



Healthcare professionals' perceptions of risk in the context of genetic testing for the prediction of chronic disease: a qualitative metasynthesis

Marie Falahee, Gwenda Simons, Karim Raza & Rebecca J. Stack

To cite this article: Marie Falahee, Gwenda Simons, Karim Raza & Rebecca J. Stack (2016): Healthcare professionals' perceptions of risk in the context of genetic testing for the prediction of chronic disease: a qualitative metasynthesis, Journal of Risk Research, DOI: [10.1080/13669877.2016.1153503](https://doi.org/10.1080/13669877.2016.1153503)

To link to this article: <http://dx.doi.org/10.1080/13669877.2016.1153503>



© 2016 The Author(s). Published by Taylor & Francis



Published online: 11 Mar 2016.



Submit your article to this journal [↗](#)



Article views: 98



View related articles [↗](#)



View Crossmark data [↗](#)

Healthcare professionals' perceptions of risk in the context of genetic testing for the prediction of chronic disease: a qualitative metasynthesis

Marie Falahee^{a,b*}, Gwenda Simons^{a,b}, Karim Raza^{a,b} and Rebecca J. Stack^{a,b,c}

^aCentre for Translational Inflammation Research, Institute of Inflammation and Ageing, College of Medical and Dental Sciences, University of Birmingham, Birmingham, UK;

^bDepartment of Rheumatology, Sandwell and West Birmingham Hospitals NHS Trust, Birmingham, UK; ^cDivision of Psychology, School of Social Sciences, Nottingham Trent University, Nottingham, UK

(Received 5 June 2015; final version received 1 February 2016)

Advances in genomic technologies and a growing trend towards stratified and preventive approaches to medicine mean that increasing numbers of individuals may have access to information about their genetic makeup, and their risk of developing diseases. This is likely to impact on healthcare professionals involved in the delivery of genetic tests, or in supporting patients who are affected by a disease with a genetic risk factor. It is therefore important to understand healthcare professionals' perceptions about providing these services, and how they feel about communicating information about genetic risk to patients. This paper provides a systematic review and metasynthesis of qualitative research exploring healthcare professionals' perceptions of genetic risk in the context of predictive genetic testing for chronic disease. Healthcare professionals expressed a range of reservations about the utility of predictive testing in this context. Professionals judged patients' understanding of risk information to be limited and subject to bias and a range of sociocultural influences. Concerns about the psychosocial impact of genetic risk information were frequently cited, both in relation to individual patients and the wider impact on their families and communities. The need for provision of multidisciplinary support was described. The concept of responsibility was also an important theme. Healthcare professionals recognized the responsibility that accompanies risk knowledge, and that ultimately this responsibility lies with the patient, not the provider. Our analysis suggests that professionals' evaluation of the utility of predictive genetic testing is influenced not only by resource deficits, but may also be interpreted as a response to challenging ethical and social issues associated with genetic risk, that are not well aligned with current medical practice.

Keywords: healthcare professionals; perceptions; genetic risk; qualitative; review

Introduction

The rapid expansion of genomic technologies has created promising opportunities for personalized approaches to health care. The identification of genetic variants that influence susceptibility to and outcome of disease facilitates preventive and stratified

*Corresponding author. Email: m.falahee@bham.ac.uk

therapeutic interventions. Consequently, the demand for and availability of genetic tests to predict the development of diseases with a heritable component are increasing (Guttmacher et al. 2010).

Genetic testing has traditionally been available in medical settings, and delivered by specialist professionals in the context of monogenic diseases. As genomic medicine advances and is increasingly applied to the prevention and treatment of common disorders with complex genetic and environmental aetiologies (Sparks et al. 2014; Wilson and Nicholls 2015), it is likely that a wider range of non-specialist health care professionals (HCPs) will become more involved in the delivery of genetic services. Therefore, it is increasingly important that HCPs develop a thorough understanding of medical genetics (Feero and Green 2011), and are equipped to communicate complex risk information in a way that is disease appropriate, and that takes into consideration the values and needs of patients (Lautenbach et al. 2013).

The availability of genetic testing is no longer restricted to health care settings. Genetic information is available to individuals via a wide range of home testing kits available directly to consumers (DTC) from private companies (Frueh et al. 2011). Public awareness and demand for such testing have grown (Su 2013; Agurs-Collins et al. 2015), though this market is facing increased regulation (Kalokairinou, Howard, and Borry 2014). As consumers become more aware of genetics, and have increased access to genetic data, it is likely that they will want to discuss issues surrounding genetic testing with HCPs. An internet-based survey by McGuire et al. (2009) found that 78% of those respondents who would consider using DTC testing would ask their physician to help them interpret the results.

The clinical translation of the expansion in genomic medicine is still at an early stage (Vassy, Green, and Lehmann 2013), and there are considerable gaps in our knowledge about the organizational requirements and clinician and patient needs (Scheuner, Sieverding, and Shekelle 2008). In order to support the integration of this knowledge into clinical practice, it is important to understand HCPs' willingness to engage with the delivery of genetic services. A systematic review by Emery et al. (1999) specifically explored the views of General Practitioners (GPs) about clinical genetics and the role of primary care in the delivery of genetic services. The review found that whilst GPs accepted their increasing role, they lacked confidence in their ability to deliver genetic services because of lack of knowledge of clinical genetics. These findings were echoed in a review by Suther and Goodson in 2003 of perceived barriers to the integration of genetics into primary care practice. Barriers included inadequate knowledge, inadequate family history gathering, lack of confidence and a lack of referral guidelines.

More than a decade later, a recent review by Mikat-Stevens, Larson, and Tarini (2015) produced similar findings. Primary care providers frequently mentioned lack of knowledge about genetics and genetic risk assessment, concern for patient anxiety, lack of access to genetics and a lack of time as important barriers to the integration of genetic medicine into routine patient care.

The existing reviews described above (Emery et al. 1999; Suther and Goodson 2003; Mikat-Stevens, Larson, and Tarini 2015) have focused on the views of primary care providers, though a wider range of professionals, including non-genetic specialists from medical and other HCP backgrounds, are likely to be increasingly involved in clinical genetics in many areas of medicine. The reviews included studies of a wide range of very different (and sometimes unspecified) kinds of genetic

testing. The context in which genetic testing occurs has an important bearing on the full meaning and impact of the test results, both for providers and patients. For example, the issues raised by a prenatal genetic test with consequences for reproductive decision-making are different from those associated with testing in a pharmacogenetic context in order to identify optimum treatment strategies. Similarly, genetic testing in paediatric settings raises different issues compared with testing adult patients.

The previous reviews described above included both quantitative and qualitative research, and employed evaluation criteria and review methods that favour quantitative methods. However, as noted by Suther and Goodson (2003), information from a relatively small number of qualitative studies provided richer descriptions of respondents' views. The aim of this paper is to provide a systematic review and synthesis of the qualitative research literature focusing on perceptions of HCPs about genetic risk pertaining to a specific context; genetic testing for the prediction of the development of chronic somatic disease in adults. We have used metasynthesis techniques (Downe 2008) to organize qualitative data from multiple papers into themes, and to provide an interpretation of recurring and inter-relating themes relating to genetic risk across studies.

Methods

Study selection criteria

Research publications meeting the following criteria were included in this review:

- (1) Primary research using qualitative research methods to explore the views of participants, with qualitative findings (quotations) reported (allowing for first order analysis to be undertaken).
- (2) Research participants were non-student HCPs. For the purpose of this review, a HCP is defined as a person working professionally in a healthcare setting offering informed health advice and health services to members of the public.
- (3) Research findings involve discussion of predictive genetic testing, and/or the communication of genetic risk information for chronic, somatic diseases in adults.
- (4) Papers written in the English language.
- (5) Peer-reviewed publications (excluding abstracts and theses).

Research publications meeting the following criteria were excluded from this review:

- (1) Research articles reporting qualitative methods that focus on the analysis of interactions between HCPs, or between patients and professionals, rather than the direct elicitation of professionals' viewpoints.
- (2) Research articles not providing identifiable findings that were explicitly related to predictive testing for chronic disease in adults. Excluded papers include those relating to genetic testing in paediatric; pharmacogenomics; prenatal; newborn; preconception/carrier; kinship; and nutrigenomic contexts. Articles relating to the prediction of psychiatric disorders and substance use

disorders were also excluded as these conditions raise very specific issues that may be distinct from those raised by other diseases.

- (3) Research articles that focused on genetic testing and/or risk communication in research settings, rather than clinical practice.
- (4) Research articles that focused mainly on the perceptions of members of the public, and little reference was made to relevant findings from HCP participants.
- (5) Research articles that focused on genetic testing after the onset of symptoms to confirm a diagnosis, or post-diagnosis testing to inform treatment.

Search strategy

The following databases were searched to identify relevant articles: Ovid MEDLINE (Pubmed; 1950 – present), CINHALL (1982 – present), PsycINFO (1967 – present), EMBASE (1947 – present), Web of Science (1900 – present) and Health Management Information Consortium (1979 – present). The search terms used are displayed in Table 1.

In total, 7687 publications were initially identified and 2822 duplicate articles were removed. The authors agreed to limit the search to research that was published on or after the year 2000, as this represents a relevant milestone in the completion of Human Genome Project (Collins and McKusick 2001), after which public awareness of genetics increased (Tambor et al. 2002). The abstracts of all 4132 remaining publications, and 169 full text publications, were read (by MF) in order to identify relevant studies. A sample of 1000 abstracts were independently read and assessed for relevance by RS. The level of agreement between researchers about relevance of studies was very high. There was continuous analysis and refinement of the application of exclusion and inclusion criteria (listed above) amongst researchers (MF, RS, and KR). Twenty-eight studies were selected for inclusion in the metasynthesis. The search process is summarized in Figure 1.

Table 1. Search terms.

professional OR	AND	genetic OR	AND	qualitative OR
provider OR		genom* OR		theme OR
practitioner OR		chromosome* OR		thematic OR
personnel OR		DNA OR		interview OR
physician OR		'deoxyribonucleic acid' OR		'focus group' OR
Doctor OR		'family history' OR		'conversation analysis' OR
Nurse OR		heredit* OR		'discourse analysis' OR
Specialist OR		inherit* OR		IPA OR
counsel* OR		'gen* sequenc*' OR		phenomenological OR
geneticist		'exom* sequenc*' OR		ethnograph* OR
		WGS OR		
		WES OR		
		CMA OR		
		'carrier test' OR		
		'incidental finding' OR		
		'unsolicited finding'		

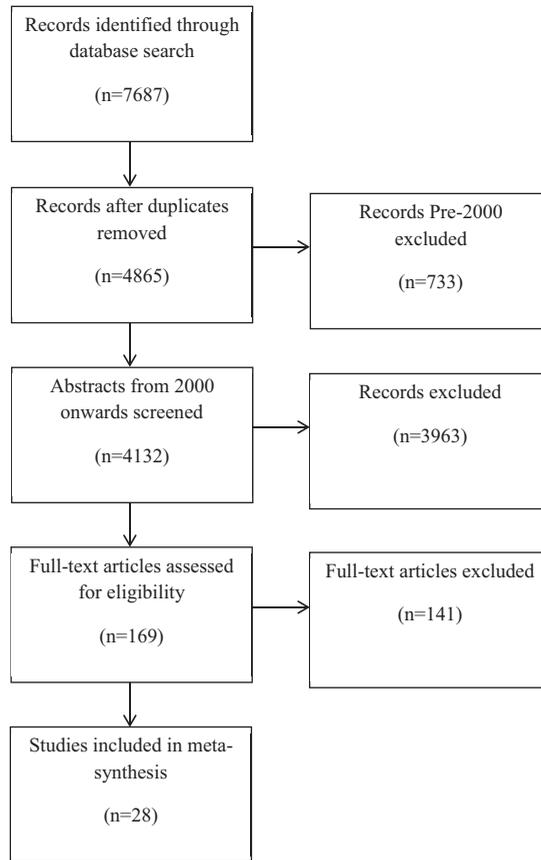


Figure 1. Summary of search process.

