

Navigating genomic testing: Evaluation of an e-learning module with general practitioners

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Abstract

Background: Primary care, or general, practitioners (GPs) are increasingly expected to communicate with and support their patients through genetic or genomic testing. However, GPs worldwide report a lack of knowledge about genetics and what constitutes a genetic or genomic test. Few Australian educational resources for medical practitioners address core capabilities in genetics or genomics.

Methods: We developed and evaluated a 90-minute online education module, *Navigating Genomic Testing in General Practice*, hosted online for 12 months. The module aimed to increase GPs' awareness and knowledge of genomics, increase confidence in their capability to support genomic testing and foster intention to utilise that knowledge. GPs residing and practising in Australia who completed the module also completed a mandatory post-module survey. They were then invited to complete a behavioural intention survey and participate in an interview.

Results: Between December 2018 and 2019, 216 Australian GPs completed the module and mandatory survey. Fourteen GPs went on to complete the optional survey. Most Australian GPs who completed the module reported gaining knowledge that was relevant to their practice (n = 215, 99.5%) and entirely met their learning needs (n = 168, 77.8%). Respondents of the optional survey (n = 14) indicated that confidence in their capability significantly increased post module, together with intention to support patients undergoing genomic testing.

Conclusion: GPs in this study reported increased confidence, ability and intent, which are factors that can lead to changes in behaviour. Our findings support use of this type of online course as an educational tool to help GPs navigate genomics as it increasingly impacts their practice.

Keywords: genetics education; genomics education; general practice; genomic testing; medical education; continuing professional education

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Introduction

Recent advances in genomic technologies have enabled faster and more accurate DNA testing at a reduced cost (Manolio et al., 2015). More and more people are accessing tests, including whole genome and exome sequencing, as part of clinical care, research studies and online DNA testing (Biesecker & Green, 2014; Phillips, 2016; Vassy et al., 2018; Williams, 2019). As primary care providers, GPs are increasingly expected to be able to communicate with and support their patients as they consider and undergo genetic and genomic testing (Carroll et al., 2016; Carroll et al., 2019). In Australia, GPs have a new and critical role in offering reproductive genetic carrier testing for fragile X syndrome, spinal muscular atrophy and cystic fibrosis (Delatycki et al., 2019), heightening the importance for GPs to keep up-to-date in this rapidly evolving field.

GPs have described their roles in genetic testing broadly, including taking family histories; identifying patients with, or at increased risk of, genetic conditions; referring patients appropriately; discussing benefits and limitations of testing; coordinating screening and facilitating risk assessment; providing post-test care and management; and responding to patient concerns (Carroll et al., 2016; Carroll et al., 2019; Hayward et al., 2017). GPs are uniquely positioned to facilitate appropriate testing and communicate genomic information (Haga et al., 2019). More than 80% of individuals in Australia see a GP at least once per year, and they are often a patient's first point of contact with the healthcare system (AIHW, 2022). In a snapshot of individuals in Australia living with a genetic condition, more than a third had discussed genomics with their GPs and thought them trusted resource (Petrie et al., 2016). Around 80% of the Australian public would consult with their GP to help them interpret a consumer genomics test result (Metcalf et al., 2019).

However, studies in Australia reflect international research that report GPs lack knowledge about genetics and genomic advances (Mikat-Stevens et al., 2015). A qualitative Australian study found that GPs were already undertaking a number of context-dependent roles in genetics/genomics, such as ordering tests, referring patients to genetic services or receiving test results, although they were not always cognisant of the genetic nature of the tests (Cusack et al., 2021).

Internationally, research has found that primary care practitioners report interest in genetics education and training that they believe will increase their knowledge and confidence, if supported by point-of-care resources (Carroll et al., 2016; Carroll et al., 2019; Paneque et al., 2016). Australian GPs also expected that genomics would have an increasing impact on their practice and expressed a need for more case-based education and training through online learning and events (Cusack et al., 2021).

In 2016, a systematic review of genetic education interventions for primary care practitioners found that around half of the included studies showed improvement in knowledge and confidence (Paneque et al., 2016). However, evidence for improved

clinical practice aligned with these knowledge gains has been scant, in part due to recruitment and measurement challenges (Talwar et al., 2017).

Methods

Study design and approach

Our clinical genomics education team aimed to build capacity in Australian GPs to improve the implementation of genomic medicine through a theory-informed education program and evaluation.

