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‘It’s much more grey than black and white’: clinical geneticists’ views on the oversight of consumer genomics in Europe

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Abstract

Aims: Direct-to-consumer (DTC) genetic tests (GT) have created controversy regarding their risks and benefits. In view of recent regulatory developments, this article aims to explore the attitudes of European clinical geneticists toward the oversight of DTCGT.

Materials & Methods: Fifteen semi-structured interviews were performed with clinical geneticists based in ten European countries. The transcripts were thematically analyzed in an iterative process.

Results & Conclusions: Respondents strongly supported quality standards for DTCGT equal to those applied within the healthcare setting. Despite participants unanimously considering the involvement of healthcare professionals to be important, mandatory medical supervision was controversial. In this regard, promoting education and truth-in-advertising was considered as being key in maintaining a balance between protecting consumers and promoting their autonomy.

Keywords: Direct-to-consumer, genetic testing, medical supervision, genetic counseling, regulation, in vitro diagnostic medical devices, Europe

1. Introduction

Since the beginning of the 2000s, commercial companies have been offering a broad variety of genetic tests directly to consumers, allowing them, in many cases to completely bypass healthcare professionals [1]. Direct-to-consumer (DTC) genetic tests include a wide variety of both health-related and non-health related genetic tests [2], ranging from susceptibility tests for Alzheimer's disease to tests revealing how much Neanderthal DNA an individual may carry.

DTC genetic tests have been criticized, among others, for their often limited clinical validity and utility and concerns have been raised regarding the potential harmful healthcare decisions made based on misconceptions or erroneous interpretation of test results by consumers [3]. Furthermore, the regulatory framework governing such services is currently complex and often unclear [4]. When it comes to the regulation of DTC genetic tests, there are several aspects that need to be considered, including the accuracy and clinical relevance of DTC genetic tests as devices, as well as the quality of the broader context within which they are provided. The latter includes the advertising of tests, the information provided along with them, as well as pre- and post- test genetic counselling and medical supervision.

Over the past years there have been extensive debates on both sides of the Atlantic regarding the appropriate way of regulating such services and whether consumers have a right to access their genetic information directly [5], [6]. In the US, recent regulatory developments were mostly focused on the clinical validity of the tests. More specifically, in November 2013, the Food and Drug Administration (FDA) sent a letter to the DTC company 23andMe requiring

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the company to halt the offer of its Personal Genome Service (PGS) until they obtain a market authorization. In the years that followed, 23andMe obtained authorization from the FDA, first, to market tests for Bloom syndrome carrier status [7], and recently genetic tests providing risk information for certain conditions including Parkinson's and Alzheimer's disease [8]. Especially concerning the latter authorization, the FDA stressed that the "tests are intended to provide genetic risk information to consumers, but the tests cannot determine a person's overall risk of developing a disease or condition." However, its decision has not been well-received by the American College of Medical Genetics and Genomics (ACMG), as it contravenes its guidelines, according to which medical supervision and genetic counselling should be an integral part of the testing process [9], [10].

On the EU level, a recent regulatory debate touched upon both matters of performance of genetic tests as well as issues of medical supervision and genetic counselling [11]. As a result of this debate, the recently adopted EU Regulation 2017/746 on In Vitro Diagnostic Medical Devices (hereinafter IVD Regulation) sets higher standards for the performance of genetic tests, in terms of clinical validity compared to the existing regulatory framework. Nevertheless, a proposal of the European Parliament to impose mandatory medical supervision and genetic counselling for a broad range of health-related genetic tests was not adopted [12]. A possible reason for this was that such provisions stirred controversy among stakeholders and were criticized by some as being overly burdensome and a potential interference of the EU in the clinical practice within Member States [11].

Especially with regard to the developments in the US, it seems likely that the case of 23andMe will pave the way for more companies to acquire authorization for tests providing risk

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information, potentially rendering DTC genetic tests more mainstream. In Europe, it is left to be seen how the new requirements will be implemented in practice, while it is open to discussion whether a chance to regulate these services in a more comprehensive way was missed.

