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Promoting the Integration of Direct-to-Consumer Genetic Testing Results Into Clinical Practice

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PROMOTING THE INTEGRATION OF DIRECT-TO-CONSUMER GENETIC TESTING RESULTS INTO CLINICAL PRACTICE

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ABSTRACT

In recent years, individuals have become interested in what insights direct-to-consumer genetic testing (DTCGT) can offer about their health. These individuals are recommended to discuss their concerns with a healthcare professional, yet most primary care professionals do not have a genetics-focused practice. This dissertation comprises five chapters to describe the integration of DTCGT results into a patient visit. Chapter I discusses the expanding demand between patients with questions about their DTCGT results and the health professionals who have been tasked to interpret these results. Chapter II is a systematic literature review that identifies what health professionals think about results from non-clinical genetic testing companies and the reasons that they are reluctant to discuss DTCGT results with patients. The literature leads to two main barriers to acceptance of DTCGT: lack of genetic/genomic literacy and the doubtfulness of clinical utility. Healthcare professionals are also concerned about legal and ethical issues that have not been addressed with DTCGT. There may be more acceptance of DTCGT in clinical practice if clinical professional societies or regulatory commissions created guidelines to its use. Chapter III is composed of the results of a survey given to osteopathic medical students that asks about their current knowledge, attitudes, and perceptions of DTCGTs. The findings show that the students were uncomfortable with the idea of discussing DTCGT results with patients due to their perceived lack of knowledge of the subject matter. The majority of students were willing to learn more about DTCGT and how it could be integrated into patient care. This sentiment mirrored the literature on established healthcare professionals.

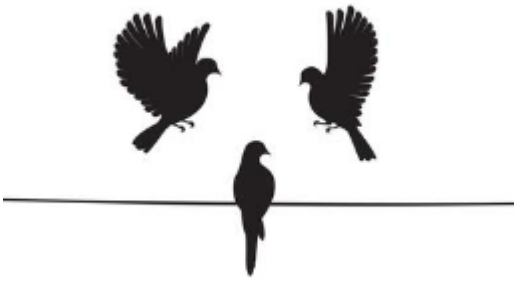
Chapter IV describes the creation of an online training module to prepare osteopathic

medical students for productive interactions with patients who have questions about their DTCGT results. This module was built to bring awareness to common clinical scenarios as well as give students applicable communication points using a virtual standardized patient. As assessed through a pre and post-survey, not only did the module enhance relevant vocabulary, but it also boosted confidence in handling patient interactions versus immediately referring patients to a genetic counselor.

Chapter V is a consolidation of the findings from Chapters II-IV. The systematic literature review and the online survey reveal that medical professionals with minimal genetic/genomic education have some trepidation about discussing DTCGT results with patients. The online training module shows that even a brief exercise can increase student awareness of how DTCGT can impact a patient's view on personal health goals. All three components (the systematic review, the survey, and the module) show that medical students are ready to learn how to integrate DTCGT results into clinical practice.

DEDICATION

This entire journey is dedicated to my three Ol' Birds: Essie Lou, Mary Lee, and Amanda and to my three Little Birds: Angela, Lillian, and Savannah. I hope I have made you all proud.



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CHAPTER ONE

INTRODUCTION

Background

Our story begins in 1996 when University Diagnostics in the United Kingdom first offered mail-order genetic testing services. In the United States, the Genetics and IVF Institute launched newspaper ads offering breast cancer testing in the United States.[1] These events marked the birth of a new chapter in what would be known as personalized medicine.

With the 21st century, new marketing techniques were introduced to expand the availability of these genetic testing services. Sciona, a nutrigenics company in the United Kingdom, started using their website to advertise and sell their genetic testing services in 2001.[1,2] By 2003, the human genome had been sequenced [3] and became a boon to more in-depth and varied medical research.[4] This research involved the identification of variations in a nucleotide at a specific position in the DNA sequence. These variations are known as single nucleotide polymorphisms (SNPs). When groups of these SNPs are inherited together or have a specific combination on the same chromosome, that is known as a haplotype. These haplotypes can be used to trace ancestry or map current and future health issues. The HapMap, a database of common haplotypes, was released to the public domain in 2004 and bolstered genome-wide association studies (GWAS).[5,6] GWAS studies have identified over 5000 diseases and traits since 2005,[4] but more samples are needed to refine the collected information.

That is where direct-to-consumer genetic tests (DTCGT) come into the picture. In 2007, companies like 23andMe, Navigenics, and Ancestry began marketing to the average person, piquing their curiosities about their genetic health history or familial connections. However, in 2010, the US Government Accountability office released a report that many of these DTCGT companies were

giving their customers unproven disease predictions by not testing for all genetic variants of certain diseases. This prompted the Food and Drug Administration (FDA) to demand that all DTCGT companies either stop testing or work with them to create guidelines and approved testing products.[2] The companies that worked to get their disease prediction tests approved by the FDA were allowed to advertise those tests to customers again. By 2017, 23andMe was the highest selling item on the Friday after Thanksgiving, known as Black Friday, in the United States.[7] This is the busiest day of the holiday shopping season and demonstrated that there was a huge public interest for personal genomic testing. The DTCGT companies all had websites that gave basic explanations about what each SNP variant meant, but for further explanation they advised seeing a personal healthcare professional.[8] This practice was done without taking into consideration that most general practitioners are non-genetic professionals and may not be prepared to discuss DTCGT results with a patient.[9] A 2016 study of 1026 DTCGT consumers showed that although 63% planned to share their results with a physician, only 27% did. Of those who did, 18% were not satisfied with their interaction. Some of the reasons for dissatisfaction were lack of knowledge, lack of interest, and skepticism from the primary care provider. Those who had a better experience sharing their test results noted their provider's ability to relate the results to the consumer's personalized care and inclusion of the results in medical records.[10] Many of these physicians started practicing well before the mapping of the genome and have done well without having to learn about or discuss DTCGT in depth therefore it is critical to promote education on the use of DTCGT for all providers and to begin early in the academic career. Therefore, this dissertation research was undertaken to address three primary aims – review of what is known about clinician knowledge and attitudes toward DTCGT, assessment of medical student knowledge, attitudes, and perception of the clinical utility of DTCGT, and the development of a training module that will reduce the barriers to communication between patient and provider.

Overview of Chapters

This dissertation contains five chapters describing the steps to creating an educational training module that will promote the facilitation of communication between DTCGT consumers and their healthcare professionals. Chapters II, and III are manuscripts that describe the knowledge, attitudes, and perceptions of practitioners and current medical students about the use of DTCGT results in clinical care. Chapter IV is the evaluation of a training module that integrates patients' DTCGT results questions into clinical care. Chapter V provides a summary of all findings and provides recommendations.

Chapter II, "Obstacles to Integration of Direct-to-Consumer Genetic Test Results Into Patient Care," aimed to uncover concerns that healthcare providers reported with integrating DTCGT results in clinical care. This was done through a systematic literature review that analyzed the statements given by current healthcare professionals who are concerned with the prospect of dealing with unsolicited genetic test results. The information found in this review is integral in seeing not only the gaps in genetic education within the medical community, but also gaps in communication that may arise between patient and provider.

Chapter III, "Osteopathic Medical Students' Attitudes About the Clinical Application of Direct-to-Consumer Genetic Test Results," focused on the gaps in current graduate medical education. Medical students at Edward Via College of Osteopathic Medicine were given a 15-question online survey about basic genetic terminology, performance expectations, and the perceived clinical utility of DTCGT results.

Chapter IV, "Navigating Direct-to-Consumer Genetic Testing: How to Approach Patients' Questions and Concerns," assessed the efficacy of an online module created to train medical students how to handle DTCGT discussions with a patient. Medical students at Edward Via College of Osteopathic Medicine were given a link to a 4-question online pre and post-module survey, an online training

module, and took part in a focus group discussion. The results of this training and discussion will be useful in creating a more thorough module that can be used among many different medical professionals.

Chapter V compiles all of the findings from Chapters II-IV and presents the implications for future research. This chapter also acknowledges the limitations found in the literature as well as the research of this dissertation. Chapter V also discusses the future of DTCGT in light of ethical issues, recent news stories, and how these two issues may change how DTCGT is perceived in the medical field.

REFERENCES

1. Hogarth S, Saukko P. A market in the making: The past, present and future of direct-to-consumer genomics. New Genet Soc. 2017;36(3):197-208. https://doi.org/10.1080/14636778.2017.1354692.

2. Ayoubieh H, Blazer K, Christopher D, et al. Direct-to-consumer genetic testing FAQ for healthcare professionals. genome.gov Web site. https://www.genome.gov/For-Health-Professionals/Provider-Genomics-Education-Resources/Healthcare-Provider-Direct-to-Consumer-Genetic-Testing-FAQ. Updated 2023. Accessed February 1, 2024.

3. Gannett L. The human genome project. The Stanford Encyclopedia of Philosophy Web site. <https://plato.stanford.edu/archives/fall2023/entries/human-genome/>. Updated 2023. Accessed February 27, 2004.

4. Loos RJF. 15 years of genome-wide association studies and no signs of slowing down. Nature Communications. 2020;11(1):5900. https://doi.org/10.1038/s41467-020-19653-5.

5. Altshuler D. A haplotype map of the human genome. Nature. 2005;437:1299-1320.

6. Helgason A, Stefánsson K. The past, present, and future of direct-to-consumer genetic tests. Dialogues in Clinical Neuroscience. 2010;12(1):61-68. https://doi.org/10.31887/DCNS.2010.12.1/ahelgason.

7. Amazon reveals its 5 top-selling items as it steals half of all online sales on Black Friday. businessinsider.com Web site. https://www.businessinsider.com/amazon-top-selling-items-on-black-friday-2017-11. Updated 2017.

8. Find genetic counseling. 23andMe Web site. https://you.23andme.com/tools/genetic-counseling/. Updated 2024. Accessed February 28, 2024.

9. McGowan ML, Fishman JR, Settersten Jr RA, Lambrix MA, Juengst ET. Gatekeepers or intermediaries? the role of clinicians in commercial genomic testing. PloS one. 2014;9(9).

10. Van Der Wouden CH, Carere DA, Maitland-Van Der Zee AH, Ruffin MT, Roberts JS, Green RC. Consumer perceptions of interactions with primary care providers after direct-to-consumer personal genomic testing. Ann Intern Med. 2016;164(8):513-522.

CHAPTER TWO

OBSTACLES TO INTEGRATION OF DIRECT-TO-CONSUMER GENETIC TEST RESULTS INTO PATIENT CARE

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ABSTRACT

Purpose

Direct-to-consumer genetic testing (DTCGT) companies have FDA approval to provide risk predictions of certain disease variants as long as they advise their consumers to discuss any concerns about their results with their healthcare provider. As a result, concerned consumers/patients present providers with unsolicited genetic test results that may lack clinical relevancy. This review aimed to uncover concerns that healthcare providers reported with integrating DTCGT results in clinical care.

Method

Five researchers searched three databases (MEDLINE, Web of Science, and CINAHL) for articles written in English between January 2009 to April 2019. Search terms included the topics of primary care provider and direct-to-consumer genetic tests. Three researchers screened articles for the inclusion criteria and the remaining two researchers performed data extraction. Researchers also identified additional articles from the reference list using a snowball approach. The team completed a thematic analysis using an iterative process.

Results

The reviewers identified four main concerns providers report about integrating DTCGT results into clinical practice. Providers felt that they did not have sufficient genomic literacy to effectively discuss findings with patients. They were unsure of the clinical utility of the test results. They were concerned about legal and ethical issues, for example, regarding upholding patient confidentiality. Without available guidelines from a governing body, providers were unsure if they should be managing genetic information instead of a genetic counselor or geneticist.

Conclusion

This study represents a gap analysis with implications for health professional education within a growing medical genetic landscape. As we move toward more genomics-informed care, training on how to clinically use DTCGT results must be included in academic and continuing medical education.

INTRODUCTION

In 2017, one of Amazon's top five Black Friday sales items was the 23andMe ancestry and health test kit.[1] The high public interest in this item was likely attributed to the fact that in April of that year, the FDA approved an application filed by 23andMe to add genetic risk testing for specific variants associated with ten conditions (including Parkinson's Disease and late-onset Alzheimer's Disease). This approval added to a panel of carrier tests for more than forty conditions already being tested.[2] While many customers have used direct-to-consumer genomic testing (DTCGT) for genealogy, more individuals have become interested in information DTCGT can offer about their health. For example, 23andMe currently provides risk reports for hereditary colorectal cancer and genetic variants associated with medication metabolism.[3] Smaller, more niche DTCGT kits have also appeared on the market. For example, consumers can purchase DNA testing to identify nutritional-related variants for weight loss purposes or variants associated with food sensitivities. Even though the slowed economy since the COVID-19 pandemic has limited the growth of DTCGT sales, companies are maintaining the interest of their substantial customer base by expanding reports to test more genes.[4] Each genetic testing company has a prominently-displayed disclaimer advising customers to seek the advice of a health provider if they have questions regarding a medical condition presented in the test results. These recommendations present a new problem for primary care providers (PCPs), who may be challenged to interpret and manage unsolicited medical test results while considering the clinical utility of what many still believe to be a novelty item.

This systematic review aims to describe providers' knowledge and attitudes about the use of DTCGT results in their practice, their perceived benefits and/or limitations in interacting with patients about test results, and the possibilities of patients and providers uniting to use DTCGT results to create a personalized health plan.

