5-1-2018

The Undergraduate Training in Genomics (UTRIG) Initiative: Early & Active Training for Physicians in the Genomic Medicine Era

Rebecca L. Wilcox

Patricia V. Adem
New York Medical College

Ebrahim Afshinnekoo
New York Medical College

James B. Atkinson

Leah W. Burke

See next page for additional authors

Recommended Citation

Wilcox, R., Adem, P., Afshinnekoo, E., Atkinson, J., Burke, L., Cheung, H., Dasgupta, S., DeLaGarza, J., Joseph, L., LeGallo, R., Lew, M., Lockwood, C., & Haspel, R. (2018). The Undergraduate Training in Genomics (UTRIG) Initiative: Early & Active Training for Physicians in the Genomic Medicine Era. *Personalized Medicine, 15* (3), 199-208. https://doi.org/10.2217/pme-2017-0077

HHS Public Access

Author manuscript

Per Med. Author manuscript; available in PMC 2018 June 19.

Published in final edited form as:

Per Med. 2018 May 01; 15(3): 199–208. doi:10.2217/pme-2017-0077.

The Undergraduate Training in Genomics (UTRIG) Initiative: early & active training for physicians in the genomic medicine era

Rebecca L Wilcox[1], **Patricia V Adem**[2], **Ebrahim Afshinnekoo**[2], **James B Atkinson**[3], **Leah W Burke**[1], **Hoiwan Cheung**[4], **Shoumita Dasgupta**[5], **Julia DeLaGarza**[2], **Loren Joseph**[6], **Robin LeGallo**[7], **Madelyn Lew**[8], **Christina M Lockwood**[9], **Alice Meiss**[7], **Jennifer Norman**[10], **Priscilla Markwood**[10], **Hasan Rizvi**[11], **Kate P Shane-Carson**[12], **Mark E Sobel**[13], **Eric Suarez**[14], **Laura J Tafe**[4], **Jason Wang**[15], and **Richard L Haspel**[6,*]

[1]Department of Pathology and Laboratory Medicine and Department of Pediatrics, University of Vermont Medical Center, Larner College of Medicine, University of Vermont, Burlington, VT, 05401, USA

[2]Department of Pathology, New York Medical College School of Medicine, Valhalla, NY, 10595, USA

[3]Department of Pathology, Microbiology and Immunology, Vanderbilt University School of Medicine, Nashville, TN, 37232, USA

[4]Department of Pathology and Laboratory Medicine, Dartmouth-Hitchcock Medical Center, Lebanon, NH,/Geisel School of Medicine at Dartmouth, Hanover, NH, 03755, USA

[5]Department of Medicine, Biomedical Genetics Section, Boston University School of Medicine, Boston, MA, 02118, USA

[6]Department of Pathology, Beth Israel Deaconess Medical Center & Harvard Medical School, Boston, MA, 02115, USA

[7]Department of Pathology, University of Virginia School of Medicine, Charlottesville, VA, 22908, USA

[8]Department of Pathology, University of Michigan Medical School, Ann Arbor, MI, 48109, USA

[9]Department of Laboratory Medicine, University of Washington School of Medicine, Seattle, WA, 98195, USA

[10]Association of Pathology Chairs, Wilmington, DE, 19801, USA

[*]Author for correspondence: rhaspel@bidmc.harvard.edu.

The opinions expressed herein are those of the authors and are not necessarily representative of those of the Uniformed Services University (USU), the Department of Defense (DOD), or the United States Army, Navy, or Air Force.

Financial & competing interests disclosure

This work was supported by the National Institutes of Health (2R25CA168544).

The authors have no relevant affiliations or financial involvement with any organization or entity with a financial interest in or financial conflict with the subject matter or materials discussed in the manuscript. This includes employment, consultancies, honoraria, stock ownership or options, expert testimony, grants or patents received or pending, or royalties.

No writing assistance was utilized in the production of this manuscript.

[11]Institue of Health Sciences Education, Barts and the London School of Medicine and Dentistry, Queen Mary, University of London, London, E1 2AD, UK

[12]Division of Human Genetics, Department of Internal Medicine, Ohio State University Comprehensive Cancer Center, Columbus, OH, 43210, USA

[13]American Society for Investigative Pathology, Rockville, MD, 20852, USA

[14]Pathology Department, Uniformed Services University, Bethesda, MD, 20814, USA

[15]Department of Pathology, UT Southwestern Medical Center, Dallas, TX, 75390, USA

Abstract

Genomic medicine is transforming patient care. However, the speed of development has left a knowledge gap between discovery and effective implementation into clinical practice. Since 2010, the Training Residents in Genomics (TRIG) Working Group has found success in building a rigorous genomics curriculum with implementation tools aimed at pathology residents in postgraduate training years 1–4. Based on the TRIG model, the interprofessional Undergraduate Training in Genomics (UTRIG) Working Group was formed. Under the aegis of the Undergraduate Medical Educators Section of the Association of Pathology Chairs and representation from nine additional professional societies, UTRIG's collaborative goal is building medical student genomic literacy through development of a ready-to-use genomics curriculum. Key elements to the UTRIG curriculum are expert consensus-driven objectives, active learning methods, rigorous assessment and integration.

