Promotion and sales of self-tests on the Internet

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16.1 Introduction

Currently, the field of medicine is confronted with new developments that allow individuals to access health information outside of a context where healthcare professionals are involved. In a recent report, the Nuffield Council on Bioethics identified this phenomenon as ‘online medicine’:

developments in digital technology, largely involving the internet, that offer new ways for individuals to obtain and share health advice, diagnosis and medication, and that provide new possibilities for storing, accessing and sharing health records, monitoring individuals’ health status and communicating with health professionals and other patients.

(2010: 22)

This definition covers developments such as online health information, online personal health records, the online sales of pharmaceuticals, teledmedicine, direct-to-consumer body imaging and personal genetic profiling (Nuffield Council on Bioethics 2010).

In this chapter, we focus on one specific type of development that was grouped under this umbrella of ‘online medicine’. In particular, we discuss the increasing offer of tests that are available and that allow consumers to measure or identify a particular disorder, risk factor or trait based on body material such as blood, saliva, urine or faeces (Grispen et al. 2011). As such, this chapter will focus on direct-to-consumer (DTC) testing that is initiated, and often interpreted, without the involvement of a healthcare professional.

As described in a Dutch report, various types of test offers are currently being sold (Weijden et al. 2007). The first type involves tests for home use, in which the consumer uses and interprets the test at home. The second type includes situations where bodily materials are sent to a laboratory which returns the results by post or online (i.e. home-collected tests). In a third type of test, the consumer has his or her sample taken at a laboratory and the results are also returned by post or made accessible online. The fourth kind comprises self-tests offered to consumers through an organization stationed in public areas, such as supermarkets (i.e., street-corner tests). In this case, the results are immediately made available and communicated to the individual, as
for example the national cholesterol test offered by the Dutch Heart Association (Deutekom et al. 2008). These different situations in DTC testing are often described under the same denominator as self-tests because they allow individuals to obtain health information about themselves without the involvement of any healthcare professional (Grispen et al. 2011; Weijden et al. 2007; Ronda et al. 2009; Ryan et al. 2006; Wilson et al. 2006; Ryan, Greenfield and Wilson 2006). Self-tests are described as tests on bodily material that are not undertaken on the advice of physicians; that are purchased and performed by the consumer; and are aimed at tracing a particular condition or predisposition that can lead to the development of a particular condition (Weijden et al. 2007).

The availability of self-tests is widespread and increasingly growing. Kearns et al. described this expanding market: ‘Currently numerous biotechnological institutes are targeting new frontiers in self-testing diagnostic devices that aim to be client-centered, technically robust and financially affordable’ (2010: 200). Already in 2006, Ryan et al. identified more than 100 unique tests in the United Kingdom for over 24 diseases sold by 19 retailers. Based on a questionnaire study among Internet users in the Netherlands, Ronda et al. (2009) reported the use of self-tests for 25 conditions. Recent research by Lovett et al. (2012) identified 127 different DTC medical tests advertised online. Among these are tests that measure markers related to cardiovascular conditions and diabetes; sexually transmitted diseases (such as chlamydia, gonorrhea and HIV); nicotine, alcohol and drug use; or cancers (such as prostate, breast or colon cancer). There are also tests available to check for male or female infertility. Since 2008, various companies have also offered genetic tests online, providing information about different types of traits or phenotypes (Borry et al. 2010). Some offer information about susceptibility to common complex disorders, whereas others provide information about non-disease-related phenotypes like eye colour. Additional tests address the metabolism of certain drugs or provide information about carrier status for autosomal recessive conditions. Furthermore, some companies provide information about genealogy or ancestry. Finally, while some companies analyse specific variants related to one or a few phenotypes, other companies analyse thousands to millions of genetic variants for a large number of different traits or disorders.

