

# Consent and confidentiality in genetics: whose information is it anyway?

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Against a background of increasing regulation regarding access to medical information and the presentation of patients' confidentiality, the case of genetic information raises interesting questions about whether the application of general rules is appropriate in all situations. Whilst all genetic information is not equally sensitive, some of it is highly predictive. It also allows deductions to be made about other family members. It may not be regarded as particularly sensitive when compared to other types of medical information and those to whom it applies may not be as anxious about preserving their confidentiality as compared with—for example, the prospect of seeing research into cause and cures for rare diseases put in hand. These distinctions also find resonance with the general public. Resolving conflicting tensions will require subtlety, not a blunt "one size fits all" model.

Against a background in which trust in the professionalism of clinicians and scientists has been shaken by a number of well publicised abuses, it is timely to discuss the issues of consent and confidentiality and the ways in which proper respect can be given to the wishes of those who seek medical treatment or who wish to participate in biomedical research.

The failure of existing regulatory mechanisms at Bristol, where professional shortcomings were known about but not acted on, and Alder Hey, where the trust of families in vulnerable situations was violated over a prolonged period for no good reason through the actions of an alleged "rogue pathologist", have resulted in a general tightening of the regulations about what may or may not be done and under what circumstances to patients and their samples. The inadequacies and competencies of the current situation are addressed in the forthcoming report of the Retained Organs Commission. This situation is likely to result in legislation, which will create offences under criminal law for those who fail to comply with the requirements of good practice in future.

The general tightening up of the regulations regarding consent and the protection of patient and family interests is a fact of life. In the light of the scandals referred to above it is hard to argue that it is not a good and a necessary development and I would be among the first to agree with the argument for a greater emphasis on seeking informed consent from patients and, in cases where individuals are unable to consent for themselves, from those who act in their interest.

In this as in other areas where the might of legislation is invoked to resolve a difficulty, however, hard cases can make bad law. Care must be taken in drawing the line between what is and what is not permissible, to ensure that an appropriate balance is struck between the interest and the rights of the individual and the legitimate interests and rights of the community to which that person belongs.

Not all medical information is equally sensitive. To treat it as if it were so is potentially to constrain some beneficial applications in ways which even those who might not wish to participate would not want to happen.

There is no absolute right to confidentiality, but there is a widely held rebuttable presumption that information obtained in the course of a medical intervention will be held in confidence by the person who obtains it unless there is a very good reason for disclosing it. This trust is the basis on which the system is able to operate and without it medical care and

research would grind to a halt. But a "one size fits all" model does not work well and in the case of genetic information, rigid application of the rules can cause significant problems which are in no one's interest, be they patient or professional.

## WHAT IS "GENETIC" INFORMATION?

By "genetic" information I mean information which may be obtained by a variety of routes that enables either a diagnosis or a prediction of either a current or a future health status that can eventually be directly related to identifiable alterations in a person's DNA. This information may be derived by direct observation of the DNA or chromosomes, through family history taking, from biochemical tests, or by clinical observations. The important element in its use in medicine or research is not the route by which it has been derived, but the reliability with which it can be used to devise and sustain valid conclusion.

Clearly, not all genetic information is equally powerful. To treat it as if it were is clumsy. Some is highly predictive of future health. Its acquisition and disclosure requires careful thought and the application of carefully designed and implemented protocols. Most is not and to treat it as if it were may create overkill. Unfortunately the limits on genetic information are not well understood. Too often it is treated, particularly by the media, as if it were like a train leaving the station and travelling along a track on which there were no stops, sidings or diversions, gathering speed inexorably until it crashes into the buffers at the terminus to the great detriment and damage of all around.

In the case of the gene for Huntington's disease there is some justice in this analogy, but even here, given our current knowledge, detection of the mutation only tells you that you will get the disease. It does not tell you when, exactly, you will start to show the symptoms, or what the precise pattern of progression will be in you—or how the interplay between you and those around you will affect their ability to provide the care and support you will need.

But Huntington's disease provides a poor model on which to base more general models for deriving consent or for protecting confidentiality. The Nuffield Council on Bioethics in its report on genetics and mental health<sup>1</sup> found that when the very rare genetic forms of mental disorder, such as Huntington's or familial Alzheimer's are eliminated, the highest increase in predictable risk of future mental health problems that could be attributed to observable genetic phenomena was

sufficient to raise the risk above that of the population as a whole by only about two to three per cent. Compare this with the predictive power of environmental features with no obvious genetic component of future mental health status—such as divorce, loss of a home, the death of a close relative, and redundancy, then the relative importance of genetics in such cases becomes apparent.

