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Compassion and equity-focused clinical genomics training for health professional learners

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Abstract

There remains an urgent need for expanded genomics training in undergraduate medical education, especially as genetic and genomic assessments become increasingly important in primary care and routine clinical practice across specialties. Physician trainees continue to report feeling poorly prepared to provide effective consultation or interpretation of genomic test results. Here we report on the development, pilot implementation, and evaluation of an elective offering for pre-clinical medical students called the Sanford Precision Health Scholars Immersive Learning Experience (PHS), which was designed leveraging genetic counseling expertise as one means to address this need. This 9-week course, piloted in Fall 2021 at UC San Diego, afforded students the opportunity to build technical skills and competencies in clinical genomics while identifying, addressing, and engaging with pervasive health disparities in genomics. Interactive exercises focused students' learning on strategies for empathic and compassionate patient interactions while supporting the application of concepts and knowledge to future practice. Upon completion of the course, participants reported increases in confidence related to skills required for clinical genomics practice. Drawing on learnings from this pilot implementation, recommendations for refining the program include deepening pedagogical engagement with ethical issues, expanding the offering to trainees across health professions, including pharmacy students, and incorporating an optional experiential learning component. Educational offerings, like PHS, that are designed with the input of genetic counseling expertise may ease pressures on the genetic counseling profession by building a more genomic-literate healthcare workforce that can better support efforts to expand access for patients.

KEYWORDS
communication, diversity, education, empathy and compassion, genetic literacy, genetic testing, health equity, precision health

Taylor J. Berninger and Ramya M. Rajagopalan should be considered joint first author.

1 | BACKGROUND AND SIGNIFICANCE: GENOMICS TRAINING NEEDS FOR MEDICAL TRAINEES

The past decade has seen rapid growth in genomics and its integration across healthcare, as new genetic health services and products have emerged in clinical, research, and consumer domains. Historically, the majority of genetic healthcare services have been provided by medical geneticists and genetic counselors, often embedded within specialty areas of clinical care such as reproductive medicine, oncology, and pediatrics (Campion et al., 2019). However, due to advances in accessibility, affordability, and clinic utility, as well as commercial and direct-to-consumer (DTC) genetic testing external to healthcare institutions, primary care practitioners are increasingly called upon to field inquiries related to genetic risks and test results (Massart et al., 2022; Ramos & Weissman, 2018). Unfortunately, undergraduate medical education continues to lag behind these developments, with a significant proportion of medical students reporting that they feel insufficiently trained in applying genetics concepts learned in core curricula to clinical practice (Association of American Medical Colleges (AAMC), 2021; Kapur et al., 2023). In a recent study, over 35% of physicians surveyed reported having encountered at least one patient who shared DTC genetic test results with them within the past year (Jonas et al., 2019), but physician trainees continue to report feeling poorly prepared to provide effective clinical consultation or interpretation of genetic test results (Eden et al., 2016). At the same time, demand for these services is likely to grow due to policy developments that have drawn attention to the need for widely available access to genetic testing and counseling, such as the White House's recent revitalization of the Cancer Moonshot Initiative (White House, 2022). As genomics becomes a more routine component of clinical practice across specialties, there is a critical need for training opportunities that strengthen genomics literacy among non-genetics health professionals, beginning at the earliest stages of education and training (Hyland et al., 2019; Myers & Bloss, 2020; Rubanovich et al., 2018; Wilcox et al., 2018).

What is known about this topic?

At most medical schools in the United States, genetics education within the undergraduate medical curriculum primarily focuses on reinforcing basic genetics knowledge, versus practice-based competencies in genomics. There continues to be a gap and an urgent need for training in clinical genomics concepts and applications of genomic information in primary and specialty care contexts.

What this paper adds to this topic?

This paper describes the implementation and evaluation of a pilot genomics training program for undergraduate medical students. The program utilizes genetic counseling expertise to emphasize clinical skills in genomics through interactive coursework and a focus on tools and strategies for bringing equity and compassion to the forefront of precision health care.

