The whole truth and nothing but the truth, but what is the truth?

Hanneke M A van den Boer-van den Berg and Anneke A Maat-Kievit

*J. Med. Genet.* 2001;38;39-42
doi:10.1136/jmg.38.1.39

Updated information and services can be found at:
http://jmg.bmj.com/cgi/content/full/38/1/39

**References**

These include:

This article cites 4 articles, 1 of which can be accessed free at:
http://jmg.bmj.com/cgi/content/full/38/1/39#BIBL

1 online articles that cite this article can be accessed at:
http://jmg.bmj.com/cgi/content/full/38/1/39#otherarticles

**Rapid responses**

You can respond to this article at:
http://jmg.bmj.com/cgi/eletter-submit/38/1/39

**Email alerting service**

Receive free email alerts when new articles cite this article - sign up in the box at the top right corner of the article

**Topic collections**

Articles on similar topics can be found in the following collections

Genetics (3948 articles)

**Notes**

To order reprints of this article go to:
http://www.bmjjournals.com/cgi/reprintform

To subscribe to *Journal of Medical Genetics* go to:
http://www.bmjjournals.com/subscriptions/
Clinical genetics in practice

The whole truth and nothing but the truth, but what is the truth?

Hanneke M A van den Boer-van den Berg, Anneke A Maat-Kievit

Abstract
The moral aspects of genetic counselling are explored in situations where the outcome of a DNA test does not lead to certain knowledge. The most frequent type of interaction between counsellor and counselee is when factual information is given, but sometimes “factual” information is difficult to obtain. How do counsellors deal with “uncertain” knowledge in genetics? Arguments and assumptions are presented and the finding of a 27 CAG repeat in the Huntington gene is used as an example. However, the questions “how far does the duty to inform reach?” and “to what extent is the doctor responsible?” are important in the whole field of genetics, and will be even more important in the future. The aims of science and clinical practice are discussed; we conclude that counsellors run the risk of taking on an infinite responsibility.

Keywords: ethics; genetics; informed consent; Huntington’s disease

For several years Huntington’s disease (HD) (MIM 143100) has served as a paradigm for presymptomatic testing for hereditary diseases. Genetic testing for HD by direct analysis of the size of the CAG repeat has been possible since 1993.1 In contrast to the earlier linkage test, testing the repeat lengths seemed to allow definitive diagnoses, enabling a yes or no answer: allele sizes of ≥40 CAG repeats lead to HD, all sizes <40 do not. For a short time all seemed crystal clear, but a more complex situation soon emerged. Allele sizes of 36–39 CAG repeats were sometimes also associated with HD. However, the HD phenotype is not always penetrant in subjects with HD alleles in this size range. There are indeed unaffected subjects aged >70 years with alleles of 36–39 CAG repeats. Empirical penetrance risks for the HD phenotype in subjects with these numbers of CAG repeats are unknown. Accordingly, subjects with these sizes of repeats cannot have predictive certainty about their HD status from their test outcome. Furthermore, it appears that allele sizes sometimes show mutability; an allele with CAG repeats <36 could indeed change into an HD allele in the next generation. For some time the “safe” limit seemed to be 30 CAG repeats, and the danger range for mutability 30-35. Then, in 1995, McGlennan et al reported an allele of 27 CAG repeats which was found in the father of an HD affected subject with 38 repeats, indicating the apparent instability of a CAG repeat of 27.2 This led to a redefinition and after 1995 the “safe” area was defined as below 27 repeats. The likelihood of an allele in the 30–35 repeat range being transmitted as a full HD allele is unknown. Potential factors include the repeat size, the sex of the transmitting parent, and whether it is identified in a (new mutation) family or in the general population.3 It is even more unclear how great the risk is in the 27-29 repeat range.

