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Changes in life decisions and in emotions following genetic testing for cancer risk

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Abstract

Genetic testing is essential for understanding the onset of advanced cancer. Women who tested positive for a *BRCA1/2* pathogenic variant experience changes in their everyday life, affecting their emotions, behaviors, and social relationships due to their "at risk" status. Moreover, a new sense of urgency arises when planning future decisions. This study aims to explore the psychological experiences of women who have undergone genetic testing through a qualitative thematic analysis. Nine Italian women who received *BRCA1/2* risk information participated in this study. Semi-structured interviews were conducted by expert psycho-oncologists. All the data were collected online via a web conferencing platform, using both video and audio. Interviews explored two areas: the first focused on emotional changes following the genetic testing results and the second is focused on the changes in daily life and personal decisions. In the first area, two themes related to negative and positive emotions and their related eight sub-themes emerged. In the second area, two main themes related to personal and social changes, which were further divided into five sub-themes, were identified. Future studies could explore how to better address emotional and behavioral challenges faced by *BRCA1/2* carriers, by providing adequate psychological support and improving doctor-patient communication.

Keywords BRCA mutation, Genetic testing, Emotions, Cancer, Cancer screening, Decision-making

Introduction

Genetic testing (GT) is rapidly advancing across the globe, equipping individuals with unprecedented insights into their biological predispositions and refining our collective understanding of personal vulnerability to disease. Literature reported that GT can be helpful to improve quality of life, thanks to increasing sensitivity training for professionals involved in testing, emotional and financial support, and additional educational resources [1]. From a psychological perspective, individuals' responses to GT results vary widely, and while some people are motivated to learn about their genetic predispositions, others may experience anxiety or apprehension about potential health outcomes. Negative emotions are commonly associated with GT for serious illnesses, particularly when individuals are uncertain about how to interpret the genetic information [2–4]. Different studies highlighted that people often feel psychologically unprepared to receive unfavorable news from GT [3]. For instance, oncological research on GT has found that women may both overestimate and underestimate their cancer risk, complicating their perception of risk [5]. In this regard, negative reactions to GT could be related to the patients' experience of a disintegration of their identity after unfavorable news. Women need to build and reintegrate a

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new identity, characterized by being a *BRCA* carrier, which requires great efforts on a psychological level [6]. Nevertheless, understanding GT and its implications is an essential first step towards heightened awareness, potentially raising additional concerns. This reframing of risk perception, along with the transition to a "not-yet" patient status, significantly redefines traditional concepts of disease and health.

An example of GT for disease susceptibility is Breast and Ovarian Cancer (*BRCA*) testing, which helps determine the likelihood of developing cancer based on genetic factors. Women who carry *BRCA1/2* pathogenic variants (PVs) face a significantly elevated risk of developing breast cancer before the age of 80 (69–72% risk), as well as a higher risk of ovarian cancer (39–44% risk for *BRCA1* and 11–17% risk for *BRCA2* PVs), compared to non-carriers [7, 8]. Recent studies have highlighted both negative and positive aspects of such awareness given the relevant impact of emotion on cancer patients' quality of life [9]. Negative aspects include higher levels of anxiety and distress among carriers - compared with non-carriers - due to the perception of an imminent future disease and its implications for both the individual and their family [10, 11]. However, carrying a PV does not necessarily guarantee the onset of disease, although it can influence life decisions and emotional well-being. A key factor in how individuals experience living with a *BRCA1/2* PV is whether they have already been diagnosed with cancer and undergone related treatments [12]. In Italy, genetic counseling for *BRCA1/2* PVs carriers is included in the public healthcare system, where patients receive both pre- and post-test consultations. These sessions provide information about test results, potential implications, and emotional support. However, the process may vary by region, leading to differences in access to timely counseling, which can influence psychological outcomes and decision-making about life changes, such as risk-reducing surgery [13].