2 | COURSE DESIGN: THE PRECISION HEALTH SCHOLARS IMMERSIVE LEARNING EXPERIENCE

We sought to address this need by designing, implementing, and evaluating an elective course in clinical genomics and precision health for pre-clinical medical students at UC San Diego School of Medicine. Course design leveraged our team's unique combination of expertise in research and teaching on genomics and health disparities, genetic counseling education, and clinical practice. In particular, leveraging a genetic counselor's experience with genomic healthcare in clinical team settings helped to frame the depth and scope of training material relevant to this learning audience. Building on this expertise, the course piloted an innovative approach to training students in skills and competencies for integrating genomics into their future practice. Course content explicitly emphasized empathy and compassion for patients and their backgrounds, as an important element of high-quality clinical care throughout the genetic testing process. This focus was motivated by our team's expertise and the goal of the Center for Empathy and Technology to promote compassionate engagement with emerging technologies, while centering issues of justice, equity, and diversity.

Course content was calibrated with the input of UC San Diego medical students obtained through two informal listening sessions. Medical students who had previously expressed interest in providing feedback on the design of an elective genomics education program were invited to participate. Three first-year students and one third-year student shared their views and offered input on topics related to pre-clinical genetics training, including genetics content integrated within the core curriculum, perceived gaps or needs in pre-clinical genetics training, and desired content. They also provided valuable feedback on possible course directions proposed by our team to help define course content and logistics. Overall, participants in these sessions highlighted perceived limitations of genomics training in the core curriculum and were enthusiastic about expanded educational opportunities in genetics and genomics. When presented with options that would deepen training in empathy and compassion, enhance understanding of patient perspectives, and contextualize genomic education within the tenets of justice, equity, diversity, and inclusion (JEDI), students responded favorably.

Findings from the listening sessions informed the learning objectives and design of a two-credit, 9-week independent study elective course called the Sanford Precision Health Scholars

Immersive Learning Experience (PHS), piloted at UC San Diego School of Medicine in Fall 2021. Course topics were oriented thematically and sequentially to mirror the phases of the genetic testing process and guide learners through consideration of the patient experience at each phase (Figure 1). Topics focused on increasing student familiarity with genetic testing platforms and clinical genomics workflows, considerations for test selection, the fundamentals of variant analysis and classification, and interpretation of sample genetic test reports from commercial and clinical laboratories. Content delivery included didactic instruction, as well as panel discussions and presentations from patient advocates, clinical genetics experts, and regional leaders in the genomics field. Teaching also leveraged a variety of pedagogical techniques, including guided discussion, case analysis and team-based learning exercises, role-play scenarios, supplemental reading, and optional reflective journaling. Within these activities, we embedded opportunities for skill development and practice delivering empathetic and compassionate care, for example facilitating conversations with patients around personal circumstances and the value of genetic testing, patient-centered analysis and interpretation of test reports, and incorporating best practices for compassionate communication of results through role-play scenarios. Case-based problem-solving exercises emphasized key learning objectives anchored in empathy and compassion, which included (1) applying a health equity lens to variant analysis and interpretation of genetic test reports from clinical and DTC services; (2) recognizing JEDI challenges in utilization of genetic testing for diverse and underserved communities historically excluded or underrepresented in medicine; and (3) identifying strategies for culturally respectful, transparent, and compassionate communication of genomic information for diverse patients and families. Session-specific learning objectives, content, and activities are listed in Table 1.

3 | COURSE IMPLEMENTATION AND EVALUATION

An announcement advertising the PHS pilot offering resulted in over 30 medical students expressing interest in receiving updates and further information about the course. Twelve students enrolled, all in their first (M1) or second (M2) year of medical training at UC San Diego; of these, five M2 students completed the course. Our informal survey of those who did not enroll or complete the course despite initial interest revealed that scheduling conflicts with competing learning opportunities, such as in-clinic shadowing experiences, were a primary reason for attrition. As we describe below, this feedback has been vital for planning future iterations of the course.