In this chapter we will first discuss the advantages and challenges posed by DTC testing, as reported in the literature and in various policy documents and recommendations worldwide. Secondly, we will discuss regulations relevant to DTC testing in Europe, notably the in vitro diagnostic medical devices legislation. We address certain drawbacks to the current regulatory framework and discuss the newly proposed Regulation on in vitro diagnostic medical devices (2012). We also refer to the Additional Protocol to the Convention on Human Rights and Biomedicine Concerning Genetic Testing for Health Purposes 2008 and national legislation enacted by different European countries that addresses genetic testing.

16.2 Policy reports: advantages and challenges of DTC tests

Direct-to-consumer testing has been the topic of various reports, guidelines, recommendations and statements. However, the number of documents addressing the specific subtype of DTC genetic testing clearly outnumbers the documents related to DTC testing in general (i.e. other types of tests offered DTC that are not genetic). The American Society of Clinical Pathology (2005) and the Dutch Society of Clinical Chemistry (2006) both addressed the latter. A national bioethics commission, the National Consultative Ethics Committee for Health and Life Sciences (2004), also discussed the challenges related to self-tests, specifically in the context of HIV.
In contrast to the few documents addressing the offer of different types of non-genetic DTC tests, numerous position statements, policies and recommendations have discussed specifically genetic testing offered direct-to-consumer (Skirton et al. 2012). Various professional societies and colleges have produced such documents, notably the American Society of Human Genetics (ASHG) (Hudson et al. 2007), the American College of Medical Genetics (ACMG) (2004), the European Society of Human Genetics (ESHG) (2010), the American College of Obstetricians and Gynecologists (2008), the American College of Clinical Pharmacology (Ameer and Krivoy 2009), the American Society of Clinical Oncology (Robson et al. 2010), the Human Genetics Society of Australasia (2012), the National Society of Genetic Counselors (2011), the Swiss Society of Medical Genetics (2009), the German Society of Human Genetics (2011) and the International Society of Nurses in Genetics (2009).

In addition, public bodies have also commented on DTC genetic testing, including the Nuffield Council on Bioethics (2010), the Belgian Advisory Committee on Bioethics (2004), the Austrian Bioethics Commission (2010), the National Council of Ethics for the Life Sciences in Portugal (2008), the French National Consultative Ethics Committee for Health and Life Sciences (2004), the Secretary’s Advisory Committee on Genetics, Health, and Society (2010) and the Human Genetics Commission (2003, 2007, 2010). A report was also prepared by the Science and Technology Options Assessment for the European Parliament (2008). Furthermore, the European Academies Science Advisory Council and the Federation of European Academies of Medicine have recently published a report (2012). In Germany, DTC genetic testing has also been discussed in a report by the German National Academy of Sciences (2010).

The existence of more policy documents addressing DTC genetic testing compared to non-genetic tests may reflect a certain type of genetic exceptionalism. Genetic tests are often treated differently than other medical tests because of the perception of various factors, including their potential familial and psychosocial impact, their potential predictive character, the fear of discrimination or stigmatisation based on genetic information and their potentially identifying nature. However, these factors are not present in all genetic tests, and many of these characteristics are also present in non-genetic medical tests. In this way, genetic tests do not differ from other medical tests in absolute terms, but rather can be viewed as having characteristics that exist on a gradual spectrum. As a European Commission Independent Expert Group asserted, ‘Genetic information is part of the entire spectrum of all health information and does not represent a separate category as such’ (McNally and Cambon-Thomsen 2004: 10).

As a consequence, we consider most potential benefits and harms related to DTC genetic and non-genetic tests as similar. Specific concerns differ based on the type of test used rather than based on whether the test is genetic. Therefore the following section offers an overview of the potential benefits and harms attributed to DTC testing based on policy documents that specifically address genetic tests as well as those that apply to general health tests.