Analysis

The application of quality criteria to qualitative research is contentious (Downe 2008; Carroll, Booth, and Lloyd-Jones 2012), and agreement between experienced qualitative researchers using standardized checklists for the appraisal of qualitative research can be low (Dixon-Woods et al. 2007). After careful reading of the 28 studies selected for inclusion in this metasynthesis, and discussion amongst the authors guided by Walsh and Downe's quality assessment tool (Walsh and Downe 2005; Downe, Simpson, and Trafford 2007), it was agreed that all of the 28 studies contained material that could usefully contribute towards the objectives of the present analysis, and that none were so flawed that the integrity and validity of the metasynthesis would be compromised by their inclusion. A summary of the 28 studies, including methodological details and reported quality assurance measures, can be found in Table 2.

Metasynthesis is a technique for the synthesis of qualitative data and the development of theoretical frameworks that is derived from meta-ethnographic methods (Noblit and Hare 1988). This method has previously been used to identify core themes in barriers to seeking help at the onset of rheumatoid arthritis, and to explore patients' experience of symptoms across multiple qualitative studies (Stack et al.

Table 2. Summary of articles included in the meta-synthesis.

Authors	Year	Country	Sample	Qualitative methods	Analysis	Quality checks	Topic of investigation
Aasen and Skolbekken	2014	Norway	Genetic counsellors in a hospital genetics outpatients department ($N = 2$)	Interviews (and transcripts of 6 counselling sessions - not included in meta-synthesis)	Interpretative phenomenological analysis	Checking themes against entire data set. Review and discussion of analysis amongst researchers	Counsellors' understanding, communication and management of uncertainty in counselling about a hereditary disposition to either colon or breast and ovarian cancer
Al-Habsi et al.	2008	UK	General practitioners who had directly referred an asymptomatic patient to a regional genetics centre ($N = 36$)	Semi-structured interviews	Thematic analysis	Independent coding	Factors associated with referral of asymptomatic individuals to cancer genetics clinic
Birmingham et al.	2013	USA	Health care providers recruited through a Cancer Institute ($n = 6$), community urology practices ($n = 8$), and community practice clinics ($n = 10$). Of these, 10 were primary care physicians and 14 were urologists or urology residents ($N = 24$)	5 focus groups, each informed by an education session	Grounded theory approach	Independent coding	Attitudes towards the utility and integration of genomic medicine for prostate cancer susceptibility into clinical practice
Bottorff et al.	2000	Canada	Physicians who had referred an ineligible		Thematic analysis	Review of analysis amongst researchers.	Referral practices for genetic testing and

patient for breast cancer risk assessment at a hereditary cancer centre ($N = 10$)	Semi-structured telephone interviews	Themes checked against data and final interviews.	experiences in counselling women about breast cancer risk and genetic testing (focusing on women's experience of inability for testing) Nurses roles in providing clinical genetic services related to adult onset hereditary disease and factors that support and limit opportunities, and influence genetic nursing practice
Bottorff et al. 2005	Canada Nurses sampled across 5 provinces who provide genetic services for adult onset hereditary disease ($N = 22$)	Thematic analysis of initial interviews and review of emerging themes	Nurses roles in providing clinical genetic services related to adult onset hereditary disease and factors that support and limit opportunities, and influence genetic nursing practice
Carroll et al. 2003	Canada Maximum variation sample of family physicians across Ontario ($N = 40$)	Constant comparative method	Family physicians' experiences, perceived role, and training needs in dealing with genetic susceptibility to cancer.
Cox and Starzomski 2004	Canada Interviews were carried out with 5 nephrologists, 3 nephrology nurses, 3 social workers, 3 medical geneticists, and 2 genetic counsellors practicing in three different	Focus groups using semi-structured interview guide 16 semi-structured interviews and one focus group	Second researcher observed focus groups and compared field notes. Independent coding of transcripts. Presentation of results to participants Independent coding
		Thematic analysis/naturalistic enquiry perspective	Social construction and clinical management of Polycystic Kidney Disease (PKD), perceptions of the role of genetics in PKD and views on pre-symptomatic testing

(Continued)

Table 2. (Continued).

Authors	Year	Country	Sample	Qualitative methods	Analysis	Quality checks	Topic of investigation
Elwyn, Iredale, and Gray	2002	Wales	regions. The focus group was attended by 2 nephrologists, 3 nephrology nurses and 2 social workers ($N = 23$) A purposive sample of 6 general practitioners (GPs) involved in postgraduate teaching, and a sample of 8 GPs with no academic role. ($N = 14$)	Two sequential focus group, each preceded by informational materials	Thematic analysis	Review and discussion of amongst researchers. Participant validation of themes	Reactions of GPs to a cancer genetics service controlled by referral guidelines and a triage system, and the perceived impact genetics will have on general practice Perceptions of genetic counselling and testing in African American women at moderate to high-risk of carrying a BRCA1/2 mutation
Graves et al.	2011	USA	Convenience and snowball sample of healthcare providers who routinely provide or refer women to genetic services, consisting of 5 genetic counsellors, 8 medical oncologists, 2 obstetrician/gynaecologists, and 4 breast surgeons ($N = 20$)	Structured interviews	Whole text content analysis	Independent coding. Review and discussion of themes amongst researchers	
Harvey	2011	UK	Genetics specialists ($n = 7$), diabetologists ($n = 5$), and general practitioners (primary	Semi-structured interviews (and relevant documentation,	Iterative thematic analysis	None reported	Views of primary, secondary and tertiary care practitioners on the possibility of using

Iredale et al.	2005	Wales	care doctors) ($n = 4$) from 2 primary care trusts and 2 hospitals in one region ($N = 16$)	not included in this meta-synthesis)	Thematic analysis	genetic testing to identify and manage susceptibility to type 2 diabetes mellitus (T2DM) within their clinical practice
Iredale et al.	2007	Wales	Professionals representing each of 9 surgeries in one region (10 doctors and 9 nurses) ($N = 19$)	Semi-structured interviews	Independent coding	Awareness of cancer genetics service and the referral guidelines used in the service, perceptions about the role that rurality plays in referral behaviour and whether or not referrals were being made to cancer genetic services outside Wales
Iredale et al.	2007	Wales	Consultants ($n = 2$), clinical associates including specialist registrars ($n = 4$) and genetic counsellors ($n = 2$) working for the national genetics service ($N = 8$)	One focus group	Discussion of main themes amongst researchers	Views of geneticists of a range of possible delivery models for cancer genetics, and how cancer genetic services might develop in the future
Kenen et al.	2011	UK	Medical geneticists ($n = 3$), oncologists ($n = 2$); cancer genetic nurse counsellors ($n = 3$); gynaecological surgeons ($n = 2$); and breast surgeons ($n = 2$). ($N = 12$)	Semi-structured interviews	Constant comparison amongst researchers	How health care professionals deal with uncertainty in counselling and treating women from hereditary breast/ovarian cancer families who receive

(Continued)

Table 2. (Continued).

Authors	Year	Country	Sample	Qualitative methods	Analysis	Quality checks	Topic of investigation
Komatsu and Yagasaki	2014	Japan	Breast specialists (oncologists/surgeons) ($n = 7$), staff physicians ($n = 5$), four nurses ($n = 4$) and one genetic counsellor ($N = 17$)	3 focus groups using semi-structured interview guide	Constant comparison	Discussion of analysis with co-author	inconclusive BRCA1/2 genetic test results (genetic tests which do not find a mutation to account for the family history) Perspectives of breastcared providers on awareness, implementation, and challenges of risk assessment and management of hereditary breast and ovarian cancer, their readiness for personalized cancer risk management in clinical practice
Lobb et al.	2001	Australia	Clinical geneticists ($n = 7$), genetic counsellors ($n = 20$) one oncologist and one nurse ($N = 29$)	Open-ended questionnaire	Constant comparative method	Review and discussion of themes amongst researchers	Perceptions of professionals working in cancer genetics about consultations and interactions with women from high-risk breast cancer families Evaluation of an initiative to integrate genetics into mainstream clinical provision
Martin et al.	2009	UK	Stakeholders (project leads and staff, steering-group members, clinicians, commissioners and	Semi-structured interviews	Thematic analysis	Independent coding. Participant validation of findings	

Mendes, Paneque, and Sousa	2012 Portugal	involved service users) ($N = 125$) Health care providers from 8 different institutions delivering cancer genetic counselling: geneticists ($n = 17$; 4 of which were interns), oncologists ($n = 2$), one obstetrician, genetic nurses ($n = 3$), psychologists ($n = 3$), and genetic counsellor trainees ($n = 4$; 3 of whom were psychologists and one nurse) ($N = 30$)	6 focus groups and 3 individual semi-structured interviews	Content analysis and constant comparison	Independent coding	Opinions about a multifamily psychoeducational programme for hereditary cancer susceptibility families, implemented at a Portuguese genetics service
Mendes, Sousa, and Paneque	2013 Portugal	Health care providers from 8 different institutions delivering cancer genetic counselling: geneticists ($n = 16$?); 4 of which were interns), oncologists ($n = 2$), one obstetrician, genetic nurses ($n = 3$), psychologists ($n = 3$), and genetic counsellor trainees ($n = 4$; 3 of whom were	6 focus groups and 3 individual semi-structured interviews	Open coding and constant comparison	Independent coding	Perceived need for the provision of psychosocial services and ways to enhance the psychosocial focus in oncogenetic service delivery

(Continued)

Table 2. (Continued).

Authors	Year	Country	Sample	Qualitative methods	Analysis	Quality checks	Topic of investigation
Miller, Giacomini, and Ahern	2008	Canada	psychologists and one nurse) ($N = 30$) Clinical geneticists ($n = 8$), non-genetic physicians (medical oncologists, surgeons, gastroenterologists; family physicians; $n = 9$), genetic counsellors ($n = 8$), nurses ($n = 4$), and scientists were involved in the development or provision of cancer genetic services ($n = 3$) ($N = 32$)	33 interviews	Case study methods and modified grounded theory approach (open coding using predetermined codes and constant comparison)	Participant validation of themes	How new and emerging genetic services should be provided, and what roles non-genetic clinicians should assume in hereditary cancer genetics
Overby et al.	2013	USA	Cancer genetic counsellors ($N = 8$)	Semi-structured telephone interviews	Open coding using interview questions as a codebook, emerging themes incorporated into the codebook inductively. Selective coding to determine major themes	Independent coding. Regular review and discussion of emerging themes amongst researchers. Participant validation of themes	What information cancer genetic counsellors seek from their patients to facilitate effective information exchange for discussing risk
Paneque et al.	2014	Portugal	Medical geneticists ($n = 9$), medical geneticist trainees ($n = 4$); psychologists ($n = 5$) ($N = 18$)	Semi-structured interviews	Thematic analysis using open coding and constant comparison	Review and consensus amongst research team	Views of professionals who provide genetic counselling services for presymptomatic testing for late-onset neurological diseases

Pfeffer, Veach, and Leroy	2003	USA	Genetic counsellors who provide familial risk counselling ($N = 25$)	Structured telephone interviews	Cross-case analysis and pattern matching method. Thematic analysis	Pilot evaluation of interview schedule by an experienced genetic counsellor. Independent coding using identified themes by an auditor	regarding relevant quality indicators and current assessment for counselling practice Views of genetic counsellors who provide familial cancer risk counselling on definitions of genetic discrimination, perceptions of patient risk for discrimination, frequency and type of discrimination experienced by their patients, sources of information about discrimination, and what they tell patients about genetic discrimination
Rees et al.	2006	Australia	Genetic counsellors ($n = 10$) and medical specialists (4 oncologists, 1 surgical oncologist, 1 clinical geneticist, and 1 medical oncologist/clinical geneticist; $n = 7$) working across 5 regional familial cancer centres ($N = 17$)	Three focus groups with genetic counsellors and one focus group and 3 individual interviews with specialists	Constant comparison	Comparison of focus group summary and final research outcomes. Member checking of focus group content. Pilot interviews with 2 genetic counsellors. Independent reading of transcripts and emerging themes	Views and practice regarding the provision of information about health-protective behaviours (e.g. exercise, alcohol consumption, diet) during genetic consultation for breast cancer

(Continued)

Table 2. (Continued).

