

Concern for families and individuals in clinical genetics

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Clinical geneticists are increasingly confronted with ethical tensions between their responsibilities to individual patients and to other family members. This paper considers the ethical implications of a "familial" conception of the clinical genetics role. It argues that dogmatic adherence to either the familial or to the individualistic conception of clinical genetics has the potential to lead to significant harms and to fail to take important obligations seriously.

Geneticists are likely to continue to be required to make moral judgments in the resolution of such tensions and may find it useful to have access to ethics training and support.

The role of the clinical geneticist might in some ways be compared to that of the general practitioner (GP) as they often look after several members of the same family and sometimes come to know the members of such families very well. As a result, in much the same way as the GP may do, clinical geneticists and genetic counsellors often develop a sense of responsibility for a number of different family members and indeed, sometimes for the future of the family itself, where this is at risk. Because genetic information is by nature both highly personal and at the same time familial, this sense of responsibility can sometimes lead to ethical tensions. The sense geneticists and genetic counsellors have of themselves as sitting at the centre of a complex web of familial moral relationships seems unlikely to diminish in the future with the increasing availability of genetic testing and an increasing number of interrelated family histories and narratives, of which they are likely to become aware as a consequence.

In this paper we want to consider the extent to which a broad "familial" conception of the clinical genetics role is workable and the extent to which it is more realistic, and more ethical, to focus on the needs and wishes of individual patients, except in exceptional circumstances. We shall argue that for both ethical and practical reasons geneticists and counsellors are likely to continue to have to work sensitively with the ethical tensions between concern for individuals on the one hand and for their families on the other.^{1,2} We shall argue that dogmatic adherence to *either* the individualistic or the familial conception of clinical genetics has the potential to lead to significant harms and to fail to take important obligations seriously. The resolution of such tensions on a case by case basis requires moral *judgment*. As a consequence, we suggest, there is likely to be an increasing need for the availability of appropriate ethics training and support in the clinical genetics setting.

RESPONSIBILITY FOR INDIVIDUAL PATIENTS AND THEIR FAMILIES

In their day to day practice, clinical geneticists and genetics counsellors come to have a range of different kinds of relationships with family members, some very involved and some very distant, and come to see these as bringing with them a wide variety of obligations and duties.³ Health professionals of all kinds will of course always have special responsibilities to the patient sitting in front of them in the

consultation.⁴ The very nature of the consultation (and of family history taking) in clinical genetics means, however, that the clinical geneticist almost inevitably becomes aware of information that creates moral relationships to others, of varying kinds and degrees, in ways that are quite unusual in most other specialties.

At one end of the spectrum there will be cases in which several members of the same family are patients of one clinician. There will be others where the geneticist, through the taking of family histories, or perhaps as a result of collaborative work with colleagues in other clinics, comes to know about family members who are not their own patients but who are in contact with health professionals elsewhere. In addition to these two categories of *professional* relationship between health professionals and family members, clinical geneticists will also often become aware of family members who are affected, at risk of being affected, or carriers of a genetic condition, but who are *not* in contact with health professionals for one reason or another. They may be unaware of their condition, or of that of other family members through lack of contact or through non-disclosure, or they may in fact be aware but have chosen not to seek genetic testing or advice. A third kind of relationship arises where a geneticist has information about family members who are not, or at least not always, biological relatives, such as spouses or other sexual partners. These may include current partners considering pregnancy and ex-partners who are no longer in contact with the patient, where—for example, information has been gained through the testing of a child.⁵

There are then a wide variety of network-like moral features emergent in the very nature of clinical genetics. Awareness of these features is likely to be most acute when the families concerned are in conflict. This is not uncommon in practice because whilst families faced with any kind of difficult health care information find this hard to deal with, it is particularly difficult for those with inherited disorders, where there can be an additional sense of responsibility for the health and illness of others. Patients may feel ashamed and guilty, or simply embarrassed about the inherited disorder and may feel unable to discuss this openly with other family members. All of this is very understandable of course and will be compounded where the family is also in conflict for other reasons, unrelated to the inherited condition. They may be going through a messy divorce or separation or they may simply not get on with each other. In some such cases, the inherited condition may itself be

a factor, such as in genetic conditions associated with aggression, anger or mental incapacity and this may make relationships in such families more difficult, compounding the difficulties faced by those suffering from the physiological effects of the condition, still further.

