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## Knowledge, attitudes and behavior of physicians regarding predictive genetic tests for breast and colorectal cancer<sup>☆</sup>

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## ABSTRACT

**Background.** Genetic testing for cancer susceptibility is an emerging technology in medicine. This study assessed the knowledge, attitudes and professional behavior of Italian physicians regarding the use of predictive genetic tests for breast and colorectal cancer, including the *BRCA1/2* and *APC* tests.

**Methods.** A cross-sectional survey of a random sample of Italian physicians was performed in 2010 through a self-administered questionnaire.

**Results.** A response rate of 69.6% (1079 questionnaires) was achieved. A significant lack of knowledge was detected, particularly for *APC* testing. Less than half of the physicians agreed on the importance of efficacy and cost-effectiveness evidence in the selection of predictive genetic tests to be offered to the patients. Multiple logistic regression analyses showed that education had a positive influence on knowledge, attitudes and, to a lesser extent, professional use. The factor most strongly related to the physicians' use of genetic testing was patients requests for breast (odds ratio = 12.65; 95% confidence interval 7.77–20.59) or colorectal cancer tests (odds ratio = 7.02; 95% confidence interval 3.61–13.64). A high level of interest for specific training was reported by almost all physicians surveyed.

**Conclusions.** Targeted educational programs are needed to improve the expertise of physicians, and, ultimately, to enhance the appropriate use of genetic tests in clinical practice.

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## Introduction

Among predictive genetic testing for complex diseases, tests for breast and colorectal cancer, if used appropriately, have been demonstrated to be efficacious and cost-effective (Becker et al., 2011). Physicians play a key role in properly incorporating emerging DNA technologies in health care (Anon, 2011; Feero and Green, 2011) because they have to be adept not only at using genetic tests in clinical care but also in explaining the test results and their limitations to patients.

Calls for enhanced genomic education for health care professionals predate the completion of the Human Genome Project (Collins, 1997).

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Despite this, several surveys performed in the U.S., Europe and Canada show that doctors are not prepared for the increasing use of genetics in clinical care (Acton et al., 2000; Batra et al., 2002; Bellcross et al., 2011; Bethea et al., 2008; Burke et al., 2009; Carroll et al., 2008; Escher and Sappino, 2000; Freedman et al., 2003; Klitzman et al., 2012; Mehnert et al., 2003; Nippert et al., 2011; Pichert et al., 2003; Sabatino et al., 2007; Shields et al., 2008; Sifri et al., 2003; Toiviainen and Hemminki, 2001; Trivers et al., 2011; Van Riel et al., 2010; Welkenhuysen and Evers-Kiebooms, 2002; White et al., 2008; Wideroff et al., 2003; Wideroff et al., 2005; Wilkins-Haug et al., 2000). Many physicians do not have any specific education and the vast majority does not feel they have the needed training and knowledge for the appropriate use of genetic testing to guide prevention or treatment decisions (Anon, 2011; Feero and Green, 2011). Recent surveys tested the effectiveness of educational interventions at improving the competency of doctors in this field (Bethea et al., 2008; Carroll et al., 2008, 2009; Drury et al., 2007).

The present study assessed the knowledge, attitudes, and professional behavior of a random sample of Italian physicians toward the use of predictive genetic testing for breast and colorectal cancer, particularly the *BRCA 1/2* and *APC* tests. A variety of determinants were explored, including education.

## Methods

In 2010, a self-administered anonymous questionnaire was e-mailed to 1670 physicians randomly selected from the registers of the Board of Physicians of Provinces of Rome and Florence. The physicians were chosen irrespective of their specialty because this information is not recorded in the registers. The online questionnaire could only be answered once. Second and third questionnaires were e-mailed to non-responders 3 and 6 months after the initial e-mail. To maximize the response rate, telephone calls were placed before each of the follow-up mailings. A total of 107 physicians could not be contacted by telephone because their numbers were not available.

The questionnaire (a copy is available upon request) comprised a series of questions designed to assess the following: i) the physicians' demographics and personal and professional characteristics; ii) their knowledge, attitudes, and professional use of genetic tests for breast and colorectal cancer; iii) their self-estimated level of knowledge and training needs.

Knowledge about predictive genetic tests for cancer was investigated through six questions using a three-point options Likert scale ("agree", "uncertain", and "disagree") [see Table 2(A) for the actual items used]. Additional four multiple-choice questions were designed to evaluate the physicians' knowledge concerning the prevalence of hereditary breast cancer and inherited forms of colorectal cancer and the penetrance of *BRCA1/BRCA2* and *APC* mutations [see Table 2(B)]. A Likert three-point scale was used to assess the physicians' attitudes through seven questions (see Table 4). In the behavior section, physicians were asked if they had administered genetic tests for breast and colorectal cancer to their patients during the previous 2 years and queried about the importance of genetic counseling and collecting information about the family and personal history of cancer. The final set of questions required the physicians to assess their own level of knowledge according to a four-answer format ("inadequate," "sufficient," "good," and "excellent") and their need for training ("yes/no" answer).