Keywords

active learning; curricula; genetics; genomics; interprofessional; medical education; NCI; precision medicine

Precision medicine has great potential for improving patient care; however, the incredible speed upon which it is being developed has left a knowledge gap between discovery and effective implementation of those discoveries into clinical practice. The goal of the Undergraduate Training in Genomics (UTRIG) Working Group is, using a collaborative and interprofessional approach, the development of nationally available resources to assist medical school educators in closing this gap.

The genomic medicine era

With the completion of the human genome project, precision medicine has transformed medical practice. Oncology is a specialty that has been quick to embrace the genomic medicine approach with oncologists routinely ordering tumor sequencing assays that analyze multiple genes in order to identify options for personalized treatment. In a 2014 study of 1007 patients with lung cancer, the use of a ten-gene panel of oncogenic drivers allowed 28% to be treated with targeted therapy. These patients survived a median of 3.5 years compared with 2.4 years for those who did not receive targeted treatment [1]. In a 2015 study of 102 children with refractory cancer, whole exome and transcriptome analysis led to

treatment modification in 14 patients with nine having ongoing partial or complete clinical remissions [2]. This emerging approach in oncology calls into question the current use of diagnostic classification systems of tumors. For example, in a patient with an unknown primary tumor presenting in the liver, the genetic profile of the tumor and the potential identification of precision medicine chemotherapeutic targets may be clinically more important than the primary location of the tumor. As stated by Dr Douglas Lowy and Dr Francis Collins in a *New England Journal of Medicine* perspective regarding the United States Cancer Moonshot Task Force, *"cancer is a disease of the genome"* [3].

The potential for personalized treatment extends to many fields in medicine. Pharmacogenomics allows physicians to customize treatment with medications including antiplatelet and antiviral agents. The need and potential for these tailored treatments is clear. A 2001 study addressing the clinical application of pharmacogenetics showed striking percentages of patients for whom drugs are ineffective ranging from 48% (migraine pharmacology) to 75% (oncology pharmacology) [4].

Genomic testing also improves diagnostics. Next-generation sequencing based assays allows for preconception carrier screening for over 100 genetic diseases. In infectious disease, genomic sequencing techniques can be important diagnostic tools when traditional methods have failed. In the case of a child with chronic meningitis, next-generation sequencing of cerebrospinal fluid led to the diagnosis of leptospirosis when culture had been negative [5]. Genomic testing has also proven clinically useful in the setting of rare diseases. In the case of a child with intractable inflammatory bowel disease, whole-exome sequencing led to the identification of a variant causing hemophagocytic lymphohistiocytosis (HLH). The patient received a bone marrow transplant to treat HLH that also cured his inflammatory bowel disease [6]. In a study of 250 patients with rare diseases, 62 had the molecular cause determined through whole-exome analysis. Demonstrating the rapid acquisition of genomic data, approximately 25% of the diagnoses were based on disease-gene discoveries made within the past 2 years prior to the study [7].

Given the clinical importance of genomic testing in clinical practice, it is not surprising that funding is being directed for the purposes of improving genomic data sharing and developing novel genomic assays. The Precision Medicine Initiative announced by President Obama in 2015 demonstrated further recognition of the importance of genomics-based oncology practice with $70 million earmarked for detection of genetic cancer drivers [8]. Consequently, there has been a surge in precision-driven initiatives and centers to research and implement these ground-breaking approaches to patient care.

Knowledge gap

Over the last two decades, research elucidating the human genome and its potential application has grown exponentially. However, the speed at which this has occurred has left a wide knowledge gap between discovery and implementation of those discoveries into clinical practice. As genomics enters mainstream clinical practice, physicians without specialized training in genetics/genomics potentially face uncertainty in ordering and interpreting genomic testing [9–13] and therefore leading to both under- and overutilization of genomics-based diagnostic tests. A number of studies have shown that genetic counselor

screening of molecular genetic testing orders led to revision of a remarkable 8–26% of orders [14,15]. In many cases, the tests were cancelled due to inappropriate ordering caused by a lack of physician comprehension of genetic testing. The cancellation of these tests prevented a waste of resources as well as potential patient distress over genetic variants of uncertain significance.

Physicians admit to this knowledge gap. In a recent survey of internists at two academic medical centers, 74% self-rated their genetics knowledge as very/somewhat poor and approximately 80% indicated a need for additional training. In spite of their acknowledged lack of training, 65% of these physicians had counseled a patient on a genetic issue and 44% had ordered a genetic test in the previous 6 months [16]. Similar results have been reported for psychiatrists and neurologists. In other studies, only approximately 15% of primary care physicians feel comfortable ordering genomic tests or explaining them to their patients [17,18]. In regard to pharmacogenomics, a nationwide survey of US physicians in 2012 explored awareness, utilization and competency in this area. While 98% of the 10,000 responding physicians agreed that genetic variations may influence drug response, only 10% felt adequately informed about pharmacogenomics testing [19]. In another study of 401 family doctors, 55% reported they had no knowledge of the Genetic Information and Non-Discrimination Act, which impacts potential discrimination in insurance and employment for patients undergoing genetic testing [20]. Although understanding the nuances of genetic and genomic testing in terms of clinical and personal issues is critical to the patients, it cannot be addressed by referral to specialists alone as there is a shortage of genetics specialists in the healthcare workforce [21]. Therefore, improving the genomic literacy of providers across specialties is a critical goal for effective implementation of approaches in genomic medicine.

