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Genetic cancer risk assessment: A screenshot of the psychosocial profile of women at risk for hereditary breast and ovarian cancer syndrome

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Abstract

Objective: There is a lack of information describing Brazilian women at risk of hereditary breast and ovarian cancer syndrome (HBOC) who undergo genetic cancer risk assessment (GCRA). This study aims to characterize the psychosocial profile of women at risk for HBOC at their first GCRA to obtain an overview of their families' profiles and the challenges of the oncogenetics setting.

Methods: This was a cross-sectional study in which interviews were conducted with 83 cancer-affected women at their first GRCA appointment after the pedigree draw. Tools to evaluate psychological outcomes were applied. The pedigree genogram and ecomap were constructed and analyzed with content analysis using the "life course perspective" theory.

Results: Individuals perceived their breast/ovarian cancer risk to be equal to that of the general population, although they were highly concerned about developing cancer. No evidence of anxiety or depressive symptoms was identified. Participants used the coping strategy of searching for religiosity. The genograms and ecomaps resulted in five major themes: support and social support; attitudes, feelings and emotions; cancer causes; communication; and relationships with relatives. Individuals between 20-29 years of age and those with no family history of cancer tended not to communicate with relatives, which may indicate future problems in the GCRA process regarding genetic testing.

Conclusions: This study demonstrated that knowing the families who undergo the GCRA process can help professionals provide more individualized and thorough attention during GCRA and genetic testing, which results in better follow-up and prevention strategies.

KEYWORDS
cancer, genetic cancer risk assessment, genogram, hereditary breast and ovarian cancer, oncology

1 | BACKGROUND

Hereditary breast cancer accounts for approximately 5%-10% of all cases of breast cancer (BC), and the most frequently associated genes are *BRCA1* and *BRCA2*.[1,2] The presence of germline pathogenic variants in these genes leads to hereditary breast and ovarian cancer predisposition syndrome (HBOC),[1] which increases the risk of BC (40%-80%),[3] ovarian cancer (OC) (11%-50%),[3] prostate cancer (9.5%-20%)[4] and other cancer types.

Due to the high risk to develop cancer it is important the identification and confirmation of HBOC, once it can help the management of risk-reducing strategies.[1] The genetic cancer risk assessment (GCRA) process is important in this context, given that it includes the pedigree drawing and analysis, the discussion of genetic testing (GT),[5] as well as patient and family education and provides support for decision making and assistance with the psychological responses that may occur.[5]

During the GCRA, some patients might develop anxiety and depression[6]; anguish due to fear of the unknown[7]; and psychological problems related to living with cancer, hereditary predisposition and family issues.[8,9] Some patients commonly seek psychological help during counseling to discuss problems concerning self-acceptance and family dynamic issues, such as difficulties in family communication, worries about stigma and effects on future family generations.[10]

In South American countries, GCRA is a relatively new field, and little is known about the profiles and needs of affected families. In Brazil, the number of GCRA services is limited.[11] It is thus important to characterize and discuss the characteristics of the probands and their families to ensure that professionals can address the challenges of the oncogenetics setting.

The principal aim of the study was to characterize the psychosocial profile of Brazilian cancer-affected women at risk for HBOC who underwent GCRA and GT. The characterization was performed by exploring BC/OC risk perceptions, worries about developing another cancer, health beliefs related to preventive exams, ways of coping with problems, anxiety and depression. In addition, family dynamics was evaluated using genograms and ecomaps with the intention to predict the possible difficulties that both patients and professionals could face during all GCRA process.

2 | METHODS

2.1 | Study scenario

The study scenario was the oncogenetics department (OD) of a Brazilian oncologic hospital (Barretos Cancer Hospital - BCH), located in the Southeast Region of Brazil. Patients at risk (due to personal and/or family history [FH] of cancer) are referred by the consultant medical doctor to the OD (it is not open to external patients). The first appointment at the OD consists of two parts of the GCRA: (a) the nurse consultation, when the oncogenetics nurse collects information regarding patients' sociodemographic data and FH to draw the pedigree and (b) the consultation with the medical geneticist, where the GT and its implications are discussed. More information about the OD organization can be found elsewhere.[11]

The study was conducted in the first appointment during the first part of the GCRA over the years 2016 and 2017. The specialist nurse in oncogenetics (N.C.) conducted the nurse consultation and then invited the patient to participate in the study. Those that accepted signed a specific consent form and answered the questionnaires and the qualitative interviews. This study was approved by the Institutional Ethics Committee (CAAE: 45128915.6.0000.5437).