Authors	Year	Country	Sample	Qualitative methods	Analysis	Quality checks	Topic of investigation
Stermer et al.	2004	UK	Primary care providers (general practitioners and practice nurses) and colorectal cancer specialists (gastroenterologists, colorectal surgeons, colorectal cancer nurse specialists) ($N = 35$)	3 focus groups with primary care providers and 10 individual interviews with specialists	Visual qualitative analysis	Participant validation of emerging themes. Discussion of emerging themes amongst researchers	Views of primary and secondary care health professionals on how to improve current services for individuals with a family history of colorectal cancer
Tan and Fitzgerald	2014	Australia	General practitioners ($n = 7$), gynaecologists ($n = 6$), gastroenterologists ($n = 6$), medical oncologists ($n = 3$) and gynaecology oncologists ($n = 6$) ($N = 28$)	Semi-structured interviews	Thematic analysis	Independent analysis of transcripts by 2 researchers	Views of general practitioners and specialists on their referral of patients with suspected Lynch syndrome to cancer genetic services
Walsh et al.	2012	USA	3 Genetic counsellors and 12 physicians (including specialists in internal medicine, medical oncology, and gastroenterology) ($N = 15$)	Focus groups using structured interview schedule and discussion of testing vignettes	Content analysis	Independent coding	Key characteristics that health professionals value regarding genetic testing and personalized medicine using the example of genetic testing for hereditary Lynch syndrome
Will, Armstrong, and Marteau	2010	UK	Snowball and opportunistic sample of general practitioners ($n = 5$), specialist	Semi-structured interviews including discussion of	Thematic analysis	Independent coding	Implications and utility of genetic testing in the case of familial hypercholesterolemia

Yamanaka and Takeda	2011	Japan	<p>physicians in lipid clinics with access to genetic testing ($n = 6$), specialist physicians in lipid clinics without such access ($n = 5$) and nurses from both types of lipid clinics who were often given the task of explaining the implications of findings ($n = 4$) ($N = 20$)</p> <p>4 diabetologist and 1 dietician workings in 3 clinics and 1 healthcare centre who routinely use predictive genetic testing for lifestyle disease ($N = 5$)</p>	Semi-structured interviews	None reported. Notes of each participants' responses and other quotations included in report	None reported	Views of clinicians using testing in primary prevention of lifestyle disease on merits and demerits of testing, and reasons for ordering testing
---------------------	------	-------	--	----------------------------	--	---------------	--

2012, 2013). Metasynthesis has also been used to understand individuals' perception of disease risk (Walter, Emery, et al. 2004). Applying this approach, the studies identified in our systematic review were analysed using thematic analysis (Braun and Clarke 2006). All 28 studies were initially coded by MF and a sample of 7 (25%) were independently coded by RS. Coding agreement between researchers was high. Initial first-order coding was applied to participant quotations and to key findings for each of the studies included. Initial codes and any discrepancies were discussed and agreed by MF and RS, and classified into second-order thematic categories. Higher (third) order themes were then interpreted to try and integrate the second-order themes and to describe inter-relationships amongst them. There was continuous review and comparison of themes amongst researchers (MF, RS, and KR) at each stage of the analysis.

Results

Of the 28 studies included in the metasynthesis, 21 explored HCPs' perceptions of predictive genetic testing and/or genetic risk counselling for cancer, 1 for familial hypercholesterolemia, 1 for polycystic kidney disease, 1 for diabetes, 1 for late-onset neurodegenerative disorders and 3 for predictive testing for more than one adult onset disease.

Nine of the studies were conducted in the United Kingdom, 5 in the USA, 5 in Canada, 3 in Australia, 3 in Portugal, 2 in Japan and 1 in Norway.

The main themes developed from the metasynthesis were: (1) Value of genetic risk information; (2) Understanding of genetic risk information; (3) Consequences of genetic risk information; and (4) Responsibility for genetic risk information. Illustrative quotations for each of these themes and associated subthemes are presented below and supplemented in Table 3 (T3; Q1–79).

Theme 1: Value of genetic risk information

This theme describes the views expressed by HCPs about the value of genetic risk information. These views relate to the clinical utility and validity of such information, use of genetic information in current and future practice, and to the potential of information about genetic risk to promote healthy behavioural change by patients.

1a. Utility and validity (T3; Q1–10)

Many HCPs felt that genetic test results did not give useful information about disease risk over and above information already available to them from family history and/or environmental risk factors (Birmingham et al. 2003; Cox and Starzomski 2004; Martin, Currie, and Finn 2009; Harvey 2011; Will, Armstrong, and Marteau 2010; Graves et al. 2011; Yamanaka and Takeda 2011; Tan and Fitzgerald 2014).

The problem with this is ... how does that change what we already know from years and years of PSA, age, family history, all the normal clinical things ...? [Provider, USA; Birmingham et al., 2003, 8]

Professionals stated that risk information from genetic tests lacked validity, and was not yet supported by sufficient evidence to be of use. The prospect of making medical decisions on the basis of information associated with a high level of uncertainty

Table 3. Quotations to support themes and subthemes.

 Theme 1. Value of genetic risk information
Subtheme 1a. Utility and validity

- Q1. The problem with this is ... how does that change what we already know from years and years of PSA, age, family history, all the normal clinical things ...? [Provider, USA; Birmingham et al. 2003, 8]
- Q2. There is a limitation on how useful testing is ... how is this going to affect this person's life? Is it going to add quality of life or quantity of life? [General practitioner; Australia; Tan and Fitzgerald 2014, 25]
- Q3. Data is not compelling enough and seems more likely to lead to dilemma in interpretation and further recommendations. [Provider, USA; Birmingham et al. 2003, 5]
- Q4. If you're going to do something which has a profound significance, you've got to be damn sure of the validity of your advice according to the results of that test. And I don't think we're in that position right now. [Family physician, Canada; Carroll et al. 2003, 49]
- Q5. We know it's [genetic testing] going to improve, but it's not here yet, and that very first of all it's frustrating, and because you like to practice evidence based medicine. [Oncologist, UK; Kenen et al. 2011, 5]
- Q6. I think that quite often having the DNA test, although it can be useful and you know in the case of a family member who wants to know are they at risk of developing high cholesterol they may want to have the genetic test, but having said that very often there isn't any particular advantage over and above just keeping an eye on their cholesterol anyway. [Consultant physician, UK; Will, Armstrong, and Marteau 2010, 913]
- Q7. Because at the moment I am looking at risks anyway, just normal family history, and if someone has a strong family history I will be emphasizing it more anyway. So yes, that would be just another one. You know, it would just be another issue. [General practitioner, UK; Harvey 2011, 319]
- Q8. We do know that there are some families in which there is definitely a genetic component. I think unfortunately anyway the treatment is the same, it doesn't change our treatment. [Diabetologist, UK; Harvey 2011, 317]
- Q9. We don't want to obsess and medicalise our patients when we have quite a lot of suspicion that there is not going to be much that can be offered by the experts to reassure them. [General practitioner, UK; Stermer et al. 2004, 50]
- Q10. I don't send them to a geneticist ... Only if it's a young woman who wants to have kids and she wants to have a different kind of discussion than I give her then I'll send them, but otherwise not ... what will they add to what I can do except freak them out? ... It's like it sounds very scary to go to medical genetics and it's like a genetic disease and all of a sudden it's got this whole other spin to it. [Nephrologist, Canada; Cox and Starzomski 2004, 146]

Subtheme 1b. Use of genetic testing in current and future practice

- Q11. The frustrating thing is all these tests become available so quickly and you're swept up into doing them or people are coming in and asking for certain things, and... you don't necessarily realize all the consequences at that point. You're being swept along in this wave of newer technology ... It's really overwhelming. It's hard to know if you're doing good by ordering these tests. [Family physician, Canada; Carroll et al. 2003, 49]
- Q12. This is the tip of the iceberg in terms of genetic testing.... As we have more genes to screen for, it's not going to be practical to send everyone to the genetics clinic. There's going to be more pressure for us to do it. ... At some stage we're going to end up having to do it. [Family physician, Canada; Carroll et al. 2003, 48]
- Q13. I think cancer genetics will, I think all predictive genetics will end up becoming part of general health care. We have, I just counted, I think 30 clinical geneticists in Ontario. Probably about 100 across the country. We cannot possibly see all these people. We just can't. So I think what has to happen is that the geneticists will end up in a consultative
-

(Continued)

role and we will kind of get things going. [Clinical geneticist, Canada; Miller, Giacomini, and Ahern 2008, 156]

Q14. Some people come and say ‘Can I be referred to find out more?’ And that’s what they want. So then it’s quite hard to say No. [General practitioner, UK; Al-Habsi et al. 2008, 755]

Subtheme 1c. Impact on health related behaviour

Q15. I guess Polycystic is a little different, because there are things you can do, there’s even diet, you can take control of your diet. And there is a sense that you’re doing something, right? [Social worker, Canada; Cox and Starzomski 2004, 145]

Q16. It might actually, if a patient sees the increased risk, increase their compliance as far as coming in for regular exams or at least being willing to get the digital rectal exam. [Provider, USA; Birmingham et al. 2003, 8]

Q17. You can’t get people to do some of these things [adhere to diet and exercise recommendations] that we have known for years and have good statistical information. We can prove it over and over again how much more beneficial it would be for them and we can’t get them to do it. You get a test like this, and I don’t know if it would help or not. [Provider, USA; Birmingham et al. 2003, 8]

Q18. You can talk until you’re blue in the face about all these things being a good thing but actually, these kind of health behaviors are very difficult if people aren’t doing them already and which is why the world is becoming a fatter place. Because people don’t do the things they’re meant to be doing. So I think although I think it’s my role to promote these things and we’ll talk about it, I feel a bit nihilistic about the fact that I don’t think people are going to change. Most people are not going to change their behaviors. [Medical specialist, Australia; Rees et al. 2006, 101]

Theme 2. Understanding of genetic risk information

Subtheme 2a. Assessment of understanding

Q19. Most women don’t know. They kind of come up with this statement of, ‘Okay, I want to know if I carry the gene or not.’ They have no idea what the implications are, what it means, what the risks are. I mean, it is sort of like a ‘yes or no’ test they are looking for. [Physician, Canada; Bottorff et al. 2000, 1453]

Q20. And sometimes I have to sit and think about ... [the results] because like you said, just in that little discussion we had, we were all looking at it [statistical computation of risk]and going, now wait a minute, what is this and what is this? [Provider, USA; Birmingham et al. 2003, 7]

Q21. The most common scenario in my office is that people come in because they have one or two or more first- or second-degree relatives with cancer and say, ‘What are my chances of getting this?’ And usually I don’t know. [Family physician, Canada; Carroll et al. 2003, 48]

Q22. I don’t think when people are thinking about their risk, they’re thinking about them in numbers the way we are, so as geneticists, we’re thinking about your risk as 15 vs. 10%, but for the average woman, they might just be feeling like, ‘Yeah, my mom and sister got breast cancer; it’s probably going to happen to me too’. [Genetic counsellor, USA; Overby et al. 2013, 245]

Q23. I define genetic counselling as recognizing and exploring issues that are relevant for a particular family member. For example, previous family history, what happened, how that has affected the client’s view of breast cancer, what the outcomes have been for the family, what position/age the person was when she (first) became aware of family history and the impact that has had on her (e.g. mother diagnosed with breast cancer when going through adolescence). [Genetic counsellor, Australia; Lobb et al. 2001, 190]

Q24. The patients do not know the type of cancer in the family; they can be remarkably unspecific. [General practitioner, UK; Al-Habsi et al. 2008, 755]

Q25. There are patients here I saw three years ago with nothing particular in their family history and they present three years later with two or three family members with breast cancer. [Nurse, Wales; Iredale et al. 2005, 201]

(Continued)

Subtheme 2b. Determinants of understanding

- Q26. I think it is important to know the personal background, where the consultands come from, their motivations for being here, what do they already know about the illness and the test, in order to allow us to deconstruct their beliefs, if needed. [Clinical psychologist, Portugal; Paneque et al. 2014, 4]
- Q27. You need to change your approach according to the sensibilities of your patients so as to assure positive effects of the test results. [Diabetologist, Japan; Yamanaka and Takeda 2011, 220]
- Q28. Patients here actually fall into categories. On the one hand very well informed patients who want to know everything... And then the other extreme who are extremely frightened, don't want to know [Consultant physician, UK; Will, Armstrong, and Marteau 2010, 913]
- Q29. And you might say that ... what you notice when you have [counselling sessions for] such large families is that people have (...) made up their minds of whether they have inherited it [the known gene fault] prior to their appointments. [Genetic counsellor, Norway; Aasen and Skölbekken 2014, 377]
- Q30. Sometimes they [African American women] just don't want to know or are not ready to know yet ... but sometimes it's really the family that doesn't want to know so they discourage it [getting tested]. [Genetic counsellor, USA; Graves et al. 2011, 684]
- Q31. Now when the media [come] out with a lot of publicity and some people aren't particularly eligible or the risk isn't as high as they think it is, [they] come in and ask me about it. We just kind of go over it. Like, your risk isn't particularly high. You can see [genetic testing programs] if you want, but your risk isn't as high as you think it is. [Physician, Canada; Bottorff et al. 2000, 1453]
- Q32. I get a lot of people who have got this stuff off the Internet who want to know, 'Where can I get genetic screening? Am I going to get cancer? [Family physician, Canada, Carroll et al. 2003, 48]
- Q33. We have some Caucasian patients that clearly don't need to be tested and are clamoring for it, and it's... the opposite in the African American community. [Breast surgeon, USA; Graves et al. 2011, 680]
- Q34. It can make choices at times when they are otherwise healthy difficult, and it could be confounded, for example, if they don't have good medical care or insurance. [Medical oncologist, USA; Graves et al. 2011, 678]