These recent regulatory developments have provoked various reactions and have revived the discussion of what is the appropriate way to regulate DTC genetic tests and how to protect public health while respecting people's right to make their own healthcare choices. In the debate regarding the optimal way of regulating such services, the views of some stakeholder groups could provide valuable insight. To date, several studies have explored the views of the general public [13]–[15] and actual users of DTC genetic tests [16]–[20], and to a lesser extent the views of healthcare professionals [21], [22], [23]. However, with regards to the latter, most studies have focused on the perspectives of healthcare professionals based in the US, while the views of European clinical geneticists remain largely unexplored. Considering that in most European countries genetic testing is deeply embedded in the traditional healthcare system and that the commercialization of medicine tends to be less prominent compared to the US, the perspective of European healthcare professionals may be particularly interesting.

In this regard, an explorative qualitative interview study was undertaken in order to explore the experiences and attitudes of European clinical geneticists towards DTC genetic testing and its regulation. To our knowledge, this is the first qualitative interview study with European healthcare professionals regarding DTC genetic testing. This study builds on the results of a survey study performed to gauge, amongst others, attitudes of European clinical geneticists towards DTC genetic tests [24], [25]. This article is focused on the attitudes of clinical geneticists towards the regulation of DTC genetic tests in Europe, while aspects related to the

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experience of the participants with patients having undergone DTC genetic tests are reported in a separate article (*reference has been removed for the purpose of the double blind peer review*). Both articles have followed the same methodology.

In this article we report on three topics relating to how respondents view the need for oversight with regard to: i) the quality of genetic testing services, ii) the (mandatory) supervision by a healthcare professional and iii) the information provided to consumers and the advertising.

2. Materials & Methods

This research project is a follow-up to a survey study aiming to explore the awareness, experiences and attitudes of European clinical geneticists regarding DTC genetic tests (*reference has been removed for the purpose of the double blind peer review*). Firstly, we identified the participants of the survey study who stated that they have had experiences with patients having undergone DTC genetic testing and invited them to participate in this follow up study. Additional participants were identified using the snowballing approach. The inclusion criteria for this study were for participants to be clinical geneticists, to be working in a healthcare institution based in a country where the IVD Regulation will apply (i.e. EU Member States, European Free Trade Association countries and Turkey) and to have been approached by at least one patient who has purchased or was considering purchasing a DTC genetic test. The identified candidates were invited to participate in the study via email.

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The interview guide was developed after an extensive review of the relevant literature. With regard to the part of the study reported herein, questions focused on the opinion of participants regarding the role of regulation regarding the quality of DTC genetic tests; direct access of consumers to genetic tests; the role of medical professionals in this context; and DTC advertising of genetic tests.

The interviews were conducted in person, via telephone or Skype between February 2015 and May 2016. All interviews were audiotaped upon obtaining informed consent from the participants. The audio files were de-identified and transcribed verbatim by the research team. For the purposes of data analysis, an iterative process was followed that combined deductive and inductive methods in order to identify common themes stemming from the interviews [26]. Codes were attributed to these themes to categorize content [27]. In the first part of this process, a subset of the interviews was coded and discussed within a team for validation. The interviews were coded three separate times with intervals among the different coding sessions in order to allow for new themes to emerge.

This study was approved by the Social and Societal Ethics Committee of the University of Leuven in January 2015.

3. Results

Interviews were performed with 15 clinical geneticists based in 10 European countries, namely in Belgium, the Czech Republic, Finland, France, Ireland, the Netherlands, Slovenia, Sweden, Turkey and the UK. In the context of the semi-structured interviews, respondents elaborated

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on a number of themes related to their experiences and attitudes towards DTC genetic testing. Herein we report only a subset of these, and in particular, we focus on those relating to the conditions under which genetic testing should be offered to the public (Table 1).

3.1 Regulation of the quality of genetic tests and services

When it comes to regulating the offer of genetic testing, a predominant theme raised by most participants was the necessity to ensure the high quality of genetic tests offered. With respect to this, most participants mentioned that the quality for DTC genetic tests should be as high as those imposed on the tests offered via the healthcare system.

