European Journal of Human Genetics (2021) 29:250–261
https://doi.org/10.1038/s41431-020-00723-7

ARTICLE

Intrafamilial communication of hereditary breast and ovarian cancer genetic information in Italian women: towards a personalised approach

Maria Luisa Di Pietro[1] · Drieda Zaçe[1] · Alessia Orfino[2] · Francesca Romana Di Raimo[3] · Andrea Poscia[4] · Elisabetta de Matteis[5] · Daniela Turchetti[6,7] · Lea Godino[6,7] · Benedetta Bertonazzi[6,7] · Marzena Franiuk[8] · Carla Bruzzone[8] · Liliana Varesco[8] · Emanuela Lucci-Cordisco[3,9] · Maurizio Genuardi[3,9]

Received: 17 October 2019 / Revised: 4 August 2020 / Accepted: 1 September 2020 / Published online: 14 September 2020

Abstract

Genomic testing expansion is accompanied by an increasing need for genetic counselling and intrafamilial communication. Genetic counselling can play an important role in facilitating intrafamilial communication and relationships. We conducted a cross-sectional, multicenter study including 252 Italian women, using a questionnaire divided in two sections, the first one to be filled after the pre-test counselling and the second after receiving BRCA test results. We assessed the factors influencing intrafamilial disclosure of genetic information for hereditary breast and ovarian cancer, family members with whom probands are more prone to share genetic information, and the perceived understanding of information received by counselees during genetic counselling. Women were accompanied to the counselling more often by their husband/partner. Among those with a positive BRCA test result, 49% intended to communicate it to their offspring and 27% to their husband/partner. Younger women, those living with their husband/partner, and those who described family communication as open/profound and spontaneous/sincere had a higher probability of being accompanied during genetic counselling and discuss about it with relatives. Spontaneous/sincere or open/profound family communication and joyful/happy familial relationships were associated with the decision to undergo genetic testing as a responsibility towards relatives. Women had a good understanding of counselling contents (mean score 9.27 in a scale 1–10). Genetic counselling providers should consider that genetic information disclosure does not depend only on the clarity of the information provided, but also on pre-existing intrafamilial communication and relationships, family structure and marital status, indicating the need for a personalised approach accounting for these factors.

Supplementary information The online version of this article (https://doi.org/10.1038/s41431-020-00723-7) contains supplementary material, which is available to authorised users.

✉ Drieda Zaçe
drieda.zace@unicatt.it

1 Sezione di Igiene, Dipartimento Universitario di Scienze della Vita e Sanità Pubblica, Università Cattolica del Sacro Cuore, Rome, Italy

2 Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome, Italy

3 UOC Genetica Medica, Dipartimento di Scienze di Laboratorio e Infettivologiche, Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome, Italy

4 UOC ISP Prevention and Surveillance of Infectious and Chronic Diseases—Department of Prevention—Local Health Authority (ASUR—AV2), Jesi, Italy

5 Oncology Unit, "Vito Fazzi" Hospital, Lecce, Italy

6 U.O. Genetica Medica, Policlinico S. Orsola-Malpighi, Bologna, Italy

7 Centro di Ricerca sui Tumori Ereditari, Dipartimento di Scienze Mediche e Chirurgiche (DIMEC) Università di Bologna, Bologna, Italy

8 Hereditary Cancer Unit, IRCCS Ospedale Policlinico San Martino, Genoa, Italy

9 Sezione di Medicina Genomica, Dipartimento di Scienze della Vita e Sanità Pubblica, Università Cattolica del Sacro Cuore, Rome, Italy

Introduction

The expansion of genomic technologies and molecular testing has been accompanied by an increasing need for genetic counselling and intrafamilial communication of genetic information. Hereditary breast and ovarian cancer (HBOC), which accounts for 1–7% and >10% of all female breast and ovarian carcinomas, respectively [1], is one the most common genetic conditions for which genetic testing is requested. The presence of a *BRCA1* disease associated high-risk variant entails a cumulative probability of ~72% for breast cancer and 44% for ovarian cancer. The corresponding estimates for *BRCA2* are about 69% and 17% [2].

Genetic information on HBOC has implications not only for those who undergo genetic testing, but also for their at-risk relatives, allowing them to make informed choices regarding treatment, prevention, changes in lifestyle and future childbearing [3, 4]. In several studies women largely anticipated that they would engage in positive health behaviour changes in response to HBOC risk disclosure [5, 6]. However, despite routine recommendations that individuals undergoing genetic testing should communicate the results to family members, about 20–40% of at-risk relatives remain unaware of genetic information of relevance to them [7]. Receiving and transmitting genetic information is a complex task, which can be influenced by several factors, including ethnicity, religion, age, gender, understanding of the contents of counselling sessions and intrafamilial communication and relationships. Detailed knowledge of these factors can be important to devise strategies aimed at improving the communication process. [8].

Many scholars and clinicians have pointed to the importance of family relationships in the dissemination of genetic information [9, 10]. Indeed, hereditary cancers are relational diseases. They can impact communication and interpersonal support within a family. The familial environment can affect an individual's decision to participate in genetic counselling and go through genetic testing. General communication and support patterns, leadership relationships and familial conflicts were found to be important in understanding the discussion of genetic counselling and testing within the family [9]. Mendes et al. [11] argue that genetic information is strongly linked to the family and that the communication of genetic risks is a process grounded within the broader milieu of family relationships and functioning. Good communication among family members allows them to develop a shared understanding and support each other, rendering them more likely to cope and adapt with the genetic condition and/or its risk [12]. The disclosure of relevant information to family members depends on the level of pre-existing relationships within the family. Claes et al. [13] found in their study that participants were most likely to communicate test results to their children (75%) and parents (58%) followed by their siblings (53%) and least likely to their aunts/uncles and cousins. Carriers of a *BRCA* high-risk variant had higher interest in genomic information and lower genetic worry and communicated genetic test results to a greater proportion of their closer (i.e., first degree) family members [14]. Furthermore, emotionally close relatives were more likely to be informed about genetic test results or the opportunity for genetic counselling [13, 14].

