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## Exploring the potential emotional and behavioural impact of providing personalised genomic risk information to the public: a focus group study

**Short title:** Impact of personalised genomic risk information

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## ABSTRACT

**Aim:** To explore the potential emotional and behavioural impact of providing information on personalised genomic risk to the public, using melanoma as an example, to aid research translation.

**Methods:** We conducted four focus groups in which 34 participants were presented with a hypothetical scenario of an individual's lifetime genomic risk of melanoma (using the term 'genetic risk'). We asked about understanding of genetic risk, who would choose to receive this risk information, potential emotional and behavioural impacts, and other concerns or potential benefits. Data were analysed thematically.

**Results:** Participants thought this risk information could potentially motivate preventive behaviours such as sun protection, and related it to screening for other diseases such as breast cancer. Factors identified as influencing the decision to receive genetic risk information included education level, children, age and gender. Participants identified potential negative impacts on the recipient such as anxiety and worry, and proposed that this could be mitigated by providing additional explanatory and prevention information, and contact details for a health professional to discuss further. Participants' concerns included workplace and insurance discrimination.

**Conclusion:** Participants recognised the potential for both positive and negative emotional and behavioural impacts related to receiving information on personalised genomic risk of melanoma.

## INTRODUCTION

The potential for personalised genomic risk information to motivate behaviour change and promote health among the public is a burgeoning area of research [1,2]. Social and behavioural theory suggests that the highly personalised nature of providing genomic risk information may be a more powerful motivator of behaviour change than standard prevention approaches [3]. However, this information might be interpreted in different ways by recipients, which could cause them to worry or feel that illness is inevitable.

An essential component for translation of genomics into improved preventive health behaviours is the assessment of emotional and psychological impacts that might arise from receipt of personalised genomic risk information, as well as broader ethical and social issues. Addressing these issues will help to anticipate and minimise possible adverse consequences [4]. Understanding the potential impact of genomic risk information can also help to contextualise behavioural effects, and plan appropriate implementation strategies for genomic risk information interventions. It is also important to predict who would elect to receive genomic risk information, and the reasons why, in order to effectively tailor the communication of genomic risk information and to maximise its relevance [5].

A recent review [6] found that most studies evaluating the impact of communicating genetic risk information have revealed either no change or a decrease in participants' negative emotions after receiving risk information. Additionally, improvements in cancer screening have been demonstrated after receiving genetic test results [3]. However, it is difficult to draw inferences about the potential impact of genomic risk information based on existing studies [3], as they have mainly focussed on rare high-penetrance gene mutations in families with a strong family history [4], or on common variants in a single gene [3,6]. Most common diseases such as cancer have complex multi-factorial causes influenced more frequently by multiple genetic and environmental factors rather than by single gene mutations [8]. Potential effects of returning risk information based on common variants across multiple genes ('genomic risk') to the wider population remain largely unexplored [1,2], and may differ to the impact of single-gene 'genetic risk' information because genomic risk is based on multiple probabilities and is not easily explained using family history [6].

We conducted focus groups to explore the issues around using genomic risk information to motivate healthy behaviour change as a novel public health disease prevention strategy. We used genomic risk of melanoma, the most serious form of skin cancer, as the disease example because: Australia has the world's highest incidence of melanoma (4<sup>th</sup> most common cancer in Australia) [9], common genomic variants are

strong predictors of melanoma risk [10,11], melanoma is highly preventable [12,13], and skin cancer prevention and detection behaviours remain sub-optimal for most Australians [14]. Focus groups are particularly suited to new areas of research because the interactive nature of discussions stimulates participants' thoughts about topics they may not normally discuss [15,16], and provides insight into how members of a target audience understand a particular topic and the language they use to talk about it [15].

The aims of this study were to:

- find out how personalised genomic risk information might be used by the public;
- determine whether people would elect to receive personalised genomic risk information and what factors may influence this decision; and,
- explore (both positive and negative) potential ethical, social, and psychological impacts of receiving genomic risk information.

## **METHODS**

### *Participant recruitment*

Participants for this study were recruited via the "Join a research study" database managed by the Cancer Council New South Wales (NSW), Australia. Members of this database have given consent to be contacted by researchers carrying out ethically approved cancer research studies. Ethics approval was obtained from The University of Sydney. The participant eligibility criteria were: age 18 years or older and no personal history of melanoma. In order to establish diversity as well as geographical representativeness, invitation letters were sent to central, western, northern and southern locations across Sydney, NSW Australia, and the focus groups 1, 2, 3 and 4 were offered in these locations, respectively, in November 2014. The central and northern locations were closer to the inner city while those in western and southern Sydney were located in outer-city suburbs.

