Family Medicine and Community Health

General practitioners' (GPs) experience, attitudes and needs on clinical genetic services: a systematic review

Cheryl Siow Bin Ong [1], Rose Wai-Yee Fok,[2] Ryo Chee Ann Tan,[3] Si Ming Fung,[2] Shirley Sun,[1] Joanne Yuen Yie Ngeow[2,3]

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[1]Sociology, School of Social Sciences, Nanyang Technological University, Singapore
[2]Cancer Genetics Service, Division of Medical Oncology, National Cancer Centre Singapore, Singapore
[3]Lee Kong Chian School of Medicine, Nanyang Technological University, Singapore

Correspondence to
Dr Joanne Yuen Yie Ngeow; joanne.ngeow@ntu.edu.sg

ABSTRACT

Objective The proliferation and growing demands of genetic testing are anticipated to revolutionise medical practice. As gatekeepers of healthcare systems, general practitioners (GPs) are expected to play a critical role in the provision of clinical genetic services. This paper aims to review existing literature on GPs' experience, attitudes and needs towards clinical genetic services.

Design A systematic mixed studies review of papers published between 2010 and 2022.

Eligibility criteria The inclusion criterion was peer-reviewed articles in English and related to GPs' experience, views and needs on any genetic testing.

Information sources The PubMed, PsycINFO, Cochrane, EMBASE databases were searched using Mesh terms, Boolean and wildcards combinations to identify peer-reviewed articles published from 2010 to 2022. Study quality was assessed using Mixed Methods Appraisal Tool. Only articles that fulfilled the inclusion criteria were selected. A thematic meta-synthesis was conducted on the final sample of selected articles to identify key themes.

Results A total of 62 articles were included in the review. Uncertainty over GPs' role in providing genetic services were attributed by the lack of confidence and time constraints and rarity of cases may further exacerbate their reluctance to shoulder an expanded role in clinical genetics. Although educational interventions were found to increasing GPs' knowledge and confidence to carry out genetic tasks, varied interest on genetic testing and preference for a shared care model with other genetic health professionals have resulted in minimal translation to clinical adoption.

Conclusion This review highlights the need for deeper exploration of GPs' varied experience and attitudes towards clinical genetic services to better facilitate targeted intervention in the adoption of clinical genetics.

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ General practitioners (GPs) are well positioned to provide patients with clinical genetic services by screening for potential patients who may benefit from genetic testing. However, GPs are faced with skills, knowledge, time and clinical constraints that hinders the effective adoption of clinical genetic services in primary care setting.

WHAT THIS STUDY ADDS

⇒ This study found that GP's views and attitudes towards adopting clinical genetic services were dependent on their experiences and context. GP's preference for a shared responsibility between them and genetic specialists may help overcome the resistance towards adoption of clinical genetic testing.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ The study highlighted the varied interest among GPs to incorporate genetic services in their clinical practice and the importance of addressing valid concerns and tailoring interventions to overcome barriers for GPs who may wish to adopt genetics in their practice.

INTRODUCTION

Advances in genetic research accompanied by the availability of a wide array of genetic tests is set to revolutionise medical practice worldwide.[1 2] General practitioners (GPs), as the gatekeepers in the healthcare systems, will need to be well informed of the benefits and risks of clinical genetic testing in order to respond to patients' requests for direct-to-consumer (DTC) genetic testing. However, clinical genetics is often regarded by GPs as a specialty arena and not a core component of generalist practice.[3 4] This discrepancy between what GPs should provide and what they perceive as within their role and competency may create confusion for primary care and clinical genetic testing healthcare providers.

Existing reviews mostly examine cancer genetics[5–10] with the most recent review that focused on general clinical genetics published in 2016.[11] Existing reviews have found that GPs experienced a lack of knowledge and confidence in basic genetics and risk assessments in the provision of clinical genetic services.[5 7 8 12 13] In addition, GPs also expressed concerns over ethical, legal and social implications (ELSI),[5 6] time pressures,[5 9] and difficulties in accessing referral

guidelines and genetic tests as barriers in the provision of cancer precision medicine. Despite these barriers, two reviews found that some GPs held positive attitudes as they recognised the importance of their increasing role in the development of primary care genetics.[9 12] Furthermore, in terms of adoption of clinical genetics, the review conducted by Paneque *et al* found that existing genetics educational interventions on patient management and policy for GPs have been poorly assessed. In order to determine the effectiveness of educational initiatives, assessment need to account for the changing primary care practices.[11]

However, little is known about whether existing educational initiatives and clinical interventions has changed GPs' experience and attitudes towards the adoption of clinical genetics. Also, most reviews focusing on primary care mostly included specialists such as oncologists, genetic counsellors, paediatricians and allied health professionals in their study population.[5–8 10 12 13] While alike, specialists would yield different experience as practice styles are heterogeneous.[14] With GPs being positioned in such milieu, it raises the need to better understand their ambivalent attitudes towards adopting clinical genetic testing, and their awareness of an increasingly salient role they could play and in advancing the utility of genetics in their clinical practice.

For this systematic review, we defined general practice to be 'the medical specialty that manages common and long-term illnesses such as asthma, diabetes and end-of-life care in children and adults, focusing on overall health and well-being.'[15] We defined genetic testing as the use of a laboratory test that comprises a broad range of testing techniques for medical care, ancestry studies or forensics, by detecting variations in an individual's DNA.[16 17] This includes the diagnosis of suspected genetic disease in symptomatic and asymptomatic newborns, children and adults (eg, Huntington's disease); risk assessment where individuals are informed of their increased or decreased risk of developing a condition (*BRCA1/BRCA2*); prediction of drug responses (eg, carbamazepine); and reproductive decision making (eg, thalassaemia).[2 17 18]

The aim of this systematic review was to examine GPs' experience and attitudes towards adopting clinical genetic services, as well as GPs' needs to provide genetic testing in their clinical practice. For this review, we defined experience as any discussion on genetics with patients and subjective experience that includes knowledge, confidence and barriers. Attitudes included views on the utility of genetic testing in their clinical practice and GPs' role in providing such clinical genetics services. Needs included strategies targeted at incorporating clinical genetic services. These included informational resources (eg, education workshops) and institutional system support (eg, practice policies, guidelines and recommendations). This review considered taking family history, recommending and ordering tests, interpreting test results, managing downstream care and referral to clinical genetic centres to fall under the umbrella of potential clinical genetic services for GPs. In this paper, the term 'GP' referred to both family physicians and primary care physicians.

METHOD

This review is reported in accordance with the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) 2020 checklist. For this review, a systematic mixed studies review (SMSR) was conducted to identify and synthesise research pertaining to the research questions. The steps taken to conduct the SMSR followed the typical process of a traditional systematic review. However, in contrast to the conventional mixed methods synthesis whereby data set from qualitative and quantitative studies were reported separately, the focal point of SMSR was the consolidation of data sets from a range of quantitative, qualitative and mixed-method studies.[19 20]

Given the complexities surrounding the adoption of clinical genetic practice, an SMSR was considered to be appropriate in synthesising the growing literature from diverse research designs.[21] While, traditional systematic reviews have been given precedence to quantitative evidence,[22] qualitative studies have been concomitantly gaining traction and recognition in healthcare related research.[23] Conducting an SMSR would thus provide a comprehensive understanding of the phenomenon and a highly practical understanding on the complex public health interventions and programmes.[20 22]

Search strategy

Four electronic databases (PubMed, PsycINFO, EMBASE and Cochrane databases) were searched systematically, and studies that fulfilled the eligibility criteria published between 2010 and 2022 were selected. The reviewers used PubMed as one of the databases as it comprises of biomedical literature from MEDLINE, life science journals and online books. In addition, 2010 was chosen as the starting date for this review in view of the emergence of the next-generation sequencing clinical genetic testing, a technology anticipated to reduce the cumulative testing costs and thereby, encouraging mainstream access to genomics.[24 25] Grey literature was not included in this review as we only considered peer-reviewed published studies. Citation search was conducted on a few studies to capture relevant articles. The exact search strategy used by the reviewer is outlined in online supplemental table 1.

Study selection

Studies were included if they addressed genetic testing related to primary care within the scope of family medicine or internal medicine. In addition, worldwide literature were included if they met the following inclusion criteria: (1) peer-reviewed articles; (2) in English and (3) focused on GPs' experience, views and needs on any genetic testing. The inclusion and exclusion criteria are outlined in online supplemental table 2. The title, abstract

and full text of the articles were screened independently by two reviewers (CO and RCAT).

Data extraction and synthesis

A meta-synthesis was conducted to integrate existing studies to identify key themes. A data-based integration approach was used, and two reviewers conducted data transformation by reconstructing quantitative data into categorical themes and narratives for comparison with qualitative data.[20 21] Studies that were included from the full text screening were grouped together if their findings addressed the GPs' experience, attitudes, or needs. Data extracted were then keyed into an Excel spreadsheet independently by two reviewers (CO and RCAT). The data included key findings related to GPs' experience, attitudes and needs in clinical genetics services as defined in the inclusion criteria (online supplemental table 2). Other data sought included authors, publication year, country of study, aims, design methods, participants, genetic type and limitations of the study. Findings from each reviewer were compared for concordance and all discrepancies were adjudicated by a third reviewer (JYYN). Finalised data were tabulated on the Excel sheet and subsequently formatted into a table in Microsoft Word for display. Data extracted from each studies can be found in online supplemental table 5. The findings have been grouped into two categories: (1) GPs' experience, attitudes, views and (2) GPs' needs.

Assessment of methodological quality

Quality assessments were conducted by two reviewers (CO and RCAT) independently. The quality of all selected studies was assessed using the 2018 version of the Mixed Methods Appraisal Tool (MMAT). MMAT was chosen as it included appraisal of various study types,[26] with five items for each study type that can be found in online supplemental table 4. Studies were first organised into their respective study type – qualitative, quantitative RCT, quantitative descriptive and mixed methods—before they were rated based on the five items specified in the chosen category. For items where the rating 'Can't tell' was given, additional comments were included. As advised by the MMAT developers, the ratings for each study have been presented in the online supplemental table to provide readers with a clearer evaluation of the quality of included studies.[26] Any missing results or data were highlighted.

RESULTS

Characteristics of studies

A total of 871 studies were identified on the database (online supplemental figure 1). After removing 213 duplicates, the titles and abstracts of 658 studies were screened. Full texts were retrieved from 160 studies. A total of 62 studies satisfied the eligibility criteria and were included in the final sample.

Of the 62 final articles included, 36 studies were quantitative studies,[27–62] 13 were qualitative studies,[3 63–74] 9 were randomised controlled trials[75–83] and 4 were mixed-methods study[70 84–86] (online supplemental table 5). The selected studies were conducted in USA (n=20), UK (n=19), Canada (n=11), Australia (n=6), Asia (n=4), New Zealand (n=1) and South Africa (n=1). These studies composed of a range of genetic scope such as general clinical genetics (n=24), oncogenetics (n=18), pharmacogenetics (n=6), cardiovascular (n=6), prenatal/neonatal genetics (n=4) and DTC testing (n=4). Forty studies reported participation of both male and female GPs in their study population. Most articles (82.3%) focused on GPs' knowledge and experiences towards genetic testing (online supplemental table 3).

Risk of bias within studies

All studies were assessed using the MMAT tool. Fifty-five studies achieved a maximum score of compliance for all five assessment items. Seven studies lacked data on non-respondents to properly assess the risk of non-response bias (online supplemental table 4).[27 30 32 40 44 47 53] One common risk across the studies was the low response rate, which was reported in 29 studies.

Uncertainty over GP's role in genetic testing

Out of the 22 studies that reviewed GP's attitudes towards their role in genetic testing, 8 studies reported that GPs felt responsible to perform genetic tasks such as taking family history to identify genetic condition,[51] assisting or counselling patients on genetic testing and results,[32 61 65] referring patients to specialists for advice and follow-up care,[51 69] and to warn families about risks in the family.[53] GPs also found themselves to be well positioned to offer genetic screening such as population-based expanded carrier screening couple test,[66 85] and in the early detection of familial hypercholesterolaemia (FH).[54]

However, 11 studies found GPs to be ambivalent towards their existing role and competency to provide genetic screening in their practice. Hussein *et al* highlighted a mismatch in attitudes and actual practice as GPs took on distinct proactive or reactive approaches. For instance, while 70% of the 271 GPs surveyed considered taking family history as an integral role of general practice,[56] another study comprising of 96 GPs found that a proportion of the GPs surveyed were sceptical if taking family history should be part of their practice due to difficulties to obtain an accurate family history.[76] While genetic concepts are part of their general practice, two qualitative studies found that they are made distinct from genetic practice and thus, not identified as core component of their practice.[3 66] As illustrated by Mathers *et al*, although GPs may appear to be more willing to document family history, the routine use of family history for general disease management was distinguished from those for genetic conceptualisation.