Another key component in the communication of genetic information is the genetic counselling process. It can affect the disclosure of genetic information to relatives in at least two different ways. First, the transmission of correct information is influenced by the level of understanding of the counselling contents by the counselee. The information given to counselees must be understandable and accessible, so that it can be correctly conveyed to their relatives, who can then make an informed decision [14]. Second, healthcare providers can help facilitate the communication of genetic counselling contents and of test results. They can do so by offering their own communication skills and services, giving tailored advice on the right time and way to disclose the information, offering to talk directly with a patient's children about their risk (with or without a parent present), consider the need for additional counselling before and after disclosure [15]. However, currently no standardised approach has been devised to this purpose, due to differences in the attitudes of probands and family members and the type and quality of interindividual relationships [15, 16]. Genetics health professionals and services are thus confronted not only with the need to provide information and care to the individuals tested, but also to help families understand and cope with genetic information [17].

Importantly, these topics have been investigated mostly in USA, Australia and Northern Europe, while there is a relative scarcity of data from other regions of the world, including Southern European countries, such as Italy. Therefore, we decided to conduct a cross-sectional study in an Italian sample, aiming to assess: (1) factors influencing the disclosure of HBOC genetic information with a focus on the pre-existing family relationships and communication, (2) whom do the probands more frequently share genetic information with and (3) the perceived understanding of the pre-test genetic counselling and the factors underlying it.

Methods

Research design and setting

This was an observational, cross-sectional, multicenter survey, which took place from September 2016 until July 2018 in four genetic centres: (1) Medical Genetics Unit, Cancer Genetics Clinic—Fondazione Policlinico Universitario A.

Gemelli IRCCS, Rome (coordinating centre); (2) Hereditary Tumors Clinic—National Institute of Cancer Research, Genoa; (3) Department of Medical and Surgical Sciences, Bologna; (4) Familial Cancer Center—"Vito Fazzi" Hospital, Lecce.

In all centres, counselling was performed by clinicians (medical geneticists in Rome, Genoa and Bologna centres; medical oncologists trained in cancer genetics in Lecce). In Genoa, a genetic nurse was also involved. All healthcare professionals providing genetic counselling were members of the "Società Italiana di Genetica Umana", which has published and follows dedicated guidelines on cancer genetic counselling (https://www.sigu.net/show/attivita/5/1/genetici).

The study was approved by the Ethical Committee of the Fondazione Policlinico Universitario A. Gemelli IRCCS.

Study population and procedures

Women aged >18 years who attended one of the collaborating genetic centres for a genetic consultation regarding HBOC were eligible to participate in the survey. In total, a convenience sample of 366 women was asked by the clinicians to be part of the study, during the enrolment period in all four centres. Those who agreed were approached by the researcher responsible for data collection, who explained the study and delivered the informed consent. After reading and signing the informed consent, participants were handed the questionnaire. This was divided in two sections: the first one was to be filled after pre-test counselling and then handed over to the researcher or delivered to a dedicated box available within each clinic. Only women who had an indication to undergo *BRCA1/BRCA2* analysis were given the second part of the questionnaire. They were asked to store it and fill it in immediately after receiving test results and leave it in the dedicated box at the clinic or send it via e-mail or ordinary post to the addresses provided by the research team. The second parts were received after a mean of 3 months following the compilation of the first parts. For each participant, the same code was written on both the first and second parts in order to link them. Overall, 252 first-part questionnaires were returned, 99 of which were also followed by the second sections (Fig. 1).

Instrument

The structuring of the questionnaire went through different phases. First, a literature review was conducted to find out tools that could be used for our aims. Although we found some questionnaires that separately assessed the influence of different factors (such as family structure, intrafamilial communication, genetic counselling, etc.) on genetic information disclosure, none collected relevant data in a comprehensive way. Moreover, we did not find any similar questionnaire administered to the Italian population. Therefore, with the

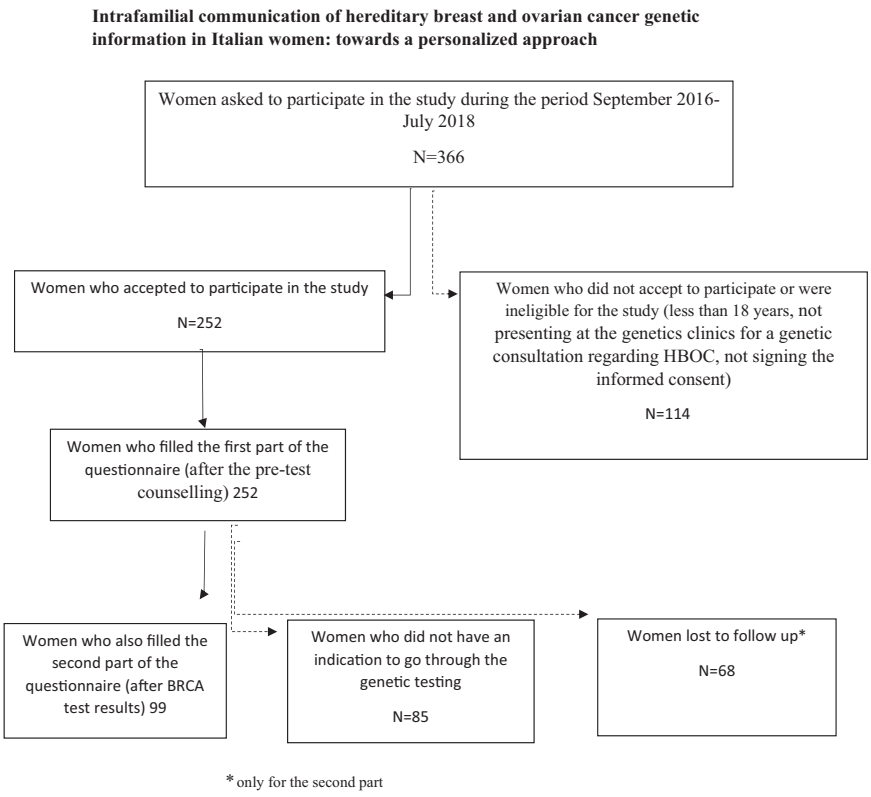

Fig. 1 Participants' flowchart. The figure shows the flow of the participants in each step of the study including enrolment, data collection process and follow-up.

aim to better adapt the questionnaire to the Italian context, we decided to create a new questionnaire based on the literature review. The first draft was presented to three different focus groups, including experts in the field of genetics, oncology, public health, epidemiology, ethics, as well as women with personal or family history of HBOC. The questionnaire's final version was then pilot-tested in a group of 20 women seen in the genetics clinic in Rome. This aimed at evaluating the feasibility, time and degree of understanding of the questionnaire. Participants suggested to add one option (ordinary but with some misunderstanding) among the answers provided to the question on family relations and one (spontaneous/sincere but sometimes difficult) to that on the communication within the family. They also suggested to group the definitions corresponding to psychological state in three groups: negative, neutral and positive feelings. Finally, they suggested to consider the possibility to send the second part of the questionnaire via e-mail or ordinary post, so these options were added at the end of the questionnaire.

