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ARTICLE

A systematic review of factors that act as barriers to patient referral to genetic services

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Patients who might benefit from genetic services may be denied access through failure to be referred. To investigate the evidence on barriers to referral to genetic services, we conducted a systematic review of empirical evidence on this topic. Nine studies were included in the review. Barriers related to non-genetic healthcare professionals were: lack of awareness of patient risk factors, failure to obtain adequate family history, lack of knowledge of genetics and genetic conditions, lack of awareness of genetic services, inadequate coordination of referral and lack of genetics workforce. Those related to individuals affected by or at risk of a genetic condition were: lack of awareness of personal risk, lack of knowledge and/or awareness of medical history of family members and lack of knowledge of genetic services. Research on access to genetic services is heterogeneous; stronger empirical evidence is needed on factors that are barriers, and further research is needed to develop 'targeted interventions' for equitable access to genetic services in a range of populations.
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INTRODUCTION

Advances in molecular, genetic and related technologies continually increase the scope for genetic screening, diagnosis, counselling and treatment/management of genetic and some complex conditions.[1,2] New advances have been made within healthcare through genetic services by providing specialized care to patients affected by, or at risk of, a genetic condition and to their families.[1,3]

Where genetic services have been incorporated into the healthcare infrastructure, primary care physicians often initiate patient access to genetic services[4] through referrals that enable patients to utilize those services. Current research on accessing genetic services is varied, with a lack of consensus on how access to genetic services should be defined. The concepts of access and utilization are often used interchangeably and although they overlap, they can be clearly distinguished. The authors of this paper carried out a systematic review of barriers that had an impact on patients' access to genetic services under two headings; those that affect patients' referral to genetic services by healthcare professionals (HCPs) and those that affect patients' utilization or uptake of genetic services post referral. This paper reports on the results of the first systematic review, the aim of which was to identify and appraise research reporting direct or indirect factors that had negative impact on patients' referral to genetic services. We excluded papers in which the sample consisted of patients who had already been referred to and received genetic services: those studies primarily reported on barriers associated with the utilization of genetic services and not referral. These will be reviewed in the second systematic review. The question for the primary investigation was: 'What factors act as barriers to accessing genetic services by influencing patient referral?'. The systematic review was carried out with the objectives to: (i) identify factors that have impact on access to genetic services for patients by influencing referral; (ii) enhance understanding of how patient-related access to genetic services has been measured thus far in the current literature; (iii) identify similarities and differences, if any, in the published research reporting evidence on barriers to patients' access

The organization of genetic services and the legal framework in which they operate vary from country to country and neither an internationally accepted definition of their aim nor a description of what constitutes these services is available. However, genetic services can include genetic testing, clinical evaluation and diagnosis of genetic conditions, genetic counselling, and management and follow up of individuals/families with, or at risk of, genetic disorders.[5] For the purpose of this review, only genetic counselling and clinical evaluation aspects of genetic services were explored, whereas patients were defined as individuals and/or families who were affected with or at risk of being affected by a genetic condition. The authors focused on the traditional face-to-face service delivery model of genetic services/counselling and excluded studies focusing on other delivery models such as 'telegenetics'.

MATERIALS AND METHODS

Study design

This systematic review was carried out using the procedure described by the Centre for Reviews and Dissemination.[6] This involved establishment of a review question, well-defined search strategy, appropriate selection criteria and quality appraisal of the included research papers.[6]

Search strategy

Brief details are provided here, the complete systematic review protocol is provided within Supplementary Material 1.

Databases

PubMed (which covers several databases), Oxford Journal, OVID, Science Direct and Google Scholar were searched. *The Journal of Genetic Counseling* and reference lists of included papers were hand searched and articles were retrieved if titles and abstracts appeared related. TD conducted the initial search, which was checked by GRW.

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Keywords
Keyword searches centred on access and referral to genetic services and/or genetic counselling (example strategies are in Supplementary Material 2). The key words 'genetic services' OR 'genetic counseling' OR 'genetic counselling' were used as the primary terms, in combination with the terms 'access', 'barriers', 'patients' and 'referral'.

Inclusion and exclusion criteria
The inclusion and exclusion criteria used to select studies are summarized in Table 1.

Empirical research studies concerning factors preventing patients from being referred to genetic services, investigating the issue from the perspective of patients and/or investigating the role and practice behaviour of HCPs were included. Studies reporting patients referred to genetic services and/or genetic counselling were excluded, as were those focusing on access to 'direct-to-consumer' genetic testing, prenatal and carrier screening programmes. Research where the design included participants responding to hypothetical situation(s) or giving self-reports on their experience or behaviour was also excluded.

