The psychosocial impact of family history screening in Australian primary care: A qualitative evaluation

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ABSTRACT

Whilst the family history is perceived as a routine part of the medical family history it is not used in a systematic way to tailor disease prevention in primary care. Family history questionnaires (FHQs) may have an important role in primary care as a screening tool to support tailored disease prevention. The potential harms and benefits of family history screening in primary care require investigation before routine adoption.

This study aimed: first to explore the experience and impact of family history collection via a novel family history questionnaire and subsequent familial risk assessment, and secondly, to assess the acceptability and feasibility of using the questionnaire in Australian primary care.

Twenty-eight semi-structured telephone interviews were conducted with patients already enrolled in a family history screening study through their family physician. Qualitative constant comparative analysis was undertaken of transcript data.

Common themes included the way in which the family unit, individual stage of life and a number of external triggers interact and contribute to how an individual comes to terms with familial disease risk. Unique findings emerged relating to the Australian perspective of participants. Living in Australia created a barrier to effective communication amongst family members about family health, and family history collection. In addition to the vast geographical distance both within Australia, and between Australia and other countries, there was an additional sense of isolation described within an historical context. The family history screening questionnaire was considered user-friendly and a worthwhile approach to

supporting disease prevention in primary care, although some participants did not retain an accurate understanding of their familial cancer risk.

In conclusion, a person's response to family history screening is reliant on a complex interplay of family, personal and external factors, which in turn are driven by their stage of life. The impact of immigration and geographic isolation from family members may further complicate a person's response to undertaking family history screening.

Keywords: Chronic disease, family history screening, psychosocial impact

INTRODUCTION

Family history is a non-modifiable risk factor for many common complex genetic conditions. Risk of ischaemic heart disease (Williams 2001), type 2 diabetes (Harrison 2003), melanoma (Gandini 2005), breast, ovarian, colorectal and prostate cancers (Collaborative Group on Hormonal Factors in Breast Cancer 2001; Pharoah 2002; Johns 2001; Lesko 1996;) are all associated with family history. Family history of a common chronic disease is associated with a 2-5 fold relative risk of developing the condition, and increases with the number of affected relatives and early age of onset (Yoon 2002). Lifestyle factors such as diet, exercise and smoking status have been shown to act synergistically with familial disease risks (Ruffin 2011). Modifiable risk factors are therefore the target of preventive interventions for those identified at increased risk.

The family physician plays a significant role in providing tailored, preventive lifestyle advice and support for behavioural change, particularly directed at high risk groups identified through family history risk assessment (Emery 2010). For those identified at increased risk, there are specific evidence-based preventive interventions available, above and beyond those offered to the general population. This often includes referral to a clinical genetics clinic for genetic counselling. The precise role of screening for familial disease in motivating change in health behaviours requires exploration as there is currently insufficient evidence to show that family history assessment in primary care improves patient outcomes (Berg 2009; Wilson 2009).

Whilst the family history is perceived as a routine part of the medical family history it is not used in a systematic way to tailor disease prevention in primary care (National Institutes of Health, 2009). Family history questionnaires (FHQs) may have an important role in primary care as a screening tool to support tailored disease prevention.

The effect of participation in a family history screening programme and exploration of beliefs about familial disease could potentially have a positive effect on motivation for preventive self-management (e.g. additional surveillance, or adopting healthy lifestyle choices). Conversely, it is possible that identifying individuals at increased risk of disease may have adverse consequences including effects on psychological coping, family relationships, employment and insurance.

Our findings relate specifically to the Australian context. In the United States there have been several major initiatives to promote awareness of the importance of the family medical history amongst the community and with healthcare professionals (U.S. Surgeon General's Family History Initiative, Genetic Alliance Family Health History, The Centers for Disease Control and Prevention). Within these initiatives routine screening in primary care for a family history of common disease has been promoted but, as with any formal screening program, the potential harms and benefits of family history screening require investigation. As part of a study funded by the National Health and Medical Research Council (NHMRC) that aimed to validate a novel FHQ for use in primary care, a qualitative sub-study was conducted to assess the impact and experience of undergoing family history screening in primary care. The study was part of a collaborative series of studies undertaken between the Discipline of General Practice at The University of Western Australia and the Department of Public Health and Primary Care's Primary Care Unit at the University of Cambridge in the United Kingdom, who have undertaken similar studies (Walter 2013; Birt 2014).

A brief, plain-language FHQ was developed to be applied to the adult primary care population. It was validated such that its performance both identified those at increased risk who required more detailed risk assessment, as well as providing reassurance to those patients at population risk (Emery et al, 2013). As part of the evaluation of the FHQ prior to recommending its routine use a routine in primary care, this sub-study aimed to both

explore the experience and impact of family history screening, using our novel FHQ, and to assess its feasibility and acceptability for use in the Australian primary care setting.