Outcomes and variables

Outcomes

We assessed the probability of transmission of genetic information through four questions: (1) did somebody accompany you at the genetic counselling session? (2) Will you talk to somebody about your genetic counselling experience? (3) Reasons for deciding to undergo genetic testing (responsibility towards other family members). (4) Reasons for choosing to know the result of the genetic test (responsibility towards relatives and children). The first two relate to the first part of the questionnaire. In questions 3 and 4, included in the second part of the questionnaire, women were asked to choose out of a list of options the reasons why they decided to undergo genetic testing and, subsequently, for choosing to be informed about the result. Women who gave affirmative answers to the first two questions and those who gave as reasons the responsibility towards their relatives were considered to have a higher probability of transmitting genetic information to their relatives.

The members of the family with whom patients more frequently intend to share their experience or genetic information were assessed using three questions: (1) if yes, who accompanied you at the genetic counselling? (2) Whom did/will you talk to about the genetic counselling? (3) If the test result was positive, whom do you intend to communicate the result to?

In order to assess the degree of perceived understanding of genetic counselling, we used three questions (Cronbach's alpha = 0.77): (1) in a scale from 1 to 10 how much of the information given did you understand? (2) In a scale from 1 to 10 how much do you think the doctor has clearly explained the medical-scientific information? (3) In a scale from 1 to 10 how much did you understand the concept of risk related to the outcome (positive/negative) of the test? The answers to these questions were given in a ten-score Likert scale, where 1 stands for "not at all" and 10 for "a lot".

Variables

We explored demographic variables (age, nationality, religion and profession), person-specific predictors (marital status, blood relatives, who they live with, reason for genetic counselling) and intrafamilial communication. The latter was assessed through three dedicated questions: (1) how would you define your family relationships (happy/joyful; ordinary, with some misunderstanding; aggressive; oppressive; superficial; absent), (2) how would you define the communication within your family (open/profound; spontaneous/sincere, sometimes difficult; problematic; occlusive; superficial; absent), (3) how would you define the importance of family relationships for you (essential/indispensable; advantageous/convenient; ordinary/trivial). Other variables were the reasons for undergoing testing, waiting time for the result and feelings during that period, test result, the moment they decided to share the result with relatives, perceived role of the genetic healthcare provider in intrafamilial communication.

Statistical analysis

Descriptive statistics was performed using frequencies, percentages, frequency tables for categorical variables and mean and standard deviation for quantitative variables. Chi square test and Student's t test were used as test of association for categorical and continuous data, respectively. To explore the factors that affected genetic information disclosure, logistic regression analysis was performed. A mean score for perceived understanding of genetic counselling was calculated based on the answers given to the three questions asked to assess it. To explore the factors that affected the mean score of counselling perceived understanding, linear regression was performed. Associations with p value < 0.05 were considered statistically significant. Statistical analysis was performed using STATA 15 software.

Results

Pre-test results

Participant characteristics and family information

Overall, 252 first parts were included in the analysis. The characteristics of the participants are shown in Table 1.

Table 1 Characteristics of the participants and family information.

	n	%
Age		
<40	32	12.7
40–60	160	63.5
>60	60	23.8
Nationality		
Italian	234	97
Other	7	3
Marital status		
Widow	9	3.7
Married	147	61
Divorced	29	12.1
Single	56	23.2
Religion		
Religious	220	94.8
Atheistic	12	5.2
Profession		
Employee	115	49.1
Unemployed	19	8.1
Retired	32	13.7
Student	3	1.3
Freelancer	26	11.1
Housewife	39	16.7
Blood relatives[a]		
Mother/father	126	54.3
Sister/brother	172	74.1
Cousins	122	52.6
Nephew/niece	113	48.7
Aunt/uncle	102	43.9
Other	68	29.3
Living with (blood and non-blood relatives)		
Husband/partner	185	76.1
Daughter/son	146	60.0
Sister/brother	13	5.4
Mother/father	22	9.1
No one	15	6.2
Parent civil status		
Married	153	88.4
Living together	5	2.9
Divorced	12	6.9
Divorced, new marriage	3	1.8
Family relations		
Happy/joyful	135	53.6
Ordinary, with some misunderstanding	99	39.3
Aggressive	5	2
Oppressive	13	5.2
Superficial	6	2.4
Absent	6	2.4
Family communication		
Open/profound	140	55.6
Spontaneous/sincere, sometimes difficult	92	36.5
Problematic/difficult	18	7.1
Occlusive	1	0.5
Superficial	4	1.6
Absent	4	1.6
Relation values		
Essential/indispensable	236	95.6
Advantageous/convenient	6	2.4
Ordinary/trivial	5	2

[a]All blood relatives, not only those whom they live with.

For the variables "blood relatives" and "living with" the sum of the percentages exceeds 100% because more than one answer could be given.

Most participants lived with their husbands/partners (76.1%), followed by 60.0% who lived with their children and the rest with their sister/brother, mother/father or alone (5.4%, 9.1% and 6.2%, respectively). Asked about their blood relatives (all of them, not only those whom they live together with), 54.1% answered mother/father, 74.1% sister/brother, 52.6% cousins, 48.7% nephews/nieces, 43.9% aunt/uncle and 29.3% others. When asked about their parents' civil status, most participants' (60.7%) answered that their parents were married.

