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Contents

Executive Summary

New breast cancer gene-testing and the perspective of clinical application of the BOADICEA^{PLUS} developed within BRIDGES imply an increased complexity of communication during the cancer genetic consultation. Many individuals who undergo cancer genetic counseling experience specific cancer genetic counseling-related psychosocial needs (Eijzenga et al., 2015; Farrelly et al., 2013). These may potentially increase with evolving breast genetic risk assessment and so it is particularly timely and relevant to monitor the effects of these advances in breast cancer risk assessment on counselees' psychosocial needs. In the Netherlands, a 26-item "Psychosocial Assessment in Hereditary Cancer" (PAHC) questionnaire has been developed for screening counselees' needs in cancer genetics clinics (Eijzenga, Bleiker, et al., 2014). To our knowledge, no empirical validation of the PAHC construct has been published. BRIDGES WP5 psychosocial study was meant to extend the application of this questionnaire in other European countries. The purpose was to rigorously translate/ and adapt the PAHC questionnaire into French, German and Spanish and to validate the factorial structure in a sample of women at high genetic risk of breast and ovarian cancer. Women at high breast cancer genetic risk were recruited in three Family History Clinics (Institut Curie, Paris, France; University Hospital, Cologne, Germany; Oncology Institute, Barcelona, Spain). Women at high breast cancer genetic risk were recruited in three Family History Clinics (Institut Curie, Paris, France; University Hospital, Cologne, Germany; Oncology Institute, Barcelona, Spain). The first step involved the translation, adaptation and pilot-testing of the PAHC in France, Germany and Spain. The pilot-testing of the PAHC was carried out with 41 patients (15 patients in Germany and in France, 11 patients in Spain), who showed overall good understanding of the three PAHC language versions. The second step comprised the validation of the PAHC factorial structure (i.e., domains of psychosocial needs) aiming at providing a scoring procedure for PAHC scales. Between October 2016 and July 2017, 428 women participated in the validation study, of which 212 women were approached in France, 180 women in Germany and 36 women in Spain. Of the 428 participants, 356 women were affected with breast cancer and 71 women were healthy. An article is under preparation to report the results of this study. The Deliverable 5.2 has been achieved in time.

Introduction and Overview

Clinical and service delivery background

Next-generation sequencing, whole-exome and whole-genome sequencing, and "gene panel testing" are recent technological advances allowing far more genes to be simultaneously tested than the *BRCA1* and *BRCA2* alone, at a reduced cost and a faster turn-around. Gene panel testing has thus entered oncology genetic services over the past few years and the number of individuals eligible for genetic testing has increased.

Within BRIDGES, the "Breast and Ovarian Analysis of Disease Incidence and Carrier Estimation Algorithm" BOADICEA^{PLUS} model is being developed to calculate cancer risks from gene panel testing, taking into account the many additional variants associated with breast cancer such as the non-*BRCA1/2* breast cancer (NBBC) genes, which confer a range of risks from "moderate" (e.g., *PALB2*) to "high" (e.g., *TP53*, *PTEN*), and common "low risk" genetic variants (SNPs). In addition BOADICEA^{PLUS} will integrate

lifestyle factors, thus allowing for computing estimates based on multigenic as well as environmental factors.

Considering these different factors, using BOADICEA^{PLUS} in clinical practice is expected to increase the complexity of breast cancer risk counseling. Variations across countries have been reported in the process of genetic testing for familial/hereditary breast cancer varies (Gadzicki et al., 2011). In addition, the communication between the clinician and counselee may also vary in terms of the information and consent procedure (e.g., communication of anticipated test results, of cancer risks; cancer risk management recommendations). These variations in cancer genetics service delivery may affect psychosocial outcomes. Assessment of these outcomes across cancer genetics delivery systems and cultural background may provide useful clinical insights.

Psychosocial background

Psychosocial studies have shown that about 25% of individuals who undergo genetic counseling and testing for cancer report clinically relevant levels of distress (Eijzenga, Aaronson, et al., 2014; Farrelly et al., 2013; Brédart, et al., 2013; Power, Robinson, Bridge, Bernier, & Gilchrist, 2011). Although most individuals who undergo cancer genetic counseling do not present general anxious or depressive disorders, the majority of them experience cancer-specific genetic counseling-related psychosocial needs (Eijzenga et al., 2015), which may then accentuate psychological distress (Farrelly et al., 2013). It is thus essential to screen, assess and monitor these psychosocial repercussions and potential needs.

Assessing counselees' psychosocial needs

Needs assessment tools have been developed to directly weight the patient's wish for (additional) care intervention. the perception of the patient's care needs clarifies where actions or resource allocation are necessary, desirable or useful to help patients to overcome their difficulties (Brédart, Kop, Griesser, et al., 2013). In oncology, monitoring psychosocial needs has become standard practice in order to reduce emotional distress, enhance quality of life, patient satisfaction and to improve patient-clinician communication. Likewise, in cancer genetic services, such a standard practice should be developed in order to appropriately refer individuals who are in need for additional health care services (e.g., extra cancer genetic consultation in case of further information needs, psychological services in case of emotional or familial problems).

Recently, a 26-item "Psychosocial Assessment in Hereditary Cancer" (PAHC) questionnaire has been developed to assess psychosocial needs in cancer genetics services (Eijzenga, Bleiker, et al., 2014). This questionnaire is intended to address the psychosocial needs that are typically observed in individuals undergoing cancer genetics counseling and testing. This questionnaire has been evaluated for its screening performance (Eijzenga, Bleiker, et al., 2014). However, to our knowledge, no empirical validation of the PAHC has been published, thus its application for the comparative evaluation of initiatives to better respond to counselees' needs is limited. The PAHC assesses hypothetical domains of

needs (i.e., needs related to genetics information, practical issues, problems with family, living with cancer, emotions, and children). The empirical assessment of the PAHC dimensionality would allow providing a validated scoring procedure for PAHC measurement scales.