Extensive pre-administration piloting was conducted with a convenience sample of physicians similar to the study population. A clear need to slim down the questionnaire emerged. Therefore, only questions concerning *APC* mutations were included among the knowledge items concerning the inherited forms of colorectal cancer, thus excluding questions regarding gene mutations associated with the Lynch syndrome. Other minor revisions included changes to the questionnaire item wording and format.

## Statistical analysis

Multiple logistic regression analysis was performed. Five models were built to identify the predictors of physicians knowledge of predictive genetic testing for breast and colorectal cancer (Models 1 and 2), attitudes (Model 3), and professional use of predictive genetic tests for breast and colorectal cancer (Models 4 and 5). For purposes of analyses, the outcome variables "knowledge" and "attitudes" in Models 1–3, originally consisting of multiple categories, were collapsed into two levels. In brief, for the variable knowledge physicians were divided in those who agreed with all correct responses versus all others, while for attitudes responders were grouped into those who showed a positive attitude in at least 70% of the questions versus all others (see Table 3 for the details of dichotomization). The following physician characteristics were initially tested in all models as predictor variables: location; gender; age; exposure to cancer genetic testing during graduate/postgraduate courses; attendance to postgraduate epidemiology and Evidence Based Medicine (EBM) courses; knowledge of the English language; internet access in the workplace; hours per week dedicated to continuing medical education; the average number of patients treated in a typical week; patient requests for genetic tests in the previous year; the presence of genetic testing laboratories in the geographical area of professional activity; and a personal or family history of breast or colorectal cancer. The variable "adequate knowledge" was also included in the model concerning attitudes, and the variables "adequate knowledge" and "positive attitudes" were included in the models concerning the professional use of predictive genetic tests (see Table 3 for the details of dichotomization).

The model building strategy suggested by Hosmer and Lemeshow (2000) was used and included the following steps: (a) univariate

analysis of each variable and inclusion if the *p*-value was lower than 0.25; (b) backward elimination of each variable that did not contribute to the model on the ground of the Likelihood Ratio Test using a cut-off of 0.05 level of significance; variables whose exclusion markedly altered the coefficient of the remaining variables were kept in the model; (c) testing of interaction terms using a cut-off of 0.15 level of significance. Adjusted odds ratio (OR) and 95% confidence intervals (CI) were calculated. All statistical calculations were performed using Stata version 8.0 (College Station, Texas, Stata Corporation, 2003).

## Results

### Study population

Of the original sample of 1670 physicians, 120 were ineligible because they were retired or no longer in clinical practice. The final sample size included 1550 physicians, of which 1079 responded (overall response rate: 69.6%). Responders and non-responders were comparable in terms of demographic characteristics (location, gender, and age; *p* > 0.05). Most responding physicians were from Rome (73.8% of responders vs. 76.9% of non-responders) and male (56.2% of responders vs. 58.9% of non-responders), with a mean age of 50.7 ( $\pm 11.5$ ) years (50.0 years for non-responders). The demographic characteristics of the sample were similar to those of all Italian physicians, as 60.6% of the members of the National Board of Physicians are male and have a similar age distribution (ENPAM, 2012). Other demographics, professional and personal characteristics of the responding physicians are listed in Table 1.

### Knowledge

Italian physicians' knowledge of predictive genetic testing for cancer appeared adequate in terms of *BRCA1/BRCA2* testing, although knowledge of *APC* testing was lacking [Table 2(A)]. Almost half of the sample (42.8%) answered all three questions about *BRCA1/2* testing correctly. This knowledge was improved if physicians were exposed to cancer genetic testing during graduate or postgraduate training, and with the increase in the amount of time dedicated to continuing medical education. Female physicians were more likely to have adequate knowledge about *BRCA1/2* testing, and this knowledge increased if genetic testing laboratories were located in the same geographical area as the physicians' workplace (Model 1 in Table 3). Only 16.9% of physicians provided correct answers to all three questions about *APC* testing. This knowledge, as in the previous case, increased with exposure to cancer genetic testing during graduate and postgraduate training and with the amount of time dedicated to continuing medical education (Model 2 in Table 3).

Physicians' knowledge was satisfactory on the penetrance of *BRCA1/BRCA2* mutations, but not regarding the prevalence of hereditary breast cancer. Most physicians knew that the absolute risk of developing breast cancer in the presence of *BRCA1/BRCA2* mutations is 40–80%, but less than one third recognized that the percentage of breast cancer cases associated with *BRCA1/BRCA2* mutations is 1–10% [Table 2(B)]. By contrast, knowledge concerning inherited forms of colorectal cancer was inadequate, as none of the surveyed physicians knew that the percentage of colorectal cancer cases associated with *APC* mutations is less than 5%, and only a small proportion of physicians recognized that the absolute risk of developing cancer in the presence of *APC* mutations is 100% [Table 2(B)].

