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Awareness of genomic testing among patients with breast cancer in Europe

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ABSTRACT

Purpose: Genomic testing, involving expression profiling of tumour tissue, is a powerful tool for determining appropriate treatments for certain cancer patients. This study aimed to evaluate awareness of genomic testing in breast cancer patients in five European countries.

Methods: The survey was initiated by Cancer Patients Europe and developed with patient associations, oncologists, and a psycho-oncologist. Participants were recruited via email and social media and completed a 42-question internet survey.

Results: Of 1383 participants in eligible countries completing the survey, 566 women with current or previous HR+/HER2- breast cancer, potentially eligible for genomic testing, were analysed. 245 (43.3 %) were aged 50–59 years and 381 (67.3 %) had received higher education. 238 participants (42.1 %) had heard about genomic testing; 122 (21.6 %) were informed of their eligibility for testing, and 104 (18.4 %) were given reasons for the test. The majority (N = 479; 84.6 %) felt they lacked sufficient information to decide, and only 139 (24.6 %) opted for testing. Overall, 246 (43.5 %) wanted more information on additional testing and 234 (41.3 %) wanted more information on treatment options. The main information sources were medical professionals (N = 363; 64.1 %) and the internet (N = 351; 62.0 %). However, 398 participants (70.3 %) indicated that their healthcare professionals did not advise them on where to find more information.

Conclusions: This study highlights insufficient awareness of, and access to, genomic testing in breast cancer. Healthcare professionals need to improve communication with patients regarding genomic testing and involve them in shared decision-making. Likewise, patient associations have a role in providing clear information to patients.

1. Introduction

In 2020, there were an estimated 2.3 million new cases of breast cancer worldwide and this disease was responsible for 0.7 million deaths. Over the last decade, breast cancer in women has surpassed lung cancer as the most diagnosed cancer worldwide [1]. In around 10 % of cases, a family history is present [2] and high-penetrance genes, notably BRCA1 or BRCA2, have been identified which are associated with a high risk of developing breast cancer [3].

Breast cancer is a heterogeneous disease. Different subtypes have

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different molecular signatures, which results in varied prognosis and response to treatment [2]. Five subtypes account for most of the diversity in breast cancer; these surrogate intrinsic subtypes are distinguished by tumour grade and by the level of expression of human epidermal growth factor receptor 2 (HER2), the two hormone receptors, oestrogen (ER) and progesterone (PR) and the proliferation marker Ki67 [4], [5,6]. The most common of these subtypes is ER+/HER2 negative breast cancer, sometimes called luminal-like. This subgroup can be further divided into luminal A-like, accounting for at least 60 % of breast cancers [2], and characterised by low proliferation and relatively better prognosis, and luminal B-like, characterised by high proliferation and worse prognosis [2,7,8]. While the standard of care for luminal-A like tumours is surgical management with adjuvant endocrine therapy with or without radiotherapy, many luminal B-like tumours require the addition of chemotherapy [7] depending on stage, age and co-morbidities.

The development of high-throughput microarray assays has made possible genomic profiling of biopsy samples from breast cancer [9–12]. These technologies have led to the development of biomarker signatures that provide prognostic information. Genomic testing can thus provide valuable information that helps healthcare providers offer treatment individualised to each patient's risk profile. For example, some of these tests can predict recurrence risk with acceptable accuracy and the potential utility of adding chemotherapy to endocrine treatment [13], [11, 12,14–19]. Current guidelines recommend the use of such biomarkers to guide treatment decisions [20–23]. The most recent European practice guidelines for the management of early breast cancer state that "defining cohorts most appropriate for [chemotherapy] increasingly depends on classifying tumours based on genomic signatures as well as other biological factors (…) that refine prognosis beyond pathology alone" and recommends that gene expression assays or endocrine response assessment be performed after confirmation of the diagnosis in patients with early HR+ (hormone receptor positive) and HER2- breast cancer (HR+/HER2-) (stage N0-1), if relevant for the therapy decision [20,21].