Efforts to improve physician genomics knowledge

Recognizing the need to improve healthcare practitioner knowledge, there have been a number of genomics education initiatives. In 2013, the National Human Genome Research Institute assembled the Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC) from 23 professional societies, 15 institutes at the NIH and organizations interested in physician education. The mission of the ISCC is to develop and share best practices for improving the genomic literacy of providers. In April 2014, the ISCC published a white paper entitled ‘Framework for Development of Physician Competencies in Genomic Medicine’ in *Genetics in Medicine*. This framework defined five entrustable professional activities specifically in genomics, each with integrated core competencies based on the Accreditation Council of Graduate Medical Education’s six core competencies [22].

Specialists in genomic medicine include medical geneticists, genetic counselors and molecular genetic pathologists. For individuals enrolled in medical genetics residency programs, there is a comprehensive graduate medical curriculum covering the full range of topics pertaining to clinical genetics, from culturally competent communication to single gene and genome-wide genetic testing strategies [23]. Pathologists have a unique role in genomic testing. Not only do they assist in directing genetic testing laboratories but they play an important role in selecting tumor and other diseased tissue for analysis and

integrating molecular findings into histopathology reports. As such, it makes sense that pathologists are also leaders in genomics education. In 2015, a College of American Pathologists work group published a list of 32 high-priority genomics competencies for practicing pathologists [24]. In 2016, to provide a framework for pathology resident education in molecular genetics and genomics, the Association for Molecular Pathology released 'A Suggested Molecular Pathology Curriculum for Residents' [25]. This document provides specific objectives related to all aspects of molecular pathology including genomic technology and informatics. The objectives are divided into 'prerequisites', 'required' and 'recommended' categories. However, although these above documents are valuable resources for guiding content for resident education, they do not provide tools for implementation of a comprehensive curriculum.

The Training Residents in Genomics (TRIG) Working Group, formed in 2010 through the Pathology Residency Directors Section (PRODS) of the Association of Pathology Chairs (APC), has found success in building a rigorous genomic curriculum with tools for implementation aimed at Postgraduate Year (PGY) 1–4 resident level trainees [26]. Recognizing that education necessitates a collaborative approach, the working group has representatives from several major pathology organizations, the National Society of Genetic Counselors and the American College of Medical Genetics and Genomics. Developed with the support of an R25 grant from the National Cancer Institute, the curriculum includes four exercises following a breast cancer patient through different levels of genetic testing: single-gene testing; prognostic gene panels; cancer gene panels; and whole-exome sequencing. Based on this curriculum, the group has held over 20 genomics workshops for residents and practicing physicians using a team-based learning (TBL) approach. The workshops have been highly reviewed with 97% of participants stating they would recommend the session to their colleagues. As one of the first examples of TBL at a national meeting, the workshop design process and assessment have been published as an educational innovation [27].

The TRIG group has also developed a train-the-trainer handbook and toolkit designed to help others implement local teaching sessions [28]. The handbook contains workshop questions and answers, detailed information on teaching using a variation of the TBL format and a preparation checklist with tips on implementation. The toolkit contains all of the necessary handouts and PowerPoint lectures. To ensure the best possible product, all materials underwent outside peer-review by a group of pathologists and genetic counselors. Online modules that translate the workshop experience to a virtual environment, including simulation-based learning of online genomics tools, have also been released on the TRIG website (http://pathologylearning.org/trig). Since the materials were made available in 2014, over 1250 individuals from 60 different countries have registered to download the material. Respondents to a 2016 downloader survey reported using at least some portion of the materials with 776 medical students, 214 residents, 76 fellows and 60 laboratory technologists. Similar evidence of utility was seen in a 2016 PRODS survey; of the 52 respondents (37% of US programs), 32% are using at least some component of the TRIG curriculum.

Demonstrating adaptability of the TRIG model, in 2017, through the ISCC, specialty-agnostic universal modules have been released based on the TRIG approach. With these

'plug and play' exercises, genes and diseases can be tailored to provide specialty-specific education. Workshops based on these modules have been held at the American Academy of Neurology, American Heart Association and American Academy of Ophthalmology annual meetings.

TBL, problem-based learning, patient simulation, flipped classroom activities and case discussions are all forms of active learning [29]. Active learning distinguishes itself from the traditional lecture by promoting student engagement in the learning process rather than passively receiving information [30]. Compared with the traditional lecture format of information delivery, active learning has been shown by Freeman *et al.* to significantly increase student performance in science, technology, engineering and mathematics (STEM) disciplines [31]. Their meta-analysis of 225 studies that reported data on examination scores or failure rates revealed that active learning in college STEM courses, when compared with traditional learning, increased examination performance (raising average grades by half a letter). They also found that failure rates under traditional lecturing were 55% higher than those documented with active learning. Based on their results, the authors called for the abandonment of traditional lectures in favor of active learning in STEM courses stating that *"if the experiments analyzed here had been conducted as randomized controlled trials of medical interventions, they may have been stopped for benefit… because the treatment being tested was clearly more beneficial."* In addition, there is growing evidence that active learning approaches can significantly improve learning for some subgroups of learners, especially women [32,33] and students from disadvantaged educational and socioeconomic backgrounds [34]. Given the success of TRIG and other existing data, active learning approaches should be particularly emphasized for genomics education.