Theoretical Framework

The possible adoption of DTCGT results for clinical use is rooted in provider acceptance of this novel innovation. Over the years, several theoretical frameworks have been created to guide the integration, acceptance, and use of technical innovations. In 2003, John Venkatesh introduced the Unified Theory of Acceptance and Use of Technology, or UTAUT, to explain factors that promote or hinder the adoption of information technology in the workplace. UTAUT is now used as a framework to determine the probability of the acceptance of new technology in classroom and clinical settings. Venkatesh describes the process put in place to facilitate the integration of new technology as a system. His model comprises four primary constructs that influence behavioral intention (a person's intent to adopt a technology) and use behavior (whether a person adopts the technology). The first construct is performance expectancy, which describes the degree to which the provider believes that using the system will help improve job performance. Effort expectancy is the degree of ease associated with use of the system. Social influence pertains to the degree to which a provider perceives that important others (e.g., supervisors) believe the individual should use the new system. Lastly, facilitating conditions influence the degree to which the provider believes that an organizational and technical infrastructure exists to support the system's use. Venkatesh and his colleagues later modified UTAUT when they concluded that gender, age, experience, and voluntariness of use influence the initial four constructs.[5]

METHODS

For this review, three reviewers (L.C., C.C., and L.S.) searched Medline, Web of Science, and CINAHL using the following keywords in text and MESH terms: primary care provider, family doctor, family practitioner, internist, healthcare provider, nurse, nurse practitioner, and personalized genomic testing, personalized genetic testing, direct-to-consumer genetic testing, and genetic testing. Reviewers limited the search to English-language articles published between January 2009 and April

2019. Articles pertaining to genetic counselors, specific genetic tests, or provider-initiated testing were excluded, culminating in 949 articles. All reviewers met to compile initial findings and delete duplicates between the three databases and create a spreadsheet of articles for full-text review. L.C., C.C., and L.S. performed full-text reviews of the 46 remaining articles, excluding those about advertising, adolescents, and one article that had never been published. L.G.C. and K.G. extracted data for each of the remaining 31 articles, based on study purpose, method/sample size, and outcomes, leaving 19 total articles that addressed PCP and patient insights on DTCGT. In January 2021, 6 additional articles were identified by doing a reference/cited-by search (snowball search), producing a total of 25 articles used in this review.

Table 2.1 Article search and selection process

RESULTS

The results of this search reveal providers' apprehensions regarding patient-provided DTCGT results. Four main concerns were identified and represent barriers to the integration of DTCGT results in clinical practice. These barriers are providers' limited knowledge of genetics/genomics, uncertainty about the clinical utility of test results, concerns about legal and ethical issues, and the lack of a single comprehensive set of guidelines put forth by a regulatory agency.

Knowledge

Because of the recommendations provided by genomic testing companies, PCPs are aware that interpretation and discussion of DTCGT results may become their responsibility.[6] Providers are even optimistic about the clinical utility of DTCGT, with most PCPs agreeing that test results could become clinically useful in the next 5-10 years.[7] However, with acceptance comes trepidation. Barriers to providers' acceptance of DTCGT as a clinical tool represent two general roadblocks: general knowledge and perceived clinical utility.

The first roadblock relates to providers' concerns about their own knowledge; specifically, gaps in their general genetic/genomic literacy, uncertainty about the validity of available DTC tests, and difficulty finding resources about how to use DTCGT results to inform clinical care.

Established providers expressed a more significant lack of genetic/genomic literacy than their younger counterparts.[8,9] This gap may reflect the fact that newer providers are more likely to have learned about precision medicine during their formal education and to have opportunities for exposure to genetic/genomic research. A survey of PCPs in the United States found that 55% of respondents felt confident in interpreting results on DTCGT reports. Still, only 22% thought they had enough training to discuss findings with a patient or make clinical recommendations based on test results.[10]

PCPs are aware of some of the better-known genomic testing companies such as 23andMe,

Navigenics (now Thermo Fisher Scientific), and the now-defunct AncestryHealth, but the number of personal genomic testing companies has increased and more disease-specific test panels are being offered. PCPs have voiced concerns about whether the labs are certified and whether the test results are accurate.[11] The most common request by PCPs regarding DTCGT is that the testing companies provide evidence-based guidelines to manage the risks identified by the test.[8]

Providers also voiced concerns regarding where to find support in clinical decision-making in the face of possibly adverse test results. As stated earlier, many providers feel confident in interpreting results but not discussing results with a patient. Only a few companies, like 23andMe, provide a dedicated website just for PCPs to access education on gene variants and pharmacogenomic relevancies. Physicians who participated in Carroll et al.'s 2016 study[10] stated that it would be beneficial to be able to consult with on-call genetic professionals, either in their healthcare system or at a local genetics center. PCPs may be accustomed to contacting an oncologist to request a consultation about cancer genetics but having someone specialized in genomics and precision medicine would be of more significant benefit.[10] Healthcare providers in a 2019 survey stated that they preferred training and information directly from the DTCGT provider (Navigenics) and their staff of genetic counselors, admitting "We (physicians) cannot keep track of the basic science - that explosion is occurring logarithmically."[12]

Healthcare professionals have pledged to be lifelong learners, and DTCGT and precision medicine provide a prime opportunity. Many PCPs are eager to learn more about DTCGT and how to integrate results into practice.[13] Providers have noted that they prefer to obtain this knowledge through journal articles, informational sessions during professional meetings, and credible websites.[14] In a survey of physicians in the Kaiser-Permanente network, over half of the 1415 respondents were willing to spend 2-4 hours per year learning about various genetic/genomic tests and how to integrate them into their practice.[13]

The second roadblock to integrating DTCGT into patient care is the perception of clinical utility. A marketing point of companies such as 23&Me is to provide "fun fact" genetics like mosquito bite frequency and earlobe type that are useless in a clinical setting. PCPs realize that those same companies target their clinical testing toward chronic diseases with lifestyle implications (cancer, diabetes, heart disease) and view those types of tests as an advantage in managing patient care.[15] Providers who have viewed the complex disease reports of their patient's results find them understandable,[6] but most providers question whether the results provide actionable information that they can translate into patient care. For example, 23andMe currently tests for only three of the >1,000 known cancer-causing BRCA variants, leaving many patients in the dark about their actual risk. If companies tested more comprehensive ranges of variants, test results could prompt earlier screenings such as mammograms, colonoscopies, or EKGs. Another serious limitation is that DTCGC companies currently do not link risk predictions with evidence-based guidelines provided by established medical organizations t to reduce or alleviate risks.[14]

Time is a factor in determining the clinical utility of any item. Given significant time constraints requiring PCPs to consult with a specified number of patients in a finite number of hours, providers express concern that there is insufficient time to thoroughly review and explain DTCGT results during a routine office visit.[15,16] Pharmacogenomic DTCGTs may be a type of time-saving test. For example, in a study of 502 physicians, most reported starting a patient on Warfarin yet 89% never ordered a pharmacogenetic test to determine optimal dosing.[6] If a patient's DTC pharmacogenomic test results were uploaded into their electronic medical record, providers could access that information in time to inform initial prescribing decisions. That might eliminate having to re-dose because of adverse reactions and save time in the long run.

Any achievement of clinical utility is wholly dependent on the interaction between patient and provider. Even though DTCGT companies suggest their customers share concerns and questions with their PCP, many customers do not feel the need to do so. In a 2016 study of 1,026 DTCGT

customers, 65% did not share results with a PCP. The most common reason for not sharing (reported by 42% of participants) was that they felt their results were not important enough to bring up. Study participants who did present their results to their PCP reported varied experiences. When asked about the interaction with their provider, 32% reported having a satisfactory discussion, 22% reported a lack of engagement from their provider, and 15% felt that their provider lacked knowledge and thus refused to engage in conversation or even acknowledge the test results.[17]

Patient-focused research brings to light issues of communication between patient and provider. Patients with DTCGT results may be prepared and ready to partner with their PCP to create a healthcare plan that is informed by genetic information, but such partnering requires providers to be receptive of the information.[4] In a survey of 1,404 family physicians, 58% felt that DTCGT results would more than likely harm a patient's general health decisions.[4] According to Dinulos and Vallee, harm can be avoided by taking the first step: to "explain DTCGT in language that the patient can understand".[18] That explanation should include letting a patient know that the results may not be fully accurate and that all genetic variants associated with a disease may not have been tested. Therefore, a family history also must be taken, and concerning test results must be confirmed by a clinical diagnostic laboratory.[19,20]

Legal and ethical issues

The Osteopathic Oath, Hippocratic Oath, and pledges taken by other PCPs all set in place the ideals of helping and not harming patients; however, four other duties also bind primary care providers: duty to inform, duty to treat, duty to follow up, and duty to protect patient confidentiality.[21] One issue that PCPs report is that DTCGT may muddy the waters regarding where their duties begin and end. Usually, a provider will order a genetic test for a particular reason, typically after a thorough explanation to the patient of possible outcomes and avenues of treatment. DTCGT removes those steps, leaving the provider to interpret unsolicited results for a test that may have been ordered without a compelling indication.[7] Suppose the testing company's results cannot provide the same

level of validity that the provider's clinical lab could provide. To what extent Is the provider obligated to discuss the results with the patient?[22]

The provider's duty to inform becomes complex in the face of DTCGT. Companies provide results for selected genetic variants associated with a disease. For example, 23andme tests for only one of the three variants strongly associated with Alzheimer's disease. For a patient who tests negative for that variant, providers may find themselves choosing whether to bring up the possibility of another variant that may be more relevant given a patient's family history. At that point, a provider may choose to order further testing. If a provider decides to inform their patient about the meaning of DTCGT results, they are obliged to make decisions about treatment and follow up. If a provider does not, or cannot, inform, they may not be honoring their duty to treat and follow up.[6]

A provider is unable to perform the duty to protect patient confidentiality when it comes to DTCGT if the patient has opted into medical or genome-wide association study (GWAS) research. At this point, the doctor/patient confidentiality agreement affords no protection against privacy breaches involving information held by genetic testing companies or entities with whom they may share data.

Practice recommendations

Looking back to Venkatesh et al.'s Unified Theory of Acceptance and Use of Technology, the social influence of important others plays a part in a provider's choice to integrate DTCGT into clinical care.[5] The American Society for Human Genetics has called for the education of providers as well as the standardized certification of all DTCGT laboratories.[23] CLIA certification has been the standard to assure clinical accuracy in most US labs, but clinically accurate does not equal clinically meaningful.[24] To facilitate clinical relevancy, the American College of Medical Genetics and Genomics has recommended minimum requirements for testing protocols. Testing results should be communicated by the laboratory in a readily understandable format and scientific evidence for each gene tested must be provided.[23] These recommendations help form the base of an organizational and technical infrastructure to support the clinical use of DTCGT results and decrease effort expectancy.

Not all governing bodies support patient use of DTCGT without provider input. In South Africa, the National Department of Health recommends that the consumer consult a genetic counselor, clinical geneticist, or clinician beforehand in order to determine the appropriateness of the test and afterward to interpret results.[23] In 2009, Germany passed the Genetics Diagnostics Act, which requires the initiation of any form of genetic testing to have physician involvement.[23] The American College of Obstetricians and Gynecologists (ACOG) stated in their 2021 committee opinion that "DTCGT should be discouraged without appropriate counseling." The ACOG further recommended that patients presenting a DTCGT, such as for carrier status, be referred to a provider skilled in risk assessment of these specific diseases and await confirmatory testing from a clinical laboratory.[25]

CONCLUSION

In the past decade, providers have seen DTCGT evolve from being just a fun introduction to genetics for the everyday person to becoming a possible catalyst for individualized healthcare. Established healthcare providers report a lack of genomic literacy when it comes to DTCGT and feel unprepared to integrate DTCGT results into patient care. This gap in provider knowledge occurs in the face of rapid advancement of genomic technologies. Luckily, these providers are willing to have genomic training to keep up with a growing population of genomic-savvy patients. Medical education regarding the clinical application of DTCGT must be implemented to quickly narrow this gap.

A common concern among providers has been about the clinical utility of these test results. Although CLIA certification of DTCGT laboratories is the present standard for clinical validity, certification does not assure that test results can be used in a meaningful manner during a patient visit.

Current medical opinion about DTCGT is varied but leaning toward capitalizing on patient curiosity and concern about disease risks to prompt? providers into following up with clinical testing.

Although studies have explored the use of DTCGT from both patient and provider viewpoints, the literature on this subject is incomplete. More data is needed on the outcomes of patient/provider collaborations and the incorporation of DTCGT test results into patient electronic health records.

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REFERENCES

1. Amazon reveals its 5 top-selling items as it steals half of all online sales on black friday. businessinsider.com Web site. https://www.businessinsider.com/amazon-top-selling-items-on-black-friday-2017-11. Updated 2017.

2. F.D.A. will allow 23andMe to sell genetic tests for disease risk to consumers . nytimes.com Web site. https://www.nytimes.com/2017/04/06/health/fda-genetic-tests-23andme.html. Updated 2017.

3. DNA reports page. 23andme.com Web site. 23andme.com/dna-reports-list. Accessed 3/2/2021.

4. Majumder MA, Guerrini CJ, McGuire AL. Direct-to-consumer genetic testing: Value and risk. Annu Rev Med. 2020;72.

5. V. Venkatesh, T. A. Sykes, X. Zhang. 'Just what the doctor ordered': A revised UTAUT for EMR system adoption and use by doctors. 2011 44th Hawaii International Conference on System Sciences. 2011:1-10. doi: 10.1109/HICSS.2011.1.

6. Bernhardt BA, Zayac C, Gordon ES, Wawak L, Pyeritz RE, Gollust SE. Incorporating direct-to-consumer genomic information into patient care: Attitudes and experiences of primary care physicians. Personalized medicine. 2012;9(7):683-692.