Launched in 2022, myC (my Cancer my Concern) is an initiative of Cancer Patients Europe (CPE) that aims to raise awareness of the benefits and values of genomic testing in cancer, educate patients and other concerned parties, and improve accessibility to genomic testing across Europe. The Genomics and Breast Cancer European Patient Survey described here was supported by Exact Sciences and is the first step of the myC initiative and part of CPE's broader goal to improve the management of cancer for patients across Europe. The objective of the survey was to describe current practice with regards to genomic testing for women with breast cancer, in particular relating to awareness, available information, and uptake of the test.

Some of the data reported here were presented at the Congress of the European Society for Medical Oncology (ESMO) held in Madrid, October 2023.

2. Methods

This study was a web-based survey, initiated by CPE and conducted in five European countries (France, Germany, Italy, Spain, and the United Kingdom). An advisory committee was established to advise on the development, implementation, and interpretation of the study, consisting of representatives of European cancer patient associations, oncologists, and a psycho oncologist. Data were collected by an independent moderator (LEAD-UP Medical Network, Paris, France) using a web-based survey software (Survey Monkey®). The survey was available over a six-week period from September 8, 2022 to October 23, 2022.

2.1. Participants

Respondents were recruited by e-mails sent to members of the patient associations involved in the study and through a social media campaign implemented by Cancer Patients Europe and the national patient associations. The invitation explained that the survey concerned breast cancer and genomic testing. The survey was open to anyone who was interested in participating, in particular patients with breast cancer and their caregivers. The data presented here are restricted to the analysis of participants who identified themselves as women with a current or previous diagnosis of HR+/HER2- breast cancer. Women with other types of breast cancer such as triple negative breast cancer, participants who identified themselves as men with breast cancer and carers or relatives were excluded from the present analysis.

2.2. Study questionnaire

The questionnaire was developed specifically for this study by CPE and the advisory committee and included 42 mandatory (response-dependant) multiple choice questions. Completion of the questionnaire took around 10 min. The questionnaire was available in five languages (English, French, German, Italian and Spanish).

The first half of the questionnaire consisted of demographic questions and knowledge of precision medicine and genomic testing, knowledge about treatment and sources of information on cancer. These questions were completed by all participants. The second half of the questionnaire was only completed by participants with current or previous breast cancer or their carers. This section encompassed experience with breast cancer diagnosis, involvement in treatment choices, patient support, worries and concerns, and personal experience with genomic testing (restricted to participants with HR+/HER2- breast cancer, who were potentially eligible for testing). The questionnaire included an open question for participants comments. An overview of the structure of the questionnaire, and which type of participant completed which type of questions is provided in Supplementary Table 1, together with the full questionnaire itself.

2.3. Statistical analysis

Data presentation is purely descriptive. Certain variables were compared between sub-groups according to country, education level and urban/rural residence using the χ^2 test.

2.4. Ethics

As data collection was anonymous and implementation of the study had no impact on patient care, ethics committee approval was not deemed necessary. The survey was conducted in accordance with European and national codes for data protection. The online survey interface provided information on the goals and procedures of the study and required participants to provide explicit consent to participate in the survey, on the basis of the information provided, before proceeding to the study questionnaire. Participants received no incentives for study participation.

3. Results

3.1. Study participants

A total of 1776 participants opened the survey and 1419 completed it (79.9 %). After excluding 36 participants not living in participating countries, 1383 remained in the study. (Fig. 1). The analysis population consisted of 566 women with current or previous HR+/HER2- breast cancer, who could have been eligible for genomic testing at the time of diagnosis (39.9 % of completers). The primary reasons for exclusion from the analysis were participants who did not personally experience breast cancer (N = 273) or had a type of breast cancer other than HR+/HER2- (N = 469) (Fig. 1).