METHODS

Study Background and Design

Informed by a systematic review of family history questionnaires developed for use in the clinical setting (Reid 2009), a 15-item FHQ was developed and evaluated for use in the primary care setting (Emery 2013). Five hundred twenty-six patients, aged 20-50, were recruited from family physician practices in metropolitan and outer metropolitan Perth, Western Australia (response rate 5.6%) to validate the FHQ. Participants completed the FHQ and had their three generation pedigrees drawn by a trained genetic counsellor. The three generation pedigree was the reference standard family history and allowed risk assessment to be performed for heart disease, diabetes, melanoma and breast, ovarian, colorectal and prostate cancers. Participants' risk status was determined as being either at increased risk or population risk for each condition based on Australian evidence-based risk assessment guidelines (The Royal Australian College of General Practitioners 2009). Participants were advised their level of risk and, if increased, suggested that they discuss this further with their general practitioner (GP).General practitioners were informed of their patients' risk assessment with advice on management of those at increased risk. A detailed account of the validation study has been reported elsewhere (Emery in press). A qualitative sub-study using semi-structured telephone interviews was conducted to assess the psychosocial

impact and experience of patients undergoing family history screening in the primary care setting.

Participants and Procedures

Semi-structured telephone interviews were conducted with 28 participants from the larger family history screening study in primary care (Emery 2013). The study was approved by the Human Research Ethics Committee, the University of Western Australia.

The qualitative component of the study commenced at the beginning of 2010 and continued until the end of recruitment into the overall study (December 2010). Within this time period, 263 participants were recruited into the main family history screening study and were consented to be approached about the interview study.

Participants who agreed to be approached were purposively sampled to reflect the larger study group's demographic and familial risk status. Sampling was conducted according to age, gender, ethnicity, familial disease and familial risk status. Interviews continued until data saturation was reached around key themes.

Telephone interviews were conducted by a research genetic counsellor. The duration of most telephone interviews was between 30 and 45 minutes. Interviews were digitally recorded and transcribed verbatim by a professional transcription service.

Instrumentation: Interview Guide

Interviews were informed by a topic guide based on previous work by Walter et al. (2005) which explored patients' understanding of their family history of common chronic diseases.. The topic guide was refined throughout the data collection period based on emerging

findings from the iterative analytic process, as is typical practice in some types of qualitative research (Pope 2006). Areas explored included:

- Initial reaction to being invited into a family history screening study;
- Exploration of any conditions that are "running in the family";
- The impact of family history screening on self, family, GP, wider community;
- Motivating factors in reducing disease risk;
- Family History Questionnaire: its perceived benefits, problems and impact.

Data Analyses

Qualitative data analyses were undertaken as an on-going iterative process. This commenced soon after the initial interviews, by applying a constant comparison technique (Lingard 2008) which allowed themes to emerge and concepts to develop; these were later mapped into a theoretical framework. Themes and hypotheses identified in early interviews were used to inform the areas of investigation in later interviews. All interviews were conducted by GTR, a trained genetic counsellor and researcher. FMW and JDE independently read all transcripts to confirm the integrity of the emerging themes and concepts.

QSR NVivo 8 was used to facilitate data coding and retrieval.

RESULTS

Response rate

Between January 2010 and December 2010, 263 people participated in the main family history study. Of these, 262 (99.6%) consented to be contacted for a follow-up telephone interview.

Purposive sampling resulted in twenty-eight participants undergoing the telephone interview. Table 1 shows the characteristics of the interview group, and compares their characteristics with the overall study group. It can be seen that the average age and gender ratio of both groups is very similar, as is the proportion of participants at increased risk of heart disease, diabetes, colorectal cancer, and melanoma. We over-sampled those at increased risk in order to optimise the richness of data. Participants with a significant family history of disease were considered likely to have stories to share that would provide insights into the lived experience of familial disease. Whilst those at population risk of all conditions were represented, early interviews with these participants revealed far fewer avenues to explore in terms of lived experience around family history of disease.

(Insert Table 1)

A number of major themes and sub-themes were identified in regard to the impact of family history screening. Prevalent themes included the way in which *feeling at risk, one's family unit,* and *individual stage of life* interact with one another to contribute to coming to terms with familial disease risk. Themes also emerged around the Australian perspective of participants. These core themes were identified as interacting with one another, and instrumental in terms of how a person comes to terms with familial disease risk. *Coming to terms with familial disease risk* is therefore presented as a subsidiary theme, which in turn encompasses themes relating to *coping and controlling* regarding familial disease risk, the participants' experiences and perceptions around the *FHQ* itself, and the *perceived impact of family history screening on the community.*

Feeling at risk

"Feeling at risk," describes the various factors that have an impact on how a person feels about their family history, and may contribute to their sense of risk or vulnerability. These

include “running in the family,” “lived experience,” “disease-specific factors,” and “external influences.”

