

# Consent and confidentiality in genetic practice

Guidance on genetic testing and sharing genetic information

A report of the Joint Committee on Medical Genetics

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April 2006



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**Royal College  
of Physicians**  
Setting higher medical standards



**Royal College of  
Pathologists**

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The British Society for Human Genetics

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# Introduction

## Overview

This report from the Joint Committee on Medical Genetics (hereafter the Joint Committee) is a response to requests from genetics professionals for clarification of issues of consent and confidentiality in clinical practice, particularly with regard to the requirements of the Data Protection Act 1998 and the Human Tissue Act 2004. Although the report specifically addresses clinical practice in the genetics services, we believe that its discussions and recommendations will be of value for all specialties.

As well as clarifying issues surrounding consent and confidentiality in genetic practice, the report identifies key practice points and lists documents which may be helpful when seeking consent. On some issues the report makes recommendations for practice.

The report covers:

- general aspects of consent as applied to genetics
- the sharing of information with other family members and between professionals
- genetic investigations performed on stored material
- the Human Tissue Act 2004, consent and DNA analysis
- the Data Protection Act 1998 and the processing of medical genetic information.

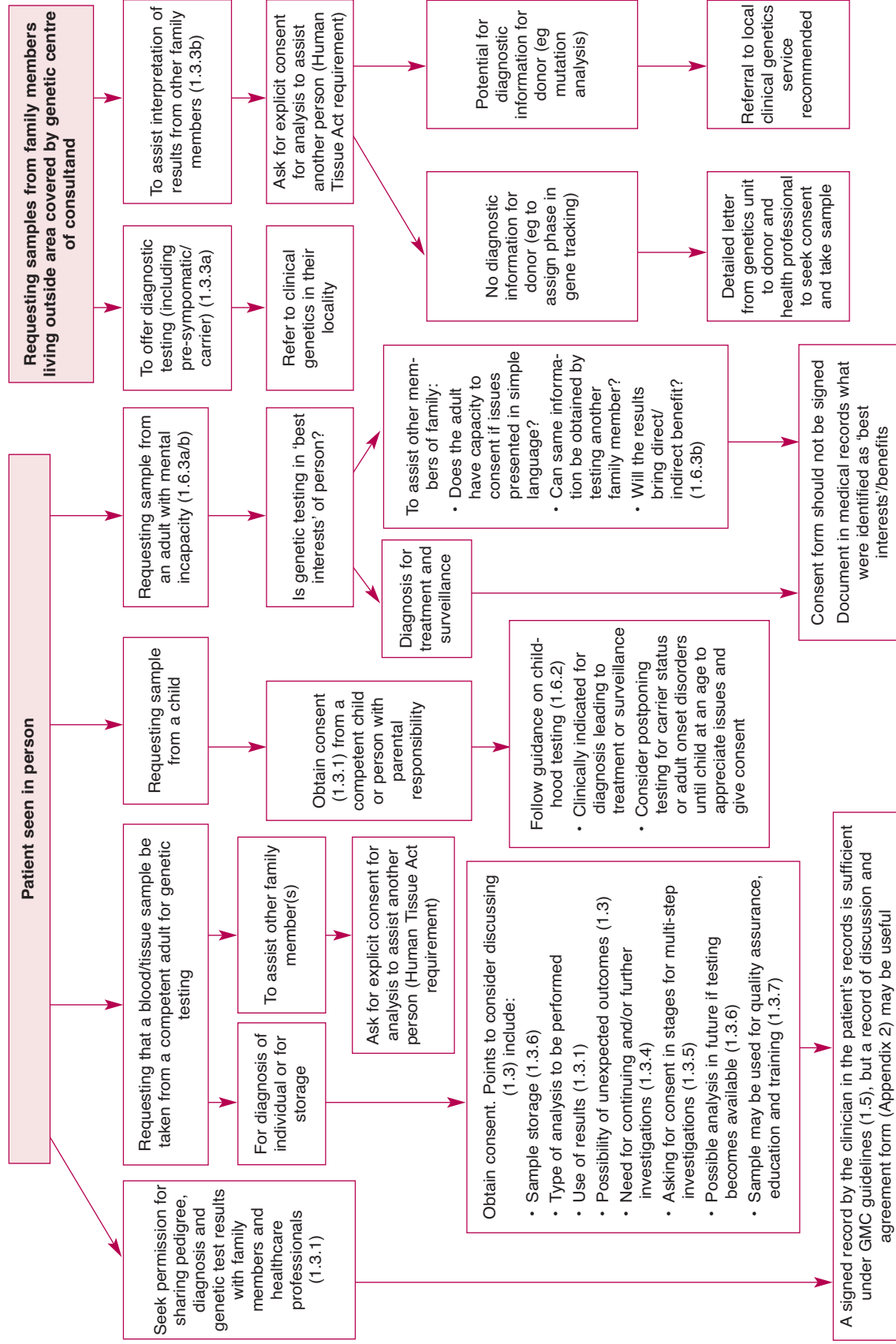
Three flowcharts (Figs 1–3, overleaf) summarise the main guidance, with cross-references to the sections in the main report where the points are covered in more detail.

The term ‘consultand’ is used to describe the individual who is requesting genetic information (not always the person affected); the ‘proband’ is the affected individual through whom a family with a genetic disorder is ascertained.

## How the report was produced

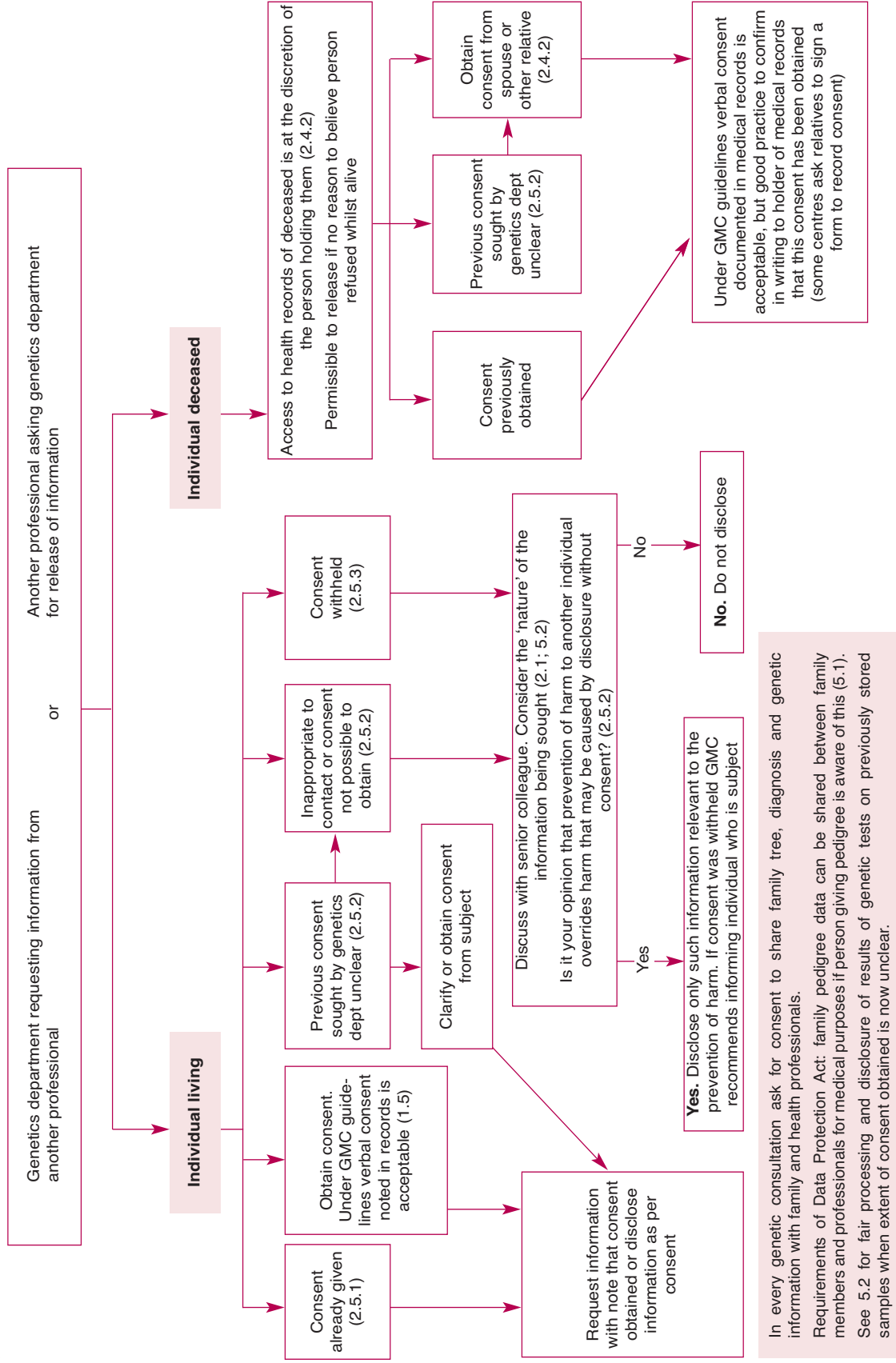
To assess current practice and areas of concern, the Joint Committee set up a working party which undertook a questionnaire survey of genetics units in the UK, and took medico-legal advice on current legislation. Issues were debated at length both within the working party and within the Joint Committee. The draft report was made available for wide consultation in the autumn of 2003 and 22 written replies were received. The preparation of the final report was delayed until 2005 to enable issues arising from the Human Tissue Act 2004 to be included. Its publication date was rescheduled for 2006 to be able to incorporate advice relating to the Human Tissue (Scotland) Bill.

