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REVIEW ARTICLE

Psychological aspects, risk and protective factors related to BRCA genetic testing: a review of the literature

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Abstract

Purpose The primary aim of the present study was to conduct a systematic review of short-, intermediate- and long-term psychological effects, such as anxiety, depression and distress, on individuals undergoing genetic testing to determine BRCA1 and BRCA2 gene mutation. The different instruments used for the measurement of each construct were reported. In addition, risk and protective factors associated with psychological outcomes of genetic tests were explored.

Methods Bibliographic databases were searched for studies published over the period 1998–2018. Using the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) method, 21 articles were selected for the current review.

Results Overall, the collected data revealed rather diverse results, although most studies reported higher levels of distress, anxiety and depression in carriers, as compared to non-carriers. The two genders were not equally represented, with men constituting only 6% of the sample. Risk factors and protective factors that may influence psychological outcomes and adjustment to genetic tests are highlighted and discussed in this review.

Conclusions The increased risk of developing cancer associated with positive genetic testing results may be experienced as traumatic by many patients, although not all individuals with positive genetic testing results will experience increased distress. Hence, future studies should consider specific risk factors in order to select those who are more likely to be in need of psychological support. Finally, it is necessary to increase the number of male samples to better understand the male experience related to genetic testing outcomes.

Keywords BRCA1/2 mutation · Genetic counselling · Cancer risk · Psychological distress · Anxiety · Depression · Review

Introduction

About 10% of all cancer diagnoses are caused by hereditary susceptibility to breast cancer (BC) and ovarian cancer (OC) [1]. The two genes more involved in these types of cancer than others are BRCA1 and BRCA2 [2].

Among healthy women, the risk to develop breast cancer is from 46 to 87% with a BRCA1 mutation and from 39 to 84% with a BRCA2 mutation, while the risk for OC is from 39 to 63% for BRCA1 and from 16.5 to 27% for BRCA2 [3]. In healthy men, the risk of developing BC is 1.2% for BRCA1 mutation and up to 8.9% for BRCA2 mutation; however, the risk of developing prostate cancer is 8.6% for BRCA1 mutation and 20% for BRCA2 mutation [3].

When there is a recurrence of disease in a family, the test for the identification of BRCA1/2 mutations may be proposed because of the higher cancer risk; this predictive genetic test can identify asymptomatic individuals who carry this mutation [4, 5]. Since genetic testing may produce a significant psycho-social impact in people undergoing it [6], the genetic counselling for these people includes both bio-medical and psycho-social issues [7, 8]. Before testing, it is necessary to evaluate and confirm the likelihood of hereditary gene mutation; therefore, medical and familial histories are collected. After that, if there is at least a 10% likelihood, a blood sample or a buccal smear is taken. There are three possible results revealed by the testing: positive, negative or inconclusive [9]. A positive result means that the individual is a mutation

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carrier; conversely, a negative result means that the individual is not a mutation carrier. An inconclusive result indicates that no known BRCA1/2 mutations were found, but a gene variant was detected that needs further investigation [10].

The literature on psychological outcomes of genetic test disclosure is conflicting: Some studies have evidenced that mutation carriers showed an increased level of psychological distress, specifically, high levels of anxiety and depression [e.g. 11], while other studies have reported no differences in distress level between carriers and non-carriers [e.g. 12]. Further studies have found higher levels of psychological distress over time in non-carriers compared to carriers [e.g. 13].

Given the lack of studies about inconclusive genetic testing results [14], we focused our attention on mutation and non-mutation carriers. Hence, the main aim of the current review was to examine psychological short-, intermediate- and long-term consequences of BRCA1/2 genetic testing. To pursue this objective, we considered cross-sectional and longitudinal studies focusing on psychological distress, anxiety and depression levels in individuals undergoing genetic testing.

A further aim of this review was to analyse risk factors and protective factors that influence psychological outcomes and adjustment to genetic testing as determined by international studies.