We developed and evaluated an online education module to improve GPs' capability to manage patients undergoing genomic testing and effectively communicate up-to-date information about genetics and genomics. The module was accredited by the Royal Australian College of General Practitioners (RACGP) and the Australian College of Rural and Remote Medicine (ACRRM).

To evaluate the module, we assessed to what extent the education was perceived to achieve knowledge gains, in addition to changes in awareness, attitude, capability and behavioural intent. Assessment and analyses were founded in behavioural theories, including the theory of planned behaviour (TPD) and Triandis' theory of interpersonal behaviour (TIB) (Légaré et al., 2014).

Module development and review

The primary outcomes of the module, established by a program logic model, were to: (1) increase knowledge about genomic testing, (2) increase awareness of the relevance of genomics and confidence in GPs' own capabilities and (3) foster stronger attitudes towards testing and an intention to implement in practice.

Core genetic capabilities were absent from the 2016 *Curriculum for Australian General Practice* (RACGP, 2016). Although core genetic capabilities are outlined in the Human Genetics Society of Australasia (HGSA) publication for medical graduates, these competencies are not specific to GPs nor have they been endorsed by Medical Deans Australia and New Zealand (Human Genetics Society of Australasia, 2022).

Based on an audit of existing international resources and genetic competencies for GPs (RACGP, 2018; Paneque et al., 2017), a content framework was developed by authors AC and BT and piloted with 27 GPs and GP educators. Most of the GPs who participated in the pilot had encountered genetic and genomic tests in practice, and some had been involved in referring patients directly to a specialist and/or genetic service. The pilot included asking GPs to rank the importance of each topic covered in the framework. Feedback from the pilot included difficulties in understanding the significance of the results and their clinical applicability, leading to challenges in communicating genomic information to patients.

As a result of the feedback from the pilot, iterations to module content and structure were undertaken and the content framework finalised (Table 1). Simulated clinical cases relevant to general practice that provided immediate feedback on GPs' choices were included as part of best practice adult genomics learning (Reed et al., 2016). Another important addition, based on the feedback from the pilot, was to develop an optional illustrated online "genetics refresher", including fundamental genomics topics of genomic testing, genetic variation and genetic inheritance. The "refresher" could be accessed from within the module by GPs who felt that they lacked the prerequisite knowledge for the module.

Written materials, figures and videos were produced in collaboration with continuing professional development (CPD) education provider ThinkGP or adapted from a concurrently developed physicians' module. All material was designed to facilitate the outcomes and aligned with professional accreditation standards (RCGP, 2016).

Table 1

Description of "Navigating Genomic Testing" Module Topics and Outcomes

Module Topics	Description
Genomics in primary care	Genomic testing, how it is used and its potential benefits, limitations and negative impacts
Types of genomic tests	Genomic testing technologies, comparing massively parallel sequencing with other available technologies
Uses and limitations of genomic testing	Applications of genomic testing (e.g., diagnostics, pharmacogenomics and consumer genomics) with discussion of their interpretation, accuracy and utility
The genomic testing process	The genomic testing process (from referral to result), including where GPs fit into the landscape of testing and communication
Understanding genomic reports & correspondence	Case examples and sample reports demonstrating primary and secondary findings, results with uncertain significance and reanalysis of genomic data
Explaining genomic information to patients	Challenges and sensitivities for patients undergoing genomic testing (e.g., privacy, insurance, equity and family implications) and common questions from patients
Outcomes	<ol style="list-style-type: none"> 1. Recognise the relevance and applications of genomic testing in primary care and the role of the GP. 2. Explain the uses and limitations of genomic testing and the ethical, legal and social issues related to genomic testing. 3. Understand genomic reports and related correspondence in order to provide patients with the information they need. 4. Identify resources for patients undergoing genomic testing and patients with genetic conditions.

