Medical Student's Knowledge and Attitude Towards Direct-To-Consumer Genetic Tests

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ABSTRACT

Aims: This study reports on the attitudes of 179 Italian Medical Students to direct-to-consumer genetic test and to participation in research practices.
Methods: Data were collected using a self-completion online questionnaire sent to 380 medical students at the faculty of Medicine of the Università Cattolica del Sacro Cuore in Rome, Italy. Questions pertained issues related to awareness and attitudes towards genetic testing, reactions to hypothetical results, and views about contributing to scientific research
Results: The response rate was 47.1%. Less than 50% of students were aware of DTC genetic test. 74% of the sample were interested in undergoing DTC genetic test and the main reason was being aware of genetic predisposition to diseases. Among those who were not willing to undergo a genetic test, the main reason was the lack of confidence in the results. In the hypothetical situations of an increased disease risk after undergoing DTC genetic testing, respondents would take actions to reduce that risk, while in the opposite scenario they would feel unaffected because of the probabilistic nature of the test.
Conclusions: We reported a good level of awareness about DTC genetic test and a high interest in undergoing DTC genetic test in our sample. Nevertheless, opinions and reactions are strongly dependent by the hypothetical good or bad result that the test could provide and by the context whereby a genetic test could be performed. Respondents seem to be exposed to the risk of psychological harms and a strong regulation regarding their use is required.

INTRODUCTION

The rapid evolution in the field of genetics has contributed to the development and implementation of genetic tests in the last 15 years. While diagnostic and presymptomatic tests have been prescribed so far by health professionals, since the 80s predictive genetic test have assumed a significant role not only in the scientific debate but also in the public opinion. One of the reasons of this clamor is that predictive genetic tests are increasingly offered in a so-called Direct To Consumer (DTC) way, which means without the intermediation of a health professional. Predictive genetic tests are designed to identify genotypes that might be associated with an

increased risk of complex diseases, such as cancer, diabetes, cardiovascular disease and others. The results are formulated in terms of disease risk probability, and provide also information on ancestry and individual response to some therapeutic drugs [1].

A strong debate has recently characterised the commercialisation of DTC genetic tests. Proponents of DTC genetic tests strongly affirm the right of individuals to freely access their genetic information and underline the value of test results in order to modify individual's lifestyle, perform further diagnostic investigations or therapeutic decisions and, more generally, to improve individuals' health [2]. Moreover, they affirm that in some selected cases the use of these test has already demonstrated the potential to provide valuable information to citizens, e.g. for breast and ovarian cancer associated with BRCA 1 and 2 gene mutations [3]. On the other hand, those opposing DTC genetic tests have raised questions about the analytical validity, the clinical validity and the clinical utility of these tests, performed outside of an evidence based clinical pathway. In fact, for most complex diseases, the potential use and contribution to health information of these tests is still unclear, and there is absolutely no scientific evidences in favor of their use [4] [5]. In addition to this, opponents argued about the ethical problems and the risk of psychological harm to which DTC test users are exposed [6].

Several surveys have been conducted in order to assess attitudes towards DTC genetic testing among physicians [7] [8] [9] [10] [11] and citizens [12] [13] [14], showing ambiguous results. Only two surveys assessing medical students attitudes on DTC genetic testing were conducted so far [15] [16]. In order to decrease this gap, we have implemented this survey aimed to assess knowledge and attitudes of medical students in Italy toward the use of DTC genetic tests.

METHODS

A self-administered anonymous questionnaire was distributed in 2014 to the students enrolled in the Faculty of Medicine and Surgery at the Università Cattolica del Sacro Cuore in Rome, Italy. A similar questionnaire was previously validated and used for a survey on a sample including medical and not medical Swiss university students [15]. The questionnaire had 29 questions in five different sections aiming to explore: demographic characteristics and personal opinions (section n 1); awareness and experience of DTC genetic test (section n 2); interest in undergoing or not DTC genetic test and/or in being enrolled in a genetic study conducted in a clinical setting, the willingness to make the data available for research and the willingness to know the results of the test (section n 3); reactions to hypothetical situations of an increased/decreased risk of disease after being subjected to a DTC genetic test (section n 4); which institutions they would support participating in a genetic study via DTC genetic test and opinion on DTC genetic test after filling out the questionnaire (section n 5). Personal opinions on religion, concern about own health and views about the influence that genetic and environment have on own health were investigated through three questions, each using a seven-point Likert scale ranging from 1 (not at all) to 7 (very much) (section n 1). Experience, awareness and attitudes towards DTC genetic tests, as well as the willingness to make the data available for research and the willingness to know the test results (section n 2) were presented as dichotomous yes-no questions. Multiple choice questions were used to study the motivations for undergoing or not DTC genetic test and the motivations for participating or not in a genetic study conducted in a clinical setting (section n 3), the reactions to the hypothetical situations of increased/decreased risk of some diseases examples and institutions the respondents would like to support by participating in a genetic study both including DTC or in the clinical setting (section n 4).