Out of the 11 studies, 7 studies found that GPs leaned towards having a minor role that focuses more on traditional genetic tasks of identifying, referring and providing psychological support rather than assessing and explaining

genetic risks, benefits and limitations.[28 31 38 51 56 60 86] Two studies found that less than 25% of the 1168 GPs surveyed were willing to discuss genetic testing,[60] and 70% of the 271 GPs did not perceive ordering genetic tests or discussing about testing implications as part of their job scope.[56] Interestingly, one study found that younger GPs were more willing to incorporate genetic tasks into their everyday clinical practice.[61]

Lack of confidence and limited knowledge

GP's uncertainty over their role in genetic testing was compounded by their lack of confidence and knowledge on genetic testing. Out of the 34 studies that reviewed GP's knowledge and experience towards genetic testing, 7 articles shed light on the limited experience and encounters with genetic testing[43 44 63 65] or genomic cases.[28 43 58] Of the 63 GPs surveyed, 77.8% had no experience with referring patients.[36]

Ten articles also highlighted GPs lack of confidence on their genetic knowledge,[40] ability to conduct genetic screening,[66] and to perform genetic tasks[48 51] such as interpreting results[62 65 84] and discussing benefits, risks, limitations and inheritance patterns.[51 62 76] The study by Bernhardt *et al*, found that only 58% of the 481GPs surveyed were confident in interpreting genetic test results. GPs also reported a lack of confidence to educate patients on genetic cardiac disease and answer patients' questions.[35]

Apart from the lack of confidence, 17 studies reported on GPs' lack of knowledge on genetic testing. GPs were found to lack necessary technical expertise and skillsets to convey results[31 67] or had minimal knowledge to interpret results or estimate risks.[50 52 70] GPs were also unfamiliar with genetic concepts,[3 32 34 42 44 46 47 69] evidently revealed through a semi-structured interviews with 15 GPs which indicated a lack of familiarity with genomic terminologies and genomic tests.[73] Rangarajan *et al* also found GP's overall knowledge of FH to be low with only 40.6% of the 133 GPs surveyed being aware of international guidelines, and 13% were cognizant of genetic services available. There was also a lack of awareness on genetic laws among GPs.[41] In addition, 52% of 90 GPs surveyed reported being unsure of how pharmacogenetics could be incorporated into their practice.[33] Insufficient knowledge on referral criteria,[31] referral pathways and appropriate centres for referrals[28 36] have also led to variation in referral patterns.[35] Teng *et al* found a wide discrepancy between GP's self-reported referral rates (87.5%) and actual referral rates (12.5%). Fiederling *et al* similarly reported that only 35% of 35 GPs would refer their patients to specific counselling centres.

Knowledge scores were found to be positively associated with comfort scores to perform genetic tasks and referrals.[86] Henceforth, minimal knowledge and lack of confidence may have discouraged GPs to feel comfortable to order genetic test[42] or adequate to provide genetic counselling. In concurrence, 65% of the 61 GPs surveyed felt that genetic counsellors, medical geneticists, or oncologists were more qualified to perform such tasks.[34] This sentiment was similarly highlighted in another study which reported 74% of the 27 GPs seeing it as others' duty to follow-up on genetic results.[86]

On the contrary, only five studies found GPs to be fairly confident about their ability to determine the need for further evaluation based on family history,[53] with 74% of 271 GPs surveyed having had contact with patients with genetic disease weekly.[61] Two articles also reported that GPs frequently refer patients for cancer genetic testing[37] and conduct cancer family history consultation and risk assessment.[38] However, it is critical to note that while 52.8% of 70 GPs surveyed felt confident to explain risks and benefits only 40% reported being confident in their genetic knowledge.[68]

Genetic education and interventions

There were 13 studies that reported on the effectiveness of clinical interventions. Five studies found an increased in comfort level with genetic testing through a multifaceted educational intervention that comprises of individualised training, supervision and additional resources such as checklists[71 80 84 85] or online genetics modules.[79] A care-based oncogenetics education that includes practical applicability, interactive sessions, small group discussions was also found to achieve a sustained improvement over 3 months after training.[78] Seventy-six per cent of the 1402 GPs surveyed also found 'pushed emails' to be useful for learning about genetics.[55] Another intervention that reported success was the use of an electronic health record coupled with family history tool which helped to increase patient awareness through system prompters that facilitated discussions.[68] Due to the rarity of genetic cases, Lemke *et al* found direct access to pharmacogenetics (PGx) testing was a good approach for GPs to obtain first-hand knowledge although more education was desired.

The importance of genetic education and training were reported in 27 studies. Five studies reported on the interest for more training and information.[28 31 37 40 62] Of which, Yu *et al* reported that 91% of the 409 GPs surveyed saw the importance to keep up with latest information on genetic disorders. Specifically, GPs expressed the need for more guidelines and timely updates on the use of genetic screening, genetic testing, genetic counselling and referrals.[34 39 63 65 70] In addition, clearer guidance that is tailored to their practice and roles as GPs were also coveted.[51 64 65 69] Greater understanding to interpret and communicate test results, care treatment,[64 67 72] and evidence on clinical utility[74] were also sought after by GPs. Of the 13 studies that reported on GPs preference to learn about genetics, 9 studies found Continuing Medical Education (CME), online medical references or journals to be useful for obtaining more information.[27 30 34 36 42 46 52 53 66] Other preferred avenues includes monthly circular on clinical and referral pathways,[35 68] grand rounds, case studies and physical seminars.[32 65]

However, Nippert *et al* reported that only 12.8% of the 1168 GPs surveyed attended CME courses on genetics. Moreover, interactive web-based CMEs were found to have minimal impact on changing clinical practices.[75] While web training has effectively increased self-reported genetic consultation and management skills, the actual number of referrals did not change.[83] Furthermore, interventions such as PGx alerts were reported to be confusing with 52% of 90 GPs surveyed not knowing how to use additional information in their practice.[33]

Varied outlook on responsibility and clinical utility

Discrepancies between interventions and actual practice could be attributed to the lack of consensus on the need for genetic training. An in-depth interview with 21 GPs revealed that the call for education, training and guidelines were not echoed by all.[3] Similarly, Schuurmans *et al* found that training may be more effective for GPs motivated to do so rather than all GPs. Nine other studies found that GPs saw genetic testing to fall under the responsibility of others. For instance, genetic specialists were highlighted to be more appropriate to provide counselling.[35 44] Genetic health professionals or pharmacists were also expected to communicate results and follow-up with patients.[32 77 86] Other than physicians, GPs also highlighted patients and family members' responsibility to follow up and adhere to recommendations,[36 56 86] which aligned with three other studies that call forth the need to educate patients and family members about genetic condition[35] using resources as such patient handouts.[65 68]

On the other hand, nine other articles emphasised on a shared care model with other healthcare professionals. This includes having opportunities to discuss with specialists or pharmacists,[32 34 65] a buddy system with geneticists or contact information on local genetic clinic.[51] GPs also appreciated summary letter, comprehensive report, interpretative comments or telephone call from genetic health professionals to help identify high risk patients and navigate through test results.[54 69 73 82] In addition, GPs placed more emphasis on the responsibility of screening centres.[36]

While GPs may not fully grasp the technicalities of genetics, some recognised the positive clinical utility of genetic results on patient care[86] and believe results would be helpful for patient care management,[58 62 64 70] especially in identifying risk and disease prevention.[40 68 71] Six articles further reported that GPs perceived genetic testing to play a bigger role in future practice.[28 42 51 61 62 65]

Yet, this positive outlook on clinical genetics was not unanimous among GPs. Resistance to integrate genetic testing into clinical practice could be attributed to the additional workload required to discuss recommendations and answer patient's questions amidst their busy practice[68 70 85] and time constraints during clinical consultation.[28 36 44 53 71 73 74 81] Furthermore, the rarity and complexity of genetic cases were perceived to have limited impact on their general practice.[3 44 47 66] Apart from the lack of clinical evidence, GPs also expressed concerns over negative patient attitudes,[43 66] language barrier with patients who did not have English as their first language,[44 71] and confidentiality and discrimination of test results.[36 46 68] Although GPs anticipate a substantial impact of genetic testing on future practice, 78% of the 1404 GPs surveyed felt that genetic testing was less appropriate to inform treatment with 58% expressing beliefs that DTC testing would likely harm patients' general health decisions.[40] Sixty-four GPs surveyed in a separate study also expressed concerns on giving patients a false sense of security or inducing unnecessary anxiety over genetic results.[62]

Moreover, resistance to adopt genetic testing was also exacerbated by organisational barriers such as rigid administration infrastructure,[66] lack of clinical guidelines on genetic practice[43 52 53] and limited access to labs that perform PGx testing.[84] Additional resources could also incur more cost that could overburden the healthcare system.[67 84]

Impact of healthcare models on GPs' attitudes

Different healthcare models in different regions may also affect GPs' willingness and expectations to adopt clinical genetic services. GPs in the UK and other regions (who were gatekeepers in their healthcare system) recognised their responsibility to provide clinical genetic services and desired greater need for genetic education relevant to their practices.[3 30 47 56 60–62 71 79 83] Within Europe, French GPs ascribed most practice responsibilities to themselves while GPs from the other UK regions assigned most tasks to a genetic specialists.[38] In contrast, GPs in the USA were generally more sceptical of the utility of clinical genetics and saw it as specialists' responsibility to perform genetic tasks.[33 40 42 46 49 52 58 75 77 86] Although Asian GPs are generally more conservative, some advocated the need for education to empower them to take on the role in the early detection of at-risk patients.[54 81]

DISCUSSION

This review highlighted the complex experience GPs face in adopting genetic practices that ranged from taking a genetic family history, recommending and ordering tests, interpreting test results, managing downstream care and referral to clinical genetic centres.

While GPs considered their role in clinical genetics to be salient, they were uncertain about what this role entails. Findings on GPs' uncertainty over their role and responsibility to provide and assess genetic results corroborates with existing literature.[87 88] Findings also concurred with literature that found GPs to perceive genetic tasks to be highly complex that requires specialists' knowledge.[89] While GPs were trained to apply multifactorial clinical risk factors in their practice to inform medication use and patient care management, they were less likely to have adequate experience or exposure to rare genetic diseases.[90] In comparison, specialists may seemed more suited, confident and better prepared to perform and

interpret DTC testing than GPs. Furthermore, a study has found that the use of DTC testing lack standardised laboratory practices and is prone to misclassification in risk assessment and should be used with caution for clinical care decisions.[91] This may pose as a risk for GPs who could be less attuned to the heightened levels of scrutiny towards DTC testing and accept genetic test results at face value.[90]

Yet, GPs remained well positioned to assist their patients in informing their family members about genetic testing options in view of their long-standing relationship. However, help from genetic counsellors is needed to ensure a smooth transition between general and specialist care. A review conducted in 2004 highlighted that GPs played an essential role in the ongoing follow-up with their patients after a genetic diagnosis has been made.[2] Aligned with the principles of generalist–specialist relations, GPs were found to favour a shared care model where specialist assistance is sought to provide comprehensive management in the complex care of clinical genetics.[92] Not just between GPs and geneticists, a national survey of primary care paediatricians also found the majority of them to be actively involved in genetic services through ordering genetic tests and referring patients to geneticists annually.[93] This interdisciplinary approach to genetic testing serves a critical role in ensuring optimal care for complex genetic cases through concerted efforts from paediatricians, genetic counsellors, therapists, nurses, social workers and psychologists to meet the medical and psychological needs of patients.[94] The need for specialists' expertise to craft a follow-up care plan after a genetic diagnosis was further reiterated in a semistructured interview with 15 GPs where they expressed that without proper follow-up actions for GPs to act on, patients may suffer from potential harms that can lead to anxiety and unnecessary investigations.[72]

Even though GPs recognised the benefits of clinical genetic testing, many were reluctant and had concerns about the adoption. First, challenges to document family history have contributed to divided opinions on adopting clinical genetic services in general practice. Time pressures and limited reimbursement for GPs may render a detailed three-generation family history impractical in their general practice.[2] Inaccuracies and gaps in information obtained from patient about their family history may also pose a problem for GPs to make appropriate genetics referral and screening recommendations. Therefore, interventions targeted at GPs' role in taking family history should seek to use family history as a triage tool to catch rare genetic cases in younger patients or potential hereditary cases with a focus on time-efficiency.

