Communicating genetic information in the family: the familial relationship as the forgotten factor

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J Med Ethics 2007;**33**:390–393. doi: 10.1136/jme.2006.017467

Communicating genetic information to family members has been the subject of an extensive debate recently in bioethics and law. In this context, the extent of the relatives' right to know and not to know is examined. The mainstream in the bioethical literature adopts a liberal perception of patient autonomy and offers a utilitarian mechanism for solving familial tensions over genetic information. This reflects a patient-centred approach in which disclosure without consent is justified only to prevent serious harm or death to others. Based on a legal and bioethical analysis on the one hand, and an examination of empirical studies on the other, this paper advocates the adoption of a relational perception of autonomy, which, in the context of genetics, takes into account the effect that any decision—whether to disclose or not to disclose—will have on the familial relationship and the dynamics of the particular family. Adding this factor to the criteria usually advocated by lawyers and ethicists will facilitate reaching a sensitive decision, which recognises the various interests of family members beyond the risk to physical health. Taking this factor into account will require a process of deliberation both between doctors and patients, and in the family. It will also require a relaxation of medical confidentiality, as the family rather than the patient is gradually perceived as the unit of care. Moreover, adopting such a relational approach will accord with current views of doctors and patients who base their decision primarily on the nature of the familial relationship.

access to it and their various interests in this regard are not fully recognised.

Focusing on the relatives' desire to know, medical confidentiality dominates the question of whether or not to disclose genetic information. The British Medical Association, for example, holds that the doctor's duty of confidentiality to the individual patient can be breached only in exceptional circumstances when serious harm to others or death can be avoided.⁵ A similar utilitarian mechanism is adopted by English medical law.⁶ In addition, there is wide agreement both in law and bioethics that doctors should have the discretion to decide whether or not to breach the patient's right to confidentiality, but no legal duty to inform the relatives.^{7–8}

Moving on to the relatives' wish to remain ignorant, the lack of cures for genetic disorders leads to doubts as to whether information should be conveyed freely within the family. The adverse psychological effect that genetic test results may have on the individual can be devastating, especially when the disorder is incurable and terminal. Within liberal discourse, patients and relatives ought to have a right not to know. This reflects the conviction that the interest in being left alone should take precedence over the interest in belonging to a close group of intimates.

However, an alternative to this liberal approach has a relational dimension. According to this perception, it is the individual's social embeddedness that nurtures his or her autonomy.⁹ Consequently, the individual has an interest in maintaining familial relationships and in living in a community with which he or she can identify.

Although this perception of relational autonomy accepts that a personal interest in making informed and free decisions is the major vehicle by which patients can lead their lives as they wish, it emphasises the conviction that the patient develops his or her autonomy by engaging with those around him or her.¹⁰

When familial tensions over genetic information arise, this alternative approach recognises that the strict rule of confidentiality should be relaxed and provide room for the ethics of the family, which is mainly based on care, commitment, intimacy, solidarity and mutual responsibility.

Support for this relational approach can be found when examining the views of doctors and patients about disclosure of medical information to family members. Research suggests that although both these groups highly value confidentiality,^{11–12} many believe that medical information can, in some circumstances, be disclosed to the relatives

Communicating genetic information to family members has been the subject of an extensive debate recently both in bioethics and law.^{1–4} In this context, two familial conflicts are investigated: (1) the conflict between the patient's interest in confidentiality and the relatives' interest in receiving the information and (2) the conflict between the patient's interest in sharing information with the relatives and their interest in remaining ignorant. The principle of autonomy and the rules of confidentiality and privacy are examined in the context of these familial tensions.

The discussion in the mainstream of the bioethical literature usually perceives the patient and the relative as two separate parties. It reflects a patient-centred approach in which the doctor-patient relationship excludes others who are closely related. Thus, despite the familial aspects of genetic information, relatives do not have free

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Received 16 May 2006
Revised 7 August 2006
Accepted 8 August 2006

without the patient's consent.¹ In the patients' view, disclosure without consent is contingent on a close and intimate relationship with the relatives, and on the effect that the information has on relatives' lives.¹³ For doctors, it may take place when the relatives' assistance is needed to improve the patient's condition, to relieve the relatives' anxiety and to prevent harm.¹⁴ Moreover, the relatives are perceived by many doctors as part of the unit of care. This emerges when comparing disclosure to relatives and to other third parties such as insurers and employers.¹² The findings also show that prevention of serious harm is not the single justification for breaching confidentiality^{15 16}; it also derives from a strong sense of moral responsibility to those who are emotionally close to the patient. This reflects the view that communication of genetic information is a family matter requiring solutions for all parties involved.¹⁷

CONSIDERING THE FAMILIAL RELATIONSHIP

As argued above, the current legal and ethical mechanism for resolving familial conflicts over medical genetic information is based on giving doctors and patients the discretion to decide whether or not to inform family members. The model suggested here does not propose to change this framework, as it aims to accord with the current structure of public healthcare systems.ⁱⁱ The suggested model, however, challenges the existing criteria that guide doctors and patients when considering disclosure of genetic information.