Building on this theoretical background, the present study aimed to qualitatively explore the thoughts, emotions, and changes in daily life of Italian women who had been diagnosed with cancer and tested positive for a *BRCA1/2* PV. Using qualitative thematic analysis, the study investigated women's experiences with GT, with a particular focus on changes in emotions and daily life and personal decisions.

Method

Participants

This study involved nine Italian women who tested positive for a germline pathogenic or likely pathogenic variant in *BRCA1/2* (see Table 1). Eight additional individuals declined to participate in this study due to personal reasons or unavailability (47%). The majority of participants ($n=7$) tested positive for the *BRCA1* gene variant, while two tested positive for the *BRCA2* variant. Only one participant was found to carry both *BRCA* genetic variants. All participants had previously been diagnosed with breast cancer. Their ages ranged from 43 to 61 years (M_{age}=50.55, SD_{age}=5.59) and most had a high level of education, with six holding university degrees. Seven participants were in a relationship and lived with their partner.

Data collection and procedure

Four researchers (I.D., V.S., G.O., and S.O.), all with extensive experience in qualitative research, conducted in-depth online video interviews to explore the experiences of participants after receiving a positive *BRCA1/2* PV result. The women were recruited from an Italian hospital, where they had previously undergone GT. A purposive selection method was used, based on the following inclusion criteria: participants had to be (a) at least 18 years old, (b) have received a positive genetic test result for a *BRCA1/2* PV, and (c) be able to speak and understand Italian. Exclusion criteria included psychological impairments that would prevent participation,

Table 1 Socio-demographic information of participants

ID (Hypothetical name)	Age	Educational level	Marital status	Child/Children (*n*; sex)	Genetic mutation	Familiarity with oncological diagnoses	Type of oncological diagnosis they have	Year of their first oncological diagnosis
#1 Elisabeth	50	University degree	Married	No	BRCA1, BRCA2	No	Endometrial cancer	36
#2 Lisa	58	High school diploma	Maiden	No	BRCA2	No	Endometrial cancer	44
#3 Leslie	46	University degree	Married	Yes (2; f, m)	BRCA2	Yes, first degree	Breast cancer	35
#4 Madeline	50	University degree	Married	Yes (2; f, f)	BRCA1	Yes, first degree	Breast cancer	28
#5 Margaret	61	University degree	Married	Yes (1; f)	BRCA1, BRCA2	Yes, first degree	Breast cancer	47
#6 Madison	49	University degree	Maiden	No	BRCA1	No	Breast cancer	39
#7 Paige	49	High school diploma	Married	Yes (2; m, m)	BRCA1	No	Breast cancer	42
#8 Sarah	49	University degree	Married	Yes (1; f)	BRCA1	Yes, second degree	Breast cancer	33
#9 Sabrina	43	High school diploma	Married	Yes (2; f, m)	BRCA1	Yes, first degree	Breast cancer	39