Second, clinical barriers such as rarity of cases, patients' psychological well-being and concerns over the accuracy of genetic results may further exacerbate their reluctance to shoulder an expanded role in clinical genetics. In contrast to taking a family history in the usual primary care context, GPs also reported a lack of clinical practice guidelines on how to assess and discuss genetic risks. Consistent with previous reviews, GPs' lack of knowledge and confidence were commonly identified to be barriers in the provision of clinical genetic services.[5 7 8 12 13] Studies have found that GPs often feel unprepared and lacked confidence due to the rarity of genetic cases in their clinical practice[95 96] and time constraints.[97] Despite these barriers, most GPs had expressed interest to further their education in genetics. Education on the ethical and moral issues surrounding genetic testing was an important proposal. As Bathurst *et al* highlighted, litigation was at the forefront of GPs' practices. Thus, education should seek to address ethical and moral issues surrounding the accuracy of tests, ability to interpret results correctly, disclosure and confidentiality in relation to inherited positive or carrier status for genetic diseases.

While existing reviews have underscored the need for educational interventions,[12 13] findings from web-based intervention studies revealed that education had minimal impact on changing clinical practices. Although web-based educational initiatives were effective in bridging the knowledge gap, such curricula may pose a challenge for time-constrained GPs.[77] Furthermore, findings on the lack of translation from knowledge to practice were not unique as educational initiatives often fail to meet the demands of the everchanging clinical practice guidelines.[11 75] This may pose a potential risk of GPs conveying genetic information to patients without having updated genetic knowledge. Future educational and training should focus on making content relevant to GPs' current clinical practice, through simple and short presentations codeveloped with GPs.[98] It is also worthwhile noting the increasing importance to include epigenetics in GP training to highlight the impact of environmental and behavioural factors which, is presently underemphasised in most genetic courses,[99] coupled with the limited research on GP's understanding of epigenetics.

Moreover, the need for genetic education and training was not echoed by all. Both Haga *et al* and Carroll *et al* found that interest in genetic testing was associated with higher confidence, a positive outlook on genetic medicine and identifying genetics as GPs' responsibility.[52 80] Concurring with this observation, one GP shared his familiarity and experience with clinical genetics as a result of his personal interest which empowered him to play a very important role in advising patients about the risk of getting inherited conditions and how best to test and manage the risk.[4] Hence, it might be more strategic to target clinical genetic interventions at GPs who have special interest in genetics rather than making it mandatory for all GPs.

Strengths of this systematic review include a broad search strategy on varied terms related to clinical genetics and GPs, which increased the likelihood of capturing relevant literature. A range of study designs were also included to increase the heterogeneity of results. However, there were some limitations to this study. Despite the inclusive approach, a limited number of studies were identified. Furthermore, most studies reported a low response rate

and selection bias of GPs with special interest in clinical genetics. In addition, it is likely that positive responses may not be reflective of all GPs views. Most studies also used quantitative methods, which may not capture the nuances in viewpoints, especially since issues revolving around clinical genetics are highly complex.

Many characteristics of GPs put them in an ideal position to facilitate clinical genetic testing. However, based on our findings, it would be unrealistic to expect GPs to adopt clinical genetic practices without adequate support and training. Moreover, not all GPs were found to incorporate clinical genetic testing into their practice due to the uncertainty of their role. Rather than integrating the entire genetic practice into clinical care, GPs may be more inclined to adopt specific genetic practices that are more aligned to their role, and relevant to their patient profile. GPs alone may not have the sufficient expertise and resources to properly engage or manage potential or diagnosed patients with genetic conditions. As such, future interventions could assess the effectiveness of having a multidisciplinary team model that provides an integrated delivery of services involving GPs, genetic counsellors and specialists to alleviate some pressure for GPs who may be daunted by the cost, time and knowledge required in providing clinical genetic services.

Furthermore, educational interventions were found to have minimal impact on GPs adopting clinical genetic practices. This raised the need for future research on alternative strategies targeted at the clinical integration and application of genetic practice. It is also worth noting that variation in healthcare models coupled with mixed attitudes on the utility of genetic testing suggested that not all GPs were receptive to the adoption of clinical genetic services. Thus, future research should examine GPs' perspectives on providing genetic information and in relation to GPs' concerns on the lack of adequate knowledge, training and other ELSI. Future interventions should also aim to understand and contextualise interventions that fit their respective healthcare models to facilitate the smooth adoption of clinical genetic practices.

CONCLUSION

Genetic testing has the potential to revolutionalise primary healthcare and GPs are expected to play a greater role in the provision of clinical genetic services. Yet, this review found that GPs were hesitant to adopt clinical genetics in their practice due to uncertainty over what their role entails which is exacerbated by their lack of knowledge, confidence and rarity of clinical genetic cases. While existing educational interventions were found to increase GPs' knowledge and confidence, they were insufficient to drive the actual adoption of genetic practices in their clinics. The presence of mixed attitudes towards adopting clinical genetics suggests a need for further in-depth research on GPs' concerns. In addition, future research should also take into consideration the variation in healthcare models across different regions, to propose interventions that are contextualised to fit the respective healthcare models.

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ORCID iD

Cheryl Siow Bin Ong http://orcid.org/0000-0002-5264-1964

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Supplementary Table 1. Search results from the databases

Search Strategy on PubMed		
S/N	**Search terms**	**Results**
1	"Genetic Testing"[Mesh]	52,659
2	"Predisposition Test*"[Title/Abstract] OR "Predisposition Genetic Test*"[Title/Abstract] OR "Predictive Test*"[Title/Abstract] OR "Predictive Genetic Test*"[Title/Abstract] OR "Predictive Screening*"[Title/Abstract] OR "Genetic Screening*"[Title/Abstract] OR "Genetic Counsel*"[Title/Abstract] OR "Genetic Service*"[Title/Abstract]	32,188
3	#1 OR #2	77,228
4	(("Physicians, Family"[Mesh]) OR "General Practitioners"[Mesh]) OR "Physicians, Primary Care"[Mesh]	31,077
5	"Fam* Practi*"[Title/Abstract] OR "Fam* Physician*"[Title/Abstract] OR "Family Doc*"[Title/Abstract] OR "General Practi*"[Title/Abstract] OR "General Physician*"[Title/Abstract] OR "Primary Care Physician*"[Title/Abstract] OR "Primary Care Practi*"[Title/Abstract] OR "Primary Care Doc*"[Title/Abstract]	125,257
6	#4 OR #5	137,934
7	"Health Knowledge, Attitudes, Practice"[Mesh]	124,634
8	"Attitude*"[Title/Abstract] OR "Sentiment*"[Title/Abstract] OR "Opinion*"[Title/Abstract] OR "View*"[Title/Abstract] OR "Perception*"[Title/Abstract] OR "Belief*"[Title/Abstract] OR "Feeling*"[Title/Abstract] OR "Experience*"[Title/Abstract] OR "Knowledge*"[Title/Abstract]	2,962,929
9	#7 OR #8	2,999,417
10	"Professional Practice"[Mesh]	265,242
11	"Physician Practice Pattern*"[Title/Abstract] OR "Clinical Practice Pattern*"[Title/Abstract] OR "Physician Prescribing Pattern*"[Title/Abstract] OR "Practice*"[Title/Abstract] OR "Referral*"[Title/Abstract] OR "Consult*"[Title/Abstract]	1,279,051
12	#10 OR #11	1,453,267
13	"Education, Continuing"[Mesh]	62,498
14	"Workshop*"[Title/Abstract] OR "Educational Activit*"[Title/Abstract] OR "Training Program*"[Title/Abstract] OR "Support*"[Title/Abstract] OR "Professional Development*"[Title/Abstract]	1,937,516
15	#13 OR #14	1,990,371
16	#3 AND #6 AND #9	315
17	#3 AND #6 AND #12	373
18	#3 AND #6 AND #15	157
19	#16 OR #17 OR #18	514

20	#19 (Filter from 2010 - 2022)	258
Search Strategy on PsycINFO		
S/N	**Search terms**	**Results**
S1	MA (Genetic Testing)	2,561
S2	TI ((Predisposition Testing*) OR (Predisposition Genetic Testing*) OR (Predictive Testing*) OR (Predictive Genetic Testing*) OR (Predictive Screening*) OR (Genetic Screening*) OR (Genetic Counsel*) OR (Genetic Service*)) OR AB ((Predisposition Testing*) OR (Predisposition Genetic Testing*) OR (Predictive Testing*) OR (Predictive Genetic Testing*) OR (Predictive Screening*) OR (Genetic Screening*) OR (Genetic Counsel*) OR (Genetic Service*))	14,954
S3	#1 OR #2	16,623
S4	MA (Family Physicians) OR MA (General Practitioners) OR MA (Physicians, Primary Care)	6,826
S5	TI ((Fam* Practi*") OR (Fam* Physician*) OR (Family Doc*) OR (General Practi*) OR (General Physician*) OR (Primary Care Physician*) OR (Primary Care Practi*) OR (Primary Care Doc*)) OR AB ((Fam* Practi*") OR (Fam* Physician*) OR (Family Doc*) OR (General Practi*) OR (General Physician*) OR (Primary Care Physician*) OR (Primary Care Practi*) OR (Primary Care Doc*))	7,463
S6	#4 OR #5	12,105
S7	MA (Health Knowledge, Attitudes, Practice)	27,856
S8	TI ((Attitude*) OR (Sentiment*) OR (Opinion*) OR (View*) OR (Perception*) OR (Belief*) OR (Feeling*) OR (Experience*) OR (Knowledge*)) OR AB ((Attitude*) OR (Sentiment*) OR (Opinion*) OR (View*) OR (Perception*) OR (Belief*) OR (Feeling*) OR (Experience*) OR (Knowledge*))	1,704,921
S9	#7 OR #8	1,711,670
S10	MA (Professional Practice)	3,502
S11	TI ((Physician Practice Pattern*) OR (Clinical Practice Pattern*) OR (Physician Prescribing Pattern*) OR (Practice*) OR (Referral*) OR (Consult*)) OR AB ((Physician Practice Pattern*) OR (Clinical Practice Pattern*) OR (Physician Prescribing Pattern*) OR (Practice*) OR (Referral*) OR (Consult*))	582,019
S12	#10 OR #11	583,422
S13	MA (Education, Continuing)	3,084
S14	TI ((Workshop*) OR (Educational Activity*) OR (Training Program*) OR (Support*) OR (Professional Development*)) OR AB ((Workshop*) OR (Educational Activity*) OR (Training Program*) OR (Support*) OR (Professional Development*))	869,843
S15	#13 OR #14	871,475
S16	S3 AND S6 AND S9	63
S17	S3 AND S6 AND S12	72
S18	S3 AND S6 AND S15	27

