THE DANISH COUNCIL OF ETHICS

Genome testing

Ethical dilemmas in relation to diagnostics, research and direct-to-consumer testing

Executive summary

The Danish Council of Ethics has adopted a position on selected ethical aspects of the use of genome testing in diagnosis and research as well as direct-to-consumer testing through private providers. The Council has focused on:

- Justification for genome testing
- The testee's self-determination in relation to feedback
- Genetic counselling and information requirements
- Consequences for the public health services

A background report forms the basis for the Council of Ethics' recommendations. The report (in Danish) is accessible on the Council's website at www.etiskraad.dk/genom-undersoegelser

The report was compiled by a working party on the Danish Council of Ethics consisting of Gunna Christiansen (Chairperson), Thomas Ploug, Lotte Hvas, Ester Larsen and Jacob Birkler. At the Council of Ethics' secretariat, Morten Andreasen, project manager for the working party, and Anne Lykkeskov, project associate, compiled the manuscript on the basis of discussions on the working party and on the Council. Ulla Hybel authored the legal analysis concerning the use of genome testing in the health care system and in research. The report was considered and adopted by the Council of Ethics at meetings in August and October 2012. The report was published in November 2012.

This summary was published in January 2013.

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Recommendations by the Danish Council of Ethics

1. Priority-setting

Where the public health services' responsibilities start and end should be clearly indicated. Citizens should be able to receive counselling in the public health services if they have developed concerns as a result of genome testing, regardless of whether the genetic information they have sourced is of dubious relevance to their health. However, they should only be offered follow-up testing or treatment if there are professional medical grounds for doing so.

RATIONALE

Studies highlight the fact that a large proportion of those opting to be genome tested, e.g. through private providers, state that they plan to consult their own GP to obtain advice and, possibly, a referral for supplementary investigations. In many cases the information will be of no clinical value and patients will therefore be putting an unnecessary strain on resources in a health system that is already under pressure to prioritize its resources.

2. Up-to-date website and upskilling

To support doctors and citizens, the authorities should ensure that they present up-to-date, comprehensive and impartial information via a web-based platform about findings that may warrant follow-up testing and treatment.

It should be ensured that counselling and referrals are given through doctors sufficiently qualified for the purpose.

RATIONALE

Genome tests can generate an unusually large amount of information of varying reliability and relevance for the testee at one go. Much of the information is interesting from a research point of view but does not meet the quality criteria normally stipulated in diagnostic work. The area is extremely complex and can be difficult to manage for non-specialists, including general practitioners. The ordinary consumer will have very limited scope for evaluating the relevance of genome testing and specific results in terms of their health implications.

3. Information and counselling

It should be a statutory requirement for genome testing to be accompanied by impartial and comprehensive information as well as counselling, both before and after testing, whether conducted in the context of diagnosis or research, or provided direct to the consumer through a private provider.

RATIONALE

Deciding to look for insights into the genetic risk factors for oneself and one's family calls for something other and more than the information and voluntariness emphasized by the present legislation. It calls for dialogue with a genetic counsellor about e.g. the possible ethical dilemmas it may lead to. In the case of predictive genetic testing, no requirement for genetic counselling as such is embodied in Danish legislation, although Denmark has signed and ratified the Council of Europe's Bioethics Convention, which does set out such a requirement. Studies suggest that consumers of genome tests are not always aware of the importance of counselling themselves.

4. Feedback and the right to know/not know

Patients undergoing genome testing should be given a reasonable degree of involvement in deciding whether, and to what extent, they are to receive feedback on any incidental findings. That should always be agreed prior to starting testing. Patients' ease of access to their files can pose a problem for those declining to receive feedback. Clinical trial subjects should not be offered information about genetic risk factors.

RATIONALE

For some trial subjects, being promised access to information about genetic risk factors can constitute an incentive to take part in genome research, even if the value of such information is doubtful in health terms. Here it should be borne in mind that the purpose of participating in the trial is research, not diagnosis.

Doctors are normally expected to offer more extensive feedback on incidental findings made in diagnostic rather than research contexts. The question of where to draw the line between relevant and irrelevant information should be seen partly as an individual matter. Given the increased likelihood that genome testing will flag up incidental findings whose relevance to health is unclear, the feedback criteria may need to be clarified to support doctors' judgement. Offering the testee feedback on personal results once the results have already been produced constitutes an undue compromise of the respect for the right of the person in question not to know.

Ideally, information that patients do not wish to have should not be generated at all, nor should it be included in their files. In the case of extended diagnostic examinations, however, incidental findings may be inevitable. Record-keeping regulations state that all health-relevant patient data coming to the doctor's knowledge must be included in the files. Such information could become important subsequently—but the patient, who does not necessarily want this knowledge, can inadvertently stumble across it by looking up his or her files through the Danish e-health portal, sundhed.dk.