### Attitudes

Attitudes toward predictive genetic testing for breast and colorectal cancer were quite heterogeneous (Table 4). Although nearly all physicians agreed that predictive genetic testing increases the chances of prevention, only a minority appeared to accept that the principles of

**Table 1**  
Demographics and professional and personal characteristics of the responding physicians. Italy, 2010.

Variables	n	(%)
Location (1079) <sup>a</sup>		
Florence	283	(26.2)
Rome	796	(73.8)
Gender (1071) <sup>a</sup>		
Female	469	(43.8)
Male	602	(56.2)
Age, years (1015) <sup>a</sup>		
<30	65	(6.4)
31–40	151	(14.9)
41–50	208	(20.5)
51–60	419	(41.3)
≥61	172	(16.9)
Exposure to cancer genetic testing during graduate training (1008) <sup>a</sup>		
No	803	(79.7)
Yes	205	(20.3)
Exposure to cancer genetic testing during postgraduate training (1009) <sup>a</sup>		
No	795	(78.8)
Yes	214	(21.2)
Postgraduate training courses about epidemiology and EBM (1007) <sup>a</sup>		
No	617	(61.3)
Yes	390	(38.7)
English language knowledge (1010) <sup>a</sup>		
Very low	113	(11.2)
Low	232	(23.0)
Intermediate	349	(34.5)
Good	221	(21.9)
Excellent	95	(9.4)
Internet available in the workplace (1004) <sup>a</sup>		
No	89	(8.9)
Yes	915	(91.1)
Hours per week dedicated to continuing medical education (890) <sup>a</sup>		
<1	0	(0)
1–5	677	(76.1)
6–10	147	(16.5)
>10	66	(7.4)
Average number of patients treated in a typical week (1011) <sup>a</sup>		
1–20	396	(39.2)
21–40	235	(23.2)
41–60	149	(14.7)
>60	231	(22.9)
Patient requests for cancer genetic tests in the previous year (1006) <sup>a</sup>		
No	829	(82.4)
Yes	177	(17.6)
Presence of genetic testing laboratories in the geographical area of professional activity (1000) <sup>a</sup>		
No	505	(50.5)
Yes	495	(49.5)
Personal or family history of breast cancer (1011) <sup>a</sup>		
No	778	(76.9)
Yes	233	(23.1)
Personal or family history of colorectal cancer (1009) <sup>a</sup>		
No	760	(75.3)
Yes	249	(24.7)
Ordering or referring patients for breast cancer predictive genetic testing in the previous 2 years (1007) <sup>a</sup>		
No	906	(90.0)
Yes	101	(10.0)
Ordering or referring patients for colorectal cancer predictive genetic testing in the previous 2 years (1004) <sup>a</sup>		
No	957	(95.3)
Yes	47	(4.7)

<sup>a</sup> Number of physicians responding to the question.

efficacy and cost-effectiveness should be used to select predictive genetic tests to be delivered to the general population. Most physicians agreed on the importance of evidence-based guidelines, genetic counseling, and the ethical, legal and social implications of predictive

**Table 2**  
Knowledge of the responding physicians regarding: (A) predictive genetic testing for breast and colorectal cancer (*BRCA1/2* and *APC* testing); (B) prevalence of hereditary forms of breast and colorectal cancer and penetrance of *BRCA1/2* and *APC* mutations. Italy, 2010.

(A) Knowledge of predictive genetic testing for breast and colorectal cancer	Agree, n (%)	Uncertain, n (%)	Disagree, n (%)
Predictive genetic tests for <i>BRCA1/BRCA2</i> mutations are able to identify patients at high risk to develop breast cancer (848) <sup>a</sup>	<b>622 (73.3)</b>	226 (26.7)	0
Women with breast cancer and strong family history should perform <i>BRCA1/BRCA2</i> testing (838) <sup>a</sup>	<b>633 (75.5)</b>	154 (18.4)	51 (6.1)
Scientific evidence recommends for <i>BRCA1/BRCA2</i> positive women clinical and instrumental surveillance starting from the age of 25 (839) <sup>a</sup>	<b>699 (83.3)</b>	124 (14.8)	16 (1.9)
Predictive genetic tests for <i>APC</i> mutations are able to identify patients who will develop colorectal carcinoma (856) <sup>a</sup>	<b>571 (66.7)</b>	246 (28.7)	39 (4.6)
<i>APC</i> testing is recommended for 10–12 years old children with a first grade relative with known <i>APC</i> mutation (780) <sup>a</sup>	<b>337 (43.2)</b>	339 (43.5)	104 (13.3)
Scientific evidence recommends for <i>APC</i> positive individuals periodic colonoscopy, starting from the age of 10–15 (809) <sup>a</sup>	<b>325 (40.2)</b>	353 (43.6)	131 (16.2)
(B) Knowledge of the prevalence of hereditary forms of breast and colorectal cancer and penetrance of <i>BRCA1/2</i> and <i>APC</i> mutations			n (%)
The percentage of breast cancer cases associated with <i>BRCA1/2</i> mutations is (766) <sup>a</sup> :			
1–10%			<b>230 (30.0)</b>
15–35%			387 (50.5)
>50%			149 (19.5)
The absolute risk of developing breast cancer in the presence of <i>BRCA1/2</i> mutations is (599) <sup>a</sup> :			
<10%			0
40–80%			<b>568 (94.8)</b>
100%			31 (5.2)
The percentage of colorectal cancer cases associated with <i>APC</i> mutations is (587) <sup>a</sup> :			
<5%			<b>0</b>
10–25%			386 (65.8)
>40%			201 (34.2)
The absolute risk of developing colorectal cancer in the presence of <i>APC</i> mutations is (583) <sup>a</sup> :			
<10%			0
40–80%			515 (88.3)
100%			<b>68 (11.7)</b>

