Ethical Management of Hereditary Cancer Information

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Acta Oncologica Vol. 38, No. 3, pp. 305–308, 1999

Genetic diagnosis yields information that is highly relevant for both the patient and the genetic relatives of the patient. In this article two ethical problems are discussed. Under what conditions should hereditary cancer information be given to a relative? It is suggested that in order to answer this question, three factors have to be considered and a balance struck: the seriousness of the condition, the existence of treatment or prevention and the reliability of the diagnosis. The second issue discussed in the article relates to the psychosocial effects of giving hereditary cancer information. It is argued that ethical management of clinical practice requires that further attention must be given to the psychosocial effects on both the individual and the family.

Received 15 September 1998
Accepted 30 December 1998

Developments in genetic medicine with pre-symptomatic testing that are taking place for an increasing number of genetically related conditions bring many difficult ethical problems to the fore. I discuss here two specific problems related to hereditary cancer information. The first problem emanates from the characteristic feature of genetic information that, in one way or another, it always also concerns the genetic relatives of the individual who is undergoing the testing. The second problem discussed in this article is the increasing role of prediction, which is an intrinsic part of pre-symptomatic testing. The ethics of risk communication needs more attention in order to ensure good ethical management of hereditary cancer information.

INFORMATION OF RELEVANCE TO GENETIC RELATIVES

Imagine a patient who has just been diagnosed with a malignant tumour. While listening to the doctor about his poor prognosis, he realizes that information about the hereditary character of this cancer was available several years earlier when a brother was diagnosed with the same disease. However, the two brothers were living in different parts of the country and did not have much contact at the time. The patient also learns that preventive measures were available at an early stage but now nothing can be done for him except palliative care. It does not require much empathy to see that this situation is morally reprehensible. How could this have happened? Whose interests were at stake?

The morally salient fact in this story is that information is acquired through genetic diagnosis that is highly relevant for the health and well-being of the proband's genetic relatives. One aspect of the problem relates to practical and bureaucratic structures setting the framework of genetic testing and counselling. For many diseases with a hereditary component, testing is still only a part of a research project and there is neither sufficient funding nor cooperation between different hospitals and counties for the test to be offered to relatives in a responsible way. Organizational problems concerning the provision of healthcare and genetic testing are not discussed in this article. However, the example concerning the two brothers indicates that there are urgent ethical problems related to the way diagnosis and counselling is organized across county and national borders. Universities, hospitals and counties must assume joint responsibility for implementing research in genetic diagnosis in ordinary healthcare programmes. In doing so, attention must be given to the psychosocial effects of genetic testing as well as other values at stake for the diagnosed individuals and their families. As is argued in the second section of this article, we still do not have sufficient empirical knowledge about these values for us to sustain an ethically responsible implementation of testing for hereditary cancer in clinical practice.

 ISSN 0284-186X

Besides the ethical management of organizational problems, there are ethical dilemmas associated with providing information to relatives who have not asked for this information and where the proband perhaps does not even want the relatives to be contacted (1). Not only the health and well-being of the relatives are at stake but also the integrity of both the proband and his genetic relatives. At bottom, there is a conflict with the fundamental duty of each doctor first to serve the interests of the patient. The primary duty of a doctor is to the patient, and for good reason. Not least, the lessons learned during the Nuremberg trials have underlined the importance of not trading patient interests for the benefits of future patients, or any other purpose, without adhering to appropriate information and consent procedures (2). However, the case of the two brothers indicates that in some situations it might be right to act in the interests of other concerned parties, perhaps even against the will of a proband as the case may be. I will come back to this issue shortly.

The development in genetic medicine does not challenge the view that the primary duty of a doctor is to his patient. However, it points to the need to examine the consequences of extending these duties to include the genetic relatives of the patient. If information is acquired through genetic diagnosis of an individual that is highly relevant for the health and well-being of a person who is genetically related to this individual, does the doctor have a duty to pass on the information even when the proband does not want it to be broadcast? If the answer to this question is in the affirmative, for what conditions and under what circumstances should this extended duty be invoked? These are the basic ethical issues regarding the handling of genetic information. Questions related to the interests of other third parties, such as public authorities, employers and insurance companies, concerning genetic information about individuals or groups of individuals will not be dealt with here.