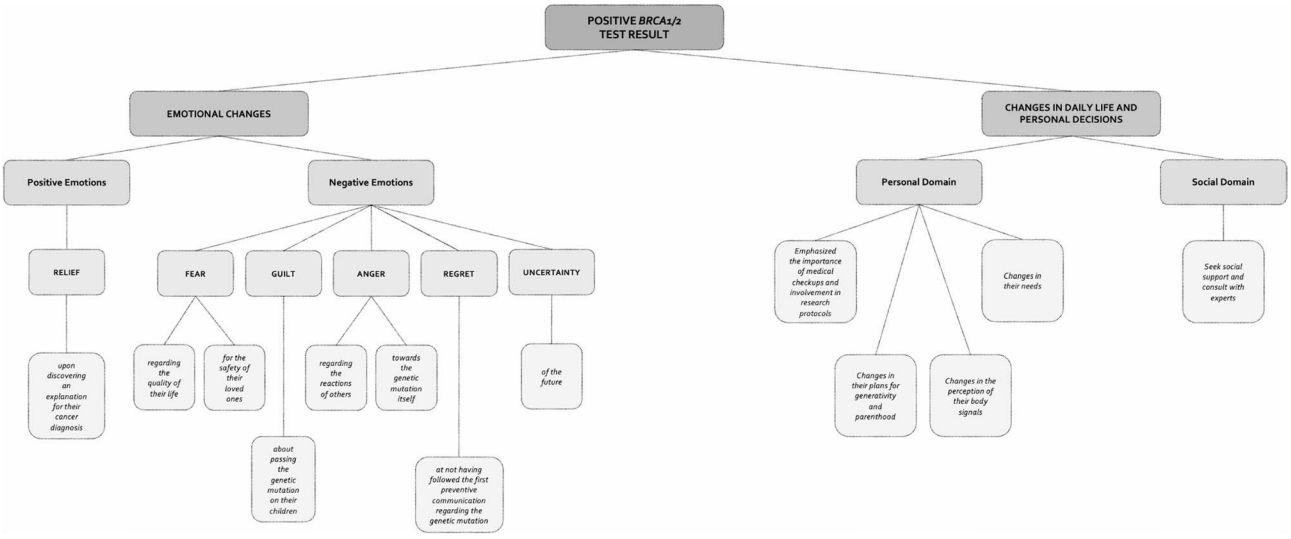

Fig. 1 Themes and sub-themes related to the impact of positive genetic test results on participants' lives

such as an inability to understand the study or sign informed consent. Women who met the inclusion criteria were contacted by telephone to assess their interest in participating in the present study. If they voluntarily agreed to participate in the study, a follow-up email was sent, containing an information sheet, an informed consent form to be signed, and a link to the online interview. Participation was entirely voluntary, and no incentives were provided. All participants were assured that their involvement would remain anonymous, and that the recorded data would be handled confidentially. Before each semi-structured interview, researchers collected socio-demographic data and explained to participants the research goals and the interview structure. The interview structure was refined through role-playing with two authors (I.D. and G.O.). All the data were collected online via a web-video conferencing platform between July and August 2022, using both video and audio. Interviews lasted between 30 and 70 min. The interview guide is provided in detail in the Supplementary Material. All semi-structured interviews were audio-recorded for transcription and analysis by expert researchers in the psycho-oncological field. We conducted a number of semi-structured interviews that allowed us to better explore the area of study, leading to adequate coverage of the themes and sub-themes related to changes in emotions, daily life, and personal decisions after receiving *BRCA1/2* PV [14, 15].

Data analysis

Data analysis was conducted by four researchers (I.D., V.S., G.O., and S.O.). A qualitative thematic analysis with a primarily inductive, "semantic" approach was used to identify the main themes, focusing on the meaning conveyed in participants' accounts [16]. This method enabled the identification of key themes that emerged from the transcripts and related to the two main areas explored in this study: emotional changes, and changes in daily lives and personal decisions. Themes and subthemes related within these two areas were explored using a data-driven approach, following the Braun and Clarke model [17]. Specifically, five steps were followed. First, the authors familiarized themselves with the contents by reading the transcripts multiple times. Second, initial data coding was conducted, and any discrepancies between raters were resolved through discussion between authors until an agreement was reached. Codes were used to identify segments of the textual reports (semantic content). Third, the codes were organized into corresponding themes and subthemes. In the fourth phase, the themes were reviewed, and a thematic map was developed. Finally, all authors reviewed the thematic map. In addition, one researcher who had not participated in the previous phases independently reviewed the entire process and the identified themes to further validate the thematic map.

Themes and subthemes were identified through repeated meetings among researchers. Quotes were chosen from text excerpts in order to illustrate emerging themes.

Results

In this study, two main areas were explored: emotional changes and changes in daily life and personal decisions. In the first area, thematic analysis revealed two themes related to emotional responses (negative and positive emotions). These themes led to the identification of eight sub-themes. In the second area, the analysis identified two main themes (personal and social changes), which were further divided into five sub-themes (Fig. 1).