BRIDGES WP5 psychosocial study (**Task 5.4;** M24**)** is aimed at providing a PAHC questionnaire in three new language versions (i.e., in French, German, and Spanish), and at assessing the dimensionality of the PAHC across linguistic versions in order to provide a scoring procedure for validated scales of empirically-derived dimensions. This report presents results of the PAHC pilot-testing of the French-, German- and Spanish- versions, and an article is under preparation to report the full psychometric analyses of the PAHC data collected in the cancer genetic services of BRIDGES WP5 partners, i.e., in France at Curie Institute Paris and in Germany at Cologne University Hospital. Thanks to a doctoral student jointly supervised at the University of Barcelona and the University Paris Descartes (Anne Brédart), an additional European centre, the Català Oncology Institute of Barcelona, could be involved providing data from a Spanish cancer genetics clinic.

Science and Technology

The protocol development, patients & clinicians information/consent forms, case report form and questionnaires in three languages were prepared in the period between September 2015 and February 2016. The protocol was submitted to ethical committees in the respective countries. Approval was obtained by summer 2016 allowing the onset of the pilot-testing. For the pilot-testing, data was collected in August and September 2016, for the validation study, data collection was undertaken between October 2016 and July 2017.

Study design

This study was performed in two steps: 1) The translation and adaptation of the PAHC into French, German and Spanish as well as the pilot-testing of the adapted versions in the respective countries; 2) the PAHC validation involving patients' accrual in three Cancer Genetics Clinics (Institut Curie, Paris, France; University Hospital, Cologne, Germany; Oncology Institute, Barcelona, Spain).

Patient sample

Eligible women were referred to genetic counselling in the participating Cancer Genetic Clinics who comply with the following criteria: 1) affected with a primary non-metastatic breast cancer (BC, uni- or bilateral), 2) aged 18 or over with no upper limit, 3) able to give informed written consent in accordance with ICH GCP and national/local regulations and procedures, and 4) to understand the questionnaire language of the participating genetic clinic. The exclusion criteria are as follows: 1) recurrent/metastatic BC, ovarian cancer (OC) or cancer of any other site; 2) unable to answer the questionnaire due to physical or cognitive disturbance.

Procedure

Patients' assessments took place first after the initial (pre-test) cancer genetic consultation and after the genetic test result disclosure (post-test) consultation. Patients were face-to-face interviewed either at

the pre- or post-test result consultation. Among other questionnaires, the PAHC questionnaire was completed by patients at home within 4 weeks after the initial consultation, either in paper or online through the CleanWeb software. Questionnaires not completed within a time period of 4 weeks after the consultation were considered missing. One reminder was made 2 weeks after providing the PAHC at the initial consultation.

Step 1: Translation, adaptation and pilot-testing of the PAHC (face validity)

The translation/adaptation of the PAHC in French, German and Spanish language was performed using the forward-backward procedure recommended by the EORTC Quality of Life Group (Kuliś, Bottomley, Velikova, Greimel, & Koller, 2017). Translations from the published PAHC English version (Eijzenga, Bleiker, et al., 2014) into the targeted language involved two psychologists experienced in oncology by language. Backward translations were performed by professional translators of each language. Divergences between the original English and the back-translations were discussed among the research team of the three institutions.

The aim of the pilot-testing was to assess the PAHC questionnaire face validity, and identify and solve problems in its administration. Pilot-testing of each PAHC language version was performed with 10 to 15 patients. Patients were invited to complete the PAHC followed by a debriefing questionnaire asking if some items were found to be difficult/confusing, disturbing/intrusive, irrelevant/redundant or missing and if so to suggest reformulation.

Step 2: Factorial structure of the PAHC (construct validity)

The PAHC questionnaire and additional data collected

The questionnaire comprises 26 items conceptually organized into six problem domains, i.e., needs related to genetics information (1), practical issues (2), problems with family (3), living with cancer (4), emotions (5), and children (6) (Eijzenga, Bleiker, et al., 2014). A 27th item inquiring about additional potential issues that could be raised by patients was not included in the factor analyses as it has an open-question format. For each item, four modalities of response are provided: 'Not at all' (1), 'A little' (2), 'Quite a bit' (3), 'A lot' (4). For 8 items, an additional "Non-applicable" modality is also provided. The "Non applicable" response option was coded as "1". For 3 items concerning children, childless patients are invited to skip the section.

Additional data were collected including socio-demographic characteristics provided by the patient (i.e., age, education level, marital and professional status, parental status), and the clinical data obtained from the medical record (i.e., breast cancer diagnosis (yes/no), type, grade, date of breast cancer diagnosis, and breast cancer surgery).

Sample size

The sample size for psychometric analyses was targeted at women tested for genetic breast or ovarian cancer risk to be recruited in the French and German institutions partners of BRIDGES (Institut Curie,

Paris and University Hospital, Cologne). The primary endpoint of the study was to evaluate the hypothesized scale structure of the PAHC, composed of 26 items. To this end, Tabachnik & Fidell (2000) recommend 10 patients per item in the questionnaire, requiring a minimum of 260 participants. To account for attrition risk, an additional 20% of participants to this minimum figure were planned to be recruited leading to a required sample of 312 participants. In addition, we expected a sample of at least 50 to 100 patients from Spain.

Statistical analyses

Baseline patients' characteristics, including clinical and socio-demographics data, were described by country and for all participants. Qualitative variables were described using number (percentage). Quantitative variables were described using mean value (standard deviation) and median with range (min-max). Study participants and non-participants were compared using chi^2 or Student's *t*-test, depending on the nature of the variable.

Face validity was assessed using the debriefing questionnaire from step 1. Acceptability, floor and ceiling effects, exploratory factor analyses and reliability of the questionnaire were performed on the validation population from step 2. Acceptability of the questionnaire was assessed by the percentage of missing data (missing items and missing forms). A high proportion of missing forms may indicate poor acceptability of the instrument (Fayers & Machin, 2007). Information provided in the debriefing questionnaire in step 1 was also used to gain additional insights into the acceptability of the questionnaire.