Taken together then, the combination of working with families, who may sometimes be in dispute, and having access to information which is predictive, interpersonal, and identifying about a *range* of types of family members both near and distant, to some of whom the clinician may have a professional duty of care, means that clinical geneticists often have moral obligations over and above those they owe to the patient in front of them. In the light of this, an important ethical question for clinical geneticists will be how to assess the appropriate weight to be given to each of these obligations in their day to day work with patients. There are many who would argue that clinical geneticists should concentrate on providing information, support, and testing to the index patient, except in exceptional circumstances such as when there is a risk of death or serious harm to identifiable others. To do otherwise, it is sometimes suggested, would be to undermine respect for patient autonomy and to step onto the slippery slope leading to unacceptably paternalistic medicine.⁶ Clinical geneticists, however, tend to see both the familial and the individual as core moral concerns and as important (and unavoidable) moral features of their practice.⁷

THREE CASES OF ETHICS IN CLINICAL GENETICS

In practice, the tensions between concern for individual patients and for other family members might arise in two different ways, either *pre-test*, or *post-test*.⁸ They arise *pre-test* in situations in which a test has been requested (or the drawing of a family history is being considered), which may, if it is carried out, have implications for, or be revealing about, people other than the person being tested. Ethical issues arise *post-test* in situations in which a test has been carried out, or where a family history has been constructed, and information has been revealed, perhaps unexpectedly, that may be of use to others. Such information may be useful because it has the potential to avoid harms, to enable these others to seek treatment, to help them to avoid harmful unnecessary treatment (such as—for example, prophylactic mastectomy), to participate in a programme of early surveillance or, perhaps, to make more informed reproductive choices.

We begin our consideration of the ethical issues by looking at those arising *post-test*.

(I) Post-test ethical issues in clinical genetics

It can sometimes become apparent after a genetic test has been carried out or a family history has been constructed, that information has been revealed that may be of use to other family members. Consider the following case in which information from a clinical diagnosis, followed by a genetic test on one family member has the potential to be useful to others. This type of *post-test* scenario arises in many inherited conditions but the one we choose to focus on here is Duchenne Muscular Dystrophy (see box 1).

This case, and others like it arising *post-test*, present a range of ethical questions. These include the question of how the clinician is to reconcile the tensions between, on the one hand, the obligation to respect the confidentiality of the index patient, and on the other, the obligation to avoid harms or to provide information that would predict health, to her sister. Arguments pulling in the former direction will tend to emphasise the duty to respect a patient's confidentiality or perhaps the importance of avoiding harms to the doctor/patient relationship that might result from any breach of confidentiality, especially if it became widely known. Arguments pulling in the other direction will tend to focus on the duty to avoid harm and the right of access to health

Box 1

A four year old boy has been diagnosed with Duchenne Muscular Dystrophy (DMD). His diagnosis is confirmed by genetic testing. Duchenne Muscular Dystrophy is a severe, debilitating and progressive muscle wasting disease in which children become wheelchairbound by their early teens and usually die in their twenties. Duchenne Muscular Dystrophy is an x-linked recessive genetic condition and whilst it is carried by girls it is only (with very rare exceptions) boys who are affected.