Note: Number and percentages referring to correct answers are in bold.

<sup>a</sup> Number of physicians responding to the question.

genetic testing. A total of 23.8% of physicians showed a positive attitude in at least 70% of the questions, and this dichotomization was arbitrarily used to identify predictors of a positive attitude. Significant predictors of positive attitudes included the following: (a) exposure to cancer genetic tests during graduate training and attendance at postgraduate training courses in epidemiology and EBM, and (b) no patient requests for cancer genetic tests in the previous year and presence of genetic testing laboratories in the local area. Female physicians were more likely to show positive attitudes, as were physicians with an adequate knowledge of predictive genetic testing for both breast and colorectal cancers (Model 3 in Table 3).

#### Professional behavior

Few physicians in our sample had either referred patients for or ordered predictive genetic testing for breast (10.0%) or colorectal cancer (4.7%) in the previous 2 years. The main determinant of professional use was the patient requests for genetic testing (Models 4 and 5 in Table 3). Other significant determinants included the following: (a) adequate knowledge of the professional use of predictive genetic testing for breast cancer (Model 4 in Table 3), and (b) the number

**Table 3**  
Determinants of physicians knowledge, attitudes and professional behavior concerning predictive genetic testing for cancer. Italy, 2010.

Variables	OR	95% CI
<i>Model 1: Knowledge about predictive genetic testing for breast cancer (BRCA1/BRCA2 mutations).<sup>a</sup></i>		
Gender (female = 0; male = 1)	0.72	0.54–0.95
Hours per week dedicated to continuing medical education (<1 = 0; 1–5 = 1; 6–10 = 2; >10 = 3) <sup>b</sup>	1.50	1.18–1.90
Exposure to cancer genetic tests during graduate training (No = 0; Yes = 1)	2.33	1.59–3.40
Exposure to cancer genetic tests during postgraduate training (No = 0; Yes = 1)	1.59	1.10–2.29
Presence of genetic testing laboratories in the geographical area of professional activity (No = 0; Yes = 1)	1.38	1.04–1.83
<i>Model 2: Knowledge about predictive genetic testing for colorectal cancer (APC mutations).<sup>c</sup></i>		
Hours per week dedicated to continuing medical education (<1 = 0; 1–5 = 1; 6–10 = 2; >10 = 3) <sup>b</sup>	1.53	1.18–1.98
Exposure to cancer genetic tests during graduate training (No = 0; Yes = 1)	1.67	1.09–2.56
Exposure to cancer genetic tests during postgraduate training (No = 0; Yes = 1)	1.74	1.14–2.64
<i>Model 3: Attitudes towards predictive genetic testing for breast and colorectal cancer.<sup>d</sup></i>		
Gender (female = 0; male = 1)	0.54	0.40–0.71
Exposure to cancer genetic tests during graduate training (No = 0; Yes = 1)	1.87	1.30–2.69
Postgraduate training courses about epidemiology and EBM (No = 0; Yes = 1)	1.71	1.28–2.28
Patient request of cancer genetic tests in the previous year (No = 0; Yes = 1)	0.64	0.44–0.93
Presence of genetic testing laboratories in the geographical area of professional activity (No = 0; Yes = 1)	1.41	1.06–1.88
Knowledge about predictive genetic testing for breast and colorectal cancer (Not adequate = 0; adequate = 1) <sup>e</sup>	1.66	1.11–2.48
<i>Model 4: Professional use of predictive genetic testing for breast cancer<sup>f</sup></i>		
Patient request of cancer genetic tests in the previous year (No = 0; Yes = 1)	12.65	7.77–20.59
Knowledge about predictive genetic testing for breast cancer (Not adequate = 0; adequate = 1) <sup>a</sup>	3.21	1.92–5.36
<i>Model 5: Professional use of predictive genetic testing for colorectal cancer<sup>g</sup></i>		
Hours per week dedicated to continuing medical education (<1 = 0; 1–5 = 1; 6–10 = 2; >10 = 3) <sup>b</sup>	1.99	1.30–3.05
Patient request of cancer genetic tests in the previous year (No = 0; Yes = 1)	7.02	3.61–13.64
Presence of genetic testing laboratories in the geographical area of professional activity (No = 0; Yes = 1)	2.05	1.01–4.21
Attitudes (Negative = 0; positive = 1) <sup>d</sup>	2.17	1.12–4.23

Note: OR: Odds Ratio. CI: Confidence Interval.