2.2 | Participants

All women older than 18 years old who met the criteria for *BRCA1/2* genetic testing (Table S1) were invited to participate in the study.

The sample calculation was performed based on the number of patients with FH suggestive of HBOC who were seen in the OD in previous years. A posteriori power analysis was conducted (Gpower 3.0.10) to verify whether the number of included patients would be representative of the true OD attendance. Based on the calculations, considering an error of 0.05, a power of 0.8 and an effect of 0.18, a value of 63 was obtained for an appropriate sample size.

2.3 | Tools

All the instruments were validated in a Brazilian-Portuguese language version.[12-15]

2.3.1 | Cancer worry

The Lerman's Cancer Worry Scale (CWS) has six items and assesses the concern about developing BC. The range for elevated cancer worry is 14 to 26.[16]

2.3.2 | Cancer risk perception

The Cancer Awareness Needs Survey (CANS) questionnaire assesses the perception of BC and OC risk.[13]

2.3.3 | Health Belief Model

The Champion's Health Belief Model Scale (CHBM) was used to evaluate health beliefs. This questionnaire uses 27 questions divided into four domains: (a) susceptibility, (b) severity, (c) benefits, and (d) barriers. Higher scores across the domains indicate that the individual considers health beliefs to be an area of focus.[17]

2.3.4 | Coping strategies

Ways of Coping Scale (EMEP) was used. It is composed of 45 items grouped into the following factors: confrontation focused on the problem, focus on emotion, search for social support, and search for

religious practices. These items evaluate thoughts and actions that people use to address a specific stressful event (in the case of GCRA), and the factor that has the highest scores indicate that the individual uses this particular methods to cope with problems.[14]

2.3.5 | Anxiety and depression

Hospital Anxiety and Depression Scale (HADS)[18] was used. Scores from 10 to 21 indicate medium to high levels of anxiety and depression.[15]

2.3.6 | Family dynamics

FH and dynamics was collected through the pedigree, genogram, and ecomap drawing (using the software GenoPro 2011) during a semistructured interview[19] (Table S2). The genogram integrates the biomedical and psychosocial histories of the patient and those of his family.[20,21] In addition, the ecomap provides an expanded view of the family, designing a support structure and portraying a connection between the family and the world/community.[22]

2.4 | Procedures

All instruments were applied at the first part of GCRA and before the GT. The application of the instruments always followed the same order after informed consent: genogram and ecomap drawing through the semistructured interview; and HADS, EMEP, CHBMS, CWS questionnaire assessment. The entire interview lasted from 30 to 60 minutes.

2.5 | Data analysis

2.5.1 | Quantitative data

Data description was performed according to the means, standard deviations, and minimum/maximum values for the quantitative variables and according to the frequencies and percentages for the qualitative variables. A chi-square test or Fisher's exact test was used to compare the categorical variables surgery, educational level, FH, cancer stage, treatment status and time since diagnosis to cancer risk perception (once the risk perception can be influenced by theses variables). For the CWS and CHBM questionnaires, which do not have single items measure, Cronbach's α was calculated to evaluate the internal consistency of these instruments. Values >0.7 were considered to indicate acceptable reliability.[23] To support the quantitative data, the software SPSS 22.0 (IBM) was used.

2.5.2 | Qualitative data

Genograms, ecomaps, and semistructured interviews were analyzed using content analysis with the technique of thematic or categorical

analysis from Laurence Bardin's framework.[24] To support the qualitative analysis, the software NVivo V.11Pro (QRS International) was used. In addition, the analyses were performed by two professionals: the nurse who conducted all the interviews and by a social scientist (L.G.), who did not participate in data collection. The qualitative data were treated using the "life course perspective" theory, that focuses on the temporal and contextual elements of an experience.[25] An example is that the age of the individual or the historical period to which individuals have been exposed may influence their future actions, such health decisions.[26] In this sense, for this study, age and FH of cancer were considered.