Subtheme 2c. Facilitating understanding

- Q35. I think people need to hear the information in several different ways, so increased risks, you know, decreased risks, no risk or population risk of breast cancer, that can mean very different things to people. [Genetic counsellor, USA; Overby et al. 2013, 244]
- Q36. In all cases, we give support materials for consultation after the visit to the centre, for calm reading ... when we are discussing more complex information I like to use metaphors, so the comprehension of genetic information will be facilitated. [Resident of medical genetics, Portugal; Paneque et al. 2014, 4]
- Q37. For polycystic kidney disease we would need to obviously have supplemental education pamphlets you can hand out to people. Give them easy to read information that they can take home with them. [Nurse, Canada; Cox and Starzomski 2004, 147]
- Theme 3. Consequences of genetic risk information

Subtheme 3a. Anxiety

- Q38. It's like why are we doing a test that raises anxiety and increases the cost of care? [Provider, USA; Birmingham et al. 2013, 8]
- Q39. People often get distressed when they realize that close relatives may also be at risk, especially younger descendants under 18 or in their twenties. Positive results in such cases are the most dramatic. [Genetic HCP; Portugal; Mendes, Sousa, and Paneque 2013, 777]

(Continued)

- Q40. Sometimes we're actually going to increase people's anxiety... and sometimes we're going to alleviate anxiety, but I mean part of our role is to define those risks for them and help them understand what's going on ... to get more information. Getting the genetic information is part of your information-gathering process, and if you don't do that, then you're not using all the tools you have. [Family physician, Canada; Carroll et al. 2003, 49–50]
- Q41. I will always try to ... I don't know if it's always [the] right [strategy] but ... at least at the beginning [of the consultation], then I always try ... not to frighten them. [Genetic counsellor, Norway; Aasen and Skolbekken 2014, 380]
- Q42. We do [evaluate when or if psychosocial support is needed] ... somehow we try to manage by ourselves the difficult moments in the consultation but it is not enough because there is no psychologist in the service! [Genetics HCP, Portugal; Mendes, Sousa, and Paneque 2013, 776]
- Q43. Well, certainly in terms of explaining basic genetics, I can explain that to anyone probably in my sleep, but if something unusual shows up on a genetic test, that is the benefit of working in a multidisciplinary team – if you don't know the answer maybe someone else does. [Nurse, Canada; Bottorff et al. 2005, 104]
- Q44. I am not trained in counselling, and I rely on members of my team to fill that vital role. [Clinical geneticist, Australia; Lobb et al. 2001, 194]
- Q45. Psychosocial issues are linked to genetic counseling ... but I need to separate what is not my expertise and guarantee a good articulation with the consultation meant for those issues with our psychologist. [Medical geneticist, Portugal; Paneque et al. 2014, 5]
- Q46. Sometimes I refer because the patient is extremely anxious. [General practitioner, UK; Al-Habsi et al. 2008, 755]
- Q47. It empowers people to find out [their risk of a BRCA1/2 mutation] and alleviate fears. [Obstetrician/gynaecologist, USA; Graves et al. 2011, 678]

Subtheme 3b. Discrimination and stigmatization

- Q48. I remember one [patient] who told me 'I don't want anybody to peek into my private life.' [Another patient] said, 'I think if I find out I have BRCA1 then ... I won't get health insurance because of discrimination'. [Medical oncologist, USA; Graves et al. 2011, 678]
- Q49. I think the potential for discrimination down the road may be substantial. [Genetic counsellor, USA; Pfeffer, Veach, and LeRoy 2003, 426]
- Q50. (Genetic testing) potentially opens cans of worms as far as the daughter's insurability is concerned ... knowing that she's got the genetic predisposition to cancer doesn't change anything. [Gynaecologist, Australia; Tan and Fitzgerald 2014, 4]
- Q51. If genetic information is included in a patient's electronic medical record, we are concerned over potential information leakage. [Breast specialist, Japan; Komatsu and Yagasaki 2014, 42]
- Q52. African American patients oftentimes tend to be more suspicious about [genetic testing] ... because they're so used to being pointed out all the time. And maybe they feel it's another way of [labeling them]. [Medical oncologist, USA; Graves et al. 2011, 680]
- Q53. I believe this can be very therapeutic as in many cases these families feel isolated and stigmatised. [Genetics HCP, Portugal; Mendes, Paneque, and Sousa 2012, 314]

Subtheme 3c. Impact on family

- Q54. Within genetics we are very aware of the family issues of genetic testing and we often feel that, you know, it can be described as being unethical to do genetic tests without consideration of the impact on other members of the family. [Genetics specialist, UK; Harvey 2011, 314]
- Q55. If we find a mutation in one of our patients, we want her relatives to take the test as soon as possible. But it is a very sensitive issue because each family's situation is different [Genetic counsellor, Japan; Komatsu and Yagasaki 2014, 41]
- Q56. Many of them [oncologists] are still stuck in the mode of treating the diseased person, not the family. The vast majority are still in that mode, because that is how they've been trained. In order to treat the person with the disease, they don't really deal

(Continued)

with the whole family who is worried about having the disease. [Clinical geneticist, Canada; Miller, Giacomini, and Ahern 2008, 155]

- Q57. Some patients do not want their families to know about their conditions. It is not easy to give the family members a telephone call [Breast specialist, Japan; Komatsu and Yagasaki 2014, 41]
- Q58. We feel sometimes is complicated for people to get in touch with them [potentially at-risk relatives] and tell them [they might be at-risk]. Sometimes conflicts prevent this happening, or people don't see each other often, or because they fear a bad reaction. [Genetics HCP, Portugal; Mendes, Sousa, and Paneque 2013, 777]
- Theme 4. Responsibility for genetic risk information

Subtheme 4a. Genetic risk as a burden

- Q59. People feel so guilty for transmitting or potentially transmitting a genetic disease which, when you think about it is, I mean they shouldn't. [Nephrologist, Canada; Cox and Starzomski 2004, 149]
- Q60. If the mother has the gene, she feels like she might have tainted her offspring. If a sister doesn't have the gene and her sister does, then the sister that doesn't have it might feel guilty. [Breast surgeon, USA; Graves et al. 2011, 681]
- Q61. For a woman who has a strong family history she may learn that she doesn't carry a familial genetic risk factor so she may be relieved from having to make various medical decisions. Sometimes gaining additional information about risk can be reassuring even if people learn that they're at increased risk because at least they have information. [Genetic Counsellor, USA; Graves et al. 2011, 681]
- Q62. [I could tell them that their cholesterol was] different from Joe Public's cholesterol which may well be raised because of a bad diet or something else. [Consultant physician, UK; Will, Armstrong, and Marteau 2010, 914]

Subtheme 4b. Responsibility to act on risk information

- Q63. In some situations, [it's] a disadvantage that the first person to be tested in the family gets the job of having to spread the information to everyone and that can be somewhat of a burden for people. [Genetic counsellor, USA; Graves et al. 2011, 682]
- Q64. It is up to the patient to decide whether to inform her relatives of the results of the genetic test. [Breast specialist, Japan; Komatsu and Yagasaki 2014, 42]
- Q65. Because potentially you could really be, you know, sort of adding to somebody's burden by raising those issues if, well one, they hadn't thought about it and then they were raised and they thought, 'Oh my goodness I really need to do something about this' or, 'I haven't been doing this and this, you know, it's going to contribute to my disease risk' and all of that sort of thing. [Genetic counsellor, Australia; Rees et al. 2006, 101]
- Q66. Secondly, you know that probably some people are having surgery unnecessarily, and thirdly ... you would feel much happier about doing the screening measures or prophylactic surgery measure that were quite extreme, if this level of certainty was much higher [Oncologist, UK; Kenen et al. 2011, 5]
- Q67. It's a two-way-street. It's our responsibility to make sure that families know that nephrology is there in terms of long-term management and care issues [Geneticist, Canada; Cox and Starzomski 2004, 153]
- Q68. Even if the counselling is done in what is now secondary care, patients will still come back to us with their letter and ask to have it interpreted. ... it will always end up back with us. ... the main pressures for us to take on counselling is going to come from the patients because if a patient is sitting in front of you and wants an answer, you feel obliged to do your best to supply what they need. [General practitioner, Wales; Elwyn, Iredale, and Gray 2002, 69]
- Q69. The follow-up should still be a physician-based thing. So I don't really have a problem with the clinics that do the genetic counselling and then send the patient to whoever would be doing the screening [Genetic counsellor, Canada; Miller, Giacomini, and Ahern 2008, 157]

(Continued)

Subtheme 4c. Non-directive approach

- Q70. I think as geneticists we can say, you know, these are the percentages, we don't know ... and it's your decision in the end, so I think in some ways we're not taking such a responsibility. I'm sure that surgeons must feel more uneasy than the geneticists. [Oncologist, UK; Kenen et al. 2011, 9]
- Q71. I usually say I can't guarantee this information won't be used against them. [Genetic counsellor, USA; Pfeffer, Veach, and LeRoy 2003, 426]
- Q72. We should adopt approaches beyond the medical- traditionalist model [in genetic counselling] because it is very paternalistic and therefore very difficult to apply straightforwardly in genetics because we deal with predictive territories. [Genetic HCP, Portugal; Mendes, Sousa, and Paneque 2013, 775]
- Q73. And the other thing I emphasized was that if you knew early that you have the disease it will become a decision. You have a choice of whether you want children. And that I tend to shy away from. I just explain to them the chances, not really telling them exactly what to do because I believe that is a very personal thing. [Nephrologist, Canada; Cox and Starzomski 2004, 149–150]
- Q74. We follow a purely clinical model based on risk measures after collecting the family history and evaluate if the genetic test is pertinent, and then it is the counselee who will decide! It is a medical consultation and our role is merely to give correct information and refer to surveillance or risk reduction options if appropriate. [Genetics HCP, Portugal; Mendes, Sousa, and Paneque 2013, 775]

was described as problematic, and likely to lead to 'dilemma in interpretation' that is at odds with the security associated with notions of evidence-based practice and professional expertise (Birmingham et al. 2003; Carroll et al. 2003; Miller, Giacomini, and Ahern 2008; Kenen et al. 2011).

If you're going to do something which has a profound significance, you've got to be damn sure of the validity of your advice according to the results of that test. And I don't think we're in that position right now. [Family physician, Canada; Carroll et al. 2003, 49]

The lack of perceived added value of genetic information to medical practice was described in relation to the current practice of the HCPs (Will, Armstrong, and Marteau 2010; Harvey 2011). There is, perhaps, an underlying assumption that current service provision should be viewed as appropriate and sufficient.

Because at the moment I am looking at risks anyway, just normal family history, and if someone has a strong family history I will be emphasizing it more anyway. So yes, that would be just another one. You know, it would just be another issue. [General practitioner, UK; Harvey 2011, 319]

Furthermore, some HCPs emphasised that the provision of genetic risk information was not always accompanied by effective additional intervention or treatment and so had limited clinical utility (Cox and Starzomski 2004; Stermer et al. 2004; Harvey 2011; Paneque et al. 2014). We do know that there are some families in which there is definitely a genetic component. I think unfortunately anyway the treatment is the same, it doesn't change our treatment. [Diabetologist, UK; Harvey 2011, 317]

Inactionable genetic information was sometimes associated with a desire to protect patients from unnecessary medicalization (Stermer et al. 2014) or fear (Cox and Starzomski 2004).

