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A critical view of the general public's awareness and physicians' opinion of the trends and potential pitfalls of genetic testing in Greece

Aim: Progress in deciphering the functionality of the human genome sequence in the wake of technological advances in the field of genomic medicine have dramatically reduced the overall costs of genetic analysis, thereby facilitating the incorporation of genetic testing services into mainstream clinical practice. Although Greek genetic testing laboratories offer a variety of different genetic tests, relatively little is known about how either the general public or medical practitioners perceive genetic testing services. **Materials & methods:** We have therefore performed a nationwide survey of the views of 1717 members of the general public, divided into three age groups, from all over Greece, and residing in both large and small cities and villages, in order to acquire a better understanding of how they perceive genetic testing. We also canvassed the opinions of 496 medical practitioners with regard to genetic testing services in a separate survey that addressed similar issues. **Results:** Our subsequent analysis indicated that a large proportion of the general public is aware of the nature of DNA, genetic disorders and the potential benefits of genetic testing, although this proportion declines steadily with age. Furthermore, a large proportion of the interviewed individuals would be willing to undergo genetic testing even if the cost of analysis was not covered by healthcare insurance. However, a relatively small proportion of the general public has actually been advised to undergo genetic testing, either by relatives or physicians. Most physicians believe that the regulatory and legal framework that governs genetic testing services in Greece is rather weak. Interestingly, the vast majority of the general public strongly opposes direct-access genetic testing, and most would prefer referral from a physician than from a pharmacist. **Conclusion:** Overall, our results provide a critical evaluation of the views of the general public with regard to genetics and genetic testing services in Greece and should serve as a model for replication in other populations.

KEYWORDS: education ■ general public awareness ■ genetic testing ■ healthcare ■ insurance cost ■ molecular genetics ■ pharmacogenomics ■ physicians' opinion ■ reimbursement

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Personalized or genomic medicine refers to the exploitation of genomic information in the context of guiding medical decision making. Examination of an individual's genome sequence can, in principle at least, enable physicians to make assessments of disease risk and arrive at decisions regarding treatment regimens. At the same time, a number of health and disease states can now be identified by distinct genotypes and/or gene-expression patterns. Hence, these molecular fingerprints can be exploited to stratify patient populations and to elucidate the pathogenesis of genetic disorders on a genome-wide basis [1,2]. We are entering an age in which individualized healthcare has become a reality by taking each person's unique genomic profile into consideration alongside their clinical profile [3]. Our new found knowledge of the molecular basis of many monogenic and complex disorders can be exploited not only in order to optimize preventive medicine strategies but also to personalize conventional therapeutic

interventions, either at an early stage in the onset of the genetic disorder or presymptomatically, leading to unprecedented opportunities for the customization of patient care [4].

Unfortunately, among the general public, awareness may often be lacking with respect to genetics and its impact on society. Similarly, physicians, who are responsible for delivering these services to the general public, can have a relatively poor perception of certain issues pertaining to genomic medicine and its potential to fine-tune conventional medical interventions to the individual patient's genomic profile. As a result, the landscape of genetic-testing services is still poorly developed in many parts of Europe and the USA despite courageous efforts to harmonize genetic testing services (e.g., EuroGenTest [101] and OrphaNet [102]). Thus, at the dawn of the genomic medicine era, understanding the general public's perception, as well as physicians' opinions, with respect to the potential societal and individual benefits – but also the problems

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and pitfalls – of genetic testing, has become an urgent goal. For this reason, we have initiated nationwide surveys to ascertain patients' and physicians' views of the genetic-testing services currently available in Greece, aiming to understand both the wishes and needs of patients and physicians with regard to the genetic testing industry and identify regulatory deficiencies and gaps in the existing legal provision that could be rectified by appropriate legislation.

We have previously reported our results from a nationwide survey of various private genetic-testing providers in Greece [5]. Here, we report our findings from a survey of the general public's perception and physicians' opinion of various issues pertaining to genetics and its impact on society at large. Our study provides a model that can be replicated in other European countries with the ultimate aim of improving public understanding of genetics and genetic testing, and facilitating the incorporation of genomic medicine into everyday clinical practice.

Materials & methods

■ Research design

A cross-sectional survey design was used for this research study that was conducted between June 2009 and September 2010. We formulated two independent questionnaires (see SUPPLEMENTARY MATERIAL; www.futuremedicine.com/doi/suppl/10.2217/pme.11.48) from which the data on individual perceptions of genetic testing and pharmacogenomics were generated.

In both questionnaires, nonrandom sampling was employed. The first questionnaire was personally given to 1717 participants from the general public, who were selected from four major cities in Greece (Athens, Patras, Thessaloniki and Larissa), two small cities (with fewer than 50,000 inhabitants) and two villages. The participants had different occupations and ranged in age. This questionnaire contained two main sections: the first part requested information such as age, gender and place of residence, whereas the second part contained nine questions regarding various aspects of genetics, such as awareness of and personal opinion about genetics, genetic tests and the use of pharmacogenomic testing from healthcare providers. The second questionnaire was distributed to 496 physicians from all medical specialties who attended the national (Greek) medical conference in May 2010. This approach was taken to ensure a truly broad coverage of physicians from all specialties and from all geographical regions within the country. Again, this questionnaire

contained two sections, the first pertaining to age and gender while the second posed five questions in order to solicit the respondents' opinions on the various potential benefits and pitfalls of genetic testing (see SUPPLEMENTARY MATERIAL). We provided the necessary clarifications to questions posed by the survey respondents when required to do so, particularly in the case of the general public, in order to ensure that a valid response was given to each question.