<sup>a</sup> Physicians were classified as those who answered correctly to all three questions on predictive genetic testing for breast cancer [first three questions in Table 2(A)] vs. all others.

<sup>b</sup> Variable modeled as ordinal because linearity was assessed.

<sup>c</sup> Physicians were classified as those who answered correctly to all three questions on predictive genetic testing for colorectal cancer [last three questions in Table 2(A)] vs. all others.

<sup>d</sup> Physicians were divided into those who showed positive attitudes in at least 70% of the questions (at least five of the seven questions listed in Table 4) vs. all others.

<sup>e</sup> Physicians were classified as those who answered correctly to all six questions on predictive genetic testing for breast and colorectal cancers [Table 2(A)].

<sup>f</sup> Physicians who referred patients for or ordered predictive genetic tests for breast cancer in the previous 2 years were grouped vs. all others.

<sup>g</sup> Physicians who referred patients for or ordered predictive genetic tests for colorectal cancer in the previous 2 years were grouped vs. all others.

of hours per week dedicated to continuing medical education, the presence of genetic testing laboratories locally, and positive attitudes about the professional use of predictive genetic testing for colorectal cancer (Model 5 in Table 3). It is interesting to note that when ordering or referring patients to predictive genetic testing for cancer for patients, almost all physicians agreed upon the importance of collecting information about the family (99.6%) and personal history of cancer (98.0%) and the importance of genetic counseling (91.8%) (data not shown).

Approximately 80% of the physicians considered their knowledge of the appropriate use of predictive genetic testing for cancer to be inadequate; almost all of the physicians (94.2%) believed that their

knowledge should be improved, and 86.0% believed that specific post-training courses in predictive genetic testing for cancer are needed (data not shown).

### Discussion

Most surveys reported in the literature reveal a lack of knowledge regarding predictive genetic testing for cancer among physicians (Acton et al., 2000; Batra et al., 2002; Bellcross et al., 2011; Escher and Sappino, 2000; Klitzman et al., 2012; Nippert et al., 2011; Pichert et al., 2003; Wideroff et al., 2005; Wilkins-Haug et al., 2000).

**Table 4**  
Attitudes of the responding physicians towards predictive genetic testing for breast and colorectal cancer.

	Agree, n (%)	Uncertain, n (%)	Disagree, n (%)
Predictive genetic tests for breast and colorectal cancer increase the prevention opportunities (913) <sup>a</sup>	<b>771 (84.4)</b>	124 (13.6)	18 (2.0)
Predictive genetic tests able to identify an increased risk of developing breast or colorectal cancer should be performed even if there are no preventive and/or curative interventions of proven efficacy (882) <sup>a</sup>	343 (38.9)	284 (32.2)	<b>255 (28.9)</b>
Predictive genetic tests for breast or colorectal cancer should be performed only if economical evaluations show cost-effectiveness ratios favorable compared to alternative health interventions (898) <sup>a</sup>	<b>387 (43.1)</b>	258 (28.7)	253 (28.2)
Authoritative and evidence-based guidelines are needed for the appropriate use of predictive genetic tests for breast and colorectal cancer (931) <sup>a</sup>	<b>875 (94.0)</b>	56 (6.0)	0
Predictive genetic tests for breast and colorectal cancer should be performed without genetic counseling informing patients of the benefits and risks of the tests (919) <sup>a</sup>	125 (13.6)	159 (17.3)	<b>635 (69.1)</b>
Predictive genetic tests for breast and colorectal cancer can contribute efficaciously to health promotion and cancer prevention only if included in wider strategies taking into account the other available health interventions (914) <sup>a</sup>	<b>806 (88.2)</b>	83 (9.1)	25 (2.7)
The implementation of predictive genetic tests for breast and colorectal cancer, being a medical matter, should not take into account ethical, legal and social implications (922) <sup>a</sup>	255 (27.7)	160 (17.3)	<b>507 (55.0)</b>

Note: Number and percentages referring to answers denoting a positive attitude are in bold.

<sup>a</sup> Number of physicians responding to the question.