Packs including an invitation letter, participant information sheet, consent form, participation card and reply paid envelope were sent via postal mail to 200 individuals; six were returned due to an incorrect address. Once a participant returned their consent form, we contacted them to allocate them to the focus group session most convenient for them. In response to the invitation mail-out, 43 (22%) people gave their consent to participate in a focus group discussion and an additional 25 (13%) gave their consent to participate in a phone interview if needed. Thirty-four participants ultimately attended the four focus groups, which were made up of 5, 12, 8 and 9 participants. A \$50 gift voucher was given to each participant to compensate them for their travel expenses and time.

### *Conducting the focus groups and discussion content*

Focus groups were conducted in two parts in a single two-hour session, including a 15 minute break. Discussion was conversational, guided by a semi-structured focus group guide with a theme list and prompts (see Discussion Guide, Appendix 1 online supplementary material). Participants were asked about their current understanding of melanoma risk and genetic risk. We used the term 'genetic risk' rather than 'genomic risk' to facilitate understanding among the public, although mentioned that the risk example was calculated based on common variations in 18 different genes. Following this, the potential impact of receiving information on personal genomic risk of melanoma as well as associated ethical, social and psychological issues were discussed.

To aid the discussion, participants were presented with a hypothetical example of personalised genetic risk information, presented graphically and in words (see online supplementary material Appendix 2). The risk information showed an 18% remaining lifetime genetic risk of melanoma for "Sarah", a hypothetical 45 year old woman living in New South Wales, Australia, compared to an average risk of 5%. The lifetime risk calculation reflected her risk of developing melanoma from now until the age of 85, and was based on her (hypothetical) genomic variation in 18 different genes, her age, sex and the state in which she lived (because melanoma incidence varies strongly by age, sex and ambient solar ultraviolet radiation). Participants were asked about what concerns people might have about receiving this information, how people could use or might respond to this information, whether they thought people would be interested in receiving it, and whether they would share their risk information with their family or friends. Preferences for different graphical and risk communication formats

and for delivery of genomic risk information were also discussed and are presented in a separate paper (manuscript under review).

#### *Data capture, coding, and analysis of qualitative data*

The focus groups were audio recorded, transcribed verbatim, and analysed thematically. QSR NVivo10 qualitative data management software supported the coding process. Initially, a working coding framework was developed, which was structured according to research questions and the Discussion Guide. Through an iterative process of reading and re-reading the transcripts, additional themes and sub-codes were identified and added to the coding framework. We used inductive reasoning to allocate phrases, words and paragraphs to both the top-level codes and sub-codes. The data within each theme were then further analysed to identify variations or patterns present. Coding was cross-checked between authors AKS and AEC, and discrepancies were resolved through discussion.

## **RESULTS**

The average age of participants was 56 years (range 19-83 years) and almost three quarters of participants held a university level qualification (Table 1).

### **Knowledge and understanding of melanoma risk and genetic risk**

Participants were asked '*Can you tell us what you already know about melanoma risk?*' In response, they listed skin type, eye and hair colour, moles, occupational and recreational forms of sun exposure as factors likely to increase melanoma risk. Knowledge of genetic risk factors for melanoma ranged from no knowledge to a sophisticated understanding. Several participants said they were previously unaware that genetic factors were related to melanoma development.

Female, focus group 2: Well I must admit, I showed my ignorance, but I had no idea that it was genetically involved. The only thing I could think about coming in the genes is if your parents or grandparents were very white, you know, white skin where you're more prone to get it and then obviously your family are probably the same skin shade...

Other participants used terms such as "family history", "DNA" and "genetic or inherited predisposition" to describe how they understood the meaning of genetic risk. One participant knew of *CDKN2A*, a high penetrance gene associated with melanoma risk.

When genetic risk was explained to participants, different views about the impact of gene variants on disease risk were expressed. Some participants noted that while genes may influence susceptibility, they do not necessarily predetermine melanoma occurrence. Others thought that if an individual has a genetic risk of melanoma, the environment and sun exposure could interact with genes and act as "triggers" for the development of melanoma. Conversely, some participants thought that genes determine the development of some diseases irrespective of external factors:

Male, focus group 3: If you think about it [your body] like a car, when out of the factory line, [it may be] resistant to engine problems where the others are not. So when I look at genetic [factors], it's what is in your body regardless of your behaviour.

