

# JOINT COMMITTEE ON MEDICAL GENETICS

Joint Committee on Medical Genetics of the Royal College of Physicians, Royal College of Pathologists and British Society for Human Genetics

**Draft document for consultation**

## Consent and confidentiality in genetic practice:

*guidance on genetic testing and sharing genetic information*

- General aspects of consent as applied to genetics
- The sharing of information to other family members and between professionals
- Genetic investigations including those performed on stored material
- The Data Protection Act and the processing of medical genetic information

This draft document is being made available for consultation until Friday January 9th 2004. Comments are welcomed and should be forwarded to the Administrator of the Joint Committee on Medical Genetics: Mr Simon Land, Royal College of Physicians, 11 St Andrews Place, Regent's Park, London NW1 4LE  
Email: [Simon.Land@rcplondon.ac.uk](mailto:Simon.Land@rcplondon.ac.uk). Telephone: 020 7935 1174 Ext 307.

October 2003

This report on Consent and Confidentiality has been compiled by Fiona Douglas and Peter Farndon and informed by a working group of the Joint Committee on Medical Genetics.

Members of the working group were John Barber, Alastair Bellingham, Carol Chu, Fiona Douglas (Chair), Judith Goodship, Penny Guilbert, Neva Haites, Alastair Kent, Sandy McCall Smith, Stewart Payne. Observers: Elaine Gadd (Department of Health), Peter Farndon (Chair, Joint Committee)

**CONSENT AND CONFIDENTIALITY IN GENETIC PRACTICE:  
GUIDANCE ON GENETIC TESTING AND SHARING GENETIC  
INFORMATION**

**October 2003**

This document is a report from the Joint Committee on Medical Genetics in response to requests from genetic professionals for clarification of issues of consent and confidentiality in clinical practice, particularly with regard to the requirements of the Data Protection Act. The Joint Committee set up a working party which undertook a questionnaire survey of genetics units in the United Kingdom to assess current practice and areas of concern. The working party took medico legal advice on current legislation. Issues were debated at length both within the working party and within the Joint Committee on Medical Genetics.

It was originally intended that this document would contain a detailed description of the current medico legal framework. This has not been included as a recent report from the Human Genetics Commission (“Inside Information: Balancing interests in the use of personal genetic data” 2002<sup>1</sup>) reviews existing legislation in detail.

Our report is now being made available for consultation. It not only identifies issues surrounding consent and confidentiality in genetic practice but also identifies key practice points and offers examples of documents which may be helpful adjuncts when seeking consent. On some issues the report makes recommendations for practice.

The four sections in this report cover:

- 1 General aspects of consent as applied to genetics;
- 2 The sharing of information to other family members and between professionals;
- 3 Genetic investigations including those performed on stored material;
- 4 The Data Protection Act and the processing of medical genetic information.

In this report, the term consultand is used to mean the individual (not always affected) who is requesting genetic information; the proband is the first person in a pedigree to be identified clinically as being affected by the disorder.

## **SECTION 1: CONSENT IN MEDICAL GENETIC PRACTICE - GENERAL ASPECTS**

### **1.1 General principles**

Consent is a process to ensure that a person understands the nature and purpose of giving a sample or undergoing an intervention. In medico-legal terms, adequate consent is ensured if anything that represents a significant risk that would affect the judgement of a reasonable person has been raised. Indeed, it is impossible for the consent process to be totally comprehensive; genetic tests may reveal unexpected results, for instance.

General guidelines for obtaining consent for examination and treatment have been issued by the Department of Health<sup>2</sup> [www.doh.gov.uk/consent](http://www.doh.gov.uk/consent) (*12 key points from these guidelines are summarised in appendix 1*) and underpin the practice of genetics just as in any other speciality. The General Medical Council has also published guidelines (GMC: Seeking patients' consent: the ethical considerations; November 1998)<sup>3</sup>

Most issues regarding consent in children and in adults lacking capacity are covered in the Guidance from the Department of Health and General Medical Council.

- **Consent should be obtained prior to a test with genetic implications being performed. Except in the exceptional circumstances detailed later, consent should have been obtained before medical genetic information is disclosed.**

### **1.2 Gaining consent in clinical genetic practice**

Guidelines from the Department of Health place the onus for consent with the clinician obtaining the genetic information or the sample.