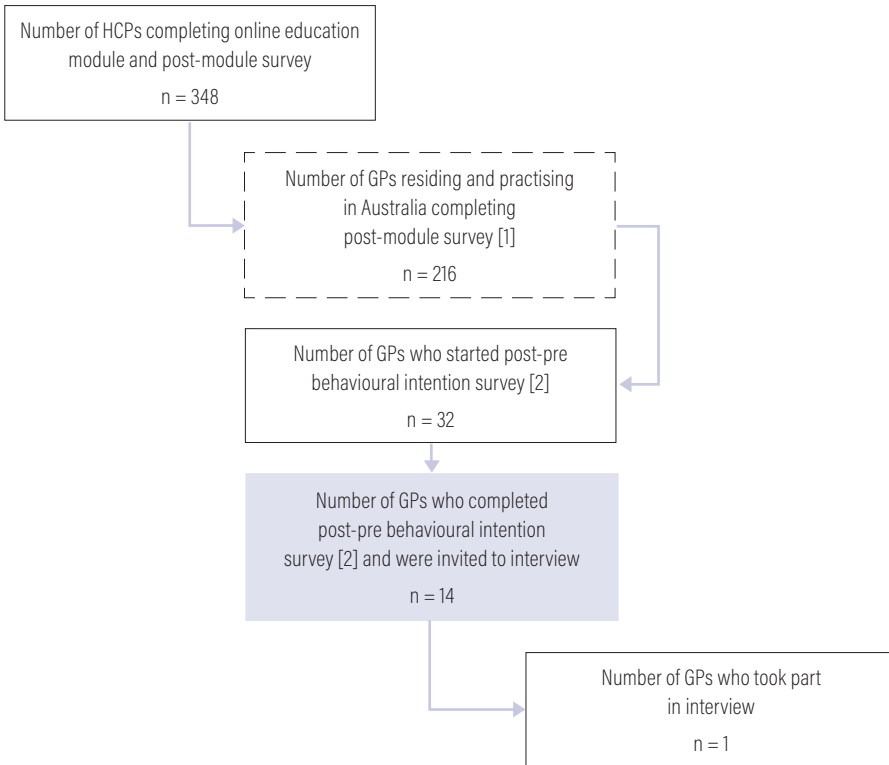
Module dissemination and evaluation

The 90-minute interactive online education module was hosted by education provider ThinkGP over a 12-month period (December 2018 – December 2019) and advertised through health network newsletters, social media and direct engagement with the ThinkGP platform. The module was supplemented with two genomics-focused blogs

by the authors (on online DNA testing and pharmacogenomics) and videos featuring genetics health professionals describing the relevance of genetics and genomics to general practice. The evaluation focused on immediate, short and medium-term outcomes (Figure 1), as outlined below.

Figure 1

Stages of the “Navigating Genomic Testing” Module Evaluation With Numbers of Respondents at Each Stage



Survey 1

A mandatory post-module survey was developed based on the RACGP Quality Improvement & Continuing Professional Development (QI&CPD) program requirements (RACGP, 2018). The survey contained nine items. Five items were measured using a 3-point Likert scale (not met, partially met, entirely met) and assessed if overall learning outcomes had been met, specifically if GPs could identify patient resources, understand genomic correspondences, explain the uses and limitations of genetic testing and recognise the relevance and application of genetic testing and their role. One item was measured on a 3-point Likert scale (not relevant, partially relevant, entirely relevant) and assessed relevance of the module to practice. One item measured

likelihood of recommending the module to colleagues, using a 5-point Likert scale (strongly disagree – strongly agree), and GPs were asked to choose the most and least useful topics covered in the module.

On completion of the mandatory survey, an additional question invited GPs who were living and working in Australia to participate in a research project to evaluate further outcomes of the module. The online participant information statements outlined that this would involve completing a 5-minute behavioural intent survey (immediately, online) and participating in a telephone interview 6 months after completing the module. GPs were offered a gift card as compensation for time and effort if interviewed.

Survey 2

The 12-item Clinical Professional Development Reaction (CPD-R) questionnaire (Légaré et al., 2014; Légaré et al., 2017) is a validated theory-informed tool used to assess the impact of CPD activities on behavioural intentions. GPs consented and completed this optional evaluation survey online, hosted by REDCap. A retrospective post-pre design was employed to maximise engagement with the module content, to enable reidentifiable information and intention data to be collected as a single session on a secure platform separate from the CPD provider and to reduce response shift bias, which may mask assessments of effectiveness (Bhanji et al., 2012; Hill & Betz, 2005).