The boy's mother, Alison, is shown to be a carrier for the mutation. Carrier women do not show symptoms of the condition, but half of their sons will inherit it from them and will be affected. Alison has a sister, Sue, who is ten weeks pregnant and who has just been scanned by a colleague in the same centre. At her first meeting, Sue told her clinician about her nephew's speech and development delay, but said she was not aware of any diagnostic label. She said she was anxious about the implications of this for her and her pregnancy and also said, later in the discussion, that she would terminate a pregnancy known to be affected with a life threatening condition. Speech and development delay are features of a wide range of conditions and would not of themselves indicate carrier testing for DMD. In addition, because the DMD gene is large and there are a number of possible mutations, testing without information about which mutation is responsible for the nephew's condition is unlikely to be informative.

Alison knows that Sue is pregnant and that the pregnancy could be affected but she has not told Sue of her son's diagnosis, or about her own carrier status, since she feels that if she tells her sister, Sue would be likely to terminate the pregnancy if it turned out to be affected. Alison feels that this would be wrong. She knows that her sister does not share her views but she has thought long and hard about the issues. She has discussed it with her family doctor and her geneticist and has decided she wants the results to remain confidential and not to be disclosed to any third party, including her sister.

Both women are patients of the regional genetics service. At the regular team meeting, clinicians and counsellors say they feel they have a duty of care to both of the sisters. To tell Sue would be to breach Alison's confidentiality. But, Sue has a one in four chance (without her carrier status being confirmed) of having an affected child. A genetic test would allow Sue to make a more informed reproductive choice.

information. It is widely accepted in most jurisdictions, that confidentiality may be breached to avoid serious harm or death to people other than the patient,¹ but is this a case in which the harms are "serious"? The interpretation of what constitutes serious harm is particularly difficult in the context of clinical genetics⁹ and the question of what constitutes "serious" harm is likely to be an ethical question of continuing practical importance in clinical practice. One's view on this matter is likely to have a significant impact upon one's view about the appropriate role of clinical geneticists in relation to families and individuals.

There are other *post-test* cases in which, in addition to the avoidance of harm, there is also a question about the *provenance* of familial information resulting from a genetic test. Consider the following case in which information is revealed *post-test* that was not expected at the time of taking consent (see box 2).

Whilst there are clearly harms that might be avoided by disclosure of the information about non-paternity to both members of the couple in this case, such as—for example, Polly going through pointless and risky prenatal testing or

Box 2

A married couple in their early thirties, Polly and Richard, are referred to clinical genetics following the diagnosis of a rare autosomal recessive condition in their newborn baby. The disorder is severe and debilitating and there is a high chance that the child will die in the first year. During their first session with the genetic counsellor, Polly and Richard are informed that there is a 25% chance that a future baby would also be affected. The couple are extremely distressed about this and Richard says he would prefer not to have any more children if they stand any chance of being affected. At the end of the counselling session, Polly and Richard make an appointment for a future meeting to discuss their reproductive options further.

Between the two meetings Polly telephones the unit to say that Richard is not the father of the child. She says she wants the clinical team to know this but is adamant that she does not want Richard to find out. As the mutation is rare in the population, this means that, if what Polly says is true, whilst Richard believes he and Polly have a one in four chance of an affected child, the risk is in fact negligible.

At their next counselling session Richard does most of the talking. He says he would like to discuss alternative options for having children, including artificial insemination by donor. The counsellor is unsure what is the right thing to do. How should the discussion be managed?

What ought to be done if the couple separate and Richard returns to the clinic with a new partner asking for reproductive advice?¹⁰

Richard deciding not to have further children on the basis of inaccurate information, an additional ethical issue here concerns the question of *who is the patient*, or perhaps *to whom does the information belong*. As Richard and Polly attended the consultation together, it might be reasonable to assume that both have a right to be informed. They came together for information about their reproductive options and thus it would seem *prima facie* at least that both Richard and Polly have a right to a direct answer to their question. But this course of action presents ethical challenges of its own.¹¹