Overall, while knowledge of environmental and behavioural risk factors was high, knowledge of genetic risk was variable, and participants usually thought about genetic risk in terms of characteristics shared within families. There was a continuum of genetic determinism from the belief that genetics plays a minor role in comparison to environmental factors, to genetics being seen as the most important risk factor.

### **Responses to hypothetical genetic risk information**

#### *Potential to cause worry*

Many participants believed that their reaction to receiving genetic risk information would be contingent on the risk level they received. Some participants stated that they would be scared or worried if they received Sarah's results and one participant stated that, "(...) even just above the average is probably enough to frighten most people." Participants also described genetic risk information as "dangerous" because "people don't understand (...) what risk is all about", and expressed concern about it potentially causing anxiety and depression.

Female, focus group 2: It [genetic risk information] makes you depressed for the rest of your life because you've got the information you're more than high risk.

Participants thought that some level of worry would be beneficial, because if a recipient of genetic risk information was not worried by their result, then participants thought they would be less likely to change their behaviour.

#### *Improved prevention*

Changes in behaviour such as getting skin checks by general practitioners (GPs), "screening", general sun protection or wearing sunscreen and hats were listed as possible responses to genetic risk information. Participants stated that if they were to receive a result that specifically indicated they were at high risk, then they would be more likely to change their behaviour. One participant said, "Well I know what would work with me, to be honest: to be told that you're high risk." It was also proposed that melanoma genetic risk would be novel information for many people, and may be more persuasive in encouraging positive behaviour change than existing sun protection and melanoma risk information, which was described by participants as "common knowledge" in Australia. Furthermore, participants framed their understanding of the potential benefits of receiving genetic risk information by identifying benefits of screening for other diseases such as breast and bowel cancer. Some participants believed that people with known genetic predispositions for breast cancer and bowel cancer generally "take better precautions," and therefore melanoma genetic risk information could similarly influence people to positively change their health behaviours.

#### *Decreased prevention*

Some participants stated that being in a low-risk category may act as a deterrent to sun protection behaviour, as a recipient may believe that they do not need to take preventive action if they have a low risk. A recipient may also interpret 'low risk' as 'no risk', which was described as "counter-productive" by some participants. Many participants believed that a low risk category result should be accompanied by an explanation that low risk does not mean 'no risk'.

#### *Anxiety for adults with children*

Participants thought that adults with children might have different emotional and behavioural responses to genetic risk information (compared to those without children), and they may be more likely to be concerned.

Male, focus group 1: The problem you're going to face is that mummy comes home and she's, oh God, my little darlings, oh better get them screened (...) and then she will spend all her time panicking, you need (...) the people to understand what it [risk] actually means [for them]...

Participants believed that most people tend to link the term 'genetic' with 'family' and therefore parents who receive genetic risk information may automatically think that their children have the same risk. It was thought that parents receiving genetic risk information may encourage their children to use sun protection and that people may be "more careful with children, knowing they might be passing it on."

Participants raised the topic of testing children for genetic diseases and there were mixed views about it. Some participants believed that if a recipient received a high-risk estimate, they would want to test their children.

Male, focus group 3: I think a mother would definitely speak to the kids about getting them tested. I think that would definitely strike a personal chord. I mean I would if I had that 18%, I'd tell - I'd ask if my kids get tested. Considering it's only a saliva test.

Other participants noted that if the recipient's children were very young, then they would be less likely to consider requesting genetic tests for them. Some participants noted that due to the permanency of a genetic test, they wouldn't want to test their children. One participant stated "once you've got it in place and you get the [genetic] testing done (...) they can't do anything about it."

#### *Depends on age and gender*

Participants suggested that age and gender could influence responses to genetic risk information. Although some of the young adults attending the focus groups were health conscious and would be interested in

their risk information, it was generally agreed that young people may be less concerned about their risk information than older people. Participants proposed that young people take more risks, are less likely to use sun protection, think they are "invincible" and wouldn't listen or change their behaviour in response to genetic risk estimates. Although participants identified melanoma as a cancer that affects young people, it was also stated that young people may not understand how melanoma could potentially affect them.

Male, focus group 2: I wonder if it's [melanoma] a bit like superannuation. I remember people coming into work talking about superannuation. I was like, I don't want to know about that for another 20 years.