■ Measures

The surveys provided the prospective data for this study. The dependent variables were derived from the questions in both surveys, scored using a binary model (0 = no, 1 = yes), whilst the independent variables comprised the demographic characteristics of respondents, particularly their age, gender and their place of residence.

■ Statistical analysis

All statistical analyses were performed using the Statistical Package for the Social Sciences, version 17.0 (SPSS Inc., IL, USA). Frequency tables were obtained and statistical analysis was performed using the χ^2 test. We also assessed the data for completeness and frequency distributions. Mean values, standard deviations and percentages were computed in order to describe the distribution of independent variables. Cross-tabulation tables (i.e., contingency tables) were created to display the relationship between two or more (nominal or ordinal) variables using the χ^2 test. Probabilities of <0.05 were considered statistically significant, when testing null hypotheses.

Results

The overall sample sizes and characteristics of the surveyed groups are shown in TABLE 1. Every effort was made in the context of both the general public and physicians' groups to be representative of the general public and medical practitioners populations, respectively, in terms of both their gender and age. In the case of the general public, only adult respondents were surveyed. The distribution of the general public regarding their place of residence broadly followed Greek demographics [103]. The questionnaires aimed to ascertain the opinions of the general public and physicians on the following three issues: awareness of genetics and genetic testing; access and various other issues pertaining to genetic testing; and direct-access genetic testing.

■ Awareness of genetics & genetic testing

We first attempted to critically evaluate the degree of education and overall awareness of the general public with respect to issues pertaining to genetics and genetic testing for both common and multifactorial genetic disorders. A significant proportion of the general public was found to be aware of the existence of DNA, the genetic material (general public question [GP-Q1]), its biological role (GP-Q2), the main sources of DNA (GP-Q3) and the existence of public and private genetic-testing laboratories (GP-Q4; TABLE 2); these elements were measured subjectively, as stated by the individuals who responded to the survey. As expected, there were significant differences when the respondents were subdivided according to their place of residence and their age. Indeed, the number of positive responses to these questions was inversely proportional to the age of the respondents, highlighting the fact that the general population is fairly uninformed about genetics. The same trend was also observed when the respondents were classified according to their place of residence, indicating reduced access of inhabitants of smaller cities and villages to information on genetics and genetic testing. Altogether, 84.3% of the general public expressed their willingness in principle to undergo genetic testing (TABLE 2).

■ Access & various other issues pertaining to genetic testing

One important parameter in personalized medicine is the access to genetic testing for both common and multifactorial genetic disorders. Our surveys of both the general public and physicians were designed to address this aspect. From the physicians' questionnaire, we discovered that 74.5% would themselves be willing, at least in principle, to undergo genetic testing (FIGURE 1A). However, a significantly lower percentage (48.5%) had encouraged their patients to undergo genetic testing (FIGURE 1B). More specifically, 42.1% had encouraged their patients to undertake a genetic test for a monogenic or multifactorial disorder, 30% a cytogenetic test, and 16.1% a pharmacogenomic test (FIGURE 1D). In the context of our own approach, molecular genetic testing was taken to refer both to monogenic and complex diseases, the latter resulting from the interaction of genetic predisposition, negative lifestyle or other environmental factors. Despite the fact that both monogenic and complex diseases involve the same sort of genetic analysis in the laboratory, the context and the interpretation

Table 1. Survey sample composition and demographic elements.

	General public, n = 1717 (%)	Physicians, n = 496 (%)
Age (years)		
<35 [†]	32.6	32.6
35–60	49.5	54.4
>60	17.9	11.9
Gender		
Male	46.9	48.3
Female	53.1	51.7
Place of residence		
City	63	
Smaller city [‡]	33.2	
Village	2.8	

[†]Our questionnaires included adult respondents with a minimum age of 18 years.
[‡]Fewer than 50,000 inhabitants.

of testing is very different in these two cases. These results are consistent with the nature of the genetic tests provided by private genetic-testing laboratories in Greece, as indicated by our previous study [5]. Surprisingly, the general public indicated that only 9.5% had been encouraged to undertake a genetic test by their physicians, friends, relatives or a genetic laboratory representative (FIGURE 1C). However, this proportion varied significantly when those who responded to the query were classified according to their place of residence, age or gender (FIGURE 1E).

We then sought to assess the general public's willingness to undertake a genetic test even if the costs would not be reimbursed by their insurance companies. It emerged that 54.8% of the general public would be willing to undertake a genetic test even if the costs would not be reimbursed (FIGURE 2B). Again the proportion of those willing to take a genetic test for a monogenic or multifactorial disorder was significantly larger (94.1%) than those willing to take a pharmacogenomic test, in a situation where the analysis costs would not be reimbursed (FIGURE 2C). In concert with this finding, 77.3% of the physicians who responded to our questionnaire were of the opinion that the cost of genetic testing services should be reimbursed by insurance companies (FIGURE 2A). However, it is noteworthy that a mere 11.9% of physicians believe that there is currently a satisfactory legal framework in Greece to cover aspects of genetic testing (FIGURE 3) such as data privacy, written informed consent, genetic testing laboratory accreditation, regulation of genetic testing costs to avoid overpricing, and so on. This finding is again consistent with our previous finding indicating considerable variation in the accreditation of the various private genetic

Table 2. Critical evaluation of the awareness of the general public on DNA and genetic testing services.