The themes and sub-themes are discussed below, and women's quotes are labeled with ID numbers.

Emotional changes

The first area explored participants' emotional changes upon receiving a positive *BRCA1/2* test result. The first theme that emerged from the data is related to negative emotions. Specifically, some participants expressed *fear regarding the quality of their life* due to the awareness of carrying a genetic mutation that increases their risk of developing cancer. For some women, this knowledge can be an emotional burden that hinders their personal goals and plans. The constant reminder of their cancer risk, along with frequent medical check-ups, led to feelings of fear and anxiety. For example, Leslie, a 46-year-old woman with a *BRCA2* PV reported: "*there is always something that reminds you of this risk, and therefore it is not easy to manage this burden*" (ID#3). This constant vigilance regarding their higher cancer risk contributed to fear and anxiety as the primary emotional responses. Some women expressed feelings of uncertainty, such as: "*now the worst possible scenario will happen*" (ID#8) or "*since I discovered this genetic mutation [...] I have anxiety and feel a little down because I have made plans for my life and I do not know how long my life will last*" (ID#7). Additionally, four participants expressed *fear for the safety of their loved ones*, especially their children. In fact, relatives of individuals with a *BRCA* mutation are strongly encouraged to undergo genetic screening themselves, given their increased risk of carrying the same variant. This awareness generated a deep emotional response in the participants, who often felt personally responsible for the potential transmission of the mutation. For example, Sabrina, a 43-years-old woman, shared: "*when I discovered the genetic mutation, I cried because I immediately thought of my children. I have a daughter, and I was afraid of being able to pass this mutation on to her. This made me feel bad*" (ID#9).

Beyond fear, some participants also reported feeling *guilty about passing the genetic mutation on their children.* This guilt often intensified when their daughters were still young or had already entered a genetic surveillance protocol. For example, Margaret, a 61-year-old woman with a *BRCA1/2* PV said: "*The only thing that distresses me the most right now is the fact that I may have passed this genetic mutation on to my daughter. This is something that really bothered me. This is truly terrible [became tearful]*" (ID#5).

In some cases, the choice to undergo medical examinations and preventive measures was not well understood by others, leading to feelings of *anger regarding the reactions of others* in one participant. One woman with a *BRCA1* mutation reported: "*Someone called my sister crazy for deciding to have a preventive operation. Someone said: If we are not sick, why remove the breasts and ovaries? This bothered me. It is a strong criticism as if this problem did not exist*" (ID#7). Additionally, two women reported *anger towards the genetic mutation itself* and the consequences this information had on their lives. As one participant stated: *"After my BRCA diagnosis, I felt angry. I do not know if it is okay to say that...but it's how I feel"* (ID#6).

Additionally, three women with a genetic mutation expressed feelings of *regret at not having followed the first preventive communication regarding the genetic mutation.* This delay affected the timing of appropriate screenings and preventive measures. For example, Madeline, a woman with a *BRCA1* mutation stated that: *"Upon receiving the BRCA diagnosis, I realized that the other doctor was right. I regret not listening to his recommendations because there may have been a protocol in the United States"* (ID#4).

Lastly, some participants emphasized the difficulty of navigating the *uncertainty of the future*. In the absence of clear medical guidance, women often felt doubt and confusion. However, connecting with others in similar situations, such as through patient associations, helped alleviate some of this uncertainty by providing both information and emotional support. Many women also sought further medical consultations and check-ups, to feel more in control of their health. Consequently, they remain highly vigilant for any bodily signals that might indicate potential health concerns, including signs of cancer. They reported the sensation of "*always being on the alert*" (ID#4) and "*continuously having in my head that I could get sick again and again*" (ID#6). Regular medical checkups and talking to professionals can help women regain a sense of control over their lives.