There was a range of understanding of what “running in the family” meant to people: some felt that just having one relative affected by a condition was enough to raise alarm about personal risk, whilst others needed to see multiple members affected, spanning a number of generations, to feel increased vulnerability:

“Within your family it’s a pressure cooker situation. So it only really takes one close family member to be affected for it to be something that is on your radar” (male, 33, high risk colorectal cancer).

“I think it has to be a few people and a few generations to be something in the family. If it’s a once off it may be external circumstances that do it” (female, 50, high risk breast/ovarian cancer, increased risk diabetes).

One woman, who had recently found out that she had distant Indigenous Australian ancestry, described that in combination with her family history, this ethnic consideration compounded her sense of vulnerability:

“Even with the sugar diabetes and with the family history of the Indigenous, they’re prone to Type 2 diabetes so it’s already there anyway. It’s like you have to be really extra careful to watch out for that. So it does make you aware.... So it hits home when you look at the family history… it even makes you a bit more scared to think, you know, I need to really do something about it” (female, 47, increased risk diabetes).

It was common for participants to express that they felt different from affected family members in crucial ways, and consequently felt less at risk of developing the condition:

*"I know my mum's [*diabetes*] is purely due to her eating habits. My habits are different to mum's so I guess it's being conscious as I grow older of what I eat, and being wary of gaining too much weight" (female, 31, increased risk diabetes).*

Many participants described the "lived experience" of witnessing the diagnosis, disease journey, or death of a family member or even a friend, as affecting their sense of risk. A number of participants retold their childhood experiences of witnessing a parent's illness. For instance, one woman described the lengthy experience of watching her father through his cancer journey:

"We had a sort of, really long road. And there were times when he was really well, and there were times that he was really sick...So it was...kind of like a roller coaster ride…. Everytime there was a diagnosis he would, well usually he would have surgery immediately and then a series of radiotherapy and other treatments, and he'd sort of gradually get better from that, we'd sort of hope that it would be the last time. But um... it invariably came back" (female, 34, father and paternal aunt died of melanoma).

Due to family member's experiences with illness, some people tended to demonstrate heightened symptom awareness within themselves:

"Because of what happened to my sister I know I'm acutely aware of my body – more than I ever was before. I'm just so aware of that now and I'm conscious of that... If I did get a, you know, feel something that, just a niggle for example in where I assume that my ovaries are, initially I would think 'Oh, what's that?'" (female, 47, high risk breast/ovarian cancer, twin sister died ovarian cancer).

A person's sense of risk was also determined by the particular condition that was present in their family. Perceived severity, cause, and controllability of different diseases were all factors that tended to contribute to participants' sense of risk.

Participants tended to have pre-conceived notions of how "serious" each condition was. Cancer was viewed as the most "serious" illness compared to heart disease, which was in turn more serious than diabetes.

"But yeah, the cancer, I think that it's deadly. It could come from anywhere at any time um... it's kind of like an evil spirit. It can just weave its way into your life and, and take hold. Um, and I think to a certain extent it can be spontaneous... like it was with my, my father. It doesn't discriminate...And so many healthy, fitness fanatics are struck down... I mean you can only take so much prevention. .. But the rest is a bit of a lucky dip I think. <laugh>" (female, 37, increased risk melanoma).

Participants tended to demonstrate a wide range of views regarding heart disease in terms of prognosis and how it could be treated.

"I guess with heart condition obviously it's serious if you get it, but it's something... that you have a bit of lead up time towards, so you can be um working to avoid it, I mean there are better treatments and things for angina these days than there were when my father died" (female, 45, increased risk heart disease).

Whilst the dangers associated with diabetes were acknowledged by some participants, it was most often ranked as the least serious condition of the three:

"I mean, diabetes, it causes, well it can certainly create the situation where you could die – no dramas there. But the death there isn't as impending in a lot of ways as in some of the other conditions" (male, 34, increased risk heart disease).

Sometimes advertising campaigns reinforced people's sense of disease risk, building upon their existing beliefs about their family history. One woman described the impact of a particular public healthcare campaign around melanoma, as she was able to directly relate to its hard-hitting message:

"Around the time dad died we heard of other people who had it..., you know you see some of those cases on TV? Of the young surfing guy who died in his 30s... And you just – the more you hear about melanoma, it's just so aggressive... it seems that once it's metastasised, it's very rare that anyone actually beats it" (female, 34, father and paternal aunt died melanoma).