The course was evaluated through a pre-course questionnaire assessing student motivations and expectations, individual session evaluations as the course progressed, a final course evaluation, and a post-course survey measuring self-reported improvement in knowledge using open-ended and Likert-scale items. Survey items were inspired by educational course evaluations rather than published survey designs or validated measures. Post-course survey responses showed that participants reported multiple competencies gained through the course (Figure 2). Specifically, all participants reported an increase in confidence related to skills required for clinical genomics practice and in their understanding of health equity challenges in clinical genomics. Most also reported increases in their preparedness to practice empathy and compassion during patient encounters in genomic medicine. In terms of student satisfaction, all students shared feedback expressing significant enthusiasm about the course content and their learning experience. Student responses to three key questions collected through course evaluation (Table 2) suggested that the pilot offering of PHS was successful in meeting the course objectives, demonstrating promise as a means of increasing clinical genomics knowledge and skills among medical students,

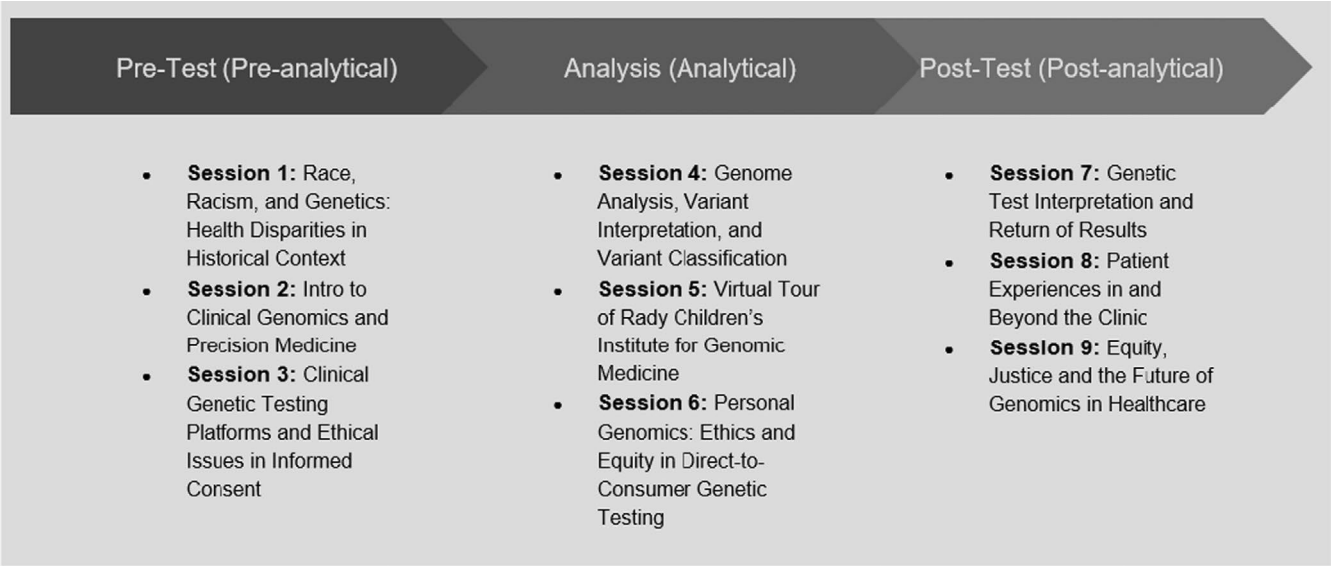

FIGURE 1 Sanford Precision Health Scholars Pilot Implementation Session Topics. Topics were sequentially ordered to mirror issues and challenges that emerge for patients and healthcare practitioners at successive phases of a patient's genetic testing journey.

TABLE 1 PHS session learning objectives and session content/activities.