Statistical Analysis

A descriptive analysis was conducted to report demographic characteristics and personal opinions of respondents. For the multiple choice questions, the answers were ranked by the number of times they were selected. To conduct a graphical evaluation of differences between motivations for not undergoing in a genetic study through DTC genetic test or through a genetic study conducted in a clinical setting, we created a radar chart bearing the dimension scores. Data has been normalised in order to make them comparable.

In order to establish associations between categorical variables, Pearson's χ^2 test was used. Statistical significance was set at a two-sided P value of <0.05. Data were analysed using STATA 13 software.

RESULTS

380 students were invited to fill in the questionnaire, of which 179 answered, (response rate 47.1%). The median age was 21 years; 59.8% of respondents were female (Table 1). There was a higher prevalence of first-year students (45.3%). Overall 45.3% of respondents were already aware of DTC genetic tests. None of the respondents has ever performed a DTC genetic test, however 73% were interested in undergoing DTC genetic tests. Among the latter, 88% would want to be informed of the results and 92% would want to make data available for research (data not shown). Around

TABLE 1. Demographic of respondents, knowledge and attitudes on DTC genetic tests.

DEMOGRAPHIC OF THE RESPONDENTS (N=179)	n[a]	%[a]
Median age, years	21	-
Gender, n*		
Females	107	59.8
Males	71	39.7
ACADEMIC YEAR		
I	81	45.3
II	32	17.9
III	32	17.9
IV	21	11.7
V	12	6.7
KNOWLEDGE ABOUT DTC GENETIC TESTS		
Already aware of the test	81	45.3
Personal experience on DTC genetic testing	0	0
ATTITUDES		
Interest in undergoing DTC genetic test	132	73.7
Interest in participating in a genetic study in a clinic	145	81.5

[a] *Sum of column did not add up to total study subjects because of missing data*

81% were interested in participating in a genetic study conducted in a clinical setting. Table 2 reports the respondents' reasons for undergoing a DTC genetic test and participate in a genetic study conducted in a clinic. The main reason for undergoing a DTC genetic test was the willingness to know if they were at risk of certain diseases (86.4%), followed by the willingness to know the risk of passing on to the children a predisposition to a disease (60.6%). Among those who were not interested in undergoing DTC genetic test, the most selected reason was the lack of confidence in the result (48.9%), followed by the concern that the result could cause worry (42.6%). Among those who were interested in participating in a genetic study conducted in a clinical setting, the main reason was the chance to contribute to the development of therapies for diseases (86.2%), followed by the interest in the knowledge of the individual genetic profile (50.3%). Among those who would not participate in this kind of genetic study, the most selected reason was the concern about their privacy (42.4%), followed by the concern that the result could cause worry (33.3%). Figure 1 shows a comparison between the reasons provided for not undergoing DTC genetic test vs. not participating in a genetic study conducted in a clinical setting. The concern about the results was an important reason for not undergoing a genetic test in both contexts. DTC genetic tests lead to more doubts about the quality of the test, while a genetic study conducted in a clinical setting leads to more concerns about the privacy of the data.

Table 3 reports on the possible reactions to different hypothetical situations of results of genetic test showing increased or decreased risk of two complex diseases examples respectively colon cancer and obesity. In the scenario of increased colon cancer risk, the most common reaction selected by the students was taking measures to reduce the risk (76.5%), followed by the option to consult a medical doctor (59.8%). 40% of respondents would take note of the result but without being affected, given the merely probabilistic nature of the test. A more consistent result was obtained in the hypothetical scenario of decreased colon cancer risk. In this particular situation 60.3% of respondents would take note of the result but without being affected, given the merely probabilistic nature of the test, followed by being reassured about own health (39.1%) and by the willingness to consult a medical doctor (35.2%). Similar results were obtained in the hypothetical scenario of DTC test performed to investigate the risk of developing obesity. In the case of an increased risk of obesity, the most selected option was to take measures to reduce the risk (75.4%), followed by the willingness to consult a medical doctor (38.6%). In the case of decreased obesity risk, 53.6% of respondents would take note of the result but without being affected, given the merely probabilistic nature of the test, followed by being reassured about own health (30.2%). Concerning the institutions that students would support by participating in a genetic study via DTC genetic test, first ranked were public institutions (72.1%), followed by not-for-profit institutions (49.7%). Only 2.2% of respondents would support for-profit institutions. One third of respondents have changed their opinion on DTC genetic test after completing the questionnaire (data not shown).