Celebrity diagnosis of illness was viewed differently to healthcare media campaigns and tended to have a greater impact on people's sense of vulnerability, interacting with their personal beliefs about their familial disease risk:

"There seems to be some families that do have a predisposition and it seems to increase your likelihood but then other people seem to get it just out of the blue... Do you remember – did you see on the news Martina Navratilova last week? She was just saying she was so fit and so healthy, and then she said she thought she'd never get it (cancer) and she did" (female, 42, increased risk heart disease and diabetes).

Family Unit

Participants' described that the success of family history screening was largely dependent upon their ability to access their family history information. Participants described their family unit in terms of its culture. Underpinning family culture were notions of ethnicity, emotional closeness and geographical distances between family members, and the impact that these could have on family health talk.

"So I don't think my mum even knew much about…. I mean she knew her sister had cancer, but she didn't know what type because it just wasn't really spoken about. And my parents came from a very conservative English stock as well <laughs>… which doesn't help! You know, the British!" (female, 33, high risk breast and ovarian cancer).

"I mean we're fairly similar in age. He's only a few years older than me. I know he's sort of had his lipids checked just to make sure he's OK, but other than that, you know, it's not the sort of thing we talk about every time we get together." (male, 34, increased risk heart disease).

Stage of life

A person's personal experiences, present situation and future plans had an impact on their understanding and interest in family history screening. People's priorities were different at each life stage. Participants described an awareness of increased age being associated with an increased risk of disease. Some participants also had an additional sense of anticipated onset, with a heightened awareness of vulnerability at the age that a diagnosis had occurred in a family member:

"So maybe it's a good point now that I stop and have a look at these things… now that I'm older I do need to assess and do maintenance" (female, 50, high risk breast and ovarian cancer, increased risk diabetes).

"So we're both coming to the 'danger zone' – the age where it (diabetes) seems to manifest itself for the others [in my family]" (male, 49, increased risk diabetes).

A number of participants were parents. The desire to be healthy for the sake of looking after their children was clear. Parents commonly expressed concerns about the implications of increased risk for future generations:

"I think before you have kids, you sort of think you're a bit immortal. And really my attitude was well… 'If it's my time, it's my time' – that's good." (female, 33, high risk breast cancer).

"If it was likely that I would develop breast cancer, I'd have a mastectomy, absolutely. Especially at my age now. I think it would be different if you found out when you were young, 20 or whatever, my body image was much more important in my 20s than it is now… now I have children, my sole goal is to stay fit and well and healthy for my kids" (female, 42, population risk).

Australian perspective

A number of participants felt that living in Australia could be isolating, and could create a barrier to discussions within the family about health and disease. Consequently, when being screened about their family history, it created greater uncertainty and concern because of their limited knowledge about the details of their family history. This was not only due to the unique aspects of geographical distance that Australian families face when family members live in different States or Territories within the country, but also when family members live in different countries. Furthermore, from an historical perspective, some participants described the impact of immigration to Australia, as well as emigration to other countries, as potential hindrances in terms of being able to trace their medical family history.

"I don't have much of a family history… my mother was adopted [born in England and adopted by an Australian family, immigrating as a baby] and my father came out from Austria when he was a young bloke and basically there wasn't a lot of family history there… I think when he separated from his family and came to Australia there was literally a… down the line separation and he never spoke about his family very much and we certainly haven't had any contact." (male, 34, increased risk heart disease).

"I've always felt that living away from family in the UK that… our family history might get a bit lost. When it comes to my children, they've got even less chance of knowing about their family history, just because of geography really." (female, 37, population risk for all conditions).

The effect of modern technology

A number of participants described that even though geographical distance can make it difficult to physically catch up with family members, communication and subsequently information is far more accessible these days compared to the pre-Internet years. This could therefore reduce the sense of isolation and uncertainty about their family history when being screened. A number of participants described the use of modern day communication technologies such as email, and social networking tools like *Facebook*, as facilitative in keeping informed about family history.

"I've got to say that they're in Europe, they're in Holland… You know, I have very, very little contact with them but you know we're on Facebook and stuff like that" (Male, 49 years, increased risk type 2 diabetes."

Despite this man's recognition that social networking has provided a conduit for family communication, there was a sense that the depth of connection is perhaps limited. He noted that whilst he was friends with his cousins in Europe, his family here in Australia were effectively estranged from his European family by virtue of living in Australia:

"So all my cousins in Europe, they may talk to each other, but they don't talk to me and my sisters because we're over here in Australia" (male, 49 years, increased risk type 2 diabetes).

