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.

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 health¹ 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 fact that the power of environmental factors with no obvi-
ous 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 hypercholes-
terolaemia 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. Develop-
ment 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 such 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 infer-
ences that can be drawn may be substantial. In such a
situation, if I know something about myself do 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 over-
ridden by their wish or need to know in order to avoid poten-
tially harmful consequences?

Work undertaken by the Genetic Interest Group 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 car-
ing 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 confi-
dentiality 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 regula-
tory 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 litiga-
tion being successful.

RESEARCH

Research ethics committees rightly require that the confiden-
tiality 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 look-
ing 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 diag-
nosis, the health authority or trust and the name of the refer-
ring 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 under-
standing 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 fami-
lies 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 inap-
propriate 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 a
difficult 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 then treating genetic information as if it were
uniquely different and so warranting special treatment, we
should be more concerned to ensure that existing mecha-
nisms work well to protect individuals and preserve confiden-
tiality 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 healthcare 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,1 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