Medical student training in genomics

Given the aforementioned knowledge gap in genomics among practicing physicians, it is not surprising that experts agree upon the importance of physician training in the effective practice of genomic medicine [21,22,35,36]. This consensus applies not only to practicing healthcare providers, but also to future physicians. Thus, introducing these concepts beginning in medical school can provide the foundation upon which a continuously expanding amount of information can be built. In addition, early exposure would allow medical students to consider a career related to genomics. Although within US and Canadian medical schools there is a trend toward increasing genetics and genomics education, a recent survey-based study found that 40% of genetics course directors still do not cover key emerging genomic medicine topics in their curriculum [37]. In addition, this study found that the overwhelming majority of taught genetics content is presented in preclinical coursework. This approach misses the opportunity to apply these concepts during the clinical phase of education where real patient cases demand an understanding of targeted therapies, the ability to appropriately order genomic tests and effectively explain test results to patients.

In a pilot study conducted at the Larner College of Medicine at the University of Vermont (VT, USA), a National Board of Medical Examiners (NBME)-customized assessment tool was designed and implemented to test students' performance in six distinct areas of genetics

and genomics to compare performance with national averages. This exam was complemented with a student self-assessment specifically addressing 25 integrated core competencies in genomic medicine selected for appropriate training level from the ISCC framework (Table 1) [22]. All class of 2017 students in their final month of the preclinical curriculum were invited to participate; 71 students (63% of the class) completed the exam and 66 completed the self-assessment. Analysis of exam performance and self-assessment revealed gaps within the curriculum including molecular techniques, mutations and cancer, online genetics resources, multifactorial conditions, microbial genomic testing and interpretation of genomic test results. The data from this pilot study served as the framework for the funded development of a genetics/genomics curriculum focused on active learning at the Larner College of Medicine.

The Association for Professors of Human and Medical Genetics (APHMG) has published a list of genetics competencies for medical students [38]. In addition, the Undergraduate Medical Educators Section (UMEDS) of the APC has developed and published the Pathology Competencies for Medical Education which includes the topic of genetic mechanisms (Disease Mechanisms and Processes Competency) and genomics (Diagnostic Medicine and Therapeutic Pathology Competency) [39]. Examples of primarily local medical student genomics curricula and teaching materials are also available on MedEdPORTAL.org and in the Genetics Education Resource Exchange (available to APHMG member institutions). There are, however, few rigorously vetted national resources to assist medical school course directors in teaching genomic medicine.

To assess the need for educational tools, in August 2016, a survey was conducted through the UMEDS of APC. Pathologists play a major role in medical student education and UMEDS members represent course directors, responsible for overseeing course content, from 115 American medical schools. Of the 32 respondents, 94% believed genomic medicine was an important curricular topic for medical students yet only 12 of 28 (42%) reported a local curriculum in genomic medicine. It is not surprising then that nearly all respondents (25 of 28, 89%), believed there was a need for nationally available educational tools to assist in teaching medical students genomic medicine. To fill this need, the UTRIG Working Group was established in 2016. The UTRIG Working Group is under the aegis of UMEDS and made up of pathology course directors, genetics experts and medical students. As with TRIG, UTRIG demonstrates a collaborative approach across specialties (Box 1).

Box 1

Undergraduate Training in Genomics cooperating organizations

- AACC, ACLPS, ACMG, AMP, APC, APHMG, ASCP, ASIP, Barts and the London School of Medicine and Dentistry, ICPI and NSGC

  AACC: American Association for Clinical Chemistry; ACLPS: Academy of Clinical Laboratory Physicians and Scientists; ACMG: American College of Medical Genetics and Genomics; AMP: Association for Molecular Pathology; APC: Association of Pathology Chairs; APHMG: Association of Professors of Human and Medical Genetics; ASCP: American Society for Clinical

Pathology; ASIP: American Society for Investigative Pathology; ICPI: Intersociety Council for Pathology Information; NSGC: National Society of Genetic Counselors.

UTRIG curriculum: key elements of development & implementation

Applying the TRIG model to undergraduate medical education will lead to increased faculty confidence in teaching genetics/genomics material as well as establishing uniform implementation across medical schools. The goal of UTRIG is to promote strong, standardized integration of genetics/genomics education into medical school curricula to prepare future physicians to practice in the age of precision medicine and empower them to teach those already in practice. The successful development of the UTRIG curriculum is based on four key elements: achieving expert consensus on goals and objectives; embracing active learning methods; rigorous assessment; and effective integration into existing curricula.

Achieving consensus

Although aligned by a common goal, the UTRIG Working Group is diverse in its areas of expertise which include laboratory medicine, surgical pathology, molecular pathology, genetics, informatics, genetic counseling, administration and education. By employing a consensus-building process, the input of this panel of experts can be maximally leveraged to support curricular goals and objectives. Given the vetting and implementation of the TRIG curriculum, the UTRIG Working Group used the TRIG exercises as a starting point. The TRIG handbook and toolkit were distributed to the UTRIG members along with a comprehensive survey. Members were asked to comment on the utility of specific TRIG components for medical school education including learning objectives, case descriptions, online genomics tools and questions. All 11 respondents agreed that the TRIG material could be adapted to medical students. Comments have been collated for distribution back to the working group with ongoing team discussion to develop a student curriculum.

Active learning

As the evidence of benefit builds, curricula across US medical schools are shifting away from traditional lectures toward active learning modalities [40–43]. Active learning, with its focus on interaction, is particularly suited to deep learning of complex genetics and genomic content including the ethical issues involved in genetic testing and the effective application of online genomics tools to clinical analyses. A study of the effects of adding short online lecture content on genetics to free up class time for in-class activities in a university biology course showed that students retained information better and scored higher on exams while faculty had more in-class time to interact with the students [44].