Female participants tended to describe women as more "health conscious" than men and therefore more likely to modify their behaviour or be concerned by risk information. Some female participants believed that preventive behaviours, such as screening tests including pap smears and mammograms, is "trained into women". Female participants generally described men as less likely to be interested in receiving genetic risk information than women. Some male participants proposed that men could have a similar reaction to women in response to genetic risk information, and that some men are likely to be interested in their genetic risk, particularly if they have a family history of a disease.

### *Depends on the 'type of person'*

Participants identified two different types of people: the type of person who takes action according to health warnings, and the type of person who is aware of health risks but does not change their behaviour.

Female, focus group 1: It would depend very much on the sort of person they are. I mean you just have to use smoking as an example. People have been told about smoking for a long time and they just ignore it, for lots of reasons, but other people immediately did something about it.

This duality framed participant discussion about how genetic risk information could influence emotions and behaviours. Smoking, wearing a helmet while riding a bike, wearing a hat while outdoors and applying sunscreen were some of the examples used to support the 'two types of people' idea. Participants mentioned other diseases such as breast cancer and bowel cancer to explain how genetic risk information can, but does not necessarily, have an impact on people's decisions about undergoing screening for various diseases.

### *Barriers to positive behavioural responses*

Potential barriers that might discourage people from changing their sun protection behaviour were mentioned, including inconvenience, concerns about appearance, and a lack of education. Participants also proposed that a risk estimate may not in itself be convincing enough for people to change their behaviour. It was mentioned that a diagnosis of melanoma would be a more persuasive way of encouraging behaviour change because "people don't think it's going to happen to them". One participant noted that she already had good sun protection habits and therefore a high genetic risk estimate would only cause her to worry, because she would not be able to do anything in addition to her normal habits in order to manage her risk.

### **How to mitigate negative impacts of genetic risk information**

While participants recognised that receiving genetic risk information, particularly via a written medium, may cause an individual to "freak out" or "panic", they also proposed various ways of mitigating this reaction.

### *Information*

Providing additional explanatory material to place the risk information in context was suggested.

Male, focus group 3: To be able to interpret the [genetic risk] data and present it with some context I think is a pretty powerful piece of the puzzle and I think it does avoid any of those spiralling off into I'm going to die, fatalistic sort of behaviour and or covering your kids in SPF [sun protection factor] at midnight.

Participants proposed that explanatory material should contain: an explanation of melanoma and other melanoma risk factors, an "action plan" detailing how melanoma risk can be minimised and managed, an outline of how behaviour can reduce risk over time, and information about the implications for their children's risk. Generally, participants expressed a desire to know more information in regards to risk and

what they could physically do. Another participant recommended that explanatory material should contain the distinction that the risk estimate is not a "cancer diagnosis".

#### *Contact with a health professional*

Participants suggested providing contact details for a health professional, either a general practitioner or a genetic counsellor, alongside the genetic risk information so that recipients who experienced worry or struggled to understand their results would be able to seek help and advice. Participants recognised that receiving a high risk level result was likely to have more impact, and suggested that a high risk level should be delivered either face-to-face or via telephone by a health professional rather than by mail.

#### **Interest in receiving genetic risk information**

Many participants indicated that they would be interested in receiving genetic risk information. They noted that "knowledge is power" and "you can't afford to be ignorant." However, others stated that some people may not be interested because it is potentially "bad news". Factors that may influence the decision to receive genetic risk information included: cost, invasiveness, having children and convenience. Some participants believed that a family history would motivate people to receive their risk, while others noted that even in the absence of a family history they would still be interested in receiving their risk information. Participants also proposed that they would be interested in finding out their genetic risk in order to inform their children. Other participants noted that they would only want to receive their risk if there were ways to reduce or control it.

Female, focus group 3: No, I've got a slightly different spin on it. I'd only be interested in knowing it if that would then lead me to know about other things I could do to reduce that risk. If it's just a risk that you can't do anything about, I think I'd rather live in ignorance.

Furthermore, participants suggested that elderly people may not be interested as they may feel as if they're "going to die from something anyway".

#### **Sharing genetic risk information**

Many participants believed that they would share the risk information with their family. Some participants thought that once a person finds out their genetic risk they are obliged to share the information. Others mentioned that they would not want to upset their family, but this would not prevent them from sharing the information. Diseases such as breast cancer, bowel cancer and osteoporosis were used by participants as examples of cases in which sharing genetic risk information can be helpful for family members. Some participants indicated that sharing the risk information with family was the responsibility of the individual receiving it but that it was up to the family member to modify their lifestyle. People with children, particularly women, were identified as being more likely to share their risk information with their family. It was also raised that children should be taught sun protection behaviour regardless of whether a genetic test might later show them to be at risk.

