Statement of the ESHG on direct-to-consumer genetic testing for health purposes

Progress in biotechnologies has led to an increasing amount of complex tests providing predictive health information. An increasing number of private companies are now offering direct-to-consumer (DTC) genetic testing services, ranging from tests for single gene, highly penetrant disorders to susceptibility testing for genetic variants associated with common complex diseases or with specific traits. With this document, the European Society of Human Genetics (ESHG) wants to provide a formal policy statement with regard to DTC sales and/or advertising of genetic tests providing predictive health information. The issue of paternity testing and ancestry testing is out of the scope of this document.

As our knowledge of the genetic basis of diseases has grown, we have seen an increasing emergence of a biotechnology industry translating this knowledge into diagnostic products and commercial genetic testing services. The ESHG is concerned about the way commercial companies are currently introducing genetic tests into the market and outside of the scope of the traditional health care system.

In line with the Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes, this Statement wants to highlight the importance of the right to information, the quality and utility of genetic testing services, individualized medical supervision, the provision of information and genetic counseling, the protection of persons not able to consent, and the respect for private life.

Right to information

The ESHG does not question that individuals are entitled to health information, and in particular genetic information, about themselves. However, the right to know must be exercised with due respect for the necessary protection of individuals from inappropriate information and testing. Various companies underline that DTC genetic testing enhances individual's autonomy and empowerment. However, the ESHG advances that this enhancement can only occur when consumers are offered adequate information, as well as genetic tests that are reliable, that offer valid interpretation possibilities and that, as such, can be useful to the health of the individuals.

DTC advertising of genetic tests

Research in the context of direct-to-consumer advertising of prescription medicine has shown that DTC advertising created an inappropriate demand for medications and/or a demand for inappropriate medications. Moreover, research showed that various advertised messages for drugs were misleading and overstating effectiveness or minimizing risks, as well as leading to inadequate changes in medication, diet or lifestyle by consumers. DTC advertising of genetic tests for health purposes runs the same risks as DTC advertising of prescription medicine in that regard. Every advertisement should conform to international standards and guidelines with regard to advertisement of drugs and medical devices. Among other issues, the advertisement should be accurate and not misleading, should support their claims with substantial evidence, and should provide information about the risks and the benefits.

Quality of genetic testing services

The ESHG wants to the stress the importance of assuring a high quality of genetic testing services before it is appropriate to commercialize them. This includes the quality of the genetic tests (in terms of validity and utility), the quality of laboratories and the quality of the persons providing the genetic services.

Although many of the tests currently offered DTC have been based on an association between a specific genetic variant and a particular disorder, it is of particular concern that their predictive value should be sufficient to meet the standards for clinical use. The clinical utility of a genetic test should be an essential criterion for deciding to offer a test to an individual or a group of individuals. The ESHG advances that genetic testing services should always meet generally accepted criteria of analytical validity, clinical validity and clinical utility. Furthermore, the ethical, legal and social implications of the provided tests should be considered extensively. In light of these criteria, the ESHG rejects the premature commercialization of various genetic tests. For recently developed tests, adequate evidence for clinical utility may not yet be available. For tests on genetic variants with a potentially high predictive value, pilot studies will be needed to generate the evidence needed. Tests for which clinical utility is not yet proven but very likely, should be performed with clinical follow-up in the context of the health care system allowing post-marketing surveillance.

All laboratories offering genetic testing services should be appropriately staffed and equipped, should implement internal and external quality controls on laboratory procedures and should be subject to regular monitoring.

All persons involved in the provision of genetic services (i.e. medical doctors, nurses, genetic counselors, and other healthcare professionals as well as non-medical staff such as biologists and technicians working in the laboratories) should have the appropriate qualifications and perform their role in accordance with professional obligations and standards.

Individualised medical supervision

It is the opinion of the ESHG that the offer of a genetic test for health purposes in the absence of medical supervision may compromise or fail to foster patient health. Key concerns underpinning the necessity of placing genetic tests in the context of a medical professional relationship are the provision of sufficient information about the purpose and appropriateness of testing, as well as the accuracy and clinical significance of testing for individuals and family members. The involvement of medical professionals could avoid the waste of money on tests that are irrelevant or not scientifically valid. In addition, the waste of money and the adverse psychosocial effects of unnecessary follow-up medical investigations could be avoided.

In view of the emerging number of tests providing predictive health information for complex disorders, clinicians will need authoritative advice on the validity and utility of the available tests that is easily accessible. As more and more medical professionals will be confronted with the potential of genetic testing and genetic information, effective training will become increasingly important. Continued and up-to-date training for clinical geneticists and genetic counselors should also be an immediate priority.

Information and genetic counseling

When an individual is considering a genetic test, this person should be provided with details about the tested condition(s), the test(s) purpose(s), the potential limitations, the validity and accuracy of the test, and the significance and use of the test result for his health. In this context truth in labeling applies, i.e. the information that is declared must be accurate, accessible, complete and comprehensible. Although some companies have well elaborated websites providing most or all of this type of information, websites have a promotional nature and are intended to sell the tests, which might compromise truth in labeling. Particularly in relation to Mendelian disorders of high penetrance genetic counselling describes the process by which information is given and individuals make decisions about testing. Therefore, websites cannot merely replace appropriate pre-test and post-test genetic counseling, which discusses a whole range of elements. Genetic counseling is a communication process, which deals with the occurrence, or risk of occurrence, of a genetic disorder in the family. The process involves an attempt by appropriately trained person(s) to help the individual or the family to understand the medical facts of the disorder; appreciate how heredity contributes to the disorder and the risk of recurrence in specified relatives; understand the options of dealing with the disorder; choose the course of action which seems appropriate to them in the view of their risk and their family goals and act in accordance with that decision; and make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder. The ESHG also points to the potential conflict of interest that may arise when the healthcare professionals involved in the counseling are employed by or linked to the companies selling the tests. In this case impartial health advice might be compromised.