With regard to diseases with a dominant pattern of heredity, the problem with managing genetic information may not be so acute. In these cases the members of the family of the proband often have extensive knowledge about the disease and the family history. Genetic relatives in these cases often approach the clinical team on their own initiative. However, the problem is more acute when it comes to recessive disorders. In these cases there may be family members who have no knowledge about the genetic risks. Even with regard to dominant diseases, the situation may be very complex. The development of genetic medicine implies a closer focus at the genetic family. However, in the society of today with new family structures and a blending of different cultures, this family may be difficult to locate. The social family and the genetic family seem to be out of phase in modern society. It is no longer enough to rely on old patterns of communication and moral bonds between kindred.

Ethical considerations refer to vital values at stake for the concerned parties, so these values must be identified as must the stake-holders. Ethical management requires, then, a balancing of these values against each other. In broad terms, the values at stake in the context of communication of hereditary cancer information are health, well-being and integrity. The proband, his close family and his distant genetic relatives are the concerned parties. Integrity refers to the interests of the proband in controlling the kind of information that is spread and the way this is done. This value is also relevant for a close family who fear being stigmatized as a 'cancer family', even if there is only evidence of a recessive disorder. For the close and distant relatives, there is the interest in terms of health as well as the interest in not knowing about the genetic risks that have to be considered. Health and well-being may be regarded as obvious values, but they both require detailed analysis and a wide spectrum of individual responses must be identified. For one individual it may only be of interest to learn about conditions where there is treatment available or when effective preventive measures can be taken. Other individuals might want to know about their condition in order to be able to plan the rest of their lives as they would wish. To be in control is an important part of one's well-being (3).

It should be noted that ethical management always requires a balancing of values. One must assume responsibility both for values that are hoped to be gained and values that must be foregone as an effect of an act. It may be right to violate the personal integrity of a patient/proband in order to help his/her genetic relative. A family member who is identified as a non-carrier may experience a sense of guilt towards family members who have been identified as carriers in a diagnostic test. Other family members may feel relieved just by knowing, even when they are identified as carriers. They feel a sense of control. Potential harm and benefits of various kinds associated with genetic testing must be considered in the ethical balancing of values that are at stake. It may in some instances be right to inflict harm such as feelings of guilt on a family member in order that a patient/proband might be helped just by knowing—to be in control. For serious conditions where there is a known treatment, I do not regard it as morally controversial to suggest that the interests of a relative are given priority and that the duty of the doctor to provide information should be extended. There are grey zones concerning how 'serious' a genetic disease is that must be taken into consideration. However, as a first approach toward ethical management I suggest the following two preconditions of ethical balancing in order to guide the proposed extension of duties.

(A) For serious conditions where there is a known treatment or when preventive measures are available, there is a stronger duty to consider the interests of

health and well-being of a genetic relative. Information should be given and treatment and counselling offered.

(B) When diagnosis is not reliable or when penetrance is low, there is a stronger duty to consider the interests of the integrity and non-maleficence of the proband and the relative. Information should not be given beyond the ordinary channels of family relationships.

COMMUNICATING RISK INFORMATION WITH UNKNOWN PSYCHOSOCIAL IMPLICATIONS

Diagnosis of hereditary cancer is a rapidly expanding area of clinical medicine. However, the current status of research concerning the psychosocial implications of communicating genetic risk information indicates that there is a substantial lack of knowledge guiding this development (4). Vital values are at stake for both the probands and their families but clinical practice seems to be developing without any profound consideration of these values. These values and the wide spectrum of individual responses to genetic risk information must be identified in order to ensure good ethical management of clinical oncogenetic practice. What sense does it make for an individual to be informed about an increased risk of 10/20/30/–100%. The answer to this question depends on both individual factors and difficulties associated with the very concept of 'risk', its measurement and its moral importance. Anyone involved in genetic medicine knows how difficult it is to give an accurate estimation of the risk. When this difficulty is mastered, the problem of risk evaluation emerges. At what risk level is a proposal for surgery, other preventive measures or just regular check-ups ethically warranted? How are we to balance the knowledge about an increased risk of acquiring cancer against the stigmatization of becoming a patient with no symptoms? The accuracy of risk estimation is the fundamental basis but subjective values and individual concerns are intrinsic parts of the evaluation. Differences in risk perception and psychological attitudes to risk-taking in general must also be taken into consideration in order to provide genetic counselling that is sensitive to individual probands.

