Genetic testing without consent: the implications of the new Human Tissue Act 2004

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Despite its focus on consent the new Human Tissue Act 2004 allows for testing without consent where a relative could benefit

In recognition of the fact that genetic test results in people can have implications for close relatives, the new Human Tissue Act 2004 allows for a direction to access a person's tissue so that testing can be carried out for the benefit of a relative, without the consent of that person. Clinical practice governed by common law and statute, before this act, acknowledged that the disclosure, without consent, of the results of a genetic test, is ethically permissible in extreme situations for the benefit of a relative. New testing for the benefit of a relative without donor consent challenges the principle that informed or adequate consent should be obtained before a genetic test with considerable predictive powers is carried out. The debate surrounding the analogous situation of HIV testing concluded that in very rare cases, disclosure of an HIV result without consent may be justified, but testing without consent is a professional misconduct. This part of the legislation is controversial and deserves further debate. The Human Tissue Authority should take measures to limit the potential harm that can be caused by this provision.

Introduction

After much debate, the new Human Tissue Act 2004 was passed by parliament in November 2004. Implementation of this act may be staggered, but it will be fully operational in May 2006. This act has several provisions that have implications in clinical practice for the use of stored tissue or samples containing cells. These provisions allow the Human Tissue Authority to permit access to human tissue for the purpose of obtaining scientific or medical information about a person (donor) for the benefit of another family member, without the consent of the donor (schedule 4, paragraph 9). The importance of this is that much current genetic testing for relatives who are at

risk is only accurate if a causative genetic change has first been identified in an affected relative.

The acknowledgement of the possible use of individual samples in predicting risk in their family members is welcomed, but the distinction between disclosure of test results to relatives, where the donor has consented to the test, but not to the disclosure to relatives, and new testing on the donor without consent is blurred by this legislation.

Shared nature of genetic information: implications for clinical practice

Certain genetic test results can provide people with diagnostic or predictive information about a particular condition or disease. For example, a woman with a strong family history of breast cancer and ovarian cancer may wish to undergo prophylactic surgery to reduce her risks if it can be shown that she has inherited the familial mutation responsible for the history. As multiple large genes may be associated, the sensitivity of genetic testing in the "at-risk" woman is greatly increased if the familial mutation can first be identified in an "affected" relative. Without this, the negative predictive value of a genetic test is currently low. If the familial mutation is known, however, a highly accurate predictive test can be available for management options such as prophylactic surgery. The outcome of such a test can have an effect on the treatment of the patient and can also have social, psychological and financial implications. Current clinical practice holds that genetic testing, especially predictive genetic testing, should be carried out only after discussing the potential implications of a result and obtaining adequate consent. This is reflected in several policy statements made by public bodies—for example, "Genetic information should not be obtained or held

without a person's consent."¹ Failure of a clinician to obtain consent would be a breach of accepted practice and policy guidelines, but can also lead to disciplinary action by the General Medical Council (GMC).

The shared nature of genetic information means that gaining access to blood or tissue of an affected relative can be important in the clarification of risks and management options for a currently healthy person. Genetic testing in people can, to a greater or lesser extent, disclose information about risk or diagnoses in their blood relatives. A person's genetic test result may clarify the risk of disease, surveillance or prophylactic measures in another member in a way that would not be possible without the use of this result. Although most families presenting to clinical genetics services are happy to share the results of tests, at least in part, because of a desire to help family members, in other families, relationships may be more strained or different members may simply have lost contact with one another. People may have a legitimate fear that they will be blamed for passing on "bad" genes, or, alternatively, they may have a genuine reason to believe that such information would harm their relatives and may withhold it to protect them. Genetics services have acknowledged such tensions since their inception and have highly skilled counsellors who are well versed in negotiating familial interactions. This is reflected in the deliberations of the members of the UK Genetics Club,² where individual cases are discussed on a regular basis. Worries about the stigma of genetic information may, of course, prevent people from attending clinical genetics services in the first place. In the hypothetical case scenario described below none of the people have had contact with genetics services and this is likely to reflect reality, as clinical genetics remains a small specialty.

Clinical example

Brenda and Sarah (fictitious names) are two sisters aged 42 and 46 years, respectively, who have a strong family history of colorectal cancer and uterine cancer. Four of their close relatives had developed and subsequently died from these cancers in their 40s and 50s. No tissues or samples are believed to be stored from any of them. Sarah was recently diagnosed with uterine cancer; she had a hysterectomy and is currently undergoing chemotherapy. She is not on the best of terms with Brenda and the two of them rarely speak. Brenda has been told by a colorectal surgeon, whom she sought advice from, that it is highly likely that the cancers in her family are

caused by a highly penetrant mutant gene. Although Brenda can herself be tested for such a mutant gene, its absence would not distinguish if she had inherited the mutant gene and whether the mutant gene was not identifiable by the current technologies. If the mutant gene can, however, be identified in an affected person in her family, then the test would accurately predict whether surveillance and possible prophylactic surgery were indicated for Brenda. Brenda attempts to contact Sarah to facilitate such testing, but Sarah is not yet ready to contemplate what she considers a highly important test, because she is unwell and because she is concerned about how her four children aged 12–22 years would receive such information. Brenda considers this unreasonable. She wishes to know whether to undertake a prophylactic hysterectomy and oophorectomy (because of the risks of uterine and ovarian cancer in such kindreds). She applies to the Human Tissue Authority for a direction to have a genetic test carried out on the stored tissue from Sarah.