Most women (53.6%) described their family relationships as happy/joyful, while for 39.3% they were ordinary but with some misunderstandings. Family communication was open/profound in 55.6% of the cases, spontaneous/sincere but sometimes difficult in 36.5% and problematic, occlusive, superficial and absent in 7%, 0.5% and 1.6% and 1.6%, respectively. Family relationships were essential for 95.6% of women answering the questionnaire, and for the rest they were advantageous or ordinary.

Genetic counselling

For nearly 40% of the counselees the reason to have genetic counselling was a personal and family history of breast/ovarian cancer. For 30%, it was one of their blood relatives who was diagnosed with breast/ovarian cancer. For 25% there was a personal history of cancer, with no relatives affected, and the remaining 5% had other unspecified reasons.

More than half of the women (59%) were accompanied at the counselling session, more often by their husbands/partners (36% of all the women who declared to live with their husbands/partners) and less by their mother/father (13% of women who reported their mother/father as being

Table 2 Members of the family with whom counselees more frequently shared their genetic counselling experience or genetic information.

	If yes, who accompanied you at the genetic counselling		Whom did you talk to about the genetic counselling		If the test was positive, whom did you communicate the result to	
Family members (blood and non-blood relatives)	*n*	%	*n*	%	*n*	%[a]
Non-blood relatives						
Husband/partner($n = 185$)	68	36%	54	29%	8	27%
Blood relatives						
Mother/father ($n = 126$)	17	13%	28	22%	9	31%
Daughter/son ($n = 146$)	34	23%	33	22%	13	49%
Sister/brother ($n = 172$)	30	17%	42	24%	9	31%

[a]In proportion to the number of positive test results (29).

still part of their blood relatives) (Table 2). In most cases the reasons for being accompanied were to not be alone (26%), for support (21.6%) or because the accompanying person was also having BRCA testing (15.1%). In 68% of cases women had already spoken to somebody about having the genetic counselling. Almost 30% had talked to their husband/partner, and a lower fraction had talked to their mother/father or daughter/son. Among those who were alone at the counselling session, 92.4% said that they would talk to somebody about it, 72% of whom would do it immediately and in most cases they would talk to their relatives (31%) and husbands/partners (24%).

The three questions assessing the degree of perceived understanding of the genetic counselling contents in a scale from 1 to 10 had a mean score of 9.2 (1.1), 9.4 (1.0) and 9.2 (1.2).

Among those who did not fully understand the information given during genetic counselling (score < 8), 30% said that this happened because of insufficient time dedicated to them during the meeting, 12% because the healthcare professional was not clear enough and 12% because they were too scared to listen. The rest chose the option "other", giving as reasons the difficulty to understand specific definitions, not paying attention and feeling insecure to ask when they did not understand something. When asked if they were satisfied with the time given to ask questions during counselling, 68.8% of the participants were very satisfied, 24.5% were moderately satisfied and 6.7% were poorly satisfied. Asked how much time the doctor dedicated to the explanation of scientific aspects and how much to listening to the counselee during the consultation, the majority (43.2%) answered "50% of time to explanation and 50% to listening".

Post-test results

Ninety-nine second parts were included in the analysis. Seventy-three per cent of the respondents going through genetic testing affirmed that they already knew what a genetic test was. Ninety-one per cent stated that they decided independently to go through the genetic test, with only two women claiming that they got pressured by the doctor. For half of the women the reason for undergoing genetic testing was the need to know the genetic cause of the disease, and for half it was the responsibility towards their relatives. All participants had chosen to know the genetic test results, justifying this decision with the opportunity to undergo appropriate prevention (67%), responsibility towards relatives and children (60%) and because health is an issue that affects everyone in the family and genetic information must be shared (18%). Most women had negative feelings while waiting for the result of the genetic test (64%), the rest had neutral (17%) or positive (37%) feelings.

Genetic test information

The test was positive for 29.3% of women who filled the second parts. Among these, 93% reported they intended to communicate the result to somebody, in 60% of cases as soon as possible. In most cases (77.7%), they would disclose the information to their relatives (without specifying which ones), in 48% to their children and 33% to their siblings. They reported they intended to communicate less the information to their husband/partner (29.6%) and friends (29.6%). None of the women said there was somebody they would prefer not communicating the information to. The reason behind intending to disclose the test result was in 88.9% of cases the sense of responsibility towards relatives and children, in 44.4% to allow relatives and/or children the freedom to decide on their own health and in 37% because health is an issue that affects everyone in the family. According to 37.4% of women, the result of the genetic test should concern blood relatives, as well as other persons who live together but are not genetically related, while 26.3% think that the result of the genetic test

Table 3 Factors affecting the probability of genetic information transmission.

	Pre-test						Post-test			
	Accompanied during genetic counselling No/Yes		*p* value	Will you talk to somebody regarding the genetic counselling No/Yes		*p* value	Reason for going through the test/ responsibility towards relatives	*p* value	Why did you choose to know the test result/ responsibility towards relatives	*p* value
	n (%)	*n* (%)		*n* (%)	*n* (%)		*n* (%)		*n* (%)	
Familial relations										
Joyful, happy	53 (39.6%)	81 (60.4%)	0.74	9 (13:4%)	58 (86.6%)	**0.04**	23 (56.1%)	**0.03**	25 (59.5%)	**0.04**
Ordinary, quiet	41 (42.3%)	56 (57.6%)	0.57	2 (3.5%)	55 (96.5%)	0.23	23 (67.6%)	0.51	28 (80.0%)	0.49
Aggressive	2 (40.0%)	3 (60.0%)	0.84	0 (0.0%)	3 (100%)	0.73	1 (100%)	–	1 (100%)	–
Oppressive	6 (46.2%)	7 (53.8%)	0.49	1 (14.3%)	6 (85.7%)	0.35	1 (50.0%)	0.67	4 (100%)	–
Superficial	4 (66.7%)	2 (33.3%)	0.39	0 (0.0%)	4 (100%)	–[a]	2 (100%)	–	2 (100%)	–
Non existing	6 (100%)	0 (0.0%)	–	0 (0.0%)	5 (100%)	–	1 (50.0%)	–	1 (50.0%)	–
Communication										
Open, profound	47 (33.8%)	92 (66.2%)	**0.02**	8 (12.1%)	58 (87.9%)	0.06	21 (52.5%)	**0.03**	28 (66.7%)	0.84
Spontaneous, sincere	42 (46.1%)	49 (53.9%)	**0.03**	3 (5.4%)	52 (94.5%)	0.34	28 (77.8%)	**0.01**	29 (78.4%)	0.74
Problematic	13 (72.2%)	5 (27.8%)	0.14	0 (0.0%)	11 (100%)	–	1 (25.0%)	0.18	0 (0.0%)	–
Occlusive	0 (0.0%)	1 (100%)	0.13	0 (0.0%)	0 (0.0%)	–	0 (0.0%)	–	0 (0.0%)	–
Superficial	2 (50.0%)	2 (50.0%)	0.59	0 (0.0%)	3 (100%)	–	3 (100%)	–	3 (100%)	–
Non existing	2 (50.0%)	2 (50.0%)	0.66	0 (0.0%)	3 (100%)	–	0 (0.0%)	–	0 (0.0%)	–
Relations value										
Essential, indispensable	95 (40.6%)	139 (59.4%)	0.9	10 (8.1%)	114 (91.9%)	0.4	47 (61.8%)	0.4	56 (70.9%)	0.8
Advantageous, convenient	3 (50.0%)	3 (50.0%)	0.29	0 (0.0%)	3 (100%)	–	2 (66.6%)	–	1 (50.0%)	–
Ordinary, banal	3 (60.0%)	2 (40.0%)	0.68	0 (0.0%)	2 (100%)	–	1 (100%)	–	1 (100%)	–