We see genetic tests in health setting as one issue. There is not a difference if it is practiced in the public health or commercial sector. The testing should fulfil the professional criteria that have been set long ago, for example by the Council of Europe and by other institutions, and in some countries by law. So this is the issue I believe.

(Interview 11)

Many participants highlighted that clinical validity and utility of genetic tests should be an essential condition for offering tests to the public. In addition, some supported that the information and counselling accompanying a genetic test should also be of the same quality as that offered in the clinical setting

[It should be ensured] that tests are offered with good counselling, that your laboratory doesn't make mistakes and that the quality of testing, how it is offered and how it is implemented is well done"

(Interview 5)

3.2 Regulation of medical supervision of genetic tests

Many respondents considered that some form of mandatory medical supervision and genetic counselling would be desirable for most genetic tests, but not as a blanket requirement for all tests. In this regard, many participants made a distinction between recreational and health-related tests. Some participants supported that when it comes to the former, consumers should be free to access them, as they can be entertaining and educational and they are less likely to cause any harm. However, when it comes to health-related genetic tests, most participants tended to be skeptical regarding whether consumers should be free to access them directly. This was particularly the case as many participants considered that, without appropriate guidance by a healthcare professional, health-related tests could have a negative impact on the health of consumers.

With regard to this, one participant explained:

I think if all that was being offered was what I would call recreational non-medical genetics, (...) I wouldn't be too worried if people were accessing that directly without going to a healthcare provider. What worries me is when people are accessing medically significant, potentially actionable

information without a healthcare provider, because I think there is a serious risk that they will misunderstand the significance of the results they get and most importantly of all, misunderstand the significance of a negative result which could well be a false negative result.

(Interview 2)

Opinions regarding the desirable degree of healthcare professional involvement in this process varied. Some participants favored a restrictive approach, according to which all health-related genetic tests should only be provided by genetics centers with appropriate pre- and post-test genetic counseling. Others stated that the above mentioned restrictions should be imposed only with regard to genetic tests that provide information with a medical significance for the clinical care of the patient, in contrast to tests that might not have a medical relevance. In this context, some participants referred to tests for monogenic disorders.

If somebody is selling a test which has a major clinical impact like a single gene disorder test, then that should be done through a medical practitioner rather than DTC.

(Interview 3)

Nevertheless, it was also acknowledged that it is not always easy to distinguish between health related and non-health related genetic tests.

“It's always very easy in [theory] to say medical or not medical but there is an awful lot that falls within a slightly grey area and I think it probably won't

be quite as easy to distinguish medical from non-medical as we hope it might be.”

(Interview 2)

With regard to the type of professionals supervising this process, various respondents mentioned that it does not necessarily need to be a medical doctor, but, depending on the test, the process could be supervised by a nurse or another qualified medical professional. Some participants considered the involvement of healthcare professionals employed by the DTC genetic tests company to be sufficient, while others were concerned by a potential conflict of interest.

[There should be pre-test genetic counseling] because then the client could make an informed decision whether they should go for the test or not, and understand the results better. And also understand that the majority of the results getting back are not related to any kind of health for themselves. But that will not happen, because I'm not going to be paid by companies saying that their test is actually [meaningless]. [...] I don't think those two things can be combined.

(Interview 4)

Nevertheless, many of the participants opposed the idea of banning DTC genetic testing outright by imposing mandatory medical supervision and genetic counseling for all tests. While all participants expressed concerns regarding DTC access to genetic tests, some stated that imposing a ban on DTC genetic tests may be paternalistic, as consumers should be the ones

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deciding what type of information about themselves they would like to have and how to spend their money. In this regard, one participant stated:

So now we are saying that, okay, you may have an interest in your health but look, this is the big brother who tells you that you cannot have these tests because we don't think that it's good. Obviously we don't think it's good but you can say that anyway without forbidding.