Even where the gene in question is significant and highly penetrant, the opportunity to intervene to reduce, prevent or treat disease alters the way in which the information should be treated. For example, in the case of familial hypercholesterolaemia or familial bowel cancer knowledge of one's genetic status can be positively beneficial in that it allows intervention and can prevent unnecessary disease and disability. Development of an appropriate model for service delivery should take account of this, as those with experience of the condition and of providing services and support for those affected will quickly tell you.

### WHAT INFORMATION IS IT ANYWAY?

Genetic information, by definition, does not apply uniquely to individuals. Knowledge of one person's genetic status allows us to draw inferences about those to whom he or she is related. In most cases these will be of so little power or reliability as to be trivial, but in a number of instances, most notably those connected with significant single gene disorders, the inferences that can be drawn may be substantial. In such a situation, if I know something about myself does my brother or sister have a right to know it too, given that it also affects them? At what point is my wish to protect my privacy overridden by their wish or need to know in order to avoid potentially harmful consequences?

Work undertaken by the Genetic Interest Group<sup>2</sup> indicates that, among those living in families where there is a diagnosis of a substantial risk of genetic disease, there is a strongly held view that such information should not be seen as the private property of the individual. Rather it should be seen as family information held in common by all those to whom it applies. Of course, like all views that are simply stated, interpretation in practice is infinitely complex and subject to all the vagaries of human nature. The ideal world notion of important information being sensitively and carefully disclosed in a caring and supportive way does not always hold true. In some situations the giving or withholding of information and the manner in which it is done can be an exercise in power or a reflection of other aspects of the family context!

The familial nature of this type of genetic information also places on the professional the obligation to define where he or she stands in relation to the maintenance of individual confidentiality or the decision to override it in the pursuit of the greater good (or perhaps the lesser harm). Again, the view from families at risk is that, in the case of severe genetic disease where there is a potentially avoidable harm, professionals ought to be willing to override the wishes of the individuals and make the information available. This is not to suggest that people should be cavalier in their attitudes and disregard patients' wishes. To do so would very quickly result in legal challenges and close attention from professional and regulatory bodies such as the Royal College and the General Medical Council (GMC). Rather it is to postulate the need for the development of protocols and frameworks within which the decisions about the appropriate course of action in a given set of circumstances can be taken and recorded, in ways that are likely to protect the best interests of all concerned. While such a framework cannot guarantee that legal action will be avoided, it will certainly minimise the chances of such litigation being successful.

### RESEARCH

Research ethics committees rightly require that the confidentiality of those participating in approved programmes of

research and development is not compromised. Whilst this is a feasible requirement in many types of research, when looking into causes and cures for rare genetic disorders it may not be possible. Indeed it may be counterproductive to try and achieve this. It may also be contrary to the expressed views of those with the condition in question.

Given the fact that it can take only two or three pieces of information to identify an individual to a very high degree of confidence, preserving the confidentiality of those with rare genetic disorders is, in many cases effectively impossible. In the case of many rare genetic disorders if you know the diagnosis, the health authority or trust and the name of the referring clinician then you can be pretty sure that you know the person. Indeed, for the research to be successful it may be essential that you are able to go back to the families and this indeed is likely to be what they would want, for it is only through the promotion of research that they will gain understanding of the situation in which they find themselves and eventually can have hope of making progress towards a cure.

Striking the right balance is difficult, particularly at the interface between research and clinical practice where initial observation of unexplained clinical signs and symptoms can move into more structured investigation almost imperceptibly, and where the nature of the referral almost begs the question about getting involved in research. The "Query xyz disorder or if not, what?" referral letter sets out both the clinical and the research agenda and it also asks the question which the families want urgently to be answered. Putting too rigid a bureaucracy around the means to answering it will frustrate these wishes. It will also act as a particular disincentive to those contemplating research with rare disorders and make the costs of doing such work rise significantly—an irony when it is often the fund raising efforts of those with the condition themselves which make it possible to contemplate doing the work in the first place.

Again, I am not arguing for *carte blanche*. Research must be regulated and it must be subject to proper ethical approval if potentially vulnerable individuals are not to be exploited unreasonably. But what works for a large scale multicentre clinical trial of a new drug for a common disease may be inappropriate for a study of a very rare genetic disorder requiring samples and case histories from clinicians throughout the UK and often further afield, each of whom may only know of one person or family with the condition in question. It is all a question of getting the balance right and of listening to the wishes of the individuals and the families who are at the heart of the problem.

### SETTING THE CONTEXT

As has been stated earlier, not all genetic information is equally sensitive. It is also important to recognise that, whilst some of my genetic information may be highly sensitive, it may not be the aspect of my medical history that I am most concerned to keep private.