Although recently established medical schools have leaned toward lecture-free curricula for the reasons stated above, the adoption of active learning educational methods in more established schools has not been without barriers. Reported faculty barriers to adoption include lack of necessary class time, a high comfort level with traditional lectures,

insufficient time to develop active learning materials and lack of control of the classroom [45,46]. Active learning methods have been shown to increase engagement, higher order learning and learning outcomes compared with the traditional lecture; however, it also decreases the amount of information delivery. Increasing pressure to cover foundational science content in shorter periods of the medical curriculum, as well of a lack of faculty protected time needed to develop strong active learning material, can potentiate conflict between active learning methods and traditional curricular frameworks. Furthermore, the topic of genomic medicine is out of the comfort zone of many course directors. The solution to these challenge lie in building targeted, multidisciplinary online education modules along with faculty handbooks that outline teaching best practices for each active in-class session. Moreover, this model lends itself well to implementation in the clinical years, a segment of the medical curriculum that is in need of further exposure to genetics [37] and that must adapt to students being assigned to disparate clinical training sites. As with the TRIG curriculum, UTRIG plans to create a genomics curriculum library of ready-to-use learning tools and faculty manuals to convert barriers into welcome additions.

Assessment

The TRIG Working Group utilized surveys and exams to determine curriculum efficacy. The Resident In-Service Exam (RISE), taken by every PGY1–4 pathology resident in the USA, has also provided valuable data. Since 2013, survey and knowledge questions related to genomic medicine have been included in the RISE [47] and a significant increase in targeted resident training in genomics has been observed. Of note, from an initial 2010 PRODS survey suggesting only 30% of residency programs provided training in genomic medicine, the result has now climbed to approximately 80% based on response for the 2017 RISE. The RISE survey and knowledge questions created by the TRIG Working Group have provided valuable data on the degree and efficacy of resident training in genomic pathology on a national scale. There are few published examples of the use of an assessment tool with the scope of the RISE to study curricular improvement.

Similar to TRIG, the UTRIG Working Group also plans a rigorous assessment of the curriculum and educational tools. The RISE-FIRST is an exam taken by over 60% of the first-year pathology residents approximately 1 month into their training. UTRIG will use this tool to assess, albeit on a smaller scale than the RISE, medical school genomics education. Since 2015, survey questions have been included on the RISE-FIRST and the data indicate students have less comfort with genomics concepts when directly compared with other areas of medicine (e.g., interpreting glucose test results). Importantly, the performance of PGY1 pathology residents on this assessment is likely an overestimate of genomics literacy compared with medical students who chose other specialties that might have less focus on genetics and genomics. Other question responses suggest a need for increased interaction with genetic counselors and medical geneticists during medical school to help develop a better appreciation of the application of genetics to clinical care. Moreover, the genetics content on the NBME shelf examinations is a minor component of the overall examination, and genomics is not listed among the topics in the NBME content outline [48]. As such, students in specialties with reduced focus on genetics may enter residencies with diminished understanding of genetics and genomics principles after limited assessment of the topics on

licensing examinations and virtually no contact with the discipline in their clinical years. The RISE-FIRST data will serve as a baseline with continued assessment as the UTRIG curricular material becomes available. The Association of American Medical Colleges (AAMC) curriculum inventory content report may also be a mechanism to obtain baseline data in regard to whether and how schools currently report teaching of genomics and whether this changes with dissemination of UTRIG educational tools.

Integration

With the rise of precision medicine, clinicians trained in genetics and genomics will be best positioned to care for patients and strengthen physician–patient trust by making informed decisions in the utilization of technologies for personalized risk assessment, diagnosis, as well as treatment selection, dosing and monitoring. A pragmatic approach with clinical integration is critical to the success of establishing this level of genetics/genomics knowledge. Just as physicians do not need to fully understand the physics of magnetism to make use of an MRI, physicians need not have advanced and in-depth education in genomic sequencing to make effective clinical use of this technology. Integration of genomics into medical education may have the additive benefit of promoting interest in medical genetics or molecular pathology as a specialty; however, the overall goal should be focused on graduating genomic-literate physicians in all specialties. The UTRIG plan is not to introduce genomics as a standalone topic, but rather to integrate it with relevant topics in the preclinical years to highlight clinical relevance and link to specific patient scenarios in the clinical years (e.g., the use of next-generation sequencing in determining cancer treatment options or requiring students to produce a three generation family tree with a standardized patient while learning about breast cancer and founder mutations). Early exploration is key to dissipating fear of the material and the use of standardized patients and clinical skill exams are excellent means to foster the development of informed physicians and enhancing patient-care skills around genetics and genomics.

To best integrate the curriculum, there is also a need to build not only student but faculty comfort with genomics concepts. In the aforementioned survey of medical school course directors, although there was a strong movement toward integrated curricula, a high percentage of the course directors overseeing the genetics content were nongeneticists [37]. This gets to the very core of the knowledge gap challenge. When education leaders themselves do not feel genomically literate, the chasm expands. As with TRIG and planned for UTRIG, the solution lies in standardized, vetted education material with faculty manuals that close this knowledge gap. Therefore, parallel to the development of the UTRIG educational tools are the building of faculty manuals and a roadmap for implementation.