Various potential advantages have been advanced in relation to DTC tests. Ease and convenience of access are two of the most obvious benefits described in the literature. Consumers are able to order tests when and where it is convenient for them (Ronda et al. 2009; Lippi et al. 2011; Ryan, Wilson and Greenfield 2010). Moreover, performing a test at home without consulting a general physician or specialist can be quick, confidential and inexpensive (Nuffield Council on Bioethics 2010; Lippi et al. 2011). DTC testing may also enable greater consumer autonomy and empowerment, allowing consumers to monitor their health status preventatively and act upon test results by making healthier lifestyle choices, or by undertaking preventive or therapeutic interventions. The right to access one’s own health information was also acknowledged in various policy documents. As expressed by the Austrian Bioethics Commission:
In principle, it can be argued that each individual has the right to obtain information about his or her state of health in order to take action in the interests of his or her own well-being on the basis of this information … In this sense, access to such tests supports the right to self-determination and the right to independent decision-making.

(2010: 29)

However, in contrast with this attention to autonomy and self-management, the offer of self-tests directly to consumers has also raised various concerns that have ethical and policy repercussions. As highlighted by the ESHG in a report about DTC genetic testing:

Individuals are entitled to health information and genetic information about themselves. However, this right to know must be exercised with due respect for the need to protect the same individuals from inappropriate genetic information and testing.

(2010: 1271)

Firstly, various concerns are being raised about the quality of many tests available and the appropriateness of offering some tests directly to consumers. As the American Society for Clinical Pathology noted:

There is concern among the medical community that tests are being conducted to screen for certain conditions (e.g., expensive total body scans to screen for cancer, a cheek swab test to screen for cystic fibrosis DNA, or an inexpensive cholesterol test that does not screen for triglycerides, an important marker for heart disease risk) in DAT [direct access testing] laboratories that would not normally be ordered by a physician. The concern here is that DAT could result in false-positives or false-negatives, possibly leading to increased health care costs as well as adverse impacts on patient health.

(2005: 2)

Many policy documents cite the potential overstatement of the actual predictive value and clinical utility of the results (ESHG 2010). Therefore, the risk that consumers might misinterpret their test results was a serious concern for most organizations. The International Society of Nurses in Genetics warned against the misinterpretation of results that are returned without a healthcare provider:

These risks include misinterpretation of information or distortion of its consequence to the overall health of the person tested due to the complexity of analytical finding implications and the vocabulary use itself. Misinterpretation of results may also lead to the failure to engage in preventive behaviors because the risk is not adequately presented.

(2009: 2)

Similar concerns revolve around the quality of test results (including accuracy and precision of measurement), the quality of laboratories (including internal and external quality assurance) and the appropriate qualifications and training of laboratory personnel (ESHG 2010).

Secondly, most policy documents recommend that trained and qualified health professionals be involved in testing to ensure that patients receive accurate information and pre- and post-test counselling. For example, the American Society for Clinical Pathology recommends:
For optimum patient health outcomes, ASCP recommends that patients consult with their physician for proper interpretation of test results. Laboratory testing helps better identify a patient’s health status. Clinicians may have access to the patient’s family history and other data that can critically affect test interpretation and can order additional tests to clarify the results or predict risk.

(2005: 2)

With regard to genetic testing, the Portuguese National Council of Ethics for the Life Sciences affirmed:

10. Genetic tests related to health should not be offered without medical indication and personalised supervision, in respect for the principles of beneficence and non-maleficence.

11. In case the test provides or may provide predictive health-related information, it should not be conducted unless genetic counseling is made available before and after the results.

(2008, p.6)

Moreover, other organizations discourage DTC testing without the supervision of healthcare professionals. The ACMG recommended that:

A knowledgeable healthcare professional should be involved in the process of ordering and interpreting a genetic test. Genetic testing is highly technical and complex. A genetics expert such as a certified medical geneticist or genetic counselor can help the consumer determine, for example, whether a genetic test should be performed and how to interpret test results in light of personal and family history. A number of risks can be reduced if a genetics professional is involved in genetic testing. These risks include lack of informed consent, inappropriate testing, misinterpretation of results, testing that is inaccurate or not clinically valid, lack of follow-up care, misinformation, and other adverse consequences.