(ii) Pretest ethical issues in clinical genetics

It might reasonably be argued, that many of the problems arising *post-test* in clinical genetics might be avoided by greater emphasis on the discussion of possible difficult implications *before* testing. This would not avoid the difficulties arising in situations where despite such discussion patients continue to refuse disclosure or where the results are truly surprising. But a full and supportive exploration prior to testing would surely avoid *many* of these problems. Whilst this is certainly going to be true in many cases, there remains a range of situations in which discussion *before* testing, or the construction of a family history, reveal difficult ethical questions. For, when a genetic test is first discussed in counselling with a client or patient, perhaps in the process of constructing a family tree, it can sometimes become apparent that the test (or family tree) is likely to produce information either of use to, or revealing about, other people. This will usually be an identical twin or an intermediate relative between one who is affected and another who wants a test. In such cases the question arises of whether consent ought to be gained from the third party and/or whether any information produced should be shared with them. Consider the case in box 3.

Had the woman not mentioned her husband's opposition to a test she would certainly have been given access to the test and she would still in practice get the test despite having mentioned her husband's reluctance. Nevertheless, her comments about her husband's anxiety mean it has now

Box 3

A woman who is eight weeks pregnant requests a direct test for Huntington's disease on her fetus. She tells her geneticist that she wants the test because she recently discovered that there is a family history of the disease in her husband's family. One of his parents is affected so he is at 50% risk of developing the condition. She is adamant that she does not want to give birth to a baby who is at risk of developing Huntington's and says that the test will help her to ensure that this is not the case. In counselling she reveals that she has discussed this issue several times with her husband but he has always said he is not willing to take a test. He does not want to know his status and is extremely anxious about the possibility that he might develop the condition. The woman and her husband are currently separated but are seeing a relationship counsellor and trying to work things out. Despite this the woman says they tend to fight a lot. When it is explained to her in counselling that testing a fetus will, if it is positive, be to test her husband, she makes it very clear that she does not want to discuss it with him as he would inevitably make a fuss and attempt to stop the test going ahead. She wants to take the test in order to make an informed decision about her pregnancy.¹²

become clear that to carry out a test on the fetus would be to carry out a highly predictive test on the husband without his consent and against his wishes in a family situation in which this might come out, perhaps in a destructive way, at a later date. And this raises the legitimate question of what the moral obligations of the geneticist are to third parties in such situations.

Ethical considerations arising pretest might include the question of whether it is ever justified to require the consent of a third party before a test can go ahead if the test will reveal the third party's disease or mutation status, or whether a person's access to a genetic test should never be dependent upon the consent of another family member and concern for the confidentiality of the index patient and her access to health care ought to trump other considerations in all cases. A further ethical consideration might be whether prior agreement, with the individual patient herself or with the wider family, should be sought about how information resulting from the test is to be shared or dealt with post-test where this is likely to have implications for others. Some have suggested that this might be achieved either through informal encouragement of family discussion or perhaps by means of a more formal family agreement such as the so called "family covenant" proposed by David John Doukas.¹³

DISCUSSION

We have described three cases which we think capture some of the different types of ethical tension in the clinical geneticist's role between concerns on the one hand for individual patients and on the other for their families. There are, we have suggested, strong arguments in favour of both a *narrow* conception and a *broader* conception of the geneticist's role. Some of the arguments in favour of the *broader* conception that have emerged are:

- Genetic testing is predictive, interpersonal, and identifying. It is generally agreed that genetic testing should only be carried out with the informed consent of the patient. To test one person can in some cases be to carry out a highly predictive test on someone else. If we are serious about informed consent in genetic testing should we not apply the same standards in both cases?
- Many people requesting genetic tests are members of families that are in dispute (or communicate poorly) and whilst

they may say that they will, or will not, discuss the results of testing with their relatives, in practice they may go on to do the opposite. They may reveal (or not reveal) difficult information in a destructive and harmful way. This goes against another core principle of clinical genetics practice, that people should not be given tests or the results of tests without adequate counselling.