(Interview 7)

Importantly, many participants stressed that a potential ban would be impractical and potentially impossible to implement. The participants who raised these pragmatic considerations stressed that the global nature of the internet makes the strict implementation of relevant national or European regulation particularly challenging.

But whatever we do we're not going to be able to effectively ban all forms DTC genetic testing because it will always be available somewhere and people will post their samples to wherever in the world it is made available. So we have to be pragmatic and acknowledge that there are some things that we can't stop.

(Interview 2)

Another related concern voiced during the interviews was that stringent regulation may increase bureaucracy and impede the clinical application of new developments in genetics.

So I think it's very nice to have laws that stimulate good work, but if the same law is hampering and delaying new developments then I think it doesn't serve the purpose that you want it to serve.

(Interview 5)

Considering the potential impracticality of imposing mandatory medical supervision and genetic counseling on genetic testing, many participants proposed alternatives in order to minimize potential harm to consumers without being overly restrictive. Such alternatives included the adoption of guidelines, encouraging people worried about their health to visit genetic clinics, promoting genetics education in the population and regulating their advertising.

With regard to the adoption of guidelines as the optimal way forward, one participant stated:

There didn't seem much point in just trying to influence the law [...] when it was becoming perfectly apparent that people could access these tests over the internet and get hold of them from anywhere in the world. So we've decided that it might be more helpful to write a series of guidelines that could be used in any country and then allow the individual countries to decide if they wanted to regulate how they wanted to do that.

(Interview 2)

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3.3 Information and advertising

Informing consumers regarding the risks, benefits and limitations of genetic testing was mentioned by many as a more realistic and efficient means of protecting consumers compared to a legal ban of DTC services.

More specifically, it was mentioned by some that, due to the complexity of genetic information, the understanding of genetics among the general population, but also among non-specialized medical professionals is rather low. With respect to this, many participants stressed the need for education for consumers as well as for other relevant stakeholders to better understand the possibilities and limitations of genetic testing. It was mentioned by some that such education should be incorporated in school curricula early in the education process. In addition, some participants mentioned the need of geneticists themselves educating the public by communicating relevant information through mass media. In the same context, it was also underlined that people should be encouraged to visit genetic clinics and use existing services in case they are worried about their health.

So if we can't stop DTC genetic tests and we can't regulate them, then perhaps it is more effective to encourage people to take up existing services that are better structured and more supportive and make sure that they know that those services exist.

(Interview 2)

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However, the practical difficulties of reaching out to the public and ensuring effective education were also acknowledged. In this context, it was stated that the vast amount of information being available to consumers by many different sources can be confusing. In addition, it was mentioned that due to lack of time and resources geneticists cannot carry the burden of educating the public by themselves and have to be supported by the state and the education system.

Similarly, in the context of placing emphasis on the ability of consumers to make informed decisions regarding genetic testing rather than banning DTC access to such services, many participants stressed the importance of truthful and balanced advertising. When asked, many respondents mentioned that they consider regulating the advertising of DTC genetic tests as a being key to protecting consumers from misleading claims and invalid tests, and at the same time empowering them to make informed decisions about their health and their access to genetic information.

So in the ideal world it would be nice and there are all these discussions about the IVD Directive, where you know, they try to have mandatory counselling everywhere, but it is not practical, it's not doable, that's the thing. So you need proper advertisement, you need to have educated public, you need someone I would say for campaigns, showing the pros and cons of genetic testing and not only these smiling and shining actors who are on TV saying how genetic testing changed their lives and so on. So it would be great [to ban DTC genetic tests], but in reality it's much more grey and not just black and white.

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(Interview 12)

Some respondents consider the way DTC genetic tests are currently advertised to be misleading and even manipulative. Some mentioned in particular that the information on the companies' websites is often inadequate.

But I find some of the [company name] advertises very manipulative and not mindful enough of the real limitations of the test they offer. They give the impression that if you have this test you're all clear to have a baby, for example.