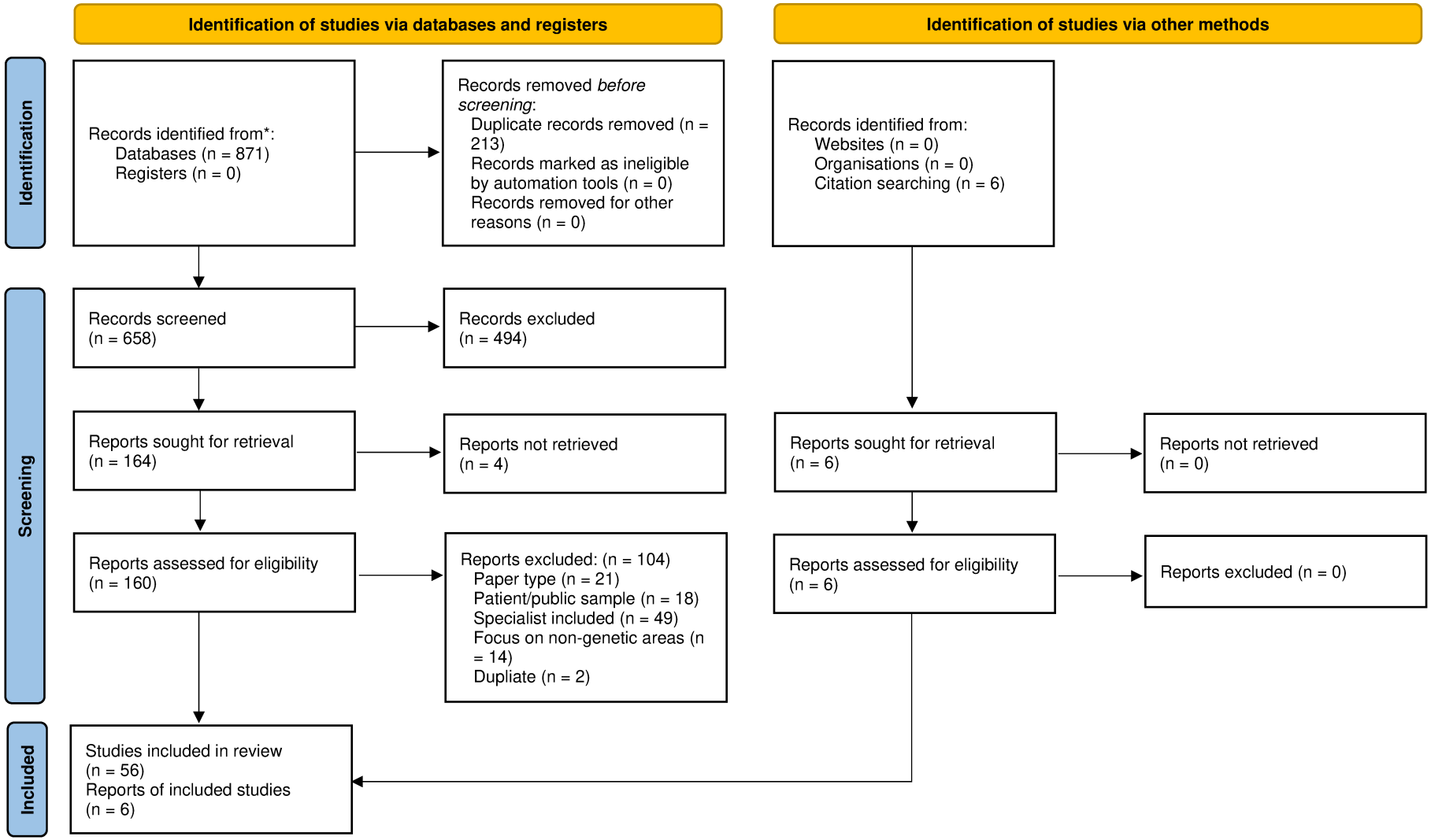
S19	S16 OR S17 OR S18	100
S20	S19 (Filter from 2010 - 2022)	48
Search Strategy on EMBASE		
S/N	**Search terms**	**Results**
1	genetic testing.mp.	48,498
2	(Predisposition Testing* or Predisposition Genetic Testing* or Predictive Testing* or Predictive Genetic Testing* or Predictive Screening* or Genetic Screening* or Genetic Counsel* or Genetic Service*).ti. or (Predisposition Testing* or Predisposition Genetic Testing* or Predictive Testing* or Predictive Genetic Testing* or Predictive Screening* or Genetic Screening* or Genetic Counsel* or Genetic Service*).ab.	38,295
3	#1 OR #2	78,986
4	(family physicians or general practitioners or primary care physician).mp.	65,542
5	(Fam* Practi* or Fam* Physician* or Family Doc* or General Practi* or General Physician* or Primary Care Physician* or Primary Care Practi* or Primary Care Doc*).ti. or (Fam* Practi* or Fam* Physician* or Family Doc* or General Practi* or General Physician* or Primary Care Physician* or Primary Care Practi* or Primary Care Doc*).ab.	153,295
6	#4 OR #5	153,676
7	Health Knowledge, Attitudes, Practice.mp.	289
8	(Attitude* or Sentiment* or Opinion* or View* or Perception* or Belief* or Feeling* or Experience* or Knowledge*).ti. or (Attitude* or Sentiment* or Opinion* or View* or Perception* or Belief* or Feeling* or Experience* or Knowledge*).ab.	3,368,287
9	#7 OR #8	3,368,347
10	Professional Practice.mp.	53,808
11	(Physician Practice Pattern* or Clinical Practice Pattern* or Physician Prescribing Pattern* or Practice* or Referral* or Consult*).ti. or (Physician Practice Pattern* or Clinical Practice Pattern* or Physician Prescribing Pattern* or Practice* or Referral* or Consult*).ab.	1,613,246
12	#10 OR #11	1,646,549
13	Education, Continuing.mp.	216
14	(Workshop* or Educational Activity* or Training Program* or Support* or Professional Development*).ti. or (Workshop* or Educational Activity* or Training Program* or Support* or Professional Development*).ab.	2,220,602
15	#13 OR #14	2,220,726
16	#3 AND #6 AND #9	407
17	#3 AND #6 AND #12	528
18	#3 AND #6 AND #15	210

19	#16 OR #17 OR #18	710
20	#19 (Filter from 2010 - 2022)	490
Search Strategy on Cochrane		
S/N	**Search terms**	**Results**
1	"Genetic Testing"[Mesh]	
2	(Predisposition Testing* or Predisposition Genetic Testing* or Predictive Testing* or Predictive Genetic Testing* or Predictive Screening* or Genetic Screening* or Genetic Counsel* or Genetic Service*).ti.ab.kw	
3	#1 OR #2	
4	(("Physicians, Family"[Mesh]) OR "General Practitioners"[Mesh]) OR "Physicians, Primary Care"[Mesh]	
5	(Fam* Practi* or Fam* Physician* or Family Doc* or General Practi* or General Physician* or Primary Care Physician* or Primary Care Practi* or Primary Care Doc*).ti.ab.kw	
6	#4 OR #5	
7	"Attitude"[Mesh]	
8	(Attitude* or Sentiment* or Opinion* or View* or Perception* or Belief* or Feeling* or Experience* or Knowledge*).ti.ab.kw	
9	#7 OR #8	
10	"Practice Patterns, Physicians'"[Mesh]	
11	(Physician Practice Pattern* or Clinical Practice Pattern* or Physician Prescribing Pattern* or Practice* or Referral* or Consult*).ti.ab.kw	
12	#10 OR #11	
13	"Education"[Mesh]	
14	(Workshop* or Educational Activity* or Training Program* or Support* or Professional Development*).ti.ab.kw	
15	#13 OR #14	
16	#3 AND #6 AND #9 (Filter from 2010 - 2022)	19
17	#3 AND #6 AND #12 (Filter from 2010 - 2022)	34
18	#3 AND #6 AND #15 (Filter from 2010 - 2022)	22

Supplementary Table 2. Inclusion/Exclusion criteria

Inclusion Criteria	
Types of Genetic Testing	All types
Paper Type	Original research, peer-reviewed journals - Full text of paper available - Global literature
Time Frame	2010 –2022
Study Population	Primary Care Physicians (PCPs), Family Physician, General Practitioner (GP)
Focus of Paper	GPs' experiences - Any or the lack thereof discussions on genetics - Subjective experiences such as confidence, comfort, knowledge, barriers GPs' attitudes - Opinions on their role in offering clinical genetic services - Awareness - General views on utility of genetic testing GPs' needs - Educative workshops or practice policies and recommendations targeted at incorporating aspects of clinical genetic services that can include taking family history, recommending and ordering tests, interpreting test results, managing downstream care, and referral to clinical genetic centres
Exclusion Criteria	
Paper Type	Commentaries, short articles, dissertations, book reviews, literature reviews, mini reviews, book chapters, editorials
Language	Any language, other than English
Study Population	Public, Patients, Specialist (Pediatricians, Ob-gyn, Oncologist, Geneticists) and Allied Health Professionals (nurses, health educators, social workers)
Focus of Paper	Testing of medical and clinical interventions, other than those targeted at genetic education

Figure 1. PRISMA flow diagram of the study selection process



Supplementary Table 3. Characteristics of included studies (n = 62)

Category	Studies, n (%)
Study Type	
Quantitative	36 (58.1%)
Qualitative	13 (21.0%)
RCT	9 (14.5%)
Mixed methods	4 (6.5%)
Country	
United States	20 (32.3%)
United Kingdom	19 (30.6%)
Canada	11 (17.7%)
Australia	6 (9.7%)
Asia	4 (6.5%)
South Africa	1 (1.6%)
New Zealand	1 (1.6%)
Sample Size	
< 50 respondents	21 (33.9%)
50 – 100 respondents	12 (19.4%)
101 – 500 respondents	21 (33.9%)
> 500 respondents	8 (12.9%)
Area of Focus *(overlaps in articles, n>100%)*	
Knowledge, experiences	50 (80.6%)
Attitudes, views, roles	38 (61.3%)
Needs (education, interventions)	43 (69.4%)
Genetic Type	
General genetics	24 (38.8%)
Oncogenetics	18 (29.0%)
Cardiovascular	6 (9.7%)
Pharmacogenetics (PGx)	6 (9.7%)
Prenatal/Neonatal	4 (6.5%)
Direct-to-consumer (DTC) testing	4 (6.5%)
Study Quality	Avg: 4.5 *(range: 4-5)*
Response rate (>50%)	11 (17.7%)
Not reported	22 (35.5%)

Supplementary Table 4. Assessment of risk of bias using the MMAT

Authors (year)	S1.	S2.	Q1	Q2	Q3	Q4	Q5
Qualitative study	Are there clear research questions?	Do the collected data allow to address the research questions?	Is the qualitative approach appropriate to answer the research question?	Are the qualitative data collection methods adequate to address the research question?	Are the findings adequately derived from the data?	Is the interpretation of results sufficiently substantiated by data?	Is there coherence between qualitative data sources, collection, analysis and interpretation?
Cusack et al. (2021)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Douma et al. (2019)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Fok et al. (2021)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Hussein et al. (2020)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Joshi et al. (2020)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Lemke et al. (2017)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Lemke et al. (2020)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Mathers et al. (2010)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
McKinn et al. (2022)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Puzhko et al. (2019)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Sebastian et al. (2022)	Yes	Yes	Yes	Yes	Yes	Yes	Yes

Sebastian et al. (2022)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Silva et al. (2022)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Tsianakas et al. (2010)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Quantitative RCT	Are there clear research questions?	Do the collected data allow to address the research questions?	Is randomization appropriately performed?	Are the groups comparable at baseline?	Are there complete outcome data?	Are outcome assessors blinded to the intervention provided?	Did the participants adhere to the assigned intervention?
Bell et al. (2015)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Bell et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Carroll et al. (2011)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Houwink et al. (2015)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Houwink et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Houwink et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Wilkes et al. (2017)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Wilson et al. (2016)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Quantitative descriptive	Are there clear research questions?	Do the collected data allow to address the research questions?	Is the sampling strategy relevant to address the research question?	Is the sample representative of the target population?	Are the measurements appropriate?	Is the risk of nonresponse bias low?	Is the statistical analysis appropriate to answer the research question?

Barrow et al. (2015)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Bernhardt et al. (2012)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Bonham et al. (2010)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Carroll et al. (2019)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Carroll et al. (2016)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Challen et al. (2010)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Dunlop et al. (2010)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Evans et al. (2020)	Yes	Yes	Yes	Yes	Yes	Can't tell (No sampling frame)	Yes
Fiederling et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Haga et al. (2019)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Haga et al. (2012)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Haga et al. (2011)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Haga et al. (2017)	Yes	Yes	Yes	Yes	Yes	Can't tell (No sampling frame)	Yes
Kadaoui et al. (2012)	Yes	Yes	Yes	Yes	Yes	Yes	Yes

Klemenc-Ketiš et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Klemenc-Ketiš et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Laedtke et al. (2012)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Leitsalu et al. (2011)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Mainous et al. (2013)	Yes	Yes	Yes	Yes	Yes	Can't tell (Lack demographic data)	Yes
Marathe et al. (2015)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Nippert et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Nippert et al. (2011)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Pelletier et al. (2020)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Powell et al. (2012)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Ram et al. (2012)	Yes	Yes	Yes	Yes	Yes	Can't tell (Non-respondent bias risk not reported)	Yes
Rangarajan et al. (2016)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Richter et al. (2013)	Yes	Yes	Yes	Yes	Yes	Yes	Yes

Saul et al. (2017)	Yes	Yes	Yes	Yes	Yes	Can't tell (No sampling frame)	Yes
Skinner et al. (2021)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
St Sauver et al. (2016)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Teng et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Truong et al. (2021)	Yes	Yes	Yes	Yes	Yes	Can't tell (No sampling frame)	Yes
Van Wyk et al. (2016)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Vande Perre et al. (2018)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Vansenne et al. (2011)	Yes	Yes	Yes	Yes	Yes	Can't tell (No data on non-respondents)	Yes
Yu et al. (2021)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Mixed Methods	Are there clear research questions?	Do the collected data allow to address the research questions?	Is there an adequate rationale for using a mixed methods design to address the research question?	Are the different components of the study effectively integrated to answer the research question?	Are the outputs of the integration of qualitative and quantitative components adequately interpreted?	Are divergences and inconsistencies between quantitative and qualitative results adequately addressed?	Do the different components of the study adhere to the quality criteria of each tradition of the methods involved?
Dressler et al. (2019)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Lemke et al. (2020)	Yes	Yes	Yes	Yes	Yes	Yes	Yes

Mazzola et al. (2019)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Schuurmans et al. (2019)	Yes	Yes	Yes	Yes	Yes	Yes	Yes

Supplementary Table 5. Summary of key findings

S/N	Author	Title	Methods, Sample size, Country	Genetic scope	Knowledge/Experience	Attitude/Views/Roles	Needs
1	McKinn et al. 2022	Clinician views and experiences of non-invasive prenatal genetic screening tests in Australia.	Qualitative (Semi-structured interview) 4 GPs 15 F, 2 M Response rate: not reported Australia	Non-invasive prenatal screening (NIPS)	- Limited experience with high chance NIPS results - Did not often identify or voice concerns about potential harms of NIPS - Reported limited time for pre-test counselling in the context of first trimester antenatal appointments		- Some GPs suggested a mandatory training on how to discuss NIPS and disseminate the results - There also needs to be more specific guidance for GPs on the use of NIPS for screening (those currently available are focused on screening for T21), and a national system to collect routine data on NIPS requests
2	Silva et al. 2022	Introducing genetic testing with case finding for familial hypercholesterolaemia in primary care: qualitative study of patient and health professional experience.	Qualitative (Semi-structured interview) 7 GPs 13 F, 11 M Response rate: not reported UK	Familial hypercholesterolaemia (FH)	- Comfortable to refer patients with results suggesting FH or a variant of unknown significance (VUS) for specialist assessment	- Positively anticipated the value of improving identification of FH, recognising potential benefits for patients and their families' long-term health	- Sought greater understanding about interpreting and communicating the range of possible test results, and more in-depth guidance on long-term care of FH (conditions, next steps by specialists) - Anticipated a need for clearer guidance about evolving roles at the primary–secondary care interface, especially guidance on who may have what clinical responsibilities or duty of care related to genetic testing for FH, and communicating and acting on results appropriately.