We don't want to obsess and medicalise our patients when we have quite a lot of suspicion that there is not going to be much that can be offered by the experts to reassure them. [General practitioner, UK; Stermer et al. 2004, 50]

Ib. Use of genetic testing in current and future practice (T3; Q11–14)

HCPs were uncomfortable with the speed with which genetic technology has advanced, in the light of their reservations about the clinical utility of genetic risk information. This concern was emphasized by the emotive language that was frequently used in this context. HCPs were wary of potential consequences and implications of genetic test results, which may outweigh any clinical benefit and which may be overlooked in the ‘overwhelming’ pervasive trend towards genetic medicine (Carroll et al. 2003; Cox and Starzomski 2004; Kenen et al. 2011).

The frustrating thing is all these tests become available so quickly and you’re swept up into doing them or people are coming in and asking for certain things, and ... you don’t necessarily realize all the consequences at that point. You’re being swept along in this wave of newer technology... It’s really overwhelming. It’s hard to know if you’re doing good by ordering these tests. [Family physician, Canada; Carroll et al. 2003, 49]

Although many HCPs referred to an inevitable future increase in the role for genetic testing (Elwyn, Iredale, and Gray 2002; Carroll et al. 2003; Cox and Starzomski 2004; Iredale, Jones et al. 2005; Iredale, Elwyn et al. 2007; Al-Habsi et al. 2008; Miller, Giacomini, and Ahern 2008; Martin, Currie, and Finn 2009; Kenen et al. 2011), this trend was associated with feeling under ‘pressure’ to respond to an increased demand for genetic services, and with feeling unable to respond without specialist support.

I think cancer genetics will, I think all predictive genetics will end up becoming part of general health care. We have, I just counted, I think 30 clinical geneticists in Ontario. Probably about 100 across the country. We cannot possibly see all these people. We just can’t. So I think what has to happen is that the geneticists will end up in a consultative role and we will kind of get things going. [Clinical geneticist, Canada; Miller, Giacomini, and Ahern 2008, 156]

Whilst the translation of genetic technology was described as a future prospect, current provision of predictive genetic testing was described as reactive by some HCPs, responding to self-selected patient’s, rather than being under the direction of the HCP (Elwyn, Iredale, and Gray 2002; Cox and Starzomski 2004; Al-Habsi et al. 2008; Martin, Currie, and Finn 2009; Paneque et al. 2014).

Some people come and say ‘Can I be referred to find out more?’ And that’s what they want. So then it’s quite hard to say no. [General practitioner, UK; Al-Habsi et al. 2008, 755]

In this situation, the risk information provided is sought out and valued by the patient rather than the provider. This is, perhaps at odds with traditional notions of professional authority and expertise.

Ic. Impact on health-related behaviour (T3; Q15–18)

A few HCPs stated that genetic risk information relating to multifactorial diseases could have a positive impact on patients’ health-related behaviour. On the one hand, such information was described as potentially ‘empowering’, by encouraging individuals to make appropriate changes to their lifestyle (Cox and Starzomski 2004; Rees et al. 2006; Will, Armstrong, and Marteau 2010; Yamanaka and Takeda 2011), whilst on the other hand this information was conceived of as potentially useful to

align patient behaviour towards ‘compliance’ with medical advice (Birmingham et al. 2003; Graves et al. 2011).

I guess Polycystic is a little different, because there are things you can do, there’s even diet, you can take control of your diet. And there is a sense that you’re doing something, right? [Social worker, Canada; Cox and Starzomski 2004, 145]

It might actually, if a patient sees the increased risk, increase their compliance as far as coming in for regular exams or at least being willing to get the digital rectal exam. [Provider, USA; Birmingham et al. 2003, 8]

However, many were pessimistic about their influence on their patients’ lifestyle choices, and felt that a positive effect of genetic risk information on patients’ health behaviours was unlikely (Birmingham et al. 2003; Rees et al. 2006). Some felt that an emphasis on genetic risk factors could even have a negative behavioural effect by detracting from the importance of lifestyle-related risk factors (Birmingham et al. 2003; Iredale et al. 2007; Yamanaka and Takeda 2011).

You can talk until you’re blue in the face about all these things being a good thing but actually, these kind of health behaviors are very difficult if people aren’t doing them already and which is why the world is becoming a fatter place. Because people don’t do the things they’re meant to be doing. So I think although I think it’s my role to promote these things and we’ll talk about it, I feel a bit nihilistic about the fact that I don’t think people are going to change. Most people are not going to change their behaviors. [Medical specialist, Australia; Rees et al. 2006, 101]

These accounts reflect a sense of powerlessness and frustration on the part of providers in relation to their inability to effect positive behavioural change by their patients, both in response to genetic information and more generally. As patients often exercise their autonomous right to choose not to adhere to medical advice, the HCPs felt ‘nihilistic’ about the effectiveness of the information they provide.

Theme 2: Understanding of genetic risk information (T3; Q19–25)

This theme describes HCPs’ views on patients’ and HCPs’ abilities to understand and make appropriate use of genetic risk information, and how this relates to the demand for and the delivery of such information.

2a. Assessment of understanding

Several HCPs noted that patients often do not understand the nature of genetic risk information, and that patients do not appreciate the uncertainty associated with risk information (Bottorff et al. 2000; Lobb et al. 2001; Elwyn, Iredale, and Gray 2002; Cox and Starzomski 2004; Graves et al. 2011; Kenen et al. 2011; Overby et al. 2013; Aasen and Skolbekken 2014).

Most women don’t know. They kind of come up with this statement of, ‘Okay, I want to know if I carry the gene or not.’ They have no idea what the implications are, what it means, what the risks are. I mean, it is sort of like a ‘yes or no’ test they are looking for. [Physician, Canada; Bottorff et al. 2000, 1453]

A level of uncertainty surrounding genetic information was also felt by HCPs. Some HCPs expressed reservations about their own understanding and confidence in genetic risk assessment (Elwyn, Iredale, and Gray 2002; Carroll et al. 2003;

Al-Habsi et al. 2008; Graves et al. 2011; Kenen et al. 2011; Birmingham et al. 2013; Mendes, Sousa, and Paneque 2013; Tan and Fitzgerald 2014).

And sometimes I have to sit and think about ... [the results] because like you said, just in that little discussion we had, we were all looking at it [statistical computation of risk] and going, now wait a minute, what is this and what is this? [Provider, USA; Birmingham et al. 2003, 7]

However, despite reservations about their own knowledge, HCPs clearly saw their own understanding of genetic risk as being qualitatively different, and superior to, the understanding of members of the public. Professional understanding is described in terms of precise, quantitative estimates, whereas public understanding is described in vague, experiential terms (Overby et al. 2013).

I don't think when people are thinking about their risk, they're thinking about them in numbers the way we are, so as geneticists, we're thinking about your risk as 15 vs. 10%, but for the average woman, they might just be feeling like, 'Yeah, my mom and sister got breast cancer; it's probably going to happen to me too'. [Genetic counsellor, USA; Overby et al. 2013, 245]

HCPs described their role as the process of making a complex evaluation of the multiple factors that could influence the understanding and beliefs of individual patients, in order to ascertain their needs (Lobb et al. 2001).

I define genetic counselling as recognizing and exploring issues that are relevant for a particular family member. For example, previous family history, what happened, how that has affected the client's view of breast cancer, what the outcomes have been for the family, what position/age the person was when she (first) became aware of family history and the impact that has had on her (e.g. mother diagnosed with breast cancer when going through adolescence). [Genetic counsellor, Australia; Lobb et al. 2001, 190]

The professional role was sometimes described as being complicated by difficulty in eliciting reliable information from patients about their family history (Cox and Starzomski 2004; Iredale et al. 2005; Al-Habsi et al. 2008; Graves et al. 2011; Overby et al. 2013; Tan and Fitzgerald 2014).

There are patients here I saw three years ago with nothing particular in their family history and they present three years later with two or three family members with breast cancer. [Nurse, Wales; Iredale et al. 2005, 201]

2b. Determinants of understanding (T3; Q26–34)

Patients' understanding of genetic risk and their reactions to risk information were described as being influenced by a wide range of cognitive processes and social factors, including patients' beliefs, expectations and motivations. HCPs described making judgements about how these factors affect the level of understanding. Furthermore, depending on the level of understanding and motives for undergoing testing HCPs would modify their approach to match the perceived needs of the patient (Lobb et al. 2001; Pfeffer, Veach, and LeRoy 2003; Cox and Starzomski 2004; Stermer et al. 2004; Rees et al. 2006; Will, Armstrong, and Marteau 2010; Yamanaka and Takeda 2011; Mendes, Paneque, and Sousa 2012; Mendes, Sousa, and Paneque 2013; Overby et al. 2013; Aasen and Skolbekken 2014; Paneque et al. 2014).

You need to change your approach according to the sensibilities of your patients so as to assure positive effects of the test results. [Diabetologist, Japan; Yamanaka and Takeda 2011, 220]

HCPs referred to variability in patients' desire to know their disease risk status, which was associated with patients' level of understanding and knowledge. (Will, Armstrong, and Marteau 2010; Graves et al. 2011; Yamanaka and Takeda 2011; Komatsu and Yagasaki 2014; Tan and Fitzgerald 2014).

Patients here actually fall into categories. On the one hand very well informed patients who want to know everything ... And then the other extreme who are extremely frightened, don't want to know [Consultant physician, UK; Will, Armstrong, and Marteau 2010, 913].

Family background, particularly personal experience of a disease, and cultural background were frequently cited as important determinants of a patient's understanding of and responses to genetic risk (Lobb et al. 2001; Carroll et al. 2003; Cox and Starzomski 2004; Graves et al. 2011; Overby et al. 2013; Aasen and Skolbekken 2014; Komatsu and Yagasaki 2014; Tan and Fitzgerald 2014).

And you might say that ... what you notice when you have [counselling sessions for] such large families is that people have (...) made up their minds of whether they have inherited it [the known gene fault] prior to their appointments. [Genetic counsellor, Norway; Aasen and Skolbekken 2014, 377]

Sociocultural influences (Cox and Starzomski 2004; Graves et al. 2011; Mendes, Sousa, and Paneque 2013) and media influences (Bottorff et al. 2000; Elwyn, Iredale, and Gray 2002; Carroll et al. 2003; Kenen et al. 2011) were cited by HCPs in several of the articles included in this analysis as important determinants of public perceptions of risk.

Now when the media [come] out with a lot of publicity and some people aren't particularly eligible or the risk isn't as high as they think it is, [they] come in and ask me about it. We just kind of go over it. Like, your risk isn't particularly high. You can see [genetic testing programs] if you want, but your risk isn't as high as you think it is. [Physician, Canada; Bottorff et al. 2000, 1453]

Cultural differences in risk appraisal meant that some groups were less likely to participate in predictive testing than others, and that there was a misalignment between need for and demand for genetic testing.

We have some Caucasian patients that clearly don't need to be tested and are clamoring for it, and it's ... the opposite in the African American community. [Breast surgeon, USA; Graves et al. 2011, 680]

Many HCPs made reference to social inequalities not only in demand for, but also in access to genetic testing services (Elwyn, Iredale, and Gray 2002; Pfeffer, Veach, and LeRoy 2003; Iredale et al. 2007; Martin, Currie, and Finn 2009; Graves et al. 2011; Mendes, Paneque, and Sousa 2012; Mendes, Sousa, and Paneque 2013).

It can make choices at times when they are otherwise healthy difficult, and it could be confounded, for example, if they don't have good medical care or insurance. [Medical oncologist, USA; Graves et al. 2011, 678]

2c. Facilitating understanding (T3; Q35–37)

The approach taken by HCPs to the provision of genetic risk information was associated with their judgement of individual patients' needs and capabilities. The importance of communicating risk information in a variety of formats appropriate for the needs of the patient was noted by several HCPs, and some described using approaches such as the use of metaphors (Lobb et al. 2001; Stermer et al. 2004; Overby et al. 2013; Paneque et al. 2014).

I think people need to hear the information in several different ways, so increased risks, you know, decreased risks, no risk or population risk of breast cancer, that can mean very different things to people. [Genetic counsellor, USA; Overby et al. 2013, 244]

The use of supplementary sources of information was often mentioned, especially accessible informational resources that patients could take away and read in their own time (Elwyn, Iredale, and Gray 2002; Pfeffer, Veach, and LeRoy 2003; Cox and Starzomski 2004; Rees et al. 2006; Mendes, Sousa, and Paneque 2013; Paneque et al. 2014; Tan and Fitzgerald 2014).