In step 2, we performed a factor analysis on the 26 PAHC items. This analysis allows studying the structure of the questionnaire (number of important dimensions and links between items and scales/dimensions, represented by factor loadings). Scale reliability was then assessed using Cronbach's α coefficients. It was expected to be higher than 0.70 (Cronbach, 1951).

The number and percentage of patients who obtained the lowest (floor effect) or highest (ceiling effect) possible score for each scale was computed. Floor or ceiling effects were considered to be present if more than 15% of the responders achieved the lowest or highest possible score respectively (Terwee et al., 2007).

Results

Step 1: Translation/adaptation and pilot-testing of the PAHC (face validity)

Translation/adaptation

Some words or expression could not be translated literally because either no corresponding word was found or because it would not represent the intended meaning. Examples of linguistic adaptations are provided below for the three languages.

French

- The word “risk” was translated as “possibilité” rather than “risque” because “risque” has a negative connotation;
- In items 1 to 5, “worried” was translated by ‘inquiète’, ‘tiraillée’ or ‘préoccupée’ as different nuances of feeling seemed more linguistically appropriate;
- Item 2 was translated into a question asking respondents to indicate their choice of undergoing genetic testing, excluding the participation in genetic counselling, since, in pilot-testing, this item was found to comprise two questions into one item;
- In item 10, “family functioning” was translated literally because no other word better representing the intended meaning could have been found; however, this word can be understood either as relational or practical functioning;
- In Item 11, “contact” could be understood as reaching a person (e.g., to obtain information) yet, since “relationships” has been the intended meaning, it has been translated into “les relations”;
- For items 13, 19, 20 and 21, the words “burdened” or “burdensome” were translated into “préoccupée”, “perturbée” or “affectée” as different nuances of feeling seemed more appropriate.

German

- In item 10, “family functioning” was translated into “family process”;
- The word “burden” or “responsibility” was considered to be too strong or negative;
- When referring to one’s family, whether the nuclear or extended family is meant, could not be specified by respondents. Nevertheless, the expression “family” has remained.

Spanish

- Item 7 was kept as in the original language version, but this question may not be relevant to the Spanish social and healthcare system;
- As in French, in item 11, “contact” could be understood as reaching a person (e.g., to obtain information) yet, since “relationships” has been the intended meaning, it has been translated into “les relations”;
- Item 12 was back-translated as follows: **“**To what extent are you worried about how your relatives will deal with a potential cancer onset in the future, in your family?” The addition of the word “potential” was found to be more acceptable in Spanish;
- Item 18 was kept as in the original language version; however, the acceptability of this item in Spanish patients was discussed among translators (i.e., the adequacy and ethical aspects of evoking/priming death-related thoughts in the setting of genetic counselling); an alternative

formulation ("Do you have questions about the sense of your life?") was suggested for pilot-testing with patients;

- Items 21 and 22 were translated literally; however, the formulation using a conditional clause was discussed; an alternative formulation was suggested for pilot-testing ("If you have been diagnosed with cancer, to what extent did/do the diagnosis and cancer treatment have an emotional impact for you?"

Three language versions

Changing moods and worries along the genetic counselling and testing process was underlined. As the PAHC was expected to be applicable at any stage along the cancer genetics counselling and testing process (i.e., from initial attendance to a cancer genetic consultation to attendance of a consultation for risk management discussion and follow-up), for items 1 to 3 ("Worry about the chance of being a carrier"; "Worry about the choice of testing"; "Worry about the choice of risk management"), a "not applicable" response option was suggested.

In contrast to the English version, the term "psychosocial worker" was translated as "psychologist" because women are referred to psychologists rather than social workers in case of psychosocial needs in France, Germany, and Spain.

Pilot-testing results

The socio-demographic and clinical characteristics of patients participating in the PAHC French, German and Spanish versions pilot-testing are provided in Table 1.

Table 1. Socio-demographic and clinical characteristics of patients participating in the PAHC French, German and Spanish versions pilot-testing

		French (N=15)	German (N=15)	Spanish (N=11)
Age (years)	Mean (SD)	44 (9)	46 (11)	49 (17)
	Range	33-61	29-65	29-71
Personal breast cancer	Yes	13 (87)	15 (100)	7 (64)
Education level	High (secondary) school or below	5 (33)	4 (27)	7 (64)
	Superior (post-secondary) school or above	10 (66)	11 (73)	4 (36)
Cancer genetics counselling stage	Initial (pre-test) consultation	11 (73)	15 (100)	5 (46)
	Genetic test disclosure consultation	4 (27)	-	6 (55)

Overall the PAHC was perceived as clear, comprehensive and useful. Respondents considered the PAHC to comprise issues that may be raised to the counselee at some time in the cancer genetics process. If an item in one of the three language versions was considered difficult to understand…this was mostly the case for only one respondent.

As shown in Table 2 and Appendix 2, eight items were commented by a range of 2 to 5 patients without requesting modification regarding the original PAHC as well as the three adapted versions.

In the PAHC German language version, **item 5** ("Worry about fulfilling plans for having children") was perceived as difficult/confusing by 4 patients and **items 7** ("Impact of testing on obtaining insurance/mortgage") and **13** ("Feelings of responsibility towards family members") were perceived as disturbing/intrusive by 3 patients.

Concerning item 7 ("Worried about the impact of genetic testing on obtaining insurance or mortgage"), some French and Spanish patients indicated that this item was not appropriate regarding their national health care system.

For the PAHC French version, **item 10** ("Worried about immediate family's functioning because of genetic testing") was perceived as difficult/confusing by 2 patients who had difficulties in understanding the full meaning of the expression "family functioning", since it could be related to both family relationships and practical or structural aspects; **items 14 to 16** ("Feel anxious"; "Feel tense"; "Feel depressed") were perceived as redundant by 5 and 4 patients; this was also expressed by one patient in Germany.

Respondents indicated that some specific issues were missing however these issues could underlie questions already in the PAHC (e.g., question on children need for help; worry about prophylactic surgery; organization of psychological help) or which could be raised on the 27th PAHC item ("other issues").