Thus my confidentiality may be severely breached by the unwarranted disclosure that I have cystic fibrosis (CF), but it is not—for example, significantly further damaged by the subsequent revelation that it is a consequence of the Delta F508 mutation. And whilst I may not mind your knowing that I have CF, I might be absolutely devastated were you to find out my HIV status, the fact that I had had several abortions and that my social father is not, in fact, my biological father. Improper disclosure of any of these could arguably do more damage than disclosing straightforward genetic information.

Rather than treating genetic information as if it were uniquely different and so warranting special treatment, we should be more concerned to ensure that existing mechanisms work well to protect individuals and preserve confidentiality in ways that are appropriate and necessary.

### WHAT DOES THE PUBLIC THINK?

The discussion about access to medical information is often predicated on assumptions about the views of the general public. Yet when the question is asked of representatives of the public, the evidence is that the public is capable of making very sophisticated distinctions between different uses for medical information. The National Health Service (NHS) is currently engaged with a pilot project in West London known as "NHS LifeHouse". This is intended to link electronic patient records held in various locations in primary and secondary care to create a comprehensive electronic health record that will deliver point of care information that is relevant and up to date, whilst at the same time providing a resource for research and for health care planning.

Mindful of the fact that there are sensitivities associated with the use and abuse of computer records holding personal health data, this project takes as its baseline a commitment to transparency and public endorsement for its aims and objectives. This led to a number of consultations,<sup>3</sup> the results of which may provide some comfort for those who seek to regulate and control access to personal medical information in an appropriate way.

In respect of the clinical uses of personal medical information, the assumption seems to be that clinicians will and should talk to one another and that relevant information ought to be passed around between professionals who share an agreed code of conduct. When it comes to research, there is more difficulty, because many people do not see the NHS as an organisation which is research orientated. If the nature and purpose of the research is explained, however, then people are generally happy for this information to be shared, provided they are asked and that the information to be shared can be shown to be relevant. Similarly for service planning purposes, where personal identifiers are stripped away. Even the private sector can get a look in if the information is anonymised and the benefits are seen to flow to the NHS, rather than in the opposite direction, leading one to the conclusion that people are aware of the complex uses that can be made of medical information, and are generally content that this should be the case when there is a clear explanation as to why, or that those who wish to regulate in order to protect confidentiality and ensure consent will have to be considerably more subtle than might have been anticipated by people seeking a quick fix to a current problem.

### CONCLUSION

To conclude, I would like to return to my starting point and to the notion that hard cases can make bad law. I would like to leave you with a problem to think about and see how you would resolve it. It takes the form of a story.

Let us suppose that my brother and I are estranged. We share the same general practitioner (GP), who is aware of the fact that we are brothers, but who does not know about the hostility between us. Let us further suppose that I receive a diagnosis of a fatal, late onset dominantly inherited, genetic disorder. This means that my brother is at 50% risk and my GP knows this. Under the terms of the Data Protection Act, data controllers are obliged to tell data subjects if they hold significant information about them. My brother is unaware of the risk. I wish my confidentiality to be respected. You are my GP. You are also a DPA data controller. What do you do? Do you contact my brother and let him know you have information about him? Do you try and ascertain if he wants to know it? What if he not only doesn't want to know, but does not want to know that there is something he does not know? And what about my confidentiality? Oh and by the way, I also do not want my wife and children to know!

Answer, on a postcard, to . . . . .!

### DISCUSSION

Julian Peto was pleased to hear Alistair Kent's views and would like to see the survey of public attitudes on data and confidentiality. He believed the vast majority of the public would be happy to allow epidemiologists and other medical researchers to use their records once they understood the potential benefits of the research and how their data would be used. But first, the public need to hear the arguments for the sharing of records. Alistair Kent agreed, adding that as long as patients understood the benefits of research they would be happy to give the go ahead to the use of their records. Such consent might be given generally, not for each specific case.

Onora O'Neill asked how a family at risk of a genetic disease could give an objective view. She also asked what was meant by family. Was it the social concept of family, or the genetic concept? In reply, Alistair Kent said that research carried out by the Centre for Family Research had revealed idiosyncratic ideas of what families are, based not just on genetics. He also advised caution before resorting to complex legislation on consent. This could result in inflexible rules that would not allow for changes to be made.

### REFERENCES

- 1 **Nuffield Council on Bioethics.** *Mental disorders and genetics: the ethical context.* London: Nuffield Council on Bioethics, 1998.
- 2 **Genetic Interest Group.** *Confidentiality guidelines.* London: Genetic Interest Group, 1998.
- 3 **Sykes W, Hedges A.** *Preliminary focus groups: provisional notes.* London: NHS LifeHouse, 2001.