

1. INTRODUCTION

Genetic testing has been the subject of public and political debate for almost two decades now. The enormous and continuous pace of scientific and technological development in this field of biomedical research and healthcare drives the ongoing discussions of the pros and cons of genetic testing. Genetic testing makes it possible to detect at a very early point in time the genetic traits of an individual that cause serious disease or disabilities for the individual himself or for his offspring, or to detect genetically based susceptibilities which indicates an increased risk of a person for developing a serious disease such as cancer. The new diagnostic options made available by genetic testing can without a doubt be helpful for detecting health risks early in order to initiate medical treatment in a timely manner. With regard to monogenetic inherited diseases, for instance, genetic testing can provide individuals with certainty as to their genetic status and thus about any increased likelihood for them to develop a disease or to pass a genetic predisposition for a disease on to their children. Without testing, persons at risk have to live with an uncertainty of a 25% or 50% risk of being a carrier.

The basic feature that genetic testing adds to medical practice for good (and at times for bad) is its “predictive” character. We gain the ability to know about our (or our offspring’s) genetic status and thus should be able to better predict our health status in the near or distant future.

Diagnostic and predictive options made available by genetic testing - despite their medical benefits - have caused debates about possible negative effects of genetic testing, among which are:

- a) The possible misuse of genetic information by third parties: Cases have been reported about employers and insurance companies discriminating against individuals on the basis of genetic testing.
- b) Information about a person’s genetic status can imply knowledge about the risks of a person’s relatives to carry the same genetic “burden”. This together with information about a person's future (particularly in cases where no therapy is at hand), which is often sensitive and psychologically problematic, has led to demands for a person's “right not to know” about his or her own genetic make-up.
- c) Testing for complex (common) diseases can only provide information about the probability (higher than average risk) of a person with a susceptibility gene to actually develop the disease. The clinical usefulness of testing is therefore considered in some cases doubtful. The only consequence of diagnosis might be to cause psychological damage.

- d) There has been criticism that the availability of more and more genetic testing options in medical practice and the high-flying visions associated with the complete identification of the human genome in 2001 could provide credence to a wrong view of “genetic determinism”, suggesting that most diseases are caused by a person's genetic makeup (and thus neglecting detrimental environmental factors) and possibly leading to a decreased social acceptance of people with disabilities or handicaps, since the availability of genetic diagnostics might make disabilities come to be regarded as avoidable.

When genetic testing first entered medical practice during the mid 1980s, it was restricted to a few inherited diseases, such as cystic fibrosis. Genetic testing and counselling were only offered by experts working at university hospitals and institutes and by a limited number of doctors who specialised in human genetics. The limited number of persons seeking genetic testing and counselling, the quite complex and expensive technical procedure of testing as well as the limited number of well-educated experts who can offer genetic testing and counselling are all factors that have contained the problematic potential of genetic testing. Many of the negative expectations connected with genetic testing were based on the assumption of an uncontrolled growth of genetic testing for a great number of common diseases, which might open the door for misuse and clinically non-indicated applications of testing. Apart from the limited number of tests available, the fact that a small group of medical practitioners and genetic counsellors has controlled the practice of testing has been regarded as guaranteeing a knowledgeable, cautious and responsible application of genetic testing, which contrasted with the negative scenarios of its widespread and clinically doubtful use. In recent years, however, some of the barriers to a growth of genetic testing beyond the “protected” realm of genetic counselling carried out in hospitals for a restricted number of persons who might be carriers of rare inherited genetic diseases have vanished or are losing strength. New technological options are available that make it both technically easier and cheaper for a genetic test to be carried out. Connected with the lowering of the technical barriers to genetic testing is a tendency for new (private) suppliers to enter the market. And last but not least, genetic testing is being offered not only for some rare Mendelian diseases but increasing for common and widespread diseases such as cancer, diabetes or cardiovascular diseases. However doubtful the clinical validity and usefulness of these tests may be, such use has the potential of making genetic testing a part of everyday health care.

A related phenomenon has been the transition to a new “business model” or “practical setting” for genetic testing since the late 1990s, namely genetic testing and counselling services offered directly to consumers. Some regard this way of by-passing the medical or healthcare setting (with a specialised doctor and its client) that previously controlled access to these services as providing free access to genetic testing, letting consumers decide on their own whether to make use of these testing options. Others consider direct-to-consumer genetic testing (DCGT) to be a possibly dangerous marketing ploy that will lead to genetic testing that is uncontrolled, scientifically unjustified, qualitatively doubtful and often intentionally misleading.

In contrast to the established practice, medical benefits and ethical and social problems of genetic testing, which have been the subject of many studies and numerous inquiries by ethical committees and other non-governmental and governmental advisory boards during the past 10 to 15 years, the debate on DCGT has just begun. DCGT is a rather new phenomenon that is apparently driven by the use of the internet. Although it is a growing market, it is still a niche market; new companies offering genetic testing via the internet currently are showing up constantly. It is however too early to tell whether they in the long and medium term will succeed to establish themselves on the market. This makes it difficult to assess the actual relevance of DCGT, which might well develop into a serious competitor to the established forms of genetic counselling and require political or statutory regulation in order to protect consumers' rights and health.

It was the objective of the STOA project "Direct-to-Consumer Genetic Testing" to explore the current use of DCGT. Starting with a discussion of the status and perspectives of genetic testing in general (section 2), the present report discusses the development of DCGT, its possible advantages and disadvantages and the arguments used by different stakeholders (3 -4) in order to explore policy options for fostering an ethically and medically reasonable offer of genetic testing to consumers (6). The concluding section (7) provides a condensed overview of the policy options at hand and of actions that could be taken into consideration at the European level.

The discussion of the pros and cons of DCGT is based on the latest available scientific literature and policy documents dealing with DCGT as well as on a systematic scan of offers of genetic testing that can be found on the internet, which was carried out in the context of the project during June and July 2008. The results of the survey (see section 5) and their possible implications for policy intervention in the field were discussed with a group of experts at a meeting hosted by the Flemish Institute for Science and Technology Assessment (viWTA) in Brussels on 22 September 2008.

The following experts participated in the meeting:

Pascal Borry, University of Leuven
Stuart Hogarth, University of Loughborough
Heidi Howard, McGill University Montreal
Alastair Kent, Genetic Interest Group
Ulf Kristoffersson, Lund University Hospital
Peter Pohl, GATC Biotech
Helen Wallace, Gene Watch U.K.

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