Table 2. Summary of the PAHC pilot-testing results

Item debriefing feed-back	Language	Item (number of patients)
Difficult to understand/confusing	French	3 (1); 7 (1); 8 (1); 10 (1); 12 (1); 17 (1); 18 (1); 23 (1); 25 (1)
	German	1 (2); 3 (1); 5 (4); 8 (1)
	Spanish	7 (1); 10 (1); 11 (2); 18 (1); 19 (1); 20 (1); 21 (1)
Disturbing/Intrusive	French	8 (1); 18 (1)
	German	4 (1); 7 (3); 12 (2); 13 (3); 18 (1); 21 (1)
	Spanish	7 (1)
Redundant	French	14 & 15 (5) & 16 (4); 17 (1)
	German	8 & 9 (1); 14 & 15 (1); 19 & 20
	Spanish	8 (1); 9 (1)

Irrelevant	French	2 (1); 4 (1); 6 (1); 7 (3); 10 (1); 21 (1)
	German	10 (1)
	Spanish	7 (2); 24 (1)
Missing	French	7 issues
	German	9 issues
	Spanish	-
Other comments	French	10; 18
	German	3; 5; 6; 8; 9; 12; 13; 24; 25
	Spanish	7; 8; 9; 11; 18; 21

Step 2: Factorial structure of the PAHC (construct validity)

For this step, at present we only report the acceptability of the PAHC. The full psychometric analyses results are reported in an article under preparation.

Acceptability of the PAHC

Among the 495 participants included in the study, 428 (86.5%) returned the questionnaires; 351 respondents (82%) fully completed the 26 items of the PAHC[1] and only 29 (6.8%) had 2 or more missing response in the questionnaire. On the whole, there were only few missing data, with the highest rate observed for the 3 items concerning worries about their close family's functioning because of genetic testing (item 10: 3.7%), feeling of being misunderstood by their partner/family/social circle with respect to genetic testing (item 8: 3.5%) and worries about the impact of genetic testing on obtaining insurance or mortgage (item 7: 3.0%). Less than 3% of missing data were observed from other items.

Discussion and Conclusion

In the BRIDGES WP5 psychosocial study (Task 5.4; M24) we have provided adapted versions of the PAHC in French, German, and Spanish language. This study highlighted the overall questionnaire acceptability reflected by the respondents' high comprehension of the items as well as low missing responses/data rates. The Deliverable 5.2 has been achieved in time.

This study comprised a large sample of women in three European regions attending genetic counseling for breast cancer risk testing. The sample is characterized by a limited number of participants with a lower level of education (below secondary school). However, in the pilot-testing phase of the study, we paid particular attention to achieving a simple item wording in each language in order to allow the PAHC to be applicable independent of the counselees' level of education.

Following rigorous translation and adaptation, we provided a French-, German- and Spanish-language version of a questionnaire designed to assess psychosocial needs in cancer genetic testing and counseling, the PAHC. Data collected in women attending an initial breast cancer genetic risk counseling consultation were used to assess the factorial structure of the PAHC in these three language versions.

[1] Excluding non-responses for items about children, when the participant had no child

Six scales and a scoring procedure were empirically derived to allow the measuring of domains of psychosocial needs. These will be reported in a separate article currently in preparation.

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Appendix

Appendix 1. PAHC language versions

Psychosocial Assessment in Hereditary Cancer (PAHC) – English version*

To help us plan better services for people attending cancer genetic consultation, we are interested in whether or not needs which you may face as a result of cancer risk have been met during the cancer genetic consultation you have had. For every item in each domain, please circle the number which best describes whether you have had difficulties or needs with this in the last week. Some questions may not be applicable to you, then answer NA (Not applicable)

		Not at all	A little	Quite a bit	Very much	
1.	Are you worried about the chance of being a carrier of a genetic mutation?	1	2	3	4	
2.	Are you worried about having to choose whether or not to go for genetic counselling and testing?	1	2	3	4	
3.	Are you worried about the choice of possible preventive options (screening or surgery)?	1	2	3	4	
4.	Are you worried about coping with the (future) DNA test results?	1	2	3	4	

5.	Are you worried about (fulfilling) your plans for having children?	1	2	3	4	NA
	Would you like to speak with a psychosocial worker in addition to the clinical geneticist/genetic counsellor about these issues 1 to 5?	Yes / No				
6.	Are you worried about the impact of genetic testing on your daily life (at home, at work, at school, or with hobbies)?	1	2	3	4	
7.	Are you worried about the impact of genetic testing on obtaining insurance or mortgage?	1	2	3	4	
	Would you like to speak with a psychosocial worker in addition to the clinical geneticist/genetic counsellor about these issues 6 to 7?	Yes / No				
8.	Do you feel misunderstood by your partner/family/social circle with respect to genetic testing?	1	2	3	4	
9.	Are you bothered by lack of support about genetic testing from your partner, family, or your social circle?	1	2	3	4	
10.	Are you worried about your immediate family's functioning because of genetic testing?	1	2	3	4	
11.	Are you worried about the contact with family members about genetic testing?	1	2	3	4	
12.	Are you worried about coping with cancer within the family?	1	2	3	4	NA
13.	Are you burdened by feelings of responsibility towards family members related to genetic testing?	1	2	3	4	
	Would you like to speak with a psychosocial worker in addition to the clinical geneticist/genetic counsellor about these issues 8 to 13?					
14.	Do you feel anxious?					
15.	Do you feel tense?					
16.	Do you feel depressed?					
17.	Do you feel insecure about the future?					
18.	Do you have questions about life and death?					
	Would you like to speak with a psychosocial worker in addition to the clinical geneticist/genetic counsellor about these issues 14 to 18?	Yes / No				
19.	How emotionally burdensome is it for you that family members have cancer?					NA

