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# **A critical appraisal of the private genetic and pharmacogenomic testing environment in Greece**

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Running head: Landscape of genetic testing services in Greece

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## **Executive Summary**

- The landscape of private genetic testing services is poorly developed in many European countries as well as the United States.
- We have performed a nationwide survey of 18 private genetic testing laboratories located in Athens and various other cities in Greece in order to obtain a better understanding of the genetic testing services that these centers provide.
- Our questionnaire included 22 questions on various aspects of genetic testing, such as the types of genetic test offered by these private laboratories, their target clientele and the marketing channels used to approach them, the cost of the analyses, and laboratory accreditation.
- 13 of the 18 private genetic testing laboratories responded to our questionnaire and analysis of their responses indicated that although molecular genetic and cytogenetic testing tend to predominate, the demand for pharmacogenomic testing is steadily growing.
- In Greece, physicians comprise the main target group for private genetic testing laboratories. Generally, they tend to be approached via the Internet, through personal contact from sales representatives or at scientific conferences.
- Although the costs of genetic testing are fairly low in Greece, most genetic laboratories either employ or collaborate with a genetic counselor. However, few laboratories appear to be properly accredited for the provision of genetic testing services.
- Our study constitutes a critical appraisal of the private genetic testing environment in Greece and provides a model for replication in other European countries.

## **Abstract**

In the post-genomic era, we are witnessing rapid progress in the identification of the molecular basis of human inherited disorders and the elucidation of genotype-phenotype relationships. The rate of progress has been driven not only by the determination and ongoing decipherment of the human genome sequence but also by the advent of new technological developments that have dramatically reduced the costs of genetic analysis. As a consequence, a considerable number of genetic testing centers have emerged, both in Europe and the United States, which offer a plethora of different genetic tests. We have performed a nationwide survey of 18 private genetic testing laboratories in Greece to acquire a better understanding of the genetic testing services that these centers provide, specifically the types of genetic test offered, the target groups, marketing channels, costs of analysis and accreditation. Molecular genetic and cytogenetic testing were found to be the predominant types of genetic testing services offered although there is an increasing demand for pharmacogenomic testing. The main target group for private genetic testing laboratories are physicians who are approached via the Internet, through personal contacts from sales representatives, and at scientific conferences. Genetic testing costs are fairly low in Greece. Although the majority of private genetic laboratories either employ or collaborate with a genetic counselor, few of them are accredited for the provision of genetic testing services. This study provides the basis for a critical appraisal of the private genetic testing environment in Greece and provides a model for replication in other European countries.

**Key Words:** Genetic testing, private laboratories, Greece, physicians, cytogenetics, molecular genetics, pharmacogenomics

## Introduction

In recent years, significant advances have been made in our understanding of the genetic basis of inherited disorders and the correlations between mutant genotype and clinical phenotype, both for monogenic and multifactorial conditions [Chen et al., 2010; Cooper et al. 2010]. These advances, in conjunction with the advent of high-throughput genetic analysis and deep resequencing, have served to reshape the field of modern medical practice [Metzker, 2010] and are reflected in the rapid development of the genetic testing industry [Ginsburg and Willard, 2009; Caulfield et al., 2010]. Nowadays, there are a wide variety of public entities and private companies that offer a broad range of antenatal and postnatal molecular genetic testing services for monogenic and multigene disorders, classical and molecular cytogenetics analysis for chromosomal rearrangements, pharmacogenomic testing and even predictive genomics for genetic disorders. In addition, many laboratories also offer molecular genetic testing services in microbiology and virology. At the same time, genetic testing services are becoming more affordable so that we can already envisage genome resequencing for as little as \$1,000 [Davies, 2010].

However, the rapid expansion of the genetic testing industry has not come without problems. In particular, some laboratories still offer genetic analysis services using in-house ('home-brew') kits rather than quality-controlled and certified assays. In addition, test results are not invariably interpreted by a qualified professional (*e.g.*, a genetic counselor), whereas other laboratories are not yet accredited for the provision of genetic testing services [Burnett, 2009]. Moreover, it transpires that, in several cases, genetic analysis is routinely conducted without obtaining informed consent from those persons requesting the test. This raises serious ethical concerns in relation to the preservation of the anonymity of the individuals tested [Gurwitz and Bregman-Eschet, 2009], the fate of

their genetic material and, most importantly, the safeguarding of test results in order to avoid genetic stigmatization [Guttmacher et al., 2010].

The landscape of private genetic testing services is still poorly developed in many parts of Europe and the United States. Hence, a number of different ethical issues often arise as a consequence. EuroGenTest (<http://www.eurogentest.org>) has recently attempted to plug this gap by initiating a drive to harmonize genetic testing services in Europe. In parallel, OrphaNet (<http://www.orpha.net>) has attempted to database the plethora of genetic testing laboratories in Europe but these efforts have often been hampered by the willingness of some laboratories to communicate the requested details of their operations. It is therefore clear that, in emerging era of personalized genomics, the task of ‘fine mapping’ genetic testing services in Europe is assuming ever greater urgency.