For polycystic kidney disease we would need to obviously have supplemental education pamphlets you can hand out to people. Give them easy to read information that they can take home with them. [Nurse, Canada; Cox and Starzomski 2004, 147]

Other suggestions for effective communication to enhance patients' understanding of genetic risk included the acknowledgement of specific cultural beliefs and practices (Graves et al. 2011), helping patients to express themselves (Mendes, Sousa, and Paneque 2013) and honest involvement of patients in the associated uncertainty (Kenen et al. 2011).

Theme 3: Consequences of genetic risk information

This theme describes HCPs concerns about the potential negative psychological and social consequences associated with genetic risk information, both for patients and for their families. These include the potential for anxiety, discrimination and stigmatization.

3a. Anxiety (T3; Q38–47)

The potential for knowledge of genetic risk to cause anxiety was a recurring theme. In some cases, the distress to patients caused HCPs to question whether doing the tests was worthwhile (Lobb et al. 2001; Cox and Starzomski 2004; Al-Habsi et al. 2008; Graves et al. 2011; Mendes, Paneque, and Sousa 2012; Walsh et al. 2012; Birmingham et al. 2013; Mendes, Sousa, and Paneque 2013; Tan and Fitzgerald 2014).

It's like why are we doing a test that raises anxiety and increases the cost of care? [Provider, USA; Birmingham et al. 2013, 8]

HCPs acknowledged that anxiety may be precipitated by awareness of the significance of genetic risk information for other family members.

People often get distressed when they realize that close relatives may also be at risk, especially younger descendants under 18 or in their twenties. Positive results in such

cases are the most dramatic. [Genetic HCP; Portugal; Mendes, Sousa, and Paneque 2013, 777]

The role of the HCP in the management of anxiety associated with genetic risk and the provision of psychosocial support to patients were frequently referred to (Bottorff et al. 2000; Elwyn, Iredale, and Gray 2002; Carroll et al. 2003; Cox and Starzomski 2004; Birmingham et al. 2013; Mendes, Sousa, and Paneque 2013; Aasen and Skolbekken 2014). The desire to protect patients from anxiety is balanced against the desire to equip patients with accurate knowledge to facilitate appropriate decision-making.

Sometimes we're actually going to increase people's anxiety... and sometimes we're going to alleviate anxiety, but I mean part of our role is to define those risks for them and help them understand what's going on ... to get more information. Getting the genetic information is part of your information-gathering process, and if you don't do that, then you're not using all the tools you have. [Family physician, Canada; Carroll et al. 2003, 49–50]

Some HCPs described feeling uncertain about the best approach to manage patients' anxiety and expressed a desire to protect people from fear. Some stated that specialist help was needed to counsel distressed patients (Mendes, Sousa, and Paneque 2013; Aasen and Skolbekken 2014).

We do [evaluate when or if psychosocial support is needed] ... somehow we try to manage by ourselves the difficult moments in the consultation but it is not enough because there is no psychologist in the service! [Genetics HCP, Portugal; Mendes, Sousa, and Paneque 2013, 776]

The need for multidisciplinary support in the delivery of genetic risk information was often acknowledged, with many HCPs feeling that they were not equipped with the necessary specialist skills to support their patients (Lobb et al. 2001; Carroll et al. 2003; Bottorff et al. 2005; Mendes, Paneque, and Sousa 2012; Mendes, Sousa, and Paneque 2013; Paneque et al. 2014).

I am not trained in counselling, and I rely on members of my team to fill that vital role. [Clinical geneticist, Australia; Lobb et al. 2001, 194]

In some instances, anxiety was referred to as a reason to engage in genetic testing (Lobb et al. 2001; Elwyn, Iredale, and Gray 2002; Al-Habsi et al. 2008; Aasen and Skolbekken 2014; Paneque et al. 2014).

Sometimes I refer because the patient is extremely anxious. [General practitioner, UK; Al-Habsi et al. 2008, 755]

Some HCPs mentioned the potential for anxiety to be relieved by information about genetic risk and described this as an empowering experience (Carroll et al. 2003; Cox and Starzomski 2004; Graves et al. 2011).

It empowers people to find out [their risk of a BRCA1/2 mutation] and alleviate fears. [Obstetrician/gynaecologist, USA; Graves et al. 2011, 678]

3b. Discrimination and stigmatization (T3; Q48–53)

The potential for genetic risk information to be used to discriminate against patients was a recurring concept (Pfeffer, Veach, and LeRoy 2003; Cox and Starzomski

2004; Al-Habsi et al. 2008; Graves et al. 2011; Birmingham et al. 2013; Komatsu and Yagasaki 2014; Tan and Fitzgerald 2014).

I think the potential for discrimination down the road may be substantial. [Genetic counsellor, USA; Pfeffer, Veach, and LeRoy 2003, 426]

Whilst healthcare providers showed awareness and concern in relation to the potential for discrimination, only rarely was this associated with discussion of their own role in the management of this issue.

If genetic information is included in a patient's electronic medical record, we are concerned over potential information leakage. [Breast specialist, Japan; Komatsu and Yagasaki 2014, 42]

HCPs also referred to the potential for patients to feel stigmatized or labelled as a result of receiving information about their genetic risk status (Cox and Starzomski 2004; Graves et al. 2011; Mendes, Paneque, and Sousa 2012).

African American patients oftentimes tend to be more suspicious about [genetic testing] ... because they're so used to being pointed out all the time. And maybe they feel it's another way of [labeling them]. [Medical oncologist, USA; Graves et al. 2011, 680]

These statements show an awareness that having an inherited predisposition for a disease can have an important and potentially negative impact on the identity of individuals and families, and how they are perceived by society. In this sense, being 'at risk' of a disease has the potential to separate the individual and their family from others, and to change the way that others relate to them.

3c. *Impact on family (T3; Q54–58)*

The fact that genetic risk information impacts on the family, and not just the individual patient was mentioned across studies (Lobb et al. 2001; Carroll et al. 2003; Miller, Giacomini, and Ahern 2008; Graves et al. 2011; Harvey 2011; Walsh et al. 2012; Mendes, Sousa, and Paneque 2013; Komatsu and Yagasaki 2014; Paneque et al. 2014; Tan and Fitzgerald 2014).

Within genetics we are very aware of the family issues of genetic testing and we often feel that, you know, it can be described as being unethical to do genetic tests without consideration of the impact on other members of the family. [Genetics specialist, UK; Harvey 2011, 314]

The need for HCPs to be aware of this wider impact of genetic information, and to respond appropriately, was described as challenging for traditional and existing models of service provision.

Many of them [oncologists] are still stuck in the mode of treating the diseased person, not the family. The vast majority are still in that mode, because that is how they've been trained. In order to treat the person with the disease, they don't really deal with the whole family who is worried about having the disease. [Clinical geneticist, Canada; Miller, Giacomini, and Ahern 2008, 155]

HCPs acknowledged that individuals may engage with genetic testing for the sake of their family (Carroll et al. 2003; Will, Armstrong, and Marteau 2010; Graves et al. 2011; Tan and Fitzgerald 2014), or may decide that they do not wish to share their genetic risk status with others (Carroll et al. 2003; Graves et al. 2011; Mendes,

Sousa, and Paneque 2013; Komatsu and Yagasaki 2014), or they may have difficulty accessing or communicating about disease risk with some family members (Mendes, Sousa, and Paneque 2013).

We feel sometimes is complicated for people to get in touch with them [potentially at-risk relatives] and tell them [they might be at-risk]. Sometimes conflicts prevent this happening, or people don't see each other often, or because they fear a bad reaction. [Genetics HCP, Portugal; Mendes, Sousa, and Paneque 2013, 777]

Although HCPs recognized the need to 'treat the family' rather than the individual in this context, the familial transmission of information about genetic risk is outside the sphere of influence of the professional, and dependent on the views of individual patients, and their autonomous choice to share, or not to share, potentially distressing, sensitive or private information. This may be particularly problematic when risk information has preventive or reproductive implications for relatives. Providing professional support and intervention for all who may be affected by genetic information may not be possible for current methods of service provision.

Theme 4: Responsibility for genetic risk information

This theme includes statements where information about genetic risk was associated with a sense of responsibility.¹ These include feelings of guilt on the part of those found to be at risk of the disease, and thus having potential to pass on this increased risk to their offspring; responsibility to share information about disease risk with others who may be affected; responsibility to take preventive action to reduce risk; and HCPs' responsibility to treat and support patients.

4a. Genetic risk as a burden (T3; Q59–62)

Genetic risk was frequently referred to as a burden (Rees et al. 2006; Graves et al. 2011; Kenen et al. 2011; Mendes, Paneque, and Sousa 2012; Aasen and Skolbekken 2014; Paneque et al. 2014) or associated with feelings of guilt (Lobb et al. 2001; Carroll et al. 2003; Cox and Starzomski 2004; Will, Armstrong, and Marteau 2010; Graves et al. 2011). HCPs described patients feeling guilty for transmitting a heritable disease, or for being unaffected by a disease when other members of the family were affected.

If the mother has the gene, she feels like she might have tainted her offspring. If a sister doesn't have the gene and her sister does, then the sister that doesn't have it might feel guilty. [Breast surgeon, USA; Graves et al. 2011, 681]

Genetic risk information was also described as potentially relieving responsibility by reducing a patient's risk status (Graves et al. 2011), or by accounting for disease status and detracting from responsibility for lifestyle choices (Will, Armstrong, and Marteau 2010).

For a woman who has a strong family history she may learn that she doesn't carry a familial genetic risk factor so she may be relieved from having to make various medical decisions. Sometimes gaining additional information about risk can be reassuring even if people learn that they're at increased risk because at least they have information. [Genetic Counsellor, USA; Graves et al. 2011, 681]

[I could tell them that their cholesterol was] different from Joe Public's cholesterol which may well be raised because of a bad diet or something else. [Consultant physician, UK; Will, Armstrong, and Marteau 2010, 914]

4b. Responsibility to act on risk information (T3; Q63–69)

Genetic risk was associated with a responsibility to decide whether or not to share risk information with other family members who may also be affected (Carroll et al. 2003; Graves et al. 2011; Komatsu and Yagasaki 2014).

In some situations, [it's] a disadvantage that the first person to be tested in the family gets the job of having to spread the information to everyone and that can be somewhat of a burden for people. [Genetic counsellor, USA; Graves et al. 2011, 682]

It is up to the patient to decide whether to inform her relatives of the results of the genetic test. [Breast specialist, Japan; Komatsu and Yagasaki 2014, 42]

Genetic risk was also associated with a responsibility to respond in other ways, such as to make lifestyle changes (Rees et al. 2006), or to decide whether or not to have children (Cox and Starzowski 2004) or whether or not to undergo prophylactic surgery (Graves et al. 2011; Kenen et al. 2011).

Because potentially you could really be, you know, sort of adding to somebody's burden by raising those issues if, well one, they hadn't thought about it and then they were raised and they thought, 'Oh my goodness I really need to do something about this' or, 'I haven't been doing this and this, you know, it's going to contribute to my disease risk' and all of that sort of thing. [Genetic counsellor, Australia; Rees et al. 2006, 101]

Responsibility was also frequently referred to with reference to the need for HCPs to provide services and support following genetic testing (Elwyn, Iredale, and Gray 2002; Cox and Starzowski 2004; Miller, Giacomini, and Ahern 2008; Graves et al. 2011; Komatsu and Yagasaki 2014; Paneque et al. 2014).

It's a two-way-street. It's our responsibility to make sure that families know that nephrology is there in terms of long-term management and care issues [Geneticist, Canada; Cox and Starzowski 2004, 153]

4c. Non-directive approach to genetic risk (T3; Q70–79)

HCPs frequently attributed ultimate responsibility to interpret or respond to genetic risk to their patients (Lobb et al. 2001; Pfeffer, Veach, and LeRoy 2003; Cox and Starzowski 2004; Kenen et al. 2011; Mendes, Sousa, and Paneque 2013; Overby et al. 2013; Tan and Fitzgerald 2014) or to other professionals (Kenen et al. 2011; Birmingham et al. 2013), rather than themselves.

I think as geneticists we can say, you know, these are the percentages, we don't know ... and it's your decision in the end, so I think in some ways we're not taking such a responsibility. I'm sure that surgeons must feel more uneasy than the geneticists. [Oncologist, UK; Kenen et al. 2011, 9]

The consensual approach was described as being non-directive, with the role of the professional being to provide information and support to empower the patient make informed decisions.