Conclusion

There is clearly a need for increased physician training in genomic medicine. There has been some success, particularly for pathology residents (through materials created by the TRIG Working Group as well as adaptation to some specialties outside of pathology) and for medical students (through materials collected and developed by the APHMG). Genomics education, however, remains an area that needs further attention in medical school curricula,

particularly in terms of genomic analysis and applied settings directly connected to clinical cases in order to lay the framework for this critical knowledge for all physicians. Building on TRIG and with continued funding through the National Cancer Institute, the UTRIG Working Group plans to take a similar approach to undergraduate medical education. Membership includes a diverse group of medical educators, genetics professionals and medical students. There is also a clear plan to obtain expert consensus on a set of active-learning-based teaching sessions with assessment and integration into existing medical student curricula. It is our goal to provide tools that can be integrated into medical curricula even by nonspecialists to increase the genomic literacy of our future physicians, a critical step to ensuring these powerful approaches continue to make inroads into patient care.

Future perspective

The TRIG/UTRIG Working Group model represents only one approach to closing the knowledge gap in genomics education. Additional tools are needed to help facilitate training not only of physicians but also nurses, physician assistants and others for which an understanding of genomics will improve patient care. It is hoped that funding organizations will also consider greater financial support for educating health professionals in genomic medicine. While projects such as the ‘Cancer Moonshot’ are critical in applying genomic medicine to the clinic, there will be difficulty with such translation without resources allocated to improving practitioner education. Furthermore, to truly encourage integration of genomics curricula into training programs, the Accreditation Council of Graduate Medical Education and Liaison Committee on Medical Education should consider developing genomics-specific requirements and competencies. As licensing and board certification exams drive learning, these should also include genomics-related questions. Lastly, as exams typically test knowledge acquisition, development of evaluation tools that directly measure skill-based objectives, such as effective provider–patient communication, are critical for determining if educational programs are having the desired effect.

References

Papers of special note have been highlighted as: • of interest; •• of considerable interest

1. Kris MG, Johnson BE, Berry LD, et al. Using multiplexed assays of oncogenic drivers in lung cancers to select targeted drugs. JAMA. 2014; 311(19):1998–2006. [PubMed: 24846037]
2. Mody RJ, Wu YM, Lonigro RJ, et al. Integrative clinical sequencing in the management of refractory or relapsed cancer in youth. JAMA. 2015; 314(9):913–925. [PubMed: 26325560]
3. Lowy DR, Collins FS. Aiming high: changing the trajectory for cancer. N Engl J Med. 2016; 374(20):1901–1904. [PubMed: 27043262]
4. Spear BB, Heath-Chiozzi M, Huff J. Clinical application of pharmacogenetics. Trends Mol Med. 2001; 7(5):201–204. [PubMed: 11325631]
5. Wilson MR, Naccache SN, Samayoa E, et al. Actionable diagnosis of neuroleptospirosis by next-generation sequencing. N Engl J Med. 2014; 370(25):2408–2417. [PubMed: 24896819]
6. Worthey EA, Mayer AN, Syverson GD, et al. Making a definitive diagnosis: successful clinical application of whole exome sequencing in a child with intractable inflammatory bowel disease. Genet Med. 2011; 13(3):255–262. [PubMed: 21173700]
7. Yang Y, Muzny DM, Reid JG, et al. Clinical whole-exome sequencing for the diagnosis of mendelian disorders. N Engl J Med. 2013; 369(16):1502–1511. [PubMed: 24088041]

8. National Cancer Institute. NCI and the Precision Medicine Initiative. www.cancer.gov/research/areas/treatment/pmi-oncology

9•. Varmus H. Getting ready for gene-based medicine. N Engl J Med. 2002; 347(19):1526–1527. Summarizes the beginning of the 'genomics knowledge gap' that occurred with the complete sequencing of the human genome and the emerging need for educating practicing physicians about the uses and shortcomings of genetic information. [PubMed: 12421898]

10. Houwink EJ, van Luijk SJ, Henneman L, et al. Genetic educational needs and the role of genetics in primary care: a focus group study with multiple perspectives. BMC Fam Pract. 2011; 12(5):1–9. [PubMed: 21223592]

11. Rinke ML, Mikat-Stevens N, Saul R, et al. Genetic services and attitudes in primary care pediatrics. Am J Med Genet Pt A. 2014; 164A(2):449–455.

12. Selkirk CG, Weissman SM, Anderson A, Hulick PJ. Physicians' preparedness for integration of genomic and pharmacogenetic testing into practice within a major healthcare system. Genet Test Mol Biomarkers. 2013; 17(3):219–225. [PubMed: 23390885]

13. Ginsburg GS. Genomic medicine: 'grand challenges' in the translation of genomics to human health. Eur J Hum Genet. 2008; 16(8):873–874. [PubMed: 18560443]

14. Kotzer KE, Riley JD, Conta JH, Anderson CM, Schahl KA, Goodenberger ML. Genetic testing utilization and the role of the laboratory genetic counselor. Clin Chim Acta. 2014; 427:193–195. [PubMed: 24084506]

15. Miller CE, Krautscheid P, Baldwin EE, et al. Genetic counselor review of genetic test orders in a reference laboratory reduces unnecessary testing. Am J Med Genet. 2014; 164A(5):1094–1101. [PubMed: 24665052]

16. Klitzman R, Chung W, Marder K, et al. Attitudes and practices among internists concerning genetic testing. J Genet Couns. 2013; 22(1):90–100. [PubMed: 22585186]