The CPD-R measures intention (planning and intention), social influence (GP perception of whether their peers supported genomic testing), belief in capabilities (GP confidence and ability to support patients undergoing genomic testing, and the ease with which this support was adopted), belief in consequences (GP perceptions of how useful it would be to support patients undergoing genomic testing and the degree to which that support would be harmful or beneficial) and moral norms (extent to which supporting patients undergoing genomic testing was ethical and acceptable). The multiple-choice items are measured on a 7-point Likert scale, where 1 = *strongly disagree* and 7 = *strongly agree*. Demographic and practice items were also included.

GPs who completed both surveys were invited by email, 6 months post module, to participate in a semi-structured interview exploring the impact of the module on their practice. If GPs did not respond, two additional email notifications were sent at 2-week intervals before ceasing contact.

Descriptive statistics and frequency analyses were used to describe the learning outcomes and relevance to practice measured in Survey 1. For likelihood of recommending the module to a colleague, the categories of strongly agree/agree and strongly disagree/disagree were combined. For the post-pre evaluation survey data, Wilcoxon signed-ranks test was used to assess changes in the CPD-R. The Wilcoxon signed-ranks test was used as the sample size was small ($n = 14$) and the variables were not normally distributed. Significance was set at .05.

All procedures followed were in accordance with the *National Statement on Ethical Conduct in Human Research* (2007 – updated 2018) in line with the Helsinki Declaration of 1975, as revised in 2008. The ethical aspects of this study were reviewed and approved by the St Vincent’s Hospital Human Research Ethics Committee (SVH file number 18/234).

Results

Mandatory module completion survey (Survey 1)

In total, 216 GPs who resided and practised in Australia completed the interactive online education module and post-module mandatory survey. Most were from New South Wales (n = 67, 31%) and Victoria (n = 65, 30.1%), with the remainder from Queensland (n = 39, 18.1%), Western Australia (n = 19, 8.8%), South Australia (n = 14, 6.5%), Australian Capital Territory (n = 8, 3.7%) and Tasmania (n = 4, 1.9%).

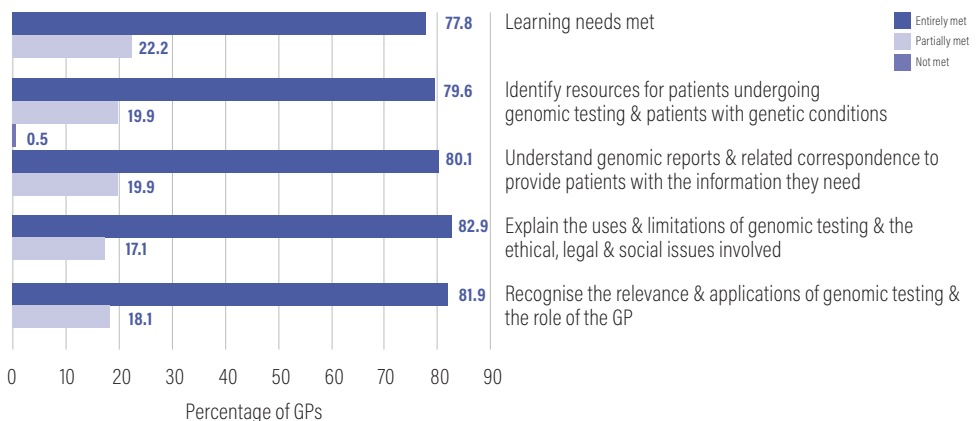
Nearly all GPs indicated the module was relevant to their individual practices; 86.6% (n = 187) noted it was entirely relevant, and a further 13% (n = 28) said it was partially relevant. Only one GP indicated it was not relevant at all. The majority agreed or strongly agreed that they would recommend the module to colleagues (n = 171, 79.2%). The remainder were neutral (n = 26, 12%) or disagreed/strongly disagreed (n = 19, 8.8%).

GPs indicated the most useful aspects of the module were the sections on uses and limitations of genomic testing, explaining genomic information to patients and case studies. The least useful aspect of the module was the section on genomic testing processes and landscape.

Most GPs indicated that their learning needs and the learning outcomes had been entirely met (Figure 2).

Figure 2

GPs’ Assessments of the Extent to Which the “Navigating Genomic Testing Module” Met the Learning Outcomes and Their Learning Needs



Post-pre evaluation survey (Survey 2)

Fourteen GPs (6.5% of eligible GPs) completed the CPD-R post and pre (completion rate of 44%). Most were specialist GPs (n = 9), were located in urban areas (n = 9) and were involved in group practice (n = 12). The average age was 51 (range 37–65 years), and GPs had been in practice, on average, for 23.4 years, although this ranged from 3–39 years.