Interestingly, some women also experienced positive emotions after receiving their test results, which emerged as a second theme. Notably, participants recognized a *sense of relief upon discovering an explanation for their cancer diagnosis* and its recurrence within the family lineage. For example, Sarah, a 49-year-old woman who tested positive for a *BRCA1* mutation, said: *"When I opened that famous envelope on one side, I was relieved, so to speak, to see that there was a scientific reason*" (ID#8). Similarly, Lisa, a 58-year-old woman with a *BRCA2* mutation shared: "*It is like she justified what happened to me, and so I have come full circle*" (ID#2). This newfound understanding gave them a sense of control and improved their ability to comprehend the potential risks.

Changes in daily life and personal decisions

The second explored area was related to the changes that participants experienced in their daily routines and life decisions, leading to the identification of two main

themes related to personal and social domains. Within the personal domain, some participants *emphasized the importance of medical checkups and involvement in research protocols* after receiving their genetic test results. In certain instances, participants opted for prophylactic surgery as a proactive measure to mitigate the risk of developing cancer. Interestingly, one participant viewed these medical checkups as an opportunity to "*take time for herself and an occasion to spend time with family*" (ID#3).

Additionally, most participants recognized a *change in their needs* after receiving genetic information, reporting a heightened awareness of their physical, psychological, and emotional well-being. This often translated into lifestyle changes, such as healthier eating habits, increased physical activity, and greater attention to mental health. As one 49-year-old woman with a *BRCA1* mutation reported: "*I have completely changed my lifestyle, not that I had such a messy lifestyle, but I have changed my way of eating, and I am much more careful in my life*" (ID#6). Some participants noted that they began to take more time for themselves. For example, a 49-year-old woman stated, "*Discovering this genetic mutation helped me pay more attention to myself and my needs. […] Now, I dedicate more time to my well-being, for example, taking physical activity or walking with a friend*" (ID#7). Additionally, some women expressed the desire to focus more on personal fulfillment and positive experiences, such as planning enjoyable trips or maintaining meaningful relationships. For example, one woman with a *BRCA1* mutation stated: "*I change my relationship with others because when I see that some people hurt my well-being, I avoid them. I want to protect myself*" (ID#7).

For some women, testing positive also meant rethinking *their plans for generativity and parenthood*. As one participant expressed, "*ugly scenarios for my future as a woman and as a mother because that was my desire*" (ID#8). In some cases, this was also determined by the induction of early menopause resulting from prophylactic surgery. Lastly, some participants experienced *changes in the perception of their body signals*. For some, minor symptoms or physical discomforts became potential signs of cancer, triggering anxiety. In contrast, others described a heightened bodily awareness that allowed them to better manage their energy and promote overall well-being. For example, a woman with a *BRCA1* mutation stated that: "*I pay attention to some aspects of my body. This helped me to overcome all my physical problems because my body is full of energy*" (ID#6).

In terms of social dynamics, five women reported a tendency to *seek social support and consult with experts* to obtain psychological support and reliable, scientifically-based information regarding their genetic mutation. For example, a woman said: "*People near me are important to me. I looked for them. When I was in difficulty, I needed to be followed by healthcare professionals and not be left alone.*" […] it is essential "*to have people who support me from a psychological and a medical point of view*" (ID#8).

Discussion

This qualitative study highlights the challenges that *BRCA1/2* carriers face in managing the uncertainty linked to genetic information and their higher risk of developing breast, ovarian and other cancers. It also offers insight into the emotional, behavioral, and lifestyle changes that follow GT results, identifying two primary domains of change: emotional changes and changes in daily habits and personal decisions. *BRCA1/2* carriers in this study described a wide spectrum of emotions triggered by the awareness of their genetic risk. Negative emotional reactions such as fear, anger, and guilt emerged as prominent themes from the collected data. These emotions were not limited to concerns for one's own health but extended to fears for relatives, especially children. This finding is in keeping with the peer-reviewed published literature on the psychological impact of *BRCA1/2* PVs among cancer patients [18]. Concerns about the increased risk of cancer recurrence are identified in the literature as the primary emotional response among carriers, often accompanied in women by a sense of guilt over the potential transmission of the variant to their offspring [19]. The fear of passing on the *BRCA1/2* variant to their children often motivates women to undergo GT and, in some cases, to opt for risk-reducing surgery [20].