	GP-Q1	p-value	GP-Q2	p-value	GP-Q3	p-value	GP-Q4	p-value	GP-Q6	p-value	GP-Q7	p-value
Place of residence												
City	93.7	0.000	80.8	0.000	87.6	0.000	72.3	0.000	85	0.535	77.4	0.375
Small city	90	0.000	77.5	0.000	82.2	0.000	65.8	0.000	82.9	0.535	74.7	0.375
Village	61.5	0.000	16.9	0.000	0	0.000	0	0.000	83.1	0.535	80	0.375
Age (years)												
<35	94.6	0.000	85.5	0.000	92.1	0.000	81.7	0.000	87.1	0.066	74.4	0.133
35–60	90.7	0.000	82.1	0.000	78.2	0.000	65	0.000	83.3	0.066	78.7	0.133
>60	86.6	0.000	60	0.000	66.2	0.000	48.1	0.000	81.8	0.066	75	0.133
Gender												
Male	88.9	0.001	74.8	0.017	80.3	0.027	63.9	0.004	81.6	0.004	74.6	0.068
Female	93.3	0.001	79.6	0.017	84.4	0.027	70.5	0.004	86.6	0.004	78.4	0.068
Overall	91.3		77.3		82.5		67.5		84.3		76.6	

Statistically significant values are shown in bold.

GP-Q: General public question.

laboratories in Greece, the type of accreditation certificate, genetic testing pricing and the absence, in most cases, of a proper procedure to communicate results to the patients [5].

■ Direct-access genetic testing

Another very important aspect of genetic testing, with serious attendant ethical and (often) legal implications is direct-access testing, also known as direct-to-consumer genetic testing [6]. From our previous analysis, it is clear that at least one genetic service laboratory in Greece offers direct-access genetic testing as part of its public outreach strategy. In addition, buccal swab sampling kits for genetic testing are sold over the counter in at least two pharmacy groups in Greece, while other pharmacies provide the same sampling kit upon request. We therefore sought the opinions of both the general public and physicians regarding direct-access genetic testing. Interestingly, only a very small proportion of the physicians (12.7%; FIGURE 4A) were in favor of direct-access genetic testing, although this proportion was slightly higher among the general public (17.9%; $p = 0.001$; FIGURE 4C). Of those individuals from the general public who were against direct-access genetic testing (82.1%), the vast majority wanted a physician to direct them to genetic testing services and then to explain the test results (96.6%; FIGURE 4D), whereas only 20.4% wanted a pharmacist to refer them to a genetic testing laboratory. These proportions varied significantly when the responders were classified according to their place of residence (FIGURE 5), underlining the fact that only 7.7% of the responders who were living in a village favored direct-access genetic testing services ($p = 0.036$). Similarly, according to the physicians who were against direct-access genetic testing, the vast majority (89.7%) believed that a physician should refer patients and/or interested individuals to a genetic testing laboratory whilst only 5% believed that a pharmacist should be allowed to perform this task (FIGURE 4B).

Discussion

The steady increase in the availability of genetic tests is a direct result of the exponential rate of discoveries in the field of human genomics, the technology now available for genome analysis [7] and our burgeoning knowledge of genotype–phenotype correlations. According to one estimate, more than 700,000 genetic tests are performed in Europe annually in both public and private genetic laboratories [8], and this is