The seeking of consent should be regarded as a means to ensure that a person understands the nature and purpose of giving a sample. Appropriate information and the opportunity to discuss issues before investigations are undertaken should be offered.

**FOR CONSULTATION**

Genetics presents some issues which appear to be specific to the clinical practice of the subject. These include the taking of a pedigree to record a family history of disease, and the using of pedigree information and genetic test results for the benefit of the whole family. Other family members may then need to be contacted to be offered testing to delineate their risk of being affected by or being a carrier of the genetic disorder. Occasionally a family member does not wish to contact other family members personally but still wishes genetic information to be available for others. It is therefore good practice to confirm that this sharing of information with other family members is acceptable to the individual being seen and to document this.

In some instances samples are taken for storage for future testing should a test become available. In other instances, a sample may be stored and/or used for quality assurance of laboratory tests for other patients. Unexpected results may be revealed by a genetic test, for instance information about parentage.

It is important that the primary health care team and other health professionals involved are kept informed about the implications of a genetic diagnosis; our questionnaire 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.

As molecular and cytogenetic 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.

- **We recommend that the issues in box 1 are discussed explicitly with patients when medical and pedigree information is being obtained and when genetic investigations are requested.**

**Box 1**

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

- The use and sharing of information (pedigree, diagnosis, affected/carrier status, test results) for the benefit of family members
- The nature of the testing to be undertaken
- The possible prolonged nature of the testing process
- The possibility that testing may reveal unexpected results (for instance information about parentage)
- The storing of samples
- Sharing information with health professionals including the primary care team

The use of samples for quality assurance does not need explicit consent under DH guidelines as long as there is a sentence explaining this in the literature available to genetics service patients (See 1.14).

### **1.3 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 particularly so 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 in Appendix 2 as a suitable example or as a record of the discussion.

- **In line with the Department of Health's guidelines on consent, the Joint Committee recommends that the health professional dealing with a patient or family should explain the genetic issues, including offering written information sheets where appropriate, and asking for formal recognition that the process had been completed by the signing of a form.**

#### **1.4 Responsibility of the clinician to obtain consent**

In current practice the assumption is that when a laboratory receives a sample the clinician responsible for the care of the patient has obtained appropriate and adequate consent; this ensures that samples which require to be processed immediately can be so treated. This particularly applies to samples requiring cytogenetic investigation.

- **The working party is seeking views on whether there is support for including on genetic laboratory test request forms a statement confirming that issues of testing and storing samples have been discussed. We suggest that a suitable statement is "In submitting this sample the clinician confirms that consent for testing and possible storage has been obtained".**

#### **1.5 Consent for testing and storing samples received from specialties other than clinical genetics**

Genetic laboratories receive a large number of samples from all branches of medicine and surgery. The Joint Committee recognises that genetics health professionals, particularly within clinical genetics, have a role in disseminating good practice regarding the issues particular to the testing and storage of genetic samples as discussed in this document. We ask genetics professionals to undertake this role. This is especially important if the request forms are amended to include a statement as in the above paragraph.

## **1.6 Children**

Parents 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 *Genetic Testing in Childhood* which highlights practical and ethical issues in more detail<sup>4</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 is postponed until the child is at an age to appreciate the issues and give consent (ie demonstrate Gillick competence).**

## **1.7 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. This situation is not changed by the draft Mental Incapacity Bill<sup>4</sup>. This Act will however give a person with power of attorney or a court appointed deputy the right to give consent on behalf of such an adult.

### **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>6</sup>. In English common law it is lawful to provide 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**

In Clinical Genetics an investigation in one individual may not change the care of that individual or be in their direct medical interests, but may be of great benefit to other family members by establishing the diagnosis or delineating a pathogenic mutation and hence recurrence risks within the wider family.

The Genetic Interest Group has argued strongly that one should not presume that an adult with incapacity would be less altruistic than a competent adult in wishing to assist other family members in genetic investigations<sup>7</sup>

The Department of Health guidelines note that care should be taken not to underestimate the capacity of a patient with a learning difficulty to understand, and many people with learning difficulties have the capacity to consent if time is spent explaining to the individual the issues in simple language.