(Interview 6)

Many participants stressed the need to ensure fair and balanced advertising of genetic tests that will accurately present the possibilities as well as the risks and limitations of each test. In this regard, some participants underlined that, given the potential health implications that genetic tests may have for consumers, additional restrictions compared to common consumers' goods may be necessary.

So you need some legislation saying, okay, if you're offering some kind of tests influencing someone's health, then it's not allowed to say something that's not [accurate]... If you're buying a car, buying a new machine etc., okay, you're spending money but that's about it... But this is about the chance of you getting ill and that should be regulated on a different level.

(Interview 4)

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Some participants made a comparison with the way food and alcohol advertising are regulated in their countries, considering that respective additional restrictions should be imposed for DTC genetic tests, taking into account their potential health impact.

I think they have to be checked in a way that you cannot claim that you have benefits or anything... Maybe what they do in food stuff as well. If you claim that a meal is healthier than anything else then you have to prove it... I would compare it to the food, the health claims in nutrition. Because it's scientifically proven and it should be solid scientifically and that would be good.

(Interview 15)

Others supported that DTC genetic tests should have a clear disclaimer clarifying that such tests are not medical and that they cannot form the basis for healthcare decision-making.

If you want to go to a palm reader you can go to a palm reader and I don't think the state should ban all palm readers because it's nonsense. Most people when they go to a palm reader they realize what it is and they don't take it as a hard fact. The same should apply to this sort of tests. It should be very clear, it's not a medical test in the sense that we understand it which is the duty of the doctors and maybe the health insurance companies and then it's really up to the people if they want to go and pay a sum of money

[...] to have a little fun with their genetics. But it must not be regarded as a medical test.

(Interview 14)

However, some participants thought that due to the complex nature of genetic tests and of the information they provide, it is not possible to communicate adequately their limitations and risks. As a result, one participant favored a ban on DTC advertising of genetic tests.

4. Discussion

The optimal way of managing DTC genetic tests has been discussed by the participants. All participants indicated that some form of regulation oversight is necessary for DTC genetic tests. Nevertheless, the opinions of participants regarding the extent of regulation and the area of focus varied.

Many participants mentioned that DTC genetic tests should adhere to the same quality standards as those applied within the healthcare system. Although most participants referred to their national public healthcare system as a gold standard, it should be acknowledged that currently there are disparities in the quality standards for genetic testing across Europe. On the EU level, Directive 98/79/EC on in vitro diagnostic medical devices (hereinafter IVD Directive) is currently regulating the quality and efficiency of IVD medical devices (including genetic tests) entering the European market. Nevertheless, the Directive has been implemented differently across EU Member States and has been widely criticized for not offering adequate safeguards when it comes to the quality of genetic tests [28], [29]. This is because currently, most genetic tests can enter the European market after being subjected only to a self-

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certification from the manufacturer. In addition, the IVD Directive has been interpreted by many as only covering the analytical and not the clinical validity of devices [30], setting the bar for quality rather low.

A greater level of harmonization regarding the safety and performance of genetic tests across Europe is expected to be achieved upon the implementation of the IVD Regulation in 2022. Regulations, as opposed to Directives, are directly applicable to the EU Member States, minimizing inconsistencies in their implementation across Europe. The IVD Regulation, which will replace the IVD Directive, sets higher standards for clinical evidence required to demonstrate compliance with the essential requirements of the Regulation.

It is likely that by explicitly requiring evidence of clinical validity in the pre-market assessment of genetic tests, the Regulation may raise the bar of quality for genetic tests in Europe and may make it more difficult for many susceptibility tests to be legally marketed in Europe. However, it is worth stressing that the clinical evidence required in this regard is limited to the intended purpose of the device as stated by the manufacturer. This constitutes an important limitation on the impact of the Regulation, as many genetic tests could be used for purposes broader than the manufacturer's claims without being supported by the appropriate clinical evidence.