Coming to terms with familial risk

Feeling at risk, family culture, stage of life and the Australian perspective were all themes that contributed to the way in which an individual came to terms with their familial disease risk. In response to familial disease risk assessment, a variety of coping and controlling

strategies were described by participants including lifestyle choices, disease screening and psychological adaptation.

"I would say diet, exercise and I guess just general well-being. You know, just being happy in ourselves and not… having too much stress in our life." (female, 34, increased risk melanoma).

"With [my] breast cancer [risk] all I can really do is my self-checks and have my annual checks with my GP. I mean, what else am I really going to do? If there is anything, make sure that I know that are changes and get onto them really early." (female, 41, population risk all diseases).

People did not seem to find cancer risk assessment very meaningful or memorable; indeed, some people did not appear to remember their disease risk information. In some cases of familial cancer, participants were assigned a "moderately increased" risk status. For these people, there was a sense of ambivalence attached to responses:

"I think I was within the slightly to moderately increased risk group. And I think for me in my mind, before I'd done this study, I was in the high risk. So I think for me… it maybe alleviated a little bit of that because it… brought me down a category. I guess in my mind… I had sort of just formulated that myself." (female, 31, moderate risk breast and ovarian cancer).

"Obviously low risk – that's good. But to be told you're moderate risk is really, well it wasn't anything that put any alarm bells in my head." (female, 33, high risk breast and ovarian cancer).

One woman failed to be reassured by being assigned "population risk" status for cancer. She described her sense of vulnerability, particularly towards less common disease that she had witnessed in a family member:

"I don't think it sort of eased any sort of fear of cancer. Even though I'm still in that average range, I can still get those rare forms of cancer or those cancers that aren't really genetically based… like my uncle has a brain tumour, and ovarian cancers…" (female, 31 , increased risk diabetes).

Feasibility and acceptability of the FHQ

Interviewees considered primary care the intuitive setting for family history screening to take place. It was acknowledged however, that the patient themselves should be aware of their own family history information, and work with their GP to address familial risk.

"I don't think that it's really the obligation of the GP, but think it's probably a positive thing if they do, 'cause many people just wouldn't do it otherwise… so for instance you might feel silly going to the doctor, but if the doctor says to you 'you know, you should have a check up once a year once you turn 50' or something then you probably would." (female, 45, increased risk heart disease).

All interviewees described the FHQ as brief and easy to complete. Some interviewees raised their difficulties in recall of family history information and most felt relieved to find out that the family history information required only included first and second degree relatives. Increased anxiety due to being "tested" and emotional upset given a recent family diagnosis or death were raised as potential negative side-effects, however, these were not directly experienced by any participants.

"The actual questionnaire is very easy to understand… it's quick, more importantly, 'cause most of us are time poor" (female, 42, population risk all diseases).

"*I found it really self-explanatory and easy to fill out and it was quite interesting because it gave me a few things to think about that I hadn't thought about before… But it was easy to fill out, very easy" (female, 41 years, population risk all diseases).*

"Maybe for someone who's had a dozen cancer deaths in their family, they might find it difficult to fill out… There will be some people where some issues are too fresh… that could be upsetting" (female, 41, increased risk melanoma).

"Some people find forms like a little bit of a test. You know, like, how's this going to be marked?" (female, 41, increased risk melanoma).

Overall, family history screening was considered an important aspect of primary care, with the potential to assist in disease prevention and even reduction in health care costs at a community-wide level. Many people felt that family history screening was an essential aspect of the holistic primary care that patients expect, yet often do not receive.

"I think it does make them (people) stop and think about it (family history)… And if they don't know, it makes them go away and investigate." (male, 49, increased risk prostate cancer).

"I just think it helps in the holistic approach to patient care." (male, 40, population risk all diseases).

A small number of potential concerns were raised about the FHQ. Some participants described the difficulty in recalling family history information or not having adequate knowledge around their family history:

"I guess with a blood test or say, a pap smear, you know, you do a test and it's either there or it's not. But with something that you're relying on your memory… there's always a chance I guess, of user error." (female, 41, increased risk melanoma, increased risk ovarian cancer).

Proposed Framework

Whilst the core themes of feeling at risk, family unit, stage of life and the Australian perspective represented distinct and unique elements of each person's life, they were considered to represent interacting aspects of a person's life that essentially feed into how they came to terms with their familial disease risk.