Controlled for age, marital status, profession, who they live with, consanguineous family members, reason for undergoing genetic counselling.
Bold values indicate statistical significance $p < 0.05$.

[a]Because of the low number of observations, a logistic regression model was not applicable.

should be of interest and communicated only to blood relatives.

Factors associated to the probability of genetic information disclosure

Table 3 shows the association of intrafamilial communication with the transmission of genetic information within the family. Women who described their family relationship as joyful/happy had significantly higher probability of talking to somebody regarding the genetic counselling, as well as giving as reason for going through the test the responsibility towards relatives and choosing to know the result of the test as a responsibility towards their relatives. Also, women who described the communication inside their family as open/profound were more often accompanied by someone at the genetic counselling and had higher probability of giving as reason for going through the test the responsibility towards relatives. Moreover, when intrafamilial communication was described as problematic, no counselee declared that the reason for going through genetic testing was also a sense of responsibility towards other relatives. Among women who described their familial relationship as joyful/happy, 86.6% answered that they would talk to someone about the genetic counselling ($p = 0.01$), and almost 60% of them declared that they chose to know the result of the genetic test also because they felt responsibility towards their relatives ($p = 0.01$).

Seventy-six per cent of those who were married answered that the reason for undergoing genetic testing was also the sense of responsibility towards other relatives, compared to 50% of those who were divorced and 36.8% of those who were single ($p = 0.02$). Women aged <40 years and those living with their husbands/partners tended to be more frequently accompanied at the genetics clinic for counselling ($p = 0.004$ and $p = 0.03$, respectively).

Table 4 Factors affecting the perceived understanding of genetic counselling contents.

	Mean score for counselling perceived understanding	*p* value
Age		0.26
<40	9	
40–60	9.3	
>60	9.2	
Intrafamilial communication problematic[a]		**0.03**
Yes	8.8	
No	9.3	
Intrafamilial communication spontaneous, sincere[a]		**0.03**
Yes	9.5	
No	9.1	
Accompanied in the counselling by[b]		**0.02**
Husband/partner	9	
Genetically related relatives	9.4	
Space given to ask questions[c]		**<0.001**
Low	8.2	
Medium	8.6	
High	9.6	
Feelings while waiting for the test results		
Negative	9.2	0.67
Neutral	9	0.85
Positive	9.1	0.67

[a]Controlled for age, marital status, religion, profession, reason for counselling, accompanied at the counselling and space to ask questions.

[b]Controlled for age, religion, profession, family relations and family communication.

[c]Controlled for age, marital status, religion, profession, who they live with, consanguineous family members, family relations, family communication, reasons for counselling and accompanied during the counselling.

Bold values indicate statistical significance $p < 0.05$.

Factors affecting perceived understanding

Women who described their intrafamilial communication as spontaneous/sincere had significantly better perceived understanding of the information given during counselling (9.5 vs. 9.1; $p = 0.03$) (Table 4). Likewise, women who were accompanied to the counselling by a blood relative rather than by their husband/partner had a significantly higher mean score of perceived understanding. On the other hand, women who described their intrafamilial communication as problematic had lower perceived understanding (8.8 vs. 9.3; $p = 0.03$). The mean score of perceived understanding was also significantly associated with the time available for questions (low vs. medium vs. high $p < 0.001$). Half of the women think the consultant should intervene in facilitating intra-family communication, mainly by listening more to the patient and facilitating a dialogue between them (40.5%), and by providing practical advice on how to communicate the received information (54.5%).

Discussion

In the present study we aimed at determining (1) whether intrafamilial communication and disclosure of genetic information are associated and (2) the type of relationship between counselees and the persons with whom they discuss genetic information. Furthermore, we wanted to assess the perceived understanding of the information received by the patients during the pre-test counselling.

The probability of transmission of genetic information was assessed using four outcomes: presence of accompanying persons at the counselling session, communication about genetic counselling with other persons, reason for undergoing genetic testing, i.e., namely, if this was prompted by a sense of responsibility towards relatives, and reasons for the willingness to accept the return of genetic test results, again especially if this was related to the same kind of familial responsibility. We figured that a positive answer to each of these questions would imply a higher chance of communication and interaction and, consequently, a higher probability of transmitting or disclosing genetic information.

The factors associated with the probability of disclosure of genetic information were women's age, marital status, type of relatives they live with, and perceived intrafamilial relations and communication. Younger women (<40 years old) are more frequently accompanied by someone at counselling, while women who belong to the age category 40–60 years old are the least probable to be accompanied. As reported, also, in literature, married women or widows state that the sense of responsibility towards their relatives is a main reason to undergo genetic testing, compared to divorced and single women [18]. This could translate into a lower probability of communication. Indeed, it has been observed that divorce and separation, along with family rifts and tensions, create barriers to communication of HBOC genetic information [8].