Methods

Systematic literature search

For data collection, reporting and discussion, we followed the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) method [15, 16]. Bibliographic databases (Scopus and PubMed) were searched for studies published in the last 20 years (from 1998 to 2018), using the following keywords individually and in combination: BRCA, BRCA1\2, BRCA mutation, genetic counselling, distress, emotion, psychological distress, anxiety and depression. The electronic search was supplemented with a manual search of reference lists in all publications found in order to identify all studies relevant to the current review.

Eligibility criteria

Studies were eligible for inclusion if they were written in the English language. For a structured approach, we assessed the eligibility criteria according to the following aspects: participants, intervention, comparison, outcome and study design (PICOS) [15].

1. Participants: We investigated human adult participants who undergo genetic testing to detect the presence of BRCA mutation.
2. Intervention: We did not focus on a specific intervention.
3. Comparison: We compared studies on BRCA mutation and non-mutation carriers.
4. Outcomes: We analysed studies that considered short- (less than 6 months from genetic test disclosure), intermediate- (from 6 to less than 36 months from genetic test disclosure) and long-term (from 36 months onwards) psychological outcomes.
5. Study design: We included observational, qualitative and quantitative studies.

Exclusion criteria were articles not written in the English language, articles not specific to BRCA mutation, literature reviews, letters to the editor, books, unpublished articles and doctoral theses, commentaries, abstracts of conferences, congresses and case-reports, and articles not reporting psychological outcomes.

Data availability The data is under control of the authors and is available on request.

Results

A total of 566 studies were screened for eligibility, and of these, 196 were identified as duplicates, and 303 were excluded because they did not meet the eligibility criteria. The abstracts of 67 studies were screened and evaluated, and of these, four were excluded because they were reviews, 35 studies did not focus on psychological outcomes, two referred to genetic mutations different from BRCA and five had an inappropriate study design. A total of 21 articles were included in this review (see flow chart in Fig. 1). These studies were published from 1998 to 2018 and were most commonly conducted in the USA and in Italy, but other countries were also represented (Spain, the Netherlands, France, Canada, Norway, Belgium, Australia and Sweden). Most studies were longitudinal (n = 13; 62%); the remainder consisted of cross-sectional studies (n = 8; 38%). Both cross-sectional and longitudinal studies were classified according to the timing of questionnaire administration (Table 1). Data was collected from 5565 participants (sample size of each study ranged from 21 to 2080 participants), and of these, 94% were women, and 6% were men. Of the reviewed articles, 17 reported the results of the genetic testing, which involved 4373 individuals. Among the reported results, 21% were positive for the BRCA mutation, 72% were negative, 6% reported inconclusive results and 1% were still waiting for the test results. In this review, we focused on psychological outcomes of mutation carriers and non-carriers.

Findings were divided into two categories: (1) distress and (2) anxiety and depression. We indicate the instruments used for the measurement of each construct in Table 2. The main