20.	How emotionally burdensome is losing a family member because of cancer?					NA
21.	How emotionally burdensome is your diagnosis or treatment for cancer?					NA
22.	Are you worried about the chance of getting cancer (again)?					
23.	Are you worried about the chance that family members will get cancer?					
	Would you like to speak with a psychosocial worker in addition to the clinical geneticist/genetic counsellor about these issues 19 to 23?	Yes / No				
	If you have children (if you do not have children please proceed to question 27)					
24.	Do you feel guilty about the chance of passing on to your children your possible genetic alterations?					
25.	Are you worried about telling your children the results?					
26.	Are you worried about the chance of your children developing cancer?					
	Would you like to speak with a psychosocial worker in addition to the clinical geneticist/genetic counsellor about these issues 24 to 26?	Yes / No				
27.	Are there any other issues related to genetic testing that bother you or that you are worried about? If yes, which issues?					
	Would you like to speak with a psychosocial worker in addition to the clinical geneticist/genetic counsellor about these issues?	Yes / No				

*Eijzenga, W., E. M. Bleiker, et al. (2014). "Psychosocial Aspects of Hereditary Cancer (PAHC) questionnaire: development and testing of a screening questionnaire for use in clinical cancer genetics." Psychooncology 23(8): 862-9.

Psychosocial Assessment in Hereditary Cancer (PAHC) – French version* V1.0

Le questionnaire ci-dessous s'intéresse aux difficultés ou besoins psychologiques et sociaux en lien avec la démarche de test génétique. Veuillez, s'il vous plaît, entourer pour chaque question le chiffre correspondant à votre situation au cours de la semaine passée. Il se peut que quelques questions ne vous concernent pas ou plus. Dans ce cas, veuillez entourer la réponse NA. NA=Non applicable à ma situation

		Pas du tout	**Un peu**	**Assez**	**Beaucoup**	
1.	Vous êtes-vous sentie inquiète de la possibilité d'être porteuse d'une mutation génétique ?	1	2	3	4	NA
2.	Vous sentiez-vous tiraillée par le choix de réaliser ou non un test génétique ?	1	2	3	4	NA
3.	Vous sentez-vous tiraillée entre les choix de mesures préventives (dépistage ou chirurgie)?	1	2	3	4	NA
4.	Vous sentez-vous préoccupée au sujet de votre réaction face au résultat du test génétique?	1	2	3	4	
5.	Vous sentez-vous préoccupée au sujet de votre projet d'enfants?	1	2	3	4	NA
	Souhaitez-vous échanger avec un psychologue, en complément de votre consultation avec le généticien/conseiller en oncogénétique, au sujet de ces questions 1 à 5?	Oui / Non				
6.	Vous sentez-vous préoccupée au sujet de l'impact du résultat du test génétique sur votre vie quotidienne (à la maison, au travail, pour les études ou pour les loisirs)?	1	2	3	4	
7.	Vous sentez-vous préoccupée au sujet de l'impact du résultat du test génétique sur le fait d'obtenir une assurance ou un prêt?	1	2	3	4	
	Souhaitez-vous échanger avec un psychologue, en complément de votre consultation avec le généticien/conseiller en oncogénétique, au sujet de ces questions 6 et 7?	Oui / Non				
8.	Vous sentez-vous incomprise par votre partenaire, entourage familial ou social en ce qui concerne le test génétique?	1	2	3	4	
9.	Vous sentez-vous préoccupée par le manque de soutien de la part de votre partenaire, entourage familial ou social au sujet du test génétique?	1	2	3	4	
10.	Vous sentez-vous préoccupée par l'impact du test génétique sur votre l'organisation familiale?	1	2	3	4	

11.	Vous sentez-vous préoccupée par les relations avec les membres de votre famille au sujet du test génétique?	1	2	3	4	
12.	Vous sentez-vous préoccupée à l'idée de faire face à un cancer chez un membre de votre famille?	1	2	3	4	NA
13.	Vous sentez-vous préoccupée par un sentiment de responsabilité vis-à-vis des membres de la famille à propos du test génétique?	1	2	3	4	
	Souhaitez-vous échanger avec un psychologue, en complément de votre consultation avec le généticien/conseiller en oncogénétique, au sujet de ces questions 8 à 13?	Oui / Non				
14.	Vous sentez-vous anxieuse?					
15.	Vous sentez-vous tendue?					
16.	Vous sentez-vous déprimée?					
17.	Ressentez-vous de l'insécurité par rapport à l'avenir?					
18.	Vous posez-vous des questions au sujet de la vie et de la mort?					
	Souhaitez-vous échanger avec un psychologue, en complément de votre consultation avec le généticien/conseiller en oncogénétique, au sujet de ces questions 14 à 18?	Oui / Non				
19.	Dans quelle mesure vous sentez-vous émotionnellement perturbée du fait que des membres de votre famille soient touchés par un cancer?					NA
20.	Dans quelle mesure la perte d'un membre de votre famille d'un cancer vous affecte-t-elle émotionnellement ?					NA
21.	Dans quelle mesure vous sentez-vous émotionnellement perturbée par le diagnostic ou le traitement de votre cancer?					NA
22.	Vous sentez-vous préoccupée par la possibilité de développer (à nouveau) un cancer?					
23.	Vous sentez-vous préoccupée par la possibilité qu'un membre de votre famille développe un cancer?					

	Souhaitez-vous échanger avec un psychologue, en complément de votre consultation avec le généticien/conseiller en oncogénétique, au sujet de ces questions 19 à 23?	Oui / Non				
	Si vous avez des enfants (si vous n'en avez pas, vous pouvez passer à la question 27)					
24.	Vous sentez-vous coupable de la possibilité d'avoir transmis une éventuelle altération génétique à vos enfants?					
25.	Vous sentez-vous préoccupée de communiquer le résultat de test génétique à vos enfants?					
26.	Vous sentez-vous inquiète de la possibilité pour vos enfants de développer un cancer?					
	Souhaitez-vous parler avec un psychologue, en complément de votre consultation avec le généticien/conseiller en oncogénétique, au sujet de ces questions 24 à 26?	Oui / Non				
27.	Est-ce que d'autres aspects liés au test génétique vous sont difficiles ou vous préoccupent? Si oui, lesquels?.....					
	Souhaitez-vous échanger avec un psychologue, en complément de votre consultation avec le généticien/conseiller en oncogénétique, au sujet de ces questions?	Oui / Non				

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Psychosocial Assessment in Hereditary Cancer (PAHC) – German version* V1.0

Wir benötigen wir Ihre Hilfe um herauszufinden, ob Ihre Bedürfnisse und Fragen in der ärztlichen Beratung zur genetischen Testung besprochen wurden.
Bitte kreisen Sie bei jeder der folgenden Fragen die Zahl ein, die am besten Ihre Situation innerhalb der **letzten Woche** beschreibt. Einige Fragen betreffen Sie möglicherweise nicht. In diesem Fall kreuzen Sie bitte "n.z." (nicht zutreffend) an.