When a decision about disclosure to relatives has to be made, various ethical and professional guidelines advise patients and doctors to consider the following criteria:

- (1) the availability of cures or preventive measures;
- (2) the severity of the disease and likelihood of onset;
- (3) the nature of the disorder;
- (4) the availability of genetic testing and its accuracy in assessing the risk;
- (5) the relative's likely emotional reaction when given the information.¹⁸

However, an additional criterion, which is omitted from these codes, is:

- (6) the effect any decision (to disclose or not to disclose) will have on the familial relationship and on the dynamics of the particular family.

This social dimension is a very important factor, often forgotten in law and bioethics. However, it reflects the sociomedical research and the views of doctors and patients concerning confidentiality. As this current research suggests, patients tend to base their decision whether or not to disclose information to family members on the quality and nature of the relationship that they have with their relatives, rather than on the availability of a cure.^{19 20} Doctors, too, do not emphasise the availability of preventive measures when considering disclosure to family members without consent. In the majority of cases, when a disease has no effective cure, and when the likelihood of onset is not certain, the criterion of familial relationship will be dominant.

It can be argued that there is no difference between the fifth and sixth criteria, as they both address the psychosocial implications of disclosing genetic information. However, there

ⁱFor a comprehensive and detailed analysis of current empirical studies on patients' and doctors' views about breach of confidentiality, see Gilbar,¹ chapter 5.

ⁱⁱModels that promote equal access to genetic information exist. Whether they compromise the individual's right to confidentiality and privacy is beyond the scope of this study. In any event, implementing these models requires significant changes in the structure of the healthcare system and its allocation of resources. For such a model, see Doukas and Berg.⁴

is a distinction between the two. The fifth criterion considers the likely emotional reaction when a decision whether or not to disclose is relevant. This does not take into account the effect of the familial relationship on the decision, but takes only the emotional stability of the individual relative into account, if he or she is informed without being asked in advance.

Laurie,² who emphasises this aspect, argues that taking this into account will protect the spatial privacy of the individual. The notion of spatial privacy is defined as a private sphere around the individual that cannot and should not be intruded upon without a justifiable cause. In the context of genetics, the relative's spatial privacy is unjustly invaded if genetic information is communicated to him or her when there is no indication of wishing to have such information. Ultimately, the assumption is that the psychological integrity of the individual relative should be respected, unless it is justifiable to invade it.²

A closer look at Laurie's argument leads to the conclusion that it is not family based. When patients consider whether or not to invade their relatives' spatial privacy, they must rely on appreciation of what the relatives' best interests are. For example, Laurie argues that the patient may decide not to invade the relative's privacy because he or she knows that the relative will not be able to deal with the news psychologically. Hence, Laurie asks the patient to stand apart from the relatives and evaluate the relevant aspects of the issue according to his or her knowledge of the relatives' interests. In such a move, the effect on the intimate relationship between the patient and the relative is overlooked. However, the patient cannot simply make a decision for the relatives according to his or her knowledge of their wishes; the patient is also affected by the nature of the relationship in the family. The sixth criterion takes this last consideration into account. Whereas the fifth criterion focuses on the emotional reaction of the relative to the information he or she receives, the sixth criterion deals with the patient's response to this reaction.

The sixth criterion will require doctors and patients to discuss the various aspects of the patient's familial relationship. These may include the relatives' reaction to the fact that the patient has developed a genetic disease: will they support the patient or will they express resentment or indifference; the patient's reaction to the relatives' decisions, such as terminating a risky pregnancy, or undergoing prenatal testing; whether the patient's decision derives from his or her relationship with the relatives; whether the nature of the familial relationship suggests that relatives will willingly agree to waive their right not to know; or whether the relationship is based primarily on individual privacy. Considering these aspects, and others, will help patients and doctors to solve familial tensions before or when they arise.