The demographic characteristics of the 566 participants are presented in Table 1. The most represented age group was 50–59 years,

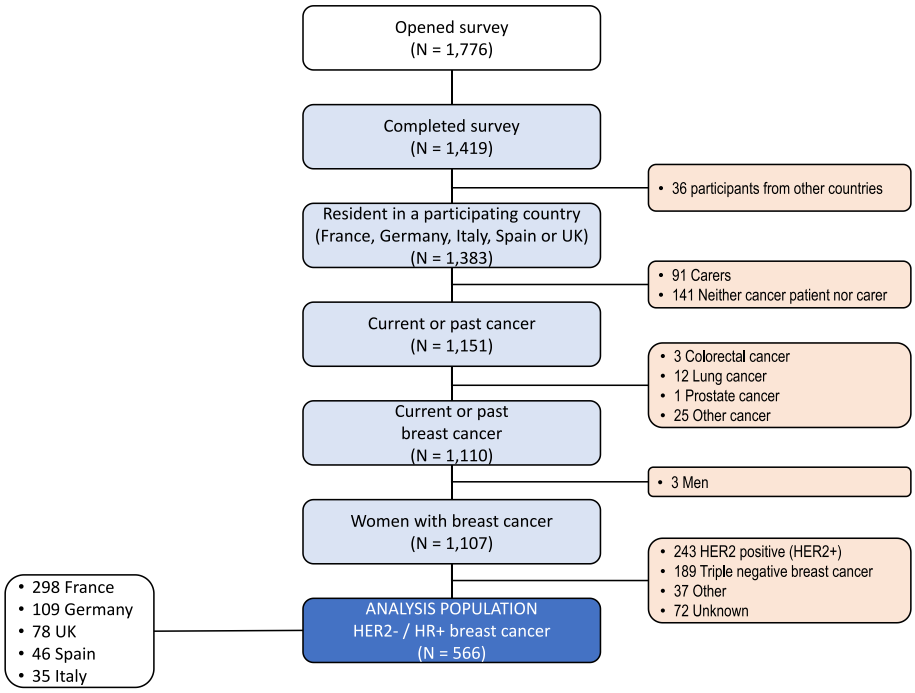

Fig. 1. Flow chart diagram.

Table 1
Demographic characteristics of the participants.

ANALYSIS POPULATION	566	(100 %)
Age		
<25 years	0	(0.0 %)
25–39 years	46	(8.1 %)
40–49 years	156	(27.6 %)
50–59 years	245	(43.3 %)
60–69 years	92	(16.3 %)
≥70 years	27	(4.8 %)
Highest level of education		
Primary school	7	1.2 %)
Secondary school	178	(31.5 %)
University	262	(46.3 %)
Postgraduate	119	(21.0 %)
Country of residence		
France	298	(52.7 %)
Germany	109	(19.3 %)
United Kingdom	78	(13.8 %)
Spain	46	(8.1 %)
Italy	35	(6.2 %)
Place of residence (population size)		
Conurbation (>500,000 inhabitants)	110	(19.4 %)
Large city (>200,000 and < 500,000 inhabitants)	52	(9.2 %)
Medium-sized city (>50,000 and < 200,000 inhabitants)	113	(20.0 %)
Small town (<50,000 inhabitants)	148	(26.2 %)
Village/rural community (<7500 inhabitants)	143	(25.3 %)

accounting for 43.3 % of participants. The sample was well-educated with 381 (67.3 %) having gone to university. Around half of women (n = 291; 51.5 %) lived in small towns or rural communities.

3.2. Awareness and perceptions of genomic testing

The results of the survey relating to awareness and perceptions of genomic testing are presented in Table 2. Almost all analysis participants thought that cancer treatment can be personalised (N = 554; 97.9 %) but less than half had heard about "personalised or precision medicine" (N = 240; 42.4 %). Three hundred and twenty participants were aware of cancer biomarkers (56.5 %) and 238 (42.1 %) declared having heard about genomic testing in cancer. Participants provided various reasons for the use of genomic testing in certain breast cancers; however, 226 women (39.9 %) said that they did not know. Less than 20 % reported that the test had been explained to them and considered that they had received enough information to make a decision.