It was originally intended that this document should contain a detailed description of the current medico-legal framework. This has not been included because a report from the Human Genetics Commission, *Inside information: Balancing interests in the use of personal genetic data*,<sup>1</sup> reviews the legislation in detail.



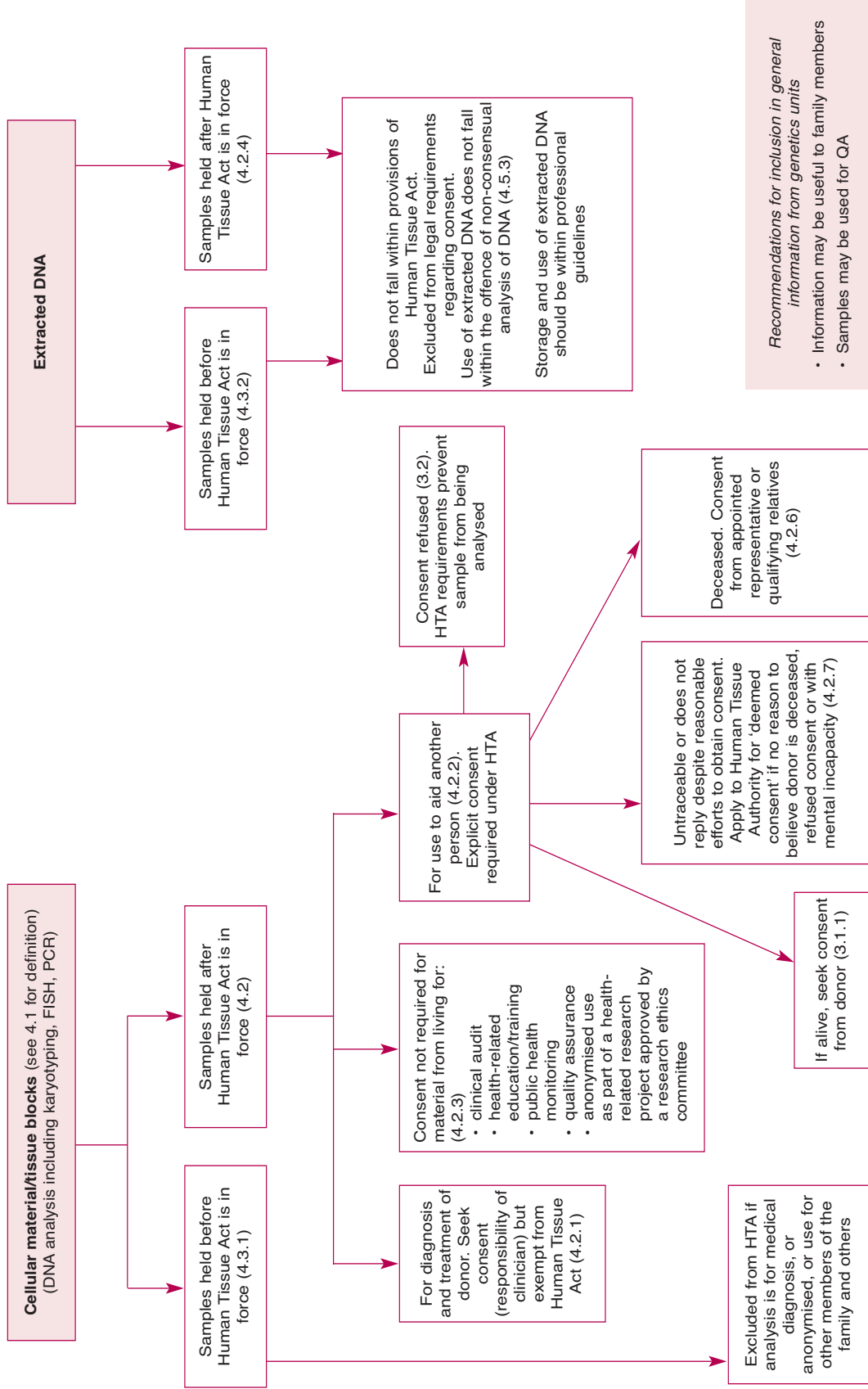
**Fig 1 Requesting information and samples during a genetic consultation: recommendations for clinical practice.** This flowchart is only a summary and should be used in conjunction with the main report; the numbers refer to the appropriate sections in the report. This flowchart may be photocopied for clinical use.

Joint Committee on Medical Genetics: Consent and confidentiality in genetic practice: guidance on genetic testing and sharing genetic information



In every genetic consultation ask for consent to share family tree, diagnosis and genetic information with family and health professionals.  
 Requirements of Data Protection Act: family pedigree data can be shared between family members and professionals for medical purposes if person giving pedigree is aware of this (5.1).  
 See 5.2 for fair processing and disclosure of results of genetic tests on previously stored samples when extent of consent obtained is now unclear.

**Fig 2 Sharing and disclosing genetic information: recommendations for clinical practice.** This flowchart is only a summary and should be used in conjunction with the main report; the numbers refer to the appropriate sections in the report. GMC = General Medical Council. This flowchart may be photocopied for clinical use.



**Fig 3 Consent for genetic analysis of stored samples or archival pathological material and the Human Tissue Act (HTA) 2004.** This flowchart is only a summary and should be used in conjunction with the main report; the numbers refer to the appropriate sections in the report. The requirements of the Human Tissue Act 2004 will not become clear until Human Tissue Authority issues guidance; our interpretation of likely effects on practice are summarised here. FISH = fluorescence in-situ hybridisation; PCR = polymerase chain reaction; QA = quality assurance. This flowchart may be photocopied for clinical use.

# ONE

## Consent in medical genetic practice: general aspects

### 1.1 Consent: general principles

The seeking of consent is a process to ensure that a person understands the nature and purpose of giving a sample or undergoing an intervention. Generally, in medico-legal terms, 'valid consent' is ensured if anything that represents a significant risk that would affect the judgement of a reasonable person has been explained. This recognises that the consent process cannot always be totally comprehensive: all eventualities cannot be covered because of unexpected clinical findings or test results.

General guidelines for obtaining consent for examination and treatment have been issued by the Department of Health (DH)<sup>2</sup> and underpin the practice of genetics just as in any other specialty. Twelve key points from the DH guidelines are provided in Appendix 1. The General Medical Council (GMC) has also published guidelines.<sup>3</sup>

The guidance from the DH and GMC also address consent in children and in adults who lack capacity. New legislation concerning the care and treatment of adults lacking capacity (the Mental Capacity Act 2005) sets out the statutory basis for obtaining consent from adults lacking capacity (see section 1.6.3).

The Joint Committee on Medical Genetics wishes to re-affirm two over-arching principles concerning consent:

- ▶ **Except in exceptional circumstances, consent should be obtained prior to a clinical or laboratory test with genetic implications being undertaken and consent should have been obtained before medical genetic information is disclosed.**

### 1.2 Gaining consent in clinical genetic practice

#### 1.2.1 Responsibility of the clinician to obtain consent

Guidelines from the DH<sup>2</sup> place the onus for gaining consent with the clinician requesting the genetic information or the sample. The request for consent may be delegated, but the clinician must ensure (under GMC guidelines)<sup>3</sup> that the person to whom it is delegated is suitably trained and qualified, has or is given sufficient knowledge of the reason for a request for information or for the proposed investigation, and understands any risks involved.