Female, focus group 3: Getting tested isn't going to change the behaviour that the kids should have, you know, the kids should wear a hat, wear sunscreen, avoid the sun between 11:00 and 3:00, all of that.

Additionally, it was acknowledged that not all people would want to share this information, and that whether or not someone shares genetic risk information depends on their relationship with their family and friends.

#### **Concerns about genetic risk information**

##### *Access to genetic counselling and health system pressures*

Many participants were not familiar with the current system for accessing genetic counselling in Australia [17]. When the facilitator explained the current system, participants raised concerns about cost and accessibility of genetic counsellors if genetic testing became more common and the existing Australian system remained.

Female, focus group 4: But if this is going to be like genetic testing on a broad scale over a number of different sort of conditions, it's probably never going to happen that you're

going to - even if it's mandatory, you're probably never going to see a genetic counsellor anyway.

There were comments regarding the feasibility of having genetic testing widely available, and concern about placing pressure on the health care system as a result of an increased demand for support for people receiving genetic risk information. Participants expressed sentiments such as, "You also wouldn't want your whole population going for that really regular early testing for melanoma..." as well as "You can't have every single person, for instance, having random genetic tests just because they just want to find out". Participants indicated that the health system would be more efficient if only high risk individuals attended genetic counselling.

#### *Potential employment discrimination*

The possibility of discrimination on the basis of receiving genetic risk information was raised by participants. It was proposed that the risk information may become accessible to people such as employers, which could disadvantage job seekers.

Female, focus group 3: And I don't know, you know, this has drawn a very long bow, but say you had your heart set on the construction industry, would they preclude you from a job on the basis of this because you know it's outside? Well I'm just - the insanity.

Employers cannot legally discriminate on the basis of genetic predisposition in Australia, however an employer can require information to determine whether an employee is able to carry out the inherent requirements of employment or to work out what reasonable adjustments to make for an employee. Some participants were unsure about whether potential employers could legally ask about genetic risk information; others observed that while employers may not explicitly state they would not hire someone at high risk, such a risk level may (possibly unlawfully) influence employers' decisions. Participants proposed that access to genetic risk information must be strictly regulated, while others raised "duty of disclosure" issues.

#### *Potential insurance discrimination*

Participants expressed concern about insurance and genetic risk information and were unsure if premiums could increase based on genetic test results. Currently in Australia, life insurers can make changes to new or updated, but not *existing*, life insurance policies on the basis of new genetic or other health information. They must, however, be able to provide actuarial data to support any loading of premiums. Health insurance in Australia is community rated, so premiums are not affected by individual characteristics, but participants were concerned that in the future these protections may be removed. There was some confusion between the potential impact on health insurance and life insurance. Some participants said that people may want to have life insurance before they receive genetic risk information.

Female, focus group 2: I just think that some people would want to make sure that they've got life insurance in case before they did it.

Male, focus group 2: Yeah, no cancer till after the test.

## **DISCUSSION**

There is a growing interest in whether personalised genomic risk information can be used as an effective motivator of behaviour change, to promote health among the population [1,2]. However, evidence of effectiveness alone is not sufficient to translate DNA-based behaviour-change interventions into routine public health policy and clinical practice. An essential component for translation is the assessment of emotional and psychological impacts as well as broader ethical and social issues that might arise from the public receiving personalised genomic risk information.

We found that many participants did not know that there were genetic risk factors for melanoma, and were unfamiliar with the term 'genetic risk'. However, other risk factors for melanoma such as sun exposure and skin characteristics were considered "common knowledge" for the Australian public, as they are usually the focus of mass media skin cancer prevention campaigns [14]. Despite this, participants noted that behaviours such as lying in the sun to become tanned are still common.



Some participants believed that presenting information on personalised genetic risk of melanoma may be more powerful in encouraging positive behaviour change than existing sun protection education. A recent study on the potential impact of giving personalised genetic risk of breast cancer to the public revealed that 85% of women would support more frequent breast screening if they were found to be at higher genetic risk of developing breast cancer [7]. A focus group study on (hypothetical) genetic testing for ovarian cancer in the population also demonstrated that participants would respond to a high genetic risk result as a "wake up call" for taking preventative action [18]. Participants in our study identified potential benefits of receiving information on genomic risk of melanoma, such as increased skin checks (early detection) and sun protection (melanoma prevention), and linked these benefits with screening for other diseases such as colonoscopies, mammograms and pap smears.