Search results
The study selection process is represented in a PRISMA flow chart in Figure 1. The initial search yielded 1184 articles, 91 duplicates were then removed. After further review, full texts of 94 articles were read: an additional 8 articles were identified through ancestral search of the bibliography. In all, 93 articles were excluded, reasons for which are summarized in Supplementary Material 3.

Quality assessment
The remaining nine articles were subjected to quality appraisal using the standard quality assessment tool developed by Kmet *et al*.[7] Two authors (TD and HS) independently assessed the papers and all authors discussed the results until consensus was reached. Studies with a score of $<70\%$ were excluded.

Data extraction and preliminary synthesis
A preliminary synthesis of extracted data from the included research studies was carried out. A review matrix table (Supplementary Table 2) was created to organize the abstracted information, including the study aims, type of genetic service/counselling, country, methodology, population sample, data analysis, results (barriers) and quality score. As thematic analysis is appropriate in the context of systematic reviews of heterogeneous data,[8] the studies' findings were thematically analysed, main themes relating to the research question were identified and the findings were summarized under these thematic headings. Narrative synthesis[9] was undertaken, which is an alternative approach for synthesizing findings from multiple studies and where statistical meta-analysis or formal meta-synthesis is not possible. However, the theory-building element was not implemented as it was not considered appropriate in the design of this systematic review.[9]

RESULTS
Supplementary Table 2 summarizes papers included in the review. Quantitative methodology was used in six studies,[10–15] and qualitative methods in three.[16–18] Four studies were conducted in the United States,[12,13,15,16] two in Australia,[11,17] two in the United Kingdom[14,18] and one in the Netherlands.[10] The majority were related to cancer genetics ($n=5$),[12–15,18] whereas one study was conducted in a general genetics clinic,[16] two in paediatric genetic clinics[11,17] and one in reproductive genetic counselling.[10]

The access-related barriers identified from the included studies[10–18] are reported under two categories:

(i) Barriers related to individuals are factors preventing patients' referral to genetic services owing to their own characteristics or situation, whereas,
(ii) Barriers related to HCPs are factors stemming from the actions and/or characteristics of non-genetic HCPs.

Table 2 provides a summary of barriers identified under these two headings.

Barriers related to individuals
Barriers related to individuals were identified in three papers.[11,16,17]

Lack of awareness of personal risk. Beene-Harris *et al*[16] reported focus group discussions with individuals affected by a genetic condition or who had children with birth defects, and was among a few that directly investigated and reported on access barriers. Reported barriers were later verified by a larger follow-up survey study.[19] Participants commented that they did not initially understand the significance of their family history and were not aware of their own risk until a diagnosis was made. In the study, demographic data were not obtained from all participants, as some feared they could be identified. As a result the range of genetic conditions is unknown, which limits the transferability of this study's findings.

Table 1 Summary of the inclusion and exclusion criteria used to select studies

Inclusion	*Exclusion*
Empirical evidence on factors preventing patient access by impeding referral to genetic services These factors are identified from studies reporting on: Empirically validated experiences of potential patients during or prior to their referral to genetic services Empirical retrospective evidence on referral barriers of patients who had received genetic services The role and practice of healthcare professionals regarding referral.	Studies reporting on the barriers affecting the utilization of genetic services by participants who have been referred to genetic services and/or genetic counselling Participants responding to hypothetical situation(s) or giving self-reports on experience or behaviour unless results verified by further analysis and evidence Quality assessment score $<70\%$
Published: in English, in peer-reviewed journals, between January 2000 and July 2013	Barriers on utilization/uptake of genetic services
Primary research papers (with quantitative/qualitative & mixed methodology) and systematic reviews	Access to 'direct-to-consumer' genetic testing, prenatal and carrier screening programmes