In current practice it is therefore assumed that when a laboratory receives a genetic sample, the clinician responsible for the care of the patient has obtained appropriate and valid consent so that the laboratory is not required to confirm and document such consent. This is particularly important for biochemical and cytogenetics laboratories where samples often need to be processed immediately.

For samples received from clinical genetics units, it is likely that issues of consent will have been

fully discussed. However, genetic laboratories receive a large number of samples from all branches of medicine and surgery. The Joint Committee wishes to encourage examples of good practice regarding issues related to the testing and storage of genetic samples, as discussed in this document. The Joint Committee recognises that genetics health professionals, particularly within clinical genetics, have a key role to play in disseminating good practice. We therefore ask genetics professionals to undertake this role.

- ▶ **The Joint Committee requests that genetics health professionals, particularly within clinical genetics, take a leading role in disseminating good practice regarding the issues particular to the testing and storage of genetic samples.**

### 1.2.2 The role of the genetic laboratory test request form

There was wide support during our consultation exercise for assisting the dissemination of good practice by including in request forms for genetic laboratory tests a statement confirming that issues relating to testing and storing samples have been discussed. As consent will need to be given by a donor specifically for DNA analysis of cellular material to be used for the benefit of others when the Human Tissue Act 2004 is in force, we recommend that a statement confirming that this has been obtained is also added to the laboratory form.

- ▶ **The Joint Committee recommends that a statement be placed on genetic laboratory test request forms such as:**  
**'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).'**

## 1.3 Issues particularly associated with the clinical practice of genetics

The seeking of consent should be regarded as a means to ensure that a person understands the nature and purpose of giving a sample or undertaking an investigation. Appropriate information and the opportunity to discuss issues should be offered as part of this process.

There are some issues which appear to be particularly relevant to the clinical practice of genetics, and which we believe need special consideration and attention. They are discussed in more detail in the sections that follow and include:

- taking a pedigree to record a family history of disease
- using pedigree information and genetic test results for the benefit of the whole family
- ensuring that colleagues involved in the care of the patient are kept informed
- information relevant to other family members being transmitted to appropriate health professionals
- contacting other family members to offer testing to define their risk of being affected by or being a carrier of the genetic disorder
- genetic laboratory (molecular, biochemical and cytogenetic) investigations often being technologically demanding and hence by their nature prolonged
- unexpected results being revealed by a genetic test, for instance information about

parentage

- some genetic tests being able to predict the future onset of disease in a currently healthy person
- storing samples for future analysis should a test become available.

### 1.3.1 Using pedigree information and genetic test results for the benefit of the whole family

Most individuals express a desire for pedigree information and genetic test results to be made available to other family members to assist in diagnosis and medical care, even if that individual would prefer not to contact family members personally but asks health professionals to take on this role. The Human Tissue Act requires that consent has been obtained specifically for the use of cellular material to assist in the care of relatives, as outlined in section 4.

- ▶ **We strongly support the good practice of confirming and documenting that it is acceptable to an individual that his or her information may be shared and samples used for the benefit of other family members.**

### 1.3.2 Ensuring that colleagues involved in the care of the patient are kept informed, and that information relevant to the care of other family members is transmitted to appropriate health professionals

It is important that the primary healthcare team and other health professionals involved are kept informed about the implications of a genetic diagnosis; our questionnaire survey of UK genetics units showed that it is common practice for genetics clinicians to seek permission for storing (and sharing) information and for sending letters to the clinicians involved in the patient's care. We strongly support this practice.

### 1.3.3 Consent for taking and testing samples from relatives living outside the area covered by the genetic centre of the consultand

#### *(a) To offer diagnostic testing to family members*

The result of a genetic test in one member of a family may allow specific testing to be offered to other family members. As the information may be used to enable diagnosis, to institute surveillance for disease complications, or to determine carrier status, we recommend that family members are referred to the clinical genetics unit in their locality which will take up their care.

#### *(b) To assist in the interpretation of a genetic test result by using information from other family members*

A sample from a family member living in another part of the country may be needed to assist in the interpretation of a test result for the consultand, who usually makes available the details of family members willing to assist.

As stated above, it is the responsibility of the health professional asking for the investigation to be undertaken to obtain consent for taking, analysing and storing the sample, after provision and discussion of appropriate information. It is unlikely that the consultand's geneticist will be

able to have direct contact with the extended family, and therefore will ask another health professional, usually the patient's general practitioner (GP), to seek consent and take the sample. The geneticist must ensure (under GMC guidelines)<sup>3</sup> that the colleague is suitably trained and qualified, has sufficient knowledge of the proposed investigation, and understands any risks involved.

*Results unlikely to provide diagnostic information for the donor of the sample*

Where the sample will be used to interpret results for the consultand but will not provide diagnostic information for the person giving the sample (for instance, in gene tracking studies), we recommend that a detailed information letter accompanies the request so that the health professional can seek valid consent. We recommend that letters be sent to the individual to give to the clinician who has been asked to take the sample, if this is known, and/or to the GP.

*Results likely to provide diagnostic information for the donor of the sample*

Where the sample may provide diagnostic information for the donor (for instance, in mutation studies), we recommend that the person should be referred to a colleague in the local clinical genetics service.

#### **1.3.4 Consent for continuing and further investigations**

The original consent for genetic testing can be assumed to remain valid if a further investigation remains within the scope of the original consent. An example would be using a new molecular technique to seek mutations in a gene where previous investigations had not detected a mutation. However, see the following paragraph.

#### **1.3.5 Consent should be requested in stages for multi-step investigations**

Molecular, biochemical and cytogenetic laboratory investigations are often technologically demanding and hence by their nature prolonged. It is good practice to point out during the process of asking for consent that results may not be available for a long period of time.

It is important that families are kept up to date with the progress of laboratory testing. A genetics laboratory may recommend that further tests are performed on a sample to generate additional useful clinical information when the results of the requested investigation are known. An example would be to determine whether important genes had been disrupted by a translocation. Good practice would be for the clinician to return to the individual from whom consent was obtained to explain the findings and to ensure consent for any continuing procedure if it is likely to reveal outcomes not covered in the original consent. This guidance is in keeping with the DH view that consent is a process and not simply a one-off event. Good liaison between clinical and genetics laboratory staff is essential in such cases. Another example would be to consider carefully any analysis undertaken on a child's sample that may lead to serendipitous discovery of an adult onset disorder.

#### **1.3.6 Seeking consent for future testing of a sample**

Samples may be taken for storage (usually as extracted DNA rather than tissue) for future analysis

should a test become available. It is helpful to make clear whether or not the patient will be re-contacted before testing is undertaken, either to obtain additional consent or to discuss the diagnoses which may then be possible.

Although it is important to obtain consent for prospective testing and re-contact, a genetics unit may not be able to give an assurance that these procedures will be performed automatically when appropriate genetic analysis becomes available. The local policy should be explained at the time of seeking consent and an explanation given of the procedure for the patient or relative to enquire whether new advances have become available.

In obtaining consent for future testing, we recommend that spoken and written information is given, and agreement confirmed by the completion of a consent form (see example in Appendix 2). The current common practice of many genetics units of asking for a relative to be named as a future contact if the donor dies is mirrored in the Human Tissue Act 2004 which allows for the appointment of a nominated representative(s) to make decisions on a donor's behalf regarding the use of tissue samples (including the release of tissue blocks). In the absence of any prior decision made by the patient, or a nominated representative having been appointed, the Act establishes a statutory list of 'qualifying relatives', any of whom may consent to the use of the patient's cellular sample (after death) for the benefit of others.

- ▶ **The Joint Committee recommends that when a sample is taken to be stored for the possibility of analysis in the future, consent is obtained and recorded for storage, future testing, and re-contacting the patient (or a relative previously specified by the patient if the patient were to be deceased). The local policy over re-contact should be explained.**

### 1.3.7 Consent for the use of samples as controls, quality assurance, audit, education and training

A sample may be stored and/or used for quality assurance (QA) of laboratory tests for other patients. The DH's model consent policy suggests that tissue samples may be used for QA purposes without specific patient consent, provided NHS bodies have an active policy of informing patients of such use.<sup>2</sup> A sample may also be used for clinical audit, education and training. Whilst it is good practice to inform all patients that their samples may be used in these ways, consent is not required under the provisions of the Human Tissue Act 2004 for samples from living patients.