In the context of discussion about behaviour change, participants used examples such as smoking despite knowing the associated risks, to explain that responses to genomic risk information are contingent on "the type of person". Age, gender and having children were also mentioned as factors that might influence behavioural responses to receiving genomic risk information. Younger people, particularly young men, and the elderly, were identified as less likely to change their behaviour. Women and parents were categorised as more likely to make positive behaviour changes. An individual's underlying risk perception has also been shown to influence motivation to change behaviour [3]. However, risk perception alone is thought to be insufficient to motivate behaviour change, and psychological and social pathways are considered important influences [3].

A study on communicating genetic risk of Alzheimer's disease found that groups more likely to seek genetic risk assessments for common disorders included women, people of European origin, individuals with a family history of a disease and adults with high education levels [5]. Participants in our study had mixed views as to whether a relevant family history would influence the choice to receive a genomic risk estimate. Women were identified as more likely, and men and younger people less likely, to opt for genomic testing. Some participants said that they would want to receive their personal genomic risk information in order to inform their children or family members, to encourage improved sun habits. Other participants maintained that having children would be a disincentive for receiving genomic risk information as it may cause parents anxiety about the implications for their children's risk levels. There were mixed views about whether or not children should get genetic testing.

Receiving genetic risk information can potentially raise emotional concerns, anxiety or feelings of fatalism [3,5]. Our focus group participants believed that a negative emotional or psychological response would be more likely if the recipient misunderstood the personalised genomic risk information. An analysis of social and behavioural research related to delivery of genomic sequencing results in clinical practice found that most studies demonstrated either no change or a decrease in negative emotions such as anxiety or depression compared to prior levels after receipt of genetic risk information, even when individuals receive results reporting increased disease susceptibility [6]. A Cochrane systematic review [4] also found no evidence that communicating DNA-based disease risk had detrimental psychological effects, however the quality of included studies was generally poor.

Participants in our study proposed that negative emotional and behavioural responses could be potentially mitigated by factors such as providing additional explanatory material to explain the risk and to describe what they could do to change their melanoma risk through preventive actions; providing a contact number for a health professional (general practitioner or genetic counsellor) to be able to discuss the risk information in more detail and ask questions; and communicating face-to-face with a health professional if the person is receiving information placing them in a 'high risk' category.

Generally participants viewed genomic testing positively. However, in line with findings of other studies [19-21], participants identified concerns about occupational, health- and life insurance discrimination. Participants were concerned that the existing protections against discrimination on the basis of genetic risk may be removed in the future. In the context of discussion about genetic counselling in Australia, participants believed that the current health system would not be able to sustain widespread feedback of

genomic risk information and that genetic counsellors would be in high demand. Participants' suggestions to address these workforce issues included expanding telephone-based genetic counselling, and restricting genetic counselling to those people in receipt of high-risk results only.

A limitation of this study is that participants' views were based on a hypothetical scenario. A study by Sanderson et al [22] found that hypothetical interest in genetic testing only modestly reflects future testing behaviour. Other studies have shown that expressed preferences for receiving genetic risk information and intentions to modify behaviour are not necessarily predictive of future preferences and behaviour change [4,6]. Therefore, participants' views in our study may differ to the actual implementation of the proposed intervention. Further research is required to investigate the actual uptake of genomic testing, and to examine how the public will respond to personalised genomic risk estimates should they choose to receive this information. Our focus group participants had a relatively high education status and had an existing interest in cancer research, which may limit the generalisability of our findings. A strength of the study is the focus on genomic risk information for a relatively common and highly preventable cancer among the wider public, extending our knowledge beyond single-gene mutation results and families with a strong family history.

Our findings provide insight into who would be interested in genomic risk information, and the potential behavioural and emotional impacts, both positive (e.g. improved prevention behaviours) and negative (e.g. anxiety). In addition, broader ethical and social issues of delivering genomic risk information were raised, such as the possibility of genetic risk discrimination and complexities of communicating risk with family members. These important issues need to be taken into account when considering provision of personalised genomic risk information to the public.