7. Mainous,Arch G.,,3rd, Johnson SP, Chirina S, Baker R. Academic family physicians' perception of genetic testing and integration into practice: A CERA study. Fam Med. 2013;45(4):257-262. http://libproxy.clemson.edu/login?url=http://search.ebscohost.com/login.aspx?direct=true&db=mnh&AN=23553089.

8. Powell KP, Cogswell WA, Christianson CA, et al. Primary care physicians' awareness, experience and opinions of direct-to-consumer genetic testing. Journal of genetic counseling. 2012;21(1):113-126.

9. McGrath SP, Walton N, Williams MS, Kim KK, Bastola K. Are providers prepared for genomic medicine: Interpretation of direct-to-consumer genetic testing (DTC-GT) results and genetic self-efficacy by medical professionals. BMC health services research. 2019;19(1):1-12.

10. Carroll JC, Makuwaza T, Manca DP, et al. Primary care providers' experiences with and perceptions of personalized genomic medicine. Can Fam Physician. 2016;62(10):e626-e635. http://libproxy.clemson.edu/login?url=http://search.ebscohost.com/login.aspx?direct=true&db=mnh&AN=27737998.

11. Loud JT. Direct-to-consumer genetic and genomic testing: Preparing nurse practitioners for genomic healthcare. The Journal for Nurse Practitioners. 2010;6(8):585-594.

12. McGowan ML, Fishman JR, Settersten Jr RA, Lambrix MA, Juengst ET. Gatekeepers or intermediaries? the role of clinicians in commercial genomic testing. PloS one. 2014;9(9).

13. Jonas MC, Suwannarat P, Burnett-Hartman A, et al. Physician experience with direct-to-consumer genetic testing in kaiser permanente. Journal of personalized medicine. 2019;9(4):47.

14. Ram S, Russell B, Gubb M, et al. General practitioner attitudes to direct-to-consumer genetic testing in new zealand. Clinical Correspondence. 2012.

15. Powell KP, Christianson CA, Cogswell WA, et al. Educational needs of primary care physicians regarding direct-to-consumer genetic testing. Journal of genetic counseling. 2012;21(3):469-478.

16. Pet DB, Holm IA, Williams JL, et al. Physicians' perspectives on receiving unsolicited genomic results. Genetics in Medicine. 2019;21(2):311.

17. van der Wouden C,H., Carere DA, Maitland-van der Zee A,H., Ruffin,Mack T.,,4th, Roberts JS, Green RC. Consumer perceptions of interactions with primary care providers after direct-to-consumer personal genomic testing. Ann Intern Med. 2016;164(8):513-522. http://libproxy.clemson.edu/login?url=http://search.ebscohost.com/login.aspx?direct=true&db=mnh&AN=26928821. doi: 10.7326/M15-0995.

18. Dinulos MBP, Vallee SE. The impact of direct-to-consumer genetic testing on patient and provider. Clin Lab Med. 2020;40(1):61-67.

19. Thrush SA, McCaffrey R. Direct-to-consumer genetic testing: What the nurse practitioner should know. The Journal for Nurse Practitioners. 2010;6(4):269-273.

20. Artin MG, Stiles D, Kiryluk K, Chung WK. Cases in precision medicine: When patients present with direct-to-consumer genetic test results. Ann Intern Med. 2019;170(9):643-650.

21. Bartlett G, Avard D, Knoppers BM. A new twist on an old problem: Primary care physicians and results from direct-to-consumer genetic testing. Personalized Medicine. 2013;10:827+. http://link.galegroup.com/apps/doc/A345821561/AONE?u=clemsonu_main&sid=AONE&xid=a24c7cb3.

22. Niemiec E, Kalokairinou L, Howard HC. Current ethical and legal issues in health-related direct-to-consumer genetic testing. Personalized Medicine. 2017;14(5):433-445.

23. Dandara C, Greenberg J, Theron M, et al. Direct-to-consumer genetic testing: To test or not to test, that is the question: Forum-clinical practice. African Journal of Health Professions Education. 2013;1

24. Mahon SM. Direct-to-consumer genetic testing: Helping patients make informed choices. Clin J Oncol Nurs. 2018;22(1).

25. Consumer Testing for Disease Risk: ACOG Committee Opinion, Number 816. Obstet Gynecol. 2021;137(1):e1-e6. doi:10.1097/AOG.0000000000004200.03(8):510-512.

CHAPTER THREE

OSTEOPATHIC MEDICAL STUDENTS'S ATTITUDES ABOUT THE CLINICAL APPLICATION OF DIRECT-TO-CONSUMER GENETIC TEST RESULTS

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ABSTRACT

Background

As personal genomics testing becomes more widespread, primary care providers have been tasked with the responsibility of handling patients' questions and concerns about direct-to-consumer genetic test (DTCGT) results. Literature shows that physicians who were not exposed to genetic/genomic training in medical school are less apt to find clinical utility in DTCGT results.

Objective

The authors sought to gauge the knowledge of and attitudes toward DTCGT in a survey of osteopathic medical students using a modified Unified Theory of Acceptance and Use of Technology (UTAUT) model format.

Methods

Between January 17 and February 3, 2023, students enrolled at Edward Via College of Osteopathic Medicine - Carolinas Campus were invited to participate in a 15-item online survey (Likert-type and multiple-choice responses). Data were analyzed on student knowledge, performance expectations, perceived clinical utility, and effort expectations in relation to possible patient interactions concerning direct-to-consumer genetic test results.

Results

One hundred sixty-five students completed the survey. Most respondents (n=115; 69.7%) reported feeling knowledgeable about basic genetic/genomic terminology; however, over half (n=86; 52.1%) were uncomfortable with discussing direct-to-consumer test results with a patient. Students see the clinical utility of the test results (n=144; 87.3%) and would like to further their education on how to use results for patient benefit (n=151; 91.5%).

Conclusion

Osteopathic medical students' uneasiness with potentially discussing direct-to-consumer genetic test results with patients has bolstered their curiosity to find ways to learn more about DTCGT. Inclusion of additional training in communicating genetic information to patients is needed in medical curricula.

INTRODUCTION

In this burgeoning age of personalized medicine, patients expect all medical professionals to have enough general genomic knowledge to answer their questions and correct misconceptions. This expectation is especially true when patients have concerns about results from direct-to-consumer genetic tests (DTCGT), which are being used more frequently. Medical professionals in non-genomic related fields often need continuing education to stay abreast of the rapidly growing world of genomic information and technology.[1,2]

In a study of 382 physicians, Powell et al.[3,4] found that physicians well-established in their practices expressed a more significant lack of genetic/genomic literacy than their less experienced counterparts. This may reflect the fact that less experienced and more recently trained physicians may have had personalized medicine integrated into their medical education and more chances for exposure to genetic/genomic research. However, that exposure mainly pertained to clinical genetic testing rather than DTCGT. The American Academy of Family Physicians' medical genetics curriculum guidelines state that residents must be able to "educate patients about the risks and benefits with DTCGT,"[5] and yet little to no training is given on DTCGT despite the various opportunities to incorporate it into the curriculum.[6]

In addition to genetic/genomic literacy, how medical professionals will accept the integration of DTCGT into clinical practice is also in question. Previous studies of medical professionals and students usually deal with the attitude toward physician-directed genomic testing,[7,8] yet the uncertainty of medical responsibility surrounding DTCGT needs further research.[9]

The aim of this study was to gauge the outlook of osteopathic medical students and their attitudes toward using DTCGT results in the clinical setting. The authors also sought to identify any factors contributing to student resistance to discussing DTCGT results with patients.

METHODS

All students enrolled at Edward Via College of Osteopathic Medicine (VCOM) - Carolinas Campus (N=652) were eligible to participate in this survey. Recruitment identified students in two phases of training: osteopathic medical students in their first two years of didactic study (OMS-I & OMS-II) and osteopathic medical students at clinical rotation sites (OMS-III & OMS-IV). A recruitment email was sent to students through the campus-wide database. Of the 652 possible participants, 165 agreed to participate.

The survey was a quantitative descriptive cross-sectional study consisting of 15 multiple-choice and Likert scale questions. There were four demographic questions relating to year in the program, number of genetic classes taken before enrolling into medical school, and if the student had taken a DTCGT. There were six questions related to the perceived knowledge and utility of DTCGT (adapted and modified from the works of Powell et al.).[3,4] The remaining questions assessed performance expectancy and effort expectancy based on the Unified Theory of Acceptance and Use of Technology (UTAUT) model.[10,11] This theory explains how performance and effort expectations influence behavioral intentions and, thus, the subsequent usage behaviors of an emergent technology. UTAUT has been used in studies on how medical professionals adapted to integrating electronic medical records systems into daily use and will best describe how medical students will approach DTCGT education.[11]

The authors of this study mapped items to the survey domains of perceived knowledge, perceived utility, performance expectancy, and effort expectancy. The survey was pilot tested by a group of healthcare professionals (i.e., a registered nurse, a physician assistant, and an osteopathic physician). Survey questions were then revised for better alignment with the survey domains based on feedback.

The 15-question online survey (See Appendix A) was conducted via Qualtrics ® survey software from January 17 until February 3, 2023, with reminders sent by email at seven and 14 days. Online consent was obtained, participants were assured of confidentiality, and no personal identifiers were collected.

Survey data were analyzed using Qualtrics XM ® for statistical analyses. Comparisons were made between class designations (OMS I through IV), the number of genetic/genomic courses taken in undergraduate school, and if the student had personally taken a DTC-GT, as well as participant responses to questions of knowledge, utility, performance expectations, and effort expectations. Both the Clemson University Institutional Review Board (IRB) and VCOM IRB provided approval through an exempt review status (See Appendix B).

RESULTS

Of 652 eligible students, 165 (25.3%) completed the survey. A slight majority of the respondents (n=89; 54.6%) were in their didactic years (years 1 and 2). Only 28 students (17%) had never taken a genetic/genomic course before entering medical school (Table 3.1). In total, 34 (20.6%) students had taken a DTCGT, with only five (3% of total students) discussing their results with a doctor.

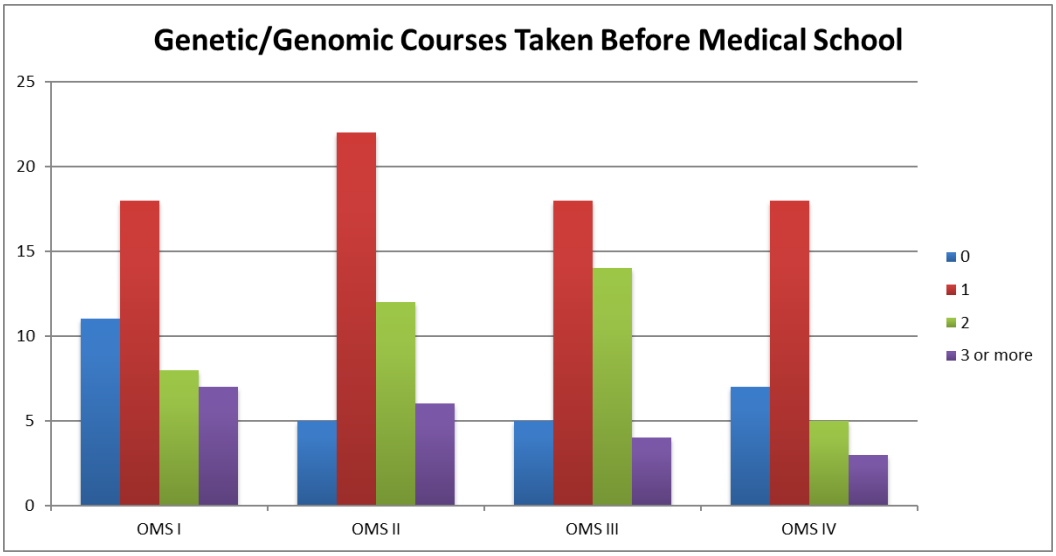
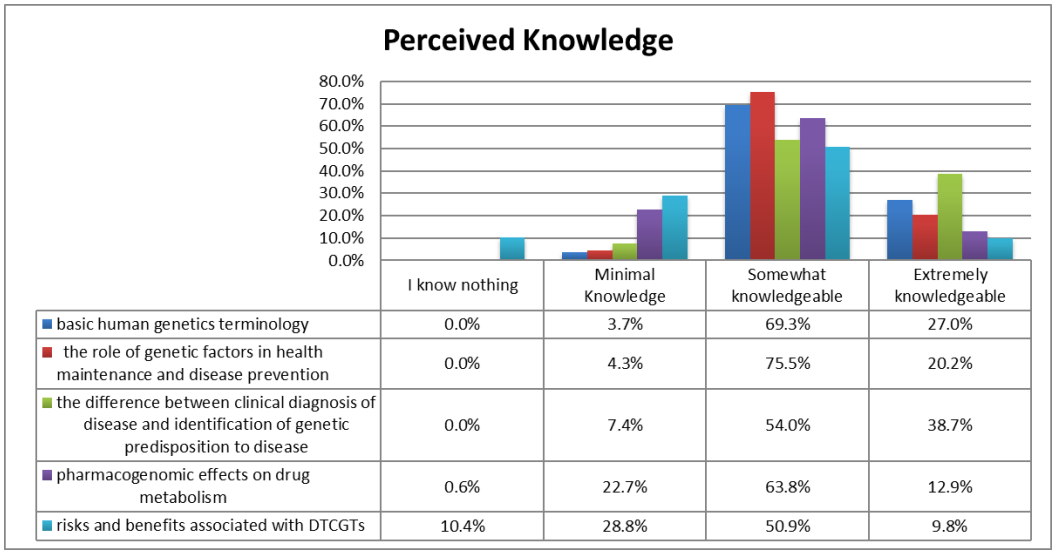


Table 3.1. Descriptive Information on Participants in the Genetics Education Survey, 2023.