The perceived quality of family relationships and communication was significantly associated with the probability of disclosure of the genetic information. None of the women who described their intrafamilial relationships as problematic declared that a sense of responsibility towards relatives was the reason to undergo testing. On the other hand, women who perceived their family relationships as joyful/happy or their family communication as open/profound and

spontaneous had higher probability to disclose genetic information. Our results are in agreement with previous findings in a French patients population, which showed that family relationships and more open family communication patterns may facilitate transmission of BRCA test results, extend HBOC information disclosure and minimise cancer-related distress [19].

On the other hand, not only the process of disclosure can be affected by the pre-existing relationships, but the reverse can also occur. Women may be worried about the potential impact that the disclosure of genetic information could have on the relationships with their relatives [20]. With respect to relatives' reactions, individuals participating in a psychoeducational group expressed concern that there could be blame, backlash or a negative impact on relationships [21]. Therefore, it is important that these issues be addressed during genetic counselling, which requires a discussion on the patterns of pre-existing family communication and relationships.

Our results indicate that in the studied sample, women undergoing the pre-test genetic counselling prefer to discuss about having the counselling and share the information given by the healthcare professionals during the counselling with their partners. This could be viewed as a request and expectation of support from them. Communication as a support-seeking behaviour has been observed in other studies [22, 23]. Moreover, they state that they do not prefer to talk to anyone else without having definitive information to disclose, i.e., having the test results. Following this logic, women tended to speak more about genetic test results with their blood relatives. Forty-nine per cent of women who declared to have at least one daughter/son, answered that they had communicated to them the result, followed by 31% of women who had communicated the result to their siblings, and 31% to their mother/father. These results are in line with other studies from the literature. A lower rate of communication of positive results to parents has previously been observed, which can be explained by the wish to avoid them to feel guilty about having passed on the genetic alteration [24]. Also, if a relative is seen as too old and no longer in a high-risk life stage, there may be no perceived benefit to inform them [25].

Considering the information flux from parents to children, it has been observed that about 40% of parents do not talk to their children about their family's condition, comparable to ~50% in our study. However, most of these parents stated that they would like to be able to discuss it with them [26]. This might also occur because some individuals inform their children only when they have reached an age where preventative measures can be undertaken or when they are at reproductive risk of passing the high-risk genetic variant on to their offspring [26]. Finally, even if women prefer to talk more to their husbands/partners about the pre-test genetic counselling, they tend to communicate less the test result to them. This could be explained by the respondents' perception that the result does not concern their husband/partner directly, since they are not genetically related. The main reason why women would consider disclosing the test results to their husbands/partners would be future childbearing. However, in our sample 62.7% of women were older than 45 years, suggesting that, in most cases, the parental project was concluded [8].

The genetic counselling process, namely the health professionals conducting it, can play an important role in the communication process. Their improved understanding of intrafamilial communication dynamics would theoretically enable them to better assist women and facilitate genetic information disclosure [27]. In our study, women were generally satisfied with the counselling, stating in most cases that they understood the information provided and the concept of risk, and that the healthcare professional was clear enough in the explanations given. Nonetheless, there is evidence that even when women are well informed, the transfer of information to relatives can be highly deficient, considering that family communication about genetics is a complex relational process, which poses practical and moral dilemmas for individuals and families [28, 29].

For those who did not understand, the primary reason was the insufficient time dedicated during counselling, followed by unclearness of the counsellor and by the fact that they were too afraid to listen. Importantly, the mean score of perceived understanding was significantly associated with the time given to ask questions during counselling. Hence, it is important that the healthcare professionals conduct the genetic counselling in such a way that allows the counselees to ask any possible questions relevant to them.

We found that the level of perceived understanding of the counselling is associated not only with the factors inherent with the counselling process itself, but also with pre-existing conditions, namely, intrafamilial communication and relationship. Women who perceived their intrafamilial communication as spontaneous and sincere had significantly higher perceived understanding of the information provided. This could also be related to the above discussed finding that women who perceived their intrafamilial communication as spontaneous and sincere also feel more responsible towards their relatives, which implies that they understand that the genetic information they will receive is relevant for their relatives, leading them to be more attentive, ask more questions and require more comprehensive explanations. Likewise, the finding that women who were accompanied during counselling by blood relatives had significantly higher mean score of perceived understanding could have at least partly the same explanation.

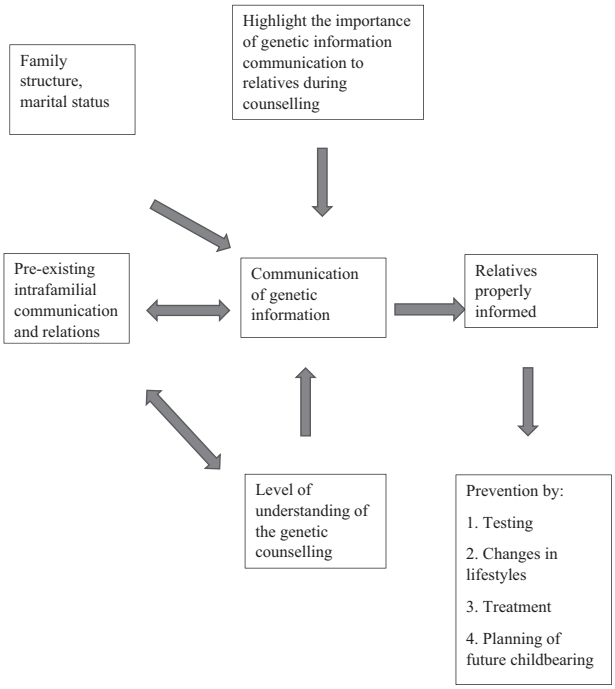

Fig. 2 Correlations between factors influencing the communication of genetic information. It is shown how communication of genetic information, pre-existing intrafamilial communication and relations, and level of understanding of the genetic counselling interconnect between them. The importance of genetic information disclosure stands in the fact that it gives the properly informed relatives the opportunity to take appropriate preventive measure.