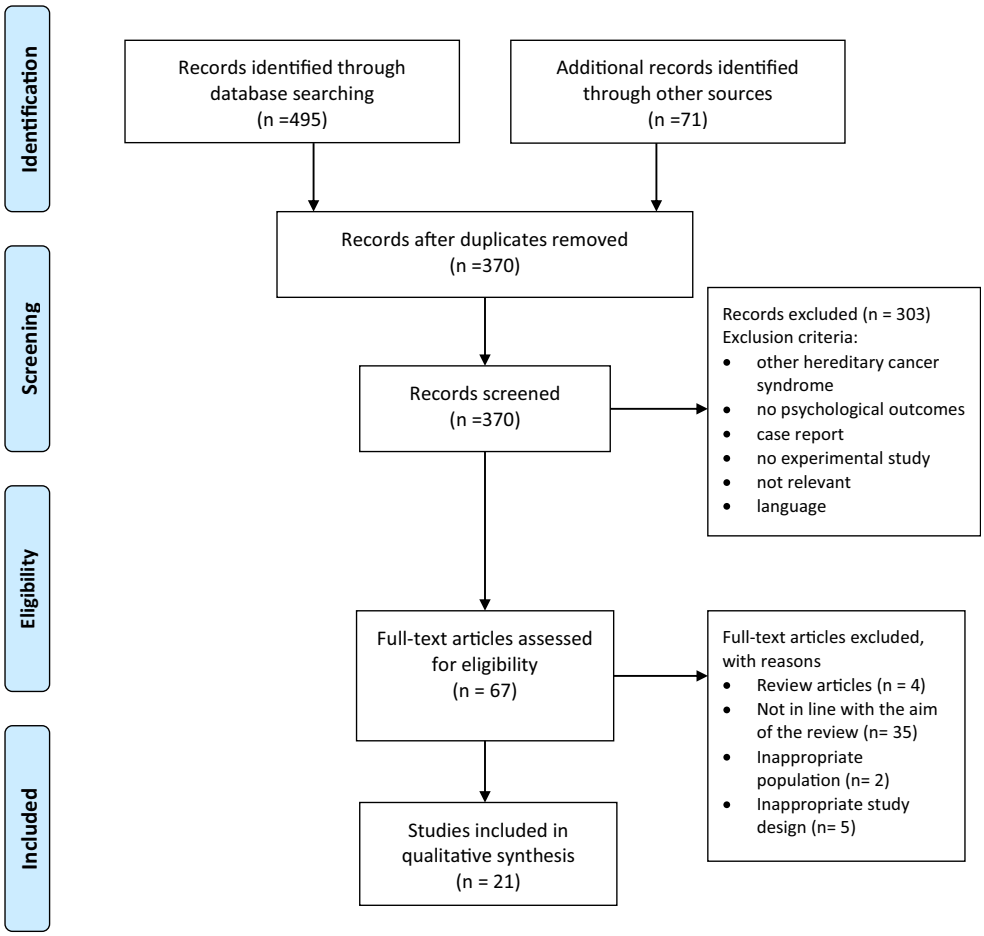

Fig. 1 Flow chart of selection and inclusion process, following the PRISMA statement

Note: PRISMA= Preferred Reporting Items for Systematic Reviews and Meta-Analysis

psychological findings of the considered articles are presented in summary form.

Distress

Seven of the included studies focused only on the measurement of distress [11, 17–22]. In addition, seven articles investigated both distress and affective states [1, 12, 23–27]. However, in this section, only the results concerning distress will be discussed.

The term *distress* refers to a negative emotional experience of a psychological nature which interferes with an individual's ability to cope with adverse life events [28]. All the reviewed studies measured the distress using the Impact of Event Scale (IES) [29], except for three studies [11, 19, 21] that, respectively, used the Psychological Symptoms Index (PSI) [30], the Hospital Anxiety and Depression Scale (HADS) [31] and the Brief Symptoms Inventory-18 (BSI-18) [32] (see Table 2).

The majority of the studies reported more increased levels of distress in mutation carriers than in non-carriers [11, 17, 18, 20, 22, 24–27], except for four studies that found no significant differences in distress levels between affected and unaffected participants [1, 12, 21, 23].

Some studies observed distress levels among mutation carriers in the short- and intermediate-terms [1, 11, 17–19, 21–27] and four of these detected increased levels of distress for up to 12 months. Meiser et al. [24] saw a specific rise in the first 10 days and hypothesised that this increase in cancer distress is long-lasting; Beran et al. [25] reported the highest levels at 1 month; Smith et al. [26] found greater levels at 3 months and Graves et al. [18] at 6 and 12 months after genetic testing disclosure. Only two studies observed long-term effects (more than 36 months after genetic testing results), detecting increased levels of distress among mutation carriers [11, 20]. Moreover, Claes et al. [23] investigated the course of distress over time as a function of the genetic test result and found that there was a significant decrease in time of cancer distress only for non-carriers.