- ▶ **We recommend that a statement about the use of samples as controls for clinical testing and in quality assurance is incorporated in the general information which genetics departments make available to patients, particularly when samples are taken. An example might be: 'After testing, part of your sample might be used anonymously to monitor the quality of laboratory results.'**
- ▶ **We recommend that the issues in Box 1 should be discussed explicitly with patients when medical and pedigree information is being obtained and when genetic investigations are requested.**

**Box 1 Genetics issues to be discussed during the consent process: recommendations.**

It would be helpful for patients if the following issues were discussed during the consent process, as appropriate:

- the use and sharing of information (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
- that samples may be used for quality assurance, education and training
- that information may be shared with health professionals including the primary care team.

*The Joint Committee recommends that these points are highlighted in a general leaflet for genetics patients or that they are included as appropriate in post-clinic summary letters.*

#### 1.4 Counselling as a part of the consent procedure for genetic testing

All health professionals agree that appropriate and intelligible information should be given to patients to enable them to assess the options and make an informed choice about genetic testing. Some health professionals believe that genetic tests should not be undertaken without counselling being an essential part of the consent process. There is consensus that counselling should be an integral component of predictive testing programmes for such single gene disorders as Huntington's disease,<sup>4</sup> or those single genes predisposing to cancer. In these cases, 'counselling' usually takes the form of several discussions over a period of time exploring the implications of taking the test.

However, in other clinical circumstances, it may not be appropriate to insist that a patient has to undertake a counselling programme before a genetic test is performed. The key is to ensure that patients have the relevant information to make an informed choice.

We agree with the Genetic Interest Group that the consent process should not be used to the detriment of patients, for example unduly increasing anxiety by presenting in great detail all possible adverse outcomes, although it is appropriate to discuss the possibility of unexpected results.

This is in keeping with the GMC's recommendation that when providing information, health professionals must do their best to find out about patients' individual needs and priorities.<sup>3</sup> Assumptions should not be made about patients' views. As patients' beliefs, culture, occupation or other factors may have a bearing on the information they need in order to reach a decision, these should be discussed with them.

Whilst it is good practice to encourage the individual to consider the issues involved, an insistence on imparting unwanted information could be interpreted as a breach of a person's right to private and family life.<sup>5</sup> For instance, some family members may be willing to give a blood sample for

the benefit of their relatives, but do not wish to receive counselling or detailed information prior to this. They may not wish to receive the results of tests on the sample either. The Joint Committee upholds such responses as being firmly within acceptable clinical practice, provided that those family members who have chosen this course understand that it has been their free decision not to receive information. A note of their wishes should be made in the medical records.

## 1.5 Recording consent

General Medical Council guidelines confirm that patients may indicate their informed consent either orally or in writing.<sup>3</sup>

A record in a patient's notes signed by the clinician that issues were discussed and that the patient agreed to particular actions is adequate for most purposes. In some cases, however, the nature and implications of the genetic information or test results to be generated make it important that a written record is available of the patient's consent and other wishes. This is considered particularly important for predictive testing.

Some respondents to our questionnaire believed that the recording of consent in genetics should be formalised through a specific consent form. Members of the Joint Committee felt that the wording of a consent form should be simple, clear and as informal as possible. The advice from the Genetic Interest Group was that families appreciated broad headings rather than detailed specifics. After considerable discussion, and examining consent forms from many genetics units, the Joint Committee offers the one shown in Appendix 2 as a suitable example or as a record of the discussion.

- ▶ **In line with the Department of Health's guidelines,<sup>2</sup> the Joint Committee recommends that during the seeking of consent from a patient or family the health professional should explain the genetic issues, offer written information sheets where appropriate and make a note of discussions in the medical records. Where appropriate, formal recognition that the process has been undertaken may be recorded by asking the patient to sign a form.**

## 1.6 Who can give consent for genetic testing?

### 1.6.1 Living competent adults

If an adult is mentally competent, consent should be sought from the person concerned.

### 1.6.2 Children

Parents, or those with parental responsibility, may give consent for genetic testing in children, but there are special issues to be considered. These are discussed in the Clinical Genetics Society report, *The genetic testing of children*, which highlights practical and ethical issues in more detail.<sup>6</sup>

Genetic testing in a child is clinically indicated in order to make a diagnosis where treatment or surveillance is available, but the Clinical Genetics Society recommends that testing for adult onset disorders or carrier testing should be postponed until the child is at an age to appreciate the issues and give consent (ie demonstrate Gillick competence). If a screening programme (for example, for haemoglobinopathies or metabolic disorders) may reveal that a person is a carrier

for a genetic disorder, the Joint Committee recommends that this should be made explicit in the information given to people undertaking the programme, and in the consent discussions.

### 1.6.3 Adults with mental incapacity

In the case of an adult lacking capacity (and who therefore is unable to give consent), a genetic test can be undertaken if it is in the best interests of the adult concerned, for instance to make a diagnosis. This situation is not changed by the draft Mental Capacity Bill<sup>7</sup> (which was in draft form when this document was prepared) but the Bill provides clarification in that the criteria for making a best interests judgement are set out. In carrying out genetic testing for the purpose of treatment or care, there is also a requirement to consult (if practicable and appropriate) certain named individuals.

#### (a) *Diagnostic testing*

The Human Genetics Commission has argued that if it is ethical for a doctor to treat a person who lacks capacity to consent – provided that the treatment is in the best interests of the person – then it is ethical to carry out genetic testing if it is medically indicated for management.<sup>1</sup> In Scotland, the Adults with Incapacity Act 2000 has clarified the law, and provides a clear legal framework for a range of decisions relating to the medical treatment and welfare of such persons.<sup>8</sup> In England, the common law currently provides the basis for providing medical treatment or care to adults who are incapable of consenting to it, and the Human Genetics Commission believes that this would apply to genetic testing provided it is necessary for treatment or care of the individual.

#### (b) *Family studies*

Establishing a clinical diagnosis or delineating a pathogenic mutation in one family member may allow diagnosis, surveillance and risk estimation for the wider family. Clinical experience confirms that the majority of people are willing to undergo investigations to help other family members, even if there is no direct medical benefit to themselves. The Genetic Interest Group has argued strongly that it should not be presumed that an adult with incapacity would be less altruistic than a competent adult in wishing to assist other family members in genetic investigations.<sup>9</sup>

Department of Health guidelines<sup>10</sup> also note that care should be taken not to underestimate the capacity to understand of a patient with a learning difficulty or cognitive decline. Many people with learning difficulties have the capacity to consent if time is spent explaining to the individual the issues in simple language. Similarly the Mental Capacity Bill establishes a functional definition of capacity and a requirement for all practicable steps to be taken to enable a patient to make his or her own decisions. A general explanation of a test or procedure will suffice.

In the case of children, the courts have ruled that those with parental responsibility can consent to an intervention which, although not in the best interests of the child, is not against the interests of such a child. We consider that it would be helpful to some families with genetic disorders if this principle were to be extended to cover adults with mental incapacity.

Currently in England and Wales, no one can give consent on behalf of an adult lacking capacity. When consent cannot be given because an adult lacks capacity, however, an intervention can be

undertaken if it is deemed to be in the best interests of that person. The draft Mental Capacity Bill formalises this by establishing that if a person lacks capacity and a person has been appointed with a lasting power of attorney or as a deputy by a court then the appointed person can give consent on their behalf (subject to the requirement to act in the patient's best interests). Those with more informal relationships (such as family members interested in the patient's welfare or carers) can also consent provided that they act in the patient's best interests.<sup>7</sup>

The House of Lords has suggested that action taken 'to preserve the life, health or well-being' of a patient will be in their best interests, and subsequent court judgements have emphasised that a patient's best interests include much wider welfare considerations. These can include indirect benefit: the wellbeing of relatives could be a valid justification, for example, if this had a positive effect on the care of the adult. The possible harm – from the testing procedure and potential harm deriving from use of the test results – to the tested person should be considered to be negligible. The Human Genetics Commission formed the view that if the intervention is in the best interests of the person concerned taking into account all the circumstances, it would be lawful.<sup>1</sup> The Mental Capacity Bill is likely to include a list of factors, all of which must be taken into account in determining 'best interests', which include the requirement to consult and to take into account the person's past and present wishes and feelings.

Before testing is undertaken on a person with mental incapacity, however, it is worthwhile considering whether the same genetic information could be obtained by undertaking tests on other family members who are capable of giving explicit consent.