Debates on the tension, posed by the prerequisite of familial information to provide accurate predictive genetic testing in another person, have tended to conclude that, in very rare circumstances, and where there is a serious harm that may be prevented, disclosure, without consent, of an already known test result may be justified.^{3,4} Opinions are, however, by no means unanimous and others would argue that disclosure may not be made under any circumstances to relatives without consent.⁵ These debates have covered the situation after people have consented to a genetic test, been informed of the result, but decline consent to disclose this result to relatives. General consensus has previously existed that new testing without consent—even when the potential benefits for family members may be large—would not be permissible because of the implications this can have for the donor. The Human Tissue Act 2004 therefore sets a surprising precedent in that it permits new testing of a stored sample without that individual's consent.

Provisions of the Human Tissue Act 2004

One of the most salient provisions for clinical genetics practice in the new Act is that the non-consensual testing of DNA is now a criminal offence under section 45. To be prosecuted for this offence, the person must have “bodily material” (material that has come from a human body, and consists of or

includes human cells) with the intent that the DNA be analysed without “qualifying consent” and the results must be used for purposes other than those listed as an “excepted purpose” (eg, medical diagnosis and treatment of that person) as stated in the Act. “Scheduled purposes” lay down where consent is generally required and include obtaining scientific or medical information about the donor that may be relevant to another person or in the future. If people were found guilty of an offence under this section, they would be liable for a fine or imprisonment, or both.

There are two situations when the Human Tissue Authority can make a direction to dispense with the requirement for consent. Firstly, when it is not reasonably possible to trace the donor of the material. This would include situations in which the donor has moved countries or has lost contact with clinical services or with the people who stand to benefit from the use of the material or the results of an analysis of DNA. Secondly, when reasonable efforts have been made to obtain the donor's consent to use the material for that purpose and the donor has not made any decision. In this case, there may be contact with the donor but he or she has not made a decision to consent to the use of the stored tissue. This situation applies in our case scenario described earlier. If the Human Tissue Authority gives a direction, then DNA analysis can be carried out on the bodily material of a person, such as Sarah, without consent, and disclosure of the results to Brenda would follow. In effect, the Act has conflated the clear distinction made in clinical practice between carrying out a genetic test and disclosure of the results of a test, because once a direction is made, both activities are permissible.

The Act says nothing about access to, or use of, genetic test results to which a donor has consented, but where there is no consent for disclosure of these results. Such cases remain governed by professional guidelines, such as those from the GMC, the common law on confidentiality, the decisions of the European Court of Human Rights and the Data Protection Act 1998.

It is also unclear whether both the holding of bodily material and the intention to analyse DNA without qualifying consent require to be prosecuted for the criminal offence of the non-consensual DNA analysis. One interpretation of the legislation is that if a laboratory holds only extracted DNA (with no remaining tissue cells, a common practice in genetic laboratories), then a person cannot be prosecuted under the act because he or she

did not have bodily material. The provision would then have a restricted application to intention to analyse DNA in stored tissues (collected after the Act comes into force). Neither the Act nor the provisional codes of practice of the Human Tissue Authority discuss whether or how Sarah should be given the result of her DNA analysis, to which she has not consented. Giving Sarah the result can be problematic because she has not asked for it; withholding the result may be equally problematic, as it may carry important information about risks or management.

Why is this provision of concern?

This provision allows a solution for people in a situation such as that of Brenda, but it also undermines many of the principles that clinical practice has been founded on. One of these is that consent must be obtained before a genetic test is undertaken. We believe that a fundamental difference exists between testing with consent, informing that person of the result, but then disclosing that result to prevent harm in relatives (without the donor's consent to disclosure), and carrying out new testing without the donor's consent for the benefit of a relative. This would also apply to other areas of healthcare. Although disclosure of a person's HIV status to contacts at risk without his or her consent may be justified in rare circumstances, it would be quite a different matter to carry out the HIV test without consent. Testing for HIV without consent has been the ground for disciplinary proceedings by the GMC. The risks associated with some kinds of genetic testing, in terms of its prediction of disease and its special sensitive nature, would be equivalent to, or of greater concern than, that of testing for HIV.^{6,7}

The provision allows bodily material collected and stored with consent for one purpose to be used without consent for another purpose, such as genetic testing. For example, if tissue is stored after an operation, a direction can be obtained for this to be used for a genetic test to benefit a relative. The donor has not consented to a genetic test but rather to the storage of the tissue, possibly as part of his or her medical record. To then use this bodily material for a genetic test, which has far-reaching implications, is contrary to the basis of the original consent.