(2008: 1)

Similarly, the ESHG stated:

The offer of genetic tests providing health-related information, in the absence of clinical indications and individualized medical supervision, may compromise patient health. Key concerns are the provision of sufficient information about the purpose and appropriateness of testing, its possibilities and limitations, as well as the clinical significance of testing. An involvement of independent medical professionals could avoid the waste of money on tests that are clinically irrelevant. In addition, the cost and adverse psychosocial effects of unnecessary follow-up or medical investigations could be avoided.

(2010: 1272)

Various documents also acknowledged that healthcare professionals might not always be adequately trained to offer and interpret new tests and, underlined the need for further education. The ASHG, for example, maintained that ‘professional organizations should educate their members regarding the types of genetic tests offered DTC, so that providers can counsel their patients about the potential value and limitations of DTC testing’ (Hudson et al. 2007: 637).
Thirdly, some organizations warned of the potential downstream impact the healthcare system may incur as a result of consumers requesting additional test interpretation, confirmatory testing and further potentially unjustified clinical interventions (e.g. biopsies, radiology, treatments) based on test results derived from DTC testing. For instance, the American College of Obstetricians and Gynecologists cautioned:

[D]irect-to-consumer genetic testing will create downstream needs for counseling, support, and care for those identified as carriers of genes associated with undesired medical conditions. In many locales, the current health care system is not sufficient to meet those needs.  

(2008: 1494)

The Austrian Bioethics Commission questioned to what extent offering DTC tests raises issues for distributive justice due to this indirect cost: ‘[a]s scarce resources cannot be available to all in equal measure, the distribution of goods must be governed by criteria that enjoy the broadest possible acceptance’ (2010: 31).

Fourthly, a number of documents address the risks associated with testing individuals without their consent or knowledge. The ESHG stated that any ‘service that requires a sample to be collected at home runs the risk of samples being submitted for testing without obtaining proper consent or without even the knowledge of the person to whom it pertains’ (2010: 1272). Indeed, tests that are performed at home could theoretically be done without the knowledge of the person tested. Given that informed consent is a key requirement to carrying out a medical intervention, various documents consider the practice of non-consensual testing unethical and suggest that the analysis of a specimen from third parties without their consent should be legally prohibited. For example, a Report by the Secretary’s Advisory Committee on Genetics, Health, and Society referred to the unclear legal situation in the USA with regard to testing without consent:

Most States do not have laws restricting surreptitious DNA testing, and those that do generally place restrictions only on nonconsensual health-related testing. Ten States have laws that broadly restrict surreptitious DNA testing for both health- and nonhealth-related purposes, such as parentage determination or ancestry. Even where State laws expressly prohibit surreptitious testing, it is unclear that these laws have ever been enforced.  

(2010: 30)

Moreover, various documents raise concerns surrounding testing of minors or persons unable to give informed consent. The Nuffield Council on Bioethics stipulated clear conditions that must be met before children should undergo genetic testing:

In the case of children, given our ethical value of the state striving to reduce harm, we recommend that companies should only analyse the DNA of children if (i) a genetic test meets the criteria of the UK National Screening Committee … and (ii) valid parental consent has been given. For such testing to take place, a condition would need to be serious, the test would need to be precise and validated, and there would need to be an effective treatment or intervention available for children identified through early detection.  

(2010: 161)

Lastly, many policy documents also discuss concerns with regard to inappropriate advertisements. They highlight the need to ensure that advertisements should be accurate and not
misleading and that claims should be transparent and supported by current evidence as well as provide correct information with regard to test limitations, risks and benefits. As expressed by the ESHG:

Research on DTC advertising of prescription medicine has shown that this has created an inappropriate demand for medications. Moreover, it has shown that various advertisements for drugs have been misleading. Overstatement of effectiveness or minimization of risk has led to inadequate or inappropriate changes in medication, diet or lifestyle by consumers. DTC advertising of genetic tests for health-related purposes runs the same risks as DTC advertising of prescription medicine in this regard. Aggressive marketing strategies and slogans for DTC genetic testing might overstate the potential for predictive information of such tests and overrate its future health implications.

