The Development and Evaluation of Online Podcast Modules as a Toolkit for Teaching Genetics and Genomics Competencies in Post-Graduate Medical Education

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Abstract

Background: The lack of comfort with core genetic and genomic competencies among medical trainees and physicians is a barrier to the implementation of precision medicine. To address this, we developed short online modules to promote genetic competencies for use post-graduate medical education.

Methods: The educational toolkit was delivered as short online podcasts accompanied by slides. Each core module is approximately 15-20 minutes, and covered basic genetics, genetic testing, counselling and consenting, and interpreting and delivering results. These were supplemented by case-based modules on cancer genetics, prenatal genetics and cardiogenetics. The modules had pre- and post-test multiple choice questions pertaining to genetic and genomic competencies, attitudes towards precision medicine, and perceived competence.

Results: Based on the pre- and post-test data, residents reported a discordance between how often they cared for patients with genetic disorders and their level of confidence with core genetic competencies. Post-module evaluations demonstrated a significant increase in confidence in interpreting a microarray, and basic genetics knowledge.

Conclusions: Our study demonstrates that podcast modules are an innovative method to promote genetic and genomic competencies to postgraduate medical trainees. Limitations to our study included a small sample size, and further work is needed identify and address barriers to implementation. We suggest that integration at the post-graduate medical education level will be crucial to further promoting the development of precision medicine competencies in medical trainees and physicians.

Introduction

One of the challenges in the implementation of precision medicine is the lack of existing genetic and genomic competencies amongst medical trainees and practicing physicians, which may be due in part to the rapid rate of advances in genomic technology with relatively slower implementation of genomic medicine (1). In order to integrate precision medicine into patient care and to effectively use and interpret genetic technologies for medical management, an understanding of the role and complexity of genetic, and in particular genomic, testing is required (2). Key competencies for physicians and trainees have been identified, which include confidence with core genetic and genomic competencies including a basic understanding of genes and inheritance, consent for testing, interpretation of test results, and application of this knowledge to clinical situations (3).

Educating the physician workforce in order to achieve these competencies is difficult due to competing curriculum priorities at the undergraduate medical education level (4). Once trainees have entered independent clinical practice, the opportunities to develop these competencies are limited and there are

again many competing priorities (5). Residency offers an ideal time to develop these competencies, as resident trainees may have a better appreciation of their utility as they gain further clinical exposure.

There is a paucity of medical literature pertaining to the teaching of precision medicine to medical residents. Much of the available literature focuses on the importance of preparing medical professionals for the precision medicine era (see systematic review by Talwar et al 2018) (6); however, very few papers present examples of educational programs or approaches to teaching this material. In most cases, the conclusions highlight the need for ensuring an educated work force, including physicians, nurses, bioinformaticians, pharmacists, and other members of the healthcare team. For example, a literature review and modified Delphi survey by Tognetto et al (2019) established key foundational components of genetics curricula, but specific steps for implementation and knowledge translation are typically not addressed (7).

McGrath *et al.* discuss the importance of medical curriculum reform, including making genetics a core competency in their 2016 article (8). They discuss the need for new continuing medical education (CME) courses, precision medicine certification, and proper staffing. Several groups have approached the teaching of precision medicine using experiential learning (9, 10). Medical students and residents were taught about genomic medicine using a hands on approach by analyzing their own genomes; however, the ethics behind this pedagogical method are debatable (11).

There are several online educational modules offered through various associations and organizations (12, 13). However, we were unable to find any examples of online learning modules teaching genetic competencies that have been evaluated to determine their effectiveness as an approach to imparting these skills. We undertook to develop a series of online precision medicine learning modules and to evaluate their effectiveness in teaching key concepts in Genetics and Genomics core competencies to non-genetics medical residents.