My job is I guess to provide the information and then the patient can decide what they want. [Gynaecological oncologist, Australia; Tan and Fitzgerald 2014, 95]

Two articles reported that although the majority of HCPs preferred a non-directive approach, some would use a directive manner regarding the adoption of health protective behaviours (Rees et al. 2006), and the recommendation of prophylactic surgery (Kenen et al. 2011).

I certainly talk about smoking if I've ascertained earlier on that the woman is a smoker. I have to admit I do discuss that, you know, in relative detail that they should try and give up or cut it down as much as they can. [Medical specialist, Australia; Rees et al. 2006, 99]

The non-directive approach adopted by the majority of HCPs in this context is reflected by the fact that the delivery of genetic risk information was often framed in terms of patient empowerment, control or autonomy (Lobb et al. 2001; Cox and Starzomski 2004; Rees et al. 2006; Graves et al. 2011; Kenen et al. 2011; Mendes, Sousa, and Paneque 2013; Paneque et al. 2014).

I think also empowering them to a degree that they can alter their risk and that they are not a passive player in the whole process, that, you know, that they can actually impact on reducing their potential risk of developing breast cancer. So I think from both those perspectives for many women it's important. [Medical specialist, Australia; Rees et al. 2006, 101]

Discussion

Previous reviews (Emery et al. 1999; Suther and Goodson 2003; Mikat-Stevens, Larson, and Tarini 2015) have identified that HCPs perceive there to be numerous barriers to the integration of genetic testing and genetic risk communication into routine clinical practice. The themes developed from this synthesis of the qualitative literature, with a specific focus on perceptions of genetic risk, help to contextualize the persistence of these barriers.

The perceived value of predictive genetic testing was often low with little to add to existing practice. HCPs were concerned about the rapid introduction of new genetic technologies without the necessary evidence base to fully inform the use of these approaches in clinical settings. Patients' understanding of genetic risk was perceived to be limited, and subject to bias and external influence. The professional's role is conceptualized as an authoritative process of evaluation to untangle these multiple influences in order to assess patients' capabilities and needs and to tailor their approach accordingly.

Concern about the potential psychological and social impact of genetic risk was a recurring theme here, and has been identified as an important barrier to the mainstream integration of medical genetics in previous reviews (Mikat-Stevens, Larson, and Tarini 2015). The need to provide comprehensive and often multidisciplinary support to patients to help them deal with the psychological distress and other negative consequences that may be associated with predictive genetic testing may be challenging when resources are stretched. HCPs were aware that the impact of genetic risk information permeates beyond the individual to families and communities with ever fluctuating needs and perspectives, and involving social processes beyond the current scope of influence of HCPs. Traditional professional–patient

interactions involve the treatment of an individual, at a particular time. The findings of this metasynthesis reflect a growing awareness of the limitations of this approach in the context of predictive genetic testing. This contextualization is challenging for traditional models of service delivery, but is essential in order to understand patients' acceptance of, understanding of and responses to genetic risk information, and has important implications for equality of access to expanding genetic services.

The concept of responsibility was clearly associated with genetic risk information. Whilst HCPs accepted responsibility to provide support and care for patients undergoing predictive genetic testing, responsibility for the resulting information about risk status and responsibility to act appropriately on the basis of this information were mostly attributed to patients, rather than providers. In this context, HCPs described their role in non-directive terms, as empowering patients to make informed decisions. However, responsibility for genetic risk for patients is associated with moral obligation towards others that may conflict with their own needs and interests (Hallowell 1999; Foster et al. 2004). A non-paternalistic, non-directive stance may have an important absolving function in this context (Salmon and Hall 2003; Schicktanz and Schweda 2012). Ideals of patient autonomy and empowerment are widely advocated in health policy, and are a valued healthcare outcome (McAllister et al. 2012), but may also be interpreted here as an appropriate response to the management of uncertainty (Kenen et al. 2011) and to the challenges of integrating genetic knowledge and technology into clinical practice (Schicktanz 2016). The construction of the HCPs role as non-directive in this context is at odds with more paternalistic notions of protecting patients from distress, and of having authority to make judgements about who can, and who cannot understand and make appropriate use of genetic information, that were identified in other themes.

Limitations

The qualitative studies reviewed here represent the views of HCPs in the USA, Canada, Europe, Australia and Japan, and may not be representative of other cultural perspectives. There are likely to be important cultural differences in responses to genetic testing (Raz and Schicktanz 2009a, 2009b). The manner in which these differences impact on HCPs' perspectives and practice is an area for future research.

The majority of the studies included in this metasynthesis explored HCPs' perceptions of predictive genetic testing for hereditary cancers. The results may therefore reflect issues that are specific to this disease context. A small number of studies included related to monogenic, autosomal dominant disorders (e.g. polycystic kidney disease), where the outcome of predictive genetic testing would be binary, and likely to be appraised differently than probabilistic estimates of risk for multifactorial conditions. The studies included reflect the conditions (mostly inherited cancers) for which predictive genetic testing is already incorporated into clinical practice. It is likely that genetic medicine will be applied in this way to a much wider range of conditions as understanding of genetics continues to grow. Heterogeneity amongst perceptions of genetic risk relating to different diseases is likely, and further research is needed to explore variation in perceptions of predictive genetic testing and genetic risk appraisal between different hereditary disorders.

The review procedures used here focused on pre-symptomatic predictive testing, and excluded articles that explicitly related to post-symptomatic genetic testing to confirm diagnosis and/or inform treatment. However, given that the majority of

articles included in this review related to cancer genetics, it is possible that some of the extracts used may refer to HCPs' experiences of genetic testing to predict recurrence of hereditary cancers in those who have already been affected by the disease.

Conclusions

The perceived resource barriers to the integration of predictive genetic testing into mainstream clinical practice that have been identified in both early and recent reviews may be symptomatic of challenging social and ethical considerations associated with genetic risk which are not well aligned with current practice. The findings of this metasynthesis contribute an in-depth analysis and integration of the qualitative research literature, and help to contextualize the persistence of these barriers, as they are perceived by health care providers.

Acknowledgements

The authors are grateful to all of the 'Mind the Risk' partners for their helpful contributions to this manuscript.

Disclosure statement

No potential conflict of interest was reported by the authors.

Funding

This work was supported by the Swedish Foundation for Humanities and Social Sciences [grant number M13-0260:1'Mind the Risk'] and the FP7 HEALTH programme [grant agreement FP7-HEALTH-F2-2012-305549 'Euro-TEAM'].

Note

1. For a philosophical account of genetic risk information and responsibility, see the contribution by Silke Schicktanz in this issue of the *Journal of Risk Research*.

References

- Aasen, T., and J. A. Skolbekken. 2014. "Preparing for and Communicating Uncertainty in Cancer Genetic Counselling Sessions in Norway: An Interpretative Phenomenological Analysis." *Health, Risk & Society* 16 (4): 370–389. doi:10.1080/13698575.2014.927838.
- Agurs-Collins, T., R. Ferrer, A. Ottenbacher, E. A. Waters, M. E. O'Connell, and J. G. Hamilton. 2015. "Public Awareness of Direct-to-Consumer Genetic Tests: Findings from the 2013 U.S. Health Information National Trends Survey." *Journal of Cancer Education* 30 (4): 799–807. doi: 10.1007/s13187-014-0784-x.
- Al-Habsi, H., J. N. W. Lim, C. E. Chu, and J. Hewison. 2008. "Factors Influencing the Referrals in Primary Care of Asymptomatic Patients with a Family History of Cancer." *Genetics in Medicine* 10 (10): 751–757. doi:10.1097/GIM.0b013e318185212a.
- Birmingham, W. C., N. Agarwal, W. Kohlmann, L. G. Aspinwall, M. Wang, J. Bishoff, C. Dechet, and A. Y. Kinney. 2013. "Patient and Provider Attitudes toward Genomic Testing for Prostate Cancer Susceptibility: A Mixed Method Study." *BMC Health Services Research* 13: 279. doi:10.1186/1472-6963-13-279.

- Bottorff, J. L., L. G. Balneaves, J. Buxton, P. A. Ratner, M. McCullum, K. Chalmers, and T. Hack. 2000. "Falling through the Cracks. Women's Experiences of Ineligibility for Genetic Testing for Risk of Breast Cancer." *Canadian Family Physician* 46: 1449–1456.
- Bottorff, J. L., M. McCullum, L. G. Balneaves, M. J. Eshen, J. Carroll, M. Kelly, and S. Kieffer. 2005. "Establishing Roles in Genetic Nursing: Interviews with Canadian Nurses." *Canadian Journal of Nursing Research* 37 (4): 96–115.
- Braun, V., and V. Clarke. 2006. "Using Thematic Analysis in Psychology." *Qualitative Research in Psychology* 3: 77–101. doi:10.1191/1478088706qp063oa.
- Carroll, J. C., J. B. Brown, S. Blaine, G. Glendon, P. Pugh, and W. Medved. 2003. "Genetic Susceptibility to Cancer – Family Physicians' Experience." *Canadian Family Physician* 49: 45–52.
- Carroll, C., A. Booth, and M. Lloyd-Jones. 2012. "Should We Exclude Inadequately Reported Studies from Qualitative Systematic Reviews? An Evaluation of Sensitivity Analyses in Two Case Study Reviews." *Qualitative Health Research* 22 (10): 1425–1434. doi:10.1177/1049732312452937.
- Collins, F. S., and V. A. McKusick. 2001. "Implications of the Human Genome Project for Medical Science." *JAMA* 285 (5): 540–544. doi:10.1001/jama.285.5.540.
- Cox, S. M., and R. C. Starzomski. 2004. "Genes and Geneticization? The Social Construction of Autosomal Dominant Polycystic Kidney Disease." *New Genetics and Society* 23 (2): 137–166. doi:10.1080/1463677042000237071.
- Dixon-Woods, M., A. Sutton, R. Shaw, T. Miller, J. Smith, B. Young, S. Bonas, A. Booth, and D. Jones. 2007. "Appraising Qualitative Research for Inclusion in Systematic Reviews: A Quantitative and Qualitative Comparison of Three Methods." *Journal of Health Services Research and Policy* 12 (1): 42–47. doi:10.1258/jhsrp.2007.000023.
- Downe, S. 2008. "Metasynthesis: A Guide to Knitting Smoke." *Evidence Based Midwifery* 6 (1): 4–8.
- Downe, S., L. Simpson, and K. Trafford. 2007. "Expert Intrapartum Maternity Care: A Meta-Synthesis." *Journal of Advanced Nursing* 57 (2): 127–140. doi:10.1111/j.1365-2648.2006.04079.x.
- Elwyn, G., R. Iredale, and J. Gray. 2002. "Reactions of GPs to a Triage-Controlled Referral System for Cancer Genetics." *Family Practice* 19 (1): 65–71. doi:10.1093/fampra/19.1.65.
- Emery, J., E. Watson, P. Rose, and A. Andermann. 1999. "A Systematic Review of the Literature Exploring the Role of Primary Care in Genetic Services." *Family Practice* 16 (4): 426–445. doi:10.1093/fampra/16.4.426.
- Feero, W. G., and E. D. Green. 2011. "Genomics Education for Health Care Professionals in the 21st Century." *Journal of the American Medical Association* 306 (9): 989–990. doi:10.1001/jama.2011.1245.
- Foster, C., R. Eeles, A. Ardern-Jones, C. Moynihan, and M. Watson. 2004. "Juggling Roles and Expectations: Dilemmas Faced by Women Talking to Relatives about Cancer and Genetic Testing." *Psychology & Health* 19 (4): 439–455. doi:10.1080/08870440410001684168.
- Frueh, F. W., H. T. Greely, R. C. Green, S. Hogarth, and S. Siegel. 2011. "The Future of Direct-to-Consumer Clinical Genetic Tests." *Nature Reviews Genetics* 12 (7): 511–515. doi:10.1038/nrg3026.
- Graves, K. D., J. Christopher, T. M. Harrison, B. N. Peshkin, C. Isaacs, and V. B. Sheppard. 2011. "Providers' Perceptions and Practices regarding BRCA1/2 Genetic Counseling and Testing in African American Women." *Journal of Genetic Counseling* 20 (6): 674–689. doi:10.1007/s10897-011-9396-3.
- Guttmacher, A. E., A. L. McGuire, B. Ponder, and K. Stefánsson. 2010. "Personalized Genomic Information: Preparing for the Future of Genetic Medicine." *Nature Reviews Genetics* 11: 161–165. doi:10.1038/nrg2735.
- Hallowell, N. 1999. "Doing the Right Thing: Genetic Risk and Responsibility." *Sociology of Health & Illness* 21: 597–621. doi:10.1111/1467-9566.00175.
- Harvey, A. 2011. "From Lab to Lifestyle: Translating Genomics into Healthcare Practices." *New Genetics and Society* 30 (4): 309–327. doi:10.1080/14636778.2011.592005.