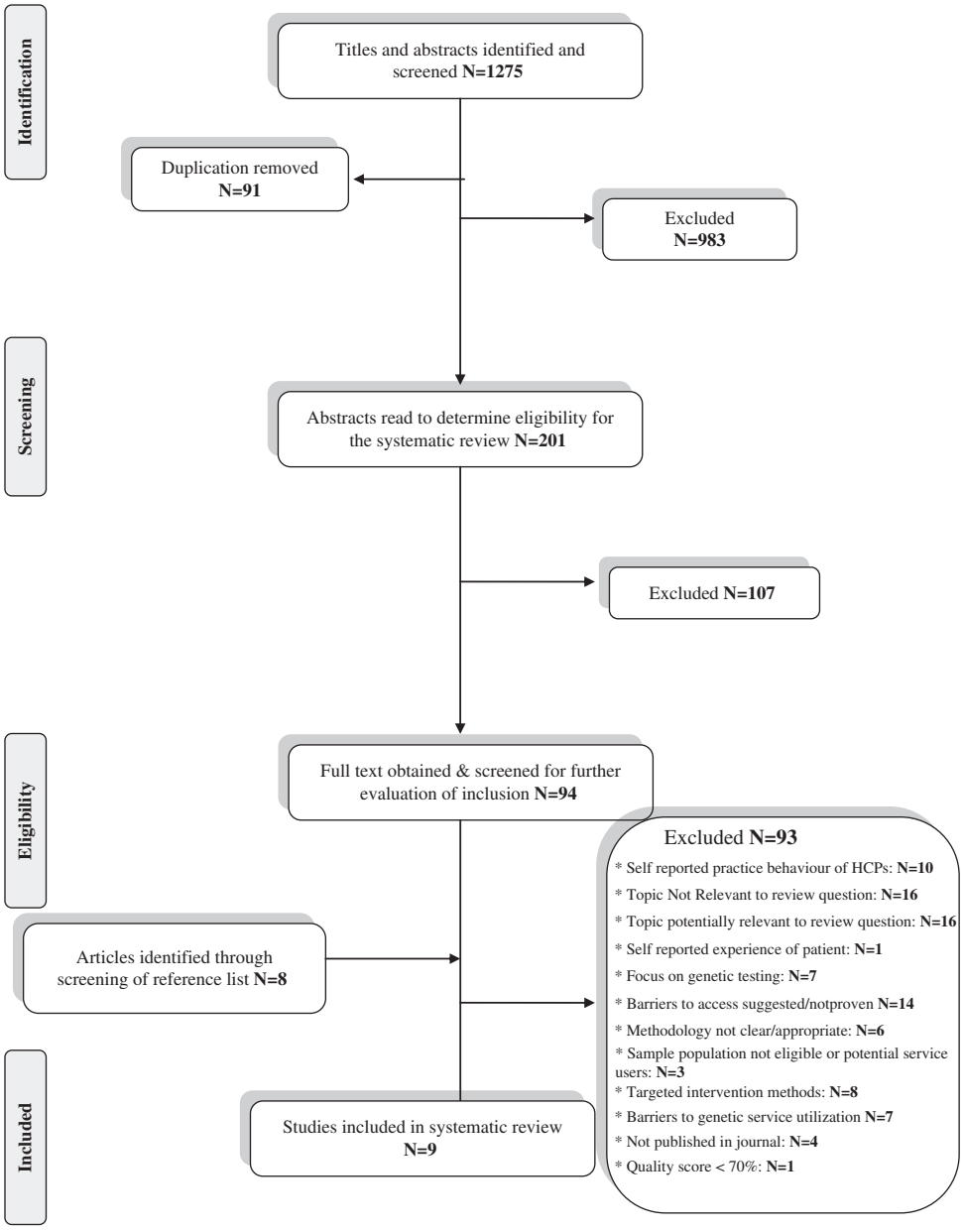

Figure 1 PRISMA flow chart illustrating study selection process.

Table 2 Summary of barriers identified in the systematic review

Barriers related to individuals	*Barriers related to healthcare professionals (HCP)*
1. Lack of awareness of personal risk	1. Non-genetic HCPs' lack of awareness of patient risk factors
2. Lack of knowledge and/or awareness of medical history of family members	2. Lack of obtaining adequate and/or accurate family history
3. Lack of knowledge of genetic services	3. Lack of knowledge on genetics and genetic conditions
	4. Lack of awareness of genetic services
	5. Inadequate coordination of referral
	6. Lack of genetics workforce

Lack of knowledge and/or awareness of medical history. This barrier was identified as significant in Beene-Harris *et al*'s study[16] and it appears that this barrier was mainly reported by participants who were affected by a genetic condition who became aware of the significance of their family medical history following a diagnosis.