Uncertainty also emerged as a central emotional experience. Participants described feeling overwhelmed in the absence of clear medical guidance or sufficient scientific understanding. This uncertainty triggered different coping strategies, such as seeking additional information, undergoing frequent medical checkups, or connecting with others in similar situations. These behaviors reflect existing evidence that individuals use a range of strategies (avoidance, information seeking, or shared decision-making) to manage uncertainty [21]. Social connection and support played a crucial role for individuals who shared similar life experiences, fostering personal motivation and positive feelings of cohesion [22, 23]. Similarly, clear and empathic communication with healthcare professionals was deemed essential for *BRCA* care continuum [24]. Interestingly, the literature shows that the detection of a *BRCA1/2* PV impacts psychological well-being (in terms of increased distress and anxiety) shortly after receiving results, while returning to pretesting levels over time [25, 26].

Changes in daily routine and life decisions emerged as the second major theme of this study. Genetic risk awareness encouraged and sometimes forced *BRCA1/2* carriers to assume a strong sense of responsibility toward their

health. This was often marked by increased body awareness, adherence to regular medical checkups, and engagement in preventive measures, including risk-reducing surgery and participation in clinical trials. Implementing action to address emotional and life-style issues can often be perceived as strategies to regain control in the face of uncertainty and risk, thanks to a higher sense of empowerment [27]. Despite concerns about adverse psychological responses, *BRCA* carriers appear to adapt well, offering them the opportunity to prioritize self-care and increase focus on their well-being. Prior studies of the long-term effects of GT have shown an increased attention to overall health as the most commonly observed behavioral changes over time [28]. Such behavioral adjustments are typically driven by a heightened perception of personal vulnerability to disease [20, 29, 30]. However, for many women, these health-centered decisions may conflict with personal life goals, particularly around fertility and motherhood [31, 32]. Genetic risk awareness also affects reproductive decision-making. These findings are consistent with the previous literature highlighting the complex and confusing reproductive challenges faced by *BRCA* carriers, especially those of childbearing age. These challenges may arise due to prophylactic surgeries or the use of oral contraceptives and tubal ligation [33, 34]. A cancer diagnosis and awareness of genetic risk can also influence attitudes and decisions regarding relationships, pregnancy, and the use of preimplantation genetic diagnosis or prenatal screening. In some instances, this may compel women to abandon reproductive plans altogether, negatively affecting their psychological well-being [34]. For example, genetic test results can create a sense of urgency for couples to make future decisions concerning motherhood and partnership. The body itself may be reinterpreted not as a resource but as a source of risk and anxiety, often becoming the focus of increased medical scrutiny and emotional ambivalence [35, 36]. Given the complexity of these issues and the multifaceted impact of such decisions, *BRCA1/2* carriers often seek emotional and social support in addition to medical information from oncologists or genetic counselors, who serve as trusted sources of guidance [37]. Support in managing and coping with *BRCA1/2 PV* should also come from psychologists, family members, or other mutation carriers. Psychological support can help people better understand their inner world - fostering self-awareness and curiosity about themselves [38–40] - and assist in managing emotions. Tailored tools are needed to help patients navigate the many of the short-and long-term challenges associated with this information over time.