3 | RESULTS

3.1 | Quantitative analysis

Of the 94 invited women, 83 agreed to participate. They were from 16 different states of Brazil (representing 61.5% of all Brazilian states), with the majority being from the Southeast Region (67.8%). The sociodemographic data showed that 73 (88%) were diagnosed with BC, 2 (2.4%) with OC, and 8 (9.6%) with BC and another primary tumor. The majority had a FH of cancer (91.6%), with 26 (31.0%) and 14 (16.8%) having first and second-degree relatives with BC, respectively and 3 (3.6%) having first- and second-degree relatives with BC (Table S3).

The worry about BC recurrence had an average score at CWS instrument of 13 (range: 6-26; SD = 5.4), meaning that the participants displayed moderate concern about developing a new cancer. However, 40% (34 cases) of our sample showed elevated levels of concern. The Cronbach's alpha of the CWS application had a value of 0.91, indicating good internal consistency.

CANS questionnaire (that measures cancer risk perception) showed that most of the women perceived their BC and OC risk as equal to that of the general population (Table 1). To verify if surgery, educational level, presence of cancer FH, cancer stage, disease status, and time since cancer diagnosis might have influenced the participants' risk perceptions, a Fisher's exact test was performed, and statistical significance was found only for the group that performed oophorectomy, which showed that women who had oophorectomy experienced lower risk perception for BC ($P = .019$) and OC ($P = .033$) (Table S4).

Health beliefs (CHBM application) had higher scores for severity, with an average score of 15.43, and for benefits, with an average score of 14.1. These results indicate that this group of women hold high perceptions of the gravity of their BC (severity scale), and they believe in the benefits of prevention strategies for BC (benefits scale). The susceptibility scale showed lower scores on the CHBM, with an average score of 8.37, showing that this group has a low perception of risk (Table 2).

The EMEP (coping strategies) showed that our population has a higher average score on the factors of confrontation focused on the problem and the search for religious practices, with respective mean

TABLE 1 Cancer risk perception

	None, very low N (%)	Lower than other people N (%)	Equal to other people N (%)	Higher than other people N (%)	Much higher than other people N (%)	Total N (%)
Breast cancer	5 (6.0)	7 (8.4)	43 (51.8)	22 (26.5)	6 (7.2)	83 (100)
Ovarian cancer	8 (9.6)	10 (12.0)	45 (54.2)	16 (19.3)	4 (4.8)	83 (100)

TABLE 2 Scores obtained in the application of the Champion's Health Belief Model

	Score	Cronbach's α
Susceptibility[a] (0-20)		
Mean	8.37	
Minimum	5	
Maximum	16	
SD	3.3	0.87
Severity[a] (0–28)		
Mean	15.43	
Minimum	7	
Maximum	26	
SD	4.5	0.75
Benefits[a] (0–20)		
Mean	14.1	
Minimum	5	
Maximum	20	
SD	3.4	0.70
Barriers[a] (0-48)		
Mean	13.9	
Minimum	12	
Maximum	20	
SD	2.1	0.35
Total	83	

[a]The domains and their respectively measures are at Table S5.

values of 4.5 (range: 2.9-5.2; SD = 0.4) and 4.2 (range: 2.4-5; SD = 0.5).

Regarding anxiety and depression, evaluated through the HADS application, the average score for anxiety and depression was 5.2 (range: 0-18; SD = 4.3), and 2.4 (range: 0-15; SD = 2.8) respectively. This finding suggests that at the time of survey completion, there was limited evidence of general anxiety and depression. However, outliers were observed, with three (3.6%) participants demonstrating high levels of anxiety (scores of 13, 16, and 18) and one (1.2%) participant demonstrating a high level of depression (score: 15).

3.2 | Qualitative analysis

The content analysis produced five main categories: (a) support and social support network (the most important entities the women

considered to give them the support); (b) attitudes, feelings, and emotions (what the women reported regarding their feelings, emotions and worries); (c) cancer causes (what women believed was the cause of their cancer); (d) communication with relatives (the quality of the women's communication with their family members); and (e) relationships with relatives (how women describe their family relationships). Subcategories were identified within these categories (Figures S1 and S2).