We have initiated nationwide surveys to assess private genetic testing services currently available in Greece, not simply from the patients’ and physicians’ point of view but also from the test providers’ standpoint. In these surveys, we have aimed to: (a) map patients’ and physicians’ needs with respect to the genetic testing industry, (b) identify regulatory deficiencies and gaps in the existing legal provision that could be rectified by appropriate legislation, and (c) provide a model for the European-wide survey of genetic testing laboratories with the ultimate goal of harmonizing genetic testing in Europe. Here, we report the results from our initial survey of genetic testing providers in Greece.

## **Methods**

This research study was conducted between March 2010 and December 2010, during which 18 private genetic testing laboratories from various cities in Greece (Table 1), and comprising the bulk of the genetic testing industry in Greece, were invited to participate. The names and websites of the participating laboratories will be made available upon request.

The survey addressed issues such as: (a) the collection of qualitative data on the types of genetic testing services provided, (b) the laboratories' clientele, (c) specific details of the approach to genetic testing, such as methods of DNA isolation and analysis, accreditation, (d) approaches to marketing, and (e) the costs to the consumer of the genetic tests being offered. A 21-point questionnaire (**Supplementary data online**) was sent to the laboratories' scientific and management personnel for self-completion, from which quantitative and qualitative data were subsequently generated. Pharmacogenomic testing formed an integral part of the questionnaire, since it has gradually become an important area of personalized medicine.

A simple binary approach (No=0, Yes=1) was employed to score the answers given. Information provided was then cross-checked from the corresponding websites, where available to ensure accuracy and consistency.

## **Results**

Thirteen of the 18 private genetic testing laboratories (72.2%) responded to the questionnaire. In almost half of all cases, several reminders had to be sent in order to elicit a response; this situation may reflect a certain degree of reluctance on the part of commercial entities to participate in such surveys (MB Petersen, personal communication).

Our data indicate that most of the surveyed genetic testing laboratories (92.3%) are involved in the provision of molecular genetic analysis for inherited disorders, followed closely by classical and/or molecular cytogenetic testing (84.6%), and molecular genetic testing for microbiology and predictive genomics (76.2%; **Fig. 1A**). By contrast, pharmacogenomic analysis was only offered by 8 of the 13 laboratories that completed the survey (61.5%). From this initial survey, it would appear that there is currently a greater demand for molecular genetic and predictive genomic testing services than other types of analysis (**Fig. 1B**). Interestingly, pharmacogenomic testing is currently the least popular among patients, probably because this is a relatively new field and people are less educated with regard to the benefits of this type of test. Usually, patients are more interested in, and informed about, their own susceptibility to disease than they are about potential adverse drug reactions or inter-individual differences in drug metabolism and disposition, and the same may well be true for the attending physicians. Ironically, modulating or even avoiding a particular drug treatment is relatively straightforward but it is not necessarily so easy to avoid the consequences of an innate susceptibility to disease. Thus, in the longer term, it may well be that pharmacogenomic testing will deliver the most in terms of clinical benefit to the patient and hence will be more widely adopted in a clinical setting.

The questionnaire responses indicated that physicians are the main target group for the genetic laboratories (92.3%), followed by the general public (43.2%) and other interested parties such as other genetic laboratories, diagnostic centres, hospitals, and pharmaceutical companies (30.8%). The main specialties that the diagnostic laboratories mainly address are obstetricians/gynecologists (92.3%), followed by pathologists (69.2%), cardiologists (69.2), psychiatrists (30.8%) and other specialties namely oncologists, pediatricians, hematologists, urologists, neurologists, and surgeons (**Fig.**



**2A).** This may not be unexpected since obstetricians and gynecologists usually order molecular genetic and, particularly, cytogenetic tests to screen for fetal malformations, particularly in cases with a family history. In addition, psychiatrists, cardiologists and oncologists are the physicians who order pharmacogenomic tests more frequently, since these are the disciplines in which pharmacogenomic testing has been most widely adopted. Of course, other factors may influence the percentage of physicians ordering the genetic tests, e.g. the numbers of clinicians in different specialties, differential funding available to practitioners in each specialty, differences in genetics education between specialties, *etc.* Interestingly, in the context of paternity testing, lawyers can also be a target group of the genetic testing laboratory.