Interestingly, while negative emotions were predominant, a sense of relief also emerged in some participants. For several women, understanding the genetic basis of their cancer allowed them to make sense of their illness and familial disease patterns. Women who received GT results appreciated the opportunity to protect others by taking preventive measures for themselves and their families [4]. Moreover, literature highlighted the relevance to communicate the genetic mutation test results to their blood relatives [41]. Future studies should explore how *BRCA* carriers' emotions and feelings could influence their willingness to share risk information with family members, as well as their intention to adopt healthy behaviors and participate in cascade screening (i.e., the process of testing blood relatives of carriers). Cascade screening is a key strategy for reducing cancer-related morbidity and mortality through early detection and prevention [42]. There is ongoing debate about whether communicating genetic test results effectively motivates healthy behavior. While some studies have suggested that GT can promote positive behavior changes [28, 43], others have produced inconclusive findings [44].

Based on our findings and the published literature, psychological support for *BRCA* carriers should be strengthened. This support should adopt an educational approach, providing essential knowledge to help individuals cope with their genetic results in clear, accessible language - even for those without a medical background. Ideally, it should be complemented by counseling that promotes understanding and informed decision-making [45]. Integrating such support into the GT process may significantly improve patients' psychological outcomes. Creating a personalized psychological intervention, people could have the possibility to receive support for the management of their emotions and the elaboration of meanings perceptions of their *BRCA1/2* PV. In this sense, it is important to take into consideration the personal characteristics of people who carry GT and the possible impact that this result could have on their life. Younger women, for example, need to address life changes related to family expectations and life planning [46]. Moreover, psychological intervention must improve health literacy regarding GT, also addressing risk-reducing surgery as one of the topics of interest and its relevant consequences on body image and the overall quality of life [47, 48]. Each type of GT performed resulted in different oncological diagnosis and, therefore, physical and psychological consequences. In this way, the creation of a personalized psychological intervention is essential to capture the personal needs and preferences.

However, it is important to acknowledge several limitations of this study. This study involves a small group of Italian women carrying *BRCA1/2* PVs and may be subject to self-selection bias: women who were more open to discussing their experiences may also be those more proactive in coping with genetic risk. Furthermore, it remains unclear how these findings generalize to unaffected *BRCA* carriers or to male carriers, both of whom

warrant further investigation. Additional socio-demographic details would have been valuable, such as information about other index cases in the family, whether participants were taking pharmacological treatments for anxiety or depression. Similarly, it would have been relevant to explore whether participants were undergoing psychological treatment, which could influence their ability to manage emotions related to the GT process. Lastly, future research could explore patients' experiences using an Interpretative Phenomenological Analysis (IPA) approach, which would help clarify how individuals make sense of their genetic test results and their implications. Despite these limitations, this study offers insights into the psychological and behavioral consequences of *BRCA1/2* PV awareness among women affected by cancer.

Conclusion

This research study explored the experiences of a small group of Italian women carrying *BRCA1/2* PVs, focusing on how awareness of genetic risk affected their emotions, daily routines, and life decisions. The findings highlighted different individual responses, including emotional distress, changes in health-related behaviors, and reconsideration of reproductive choices. These results underscore the need for comprehensive support strategies (including clear communication, emotional support, and personalized care) to help *BRCA1/2* carriers manage uncertainty, navigate complex life decisions, and maintain psychological well-being in the face of inherited risk.

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s40359-025-03524-z.

Supplementary Material 1.

Authors' contributions

I.D. conceptualized the study and wrote the first draft of the manuscript. I.D., G.O., S.O. and V.S. contributed to the writing, data-collection, and data analysis. All authors edited and reviewed the manuscript. G.P. contributed to the important intellectual content and supervised the whole process. All authors read and approved the final version of the manuscript.

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Data availability

The dataset is available from the corresponding author upon reasonable request.

Declarations

Ethics approval and consent to participate

This study was approved by the institutional review board of the European Institute of Oncology (approval number R1249/20-IEO1314) and was conducted in accordance with the Declaration of Helsinki. All participants agreed to participate in the research and gave written and verbal informed consent before starting the interview.

Consent for publication

Not applicable.

Competing interests

The authors declare no competing interests.

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