Session topic	Learning objectives	Content/activities
Session 1: Race, Racism, and Genetics: Health Disparities in Historical Context	• Contextualize three main challenges to health equity that precision health needs to address • Explore the ethically fraught history of genetics and race, including the role that the science of heredity has played in inflicting trauma • Recognize how the flawed assumption that racial health disparities result from biological deficiencies can perpetuate disparities	• Didactic lecture • In-class Exercise: Henrietta Lacks Case Study • Optional Reflective Journaling
Session 2: Intro to Clinical Genomics and Precision Medicine	• Differentiate genetics from genomics • Review core concepts of Mendelian genetics • Explore the complexity of clinical genomics and current utilization in healthcare	• Kahoot Quiz Game: Basics of Genetics and Genomics • Didactic Lecture • Case Discussion: Interpretation of Tay-Sachs disease carrier screening in individuals with non-Jewish, non-European ethnic background • Optional Reflective Journaling
Session 3: Clinical Genetic Testing Platforms and Ethical Issues in Informed Consent	• Review genetic testing techniques and utility in clinical practice • Discuss possible genetic test results and facilitate anticipatory guidance for patients • Delineate essential elements of the informed consent process • Reflect on the impact of potential genetic test results on patients and families	• Didactic Lecture • Breakout Activity: Decoding informed consent documents and making meaning for patients • Case Discussions: Key elements of informed consent • Optional Reflective Journaling
Session 4: Genome Analysis, Variant Interpretation, and Variant Classification	• Delineate the essential steps in the variant curation process • Unpack the challenges and limitations of using a singular reference sequence • Identify tools and guidelines for supporting variant curation and classification	• Didactic Lecture • In-class Exercise: Curate a Variant • Optional Reflective Journaling
Session 5: Virtual Tour of Rady Children's Institute for Genomic Medicine	• Explore the steps for processing and assaying a sample via next-generation sequencing • Outline the process of ordering a clinical genetic test • Utilize the NIH Genetic Test Registry to identify appropriate testing to offer patients	• Virtual Lab Tour • Q&A with Lab Genetic Counselor • In-Class Exercise: Navigating Genetic Test Menus and using the NIH Genetic Test Registry • Optional Reflective Journaling
Session 6: Personal Genomics: Ethics and Equity in Direct-to-Consumer Genetic Testing	• Compare and contrast clinical genetic tests with Direct-to-Consumer (DTC) test offerings • Discuss the implications of DTC genetic testing for individuals and healthcare providers • Identify issues of ethics and equity in DTC genetic testing for health and ancestry	• Didactic Lecture • In-Class Exercise: DTC Website Investigation • Case Discussion: Provider interpretation and facilitating discussion of DTC test report • Optional Reflective Journaling
Session 7: Genetic Test Interpretation and Return of Results	• Define the essential elements of a genetic test report and the implications for clinical interpretation • Analyze result classifications and their impact on patient risk assessment and management recommendations • Discuss strategies and best practices for disclosing results to patients • Identify appropriate post-test support resources for patients and families	• Didactic Lecture • Case Discussion: Sample report analysis • In-class Exercise: Return of Results role play • Optional Reflective Journaling
Session 8: Patient Experiences in and Beyond the Clinic	• Reflect on patient and family experiences with genetic testing, diagnosis, and healthcare implications • Highlight the importance of clinician empathy and compassion in patient care and advocacy • Discuss the role of clinicians in patient advocacy and equity/access efforts	• Guest Speakers: Patient Presentation • Fireside Chat: Discussion with speakers • Optional Reflective Journaling
Session 9: Equity, Justice, and the Future of Genomics in Healthcare	• Brainstorm strategies for increasing access and equitable utility of clinical genomics services and technology • Explore the current utility of integrating precision medicine and genomics into routine healthcare settings • Envision the landscape of precision medicine and genomics in future healthcare practice	• Guest Speakers: Panel Discussion • Optional Reflective Journaling

