

Ethical implications and legislative control of direct-to-consumer genetic testing in Europe

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Since their first commercialization in 2007, the use of Direct To Consumer (DTC) genetic testing (GT) has rapidly increased. From the consumer viewpoint, the possibility to access genetic information without supervision of a medical doctor could allow for free choice and self-diagnosis (1), and in principle leads to an increased attention on the lifestyle choices and environmental stressors.

Currently several issues surround the use of DTC-GT, spanning from the ethical and legal context, to their clinical utility and interpretation of results. Whereas such tests were introduced in the market to predict the risk of monogenic disorders such as cystic fibrosis and Tay-Sachs disease, this testing method is currently turning towards the prediction of complex multi-factorial diseases (2, 3) for which DTC-GT offers less predictive value and preventive clinical options. Additionally, DTC-GT is marketed without the obligation for medical counselling, which has the potential to lead the user to: an overestimation of the risk of developing a certain disease for which a slight increased risk is reported or a false sense of reassurance for the negative results (4). In the worst scenario, a user for which the genetic test for detection of the BRCA mutation is negative, could fail to undergo regular mammographic screening, ignoring that his/her baseline risk is not zero due to interplay of environmental factors in the disease occurrence.

The lack of communication with a physician is an important ethical issue of DTC- GT especially for the diagnosis of non-curable diseases which require a sensitive approach when communicating the test results to the patients (5). Additionally, the marketing of DTC-GT available on the internet can lead to the use of samples from minors or third parts without consent, also for tests with no current benefits and preventive options for children (6). The European Society of Human Genetics stated that when a parent requests to have a child tested for a genetic disease without immediate medical benefits, “an assessment should be made of the balance of harms and benefits”, and also that DTC-GT should be permitted only after the patient reaches the age of majority (7). Lastly, genetic data represents a sensitive data source, which should be safeguarded to avoid social stigmatizations or work and medical insurance discriminations (8). The Oviedo Convention on Human Rights and Biomedicine stated that tests which are predictive of a genetic disease or susceptibility may be performed only for health purposes and subject to appropriate genetic counselling (9). Although several Bioethical groups have provided opinions about DTC (10), the DTC-GT industry defends their products by supporting patient empowerment and the right of each individual to access their genetic information. By arguing that genetic data will be used for research, they manage to bypass the ethical documents and the services of health professionals (11).

Issues surrounding the regulation of DTC-GT are worth discussing. A recently published report from the Federation of European Academies of Science shows a largely fragmented legislation in the European countries (10), and underlines that a common core of good practices and recommendations is currently missing. Although in some countries informed consent and counselling of a specialist before and after genetic testing is mandatory, at the time, no European country has developed specific laws (12) to regulate DTC-GT. In Germany the Genetic Examination Act (13) reported in 2012, that a diagnostic genetic examination

and a predictive genetic examination can be carried out only by medical doctors, and similar limitations have been established in France in 2011, and in Switzerland already since 2004 (14). The Spanish law (Ley 14/2007 de Investigación Biomédica) foresees that “*Predictive tests of genetic diseases shall only be carried out for medical or medical research purposes and with genetic counselling*” (art. 9) (15). The Chapter 5 of the Biotechnology Act of 2003 (16), reports in Norway that genetic testing is limited to medical purposes, specifically for diagnosis or therapeutic aims. As for Belgium, an extensive interpretation of article 2 of the Law on the practice of health-care professions (Royal Decree 78/1967) seems to be the only one legal provision applicable to DTC-GT. Indeed the Belgian law prescribes that a physician should be involved in the practice of medicine, but DTC companies declare that there is no practicing of medicine involved in genetic testing, and that their tests should not be considered medical information but only informational tools (12). In the Netherlands, the Dutch Act on Population Screening issued in 1996, permits the companies to sell DTC-GT if they obtain a proper permit from the Ministry of Health and Sport (17).

In Italy there is no specific law for the regulation of the DTC-GT. The Italian consumers’ code, issued in 2005 and updated in 2012, presents general considerations on the safety of products directly sold to individuals, but there is no particular provision on the selling of DTC-GT in the country (18). The only official document published on DTC-GT is the report titled “*Test genetici di suscettibilità e medicina personalizzata*”, issued by the National Committee for Bioethics in 2010 (19). The Committee outlined in this document some best practices that the Italian legislative body shall follow in order to control the phenomenon. The core of the advices is to minimize the use of DTC-GT for two main reasons: (i) at the actual state of science, it seems that the technology is not precise and reliable; and (ii) a misuse of the tool could lead to useless request for further diagnosis, with an increase in the overall expenditure for the Italian National Health System, with a consumption of resources that would have no clinical justification. Moreover there is a lack of regulations about the safety and quality of implementation of the tests, which should be conducted not only by using strict and standardized methodology, but also with a professional interpretation of results (10). A legislative intervention is needed in the general organization of genetic structures in Italy, which should be rationalized in accordance with the national guidelines, and should be combined with a constant training of general practitioners and education of consumers to the appropriate use of genetic testing (20) and to the importance of a genetic counselling before and after their use.

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