3	Cusack et al. 2021	General practitioners' views on genomics, practice and education: A qualitative interview study.	Qualitative (Semi-structured interview) 28 GPs 12 F, 16 M Response rate: not reported Australia	Genomic testing	- Only 3 GPs attended continuing professional development activities on genetics/genomics - Most GPs reported little experience with genetic or genomic testing - 3/4 reported referring patients to genetic services or specialists - Lack of evidence and reliability of online DNA testing was a concern for some GPs, who stated they lacked confidence interpreting results to support their patients - Challenge such longer consultation times, cost of genomic tests	- Most felt their role was to assist or counsel patients to help them understand these types of tests and results, and to refer or seek advice from genetic specialists as required - Predict genomics to play a bigger part in their future practice, especially for risk prediction and to inform treatment and management; but a small number were uncertain.	- Need for more education, training and support resources such as clear, up-to-date guidelines on genomic testing; decision supports; RACGP resources; patient handouts; and opportunities to discuss issues with a genetic specialist - Interested to learn about genomics with relevance to their practice - Prefer case studies, face-to-face events (seminar, workshops), online learning, journals, accredited CPD events, webinars and podcasts.
4	Fok et al. 2021	How practice setting affects family physicians' views on genetic screening: a qualitative study.	Qualitative (Semi-structured interview) 30 FPs 14 F, 16 M Response rate: 75% Singapore	Genetic screening	- Perceived level of confidence to conduct GS was low due to lack of training and knowledge - Public barriers (Lack of control, Lower patient socioeconomic status and literacy, Rigid administrative infrastructure) - Private motivations (Strong longitudinal patient relationship, Practice autonomy, Higher patient literacy)	- Generally perceived themselves to be well-positioned to offer GS but expressed ambivalence about their current roles and competency to practise GS - Some perceived that offering GS was not core to their scope of practice due to rarity of genetic conditions. - Negative patient attitudes as a potential barrier (emotional and psychological burden)	- GS adoption would be greater if Continuing Medical Education (CME) and other educational and systems support were offered

5	Truong et al. 2021	Genetic Referral Patterns and Responses to Clinical Scenarios: A Survey of Primary Care Providers and Clinical Geneticists.	Quantitative (Survey) 95 PCPs 61 F, 34 M Response rate: not reported US	General genetic testing	- 48% indicated that they would recommend genetics evaluation, genetic counselling, and/or genetic testing for developmental delay - 71% would recommend for colon and uterine cancer - Concerns for financial cost to patients was the most common barrier		- 78% preferred either or both online continuing medical education (CME) activities and online medical references sites as methods for obtaining genetic information

6	Yu et al. 2021	Preparing genomic revolution: Attitudes, clinical practice, and training needs in delivering genetic counseling in primary care in Hong Kong and Shenzhen, China.	Quantitative (Survey) 151 Hong Kong PCPs 48 F, 103 M Response rate: 8% 258 Shenzhen PCPs 145 F, 113 M Response rate: 37% Hong Kong, Shenzhen, China	Genomic medicine	- 17% HK-PCPs and 40% SZ-PCPs had encountered patient cases related to genomic medicine in the past 6 months - HK-PCPs were most confident in "obtaining information about genetic disorders from FH" and least confident to decide which "genetic testing should be done" - SZ-PCPs were most confident in referring patient to "a relevant specialist for suspected genetic disorders" and least confident in "explaining to patients on genetic testing results" and "advising patients whether they should do genetic test". - 55% expressed insufficient time during clinical consultation to discuss genetic issues - 78% were unaware of the referral pathway for patients with suspected and confirmed genetic disorder	- 91% agreed that it was important to keep up to date with the latest information on genetic disorders - 86% agreed that personalized medicine is the future of healthcare - About 80% of PCPs felt that breast, ovarian and colorectal cancers and congenital anomalies were conditions worth performing genetic testing - 68% perceived ethical controversies associated with genetic testing	

7	Joshi et al. 2020	Primary care provider perspectives on using genomic sequencing in the care of healthy children.	Qualitative (Semi-structured interview) 11 FPs and 5 primary care pediatricians Response rate of 69% (11/16), 31% (5/16) Canada	Genome sequencing (GS)	- Many providers felt they lacked the necessary technical expertise and skills to convey GS results to the parents (felt unfamiliar with genetic concepts and expressed discomfort with interpreting and using GS results)	- Most PCPs saw value in using GS in research for healthy children but diverged in opinion on using results in primary care for children - Proponents saw value in informing their patients' preventative care and benefiting scientific research as a whole - Had more dynamic definitions of actionability (interventions to reduce morbidity and mortality) - Skeptics were driven by providers' ambivalence about using a research test and uncertainty about what to do with the result - Did not feel they had a professional obligation to use GS results in primary care - Aligned with traditional, restrictive definitions of actionability	- Additional resources required to facilitate GS testing, pretest and posttest counseling, and additional support or training for themselves - Additional resources incurred costs, which could over-burden the healthcare system

8	Pelletier et al. 2020	Survey of primary care physicians' views about breast and ovarian cancer screening for true BRCA1/2 non-carriers.	Quantitative (Survey) 134 FPs 76 F, 58 M Response rate: 45% Canada	Breast and ovarian cancer screening (*BRCA1/2*)	- FPs were more likely than gynecologists to recommend unproven ovarian cancer screening to a carrier but less likely to recommend proven MRI screening.		
9	Lemke et al. 2020	Primary care physician experiences utilizing a family health history tool with electronic health record-integrated clinical decision support: an implementation process assessment.	Qualitative (Semi-structured interview) 24 PCPs 19 F, 5 M Response rate: not reported US	Genetic and Wellness Assessment (GWA), CDS alert tool	- Expressed concern on the amount of time needed to discuss the alert recommendation due to busy practices and patient having difficulties answering family history questions - Lack of follow-up on the testing and referrals due to cost, insurance concerns, fear, stigma, lack of interest, and logistical issues - Alert fatigue; CDS recommendations differing from their clinical judgment; and technical issues		- GWA helped increase patient awareness of the importance of their family history - Facilitated patient-physician discussions about disease risk by providing CDS alert so that PCPs receives specific information about genetic testing, personalized medicine services available, and next steps within the health system - Need for more physician education about the GWA CDS recommendations

10	Evans et al. 2020	How genomic information is accessed in clinical practice: an electronic survey of UK general practitioners.	Quantitative (Survey) 159 GPs Response rate: not reported UK	General genetic testing			- Majority wanted to keep up to date with genomic medicine via online educational modules (70%); willing to spend 30min to 1 hour (78%) on it - More than 60% choose NICE Clinical Knowledge Summary (CKS) and GP notebook for FH and FBC scenarios; Internet search engines was next most popular; far fewer (19%) access government webpage for information - Local clinical genetic services or seeking advice from specialists/secondary care colleagues were most common
11	Dressler et al. 2019	Implementing pharmacogenetic testing in rural primary care practices: a pilot feasibility study.	Mixed methods (Survey & interviews) 4 PCPs Response rate: not reported US	Individualised training and education on PGx testing	- None of the PCPs ordered a PGx test prior to the study as they did not know/have access to a lab that performed PGx testing, not seeing convincing evidence of clinical utility, and not feeling confident to interpret and apply results in treatment decision - Pricing continues to be concern and barrier for physicians	- Different views on how PGz can enhance patient care; some prefer pre-emptive testing while some prefer testing at point of care when needed to predict response to drug	- Individualized PGx training provided by the PM pharmacist showed a boost in physician's comfort level with PGx testing - Shift in barriers from lack of expertise, lack of comfort to more practical issues of costs, and issues related to electrical medical records - Clinical interpretative summaries provided by the PM Pharmacist were very helpful

12	Schuurmans et al. 2019	Feasibility of couple-based expanded carrier screening offered by general practitioners.	Mixed methods (Survey & interviews) 10 GPs (interview), 116 GPS (checklist) Response rate: 90% The Netherlands	Expanded carrier screening (ECS)	- Over time they developed a routine for conducting the counselling, which reduced the time required for preparation and counselling itself - GPs did not experience any barriers in communicating the normal results or to referring any couples at normal risk to Clinical Genetics for additional pre- or post-test counselling	- Most GPs were positive about combining ECS pre-test counselling with GPC - GPs considered themselves as the most suitable providers for a population-based ECS couple-test. - ECS-provision as standard care by all GPs might not be feasible because not all may be able to keep up with technological advances; only motivated GPs willing to do so should be trained to provide ECS - Some were resistant to additional workload due to busy practices	- All GPs interviewed said they felt able to provide the pre-test counselling mainly because of the training, supervision and additionally provided materials such as study checklist as a practical guidance
13	Douma et al. 2019	Information exchange between patients with Lynch syndrome and their genetic and non-genetic health professionals: whose responsibility?	Qualitative (Interview) 6 GPs Response rate: not reported The Netherlands	Lynch syndrome (LS)	- Generally followed the patient's request to be referred for genetic counselling and rely on the cancer family history that patients provide on their own initiative - Provide very little explanation about LS to their patients at the time of referral, as they lacked the knowledge - Several GPs were not regularly informed by GEs about the endoscopic surveillance, while others reported to receive letters or were unsure about whether	- Felt responsible for referring patients for follow-up care and also for providing support. - Did not perceive this to be their responsibility	- Like to have rapid access to information and information specifically tailored for GPs. - GPs appreciated the letter from the genetic HP; generally, they only had contact with the gastroenterologists via letters.

					they were informed by the gastroenterologists.		
14	Vande Perre et al. 2018	Role of the general practitioner in the care of BRCA1 and BRCA2 mutation carriers: General practitioner and patient perspectives.	Quantitative (Survey) 58 GPs 24 F, 34 M Response rate: 38.2% UK	BRCA1/2	- 81% collected the family history - 24% considered they had sufficient knowledge of the indication criteria for genetics consultation - 69.7% considered that they were not able to answer patients' questions about BRCA1/2 guidelines - 75.9% were not familiar with the criteria for referring patients to cancer genetics consultations	- Many (72.4%) felt not included or that they had a minor role (31%) in the care of their patients - 72.4% saw their role in caring for these patients is limited to psychological support and to motivate relatives to undergo screening (70.7%).	- 27.5% were trained during their initial training to care for patients with a BRCA1/2 mutation - Only 11.8% of the GPs attributed their knowledge on the subject to the referral guidelines of the French national cancer institute (INCa). - 32.8% reported receiving a letter from the geneticist - 79.6% are interested in training

15	Wilkes et al. 2017	Increasing confidence and changing behaviors in primary care providers engaged in genetic counselling.	RCT (survey) 121 PCPs - 60 intervention; 61 control 40.5% F, 59.5% M Response rate: 3.5% US	BRCA, genetic testing, perinatal			- Interactive web-based CME was more effective at improving knowledge and shared decision making behaviors but had a small effect on attitudes and minimal impact on clinical behaviours on ELSI discussions - Intervention showed greater increase in knowledge, more satisfied with educational materials and more confident in their ELSI genetic knowledge and skills - Self-efficacy improved in both groups; intervention showed significantly higher improvements

16	Lemke et al. 2017	Primary care physician experiences with integrated pharmacogenomic testing in a community health system.	Qualitative (Semi-structured interview) 15 PCPs 60% F, 40% M Response rate: not reported US	Pharmacogenetic (PGx) testing	- PGx testing results were used to adjust patient medications to increase effectiveness and reduce side effects - Lack of understanding of the pharmacogenomics test report and how to interpret, not adequately prepared to communicate complex results - Delay to receive results was a barrier in providing timely patient feedback - Time constraints as a challenge and the need for an in-office follow-up appointment to discuss results	- PGx testing could help individualize medication treatments for their patients - Utility of PGx testing was helpful for patients to potentially avoiding medication side effects and guide decision-making for patients starting a new medication - Using PGx direct access testing can foster increased patient autonomy and satisfaction (more efficient and save the additional costs), and assurance on medication plan - Few did not think PGx testing was useful in their patient population now but will be more valuable in the future - High cost and lack of reimbursement for patients	- Undergoing direct access PGx testing themselves was a useful teaching tool and that it was helpful for them to have first-hand knowledge of the testing and resulting process - Desire for clarification on the results report and preferred certain formats for results display as well as a paper copy of the results - More PGx education (such as in-services, case studies, and online training) to guide on how to address cost and insurance issues with patients - Further training specific to results report interpretation - Interested in receiving both provider and patient education materials (colourful pamphlets, etc.)