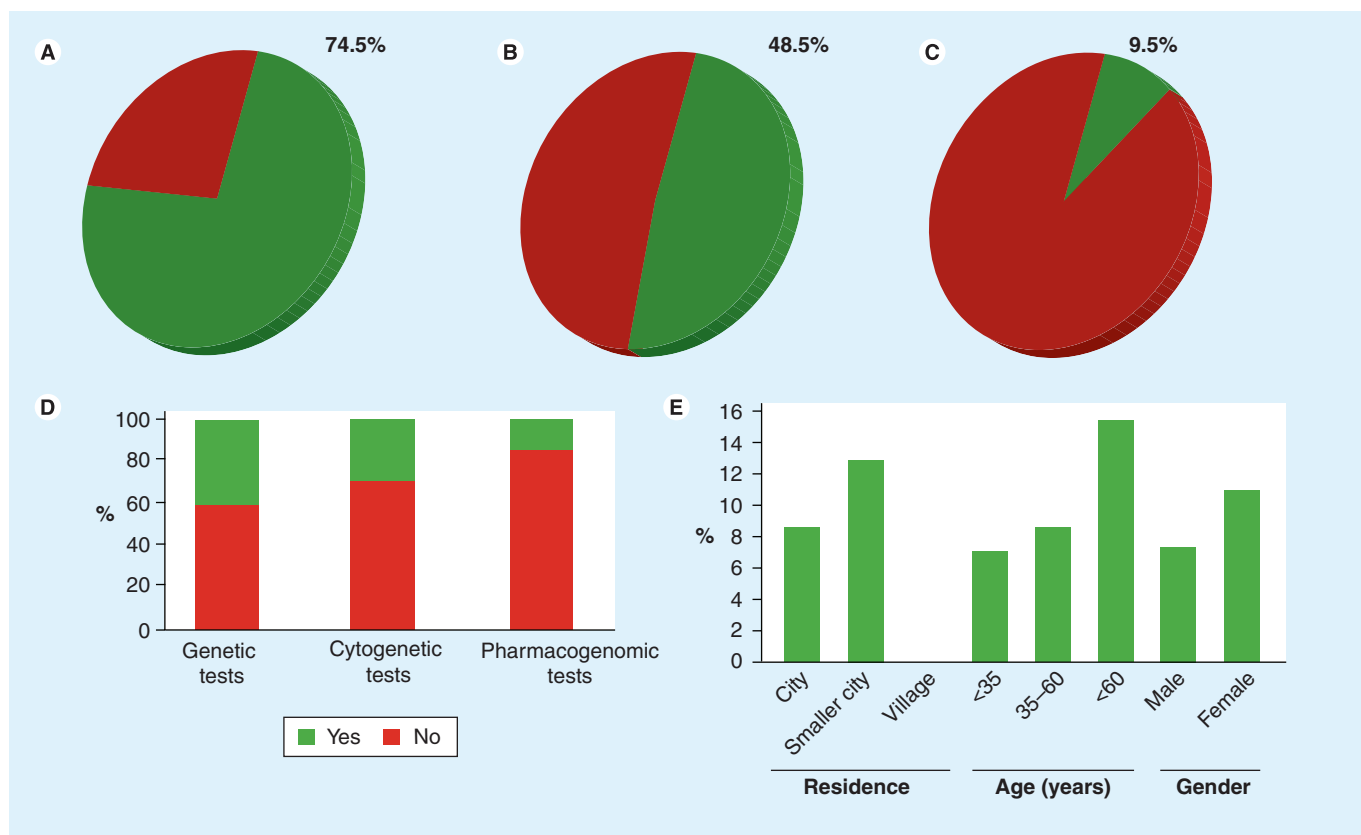


Figure 1. Appraisal of access by physicians and the general public to genetic testing services. Outcome of the survey of physicians' willingness to personally undertake a genetic test (physician question [P-Q]3; [A]) and to direct their patients to genetic testing services (P-Q4; [B]), with a particular focus on genetic (P-Q4A), cytogenetic (P-Q4B) and pharmacogenomic testing (P-Q4C; [D]). Outcome of the general public's feedback with respect to whether their physicians have recommended them to take a genetic test (general public question 5; [C]) and detailed depiction of their positive replies in the various subgroups, regarding their place of residence, age and gender (E).

expected to increase in the years to come. In contrast to the USA, in Europe there are significant differences between individual countries, even between EU member states, regarding genetic testing services. In other words, in some countries, there are established regulatory frameworks and provisions for genetic testing services, whereas in others the area is still not properly regulated (for an overview of the existing regulatory frameworks on genetic testing services, see the European Society of Human Genetics' website [104]). Although there have been some attempt to harmonize genetic testing practices across Europe, an in-depth analysis, based on comprehensive surveys of the current situation in European countries is still lacking. Presently, only a handful of studies have been performed in European populations to assess the attitude of the general public towards genetics and genetic-testing services, namely in Finland [9], Germany [10] and Russia [11].

The present study complements our previous work on the private genetic testing environment in Greece [5], by attempting to explore how both

the general public and medical practitioners perceive genetics and genetic-testing services in the country. It was designed to explore how the Greek general public and their attending physicians perceive genetics and genetic testing, what their opinions are with respect to the regulatory and legal frameworks that oversee these services and what they think about the concept of direct-access genetic testing, which has recently gained significant popularity. These surveys are not only among the very first of their kind performed in Europe, along with namely Finland and Germany, but also paid special attention (for the first time in Europe) to attitudes towards pharmacogenomic testing, since this emerging discipline is anticipated to have a central role in translational medicine in the future.

Our surveys included a large number of participants from the general public and physicians. We opted to carry out personal interviews rather than acquire information through electronic surveys since, from our own experience, the latter approach would not have yielded a satisfactory number of responses, particularly

