Direct-to-Consumer Genetic Tests

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TYPE STATUS	Study published	LAST UPDATE	October 2021	European countries

PROBLEM & OBJECTIVE

PROBLEM Genetic determinants of multifactorial diseases could be exploited to predict risks in patients/citizens. The increasing availability of DTC-GT is mirrored to the increasing use of them. This could lead to erroneous interpretation of risk, unnecessary worries and clinical investigations based on the test. Eventually, DTC-GT could cause an excessive burden on healthcare systems, wasting scarce resources

OBJECTIVE To provide the landscape of DTC-GT in EU, considering citizens' literacy, healthcare professionals' knowledge and legislative frameworks about DTC-GT, is fundamental to face the emerging challenge for national healthcare system

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KEY COMPONENTS / STEPS

- Direct-to-consumer genetic tests (DTC-GTs) are genetic tests for a medical or non-medical trait that are sold directly to the public, usually ordered without the engagement of a healthcare professional.
- The introduction of DTC-GT could be dated back to 2003 in USA and in about a decade, DTC-GT spread across continents.
- Scientists, professional societies and others have expressed varying views about what and how to regulate with regard to DTC-GT.
- In 2008, they were named "retail product of the year", thanks to the progressive empowerment of citizens and the wide range of applications of personalized medicine, the collapsing costs of required technologies and the shortening of time for each test.
- In 2013 FDA sent a "cease and desist" letter preventing 23andMe, a major company of DTC-GT to sell test concerning medical aspects.
- In 2015, FDA authorized the first medical application of DTC-GT for Bloom Syndrome, and DTC-GTs for common conditions, like Parkinson and Alzheimer and breast cancer, were authorized in the following years.
- The European Society of Human Genetics (ESHG) set a statement identifying the needs for evidence about clinical and analytical validity, utilities, medical supervision.

KEY CONTEXTUAL FACTORS

The rise of DTC-GT exemplifies some of the wider changes affecting healthcare and public health:

- growth of a globalized industry;
- less public deference to traditional, physician-led, professional forms of authority;
- familiarity with the internet; an increasing desire by the individual to have information;
- various pressures to exercise personal choice and responsibility.

A clear uniform regulation of the DTC-GT provision is still lacking. The actual legislative and ethical framework covered aspects about genetic testing in traditional healthcare, but were quite ineffective on global online market.

MAIN IMPACTS / ADDED VALUE

- Several doubts on clinical utility and validity of DTC-GT expressed, but the market is steadily growing.
- EU directive 2005/29 on Unfair Commercial Practices and EU directive 98/79 on in vitro diagnostic medical devices regulated provision of medical devices in European countries.
- The Convention on Human Rights and Biomedicine and its additional protocol on genetic testing is the international instrument to set a basic framework to regulate DTC-GT.
- Several European agencies/societies (EASAC, FEAM, ESHG) released position papers.
- The implementation of genomics in clinical practice is planned in National Plans. Several Member States have more stringent legislation on DTC-GT services.

LESSONS LEARNED

- Legislative framework is fragmented, it is necessary to adopt international guidelines, standards and codes of practice based on greater transparency of information provision.
- It is critically important to address common public misconceptions about what genetic tests can offer in terms of medically relevant information so as to inform and empower the consumer to decide for themselves when faced with offers of DTC-GT.
- It is vital for Europe to do better in educating medical and other health professionals about genetics, for example to improve the confidence of primary care physicians to interpret and explain risk and benefit based on genetic information. Scientific studies have been performed on health professionals' knowledge. Still much unpreparedness to face citizens' needs emerged.
- European citizens, overall, have a low level of knowledge on DTC-GTs and a high interest in their purchase. This understanding might contribute to the development of educational programs in order to the increase of general public capabilities to make appropriate health decisions.
- in order to protect the citizens from incompetent and harmful services, all of the aspects of DTC-GTs should be taken into consideration in the decision processes regarding the regulation of these tests both on national and international level.
- The involvement of multiple-decision makers is required in the decision whether to introduce these tests in clinical practice.

REFERENCES & DOCUMENTATION

- A review of the legislation of direct-to-consumer genetic testing in EU member states - PubMed (nih.gov)
- European citizens' perspectives on direct-to-consumer genetic testing: an updated systematic review - PubMed (nih.gov)
- Internet-Based Direct-to-Consumer Genetic Testing: A Systematic Review - PubMed (nih.gov)
- Statement of the ESHG on direct-to-consumer genetic testing for health-related purposes - PubMed (nih.gov)

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