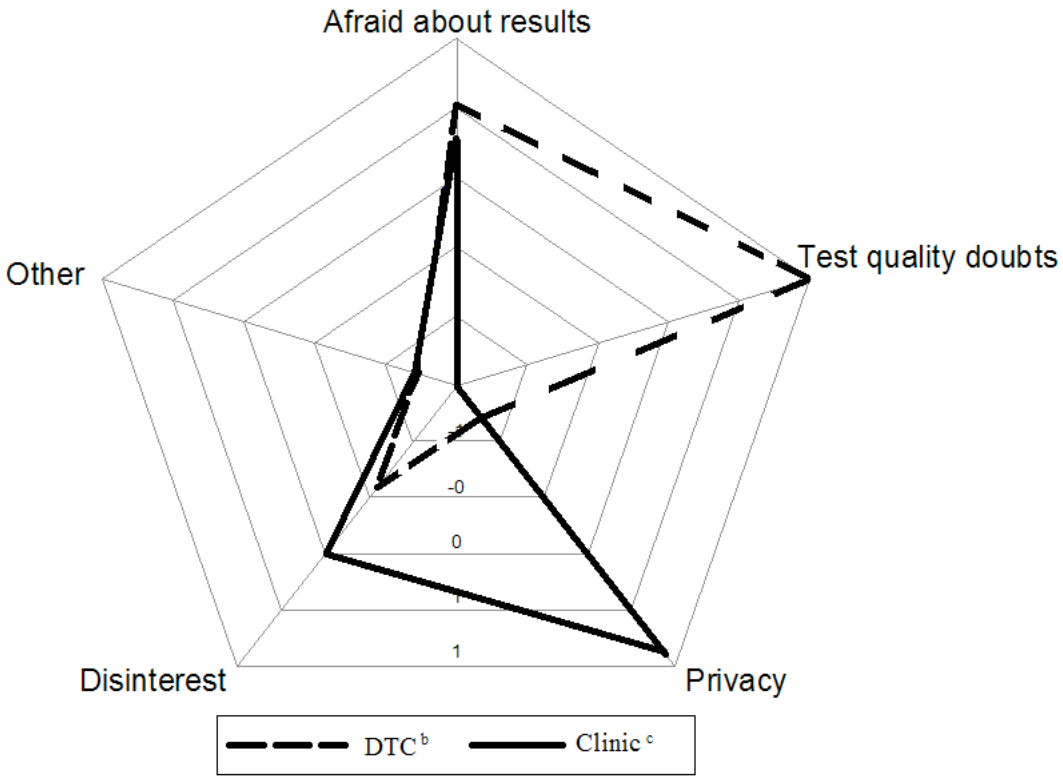
Table 4 reports on the consistent relationship between demographic characteristics, attitudes and opinions on DTC genetic tests, and interest among the participants in participating in a genetic study conducted in a clinical setting. Older students of the sample (>21 years) showed to be more likely to have heard of DTC genetic test before the survey compared to the youngest ($p = 0.009$). Females were more likely to participate in a genetic study conducted in a clinic than males ($p = 0.011$). A consistent association was also found between the agreement to make results of DTC genetic tests available for research and interest in participating in genetic researches conducted in a clinical setting ($p = 0.003$). Lastly older students, as well as those who were already aware of DTC genetic test before the questionnaire, were less likely to change their opinion after completing the questionnaire ($p = 0.01$).

DISCUSSION

Our study shows a high interest and a fair level of awareness on DTC genetic tests in a sample of medical students in Italy. Our survey shows that opinions and reactions on DTC genetic tests are heavily dependent by the hypothetical good or bad result that the test could provide, and by the context whereby a genetic test can be performed.