Lack of knowledge of genetic services. Lack of knowledge of genetic services was identified as a barrier in three studies.[11,16,17] Beene-Harris *et al*[16] reported that almost all of their focus group participants mentioned lack of knowledge of genetic services and other genetic resources. Participants experienced difficulty in obtaining information,

including knowing where to go to have their condition diagnosed. However, it is not evident whether there was a difference in the range of difficulty experienced based on which genetic condition was involved. Similarly, Collins *et al*[17] found that some parents of children with Down syndrome did not receive genetic counselling as they were not aware of the existing genetic services, and were not referred appropriately by other HCPs. The study showed differential experience in accessing services based on genetic condition: parents of children with cystic fibrosis were referred to a genetic counsellor as part of a newborn screening programme but no such process was available for parents of children with Down syndrome. The data, however, came from a group of only 4 parents (out of 14, where remaining majority did receive genetic counselling). The same researchers carried out a larger study targeting a population-based sample of parents of children with Down syndrome[11] and again found that lack of awareness of genetic counselling was indicated as a barrier to gaining access to genetic services.

Barriers related to HCPs

Eight of the nine included[10,12–18] papers reported empirical evidence on barriers associated with HCPs. The HCPs in these studies were not specialized in genetics but were 'gatekeepers' to specialist genetic services and/or genetic counselling. All of the barriers identified overlap and collectively contribute to patients' not accessing genetic services. Although reported separately below, their direct association with each other is evident.

Non-genetic HCPs' lack of awareness of patient risk factors. Non-genetic HCPs' failure to recognize patients' hereditary risk factors resulted in patients not being referred to appropriate genetic services. Aalfs *et al*[10] focused on why at-risk pregnant women, referred for prenatal genetic counselling, had not been referred before their pregnancy, as reproductive genetic counselling would have provided opportunistic and timely risk assessment. They found that in 71% of the cases, the main reason for patient non-referral during pregnancy resulted from their GPs' lack of awareness of patients' potential genetic risk factors before pregnancy. In some cases referral took place without the knowledge of the GP, but reasons for this could not be explored in the study. Lack of awareness of patients' risk factors was also identified in studies focusing on HCPs' competence in collecting family history, reported below.

Lack of obtaining adequate family history. A complete and accurate family history is an essential tool used in diagnosing hereditary conditions, determining personal risk for genetic conditions or assessing risk to the next generations. Lack of obtaining adequate family history was reported as a barrier in three studies.[12,14,15] In all of these, patients' medical notes were reviewed to assess whether the recorded family history by physicians was complete. Grover *et al*[12] compared concordance between physician notes on family history, obtained during clinical visits in a gastroenterology cancer clinic, with the corresponding patient's self-report of family history in a written questionnaire. They explored whether high-risk patients were appropriately selected through risk assessment and referred onto cancer genetic evaluation. With a 98% response rate from patients, their corresponding medical records were scrutinized and physicians did not document 32% of cancer cases reported by participants in their family history: significantly, nearly one-third of these unreported cancer cases occurred in first-degree relatives. Also only 17% of patients at risk for a hereditary cancer syndrome were documented to have been referred for a genetic evaluation. It is evident that the significance of family history and hereditary cancer risk was not being recognized by the treating physician, even when family history was adequately recorded in the medical notes. In a similar study design, Sweet *et al*[15] compared family history information obtained by physicians with touch-screen data entered directly by patients at a comprehensive cancer clinic. They reported that only 69% of family history information from the patients' medical record was available compared with self-reported computer entry information. They also found that, frequently, family history information was not routinely updated at follow-up clinics with patients and documentation of risk assessment was found in only 14 high-risk patients' charts, of which only 7 had been referred to genetic counselling. Lanceley *et al*[14] also extracted family history and related data from ovarian cancer patients' notes in three UK gynaecological cancer centres and found that family history was not consistently recorded in the 114 case notes analysed, whereas in 15 cases, family history was not adequately obtained and was insufficient to carry out a risk assessment. Twenty-two women were identified as high risk by the research team: the risk was documented in 15 cases but no further action was noted.

Reviewing medical notes as evidence of HCPs' daily practice may be a laborious approach; however, it does provide an insight into the HCPs' actual practice behaviour. None of the studies explored the findings from medical records from HPCs' perspectives concerning why family history was insufficiently collected or why appropriate risk assessment or referral to genetics was lacking.