17	Haga et al. 2017	Primary care providers' use of pharmacist support for delivery of pharmacogenetic testing.	Quantitative (Survey) 12 PCPs US	Pharmacogenetic (PGx) testing	- 58% reported ordering genetic testing for disease diagnosis one-time or two-times per year - All 12 GPs indicated that they did not feel well informed about genetic testing in general nor about PGx testing specifically - 2 GPs felt comfortable to discuss PGx testing prior to ordering test, 3 GPs felt somewhat comfortable to discuss PGx test results, 3 GPs felt comfortable using PGx test to inform treatment decisions	- 83% believed that pharmacists would have some or a large role in delivering PGx; 75% believed that geneticists/genetic counselors would have some or a large role in delivering PGx testing - 42% believed that the physician who ordered a PGx test should communicate test results to the patient - 5 GPs believed that either the ordering physician, a genetic counselor or a pharmacist could communicate PGx results	- Awareness on PGx was gained from professional meetings, drug or laboratory representative, publications, CME learning, grand rounds or point-of-care notification - 75% prefer to learn about PGx through grand rounds or other in-house seminars - 92% indicated having some assistance in interpretation would increase likelihood to order a PGx test - 63% consulted pharmacist; providers who did not consult the pharmacist did so because they did not feel they needed pharmacist's input or they did not have time - All agreed that having a pharmacist available is helpful (meet patients, more learning opportunities with pharmacist, mail written summary of test results)
18	Wilson et al. 2016	Supporting genetics in primary care: investigating how theory can inform professional education.	RCT (survey) 96 PCPs Response rate: 76.8% Canada	Hereditary breast and ovarian cancer (HBOC)	- FPs' intentions were lower for 'making a risk assessment' (perceived as the most difficult, saw no value, felt no pressure or confidence to do it) than for the other two behaviours ('taking family history' and 'making referral')	- Taking family history seen positively as a normal activity for FPs; but a proportion were sceptical if this should be part of their practice due to lack of confidence to take FH	

19	St Sauver et al. 2016	Integrating Pharmacogenomics into Clinical Practice: Promise vs Reality.	Quantitative (Survey) 90 PCPs Response rate: 57% US	Pharmacogenomics (PGx)			- 52% did not expect or know how to use pharmacogenetic information in future practices - Of those that received alert, 53% felt that alerts were confusing, irritating or difficult to find additional information - Only 30% changed their prescription in response to PGx alert
20	Van Wyk et al. 2016	Knowledge regarding basic concepts of hereditary cancers, and the available genetic counselling and testing services: A survey of general practitioners in Johannesburg, South Africa.	Quantitative (Survey) 61 PCPs Response rate: 31.1% South Africa	Hereditary cancer (HBOC, LS, FAP)	- GPs have limited knowledge about basic concepts of hereditary cancers and local genetic facilities available. - Majority were unsure how to perform risk assessments; only 36% would refer to appropriate cancer genetic services - 65% felt that they were not sufficiently qualified and equipped to provide genetic counselling; and agreed that genetic counsellors (100%), medical geneticist (85%) and oncologist (68%) were more qualified	- Most GPs are interested to learn more or become more involved in referring at-risks patients appropriately. - Most agreed that patients should have counselling before testing.	- Important resources includes: CME (86%), discussion with colleagues (82%), guidelines (82%), published data, journals (88.5%)

21	Marathe et al. 2015	General Practitioners' knowledge and use of genetic counselling in managing patients with genetic cardiac disease in non-specialised settings.	Quantitative (Survey) 144 GPs 73 F, 71 M Response rate: 21% Australia	Genetic cardiac diseases (GCDs)	- 51.4% feel confident in educating patients with GCDs but 29.3% were unsure - 39.6% were not confident to answer patient's questions about GCD - 56% did not feel confident with the knowledge they have regarding GCDs but 56% were confident with their knowledge in appropriately managing GCDs in their clinical practice - 76.1% routinely educated patients and their relatives - 86.7% had heard about the Tasmanian Genetic Counselling Service but 52.8% knew little about the service provided - Variations in referral: 37.3% said that they sometimes referred, 26.8% did routinely refer, 14.8% did not routinely refer, and 10.6 % only referred if the patient asked for it	- 100% agreed that it is important to educate patients about their genetic condition - 95.1% also agree that it is important to educate family members about genetic conditions - 94.3% mentioned cardiologist or specialist as being most important in the team of GCD care providers for guidance, 2 GPs also saw it as the cardiologists' role to refer	- Education was needed through monthly newsletter or in the form of creating clinical pathways to assist in referring appropriately

22	Barrow et al. 2015	Improving the uptake of predictive testing and colorectal screening in Lynch syndrome: a regional primary care survey.	Quantitative (Survey) 63 GPs Response rate: 29.2% UK	Lynch syndrome (LS)	- 77.8% had no previous experience of referring a patient/family with suspected LS to the Regional Genetics Service, 79.4% were unclear which patients should be referred for investigation - 73.0% were unaware of the Regional Lynch Syndrome Registry - 61.9% had no experience of discussing cancer risk, 38.1% had no experience discussing screening recommendations - 87.3% did not feel confident to discuss the details of LS - 57.1% had concerns over confidentiality which would prevent them from approaching at-risk relatives - Barriers includes lack of knowledge and time constraints (41.3%)	- 49.2% did not feel this was part of their role - 90.5% felt that patients themselves had the most responsibility for adhering to the recommended screening guidelines although 50.8% identified this as part of their role also - Shared responsibility among healthcare professionals, including the Regional Genetics Service, the gastroenterologist/colorectal surgeon and GP, with most responsibility for screening lying with the screening centres.	- 74.6% highlighted the lack of supporting literature to facilitate the discussion

23	Bell et al. 2015	Impact of a randomized controlled educational trial to improve physician practice behaviors around screening for inherited breast cancer.	RCT (survey) 121 PCPs - 60 intervention; 61 control 40.5% F, 59.5% M Response rate: 3.5% US	BRCA			- Intervention had minimal impact on practices to offer genetic counselling but with a few exceptions: - Intervention-physicians were more likely to explore genetic counseling benefits; advise for a test decision after counselling; and inform that postiive results would indicate increased risk of prostate cancer for male relatives - Intervention-physicians were less like to ask about Ashkenazi heritage - Specific questions about cancers in the family, including ovarian, breast, and prostate cancers, were not usually asked. - Cost, implications of treatment, and limitations of current genetic testing were not usually discussed. - Majority offered referral to geneticists

24	Teng et al. 2014	Attitudes and knowledge of medical practitioners to hereditary cancer clinics and cancer genetic testing.	Quantitative (Survey) 32 GPs Response rate: 25% Australia	Cancer genetic testing	- 87.5% have referred patients for cancer genetic testing (GPs referred 1 in 790 patients) - 60% correctly estimated the cost of the first family member (proband) to undergo cancer genetic testing - 20% correctly estimated turnaround time for routine cancer genetic testing, and 30% for urgent cancer genetic testing - Wide discrepancy between the self-reported GP referral rate (87.5 %) and the actual referral rate calculated from patient files (12.5 %) - Reasons for not referring: no treatment, no patient request for it		- 84 % wanted more information

25	Houwink et al. 2014	Effectiveness of oncogenetics training on general practitioners' consultation skills: a randomized controlled trial.	RCT (survey) 56 GPs (38 intervention, 18 control group) 41 F, 15 M Response rate: 64% (56/88) The Netherlands	Oncogenetics			- Case-based oncogenetics education can achieve sustained improvement (3 mths after the training) - Positive results for active and interactive sessions, single-group and smaller-group sessions - Participating GPs seemed to be more comfortable incorporating oncogenetics into patient consultation skills (high applicability skills)
26	Houwink et al. 2014	Sustained effects of online genetics education: a randomized controlled trial on oncogenetics.	RCT (survey) 44 GPs 39 F, 5 M Response rate: 55% The Netherlands	Oncogenetics			- Online genetics CPD module can result in sustained improvement of genetics knowledge - More than 90% applied newly acquired knowledge at least once a month - Self-reported applicability aspects focused indicates that the G-eCPD mainly improved genetics knowledge rather than skills

27	Nippert et al. 2014	Cancer risk communication, predictive testing and management in France, Germany, the Netherlands and the UK: general practitioners' and breast surgeons' current practice and preferred practice responsibilities.	Quantitative (Survey) 1197 GPs Gender reported UK (France, Germany, the Netherlands and the UK)	BRCA 1/2	- Majority reported that a cancer family history is raised in a consultation "at least once a week"/"once a month" - GPs from Germany (76.6%) and France (74.3%) reported that they would always take a family history whereas only 36.0% of the Dutch and 40.1% of the British GPs reported always taking FH. - Majority reported that they "always"/"frequently" provide risk assessment	- Majority of the GPs from Germany, the Netherlands and the UK considered practice responsibility should be "to provide support after breast cancer testing" - GPs from France ascribed to the following tasks: "explain the inheritance pattern of familial breast cancer", "inform about breast cancer genetic risk for the relatives", "inform about breast cancer genetic testing", "provide support after breast cancer genetic testing", and "inform about possible management options available after the results of breast cancer genetic testing". - GPs from all countries unanimously agreed that "disclose breast cancer genetic test results to the patient" should be undertaken by a genetic specialist.	
28	Fiederling et al. 2014	Consideration of family history of cancer in medical routine: a survey in the primary care	Quantitative (Survey) 35 GPs Response rate: 70% Germany	Family history of cancer (FHC)	- 53% reported that they only ask for FHC in general, but not for a specific cancer site. Those who noted asking for specific cancer sites most frequently asked for a family history of breast or CRC - 97% would screen according to general guidelines and		- 57% did not feel there is a need for standardized tool to collect information on FHC - 60% feel that there is a need for further information or guidelines regarding preventive counselling of individuals with a FHC - Most prefer either

		setting in Germany.			79% would give recommendations for a healthy lifestyle, only 35% would refer to a specific counselling centres		flyer/booklet (17%) or computerized tool (14%) for preventive counselling
29	Mainous AG 3rd et al. 2013	Academic family physicians' perception of genetic testing and integration into practice: a CERA study.	Quantitative (Survey) 1,404 PCPs 45% F, 55% M Response rate: 45.1% Canada, US	Heart disease, breast cancer, diabetes, hemochromatosis, alzheimer, DTC	- Majority were not confident in their knowledge on available genetic testing even though they anticipate GT to have substantial impact on future clinical practice.	- Majority (71.8%) felt that genetic testing was valuable to test patient's risk for disease but less so to determine suitable treatment for patient - Self-perceived knowledge was positively associated with prediction on impact of GT, and importance of GT curriculum - 58.1% felt that DTC was more likely to harm patients' general health decisions	- Many felt that GT education is important.
30	Laedtke et al. 2012	Family physicians' awareness and knowledge of the Genetic Information Non-Discrimination Act (GINA).	Quantitative (Survey) 383 FPs 130 F, 266 M Response rate: 26.9% US	Genetic Information Nondiscrimination Act of 2008 (GINA)	- 54.5% indicated they had no prior awareness of GINA, 35.2% were aware of GINA but had limited knowledge, 10.3% were aware of GINA and claimed a basic understanding - Most common concern for discrimination was on life insurance (49.6%)		

31	Haga et al. 2012	Primary care physicians' knowledge of and experience with pharmacogenetic testing.	Quantitative (Survey) 40.58% Fam medicine, 58.21% internal med, 1.21% other 34.04% F, 65.96% M Response rate: 15% (597) US	Pharmacogenetic (PGx) testing	- 51.4% strongly or somewhat disagreed that they felt well-informed about genetic testing - 73.0% did not feel that their genetics training adequately prepared them to appropriately order or use genetic tests. - 43.7% strongly or somewhat disagreed that they felt comfortable ordering a test to predict disease susceptibility - Only 13% felt well-informed about the role of PGx testing in therapeutic decision-making	- 64.5% agreed that PGx testing is or will soon be a valuable tool to predict risk of adverse events or likelihood of effectiveness - Most (62.9%) believed that they should have primary responsibility for making patients aware of a PGx test - 57.5% believed it was their responsibility as a primary care provider to discuss PGx test results with the patient	- Preferred methods to educate PCPs were CME (in-person courses) 36.5%, training in residency 15.5. - Most PCPs learned about PGx through journals (46.9%) or professional meetings, CME, or grand rounds (46.61%).