Perceived Knowledge

Most students (n=115; 69.7%) felt somewhat knowledgeable about basic human genetics terminology. More students ranked themselves as knowledgeable about the role of genetic factors in health maintenance and disease prevention (n=125; 75.8%). Slightly over one-third of the students (n=64; 38.8%) felt they were extremely knowledgeable about the difference between the clinical diagnosis and the genetic predisposition of a disease. A total of 39 students (23.6%) stated that they had minimal to no knowledge of the pharmacogenomic effects on drug metabolism despite pharmacogenomics being a part of their first-semester medical school curriculum. When asked to rank their knowledge about the risks and benefits associated with DTCGTs, 65 students (39.4%) reported minimal to none (Table 3.2).



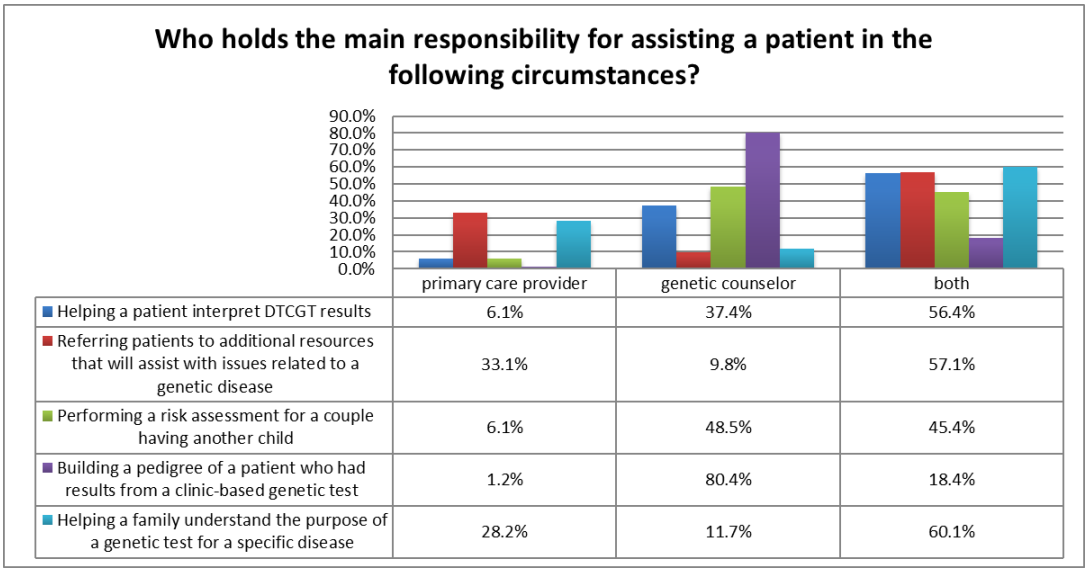
	I know nothing	Minimal Knowledge	Somewhat knowledgeable	Extremely knowledgeable
basic human genetics terminology	0.0%	3.7%	69.3%	27.0%
the role of genetic factors in health maintenance and disease prevention	0.0%	4.3%	75.5%	20.2%
the difference between clinical diagnosis of disease and identification of genetic predisposition to disease	0.0%	7.4%	54.0%	38.7%
pharmacogenomic effects on drug metabolism	0.6%	22.7%	63.8%	12.9%
risks and benefits associated with DTCGTs	10.4%	28.8%	50.9%	9.8%

Table 3.2. Perception of Current Knowledge of Participants in the Genetics Education Survey, 2023

Performance Expectancy

Venkatesh defines performance expectancy as the degree to which an individual believes that using the system will help him or her attain gains in job performance.[10] To assess individual performance expectancy, the students were asked to rate how comfortable they would be performing certain clinical tasks. Only half of the students (n=83; 50.3%) felt somewhat comfortable taking and

interpreting a patient's family genetic history, while (n=86; 52.1%) were uncomfortable discussing DTCGT results with a patient. Explaining DTCGT risk assessments of disease also put students at unease, with 91 (55.2%) reporting being uncomfortable with the task. Nearly half of the respondents felt somewhat comfortable using information technology to obtain current and credible information about genetics (n=81; 49.1%) and integrating genetic test results into patient management (n=84; 50.9%). When asked about the coordination of care and responsibility between a clinician and a genetic counselor in certain circumstances, students often responded that responsibility belonged to the genetic counselor or to both parties (Table 3.3).



	primary care provider	genetic counselor	both
Helping a patient interpret DTCGT results	6.1%	37.4%	56.4%
Referring patients to additional resources that will assist with issues related to a genetic disease	33.1%	9.8%	57.1%
Performing a risk assessment for a couple having another child	6.1%	48.5%	45.4%
Building a pedigree of a patient who had results from a clinic-based genetic test	1.2%	80.4%	18.4%
Helping a family understand the purpose of a genetic test for a specific disease	28.2%	11.7%	60.1%

Table 3.3. Performance Expectancy of Participants in the Genetics Education Survey, 2023

Perceived Utility

An overwhelming majority of students (n=144; 87.3%) reported seeing the clinical utility of DTCGT, with most choosing reasons such as offering screening tests at an earlier age (n=132; 91.6%), to individuals found to be at an increased risk (n=119; 82%), and/or recommending lifestyle changes based on risk predictions (n=118; 81.9%; Table 3.4). The students who did not see the clinical utility

did not trust the clinical certification of an unknown genetic testing facility (n=14; 67%), the lack of formal guidelines to reduce or alleviate risks (n=13; 61.9%), or increased patient anxiety (n=13; 61.9%). Altogether, a little over one-fourth of the respondents (n=44; 26.6%) reported DTCGT results from a patient would not influence how they would provide care. An even lower amount (12.7%) stated that they found no clinical utility in DTCGT and did not want to learn more about it because the companies "don't test for as many genetic variants and are riddled with disclaimers" or "I don't trust the clinical certification of an unknown genetic testing facility" which mirror the sentiments of many clinicians.[4]

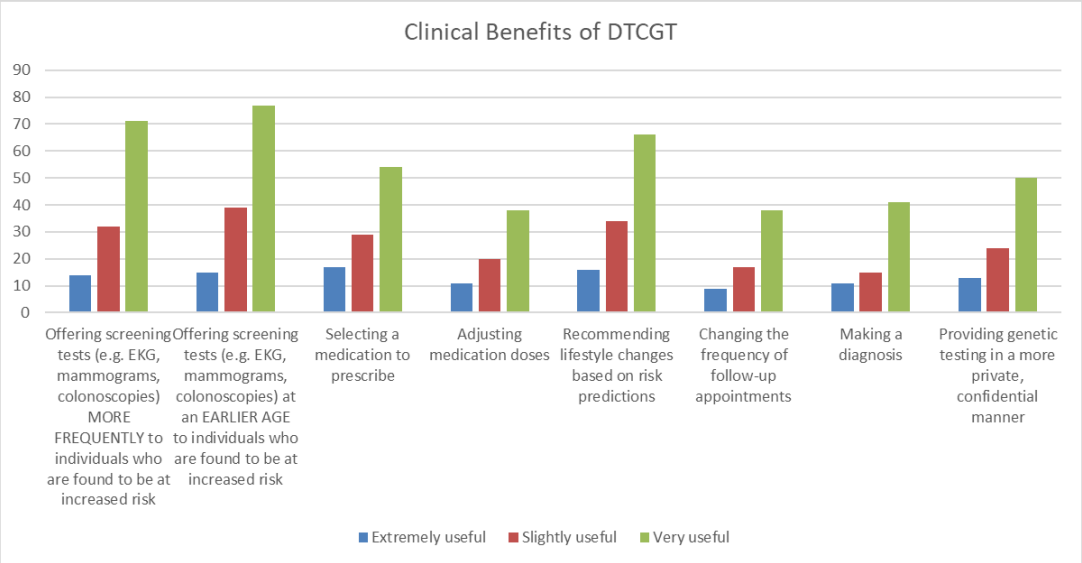


Table 3.4. Perceived Utility of DTCGT among Participants in the Genetics Education Survey, 2023

Effort Expectancy

To gauge the effort expectancy, or the degree of ease associated with the use of the system,[10] participants were asked if they would like to know more about DTCGT and which topics were of interest. A total of 151 (91.5%) respondents reported that they would like to learn more, with 135 (89.4%) wanting to know how to interpret test results and 129 (85.4%) wanting to know where to find credible information about the tests (Table 5). Of the 14 students who did not want to learn more

about DTCGT, eight (57.1%) reported that they would learn more if it ever came up during a patient visit, and four (28.6%) were not interested in the topic at all.

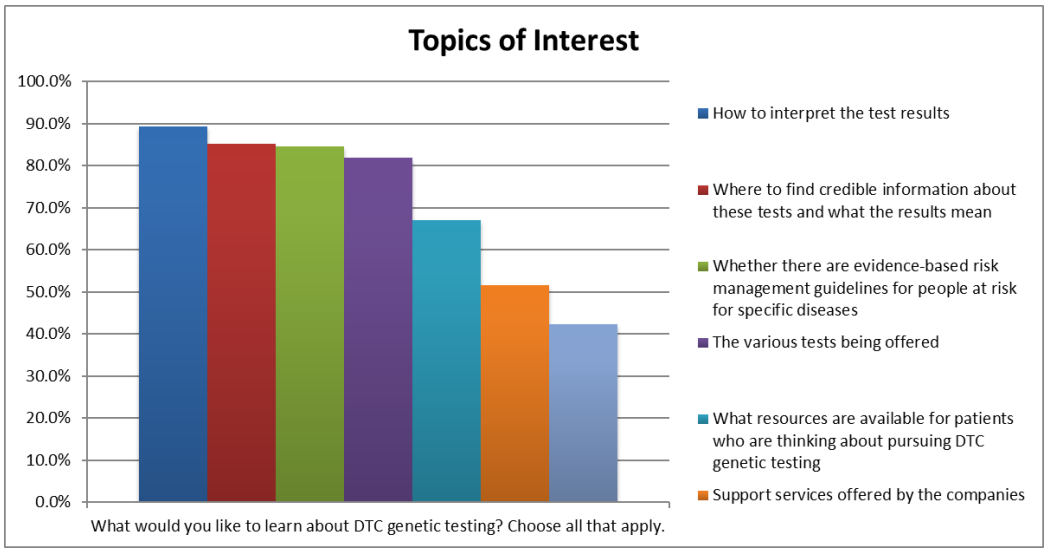


Table 3.5. Educational Interests Chosen by Participants in the Genetics Education Survey, 2023

DISCUSSION

In this survey encompassing 165 osteopathic medical students across their four years of training, students reported feeling competent and eager to learn more about DTCGT and how to use it to enhance clinical care. At VCOM, all students take a genetics course in the Fall semester of their first year. This is similar to findings by Thurston et al.'s study of US and Canadian medical schools,[12] suggesting that our findings are applicable across medical school curricula and medical training programs. In addition to the stand-alone genetics course required at the beginning of the first didactic year, most respondents took at least one genetics course before attending VCOM. This may have contributed to the amount of comfort reported in knowledge, the positive acceptance of clinical utility, and the willingness to learn more about DTCGT. This eagerness to learn may also be rooted in the need for clarity of performance expectancy. Students want to know if they are responsible for handling a clinical genetic task or if it should fall to a genetic counselor.

However, DTCGT may require more emphasis in current curricula for students to be proficient. A study of 112 medical genetics course director respondents indicated that DTCGT was included in less than 50% of allopathic and osteopathic medical schools in the United States and Canada in 2013-14.[13] The proportion of medical schools including instruction on DTCGT in recent years is unknown. DTCGT is currently only mentioned in the VCOM genetics course as an avenue of personalized genetic testing and not as a clinical tool like physician-directed genomic testing. Future research of how DTCGT is taught in the current curriculum of medical schools is needed.

Results of this study indicate that student responses aligned with many of their peers in that a majority feel somewhat knowledgeable about how pharmacogenomics relates to drug metabolism and how its usage is beneficial in clinical care.[8] The US Food and Drug Administration (FDA) granted 23andMe approval for select pharmacogenomic testing in October 2018,[14] so it may be likely that additional research on how this topic will come up in patient visits will be needed.

Limitations and Future Implications

While the sample size of respondents was 165, this study was conducted with medical students based at one campus. Replicating this study on a larger scale with additional healthcare clinicians (i.e., nurse practitioners and physician assistants) may yield stronger data and results.

An implication for practice is that in the absence of medically regulated guidelines for the use of DTCGT in clinical practice, formalized training programs are needed. These programs should address the common concerns/questions that may arise when discussing DTCGT results during routine office visits and how to handle them.

CONCLUSION

The osteopathic medical students were comfortable with their genetic/genomic knowledge, yet their confidence was lower when discussing genetic test results with patients. That lack of confidence

appears to have bolstered a willingness to learn how to integrate patient-initiated genomic testing into clinical care. Medical educators need to implement standardized educational interventions on precision medicine, including interpretation of direct-to-consumer genetic testing, that capitalize on the eagerness to learn to positively impact the care of patients and the comfort of clinicians.

REFERENCES

1. Soller HB. *The acquisition of clinical genetics knowledge in medical students.* [M.Sc.]. University of Pittsburgh; 2022.

2. Crellin E, McClaren B, Nisselle A, Best S, Gaff C, Metcalfe S. Preparing medical specialists to practice genomic medicine: Education an essential part of a broader strategy. *Frontiers in genetics*. 2019;10:789.

3. Powell KP, Christianson CA, Cogswell WA, et al. Educational needs of primary care physicians regarding direct-to-consumer genetic testing. *Journal of genetic counseling*. 2012;21(3):469-478.

4. Powell KP, Cogswell WA, Christianson CA, et al. Primary care physicians' awareness, experience and opinions of direct-to-consumer genetic testing. *J Genet Couns*. 2012;21(1):113-126.