Informed consent

A genetic test for health purposes may only be carried out after the individual concerned has given free and informed consent. Although DTC genetic testing companies usually require a consent form to be signed when ordering a test, the ESHG considers that an informed consent procedure cannot be reduced to a written consent form. Although such a document might be necessary to document the process of informed consent, it can never be a substitute for a process that is in place to ensure that individuals comprehend the disclosed information, act voluntary, are competent to act and are able to consent to all elements of the consent form. An informed consent procedure should be a process in which individuals are informed about the purpose of a test, prospects of prevention or treatment, inheritance pattern if appropriate, risk of the disease, available choices, reliability and limitations of the test concerned, and possible psychological impact and other consequences of the test for the individual and his/her relatives. Privacy and confidentiality of the results, as well as possible consequences related to its disclosure to third parties, such as insurance companies and employers, should be discussed, when appropriate.

Moreover, any genetic testing service that requires a sample to be collected at home runs the risk of samples being submitted for testing without proper consent: DTC genetic testing companies do not have tools to assure that the biological sample provided for testing is obtained from the person claiming to be the sample provider.

Genetic testing in minors

The principles set out in the recommendations on minors issued by the ESHG apply here as well. However, although in a clinical context the opinion of a minor should be taken into consideration as an increasingly important determining factor in proportion to his age and degree of maturity, the context of a DTC genetic testing does not allow an adequate assessment of the competence of a minor. Therefore, the ESHG considers that genetic tests that are offered without medical supervision should never be available to individuals who have not reached the age of legal majority.

Respect for private life

Genetic tests should always be performed with respect for private life. In particular, companies offering DTC genetic tests should keep the customer's data confidential, should inform the customers of their procedures for ensuring confidentiality of the data, should explain what will happen with the sample and the data when the testing process is concluded and should have a clearly laid out plan as to what will happen to the samples and data should the company be sold or go bankrupt. Companies inviting their customers to share their genetic information via a web community or forum should inform people about potential drawbacks of disclosing this type of sensitive information. The ESHG rejects the use of personal details and genetic information by test providers (or affiliated companies) for direct-to-consumer marketing of medicines, vitamins or dietary supplements.

Research

Various companies that are offering DTC genetic services are performing research activities on the biological samples and the respective genetic information of their customers. The ESHG is concerned about the inadequate consent process with which costumers are enrolled in the research activities of these companies. Companies should make it clear to consumers if their sample or data will be used in further research and a separate and clear consent procedure should take place. Informed consent documents for participation in research should include information on the fact that the research may lead to patentable inventions.

Oversight of genetic testing

In order to overcome the immature translation of some genomic services to the market and the clinic, standards of best practice will be necessary. This may require regulatory oversight, professional and quality control systems. Professional initiatives will become more and more important in order to synthesize available data on the clinical validity and utility of specific genetic tests and identify gaps in knowledge as well as the studies needed to resolve them. Regulatory mechanisms, and in particular the adaption of the European In Vitro Diagnostic Devices Directive and the implementation of the Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes, will be necessary in order to allow the appropriate introduction of reliable genetic services in the healthcare system.

Impact on healthcare system

The current provision of genetic testing services outside the healthcare system creates concerns that this will lead to increased burdens on public health resources, due to increased calls on primary care physicians' or clinical geneticists' time, as well as adverse impacts on patient health (such as harm caused by overtreatment or adverse psychological consequences caused by false positive test results). In contrast, the ESHG wants to advocate the introduction of genetic tests, which meet the necessary quality criteria and are considered relevant for prevention or health, into the healthcare system and urge for their reimbursement by social security systems. No inequity should occur when introducing tests with proven clinical utility in the healthcare system.

Conclusion

The ESHG wants to underline in this document some fundamental considerations with regard to the delivery of genetic services to the public:
(a) the provision of test with proven clinical utility, performed in laboratories complying with relevant quality standards and supported by qualified personnel;
(b) the importance of medical supervision;
(c) the provision of appropriate information and counseling;
(d) respect for minors;
(e) respect for private life;
(f) respect for research ethics principles; and
(g) the need for an appropriate oversight of genetic tests.
These considerations are especially true for tests that are capable of providing information that have important implications for the health of the person concerned or for members of her family, as well as for tests that have important implications concerning reproductive choices. This being said, these considerations remain valid for all tests in which health claims are being made. These considerations are crucial in the successful translation of genetic information and tests from research to the clinic and society. DTC genetic testing or advertising of tests of unproven benefit, and without adequate counseling, goes against the professional standards the ESHG advances and might negatively affect the reputation of the genetic testing field. Policy makers are urged to protect the population by adapting the European IVD Directive and implementing the Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes.