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“Naked genes” as health psychological innovation: A challenge for research and intervention

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Abstract

The identification of hereditary breast/ovarian cancer predisposing genes enabled the introduction of genetic testing in clinical practice. This created new psychological scenarios related to the many choices these women face. To explore the psychological effects of BRCA1/2 test on Italian healthy women, the following themes were investigated: childbearing intentions, feelings about the future and future projects, family support, feelings towards children and partners, risk perception, and attitudes towards risk management strategies. The participants were 21 young women (average age 35 years) who had undergone BRCA1/2 testing, recruited from a hospital in a medium city in northwestern Italy. A qualitative approach was chosen to explore these complex and sensitive issues. In depth interviews were developed after a review of the literature. The interviews were transcribed verbatim and were analysed using NVivo9. The analysis of the interviews highlighted overall good adjustment to the test results. However, some women underlined a need for long-term psychological support. The main issues were: awareness of psychological support, managing uncertainty, impact on life planning, childbearing intentions, and choice of prophylactic surgery interventions. These results may enable better understanding of the experiences of Italian women who underwent BRCA1/2 testing in order to develop effective interventions in the area of support and care.

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1. Introduction

About twenty years ago, the discovery of BRCA1 and BRCA2, two of the major genes that when mutated are implicated in the predisposition for breast and tubes/ovarian cancer, enabled the introduction of genetic testing in clinical practice. These genes represent a challenge for these women not only from a medical point of view, but also from a psychological one. When looking for a way to reduce uncertainty about their level of risk by undergoing genetic testing, women who receive a positive test result are paradoxically immersed in a situation that on the one hand eliminates some uncertainties, but eventually adds new ones to the other (DiMillo et al. 2013). Therefore this type of test discloses information about probabilities. The new technologies create the need for the individual to internalise an idea of inevitable risk 'to be where you have not yet been and where maybe you will never be' (Duden, 2006, p. 173). Within this condition, for some women the illness and the risk of illness may become the same (Aronowitz, 2007); the borders between the person with symptoms and those with no symptoms yet (Konrad, 2005) are fluid (Nowotny & Testa, 2012): the result of genetic testing leads the individual to identify with the diagnosis of "his/her being so", not something that "he/she feels" or that "he/she owns" but that "he/she is" (Duden, 2006). The World Health Organization defines health as the state of complete physical, mental and social well-being, and not only as an absence of disease (WHO, 1948). The self-perception of health, characterised by positive feelings toward oneself, a feeling of self-control and an optimistic outlook on the future, not only provides a leading force for coping with the difficulties of everyday life, but also with those stressful situations that threaten existence (Vazquez, Hervas, Rahona, & Gomez, 2009). In particular, the literature indicates that symptoms of anxiety, depression and stress related to cancer risk is higher among women with a family history of breast cancer, and that having a mother who died of cancer and a transmitted mutated gene leads to higher perceived stress, anxiety, and a lower quality of life (Wenzel et al., 2012). The present work seeks to understand how a positive BRCA1/2 test result affects the psychological well-being and the planning of future life. Furthermore, few many studies with qualitative approaches have addressed these issues; in particular the themes of our study have emerged from more extensive research involving other dimensions which have not been specifically studied. One recent work explores the uncertainty that women carrying a BRCA1/2 genetic mutation experience (DiMillo et al., 2013); additionally den Heijer and colleagues (2011a; 2012) examined the impact of social and personal resources on psychological distress. However, little research has been done so far in Italy on the consequences of BRCA1/2 testing on young women and on the resulting need for psychological support. The aim of this work was, therefore, to access the experiences, behaviours and expectations for the future of women in order to analyse their specific needs for psychological support.

2. Methods

A qualitative approach was appropriate for exploring these complex and sensitive issues. In particular, several studies have been conducted using qualitative procedures based on Grounded Theory (Glaser & Strauss, 1967) in the field of genetic counselling for BRCA1/2 (DiMillo et al., 2013; Grubs & Piantanida, 2010; McAllister, 2001). An in-depth interview was developed after a review of the literature and from previous personal experience of the research group. Questions were fine-tuned during a pilot interview.

2.1 Participants

Eligible participants were women aged 18–40 years who had undergone genetic testing in the preceding 10 years for a pathogenic BRCA1 or BRCA2 mutation. Participants may have had a family history of cancer (mother, sister, aunts ill or died from cancer) but no personal cancer history. Participants were recruited from a hospital in a medium city in northwestern Italy. Eligible women were considered by their clinical team to have no mental health contraindications, and were contacted more than 7 months after their BRCA1/2 test result. A total of 36 eligible women were identified and contacted, of which 21 women joined the project. The average age of participants was 35 (range 24–40).

Data collection and analysis

Women were invited to participate via a telephone call in which they were asked for permission to be sent an e-mail outlining the aims of the study. This was followed by a phone call to verify acceptance and eventually arrange the date of the meeting. Each interview lasted between 30 and 120 minutes. Data were collected at the participants' homes or a location of their choosing in order to increase participation, create a positive climate and put the subject at ease. Participation in the study was voluntary; anonymity and confidentiality were guaranteed. The informed consent protocol was provided to the subjects while presenting the aims of the research and before the participants started the interview. The Ethics Committee of the hospital approved the study. Each interview was audio recorded and transcribed verbatim. The names of participants were modified to preserve their privacy. The research group analysed the study material: the interview results were analysed and compared for common themes using NVivo9 software based on a Grounded Theory approach (Glaser & Strauss, 1967). A thematic analysis of the transcripts was conducted according to an inductive theoretical framework. Two researchers coded all the interview transcripts privately and independently of one another. They used a coding scheme in which they looked for emergent themes, or recurring domains of meanings across the interviews (Grubs & Piantanida, 2010; Lofland & Lofland, 1995; Rossman & Rallis, 1998). The analysis involved a multi-step process that integrated procedures suggested by Strauss and Corbin (1998): a coding scheme was developed inductively from initial readings of the transcripts; this coding scheme guided the systematic review of the qualitative data and organisation into analytical categories.

3. Results

The present work explores the psychological effects of BRCA test on healthy women. The themes that emerged from the interviews are different and include several dimensions of well-being in women's processes. In this paper we focus on the aspects and significant moments in which women have highlighted the need for psychological support in order to cope better with the condition of uncertainty by being a BRCA1/2 mutation carrier or by being part of a family at high breast/ovarian cancer risk. The participants of this study with positive or negative results described three main effects of genetic testing on the planning of their future life: a) some women affirmed that the mutation has had no effect on the planning of their lives, particularly those with a positive result stating they do not consider it acceptable that their lives can be influenced by a mutated gene; b) some women admitted that the result had effects on the planning of their lives, particularly those with a positive result stating they have planned for the future taking into account the possible occurrence of the disease; this strategy enables them to more readily react in the case of illness; c) the last group of women argued that the test result was disadvantageous in planning their life, because planning for the future is perceived as a futile effort since the disease could destabilise their desired plans. This is considered particularly important by women who, despite having had a negative test result, have been deeply and psychologically marked by a family background of cancer disease. The analysis of the interviews highlighted, in general, overall good adjustment to the test results both for women with a positive or negative result. In fact, it seems that more than the test result, a family history of disease, lived as close in particular the disease of mothers, leads to thoughts about cancer and uncertain futures. However, some women underlined a need for long-term psychological support. The main issues in need of support were: awareness, uncertainty, impact on life planning, childbearing intentions, choice of prophylactic surgery (Tab.1).

Tab. 1 Categories and quotations illustrating participants' actual views regarding psychological support

Categories	Sub-categories	Representatives quotes
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Need psychological support	awareness	<p>Lucia (36 years old): <i>"Make a lot of psychological support for people who are ill is right but also for the people who have family histories so important (holds back tears) the psychological support would be important. I think it is right to propose psychological support because maybe they don't realize the need for it, but make sure that there is also a path for them."</i></p> <p>Maria (36 years old): <i>"I think it would take a psychological pathway, psychological help to really understand our moods because many times, as I said, life is hectic, banging here and there, do not really know what you have inside. In fact, we should stop a moment and think, now let's talk about this thing"</i>.</p> <p>Sandra (40 years old): <i>"Often, maybe you give importance only to the aspect of the disease, that is, though, as in all things, the psychological aspect is crucial, so how we deal with things, I think it is. That is, it is also important to give importance to these aspects that perhaps things may seem superficial analysis of secondary importance, whereas they are not"</i>.</p>
	uncertainty	<p>Elisabetta (35 years old): <i>"Having more knowledge can help you prepare mentally for the bad things that can happen, preparation, psychological support"</i>.</p>
	impact on life planning	<p>Paola (29 years old): <i>"The psychological support was useful, that is, did not make me change my mind, but it put me a little in front of all perspectives. So, I said to myself 'Let's think carefully because it could be that ...'. The psychological support put me in front of all the real possibilities that could have happened. I thought if I could feel myself accepted by others. I thought if I could feel myself accepted by others if others knew that (I have the modified gene)...could I have told them the situation or could I not have told them the situation"</i>.</p> <p>Lucia (36 years old): <i>"This support is a different kind, certainly less serious. Until you're not sick surely you have psychological support need in a different way; it's not that you do not need, however, is not to be underestimated the impact these modified genes have on your life certainly..."</i>.</p>
	childbearing intentions	<p>Paola (29 years old): <i>"could I have a son knowing that I have this mutation? The psychological support had put me in front of all the possibilities and then I think it's been helpful. Then in spite of this I have not changed my mind because now my inner choice I had already made"</i>.</p>
	choice of prophylactic surgery	<p>Maria (36 years old): <i>"I told you, I mean, it would be even nicer to be able to deal better with this situation, make a psychological pathway, because, you know that any psychological help, I think it would be a beautiful thing, a positive thing, but .. one more thing .. that helps."</i></p>

Because, however, test choice of prophylactic surgery is a trauma ... it is something that perhaps is bigger than you can imagine, than you are, what you will and also it takes psychological support. However, it is necessary to continue to be followed in time by a psychologist, not just after the surgery and then just end game however good or bad it would take a path of control”.