In the case of the age range of the people undergoing genetic testing, the majority are aged between 35 and 60 years (92.3%), followed by people younger than 35 years (76.9%). Perhaps unsurprisingly, people older than 60 years of age are less likely to undergo genetic testing (46.2%; **Fig. 2B**), most likely due to their lack of knowledge about the potential benefits of genetic analysis (Koromila and coworkers, in preparation). It might also be that people older than 60 are less likely to want genetic disease testing, as they are probably already quite knowledgeable about their existing pathologies from personal experience and less likely to want paternity testing for obvious reasons. However, one would perhaps expect this group to be more likely to avail themselves of genetic testing in an oncology context. As far as the source of the genetic material is concerned, peripheral blood was found to be the most commonly used DNA source (92.3%), followed by buccal swab samples (84.6%), tissue samples (e.g. paraffin-embedded tissue or fresh biopsies; 76.9%), saliva (53.9%) and other sources such as semen, urine, cell cultures (**Fig. 3**). The choice of sample is going to be very much test-dependent and hence very much a function of what tests the different

companies perform. For example, buccal swabs and saliva samples may be more frequently used as a DNA source for predictive genomic testing, while peripheral blood will be used for molecular genetic testing services. Although the results obtained from the isolated DNA do not differ, physicians and the general public generally tend to prefer peripheral blood as the DNA source for genetic tests, since they appear to feel that this sampling and DNA isolation approach somehow has a more solid scientific basis as compared to the other DNA sources.

An integral part of the questionnaire was the issue of the cost to the consumer of the available tests. The majority of these tests were cheaper than 300 EUR (**Fig. 4**) whereas the number of genetic tests performed decreased as the corresponding analysis costs increased (**Fig. 4** and **Supplementary information online**). All pharmacogenomic tests offered were cheaper than 300 EUR, while in some cases the cost of the test was as low as 50 EUR when such tests are ordered and performed in bulk.

Although the majority of private genetic testing laboratories performed genetic analysis with informed written consent from the patients (76.9%), several genetic laboratories did not fulfill this requirement, according to their responses to our survey. This finding underlines the need to make informed written consent a obligatory requirement for genetic testing in Greece and other European countries.

It was however encouraging that all private genetic testing laboratories that responded to our survey had a genetic counselor working or collaborating with them. Although the majority of genetic testing laboratories have an ISO certificate (84.6%), and hence may be considered accredited, very few have been certified for the provision of genetic testing services specifically (ISO-15189 and/or ISO-17025; Burnett et al., 2009), namely ISO-17025 (15.4%) and ISO-15189 (15.4%). The accreditation system in Greece follows that adopted in other European countries. The introduction of new

genetic and pharmacogenomic tests occurs after consultation of the scientific literature for new research studies on genotype-phenotype correlations (76.9%), followed by recommendations from an internal scientific review group, to the company, Scientific Advisory Board or scientists (69.2%), external physicians (61.5%) or advisors (38.5%).

As stated above, 61.8% of the private genetic laboratories offer pharmacogenomic testing services. These tests relate mostly to anticoagulant drugs (53.9%), followed by antidepressants and antipsychotic drugs (38.5%), cholesterol lowering drugs (30.8%), oncologic drugs (15.4%) and analgesics (30.8%; **Fig. 5**). Finally, we wished to understand the various marketing approaches adopted by each laboratory to attract test samples. According to our survey, the laboratory's own website constituted the most frequently used means to inform both the general public and physicians about new genetic and pharmacogenomic tests being offered (both 92.3%; **Fig. 6A, B**). Physicians were also informed by attendance at scientific conferences (63.2%) and direct contact with sales representatives (63.2%), while only 23.1% obtained their information through educational seminars organized by the laboratories (**Fig. 6A**). Despite this, 46.2% of the genetic laboratories had participated as sponsors and 30.8% as exhibitors in a recent (2010) Greek medical or genetics-related conference, in which physicians mostly participate. By contrast, 61.5% of the genetic laboratories inform the general public of their available genetic and pharmacogenomic tests via custom produced advertising material (e.g. leaflets, brochures that are mailed to them directly, while only 23.1% employed advertisements in newspapers or magazines (**Fig. 6B**). According to the survey (**Fig. 6B**), none of the responding genetic laboratories uses a call center to directly communicate their services to the general public by making unsolicited calls to tout for business. However, we are aware that at least one of the laboratories that failed to respond to our questionnaire uses this approach as part of its

public outreach strategy. In addition, buccal swab sampling kits for genetic tests are sold over the counter in at least one chain of pharmacies in Greece, while other pharmacies provide the same sampling kit upon request.