5. American Academy of Family Physicians. Recommended curriculum guidelines for family medicine residents: Medical genetics aafp.org Web site. www.aafp.org/dam/AAFP/documents/medical_education_residency/program_directors/. Updated 2019. Accessed 1/20/, 2022.

6. Hagiwara N. Application of active learning modalities to achieve medical genetics competencies and their learning outcome assessments. *Advances in medical education and practice*. 2017;8:817.

7. Carver RB, Castéra J, Gericke N, Evangelista NAM, El-Hani CN. Young adults' belief in genetic determinism, and knowledge and attitudes towards modern genetics and genomics: The PUGGS questionnaire. *PloS one*. 2017;12(1):e0169808.

8. Rahma AT, Elsheik M, Elbarazi I, et al. Knowledge and attitudes of medical and health science students in the United Arab Emirates toward genomic medicine and pharmacogenomics: A cross-sectional study. *Journal of Personalized Medicine*. 2020;10(4):191.

9. Bernhardt BA, Zayac C, Gordon ES, Wawak L, Pyeritz RE, Gollust SE. Incorporating direct-to-consumer genomic information into patient care: Attitudes and experiences of primary care physicians. *Personalized Medicine*. 2012;9(7):683-692.

10. Venkatesh V, Morris MG, Davis GB, Davis FD. User acceptance of information technology: Toward a unified view. *MIS quarterly*. 2003:425-478.

11. Venkatesh V, Sykes TA, Zhang X. 'Just what the doctor ordered': A revised UTAUT for EMR system adoption and use by doctors. . 2011:1-10.

12. Thurston VC, Wales PS, Bell MA, Torbeck L, Brokaw JJ. The current status of medical genetics instruction in US and Canadian medical schools. *Academic Medicine*. 2007;82(5):441-445.

13. Plunkett-Rondeau J, Hyland K, Dasgupta S. Training future physicians in the era of genomic medicine: Trends in undergraduate medical genetics education. *Genetics in Medicine*. 2015;17(11):927-934.

14. Gammal RS, Mayes J, Caudle KE. Ready or not, here it comes: Direct-to-consumer pharmacogenomic testing and its implications for community pharmacists. *Journal of the American Pharmacists Association*. 2019;59(5):646-650.

Appendix A

Osteopathic Medical Student Attitudes Survey Questions

What is your academic designation?

- ○ OMS I (1)
- ○ OMS II (2)
- ○ OMS III (3)
- ○ OMS IV (4)
- ○ I am not an enrolled student (5)

Skip To: End of Survey If What is your academic designation? = I am not an enrolled student

How many genetic/genomic courses did you take before entering medical school?

- ○ 0 (1)
- ○ 1 (2)
- ○ 2 (3)
- ○ 3 or more (4)

Have you ever taken a direct-to-consumer genetic test (DTCGT)?

- ○ No (1)
- ○ Yes (2)

Skip To: Q2 If Have you ever taken a direct-to-consumer genetic test (DTCGT)? = No

Did you discuss any of your DTCGT results with a doctor?

- ○ No (1)
- ○ Yes (2)

How would you rank your knowledge of...

	I know nothing (1)	Minimal Knowledge (2)	Somewhat knowledgeable (3)	Extremely knowledgeable (4)
basic human genetics terminology (1)	○	○	○	○
the role of genetic factors in health maintenance and disease prevention (2)	○	○	○	○
the difference between clinical diagnosis of disease and identification of genetic predisposition to disease (3)	○	○	○	○
pharmacogenomic effects on drug metabolism (4)	○	○	○	○
risks and benefits associated with DTCGTs (5)	○	○	○	○

How comfortable are you...

	Extremely uncomfortable (1)	Somewhat uncomfortable (2)	Somewhat comfortable (3)	Extremely comfortable (4)
taking and interpreting a patient's family genetic history (1)	○	○	○	○
discussing DTCGT results with a patient (3)	○	○	○	○
explaining DTCGT risk assessments of diseases to a patient (2)	○	○	○	○
using information technology to obtain current and credible information about genetics for oneself, patients, and colleagues (4)	○	○	○	○
incorporating genetic test results into patient management (5)	○	○	○	○

In your opinion, who holds the main responsibility for assisting a patient in the following circumstances?

	primary care provider (1)	genetic counselor (2)	both (3)
Helping a patient interpret DTCGT results (1)	○	○	○
Referring patients to additional resources that will assist with issues related to a genetic disease (2)	○	○	○
Performing a risk assessment for a couple having another child (3)	○	○	○
Building a pedigree of a patient who had results from a clinic-based genetic test (4)	○	○	○
Helping a family understand the purpose of a genetic test for a specific disease (5)	○	○	○

In general, do you think DTCGTs are currently clinically useful, meaning the information DTCGTs contain could enhance your level of patient care?

- ○ YES (1)
- ○ NO (2)

Skip To: Q9 If In general, do you think DTCGTs are currently clinically useful, meaning the information DTCGTs c... = NO

In your opinion, how clinically useful is DTCGT?

	Not at all useful (1)	Slightly useful (2)	Very useful (3)	Extremely useful (4)
DTCGT is (1)	○	○	○	○

Which of the following do you see as a clinical benefit of DTC genetic testing? Choose all that apply.

- ☐ Offering screening tests (e.g. EKG, mammograms, colonoscopies) MORE FREQUENTLY to individuals who are found to be at increased risk (1)
- ☐ Offering screening tests (e.g. EKG, mammograms, colonoscopies) at an EARLIER AGE to individuals who are found to be at increased risk (2)
- ☐ Selecting a medication to prescribe (3)
- ☐ Adjusting medication doses (4)
- ☐ Recommending lifestyle changes based on risk predictions (5)
- ☐ Changing the frequency of follow-up appointments (6)
- ☐ Making a diagnosis (7)
- ☐ Providing genetic testing in a more private, confidential manner (8)

Skip To: Q6 If Condition: Selected Count Is Greater Than or Equal to 1. Skip To: If a patient were to bring their DTCG....

Why do you feel DTCGTs are not clinically useful? Choose all that apply.

- ☐ It is too difficult to interpret what the results mean regarding patient care (1)
- ☐ I would not change a patient's management based on DTC testing (2)
- ☐ It will cause more patient anxiety (3)
- ☐ No guidelines exist to reduce or alleviate the risk for many diseases (4)
- ☐ I don't trust the clinical certification of an unknown genetic testing facility. (6)
- ☐ Other (5) _____

If a patient were to bring their DTCGT results to discuss potential health risks with you during an office visit, how likely is it that those test results would influence how you care for that patient?

	Extremely unlikely (1)	Somewhat unlikely (2)	Somewhat likely (3)	Extremely likely (4)
Influence would be (1)	○	○	○	○

Would you like to learn more about DTC genetic testing?

- ○ YES (1)
- ○ NO (2)

Skip To: Q10 If Would you like to learn more about DTC genetic testing? = NO

What would you like to learn about DTC genetic testing? Choose all that apply.

- ☐ The various tests being offered (1)
- ☐ The different testing companies (2)
- ☐ Support services offered by the companies (3)
- ☐ How to interpret the test results (4)
- ☐ Where to find credible information about these tests and what the results mean (5)
- ☐ Whether there are evidence-based risk management guidelines for people at risk for specific diseases (6)
- ☐ What resources are available for patients who are thinking about pursuing DTC genetic testing (7)

Skip To: End of Survey If Condition: Selected Count Is Greater Than or Equal to 1. Skip To: End of Survey.

Why are you not interested in learning more? Choose all that apply.

- ☐ I do not think that my patients will be asking about DTC genetic testing (1)
- ☐ I do not have time to learn more about DTC genetic testing (2)
- ☐ I will learn more about DTC genetic testing if it comes up in a patient visit (3)
- ☐ I am not interested in DTC genetic testing (4)

End of Block: Default Question Block

Appendix B

IRB Documents for Attitude Survey

Informed Consent

Information about Being in a Research Study
Clemson University

Osteopathic Medical Students' Attitudes About the Clinical Application of Direct-to-Consumer Genetic Test Results

KEY INFORMATION ABOUT THE RESEARCH STUDY

Kanesha Glenn, PhD(c), MS is inviting you to volunteer for a research study. Kanesha Glenn is a graduate student at Clemson University conducting the study with Linda D. Ward, PhD, APN, FNP-C in the Clemson University School of Nursing

Study Purpose: The purpose of this research is to examine Edward Via College of Osteopathic Medicine students' knowledge, attitude, and perception of the clinical use of direct-to-consumer genetic testing.

Voluntary Consent: Participation is voluntary and you have the option not to participate. Declining to participate will not affect your grade.

Activities and Procedures: Your part in the study will be to complete a survey about your current genetic/genomic knowledge and perceptions about the clinical usage of direct-to-consumer genetic testing.

Participation Time: It will take you about 5 minutes to complete this survey.

Risks and Discomforts: There are no known risks involved with participation in this study.

Possible Benefits: You may not benefit directly from taking part in this study, however the researchers hope the results of this study can provide useful information for educators working with future osteopathic medical students to create an effective learning tool for medical education.

INCLUSION REQUIREMENTS: In order to participate in this study, you must be an osteopathic medical student currently at Edward Via College of Osteopathic Medicine's Carolina Campus. If you are not an enrolled student, you are not eligible to participate in this study.

PROTECTION OF PRIVACY AND CONFIDENTIALITY: The results of this study may be published in scientific journals, professional publications, or educational presentations, but all data will be presented in aggregate. All responses will be recorded anonymously and the researchers will do all that they can to maintain privacy and confidentiality. Responses will be collected via Qualtrics, which has been set to not record any personal information from participants. Data will be stored in Qualtrics and in a Google Drive folder only accessible to Linda Ward and Kanesha Glenn.

The information collected during the study could be used for future research studies or distributed to another investigator for future research studies without additional informed consent from the participants or legally authorized representative. No identifiable information will be collected during the study or on the research study instruments.

CONTACT INFORMATION: If you have any questions or concerns about your rights in this research study, please contact the Clemson University Office of Research Compliance (ORC) at 864-656-0636 or irb@clemson.edu. The Clemson IRB will not be able to answer some study-specific questions. However, you may contact the Clemson IRB if the research staff cannot be reached or if you wish to speak with someone other than the research staff.

If you have any study related questions or if any problems arise, please contact Kanesha Glenn at 864-327-9857or kaneshn@g.clemson.edu.

CONSENT: By participating in the study, you indicate that you have read the information written above, been allowed to ask any questions, and you are voluntarily choosing to take part in this research. You do not give up any legal rights by taking part in this research study.

Email Invitation to Participate in Survey

As a student enrolled at Edward Via College of Osteopathic Medicine, you have been identified as a potential participant in a research study about direct-to-consumer genetic testing.

This study is being conducted by Ms. Kanesha Glenn, a PhD candidate at Clemson University and Instructor at Edward Via College of Osteopathic Medicine, and Dr. Linda Ward, an Associate Professor at Clemson University. The purpose of the study is to investigate OMS knowledge, attitude, and perceptions of the clinical use of direct-to-consumer genetic testing. Data collection will occur using an online survey that should take you approximately 5 minutes to complete. Eligibility to participate in this survey is restricted to enrolled osteopathic medical students at Edward Via College of Osteopathic Medicine's Carolinas Campus. There are no rewards for participating in this survey, and there are also no risks. The results of this survey will be used for future conference, journal publications, and will be used to create an evidence-based learning tool for medical education. No personally identifiable information will be collected.

To participate, please click on the link to the Qualtrics survey below, where you will find additional information on consenting to participate and can complete the study. If you have any questions about participating in this study, please contact Kanesha Glenn (kaneshn@g.clemson.edu) or Linda Ward (ldward@clemson.edu). This survey will close on Friday, February 3, 2022.

IRB Approvals

Division of RESEARCH
Research Compliance

To: Linda D Ward
Re: Clemson IRB Number: IRB2022-0587
Review Level: Exempt
Review Category: D2
Determination Date From: 09-Nov-2022
Determination Date To: 30-Nov-2025
Funding Sponsor: N/A
Project Title: Osteopathic students' attitudes about the clinical application of direct-to-consumer genetic test results

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The Office of Research Compliance determined that the proposed activities involving human participants meet the criteria for Exempt level review under 45 CFR 46.104(d). The Exempt determination is granted for the certification period indicated above.

Principal Investigator (PI) Responsibilities: The PI assumes the responsibilities for the protection of human subjects as outlined in the Principal Investigator's Responsibilities guidance.

Non-Clemson Affiliated Collaborators: The Exempt determination only covers Clemson affiliated personnel on the study. External collaborators have to consult with their respective institution's IRB office to determine what is required for their role on the project. Clemson IRB office does not enter into an IRB Authorization Agreement (reliance agreement) for Exempt level reviews.

Modifications: An Amendment is required for substantial changes to the study. Substantial changes are modifications that may affect the Exempt determination (i.e., changing from Exempt to Expedited or Full Board review level, changing exempt category) or that may change the focus of the study, such as a change in hypothesis or study design. **All changes must be reviewed by the IRB office prior to implementation.**

PI or Essential Study Personnel Changes: For Exempt determinations, submit an amendment ONLY if the PI changes or if there is a change to an essential study team member. An essential team member would be an individual required to be on the study team for their expertise or certification (i.e., health expert, mental health counselor). Students or other non-essential study personnel changes DO NOT have to be reported to the IRB office.

Reportable Events: Notify the IRB office within three (3) business days if there are any unanticipated problems involving risk to subjects, complications, adverse events, complaints from research participants and/or incidents of non-compliance with the IRB approved protocol. Incidents may be reported through the IRB online submission system using the Reportable Incidents eform or by contacting the IRB office.