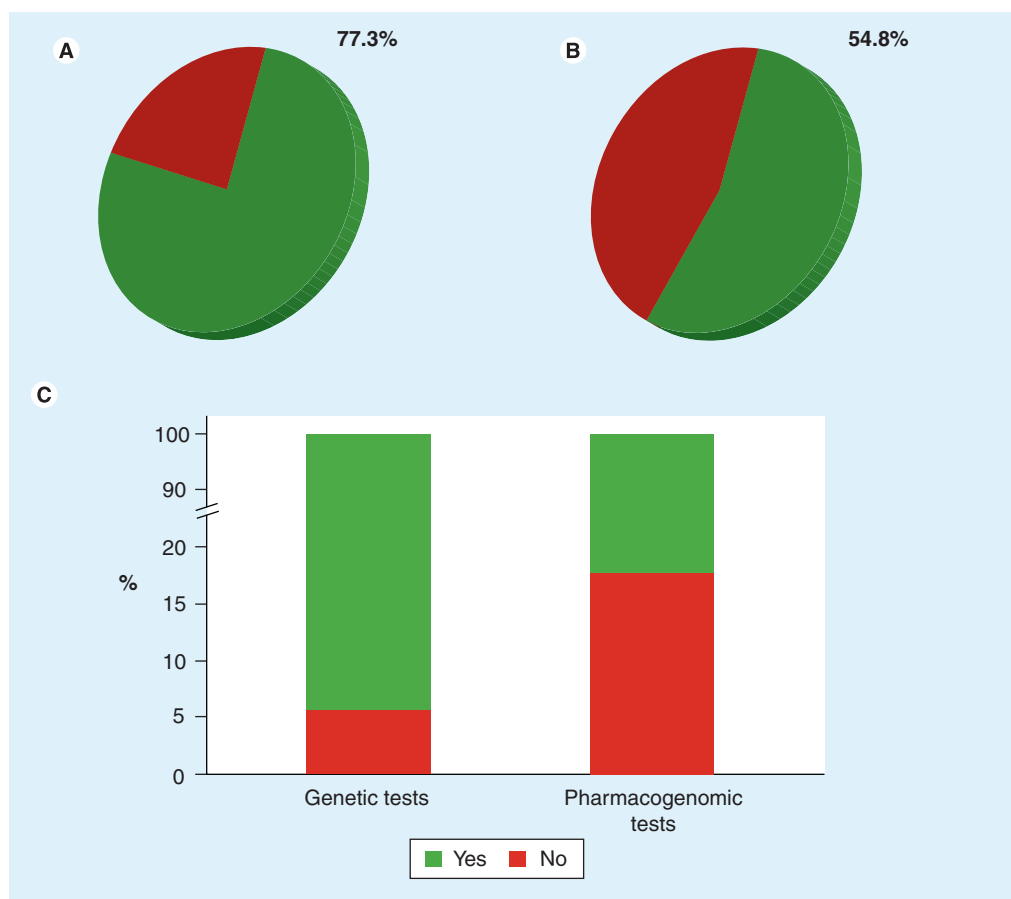


Figure 2. Opinions of the physicians and the general public with regard to genetic testing and reimbursement of the corresponding costs. Physicians' opinion of the reimbursement of genetic testing costs (physician question [P-Q]1; **[A]**), willingness of the general public (GP) to undergo genetic testing when analysis costs are not reimbursed by insurance companies (GP-Q8; **[B]**) and differences with respect to genetic and pharmacogenomic tests (GP-Q8A and GP-Q8B; **[C]**).

from older people and those living in smaller cities and villages who are likely to be less computer literate. Moreover, it would have probably introduced bias since those people who are most computer literate are also likely to be the most informed about genetic testing for a variety of reasons. Participants from the general public were therefore approached in several public places, such as pharmacies, supermarkets, cafes and restaurants, while physicians were selected while attending the 36th Panhellenic Medical Conference in Athens, Greece, on the 4–8 May 2010. We fully appreciate that few of our questions required a simple 'yes/no' answer, a study design that may not always provide the most useful insights when seeking to identify whether the public is aware of some specific fact or issue. On the other hand, we wished to avoid eliciting stereotypical responses and kept the questionnaire simple and easy to answer in order to encourage maximum participation on the part of our respondents, particularly the main target groups.

■ Overall perception of genetics & genetic testing

Our first goal was to critically evaluate the degree of education and overall awareness of the general public with respect to issues pertaining to genetics and genetic testing. As indicated in TABLE 2, the majority of the general public was aware of the nature and role of the genetic material, as well as the various different sources from which an individual's DNA can be obtained. They were also well aware of the existence of both public and private genetic testing laboratories, while 84.3% of the general public expressed their willingness to undergo genetic testing (TABLE 2). This proportion was markedly similar to that observed for a similar sized urban Russian population, where 85% of the 2000 respondents answered positively to a question regarding their own willingness to undergo predictive genetic testing for preventable health conditions [11]. However, only a small proportion of people have actually been encouraged to undergo genetic testing by a physician, a relative

or even a laboratory representative (FIGURES 1C & E). Intriguingly, this proportion is somewhat lower than the professed willingness of the physicians to direct their patients to genetic-testing services if deemed necessary (FIGURE 1B). This finding can be explained by the fact that although physicians are willing in principle to recommend genetic testing to their patients, in practice they often fail to do so. One possible explanation could be a lack of understanding and/or poor education with respect to the potential benefits of genetic testing for monogenic and complex disorders, classical or molecular cytogenetics and pharmacogenomics. This concurs with a previous study that indicated that only 5% of prescribers of azathioprine had requested DNA testing for variants in the *TPMT* gene to determine the ability of their patients to respond to the treatment [12]. Furthermore, significant differences were observed among the responses of the general public according to their places of residence, their age and, in certain cases, their gender (TABLE 2), which can again be explained by a lack of awareness of genetics and issues pertaining to genetic testing. These data are comparable to a similar sized study conducted in Russia, indicating that gender and age significantly influenced responses of the 2000 respondents who participated in this survey [11]. Our results also indicated that genetic tests for a monogenic or multifactorial disorder are strongly preferred over cytogenetic and pharmacogenomics tests (FIGURE 1D), as indicated by the physicians' responses; these findings are concordant with our companion study of private genetic-testing laboratories in Greece [5]. Similar results have also been reported from a comparable study in the UK, albeit involving a significantly smaller number of individuals [13].