Conducting a serious discussion about these aspects will require the doctor to have close knowledge of the nature of the patient's familial relationship. In the area of genetics, this can be gained in clinics that employ therapists and counsellors, who can discuss the various implications that the genetic information has for the family.ⁱⁱⁱ²¹ Moreover, at the primary-care level, general practitioners are expected to know their patients and their families well.²² In many cases, family members have the same general practitioner, who has known them for years and is trained to mediate familial disagreements.

ⁱⁱⁱSee Harper,²¹ chapter 1. Notably, there are various formats of genetic counselling, such as: (1) mutation testing on one individual, which suggests that his or her relatives carry the same mutation; (2) mutation testing for the purpose of enabling healthcare professionals to provide risk assessment to relatives; and (3) providing a clinical diagnosis or risk assessment to the individual patient, which clarifies the risk assessment given to the relatives. These various settings of genetic counselling have different implications for the family.

The implementation of this social criterion requires a process of communication and deliberation, which should be conducted both at the doctor's clinic and within the family. As Parker¹⁰ argues, ethical dilemmas in genetics require interpersonal deliberation. Thus, through discussions with the doctor and in the family, the patient and the relatives will become aware of each others' core values and beliefs regarding confidentiality, privacy and disclosure of genetic information. For example, in communicating with the patient, the doctor will learn whether the patient wishes to inform the relatives, whether there are relatives he or she is reluctant to inform, and whether he or she knows the relatives' views about disclosure. In communicating with the relatives, the patient will become aware of their opinions: whether they wish to be informed or not.

The doctor–patient relationship

Focusing on the deliberative process between doctors and patients, the patient will gradually become aware of the implications the information has for the relatives. France and Clarke²³ discuss one case of subarachnoid haemorrhage and two cases of *BRCA* carriers who were initially reluctant to disclose genetic information to their family members. The reason for their initial refusal to disclose was their suspicion that the relatives would “react badly”. However, following a process of deliberation with the genetic counsellors, the patients decided to communicate the information.

France and Clarke emphasise the benefit of allowing patients time to make a decision as to whether or not to disclose information to their relatives, and argues that if disclosure is forced, it might damage the familial relationship. This leads to the assertion that for many patients the choice whether or not to inform relatives is not between “now or never” but between “now and by the time we next meet”. In addition, as other studies show, it is not a choice between disclosure to all and disclosure to no one, but a decision whether to disclose to one relative or more.^{24–25} Another alternative is a partial disclosure. This means, for example, disclosing the information without exposing the patient's identity. This method is used to avoid damaging the status of the patient in the family and the community. Thus, the issue of disclosure, which is perceived by many as an absolute question of all or none, is in practice more flexible, with the decision being made according to the social and familial circumstances of the case.

However, how can a deliberative model deal with a conflict between the interests of the patient and those of the relatives when a compromise cannot be achieved? In this context, we should analyse two examples discussed in the literature. One is the case of an 8-week pregnant woman who wants to undergo prenatal testing for Huntington's disease, knowing that her husband, with whom she has a fragile relationship, refuses to take the test or know the results.^{iv26} The second example is that of a patient with breast cancer who was tested to know whether she is a *BRCA* carrier, but was resolved not to inform her two young adult daughters if the results were positive.²⁷ In these cases, one may wonder how a process of deliberation would help to solve the familial tensions.

In the first example, in which the disease is terminal, an awareness of the nature of the difficult relationship between the woman and her husband is crucial in considering the patient's decision to undergo testing. The patient's right to undergo testing should be considered alongside the possibility that such a decision may destroy the relationship of this family and may have a serious adverse effect on the lives of the husband and the child, if born. The child may not be able to

have the benefit of living with both biological parents. In addition, it will be difficult for the mother to conceal the information from her spouse, especially if they do remain in a close relationship. And when the husband is accidentally or intentionally informed, this may cause substantial psychological harm to him.

Admittedly, the proposed model does not presume to solve such a serious conflict. However, by embarking on a deliberative process with the doctor or the genetic counsellor, the patient may gradually realise the implications of her decision to undergo prenatal testing in terms of her relationships with her husband and unborn baby. Although the patient will have the final say on this issue, by having a thorough discussion with her doctor, she may come to appreciate that to be tested despite her husband's refusal may compromise her other life plans and the relationships that matter to her the most.