In a study that considered only men, it was observed that the levels of distress before the result were low, and those who

Table 1 Time of questionnaire administration in longitudinal and cross-sectional studies

Author	Genetic test	Test disclosure	< 1 m	≥ 1 m	≥ 3 m	≥ 6 m	≥ 12 m	≥ 36 m
Arver et al. (2004)	+		+	+		+	+	
Beran et al. (2008)	+			+		+	+	
Bjørnslett et al. (2015)		+						
Bonadona et al. (2002)				+				
Borreani et al. (2013)				+			+	
Bosch et al. (2012)					+		+	
Cicero et al. (2017)		+						
Claes et al. (2005)	+						+	
Cukier et al. (2012)	+							
Dorval et al. (2008)	+			+			+	+
Graves et al. (2011)	+					+	+	
Kinney et al. (2005)	+			+	+		+	
Listøl et al. (2017)	+		+					
Lodder et al. (1999)	+							
Lodder et al. (2001)	+	+						
Meiser et al. (2002)	+		+		+		+	
Mella et al. (2017)				+				
Metcalfe et al. (2012)							+	+
Power et al. (2011)			+					
Smith et al. (1999)			+					
Smith et al. (2008)	+		+		+	+		

We used the symbol + to highlight the time of psychological questionnaire administration

Genetic test = at the blood test or buccal smear test

Test disclosure = at the result of genetic test

< 1 m = within 1 month after the genetic test result

≥ 1 m = from 1 to 2 months after the genetic test result

≥ 3 m = from 3 to 5 months after the genetic test result

≥ 6 m = from 6 to 11 months after the genetic test result

≥ 12 m = from 12 to 35 months after the genetic test result

≥ 36 m = from 36 months onwards

had daughters showed significantly more distress than those without daughters [19].

Regarding gender differences in levels of distress, overall, women reported higher levels of distress than men [18, 22]. Specifically, the study of Graves et al. [18] showed that carrier women reported higher levels of distress than carrier men before genetic counselling and 6 months after receiving test results, while no gender differences were detected at 12 months after genetic testing. Moreover, non-carrier women showed significant differences in comparison to men only before the genetic test.

In the study of Smith et al. [22], men showed high levels of distress only when they had a sibling with the mutation, while women always experienced adverse short-term psychological reactions after knowing the positive result of the genetic test regardless if they knew the sibling's test results. Another study observed that the distress experienced by individuals undergoing genetic testing is similar to the symptoms of post-traumatic stress disorder (PTSD), a condition that occurs in people who experience traumatic events during their lifetime [27].

In the reviewed studies, the following variables were discovered as potential risk factors: being the first subject tested in the family [17, 22], being unmarried [18], having a family history of breast/ovarian cancer [18], being in a family where one member has already received a positive result [33] and knowing of a sibling's positive test result [22].

It was also found that some protective factors can reduce distress levels over time in mutation carriers and non-carriers, such as having pre-test information and education [11] and

