The role of social support and stress coping strategies in genetic screening for cancer predisposition – BRCA testing as an example

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Abstract

Introduction: Individuals with monoallelic pathogenic variants in BRCA1/BRCA2 genes are at an increased risk of developing breast or ovarian cancer. The results of genetic tests may present a stressor related to the fear associated with the impact of the diagnosis on an individual's future.

Aim: The aim of the study was to verify the level of depression, anxiety, and stress in individuals waiting for the results of BRCA genetic tests. How personality traits, stress coping strategies and social support influence the level of mental discomfort was examined.

Material and methods: 82 consecutive individuals completed an anonymous online survey consisting of basic information and five questionnaires: the Inventory for Measuring Coping with Stress (Mini-COPE), the Depression, Anxiety and Stress Scale (DASS-21), the Generalized Anxiety Disorder 7-Item Scale (GAD-7), Polish adaptation of the Ten Item Personality Inventory test (TIPI-PL), and the Berlin Social Support Scales (BSSS).

Results and discussion: Participants obtained the highest scores in Mini-COPE strategies: active coping, planning, positive reframing, acceptance, and seeking emotional support. The results of DASS-21 and GAD-7 indicate the level of depression, anxiety, and stress mainly in the normal range. The highest scores in the TIPI-PL test were obtained on the conscientiousness, agreeableness, and extraversion scales. Participants, who have already obtained the results had higher scores on the Need for Support Scale (BSSS).

Conclusions: In general, the situation of BRCA genetic testing is not causing significant negative mental discomfort. Nonetheless, the results indicate a significant role of social support in coping with stress in individuals who have already obtained the results of their genetic test.
1. INTRODUCTION

Breast cancer is the most common type of cancer that affects women.\(^1\) The study carried out by Cardoso et al. (2012) shows that breast cancer is not a frequent disease in population under 40 years old. Despite this, the authors noticed a clear growth in morbidity in this group in recent years, especially in the United States.\(^2\) According to the study of Shoemaker et al. (2019) incidence trends increased from 2004 to 2013 for Asian or Pacific islander (API) women and white women aged 20–34 years.\(^3\) Moreover, the other study shows that in South Korea from 2000 to 2017 individuals aged 40–50 years old are getting breast cancer more often, and the peak point on the graph is gradually shifting to the right along the X-axis.\(^4\) It shows, that breast cancer is an increasingly common disease that is beginning to affect younger and younger people. Of all cases, 5%–10% are caused by highly penetrant mutations, for example, monoallelic pathogenic variants in \(BRCA1\) or \(BRCA2\) genes.\(^5,6\) Individuals with \(BRCA1\) defect are at the risk of having breast cancer at the level of 55%–72%.\(^7\) It also increases the likelihood of developing ovarian cancer, which is estimated as being 39%–44%.\(^7,8\) Due to such significant probability, it is highly recommended for women with breast or ovarian cancer to undergo \(BRCA\) genetic testing. Knowing whether or not they have an underlying genetic cause of their disease gives them an opportunity to take steps towards preventing further consequences, e.g. undergoing prophylactic mastectomy or salpingo-oophorectomy or more frequent clinical follow-up. The studies of Yadav et al. (2017) and Petrie et al. (2015) show that this knowledge makes women with cancer choose mastectomy of both breasts more willingly.\(^9,10,11\) Moreover, genetic testing can be the beginning of lifestyle changes for disease prevention or stress release from lack of the genetic burden.\(^12\)