- ▶ **In 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. In these circumstances, the standard consent form should not be signed, but it is good practice to make a note in the medical records as to why the action was believed to be in the patient's best interests.**

In Scotland the law now allows consent to be given on behalf of an adult with incapacity,<sup>8</sup> but in Scotland carers of such an adult can refuse on his or her behalf even if family members regard an intervention as being likely to provide the individual with indirect benefit.

## 1.7 Consent to clinical photography and video recording

As well as being part of the medical record, clinical photographs and video recordings are important for teaching, audit and research. A particularly valuable role in genetics is in delineating and understanding the natural history of dysmorphic syndromes. The purpose and possible future use of the photographs and video must be clearly explained to the person (or parent), before their consent is sought. If the photographs or video are to be used for teaching, audit or research, patients must be aware that they can refuse without their care being compromised, or they can require that photographs or video be used only if the material can be anonymised. A specimen consent form is provided in Appendix 3.

## 1.8 Issues of consent and confidentiality in complex clinical cases

The responses received during the consultation identified a need for urgent clinical ethics advice on issues of consent and confidentiality in those complex genetics cases where local consensus

has not been achieved. However, it was acknowledged that a high level of commitment and acceptance would be required by a group willing to undertake this role and that it should be seen not as an erosion of clinical autonomy but as an aid to decision making.

The Joint Committee on Medical Genetics asks the British Society for Human Genetics, in liaison with the Genethics Club,<sup>11</sup> to decide how best to provide urgent advice. The Genethics Club is an existing UK forum which currently meets three times a year to discuss practical ethical problems encountered in clinical genetics departments.

## TWO

# Giving and sharing genetic information

### 2.1 The use of existing genetic information to facilitate accurate genetic testing

Clinical 'information' in genetics may include data from the pedigree, the name of a genetic disorder, the genetic status of a family member (eg carrier/affected) or the result of a clinical or laboratory test.

For some genetic disorders, best clinical practice requires a detailed family history – to assist in making a precise diagnosis or in confirming or determining the most likely mode of inheritance. The information available to the family member giving the pedigree is most likely to have been obtained from other family members who may also have suggested a referral to a genetic clinic. The information contained in a pedigree, including whether family members are affected or unaffected, may be known to many people, including friends and acquaintances. The use of the family history is further discussed in sections 2.3 and 5.1.

When a person asks for genetic testing it is also usually because of knowledge that a family member (usually the proband) has had a molecular or cytogenetic test performed. The request for testing may be to determine carrier status (eg for cystic fibrosis or Duchenne muscular dystrophy), to assist with reproductive decisions or to determine whether someone with a genetic predisposition (eg to breast or bowel cancer) needs to have surveillance and treatment. The consultant has often been informed which genetics laboratory performed the test, and may even have the precise result. This allows the laboratory to perform a specific test for the known genetic anomaly.

Indeed, the best medical practice is to test the consultant for the specific mutation or genetic anomaly which has been found in the affected family member. It is not sensible to undertake a general screen of the consultant's sample in such circumstances to see if an abnormality can be found. If no anomaly were to be identified, this could be because the person had not inherited the disorder, or it could be because the technique employed in the laboratory testing the consultant's sample was not able to detect the particular type of mutation.

It is therefore entirely appropriate for best clinical care that technical information and laboratory reports should be shared between the heads of those laboratories undertaking the testing, be transferred from one family member's record to another in the same laboratory to permit testing, or be available to clinical staff in genetics units.

### 2.2 Legal background

The legal basis for the use and disclosure of medical information is the common law of confidence. The interpretation of this law by bodies such as the GMC is important for healthcare professionals. However, the processing of the information is regulated by statute, namely the provisions of the

Data Protection Act 1998. Clarification has been sought from the Information Commissioner about the fair processing of information for genetic diagnostic purposes when this requires the use of information about other family members. This is discussed in section 5.

### **2.3 The family history as an aid to genetic diagnosis**

Documenting an accurate family history (or pedigree) of disease may be necessary to make an initial specific genetic diagnosis or to form a view about mode of inheritance and recurrence risks. This family history, usually obtained from one member and given in good faith, contains information about other family members, who may not be aware that it is being given and recorded.

However, a family member is unlikely to know confidential medical information other than that which has been given by the person concerned. Most of the information given to construct a pedigree during a genetic consultation is likely to be known to a wide circle of people.

Nevertheless, family information held by regional genetics centres is held in confidence and is used to aid diagnosis and estimation of risk for the person giving the pedigree.

As the information may be used to advise other family members, it is good practice to gain consent for information sharing as discussed in the section on seeking consent (section 1.3.1). We recommend that either a note is written in the medical record, or a consent form used.

### **2.4 Use of medical records to confirm diagnosis**

Information about the diagnosis and clinical status of other family members can be vital in determining risks, particularly when there appears to be an inherited predisposition to cancer. Access to information about other family members is governed by the Data Protection Act 1998<sup>12</sup> and the Access to Health Records Act 1990.<sup>13</sup>

#### **2.4.1 Family members who are living**

Current practice is to obtain written consent from living family members to access their medical information, usually via the individual within the family who is requesting genetic information. Family members are usually asked to sign a form confirming their permission.

#### **2.4.2 Deceased family members**

Once a deceased person's estate has been dispersed, access to health records in hospitals, general practices and other healthcare settings under the Access to Health Records Act 1990 is at the discretion of the person holding them, unless the deceased refused permission. Some hospitals exercise this discretion by choosing to allow access to such records only with consent from the spouse, but the information contained in the records may be of great importance for the optimal medical management of a blood relative, a possibility not considered in the Act. For example, in making the diagnosis of a hereditary predisposition to cancer or in deciding on which gene to perform mutation analysis in a blood relative, it may be necessary to know details of the histology of the cancer.

The importance of sharing genetic information for the medical care of relatives is increasingly being recognised. The Human Genetics Commission has recommended an extension of the limited circumstances for the revealing of confidential information obtained in a medical context to include disclosure in the interests of relatives. The Human Tissue Act has introduced the concept of ‘any qualifying relative’ in giving consent for the release of bodily material posthumously for genetic analysis (as discussed in section 4). We recommend that a similar concept of consent from qualifying relatives be accepted by healthcare facilities in exercising their discretion to release medical records of deceased patients. Under GMC guidelines, verbal consent given to the genetics department and documented in the genetics medical records is acceptable, but we recommend that the genetics department confirms that it has consent in a written communication to the medical records department from whom the records are being requested.

- ▶ **We strongly recommend that medical records departments of healthcare facilities 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 in the family. Under GMC guidelines, verbal consent documented in the genetics department notes is acceptable.**

## 2.5 Disclosure of information

### 2.5.1 Where consent for releasing information has been obtained

Medical information can be disclosed when consent has been obtained. As most family members wish their information to be available to help relatives, we recommend the good practice of obtaining and documenting consent for disclosure in routine clinical practice.

### 2.5.2 Where the basis of consent for releasing information is unclear

There are clinical circumstances where the scope of consent sought previously for the sharing of information or for the testing, storage and use of samples may no longer be clear. This is usually because the information which patients and clinicians discuss during the consent process is continually changing with technological advances, and some patients may have been seen many years earlier.

Attempting to verify past consent or newly seeking consent from the information or sample donor is considered the optimal situation, but this may not be possible because contact has been lost or may not be clinically appropriate because the family member seeking information may be concerned about compromising his or her confidentiality. For example, an individual wanting to undertake prenatal diagnosis may not wish anyone to know of the pregnancy until the test results are available. The necessity to seek consent from another family member for release of the information will lead to a breach of confidence for the pregnant woman. The public interest in keeping the consultant's pregnancy confidential may be more important than the public interest in requiring consent to the disclosure of a test result from a family member; this concept is recognised legally.

The balance, then, must be carefully considered by the health professional and the clinical judgement documented: 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. In these circumstances, a sample of extracted DNA and/or the result may be used. (See section 4 for constraints over the use of tissue containing cells when the Human Tissue Act is in force.) This advice is based on current GMC guidelines.<sup>14</sup> We recommend that as far as possible the information from the sample remains confidential and is used to inform care for the consultand, without releasing specific information about the sample donor. For instance, it is not necessary to release the technical description of a family mutation to a person who has been tested and shown not to have inherited the mutation. If a family member is shown to have a mutation, this is now their personal medical information and may be divulged to them.