32	Powell et al. 2012	Primary care physicians' awareness, experience and opinions of direct-to-consumer genetic testing.	Quantitative (Survey) 382 PCPs 115 F, 263 M Response rate: 16.2% US	DTC testing	- 61.3% had never heard or read about DTC genetic testing - Among those that had read, common sources of information were medical or scientific journals (35.1%), television (33.1%), a newspaper article (28.4%) and the Internet (27.0%) - Older PCPs (41 and above) were almost twice as likely to be aware of DTC genetic testing than younger PCPs. - 81.1% had never discussed DTC tests with a patient or had a patient bring in results of DTC genetic tests - 33.8% felt DTC genetic test results were likely to influence the care of patients in their practice - 85% did not feel prepared to answer their patient's questions regarding DTC genetic testing	- Among the 63 respondents (42.6%) who thought that testing was clinically useful when formulating medical management plans, most frequently endorsed benefits were the ability to: 1) offer screening tests at an earlier age to individuals at an increased risk (82.5%, n = 52), and 2) offer screening tests more frequently to individuals who are found to be at an increased risk (81.0%, n = 51). - Among the 85 respondents who thought that it is not clinically, reasons endorsed were 1) no guidelines exist to reduce or alleviate the risk for many diseases (80.0%, n = 68), 2), it is too difficult to interpret what the results mean regarding patient care (58.8%, n = 50), 3), it will cause more patient anxiety (51.8%, n = 44), 4), they would not change a patient's management based on DTC testing (35.3%, n = 30)	

33	Ram et al. 2012	General practitioner attitudes to direct-to-consumer genetic testing in New Zealand.	Quantitative (Survey) 113 GPs 49 F, 64 M Response rate: 38% New Zealand	DTC testing	- Only half of respondents had heard about DTC genetic testing. - GPs who had received training disagree that DTC is a useful service of healthcare - Lack of knowledge, experience and time were all considered barriers to GPs providing genetic counselling	- Respondents were ambivalent on benefits of DTC but agreed with risks and barriers presented; those without training emphasised on proposed benefits while those with training emphasised on proposed risks. - Genetic specialist was highlighted as the most appropriate to provide counselling.	
34	Kadaoui et al. 2012	Breast cancer screening practices for women aged 35 to 49 and 70 and older.	Quantitative (Survey) 460 GPs 247 F, 206 M Response rate: 36% Canada	Breast cancer	- For women aged 35 to 49 years, more than 80% of physicians reported using practices deemed adequate, except for instruction in BSE and referral for genetic counseling (60% and 54%). - For women 70 years of age and older with GLE, only 50% of general practitioners prescribed screening mammography. - For the 70 years and older age group without GLE, for whom screening is not indicated, nearly half of physicians continued to perform CBE and more than one-third continued to review family history		

35	Haga et al. 2011	Genomic risk profiling: attitudes and use in personal and clinical care of primary care physicians who offer risk profiling.	Quantitative (Survey) 79% Internal med, 19.1% family medicine, 1.9 other 14.6% F, 85.4% M Response rate: 44% (167) US	Genetic testing	- 45% strongly or somewhat strongly agreed that they felt well-informed about genetic testing - 52% strongly or somewhat strongly agreed that they would feel comfortable ordering genetic testing for disease susceptibility - Significant association between feeling well-informed and feeling "comfortable" ordering a genetic test - those who felt well-informed were more likely to feel comfortable (78.6%) than those who did not feel well-informed (29.8%). - 49% did not believe that their genetics training was adequate.	- 53% expressed concerns about life and long-term/disability insurance discrimination, 50% about health insurance discrimination, 43% about confidentiality, 41% about inadequate knowledge of testing, and 36% indicated they did not believe testing would provide useful information	- Preferred educational resources to learn about genomic risk profiling: CME courses (69%), medical journals (57%), professional medical meetings (53%), and educational programs offered by testing companies (47%)

36	Carroll et al. 2011	GenetiKit: a randomized controlled trial to enhance delivery of genetics services by family physicians.	RCT (survey) 80 PCPs - 47 intervention; 33 control 49 F, 31 M Response rate: 64% Canada	Hereditary breast and ovarian cancer (HBOC)			- Multifacted educational intervention could significantly improve referral decisions, to be more consistent with guidelines and, instil greater confidence in core genetics competencies - Intervention-physicians showed lower decisional difficulty and higher appropriate referral decisions score; higher confidence across all competencies - Among intervention-physicians: materials (Gene messenger) were generally useful; 93% would like to continue receiving information, 93% would recommend to their colleagues; 76% said that practice changed 'a little' with 9% stating changing 'a lot'
37	Vansenne et al. 2011	Providing genetic risk information to parents of newborns with sickle cell trait: role of the general practitioner in neonatal screening.	Quantitative (Survey) 131 GPs 59 F, 72 M Response rate: 49% unadjusted The Netherlands	Neonatal screening (Sickle Cell)	- Few GPs were aware of primary goals of reporting carriers was identify and guide reproductive decisions of parents. - Barriers includes intrinsic (lack of clinical experience) and extrinsic (rarity of sickle cell) - Majority reported the lack of specific clinical experience		

					and knowledge on disease and inheritance		
38	Nippert et al. 2011	Confidence of primary care physicians in their ability to carry out basic medical genetic tasks-A European survey in five countries-Part 1.	Quantitative (Survey) 1168 GPs 1454 F, 2226 M Response rate: not reported UK (France, Germany, the Netherlands, and the UK)	Genetic tasks	- 64.4% were not confident to perform basic genetic tasks (take and identify FH, identify and explain autosomal family patterns, estimate risk, recognise malformations, provide psychosocial counselling, identify patient support groups, identify relevant information, identify specialist genetic services) - 19.3% did not receive any genetic training and 61.1% had only undergraduate training. - 34.2% have at least one patient per month with a genetic condition and 17.9% report more than one patient contact due to a genetic condition per week		- 12.8% attended CME/CPD courses in genetics

39	Bonham et al. 2010	Patient physical characteristics and primary care physician decision making in preconception genetic screening.	Quantitative (Survey) 968 PCPs - 495 saw black patient, 473 saw white patient 324 F, 668 M Response rate: 10%; unadjusted US	Genetic screening	- Majority of physicians reported that they would not offer genetic screening but race was a significant factor in their decision - Physicians were 1.5 times more likely to offer genetic screening to black patient in clinical vignette compared to white patient. - 88% reported age as a factor that influenced their decision to offer screening		
40	Tsianakas et al. 2010	Offering antenatal sickle cell and thalassaemia screening to pregnant women in primary care: a qualitative study of GPs' experiences.	Qualitative (in-depth interview) 25 PCPs - 17 intervention; 8 control Response rate: not reported UK	Antenatal sickle cell and thalassaemia (SC&T) screening	- Organisational barriers: lack of time, best left to midwives to inform patients, inability to understand English	- GPs saw the benefits of offering antenatal screening in primary care, as early screening will provide additional options for pregnant women therefore improving healthcare.	- Materials and trainings were found to be helpful for future screenings

41	Sebastian et al. 2022	Widening the lens of actionability: A qualitative study of primary care providers' views and experiences of managing secondary genomic findings.	Qualitative (Semi-structured interview) 15 FPs (3 patient, 12 hypothetical patient) 10 F, 5 M Canada	Secondary genomic findings (SFs)		- PCPs in both groups approached SFs through the lens of actionability: by looking for clinical actions that could be taken based on this information - Did not consider all SFs to be beneficial because they did not perceive all SFs to be actionable. - All PCPs saw the benefit of medically actionable and pharmacogenomic SFs such as referrals, alternative medications or dosages, and entering this information prominently into the EMR for future clinical decision making	- Without actionability, PCPs described that patients were only left with the potential harms of learning SFs (anxiety from not knowing what to do, potential for unnecessary follow-up investigations with physical and psychological patient harm, escalating cost)
42	Sebastian et al. 2022	Challenges and practical solutions for managing secondary genomic findings in primary care.	Qualitative (Semi-structured interview) 15 FPs (3 patient, 12 hypothetical patient) 10 F, 5 M Canada	Secondary genomic findings (SFs)	Challenges related to clinical practice: - Lack of time to manage SFs in a busy practice (time required to discuss results) - Lack of familarity/knowledge with genomics terminology and genomic tests (knowledge challenges) - Technology (EMR) - inability to appropriately store genomic information	- Most providers described feeling responsible for incorporating secondary findings into their practice, but a limited capacity to manage these finding	- Innovative practice solutions - clinical decision support tools, web-based patient portals, chatbots - Comprehensive letter and report - make results easier to understand and navigate - New EMR feature to store genomic information

43	Skinner et al. 2021	Interpretation and management of genetic test results by Canadian family physicians: a multiple choice survey of performance.	Quantitative (Survey) 67 FPs Response rate: not available Canada	Genetic testing	- FPs are more likely to misinterpret or mismanage basic genetic information - 49% of FPs were unable to correctly estimate carrier status for an autosomal recessive condition, although they tended to err on the side of overestimating risk in this scenario - 69% of the responses to the scenario were inappropriate with microarray testing replacing karyotype		
44	Hussein et al. 2020	Is family history still underutilised? Exploring the views and experiences of primary care doctors in Malaysia.	RCT (focus group, in-depth interview) 25 PCPs 18 F, 7 M Response rate: not reported Malaysia	Family history	- FH not collected consistently and systematically but only if GPs felt it was necessary or relevant to patients either: - Proactively for health screening; prevalent multifactorial conditions (diabetes, cardiovascular disease); newly registered patients - Reactively when specific genetic symptoms appears - GPs seldom draw pedigree as EMR is not user-friendly; difficult and time consuming; patients having difficulty recalling their FH	- Mismatched in attitudes and practice where taking FH is an important part of clinical assessment to identify hereditary conditons; GPs have a role to play but approaches varies	

45	Lemke et al. 2020	Primary care physician experiences with integrated population-scale genetic testing: A mixed-methods assessment.	Mixed methods (Survey & interviews) 17 PCPs (interview) 70 PCPs (survey, 67.3%) 35 F, 34 M US	Genetic testing	- Most PCPs (74.3%) reported feeling concerned about the privacy of their patients' genetic test results and the potential for health (60.3%) and life (91.5%) insurance discrimination - 52.8% feel confident explaining the risks and benefits of genetic testing to their patients - cancer risk (42.9%), cardiac risk (27.2%) and PGx (32.8%) - Confidence to explain results was slightly higher than their reported ability to articulate clear next steps - 86.8% reported that the genetic testing program has increased their workload - Only 28.9% agreed that they have received adequate training to offer genetic testing in their practice - 40.0% reported being confident in their knowledge of genetics, their ability to explain genetic concepts (47.1%) and results to patients (34.8%) and their ability to respond to patient questions about genetic technologies (27.9%)	- PCPs highlighted the value of genetic testing in identifying risk to detect and prevent disease in patients and their families - 77% somewhat or strongly agreed that the genetic testing program is useful to change their current management of patients' care - 81.4% agreed that the genetic testing program has value in identifying the need for increased disease screening and supporting patient care management (69.6%)	- Suggested the need for both patient and provider educational resources such as patient education handouts (78.6%) and physician reference sheets (78.5%) - 56.5% were satisfied overall with the DNA-10K program - Additional education on medical management options for patients with a positive result (88.4%) and clinical testing guidelines (86.6%).

