MINISTRY OF
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Ethical observations on offering commercial genetic tests to consumers

Commercial, direct-to-consumer genetic tests have been discussed in the media, and questions concerning the reliability of the tests and how the results should be used have been brought forward. Biobanking and Biomolecular Resources Research Infrastructure, BBMRI.fi organized an extensive seminar in Finland on 5-6 October, 2011. The seminar also touched upon direct-to-consumer genetic tests. The National Advisory Board on Social Welfare and Health Care Ethics ETENE decided to discuss the topic and invited Helena Kääriäinen, MD, PhD, Research Professor at the National Institute for Health and Welfare to be heard at its meeting.

The number of companies offering direct-to-consumer genetic tests has increased significantly. At present, the number of companies offering various genetic tests through the internet is estimated to exceed 100. The tests may include paternity or family background tests, but in terms of healthcare, gene tests assessing the risk of becoming ill are the most significant. Frequently offered are test packages used for profiling the risk of developing one of several diseases of public health importance.

The reason for the growth of this business is first of all the strong development of gene research which can also be seen in the diversification and expansion of commercial gene tests. The second reason is that DNA material withstands shipping well, and even small amounts of DNA are sufficient for conducting a large number of tests. The third reason is the growth in the popularity of the internet as a tool for purchasing services. In addition, commercial gene tests afford an individual the opportunity to obtain information on his or health, disease or family relationships that previously required the assistance of a physician or another healthcare professional.

Commercial gene tests have also been a cause for concern for many different reasons. Many of the Gene tests for common diseases of public health importance provide information on the risk of developing a disease. However, they do not provide information on the certainty of a person developing a certain disease of public health importance or not. These diseases are multifactorial and therefore the existence of a certain gene or gene combination by itself does not explain why a person develops a disease. For example: Even if a gene test indicated that the risk of developing adult-onset diabetes is elevated, other factors can act to reduce this risk. Correspondingly, lifestyle factors alone can increase the risk manifold, even if such a risk was not found in gene tests. If gene tests had been proven effective in controlling diseases of public health importance, they would most likely be used in health care service provision. On the other hand, it is customary to assess the risk of becoming ill by means of conventional laboratory tests, such as measuring the serum cholesterol level, which on an individual level, does not indicate who actually becomes ill and who does not.

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Some of the commercial tests investigate matters other than disease, for example, predicting balding. The biological parentage of a child can also be determined using commercial gene tests. Because only a little DNA is needed, it is possible to take a sample from another person without his or her knowledge. However, the results of direct-to-consumer paternity tests are not legally valid because they were not conducted in a controlled manner, and it is possible that the samples were exchanged, or for some other reason, are not those of a given person. This substantially reduces the reliability of the tests. Risks of this type should be taken into consideration before deciding to obtain a test through the internet, rather than having it performed under controlled circumstances as part of public healthcare service provision. In principle, however, it is possible to determine one's biological roots through these tests.

Reading the entire genome may already be less expensive than conducting individual gene tests. Substantially new, definite information on becoming ill will be introduced in this way in the future. Some of the direct-to-consumer companies offer or plan to offer such services, even if they did not report all the results. The whole genome could be used to determine mutations of a few genes only, and report those to the customer. Reading the entire genome would not only provide information on a certain selection of gene mutations that increase the risk of becoming ill, but also on genetic defects in what are known as monogenic disorders that do not affect the risk of becoming ill, but unequivocally cause a certain disease. At present, it is difficult to predict how genetic information obtained through whole genome sequencing will be offered to consumers in the future.

In principle, gene tests can be conducted at any stage of a person's life. Direct-to-consumer tests are intended for adults, and companies usually inform customers that they do not perform tests for children. Because of the nature of the sample taking for gene tests performed on an embryo or during the foetal period, these tests are performed exclusively within the health care system.

The advantages and disadvantages of direct-to-consumer gene tests depend on how they are offered and what kind of information and interpretation is used to accompany the results. At its best, genetic profiling or reading the entire genome can help in diagnostics, planning and implementing targeted treatments and acquiring information on factors that predispose a person to the side effects of rarely used drugs. It can also be helpful in controlling certain lifestyle risks (for example, knowledge of an elevated risk of embolism helps a frequent flyer take measures to ensure good circulation during the flight). In the worst case scenario, a child or a young person may hear about his or her state of health, and this may affect his or her entire outlook on life. Parents can also receive painful information concerning their children becoming ill in the future.

Because the spectrum of direct-to-consumer tests/test packages is quite extensive, it is impossible to say anything general about their supply. Some of the tests are easy for the consumer to understand on the basis of the interpretation the seller of the test offers on its website. However, some tests contain information which in order to be understood requires a face-to-face conversation with a healthcare professional. A need of this type could indirectly increase the use of healthcare services, and finding a profes-

sional knowledgeable on genetic testing and interpreting its results is not necessarily easy. In extreme cases, knowledge about a serious future threat to one's state of health may be a heavy or even an overwhelming psychological burden to bear. This would also necessitate the availability of special support that is not readily available within the social and healthcare system at present. Acquiring the carrier tests to assess the need for conducting gene tests during the foetal period also raises questions about justice. Should all willing expectant parents have the right to such testing, and what would be the result? Would the number of severely ill children be reduced and on the other hand, would the number of selective abortions increase? How would society attempt to make an effort to guide such decisions indirectly? And what would be the consequence if the most affluent expectant parents received such information and those with lower incomes did not?

In healthcare, gene tests are used primarily to further define the diagnosis of an already detected disease. In addition, they can be used in the identification and early diagnosis of some diseases that run in certain families – the most notable example being families with hereditary cancer predisposition. In some families, gene tests are used for carrier and foetal diagnostics. In all such cases, it is a matter of diseases that are relatively uncommon. This development has been received by the healthcare service system as a step forward in the treatment and early diagnosis of these diseases, and in providing genetic counselling for families.

In terms of profiling the risk of more common health hazards and major diseases of public health importance, the situation has developed more recently. Even in this group, the risk of some diseases can be assessed sensibly already but in regard to most diseases, genetic information is only now being collected. Tests may encourage people to make lifestyle changes; however, to date, no long-term follow up is available in this regard.

It is clear that information transmitted through gene tests can lead to either needless worry or very-well-founded worry about one's own future health status or that of relatives or other loved ones. Therefore, ETENE's view is that everyone contemplating on such a test should understand the nature of the information acquired and consider whether or not he or she is ready to receive it and what the consequences would be. For the sole purpose of a possible increase in one's life management ability, commercial gene tests should not be undertaken. Anyone considering buying a test should determine the reliability of the service provider and also ensure that he or she has help available to interpret the results. The more extensively the genome is being tested, the more difficult it is to conceptualize in advance the effects of the results on one's life. The threshold for testing children must be particularly high, because it is impossible to make a decision on behalf of another person concerning whether he or she would like to know at a young age about developing a hereditary disease later in life.

Another question that remains unanswered at present is how the quality of direct-to-consumer gene tests can be controlled. Because this activity is likely to increase, ETENE's view is that realistic preparations should be made with regard to its supervision. At present, legislation on direct-to-consumer tests is lacking in Europe, and there are no guidelines for high-quality implementation of these activities. Therefore,

ETENE's view is that the Ministry of Social Affairs and Health, in cooperation with its expert agencies, should ensure that citizens have access to information that is independent of the test manufacturers at least in the electronic format. Offering commercial gene tests directly to the consumer is quite new and ETENE's opinion is that it is important to collect information on the use of these tests and the results of their use.

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