

# Recording, using and sharing genetic information and test results: consent is the key in all medical specialties

Peter A Farndon, on behalf of the Joint Committee on Medical Genetics

**Peter A Farndon**

MSc MD FRCP,  
Past Chairman,  
Joint Committee on  
Medical Genetics;  
Professor of  
Clinical Genetics,  
University of  
Birmingham

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*‘You can’t draw and record a family tree without the permission of all the members shown on it – the Data Protection Act won’t allow it.’*

*‘You can’t take blood from an adult with mental incapacity for a genetic test just to help other members of the family.’*

*‘Medical confidentiality won’t allow my laboratory to release a DNA result to allow genetic testing to be offered to another family member.’*

These statements are not wholly correct but were in wide circulation a few years ago. Some health professionals appeared to believe that genetic information in itself was different from other healthcare information and should be handled differently, or preferably not handled at all. Others believed that such matters were solely the province of the specialist genetic services. According to reports received by the Joint Committee on Medical Genetics, uncertainty appeared to be affecting clinical practice. The concerns of some health professionals appeared to be mirrored in public concern about the collection, storage and use of personal genetic information in general, but use for clinical purposes was strongly supported by the public.<sup>1</sup> Clinical experience in genetics centres also stressed that the majority of individuals wished their genetic information to be used for the benefit of other family members.

The majority of testing for single gene disorders and inherited chromosomal anomalies continues to be organised through regional genetics centres but patients are often identified through non-genetics colleagues. How should they deal with the implications of the result to the wider family? What is the

status of a family tree taken from one family member, and can its information be shared? These are issues which involve all specialties dealing with patients with genetic disorders. How can they be incorporated into clinical practice?

Fortunately, the Joint Committee on Medical Genetics set up a working party to review current clinical practice and to offer guidance and has just published its report, *Consent and confidentiality in genetic practice: guidance on genetic testing and sharing genetic information*.<sup>2</sup> The Working Party carried out a questionnaire survey of genetics units in the United Kingdom, took medico-legal advice on current legislation and encouraged wide debate and consultation. As well as detailed guidance in the main text, flow charts summarise recommendations for clinical practice when requesting information and samples during a consultation and when sharing and disclosing genetic information. A third flow chart outlines the implications of the provisions of the Human Tissue Act 2004 for genetic analysis of stored samples or archival pathological material. Although the Working Party was set up in response to concerns from genetics professionals, the discussions and recommendations of the report are applicable to all specialties.

**General aspects of consent and genetics in practice**

The accepted principles of medical consent and confidentiality<sup>3–5</sup> underpin the uses of personal genetic information in clinical practice. Apart from in exceptional circumstances, consent should be obtained prior to a clinical or laboratory test with genetic implications being undertaken and before medical genetic information is disclosed. There are additional issues to consider, however, particularly when pedigree information and genetic test results need to be shared for the benefit of other family members. These are discussed in the following sections.

**Potential health benefits for other family members through sharing pedigree information and genetic test results**

A family history (or pedigree) may assist in making an initial specific genetic diagnosis or to form a view

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about mode of inheritance and recurrence risks for the person originally referred, but it can also identify other family members who are at risk of being affected by, or being a carrier of, a genetic disorder. In families with an inherited predisposition to breast or bowel cancer, this can save lives.

Understanding the genetic relationships of family members may be required to determine whether a genetic test result from one family member can be used to generate information for another member. Sharing the laboratory result allows a specific test to be performed for the known genetic anomaly, rather than having to repeat a general screen to see if an abnormality can be found.

It is therefore entirely appropriate for best clinical care that relevant information is transmitted to appropriate health professionals. Family members may need to be contacted to offer testing to define their risk of being affected or of being a carrier of a genetic disorder, or to arrange for samples to be stored for future analysis should a test become available. It is therefore vital to have discussed the potential sharing and use of personal genetic information and samples and to have obtained appropriate consent.

### Obtaining consent to share genetic information or genetic test results

As most individuals express a positive desire that pedigree information and genetic test results should be available to other family members to assist in diagnosis and medical care, including assessment of genetic risk, it is good practice to document this. The Human Tissue Act 2004 requires that consent has been obtained specifically for the use of cellular material to assist in the care of relatives. A handwritten note in the medical records is sufficient, or a signed consent form may be used.

