physicians

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Background The importance of sleep for physicians in training has been well studied and described. Here, we consider the effect of interrupted sleep on those that serve the medical community through home-based call as attending-level providers. Previously, two subspecialty populations, urology and otolaryngology have evaluated the impact of home-based call on fellows. As the number of rare disease patients diagnosed increase, the need for immediately-available experts to support the care for these patients is increased. Thus, the role of home-based call for subspecialists continues to grow.

Methods Here, we describe a pilot survey of 25 physicians who perform home-based call and the effects that this responsibility has on their sleep patterns. This survey was reviewed by the Children's National Hospital IRB. The survey was distributed via RedCap to physicians, using targeting to attending level physicians.

Outcomes In the first 25 participants, the average sleep not on call (7.44 hours) was significantly more than that while on home-based call (5.28 hours, $p<0.005$). The number of calls per night leading to this sleep disturbance was less than 5 calls from midnight to 6 am. Respondents most commonly reported that the home call had "some effect" on sleep followed by "moderate effect". Extensive and little effect were also chosen. No effect was not chosen by a respondent. We also asked about the effects of home call on adaptive behaviors. This included napping, decreasing social activities, decreased time spent with children, decreasing self care habits (exercise, spouse), increasing home-based supports (ordering food, child care). The most emotionally charged responses were noted to be about having less or worsened interactions with provider's children.

Conclusion Home-based call impacts the sleep, wellness, and requires accommodations for the physical, social, and mental impacts of this on the providers studied.

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