The aim of genetics
The aim of genetics is, like every branch of science, to gather “true knowledge”. Geneticists hope that knowledge about genes and mutations may help to explain causes of diseases and that understanding the cause can be used to assist in treatment. Unfortunately, nature often behaves like Hydra, the many headed monster: when science cuts a head from this monster, two other heads appear. Otherwise formulated: every new solution to an old problem creates two new problems. In science, “true” is what “has been proven”, but scientists often have to live with ambiguous and limited knowledge. Scientific progress does not only mean “gaining certainty”. In a way, the uncontrollable field does not become smaller. Geneticists are also confronted with this paradox. They attempt to make our world more understandable, they want to explain reality and try “to handle” it. The discovery of the nature of the Huntington gene mutation with its CAG repeat mechanism initially created more certainty, enabling “reliable” presymptomatic testing. However, the reverse became true also: the dynamic nature of the mutation brought more and new uncertainties. For example, we do not know what a safe margin is in the case of mutable CAG repeats. We do not know if, when, how, or why an allele becomes unstable. Opinions about this subject have changed and will probably change again in the future, but in spite of this geneticists want to try to draw “safe
The aim of clinical genetics

Genetic counselling is defined as “a communication process which deals with the human problems associated with the occurrence, or risk of occurrence, of a genetic disorder in a family”. Among other things, this process involves an attempt to help the person or the family “to comprehend the medical facts, including the diagnosis, the probable course of the disorder and the available management.”. Providing factual information is a very central interaction between counsellor and counsellee. That part of the counselling process is meant to help a person “choose the course of action which seems appropriate to him and act in accordance with that decision”.

However, respecting the patient’s autonomy does not only mean “informing”, but also “sharing responsibility”. We know it is important that counsellors get full support from their counsellors in the decision making process. So, respect also involves “treating persons to enable them to act autonomously”; requiring values such as empathy and compassion.

The duty to inform

Factual information should at least consist of true statements. A true statement is: “allele sizes of ≤26 CAG repeats have never been associated with an HD phenotype”;

be interpreted is sometimes ambiguous. A true statement is also: “allele sizes of ≥40 CAG repeats who died, disease free, after living up to or past the normal life expectancy”. This means for those tested, a yes answer is possible, in contrast to the group of people with 36-39 CAG repeats. In that group, a yes or no answer is not possible, as was shown above. The number of CAG repeats is a fact, but how this fact has to
Truth telling in clinical genetics

INFORMATION IN A NON-DIRECTIONAL WAY

Assumptions

(1) ONE OUGHT TO AVOID FUTURE REGRET
In general, people regret more what they did not do or what they have forgotten to do than what they did, even when it went wrong. Similarly, doctors appear better at providing arguments justifying a wrong action than when they omit something. Fear of future regret directs a counsellor towards informing and a counsellor towards doing something with the information because they both experience the same “anticipated decision regret”. However, the moment a counsellor explores a counsellor’s wish to know a certain test result, the counsellor’s right not to know is in a way violated. More precisely, is it necessary to explore whether there is a wish to be informed if a CAG repeat of 27 is found in an unaffected parent or in the partner of a carrier? “Do you want” questions may lead to supposed wants and needs, while informing may direct towards the use of the information rather than to neglect it. Are counsellors always obliged to increase the options available and to present every possible option? What is the aim? Perhaps the aim lies in the starting point of genetic counselling.

(2) A COUNSELLOR OUGHT TO PROVIDE FACTUAL INFORMATION IN A NON-DIRECTIONAL WAY
Sometimes, strictly adhering to the admonition to be morally neutral in the selection and transmission of information leads counsellors simply to dump information onto their clients, as stated by Caplan. But is “informing about the facts” in order “to choose and act” an exercise in truth dumping in which every fact, every option, every risk, and every benefit is unleashed? Is it really possible to be non-directional and inform about every possible event? Non-directive is not the same as morally neutral. Informing is not as value free as it sometimes seems to be, certainly not for the one who receives the information. Information from a doctor has a prescribing element. If a genetic counsellor thinks he/she ought to inform a couple of all findings, even if the findings are uninformative or difficult to interpret, he/she creates an environment in which the decision “to do” something with the test results seems wiser than “to do nothing”.