Closing IRB Record: Submit a Progress Report to close the IRB record. An IRB record may be closed when all research activities are completed. Research activities include, but are not limited to: enrolling new participants; interaction with participants (online or in-person); collecting prospective data, including de-identified data through a survey; obtaining, accessing, and/or generating identifiable private information about a living person.

New IRB Application: A new Exempt application is required if the research activities continue for more than 3 years after the initial determination. **Exempt determinations may not be renewed or extended and are valid for 3 years only.**

Non-Clemson Affiliated Sites: A site letter is required for off-campus non-public sites. Refer to the guidance on research site/permission letters for more information. Submit the Amendment eform to add additional sites to the study.

International Research: Clemson's approval is based on U.S. human subjects protections regulations and Clemson University human subjects protection policies. Researchers should become familiar with all pertinent information about local human subjects protection regulations and requirements when conducting research internationally. We encourage you to discuss any possible human subjects research requirements that are specific to your research site with your local contacts, to comply with those requirements, and to inform Clemson's IRB office of those requirements. Review the FAQs for more information about international research.

Contact Information: Please contact the IRB office at IRB@clemson.edu or visit our webpage if you have questions.

Clemson University's IRB is committed to facilitating ethical research and protecting the rights of human subjects. All research involving human participants must maintain an ethically appropriate standard, which serves to protect the rights and welfare of the participants.

Institutional Review Board
Office of Research Compliance
Clemson University

IRB Number: IRB00000481
FWA Number: FWA00004497

Institutional Review Board

Edward Via College of Osteopathic Medicine

DATE:	December 9, 2022
TO:	Kanesha Glenn, MS, PhD(c)
FROM:	Edward Via College of Osteopathic Medicine Institutional Review Board
PROJECT TITLE:	[1987268-1] Osteopathic Medical Students' Attitudes About the Clinical Application of Direct-to-Consumer Genetic Test Results
VCOM IRB RECORD #:	2022-107
SUBMISSION TYPE:	New Project
ACTION:	DETERMINATION OF EXEMPT STATUS
APPROVAL DATE:	December 9, 2022
REPORT DUE:	December 9, 2024
REVIEW CATEGORY:	Exemption category #2

Thank you for submitting your proposal to the Edward Via College of Osteopathic Medicine Institutional Review Board. The proposed research has been subject to exempt review according to the specifications authorized by 45CFR 46.101 and 21 CFR 56.110. The Edward Via College of Osteopathic Medicine Institutional Review Board has deemed your project **exempt**.

As your protocol is exempt, there is no required continuing review date, however, the VCOM IRB requires a biennial Status Report to be submitted for review. Please use Form D for this procedure. Your next Status Report must be received by the report due date of **December 9, 2024**. If the study concludes prior to the date of the next Status Report, please report the study as closed using Form D.

Should your protocol change, you (as the PI) are responsible for reporting proposed changes to the IRB **before their implementation**. All modifications must be reviewed and approved by the IRB before they may be enacted. Please use the appropriate Modification Request (Form G) for this procedure.

Please retain a copy of this correspondence with your records.

If you have any questions, please contact W. Eryn Perry at eperry@vcom.edu. Please include your project title and VCOM IRB Record # in all correspondence with this committee.

This letter has been issued in accordance with all applicable regulations, and a copy is retained within Edward Via College of Osteopathic Medicine Institutional Review Board's records.

CHAPTER FOUR

NAVIGATING DIRECT-TO-CONSUMER GENETIC TESTING: AN INTERACTIVE MODULE ON HOW TO APPROACH PATIENTS' QUESTIONS AND CONCERNS

Kanesha N. Glenn, MS, PhD(c)

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ABSTRACT

Background

Direct-to-consumer genetic testing has become a common method for the average person to learn more about their personal genetic make-up. The testing companies that offer these genetic tests ask their customers to discuss any concerns about their results with their personal physician, but these physicians are not usually genetic professionals and may have less than minimal genetic training about direct-to-consumer genetic testing. Currently, medical students receive basic genetic training in their didactic years. This training includes how to initiate clinical genetic testing, but students may not receive training on how to advise patients on the results of direct-to-consumer genetic testing.

Objective

This project has two purposes. First, to assess the effectiveness of an interactive module on medical students' knowledge. Two, to assess the adeptness of medical students to manage clinical scenarios involving direct-to-consumer genetic test results.

Methods

This pilot study was conducted on November 14th and 16th, 2024 at Edward Via College of Osteopathic Medicine - Carolinas Campus. First and second-year students were given an online pre-survey consisting of one knowledge-based question about genomic vocabulary and three patient scenarios. The scenarios assessed the students' choices about giving clinical advice to a patient with specific genetic questions. Next, students followed a link to a short training module that covered the pros and cons of direct-to-consumer genetic testing, pertinent vocabulary, and interactive patient scenarios. Then, the students were routed to an online post-survey given to evaluate changes in knowledge and assess how the students chose to handle the patient

interactions. The study finalized with a focus group discussion about the module and its effectiveness.

Results

Out of the 327 students invited, 17 (5%) chose to participate. Each of those 17 students was given three patient scenarios to evaluate. Before completing the training module, 11 out of 51 (22%) total responses indicated a referral to a genetic counselor would be necessary. After the educational module, the referral responses decreased to 1 out of 51 (2%) total responses. Each student was also given 5 genetic vocabulary words to match to the correct definition. Genetic vocabulary knowledge improved from 69 of 85 (81%) total answers to 81 out of 85 (95.3%) total answers. In the focus group, students provided positive feedback regarding the usefulness of the modules and expressed interest in future topics.

Future Implications Conclusion

This interactive module can be an effective way to increase students' knowledge and understanding of how to navigate clinical situations involving direct-to-consumer genetic test results and can be used to train other health professionals on pertinent patient related topics.

INTRODUCTION

As clinical genomic technologies create a clearer path to precision medicine, medical educators are challenged to narrow the informational learning gaps between bench and bedside.[1] With the introduction of direct-to-consumer genetic testing, medical educators now have to bridge a gap between patient and provider. A 2015 study of medical school genetic course directors in the United States (US) and Canada found that when genetics courses are taught in medical school, the most common teaching method is a lecture-based course in the first year of the curriculum, with integration into additional courses such as biochemistry or nutrition.[2] In cases where genomics training is not integrated into medical school curriculum, students may lack time and resources to obtain training due to competing priorities. This project has two purposes. First, to assess the effectiveness of an interactive module on medical students' knowledge. Second, to assess the adeptness of the students in handling clinical scenarios involving direct-to-consumer genetic test results.

Background

In parallel with the rapidly evolving sector of genomics and personalized medicine, direct-to-consumer genetic testing (DTCGT) has also expanded over the last several years. In 1996, genetic testing for limited purposes was first marketed to consumers through newspaper ads and mail orders in the United Kingdom and US. Since the initial inception of DTCGT, companies have increased marketing to consumers. In 2017, the US Food and Drug Administration (USFDA) approved DTCGT for genetic risk testing. By 2018, an estimated 26 million consumers had sent genetic samples in to be processed for health and/or ancestry results.[3] Genetic testing results are typically delivered to the consumer by email, with directions to

contact their personal healthcare professional (if they have one) for questions pertaining to the results.

As individuals who are motivated to maximize their health outcomes continue to utilize DTCGT, it is important for healthcare providers to be prepared to answer questions, and to be familiar with the benefits, risks, and limitations of DTCGT. One challenge is that medical professionals often feel unprepared to discuss DTCGT results with their patients due to a lack of genomic education.[4,5,6] However, in recent surveys, most indicated they were willing to learn more about genomics if it was clinically beneficial and would easily fit into their limited amount of time with patients.[7,8]

The Association of Professors of Human and Medical Genetics, Accreditation Council for Graduate Medical Education, and American Academy of Family Physicians all have guidelines on which genetic topics should be prominent in the curriculum of medical professionals, although little attention has been directed toward DTCGT and how to handle clinical discussions about indications and results with patients.[9,10,11] To effectively integrate genomics training into a busy curriculum, online training for DTCGT with a brief online module may prove to be a viable and effective alternative delivery modality.

METHODS

A quasi-experimental survey design was selected to test the effectiveness of an educational module in improving student knowledge and ability to manage patient scenarios. The study was conducted at Edward Via College of Osteopathic Medicine (VCOM) - Carolinas campus. Study students were first and second-year osteopathic medical students who were recruited to participate through campus email and video flyers posted on campus informational television screens. Participation included both an online and in-person portion, both of which were

completed on campus, outside of class time, and divided into two groups of students. Consent was obtained prior to beginning the survey and subjects were assured that their responses were confidential and de-identified. VCOM was not informed who either did or did not participate. Both Clemson University's and VCOM's Institutional Review Boards granted this study exempt status (IRB #2023-0384 and 2101012-1 respectively, see Appendix E).

This study was created using the Reporting Item Standards for Education and its Evaluation in Genomics (RISE2 Genomics).[8] RISE2 Genomics is a tool that was developed in 2020 to formalize the creation and evaluation of genomic education interventions. The guidelines in RISE2 Genomics outline an intervention from design and development through evaluation impact of the intervention.

Students met on November 14th and 16th, 2023, in a lecture hall outside of class time and were given a link to an online consent form. Participation was voluntary, and there were no incentives for participation provided. Once consent was obtained, students were automatically routed to the pre-survey, a short microlearning module, and a post-survey. The pre and post-surveys consisted of four questions and were conducted via Qualtrics® survey software (see supplemental file 1). The first part of the survey assessed student actions toward three initial patient scenarios. The first scenario involved a patient with no findings of breast cancer variants on her DTCGT results even though she lost three family members to the disease. The second scenario dealt with an adult adoptee with no family history who wanted to know if they should take a DTCGT. The third scenario is a patient whose DTCGT results uncovered an Alzheimer's genetic variant, even though no one in his family had the disease (see Appendix C).

The second part of the survey assessed knowledge of basic terminology pertaining to genetic risk factors. The terms that the authors selected to test the student's knowledge are found on DTCGT

results, so it is important that physicians are familiar with the terms. The section consisted of matching definitions with the following terms: *absolute risk*, *risk factor*, *predisposition*, *polygenic risk score*, and *relative risk.*

The online microlearning course "Navigating Direct-to-Consumer Genetic Testing: How to Approach Patients' Questions and Concerns" was created using the Articulate360® Rise360 e-learning development platform in conjunction with the Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG) Point of Care Tool in development at the National Human Genome Research Institute. This Point of Care Tool is being created to help healthcare professionals handle patient questions about their DTCGT results.

The microlearning course is divided into three sections. The first section is knowledge of the benefits and limitations of DTCGT and defines pertinent genetic/genomic terms. The second section is a simulated interactive patient visit. In this part, how a student chooses to handle a situation will elicit a positive or negative response from the patient. The student must properly answer a patient's concern or start over from the initial question. Each of these patient scenarios has been chosen because they can be managed by a non-genetic healthcare professional, such as a general practitioner. The third section consists of available resources for provider use or to pass along to a patient in need. Below is a link to the microlearning course and it can be found in Appendix D. (https://rise.articulate.com/share/7vcUHKGngoA1rsZGKHSzD7b4dsqs_KLT).

After watching the microlearning course, the student was given the post-survey.

The online portion was followed by a guided group discussion facilitated by one of the research team members. Day 1 consisted of 4 students, and day 2 consisted of 13 students. No personally identifiable information was collected or retained.

RESULTS

Pre/Post-survey

Of the 327 students invited via email, 17 (5%) agreed to participate (13 OMS-I and 4 OMS-II). Participation time took an average of 22 minutes. From pre- to post-survey, there was a noticeable change in how students reacted to the patient scenarios. Each of the 17 student was given 3 scenarios to evaluate. Initially, 11 out of 51 (22%) of the students' total responses were to refer each patient to a genetic counselor. This number dropped to 1 out of 51 (2%) after going through the learning module. Many of the students stated in the post-survey discussion that explaining risks, benefits, and options for DTCGT makes for a better patient interaction.

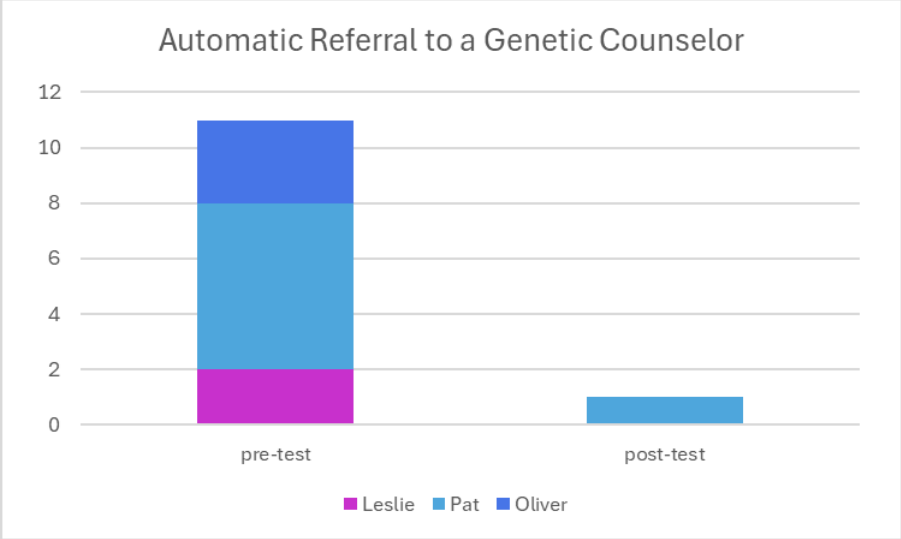

Figure 4.1. Number of students who would automatically refer each patient to a genetic counselor pre and post module.