## **Discussion**

The rapid pace of development in the field of human genomic technologies has led to an exponential increase in the number of available genetic tests and a decrease in the relative cost of each genetic analysis. As a result, genetic testing services have become readily available to patients and the general public alike, whether to assess the risk of developing a life-threatening inherited disorder or to predict the efficacy of (or toxicity to) a specific drug. Indeed, it is estimated that more than 700.000 genetic tests are performed in Europe on an annual basis [Grimaldi et al., 2011], and this figure is expected to grow rapidly over the next few years. Although the number of public and private genetic testing laboratories continues to grow, little is known about the general landscape in which genetic testing services operate in many countries. Indeed, in many European countries, there are gaps in legislation covering genetic testing (<https://www.eshg.org/fileadmin/www.eshg.org/documents/Europe/LegalWS/ReportESHG-LegalWorkshop2010.pdf>), and hence the different parties involved are not fully protected from unethical practices [Hogarth et al., 2008]. In Europe, there are significant differences between individual countries as far as genetic testing services are concerned. Although in many countries, there is an established regulatory framework and provisions for genetic testing, in other countries the area is still not properly regulated. The EuroGenTest Network (<http://www.eurogentest.org>) and EuroGenGuide (<http://www.eurogenguide.org.uk>) are some of the efforts that have been attempting not only to harmonize genetic testing services across Europe but also to provide guidance on

genetic testing/analysis for patients and physicians. These commendable efforts notwithstanding, a detailed analysis of the current situation in European countries is currently lacking. For most of these countries, there are some reports, available through newsletters from the European Society of Human Genetics (<http://www.eshg.org>), but no comprehensive survey has ever been conducted.

In a Greek context, the first genetics units appeared in Athens in the early 1960s and cytogenetic laboratories became the first to offer their services to the general public. Since 2003, at which time only 5 private genetic laboratories offered (a range of) genetic services in Greece, their number has increased significantly. However, the country still lacks formal genetics centers organized within a national genetic testing network, as in the United Kingdom (the UK Genetic Testing Network, which advises the NHS on genetic testing across the whole of the UK; <http://www.ukgtn.nhs.uk/gtn/Home>). Also, the number of physicians in full-time clinical genetics is extremely limited in the public sector, while at the same time there are a large number of well trained clinical scientists in genetics who cannot be absorbed despite the growing need for genetic services. University departments and public hospitals are usually under-staffed and under-paid owing to the lack of resources to support trained personnel. Recently, we initiated a major nationwide survey to try to understand the context of private genetic testing services in Greece and to explore how both the general public and physicians perceive genetics and genetic testing services. Such an analysis has not previously been performed and it was designed to address key aspects of genetic analysis such as ethics and education as well as insurance and confidentiality issues. In addition, our survey paid particular attention to pharmacogenomic testing since this emerging discipline is anticipated to have a central role in translational medicine.

Our results from surveying the provision of genetic testing services from private laboratories showed that, at present, demand leans towards molecular genetic and cytogenetic testing, following many years of successful application of these approaches. According to the views of certain laboratories who offer this service, pharmaceutical companies in Greece attempt to discourage physicians from recommending pharmacogenomic testing, since this will tend to reduce the pharmaceutical companies' profit margins. The argument that they use is that the pharmacogenomic test costs significantly more than simply trying out the drug in question. Moreover, given the recent emergence of this field, pharmacogenomic tests currently tend to be a low priority for customers of genetic testing companies (**Fig. 1**). This is however likely to change over time as a consequence of integrating pharmacogenomics into mainstream medical practice.

Another interesting finding is the nature of the customer target group of private genetic testing laboratories, who are primarily physicians, in particular gynecologists (**Fig. 2**) and to a lesser extent pathologists, cardiologists, oncologists and psychiatrists. This result is not so unexpected given that the general public in Greece is strongly opposed to direct-access testing (Koromila and coworkers, in preparation). However, a significant fraction of laboratories (43.2%) receive test requests directly from patients. In this latter case, the presence of a qualified genetic counselor is required to accurately and reliably communicate the test result to the patient who requested the test. According to the laboratory personnel, physicians that refer patients to them for testing are not in a position to explain the test result, due to lack of the appropriate education and training. This finding concurs with the results obtained from the physicians' survey (Koromila and coworkers, in preparation), where a significant proportion of physicians (particularly the older physicians) display a remarkable lack of knowledge of genetics. According to

the laboratory personnel, the younger new generation physicians understand genetics much better and hence are not only in a better position to explain test results but also eager to encourage their patients to undergo genetic testing in the first place. We must be aware that it has only been relatively recently that molecular biology and genetics has been incorporated into mainstream university curricula as an integral part of medical, pharmacy and nursing studies at the undergraduate and graduate level.

Another interesting finding from the genetic laboratories survey indicates that the means of communication with their target group is primarily via the company website (92.3%, both for physicians and patients), followed by advertising, scientific conferences and visits to physicians from sales representatives. The companies who responded to our survey indicated that they do not use call centers to make unsolicited calls to the general public to tout for business. However, the authors are aware of at least two private genetic laboratories in Athens who have either used this approach in the past or are currently using this approach in order to attract their clientele. We believe that this approach is wholly unethical since the general public is most unlikely to acquire a detailed understanding of the pros and cons of taking a particular genetic test by these means. Indeed, even if the ‘worried well’ are not actually misled by the company’s sales procedures, they will probably not be clear as to how they might benefit from the genetic test.