If genetic testing is carried out in the interest of a relative, it is highly likely that the result will also have implications for the donor. For example, in Sarah's case, her risk of future colorectal or other cancers can be inferred from the test to which she has not consented,

and there could be psychological and social consequences (feelings of blame, perceived stigma and worries about insurance to name but a few). Good clinical practice requires voluntary adequate consent of the people requiring a detailed discussion of the implications of the test. Not everyone at risk of a genetic condition will want to know their genetic status, or find this an easy decision to come to. The right not to know should be respected.⁸ There may be good reasons why people decide not to have a genetic test at a particular point in their lives, but such reasons may be difficult to convey to relatives with whom relationships are very strained. Dispensing with the need for informed consent by a direction from the Human Tissue Authority has the potential to severely undermine good clinical practice and can place people who are the subject of such a direction in an impossible position.

If a direction is applied for, donors must be given notice of the application, with the resultant probability that they will be forced into making a decision that they are not ready to make. They may decline consent when a more informed discussion with clinical services can facilitate it. Such uninformed decisions may be more likely to inflame other family members, entrench bad relationships, or lead to the donor's decision not to have the genetic test. The treating clinicians may also be placed in a difficult situation, as they may be compelled to advise the donor to refuse to consent to negate the direction made by the Human Tissue Authority. This assumes that a clinician would necessarily be participating in such negotiation, a clarification of which we would welcome. Further adverse consequences may include difficulty in giving the donor the results of the genetic test or the same opportunities for counselling those people who have made a positive decision to have a genetic test. It also places the decision-making process outside the clinic and the procedures and safeguards that operate in this context.

It is unclear what will constitute a reasonable effort to contact the person under the act and therefore how the Human Tissue Authority will interpret this requirement. Under the act, anyone can apply for a direction from the Human Tissue Authority to access human tissue to carry out genetic analysis. As the act stands, this does

not need to be a clinician nor does the request need to be channelled through a National Health Service clinical genetics clinic. Such details have not yet been clarified in the draft guidelines of the new Human Tissue Authority that were put out to public consultation until late 2005. Although these guidelines state that such circumstances are expected to arise infrequently, it does not provide guidance on how they would be handled when they did arise, or what would happen if they arose more frequently than rarely.

Before the Human Tissue Act, careful deliberations on whether to disclose the results of a genetic test that has been undertaken in a clinical setting would have been decided by the clinical team on a case-by-case basis. According to Liddell and Hall,⁹ such decisions would have been lawful, provided that this conformed with the common law public interest defence to the breach of confidence doctrine, Human Rights Act and the Data Protection Act 1998. The Human Tissue Act now hands such deliberations to the Human Tissue Authority, a body that does not have an intimate knowledge of the case at hand, does not participate in the care of the patient and does not currently have clinical genetic expertise.

Conclusion

We are concerned about the provision in the new Human Tissue Act that allows the analysis of DNA without consent when someone has not yet made a decision. With this provision now enshrined in law, the Human Tissue Authority will have to carefully formulate its guidelines to determine:

- who can apply for a direction—whether it is a person or it must be channelled through a National Health Service clinic;
- what would be considered reasonable efforts to get the donor to decide, bearing in mind that coercion may be perceived; and
- whether the individual will be informed about the results of a genetic test carried out without consent.

Disclosure of a genetic test result without consent is never taken lightly in clinical practice and clinicians must be satisfied that they have made all efforts to obtain consent and that they are preventing serious and imminent harm to a relative. This provision in the

Human Tissue Act allows a new genetic test to be carried out, without consent and without the safeguards that have existed in clinical practice to protect patients and families. Although we recognise that the law before the Human Tissue Act did not always provide clear directions on how to resolve the dilemmas raised by the shared nature of genetic information, we question whether these new provisions adequately deal with this gap. We suggest that these provisions are a radical departure from the principles that underpin clinical practice and would welcome a wider debate on this issue.

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REFERENCES

- 1 **Human Genetics Commission**. *Inside information*. London: Human Genetics Commission, 2002.
- 2 **Genethics club**. <http://www.genethicsclub.org> (accessed 1 May 2006).
- 3 **Lucassen AM**, Parker M. Confidentiality and serious harm in genetics—preserving the confidentiality of one patient and preventing harm to relatives. *Eur J Hum Genet* 2004;**12**:93–7.
- 4 **Lucassen AM**, Parker M. Genetic information: a joint account? *BMJ* 2004;**329**:165–7.
- 5 **Ashcroft A**, Lucassen A, Parker M, et al. *Case analysis in clinical ethics*. Cambridge: Cambridge University Press, 2005.
- 6 **General Medical Council**. *Serious communicable diseases*. London: General Medical Council, 1997.
- 7 **Dyer C**. GP reprimanded for testing patients for HIV without consent. *BMJ* 2000;**320**:135.
- 8 **Laurie G**. Protecting and promoting privacy in an uncertain world: further defences of ignorance and the right not to know. *Eur J Health Law* 2000;**7**:185–91.
- 9 **Liddell K**, Hall A. Beyond Bristol and Alder Hey: the future regulation of human tissue 13. *Med Law Rev* 2005;**170**:186.