It should be observed that the situation is not entirely new. There is a long clinical experience to rely on concerning the communication of sensitive information and medical information that a patient and his/her relatives might find difficult to bear. There are also experience and results from clinical studies available on the care of cancer patients. However, there are certain elements of genetic risk information where we cannot rely upon this experience. Other genetically related individuals are concerned to an extent that has not been experienced before. Even if there is some experience in communicating predictive information about health hazards based on biochemical predictors, genetic risk information is specific to an extent that has not been seen earlier. There may also be perceptions associated with the notion of heredity that compound the difficulty in giving information to an individual based on risk assessment for a population of individuals.

A review of the current status of research concerning the psychosocial implications of communicating genetic risk information reveals the following questions that need further attention.

1. *How is the information passed on to family members and what are the reactions*? The current practice implies that doctors have to rely on the proband/patient for passing on information to family and relatives. However, there is no empirically sustained documentation regarding the giving and receiving of this second-hand information. There is a substantial risk that complicated genetic information and risk estimates will be misunderstood. There is, furthermore, no control over psychological reactions when communication of genetic risk information is not supported by a professionally trained genetic counsellor (5).
2. *How are relations with family and other close associates affected*? There is evidence from clinical experience in genetic counselling for pre-symptomatic diseases indicating complex reactions within the family circle when one individual is tested. The evidence is anecdotal in nature and needs empirical proof in studies that recognize the diversity and the complexity of the consequences of genetic risk information in the families. Family members who do not carry the defective gene experience a sense of guilt that in other contexts has been described as the 'survival guilt' (6). The identification of a child with a parent may change when a hereditary similarity is confirmed or disproved. One branch of the family may be accused of introducing a bad element into the inheritance. There is also a risk of families being stigmatized as 'high-risk-families' or as 'cancer families' that must be investigated in psychological and social studies.
3. *To what extent is the capacity of correctly estimating the risk dependent on the cultural context of genetic consultation*? Evans and associates found that of those who had been subject to genetic counselling after diagnosis of breast cancer, only 33% were afterwards able to give the correct figure for risk of acquiring breast cancer for women in general (7). Only 41% could correctly estimate their own risk. These findings emphasize the need for investigating the cultural context of communicating genetic risk information and the relationship between the counsellor and the counselled. It further underlines the problems associated with relying exclusively on probands/patients to pass on information to family and relatives.
4. *Why are some individuals depressed despite reassuring information and why do they experience guilt*? There

seems to be no simple stimulus-response mechanism in communicating genetic risk information, a mechanism asserting that low risk implies a low degree of anxiety (8). On the contrary, there seem to be complex patterns of reactions involved where different components of an individual's life-view are affected. The idea of 'heredity' itself may shake fundamental conceptions of life and individual destiny. The association of risk perception with guilt is also connected with moral values peculiar to a given sociocultural context. Psychometric studies and structured interviews are needed in order to understand how the proband assesses his or her own situation of risk and how quality of life is affected.

5. *Are there any positive consequences for the individual and what exactly are they*? The small number of psychosocial studies available have focused on problems and negative consequences associated with the communication of hereditary cancer information. This approach is important and must continue. However, ethical management implies that a balance must be struck between negative and positive consequences. Furthermore, the positive values must be identified and fully appreciated in the ethical balancing of values. Increased levels of anxiety or depression or emotional disturbance are alarming factors but they may be worth while if the benefits are good enough. It is important to assess positive consequences such as experiencing a sense of being in control, the relief from being in a state of not knowing, the possibility of taking preventive measures or offering treatment and the security that is offered through regular check-ups. In true ethical balancing there are always some values that will be violated. However, it is only after the values at stake for all those concerned have been accurately identified that this balance can take place.

CONCLUSION

Ethical management of hereditary cancer information requires careful attention to vital values at stake for the relatives of the proband. Doctors must consider their duty not only to the patient but also to the relatives of the patient. In the elaboration of this extended duty in clinical medicine three factors should be taken into consideration: the seriousness of the hereditary condition, the existence of treatment or prevention and the reliability of the diagnosis. With regard to the psychosocial effects of diagnosis for hereditary cancer, multidisciplinary competence and support are needed in order to address the problems. Individual reactions as well as the effects on the family must be given greater attention in future research on clinical ethics.

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