European Journal of Public Health, 1–4

https://doi.org/10.1093/eurpub/ckad189

Public attitudes, knowledge and educational needs toward genetic testing and omics sciences: a pilot survey conducted in Italy

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Advancements in genomics have significant implications for public health, making citizens' education vital for informed decision-making. Based on two literature reviews' findings and a survey conducted with experts from the Italian Network of Genomics in Public Health, we conducted a pilot survey on Italian citizens' attitudes, knowledge and educational needs toward genetic testing and omics sciences. Our results demonstrate a widespread interest in genetic testing and uncertainties regarding associated risks, with 99% of participants acknowledging insufficient knowledge of genetic testing. There is an urgent need for educational tools to improve citizens' literacy and engagement in this rapidly evolving field.

Introduction

The omics revolution encompassed diverse scientific disciplines, making omics sciences such as genomics, transcriptomics, proteomics, metabolomics, microbiomics and nutribiomics, increasingly relevant and resulting in a surge in knowledge and availability of accessible technologies like next-generation sequencing. These were pivotal in developing novel diagnostic tests, discovering new biomarkers and drugs, and advancing personalized medicine.[1]

Public health genomics' implementation in European healthcare systems is limited despite some countries adopting targeted health policies.[2] Italy is a frontrunner, incorporating precision health as a dedicated pillar in National Prevention Plans since 2010, publishing the first-ever Guidelines on Genomics in Public Health in 2013,[3] and being among the first 13 countries to sign the Declaration of Cooperation 'Towards access to at least 1 million sequenced genomes in the European Union by 2022'.[4] In 2017, the Italian National Innovation Plan of the Healthcare System[5] recognized the need for educational initiatives targeting professionals, citizens, and decision-makers to integrate omics sciences into healthcare. The Italian Ministry of Health funded several projects, including a collaborative survey with a civic organization. This pilot survey is the first in Italy evaluating citizens' attitudes, knowledge and educational requirements in the omics sciences field; the findings are presented in this paper.

Methods

Based on the findings from two literature reviews[1,6] and a survey conducted with experts from the Italian Network of Genomics in Public Health (GENISAP),[7] we designed a questionnaire evaluating existing knowledge and awareness among Italian citizens regarding genetic testing and omics sciences.

The survey consisted of 57 questions divided into three sections: 'Personal knowledge of omics sciences', 'Attitudes' and 'Educational needs'. The questionnaire and a brief description of the project were disseminated on the channels of CittadinanzAttiva, a citizen activism organization, from 29 October 2020 to 15 April 2021. A total of 359 participants were recruited, and all completed the survey.

Determinants of personal knowledge and attitudes were assessed using two multivariable logistic regression models. The variables 'knowledge' and 'attitudes' were categorized into two levels, with respondents classified as having knowledge or attitudes if they provided appropriate responses to at least 50% of the questions (e.g. 'reporting knowledge of personalized medicine', 'being informed about genetic testing'). Those who could not provide an answer or judgment were classified as having 'no knowledge' or 'no positive attitude'. The covariates included age, gender, personal or family history of a genetic disease, education level and genetics education at school and/or university. Multivariable logistic regression models were built using the strategy suggested by Hosmer and Lemeshow.[8] The univariable analysis examined each variable and included it in the multivariable logistic model when P values < 0.15. Independent variables' influence on each investigated binary outcome was expressed as adjusted odds ratios (OR) and 95% confidence interval (CI).

Statistical significance was set at P values < 0.05. The statistical analysis was performed using the STATA 16.0 software. The Ethics Committee of Fondazione Policlinico Gemelli approved this survey under ID 3504.

Results

Demographic and general information

All 359 citizens completed the survey. Results are reported in Table 1. The participants' mean age was 46 (SD = 16), and 65.2% were female. Most held a university degree (42.9%) or higher education level (22.3%). Regarding personal or family history, most respondents reported no

Table 1 Survey questions and public responses on knowledge, awareness and educational needs on genetic testing and omics sciences