Table 2 Characteristics of included studies

Author (year)	Country	Aim	Study design	Sample size	Psychological measurements	Genetic test result
Arver et al. (2004)	Sweden	Investigate psycho-social consequences after genetic tests	Longitudinal	Total sample = 87 Women = 87 Men = 0	HADS; QoL	Positive = 31 Negative = 56 Inconclusive = 0
Beran et al. (2008)	USA	Investigate psychological impact of genetic tests	Longitudinal	Total sample = 155 Women = 155 Men = 0	CES-D; STAI; PANAS; IES-R	Positive = 38 Negative = 21 Inconclusive = 96
Bjørnslett et al. (2015)	Norway	Measure potential psychological distress	Cross-sectional	Total sample = 354 Women = 354 Men = 0	HADS; IES; MICRA	Positive = 32 Negative = 322 Inconclusive = 0
Bonadona et al. (2002)	France	Evaluate the consequences of positive results on genetic tests	Longitudinal	Total sample = 23 Women = 17 Men = 6	Semi-structured interview; MICRA	Positive = 16 Negative = 7 Inconclusive = 0
Borreani et al. (2014)	Italy	Describe the impact of preventive options on the psychological condition of *BRCA1*/*BRCA2* carriers	Longitudinal	Total sample = 79 Women = 79 Men = 0	HADS; Digital Body Photo Test; QoL; cancer worry scale; cancer risk perception; SF-12	Positive = 79 Negative = 0 Inconclusive = 0
Bosch et al. (2012)	Spain	Identify potential predictors of pathological anxiety at 3 months and 1 year after the genetic test	Longitudinal	Total sample = 364 Women = 312 Men = 52	HADS	Positive = 114 Negative = 60 Inconclusive = 117
Cicero et al. (2017)	Italy	Examine the relation between cancer risk perception and genetic risk before receiving genetic test results, considering the influence of some psychological variables	Cross-sectional	Total sample = 120 Women = 108 Men = 12	HADS; CRP; GRP; DT	Not specified
Claes et al. (2005)	Belgium	Evaluate distress and illness representations 1 year after test result	Longitudinal	Total sample = 68 Women = 68 Men = 0	Semi-structured interview; STAI; UCL; IES; Illness representation; SCL-90	Positive = 34 Negative = 34 Inconclusive = 0
Cukier et al. (2012)	USA	Examine the distress related to oncological disease and depressive symptoms	Cross-sectional	Total sample = 148 Women = 148 Men = 0	IES; CES-D	Not specified
Dorval et al. (2008)	Canada	Compare lifestyle behaviours and psychological distress between women initiating BRCA1/2 testing and women of general population	Longitudinal	Total sample = 640 Women = 640 Men = 0	Interview; PSI	Not specified
Graves et al. (2011)	USA	Describe and compare the psychosocial outcomes following the genetic test	Longitudinal	Total sample = 321 Women = 227 Men = 94	IES; BSI; MICRA	Positive = 123 Negative = 198 Inconclusive = 0
Kinney et al. (2005)	USA	Evaluate levels of psychological distress before and after receiving the test results	Longitudinal	Total sample = 85 Women = 57 Men = 28	CES-D; STAI; Cancer Worry Scale; IES	Positive = 19 Negative = 66 Inconclusive = 0
Listøl et al. (2017)	Norway	Evaluate how an education course about heredity breast and ovarian cancer can influence anxiety and depression levels	Longitudinal	Total sample = 100 Women = 100 Men = 0	HADS; BGCSES; TMSI	Positive = 100 Negative = 0 Inconclusive = 0
Lodder et al. (1999)	the Netherlands		Cross-sectional	Total sample = 85	HADS; IES; SAQ-N	Not specified

Table 2 (continued)

Author (year)	Country	Aim	Study design	Sample size	Psychological measurements	Genetic test result
		Identify subjects with high levels of distress before the genetic testing		Women = 85 Men = 0		
Lodder et al. (2001)	the Netherlands	Investigate levels of psychological distress	Longitudinal	Total sample = 21 Women = 0 Men = 21	Semi-structured interview; HADS; IES; Life Orientation Test	Positive = 4 Negative = 24 Inconclusive = 0
Meiser et al. (2002)	Australia	Investigate the long-term psychological impact of genetic testing, in carriers and non-carriers	Longitudinal	Total sample = 90 Women = 90 Men = 0	IES; BDI; STAI; Miller:	Positive = 30 Negative = 60 Inconclusive = 0
Mella et al. (2017)	Italy	Investigate the emotional states in association with affective states	Cross-sectional	Total sample = 89 Women = 89 Men = 0	HADS; POMS, Emotional Thermometers	Positive = 10 Negative = 79 Inconclusive = 0
Metcalfe et al. (2012)	Canada	Identify levels of distress related to the risk of developing cancer	Longitudinal	Total sample = 2080 Women = 2080 Men = 0	Study-specific Questionnaire; IES	Positive = 23 Negative = 2057 Inconclusive = 0
Power et al. (2011)	Canada	Examine levels of distress in people undergoing genetic test	Cross-sectional	Total sample = 318 Women = 286 Men = 32	History Questionnaire; Feelings about Test Results Measure; Psychosocial Needs Questions; BSI-18	Positive = 85 Negative = 51 Inconclusive = 33 Attending result = 49
Smith et al. (1999)	USA	Examine psychological distress among individuals tested for a *BRCA1* mutation and if it is influenced by their siblings' test results	Cross-sectional	Total sample = 212 Women = 125 Men = 87	IES; STAI	Positive = 75 Negative = 137 Inconclusive = 0
Smith et al. (2008)	USA	Evaluate psychological consequences of genetic test results	Longitudinal	Total sample = 126 Women = 126 Men = 0	GSI; IES; PSS; STAI; CES-D; QoL	Positive = 20 Negative = 67 Inconclusive = 13