The largest change was with the scenario where an adoptee asks for an opinion about using DTCGT to find unknown health issues. Pre-survey, only 9 (53%) students thought that they should explain that DTCGT will not give a complete view of genetic health, but it may find genetic relatives with more family history information. Post-survey, that number rose to 16 (94%), a 41% increase.

The vocabulary portion of the survey was a matching section where commonly anticipated terms were matched to their definition. The section consisted of 5 terms that patients may see on DTCGT results and will possibly have questions about. A McNemar's test was used to investigate whether the modest increase was statistically significant between pre and post-survey vocabulary scores. Pre-survey, 69 out of 85 (81%) of the total terms were matched correctly, post-survey, that number had risen to 81 (95%). The results were not significant.

X^2 (1) = 0.96, p = .33

Group Discussion

After the post-survey, a brief group discussion was held to discuss the module. The discussion themes were broken down into two main questions:

1. What do you believe are the pros and cons of this specific training module?
2. What are your feelings toward taking elective online training of this type in the future?

The students reported that they enjoyed the fact that the 15-minute training module was "concise" and "easy to digest," and the majority of the group liked that the module was interactive. One student stated, "I thought I knew the right thing until…I watched the video and then went through the module. After I went through the module where I talked to the patient, I kind of started to understand where they could be."

The main con brought up by students was the surveys and the module all had the same patient

scenarios. One student said this study design made it a conflict for a proper assessment because everything was the same, while others said it was hard to think about the scenarios differently so they were "picking the same things I did before," or "I just memorized 'ohh this is the right answer.'" A student mentioned that this module could have been improved if they could see what an actual DTCGT report would look like.

When asked about more online training similar to this module, the students all agreed that it would be a good resource as long as the topics were relevant to their needs and the modules were short enough to maintain their interest. One student stated that he/she would like to know more about the legal and ethical issues related to DTCGT while another said that this type of module would be great for learning about unusual clinical cases.

DISCUSSION

This study evaluated the implementation of an interactive online module among osteopathic medical students in the first two years of study. Results indicated that the module increased genetic vocabulary knowledge as well as the ability to manage patients with basic genetic questions. A 2016 study showed that simulation-based virtual learning in medical genetics can increase student confidence in patient relations.[12] This was mirrored in the fact that students in this study were more likely to choose to explain the risks and benefits of DTCGT to a patient after completing the module than to refer the patient to a genetic counselor. During the group discussion, the students found this method of training was best suited for short, clinical cases to be reviewed outside of instructional time. This module can be created and updated on Articulate 360 to stay current with the ever-changing landscape of genetic/genomic medicine.

Limitations and Future Implications

One limitation of this pilot study was the small number of students. This may be due to the study

being scheduled the day before a test. Recommendations for future implementation are to disseminate modules online for the students to take at their convenience and the group discussion changed to open-ended questions at the end of the post-survey.

CONCLUSION

Interactive online modules could be an effective method to increase osteopathic medical students' knowledge of DTCGT and ways to manage patients when they get results, and/or manage other genetic/genomic issues that may arise in a clinical visit.

REFERENCES

1. Campion M, Goldgar C, Hopkin RJ, Prows CA, Dasgupta S. Genomic education for the next generation of health-care providers. *Genetics in Medicine*. 2019;21(11):2422-2430.
2. Plunkett-Rondeau J, Hyland K, Dasgupta S. Training future physicians in the era of genomic medicine: Trends in undergraduate medical genetics education. *Genetics in Medicine*.2015;17(11):927-934. https://www.sciencedirect.com/science/article/pii/S1098360021031464. doi: 10.1038/gim.2014.208.
3. Ayoubieh H, Blazer K, Christopher D, et al. Direct-to-consumer genetic testing FAQ for healthcare professionals. genome.gov Web site.
4. https://www.genome.gov/For-Health-Professionals/Provider-Genomics-Education-Resources/Healthcare-Provider-Direct-to-Consumer-Genetic-Testing-FAQ. Updated 2023. Accessed 2/01/, 2024.
5. Powell KP, Cogswell WA, Christianson CA, et al. Primary care physicians' awareness, experience and opinions of direct-to-consumer genetic testing. *J Genet Couns*. 2012;21(1):113-126. http://libproxy.clemson.edu/login?url=http://search.ebscohost.com/login.aspx?direct=true&db=m nh&AN=21769569. doi: 10.1007/s10897-011-9390-9.
6. Mainous,Arch G.,,3rd, Johnson SP, Chirina S, Baker R. Academic family physicians' perception of genetic testing and integration into practice: A CERA study. *Fam Med*. 2013;45(4):257-262. http://libproxy.clemson.edu/login?url=http://search.ebscohost.com/login.aspx?direct=true&db= m nh&AN=23553089.

7. Powell KP, Christianson CA, Cogswell WA, et al. Educational needs of primary care physicians regarding direct-to-consumer genetic testing. *Journal of genetic counseling*. 2012;21(3):469-478.
8. Jonas MC, Suwannarat P, Burnett-Hartman A, et al. Physician experience with direct-to-consumer genetic testing in kaiser permanente. *Journal of personalized medicine* 2019;9(4):47.
9. Nisselle A, Janinski M, Martyn M, et al. Ensuring best practice in genomics education and evaluation: Reporting item standards for education and its evaluation in genomics (RISE2 genomics). *Genetics in medicine*. 2021;23(7):1356-1365.
10. Massingham LJ, Nuñez S, Bernstein JA, et al. 2022 Association of professors of human and medical genetics (APHMG) consensus–based update of the core competencies for undergraduate medical education in genetics and genomics. *Genetics in Medicine*. 2022;24(10):2167-2179.
11. Blitzer M, Cichon M, Cusmano-Ozog K, et al. ACGME medical genetics and genomics milestones .chrome-extension://efaidnbmnnnibpcajpcglclefindmkaj/https://www.acgme.org/globalassets/pdfs/milestones/medicalgeneticsmilestones.pdf. Updated 2019. Accessed 2/12/, 2024.
12. American Academy of Family Physicians. Recommended curriculum guidelines for family medicine residents medical genetics. *Recommended curriculum guidelines for family medicine residents–Medical genetics.2019*

13. McGrath SP, Walton N, Williams MS, Kim KK, Bastola K. Are providers prepared for genomic medicine: Interpretation of direct-to-consumer genetic testing (DTC-GT) results and genetic self-efficacy by medical professionals. *BMC health services research*. 2019;19(1):1-12
14. Makransky G, Bonde MT, Wulff JS, et al. Simulation based virtual learning environment in medical genetics counseling: An example of bridging the gap between theory and practice in medical education. *BMC medical education*. 2016;16(1):1-9.

Appendix C

Interactive Module Survey Questions

Leslie, 46, comes to you confused by her findings. The DTC-GT states that no BRCA variants were detected, yet two of her aunts (paternal) and a grandmother (maternal) died of breast cancer. She has been worried for years about developing this disease and wonders if this is a sign that she can ease her mind.

- ○ Immediately schedule her for a full BRCA screening
- ○ Go over her medical history and look for red flags
- ○ Explain to her the clinical benefits and limitations of DTC-GT
- ○ Discuss the choice of pre-emptive mastectomy
- ○ Refer her to a genetic counselor

Pat, 26, is an adoptee (closed adoption) and is concerned about unknown health issues. They are currently in good health and do not meet any criteria for clinical genetic screening through their insurance. They are considering DTC-GT to find potential elevated risks and/or relatives and they want to know your ideas on the matter.

- ○ Let Pat know that a DTC-GT won't give much more information than they already get through consistent health screenings
- ○ Explain to Pat that most DTC-GT will not give a complete view of genetic health, but finding relatives with more health information would definitely be helpful
- ○ Tell Pat that finding close relatives may be unlikely, but give them the names of a few CLIA certified/FDA approved labs.
- ○ Refer them to a genetic counselor

Oliver, 28, wants to know about ways to prevent or deter the effects of Alzheimer's disease. His DTC-GT detected 1 copy of the ε4 variant in the APOE gene. The test results say that he has an increased risk of developing late-onset Alzheimer's, but no one in his family has ever been diagnosed.

- ○ Refer him to a genetic counselor
- ○ Tell him that most people with one copy of this variant never get Alzheimer's disease, but discuss how lifestyle and environment contribute to the disease.
- ○ Even though there is no family history of dementia, order a clinical genetic test to be certain.
- ○ Explain the difference between an "increased risk" and a "confirmed diagnosis"

	predisposition	polygenic risk score	absolute risk	relative risk	risk factor
⊗The likelihood an individual will develop a disease over a specific amount of time.	○	○	○	○	○
⊗Any variable that increases an individual's chances of getting a disease.	○	○	○	○	○
⊗An increased chance that an individual will develop a disease based on their genetic makeup.	○	○	○	○	○
⊗An assessment of the risk of a specific condition based on the collective influence of many genetic variants.	○	○	○	○	○

⊗The likelihood an individual with a risk factor will develop a disease compared with an individual with a similar background without that risk factor.	○	○	○	○	○

Appendix D

Website Link to Interactive Module

https://rise.articulate.com/share/7vcUHKGngoA1rsZGKHSzD7b4dsqs_KLT

Appendix E

IRB Documents for Interactive Module Study

Informed Consent

Information about the Research Study
Clemson University

Navigating Direct-to-Consumer Genetic Testing:
How to Approach Patients' Questions and Concerns

KEY INFORMATION ABOUT THE RESEARCH STUDY

Kanesha Glenn, PhD(c), MS is inviting you to volunteer for a research study. Kanesha Glenn is a graduate student at Clemson University conducting the study with Kim A. Pickett, PhD, FNP-BC, BC-ADM in the Clemson University School of Nursing

Study Purpose: The purpose of this research is to highlight how to handle patient-initiated conversations about direct-to-consumer genetic test results, as well as ways to integrate these results into the patient's health plan.

Voluntary Consent: Participation is voluntary, and you have the option to not participate.

If you decide not to take part or to stop taking part in this study, it will not affect your grade in any way.

Activities and Procedures: Your part in the study will be to:

1. Watch a brief educational module about direct-to-consumer genetic test results.
2. Take a pre and post module test.
3. Participate in a group discussion about the module.

Some of the information shared during the group discussion may be personal, we ask that you respect others in the group and keep the information shared confidential. Please do

not share any information that may be sensitive or make you uncomfortable. You may refuse to answer or leave the discussion at any time if you become uncomfortable.

Participation Time: It will take you about 1 to 2 hours to be in this study.

Risks and Discomforts: We do not know of any risks or discomforts to you in this research study.

Possible Benefits: You may not benefit directly from taking part in this study, however the information gained will be of use to you in your future medical endeavors.

AUDIO/VIDEO RECORDING AND PHOTOGRAPHS

The post-module group discussion will be recorded. We will use Microsoft Word to record and transcribe all comments. Both will be stored on VCOM's secure OneDrive server. The recording will be stored for 30 days just to ensure that the transcription is accurate.

PROTECTION OF PRIVACY AND CONFIDENTIALITY

The results of this study may be published in scientific journals, professional publications, or educational presentations.

> The information collected during the study could be used for future research studies or distributed to another investigator for future research studies without additional informed consent from the participants or legally authorized representative. No identifiable information will be collected during the study or on the research study instruments.

CONTACT INFORMATION

If you have any questions or concerns about your rights in this research study, please contact the Clemson University Office of Research Compliance (ORC) at 864-656-0636 or irb@clemson.edu. The Clemson IRB will not be able to answer some study-specific questions. However, you may contact the Clemson IRB if the research staff cannot be reached or if you wish to speak with someone other than the research staff.

If you have any study related questions or if any problems arise, please contact Kanesha Glenn at 864-327-9857 or kaneshn@g.clemson.edu.

CONSENT

By participating in the study, you indicate that you have read the information written above, been allowed to ask any questions, and you are voluntarily choosing to take part in this research. You do not give up any legal rights by taking part in this research study.

Flyer Invitation to Participate in Study

Email Invitation to Participate in Study

As a student enrolled at Edward Via College of Osteopathic Medicine, you have been identified as a potential participant in a research study about direct-to-consumer genetic testing.

This study is being conducted by Ms. Kanesha Glenn, a PhD candidate at Clemson University and Instructor at Edward Via College of Osteopathic Medicine, and Dr. Kim Pickett, an Associate Professor at Clemson University.

The purpose of the study is to learn how to handle patient-initiated conversations about direct-to-consumer genetic test results as well as ways to integrate these results into the patient's health plan. Data collection will occur using an online module that should take you approximately 15 minutes to complete.

Eligibility to participate in this survey is restricted to enrolled osteopathic medical students at Edward Via College of Osteopathic Medicine's Carolinas Campus. There are no rewards for participating in this survey, and there are also no risks.

The results of this study will be used to create an evidence-based learning tool for medical education and may be used for future conference presentations or journal publications.

Participation in this survey is anonymous and no personally identifiable information will be collected.

To participate, please bring your laptop to (date/time/location TBA).

If you have any questions about participating in this study, please contact Kanesha Glenn (kaneshn@g.clemson.edu) or Kim Pickett (kpicket@clemson.edu).

IRB Approvals

Division of RESEARCH
Research Compliance

To: Kim Ann Pickett
Re: Clemson IRB Number: IRB2023-0384
Review Level: Exempt
Review Category: 1
Determination Date From: 25-Aug-2023
Determination Date To: 31-Aug-2026
Funding Sponsor: N/A
Project Title: Navigating Direct-to-Consumer Genetic Testing: How to Approach Patients' Questions and Concerns

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The Clemson University IRB office determined that the proposed activities involving human participants meet the criteria for Exempt level review under 45 CFR 46.104(d). The Exempt determination is granted for the certification period indicated above.