Study Design

Our group developed a series of online video podcasts to be used as an online toolkit to promote genetic and genomic competencies for use in the education of Postgraduate Medical trainees at the Cumming School of Medicine. The objective of the toolkit was to increase core genetic and genomic competencies, with the goal of helping to prepare non-genetics residents for clinical scenarios involving precision medicine. The group developing the podcasts was composed of residents in the Medical Genetics and Genomics residency program at the University of Calgary, Cumming School of Medicine, with oversight from one of the clinical staff. The study was open August 6, 2018 to April 21, 2020.

The toolkit is delivered as short online podcasts accompanied by slides, which provide a collection of clear, concise educational modules available to learners for their ongoing reference. Each core module is approximately 15-20 minutes, and covers basic concepts in genetics, genetic testing, counselling and consenting, and interpreting and delivering results. These are supplemented by shorter case-based modules. Development of the curriculum was based on genetic and genomic competencies deemed to be

important for a general practitioner and which fulfilled various aspects of the CanMEDS framework of essential physician abilities, outlined by the Royal College of Physicians and Surgeons of Canada (14) (Figure 1).

Participants were recruited through email or by attending the resident academic half days. The toolkit and evaluations are password-restricted to postgraduate medical trainees at the Cumming School of Medicine. The website was designed in a manner which did not force participants to complete the questions and participants could advance to the next module without completing the questionnaire. The responses were collected anonymously using Google forms.

We then evaluated the effectiveness of the online podcast modules as a technique to teach these competencies. Each module has pre- and post-module questions pertaining to genetic and genomic competencies, attitudes towards precision medicine and perceived competence. Questions were primarily multiple choice (for knowledge-based questions) or modified Likert scale (for confidence-based questions) (Figure 2). Full pre- and post-tests are available for review (**Supplementary Figures**). Analysis of responses was performed in aggregate.

Methods And Results

Demographics

A total of 166 pre-tests and 110 post-tests for Modules 1-4 were completed by residents in at least 8 disciplines (Figure 3). The majority of residents were from family medicine, pediatrics, and pediatric neurology, and most were in their first or second year of residency. Due to the website structure, residents who completed more than one module were counted independently for each module.

Level of Exposure to Genetics in a Clinical Setting

Participants were asked two questions to gauge their level of exposure to genetics in a clinical setting: "How often do you care for patients with genetic disorders?" and "How often do you see the results of genetic tests?" for each pre-test. Responses were graded on a 5-point Likert scale. Due to an error, this information was not available for participants who completed Module 1. There were three individuals who answered the question "How often do you see the results of genetic tests?" but not "How often do you care for patients with genetic disorders?".

Responses from Modules 2-4 demonstrated that 24/104 (23%) of participants "Very often or Often" care for patients with genetic disorders, 37/104 (36%) "Sometimes" and 43/104 (41%) "Rarely or Never" (Figure 4). Surprisingly, only 12/107 (11%) of participants "Very often or Often" see the results of genetic testing, 23/107 (21%) "Sometimes" and 72/107 (68%) "Rarely or Never". Four of the respondents in pediatrics reported that they "Very often" care for patients with genetic disorders but "Never" see the results of genetic testing.

Level of Confidence with Medical Genetics and Genomics Competencies

Participants were asked two questions to gauge their level of confidence with genetic competencies commonly encountered in clinical settings: "How confident are you in interpreting the results of a microarray?" and "How confident are you in discussing specific genetic concerns with patients and families?" in each pre- and post-test. Responses were graded on a 4-point Likert scale. Due to an error, this information was not available for participants who completed Module 1.

We then compared responses pre- and post-test, to determine whether confidence levels improved with the modules. A 2-sample proportion test was applied to those who reported "Not at all confident" and those who reported "Not very, Somewhat and Very confident". A p value of <0.05 was considered statistically significant.

Confidence levels improved significantly for interpreting the results of a microarray ($z=3.33$, $p=0.00086$). While there was a trend to improvement, the change in confidence in discussing genetic concerns with patients and families was not significant ($z=1.84$, $p=0.066$).