For the category "support and social support network," the subcategory "religiosity and/or spirituality" was the most frequent. The frequency of each category and examples of the material categorized from the interview narrative are in Table 3.

The data were treated using the "life course perspective" theory. Age was used as an important factor to evaluate whether the participant's age interfered with the form of speech and with the construction of genograms and ecomaps. Table S6 shows the categories and subcategories found through this analysis by age group.

The majority of women between the ages of 30 and 39 were more worried about work and finances than were other groups of women. The category "communication with relatives," did not appear in the age group between 20 and 29 years. This omission leads us to suggest that this group may experience greater problems in sharing information with their relatives.

Analysis was performed to verify the frequency of all categories and subcategories that appeared while considering the presence and absence of a FH of cancer, indicating that participants who did not have a FH did not report issues related to "concern about future generations" and "promotion of communication between members" (Table S7).

4 | DISCUSSION

This study provided an overview of the psychosocial profile of patients at risk for HBOC seeking GCRA in a Brazilian reference cancer center.

Communication process is very important for effective GCRA. As the majority of our population of first-time visitors to an OD estimate their cancer risk to be equal to that of the general population, it is important to ensure that the women understand the facts regarding their risk and consider their personal and FH of cancer to help them make sense of GT.

The cancer risk perception is a subjective and complex matter. The literature shows that risk perception encompasses many factors of life experience. A study conducted by Peipins et al, evaluated 2524 women and compare risk perception to several factors, including the

TABLE 3 Qualitative analysis of genogram and ecomap

Categories and subcategories	Frequency (n/83)	Example
Support and social support network		
Religiosity and/or spirituality	74/83	"The church is the social support for times of difficulty"
Community	19/83	"There are friends at work who are a great source of support"
Family	16/83	"The source of support is my father, mother and siblings"
Health service	6/83	"The hospital where I am getting the treatment is a place where I can have a great source of support"
Attitudes, feelings, and emotions		
Realistic or neutral attitudes	11/83	"I faced the cancer diagnosis well, and the cancer for me was a disease like any other"
Reserved personality	5/83	"I do not like to talk about my feelings"
General concerns	43/83	"I am anxious about the curative surgery in my breast"
Worry about future generations	17/83	"I want to do the genetic test to help my daughter and nieces"
Negative feelings	21/83	"Feel fear and anguish"
Self-esteem/self-image	4/83	"Sad in relation to my vanity since I do not feel pretty"
Cancer causes		
Stress and/or emotional aspects	14/83	"Cancer is related to sorrows and sufferings caused by past moments"
Chance or destiny	6/83	"Cancer is due to destiny and luck"
Genetics and family history	4/83	"Cancer is related to genetic aspects of the family"
Lifestyle	9/83	"Cigarettes and diet are related to cancer occurrence"
Other	1/83	"I am a good person, and for me, cancer comes to bad people. I do not understand what happened"
Communication with relatives		
Deprivation of communication	7/83	"I do not talk too much about my problems with my family. They do not talk too much"
Promotion of communication	13/83	"I talk about my problems with my family when I need, but I prefer to talk with my mother and husband"
Concern about privacy	5/83	"Family members always want to share information about my health condition"
Relationship with relatives	13/83	"I have a conflicting relationship with my first-degree relatives" "I have close and good relationships with my first-degree relatives"

experience of cancer, showed that risk perception is not only related to cognitive and level factors but also with family and personal experiences.[27]

While some studies demonstrate that women with a FH of cancer tend to overestimate their risk in general,[28-32] our sample did not reflect this behavior; in fact, they perceived their risk of breast and OC as being similar to that of the general population. One reason why they may have felt more reassured regarding risk is because all of them have had a previous cancer diagnosis, and the vast majority had already undergone curative surgeries, which can bring, in some cases, a false sense of relief. This explanation is consistent with the analyses between surgery and cancer risk perception, where women who underwent oophorectomy had lower perceptions of breast and OC risk. It is important for professionals to be aware of the factors that may influence cancer risk perceptions as patients' beliefs influence their risk management decisions.[33]

Corroborating the risk perception obtained data, the "susceptibility" domain had a lower score on the CHBM, reflecting the participants' low risk perception. Despite that, the CWS showed that worry about developing cancer again tended to be moderate to high.