- Iredale, R., L. Jones, J. Gray, and J. Deaville. 2005. "The Edge Effect": An Exploratory Study of Some Factors Affecting Referrals to Cancer Genetic Services in Rural Wales." *Health & Place* 11 (3): 197–204. doi:10.1016/j.healthplace.2004.06.005.
- Iredale, R., G. Elwyn, A. Edwards, and J. Gray. 2007. "Attitudes of Genetic Clinicians in Wales to the Future Development of Cancer Genetics Services." *Journal of Evaluation in Clinical Practice* 13 (1): 86–89. doi:10.1111/j.1365-2753.2006.00657.x.
- Kalokairinou, L., H. C. Howard, and P. Borry. 2014. "Changes on the Horizon for Consumer Genomics in the EU." *Science* 346 (6207): 296–298. doi:10.1126/science.1256396.
- Kenen, R., A. Ardern-Jones, E. Lynch, and R. Eeles. 2011. "Ownership of Uncertainty: Healthcare Professionals Counseling and Treating Women from Hereditary Breast and Ovarian Cancer Families Who Receive an Inconclusive BRCA1/2 Genetic Test Result." *Genetic Testing and Molecular Biomarkers* 15 (4): 243–250. doi:10.1089/gtmb.2010.0071.
- Komatsu, H., and K. Yagasaki. 2014. "Are We Ready for Personalized Cancer Risk Management? The View from Breast-Care Providers." *International Journal of Nursing Practice* 20 (1): 39–45. doi:10.1111/ijn.12115.
- Lautenbach, D. M., K. D. Christensen, J. A. Sparks, and Robert C. Green. 2013. "Communicating Genetic Risk Information for Common Disorders in the Era of Genomic Medicine." *Annual Review of Genomics and Human Genetics* 14 (1): 491–513. doi:10.1146/annurev-genom-092010-110722.
- Lobb, E., P. Butow, B. Meiser, K. Tucker, and A. Barratt. 2001. "How Do Geneticists and Genetic Counselors Counsel Women from High-Risk Breast Cancer Families?" *Journal of Genetic Counseling* 10 (2): 185–199. doi:10.1023/A:1009447932274.
- Martin, G., G. Currie, and R. Finn. 2009. "Bringing Genetics into Primary Care: Findings from a National Evaluation of Pilots in England." *Journal of Health Services Research & Policy* 14 (4): 204–211. doi:10.1258/jhsrp.2009.008158.
- McAllister, M., G. Dunn, K. Payne, L. Davies, and C. Todd. 2012. "Patient Empowerment: The Need to Consider It as a Measurable Patient-reported Outcome for Chronic Conditions." *BMC Health Services Research* 12: 157. doi:10.1186/1472-6963-12-157.
- McGuire, A. L., C. M. Diaz, T. Wang, and S. G. Hilsenbeck. 2009. "Social Networkers' Attitudes toward Direct-to-consumer Personal Genome Testing." *The American Journal of Bioethics* 9 (6–7): 3–10. doi:10.1080/15265160902928209.
- Mendes, Álvaro, M. Paneque, and L. Sousa. 2012. "Are Family-oriented Interventions in Portuguese Genetics Services a Remote Possibility? Professionals' Views on a Multifamily Intervention for Cancer Susceptibility Families." *Journal of Community Genetics* 3 (4): 311–318. doi:10.1007/s12687-012-0079-z.
- Mendes, A., L. Sousa, and M. Paneque. 2013. "From Constraints to Opportunities? Provision of Psychosocial Support in Portuguese Oncogenetic Counseling Services." *Journal of Genetic Counseling* 22 (6): 771–783. doi:10.1007/s10897-013-9612-4.
- Mikat-Stevens, N. A., I. A. Larson, and B. A. Tarini. 2015. "Primary-Care Providers' Perceived Barriers to Integration of Genetics Services: A Systematic Review of the Literature." *Genetics in Medicine* 17 (3): 169–176. doi:10.1038/gim.
- Miller, F. A., M. Giacomini, and C. Ahern. 2008. "Contending Visions in the Evolution of Genetic Medicine: The Case of Cancer Genetic Services in Ontario, Canada." *Social Science & Medicine* 67 (1): 152–160. doi:10.1016/j.socscimed.2008.03.035.
- Noblit, G. W., and R. D. Hare. 1988. *Meta-Ethnography: Synthesizing Qualitative Studies*. London: Sage.
- Overby, C. L., W. K. Chung, G. Hripcsak, and R. Kukafka. 2013. "Cancer Genetic Counselor Information Needs for Risk Communication: A Qualitative Evaluation of Interview Transcripts." *Journal of Personalized Medicine* 3 (3): 238–250. doi:10.3390/jpm3030238.
- Paneque, M., Á. Mendes, L. Guimarães, J. Sequeiros, and H. Skirton. 2014. "Genetics Health Professionals' Views on Quality of Genetic Counseling Service Provision for Presymptomatic Testing in Late-Onset Neurological Diseases in Portugal: Core Components, Specific Challenges and the Need for Assessment Tools." *Journal of Genetic Counselling* 24 (4): 616–625. doi: 10.1007/s10897-014-9784-6.
- Pfeffer, N. L., P. M. Veach, and B. S. LeRoy. 2003. "An Investigation of Genetic Counselors' Discussions of Genetic Discrimination with Cancer Risk Patients." *Journal of Genetic Counseling* 12 (5): 419–438. doi:10.1023/A:1025816820243.

- Raz, A. E., and S. Schicktanz. 2009a. "Diversity and Uniformity in Genetic Responsibility: Moral Attitudes of Patients, Relatives and Lay People in Germany and Israel." *Medicine Health Care and Philosophy* 12 (4): 433–442. doi:10.1007/s11019-009-9215-x.
- Raz, A. E., and S. Schicktanz. 2009b. "Lay Perceptions of Genetic Testing in Germany and Israel: The Interplay of National Culture and Individual Experience." *New Genetics and Society* 28 (4): 401–414. doi:10.1080/14636770903314533.
- Rees, G., M. Young, C. Gaff, and P. R. Martin. 2006. "A Qualitative Study of Health Professionals' Views regarding Provision of Information about Health-Protective Behaviors during Genetic Consultation for Breast Cancer." *Journal of Genetic Counseling* 15 (2): 95–104. doi:10.1007/s10897-005-9009-0.
- Salmon, P., and G. M. Hall. 2003. "Patient Empowerment and Control: A Psychological Discourse in the Service of Medicine." *Social Science and Medicine* 57 (10): 1969–1980. doi:10.1016/S0277-9536(03)00063-7.
- Scheuner, M. T., P. Sieverding, and P. G. Shekelle. 2008. "Delivery of Genomic Medicine for Common Chronic Adult Diseases." *JAMA* 299 (11): 1320–1334. doi:10.1001/jama.299.11.1320.
- Schicktanz, S. 2016. "Genetic Risk and Responsibility: Reflections on a Complex Relationship". *Journal of Risk Research* Manuscript under review.
- Schicktanz, S., and M. Schweda. 2012. "The Diversity of Responsibility: The Value of Explanation and Pluralization." *Medicine Studies* 3: 131–145. doi:10.1007/s12376-011-0070-8.
- Sparks, J. A., M. D. Iversen, R. M. Kroouze, T. G. Mahmoud, N. A. Triedman, S. S. Kalia, M. L. Atkinson, et al. 2014. "Personalized Risk Estimator for Rheumatoid Arthritis (PRE-RA) Family Study: Rationale and Design for a Randomized Controlled Trial Evaluating Rheumatoid Arthritis Risk Education to First-Degree Relatives." *Contemporary Clinical Trials* 39 (1): 145–157. doi:10.1016/j.cct.2014.08.007.
- Stack, R. J., K. Shaw, C. D. Mallen, S. Herron-Marx, R. Horne, and K. Raza. 2012. "Delays in Help Seeking at the Onset of the Symptoms of Rheumatoid Arthritis: A Systematic Synthesis of Qualitative Literature." *Annals of the Rheumatic Diseases* 71 (4): 493–497. doi:10.1136/ard.2011.155416.
- Stack, R. J., M. Sahni, C. D. Mallen, and K. Raza. 2013. "Symptom Complexes at the Earliest Phases of Rheumatoid Arthritis: A Synthesis of the Qualitative Literature." *Arthritis Care and Research* 65 (12): 1916–1926. doi:10.1002/acr.22097.
- Stermer, T., S. Hodgson, F. Kavalier, S. Watts, and R. Jones. 2004. "Patients' and Professionals' Opinions of Services for People at an Increased Risk of Colorectal Cancer: An Exploratory Qualitative Study." *Familial Cancer* 3 (1): 49–53. doi:10.1023/B:FAME.0000026813.73472.9d.
- Su, P. 2013. "Direct-to-Consumer Genetic Testing: A Comprehensive View." *The Yale Journal of Biology and Medicine* 86 (3): 359–365.
- Suther, S., and P. Goodson. 2003. "Barriers to the Provision of Genetic Services by Primary Care Physicians: A Systematic Review of the Literature." *Genetics in Medicine* 5 (2): 70–76. doi:10.1097/01.GIM.0000055201.16487.61.
- Tambor, E. S., B. S. Bernhardt, J. Rodgers, N. A. Holtzman, and G. Geller. 2002. "Mapping the Human Genome: An Assessment of Media Coverage and Public Reaction." *Genetics in Medicine* 4: 31–36. doi:10.1097/00125817-200201000-00006.
- Tan, Y. Y., and L. J. Fitzgerald. 2014. "Barriers and Motivators for Referral of Patients with Suspected Lynch Syndrome to Cancer Genetic Services: A Qualitative Study." *Journal of Personalized Medicine* 4 (1): 20–34. doi:10.3390/jpm4010020.
- Vassy, J. L., R. C. Green, and L. S. Lehmann. 2013. "Genomic Medicine in Primary Care: Barriers and Assets." *Postgraduate Medical Journal* 89 (1057): 615–616. doi:10.1136/postgradmedj-2013-132093.
- Walsh, D., and S. Downe. 2005. "Meta-Synthesis Method for Qualitative Research: A Literature Review." *Journal of Advanced Nursing* 50 (2): 204–211. doi:10.1111/j.1365-2648.2005.03380.x.
- Walsh, J., M. Arora, C. Hosenfeld, U. Ladabaum, M. Kuppermann, and S. J. Knight. 2012. "Preferences for Genetic Testing to Identify Hereditary Colorectal Cancer: Perspectives of High-Risk Patients, Community Members, and Clinicians." *Journal of Cancer Education* 27 (1): 112–119. doi:10.1007/s13187-011-0286-z.

- Walter, F. M., J. Emery, et al. 2004. "Lay Understanding of Familial Risk of Common Chronic Diseases: A Systematic Review and Synthesis of Qualitative Research." *The Annals of Family Medicine* 2 (6): 583–594. doi:[10.1370/afm.242](https://doi.org/10.1370/afm.242).
- Will, C. M., D. Armstrong, and T. M. Marteau. 2010. "Genetic Unexceptionalism: Clinician Accounts of Genetic Testing for Familial Hypercholesterolaemia." *Social Science & Medicine* 71 (5): 910–917. doi:[10.1016/j.socscimed.2010.05.018](https://doi.org/10.1016/j.socscimed.2010.05.018).
- Wilson, B. J., and S. G. Nicholls. 2015. "The Human Genome Project, and Recent Advances in Personalized Genomics." *Risk Management and Healthcare Policy* 8: 9–20. doi:[10.2147/RMHP.S58728](https://doi.org/10.2147/RMHP.S58728).
- Yamanaka, H., and K. Takeda. 2011. "Practice of Personalized Primary Prevention of Lifestyle Diseases: Associated Problems and Issues in Japan." *Personalized Medicine* 8 (2): 215–224. doi:[10.2217/pme.10.85](https://doi.org/10.2217/pme.10.85).