The present survey confirmed these findings, particularly concerning the prevalence of inherited forms of breast and colorectal cancer and the penetrance of *APC* mutations. These results have important practical implications because overestimating the prevalence of inherited forms of breast and colorectal cancer may result in the inappropriate and unnecessary use of predictive genetic tests. Conversely, if physicians underestimate the penetrance of the *APC* mutations, they may be less inclined to advise family members about the inherited risks, or less likely to refer patients to clinics that could provide optimum care. It is interesting to note that the items concerning education in the current survey were among the most important determinants of good knowledge of predictive genetic testing, confirming that education and specific training are fundamental issues that need to be addressed.

Physicians' attitudes usually have a vital impact on the process of technology diffusion. Many Italian physicians believed that predictive genetic testing for cancer should be performed without clear scientific evidence regarding the efficacy and cost-effectiveness of such interventions. These beliefs are in line with the findings obtained in more general terms by other Italian surveys (De Vito et al., 2009a, 2009b) and represent an obstacle to the appropriate use of predictive genetic tests because they are often introduced into clinical practice for commercial purposes, in the absence of rigorous evaluation of efficacy and cost-effectiveness (Col, 2003; EASAC and FEAM, 2012). Items concerning education and adequate knowledge had a positive impact on attitudes. The availability of local genetic testing laboratories increased the likelihood of a positive attitude. Unexpectedly, patient inquiries about cancer genetic testing during the previous year appeared to have a negative effect on attitudes. Female physicians were more likely to have a positive attitude (and adequate knowledge) than males, and this is in line with a greater attention of the female gender to predictive genetic testing for cancer ascertained in other surveys (Escher and Sappino, 2000; Geller and Holtzman, 1995; Wertz, 1993).

Concerning professional use of predictive genetic testing for cancer, approximately 10% of physicians declared that they had referred patients for or ordered predictive genetic testing for breast cancer (5% for tests for colorectal cancer) in the previous 2 years. These figures are similar to, or somewhat lower than, those reported in others surveys (Acton et al., 2000; Bellcross et al., 2011; Klitzman et al., 2012; Mehnert et al., 2003; Shields et al., 2008; Sifri et al., 2003; Welkenhuysen and Evers-Kiebooms, 2002; Wideroff et al., 2003). Even if nearly all the physicians acknowledged both the importance of collecting information regarding a familial and personal history of cancer and of genetic counseling in prescribing or referring patients for predictive genetic testing, the current survey was not designed to investigate whether the professional use of the tests was appropriate. The factors most strongly related to physicians' use of predictive genetic tests for cancer were patient requests during the previous year and, to a lesser extent, the presence of local genetic testing laboratories locally. Adequate knowledge, positive attitudes, and time spent for continuing medical education also had an impact on the likelihood of professional use. The importance of patient inquiries has been reported in the literature (Klitzman et al., 2012; Sifri et al., 2003; White et al., 2008; Wideroff et al., 2003). In the current survey, physicians caring for patients who asked for cancer predictive genetic testing during the past year reported a 13-fold and 7-fold greater use of tests for breast and colorectal cancer, respectively. The fact that the physicians' use of genetic tests appears to be guided, at least in part, by patient requests suggests that their decisions may be driven by factors other than clinical indications or clinical utility. These findings underscore the importance of the physician being ready to respond to patient requests for testing by providing patients with information about the advantages and limitations of such tests in addition to offering genetic counseling when appropriate or suggesting other alternatives when testing is not indicated.

This study has several limitations. First, a high percentage of non-responders (approximately 20%) was registered for questions concerning knowledge. Therefore, knowledge estimates reported in this study (calculated on responders) may be overestimated because non-responders may be less informed. Second, because information about specialties was not available from the registries of the Italian Boards of Physicians, the survey could not be designed to assess the likely differences that may exist across specialties. Although physicians were queried about their specialty in the questionnaire, the number of physicians in most specialties was too low to perform meaningful comparisons, therefore, the variable "specialty" was not included in the analyses. Finally, because a clear need to slim down the questionnaire emerged in the pilot study, only questions concerning *APC* gene mutations were included in the knowledge items concerning inherited forms of colorectal cancer, and questions on other gene mutations (e.g., for Lynch syndrome) were not included. *APC* mutations are less frequent but occur with a higher penetrance than other gene mutations. Previous surveys in the U.S. showed that physician's awareness of commercial availability was higher for *APC* tests than for tests for genes associated with Lynch syndrome (Batra et al., 2002; Wideroff et al., 2003). However, it should be acknowledged that there are no data available in the Italian context to conclude if knowledge about *APC* tests is equal or different from knowledge about tests for genes associated with Lynch syndrome.