(2010: 1271)

Based on the aforementioned concerns, it is not surprising that various organizations have warned against the use of DTC testing. For example, the US Food and Drug Administration (FDA) stated:

Despite the benefits of home testing, you should take precautions when using home-use tests. Home-use tests are intended to help you with your health care, but they should not replace periodic visits to your doctor ... Most tests are best evaluated together with your medical history, a physical exam, and other testing. Always see your doctor if you are feeling sick, are worried about a possible medical condition, or if the test instructions recommend you do so.

(2010: 1)

Similarly, the US Federal Trade Commission, aimed at preventing fraudulent, deceptive and unfair business practices in the marketplace and, to increase consumer awareness, also warned against DTC genetic testing on its website: ‘Some of these tests lack scientific validity, and others provide medical results that are meaningful only in the context of a full medical evaluation’ (2006: 1).

16.3 Regulation

16.3.1 In vitro diagnostic medical devices legislation

At the regulatory level, the self-tests described herein usually fall under the statutory regulation of medical devices. In Europe, three directives regulate medical devices, specifically Council Directive 90/385/EEC on the approximation of the laws of member states relating to active implantable medical devices, Council Directive 93/42/EEC concerning medical devices and Directive 98/79/EC on in vitro diagnostic medical devices. Directive 98/79/EC, which was published in 1998 and which came into force in all EU Member States in 2003, governs the safety, quality and performance of in vitro diagnostic medical devices. Specifically, it outlines the requirements for placing a product on the market (e.g. labelling, analytical and diagnostic performances) and imposes an obligation of post-marketing surveillance. Although generally less burdensome than regimes governing pharmaceutical products, both share a number of key features, including a duty to ensure the safety and performance of healthcare products. Moreover, regulatory authorities may remove existing products from the market should serious problems arise.
The above-mentioned directives regulate the safety and marketing of medical devices in the EU. Each member state must transpose certain provisions of the directives into their national laws. This legal framework is complemented by a variety of non-binding ‘guidance documents’ and ‘implementing measures’. These documents seek to ensure a harmonised approach throughout the EU, and promote a shared approach for manufacturers and notified bodies (typically commercial entities licensed to perform conformity assessments of medical devices) involved in conformity assessments (see below) (Castle and Blaney 2010).

In the current Directive, medical devices are defined as:

any instrument, apparatus, appliance, material or other article, whether used alone or in combination, including the software necessary for its proper application, intended by the manufacturer to be used for human beings for the purpose of: diagnosis, prevention, monitoring, treatment or alleviation of disease; diagnosis, monitoring, treatment, alleviation or compensation for an injury or handicap; investigation, replacement or modification of the anatomy or of a physiological process; control of conception, and which does not achieve its principal intended action in or on the human body by pharmacological, immunological or metabolic means, but which may be assisted in its function by such means.


In vitro diagnostic medical devices are defined as:

any medical device which is a reagent, reagent product, calibrator, control material, kit, instrument, apparatus, equipment, or system, whether used alone or in combination, intended by the manufacturer to be used *in vitro* for the examination of specimens, including blood and tissue donations, derived from the human body, solely or principally for the purpose of providing information: concerning a physiological or pathological state; or concerning a congenital abnormality; or to determine the safety and compatibility with potential recipients; or to monitor therapeutic measures.

(*Directive 98/79/EC, article 2(b)*)