Furthermore, on the CHBM, a higher mean was found in the "severity" domain, indicating that even if risk perception is not high, the worry about and the perception of the cancer severity is present, showing how risk perception is a complex issue that should be considered during the GCRA process.[34]

In our population, most women did not display symptoms of anxiety and/or depression. However, care should be taken to interpret these findings. An European study found that at the pre-GT moment there was not the presence of anxiety and depression symptoms.[35] On the other side, previous data from the literature showed that the HADS application is not enough to identify psychological alterations, and need the application of other tools to complement the evaluation.[36] This is why it is important to highlight the data from the qualitative analysis that reveal the presence of the "attitudes, feelings and emotions" category, which shows that the participants experience worries and negative feelings that might interfere with aspects of anxiety and depression.

A coping strategy identified in our sample was the religiosity (highest score at EMEP). These data corroborate with the genograms and ecomaps analyses that showed the "support and social support

network" category. A study conducted in the chemotherapy sector of the same hospital where this study was performed showed that patients have a high level of spirituality and consider addressing their current disease situation an important issue.[37] Another important point to be considered is that women with high levels of spirituality or religious are more negative about GT.[38,39] A study by Botoseneanu and coworkers showed that among women testing for *BRCA1/2*, those with high level of religiosity hold more negative attitudes toward GT.[39] In addition, Schwartz and coworkers[38] found that women with higher levels of spiritual faith were significantly less likely to be tested.

The presence of close relationships and the promotion of communication can facilitate information's transmission among family members and streamline prevention and control strategies. Moreover, family dialogue tends to reduce the anxieties generated by GT.[10] In our study we could identify the presence of conflicts with first-degree relatives, that can difficult the process of GCRA once information about cancer risk, prevention and GT results may not be disseminated.

The qualitative data showed that factors such as the presence of FH of cancer and the age of the individual can interfere with how individuals address situations and decide upon their health-disease processes. Participants who had no FH of cancer did not demonstrate "worry about future generations," do not have the "promotion of family communication" and do not consider the "health service" as a social support. This information is important for the GCRA because it can be more difficult to interact with women who are the first in their families to have such experiences.[9]

Besides, women in the age range of 20 to 29 years did not present the category "communication with relatives," but they did present "relationships with relatives," which suggests that even in a positive relationship with relatives, communication regarding attitudes of cancer prevention or GT may not occur. This group of women may block information from the family regarding cancer risk and preventive attitudes that are usually approached at GCRA.

In conclusion, this study characterized patients at risk for HBOC undergoing GCRA, showing that according to the group characteristics, the GCRA team should be prepared to different responses and with that improve communication, offering support in the decision-making process and may even reduce problems related to family dynamics and psychological distress.

4.1 | Study limitations

The instruments used to assess cancer risk perception, worry, among other measures, are not specific for GCRA. Despite that, the instruments have been validated for the Brazilian population, and, they result in an important information that was corroborated from qualitative data.

Although the center where the study took place receives patients from all Brazilian territory, this sample cannot be considered representative of the Brazilian population, and given the high level of diversity associated with the Brazilian people, caution should be exercised when extrapolating these findings to other regions or populations.

The cross-sectional study design has several limitations, as it cannot analyze behavior over a period of time, does not help determine cause and effect and, the timing evaluated could not be representative. It will be important for future studies to include a longitudinal design to mitigate these limitations.

4.2 | Clinical implications

It is important for health professionals to know the profile of the patients/families seeking GCRA. In this study, despite patients' great concern about developing cancer, most consider their risk of cancer equal to that of the general population. This situation can have potential implications for cancer prevention adherence.

The presence of religiosity to cope with problems, in our population, may influence health decisions. This finding supports the importance of health professionals to develop adequate communication strategies during the GCRA process.

To understand the family dynamics may facilitate the communication process among the proband, relatives, and health professionals allowing the professional to decide the best approach to convey health and genetic related information to each family.

CONFLICT OF INTEREST

The authors declare no potential conflict of interest.

DATA AVAILABILITY STATEMENT

The datasets used and/or analyzed during the current study are available from the corresponding authors on reasonable request.

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SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section at the end of this article.