### 2.5.3 Where consent to release information has been refused

The Human Genetics Commission,<sup>1</sup> the Nuffield Council on Bioethics<sup>15</sup> and the GMC<sup>14</sup> have all expressed the view that the rule of confidentiality is not absolute. In special circumstances it may be justified to break confidence where the aversion of harm by the disclosure substantially outweighs the patient's claim to confidentiality.<sup>1,14</sup> Examples may include a person declining to inform relatives of a genetic risk of which they may be unaware, or to allow the release of information to allow specific genetic testing to be undertaken.

Before disclosure is made in such circumstances, an attempt should have been made to persuade the patient in question to consent to disclosure; the benefit to those at risk should be so considerable as to outweigh any distress which disclosure would cause the patient; and the information should be anonymised and restricted as far as possible to that which is strictly necessary for the communication of risk.<sup>1</sup>

We recommend that before disclosure is made when consent has been withheld, the situation should be discussed with experienced professional colleagues and the reasons for disclosure documented. Current GMC guidance states that the individual should generally be informed before disclosing the information.<sup>14</sup>

## 2.6 Related issues

### 2.6.1 Charges

Under the Data Protection Act and the Access to Health Records Act, holders of records (hospitals and general practices) are permitted to levy a charge for access to information contained in the medical record. As the NHS is a mutual service and the information is for clinical care, it is not in the interests of patients, families or service provision that charges are made when genetics units seek information on the proband or other family members. We strongly recommend that any charges be waived in these circumstances.

### 2.6.2 Destruction of medical records

Valuable information about specific diagnoses (for instance, to confirm the types of cancer occurring in families with inherited forms of cancer) may be lost when paper records are destroyed. We recommend that the NHS IT strategy takes into account the necessity for storing such diagnoses. In the interim, it would be ideal for genetic purposes if they could be stored in an alternative medium when the paper records are destroyed.

### **2.6.3 Retention and storage of pathology and genetic samples in laboratories**

In March 2005 the Royal College of Pathologists revised its document, *The retention and storage of pathological records and archives: guidance from the Royal College of Pathologists and the Institute of Biomedical Science*, which is available through [www.rcpath.org](http://www.rcpath.org). The document considers the issues associated with the storage of all pathology tissues, including DNA, in the light of the Human Tissue Act 2004. The Royal College will update this document once the relevant guidelines from the Human Tissue Authority have been published.

### **2.6.4 Cancer registries**

Cancer registries are extremely valuable sources of information in determining whether a family is likely to have an inherited predisposition to cancer. If the person is alive, consent is sought and documented, usually by the clinical genetics service through the consultant, before the release of information is requested. UK cancer registries will release information on deceased relatives to clinical genetics units under the mutual understanding of respect for confidentiality. We strongly support this vital practice for clinical care.

### **2.6.5 The use of results when samples are tested in research laboratories**

Many aspects of clinical and laboratory genetics remain at the interface between service and research. For some diseases there is no clinical testing service. Using results generated in a research laboratory is acceptable as long as the patient appreciates that the results have been generated in a non-clinical service setting. It is considered best practice to confirm the results in an NHS laboratory if possible.

## THREE

# Genetic investigations on stored samples or archival pathological material

In order to make or confirm a genetic diagnosis it may be vital to perform tests on stored samples, most commonly samples of extracted DNA and histological material. Testing archived pathological material from an affected family member is particularly useful where an inherited predisposition to cancer is suspected. The removal, storage and use of human tissue containing cells will fall within the provisions of the Human Tissue Act, but extracted human DNA will not (see section 4 for a full discussion).

### 3.1 Samples where the basis of consent is unclear

Our recommendations on consent and storage of new samples for possible future use are outlined in section 1.3.6, but there are collections of samples, including pathological archive samples, for which prospective consent may not have been sought when the sample was taken because specific tests were not available at that time.

#### 3.1.1 Stored samples from a living patient

If the person is still alive, attempts should be made to contact them to seek their permission for testing the sample (as discussed in section 2.5.2). Often contact can be re-established through the family member who is seeking advice from the genetic clinic, and who may stand to benefit from the results of the testing.

If the sample is of extracted DNA, and the person cannot be contacted, or does not respond, an assessment of the balance of harm as discussed in section 2.5.2 should be undertaken to decide whether the testing can proceed.

If the donor of a stored cellular/tissue sample fails to respond to repeated requests to consent to testing or cannot be traced, samples of cellular material/tissue blocks may still be tested with the 'deemed consent' of the Human Tissue Authority (see Section 4), or in Scotland, the Court of Session. If a living patient has refused permission for testing, however, the sample of cellular material may not be lawfully tested.

#### 3.1.2 Samples collected as part of the Newborn Screening Programme

When it is necessary to perform a diagnostic test on a sample which was collected as part of the Newborn Screening Programme (for instance, a dried blood spot sample), specific consent outwith the screening programme must be obtained.

### 3.1.3 Samples taken at post-mortem examination

The use of post-mortem material is covered by the Human Tissue Act (HTA) 2004<sup>16</sup> which, once it takes effect (probably in April 2006), will repeal the Human Tissue Act 1961.<sup>17</sup> Under the HTA 1961, the person lawfully in possession of the body could authorise use of ‘any part of the body’ for ‘therapeutic, educational and research purposes’ if s/he has no reason to believe that the deceased or surviving spouse or relatives would object.<sup>16</sup> There are separate but similar arrangements for Scotland. It has been common practice, therefore, for a pathologist to ask a genetics department to confirm that it had obtained consent from a relative to demonstrate that there was no reason to believe that there would be an objection before releasing material. Under GMC guidelines, this consent can be verbal consent documented in the genetics notes, or formal written consent.<sup>3</sup>

The analysis of posthumous tissue samples is to be regulated by the Human Tissue Act 2004 which provides that the wishes of the donor immediately before he died or, if none are known, the consent of a person appointed by the donor or of any ‘qualifying relative’, takes effect. The consent of any qualifying relative suffices for the purposes of DNA analysis; this provision is explained more fully in section 4. Codes of practice are to be issued by the Human Tissue Authority which will provide more guidance on the scope and specificity of consent and what should happen in difficult cases, such as where relatives disagree. In the meantime, existing guidance on post-mortem examinations will be relevant.

The value of post-mortem tissue in genetic diagnosis for families is highlighted in draft information leaflets about post-mortem examination of a baby or child, and of an adult (available on the Department of Health website).<sup>18</sup>

The draft consent form (September 2002) for adult post-mortem examination<sup>18</sup> does not ask specifically about genetic tests, but notes that tissue and body fluids may be removed and stored indefinitely as part of the medical record. The draft consent form for a hospital post-mortem examination on a baby or child specifically asks for consent to ‘genetic tests’ being performed (including karyotyping). The consent form notes that samples may be kept indefinitely as part of the medical record and ‘may be used in the future for the care of other members of your family’.

## 3.2 Samples where consent to store or use human cellular material containing DNA has been refused

If a competent living person refuses consent to the storage and/or use of their cellular tissue samples for the benefit of another (including a family member) then the requirements of the Human Tissue Act prevent the sample from being analysed. In this circumstance, genetics departments may be able to assist the family by offering to act as a source of information and explanation for the donor.

## FOUR

# The Human Tissue Act 2004, consent and DNA analysis

### 4.1 Framework for regulating the storage and use of tissue

This introductory section describes the overall legal framework for storage and use of tissue. Specific regulations relating to DNA analysis are covered in subsequent sections.

The Human Tissue Act 2004 provides a legal framework for regulating the storage and use of tissue from the living, and removal, storage and use of tissue from the deceased for 'scheduled purposes', underpinned by consent from the appropriate person. Tissue in this context is defined as material from a human body that consists of or includes human cells, with the exception of gametes, embryos outside the body, and hair and nail from a living person. Chromosome preparations in fixative, dried blood spots, and unfixed tissues stored at  $-20^{\circ}\text{C}$  fall within the provisions of the Act, but cell lines (including lymphoblastoid) and extracted nucleic acid are excluded, as is any other human material created outside the human body. Anonymised material from living people used in a health-related research project approved by a research ethics committee is also excluded.

The Act establishes that consent from an appropriate person is required for the storage and use of human organs and other tissue from living persons, and for removal, storage and use of such material from the deceased. The Act specifies in Schedule 1 that purposes normally requiring consent by law (where tissue is from the living or the deceased) include:

- anatomical examination and determining the cause of death
- obtaining scientific or medical information about a living or deceased person which may be relevant to any other person (including a future person)
- research in connection with disorders, or the functioning, of the human body
- transplantation.

The legal requirement introduced by the Human Tissue Act to obtain consent where cellular material is to be used to obtain genetic information for another person reinforces the need to ensure that this is explicitly discussed and documented in clinical practice.