One Greek pharmacy group has achieved a degree of notoriety by promoting genetic tests and selling DNA sampling kits to the public over the counter while other pharmacies are generally willing to order these sampling kits upon request. We have made enquiries with several pharmacies about the demand for these kits but it would appear that the demand is very low. Again, this finding concurs with the results from our general public survey (Koromila and coworkers, in preparation) indicating that only a

small fraction of the general public would prefer a pharmacist to recommend a genetic test to. The Hellenic Society of Medical Geneticists (<http://www.sige.gr>; content in Greek) have recently published a warning about direct-access testing services being offered by Greek private genetic laboratories using call centers, stressing that these are highly specialized tests whose benefits and results cannot possibly be communicated by phone and by people who have not received the appropriate training. The Society has also stressed that if there were proper regulatory and legal frameworks in place, they would prevent such practices in what is still fortunately a fairly small number of genetic laboratories. It is noteworthy that the first law on the regulation of the practice of medical genetics was passed by the Greek Parliament back in 1980, but it has never been implemented. Cost-wise, the provision of genetic testing services is reasonably cheap with the majority of genetic tests costing less than 300 EUR, and in certain cases below 50 EUR. Bearing in mind the continuously falling costs of genetic analyses, the current profit margins of these laboratories are likely to be still high and hence there is considerable room for price reductions in the future (at least in a truly competitive market).

From our current study, it seems quite evident that the provision of genetic services in Greece has not yet benefited from any central planning, nor has it yet acquired an appropriate regulatory framework. This of course may resemble the situation pertaining in other European countries, although for some of them (like the UK, Germany, the Netherlands and several others), genetic testing is well organized. There are no professional guidelines concerning quality assessment of genetic services in Greece, although recently, more optimistically, many molecular genetic laboratories have joined quality assessment schemes for genetic disorders organized by the European Molecular Genetics Quality Network [EMQN; <http://www.emqn.org/emqn/Home>;



Dequeker et al., 2001]. Despite this, very few Greek genetic laboratories have been accredited with an ISO-15189 or ISO-17025 while other genetic laboratories are accredited with an ISO-9001 or equivalent, and hence are not properly certified for genetic testing.

In essence, our study provides the basis for a critical appraisal of the private genetic testing environment in Greece and provides a model for replication in other European countries to assess the landscape of genetic testing services.

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The authors declare no competing financial interests.

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## Figure legends

### Figure 1

The various types of genetic testing analysis offered by private genetic laboratories in Greece (**A**) and their relative proportions in terms of the demand for these tests (**B**).

### Figure 2

Overview of the main target group of private genetic laboratories in Greece. **A**.

Summary of the key medical specialties that refer patients to private genetic laboratories for genetic testing. **B**. Relative proportions of the age range of patients undergoing private genetic testing in Greece.

### Figure 3

Relative proportions of the main DNA sources used for genetic analysis; \*: Refer to text for details.

### Figure 4

Marketing channels employed by the private genetic laboratories to contact their main target groups, namely physicians (**A**) and the general public (**B**); \*: Refer to text for details.

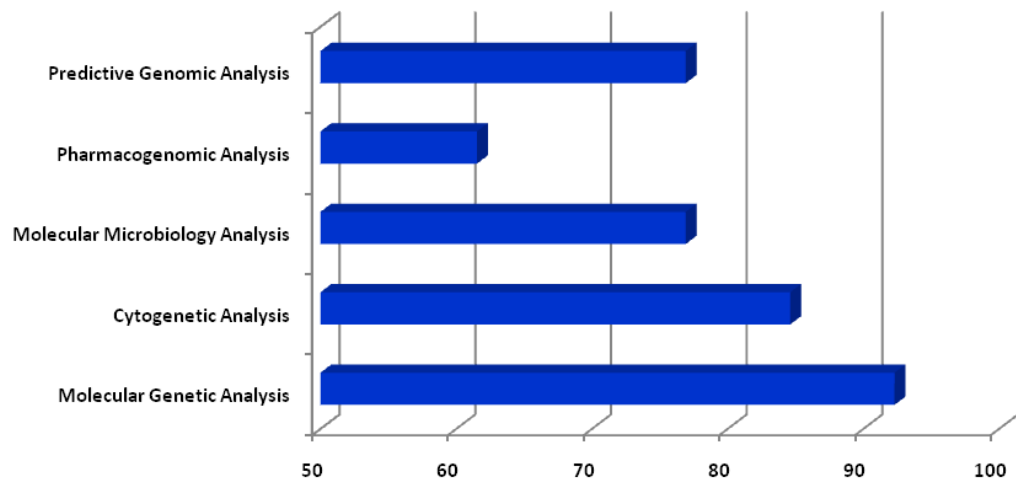
### **Figure 5**

Breakdown of the costs of genetic analysis by type evaluated in this survey.