Unfortunately, a positive result of a genetic test can be potentially a reason for increased stress or anxiety. Individuals who are aware of having \(BRCA\) pathogenic variant worry about passing it on to their children.\(^13\) According to the research of Lodder et al. (2001), 20% of women who had been diagnosed to have the hereditary disease and 11% who did not, reported high anxiety after receiving the result of a test.\(^14\) The level of anxiety after receiving the result was significantly connected with a high level of anxiety while waiting for the result. This study also highlighted the possible influence of awareness of the presence of a pathogenic variant in the family. Non-carriers, whose sister was diagnosed as having a pathogenic variant, had a higher level of depression after genetic testing than the other non-carriers. Wenzel et al. (2012) examined another issue: the impact of a death of a family member from \(BRCA\)-associated morbidity.\(^15\) This experience makes individuals diagnosed to have a \(BRCA\) defect feel more stressed and anxious and report lower quality of life than patients, whose family member is still alive.\(^13\) Makhnoon, Arun, and Bedrosian (2022) showed that subjects with positively diagnosed first-degree relatives felt more anxious after getting the results of a genetic test for up to 6 months.\(^16\) The researchers, same as Hesse-Biber and An (2016), indicate the matter of social support too.\(^16\) The satisfying social support makes persons more likely to undertake genetic testing, take preventive disease actions, and have a lower level of distress connected with cancer.\(^17\) However, there is a deficit of studies that focus specifically on the impact of social support in genetic testing.

It should be mentioned however that some of the studies do not indicate any significant negative psychological effects after receiving the diagnosis. One of them is a systematic review written by Oliveri et al. (2018).\(^18\) The main conclusion of their work was that individuals who decided to undergo genetic testing do not experience the growth of anxiety or a negative impact on their quality of life (it does not apply to Huntington’s disease).\(^18\) Marteau and Croyle (1998), in their research show that the level of stress in patients after receiving positive test turned out to be still in a normal range.\(^19\) What is more, National Cancer Organization (NCO) indicates that many participants of their study feel more competent and less anxious after receiving their genetic test results.\(^20\) Nevertheless, the latest review of Makhnoon shows that although the negative effects of the tests are very few, mild, and temporary, but they do occur.\(^17\)

2. AIM

The aim of this study was to verify the level of stress, anxiety, and depression in individuals waiting for the \(BRCA\) genetic test result, in comparison to those who have already received it. The context of the disease and family history has also been taken into account. The study evaluated also how personality traits and stress coping strategies influence the level of stress, anxiety, and depression. Furthermore, the study aimed to test the needs and satisfaction of social support in individuals waiting for the genetic test results.

3. MATERIAL AND METHODS

3.1. Participants

82 consecutive adult individuals from a single genetic out-patients clinic at the Center for Rare Diseases, University Clinical Center in Gdansk, Poland, eligible for \(BRCA\) testing according to criteria as in the Polish National Health Programme were enrolled in the survey. The majority of participants were female (64 vs. 18). The median age was 39.5 years (range: 19–74 years). The majority of the respondents had a higher education degree (65%), followed by 26% having secondary education, 8% vocational education, and 1% basic education. One third of patients (22%) declared that they have or had cancer. Most of the respondents (71%) had a 1st degree relative diagnosed with cancer, whereas 18 (26%) had a family member already diagnosed to have a \(BRCA\) pathogenic variant. Among the respondents, 33 people have already received the results of genetic tests, and 49 are still waiting for them.
3.2. Procedure
The material for analysis was collected for over a year, from February 2021 to March 2022. Respondents completed an anonymous survey consisting of basic demographic information related to age, gender, and level of education as well as three questions about patients’ and their family’s history of the disease: ‘Do you have or have you had cancer?’, ‘Is any of your relatives (only parents, siblings, children) diagnosed with cancer?’, ‘Has any of your relatives a diagnosed mutation associated with hereditary cancer (e.g. BRCA1)?’. The main part of the study consisted of five standardized questionnaire methods.