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 note that it would be helpful to some families with genetic disorders if this 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. An intervention can be undertaken, however, if it is deemed to be in the best interests of that person, when consent is not necessary. The draft Mental Incapacity Bill changes this. If a person lacks capacity and a person has been appointed with power of attorney or as a deputy by a court then the appointed person can give consent on their behalf. However an intervention must still be in the person's best interests<sup>5</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 well being 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 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.

- **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 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>6</sup> but in Scotland carers of such an adult can refuse on his or her behalf even if family members regard the consent to provide the individual with indirect benefit.

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

It is sometimes desirable to store a sample with the expectation that genetic analysis may become available in the future.

- **The Joint Committee recommends that consent for storing the sample, prospective consent for testing, and consent to re-contact either the patient or a specified relative be obtained when a sample is taken.**

This will also ensure that in the event of that person's death the sample may be available and used for the benefit of other family members. We recommend that this is by spoken and written information supported by a consent form (example in appendix 2).

Depending on the policy of the genetics unit, it may be necessary to include an explanation that it may not be possible to offer automatic testing and recontact if genetic analysis becomes available.

### **1.9 Counselling as a part of the consent procedure for 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 this should be so in predictive testing programmes for single gene disorders such as Huntington's disease or those single genes predisposing to cancer.

In some circumstances, however, an insistence on counselling may not be appropriate. We agree with the Genetic Interest Group that the consent process should not be used to the detriment of patients, such as 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 General Medical Council's recommendation that when providing information, health professionals must do their best to find out about patients' individual needs and priorities. For example, patients' beliefs, culture, occupation or other factors may have a bearing on the information they need in order to reach a decision. Assumptions should not be made about patients' views, but these matters should be discussed with them.

Whilst good practice is always 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>8</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 result from the sample either. The Joint Committee recognises that this is acceptable providing that the family members understand that it is their decision not to receive information, and a note of their wishes is made in the medical records.

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

It is important that families are kept up to date with the progress of laboratory testing. A genetics laboratory may recommend that additional 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 if 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 inform them of 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 Department of Health 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.

### **1.11 Consent to clinical photography and video recording**

Clinical photographs and video recordings may be used both as a medical record and as a tool for teaching, audit and research. 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 and that when required or appropriate the photographs or video can be anonymised. A specimen consent form is in appendix 3.

### **1.12 Duration of the consent**

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 also 1.7.

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

**a) To offer testing to family members**

A test result in one member of a family may allow specific testing to be offered to other family members, for diagnosis, surveillance for disease complications or for carrier status. We recommend that family members are referred to the local Genetics Department in these instances.

**b) To assist in the interpretation of the proband's result**

A sample from a family member who lives in another part of the country may be necessary to assist the interpretation of a test result for the consultand. The details of family members who are willing to assist are usually made available via the consultand. Clinical geneticists then seek the help of a clinical colleague to take the sample, most frequently the patient's general practitioner.

As already stated, it is the responsibility of the person directly taking the sample to obtain consent for taking, analysing and storing the sample.

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

**1.14 Consent for the use of samples as controls and in quality assurance**

Genetics laboratories use samples in which DNA mutations have been identified as quality controls in the analysis of other samples to maintain standards and ensure good practice.

The Department of Health'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.

- **We recommend that a statement is incorporated in the general information which Genetics Departments give to patients when samples are taken. An example might be: "So that our laboratories can continually monitor the quality of their results, they may anonymise a sample after it has been tested for use in quality assurance".**

#### **1.15 Issues of consent and confidentiality in complex clinical cases**

- **The Joint Committee is seeking views on a suggestion it has received to recommend the setting up of a clinical ethics committee for issues of consent and confidentiality regarding complex clinical cases that lead to an ethical dilemma such that local consensus could not be achieved.**

It is proposed that this national multi-professional and multi-disciplinary committee should involve geneticists, genetic counsellors, ethicists, members of patient interest groups and also representation from the wider public. The use of discussion by e-mail may allow a rapid response for urgent situations. However it is acknowledged that a high level of commitment and acceptance by all the relevant professional groups would be required. It is important that this is not seen as erosion of clinical autonomy but as an aid to decision making.

---

## **SECTION 2: GIVING AND SHARING GENETIC INFORMATION**

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 test.