Demographic and general information ($N = 359$)		
	n	**%**
Gender		
Female	234	65.2
Male	125	34.8
Age (years)	Mean = 46	SD = 16
Educational degree		
Compulsory school or lower level	125	34.8
University degree	154	42.9
Postgraduate training	80	22.3
Have you ever formally studied genetics at school?[a]	123	34.3
Have you ever formally studied genetics at university?[a]	61	17.0
Do you have relatives (or friends) who are carriers of a genetic disease?[a]	88	24.5
Are you a carrier of a genetic disease?[a]	9	2.5
Are you suffering from a chronic disease?[a]	51	14.2
Knowledge ($N = 359$)		
	n	**%**
Do you know what chronic diseases, also called multifactorial ones, are?[a]	244	68.0
Are you informed about the possibilities of using genetic testing to prevent disease?[a]	234	65.2
Are you informed about the possibilities of using genetic testing to treat a disease?[a]	253	70.5
Are you informed about the possibility of being diagnosed with a DNA test?[a]	262	73.0
Indicate which of the following genetic testing applications you have heard of[a]		
Determination of the risk or likelihood of contracting a particular disease	262	73.0
Determination of the treatment of the disease after diagnosis	191	53.2
Determination of the efficacy of drugs in an individual	198	55.2
Determination of the probability of transmitting on a hereditary disease to children	306	85.2
Have you ever heard of gene therapy?[a]	221	61.6
Have you ever heard of personalized medicine and/or precision medicine?[a]	200	55.7
Have you ever heard of omics sciences?[a]	95	26.5
Have you ever heard of direct-to-consumer genetic testing (DTC-GT)?[a]	120	33.4
Attitudes ($N = 359$)		
	n	**%**
Where there are effective tools to prevent certain diseases in a targeted way, would you like to know your risk of contracting them?		
Yes	286	79.7
No	17	4.7
I don't know	56	15.6
Would you undergo a DTC genetic test?		
No	37	10.3
I don't know what they are	144	40.1
Yes	147	41.0
Yes but only if free	31	8.6
How much do you agree with the statement 'doctors should be involved in prescribing tests and explaining the results of genetic tests'?		
Agree	302	84.1
Disagree	3	0.8
Neither agree nor disagree	54	15.1
Would your lifestyle change according to the results of a genetic test?[a]	318	88.6
In which cases would you like to know the results of a genetic/omic test you have undergone?		
Never	4	1.1
Always	273	76.0
Only in case of treatable illnesses	22	6.1
Only in case of serious illnesses	21	5.9
Only in case of serious illnesses but treatable at the same time	39	10.9
If you are undergoing a genetic/omic test, would you like to know the risk of disease for your family members and the risk of having passed on the predisposition for a disease to your children?[a]	342	95.3
Would you share genetic test results for research purposes?		
Yes	308	85.8
No	10	2.8
I don't know	41	11.4
Would you participate in genomic research studies?[a]	267	74.4
Do you think the privacy of individuals participating in genomic studies is adequately protected?		
Yes	102	28.4
No	25	7.0
I don't know	232	64.6
Do you agree with the statement 'all women planning a pregnancy must be tested for genetic risk'?		
Agree	188	52.4
Disagree	43	12.0
Neither agree nor disagree	128	35.6
Educational needs ($N = 359$)		
	n	**%**
Do you think citizens are adequately informed about the omics sciences?[a]	5	1.4
Do you think there is a need for more training/information initiatives on omics sciences aimed at citizens?[a]	343	95.5
Do you think citizens are adequately informed about DTC-GTs?[a]	13	3.6
Do you think there is a need for more training/information initiatives on genetic tests and DTC-GT aimed at citizens?[a]	341	95.0

Predictors of knowledge of the omics sciences

Explanatory variable	**OR (95% CI)**	***P*-value**
Age (years)	0.99 (0.98–1.01)	0.48
Gender, Female	1.00 (0.61–1.65)	0.99
Education degree (ref. Compulsory school or lower level)		
University degree	4.31 (2.46–7.53)	<0.0001
Postgraduate training	1.99 (1.04–3.80)	0.037
Italy: region of origin (ref. South)		
Central	1.63 (0.88–3.03)	0.12
North	1.06 (0.55–2.03)	0.87
Genetics training at school and/or university	1.95 (1.19–3.18)	0.008
	3.67 (2.09–6.46)	<0.0001

(continued)

Table 1 Continued

Predictors of knowledge of the omics sciences		
Explanatory variable	**OR (95% CI)**	***P*-value**
Carrier of a genetic disease and/or with relatives (or friends) who are carriers of a genetic disease		
Carrier of a chronic disease	1.14 (0.58–2.25)	0.71
Predictors of positive attitudes toward the omics sciences		
Explanatory variable	**OR (95% CI)**	***P*-value**
Age (years)	1.02 (1.01–1.04)	0.003
Gender, Female	1.76 (1.08–2.85)	0.023
Education degree (ref. Compulsory school or lower level)		
University degree	2.51 (1.49–4.21)	0.001
Postgraduate training	4.21 (2.19–8.09)	<0.0001
Italy: region of origin (ref. South)		
Central	0.57 (0.31–1.05)	0.071
North	0.78 (0.41–1.47)	0.44
Genetics training at school and/or university	1.58 (0.96–2.60)	0.069
Carrier of a genetic disease and/or with relatives (or friends) who are carriers of a genetic disease	0.98 (0.58–1.67)	0.94
Carrier of a chronic disease	1.11 (0.57–2.15)	0.75

a: total number of respondents who answered "Yes".

genetic disorders (97.5%, 75.5%). Around 34% of participants studied genetics in school, while 17% studied genetics in university.

Knowledge, attitudes and educational needs

More than half of the respondents had heard about personalized medicine (55.7%); most were unaware of omics sciences (73.5%), while 33.4% had heard about direct-to-consumer-genetic-testing (DTC-GT) (see Table 1). Respondents were informed about the use of genetic knowledge to prevent diseases (65.2%), treat diseases (70.5%), and be diagnosed with DNA testing (73.0%). Additionally, 61.6% of respondents had heard about gene therapy.