Antonella (40 years old): "A small glimpse of someone who says if the person might have understood or less what might be the implications (both prophylactic surgery than on maternity), because once the things you know, you know, and at that point one can have underestimated”.

Discussion

The resulting data suggest that the genetic test result drags women into a situation of great uncertainty. Initially, many women made the decision to undergo genetic testing in order to obtain a response about their state of health. However, they later realised that even though they received a confirmation of a genetic predisposition to breast/ovarian cancer, they have been plunged into a new situation of uncertainty. This pattern emerges also in the case of a predictive negative test result. The family history and the statistical probability, as explained during the consultation, represent a psychological framework in which women are embedded. In addition, women who having a negative test result, they don't will insert in prevention path by clinician but left alone them with their family history of cancer strongly impacting on their psychological experience. Instead, for some, which have a positive result, can have the possibility to be followed by clinicians in a prevention path, and this made them more reassured. As also observed by and colleagues (2013), some women live with fear constantly waiting to receive the "bad news" for this reason, thinking in advance about the possible effects that an oncological disease could have on their lives. Uncertainty pervades decisions about preventive actions, and the burden of responsibility for these choices turns out to be very "overwhelming", as anticipated by Klitzman and Chung (2010). Moreover, the analysis of the interviews underlines the presence of a specific anxiety related to the possibility of developing cancer, as theorised by Meiser and colleagues (2006-2007). Despite a positive or negative test result, family history of disease (Hoskins, Roy, Peters, Loud, & Greene, 2008) has stronger repercussions on daily life and future prospects. Many women say they are scared of developing cancer, and this specific distress has also been identified by Beran and colleagues (2008). A high perception of risk for contracting cancer and the well-being of sons are the strongest motivations that encourage the decision to undergo prophylactic surgery interventions (Bebbington Hatcher & Fallowfield, 2002; Fang et al., 2003; Fry, Rush, Busby-Earle, & Cull, 2001; Hallowell, 1998; Miller, Fang, Manne, Engstrom, & Daly, 1999). The problems emerging from the research participants underline several points of contact with the findings of Hallowell (1998): in particular women feel they need to know in advance the physical and emotional consequences of the surgery, because the interventions are going to affect the natural mind-body balance.

4. Conclusion

Receiving a BRCA test result is an important experience in a woman's life with lifelong implications that represent a challenge for research and intervention in order to promote operators' reflections on need for specific support. The present study aimed to investigate how the test result impacts the future lives of these women. According to several authors, the complex psychological and existential issues related to the experience of having to adapt to a genetic diagnosis is going to always appear more frequently in clinical practice (Farkas-Patenaude, Guttmacher & Collins, 2002; Lerman, Croyle, Tercyak, & Hamann, 2002; Zinzi, 2004). In this framework, multidisciplinary health care equips (Hoskins et al., 2008), psychosocial interventions or support groups for women at risk of hereditary breast cancer should be present not only during the path of genetic counselling but also later in the different phases of decision-making related to prophylactic and preventive strategies (Hatcher & Fallowfield, 2003; Wevers et al.

2012). In this regard, the development of test protocols that take into account a range of possible differentiated cultural backgrounds and beliefs related to health and medical culture in various social groups could be fruitful for research and intervention (Meiser et al., 2006-2007). In addition, the social support of people outside the family could be a valuable ingredient in the path of these women reinforced through the perception of being fully understood (Bebbington et al., 2002; den Heijer et al., 2011b; Kenen, Shapiro, Friedman, & Coyne, 2007).

By the end of the 90s, the American Psychological Association (APA) set up a special committee, the Advisory Board on Genetic Issues, and has recently emphasised the need for more active involvement of researchers and psychologists in the field of human genetics and its applications from a multidisciplinary perspective. On the clinical side, according to the authors, interventions should focus, in particular, on self-esteem, feelings of stigmatisation and isolation, and should adapt to individual needs and characteristics (den Heijer et al., 2011a).

This study represents a first attempt towards an enhanced understanding of these themes within the context of Italian young women, an understanding of well-being processes that may lead to considerable improvements in the social and psychological support strategies and in the quality of life of BRCA mutation-positive members of mutation positive families with breast/ovarian cancer family history. Furthermore, the paper also contributes to the psychological needs of women with a negative test result who seem to need support regardless of the outcome, but for the family history of the disease that has strongly impacted them. In contrast to patients who are fighting cancer, the nature of the genetic predisposition BRCA1/2 naturally generates uncertainty. The support of the operators is therefore valuable to boost the resilience of these women in order to help them deal with more serenity with everyday life, and to cope with the discomfort that involves the feeling of living under a sword of Damocles (DiMillo et al., 2013).

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