HADS Hospital Anxiety and Depression Scale, *QoLS* Quality of Life Scale, *CES-D* Center for Epidemiologic Studies Depression Scale, *STAI* State-Trait Anxiety Inventory, *PANAS* Positive and Negative Affect Schedule, *IES-R* Impact of Event Scale Revised, *IES* Impact of Event Scale, *MICRA* Multidimensional Impact of Cancer Risk Assessment, *MOS* Medical Outcomes Study, *SF-12* Short Form Health Survey-12, *CRP* cancer risk perception, *GRP* genetic risk perception, *DT* Distress Thermometer, *UCL* Utrecht Coping List, *SCL-90* Symptom Checklist-90, *PSI* Psychiatric Symptom Index, *BSI* Brief Symptom Inventory, *BGCSES* Bergen Genetic Counselling Self-Efficacy scale, *TMSI* Threatening Medical Situations Inventory, *SAQ-N* Self-Assessment Questionnaire-Nijmegen, *BDI* Beck Depression Inventory, *POMS* Profile of mood state, *BSI-18* Brief Symptom Inventory-18, *GSI* Global Severity Index, *PSS* Perceived Stress Scale

talking within the family with direct communication exchanges, specifically, between siblings who undergo genetic testing [22].

Anxiety and depression

Twelve of the included studies focused on both anxiety and depression [1, 2, 12, 13, 24–26, 33–37], one focused only on anxiety [23] and one focused only on depression [27].

Eight studies assessed depression and anxiety symptoms using the HADS [1, 2, 13, 33–37], four assessed depression using the Center for Epidemiologic Studies Depression Scale (CES-D, [38]) [12, 25–27], one study assessed depression using the Beck Depression Inventory [39] [24] and five assessed anxiety using the State Trait Anxiety Inventory (STAI; [40]) [12, 23–26] (see Table 2).

Six studies found more increased levels of anxiety and depression in mutation carriers compared to non-carriers [1, 24–27, 35]. One study identified a decrease in anxiety levels in both carriers and non-carriers [23]. Surprisingly, Arver et al. [13] observed decreased levels of anxiety and depression over time in mutation carriers and increased levels in non-carriers; in addition, one study found no clinically meaningful variations in the anxiety and depression scores over time on a sample of cancer-affected and -unaffected mutation carriers [2]. Other studies did not find significant differences between carriers and non-carriers [12, 34, 36, 37].

Higher levels of anxiety and depression have been observed at 1, 3 and 6 months. At 6 and 12 months, the effects of test result disclosure disappeared, and the levels returned to baseline [25, 26, 34]. Moreover, some studies showed that gender is a factor that can influence anxiety and depression levels in people undergoing genetic testing. Women reported higher levels of anxiety and depression than men [22, 34], specifically, at 3 and 6 months after the genetic test disclosure [34].

Some factors can positively or negatively influence levels of anxiety and depression. Some potential risk factors are the following: having a cancer diagnosis [12, 26, 34, 35, 37], communicating the test result to the family [25], being younger than 40 years of age, having an unoptimistic personality and using suppression as an emotional regulation strategy [37, 41]. Among the protective factors are having pre-test information and education [13, 36] and making decisions about preventive prophylaxis [25, 37].