Knowledge of Genetics and Genomics Core Competencies

Participants were asked a short series of multiple choice questions examining knowledge of the core competencies and application in clinical scenarios for each pre- and post-test. For each module, more participants completed the pre-test than the post-test (116 pre-tests and 110 post-tests completed), and Module 1 had the largest number of respondents (59 pre-tests and 35 post-tests).

We compared mean scores for each pre- and post-test for all modules separately, and for the 4 modules combined (Figure 5). An independent two-tailed t-test was applied for each scenario. A p value of <0.05 was considered statistically significant.

Module 1 responses demonstrated a significant improvement in mean score ($t=-3.04$, $p=0.0031$). A trend for improvement in scores was observed for all of the remaining modules and for the 4 modules combined, not statistically significant (Module 2 $t=-0.96$, $p=0.34$; Module 3 $t=0.36$, $p=0.72$; Module 4 $t=-0.50$, $p=0.62$; Combined modules $t=-0.84$, $p=0.40$).

Discussion

We developed a series of online video podcasts to be used as a toolkit to promote genetic and genomic competencies for non-genetics residents. While a clear need for teaching of Genetics and Genomics key competencies has previously been demonstrated, prior to our study, evidence regarding the effectiveness of existing online modules as a tool for teaching precision medicine competencies was limited.

Our study identified a marked discordance between residents' reported exposure to patients with genetic diseases and their level of confidence in basic genetic competencies, in keeping with the previously

identified need to prepare physicians-in-training for genomics-based healthcare. While 59% reported seeing patients with genetic diseases "Sometimes, Often, or Very Often" in their practice, prior to our modules, 59% reported they were "Not at all confident or Not very confident" in discussing specific genetic concerns with their patients. Similarly, we noted that exposure to patients with genetic conditions was reported to be higher than exposure to genetic test results; the majority of residents (68%) reported that they "Rarely or Never" see the results of genetic testing. We speculate that this may reflect local reporting practices, as results of genetic testing in our centre are not available on the electronic medical record and are typically sent to the ordering staff physician.

There was a trend towards improvement in genetics and genomics confidence and knowledge with the completion of the modules, with some areas demonstrating statistically significant improvements. Prior to participating in the study, 75% reported that they were "Not at all confident or Not very confident" in interpreting the results of a microarray. Participants' confidence in interpreting a microarray demonstrated a statistically significant improvement ($z=3.33$, $p=0.00086$) following completion of our toolkit modules. Our study showed significant improvement in residents' knowledge of basic genetic concepts between Module 1 pre- and post-test responses. Although results for the other modules did not reach statistical significance, the scores all trended up between the pre- and post-test modules. Similarly, while there was a trend to improvement, there was no statistically significant increase in confidence in discussing specific genetic concerns following completion of the podcasts. We speculate that this may be in part due to the wide range of genetic disorders seen by participants, not explicitly covered by the four modules.

One of the strengths of our study was the wide range of participants who enrolled. Residents participated from multiple programs, including dermatology, family medicine, internal medicine, neonatology, neurology, pediatrics, pediatric nephrology, pediatric neurology, and other (22 remained unspecified). We had participation from residents in their first year of training, to their 6th year into residency. Comparison by specialty demonstrated that residents in pediatrics and pediatric neurology are most likely to care for patients with genetic disorders and see the results of genetic testing, while patients in family medicine and dermatology report doing so less often.

Another highlight of our study includes some of the important gaps in participants' knowledge of core genetic concepts that were identified. For example, in the Module 1 pre-test, 58% of residents failed to identify that mitochondrial diseases can also be inherited in autosomal and X-linked patterns, and 12% incorrectly identified the correct number of autosomes and sex chromosomes in a normal human karyotype. We also noted several gaps in knowledge in the pre-test responses in Modules 2-4. For example, 21% of residents incorrectly identified the main type of genomic variation detected by a microarray. These knowledge gaps have the potential to seriously impact clinical care for patients with suspected genetic disorders, and again highlight the importance of ongoing genetics and genomics education at the undergraduate medical education and post-graduate medical education level.