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**Table 1:** Demographic characteristics of participants

Characteristics	Number n=34 <sup>1</sup>	(%)
Gender		
Males	9	(27)
Females	24	(73)
Age (years)		
18-45	10	(30)
46-65	12	(36)
66-85	11	(33)
Education		
Some high school	0	(0)
High School	4	(12)
Technical college certificate or diploma	5	(15)
University diploma or degree	24	(73)
Country of birth		
Australia	24	(73)
Other	9	(27)
Ethnic background		
Caucasian/white	26	(79)
South Asian	3	(9)
Middle Eastern	1	(3)
Pacific Islander	1	(3)
Other	2	(6)
Hair colour at age 18		
Red	2	(6)
Blonde	3	(9)
Light or mouse brown	12	(36)
Dark brown	13	(39)
Black	3	(9)
Eye colour		
Black/brown	15	(46)
Blue or grey	12	(36)
Green or hazel	6	(18)

<sup>1</sup> One participant attended the focus group but did not complete a questionnaire

## **APPENDIX 1 (SUPPLEMENTARY MATERIAL)**

### **Exploring the potential emotional and behavioural impact of providing personalised genomic risk information to the public: a focus group study**

#### **FOCUS GROUP DISCUSSION GUIDE**

**Script for focus group facilitator:** This will be spoken by the facilitator

Welcome to today's Focus Group session!

My name is [facilitator's name] and this is [other researcher], who are part of the [name of research team] with the [name of institute]. You have been invited to participate in today's focus group session to help us develop an easy to understand method of talking about a person's individual genetic risk of melanoma.

We would like to hear everyone's ideas and feedback, and we encourage all of you to participate and contribute to the session. We do have quite a lot to get through within the 2 hours so we ask for your understanding and cooperation, as we may sometimes have to interrupt discussions in order to move onto our next talking point. We are not looking for you to agree about anything so it is completely fine for you to disagree with each other, but obviously please try not to criticise others.

*[Explain that we are not doctors and cannot give medical advice but we will answer questions to the best of our ability and they can follow up with their own doctor.]*

*[Request participants to switch off mobile phones or put them on 'silent'.]*

Our discussion today will have two parts. The first part will explore how people understand melanoma risk and different ways of communicating genetic risk as well as the potential impact of receiving information on personal genetic risk of melanoma for you and your family. We will use made-up examples only, you will not be given your own personal genetic risk of melanoma. We will then have a short break. Light refreshments, tea and coffee will be available during the break. The second part of the session will explore whether a genetic counsellor could help with the communication process.

We would like to make an audio recording of this session, just to make sure we can accurately summarize the discussion. Everything that's said or written today will be kept strictly confidential. When the study results are reported, it will not be possible to identify any individual participants.

*[To discuss location of toilets, fire exits, practical issues]*

Are there any questions?

We will start off the session by getting you to fill out a short questionnaire about yourself. If anything is unclear in the questionnaire, please feel free to ask one of us to clarify for you.

## Description of Focus Group sessions

THEMES	FLOW OF DISCUSSION AND KEY QUESTIONS	ADDITIONAL PROMPTS	Estimated time
<p>PART 1</p> <ul style="list-style-type: none"> <li>• Risk Understanding</li> <li>• Risk Presentation</li> <li>• Ethical, Social and Psychological implications</li> </ul>	<p><i>Script above and distribute basic questionnaire to collect information about age, sex, skin colour, eye colour, hair colour, ethnicity, city/country of birth, sunburn, current sun protection behaviours (Recorded individually)</i></p> <p><i>[With participants' consent, <b>start recording</b>. Request that people speak up and avoid speaking at the same time as others.]</i></p> <p><i>[Ask people to introduce themselves <b>briefly</b> – first names.]</i></p> <p>INTRODUCTION:            As you may be aware, melanoma is the most serious type of skin cancer in Australia and we are trying to improve its prevention and early diagnosis. We know that a person's genetic make-up can contribute to their risk of getting melanoma, even when a person has no family history of melanoma. These genetic changes can be inherited and passed on to future generations. We now know quite a lot about the genetic changes that increase a person's risk of melanoma, and we can test for some of these using a blood or saliva sample.</p> <p>Currently genetic testing through the public health system is only available for a rare gene change that puts you at a high risk of developing a melanoma. However, as we find more common gene changes that contribute to the risk of developing melanoma, in future, we can expect testing to be available for these more common gene changes.</p> <p>We are going to be talking a lot about <u>melanoma risk</u>, so, to clarify: in today's session when we are talking about risk, we are talking about the chance of a person developing melanoma over their lifetime.</p>		1 Hour