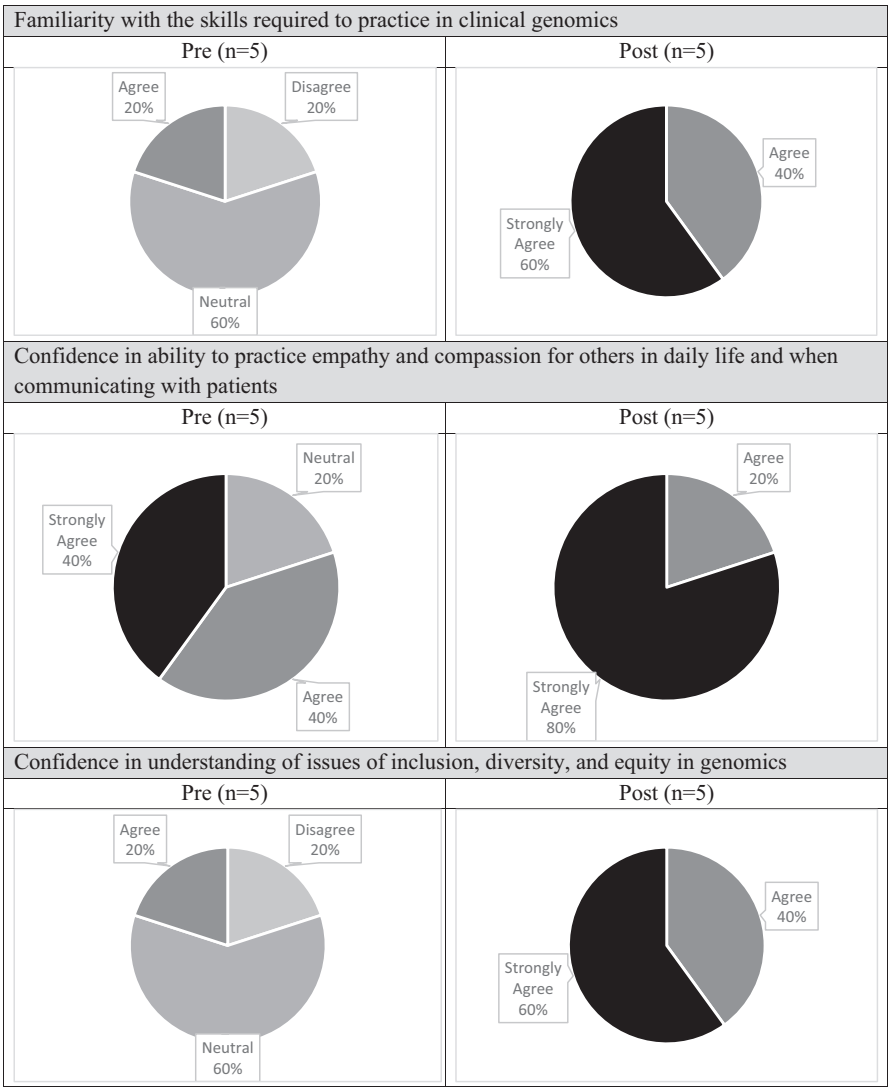

FIGURE 2 Pre- and Post-Course Evaluation Responses from PHS Pilot Participants.

while building recognition of and attention to the importance of health equity, empathy, and compassion in clinical genomics practice.

4 | DISCUSSION AND FUTURE DIRECTIONS

As a proof of concept, and for the small number of students who completed the course, the pilot iteration of the PHS program appeared effective in addressing gaps in genomics education and stimulating student interest in clinical genomics and its relevance to future professional practice. At the same time, one of the major takeaways from our experience is that while there is a need for training opportunities in clinical genomics for medical students, pre-clinical curricula and medical student schedules are already full, presenting ongoing challenges to students' ability to pursue multiple training interests, especially when faced with a wide variety of elective choices. This suggests that to be competitive with other elective offerings, genomics content should be delivered in innovative ways that are mindful of constraints on students' time.

We take additional learnings from this pilot implementation that we will leverage in guiding refinements for future offerings. First, we aim to be responsive to students who emphasized their interest in expanding and deepening course content focused on engagement with ethical issues in genomics, including more opportunities for group discussion of emerging topics and debates in genetics. Second, when presented with the idea of integrating an optional personal genetic testing experience into the course curriculum, students in both the pre-course listening sessions and those who enrolled in the course responded positively. This suggests that experiential learning opportunities might serve as a novel modality through which to spur student interest and retention. This approach could advance several aims, including cultivating understanding and empathy for the patient experience, strengthening skills for compassionate interaction and engagement with patients, and providing an additional learning methodology for reinforcing concepts related to genomics education. While mock reports and exercises can be utilized to simulate the genetic testing experience (and should be made available as an option to provide a comparable learning experience for students

TABLE 2 Students' assessment of course content and learning experience.