Going beyond the regulation of DTC genetic tests as devices, participants elaborated their positions on the regulation of the broader context in which these devices are offered. Regarding mandatory medical supervision, many participants were of the opinion that this would be a desirable regulatory option for health-related tests. Medical supervision was interpreted by most participants in a broad way, which encompasses both the provision of genetic tests upon medical prescription and genetic counselling. Given that health-related genetic tests can

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encompass a wide variety of tests, some participants specified that the degree of medical supervision (i.e. for example whether both pre-and post- test counselling should be mandatory) should be proportionate to the health impact a test may have. In this regard, some participants mentioned that increased supervision should be in place only for medically significant genetic tests. Medical supervision for medically significant test has also been supported by several guidelines [31], [32] and has been favored by healthcare professionals and patients participating in previous studies [24], [33], [34].

However, as also stressed by one participant, the line between what is considered to be a health-related and what a lifestyle test is often blurry. While there are tests that are clearly not meant to advise healthcare management, such as ancestry or paternity tests, there are other tests, that while they don't primarily aim to diagnose a disease and guide clinical decision making, they can still deliver claims that are health-related. This is the case, for example, with nutrigenomics tests that provide dietary advice. In addition, many companies offer different “packages” of genetic tests, providing at the same time both health-related and non-health-related information.

Scepticism towards direct access of consumers to specific types of genetic tests was reported in a previous study with European clinical geneticists conducted by Howard and Borry [24]. In this study, the majority of participants supported that a subset of DTC genetic tests should be banned. Two other empirical studies in patient groups also indicated that patients tend to be sceptical regarding the provisions of DTC genetic tests and tend to favor the option of mandatory medical supervision [33], [34]. The findings of the above mentioned studies may indicate that healthcare professionals and patients, being more familiar with the healthcare

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context, tend to value it more and may be less likely to support access to health-related genetic testing without the involvement of a healthcare professional.

At the same time, some empirical studies indicate that DTC genetic testing consumers tend to favour increased access on genetic testing and be less supportive of increasing government regulation in this field. More specifically, a survey study by Gollust et al. [35] on 941 consumers of 23andMe and Pathway Genomics reported that the vast majority of participants (80%) believed that individuals have the right to directly access their genetic information. Furthermore, a minority of participants (28%) were in favor of government regulation on DTC genetic tests and only 14% favored restricting genetic testing to a clinical setting (however, the study also reported that consumers that had a negative experience with such tests were more likely to favor the involvement of healthcare professionals in this context). Similarly, a survey study performed by Bollinger et al. on 1.047 consumers of 23andMe, Navigenics and deCODEme regarding their attitudes on regulation, revealed that, by a majority of 66%, consumers supported that DTC genetic tests should be made available without governmental oversight [19].

Either as a way of compensating for the potential absence of a healthcare provider or as an additional safeguard, setting high standards for quality of DTC genetic tests advertising and the information accompanying them was a prevalent theme in our study. Accurate and adequate information is a cornerstone in exercising autonomous decision-making. In the context of DTC genetic tests, it is only by understanding the potential, risks and limitations of such services, as well as the existing alternatives, that one can take a truly informed and autonomous decision regarding the type of genetic information they want to receive and their healthcare

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management. Research on the public's understanding of genomics is, to this day, rather limited [36]. Despite the lack of conclusive evidence in this regard, several studies suggest that the general public in western countries are able to acknowledge the fact that disease may be the result of several genetic and non-genetic factors [36]. When it comes to actual consumers of DTC genetic tests, some studies indicate that, on average, consumers exhibit an overall accurate understanding of DTC genetic testing results [14], [18], [37], [38].

Ensuring balanced advertising as well as adequate and accurate information accompanying the tests could have been a way of ensuring the informed consent of consumers. Nevertheless, advertising, is, by its very nature, promotional and, therefore, biased [39].

In this regard, most respondents were of the opinion that currently information conveyed through advertising of DTC genetic tests is not always accurate and balanced. This view is also supported by a study performed by Singleton et al. [40], which performed a content analysis on the websites of 23 companies. According this study, the statements regarding the benefits of their products, on those websites, outnumbered statements on risks and limitations by 6:1 ratio. This tactic, according to the authors, may lead consumers to acquire an inaccurate understanding of risks and benefits of such tests and take ill-informed decisions [40], [41].