The reimbursement of genetic testing costs by insurance companies is another important parameter of genetic testing to consider. The lack of any reimbursement could discourage interested parties from undergoing genetic testing, especially when costs are rather high or the patients are from a low-income bracket. A significant proportion (54.8%) of the general public expressed their willingness to take a genetic test even if the costs would not be reimbursed (FIGURE 2B), and again a preference for genetic testing for a monogenic or multifactorial disorder over pharmacogenomic testing was evident (FIGURE 2C). Of course, this proportion is critically dependent on the health benefits, as these are perceived by the patient, and hence the latter percentage is likely to be smaller in relation to

those tests that do not provide such information, for example, pharmacogenomic tests predicting drug toxicity versus efficacy. These findings are in contrast with those from a recent survey in Canada that indicated that very few respondents were willing to pay for genetic testing to acquire information about genetic factors related to clinical disorders; 62% indicated that the public healthcare system should reimburse these tests [14]. It should be noted that the latter survey performed in Canada distinguished genetic tests by the type of information that could be gained from them (e.g., genetic factors related to manageable conditions or serious, unpreventable disease), and specified how much one is willing to pay (in cash brackets), and as such is not directly comparable to our present study.

Importantly, over 75% of the physicians questioned were on the belief that the costs of genetic testing services should be reimbursed by insurance companies (FIGURE 2A). Our questionnaire that was presented to physicians did not distinguish between genetic testing for inherited disorders and pharmacogenomic testing; it would therefore be interesting to see if physicians differ from the general public in terms of their beliefs on this issue. These findings should constitute a major driving force behind efforts to establish the necessary regulatory framework so that genetic testing costs can be reimbursed. In their responses, the physicians emphasized the lack of a satisfactory legal framework to cover genetic testing (FIGURE 3), such as the accreditation of genetic testing laboratories, data privacy, written informed consent and the regulation of genetic testing costs. In Greece, genetic testing services are mainly regulated through the legal framework which applies to the Greek national

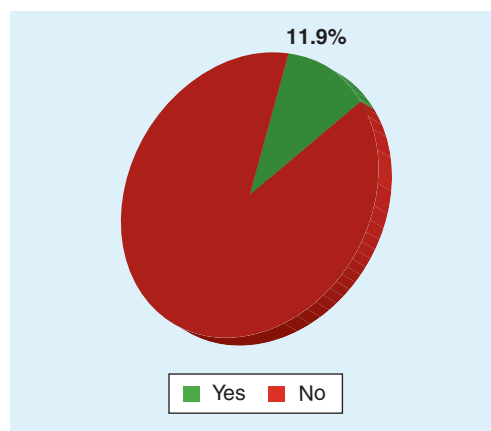


Figure 3. Physicians' opinion with regard to the existence of the necessary legal framework covering genetic testing in Greece (physician question 2).

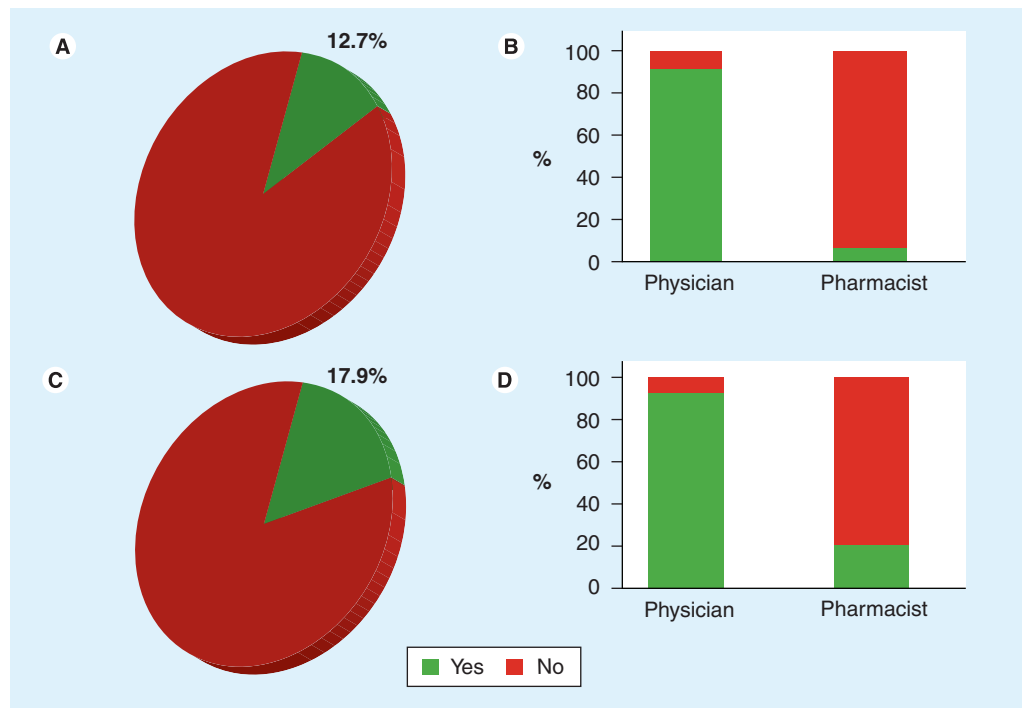


Figure 4. Opinions regarding direct-access genetic testing in Greece. Physicians' overall views on the concept of direct-access genetic testing physician question (P-Q)5, P-Q5A and P-Q5B; **(A & B)** and the corresponding view of the general public (general public question [GP-Q]9, GP-Q9A and GP-Q9B; **(C & D)**).