It is lawful for cellular material from a living person to be stored and used without any consent for clinical audit, quality assurance and performance assessment (which could include evaluations of *in vitro* diagnostic devices), public health monitoring and health-related education and training.

The Act is unlikely to come into force before April 2006. It will extend to England, Wales and Northern Ireland, except for section 45 and Schedule 4 (non-consensual DNA-analysis), which will apply throughout the UK. The requirements of the Act will not become clear until relevant regulations and codes of practice have been published by the Secretary of State and the Human

Tissue Authority, and the impact of other legislation such as the Mental Capacity Act and the Coroners Rules is known. However, we present here our interpretations of the likely effects of the Act on practice in genetics.

Although the Act imposes a requirement to obtain a licence for certain activities relating to material which has come from the body of a deceased person, it should be noted that there is provision for regulations to be made to remove this requirement for scheduled purposes. Further guidance for genetic laboratories must be awaited.

## **4.2 Consent requirement for cellular material after the Human Tissue Act is in force**

Professional guidelines outlined in this document take into account accepted best practice and the legal requirements for consent under the Human Tissue Act and are therefore recommended as a general framework for practice.

### **4.2.1 Consent requirement for cellular material used in the diagnosis and treatment of its donor**

Use of cellular material in the diagnosis and treatment of its donor is not subject to the Human Tissue Act 2004.

### **4.2.2 Consent requirement for cellular material to assist in the care of other relatives**

Specific consent under the Act is required to use cellular material to assist in the care of other relatives (but not extracted DNA whose use will fall within professional guidelines).

### **4.2.3 Consent requirement for cellular material from the living for audit, research, and quality assurance**

Consent under the Act is not required for material from the living to be analysed for certain purposes including clinical audit, education or training relating to human health, public health monitoring and quality assurance (the purposes listed in Schedule 4, Para 8, of the Human Tissue Act).

### **4.2.4 Consent requirement for extracted DNA**

When nucleic acid has been extracted from the cellular material, then the legal requirements of the Act regarding consent, storage and use no longer apply (although professional guidelines for its use should be followed).

### **4.2.5 Consent requirement for cellular material for research**

Consent is required to obtain cellular material from a person for the purpose of DNA analysis for research, except where the tissue comes from a living person, the research has ethical approval, and the person carrying out the analysis is not able to identify the donor, and is not likely to do

so in the future. This does not preclude the use of linked data provided that the person holding the encryption key is not a member of the research team.

#### 4.2.6 Who can give qualifying consent for analysis of DNA in cellular tissue?

The Act lists those who can give ‘qualifying consent’ for analysis of DNA in cellular tissue (Part 1, Schedule 4). 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. As already noted, specific consent must be obtained for the use of cellular material for the benefit of another family member. The gaining of such ‘qualifying consent’ is sufficient to prevent an offence of ‘DNA theft’ under the Act. Those with parental responsibility can give consent for a child.

The analysis of posthumous cellular samples is regulated by the wishes of the donor (adult or child) immediately before s/he died or, if no such decision was made, by the consent of a representative appointed by the donor or of a ‘qualifying relative’. Qualifying relatives are:

- spouse or partner
- parent or child
- brother or sister
- grandparent or grandchild
- child of a brother or sister
- stepfather or stepmother
- half-brother or half-sister
- friend of long-standing.

The similar list in Scotland is more extensive.

For the purposes of genetic analysis on posthumous cellular material, the consent of any qualifying relative will suffice – the list is unranked. 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.

However, for the purposes of removal, use and storage of tissue, these relationships are ranked such that consent should be obtained from the person who is at the top of the list. Where two or more people have equal ranking it is sufficient to obtain the consent of any of them. (Consent to the removal of tissue from the living person is not regulated by the Human Tissue Act and remains a matter for the common law.)

#### 4.2.7 Consent where a family member cannot be traced, after the Act comes into force

Where it is in the interests of another person (even a future person) that the analysis of DNA (including by karyotyping, fluorescence in-situ hybridisation and polymerase chain reaction) in cellular material be undertaken to provide scientific or medical information about the donor, the Act provides that:

- Where a donor cannot be traced and there is no reason to believe that s/he has died, has refused consent or is incompetent, then the analysis can be carried out following

application to the Human Tissue Authority which may ‘deem consent’, or in Scotland, following application to the Court of Session.

- Where the donor can be traced, but consent/lack of consent is not forthcoming despite reasonable efforts to obtain it, and there is no reason to believe that s/he has died, has refused consent or is incompetent, then the analysis of the cellular material can be carried out where the Human Tissue Authority ‘deems consent’, provided that the donor has been given notice of the application.

### 4.3 Consent requirement for cellular material and DNA stored before the Human Tissue Act is in force

#### 4.3.1 Cellular material held on the day before the Act comes into force

Existing holdings of cellular material are excluded from the consent requirements of the Act where they are to be analysed for scheduled purposes, or medical diagnosis, or for any purpose if the material is anonymised. It should be noted that included in the scheduled purposes under this provision is the use for other members of the family.

#### 4.3.2 Existing holdings of DNA

Existing holdings of DNA (extracted from the human cells from which they derive prior to the Act coming into force) are excluded from the legal requirements of the Act regarding consent, as they do not constitute ‘bodily material’. Neither does extracted nucleic acid come within the provisions of the offence of the non-consensual use of DNA. The storage and use of this material, however, should remain within professional guidelines such as those outlined in this document.

### 4.4 The use of information relating to DNA analysis

The Human Tissue Act does not, as such, apply to the use of information relating to DNA analysis, and current law relating to data protection, confidentiality and human rights law will continue to operate where a geneticist refers to records made for the clinical treatment of the proband with the intention of assisting a relative.

### 4.5 The offence of non-consensual analysis of DNA

In addition to the legal requirements for consent and for the taking, storage and use of human tissue, the Act made special provisions regarding the non-consensual analysis of DNA.

The Human Genetics Commission’s concerns over the possibility of DNA theft and its use for malicious or prurient reasons prompted the inclusion in the Act of a criminal offence, prohibiting a person from having human material with a view to analysing its DNA without consent. It is not the intention of the Act to prevent the storage and use of tissue for DNA analysis for medical and research purposes.

For the purposes of the offence of non-consensual DNA analysis, the term ‘DNA analysis’ (*Hansard*, 11 October 2004, col GC31) includes any process intended to provide information about the DNA in the bodily material and includes DNA sequencing, generation of DNA markers,

chromosome visualisation. Methods of deducing information about the DNA from RNA, protein and metabolites are also included.

The offence will apply only to the holding of cellular material with the intention of DNA analysis without consent; it will not apply to the holding and use of extracted DNA. DNA analysis of bodily material undertaken as part of the medical diagnosis or treatment of the person whose body manufactured the DNA is excluded from the criminal offence.

**4.5.1 Analysis of DNA in cellular material (including karyotyping, FISH and PCR on the cellular material)**

No offence is committed if consent has been obtained or the analysis is for medical diagnosis or treatment of the person whose body manufactured the DNA, or for other purposes (including the functions of a coroner and the prevention or detection of crime).

**4.5.2 Analysis of DNA in cellular material in holdings before the Act comes into force**

No offence is committed if consent has been obtained or if the analysis is for other scheduled purposes including clinical audit, education or training, obtaining information which may be relevant to any other person (including a future person), quality assurance and research (where the donor of the sample has been anonymised and is unlikely to be identified).

**4.5.3 Analysis of extracted DNA**

No offence is committed by the analysis of extracted DNA.

## FIVE

# The Data Protection Act 1998 and the processing of medical genetic information

### 5.1 Disclosure of information from family pedigrees

Our interpretation of detailed advice from the Information Commissioner is that the information shown on a family pedigree can be passed between health professionals (under Schedule 3 of the Data Protection Act 1998) without the explicit consent of all those shown on the pedigree if the processing is necessary for medical purposes (including the purposes of preventative medicine, medical diagnosis, medical research, the provision of care and treatment, and the management of healthcare services). However, when obtaining the family history, the health professional should advise the person giving the pedigree that it may be used to determine the mode of inheritance of a disorder, and that it may be shared with other members of the family if they seek advice, and with other health professionals (clinical and laboratory) if necessary for the care of family members. It is good practice to review the information on the pedigree before it is shared, to try to ensure that only information relevant to the clinical purpose is released; for instance, in some clinical situations it may not be necessary to give names on parts of the pedigree.