All the above-mentioned aspects, summarised in Fig. 2, should be considered by the healthcare professional conducting genetic counselling. Our results indicate that it is important to have information on the pre-existing intrafamilial communication and relationships to better understand the background interacting on. In this regard, we saw that family structure and marital status are important factors. Pre-existing intrafamilial communication and relationships can directly affect the way a woman approaches genetic counselling and the probability of genetic information disclosure (i.e., women were more responsible towards their relatives when good communication and relationships existed). In turn, literature suggests that the latter can affect family relationships in two opposite ways, by strengthening them or by leading to dysfunction [20]. Furthermore, our results indicate that intrafamilial communication can play a role in the understanding of the genetic counselling and, consequently, it can affect the level of disclosure of genetic information and genetic test results. All the above-mentioned factors would help the relatives be more informed. In this way as literature suggests, they would be more prone to implement prevention strategies like testing, changes in lifestyle, planning of future childbearing [5, 6].

In conclusion, healthcare professionals should keep in mind that the disclosure of genetic information to the relatives does not depend only on the clarity of the information they give during counselling, but also on the level and quality of pre-existing intrafamilial communication and relationships, family structure, marital status and time given to ask questions. A proper genetic counselling impacts not only the counselee, but her whole family and can help in facilitating intrafamilial communication, while inappropriate counselling could be a source of worry, misunderstanding and dysfunction in intrafamilial communication and relationship. On the other hand, the involvement of relatives in genetic counselling is a complex matter and often it may need more than one or two counselling sessions. Increased time of clinical consultations could probably be helpful, but may not be sufficient to devise the best strategy for the single proband, considering also the different types of familial relationships, which may require different solutions tailored individually for family members [15, 16]. The use of supportive actions and resources, including lay summaries and pamphlets, learning on previous experiences of other counselees, and education on how to approach relatives, including providing information on potential reactions and how to cope with them may facilitate communication [16].

Genetic counselling is inspired by the principle of non-directiveness, which, however, should not be viewed as an obligation to act according to patient's views for the health professional involved [30]. One of the essential goals of cancer genetic counselling is to reach out to all biological relatives of a disease associated variant carrier in order to identify all high-risk individuals and maximise the efficacy of preventative measures. Family communication is a key issue in this regard, and we have shown that relational barriers may hamper this task also in a Southern European country, like Italy. Overall, our findings highlight the importance of a personalised counselling approach, focused not only on individual issues and biomedical information, but also largely family oriented and aimed at providing psychological support [31, 32].

These findings are very important, considering that this is the first study conducted in Italy regarding these issues. In general, we found that the attitudes towards familial communication of genetic counselling and testing for HBOC do not significantly differ from other countries where some of the issues examined in our study were previously considered. Importantly, genetic counselling for cancer predisposition in Italy is usually provided by medical doctors or graduates in biosciences who have completed a specialty in medical genetics. In the field of cancer genetics, in some centres medical oncologists trained in hereditary cancer are also involved, while there is not a dedicated figure such as a genetic counsellor. In several countries, in Europe and worldwide, genetic counsellors have an important role in counselee education and information on the specific

condition present in the family and encourage discussions about personal and reproductive choices [33]. The availability of professional figures primarily involved in keeping contacts with counselees and facing their emotional issues could be useful to implement improved strategies or interventional trial, i.e., telephone follow-up with counselees, direct contact of family members following proband's consent, counselling sessions involving multiple family members simultaneously or multi-family intervention groups [34–36]. Other peculiarities of the Italian situation pertain to familial relationships, including the tendency of nuclear family members, namely children, to spend more time in the origin household compared to other European countries, which could affect family members' relations and communication.

Limits and future perspectives

The results of this study should be considered in the light of some limitations. First, we had a low number of second parts of the questionnaire returned, mainly due to logistic reasons. Some women did not deliver the second parts and it was difficult to trace them back. We had very small sample size for certain findings, making it impossible to perform statistical analyses. Second, genetic information and its disclosure are sensitive topics for many patients, so reporting bias could have distorted the information declared by women. Furthermore, the results of this study concern the Italian context and may not be applicable to other populations. Lastly, the findings that certain factors are associated to perceived genetic counselling understanding and genetic information disclosure could be due to confounding that we did not control for. In addition, in order to verify the impact of counselling on the family and the expectations of relatives, it would be important to consider counselees' family members or first-degree relatives for future studies.

Compliance with ethical standards

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

Publisher's note Springer Nature remains neutral with regard to jurisdictional claims in published maps and institutional affiliations.

