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A Brief Study on Medical Genetics Teaching For Paediatric Residents

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Abstract

Genetic testing is widely used in medicine and is frequently requested or ordered by patients, non-genetics subspecialists, and primary care physicians. Other research has demonstrated that ordering genetic testing is frequently uncomfortable for clinicians. A consistent training programme for teaching resident physicians about genetic testing does not yet exist, despite efforts to teach these ideas through continuing medical education. Methods: From September to October 2020, we used email to find every paediatrics resident at our facility (N=102). Residents were invited to participate in a Qualtrics web survey that asked about their perceived level of awareness about the fundamental ideas behind genetic testing and their reported level of comfort discussing these ideas with their family. The percentage of respondents who said they were unprepared ranged from 28% (basic genetics principles) to 80%. The majority of paediatrics residents concurred that they would benefit from a course that covered the fundamentals of genetic testing.

Keywords: Medical genetics • Paediatric residents • Genomic

Introduction

There are numerous combined paediatrics residency training programmes at Indiana University, and these programmes have a long history of coordination, collaboration, and interdisciplinary teamwork. The Department of Medical and Molecular Genetics and the Department of Paediatrics at Indiana University School of Medicine collaborated to create the Paediatrics Medical Genetics Residency. A four-year combined paediatrics and medical genetics training programme is available through this programme. Participants receive access to cutting-edge genetics research possibilities in addition to their clinical training in paediatrics and medical genetics. Since the discovery of DNA in 1953 and the completion of the Human Genome Project in 2003, the discipline of genetics has made impressive strides. Due to this remarkable advancement, all healthcare professionals now need to be conversant in the fundamental concepts and practical applications of genetics. It is essential to start incorporating the subject into undergraduate medical education in order to ensure that doctors have a solid understanding of human genetics and are always up to date. This learning must continue throughout the additional training years in addition to the years spent as a practising physician due to the astounding rate of advancement in genetics [1].

Medical genetics, which is both a clinical specialty and a basic science, offers a distinctive viewpoint on how the human body functions in both health and disease. The teaching of medical genetics must last throughout the undergraduate medical school curriculum and into the graduate years. The programme must specifically cover medical genetics. Specific learning objectives in medical genetics must be created, even though some aspects of the subject are similar to and may be covered in other disciplines. Each medical school's genetics curriculum should be implemented by a highly trained medical genetics specialist, or by a small committee of medical geneticists. This duty should encompass participation in all genetics-related courses throughout the undergraduate medical programme [2].

In an era marked by a tremendous explosion of new medical knowledge, the traditional two-plus-two, largely lecture-based medical curriculum (two years of didactic basic science followed by two years of clinical immersion) was replaced by an understanding of the significance of educating physicians who are adaptable, life-long learners who have the skills and attitudes necessary to remain competent, and humanistic physicians. With this modification, the emphasis of medical education has shifted to teaching students "how to learn." The Double Helix Curriculum (DHC), which was created as a result of the curriculum reform, is so titled because it encompasses "intertwining strands" of basic and clinical scientific instruction over the course of four years [3].

Description

The actual number of students enrolled in these training programmes is dwarfed by the number of monogenetic health care professionals already working in the field, despite the fact that there have been a good number of studies. Each programme had between 10 and 710 "students," while 60% of them had fewer than 100 members. Our calculations indicate that less than 5,000 monogenetic health practitioners throughout a 16-year period. The influence of these factors on the success of the programmes cannot be ascertained because there are few statistics on the participants' age, ethnicity, or experience. Importantly, only around 30% of the studies investigated changes in clinical practise, and frequently, participant self-report was used. Beginning first-year students in their medical school are introduced to topics that are crucial to understanding genetics in the introductory course, Mastering Medical Information. In addition to learning about prevalent complicated conditions, the students in this epidemiology, biostatistics, evidence-based medicine, and clinical trial design course are taught the statistical concepts that serve as the basis for understanding population genetics and genetic screening. Similar to this, embryology is introduced for the first time in Human Structure and Function, a Year 1 Anatomy-Physiology course. The chi-squared approach is used to examine data on population and family genetics, Bayesian statistics are used to resolve genetic counselling issues, and normal distribution is used to explain common complex genetics. These topics are specifically covered during the Molecules-to-Cells course [4].

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The goal of the majority of these training initiatives is to expand access to care in the long run, but little is known about their costs or the degree of proficiency attained by their graduates. These articles do not discuss the expenses of the educational programmes anywhere. These expenses include those related to programme development and execution (equipment, travel, supplies, and lost clinical practise time), in-kind contributions, and costs related to programme evaluation. The description of participant incentives, which were offered in roughly half of the studies in the form of salary, coffee coupons, book vouchers, monetary compensation, training certificates, and continuing education units, was the sole reference to costs in this analysis [5].

The lack of genetic counsellors, clinical geneticists, and laboratory geneticists is one of the reasons why health care practitioners should focus more on genetics education. In 2004, a research from the United Kingdom indicated that 1 full-time equivalent clinical geneticist was required for every 250,000 people. 3 According to the American College of Medical Genetics and Genomics (ACMG), there is only one full-time equivalent for every 600,000 Americans, which is 2.5 times less than the country's anticipated requirement. The extraordinarily long wait times for a new appointment with a medical geneticist, up to a year or longer at some institutions, provide supporting evidence of this shortage (personal communication).

Conclusion

You have access to a wide choice of training possibilities on our lovely, academic children's hospital campus. Nationwide Children's is a national leader in creative approaches to population, school, and community health at one end of the child health care spectrum. At the Abigail Wexner Research Institute, we are developing life-saving treatments for single gene diseases including spinal muscular atrophy and muscular dystrophy. At the other, we are integrating next-generation genomics into paediatric medicine practise. Outstanding general paediatric experiences in our primary care practises and general hospital services form the basis of our residency training programme. 60% of our residents participate in nationally recognised clinical and research programmes for subspecialty training, which are run by committed mentors and faculty members who are eager lecturers.

Acknowledgement

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Conflict of Interest

None.

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