Walter's "Model of Familial Risk Perception" (Walter 2005) was used as a template for the development of a framework that shows how these themes interacted, with particular attention to the impact of living in Australia (see Figure I below). The Model of Familial Risk Perception suggests that once a person acknowledges that an illness runs in their family, the family history grows in meaning through many routes until a sense of vulnerability to that disease is gained. The person will then attempt to cope with or control that sense of vulnerability:

[Insert Figure I]

Walter's model was informed by a systematic review of qualitative studies that explored how people understand a family history of chronic disease (Walter 2004). A further interview study (Walter 2005) enhanced the findings of the review and confirmed the Model of Familial Risk Perception. We found considerable overlap between the themes identified in the UK study and those identified in the present Australian study. We were therefore able to confirm Walter's model (Walter 2005), and develop a new theoretical framework for how Australians come to terms with familial disease risk (see Figure II).

[Insert Figure II)

DISCUSSION

Core themes

Core themes around feeling at risk, family unit, and stage of life were shown to interact with one another and feed into how someone comes to terms with familial risk. The Australian perspective, which was underpinned by the isolation of Australia both in global terms and within the country itself, was described as a key barrier to meaningful family health talk and ultimately the collection of family history information.

The Australian perspective

Australia is essentially a young, migrant population. A nation of renowned cultural diversity, the Australian Bureau of Statistics suggests that almost 1 in 4 Australians were born overseas (Australian Bureau of Statistics 2006). Therefore, people living in Australia are often likely to have family members living overseas, and family health talk amongst these families is potentially hindered. Our findings confirmed previous work that associates a lack of geographical proximity with lack of family communication regarding health (Lindenmeyer 2010). Some participants described the difficulty of staying in touch with family members overseas, let alone being up to date with family health information. To a limited degree, the existing literature has noted this effect of having relatives spread across continents, even mentioning Australia and New Zealand as 'distant' locations in relation to nations in the northern hemisphere (Lindenmeyer 2010).

Australia is not only geographically far from other countries but it is also a huge country with two thirds of its population clustered in coastline metropolitan areas and the remaining population scattered across often remote areas. This study was conducted in Western Australia, the largest state of Australia, yet inhabited by only about 10% of the country's

population. The majority of Australia's inhabitants live in New South Wales and Victoria – states found on the eastern side of the country, whose capital cities are some three to four thousand kilometres from Western Australia's capital city, Perth.

The third important Australian factor is that Australian people's family history knowledge can also be limited due to historical events. For instance, during and post-World War II, a significant cohort of children were adopted from the war-torn United Kingdom, to provide a 'better life' for them in Australia. Whilst some of these Australians have been able to trace back to their European roots, many are left with gaps in information around their family history .

Our research confirms previous work from North America that correlates geographical separation with reduced family communication (Lindenmeyer A 1998). A number of participants described the difficulty of staying in touch with family members overseas, let alone being up to date with family health information. Whilst modern information technology facilitated communication between geographically distant family members, particularly for younger generations, it was less helpful for older generations. Despite developments such as Facebook Twitter and Skype which should reduce the effect of geographical separation, whether this is enough to harness the closeness required for effective family health talk warrants further research.

Feeling at risk

The findings showed that there is a variable understanding of what people considered about a condition "running in the family," and their understanding was influenced by numerous factors previously described, including the number of relatives affected, the age of diagnosis of affected relatives, and the severity of the illness (Walter 2005).

Walter and Emery described an individual relating to an affected family member in terms of their “likeness” in appearance and personality, and that this could raise concerns about their own vulnerability to the same condition (Walter 2005). These results confirm this finding, and suggest that “likeness” may extend to other aspects including stage of life, role, lifestyle or personal situation.

Conversely, feeling “different” in crucial ways to the affected relative has also been described as a mechanism employed to decrease a person’s perceived risk (Hughes 2003). This was particularly evident when participants described why affected family members were obvious ‘candidates’ for the diseases they developed. Lifestyle habits such as smoking, poor diet and lack of exercise were often cited as reasons for heart disease and diabetes, and to a lesser degree cancer.

A number of participants had experienced the “lived experience” of a disease, namely diagnosis, disease trajectory, and sometimes death, of family members. These experiences impacted on their disease perceptions and their own sense of risk and confirmed that the way an individual perceives the importance of their family history and comes to understand their familial risk can be affected by their experience in witnessing a relative’s illness and suffering and the emotional impact this has (Lindenmeyer 2010, Matthew 2011, Walter 2005).

Disease-specific factors were also important. Overall, people considered cancer to be more serious than heart disease, which is in turn more serious than diabetes. Confirming previous findings (Walter 2006, Wang 2009), cancer was almost universally considered the most severe of the three illnesses, often considered an inevitable life sentence. Diseases were largely considered to be multi-factorial in cause and the shared environmental, behavioural and inherited factors were also considered to be important.