References

1. Balmana J, Diez O, Rubio IT, Cardoso F, ESMO Guidelines Working Group. BRCA in breast cancer: ESMO clinical practice guidelines. Ann Oncol. 2011;22:vi31–4.
2. Kuchenbaeker KB, Hopper JL, Barnes DR, Phillips KA, Mooij TM, Roos-Blom MJ, et al. Risks of breast, ovarian, and contralateral breast cancer for *BRCA1* and *BRCA2* mutation carriers. JAMA. 2017;317:2402–16.
3. Domchek SM, Friebel TM, Singer CF, Evans DG, Lynch HT, Isaacs C, et al. Association of risk-reducing surgery in BRCA1 or BRCA2 mutation carriers with cancer risk and mortality. JAMA. 2010;304:967–75.
4. Katapodi MC, Northouse RN, Pierce P, Williams RA. Differences between women who pursued genetic testing for hereditary breast and ovarian cancer and their at-risk relatives who did not. Oncol Nurs Forum. 2011;38:572–81.
5. Meisel SF, Fraser LSM, Side L, Gessler S, Hann KEJ, Wardle J. Anticipated health behavior changes and perceived control in response to disclosure of genetic risk of breast and ovarian cancer: a quantitative survey study among women in the UK. BMJ Open. 2017;7:e017675.
6. Mahoney MC. Breast cancer risk reduction and counselling: lifestyle, chemoprevention, and surgery. J Natl Compr Canc Netw. 2007;5:702–10.
7. Hodgson JM, Metcalfe SA, Aitken M, Donath SM, Gaff CL, Winship IM, et al. Improving family communication after a new genetic diagnosis: a randomised controlled trial of a genetic counselling intervention. BMC Med Genet. 2014;15:33.
8. Nycum G, Avard D, Knoppers BM. Factors influencing intrafamilial communication of hereditary breast and ovarian cancer genetic information. Eur J Hum Genet. 2009;17:87–880.
9. Koehly LM, Peterson SK, Watts BG, Kempf KKG, Vernon SW, Gritz ER. A social network analysis of communication about hereditary nonpolyposis colorectal cancer genetic testing and family functioning. Cancer Epidemiol Biomarkers Prev. 2003;12:304–13.
10. Rolland JS. Genetics, family systems, and multicultural influences. Fam Syst Health. 2006;24:425–41.
11. Mendes A, Metcalfe A, Paneque M, Sousa L, Clarke AJ, Sequeiros J. Communication of information about genetic risks: putting families at the center. Fam Process. 2017;57:836–46.
12. Hoskins LM, Roy K, Peters JA, Loud JT, Greene MH. Disclosure of positive BRCA1/2-mutation status in young couples: the journey from uncertainty to bonding through partner support. Fam Syst Health. 2008;26:296–316.
13. Claes E, Evers-Kiebooms G, Boogaerts A, Decruyenaere M, Denayer L, Legius E. Communication with close and distant relatives in the context of genetic testing for hereditary breast and ovarian cancer in cancer patients. Am J Med Genet. 2003;116A:11–9.
14. Elrick A, Ashida S, Ivanovich J, Lyons S, Biesecker BB, Goodman MS, et al. Psychosocial and clinical factors associated with family communication of cancer genetic test results among women diagnosed with breast cancer at a young age. J Genet Couns. 2017;26:173–81.
15. Keenan KF, McKee L, Miedzybrodzka L. Genetics professionals' experiences of facilitating parent/child communication through the genetic clinic. J Genet Couns. 2020;29:44–55.
16. Pollard S, Kalloger S, Weymann D, Sun S, Nuk J, Schrader KA, et al. Genetic testing for hereditary cancer syndromes: patient recommendations for improved risk communication. Health Expect. 2020;00:1–9.
17. McDaniel SH, Rolland JS, Feetham SL, Miller SM. "It runs in the family": family systems concepts and genetically linked disorders. In: Miller SM, McDaniel SH, Rolland JS, Feetham SL, editors. Individuals, families, and the new era of genetics: biopsychosocial perspectives. New York: W.W. Norton & Company; 2006. p. 118–318.
18. Kenen R, Arden-Jones A, Eeles R. Healthy women from suspected hereditary breast and ovarian cancer families: the significant others in their lives. Eur J Cancer Care. 2004;13:169–79.
19. Julian-Reynier C, Eisinger F, Chabal F, Lasset C, Nogues C, Stoppa-Lyonnet D, et al. Disclosure to the family of breast/ovarian cancer genetic test results: patient's willingness and associated factors. Am J Med Genet. 2000;94:13–8.

20. Gilbar R. Communicating genetic information in the family: the familial relationship as the forgotten factor. J Med Ethics. 2007;33:390–93.
21. Speice J, McDaniel SH, Rowley PT, Loader S. Family issues in a psychoeducation group for women with a BRCA mutation. Clin Genet. 2002;62:121–7.
22. Hughes C, Lerman C, Schwartz M, Peshkin BN, Wenzsel L, Narod S, et al. All in the family: evaluation of the process and content of sisters' communication about BRCA1 and BRCA2 genetic test results. Am J Med Genet. 2002;107:143–50.
23. Patenaude AF, Dorval M, DiGianni LM, Schneider KA, Chittenden A, Garber JE, et al. Sharing BRCA1/2 test results with first degree relatives: factors predicting whom women tell. J Clin Oncol. 2005;24:700–6.
24. D'Agincourt-Canning L. Experiences of genetic risk: disclosure and the gendering of responsibility. Bioethics. 2001;15:231.
25. Metcalfe A, Coad J, Plumridge GM, Gill P, Farndon P. Family communication between children and their parents about inherited genetic conditions: a meta-synthesis of the research. Eur J Hum Genet. 2008;16:1193–200.
26. Van Oostrom I, Meijers-Heijboer H, Duivenvoorden HJ, van Gool AR, Seynaeve C, Meer CA, et al. Social and behavioral research in clinical genetics. Clin Genet. 2007;71:35–42.
27. Black L, McClellan KA, Avard D, Knoppers BM. Intrafamilial disclosure of risk for hereditary breast and ovarian cancer: points to consider. J Community Genet. 2013;4:203–14.
28. Foster C, Eeles R, Arden-Jones A, Moynihan C, Watson M. Juggling roles and expectations: dilemmas faced by women talking to relatives about cancer and genetic testing. Psychol Health. 2004;19:439–55.
29. Chalmers K, Marles S, Tataryn D, Scott-Findlay S, Serfas K. Reports of information and support needs of daughters and sisters of women with breast cancer. Eur J Cancer Care. 2003;12:81–90.
30. Elwyin G, Gray J, Clarke AJ. Shared decision making and non-directiveness in genetic counselling. J Med Genet. 2000;37:135–8.
31. Mendes Á, Paneque M, Sousa L, Clarke A, Sequeiros J. How communication of genetic information within the family is addressed in genetic counselling: a systematic review of research evidence. Eur J Hum Genet. 2016;24:315–25.
32. Jacobs C, Patch C, Michie S. Communication about genetic testing with breast and ovarian cancer patients: a scoping review. Eur J Hum Genet. 2019;27:511–24.
33. Gaff C, Hodgson J. A genetic counselling intervention to facilitate communication about inherited conditions. Eur J Hum Genet. 2014;23:914–23.
34. Abacan MA, Alsubaie L, Barlow-Stewart K, Caanen B, Cordier C, Courtney E, et al. The global state of the genetic counsling profession. Eur J Hum Genet. 2019;2:183–97.
35. SPRinG Collaboration. Developing an intervention to facilitate family communication about inherited genetic conditions, and training genetic counsellors in its delivery. Eur J Hum Genet. 2016;24:794–802.
36. Mendes A, Chiquelho R, Santos T, Sousa L. Family matters: examining a multifamily group intervention for women with BRCA mutations in the scope of genetic counselling. J Community Genet. 2010;1:161–8.