3.3. Methods
In order to assess the patients’ mental discomfort, five standardized questionnaires were used: the Depression, Anxiety and Stress Scale (DASS-21), the Generalized Anxiety Disorder 7-Item Scale (GAD-7), the Inventory for Measuring Coping with Stress (Mini-COPE), Polish adaptation of the Ten Item Personality Inventory test (TIPI-PL), and the Berlin Social Support Scales (BSSS). The BSSS was added after receiving the completed questionnaires from 14 participants. DASS-21 is a method of measuring psychological distress along the constructs of depression, anxiety, and stress. This questionnaire consists of 21 questions that relate to the previous week. GAD-7 was designed in 2006 by Spitzer et al (2006). This 7-point method is used to assess a patient’s anxiety level during the last 2 weeks. Mini-COPE examines 14 strategies for coping with stress: active coping, planning, positive reframing, acceptance, sense of humor, turning to religion, seeking emotional support, seeking instrumental support, self-distraction, denial, venting, substance use, behavioral disengagement, and self-blame. TIPI-PL is a short method of assessing the Big Five personality traits: openness to experience, conscientiousness, extraversion, agreeableness, and emotional stability. BSSS are self-report measures of perceived available emotional support, perceived available instrumental support, need for support, support seeking, actually received support, and protective buffering scale.

3.4. Study algorithm
Firstly, the authors tested a pilot group of 14 subjects. In view of the results the BSSS and 2 additional questions about patients’ and their family’s medical history were added to the survey. In the subsequent analyses a group of patients who have been waiting for the BRCA genetic testing results (Before result – BR, n = 49) were compared to group of patients who have already received the results (After result – AR, n = 33). Male to female ratio and age distribution were similar in both subgroups.

3.5. Statistical analysis
Analyses were performed using Statistica 13.3 software (StatSoft, Polska, Kraków, Poland) data analysis software system. Frequencies were compared using Fisher’s exact test. For continuous variables, differences between groups were evaluated using the Mann-Whitney U test for pairwise comparisons. The r-Spearman’s correlation method was used to assess the relationship between waiting time and other variables. The level of statistical significance was assumed P < 0.05. Due to the small sample size and exploratory nature of the study Bonferroni corrections for multiple comparisons were not made.

4. RESULTS

4.1. DASS-21
The results of all 3 scales were in the range of norm among the majority of the respondents (Figure 1). The scores were as follows: depression (median 3, range 0–20), anxiety (median 2, range 0–16), and stress (median 5, range 0–20). All 3 scales were very strongly and positively correlated with each other and the result of the GAD-7 questionnaire was at a significance level of P < 0.001. The Mann-Whitney U test
showed that no significant differences between the groups BR and AR. The same applies to differences in gender, level of education, and age.

4.2. GAD-7
Most of the respondents’ results were between the minimum and mild anxiety levels (median 5.5, range 0–20) (Figure 2). The Mann-Whitney U test showed significant differences between the genders (Figure 3; \( P = 0.009 \)). Differences between BR and AR groups; the level of education and age were not statistically significant (\( P > 0.05 \)).

4.3. TIPI-PL
The results for groups BR and AR are shown in Figure 4. Moderate, but still statistically significant, correlations were found between extraversion and conscientiousness (\( r = 0.37; P < 0.001 \)), extraversion and emotional stability (\( r = 0.4; P < 0.001 \)), agreeableness and conscientiousness (\( r = 0.38; P < 0.001 \)) and extraversion and openness to experience (\( r = 0.28; P < 0.01 \)).

Significant differences related to gender were observed (Figure 5). On the contrary, the were no significant distinctions between the BR and AR, just like for age and level of education.

4.4. BSSS
The results of the BSSS questionnaire for the whole group were as follows: perceived available emotional support (median 3.5, range 1.75–4.00), perceived available instrumental support (median 4, range 1.75–4.00), need for support (median 2.8, range 1.3–3.8), support seeking (median 2.8, range 1–4), actually received emotional support (median 3.9, range 1.6–

Figure 2. The distribution of results for the GAD-7 questionnaire for both groups: BR, and AR.

Figure 3. The level of anxiety according to the GAD-7 questionnaire with respect to gender.

Figure 4. Personality dimensions according to the TIPI-PL questionnaire. Median, non-outlier range, and the quartile distribution of the scores in the subgroups BR and AR. Outliers are marked with dots, and extreme values are marked with an asterisk. All comparisons were statistically insignificant (\( P > 0.05 \)).