Walter and colleagues (Walter 2005) suggested that a person's ability to control or cope with their perceived familial risk often depended upon their perceived severity of the disease. Our data were consistent with this finding, in that the development of cancer and its trajectory were largely out of one's control, whereas there were more aspects of participants' lives that they could modify to control heart disease or diabetes.

External factors also contributed to feeling at risk.A number of participants mentioned that the media tends to trigger a heightened awareness of risk, and often gives a 'bombardment' of information. Whilst some studies (Hughes 2003,Wang 2009) have suggested that this "bombardment" can increase a person's sense of vulnerability, some participants in this study described the counter effect, and tended to tune out of what was considered to be "white noise."

Interestingly, the one media campaign that did resonate with a participant used a real life story of a young male surfer who was diagnosed with melanoma and died in his thirties. The common theme of "real people" being affected seemed to be a strong trigger that impacted upon some participants' senses of vulnerability. Whilst Walter and colleagues found that a few participants mentioned the influence of healthcare professionals or media in informing perceived threat of illness, the more common tendency was to draw on personal experience (Matthew 2011).

Family Unit

The family unit was described as pivotal to both the health of a family and how family health history was communicated amongst its members. A person's ethnic or cultural background was shown to influence and challenge their adult lifestyle choices. Within some family cultures some aspects of health talk, especially around female-oriented health, were considered inappropriate or "taboo" topics of discussion.

There is an extensive literature surrounding patterns of family communication around health and familial risk (Hunt 2001, Bylund 2011, Gaff 2007, Lindenmeyer 2010), with one study suggesting that health talk can even be an indication of care and closeness amongst family members (Harris 1998). Our interview data, however, did not link health talk with care amongst family members, with a number of participants suggesting that it was not something families would consider as a topic of 'normal' conversation.

Life stage

"Life stage" has been described in previous studies and draws from a common understanding that any point in the life span should be viewed dynamically as the consequence of past experiences and future expectations (Benjamin 2008, Kenen 2003). Participants' reactions to family history screening appeared to be influenced by the stage of their life journey. This confirms what others have noted, that with time and age, people are able to reassess their lives, priorities, decisions and plans (Appleton 2000).

Having children was a common motivator to maintain healthy lifestyle choices and undertake screening for chronic disease. Consistent with Kenen and colleagues' findings (Kenen 2003), parents commonly expressed concerns about the implications of increased risk for their children.

Participants described vulnerability due to "anticipated onset." Confirming findings from the relevant published literature, anticipated onset describes how an individual feels when they acknowledge that they perceive a threat to their own health from a disease at a specific life stage (Benjamin 2008, Lim 2011).

Coping with and controlling familial risk

Considering the themes that have emerged around feeling at risk and the ways of coping with and controlling familial risk, the results confirmed Leventhals' Self Regulatory Model (SRM) (Leventhal 1997), upon which Walter's "Model of Familial Risk Perception' was based (Walter 2005). Based on people's own representations of their family history as a potential threat, individuals devise action plans to cope with perceived threats. As Rees (Rees 2004) described, whether a person ends up actively pursuing positive action regarding their familial risk is dependent upon their personal sense of vulnerability. The more vulnerable a person feels, usually the more likely they are to take action.

Interviewees generally expressed the belief that a healthy diet, positive attitude and reduction in stress were key factors in controlling their disease risk. The data confirmed literature describing that people believe happiness, a balanced life where work, family and health were all balanced is usually more of an ideal than a reality (Kenen 2003).

Family history screening in primary care

The GP was considered a source of screening information, and interviewees generally expected that the GP would enquire about a patient's family history. However, it was also acknowledged that the patient themselves should be aware of their own family history information, and work with their GP to address potential familial disease risk. This notion of a "two-way partnership" between GP and patient to allow for optimal family history screening confirmed Harris and colleagues' findings from a study of first degree relatives of people with colorectal cancer (Harris 1998).

Acceptability of family history screening

The findings around the acceptability of family history screening resonated with previous work by Bylund and colleagues (Bylund 2011) who used the Extended Health Belief Model to

understand family members' perceptions of risk for a genetic condition. A number of the factors described as perceived benefits of genetic testing can be translated across to perceived benefits of family history screening: people will consider the health of other members of their family through passing on information, their personal future health, the benefits of having peace of mind through confirming or resolving the unknown, and finally whether there may be some urgent resolution where symptoms may be present. It is important to distinguish however, between the genetic testing for single gene conditions whereby the patient receives a definitive result surrounding the presence or absence of a genetic mutation that Bylund and colleagues describe (Bylund 2011), and family history screening for polygenic, complex conditions whereby only a risk category is provided.