All respondents (6.5% of eligible GPs) were currently practising in Australia. Most were specialist GPs: 64% FRACGP, 14% GP registrars, with the remaining listed as "other". GPs were located in urban (64%) and rural (36%) areas from across Australia. Most were located in NSW (50%), QLD (14%) and VIC (14%). Nearly all GPs were involved in group practices (92%).

Significant differences were evident in the post-pre data relating to GPs’ confidence and intent to implement learnings into practice (Table 2). For the belief in capabilities subscale (confidence and adoption), the Wilcoxon signed-ranks tests indicated that the median post-test ranks (5.83) were statistically significantly higher than the pre-test rank (median = 4.67), $Z = -2.06, p = .039$. Two pairings were tied, and one was lower post versus pre test. Two items in Survey 2 measured planning and intention, forming the subscale of behavioural intent. The Wilcoxon signed-ranks tests indicated that the median post-test ranks (6.75) were statistically significantly higher than the pre-test rank (median = 6.50), $Z = -2.03, p = .042$. Nine pairings were tied, and five were positive.

There were no significant differences in post-pre scores for the subscales of social influence, moral norms and belief in consequences: there were no significant differences in GPs’ perception of their peers exhibiting support for patients undergoing genomic testing; they also felt that supporting patients was useful, beneficial, ethical and acceptable prior to undertaking the education module and, hence, no gains were reported post module.

Table 2

Wilcoxon Signed-Ranks Tests for the CPD-R Subscale and Items From the Post-Pre Behavioural Intention Survey

Item	Pre-Module			Post-Module			Z	p
	M	SD	Mdn	M	SD	Mdn		
Social influence								
Percentage of my colleagues who support patients undergoing genomic testing	3.36	1.50	3.5	3.36	1.50	3.5	-	NS
My co-worker supports patients undergoing genomic testing [never-always]	5.36	1.95	6	5.43	1.83	6	-	NS
Important people in my profession support people undergoing genomic testing [SD-SA*]	5.07	1.94	5.5	5.29	1.86	7	-	NS

Item	Pre-Module			Post-Module				
Beliefs about consequences								
Supporting patients undergoing genomic testing would be [harmful-beneficial]	6.07	1.33	7	6.29	.83	6.5	-	NS
Supporting patients undergoing genomic testing would be [useless-useful]	6.00	1.44	6.5	6.21	1.12	7	-	NS
Moral norms								
Supporting patients undergoing genomic testing is the ethical thing to do [SD-SA]	5.14	1.99	5.5	5.57	1.91	6.5	-	NS
It is acceptable to support patients undergoing genomic testing [SD-SA]	6.43	1.09	7	6.29	1.73	7	-	NS
Beliefs about capabilities								
I am confident I could support patients undergoing genomic testing [SD-SA]	4.23	1.42	4	5.50	1.95	6.5	2.11	.035
Supporting patients undergoing genomic testing would be [extremely difficult-easy]	5.14	1.99	5	5.57	1.91	5.5	-	NS
I have the ability to support patients undergoing genomic testing [SD-SA]	4.21	1.63	4	5.14	1.61	5	-2.49	.013
Behavioural intention								
I intend to support patients undergoing genomic testing [SD-SA]	5.29	2.23	6.5	5.86	1.96	7	-2.06	.039
I plan to support patients undergoing genomic testing [SD-SA]	6.07	1.44	7	6.43	.94	7	-	-

* Strongly disagree-strongly agree

Semi-structured interview

Only one GP who completed both surveys was interviewed about how the education module had impacted her practice. Due to low recruitment, these results are not reported here.

Discussion

Findings from this study suggest that the online education module's aims of improving knowledge and awareness were met, as most GPs agreed their learning needs were met and the content was relevant to their practice. Topics that GPs rated as most useful reflected both the needs assessment and previous literature (Carroll et al., 2019), with GPs' interest highest in learning about utility and limitations of genetic and genomic testing and communication of genomic information to patients. GPs expressed lower interest in the processes of genetic testing, which could be seen to be less relevant to their day-to-day practice (Cusack et al., 2021). In contrast, and aligning with previous studies

(Paneque et al., 2016; Reed et al., 2016), GPs favoured inclusion of case studies modelling pivotal roles for GPs, such as taking an accurate family history and referring appropriately to genetics (Carroll et al., 2019; Carroll et al., 2021; Cusack et al., 2021).