The use and disclosure of medical information comes under the guidelines of bodies such as the General Medical Council. However, the processing of the information comes under the provisions of the Data Protection Act. Clarification has been sought from the Information Officer 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 4.

### **2.1 The family history**

Documenting an accurate family history (or pedigree) of disease may be necessary to make an initial specific genetic diagnosis. 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 has been given. The 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. We recommend that either a note is written in the medical record, or a consent form used.

A family member is unlikely to know confidential medical information other than that which has been given by the person concerned. That a person has a genetic disorder is likely to be known within a person's friends and acquaintances. Therefore much of the information given in a pedigree during a genetic consultation may be freely available, although it should still be treated in confidence.

## **2.2 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) and the Access to Health Records Act (1990)<sup>9,10</sup>.

### **a) Family members who are living**

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

### **b) 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 is at the discretion of the person holding them, unless the deceased refused permission. Some hospitals choose to allow access to such records only with consent from the spouse. The possibility that information within the records would be of benefit to the health of surviving relatives is 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.

- **We recommend that hospital medical records departments accept consent from blood relatives where there is a risk of an inherited genetic disorder in the family.**

The Human Genetics Commission has extended the limited circumstances for the revealing of confidential information obtained in a medical context by adding where disclosure is in the interests of relatives.

### **2.3 The use of existing genetic information to facilitate accurate genetic testing**

A person usually asks for genetic testing because of knowledge that a family member (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 consultand 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 consultand 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 consultand'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 due to the technique employed in the laboratory testing the consultand's sample not being able to detect the particular type of mutation.

Technical information therefore needs to be shared confidentially between the heads of laboratories undertaking the testing, or transferred from one family member's record to another in the same laboratory to permit testing.

### **2.4 Disclosure of information**

#### **a) Where consent has been obtained**

Disclosure of medical information is of course acceptable when consent has been obtained. Most family members wish their information to be available to help relatives. We recommend the good practice of obtaining consent for this disclosure from family members wherever they are seen.

**b) Where the basis of consent is unclear**

As discussed earlier in the section on obtaining consent, we recognise that there are clinical circumstances where the basis of consent previously sought is unclear. This usually involves the use of stored samples or the use of test results, particularly technical information about a mutation which has been identified in a family member.

In some instances seeking specific consent for the use of a stored sample or results may not be possible, or there may be good reason to believe that more harm may result to a family member in not using the sample than to another from the use of the sample or result without confirming that consent has been granted.

In some cases the confidentiality of the family member seeking genetic testing would be compromised by actively seeking consent to use information derived from another family member. For example, an individual who is seeking prenatal diagnosis may not wish anyone to know of the pregnancy until the test results are available. The necessity to seek consent for release of the information leads to a breach of confidence for the pregnant woman.

Both of these concepts are legally recognised.

The balance, then, must be carefully considered by the health professional and the clinical judgement documented. In these exceptional circumstances, the sample and/or result may be used. This advice is based on current GMC guidelines, but 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.

**c) Where consent to release information has been refused.**

The Human Genetics Commission<sup>1</sup>, the Nuffield Council on Bioethics<sup>13</sup> and the General Medical Council<sup>11</sup> have all expressed the view that that the rule of confidentiality is not

absolute. In special circumstances it may be justified to break confidence where a person declines 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.

This would arise where the benefit of disclosure substantially outweighs the patient's claim to confidentiality<sup>1,11</sup>

Such disclosure in these circumstances should be on the proviso that an attempt has been made to persuade the patient in question to consent to disclosure; the benefit to those at risk is so considerable as to outweigh any distress which disclosure would cause the patient; and the information is, as far as possible, anonymised and restricted to that which is strictly necessary for the communication of risk<sup>5</sup>.

We recommend that before disclosure is made when consent has been withheld, the situation should be discussed carefully with professional colleagues and the reasons for disclosure documented. Current GMC guidance states that the individual should generally be informed before disclosing the information<sup>11</sup>.

## **2.5 Related issues**

### **a) 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.

**b) 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.

**c) Cancer registries**

Cancer registries are extremely valuable sources of information. If the person is alive, consent is sought and demonstrated before information is released; for deceased relatives the UK cancer registries will release information to clinical genetics units under the mutual understanding of respect for confidentiality. We strongly support this vital practice for clinical care.

