

EXECUTIVE SUMMARY

At the end of the 1990s, genetic testing offered directly to consumers came onto the market as a new “business model”. Up until then, genetic testing had been carried out by specialised institutes in the medical sector upon referral by a medical doctor. In recent years, new companies offering direct-to-consumer genetic testing (DCGT) via the internet alone are emerging constantly.

This method of “bypassing” the medical sector with its established ethical and quality standards has given rise to concerns regarding an uncontrolled growth of the market for genetic testing. Tests are offered whose clinical validity and utility is doubtful and thus could do harm to consumers who might be misled and insufficiently informed by the DCGT companies' advertisements.

The present report provides an overview of the current discussion on DCGT among experts and public authorities and on the current status of DCGT offers on the internet. Guided by an analysis of the market development and the pros and cons of DCGT, the report discusses possible options and needs for political intervention.

The increasing number of DCGT offers can be regarded as being driven by the following trends that currently characterise genetic testing in general:

- The availability of genetic tests for common diseases and susceptibilities to common diseases represents a promising economic option for companies developing genetic testing assays or kits as well as for companies offering services on a private basis directly to customers.
- Technical achievements such as the development of DNA microarrays reduce the technical and financial barriers to a private market for genetic testing.
- Genetic testing is on its way to becoming an option for preventive medicine in general. It is discussed as a new important public health option, and the perspectives of new applications such as pharmacogenetics and nutrigenomics indicate new business opportunities.

The central difference between DCGT and the standard genetic testing situation in the context of the established system of genetic counselling is the way informational support is (or rather is not) provided in internet offers of testing. It may well be that there is no provision for counselling at all except for the written advice on the webpage. Counselling may be offered as an additional special service at extra costs and at the customer's request. It may also be that a recommendation or at least an offer is given for the customer to contact a doctor or health practitioner from the company via phone for counselling. In other cases, the customer may be recommended to consult his own doctor on the test results. It may also be the case that the entire process follows a standardised non-personal web-exchange procedure. Even the report containing the results of the diagnosis and their interpretation as well as recommendations to the client can be produced by software that automatically combines information from the DNA diagnosis with information read from a questionnaire on the customer's lifestyle.

The most obvious problem of DCGT is that - as is supported by an assessment of 38 DCGT websites carried out in the context of the project - the majority of tests offered to consumers directly are tests for susceptibilities for disease based on so-called SNPs (single nucleotide polymorphisms). These tests are most interesting from a commercial point of view since they are related to widespread common diseases (such as cancer). Experts regard most offers of testing based on SNPs to be meaningless from a scientific point of view, since the clinical validity of most of the tests has not (yet) been sufficiently proven. However, since recommendations that can be drawn (and are drawn by providers) from positive test results usually do not go beyond what a doctor would recommend to any patient as being good for his/her health (e.g. practise sports, avoid fatty foods), some consider offering this directly to consumers to be harmless. Others, however, opine that this kind of testing may harm clients. If results are negative, the client may gain the false impression of being safe with regard to developing a certain disease and might not see the need for adopting a healthy lifestyle; this would be totally misleading, as the absence of "negative" SNPs tested does not imply an absence of the risk of developing e.g. high blood pressure from bad dietary habits, other behavioural and environmental factors or other (so far unknown) genetic traits (that were not tested).

The internet survey supported the notion that,

- many DCGT offers do not meet a minimum set of quality criteria that can be regarded to be necessary for ensuring adequate information and protection of customers against misleading interpretation of the need for as well as the possible consequences of genetic testing,
- most DCGT offers fail to provide proper information on the scientific evidence behind genetic testing services offered to customers (clinical validity and utility),
- many of the companies offering genetic testing services via internet do not include genetic counselling at all in their services. Only a few urge customers to involve an expert before purchasing a gene test, and "counselling" in most cases only is provided as written information via mail or via web-log.

Due to the complexity of genetic information that could well mislead consumers or be used to mislead them, and due to the likely serious health and psychological consequences of this, there is a consensus that principles such as informed consent and quality standards of testing and counselling must be ensured since DCGT offers via the internet can obviously be associated with consumer protection problems. Thus it is widely regarded to be legitimate to regulate the market for DCGT. It is, however, a matter of discussion to what extent governmental intervention is needed, and whether regulations should apply in the same way to all different types or purposes of DCGT services.

At the centre of discussions on possible regulatory interventions, there are two options:

- Statutory restriction of genetic testing to the medical context (e.g. by making the referral by a medical doctor mandatory) could ensure a minimum standard of quality of testing and counselling. This is for instance suggested by the Council of Europe's recently released "Additional Protocol on Genetic Testing" which stipulates that "a genetic test for health purposes may only be performed under individualized medical supervision". It is, however, discussed to what extent all types of genetic testing should be covered by such a regulation or whether "non-risk" tests should be openly available commercially.
- As companies offering DCGT so far are not obliged to provide any scientific evidence regarding the clinical validity and utility of tests offered and as the evidence for many tests is regarded to be doubtful by experts, a system of pre-marketing approval of genetic tests is argued for. The European In-Vitro-Diagnostics Devices Directive which stipulates the marketing of in-vitro diagnostic does not cover genetic testing so far or treats gene tests as "low-risk" devices for which no pre-marketing approval is provided.

At the European level, the following options for policy interventions are conceivable in order to ensure high standards of genetic testing services and to hinder misuse and uncontrolled growth:

- The IVD Directive is currently undergoing a process of amendment. To provide for a broad scope of gene tests being covered by the directive would allow the establishment of a European system of pre-market approval of gene tests which might drastically restrict the leeway for DCGT.
- At the national level, there are discussions of setting up a code of practice for DCGT to ensure minimum quality standards. It must be considered whether such a code could be established on the European level, and could be enforced by monitoring by a European public authority.
- In order to ensure the "technical" quality of testing services, it could be envisaged to establish a European system of control and accreditation of laboratories carrying out molecular testing, as is demanded by guidelines recently published by the OECD.