Figure 5. Personality dimensions compared with respect to gender according to the TIPI-PL questionnaire. Median, non-outlier range and the quartile distribution of the scores in both groups. Outliers are marked with dots.
4.0), actually received instrumental support (median 3.7, range 1–4), actually received informational support (median 3.5, range 1–4), satisfaction of received support (median 4, range 2–4), protective buffering scale (median 2, range 1.0–3.5). The results for BR and AR groups are shown in Table 1.

Significant differences between the results of BR and AR subgroups on scales need for support (Figure 6a; \( P = 0.01 \)) and actually received instrumental support (Figure 6b; \( P = 0.02 \)) were observed.

In the study the authors also noticed a significant difference between the results of groups of participants younger than 40 years old and older in the protective buffering scale (Figure 6c; \( P = 0.05 \)). Differences in the results of women and men in support seeking also turned out to be statistically significant (Figure 6d; \( P = 0.02 \)).

### 4.3. Mini-COPE

The results of participants on each scale were: active coping (median 2, range 0.5–3), planning (median 2, range 0.5–3), positive reframing (median 2, range 0–3), acceptance (median 2, range 0–3), sense of humor (median 0.5, range 0–3), turning to religion (median 1, range 0–3), seeking emotional support (median 2, range 0.5–3), seeking instrumental support (median 2, range 0–3), self-distraction (median 2, range 0–3), denial (0.5, range 0–3), venting (median 1.5, range 0–3), substance use (median 0, range 0–3), behavioral disengagement (median 0.5, range 0–2), self-blame (median 1, range 0–3). No significant differences were observed between the groups BR and AR.

The Mann-Whitney \( U \) test presented significant differences between genders in strategies of: planning, acceptance, seeking instrumental support, behavioral disengagement, and venting (Table 2). Also, significant differences between groups of participants younger than 40 years old and older in strategies of: positive reframing, acceptance, and self-blame were found (Table 3).

### 5. DISCUSSION

In this study the authors aimed to examine whether individuals waiting for the \textit{BRCA} genetic test results experience a greater level of depression, anxiety, and stress, in comparison...
Table 2. Median, the quartile distribution and statistical significance in scales planning, acceptance, seeking instrumental support, behavioral disengagement and venting of the Mini-COPE questionnaire with respect to gender.

<table>
<thead>
<tr>
<th>Coping with Stress Strategy</th>
<th>Females Median</th>
<th>Females Q1</th>
<th>Females Q3</th>
<th>Males Median</th>
<th>Males Q1</th>
<th>Males Q3</th>
<th>P</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acceptance</td>
<td>2.0</td>
<td>0.5</td>
<td>3.0</td>
<td>1.8</td>
<td>0.0</td>
<td>2.5</td>
<td>0.007</td>
</tr>
<tr>
<td>Seeking instrumental support</td>
<td>2.0</td>
<td>0.0</td>
<td>3.0</td>
<td>1.0</td>
<td>0.5</td>
<td>2.0</td>
<td>0.002</td>
</tr>
<tr>
<td>Self-distraction</td>
<td>2.0</td>
<td>0.0</td>
<td>3.0</td>
<td>1.5</td>
<td>0.0</td>
<td>2.5</td>
<td>0.001</td>
</tr>
<tr>
<td>Venting</td>
<td>1.5</td>
<td>0.0</td>
<td>3.0</td>
<td>0.8</td>
<td>0.0</td>
<td>1.5</td>
<td>0.00001</td>
</tr>
</tbody>
</table>

Table 3. Median, the quartile distribution, and statistical significance on scales positive reframing, acceptance, and self-blame of the Mini-COPE questionnaire for groups of participants younger than 40 years old and older.