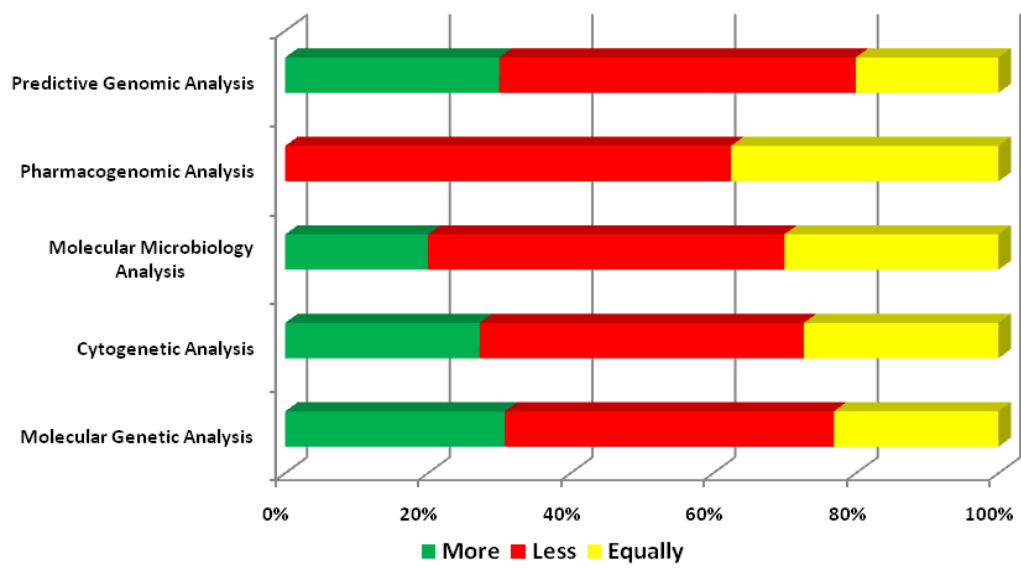
### **Figure 6**

Relative proportions of the main types of drugs for which pharmacogenomic tests are offered by private genetic laboratories; \*: Refer to text for details.