If effective prevention tools were available, 79.7% of participants expressed interest in knowing their disease risk, and 49.6% would consider undergoing DTC-GT. That physicians should be involved in prescribing genetic tests and explaining the results were agreed by 84.1%.

After undergoing genetic testing, most respondents would always like to be informed of the results (76.0%) or only if pathologic findings are present (22.9%). Also, 95.3% would like to know their family members' disease risk and the risk of passing on disease predisposition to their children. Additionally, 88.6% reported being willing to make lifestyle changes based on the results of a genetic test. Fewer respondents (52.4%) agreed that all women planning a pregnancy should be tested for genetic risk, with many undecided (35.6%). On privacy, 85.8% would share their genetic test results for research purposes, but 64.6% were unsure whether the privacy of individuals participating in genomics studies is adequately protected.

Almost all (98.6%) agreed that citizens are not adequately informed about omics sciences and DTC-GT, emphasizing the need for more training and information initiatives.

Multivariable analysis

Factors associated with knowledge of genetic testing and omics sciences included education level higher than compulsory schooling [university degree—OR 4.31 (2.46–7.53), $P < 0.0001$; postgraduate training—1.99 (1.04–3.80), $P < 0.037$], genetics training in school and/or university [OR 1.95 (1.19–3.18), $P = 0.008$] and being a carrier of a genetic disease and/or having relatives (or friends) who are carriers [OR 3.67 (2.09–6.46), $P < 0.0001$].

A positive attitude toward omics sciences was associated with age [OR 1.02 (1.01–1.04) $P = 0.003$], female gender [OR 1.76 (1.08–2.85), $P = 0.023$] and an education degree higher than compulsory school [university degree—OR 2.51 (1.49–4.21), $P = 0.001$; postgraduate training—OR 4.21 (2.19–8.09), <0.0001]. There was also a positive, borderline non-significant association with receiving genetics training in school and/or university [OR 1.58 (0.96–2.60), $P = 0.069$].

Discussion

Our survey explored the Italian public's knowledge, attitudes and educational needs with regards to genetic testing and omics sciences. Participants have low perceived knowledge, suggesting more educational efforts; they recognize the potential benefits of genetics in disease prevention, diagnosis and treatment.

Participants demonstrated favorable attitudes toward genetic testing and omics sciences. Most wanted to understand their disease risk if effective preventive measures were accessible; slightly less than half indicated a willingness to undergo DTC-GT. Most participants agreed that healthcare professionals should be involved in genetic test recommendation and results interpretation. Nearly all wanted to learn about their family members' disease risks and potential transmission of predispositions to their children. Most were willing to make lifestyle changes following genetic testing results.

Privacy and data protection emerged as significant concerns. Most respondents were willing to share their genetic test results for research purposes, and over half expressed uncertainty about the adequacy of privacy protections for individuals participating in genomics studies.

Almost all respondents concurred that more public awareness regarding omics sciences and DTC-GT is needed, emphasizing training and information initiatives to educate the public.

Respondents strongly agreed on the need for accessible and reliable information. In addition, the highly educated showed more positive attitudes toward omics sciences.

Our survey's results show citizens have a positive attitude toward omics sciences and genetic testing, reporting the need for more educational initiatives; these are in line with the findings of our prior systematic review[1] and international literature.[9,10] However, sample size could impair the possibility of drawing generalized conclusions, and high representation of highly educated participants may impact responses, e.g. on knowledge and awareness. We are conducting a second survey in eight European countries (Italy, the Netherlands, France, Germany, Spain, Hungary, Romania and Poland) for more complete data.

Omics sciences' advancements have far-reaching implications for public health, highlighting the need for a strategic approach to promote and govern this disruptive innovation. Educating citizens becomes essential in enabling them to make informed decisions regarding their health.

Acknowledgements

We thank the Italian Ministry of Health for supporting the activities of the National Center for Disease Prevention and Control (CCM) projects related to genomics and omics sciences.

Funding

This study was funded by the National Center for Disease Prevention and Control (CCM), Italian Ministry of Health (CCM2021, CUP B85F21004970001).

Conflicts of interest: None declared.

Data availability

The data underlying this article will be shared on reasonable request to the corresponding author.

Key points

- This survey addressed to Italian citizens demonstrated a widespread interest in genetic testing and its potential to improve health outcomes, with over half of the respondents being aware of possible uses of genetic testing. However, uncertainties regarding its risks prevail, with over 80% of respondents indicating that doctors should be involved in prescribing tests and explaining the results of genetic tests.
- Respondents (98.4%) indicated they were not adequately informed about omics sciences and 95.5% expressed the need for more training initiatives for citizens. These results are informative because the appropriate and conscious utilization of new omics technologies and services by citizens requires correct information of users, regarding not only genetic testing, but also omics sciences and their possibilities and limitations.
- Based on our findings, we argue that citizens will be more receptive to adopting new behaviors and practices and contribute to the realization of precision health only if educational, socioeconomic and cultural hurdles are correctly addressed.

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