<table>
<thead>
<tr>
<th>Coping with Stress Strategy</th>
<th>&lt;40 years old Median</th>
<th>&lt;40 years old Q1</th>
<th>&lt;40 years old Q3</th>
<th>&gt;40 years old Median</th>
<th>&gt;40 years old Q1</th>
<th>&gt;40 years old Q3</th>
<th>P</th>
</tr>
</thead>
<tbody>
<tr>
<td>Positive reframing</td>
<td>1.5</td>
<td>0.0</td>
<td>3.0</td>
<td>2.0</td>
<td>0.0</td>
<td>3.0</td>
<td>0.006</td>
</tr>
<tr>
<td>Acceptance</td>
<td>2.0</td>
<td>0.0</td>
<td>3.0</td>
<td>2.5</td>
<td>1.5</td>
<td>3.0</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>Self-blame</td>
<td>1.0</td>
<td>0.0</td>
<td>0.5</td>
<td>0.0</td>
<td>2.5</td>
<td>0.0</td>
<td>0.03</td>
</tr>
</tbody>
</table>

Table 4. r-Spearman’s correlations between scales of the DASS-21, Mini-COPE and TIPI-PL questionnaires.

<table>
<thead>
<tr>
<th></th>
<th>DASS-21</th>
<th>Mini-COPE</th>
<th>TIPI-PL</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Depression</td>
<td>Active coping</td>
<td>Planning</td>
</tr>
<tr>
<td>Depression</td>
<td>—</td>
<td>-0.28**</td>
<td>n.s.</td>
</tr>
<tr>
<td>Active coping</td>
<td>-0.28**</td>
<td>0.70***</td>
<td>n.s.</td>
</tr>
<tr>
<td>Planning</td>
<td>n.s.</td>
<td>0.70***</td>
<td>—</td>
</tr>
<tr>
<td>Positive reframing</td>
<td>n.s.</td>
<td>n.s.</td>
<td>0.44***</td>
</tr>
<tr>
<td>Seeking emotional support</td>
<td>n.s.</td>
<td>0.75***</td>
<td>0.64***</td>
</tr>
<tr>
<td>Behavioral disengagement</td>
<td>0.31**</td>
<td>-0.55**</td>
<td>-0.50**</td>
</tr>
<tr>
<td>Self-blame</td>
<td>0.23*</td>
<td>-0.31**</td>
<td>-0.43**</td>
</tr>
<tr>
<td>Extraversion</td>
<td>n.s.</td>
<td>n.s.</td>
<td>n.s.</td>
</tr>
<tr>
<td>Conscientiousness</td>
<td>n.s.</td>
<td>n.s.</td>
<td>0.32**</td>
</tr>
<tr>
<td>Emotional stability</td>
<td>-0.23*</td>
<td>0.36***</td>
<td>0.34**</td>
</tr>
</tbody>
</table>

Comments: *P < 0.05; **P < 0.01; ***P < 0.001; n.s. – not specified.
they are less stable, less balanced, and can experience easier negative emotions. That results are in line with the other studies.17–19

In the presented study we tested the hypothesis whether the patients while waiting for the results would be more depressive, anxious, and stressed when compared to individuals who have already received them. As the results present, the differences in scores on each scale of the DASS-21 between these two groups were not significant. Most patients in a situation of genetic testing received scores that are within the normal range, namely 64% on the depression scale, 66% on the anxiety scale, and 67% on the stress scale (Figure 1). According to the results of the GAD-7 questionnaire most (80%) of the individuals obtained results that indicate minimal and mild levels of anxiety. The same conclusion comes from a study by Hallowell et al. (2002), in which 30 women going through BRCA screening were interviewed. The situation of BRCA screening and waiting for the results was not seen as anxiety-provoking.16 Several other studies confirm that genetic testing does not have a significant negative psychological impact on individuals during waiting for the results.17,19,20