Discussion

In this systematic review, we aimed to investigate the consequences of genetic test results disclosure on psychological distress, anxiety and depression levels. To our knowledge, this is the first systematic review to focus on the psychological outcomes in both males and females who undergo BRCA genetic testing, as previous reviews were mainly based on women and/or on cancer patients [42–44]. After a selection procedure following the PRISMA method [15, 16], we considered 21 studies that analysed the psychological effects among people who underwent genetic testing for BRCA1/2 mutation.

Overall, we found rather diverse results. Most studies reported higher levels of distress, anxiety and depression in mutation carriers, compared with non-mutation carriers. Most probably, these high levels can be explained by an increased risk of future diseases and by implications not only for the tested individuals but also for their whole family [17, 25]. Other studies do not report differences between carriers and non-carriers [12, 21, 23, 34, 36, 37]. Surprisingly, one study reported increased levels of anxiety and depression in non-mutation carriers and decreased levels in mutation carriers [13]. A possible explanation that the authors give for this last finding was the high motivation and interest of the study participants who had required the genetic test and had been already involved in preventive programs. As also Fine suggests, there might be psychological benefits associated with the satisfaction to participate in clinical trials and contribute to research; furthermore, sharing with family members both the information and the emotions may improve relationships among them [45].

Some of the reviewed articles investigated if there were gender differences in levels of distress, anxiety and depression. They found that females have higher levels of distress, anxiety and depression than males. This could depend on the different ways men and women cope with their genetic testing result; in fact, men, unlike women, tend to minimize the emotional impact of their result, interpreting the information more in terms of the family, rather than in personal terms [18, 46]. Moynihan et al. [47] stated that men with BRCA mutation experience sadness but tend to suppress it; moreover, they are much more worried about their family, for example, for their children, as they feel guilty and responsible for them.

A possible explanation for the inconsistency in the international literature may be the use of different tools to detect anxiety, depression and distress, as we suggested in the above paragraphs. Besides, not all the papers considered other risk factors that may affect the emotional reaction to the genetic test.

Following this last consideration and our further objective, we analysed some risk factors and protective factors that, according to previous literature, may influence psychological outcomes and adjustment to genetic test results. The main risk factors comprised the following: having a cancer diagnosis [12, 22, 26, 34, 35, 37], being the first subject tested in the family [17], having a greater family history of breast/ovarian cancer [18], being unmarried [18], knowing a sibling's test result [22],

communicating the test result to the family [25], being in a family where one person has already received a positive result from the genetic test [33], being younger than 40 years of age, having an unoptimistic personality and using suppression as an emotional regulation strategy [37, 41]. Instead, having pre-test information [13, 36] and having direct exchanges in the family, especially between siblings who undergo genetic testing [22], might be protective factors. There are some gender differences in the factors that may influence levels of distress in mutation carriers. Among men, some risk factors are being unmarried and unemployed; in women, they are having daughters, a low income and a personal history of cancer [18].

Moreover, one of the main factors that influences and predicts high levels of anxiety and depression after the test is the presence of high levels of anxiety and depression before carrying out the genetic test, regardless of the test result [19, 23, 24, 26, 34, 37, 48, 49].

One study, however, shows that the levels of anxiety and depression were not associated with being or not being a mutation carrier, being or not being under oncological treatment, age, level of education, marital status and having children [33]. In particular, with regard to having an oncological diagnosis, some studies have shown that patients with a recent cancer diagnosis are more psychologically vulnerable and report higher levels of anxiety, depression and distress, probably because of the recent diagnosis and its treatment [35, 42].

Also, Beran et al. [25] found high levels of cancer-specific distress. In fact, for mutation carriers, the heightened distress observed during the months after test result receipt may be explained by decisions about prophylactic options,[1] communication of results to the family and one's own emotional and cognitive processing of the result. In addition, Bosch et al. [34] concluded their research by stating that the most psychologically vulnerable population is the one that presents higher concerns related to developing cancer.