17. Haga SB, Burke W, Ginsburg GS, Mills R, Agans R. Primary care physicians' knowledge of and experience with pharmacogenetic testing. Clin Genet. 2012; 82(4):388–394. [PubMed: 22698141]

18. Musunuru K, Haspel RL. Improving genomic literacy among cardiovascular practitioners via a flipped-classroom workshop at a national meeting. Circ Cardiovasc Genet. 2016; 9(3):287–290. [PubMed: 27329653]

19. Stanek EJ, Sanders CL, Taber KA, et al. Adoption of pharmacogenomic testing by US physicians: results of a nationwide survey. Clin Pharmacol Ther. 2012; 91(3):450–458. [PubMed: 22278335]

20. Laedtke AL, O'Neill SM, Rubinstein WS, Vogel KJ. Family physicians' awareness and knowledge of the Genetic Information Non-Discrimination Act (GINA). J Genet Couns. 2012; 21(2):345–352. [PubMed: 21927977]

21. McGrath S, Ghersi D. Building towards precision medicine: empowering medical professionals for the next revolution. BMC Med Genomics. 2016; 9(23):1–6. [PubMed: 26729011]

22••. Korf BR, Berry AB, Limson M, et al. Framework for development of physician competencies in genomic medicine: report of the Competencies Working Group of the Inter-Society Coordinating Committee for Physician Education in Genomics. Genet Med. 2014; 16(11):804–809. The National Human Genome Research Institute convened the Inter-Society Coordinating Committee for Physician Education in Genomics (ISCC) to develop and share best practices in the use of genomics in medicine. This ISCC consensus paper provides a framework for development of genomics practice competencies for formulation of competencies for physicians in various medical disciplines and assessment of current practice or knowledge base. [PubMed: 24763287]

23. Korf BR. The medical genetics residency milestones. J Grad Med Educ. 2014; 6(Suppl 1):87–90. [PubMed: 24701269]

24. Laudadio J, McNeal JL, Boyd SD, et al. Design of a genomics curriculum: competencies for practicing pathologists. Arch Pathol Lab Med. 2015; 139(7):894–900. [PubMed: 26125429]

25. Aisner DL, Berry A, Dawson DB, Hayden RT, Joseph L, Hill CE. A suggested molecular pathology curriculum for residents: a report of the association for molecular pathology. J Mol Diagn. 2016; 18(2):153–162. [PubMed: 26857063]

26. Haspel RL, Atkinson JB, Barr FG, et al. TRIG on TRACK: educating pathology residents in genomic medicine. Per Med. 2012; 9(3):287–293. [PubMed: 29758790]

27. Haspel RL, Ali AM, Huang GC. Using a team-based learning approach at national meetings to teach residents genomic pathology. J Grad Med Educ. 2016; 8(1):80–84. [PubMed: 26913108]

28••. Training Residents in Genomics (TRIG) Working Group. Training Residents in Genomics (TRIG). http://pathologylearning.org/trigIn 2010, the Training Residents in Genomics (TRIG) Working Group was formed through the Pathology Residency Directors Section of the Association of Pathology Chairs. This website provides the Training Residents in Genomics (TRIG) Working Group Resident Genomic Pathology Workshop Instructor Handbook and Toolkit, materials and guidance needed to implement a structured and field-tested introductory resident genomic pathology curriculum, free of charge

29. Huggett, KN., Jeffries, WB. An Introduction to Medical Teaching. 2. Springer; Dordrecht, The Netherlands: 2014.

30. Bonwell, CC., Eison, JA. ASHE-ERIC Higher Education Report No. 1. The George Washington University; Washington DC, USA: 1991. Active learning: creating excitement in the classroom.

31•. Freeman S, Eddy SL, McDonough M, et al. Active learning increases student performance in science, engineering and mathematics. Proc Natl Acad Sci USA. 2014; 111:8410–8415. One of the foundational papers supporting the use of active learning in science, technology, engineering and mathematics-based education. [PubMed: 24821756]

32. Lorenzo M, Crouch CH, Mazur E. Reducing the gender gap in the physics classroom. Am J Phys. 2006; 74:118–122.

33. Gross D, Pietri ES, Anderson G, Moyano-Camihort K, Graham MJ. Increased preclass preparation underlies student outcome improvement in the flipped classroom. CBE Life Sci Educ. 2015; 14(4):ar36. [PubMed: 26396151]

34. Haak DC, HilleRisLambers J, Pitre E, Freeman S. Increased structure and active learning reduce the achievement gap in introductory biology. Science. 2011; 332:1213–1216. [PubMed: 21636776]

35. Manolio T, Ward R, Ginsburg GS. Clinical implementation of genomic medicine: the importance of global collaboration – editorial. Expert Rev Precis Med Drug Dev. 2016; 1(4):349–351.

36. Nelson EA, McGuire AL. The need for medical education reform: genomics and the changing nature of health information. Genome Med. 2010; 2(18):1–3. [PubMed: 20193046]

37. 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; 17(11):927–934. [PubMed: 25674779]

38. Hyland, KM., Dasgupta, S., Garber, KB., et al. APHMG medical school core curriculum in genetics. http://media.wix.com/ugd/3a7b877064376a9eb346cfa1b85bc2f137c48f.pdf

39. Knollmann-Ritschel BEC, Regula DP, Borowitz MJ, Conran R, Prystowsky MB. Pathology competencies for medical education and educational cases. Acad Pathol. 2017; 4:1–36.