		Gar nicht	**Etwas**	**Ziemlich**	**Sehr**	**Nicht zutreffemd**
1.	Machen Sie sich Sorgen über die Möglichkeit, Trägerin einer genetischen Mutation zu sein?	1	2	3	4	NZ
2.	Machen Sie sich Sorgen darüber, sich entscheiden zu müssen, eine genetische Beratung oder Testung in Anspruch zu nehmen oder nicht?	1	2	3	4	NZ
3.	Machen Sie sich Sorgen über die Wahl einer möglichen präventiven Maßnahme (Früherkennung oder Operation)?	1	2	3	4	NZ
4.	Machen Sie sich Sorgen, mit dem Gentestergebnis umgehen zu können?	1	2	3	4	
5.	Machen sie sich Sorgen darüber, ob Sie Ihren Kinderwunsch erfüllen können?	1	2	3	4	NZ
	Möchten Sie in Bezug auf die Fragen 1-5 zusätzlich zu der ärztlichen Beratung eine psychologische Beratung in Anspruch nehmen?	Ja / Nein				
6.	Sind Sie darüber besorgt, dass der Gentest Ihr Alltagsleben beeinflusst? (z.B. zu Hause, im Beruf, in der Schule oder in der Freizeit)	1	2	3	4	
7.	Sind Sie besorgt, dass der Gentest einen Einfluss auf den Abschluss einer Versicherung oder eines Hypothekendarlehens haben wird?	1	2	3	4	
	Möchten Sie in Bezug auf die Fragen 6 und 7 zusätzlich zu der ärztlichen Beratung eine psychologische Beratung in Anspruch nehmen?	Ja / Nein				
8.	Fühlen Sie sich in Bezug auf die Gentestung unverstanden von Ihrem Partner, Ihrer Familie oder Ihrem sozialen Umfeld?	1	2	3	4	
9.	Fehlt Ihnen im Umgang mit dem Gentest Unterstützung von Ihrem Partner, Ihrer Familie oder Ihrem sozialen Umfeld?	1	2	3	4	
10.	Sind Sie darüber besorgt, dass der Gentest die Abläufe in Ihrem engeren Familienkreis belastet?	1	2	3	4	

11.	Sind sie darüber besorgt, dass der Gentest die familiären Beziehungen belastet?	1	2	3	4	
12.	Wie sehr sind Sie darüber besorgt, mit Krebserkrankungen in Ihrer Familie zurecht zu kommen?	1	2	3	4	NZ
13.	Sind Sie belastet, weil Sie sich aufgrund der genetischen Testung gegenüber Ihren Familienmitgliedern verantwortlich fühlen?	1	2	3	4	
	Möchten Sie in Bezug auf die Fragen 8-13 zusätzlich zu der ärztlichen Beratung eine psychologische Beratung in Anspruch nehmen?	Ja / Nein				
14.	Fühlen Sie sich ängstlich?					
15.	Fühlen Sie sich angespannt?					
16.	Fühlen Sie sich deprimiert?					
17.	Fühlen Sie sich in Bezug auf die Zukunft verunsichert?					
18.	Stellen Sie sich Fragen zu Leben und Tod?					
	Möchten Sie in Bezug auf die Fragen 14-18 zusätzlich zu der ärztlichen Beratung eine psychologische Beratung in Anspruch nehmen?	Ja / Nein				
19.	Fühlen Sie sich durch Krebserkrankungen in Ihrer Familie emotional belastet?					NZ
20.	Wie belastend ist es für Sie, ein Familienmitglied durch Krebs zu verlieren?					NZ
21.	Wie belastend ist die Diagnose und Behandlung Ihrer Krebserkrankung?					NZ
22.	Machen Sie sich Sorgen um Ihr Krebserkrankungsrisiko (Wiedererkrankungsrisiko)?					
23.	Sind Sie darüber besorgt, dass Familienangehörige an Krebs erkranken könnten?					
	Möchten Sie in Bezug auf die Fragen 19-23 zusätzlich zu der ärztlichen Beratung eine psychologische Beratung in Anspruch nehmen?	Ja / Nein				
	Falls Sie Kinder haben, beantworten Sie bitte die Fragen 22 bis 24. Wenn Sie keine Kinder haben, bitte fahren Sie mit Frage 25 fort.					
24.	Fühlen Sie sich schuldig, dass Sie eine Genmutation an Ihre Kinder weitergegeben haben könnten?					

25.	Sind Sie darüber besorgt, das Gentestergebnis Ihren Kindern mitzuteilen?					
26.	Sind Sie darüber besorgt, dass Ihre Kinder an Krebs erkranken könnten?					
	Möchten Sie in Bezug auf die Fragen 24-26 zusätzlich zu der ärztlichen Beratung eine psychologische Beratung in Anspruch nehmen?	Ja / Nein				
27.	Gibt es weitere Fragen im Zusammenhang mit der Gentestung, die Sie beschäftigen oder die Ihnen Sorgen bereiten? Wenn ja, welche?					
	Möchten Sie in Bezug hierauf zusätzlich zu der ärztlichen Beratung eine psychologische Beratung in Anspruch nehmen?	Ja / Nein				

**Eijzenga, W., E. M. Bleiker, et al. (2014). "Psychosocial Aspects of Hereditary Cancer (PAHC) questionnaire: development and testing of a screening questionnaire for use in clinical cancer genetics." Psychooncology 23(8): 862-9. Translated by Sherer Anne, Family History Clinic, Cologne University Hospital, Cologne, Germany, 2016*

Psychosocial Assessment in Hereditary Cancer (PAHC) – Spanish version* V1.0

Con el objetivo de mejorar la atención a las personas que realizan una consulta de asesoramiento genético, nos interesa saber en qué medida las necesidades que le han surgido como resultado de su riesgo de cáncer han sido atendidas durante dicha consulta. Rodee con un círculo el número que mejor describa el grado en el que ha tenido dificultades o necesidades en los siguientes aspectos a los largo de la última semana. Algunas preguntas puede que no se apliquen a su caso particular, responda entonces NA (No aplicable).