The proportion of participants who declared having heard of genomic testing increased significantly with the level of education ($p = 0.0016$; Fig. 2). No difference was observed between women living in urban and rural environments ($p = 0.701$; data not shown).

3.3. Use of genomic testing

Overall, 139 (24.6 %) women decided to proceed with genomic testing. The principal reasons for not taking the test were not having received enough information (32.3 % of patients), but of the 211 participants who replied 'Other', 108 participants stated that they were not aware of the test and a further 73 that they had not been offered it by their physician. Twelve women stated that the test was not available when they were diagnosed with breast cancer.

Only 122 women (21.6 %) were told that they were eligible for genomic testing (Table 2) and 105 of these subsequently proceeded with the testing (86.1 %). Fourteen women who were eligible did not proceed with testing, citing reasons including lack of information (7 participants), because the test was not thought to be necessary by the healthcare professional (4 participants), because the test was not covered by health insurance (2 participants), or because the quality of the tumour sample was insufficient (1 participant). In three cases, the physician ordered the test without consulting the patient.

3.4. Country variations

Significant differences were observed between countries in both awareness and use of genomic testing (Fig. 3). The proportion of patients who had heard of genomic testing ranged from 36.7 % in Germany to 74.3 % in Italy ($p = 0.009$). The proportion of patients who were told they were eligible for genomic testing ranged from 17.4 % in Spain to 37.1 % in Italy, and the proportion who actually took the test from 19.5 % in France to 40.0 % in Italy.

Table 2
Awareness and perceptions of genomic testing.

ANALYSIS POPULATION	566	(100 %)
Have you heard about the concept of personalised or precision medicine?[a]	240	(42.4 %)
If so, what do you think it means?		
A personalised treatment schedule.	137	(57.1 %)
A medical assessment based on a person's likelihood of getting a disease.	48	(20.0 %)
Personalised medical appointments.	28	(11.7 %)
A method for tailoring the right treatment for the right person.	184	(76.7 %)
Do you think that cancer treatment could be personalised?	554	(97.9 %)
Do you know what cancer biomarkers are?	320	(56.5 %)
Have you heard genomic testing in cancer?	238	(42.1 %)
Do you know why genomic testing is used in some breast cancers?[a]		
To know if you might benefit from chemotherapy	125	(22.1 %)
To help estimate the risk of disease returning	117	(20.7 %)
To help choose the appropriate treatment	160	(28.3 %)
All of the above	142	(25.1 %)
I don't know	226	(39.9 %)
Were you told you were eligible for genomic testing? Yes	122	(21.6 %)
If yes, the reasons for genomic testing were explained	104	(85.3 %)
Did someone explain how the test is performed?	85	(15.0 %)
Did you have enough information to make a decision about having a genomic test?	87	(15.4 %)
Did you decide to take a genomic test?	139	(24.6 %)
If not, why not?[a]		
I was scared of taking the test	2	(0.5 %)
I did not have enough information	138	(32.3 %)
My healthcare professional did not think it was necessary	66	(15.5 %)
The test was not covered by my health insurance	4	(0.9 %)
I could not afford the test	6	(1.4 %)
Other (please specify).	211	(49.4 %)

[a] Multiple responses possible.

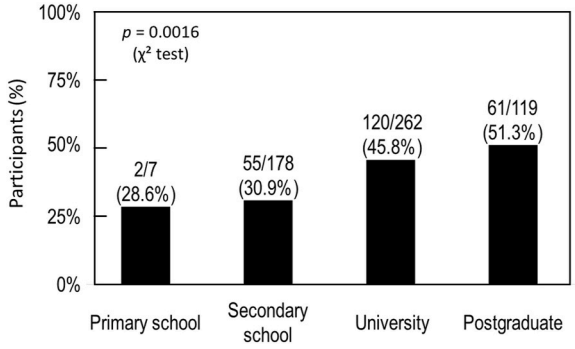

Fig. 2. Awareness of genomic testing by level of education.