Survey question	Feedback offered through course evaluation ($n = 5$)	Relevance to course objectives
What will you remember or take away from this course?	"This was one of the best (if not THE best) electives I've taken at UCSD SOM. I found the topics that we learned to be super clinically relevant. The class also afforded me the opportunity to delve more deeply into social factors involved in understanding genetics and genetic testing." [Participant 1] "The course deepened my knowledge of the disparities in utility of genetic results and many of the shortcomings of current methods. It also taught me how to handle patients who bring genetic testing results to me with questions, and will make me much more comfortable discussing genetic testing and results with patients in the future." [Participant 2] "I have learned how to better discuss genetic counseling options with patients and how to navigate the genetics system better." [Participant 3] "I will remember to partner with my entire healthcare team and reach out for help when I am uncertain about the fast-paced changes in genetic testing and precision med." [Participant 4] "I will remember that many genetic tests are only useful for people of certain ancestry groups, and to look at the fine print when making decisions." [Participant 5]	Responses suggest that the course fostered a deeper understanding of clinical genetic services and referral pipelines, strategies for empathetic and compassionate patient interactions, and awareness of health equity concerns and limitations related to current genomics methods
Please list aspects of the course you particularly enjoyed	"Discussion was really good! Class felt less like a boring monologue/lecture and more like a constant conversation – hearing from experts in the field and learning about what their [sic] day to day looks like (and how we can work better with them in the future as physicians!) – knowledge of cutting-edge technology and new advances in the field of genetics" [Participant 1] "I really enjoyed the conversations that came out during the classes that were prompted by the topics of discussion that were being lectured on that day. We really got into complex ethical issues and I walked away with brand new perspectives on many of these issues thanks to the engaging conversations we had." [Participant 2] "I enjoyed the practical exercises we did to apply what we learned, and the outside speakers." [Participant 3] "I enjoyed examining everything through a DEI lens. I think our conversations that sprung from the lectures were very thought provoking and engaging" [Participant 4] "I liked the organic, intimate discussions. These often yielded the most interesting points." [Participant 5]	Responses suggest that the interactive learning modes built into the course helped solidify knowledge acquisition around clinical genetic services delivery, issues of equity and inclusion in genomics, and technology underlying the latest genomic advances
Please list particular skills and knowledge you learned in this course	"I learned SO much in this class! This class delved into the nitty gritty of genetic testing and counseling, but also taught me a lot about how to talk to patients about testing results, be cognizant of social factors playing into genetic testing/test design, etc. things I wasn't learning in my preclinical more science-focused classes. I feel like this class made me a better scientific communicator." [Participant 1] "How to evaluate genetic tests, how to identify deficiencies in the useful clinical information that certain genetic tests provide for minority groups, how to compare available tests and how to select the most appropriate test for a given clinical situation, how to think critically about the results of these tests and how to put results into context given a clinical picture or a life situation. I also found it very useful to go through the exercise of sitting with mock patients and giving results of genetic tests." [Participant 2] "Talking to patients about genetic counseling options, the process of genetic sequencing, how labs sequence and interpret the data from sequencing and deliver it as physician recommendations." [Participant 3] "Disclosure of genetic testing results. Critically analyzing genetic reports" [Participant 4] "I learned what to consider when ordering a genetic test and how to better counsel patients on genetic info." [Participant 5]	Responses suggest that concrete skills were acquired, including strategies for successfully working with clinical care teams and patients to compare, evaluate, select, and integrate genetic and genomic testing options to advance personalized patient care, skills for socially aware and compassionate patient communication of test options and results, and critical assessment and management of health equity concerns in the interpretation and communication of results

who do not elect personal genetic testing), prior studies have shown that integrating genetic testing opportunities into student coursework enhances appreciation for the patient experience and increases knowledge surrounding applications of genomic technologies in patient care, without significant harms to learners (Knoell et al., 2009; Linderman et al., 2018; Salari et al., 2013; Vernez et al., 2013).