Similar studies have shown a fair level of interest in undergoing DTC genetic test [15] [16] [17] [18]. The opportunity to research participation was the key reason for undergoing DTC genetic test among different populations [15] [19], while concerns on the privacy of personal data [20], doubts on validity of the results, and the fear that the result may be of concern [21] [22] [23] were the main reasons for not undergoing DTC testing among several studies conducted in US, Canada and Australia. As few persons have had experience with DTC genetic tests, previous studies used to create hypothetical situations of a genetic results after being tested [15] [17] [23] [24]: in the case of high risk the main reaction of the respondents was to take actions to reduce such risk, while in the case of low risk main reaction was to feel unaffected due to the probabilistic nature of the test. Other surveys already reported a fair level of interest in undergoing DTC genetic test, nevertheless none of these were conducted among a sample consisting of only medical students, and this may be the reason why we reported a higher level of interest. There might be a trend between people interested in undergoing DTC genetic test and those who most care about their own health. Consistent with this finding, the key reason for undergoing DTC genetic tests was found to be the opportunity to know the genetic predisposition to disease. It is noteworthy to note that research participation, a motivation that was studied and found as favorable for undergoing DTC genetic test [19], was not highly ranked in our study population. Vayena et al. previously reported that "…students engaging with scientific research (e.g. lab, clinic) might be more aware of the issues around research participation and may feel more inclined to see themselves contributing to such research" [15]. Our population has placed the interest to know any personal predisposition to a disease, rather than the opportunity to participate in scientific research, as the main motivation for undergoing DTC genetic testing. Conversely, among those who were not interested in undergoing DTC genetic test, the main reasons of this lack of interest were doubts about their clinical validity followed by the fear that the results may be of concern. This result is consistent with the literature data

FIGURE 1. Comparison of reasons for not undergoing DTC genetic test vs. not participating in a genetic study conducted in a clinic



[b] *DTC genetic testing*
[c] *Study conducted in a clinic*

explaining why people would refrain from testing: firstly their real clinical value, given the lack of a professional supervision in the context of an evidence based clinical path; secondly the possible risk for DTC tests' users to be unjustifiably exposed at psychological harm and distress given their merely probabilistic nature and, generally, their low predictive value [21] [22] [23]. This is a big issue, mainly because it is still possible that test-takers who do not think they will be concerned about the results, will become when they actually receive them.

Privacy concern, another issue that was studied and found as daunting for genetic testing and genetic participation in other studies [20], was ranked low in our study population. This can be explained by several factors. First, our sample consisted of very young people (median age 21 years). Previous studies [25] have shown that young people tend to have a lack of knowledge about privacy, which may thus justify why in our study they confirmed to be less concerned about it. Another explanation may be caused by the fact that

TABLE 2. Respondents' ranked reasons for undergoing or not DTC genetic test and for participanting in a genetic study in a clinic.

	RESPONSE	
REASONS FOR UNDERGOING DTC GENETIC TESTING (n=132)	**n**	**%**
I would like to know if I am at risk of certain diseases	114	86.4
I would like to know the risk of my passing on to my children a predisposition to disease	80	60.6
I would like to know my personal characteristics	69	52.3
I would like to know my sensitivity to certain medication	65	49.2
I would like to contribute my genetic data to scientific research	63	47.7
I would like to find out about my genetic traits	50	37.9
I am interested in genetics in general	21	15.9
Only if I can access the test free of charge or at a significant discount	21	15.9
To find out how these tests are performed	8	6.1
Just for fun	5	3.8
Other	1	0.8
REASONS FOR REFRAINING FROM DTC GENETIC TESTING (n=47)	**n**	**%**
I do not think the test results are valid	23	48.9
I am concerned that the results will worry me	20	42.6
I do not see any utility in such tests/I am not interested in my genetic profile	13	27.7
I am concerned about the privacy of my data	9	19.1
Cost is an obstacle to undergo testing	7	14.9
Other	2	4.3
I am skeptical about genetic testing in general	0	0.0
REASONS FOR PARTICIPATING IN A GENETIC STUDY IN A CLINIC (n=145)	**n[a]**	**%***
To contribute to the development of therapies for diseases, but neither I nor my dear could be affected	125	86.2
To know my genetic profile	73	50.3
To allow the development of therapies for diseases that could affect me or my dear	67	46.2
In order to contribute to a greater informative value of genetic testing	48	33.1
I am interested in genetics and research in general	38	26.2
REASONS FOR NOT PARTICIPATING IN A GENETIC STUDY IN A CLINIC (n=33)	**n[a]**	**%***
I am concerned about the privacy of my data	14	42.4
I am concerned that the results will worry me	11	33.3
Time and organisation would be an obstacle to participation	10	30.3
I am skeptical about genetic testing	5	15.2
I am skeptical about genetics and genomics	4	12.1
Other	2	6.1
I am not interested in genetic and genomic in general	0	0.0

[a] *Total is >100% as respondents could chose answers from multiple-choice questions.*

TABLE 3. Reactions on the interviewed students to hypothetical high/low genetic risk.