3.5. Sources of information

Two-thirds of patients considered that they had received enough information about their breast cancer and that their physician had discussed treatment with them (Supplementary Table 2). When treatment options were discussed with the patients, surgery was mentioned in 93.9 % of cases as well as radiotherapy (85.0 %), hormone therapy (82.7 %) or chemotherapy (71.3 %).

When searching for more medical information, participants most frequently consulted healthcare professionals or the internet. In only a minority of cases did the healthcare professional suggest where to find more information (29.7 %). Nonetheless, participants would have liked to have received more information on long-term treatment effects (57.4 %), additional testing (43.5 %), treatment options (41.3 %), type of cancer (23.3 %) or local patient support groups (27.7 %).

4. Discussion

The principal finding of this study was a low level of awareness of genomic testing in women with HR+/HER2 cancer in Europe, with 57.8 % of participants not having heard of this technology. The degree of awareness increased by education level. An American study published over a decade ago reported a low level of awareness of genomic testing [24]. Despite mounting evidence, endorsement in practice guidelines and wider test use, our study, together with recent studies performed in other non-European countries [25,26], demonstrate that awareness has not increased over the intervening decade.

We observed significant differences between patients from the five European countries surveyed, with the proportion of women declaring that they had heard of genomic testing in Italy being >20 % higher than in the other four countries. It should nevertheless be noted that patient associations in different countries may have different extent of involvement with patients and may represent a more-or-less wide patient base. Additionally, the level of utilisation of social media campaigns differed across patient associations, with some being more active than others. These differences may contribute to the very different numbers of patients recruited in each country. For this reason, between-country differences should be interpreted with caution.

The study population was generally highly educated (67.3 % having gone to university) with a potential link to a patient association, and this clearly does not reflect the actual structure of education levels in the European population in general. Our population probably has a higher level of health literacy in such patients as compared to the general population. In consequence, it may be assumed that the level of awareness of genomic testing in the general population of women with ER+/HER2- breast cancer is even lower than that reported in the present study. It would be important in future studies to attempt to harness this information from a more educationally representative sample of breast cancer patients in Europe, for example by providing a questionnaire to be completed by women at the point of treatment assignment following diagnosis.

In principle, women constituting the analysis population were all potentially eligible for genomic testing as they had HR+/HER2- breast cancer. However, in current practice, only a minority undergo testing. In the present study, only 21.6 % were told that they were eligible for genomic testing and only 24.6 % were tested. Again, country differences in uptake rates were observed with the highest reported use in Italy. A consequence of such a low level of genomic testing may be the prescription of adjuvant chemotherapy for women unlikely to benefit from it, with the associated risks and inconvenience to the patient and unnecessary cost to the health system for the expense of chemotherapy, associated supportive therapies and non-medical costs [27]. On the other hand, not performing genomic testing could also lead to under-treatment by not prescribing chemotherapy in patients who would benefit from it, and high costs from potential relapse or metastasis. [27]. A previous American survey has reported a strong interest in genomic