However, as these prior studies have emphasized, several ethical considerations need to be addressed in order to safely implement optional personal genetic testing within the context of an elective course. These include selecting a testing service whose products afford high confidence in the accuracy and clinical utility of the results provided; confirming that the service has robust and clearly communicated genomic data security policies; ensuring autonomous and private decision-making around the pursuit of testing; and ensuring the availability and accessibility of post-test counseling support and resources. Offering students opt-in access to clinical-grade genetic testing through a consumer-initiated service is one potential option for such a course. Participants who opt-in would engage with the service and their test results privately and independently of instructional personnel. Participants would also have access to complimentary genetic counseling services in the event of an actionable finding and could engage with data and reports that are more reflective of genetic tests they are likely to encounter within healthcare systems in their future professional capacity. Financing for such a test experience would need to be structured to ensure all students have equal access to participate if they desire. Supplemental exercises to help students connect with the patient experience regardless of whether they opted in or out of personal genetic testing would be needed in order to ensure that all learners have opportunities to sharpen their skills related to empathy and compassion.

Overall, student feedback illuminates promising directions for reimagining course content and delivery to expand the reach of future offerings. We are in the process of leveraging these insights into the design of a new workshop format that builds on the pilot PHS course, but deemphasizes didactics, prioritizes discussion and active learning in a minimum of in-person sessions, focuses content delivery through a series of asynchronous and flexible learning modules, and offers an optional personal genetic testing experience that is thoughtfully integrated to address potential ethical concerns. This redesign will be structured to better complement students' already full schedules and address the bandwidth limitations that pose a key barrier to medical students' participation in such elective offerings.

5 | LIMITATIONS

Evaluation of the pilot offering we describe is limited by the small number of students from whom data could be collected. Future offerings could incorporate validated measures in pre- and post-course surveys to assess course impacts. Since course content was tailored to the prior pre-clinical genetics training and needs of medical students at UC San Diego, implementation of similar content at other institutions would likely benefit from adapting or modifying course content to align with the specifics of existing genetics training within their programs. Importantly, articulating the value of structured learning around JEDI, empathy, and compassion in clinical genomics is likely to be most effectively accomplished with reference to an institution's unique curriculum and the professional development goals and expectations of its trainees.

6 | GENOMIC LITERACY FOR THE HEALTHCARE WORKFORCE

Our experience with PHS suggests that interactive learning experiences offered through the lens of health equity, empathy, and compassion may be a valuable approach for developing student competencies and addressing the genomic knowledge gap for clinician trainees. In moving beyond primarily didactic modes of student engagement and the more historically static content typical of some pre-clinical genetics curricula, innovations implemented in PHS piqued student interest and were experienced as sharpening students' grasp of essential technical concepts while cultivating empathy, compassion, and connections to patient-facing practice.

For the genetic counseling profession, offerings like PHS have strong potential to aid in building a more genomic-literate healthcare workforce, which could ease pressures on the profession while also expanding genomic health care access for patients. Furthermore, genetic counselors bring important strengths to the design and implementation of such trainings. Specifically, these include skills in empathic communication of genetic information, a culture of interprofessional collaboration, and up-to-date knowledge of the current genomic healthcare landscape and patient needs. Enhancing opportunities and resources through which perspectives from genetic counseling can continue to inform genomics education for diverse health professional learners thus holds promise for bettering clinical genomic patient care.

AUTHOR CONTRIBUTIONS

Taylor Berninger and Ramya Rajagopalan confirm that they have full access to all the data reported in the manuscript and take responsibility for the integrity of the data and the accuracy of data analysis. All of the authors gave final approval of this version to be published and agreed to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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CONFLICT OF INTEREST STATEMENT

Taylor Berninger, Ramya M. Rajagopalan, and Cinnamon S. Bloss declare that they have no conflict of interest.

ETHICS STATEMENT

Human studies and Informed Consent: Course evaluation protocol was submitted to the UC San Diego Institutional Review Board and was deemed not to constitute human subjects research.

Animal Studies: No non-human animal studies were carried out by the authors for the article.

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