40. Association of American Medical Colleges. Use of instructional methods by US and Canadian medical schools. www.aamc.org/initiatives/cir/456458/ci07.html

41. Association of American Medical Colleges. Medical schools reporting use of selected teaching formats in 2013–2014, by topic area. www.aamc.org/initiatives/cir/406470/06c.html

42. Prober CG, Health C. Lecture halls without lectures- a proposal for medical education. N Engl J Med. 2012; 366(18):1657–1659. [PubMed: 22551125]

43. Schwartzstein RM, Roberts DH. Saying goodbye to lectures in medical school- paradigm shift of passing fad? N Engl J Med. 2017; 377(7):605–607. [PubMed: 28813217]

44. Prunuske AJ, Batzli J, Howell E, Miller S. Using online lectures to make time for active learning. Genetics. 2012; 192(1):67–72. [PubMed: 22714412]

45. Miller CJ, Metz MJ. A comparison of professional-level faculty and student perceptions of active learning: its current use, effectiveness and barriers. Adv Physiol Educ. 2014; 38(3):246–252. [PubMed: 25179615]

46. Michael J. Faculty perceptions about barriers to active learning. Coll Teach. 2007; 55(2):42–47.

47. Haspel RL, Rinder HM, Frank KM, et al. The current state of resident training in genomic pathology: a comprehensive analysis using the resident in-service examination. Am J Clin Pathol. 2014; 142(4):445–451. [PubMed: 25239410]

48. National Board of Medical Educators (NBME). Exam content outline. www.nbme.org/Schools/Subject-Exams/Subjects/basicscibio.html

Executive summary

The genomic medicine era

- Precision medicine has great potential for improving patient care; however, the incredible speed upon which it is being developed has left a knowledge gap between discovery and effective implementation of those discoveries into clinical practice.
- Survey studies have shown that physicians openly admit this knowledge gap, self-rating their genomics knowledge base as low with acknowledgment of the need for additional training, despite the fact that ordering, interpreting and explaining genomic test results to their patients have entered mainstream medical practice.

Efforts to improve health practitioners' genomics knowledge

- Important initiatives to improve health practitioner genomics education include the Inter-Society Coordinating Committee for Physician Education in Genomics white paper entitled 'Framework for Development of Physician Competencies in Genomic Medicine', the College of American Pathologists Genomics Curriculum Work Group's 32 high-priority genomics 'Competencies for Practicing Pathologists', the Association for Molecular Pathologists 'A Suggested Molecular Pathology Curriculum for Residents' and the Association for Professors of Human and Medical Genetics 'Medical School Core Curriculum in Genetics'.
- The Training Residents in Genomics (TRIG) Working group, formed in 2010 through the Pathology Residency Directors Section of the Association of Pathology Chairs and supported by an R25 grant from the National Cancer Institute, has successfully built a rigorous genomics curriculum with tools for implementation aimed at resident level trainees.
- To close the knowledge gap it is necessary to start at the beginning. Building genomic literacy in our medical students is critical to not only preparing of future physicians to practice in the age of precision medicine but to empower them to teach those already in practice.
- The Undergraduate Training in Genomics (UTRIG) Working Group, organized in 2016 under the aegis of the Undergraduate Medical Educators Section of the Association of Pathology Chairs, is an interprofessional working group with a collaborative goal of building genomic literacy in medical students through the development of a standardized, ready-to-use genomics curriculum.

UTRIG curriculum: key elements of development & implementation

- Consensus: employing a consensus-building process, the input of a team of interprofessional experts can be maximally leveraged to support curricular goals and objectives.

- Active learning (AL): as the evidence of benefit builds, US medical schools are shifting away from traditional lectures toward AL. AL modalities are particularly suited to complex genetics/genomics content such as ethical issues and the effective application of online genomic tools. UTRIG plans to create a genomic curriculum library of learning tools and faculty manuals for ready-to-use AL sessions.
- Assessment: rigorous assessment of the curriculum and educational tools is essential to providing a standardized product and for continuous improvement. On a national level, the RISE-FIRST exam, taken by first year pathology residents, will allow assessment of medical school genomics education.
- Integration: a pragmatic approach to this curriculum with clinical integration is key to its success. Although there may emerge an additive benefit of promoting interest in a career in medical genetics or molecular pathology, the overall goal of this curriculum is graduating genomic-literate physicians in all specialties.

Table 1

Students were asked to indicate their level of confidence in their ability to do each of 25 selected ISCC-integrated core competencies in genomic medicine.

Competency	Average confidence[†]
Use online genetics-specific resource	2.25 ± 0.15
Use empirical risk figures to provide information for multifactorial conditions	2.37 ± 0.13
Explain the core strategies for genomic testing for microbial disease	2.47 ± 0.13
Explain reasons for false-positive and false-negative microbial genomics tests	2.50 ± 0.14
Explain analytic validity, clinical validity and clinical utility as they relate to genomic testing	2.50 ± 0.15
Describe the basic patterns of Mendelian inheritance	3.97 ± 0.11
Appreciate the importance and abundance of genetic diversity in humans	4.19 ± 0.10

[†]Lowest: 1 (Not at all confident); Highest: 5 (Extremely confident).

The five lowest and two highest scoring competencies of the 25 tested with average confidence level ± standard error of the mean (n = 62). ISCC: Inter-Society Coordinating Committee for Physician Education in Genomic.