The authors also verified the second hypothesis, which concerns whether patients who are waiting for receiving BRCA test results need more social support than patients after receiving them. Our study proves that it is the opposite: individuals who have already diagnosed need more support than patients who are before receiving the genetic test results. The probable explanation is that the AR group includes some individuals, who have just learnt about having a BRCA disease-causing variant. Subjects who have a BRCA pathogenic variant report a clinically significant feeling of helplessness, stigmatization, and anxiety for health.16 More to the point, the anxiety level after diagnosis generally increases in women with positive results compared to women with a negative test. In order to reduce the level of anxiety they may undertake a number of cancer preventive actions, like risk-reducing surgeries. Bilateral risk-reducing mastectomy provides a 90%–95% risk reduction in getting breast cancer.44 McCalfe et al. (2020) in their study were trying to assess if a mastectomy surgery can reduce the psychological discomfort in women with a BRCA diagnosis.16 It turned out that undergoing this kind of treatment made women feel less distress than those who have not done it yet.42 In the article of Hatcher et al. (2001) the authors came to the same conclusion. The high level of anxiety before mastectomy surgery reduced significantly after surgery.45 According to the study by Hesse-Biber and An, the cancer preventive actions were taken more diligently by individuals who had a BRCA pathogenic variant when they received strong social support.17 The participants of our study were mainly using the seeking emotional support and seeking instrumental support strategies while coping with stress. It points to the significant role of social support in the situation of BRCA testing, that unfortunately is not yet sufficiently evaluated in the literature. Li and Nishikawa in their study (2012) associated an active coping strategy with seeking social support.46 According to our study the results on active strategies of stress coping scales, like active coping, planning, and positive re-framing were positively correlated with the result on the seeking emotional support scale. Moreover, our study presents that in general the results on personality traits scales like agreeableness, conscientiousness and extraversion were also positively correlated with results on active stress coping strategies scales, and negatively with results on scales of self-blame and behavioral disengagement. Additionally, the result on the scale of self-blame was positively correlated with the depression scale result (DASS-21).

According to our study, participants scored the highest results on agreeableness, conscientiousness, and extraversion scales on the TIPI-PL questionnaire and on healthy stress coping strategies scales such as active coping, planning, acceptance, positive reframing, and seeking emotional support. The correlations show that such results may favor an effective coping with stress in a situation of BRCA genetic testing. Dysfunctional coping strategies, which are: behavioral disengagement and self-blame can be an important indication for undergoing psychological therapy to improve the quality of life.32

5.1. Study limitations

The main limitation of our study is the small number of participants. That could make a difficulty in identifying significant relationships in the results. The same comes with the bias proportions of female and male responders. Women were in a great majority (78%) compared to men (22%). It is also noticeable in the other studies in this field, due to the greater number of women who are referred for genetic tests. The AR group was not divided into groups of patients with positive and negative genetic test results. That division would make it possible to verify the differences in psychological discomfort between these two groups. Lastly, the authors of the BSSS questionnaire noted that there is no information available about the usefulness of the actually received support and protective buffering scales in non-stressful situations.47 Also, the measurement of these scales can be distorted, because of the social desirability factor, which is a tendency to report answers in a way to present oneself in a better light rather than in a truthful manner.48 That can lead to inaccurate self-reports and hence mistaken conclusions.

5.2. Future research directions

There is a lack of studies that verify the negative psychological implications on people who are awaiting the results of the BRCA genetic test. The articles about the psychological impact of genetic testing on women are prevailing, while studies including male participants are scarce. It is also noticeable that there is a lack of studies evaluating aspects of social support. Getting to know the needs of patients in this matter will provide information on whether they need psychological support before knowing the result of genetic testing.
6. CONCLUSIONS

(1) The situation of waiting for BRCA genetic testing did not cause a significant negative impact on patients.
(2) A higher need for social support was observed in patients, who already received the genetic diagnosis.
(3) In this study, an important role of social support in coping with stress was also noticed.
(4) The results show significant differences between women and men.
(5) Extending the topic in further research is recommend-
ed, especially by using tools that apply the aspect of so-
cial support and to examine more male subjects.

Conflict of interest
The authors declare that they have no known competing fi-
nancial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Ethics
The procedure was approved by the Board of the University Clinical Center in Gdansk and the Independent Bioethics Committee for Scientific Research at the Medical University of Gdansk (NKBBN/280/2021). All participants gave informed consent to participate in the study.

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