Manufacturers wanting to place a medical device on the market or put it into service must first classify the product according to one of the three risk categories, namely low, moderate or high. Currently, Annex II to Directive 98/79/EC lists a small number of tests that have been classified as high risk (List A) or moderate risk (List B). Only devices listed in Annex II and devices intended for self-testing are subject to a conformity assessment by a third party. These third parties, called ‘notified bodies’, are private commercial companies licensed by national regulators to perform conformity assessments of medical devices, including *in vitro* diagnostic medical devices (Castle and Blaney 2010: 238; Directive 98/79/EC article 15). During this conformity assessment, medical devices receive a ‘CE mark’. The CE mark symbolises that the manufacturer has declared the product to meet all legislative requirements, including safety requirements, and that the medical device has been assessed following the required procedure (French-Mowat and Burnett 2012: S23). The CE mark further indicates that the device ‘can be freely marketed anywhere in the European Economic Area (EEA) without further control’ (French-Mowat and Burnett 2012: S23). This pre-market review is one way to attempt to ensure truth-in-labelling (i.e. the manufacturer’s intended use for the product is supported by the clinical data on the test’s performance as set out in the technical file and summarised in the product label and in
promotional material). Unlike devices classified as moderate or high risk, low-risk devices do not go through pre-market review and only need to be registered. Despite these risk category distinctions, Directive 98/79/EC requires manufacturers to report all serious adverse incidents involving devices, regardless of their risk category, as per the mandatory vigilance procedure ensuring post-market surveillance (article 11).

The DTC genetic tests described in the first part of this chapter are considered in vitro diagnostic devices. However, technically speaking, they are not considered devices for self-testing as described by the directive because individuals must submit samples to a laboratory that then returns the results to the consumers. This does not change the fact that they are self-tests in the general definition of the term.

In 2008, the European Commission held a public consultation concerning the recast of the Medical Device Directives. Another public consultation was held in 2010. Based on these consultations, in September 2012, the European Commission proposed a new legal instrument to replace the current legislation (European Commission 2012a). An amended version of the Regulation was put to a vote in the European Parliament on 22 October 2013. At the time of writing, the proposed regulation was under discussion at the Council of Ministers.

A number of changes from the original Directive have already received sufficient support and will likely remain in the definitive version of the legislation. Major critiques were made to the existing classification system in the Directive, due to inconsistencies in classifying low risk versus moderate or high risk. For example, some tests listed in Annex II, List B, as having moderate risk raise questions of coherence: chlamydia tests were listed but no other tests for sexually transmitted diseases were included; testing for phenylketonuria (PKU) was also included in this list, but there were no other tests for heritable disorders; and prostate specific antigen (PSA) testing was listed, but no other tests for cancer were included. As a consequence of the current list-based system, most devices were not subject to pre-market review.

In response to these problems, the proposed Regulation suggests the adoption of a four-class risk-based classification system based on the Global Harmonisation Task Force (GHTF) model whereby in vitro devices would be divided into four categories ranging from high to low risk, depending on their potential impact on public health and/or the individual patient (European Commission 2012a). All genetic tests (including DTC genetic testing), for example, would fall under class C (i.e. moderate to high risk) and would be subject to pre-market review by a notified body unless they fall under a health institution exemption (European Commission 2012a). This categorisation is significant since most genetic tests to date would generally not fall within Annex II and therefore would not be subject to conformity assessments.

Despite the proposed changes to the Directive, the newly proposed Regulation on medical devices (European Commission 2012b) has been criticised for not sufficiently strengthening the evidentiary requirements on safety and efficiency before market introduction (Storz-Pfennig et al. 2013). In 2013, a group of experts submitted a petition to the European Commission, European Parliament and European Council, requesting that they ‘enforce the rigorous clinical evaluation of medical devices’ (Eikermann et al. 2013: 1; Petition 2013), noting:

Currently, there is no requirement that approval of high and medium risk devices should be based on high quality evidence of benefits that are relevant to patients. We recommended that patient safety should be improved by requiring assessment of short and long term benefits and harms in well designed randomised clinical trials and other high quality clinical studies. Post-marketing surveillance should also be compulsory to ensure that benefits and harms of the device in real world settings are similar to those shown in clinical trials.

(Eikermann et al. 2013: 1)
The debate about these requirements also surrounds *in vitro* diagnostic medical devices in the proposed Regulation regarding the inclusion of the concept of clinical utility. Ultimately, the reason for undergoing (genetic) testing for health-related purposes should be based on the clinical utility of the test. Clinical utility refers to the ways in which the results of testing for a genetic marker (that is known to increase the risk of developing a disease) can be useful in clinical practice (i.e. treatment or prevention options). Moreover, clinical utility informs how results can be utilised to reduce the patient’s risk of developing the disease and the extent to which this information contributes to our knowledge of what should be done to prevent disease.