	<p>1) Before we begin, can you tell us what you already know about melanoma risk?</p> <p>2) Does anyone know what the risk factors of melanoma are?</p> <p>3) What do you think we mean when we say “genetic risk of melanoma”?</p> <p>We will now look at a hypothetical risk scenario, and a few risk presentation formats. We would like you to tell us what the risk presentations mean to you, and whether or not they are easy to understand. We know people don't always think about risk in terms of numbers and graphs, and some people find graphs confusing, that is why we would like to know how you interpret these presentations of melanoma risk.</p> <p><b><i>Distribute hypothetical scenarios with risk presentation formats (5 formats)</i></b></p> <p>Let's look at the Scenario and Format 1 – <i>read out scenario and presentation format</i></p> <p>4) What does the graph mean to you?  5) What do you think Sarah's risk is?  6) Do you have any comments on the way it is presented?</p> <p>7) What do you like about this format? 8) What don't you like about this format? 9) Can you make suggestions for improving it?</p> <p><b><i>Repeat questions for other formats (2, 3, 4, 5) – wording is the same</i></b></p> <p><b>Can you please indicate on this form (show paper) your preferences for the different risk presentation formats, ordering 1 as your favourite to 5 as your least favourite.</b></p>	<p>Is it something you ever think about? Talk to others about?</p> <p>Do you think there may be genetic or inherited risk factors?</p> <p>Is it easy to understand?  Is there anything unclear?</p> <p>How do you think people might respond to this information?</p>	
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	<p>10) Would it matter if this information was presented online or on paper?</p> <p>One of our research questions is whether this type of information would lead to people making better decisions about preventing melanoma, so we would like your views on this also.</p> <p>11) How do you think people might respond to this information about their own genetic risk?</p> <p>12) What concerns might people have about receiving this information?</p> <p>13) Do you think it will influence how much time people spend in the sun or how they use sun protection?</p> <p>14) Do you think people would be interested in knowing this information?</p> <p>15) How do you think people could use this information?</p>	<p>Do you think people would feel any different after receiving this information?</p> <p>How might receiving this information make people feel? Anxious? Stressed? Relieved? No Difference?</p> <p>In what way?</p> <p>Why or why not? If so why? At what ages/stage in life?</p> <p><i>Practical:</i> Sun protection behaviours</p>	
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	<p>16) Do you think anyone should be able to find out this information for themselves, or should it only be given by a health professional?</p> <p>17) Do you think a person who gets this information would share it with their family? Why or why not?</p>	<p>Skin examination behaviours Time spent in the sun <i>Psycho-social</i>: insurance, family communication</p> <p>Would they feel comfortable? Important for family to know?</p>	
BREAK			5-10 mins
<p>PART 2</p> <p>Genetic counsellor assistance</p>	<p><i>Reanu</i>: Briefly introduce the role of a genetic counsellor A Genetic counsellor is someone who is able to provide information to individuals and families about genetic conditions including certain types of cancer. They organize and assist with genetic testing and screening and deliver genetic test results and risk information about genetic conditions. They also provide options and support for the decision-making process and help with coming to terms with the impact of genetic test results and communicating this information within families.</p> <p>18) If people were sent out information about their personal genetic risk of melanoma, do you think they would be interested in talking to a genetic counsellor about it?</p> <p>19) Let's think back to Sarah, whose risk we looked at earlier. Imagine that when she gets her genetic risk information, Sarah makes an appointment to see a genetic counsellor. What sorts of questions do you think she might want to ask the genetic counsellor?</p>	<p>Specific concerns? Inheritance? Potential concerns of passing on this genetic risk? Genetic testing?</p>	30 mins

	<p>20) Do you think it would be better to talk to a genetic counsellor or other health professional over the phone or in person?</p> <p>21) Currently in Australia, through the health system, genetic risk information is only given to people with lots of cancers in the family, and can only be given through a genetic counsellor. What do you think about this?</p> <p>22) Is there anything about what we have discussed today that concerns or bothers you?</p> <p>23) Is there anything else that you would like to say or ask that we haven't already discussed today?</p> <p><i>THANK ALL PARTICIPANTS FOR THEIR CONTRIBUTION TODAY.</i></p>	<p>Concerns about sharing information with family?</p>	
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End of Session (1 Hour 45 mins)