Principal Investigator (PI) Responsibilities: The PI assumes the responsibilities for the protection of human subjects as outlined in the Principal Investigator's Responsibilities guidance.

Non-Clemson Affiliated Collaborators: The Exempt determination only covers Clemson affiliated personnel on the study. External collaborators have to consult with their respective institution's IRB office to determine what is required for their role on the project. Clemson IRB office does not enter into an IRB Authorization Agreement (reliance agreement) for Exempt level reviews.

Modifications: An Amendment is required for substantial changes to the study. Substantial changes are modifications that may affect the Exempt determination (i.e., changing from Exempt to Expedited or Full Board review level, changing exempt category) or that may change the focus of the study, such as a change in hypothesis or study design. **All changes must be reviewed by the IRB office prior to implementation.** Instructions on how to submit an Amendment is available on the IRB webpage

PI or Essential Study Personnel Changes: For Exempt determinations, submit an amendment ONLY if the PI changes or if there is a change to an essential study team member. An essential team member would be an individual required to be on the study team for their expertise or certification (i.e., health expert, mental health counselor). Students or other non-essential study personnel changes DO NOT have to be reported to the IRB office.

Reportable Incidents: Notify the IRB office within three (3) business days if there are any unanticipated problems involving risk to subjects, complications, adverse events, complaints from research participants and/or incidents of non-compliance with the IRB approved protocol. Incidents may be reported through the IRB online submission system using the Reportable Incidents eform or by contacting the IRB office. Review the IRB policies webpage for more information.

Closing IRB Record: Submit a Progress Report to close the IRB record. An IRB record may be closed when all research activities are completed. Research activities include, but are not limited to: enrolling new participants; interaction with participants (online or in-person); collecting prospective data, including de-identified data through a survey; obtaining, accessing, and/or generating identifiable private information about a living person.

New IRB Application: A new Exempt application is required if the research activities continue for more than 3 years after the initial determination. **Exempt determinations may not be renewed or extended and are valid for 3 years only.**

Non-Clemson Affiliated Sites: A site letter is required for off-campus non-public sites. Refer to the guidance on research site/permission letters for more information. Submit the Amendment eform to add additional sites to the study.

International Research: Clemson's determination is based on U.S. human subjects protections regulations and Clemson University human subjects protection policies. Researchers should become familiar with all pertinent information about local human subjects protection regulations and requirements when conducting research internationally. We encourage you to discuss any possible human subjects research requirements that are specific to your research site with your local contacts, to comply with those requirements, and to inform Clemson's IRB office of those requirements. Review the FAQs for more information about international research.

Contact Information: Please contact the IRB office at IRB@clemson.edu or visit our webpage if you have questions.

Clemson University's IRB is committed to facilitating ethical research and protecting the rights of human subjects. All research involving human participants must maintain an ethically appropriate standard, which serves to protect the rights and welfare of the participants.

Institutional Review Board
Office of Research Compliance
Clemson University

IRB Number: IRB00000481
FWA Number: FWA00004497

Edward Via College of Osteopathic Medicine
VIRGINIA • CAROLINAS
AUBURN • LOUISIANA

Institutional Review Board
Edward Via College of Osteopathic Medicine

DATE:	October 18, 2023
TO:	Kanesha Glenn, MS, PhD(c)
FROM:	Edward Via College of Osteopathic Medicine Institutional Review Board
PROJECT TITLE:	[2101012-1] Navigating Direct-to-Consumer Genetic Testing: How to Approach Patients' Questions and Concerns
VCOM IRB RECORD #:	2023-165
SUBMISSION TYPE:	New Project
ACTION:	DEFERRED
APPROVAL DATE:	October 18, 2023
REPORT DUE:	October 18, 2025
REVIEW CATEGORY:	Administrative Review & Deferral

Thank you for submitting your proposal to the Edward Via College of Osteopathic Medicine (VCOM) Institutional Review Board (IRB). VCOM IRB has has elected to cede oversight for this exempt project to the Clemson University IRB.

You (as the PI) are responsible for following guidance set forth by the Clemson IRB. Should your protocol change, you (as the PI) are responsible for reporting proposed changes to the Clemson University IRB **before their implementation**. All modifications must be reviewed and approved by the Clemson University IRB before they may be enacted. You are respsonsible for reporting changes made to the approved Clemson protocol to the VCOM IRB within 14 days. Additionally, you are responsible for forwarding all reports (e.g., progress reports, adverse event reports, closure reports, etc.) and additional Clemson IRB determination letters to the VCOM IRB in a timely manner.

Please retain a copy of this correspondence with your records.

If you have any questions, please contact Katherine Hanson at kbaumgarner@vcom.edu. Please include your project title and VCOM IRB Record # in all correspondence with this committee.

This letter has been issued in accordance with all applicable regulations, and a copy is retained within Edward Via College of Osteopathic Medicine Institutional Review Board's records.

CHAPTER FIVE

CONCLUSION

The Association of Professors of Human and Medical Genetics and the National Coalition for Health Professional Education in Genetics have genetic competency guidelines for medical schools to integrate into their curriculum, but each of these deals with physician initiated testing.[1,2] In order to keep up with the growing genetic education of the public, up primary care providers need to know how to offer communication and patient care for a situation that is becoming more common. The American Academy of Family Physicians' guidelines for residents has one medical genetic skill stating that a physician should be able to "educate patients about DTC genome-wide association study testing as a risk-stratification strategy, its benefits, and its risks." [3] With the ever-growing amount of genetic information loaded crammed into the medical school curriculum, this dissertation explored a way to create an alternate educational training module that teaches the knowledge and communication skills needed to educate those patients.

Chapter I provides a brief history of DTCGT and the stakeholders involved in its continued use (testing companies, FDA, customers/patients, and healthcare professionals). This chapter also brings to light the issue that patients are becoming more genetically educated, so healthcare professionals need to maintain a level of genetic literacy. This dissertation had three aims. The first one was to uncover concerns that healthcare professionals reported with integrating DTCGT into clinical care. This was addressed in the systematic literature review provided in Chapter II. The second aim was to assess the attitudes of medical students about the clinical application of DTCGT results

and was addressed in the survey described in Chapter III. The final aim was to assess the efficacy of an online module created to train medical students how to handle discussions about DTCGT results with a patient and is provided in Chapter IV.

Chapter II

Chapter II, "Obstacles to Integration of Direct-to-Consumer Genetic Test Results Into Patient Care," was a systematic literature review about healthcare professionals' knowledge and attitudes about DTCGT and its clinical utility. This chapter used John Venkatesh et al.'s Unified Theory of Acceptance and Use of Technology (UTAUT) to explain the factors used to accept DTCGT as a clinical tool: performance expectancy (how well it will improve job performance), effort expectancy (ease of use), social influence (perception that important others use the system), and facilitating conditions (how much support exists to help use the system).[4] These four constructs brought our four themes in the literature that explain the barriers to accepting DTCGT: knowledge, clinical utility, legal and ethical issues, and lack of practice recommendations.

Limitations

There was a paucity of literature about DTCGT and primary care provider interactions, especially from the perspective of the healthcare professional. This may be due to the fact that most of the articles were geared towards genetic professionals and genetic counselors.

Recommendations

Among the articles found, there were a few recommendations about what is needed to bridge the educational gap for providers, but none were about actually creating a solution. Educational interventions need to be developed to address this knowledge gap taking into consideration a healthcare provider's limited time availability.

Future Research

More data is needed on the outcomes of patient/provider collaborations and the incorporation of DTCGT test results into patient electronic health records. An assessment needs to be done among medical students as they would be considered early adopters of new genomic innovations. This assessment should include how medical students view the clinical validity of DTCGT as well as their confidence in patient interactions concerning DTCGT results. Findings from this paper served as the basis for the next two research studies (Chapter III and Chapter IV).

Chapter III

Chapter III, "Osteopathic Medical Students' Attitudes About the Clinical Application of Direct-to-Consumer Genetic Test Results," was the development and assessment of an online survey that used a modified version of the UTAUT model format to measure student knowledge, performance expectations, perceived clinical utility, and effort expectations in relation to possible patient interactions concerning DTCGT results. This survey was emailed to all students enrolled at Edward Via College of Osteopathic Medicine (VCOM) - Carolinas Campus. These students were comfortable with their

current genetic/genomic knowledge and many saw positive clinical usefulness of DTCGT. However, the students showed low confidence in their ability to discuss DTCGT results with patients, preferring to leave that task to genetic counselors. Even though there was a lack of confidence, there was a desire to learn more about how to integrate patient-initiated testing into clinical care.

Limitations

The main limitation of this study was that it was only done at one of VCOM's four campuses. Expanding this study could have given a much larger sample size.

Recommendations

This study could have also been altered to include other healthcare professionals such as nurse practitioners and physician assistants to give a more comprehensive idea of how DTCGT is viewed in the medical field.

Future Research

This study points to the need for a standardized educational intervention on DTCGT that capitalizes off of medical students' eagerness to learn about prospective technologies. The Reporting Items Standards for Education and its Evaluation in Genomics (RISE2 Genomics) is a standardized procedure that describes how to plan and report educational interventions in genomics.[5] Information garnered from this study was used to inform the creation of the module in Chapter IV.

Chapter IV

Chapter IV, "Navigating Direct-to-Consumer Genetic Testing: How to Approach Patients' Questions and Concerns," is a brief report on the efficacy of an educational module that trains medical professionals how to handle patient interactions concerning DTCGT. This module was created using the RISE2 Genomics guidelines and was evaluated through a pre-survey, post-survey, and a focus group. Students in their first two years at VCOM - Carolinas Campus were invited to attend and were given a website link to begin the study. The module contained interactive elements that allowed the students to receive feedback from the patient throughout the clinical scenario. The focus group revealed that students underestimated their ability to handle the scenarios, but they would like to have firmly established guidelines to follow in such situations. Analysis of the surveys showed the module did enhance vocabulary and changed how they would initially respond to the virtual patients.

Limitations

Even though video flyers invitations were posted on television screens around the VCOM campus in addition to the email invitation to combat what was deemed as a low response to the previous survey, only seventeen students accepted the invitation to participate. This may have been due to the study being done at the end of the day one day before a test for the OMS I students and the misconception that a study of this type would take up too much time.

Recommendations

This study could have been given to all four of VCOM's campuses during the semester of the actual genetics course. Scheduling the study to take place at the end of class time or allowing the students to take the study on their own free time may garner a greater number of test subjects. Focus group questions can be changed to open-ended response questions on the post-survey.

Future Research

This module could be a useful tool to use as a curriculum supplement for any medical genetic program. Institutions could also modify this module to be utilized for a variety of genetic topics in the medical curriculum or continuing medical education for current professionals.

Conclusion

As of October 2023, one of the biggest fears of DTCGT consumers and potential consumers came to fruition. The company 23andMe had its genomic database infiltrated by hackers.[6] Media outlets have posted that hackers could use this information to blackmail people with genetic family secrets to hide or to create bioengineered weapons based on genomic data. Some have even gone as far as to say that the stolen genetic data could be used to bypass biometric security systems,[7] however the technology has not been invented for that level of crime. Even as this company fails due the novelty of personal genetic control wears off[8] and the loss in confidence in 23andMe's ability to keep genetic information secure[7], genomic testing companies are going to do what they

are known to do: adapt. What started out as a way for physicians to test for specific SNPs turned into a way for everyone to learn about how their genes affect their lives and the lives of family members known and yet to be known.

Medical professionals need to keep up with their genetic/genomic literacy throughout their careers. This dissertation shows that educational modules are a viable option to easily disseminate information to medical professionals so that the conversations between patient and provider can remain positive and productive.

REFERENCES

1. Massingham LJ, Nuñez S, Bernstein JA, et al. 2022 Association of Professors of Human and Medical Genetics (APHMG) consensus–based update of the core competencies for undergraduate medical education in genetics and genomics. *Genet Med. 2022;24(10):2167-2179. doi:10.1016/j.gim.2022.07.014*

2. National Coalition for Health Professional Education in Genetics, (NCHPEG). Core competencies in genetics. The Jackson Laboratory Web site. https://www.jax.org/education-and-learning/clinical-and-continuing-education/ccep-non-cancer-resources/core-competencies-for-health-care-professionals. Updated 2007. Accessed Oct 3, 2020.

3. Recommended curriculum guidelines for family medicine residents/medical genetics. American Academy of Family Physicians Web site. https://www.aafp.org/dam/AAFP/documents/medical_education_residency/program_directors/Reprint258_Genetics.pdf. Updated 2016. Accessed Apr 28, 2019.

4. Venkatesh V, Sykes TA, Zhang X. 'Just what the doctor ordered': A revised UTAUT for EMR system adoption and use by doctors. . 2011:1-10.

5. Nisselle A, Janinski M, Martyn M, et al. Ensuring best practice in genomics education and evaluation: Reporting item standards for education and its evaluation in genomics (RISE2 genomics). Genetics in medicine. 2021;23(7):1356-1365.

6. Brown KV. 23andMe hack breaches 6.9 million users' info, including some's health data. TIME Web site. https://time.com/6342551/23andme-hack-health-data-profiles-compromised/. Updated 2023. Accessed Feb 29, 2024.

7. Battle P. Hackers could use your stolen 23andMe DNA data as a weapon. TheStreet Web site. https://www.thestreet.com/technology/hackers-could-use-your-stolen-dna-data-as-a-weapon. Updated 2023. Accessed Feb 29, 2024.

8. Winkler R. 23andMe's fall from $6 billion to nearly $0 --- once-hot DNA-testing company is struggling to pivot to healthcare. Wall Street Journal. 2024. Available from: https://www.proquest.com/newspapers/23andmes-fall-6-billion-nearly-0-once-hot-dna/docview/2920418709/se-2?accountid=6167.