46	Carroll et al. 2019	Informing Integration of Genomic Medicine Into Primary Care: An Assessment of Current Practice, Attitudes, and Desired Resources.	Quantitative (Survey) 361 FPs Response rate: 26.4% adjusted Canada	Genetic testing	- Lack knowledge and confidence in GM skills needed - Involvement in key tasks to deliver traditional GM: Majority were highly involved in some aspects of traditional GM tasks (identifying; referrals; providing support) but less so in others (evaluating results; discussion on benefits, risk and limiations) - Low confidence: Self-reported confidence on GM skills were moderate to low - participants who indicated interests were more likely to have a higher confidence score; agree in advances of GM; seeing it as their responsibility	- FPs see a role for themselves in taking FH, identifying genetic condition, making appropriate referrals, supporting patients - Mixed attitudes (somewhat optimistic and cautious about current clinical benefits). - Mixed attitudes: Majority expect advances in GM to improve patient's health outcomes but fewer than half agreed it was important to learn about personalised patient care based on genomics; it was their responsibility; genomics as an exciting part of practice	- Resources: Very few could find useful information regarding genetic services with regards to their own practice. - Useful resources includes local genetic clinic contact information, genetic referral, testing and guidelines; most popular suggestion for integration was contact (telephone/fax/email) or buddy system with geneticists

47	Haga et al. 2019	Primary care physicians' knowledge, attitudes, and experience with personal genetic testing.	Quantitative (Survey) 82 FPs, 48 Internal Medicine 64 F, 66 M Response rate: not reported US	DTC testing	- 62% did not receive any type of formal education in genomic medicine - 42% had referred 1-3 patients for a genetic consultation in the past year - 44% have never ordered a genetic test - Top 3 concerns were the lack of established clinical practice guidelines (72%), uncertain clinical utility (65%), and personal lack of knowledge to interpret the information (56%) - 92% had none or minimal knowledge of GWAS - 61% had minimal knowledge about when and how to integrate genomic medicine into practice - 59% reported that testing experience improved their knowledge of genomic medicine a little	- Positive experience with a novel application or service may improve future knowledge acquisition regarding this specific test and related applications, as well as potentially alter practice behaviors - Attitudes improved significantly following testing regarding confidence in discussing results of DTC genetic testing, knowledge about discussing risks, benefits and results of DTC genetic testing as well as patients' ability to understand their results and perceived benefit	- Preferred mode of education for genomic medicine is online CME programs (42%), followed by professional meetings (21%), and in-person CME such as grand rounds (18%).

48	Puzhko et al. 2019	Health professionals' perspectives on breast cancer risk stratification: Understanding evaluation of risk versus screening for disease.	Qualitative (Interview) ~11 PCPs Canada	Breast cancer	- Time restriction due to the lack of time at a typical appointment was among the most important concerns - Major concern of PCPs was the interpretation of the meaning of the new breast cancer risk stratification approach and its advantages	- PCPs agreed that implementation of this new program could be beneficial for women.	- Use public campaigns, invitation perceived as being issued by the government would add to the chances of being accepted - More evidence that the risk stratification model is beneficial and provide justification of the value - Suggested engaging a nurse other trained personnel, or the creation of a helpful online tool - Being able to use a validated tool for guiding screening practices, rather than being influenced by women's anxiety, would be beneficial

49	Saul et al. 2017	Survey of family history taking and genetic testing in pediatric practice.	Quantitative (Survey) 349 PCPs 224 F, 124 M US	Genetic testing	- 99% collected information about the family health history - 88.3% felt confident in their ability to determine the need for further evaluation based on the results of the FH - 50.6% refer many or most of their patients identified as at-risk for a genetic related disorder to geneticists or other specialist - 95% had referred patients for genetic consultation - Lack of training on genetic risks and choosing appropriate tests (53.1%), inadequate time during typical office visit to interpret tests (48.9%), lack of training in genetic interpretation (60.2%), and lack of guidelines for care management (57.4%).	- 84.8% agreed that PCPs have a duty to warn families about risks in the family. - 71.8% felt there are situations in which it is the role of the PCP to provide genetic testing and evaluation	- 3/4 were interested in CME programs having to do with genetics in primary care

50	Rangarajan et al. 2016	Knowledge and awareness of familial hypercholesterolaemia among registered medical practitioners in tamil nadu: Are they suboptimal?.	Quantitative (Survey) 133 PCPs Response rate: 77.37% India	Familial hypercholesterolaemia (FH)	- Significant shortfall in awareness, knowledge and practices on FH among GPs; role of primary care in FH has not been adequately defined - Overall knowledge on FH among GPs was low (40.6% aware of international guidelines; 12.8% aware of preventive, management and referral services of FH) - 41.4% were unsure if they had FH patients; FH is undiagnosed in the community	- 82% saw GPs as most effective in the early detection of FH	- 69.2% prefer interpretative comments and alerts from labs to highlight at-risk patients
51	Carroll et al. 2016	The Gene Messenger Impact Project: An Innovative Genetics Continuing Education Strategy for Primary Care Providers.	Quantitative (Survey) 1402 FPs 842 F, 560 M Response rate: 7.4% Canada	Genetic testing			- 92% indicated that their practice would be changed or improved by at least one of the rated Gene Messengers - 79% of the Gene Messenger ratings indicated FPs had learned something new - 88% were satisfied with Gene Messengers, 76% found this method of pushed emails useful for learning about genetics and found Gene Messengers useful for clinical practice - 94% wanted to continue to receive them - FPs commented that this method was an ideal way to stay up to date in an evolving field such as genomics, and that the email push “forced”

							them to learn about genomics topics that they might not have sought out
52	Klemenc-Ketis et al. 2014	Family physicians' management of genetic aspects of a cardiac disease: A scenario-based study from Slovenia.	Quantitative (Survey) 271 FPs 75.6% F, 24.4% M Response rate: 27.1% UK	Hereditary cardiomyopathy (HCM)	- Only 50% feel competent to interpret genetic risks; 25% will give genetic testing information; 6% will interpret results - Younger FPs more willing to include genetic tasks in everyday practice - FPs with more genetic education more willing to refer patients to genetic/cardiovascular assessment	- More than 70% believe taking FH is part of their job but 70% also believe that ordering and discussing genetic test/implications is not part of their job - FPs believe it is a family responsibility to inform their relatives of risk but almost 70% would choose not to respect patients' wishes and inform relatives themselves	

53	Bell et al. 2014	Detecting familial hypercholesterolaemia in the community: Impact of a telephone call from a chemical pathologist to the requesting general practitioner.	RCT (Case-historical control study) 82 GPs (intervention), 83 GPs (control) Australia	Familial hypercholesterolaemia (FH)			- A telephone call from a chemical pathologist to the requesting GP of a patient at high risk of FH significantly improves FH detection and specialist referral rates in addition to interpretative comments
54	Richter et al. 2013	Variants of unknown significance in BRCA testing impact on risk perception, worry, prevention and counseling.	Quantitative (Survey) 21 FPs Response rate: 44% US	BRCA 1/2	- 24% 'always/consistently' mention VUS as a possible test result upon referral		

55	Bernhardt et al. 2012	Incorporating direct-to-consumer genomic information into patient care: Attitudes and experiences of primary care physicians.	Quantitative (Survey) 315 Internal medicine, 187 Fam Med. 98 F, 401 M Response rate: 23.3% US	DTC testing	- Only 50% of respondents ordered a genetic test more than once a year, and only 16% ordered tests once a week or more. - 58% of respondents reported feeling confident in interpreting genetic test results - 20% had no genetics education, while 56% had a genetics course in medical school - 22% felt their training in genetics was sufficient to work with their patients who have had genetic testing	- 40% agreed that such results would be helpful in patient management - 49% of respondents agreed that this kind of testing will be commonplace in the next 5 years (respondents who ordered genetic tests at least once a month were significantly more likely to agree) - 43% of respondents indicated they would be likely or very likely to change the management of the hypothetical patient (approximately one-third did not mention the disorders they would address and gave nonspecific response)	
56	Dunlop et al. 2010	'Start the conversation': the New South Wales (Australia) family health history campaign.	Quantitative (Survey) 138 GPs 57 F, 53 M Response rate: 23% Australia	Family history			- 30% reported that they had heard about the campaign through one or more sources: the newsletter of the Division of General Practice (60%), mail or e-mail (48%), an article in 'Australian Dr' (40%), general media including television interview and newspaper articles (40%), and other which included patient, family, or friends (5%). - Only 18% reported that they had seen or currently had one or more of the campaign

							resources: pads of 'Tips on collecting a family health history' (n = 20), the FHH collection tool 'My Family Health Record' (n = 22), and the poster (n = 14)
57	Challen et al. 2010	General practitioner management of genetic aspects of a cardiac disease: a scenario-based study to anticipate providers' practices.	Quantitative (Survey) 1,168 PCPs 404 F, 764 M Response rate: 28.6% France 236, Germany 251, Netherlands 254, Sweden 262, UK 165 UK	Hereditary cardiac disease	- 38% willing to explain inheritance; 28% willing to carry out other tasks - German, Swedish and UK more likely to do initial tasks (taking FH) while French would either carry out most tasks or refer for the entire genetic package	- Although 61% consider it part of their role to take a FH, far fewer (less than 25%) would be willing to discuss specific genetic tests or their implications. This results also vary according to the specific country context.	

58	Houwink et al. 2015	Effect of comprehensive oncogenetics training interventions for general practitioners, evaluated at multiple performance levels.	RCT (survey) 92 GPs - 42 in G-eCPF; 50 in live training program Response rate: 52% (G-eCPD); 57% (live training) The Netherlands	Oncogenetics			- For G-eCPF, self-reported genetic consultation skills and consideration of referral to clinical genetics centres increased after one year but number of regional referrals did not change - 88% of GPs who attended live training session more frequently considered referring patients to genetic centres than those who attended online CPD (64%)
59	Klemenc-Ketis et al. 2014	Family physicians' self-perceived importance of providing genetic test information to patients: a cross-sectional study from Slovenia.	Quantitative (Survey) 271 FPs 205 F, 66 M Response rate: 27.1% UK	Genetic testing	- Majority of FPs received education from undergraduate studies - O6674% reported having contact with patients with genetic disease weekly	- FPs expressed clear role in genetics and perceived genetics to be highly important - More than 90% felt that it was their duty to discuss genetic testing issues with their patients; especially positive and negative test results, and risk of inheritance - FPs expressed lower interests on ethical issues	
60	Leitsalu et al. 2012	Giving and withholding of information following genomic screening: challenges identified in a study of primary care	Quantitative (Survey) 64 PCPs Response rate: 41.54% UK	Genetic screening	- PCPs do not show great confidence in their own ability to discuss genetic test results with patients and families but tend to provide risk information for specifc conditions regardless of circumstances - Majority feel comfortable to talk about basic genetics and	- There was postive attitudes among PCPs regarding the introduction of genetic information into clinical practice and receiving additional training in genomics, but varies based on patient. - Majority believe that	- Majority agree that training program on GT is necessary

		physicians in Estonia.			take FH but most were not comfortable to talk about inheritance patterns - False security, unnecessary anxiety were two common concerns	predictive genetic testing will improve healthcare	
61	Mathers et al. 2010	Family history in primary care: understanding GPs' resistance to clinical genetics--qualitative study.	Qualitative (In-depth Interview) 21 GPs 12 F, 9 M Response rate: not reported UK	Genetic testing	- GPs also admit that they are not confident about their genetic knowledge - Routine use of FH for clinical decision making is distinguised from genetic conceptualisation; FH is an integral part of general practice and not just for diagnosis or risk-assessment but also psychosocial dimensions - GPs expressed concern over being right, being updated with evidence, and making appropriate management decisions	- Although genetic concepts are part of GP practice, they are made distinct from genetics and genetic practice; not identified as core component of their practice. - Genetics/genetic practice not perceived to have significant impact on their practice; which are seen as rare, complex and specialist	- Call for education, training and guidelines; but need is not echoed by all

62	Mazzola et al. 2019	Primary care physicians' understanding and utilization of pediatric exome sequencing results.	Mixed methods (Survey & interviews) 27 PCPs Response rate: 12.6% US	Exome sequencing (ES)	- Knowledge scores were positively associated with comfort score to perform genetics tasks and referrals; more recent genetic training showed higher knowledge and confidence scores	- Even though PCPs may not fully understand ES, majority found ES beneficial for their patient's care and identified and recognise positive clinical utility of ES results - PCPs look to GHPs to communicate results and manage follow up directly with patients; 74% of PCPs agree that its family responsibility to follow up on results	