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Research paper

Making sense of genetic risk: A qualitative focus-group study of healthy participants in genomic research

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ABSTRACT

Objective: It is well known that research participants want to receive genetic risk information that is about high risks, serious diseases and potential preventive measures. The aim of this study was to explore, by qualitative means, something less well known: how do healthy research participants themselves make sense of genetic risk information?
Method: A phenomenographic approach was chosen to explore research participants' understanding and assessment of genetic risk. We conducted four focus-group (N = 16) interviews with participants in a research programme designed to identify biomarkers for cardiopulmonary disease.
Results: Among the research participants, we found four ways of understanding genetic risk: as a binary concept, as an explanation, as revealing who I am (knowledge of oneself) and as affecting life ahead.
Conclusion: Research participants tend to understand genetic risk as a binary concept. This does not necessarily imply a misunderstanding of, or an irrational approach to, genetic risk. Rather, it may have a heuristic function in decision-making.
Practical implications: Risk communication may be enhanced by tailoring the communication to the participants' own lay conceptions. For example, researchers and counselors should address risk in binary terms, maybe looking out for how individual participants search for threshold figures.

1. Introduction

New technologies in the field of genomics, such as next-generation sequencing (NGS), enable a vast number of biomarkers to be measured easily with simple blood samples. By looking at patterns of change in the genome, and in relation to people's lifestyles and viral infections, extensive 'omics' analysis is helpful for investigating disease progression [1]. Exploration of these disease mechanisms helps scientists to identify new biomarkers that allow identification of high-risk individuals more precisely and at an earlier stage, all of which may contribute to improving health and saving lives [2].

These genomic technologies are advancing rapidly, costs of testing are declining and analysis of results is becoming faster [3]. Genomic testing is expected to become more common in research (as well as in clinical practice). High-throughput investigation of this kind generates large quantities of data, some of which involve potentially unexpected genetic risk information that goes beyond the scope of the original research aims. This development opens up a new pathway into health care. As a result of having received genetic risk information, healthy research participants may approach health care seeking genetic counselling. There is therefore a need to understand more about how this potential patient group understands genetic risk.

Several studies have investigated whether research participants wish to be informed about genetic risk. These studies indicate that the overwhelming majority of individuals participating in research, and of members of the public, want to receive information about individual genetic findings. Respondents emphasize the desirability of information that a) indicates a high risk of developing disease, b) concerns serious diseases and c) can be acted upon [4–8]. Since such characteristics (high risk, disease severity and actionability) vary between different incidental findings, the desire to get feedback about results is expected to depend on the research context. In one study, willingness to receive information about a grave but preventable condition with a

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Table 1
Description of the SCAPIS programme.

The SCAPIS (Swedish CArdioPulmonary bioImage Study) research programme is a population study involving extensive measurements of 30,000 Swedes aged 50–64. The aim of the project is to find risk markers that allow prediction of who is at risk of cardiopulmonary disease, and prevention of this disease before it occurs. The study is a collaboration among six university hospitals in Sweden, funded primarily by the Swedish Heart-Lung Foundation [2].

90% probability of incidence decreased from approximately 96% to 62% when the risk of incidence fell to 1% [6].

The characteristics of genetic information that research participants value the most are thus reasonably well known. How well research participants or patients can recall genetic risk information and whether they understand the probability notion of genetic risk have also been studied [8–10]. It has been pointed out that people in general have a limited capacity for grasping and assimilating the notion of risk in terms of the probability of an undesired event [11–13]. Also, research has emphasized that participants may have their own lay understandings of genetic risk that diverge from scientific notions related to numeric probabilities [14,15] and that their perception may be shaped by their own life situations, such as their experience of disease [16,17], their attitudes and beliefs, and their psycho-social situations [18,19]. However, less is known about their own ways of conceptualizing genetic risk. As a complement to studies that investigate how well people understand and recall genetic risk in the probabilistic sense, and to studies on psycho-social consequences of genetic risk information, we therefore want to explore how participants themselves make sense of genetic risk. In this study, our aim is to investigate what genetic risk information means for the individual, i.e. how they make sense of the information within their own lives. For this reason a phenomenographic methodological approach was selected.

2. Method

2.1. Design and theoretical framework

A phenomenographic approach was chosen to explore research participants' conception of genetic risk. The purpose of this approach is to map the qualitatively different ways in which they think about, conceptualize and understand phenomena in the world around them. A phenomenographic approach focuses on people's thought content and its variation among individuals. The variation in their views are fundamental to understand their various ways of acting, forming beliefs and experiencing the world [20]. Thus, in this study, we sought to investigate the phenomenon of genetic risk as conceived by research participants.

2.2. Participants

Sixteen research participants with experience of taking part in SCAPIS (see Table 1), a research programme aimed at finding biomarkers for cardiopulmonary disease, participated in this study in four different focus groups (consisting of: 4 women and 2 men; 2 women and 2 men; 2 women and 1 man; 1 woman and 2 men respectively). The demographics are given in Table 2. To obtain a wide range of viewpoints in the discussions, we tried as far as possible to recruit participants representing all ages of the programme, with both high and low levels of educational attainment, and with and without prior experience of disease and genetic diagnosis. One or two in each group had prior experience, themselves or through a close relative, of severe diseases requiring extensive treatment, e.g. cancer or Parkinson's disease. Only one informant had experience of a close relative living with a monogenetic disorder (Huntington disease). The study population for SCAPIS is a representative sample of the general public (50–64 years of age). In our study, however, they are of interest as healthy participants in research that is expected to generate risk information about them.

Table 2
Demographic characteristics of research participants (n = 16).

Background variables	*n*
Sex	
Female	10
Male	6
Age (years)	
50–54	3
55–59	4
60–65	9
Education level	
Compulsory school (9 years completed)	2
Upper secondary school (12–13 years completed)	5
Post-secondary and/or higher education	9

2.3. Data collection

Data were collected in March and April 2016 by the first (JVJ) and the third author (UHU). The focus-group interviews were recorded. The interviews were held primarily by the first author. The third author engaged in the interviews, when necessary, by asking follow-up questions. The interviews lasted 47–77 min each and were conducted in Swedish. The interviews were conducted in a familiar room at the hospital, visited by the informants when they first joined SCAPIS. The interviewer emphasized that there were no right or wrong answers, just different perspectives on the issue. To enhance the participants' confidence and put them at ease, each interview began with asking why they had chosen to participate in the SCAPIS research programme and how they had experienced the extensive testing. A semi-structured interview guide with open-ended questions was developed with recommendations from Krueger and Casey [21], see Table 3. Probing questions were also asked, to explore specific topics expected to have a bearing on the research participants' general understanding of the concept of genetic risk. For example, if an informant said that it is important that disclosed information is about something real, the interviewer asked a probing question like "What do you mean by 'real'?" and asked the informant to describe a real genetic risk.

Prior to the focus-group interviews, one pilot interview with seven interviewees (colleagues and friends) was conducted to assess whether the questions were appropriate for stimulating discussion and enhancing reflection. Minor adjustments to the order of the questions were made. Group size underwent a major change: we decided to make the groups smaller ('mini focus groups' as Krueger and Casey [21] call it), to make it possible for all the informants to better share their personal experiences and thoughts about the topic.

2.4. Analysis

The recorded interviews were transcribed verbatim by a professional transcription company. The first author (JVJ) then

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