Our study had a number of limitations. Since the responses were anonymous, the pre- and post-questionnaire for each participant could not be matched to determine if an individual participant

improved their genetic and genomic competencies. Similarly, it could not determine how many modules each participant completed. This impacted our ability to detect and analyze differences in outcomes between pre- and post-module tests. We also had a small sample size, particularly for Modules 2-4. We speculate that this is because most residents were asked to complete the modules on their own time, rather than being provided dedicated time (for example, at an academic half-day). We suggest that integration at the post-graduate medical education level, including protected teaching time, is crucial to promoting the development of competencies in precision medicine.

We identified several future areas of development. While the toolkit included clinical case modules in a variety of disciplines (cancer genetics, prenatal genetics, cardiogenetics), these were not evaluated in our study. Addressing barriers to completion of the online modules will be necessary to determine if they are effective in promoting additional genetics and genomics competencies among residents. At our centre, physicians in many non-genetic specialties frequently order genetic tests for their patients and deliver the results. Without effective and innovative educational solutions, the genetics knowledge gap among residents will remain a barrier to successful implementation of precision medicine in healthcare. Finally, while these modules were developed prior to the COVID-19 pandemic, there has been an overwhelming adoption of e-learning techniques during the pandemic, and these modules are easily adapted for use in this context.

In conclusion, podcast modules are an innovative method to promote genetic and genomic competencies to postgraduate medical trainees. Developing confidence with core genetic and genomic competencies is essential for effectively implementing precision medicine into patient care. Teaching these competencies during post-graduate medical training remains a challenge due to competing curriculum priorities as well as limited time. Self-directed learning through online modules and podcasts offers an opportunity to overcome some of these challenges.

Declarations

Ethics approval and consent to participate

This study was approved by the Conjoint Health Research Ethics Board at the University of Calgary, REB18-0089. Informed consent was obtained from all participants, and data was de-identified.

Consent for publication

N/A

Availability of data and material

Data available upon request.

Competing Interests

The authors declare no conflicts of interest.

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Author's contributions

Conceptualization: CC, EP, CC, MM, XRY; Formal Analysis: EP; Funding acquisition: CC, EP, MM, XRY; Investigation: CC, EP, CC, LB, MM, XRY; Methodology: CC, EP, CC, MM, XRY; Supervision: MM, RS; Visualization: EP; Writing original draft: EP, MM; Writing review and editing: CC, EP, LB, MM, RS, XRY

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Figures

CanMEDS Framework	Precision Medicine Toolkit Objectives
Medical Expert	<ol style="list-style-type: none"> 1. Recognize the basic principles of genetic inheritance, heredity, variation, and genomics 2. Be able to take an appropriate family history 3. Describe the role of genetic investigations in patient management 4. Recognize the types of genetic investigations that can be ordered 5. Interpret a genetic testing report 6. Distinguish the role of diagnostic testing versus predictive testing
Health Advocate	<ol style="list-style-type: none"> 1. Identify the role of genetic testing in the prevention or early detection of genetic disease 2. Recognize the importance of counselling and consenting of patients prior to genetic testing
Communicator	<ol style="list-style-type: none"> 1. Recognize the principles of genetic counselling 2. Recognize the principles of consent
Collaborator	<ol style="list-style-type: none"> 1. Recognize the role of genetic counsellors, medical geneticists, laboratory scientists and bioinformatics specialists in Precision Medicine
Scholar	<ol style="list-style-type: none"> 1. Locate appropriate genetic resources 2. Re-appraise the literature to reinterpret various genetic findings
Leader	<ol style="list-style-type: none"> 1. Be aware of innovation in the field and set an example regarding appropriate use of these tests

Figure 1

The curriculum objectives covered by the precision medicine toolkit fulfill key competencies, as outlined by the CanMEDS framework for Canadian physicians.