**A**

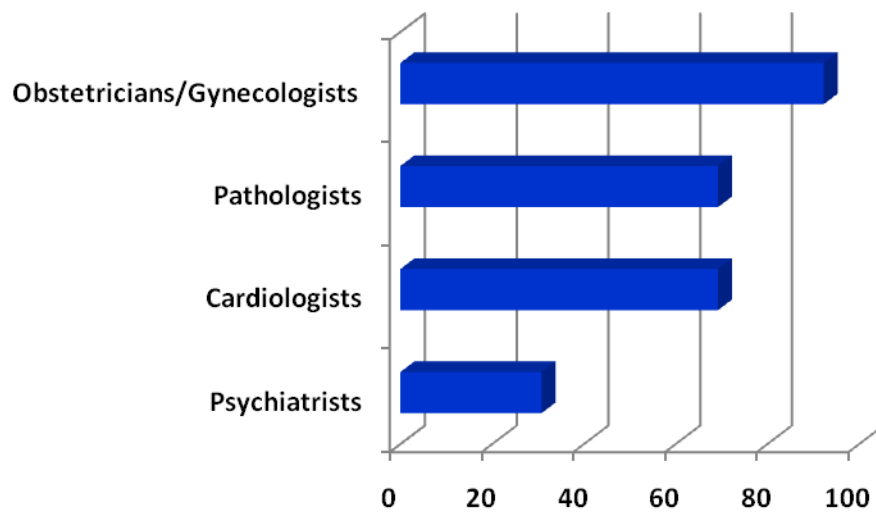


**B**

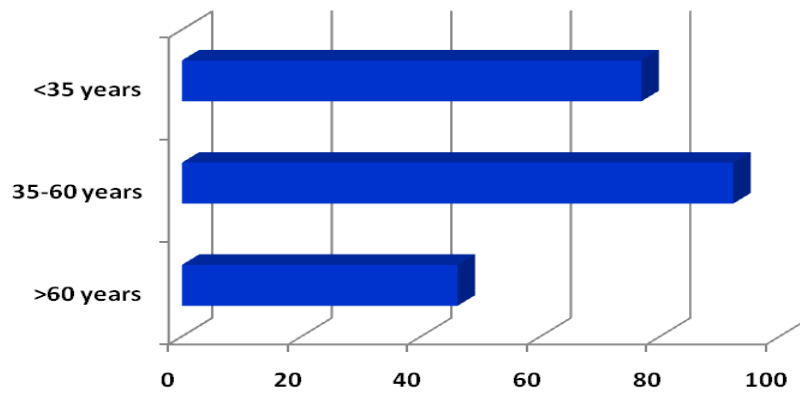


**Figure 1**

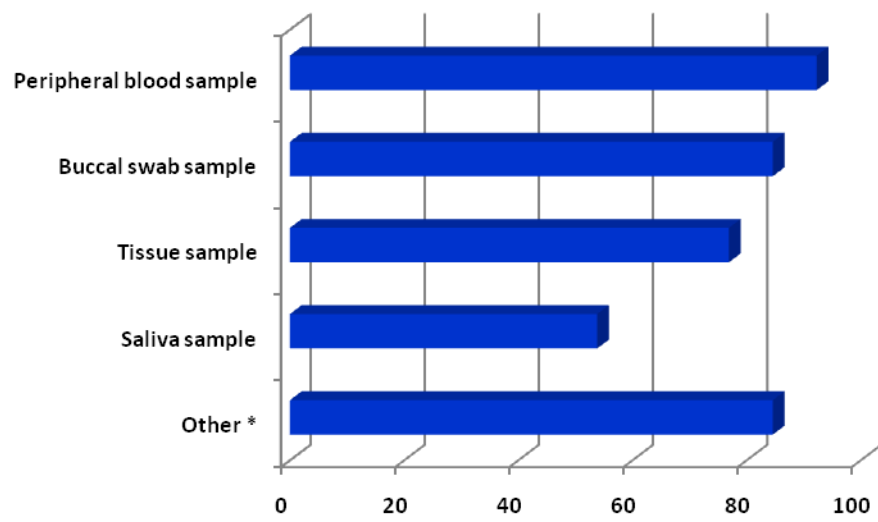
**A**



**B**



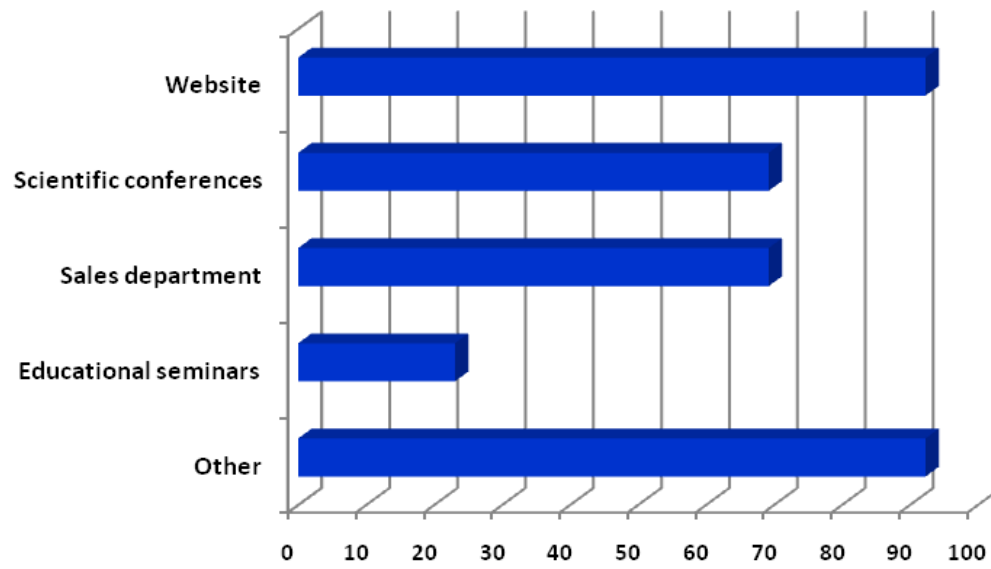
**Figure 2**



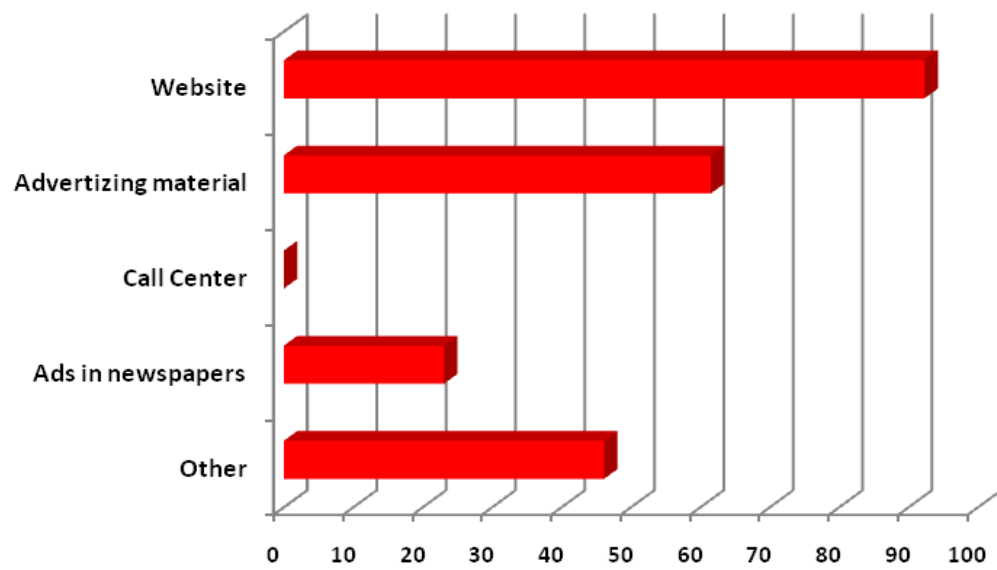
**Figure 3**



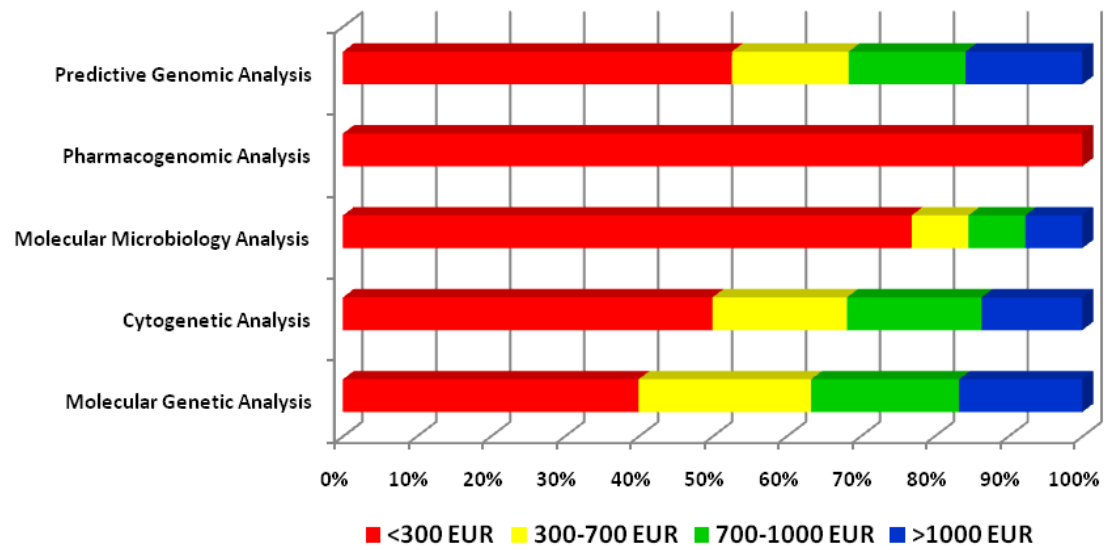
**A**



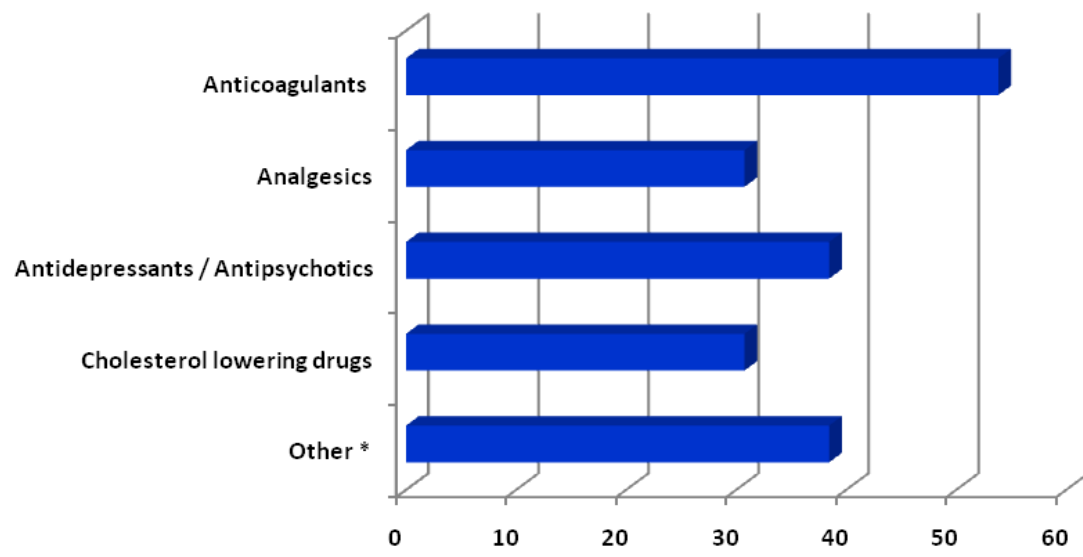
**B**



**Figure 4**



**Figure 5**



**Figure 6**

**Table 1**

Locations of the private genetic laboratories invited to participate in the study.

	Number of laboratories		
City	Invited	Responded	%
Athens	12	10	83.3
Piraeus	2	0	0
Thessaloniki	1	1	100
Patras	1	1	100
Alexandroupolis	1	0	0
Heraklion	1	1	100
<b>TOTAL</b>	<b>18</b>	<b>13</b>	<b>72.2</b>