## Conclusions

The results of this survey indicate that physicians in Italy are not yet ready to play a definitive, appropriate role in the context of cancer genetic predictive testing. Many survey items related to education had a positive influence on knowledge, attitudes and, to a lesser extent, professional use. The professional use of cancer predictive genetic tests in Italy might be not completely appropriate, and physicians reported a high level of interest in receiving additional specific training in the field. Overall, this study clearly indicates that priority must be given to targeted educational programs (Mazzucco et al., 2012). However, lessons drawn from many other areas of medicine indicate that education alone may not translate into the effective and appropriate adoption of innovative practices (Greco and Eisenberg, 1993; Grol and Grimshaw, 2003). A specific policy regarding public health genomics needs to be developed at the national level, which is currently being undertaken in Italy by the Ministry of Health (Simone et al., 2013). Additional research is needed to characterize further the contextual factors that influence the incorporation of cancer predictive genetic testing into clinical practice, and the organizational changes needed within the health care system to provide these services both effectively and efficiently.

## Conflict of interest statement

The authors declare that there are no conflicts of interest.

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## References

- Acton, R.T., Burst, N.M., Casebeer, L., et al., 2000. Knowledge, attitudes, and behaviors of Alabama's primary care physicians regarding cancer genetics. *Acad. Med.* 75, 850–852.