Participants generally described family history screening as important in contributing to the prevention of common chronic diseases. Furthermore, family history screening was considered by many to be a benefit to the provision of a holistic primary healthcare service. The FHQ was considered an acceptable instrument to support family history screening, being easy to complete and not time consuming.

The family physician could play a significant role in providing tailored, preventive lifestyle advice and support for behavioural change, particularly directed at higher risk groups identified through family history risk assessment. For those identified at increased risk, there are specific evidence-based preventive interventions available, above and beyond those offered to the general population. The precise role of screening for familial disease in motivating change in health behaviours requires exploration as there is currently insufficient evidence to show that family history assessment in primary care improves patient outcomes (O'Neill 2008, Williams 2011).

Limitations

While this study was the first to explore the Australian perspective around family health history and provides important and novel findings, it is important to acknowledge the potential recruitment bias within the main family history screening study. There was only a 5% response rate to invitations sent out by primary care practices into the study so participants may have had a particular interest in their family history and an increased awareness of familial risk. The purposive sampling undertaken for the interview study was an important process in optimising the extent to which findings could be generalised with broad representation of gender, age group, familial disease and risk status.

As a single researcher/ genetic counsellor conducted the baseline family history screening appointment as well as the follow-up telephone interviews, any bias in the data collection and interpretation was minimised. It is possible however, that the researcher's clinical training could have influenced data collection and interpretation therefore the research team critically examined their own role, potential bias, and influence on the findings. It is also possible that the content and quality of the follow-up interview would have differed had the interviewer been a "stranger" to the participant.

Implications for genetic counselling

Our findings on how people make sense of their family history and their responses to being screened for disease risk using an FHQ, have implications for training and clinical practice. Genetic counsellors need to understand different responses to discussion about risk of common diseases such as cancer and heart disease including behavioural changes to reduce risk. This could include exploration of a person's perceptions about disease vulnerability as a motivator to behavioural change. Our findings relating to Australian migrants, the sense of isolation from family and from their family history is relevant to other geographically large countries with large migrant populations such as the US and Canada.

Research Recommendations

Our findings relate specifically to the Australian context. In the US there have been several national initiatives to raise awareness about the importance of the family medical history and it is possible therefore that one would find different perceptions in a US population. A larger study that compared the psychosocial impact of family history screening across different countries would provide better insight into the applicability of these findings to other populations. Future research is also needed to test a range of implementation strategies aimed at systematically applying the FHQ to clinical populations and measuring their effects on patient and health service outcomes.

Conclusion

This study has provided an in-depth analysis of the psychosocial impact of family history screening. It revealed that a person's response to family history screening depends on a complex interplay of family, personal and external factors, which in turn are driven by that person's stage of life. An explanatory model was suggested to explain the Australian perspective of coming to terms with family history disease risk. Unique themes were identified relating to the Australian context within which the family history screening study was conducted. The isolation of Australia both in global terms and within the country itself were described as key barriers to meaningful family health talk and ultimately the collection of family history information. The study also demonstrated that the FHQ was easily completed, and that people felt that family history screening should be undertaken in primary care.

Conflict of Interest

Author G Reid, FM Walter and J Emery declare that they have no conflict of interest.

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Table 1. Demographic Characteristics of Interview group compared and overall study group

Category	Interviewees ***n* = 28**	Overall study group ***N* = 526**
Female	21 (75%)	406 (77%)
Male	7 (25%)	120 (23%)
Age: mean, range	40.7, 31-50	40.1, 20-50
Population risk for all conditions	8 (29%)	279 (53%)
Increased risk heart disease	5 (18%)	89 (17%)
Increased risk diabetes	8 (29%)	121 (23%)
Increased risk breast/ovarian cancer	6 (21%)	65 (12%)
Increased risk colorectal cancer	2 (7%)	37 (7%)
Increased risk prostate cancer	1 (4%)	6 (1%)
Increased risk melanoma	4 (14%)	53 (10%)
Jewish Ancestry	1 (4%)	4 (0.8%)
Indigenous Australian Ancestry	1 (4%)	3 (0.6%)
Consanguinity	1 (4%)	2 (0.4%)

Figure I 'Walter's Model of Familial Risk Perception (Walter 2005)

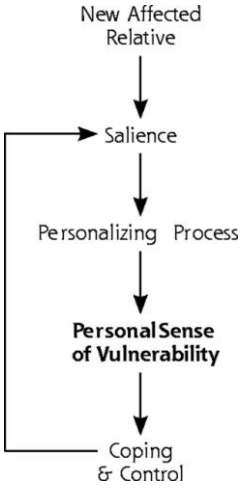

Figure II: Coming to terms with familial risk: An Australian perspective