Furthermore, it was shown that the anxiety levels of those recently diagnosed with cancer decreased after genetic counselling, becoming similar to the levels of patients diagnosed more than 1 year before. Hence, individuals with a recent diagnosis could benefit from genetic counselling [42, 44], as medical treatment protocols and surveillance for mutation carriers are well-defined and make treatment processes predictable and understandable [44]. Another study has highlighted the psychological benefit of undergoing the genetic test, regardless of its result. In fact, a study by Smith et al. [26] has shown that people who decide not to undergo the genetic test perceive greater risk and have more intrusive and avoidant thought over time. Also, Bonadona et al. [17] suggested that the opportunity for individuals to discuss with professionals about how they can share results and what potential reactions they can expect from relatives may facilitate the transmission of information, as well as help them cope with their concerns about their own high risk of developing cancer and the risk for their children. Overall, our data shows that individuals who undergo pre-testing genetic counselling and educational programs reported lower levels of distress, anxiety and depression than individuals who do not receive a pre-test education, as genetic and educational counselling may improve their ability to cope with the test result, with the implications for other family members, and with the different choices to consider [11, 13, 36].

Study limitations and further considerations

In the current review, there are critical issues about the long-time range of the reviewed studies (1998 to 2018) because scientific and medical discoveries in the last 20 years might have changed the patients' perception of genetic testing, influencing emotional experiences and surveillance options. Furthermore, the sample size is unbalanced between men and women; in fact, women represented a great majority as compared with men. Moreover, the population is not homogeneous because some studies included individuals with a cancer diagnosis and others only healthy people, and this may have affected the data, as a prior cancer diagnosis may influence the psychological outcomes. Another limitation is the high variability of the time range between genetic testing and administration of the psychological tools (with studies evaluating psychological outcomes before or at the same time of the genetic testing and studies up to 48 months after the genetic testing result), as this may influence the levels of the studied variables. Besides, the selected papers comprise samples exclusively from Western cultures, as studies representative of other cultural contexts were not found. Lastly, the analysed studies include standardized tools that sometimes do not distinguish different variables (as for HADS that considers both anxiety and depression) and were not specifically developed inside this topic. All these limitations lead us not to generalize our results.

However, our findings do provide a fuller picture of how genetic testing impacts individuals and may help genetic counsellors, health professionals and researchers care for people who undergo genetic testing. This review may also contribute to the knowledge of the experiences related to the fear of cancer and to being carriers of a genetic mutation, suggesting what psychological aspects to consider when working with these patients. In the light of these considerations, it is important that clinical

[1] The impact of decision to undergo (or not undergo) prophylactic surgery on post-testing distress, anxiety and/or depression has been analysed only in three studies (Borreani et al. 2014; Metcalfe et al. 2012; Claes et al. 2005) with inconsistent results. Borreani found that women who undergo prophylactic surgery have higher anxiety than women who decide not to undergo; Metcalfe (2012) found exactly the opposite, and Claes did not observe differences between the two groups.

psychologists could participate to genetic counselling in order to both detect early signals of risk factors and offer psychological supports to patients who need it. Furthermore, our data seems to underline the necessity for a major focus on males' emotional needs, also considering that the clinical intervention should be specifically tailored to the different needs of men and women. Finally, a clinical psychologist may offer an important contribution also in the decisions about how to communicate the genetic testing results to relatives, as previous studies highlighted the psychological benefits associated with an adequate communication within the family.

The increased risk of developing cancer may be experienced as traumatic [27]. In fact, as Mella et al. [33] suggested, individuals receiving the test result may need psychological support because of the high risk of depression and anxiety. As our review showed, this risk does not involve all the people undergoing genetic testing. Hence, future studies should consider all the risk factors we analysed in order to select people who are more likely to need psychological support. Moreover, in respect to our data collection, all the questionnaires used to detect anxiety, depression and psychological distress have been used world-wide but were not specifically developed for the context of oncological risk research. Hence, future research should consider developing and using tools more specific and sensitive to this field. Additionally, our data should be useful for future research in suggesting the inclusion of more male samples in the research on this topic. Finally, our work could encourage researchers to carry out more studies focusing on the psychological impact of an inconclusive result at the genetic testing.

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Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

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