5. Children's and young people's access to genome testing

The Danish Council of Ethics is split on the question of whether society should restrict the possibilities for genome testing children and young people. Some members think that children and young people should only be genome tested as an exception—where there are weighty health-related reasons for doing so—while others feel that parents' freedom to genome test their under-age children should not be restricted.

RATIONALE

Children and young people under the age of 18 cannot be expected to be independently capable of taking a stance on the prospects of being genome tested. The decision to obtain a diagnosis on behalf of a child must rest with the parents when the child is under 15. Research is subject to an 18-years-of-age limit. Information about risk factors can create undue concern on the part of the child's parents and the child when he or she is able to access this information at a later date. The question, however, is whether it should be the responsibility of the parents alone to determine the expediency of genome testing children and young people in specific cases or whether society should curtail parents' freedom to genome test their children.

Legal status unclear

The recommendations set out reflect the Council's view that a common set of guidelines that can safeguard citizens' and doctors' legal status is lacking in this particular area. The use of genetic testing is governed by a large number of general laws and rules, which can give rise to interpretative uncertainty and the development of varying practice.

Summary of Chapters 1-3 of the background report

Chapter 1: Introduction

- Genome tests can generate large volumes of information about possible genetic risk factors. In some people's opinion that creates promising scope for prevention, while others point out that the information will primarily be onerous for some people. Some will wish for the information, others will not. That gives rise to a dilemma with regard to how to respect some people's right to know and others' right not to know. To this must be added the question, amongst others, of how to prepare the public health services for a possible future in which citizens have far easier access to personal genetic information. The Council of Ethics has focused on four questions:
- Justification for genome testing
- The testee's self-determination in relation to feedback
- Genetic counselling and information requirements
- Consequences for the public health services
- Within the space of a few years, technological development has made it far cheaper and faster to generate genetic data, but interpreting those data is challenging
- Genome tests are already being used in a lot of research, offered through private providers and taken into service in Denmark's hospitals. Genome testing can be relevant for up to almost half the patients currently being referred for genetic investigation
- Genome tests can also produce reliable knowledge about severe hereditary disease. However, the Council of Ethics' work has focused on the wealth of uncertain information.

Chapter 2: Significance and relevance of genome testing in health terms

- Genome tests can generate large volumes of uncertain information about the testee's possible risks of disease—risk factors that can have varying relevance for health. This uncertainty relates not least to predicting widespread diseases like type 2 diabetes, cancer and cardiovascular illness, and it is uncertain whether that will change noticeably with more research, since the development of these diseases is largely conditioned by environment rather than heredity
- Patients and trial subjects must be informed about findings that are clearly of essential relevance to health. Through their files, patients have access to all health-relevant information produced about them, including information they may have opted out of.

Trial subjects do not have the right to information about individual results, but are often offered certain items of information as a kind of consideration for taking part

- Most genome information is located in a grey area between what can be regarded as clinically clearly essential and clearly inessential. Often that information will be of interest to research but insufficiently reliable for what has traditionally been regarded as forming a suitable basis for health decisions. Hospitals normally narrow their focus to the patient's diagnosis, but when the whole genome is examined, it increases the probability of incidental findings
- Genome testing will presumably enable most people who undergo it to obtain reliable, new knowledge about at least one disease to which they have a markedly above-average disposition
- The relevance of genome information to the individual depends on specific aspects of both the finding and the testee. For many people, reaching the level of understanding required by genuine self-determination will be a taxing prospect
- The legislation attaches importance to information rather than counselling as a basis for making one's decision. In practice, however, some importance is given to counselling, but there are only a few clinical geneticists in Denmark
- Fundamentally, genetic counselling must be non-directive, but that is not possible realistically; nor do many patients wish for that. As a result, having the doctor convey genetic risk information is to some extent instrumental in indirectly defining whether acquiring knowledge of one's own genes is a good or a bad thing
- Existing evidence about testees' interpretation and handling of genome information can neither confirm nor disconfirm that genome tests arouse concern, or that such information motivates them to live a more healthy life.

Chapter 3: Genome tests – ethical deliberations

- Information generated by genome testing about possible risk factors gives rise to a number of ethical dilemmas
- For the doctor or researcher those dilemmas occur particularly in terms of whether to respect the patient's or the trial subject's autonomy by providing them with the most information possible generated by the sequencing, or by respecting the person's right not to know about this uncertain information. That dilemma can be managed by involving the patient or trial subject beforehand in the decision as to what information he or she wishes to receive
- There is an additional dilemma relating to patients and trial subjects wishing for detailed knowledge, including a knowledge of uncertain information, as well as those undergoing genome testing through private providers on their own initiative: Whereas in some instances this information can probably enable the person concerned to prevent disease in a timely manner, in many other instances that information will be so vague as to make the testee feel compelled to consult his or her own GP and possibly even demand referrals to specialists on an inadequate basis. Other things being equal, that will place a burden on the public health system, as resources will be deployed on this to the detriment of other areas.

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