- Anon, 2011. Physicians as guardians of genetic knowledge. *Lancet* 377, 967.
- Batra, S., Valdimarsdottir, H., McGovern, M., Itzkowitz, S., Brown, K., 2002. Awareness of genetic testing for colorectal cancer predisposition among specialists in gastroenterology. *Am. J. Gastroenterol.* 97, 729–733.
- Becker, F., van El, C.G., Ibarreta, D., et al., 2011. Genetic testing and common disorders in a public health framework: how to assess relevance and possibilities. Background Document to the ESHG recommendations on genetic testing and common disorders. *Eur. J. Hum. Genet.* 19 (Suppl. 1), S6–S44.
- Bellcross, C.A., Kolor, K., Goddard, K.A., Coates, R.J., Reyes, M., Khoury, M.J., 2011. Awareness and utilization of BRCA1/2 testing among U.S. primary care physicians. *Am. J. Prev. Med.* 40, 61–66.
- Bethea, J., Qureshi, N., Drury, N., Guilbert, P., 2008. The impact of genetic outreach education and support to primary care on practitioner's confidence and competence in dealing with familial cancers. *Community Genet.* 11, 289–294.
- Burke, W., Culver, J., Pinsky, L., et al., 2009. Genetic assessment of breast cancer risk in primary care practice. *Am. J. Med. Genet., Part A* 149A (3), 349–356.
- Carroll, J.C., Cappelli, M., Miller, F., et al., 2008. Genetic services for hereditary breast/ovarian and colorectal cancers – physicians' awareness, use and satisfaction. *Community Genet.* 11, 43–51.
- Carroll, J.C., Rideout, A.L., Wilson, B.J., et al., 2009. Genetic education for primary care providers: improving attitudes, knowledge, and confidence. *Can. Fam. Physician* 55, e92–e99.
- Col, N.F., 2003. The use of gene tests to detect hereditary predisposition to chronic disease: is cost-effectiveness analysis relevant? *Med. Decis. Making* 23, 441–448.
- Collins, F.S., 1997. Preparing health professionals for the genetic revolution. *JAMA* 278, 1285–1286.
- De Vito, C., Nobile, C.G., Furnari, G., et al., 2009a. Physicians' knowledge, attitudes and professional use of RCTs and meta-analyses: a cross-sectional survey. *Eur. J. Public Health* 19, 297–302.
- De Vito, C., Carmelo Nobile, G., Furnari, G., et al., 2009b. The role of education in improving physicians' professional use of economic evaluations of health interventions: some evidence from a cross-sectional survey in Italy. *Eval. Health Prof.* 32, 249–263.
- Drury, N., Bethea, J., Guilbert, P., Qureshi, N., 2007. Genetics support to primary care practitioners – a demonstration project. *J. Genet. Couns.* 16, 583–591.
- EASAC, FEAM, 2012. Direct-to-consumer genetic testing for health-related purposes in the European Union: the view from EASAC and FEAM. EASAC policy report 18. The Clyvedon Press Ltd., Cardiff (UK).
- ENPAM, 2012. *Annuario Statistico 2010. Statistical yearbook 2010.* ENPAM Foundation, Rome (Available at: [www.enpam.it/wp-content/uploads/Annuario/P\\_annuariostatistico\\_2010\\_LUGLIO.pdf](http://www.enpam.it/wp-content/uploads/Annuario/P_annuariostatistico_2010_LUGLIO.pdf). Last access: January 10, 2013).
- Escher, M., Sappino, A.P., 2000. Primary care physicians' knowledge and attitudes towards genetic testing for breast-ovarian cancer predisposition. *Ann. Oncol.* 11, 1131–1135.
- Feero, W.G., Green, E.D., 2011. Genomics education for health care professionals in the 21st century. *JAMA* 306, 989–990.
- Freedman, A.N., Wideroff, L., Olson, L., et al., 2003. US physicians' attitudes toward genetic testing for cancer susceptibility. *Am. J. Med. Genet., Part A* 1 (20A), 63–71.
- Geller, G., Holtzman, N.A., 1995. A qualitative assessment of primary care physicians' perceptions about the ethical and social implications of offering genetic testing. *Qual. Health Res.* 5, 97–116.
- Greco, P.J., Eisenberg, J.M., 1993. Changing physicians' practice. *N. Engl. J. Med.* 329, 1271–1273.
- Grol, R., Grimshaw, J., 2003. From best evidence to best practice: effective implementation of change in patients' care. *Lancet* 362, 1225–1230.
- Hosmer, D.W., Lemeshow, S., 2000. *Applied Logistic Regression*, 2nd ed. Wiley & Sons, New York.
- Klitzman, R., Chung, W., Marder, K., et al., 2012. Attitudes and practices among internists concerning genetic testing. *J. Genet. Couns.* <http://dx.doi.org/10.1007/s10897-012-9504-z>.
- Mazzucco, W., Boccia, S., Ricciardi, W., 2012. Addressing the gap between genetics knowledge and clinical practice: a pilot study to implement genetics education among physicians in Italy. *Ital. J. Public Health* 16 (e8673), 1–3.
- Mehnert, A., Bergelt, C., Koch, U., 2003. Knowledge and attitudes of gynecologists regarding genetic counseling for hereditary breast and ovarian cancer. *Patient Educ. Couns.* 49, 183–188.
- Nippert, I., Harris, H.J., Julian-Reynier, C., et al., 2011. Confidence of primary care physicians in their ability to carry out basic medical genetic tasks—a European survey in five countries—Part 1. *J. Community Genet.* 2, 1–11.
- Pichert, G., Dietrich, D., Moosmann, P., Zwahlen, M., Stahel, R.A., Sappino, A.P., 2003. Swiss primary care physicians' knowledge, attitudes and perception towards genetic testing for hereditary breast cancer. *Fam. Cancer* 2, 153–158.
- Sabatino, S.A., McCarthy, E.P., Phillips, R.S., Burns, R.B., 2007. Breast cancer risk assessment and management in primary care: provider attitudes, practices and barriers. *Cancer Detect. Prev.* 31, 375–383.
- Shields, A.E., Burke, W., Levy, D.E., 2008. Differential use of available genetic tests among primary care physicians in the United States: results of a national survey. *Genet. Med.* 10, 404–414.
- Sifri, R., Myers, R., Hyslop, T., et al., 2003. Use of cancer susceptibility testing among primary care physicians. *Clin. Genet.* 64, 355–360.
- Simone, B., Mazzucco, W., Gualano, M.R., et al., 2013. The policy of public health genomics in Italy. *Health Policy* 110, 214–219.
- Toiviainen, H., Hemminki, E., 2001. Finnish physicians' interest in genetic screening. *Community Genet.* 4, 27–35.
- Trivers, K.F., Baldwin, L.M., Miller, J.W., et al., 2011. Reported referral for genetic counseling or BRCA 1/2 testing among United States physicians: a vignette-based study. *Cancer* 117, 5334–5343.
- Van Riel, E., Wärläm-Rodenhuis, C.C., Verhoef, S., Rutgers, E.J., Ausems, M.G., 2010. BRCA testing of breast cancer patients: medical specialists' referral patterns, knowledge and attitudes to genetic testing. *Eur. J. Cancer Care (Engl.)* 19, 369–376.
- Welkenhuysen, M., Evers-Kiebooms, G., 2002. General practitioners and predictive genetic testing for late-onset diseases in Flanders: what are their opinions and do they want to be involved? *Community Genet.* 5, 128–137.
- Wertz, D.C., 1993. Providers' gender and moral reasoning: a proposed agenda for research on providers and patients. *Fetal Diagn. Ther.* 8 (Suppl. 1), 81–89.
- White, D.B., Bonham, V.L., Jenkins, J., Stevens, N., McBride, C.M., 2008. Too many referrals of low-risk women for BRCA1/2 genetic services by family physicians. *Cancer Epidemiol. Biomarkers Prev.* 17, 2980–2986.
- Wideroff, L., Freedman, A.N., Olson, L., et al., 2003. Physician use of genetic testing for cancer susceptibility: results of a national survey. *Cancer Epidemiol. Biomarkers Prev.* 12, 295–303.
- Wideroff, L., Vadaparampil, S.T., Greene, M.H., Taplin, S., Olson, L., Freedman, A.N., 2005. Hereditary breast/ovarian and colorectal cancer genetics knowledge in a national sample of US physicians. *J. Med. Genet.* 42, 749–755.
- Wilkins-Haug, L., Hill, L.D., Power, M.L., Holzman, G.B., Schulkin, J., 2000. Gynecologists' training, knowledge, and experiences in genetics: a survey. *Obstet. Gynecol.* 95, 421–424.