RESPONSE, N (%)	↑ COLON CANCER	↓ COLON CANCER	↑ OBESITY	↓ OBESITY
I would consult a medical doctor	107 (59.8)	63 (35.2)	69 (38.6)	47 (26.3)
I would discuss results with family/friends	7 (3.9)	4 (2.2)	6 (3.4)	5 (2.8)
I would ignore the result	0	12 (6.7)	10 (5.6)	22 (12.3)
I would take note of the results but they would not affect me due to their merely probabilistic nature	73 (40.8)	108 (60.3)	52 (29.1)	96 (53.6)
I would worry about my health	31 (17.3)	-	19 (10.6)	-
I would be reassured about my health	-	70 (39.1)	-	54 (30.2)
I would take measures to reduce the risk	137 (76.5)	-	135 (75.4)	-
I would become lax about taking care of my health	-	6 (3.4)	-	22 (12.3)
Total[a], n	355	263	291	246

[a] *Total is >100% as respondents could chose answers from multiple-choice questions.*

TABLE 4. Significant relationship between demographic characteristics, attitudes and opinions on DTC genetic tests, interest in participating in a genetic study conducted in a clinical setting.

			N TOT	P-VALUE
KNOWLEDGE OF DTC GENETIC TEST BEFORE THE QUESTIONNAIRE				
	NO	**YES**		
Age				
≤ 21	50 (65.8%)	26 (34.2%)		
> 21	47 (46.1%)	55 (53.9%)	178	0.009
INTEREST IN PARTICIPATING IN A GENETIC STUDY IN A CLINIC				
	NO	**YES**		
Gender				
Female	13 (12.1%)	94 (87.8%)		
Male	19 (27.1%)	51 (72.8%)	177	0.011
INTEREST IN PARTICIPATING IN A GENETIC STUDY IN A CLINIC				
	NO	**YES**		
Make data available for research				
No	4 (40%)	6 (60%)		
Yes	11 (9.01%)	111 (90.9%)	132	0.003
OPINION CHANGED AFTER THE QUESTIONNAIRE				
	NO	**YES**		
Age				
≤ 21	42 (56%)	33 (44%)		
> 21	76 (74.5%)	26 (25.5%)	177	0.01
OPINION CHANGED AFTER THE QUESTIONNAIRE				
	NO	**YES**		
Knowledge of DTC genetic test before the questionnaire				
No	56 (58.3%)	40 (41.7%)		
Yes	62 (76.5%)	19 (23.5%)	177	0.01

the genetic information could be perceived by medical students as having a low predictive power especially if the interaction with the environment is not considered. It's not unlikely that privacy concerns will increase along with the amelioration of predictive value of DTC genetic test. This could be the reason why students are concerned more about the privacy of the data in a genetic study conducted in the clinic rather than via DTC genetic test (Figure 1). Consistent with the result of another study [15], participants have in fact considered a genetic study conducted in the clinic as more predictive than the DTC genetic test.

The hypothetical situation of increased colon cancer risk showed active reactions from respondents, including involving a medical doctor, while increased obesity risk didn't show to result in such active response among participants. This should be easily explained, as another study showed [15], by the perception of colon cancer risk with respect to obesity as more dangerous and life threatening condition. A different result was obtained in the scenario of a decreased risk. In this case reactions were more similar for the two hypothetical conditions: main reaction was to take note of the results but not feel affected by them due to their merely probabilistic nature. It is also important to note that conversely, in the case of higher risk regarding both conditions, respondents would take actions to reduce risk. One explanation may be that bad news tends to be more believed than good news [26] and are therefore more alarming.

The first limitation in our study is the small sample size. Furthermore the survey includes questions on hypothetical scenarios, responses to which may differ from actual decision and experience [25].

Despite those limitations, final consideration is the good level of awareness and the high interest toward the world of DTC genetic test among medical students enrolled in our study. This is of particular concern, as DTC genetic tests have become very popular in recent years and doctors often don't feel adequately prepared to face this phenomenon [27] [28] [29] [30] [31] [32] [33] [34]. Interest in DTC tests may be the first step to spread knowledge among the doctors of the future years and to make them aware of the opportunities and the risks related to their use. This is of outstanding importance both for the single user and for the community given that if medical doctors and public health professionals will not find the way to lead and govern the use of genetic test, other figures will do it for commercial purposes and outside the context of a good clinical practice [35].

Competing interests

The authors declare that they have no competing interests.

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