healthcare system as a whole and as such there are no dedicated laws specifically intended to cover genetic testing services. The regulations on patient rights are readily applicable as rights of genetic services users. The authority of the Greek Bioethics Committee is restricted to provide some important recommendations that complement the existing legislation. Our companion study [5] demonstrated that there is considerable variability in terms of: the accreditation of the various private genetic laboratories in Greece; the nature of their accreditation certificate; the cost of genetic testing and; the proper means (or not, in most cases) to communicate test results to the patients. There is a clear gap in current Greek legislation regarding direct-access genetic testing (see 'Direct-access genetic testing & society' section). Indeed, the Hellenic Society of Medical Geneticists [105] (content in Greek language) and the Hellenic Bioscientists Association [106] (content in Greek language) have both published warnings regarding direct-access testing services being offered by Greek private genetic laboratories (using call centers or advertizing these tests over the internet), stressing that these are highly specialized tests whose potential benefits and results cannot possibly be communicated by these means and by people who have not received the appropriate training.

■ Direct-access genetic testing & society

Direct-access genetic testing represents a very controversial issue with serious ethical and societal implications [15]. More than 1000 genome variants are associated with susceptibility to genetic disorders and as a result, since 2007, an increasing number of genetic tests for common disorders and 'predictive markers' are available, most of which lack sufficient evidence of clinical validity, any proper meta-analysis of the marker(s) in question, and hence their utility in a clinical setting is doubtful [16]. These tests can be purchased via the internet or over the counter in pharmacies in the USA and certain European countries, without the need for a medical specialist or biomedical scientist as intermediary. The results of these tests may therefore confuse the purchasers, and may falsely raise concern or even distress, or conversely provide false reassurance, while the lack of proper communication of the test results by a medical specialist deprives people of an adequate explanation for the potential consequences of the test result or of possible courses of remedial action in relation to their health. In other words, taking such tests may simply be a waste of money, thereby negatively impacting upon the public's opinion and diminishing their trust in genetic testing for *bona fide* medical purposes [17].

Our companion survey of private genetic laboratories indicated that at least one genetic laboratory in Greece offers direct-access genetic testing services [5], which had unfortunately not responded to our survey. Since direct-access genetic testing is a rather controversial issue and very few studies have previously been performed to canvas the general public's and physicians' opinions, we decided to include this topic in our surveys. Our results indicate that very few physicians (12.7%) favor direct-access testing (FIGURE 4A), although a slightly larger proportion of the general public hold the same view (17.9%; FIGURE 4C). In the latter case, this percentage was significantly higher compared with that obtained from a study performed in the UK [18], indicating that only 5% of the respondents would be willing to undertake such a test, even if the price were less than £250. In the same study, a surprising 50% responded positively to the hypothetical question of whether or not they would be willing to undertake such a test even if it were offered free of charge. These data concur with similar results from the USA [19,20], indicating that individuals undergoing direct-access genetic testing have significant concerns about the entire process. Such information was not previously available in European populations [21], which was another important outcome from our surveys. Thus, the majority of both physicians and the general public are largely against direct-access genetic testing. This of course raises serious concerns with respect to the marketing channels that these laboratories employ to attract new customers. For example, advertizing these tests through the internet is the norm, whereas other marketing channels include cold-calls, advertisements in newspapers, or even information days at various venues (e.g., schools and municipal health centers, amongst others) from scientists collaborating with these laboratories. Preliminary data from our meta-analysis of several genes and DNA variants included in the tests offered indicate that, in the case of at least two genes and their accompanying variants, there is insufficient scientific evidence to include data pertaining to these genes/variants in the calculation of the overall risk [PAVLIDOU AND COWORKERS, MANUSCRIPT IN PREPARATION]. This of course poses some serious concerns regarding the scientific accuracy of the results obtained.

In general, those physicians and members of the general public who intimated that they were against direct-access genetic testing also indicated that they preferred a physician rather than a pharmacist to refer the interested parties to a genetic laboratory. In general, physicians

and pharmacists are the key interlocutors for the general public and this indeed was our reason for including them in the survey. In particular, 96.6% of the general public wished a physician to refer them to a genetic laboratory and to explain the test results to them, with a significantly smaller percentage (20.4%) being content to go through a pharmacist. These percentages varied significantly when the responders were classified according to their place of residence (FIGURE 5); only 7.7% of the responders who were living in a village favored direct-access genetic testing services. Similarly, with respect to the physicians who were against direct-access genetic testing, the vast majority (89.7%) believed that only a physician should refer patients and interested individuals to a genetic testing laboratory, whereas only 5% believed that it was appropriate for a pharmacist to undertake this task (FIGURE 4B). These findings should be considered alongside the results of our companion study [5], which demonstrated that one Greek pharmacy group promotes genetic tests and sells DNA sampling kits to the public over the counter whereas other pharmacies are generally willing to order these sampling kits upon request. We have made enquiries to several pharmacies regarding the demand for these kits, and it would appear that the demand is very low.