Lack of knowledge of genetics and genetic conditions. Three studies indicated lack of knowledge on genetics and/or genetic conditions as a barrier.[10,16,18] Aalfs *et al*[10] reported that GPs who participated in the study considered their level of knowledge in genetics to be limited. Iredale *et al*[18] also found that knowledge on cancer genetics was generally low among physicians practising in a rural part of Wales (UK). From the service users' perspective, participants affected by genetic conditions in Beene-Harris *et al*'s study[16] pointed out that the HCPs' lack of knowledge of genetic conditions made it difficult for them to receive the appropriate medical management and treatment. However, this barrier emerged from the experiences of patients affected by genetic conditions and was not obtained directly from research including HCPs.

Lack of awareness of genetic services. Lack of awareness of genetic services by non-genetic HCPs clearly prevents them from carrying out their 'gatekeeper' role. Iredale *et al*[18] investigated the effect of 'rurality' on the referral behaviour of HCPs by stratifying sample groups on the basis of clinical practice location (ie, rural *vs* urban/suburban) for comparison: the level of awareness of the existing cancer genetics service among physicians practising in rural areas was low. Similarly, Koil *et al*[13] investigated why patients referred for hereditary breast cancer indication from rural areas made up $<1\%$ of their patient population. They found that physicians practising in such areas were more likely to refer patients to an oncologist than to a genetic counsellor and that the lack of awareness of services was identified as a more significant barrier among the rural HCPs. Participants affected by genetic conditions in Beene-Harris *et al*'s study[16] complained that their physicians did not have the right knowledge regarding genetic services to refer them appropriately.

Inadequate coordination of referral. As mentioned previously, Aalfs *et al*[10] reported on women at genetic risk, who were referred for genetic counselling during, rather than before pregnancy. The coordination of referral to genetic services in this study seems inadequate, as referral before a pregnancy is advantageous.

Iredale *et al*[18] and Koil *et al*[13] investigated why physicians practising in rural areas referred fewer patients to genetics services. Geographical location of the genetic service, travel costs, time limitation, patient preference, patient disinterest and attitude of HCPs were found to be contributory factors to inadequate coordination of referral in these two studies. Collins *et al*[17] reported that some parents of children with Down syndrome had never received genetic counselling because they were not referred by their physician.

Lack of genetic workforce. There is an increased expectation of genetic service providers owing to advancing genetic technology and literature clearly shows that there is a lack of specialist workforce in genetics to meet such needs. Beene-Harris *et al*[16] reported that patients affected with a genetic condition voiced the need of more genetic specialists as, for some, none existed where they lived. Similarly, Koil *et al*[13] and Iredale *et al*[18] reported that the lack of genetic specialist in rural areas contributed to barriers in accessing those in urban cities.

DISCUSSION

In this systematic review, we only retrieved and included studies that were published in English. The majority of research in genetic services, including genetic counselling, is from countries where English is the main language but we cannot rule out the possibility that this review excluded research published in other languages. Narrative synthesis was used to analyse and report the emerging themes as research on access to genetic services was found to be heterogeneous in terms of methodology and data and analysis via other methods was not possible.

In all of the countries represented by the papers included in the review, genetic services have emerged as a branch of specialized healthcare that has been well integrated into the healthcare infrastructure for many years.[20] In recent years, publications from different countries such as Brazil and Philippines reporting on the need, establishment, practice and cultural characteristics of genetic services have appeared[21–24] but do not address access to services.

Half of the studies[12–15,18] included in this review focused specifically on cancer genetic services, a subspecialty of genetic services. There is clear evidence that if individuals at risk of hereditary cancer syndromes are identified early enough, they would greatly benefit from early cancer detection screening programmes. This may explain why there is significant research investigating appropriate access to cancer genetic services by all those eligible and whether the outcomes of these services are producing favourable results such as prevention of cancer.[13,15,25–36]

It was not surprising that the majority of studies focused on the role or practice of HCPs in patients' access to genetic services. The translation of new genetic information and technology into medical care is challenged by several factors, including lack of adequate genetic workforce and increased demand of specialist care in genetics, which was also reported as a barrier in this review.[16] Primary HCPs are now 'gatekeepers' to specialist genetic services and need to be able to identify, assess and refer patients, at hereditary risk, to relevant genetic services.[13,31,37,38] However, research has shown that many HCPs practising in primary care do not have genetic competence, that is, they do not possess the necessary genetic knowledge or skills to fulfil such roles.[37–46]