Considering the inherent bias in the messages conveyed by advertisements, some respondents stressed that genetic testing should not be seen as a common commercial good and that additional safeguards, akin to those applied in alcohol or food should be in place. If the paradigm of food and alcohol advertising, as regulated in most European countries, is followed,

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this would signify restrictions in the volume and means of such advertising, as well as limitations in its content, in order to protect vulnerable groups, especially children [42], [43].

Taking into account the potential health implications of genetic testing, even stricter approaches have been supported by stakeholders in the past years. In line with the approach followed for prescription drugs, the DTC advertising of which is forbidden in Europe, Eurogentest has argued in the past in favor of allowing DTC advertising of genetic tests only for certain types of tests, while for others advertising should be addressed only to healthcare professionals [44]. Going one step further, during the discussion on the adoption of the IVD Regulation, the European Parliament had proposed a complete ban on DTC advertising of all genetic tests with a direct or indirect health purpose [12]. Even though this proposal was not integrated in the final text, the Regulation, for the first time includes an article on claims and misleading advertising for IVD medical devices [45]. This is the first time that advertising of medical devices is explicitly addressed by EU Regulation and this may be considered an acknowledgment that the advertising of such devices is worth particular attention [39].

Overall, promoting genetics education in school and University curricula and raising awareness in the general public was seen by many as way of striking a balance between protecting consumers and not overly restricting their autonomy.

5. Conclusions

Kalokairinou, L., Borry, P., Howard, H.C. (2020). 'It's much more grey than black and white': clinical geneticists' views on the oversight of consumer genomics in Europe. *PERSONALIZED MEDICINE*, 17 (2), 129-140. doi: [10.2217/pme-2019-0064](https://doi.org/10.2217/pme-2019-0064)

This study reveals through qualitative data that the clinical geneticists who participated are very sceptical of the quality and benefits of DTC genetic testing; they are concerned about their potential negative implications on the health of consumers; and they tend to favour strict oversight of such services. Almost in their totality, participants supported high standards for analytical validity, clinical validity and clinical utility, as well as for the quality of information provided along with the tests and for their advertising. However, even though all participants consider the involvement of a healthcare professional of vital importance, they were divided as to whether mandatory medical supervision would be desirable taking into account pragmatic obstacles and the right of choice of consumers. Similarly, an absolute ban on DTC advertising or access to all DTC genetic tests was not deemed desirable by the majority of participants, although strict control in this regard was favoured.

Conflict of interest

The authors declare that there is no conflict of interest.

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Table 1 Summary of the views of participants regarding the oversight of DTC genetic testing

Regulation of the Quality of Genetic Tests

- Quality as high as for tests within the public healthcare system
- Clinical validity and utility of tests may be precondition

Regulation of Medical Supervision

- No consensus regarding the types of tests for which supervision should be required:
 - For all health related tests
 - Only for medically significant tests
- No consensus regarding the desirability of a legal ban on DTC genetic testing:
 - Desirable for some genetic tests
 - Paternalistic
 - Impractical and potentially impossible to implement
- Alternatives banning DTC genetic testing by law:
 - Adoption of guidelines
 - Promotion of genetics education
 - Regulation of the advertising of genetic tests

Information and Advertising

- Empowering consumers to make own decisions by informing s them regarding risks and benefits of DTC genetic testing
 - Promotion of education
 - By incorporating genetics in school curricula at an early stage
 - Through public outreach of clinical geneticists
 - Raise awareness on genetics among non-specialist healthcare professionals
 - Practical difficulties of educating the public
 - Vast amount if information from different sources
 - Experts do not have the time or resources to inform/teach
 - Need for balanced and truthful advertising
 - Legislation may be a key in protecting consumers from invalid tests
 - Current advertising practices often misleading
 - Need for stricter regulation compared to common consumer goods
 - A disclaimer for some tests clarifying that they should not be used for medical decision-making
 - Challenges in adequately communicating the limitations and risks of genetic tests acknowledged
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