- Genetic testing of one person can produce information that will make predictive testing available to other family relatives that would not otherwise have been possible. This information may enable them to make more informed reproductive choices, to make choices about surveillance, or to avoid unnecessary treatment. If this information is not made available relatives may die or suffer serious harms as a result.
- In many cases, such as in cases of non-paternity combined with inherited disorders, particularly where couples come for testing together, the provenance of information is not clear. Why should one person be assumed to have sole right to the information?

Whilst these arguments are powerful, particularly in relation to cases such as the ones described above, there are also strong arguments in favour of the *narrower* conception of the geneticist's role. Some of these arguments are:

- To require consent from third parties before carrying out a test, even if this is predictive of others, would effectively be to offer them a veto over the availability of testing for the index patient and would in some cases mean that patients would have to be turned away and refused a helpful genetic test. Patients may die or suffer serious harm as a result.
- To require discussion with other family relatives would be to undermine the patient's right to confidentiality, which is a key to trust in the physician/patient encounter. It might mean that potential patients would not come forward for testing and treatment.
- To start on this road is to start down the slippery slope towards "paternalistic" medicine in which other family members and health professionals would be able to decide what is best for patients. Patients should be able to attend clinical genetics clinics in their own right without interference from outside and should be enabled to make free and informed choices about their health care and about whether they do or do not take genetic tests.
- In practice the broader conception would be likely to be unworkable. How would the genetics service ensure that all family members were adequately informed? What would be the practical and ethical difficulties of approaching and informing relatives who had not been referred for testing and had perhaps not even previously considered the issue?

What these cases and arguments show is that neither the narrow nor the broad conception of the geneticist's role is free of significant ethical difficulty. To hold strictly to either position has the potential to lead to harms of various kinds and to fail to take important obligations seriously.

CONCLUSION

Two key conclusions emerge from this discussion.

Firstly, the relative strength of the moral arguments in favour of the narrower and the broader conceptions will differ both between cases and within cases—that is, over time, where counselling is a long term affair. The assessment of the merits of a particular course of action will require moral judgment and some degree of agreement among clinical teams

about what constitutes good practice. This suggests that rather than formal ethics guidance or policy of a very *prescriptive* kind, geneticists and genetics teams will need to develop their own fairly sophisticated skills of ethical argument and reasoning and may perhaps benefit at least sometimes from support from an ethics committee or clinical ethicist in the resolution of such dilemmas.

Secondly, sensitive discussion of many of these issues prior to testing and a clarification of the centre's policy on them may help to make these difficulties easier to deal with when they do arise. In order for this to help with those issues arising pre-test any such discussion will have to take place fairly early on in the counselling process. The feasibility of this kind of approach depends in practice upon the achievement of broad agreement about what constitutes good ethical practice in cases such as these and this implies that this decision making, whilst made on a case by case basis, will need to be informed both by policy principles drawn up in the light of the development of broad public agreement about how the nature and role of clinical genetics ought to be defined, and by good communication between genetic centres. Such agreement will require the promotion of informed public policy debate. The increasing availability of genetic testing outside the clinical genetics setting (by other clinicians such as neurologists, cardiologists, paediatricians etc) will also mean that broader education and guidance will need to be developed here too and, given the support described above, and their substantial experience of dealing with and discussing these matters, clinical geneticists may perhaps be in an ideal position to offer such advice and guidance to other specialities.

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- 5 Clinical geneticists may also have a range of additional obligations with regard to the testing of children and fetuses but discussion of this issue lies outside the scope of this paper.
- 6 **Rhodes R**. Confidentiality, genetic information, and the physician-patient relationship. *Am J Bioeth* 2001;**1**:26–8.
- 7 This observation is based on the experiences of the authors in running ethics discussions in clinical genetics teams in Southampton and Oxford, and in the running of a national "Genetics Club", with Angus Clarke, at which members of clinical genetics teams from around the United Kingdom discuss the ethical issues arising in their daily practice.
- 8 For the purposes of this paper we take "pretest" to include not only situations before anticipated testing, but also situations where no test is available, or where testing has not been considered, but where a family history is to be constructed.
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