During the 2010 public consultation on *in vitro* diagnostic medical devices, 67 per cent of respondents affirmed that clinical utility should not constitute part of the pre-market assessment process (European Commission 2012c). Rather, they considered it to be a ‘moving concept’ more effectively regulated at the member state level (European Commission 2012c: 22–3). Respondents also believed ‘clinical utility should not be demonstrated by the manufacturer, but should be assessed by the user. The user would have to decide on the clinical utility of a specific IVD [*in vitro diagnostic*] medical device in a specific context or a specific population’ (European Commission 2012c: 23). Furthermore, respondents stated that ‘it would be impossible to demonstrate the clinical utility and therefore, it will limit the market access for innovative IVD medical devices’ (European Commission 2012c: 23).

Despite this controversy, during the vote of the European Parliament in October 2013, clinical utility was introduced as a performance requirement for IVD devices that has to be taken into account where appropriate (Amendment 204). Even though this development could have a positive impact for public health by deterring medically irrelevant tests from reaching consumers, the proposed Regulation does not include a definition of clinical utility or any criteria for its assessment. Consequently, given the existence of different definitions of clinical utility and the subjective dimension these definitions may entail, as well as the lack of sufficient guidance regarding its interpretation, the new Regulation on IVD medical devices might fail, like the previous Directive, to provide a framework in which the quality of tests is sufficiently assessed before these tests are provided to individuals.

An additional concern focuses on the inadequacy of notified bodies to assess devices before market introduction (Cohen 2012a, 2012b; Godlee 2012). For example, the aforementioned petition urged that

> approval of high and medium risk devices (category III and IIb) as well as *in vitro* diagnostic devices should be done by a new public body similar to the European Medicines Agency or that the EMA is given an extended mandate to carry out these assessments.

(Eikermann et al. 2013: 1)

### 16.3.2 Canalisation of self-tests through healthcare professionals

Currently, Directive 98/79/EC does not permit *in vitro* diagnostic medical devices bearing the CE mark of conformity to be blocked from being placed on the market, but allows national measures to canalise the provision of devices. The Directive does not affect national measures that require a medical prescription for a specific device (Directive 98/79/EC, article 1(6)). However, the proposed Regulation attempts to introduce a change in the provision of genetic tests. During the vote of the European Parliament, a new article was integrated whereby genetic testing would only be performed by a medical professional, after appropriate genetic counselling and informed consent (2013). The approach of canalising genetic tests through healthcare professionals and emphasising the importance of genetic counselling and informed consent seems consistent with the national legislation of several European countries.

More specifically, in the Netherlands, some self-tests (e.g. for HIV) must be canalised through a doctor or a pharmacist. Only after the user receives certain information (i.e. the possibility of being tested anonymously in the framework of medical supervision, the importance of medical supervision if the test is positive, the correct use of the test and the correct interpretation of test results) may the test be provided (Health Council of the Netherlands 2007). This is not considered the regulation of a ‘service’, but rather an integral part of the provision of a ‘good’. This canalisation procedure provides the framework under which the ‘good’ can be provided (Health Council of the Netherlands 2007).

Along these lines, various other European countries have enacted legislation stipulating that genetic testing for health purposes may only be available with medical supervision, informed consent prior to testing and genetic counselling (Borry et al. 2012). For example, the German Human Genetic Examination Act (Genetic Diagnosis Act – GenDG) 2009 requires that diagnostic genetic testing be conducted by a physician and that predictive genetic testing be conducted by a certified medical specialist (section 2 §7). The Act specifies that these tests can only be conducted after sufficient information is given concerning the nature, meaning and consequences of the test, and after having obtained consent. Similarly, French legislation only permits genetic testing in the context of a clinical relationship and integrates specific requirements relating to genetic counselling and informed consent (see Code Civil 2013, article 16-1; Code de la Santé Publique 2013, article R1131-1).

