Clinical genetic testing in medical education

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ABSTRACT

Although clinical geneticists have traditionally monopolized the administration of genetic tests, several genetic test companies have started producing affordable consumer tests for genetic screening and ancestry analysis. With genetic test kits readily available, patients are increasingly using personal genetic testing resources with the intention of sharing their results with their primary care physicians. Therefore, both medical professionals and patients need information and guidelines to interpret and act on personal genetic data. However, medical students, residents, and attending physicians have limited experience in handling the complexities involved in interpreting consumer genetic testing. Medical students, though, have shown significant interest in genetic testing and its application in their patients. Given the interest and need, medical education programs would best serve their students and patients through early education and training in clinical genetic testing.

KEYWORDS Genetics; medical education; personal and consumer genetic testing

Clinical geneticists remain integral in diagnosing, managing, and counseling patients and families on various genetic conditions.¹ Traditionally, clinical geneticists and other health care professionals have been the sole administrators of genetic tests. However, genetic testing companies, such as 23andMe (Mountain View, CA), have produced readily available and affordable personal genetic tests for genetic screening and ancestry analysis.² As a result, patients are increasingly using personal genetic testing and plan to share their genetic data with their primary care physicians.³ This is particularly true “when customers face ambiguous or alarming results needing expert interpretation, and they heed the timeless suggestion: ask your health care provider for more information.”⁴ In response, the genetic counselor workforce has rapidly expanded, encouraging further genetic testing and referrals from primary care physicians.^{5,6} Although physicians and residents are interested in personal genetic testing, most feel deficient in handling genetic data and assessing a patient’s health risks.^{7,8}

Most clinical geneticists spend two or more years in fellowship training and research mastering the essentials of genetic testing and counseling.⁹ According to the American Board of Medical Genetics and Genomics, clinical genetics requires “understanding genetic tests and results, ability to

develop differential diagnoses for constitutional and somatic genetic disorders, and effective communication, management, and professional skills.”⁹ Additionally, the interpretation of genetic data requires consideration of both the context and application of a specific genetic test and communication between various medical specialties.¹⁰ Most primary care physicians and specialists are either unable or unwilling to pursue additional postgraduate education in clinical genetic testing.^{9,11} Therefore, there is a concern in the medical community about providing medical staff and patients with the resources needed to interpret and act on personal genetic data.^{12,13} This concern is compounded by the shortage of clinical geneticists. According to the American Medical Association, “58% of clinical genetics GME [graduate medical education] slots are unfilled. 17 states currently have shortages and the 5 to 15 year forecast predicts more shortages.”¹⁴ The clinical genetics workforce also faces several supply and demand challenges: “On the supply side, the profession [clinical genetics] faces challenges with the three Rs—recruitment, retention, and retirements. On the demand side, the convergence of profound shifts in population demographics, legislative and regulatory reforms, and major advances in medical diagnostics and treatments will be significant influences.”¹⁵ In response to these shortages, several authors have proposed initiatives to increase

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laboratory professional outreach and education programs, legislative proposals, and collaborative efforts between clinical geneticists and other clinical departments.¹⁵

Most medical students and residents do not have the skills required to interpret and manage personal genetic testing provided by patients.⁴ One study reported that only 11% of US and Canadian medical schools incorporate clinical genetics training in their curricula.¹⁶ Despite this gap, medical students have shown interest in genetic testing and have a strong interest in learning to handle this information for their patients.¹⁷ Several studies showed that improvements in physician understanding of genetic data correlated with improved patient–physician communication and interaction.^{10,18} Therefore, early medical education on basic clinical genetics could impact patient outcomes.⁴ “The most basic skill with which every medical student should be invested is how to ... assess genetic risk ... [and] learn how to synthesize this information to determine the clinical contexts where genetic/genomic testing or referral to a genetic counselor is warranted.”⁴ Medical education programs would best serve their students and patients through early educational exposures in clinical genetic testing and counseling.

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