(3) COUNSELLORS OUGHT TO TELL THE TRUTH AND RESPECT THE AUTONOMY OF THE COUNSELLEE
It seems a universal rule to tell the truth, the whole truth, and nothing but the truth, in order to give the patient the opportunity to act freely. But when it is unclear what “the truth” is, this rule can easily lead to a sort of technical imperative. Not an imperative in the sense of “we have got the technology so we ought to use it” but in the sense of “we have used the technology so we ought to inform about all the results in every possible detail”. However, informing a counsellor does not always promote his autonomy, because autonomy is impossible in cases of inadequate and incomplete understanding.

(4) ONE OUGHT TO AVOID A LEGAL CLAIM
Fearing legal claims will probably direct the counsellor’s choices too. Our medicine seems to become more and more “defensive”. Globalisation of our world has greatly speeded up transfer of knowledge. Nowadays, scientists strive to use “the best method in the world” to solve a given problem. There is a continuous exchange of knowledge and, as a result, also of moral rules and values. However, moral ideas and ideals are not universal and depend, among other things, on law, insurances, economic factors, public policy, etc. Exchanging opinions, arguments, reasons, and motives remains important, but we must avoid the possibility of legal claims in one country directing the decisions and actions in another. In the contact between counsellor and counsellor moral values are often more important than legislation. We must prevent legal rules from harming those who are supposed to be protected by those rules.

Conclusion
Of course, respecting the counsellors’ autonomy means informing them of the medical facts, in order to give them the opportunity to choose between alternatives. However, to repeat our questions: what are the relevant medical facts and, most of all, to what extent should the doctor be responsible? Sometimes it seems “chance” no longer exists nowadays, it seems “fate” has completely changed to “fact”. The danger that one, as a counsellor, risks to feel infinitely responsible, not because one has caused something, but because one did not try to avoid it. If we feel responsible for all the facts, we want to deal, one way or another, with those facts. However, we have to realise that our capacities are limited, particularly the
capacity to understand and foresee all the consequences of our actions. We should not take more responsibility than we are able to bear.

As long as we do not know what the right decision is, we have to proceed carefully. Thousands of years ago the Greek physician Gorgias stated: “man can not know the past entirely, he can not overlook the present entirely and he can not foresee the future”. Nowdays everything seems to be calculable and predictable, but destiny is still capricious and life is still precarious. Counsellors want to tell the truth but unfortunately they do not always know what the truth is, as illustrated by the mutability of CAG repeats. They do not know how great the chance is that the repeats will expand. If counsellors do not know how to interpret certain facts, they are not able to give their counsellees the right interpretation either. The field of genetics becomes more and more a field of risk estimates and therefore the question will become more how “certain” a risk percentage is, and most of all which uncertain percentage is worth mentioning. In “minimal risk” situations (in this example when unaffected parents or partners are involved) it is wise for the counsellor to refrain, to keep his uncertainty to himself, to tell his patients the things he is pretty much sure of, and to keep silent about the things in which “chance” and “luck” play an important role. This is because the advantages for the counsellee are limited in those kinds of situations. If a counsellor has doubts about the value of ambiguous knowledge, the solution cannot be left to the responsibility of the counsellor. The question is if it is always justified to give “all available” information to counsellees, presuming that “knowing” is always better than “not-knowing”, and on the assumption that counsellees are able to cope with and master all kinds of uncertainties.

This point of view does not only have consequences for counsellors, but also for counsellees and, ultimately, for the whole of society. Do the latter accept and respect the limitation of the doctor’s responsibility and accept the limitations of technology? Resources should be invested into researching the wants, needs, and expectations of (potential) counsellees. Do they want information about every single hypothesis and every single exception? What do they really want and expect from genetics and what are counsellors able and willing to give?

We thank Prof Dr M F Niermeijer and Prof Dr M Breuning for their critical and stimulating remarks and Dr R Giles for language editing.