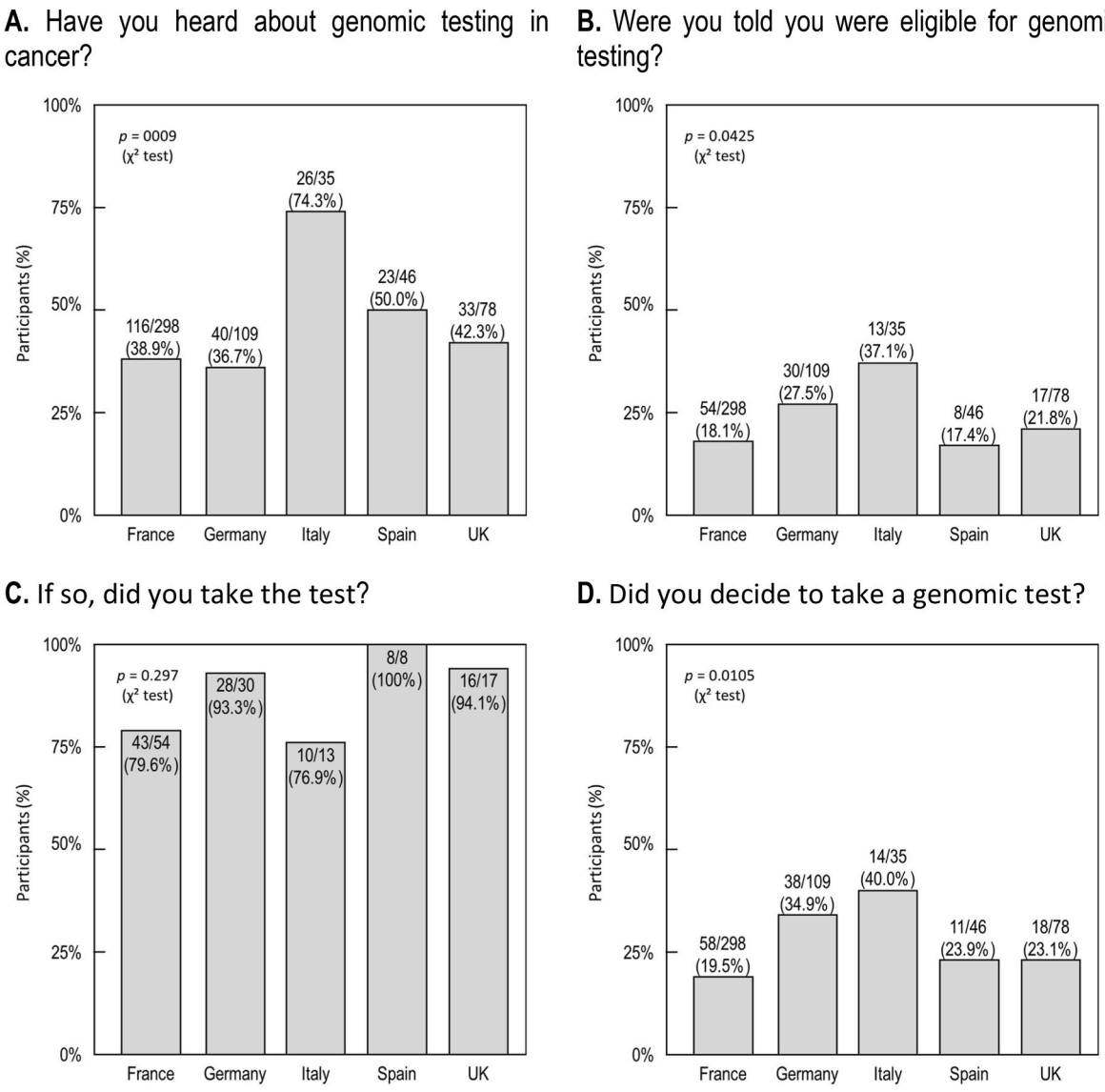

Fig. 3. Awareness and use of genomic testing by country.
Data are reported for the analysis population of 566 women potentially eligible for genomic testing.

testing from eligible women, with 76 % of participants stating they would "definitely" want to be tested [28]. Participants believed in using the results to inform treatment decision making.