		Not at all	A little	Quite a bit	Very much	
1.	¿En qué medida se siente preocupada ante la posibilidad de ser portador de una mutación genética?	1	2	3	4	NA
2.	¿En qué medida se siente preocupada ante la decisión de acudir o no a la consulta de asesoramiento genético y realizar un estudio genético?	1	2	3	4	NA
3.	¿En qué medida se siente preocupada por el hecho de decidir si realizar posibles medidas preventivas de cáncer (revisiones médicas o cirugía profiláctica)?	1	2	3	4	NA
4.	¿En qué medida se siente preocupada de cómo reaccionará ante los resultados del estudio genético?	1	2	3	4	
5.	¿En qué medida le preocupa poder cumplir sus planes de tener hijos?	1	2	3	4	NA
	Además de la visita con el médico o asesor/a genético, ¿estaría interesada en poder hablar con un psicólogo acerca de las preguntas 1 a 5?	Sí / No				
6.	¿En qué medida se siente preocupada de que los resultados del estudio genético puedan afectar su vida diaria (familia, trabajo, estudios, ocio)?	1	2	3	4	
7.	¿En qué medida se siente preocupada de que los resultados del estudio genético puedan influir negativamente a la hora de obtener o no un seguro o una hipoteca?	1	2	3	4	
	Además de la visita con el médico o asesor/a genético, ¿estaría interesada en poder hablar con un psicólogo acerca las preguntas 6 y 7?	Sí / No				
8.	¿En qué medida se siente incomprendida por su pareja, familia o amigos con relación a su interés por el estudio genético?	1	2	3	4	

9.	¿En qué medida se siente molesta por la falta de apoyo con relación al estudio genético por parte de su pareja, familia o amigos?	1	2	3	4	
10.	¿En qué medida se siente preocupada por el funcionamiento normal de su familia con respecto al estudio genético?	1	2	3	4	
11.	¿En qué medida le preocupa la relación con los miembros de su familia con respecto al test genético?	1	2	3	4	
12.	¿En qué medida se siente preocupada por cómo su familia afronta el cáncer?	1	2	3	4	NA
13.	¿En qué medida se siente sobrecargada por la responsabilidad que implica el hecho de realizar el estudio genético de cara a su familia?	1	2	3	4	
	Además de la visita con el médico o asesor/a genético, ¿estaría interesada en poder hablar con un psicólogo acerca de las preguntas 8 a 13?	Sí / No				
14.	¿Se siente nervioso?					
15.	¿Se siente tenso?					
16.	¿Se siente deprimido?					
17.	¿Se siente inseguro acerca del futuro?					
18.	¿Le surgen a veces preguntas sobre la vida y la muerte?					
	Además de la visita con el médico o asesor/a genético, ¿estaría interesado/a en poder hablar con un psicólogo acerca de las preguntas 14 a 18?	Sí / No				
19.	¿En qué medida le afectaría emocionalmente que un miembro de su familia fuera diagnosticado de cáncer?					NA
20.	¿En qué medida le afectaría emocionalmente la pérdida de un miembro de su familia debido al cáncer?					NA
21.	¿En qué medida le ha afectado emocionalmente su diagnóstico y tratamientos para el cáncer?					NA
22.	¿En qué medida se siente preocupada por el riesgo de padecer cáncer (otra vez)?					
23.	¿En qué medida le preocupa la posibilidad de que un miembro de su familia pueda tener cáncer?					

	Además de la visita con el médico o asesor/a genético, ¿estaría interesado/a en poder hablar con un psicólogo acerca de las preguntas 19 a 23?	Sí / No				
	En caso de que tenga hijos (si no los tiene pase directamente por favor a la pregunta 27):					
24.	¿En qué medida se siente culpable ante la posibilidad de transmitir la posible mutación genética a sus hijos?					
25.	¿En qué medida se siente preocupada por tener que decirle a sus hijos los resultados del estudio genético?					
26.	¿En qué medida le preocupa laposibilidad de que sus hijos puedantener cáncer?					
	Además de la visita con el médico o asesor/a genético, ¿estaría interesado/a en poder hablar con un psicólogo acerca de los aspectos anteriores?	Sí / No				
27.	¿Encuentra otros aspectos relacionados con el estudio genético que le molesten o le preocupen? Si es así, ¿cuáles son?					
	Además de la visita con el médico o asesor/a genético, ¿estaría interesada en poder hablar con un psicólogo acerca de los aspectos anteriores?	Sí / No				

**Eijzenga, W., E. M. Bleiker, et al. (2014). "Psychosocial Aspects of Hereditary Cancer (PAHC) questionnaire: development and testing of a screening questionnaire for use in clinical cancer genetics." Psychooncology 23(8): 862-9.* Traducción española Gil, Cano, Institut Català, Universitat AutÒnoma de Barcelona, 2015