Still, should the doctors refuse to perform the prenatal test, they may have to justify the decision if challenged by the mother. At this stage, the question is whether prevention of future physical harm supersedes the potential destruction of the familial relationship. It is a dilemma with no correct answer. However, the above-suggested social criteria provide the doctors at least with a valid justification for their refusal to perform the test.

The second example reflects other difficulties for the doctor and the patient. If the results are positive and the patient insists on her refusal to disclose the information to her two daughters, the availability of some measures to reduce the risk of the daughters' developing inherited breast cancer should be taken into account, together with the nature of the relationship of the particular family. Thus, the mother's instinct to protect her daughters from the bad news may be altered through a process of deliberation with her husband and the doctor. Investigating the familial relationship, the shared values of the patient's family and the level of communication among its members, the doctor can indicate to the mother that her initial decision not to disclose may endanger her relationship with her daughters and with her husband, who may favour disclosure to protect his daughters' physical health. The doctor may also point out to the patient that her responsibility towards her daughters may entail disclosure, especially when they can take measures to reduce their risk if they test positive. However, if the deliberative process does not yield an agreement and the doctor decides to disclose information without consent, the social criterion may add another layer to the utilitarian justification of prevention of harm.

Regarding the right not to know, the model is sensitive to a certain degree to the relatives' interest in not receiving genetic information. The doctor and the patient may come to a decision not to disclose, following serious consideration of the moral, psychological and social implications of such a decision. However, unlike other models, the present model does not aim to bar a free flow of information in the family. Rather, the underlying perception is that familial relationships are based mainly on communication and exchange of thoughts, views, feelings and experiences, which leave little room for relatives to fulfil their interest in being left alone and remaining ignorant. Nevertheless, the interest in not receiving information can be satisfied when the relatives' views are known or in exceptional cases—for example, when the relative suffers from severe depression. Ultimately, reinforcing a sense of moral responsibility in the family implies that relatives will have to compromise their personal interest in being in a state of separateness.

The patient–relatives relationship

Moving on to the deliberative process within the family, it must be admitted that in many families such a process is not possible

^{iv}See box 3 in Parker *et al*²⁶. The authors raise several ethical questions in this context.

either before or after testing. A family whose relationship is dominated by resentment and anger will not be willing to conduct a serious discussion about the communication of genetic information. This will impose a considerable burden on the doctor, who will have to consider carefully whether the interests of all parties concerned are not overly compromised by disclosure. In that sense, the desire to maintain social relationships in their own right may be outweighed by the interests of the individual family member.

SUMMARY

To sum up, the aim of the deliberative process is to change the ethical and legal position by adopting a relational perception of autonomy. Such a perception must lead to relaxation of medical confidentiality and privacy by including a social dimension in the utilitarian criteria. Consequently, the various interests of family members beyond the promotion of physical health would be recognised.

Admittedly, this deliberative process is not free of difficulties. With regard to the doctor–patient relationship, it will require extra resources, as the doctor will have to dedicate time to learn the patient's familial relationships; discuss the decision with the patient as to whether or not to disclose; and establish a high level of trust. Another significant difficulty is whether this suggested deliberative model accords with unhappy families whose relationships are complicated, or with patriarchal families, who are dominated by a single family member. In these circumstances, an effective process of deliberation is difficult to attain, and lawyers, ethicists and medical practitioners tend to give preference to patient's rights of autonomy, privacy and confidentiality.²⁸ Clarke wonders whether the pregnant woman, in the example presented above, would be able to keep the test result from her husband and not show it when they argue, and she thinks his behaviour is irrational. Would this information, asks Clarke, not damage their relationship permanently?²⁹

CONCLUSION

Genetic information has considerable impact on people's self-image and identity. It prompts them to rigorously exercise their rights to autonomy, confidentiality and privacy. Instinctively, they wish to control the flow of this sensitive information. However, genetic information poses a challenge to this individualistic interest, for it derives from genetic material that people share with their relatives, and, as studies show, it has a considerable effect on blood and non-blood relatives alike. Family members believe that their interests with regard to this information should be legally and ethically recognised.

The area of genetics elicits complex tensions in families. The deliberative model proposed in this study aims to provide a flexible and workable framework for doctors and patients to promote the interests of family members. The deliberative model, which attempts to accord with the current settings of the National Health Service and other public healthcare systems, suggests that when familial tensions over genetic information arise, both law and professional codes of ethics

should consider the social dimension of these conflicts. As research develops in the area of genetics, the family, rather than the individual, will be seen as the unit of care.

Competing interests: None declared.

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