The study has several strengths and limitations. The strengths include the relatively large sample size and the high completion rate for this type of survey. In addition, the multinational scope enabled potential differences between countries to be evaluated. Limitations include the fact that the data represent the declarations of participants, and it is not possible to verify the information independently. In addition, no clinical information was collected nor any other clinicopathological markers that contribute to the treatment decision. Our survey was recruited using patient associations and online social media campaigns which might cause a bias towards people with higher knowledge of breast cancer and internet use. In consequence, participants who were more computer-literate may have been more likely to complete the survey. This might explain the relative young age of participants in the survey compared to breast cancer patients in general. In addition, the survey did not collect information concerning the date of breast cancer diagnosis. This could mean that the testing was not an option for patients diagnosed before the tests were introduced or reimbursed by health authorities. However, given that the median age of diagnosis of breast cancer is now in the early 60s [29], the fact that only one in five of our sample was over age 60 would suggest that most participants had been diagnosed recently. Another limitation may relate to the understanding of the terminology used for genomic testing; there is no standardised vocabulary, and this is reflected in the related scientific literature where a range of terms including, but not limited to, 'microarray testing', 'expression profiling of tumour tissue', 'gene expression analysis', 'recurrence biomarkers' are used. We used the terms 'precision' and 'personalised' to refer to genomic testing in recognition of this common usage, although our preference is to refer to 'precision medicine' when reflecting molecular or genomic testing [30]. Many women (68 %) believed that that 'personalised' or 'precision' medicine may refer to treatment schedules or appointments demonstrating the need for clarity in language. There is also a possible confusion with genetic tests for breast cancer predisposition (especially BRCA1 or BRCA2 mutations). Differences in how physicians refer to the technology with their patients in different languages may affect recognition by the patient, and this may have influenced some of the country differences observed.

The findings emphasise the need for physicians to integrate genomic testing in their dialogue with patients when indicated about their type of breast cancer and the treatment options available. The current European

practice guidelines indicate that the management of early breast cancer should be multidisciplinary and provided by specialised breast units [20,21,31] and it is important that genomic testing should be considered for all patients with profiles for which testing is recommended in practice guidelines. Discussion about risks and benefits of systemic therapy is usually performed by an oncologist but the decision should be taken by a multidisciplinary tumour board. Surveys of healthcare professionals involved show that confidence in genomic testing grows with greater experience and volume of testing [32].

Study participants showed high levels of using the internet for medical advice in line with previously published studies [33–36]. However, information available may not be directly relevant, inaccurate or hard to find [34] and can increase worry [37]. In this context, we would recommend that healthcare professionals help patients navigate to reliable resources and that patient associations collaborate to produce independent and accurate information codesigned with patients. Moreover, patient associations could provide a valuable channel to provide this information directly to patients.

In conclusion, in this study, four out of five women with HR+/HER2– breast cancer were not told that they were eligible for genomic testing and three out of five were not aware of it. Given the education level of participants, this is probably an upper estimate of the true level of awareness in Europe. This shortfall should encourage health service providers to ensure that genomic testing is more widely discussed with patients and offered to those who could benefit, in order to ensure appropriate prescription of adjuvant chemotherapy to breast cancer patients. Furthermore, patient associations have a crucial role to play as accessible portals for providing patient-friendly information.

CRediT authorship contribution statement

Antonella Cardone: Writing – review & editing, Writing – original draft, Validation, Supervision, Project administration, Methodology, Funding acquisition, Conceptualization. **Dany Bell:** Writing – review & editing, Validation, Methodology, Conceptualization. **Conchi Biurrun:** Writing – review & editing, Validation, Methodology, Conceptualization. **Francesco Cognetti:** Writing – review & editing, Validation, Methodology, Conceptualization. **Fatima Cardoso:** Writing – review & editing, Validation, Methodology, Conceptualization. **Ana Ramirez Piris:** Writing – review & editing, Validation, Methodology, Funding acquisition, Conceptualization. **Csaba Degi:** Writing – review & editing, Validation, Methodology, Conceptualization. **Michael Patrick Lux:** Writing – review & editing, Validation, Methodology, Conceptualization. **Richard Simcock:** Writing – review & editing, Validation, Methodology, Conceptualization. **Johanna Wassermann:** Writing – review & editing, Validation, Methodology, Conceptualization. **Rosanna D'Antona:** Writing – review & editing, Validation, Methodology, Conceptualization. **Isabel T. Rubio:** Writing – review & editing, Validation, Methodology.

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Declaration of competing interest

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Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.breast.2025.104436.

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