Appendix 2. PAHC pilot-testing detailed results

		French (N=15)	**German (N=15)**	**Spanish (N=11)**
1.	Are you worried about the chance of being a carrier of a genetic mutation?	-	Difficult/confusing: N=2 (Need for timeframe of worry)	-
2.	Are you worried about having to choose whether or not to go for genetic counseling and testing?	Relevance depend on the time in the cancer genetic counseling trajectory – May not be relevant after initial genetic counseling	Irrelevant: N=1	-
3.	Are you worried about the choice of possible preventive options (screening or surgery)?	Difficult/confusing: N=1 – word possible not necessary	Difficult/confusing: N=1 (word "option") Comments: "Is it difficult for you to decide on a preventive option" -> two questions (one about decision difficulty, one about worry)	-
4.	Are you worried about coping with the (future) DNA test results?	Irrelevant: N=1 (No doubt about wish to be tested)	Disturbing/intrusive: N=1 (Better to say expected result)	-
5.	Are you worried about (fulfilling) your plans for having children?	-	Difficult/confusing: N=4 Comments: Would prefer a positive wording	-

6.	Are you worried about the impact of genetic testing on your daily life (at home, at work, at school, or with hobbies)?	Irrelevant: N=1	Comments: Two questions, work and private life separately (N=2)	-
7.	Are you worried about the impact of genetic testing on obtaining insurance or mortgage?	Difficult/confusing: N=1 - Irrelevant: N=3	Disturbing/intrusive: Q about mortgage; had never thought of before (N=3)	Disturbing: N=1/confusing: N=1/ irrelevant: N=2 Comments: Doesn't adjust with the Spanish social and healthcare system given that there is no obligation to disclose such information Word impact has a negative connotation so translation adaptation made
8.	Do you feel misunderstood by your partner/family/social circle with respect to genetic testing?	Difficult/confusing: N=1 Disturbing/intrusive: N=1	Difficult/confusing: N=1 (core family or whole family) Comments: Reverse polarity of response; More positive response (N=3)	Redundant: N=1 Comments: Found less important – Would not expected that family or social environment would not support decision about genetic counseling or this being an obstacle to them
9.	Are you bothered by lack of support about genetic testing from your partner, family, or your social circle?	-	Redundant: Q8&Q9 (N=1) - Comments: Reverse polarity of response; More positive response (N=3); Separate Q for partner, family, social circle (N=1)	Redundant: N=1 Comments: Found less important- Would not expected that family or social environment would not support decision about genetic counseling or this being an obstacle to them

10.	Are you worried about your immediate family's functioning because of genetic testing?	Difficult/confusing: N=2 Comment: don't see why genetic test may affect family functioning (N=2) Functioning translated by word "organisation"	Irrelevant/redundant: N=1 Functioning translated by word "Abläufe" (≈ process, routine)	Confusing: N=1
11.	Are you worried about the contact with family members about genetic testing?	- Contact to be translated by word "relationship"	-	Difficult/confusing: whether here 'contact' means 'searching for getting hold of someone', or the 'action itself of interacting with others' Contact to be translated by word "relationship"
12.	Are you worried about coping with cancer within the family?	Difficult/confusing: N=1 - Comment: cancer current or past?	Disturbing/intrusive: N=2 Comments: Didn't think about it, but have to now!	-
13.	Are you burdened by feelings of responsibility towards family members related to genetic testing?	-	Disturbing/intrusive: N=3 Comments: Don't use guilty; my genes are not my fault	-
14.	Do you feel anxious?	Redundant: N=5	Irrelevant/redundant: Q14 &Q15 (N=1; tense/anxious similar)	-
15.	Do you feel tense?	Redundant: N=5	-	-
16.	Do you feel depressed?	Redundant: N=4	-	-
17.	Do you feel insecure about the future?	Difficult/confusing: N=1 Redundant: N=1	-	-

18.	Do you have questions about life and death?	Difficult/confusing: N=1 Disturbing/intrusive: N=1 Comment: difficult subject	Disturbing/intrusive: N=1 Comments: Difficult Q; because of family, not because of test	Difficult: N=1 Comments: debate about how adequate/ethical is to induce death-related thoughts in the setting of genetic counseling. Would like to propose a more existentialism-orientated question like 'Do you have questions about the sense of your life?'
19.	How emotionally burdensome is it for you that family members have cancer?	-	Comments: Q19 & Q20 redundant	Difficult: N=1
20.	How emotionally burdensome is losing a family member because of cancer?	-	-	Difficult: N=1
21.	How emotionally burdensome is your diagnosis or treatment for cancer?	Irrelevant: N=1	Disturbing/intrusive: N=1	Difficult/confusing: N=1 Comments: Which time frame (since they had gone through different mood states, and past and present feelings were intertwined); Make two separate questions Translation: add “possible” cancer diagnosis
22.	Are you worried about the chance of getting cancer (again)?	-	-	-
23.	Are you worried about the chance that family members will get cancer?	Difficult/confusing: N=1	-	Comments: the impact of cancer in their family members to be their major concern

24.	Do you feel guilty about the chance of passing on to your children your possible genetic alterations?	-	Comment: difficult because of guilt; better use the word “responsible”	Comments: Found less important
25.	Are you worried about telling your children the results?	Difficult/confusing: N=1	Comment: difficult because of guilt/responsibility	-
26.	Are you worried about the chance of your children developing cancer?	-	-	-
	Perceived item missing	Question on help for children; worry about prophylactic surgery; worry about multiple gene testing and other cancer risks; guilt about transmission; family communication as a barrier for testing; when possible to ask for psychological help; impact of salpingo-oophorectomy on child bearing	Question about information material; who supports me; where do I get help; questions about medical tests; how long it will take to decide; cancer risk in the future is hard to understand; advantages of prophylactic mastectomy; whether need for chemotherapy if tumour diagnosed; question about impact on job, charity work, other duties in society	-
	Overall comments	Very comprehensive; very clear, relevant, useful; some questions not relevant now but could be relevant later - Comments: Q14-Q16 redundant (N=2); why ask whether need for psychological help after each question and not only at the end; Did not think of some questions because had not yet received test result, e.g., guilt for children, but now worry about these questions.	Comments: Have separate questions for family, husband, daughters (N=1); All very negative/have more positive wording, e.g., responsibility vs guilt (N=3); Very comprehensive (N=1); Too many Q on family is upsetting (N=1); Need well defined timeframe (N=2); Reverse response polarity on family Q	Comments: Items about interest to speak with a psychologist or other mental health professionals appear redundant