Conclusion

We provide here the results from two nationwide surveys which assesses the general public's awareness of, and physicians' opinion on, genetic testing services in Greece. Participants in our surveys were relatively well informed about genetics; however, at the same time, therefore more skeptical towards certain aspects of genetic testing, particularly direct-access genetic testing. In other words, it

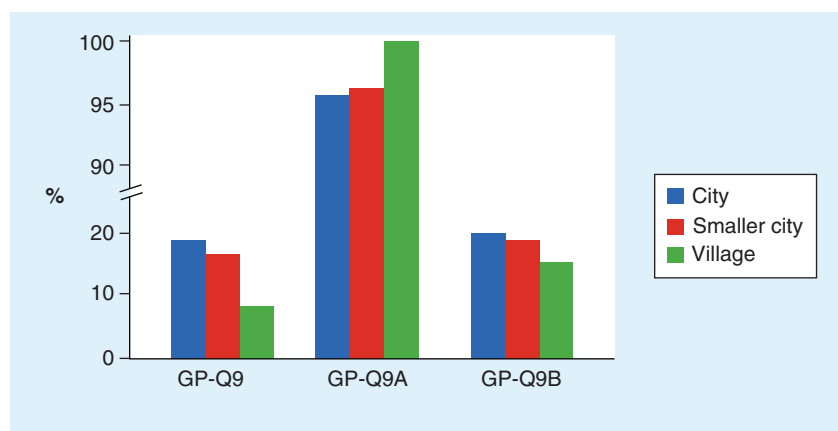


Figure 5. Differences in the views expressed by the general public with respect to direct-access genetic testing, according to their place of residence (see text for details).

GP-Q: General public question.

would appear that a better-informed general public harbors more critical views and is not necessarily more supportive of new genetics research and discovery. Our results provide significant new insights into the potential benefits and pitfalls of genetic testing in Greece. Our future goal is to expand this study in order to acquire further insight into both the publics and physicians' attitudes towards genetic testing so that legal issues and regulatory weaknesses may be addressed with the aim of ensuring that the field will come to be adequately and appropriately regulated. To this end, it is hoped that the existing gap between the overall provision of genetic testing in Europe and the USA will somehow be bridged. Our study not only provides the basis for a critical appraisal of the genetic testing environment in Greece, but also stands as a model for replication in other countries to assess the landscape of genetic testing services.

Acknowledgements

The authors are indebted to the participants of their surveys, without whom this study would not have been possible. The

authors also cordially thank Marianna Kouskou, Sofia Koutoglou, Maria-Theodora Melissari, Christina Paliou and Chris Loizos for their valuable assistance during this project.

Financial & competing interests disclosure

This work was partly funded by the Golden Helix Institute of Biomedical Research and by the University of Patras research budget. The authors have no other relevant affiliations or financial involvement with any organization or entity with a financial interest in or financial conflict with the subject matter or materials discussed in the manuscript apart from those disclosed.

No writing assistance was utilized in the production of this manuscript.

Ethical conduct of research

The authors state that they have obtained appropriate institutional review board approval or have followed the principles outlined in the Declaration of Helsinki for all human or animal experimental investigations. In addition, for investigations involving human subjects, informed consent has been obtained from the participants involved.

Executive summary

- In the postgenomic era, we are witnessing significant advances in the functional decipherment of the human genome sequence. This has been made possible by new technological developments in the field of genomic medicine, which in turn, has facilitated the incorporation of genetic testing services into mainstream clinical practice. However, in many European countries, there is very little knowledge regarding how either the general public or medical practitioners perceive genetics and genetic testing services.
- For this reason, we initiated a pilot nationwide survey involving 1717 individuals from the general population in Greece, divided according to age and residence, in order to better understand how the general public perceives genetic testing. At the same time, we performed a similar survey involving 496 Greek physicians.

Results

- Our analysis indicated that a significant number of respondents from the general public are aware, at least in principle, of the nature of DNA and genetic disorders, as well as the potential benefits of genetic testing. Moreover, a large proportion of the respondents were willing to undergo genetic testing even if the costs of analysis would not be reimbursed. Perhaps surprisingly, only a relatively small proportion of the general public has actually been advised to undergo genetic testing, either by relatives or physicians.
- In addition, a large proportion of the physicians who participated in this survey believe that the regulatory and legal frameworks that govern the provision of genetic testing services in Greece are rather weak. The vast majority of both the general public and physicians strongly oppose direct-access genetic testing, and would generally prefer referral to be from a physician rather than from a pharmacist.

Conclusion

- Overall, these results provide the basis for an assessment of the views of the general public and physicians on genetics and genetic testing services in Greece. These surveys could be readily replicated in other populations.

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