<p>You are in clinic seeing Mason, a 2 year old boy diagnosed with cystic fibrosis. He has 2 older sisters who are both healthy. His parents, John and Lisa, are pregnant with a fourth child, a boy, and ask you what the chances are that this child will also have cystic fibrosis. Given that this is an autosomal recessive condition, you tell them:</p> <ul style="list-style-type: none"> ☐ There is a 1/4 chance of inheriting an autosomal recessive condition when both parents are carriers. Since John and Lisa already have three children, one of whom is affected, there is a 0% chance that the fourth child will be affected. ☐ There is a 1/2 chance of inheriting an autosomal recessive condition when both parents are carriers. There is a 50% chance that their unborn child will be affected. ☐ There is a 1/4 chance of inheriting an autosomal recessive condition when both parents are carriers. There is a 25% chance that their fourth child will be affected. ☐ In autosomal recessive conditions, boys have a 50% chance of inheriting the condition, and girls have a 0% chance. Since John and Lisa are expecting a boy, there is a 50% chance that he will be affected. 	<p>You ordered a microarray on a pediatric patient with intellectual disability, and the results came back as normal. You disclose the normal result to the parents, and they ask you, "does this mean that our child's intellectual disability is not due to a genetic condition?" How might you counsel them?</p> <ul style="list-style-type: none"> ☐ No, there could still be an underlying genetic condition causing the intellectual disability ☐ Microarray testing is performed as a first line test for intellectual disability, so yes, that is correct. ☐ This test result rules out intellectual disability in the child. ☐ It's uncertain at this time. However, we can safely assume that the risk of recurrence of intellectual disability in your family is very low (i.e. less than 1%) since we know that the microarray result was negative.
<p>You are seeing Kate, a 6-year-old girl with autism and mildly dysmorphic facial features. You suspect that there may be a genetic cause for her clinical presentation, and decide to order achromosomal microarray. This test is best for looking at:</p> <ul style="list-style-type: none"> ☐ DNA rearrangements (translocations) ☐ Missing or extra segments of DNA (microdeletions and microduplications) ☐ Mutations within a gene (single nucleotide variants) ☐ Inverted segments of DNA (inversions) 	<p>You ordered a genetic test for polycystic kidney disease and obtained the following result: PKD1 c.9829C>T, p.(Arg3277Cys), heterozygous, pathogenic variant (paternally inherited), AD Based on this result, answer the following questions:</p> <p>What is the gene that this variant is in?</p> <ul style="list-style-type: none"> ☐ Cys ☐ PKD ☐ AD ☐ PKD1

Figure 2

Sample questions from each module.

Specialty	Pre-test	Post-test
Dermatology	11	5
Family Medicine	83	54
Internal Medicine	1	0
Neonatology	2	0
Neurology	1	0
Pediatric Nephrology	1	0
Pediatric Neurology	16	11
Pediatrics	29	21
Unspecified	22	19
Year of Residency	Pre-test	Post-test
PGY1	24	14
PGY2	94	65
PGY3	19	15
PGY4	8	5
PGY5	10	8
PGY6	2	0
Unspecified	9	3
	166	110

Figure 3

A total of 166 pre-tests and 110 post-tests were completed by residents in at least 8 specialties, including residents across all levels of training.

Figure 4

Reported frequency of seeing the results of genetic tests, and caring for patients with genetic disorders (A, B). Resident confidence with discussing specific genetic concerns, interpreting results of a microarray (C, D).

	Pre-Test Participants (n)	Pre-Test Mean Score (%)	Post-Test Participants (n)	Post-Test Mean Score (%)
Module 1	59	80.68	35	90.56
Module 2	48	79.35	31	84.90
Module 3	30	69.40	23	71.65
Module 4	29	73.59	21	77.48
Combined	166	79.45	110	82.58

Figure 5

Mean scores for knowledge in Modules 1-4, and combined scores for Modules 1-4.

Supplementary Files

This is a list of supplementary files associated with this preprint. Click to download.

- [Module1TestPretest.pdf](#)
- [Module1TestPosttest.pdf](#)
- [Module2TestPretest.pdf](#)
- [Module2TestPosttest.pdf](#)
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- [Module3TestPosttest.pdf](#)

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