### What should be included in the discussion requesting consent?

The report recommends that the following genetics issues are discussed during the consent process:

- the use and sharing of information (eg pedigree, diagnosis, affected/carrier status, test results) with other family members for their benefit
- the nature of the testing to be undertaken and its implications
- the possible prolonged nature of the testing process
- the possibility that testing may reveal unexpected results depending on the particular analyses used
- the storing of samples
- the use of samples for quality assurance, education and training
- the sharing of information with health professionals including the primary care team.

The Joint Committee has recommended that genetic laboratories consider adding the following statement on laboratory test request forms to assist clinicians:

In submitting this sample the clinician confirms that consent has been obtained:

- a) for testing and possible storage
- b) for the use of this sample and the information generated from it to be shared with members of the donor's family and their health professionals (if appropriate).

### Consent versus confidentiality

Where consent for the above activities has not been requested, or its extent is unknown, genetic testing for other family members can be compromised. Attempting to verify past consent or newly seeking consent from the information or sample donor is considered the optimal situation. This may not be possible because contact has been lost. Alternatively, it may not be clinically appropriate because the family member seeking information may be concerned about compromising his or her confidentiality – for instance, a pregnant relative wishing to undertake prenatal diagnosis. As in several other situations outlined in the report, a clinical judgement has to be made case by case about the balance of harm to family members – a balance between seeking consent and preserving confidentiality.<sup>5</sup> There may be good reason to believe that more harm may result to a family member by not using the DNA sample or test result than would result to another member through their use without confirmation that consent had been granted. By routinely requesting and documenting consent to share information and use stored samples, such situations would be avoided; the Joint Committee asks those ordering a genetic test or recording a detailed pedigree to encompass this extension to their responsibility.

### Disclosure of information from family pedigrees and of test results

The Information Commissioner gave advice that information on a family pedigree can be passed between health professionals if necessary for medical purposes without the explicit consent of all those shown on the pedigree (under Schedule 3 of the Data Protection Act 1998). The person giving the pedigree, however, should be informed that it may be used to determine mode of inheritance, shared with family members if they seek advice and shared with other health professionals if necessary for the care of family members. It is good practice to release only information relevant to the clinical purpose; for instance, it may not be necessary to give names on parts of the pedigree.

Medical information is sometimes needed about deceased family members. Some medical records departments have declined access except with the consent of the deceased's spouse, but they may use their discretion under the Access to Health Records Act 1990 to disclose medical information upon request from the blood relatives of a deceased person where there is a risk of an inherited genetic disorder.

When releasing test results, it may be possible to release technical information about a 'family mutation' rather than specific personal information about the sample donor.

### Adults with mental incapacity

The majority of people are willing to undergo genetic investigations to help other family members, even if they derive no direct benefit themselves. The Genetic Interest Group argued strongly that it should not be presumed that an adult with incapacity would be less altruistic,<sup>6</sup> and many people with learning difficulties have the capacity to consent if time is spent explaining the issues in simple language.<sup>7</sup> When considering the taking of a sample from an adult with mental incapacity to aid the wider family, the benefit to the incapacitated adult must be clearly identified (which might include the positive effect on the care of the adult through wellbeing of the relatives<sup>8</sup>) and it should not be possible to obtain the same genetic information through tests on other family members. The report discusses these issues and the genetic testing of children.

### Consent for analysis of DNA in cellular tissue and the Human Tissue Act

In general, a living competent adult or child must give consent to the analysis of his or her own DNA in cellular material and the analysis may be for any purpose as long as specific consent has been given. The gaining of such 'qualifying consent' is sufficient to prevent an offence of 'DNA theft' under the Human Tissue Act. Those with parental responsibility can give consent for a child.

Specific consent is required under the Act to use cellular material to obtain genetic information for another person (but not extracted DNA whose use falls within professional guidelines). This legal requirement reinforces the need to ensure that using cellular material to assist relatives is explicitly discussed and documented in clinical practice.

For the purposes of genetic analysis on posthumous cellular material, the consent of any qualifying relative (as defined by the

Act) will suffice. This will be particularly helpful for families who wish to obtain stored tissue for extraction of DNA from a deceased family member, usually for mutation detection.

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