### 5.2 Fair processing and disclosure of results of genetic tests

We asked the Information Commissioner for guidance on the fair processing of results, particularly from samples stored in genetics units during the development of the clinical DNA services over the last 20 years. It may prove impossible now to determine the extent of verbal consent sought when the sample was taken. It may also no longer be possible to contact the person who gave the sample, and if it is, by doing so the consultand may have to give up his or her own right to a private life under the Data Protection Act (for instance, if a pregnancy was involved, or a wish to have a prophylactic mastectomy if found to have a BRCA1 mutation).

The advice was that each case should be considered on an individual basis.

If a patient could not have envisaged that a sample could be used to help family members, then generally under the Data Protection Act, s/he must be informed of this fact. However, if providing this information to the person who gave the sample would involve 'disproportionate effort' then an exemption is available.

The term 'disproportionate effort' is not defined in the Act. What does or does not amount to disproportionate effort is a question of fact to be determined in each and every case. The Information Commissioner has advised us that a number of factors should be taken into account, including the nature of the data, the length of time and the cost involved to the data controller in contacting the original patient to provide the fair processing information.

The Joint Committee believes that it is particularly important to consider the 'nature' of the data to be used in helping another family member in this situation. Two points which may be useful

to consider are the extent to which the result in itself reveals information about the sample donor, and its likely effect on the individual as a consequence of its disclosure. We note from talking to heads of laboratories that a description of a change in DNA structure causing a genetic disorder is currently unlikely to reveal any information about the person who gave the sample apart from the fact that s/he has the disease for which the test was taken in the first place.

If, having considered all the factors, a health professional wishes to share information for clinical care (for instance, scientific information about a mutation) to another health professional under this exemption of the Data Protection Act, a note should be kept of the grounds, as outlined above.

For new samples, the requirements of the Data Protection Act can be met by the health professional ensuring that the patient is made aware that the test results could be used to provide appropriate management for other family members. We believe that this is covered by statement 5 on the Record of consultation form in Appendix 2.

## APPENDIX I

# Twelve key points on consent: the law in England

The following points are taken from Appendix A of the Department of Health *Reference guide to consent for examination or treatment*.<sup>2</sup>

### When do health professionals need consent from patients?

1. Before you examine, treat or care for competent adult patients you must obtain their consent.
2. Adults are always assumed to be competent unless demonstrated otherwise. If you have doubts about their competence, the question to ask is: 'can this patient understand and weigh up the information needed to make this decision?' Unexpected decisions do not prove the patient is incompetent, but may indicate a need for further information or explanation.
3. Patients may be competent to make some healthcare decisions, even if they are not competent to make others.
4. Giving and obtaining consent is usually a process, not a one-off event. Patients can change their minds and withdraw consent at any time. If there is any doubt, you should always check that the patient still consents to your caring for or treating them.

### Can children consent for themselves?

5. Before examining, treating or caring for a child, you must also seek consent. Young people aged 16 and 17 are presumed to have the competence to give consent for themselves. Younger children who understand fully what is involved in the proposed procedure can also give consent (although their parents will ideally be involved). In other cases, someone with parental responsibility must give consent on the child's behalf, unless they cannot be reached in an emergency. If a competent child consents to treatment, a parent **cannot** override that consent. Legally, a parent can consent if a competent child refuses, but it is likely that taking such a serious step will be rare.

### Who is the right person to seek consent?

6. It is always best for the person actually treating the patient to seek the patient's consent. However, you may seek consent on behalf of colleagues if you are capable of performing the procedure in question, or if you have been specially trained to seek consent for that procedure.

### What information should be provided?

7. Patients need sufficient information before they can decide whether to give their consent: for example information about the benefits and risks of the proposed treatment, and alternative treatments. If the patient is not offered as much information as they reasonably need to make their decision, and in a form they can understand, their consent may not be valid.

### Is the patient's consent voluntary?

8. Consent must be given voluntarily: not under any form of duress or undue influence from health professionals, family or friends.

### Does it matter how the patient gives consent?

9. No consent can be written, oral or non-verbal. A signature on a consent form does not itself prove the consent is valid – the point of the form is to record the patient's decision, and also, increasingly, the discussions that have taken place. Your Trust or organisation may have a policy setting out when you need to obtain written consent.

### Refusals of treatment

10. Competent adult patients are entitled to refuse treatment, even where it would clearly benefit their health. The only exception to this rule is where the treatment is for a mental disorder and the patient is detained under the Mental Health Act 1983. A competent pregnant woman may refuse any treatment, even if this would be detrimental to the fetus.

### Adults who are not competent to give consent

11. **No-one** can give consent on behalf of an incompetent adult. However, you may still treat such a patient if the treatment would be in their best interests. 'Best interests' go wider than best medical interests, to include factors such as the wishes and beliefs of the patient when competent, their current wishes, their general well-being and their spiritual and religious welfare. People close to the patient may be able to give you information on some of these factors. Where the patient has never been competent, relatives, carers and friends may be best placed to advise on the patient's needs and preferences.
12. If an incompetent patient has clearly indicated in the past, while competent, that they would refuse treatment in certain circumstances (an 'advance refusal'), and those circumstances arise, you must abide by that refusal.

This summary cannot cover all situations. For more information, see the pages on the Department of Health website on consent:

[www.dh.gov.uk/PolicyAndGuidance/HealthAndSocialCareTopics/Consent/fs/en](http://www.dh.gov.uk/PolicyAndGuidance/HealthAndSocialCareTopics/Consent/fs/en)

## APPENDIX 2

### Record of consultation and agreement over genetic testing and sharing of information: sample form

Genetics record number:

Name:

Date of birth:

Address:

During this consultation we have discussed the following issues, and you have agreed to the uses shown below. Please cross out the words 'Discussed', 'Agreed', 'Not applicable' as appropriate for each use.

1	I agree to analysis of the sample for .....	Discussed	Agreed	Not applicable
2	I agree to the sample being stored in case future checks or tests are needed	Discussed	Agreed	Not applicable
3	I would like to be contacted <u>before</u> further tests are done on the stored sample if new tests become available	Discussed	Agreed	Not applicable
OR				
4	I am happy for further diagnostic tests on the stored sample to be undertaken without being contacted	Discussed	Agreed	Not applicable
5	I agree that information and test results may be shared to help other family members	Discussed	Agreed	Not applicable

Signed..... Date.....  
(Clinician)

Please confirm your agreement by adding your signature to this form below:

Signed.....  
(Patient/Parent)

We may keep any leftover samples to check the quality of our results for other patients. We make sure that nobody knows whose sample is helping us to do this.

Copy:   Records  
          Patient/Parent

## APPENDIX 3

### Agreement to a photographic record (still or video): sample form

Date:	Consultant:
First names:	
Surname:	
Date of birth:	Record number:

I agree that the photographic images of

.....

(a)	can be stored as a confidential medical record and used as an aid to diagnosis	Discussed	Agreed	Not applicable
(b)	may be shown to appropriate health professionals to aid medical teaching and research	Discussed	Agreed	Not applicable

Please cross out the words 'Discussed', 'Agreed', 'Not Applicable' as appropriate.

If we think it would be valuable for the images to be published in a journal, textbook, as part of a display or information leaflet or on an open access website (which may be seen by the general public, as well as medical professionals), we would contact you again.

Signature(s) ..... Parent/Guardian/Patient

The type of permission you give will not affect your treatment in any way. If in the future you wish to change your mind, you have the right to do so at any time by writing to the Clinical Genetics Unit.

Joint Committee on Medical Genetics: *Consent and confidentiality in genetic practice: guidance on genetic testing and sharing genetic information*

# References and further reading

## References

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## Further reading

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- Department of Health. The Caldicott Committee. *Report on the review of patient-identifiable information*. London: DH, 1997.
- Department of Health. *Code of practice and guidance on genetic paternity testing services*. London: DH, March 2001.
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## Additional legal documents of interest

- Health and Social Care Act 2001.
- Department of Health. *Health service guideline* (96) 18, The protection and use of patient information, 7 March 1996.
- The Children Act 1989.
- Children (Scotland) Act 1995.
- Legal Capacity (Scotland) Act 1991.
- Re Y (mental patient: Bone marrow donation)[1997].
- Gillick v West Norfolk and Wisbech AHA* [1986] AC 112.
- The Medicines for Human Use (Clinical Trials) Regulations 2004, available on [www.legislation.hmso.gov.uk/si/si2004/20041031.htm](http://www.legislation.hmso.gov.uk/si/si2004/20041031.htm)