The Additional Protocol to the Convention on Human Rights and Biomedicine Concerning Genetic Testing for Health Purposes contains similar requirements: ‘[a] genetic test for health purposes may only be performed under individualised medical supervision’ (article 7). The Additional Protocol also emphasises the importance of genetic counselling, informed consent, the protection of persons unable to consent, respect for private life and the right to information, as well as the right not to know.

16.3.3 Screening legislation

Some countries have developed screening legislation that may also impact the provision of self-tests. The Dutch Act on Population Screening 1992, for example, provides a legal framework which evaluates a test/examination before it is offered to the population: ‘population screening is defined as “a medical examination which is carried out in response to an offer made to the entire population or to a section thereof and to detect diseases of a certain kind or certain risk indicators, either wholly or partly for the benefit of the persons examined”’ (Borry et al. 2012: 718). For such tests, the Dutch Minister of Welfare and Sports issues a permit. If performed without a permit, tests that detect (risk factors of) cancer and (risk factors of) ‘incurable’ diseases – which can neither be treated nor prevented – are illegal in the Netherlands. Based on article 7, the Minister can refuse a licence if a test is scientifically unsound, if it is not in accordance with the professional medical practice standards or if the potential health risks outweigh the expected benefits. However, problems remain in interpreting and enforcing the Act, notably for self-tests available via the Internet.

16.4 Conclusion

In the first part of this chapter, we discussed the main advantages and disadvantages of health tests offered directly to consumers, as expressed in national and international policies. Although we focused on documents specifically addressing DTC genetic testing, many issues are also applicable
to other types of DTC clinical tests. In the second half of this chapter, we discussed European
laws that regulate various aspects of self-testing and genetic testing, including the European in
vitro diagnostic medical devices legislation and the Additional Protocol to the Convention on Human
Rights and Biomedicine. Together, these two sections provide an overview of the ethical and legal
issues surrounding self-testing and, more specifically, DTC genetic testing.

Looking toward the future, what can we expect from self-tests? As patients are becoming
more engaged in their own healthcare (Rozenblum and Bates 2013) and an increasing propor-
tion of the population can access the Internet, there is reason to believe that more users will
perform self-tests. In fact, some predict that self-tests will become widely used and even more
readily available in years to come (Ronda et al. 2009).

In the narrower realm of DTC genetic testing, realistic or plausible future predictions are
slightly more complicated. In 2010, Wright and Gregory-Jones believed the DTC genetic test-
ing market to be relatively small. In recent years, a number of companies ceased selling DTC
genetic testing services (Vorhaus 2012), while others have changed their policies for providing
genetic testing to involve healthcare professionals (Howard and Borry 2012). Indeed, certain fac-
tors will influence the future of DTC genetic testing including, among others, public demand
and general social acceptance, consensus or advocacy among healthcare professionals and other
stakeholder groups regarding service models and legal regulations that may limit the activities
of DTC genetic testing companies in certain jurisdictions. This last issue was recently highlighted
by the FDA when it limited the activities of the DTC company 23andme (FDA 2013).

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References

testing’, Genetics in Medicine, 6 (1).
American College of Medical Genetics (ACMG) (2008) ‘ACMG statement on direct-to-consumer
Borry, P., Cornel, M. C. and Howard, H. C. 2010, ‘Where are you going, where have you been? A recent
‘Legislation on direct-to-consumer genetic testing in seven European countries’, European Journal of
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Legislation

Code Civil 2013 (France).

Code de la Santé Publique 2013 (France).


Gesetz über genetische Untersuchungen bei Menschen (Genetikgesetz-GenDG) (Human Genetic Examination Act (The Genetic Diagnostics Act – GenDG)) 2009 (Germany).

Wet op het bevolkingsonderzoek (the Dutch Act on Population Screening) 1992 (Netherlands).