Our findings also suggest that the module fostered an intention to implement support for genomic testing in practice. According to behaviour change theories, improving uptake of new practices involves multiple factors that can include an individual's attitude and their belief that they have control over its implementation (Azjen, 1991; Légaré et al., 2014). In the post-pre survey completed after the module, GPs reported significant self-perceived increases in ability and confidence to support patients undergoing genomic testing. GPs' intent to support patients through the genomic testing process also increased significantly, while their perception of the ease of supporting these patients also increased, but not significantly. Behaviour change is also influenced by peers' views and perception of importance (Azjen, 1991; Légaré et al., 2014). As this module was undertaken by individuals online, GPs' thoughts about their peers' views of genomic testing were unlikely to alter considerably over the course of the module, however most GPs who completed the module would recommend it to a colleague. Participating GPs' perceptions of the usefulness, consequences of, ethics and acceptability of patients undergoing genomic testing were already high prior to the module, so this created a ceiling effect post module.

Our assessment of behavioural change was halted by low GP recruitment for the interview 6 months after completion of the module. This may have been due to busy GPs' perceived lack of time to schedule and complete an interview rather than a survey (Brodaty et al., 2013), a lack of perceived value in participating further in research without additional professional development (Brodaty et al., 2013) or a rarity of genetic cases to practise what they had learnt between surveys and interview (Ong et al., 2022). Evidence for improved clinical practice after a medical genomics education intervention is scant in the literature (Paneque et al., 2016; Talwar et al., 2017). However, future research may benefit from the new roles for Australian GPs in genetic carrier screening (Delatycki et al., 2019), providing additional incentive for GPs to undertake genomics education and participate in its evaluation. Longer-term research with GPs may also be facilitated through additional communication and/or offering additional education updates as part of any follow-up (Ong et al., 2022) to increase the perceived value of the GPs' participation.

GPs in this study reported increased confidence and an improved ability to support patients undergoing genomic testing. By combining the survey data, we can demonstrate a continuum of participant satisfaction, perceived increases in confidence and ability and intent to implement, which are factors that may lead to further practice change (Legaro et al., 2014).

We can also model an evidence-based approach to design, development, delivery and evaluation of an educational intervention. To effectively integrate genomics and precision medicine into routine healthcare in Australia, upskilling the non-genetics workforce

is a priority (O'Shea et al., 2022). However, web-based interventions may bridge knowledge gaps but rarely translate into behaviour change (Ong et al., 2022) without additional workplace learning, access to communities of practice (Reed et al., 2016) or multidisciplinary team models. These findings are reported in accordance with the *Rise2 Genomics* reporting standards for genomics education and evaluation (Nisselle et al., 2021) to contribute to evidence for effective genomics education across different specialties, contexts and modalities.

There were a number of limitations to this study. The module was offered as self-directed CPD as one of many available on the ThinkGP CPD platform, creating a response bias in the sample, as only GPs who were already interested in the topic would have participated. After the first survey, there were only small numbers of GPs responding, potentially skewing participants towards GPs who perceive the module and its subject matter positively and were more inclined to participate in each stage. In addition, there are limitations introduced by the retrospective post-pre survey, which relies on the accuracy of self-reporting perceived learning before and after the intervention. Lack of recruitment for interviews also halted assessment of behaviour change.

Conclusion

Overall, the findings of this evaluative study indicate that GPs who completed the module gained knowledge about genomics and its relevance and application to general practice. Most GPs reported that their learning needs were met in key areas relevant to GPs' anticipated roles in genomics, including communicating with patients about genomic information and supporting patients through genomic testing by identifying patient resources and understanding the uses, limitations and issues of genomic testing. The findings also indicate that GPs' perceived capability and clinical behavioural intentions improved after completing the education module. GPs' confidence, ability and intention to support patients undergoing genomic testing also increased significantly. These findings support the importance and efficacy of this type of short online course as an effective educational tool to assist GPs to navigate genomics, as it impacts their practice.

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A non-accredited version of the module is available for self-enrollment and review at <https://learn.garvan.org.au/courses/navigating-genomic-testing-primary-care/>

Conflicts of interest and funding

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