Researchers investigating HCPs' genetic competence, practice and/or behaviour, have mainly collected data via surveys and questionnaires where HCPs self-report information.[27,37,43–51] There are limitations in these types of studies regarding the validity of the data obtained in terms of how much they reflect the actual situation. Van Riel *et al*[27] explored the knowledge and attitudes of medical specialists on BRCA testing and their referral pattern to genetic services through questionnaires, suggesting a substantial proportion of patients at risk of hereditary breast cancer were not being referred. However, they acknowledge that their findings may not be a true representation of daily practice and they could not identify the reasons for the low referral rate. There appears to be a gap in research that further examines such initial findings in order to determine how much of what physicians say or do matches their real actual behaviour. Lanceley *et al*,[14] Grover *et al*[12] and Sweet *et al*[15] tried to explore the genetic competence of HCPs by analysing the patient medical records, and by comparing them with the information given by patients. They found that often the data did not match and were inaccurate. Grover *et al*[12] found that even when the adequate information on family history was obtained, only very few of the eligible patients were in fact referred, because of the HCPs' lack of knowledge of cancer genetics. These studies were more evidence-based compared with other studies that relied solely on HCPs' self-reports. They are also among the few that assess the quality of family history, recorded by HCPs, with respect to access to genetic services. However, none of the studies included interviews or discussions with HCPs concerning inadequate family recording. This would have been an added information from the perspective of the HCPs.

Genetic knowledge is an essential component of competence of HCPs and has received significant attention within research, where physicians' level of knowledge has been reported to vary from minimal to moderate and to vary with their subspecialty.[27,45,47,52] Nippert *et al*[1] reported on the results of a survey administered in 2005 in five European countries as part of a larger study where researchers explored genetic knowledge and skills of primary HCPs through their self-reported confidence in taking on genetic tasks. They found that primary care physicians who reported the lowest levels of confidence in carrying out genetic tasks were those who had been least exposed to medical genetics during their specialist training; hence their knowledge of genetics was minimal. Obstetricians/gynaecologists and paediatricians had received more education in medical genetics in their education compared with GPs. Similarly, GPs were the specialists who engaged least in continued education programmes or seminars in genetics. Acton *et al*[45] also reported higher levels of knowledge among obstetrician/gynaecologists, who were also referring more patients to genetic testing for cancer compared with other physicians in primary care in Alabama (USA). However, such conclusions drawn from self-reported data need further validation.

Lack of awareness of genetic services by the non-genetic HCPs was reported both in the extensive literature and in the findings of this review. Awareness to existing services appeared to be associated with geographical barriers as it was reported among physicians practising in rural areas in two studies.[13,18] Geographical location of the genetic services and the practices of the HCPs (urban *vs* rural) clearly affect awareness as well as access to genetic services and resources by HCPs in the literature.[23,53]

The attitude of primary care physicians has also been found to have an impact on patients' access to genetic services. Research has shown that physicians who have negative attitudes or misconceptions about the clinical utility and benefits of a genetic service are less likely to suggest genetic counselling to their patients or refer them to the genetic services.[13,16,40,54] Several studies have shown that HCPs can have misconceptions or assumptions regarding their patients' acceptance and/or utilization of genetic services and/or counselling if referred and reported on HCPs' opinions as to why their patients would not uptake genetic counselling (eg, fear of insurance discrimination, cultural and religious differences between service providers and

themselves, etc).[54,55] Such perceptions clearly influence HCPs' practice behaviour in terms of referring for genetic services and/or genetic counselling.

Research seems to be more focused on the access or uptake of genetic testing than on genetic services and/or counselling. In majority of these studies, patients have already gained access to genetic counselling as it is prerequisite to genetic testing.[25,27,56–58] As a result there appears to be a gap in research directly looking at factors influencing patients' gain of access to genetic services.

CONCLUSION

The findings of this review indicate that very few studies focused directly on access to genetic services. Barriers often emerge as coincidental findings, or are not proven by empirical evidence. Authors who do explore issues of access do not always differentiate between being referred to and accessing genetic services in terms of utilization (eg, attending the appointment). In particular, there is a lack of research in subspecialties other than cancer genetics and therefore more research in various types of genetic services is needed. Authors of future studies need to integrate methodologies that further evaluate the validity of self-reported behaviour of HCPs in their research (eg, comparing survey results with medical notes). In conclusion, there is a need for research that is designed to directly explore barriers to access in various subspecialties of genetic counselling and services with clearer methodology, in order to develop and implement intervention methods to overcome them.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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