**d) 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. For extremely rare diseases, this testing may fall within the remit of the National Genetics Reference Laboratories.

---

**SECTION 3: GENETIC INVESTIGATIONS ON STORED SAMPLES OR ARCHIVAL PATHOLOGICAL MATERIAL**

Performing genetic tests on stored samples may be vital for diagnosis, and most commonly involves DNA samples and histological material. Testing archived pathological material is particularly useful where an inherited predisposition to cancer is suspected.

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

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 then.

**a) Stored samples from a living patient**

If the person is still alive, attempts should be made to contact them to seek their permission. Often contact can be re-established through the family member who may stand to benefit from the results of the testing.

We recognise, however, that there are clinical circumstances where seeking specific consent for the use of a stored sample or results may not be possible, or there may be good reason to believe that more harm may result to a family member in not using the sample than to another from the use of the sample/result without consent. These circumstances and possible resolutions have been discussed in Section 2.4 on disclosure.

**b) Samples taken at post mortem examination**

Post mortem material is covered by the Human Tissue Act 1961 – the person lawfully in possession of the body may 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>12</sup>. The pathologist may wish to ask the genetic department to seek consent from at least one surviving relative to demonstrate there was no reason to believe that there would be an objection before releasing material. Although

verbal consent, documented in genetics notes, is medico-legally adequate, some genetics departments formalise this with written consent.

The Joint Committee on Medical Genetics noted that the existing Act could preclude the use of tissue to aid genetic diagnosis in the blood relatives of the deceased, for example because of a surviving spouse vetoing that use for step children. The Joint Committee strongly recommends that any future legislation recognises the needs of blood relatives regarding genetic testing.

### **3.2 Governmental reviews of the legal status of samples and proposed regulatory framework**

The legislative framework relating to post-mortem tissues is under review and the proposed new legal framework can be found in the Government's consultation document "Human Bodies, Human Choices": [www.doh.gov.uk/tissue/review\\_of\\_law.htm](http://www.doh.gov.uk/tissue/review_of_law.htm).

This proposes either "respectful disposal" or "samples being made available for respectful use in ways which ensure that the deceased can contribute to the benefit of either their own families or to the communities as a whole". This view is endorsed by the report of the Human Genetics Commission<sup>5</sup>.

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 (<http://www.doh.gov.uk/tissue/pmadultinfo.pdf>).

The draft consent form (September 2002) for adult post mortem examination 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" ([www.doh.gov.uk/tissue/family.htm](http://www.doh.gov.uk/tissue/family.htm))

The Retained Organs Commission has been seeking views on the future legal status of tissue blocks and slides and (a) whether specific consent (in addition to that obtained for an autopsy or surgical procedure) for the retention and use of tissue blocks and slides is required and/or (b) whether they should be counted as part of the patient's medical record and therefore outside the constraints of specific consent. The consultation period ended in February 2003 and the results are awaited. [www.nhs.uk/retainedorgans/index.htm](http://www.nhs.uk/retainedorgans/index.htm)

In all three consultation documents there is some recognition of the importance of genetic testing utilising both surgical as well as autopsy derived tissues. However, there is no explicit recognition that tissues for genetic testing are often taken in addition to tissue biopsies destined to be "tissue blocks and slides". Tissues (and their derivatives) taken for genetic testing and storage include chromosome preparations in fixative, fibroblast cell lines, unfixed tissues stored at  $-20^{\circ}\text{C}$  and also cell lines (fibroblasts and lymphoblastoid) stored in liquid nitrogen. It is not known whether the legal status of tissues, DNA pellets, cell lines and other stored genetic material will be included in the final recommendations from the consultations.

Furthermore, the concept that genetic testing although possibly not directly benefiting the patient may nevertheless have profound consequences for the extended family in future generations, is barely recognised in the above documents. It is essential that the legal status of tissues (from surgical as well as autopsies) destined for genetic testing must have a clearly defined legal status which corresponds to that finally decided for tissue blocks and slides. If all tissues were legally defined as part of the patient's medical record and therefore kept indefinitely, they could be used for proactive testing as well as altruistic quality assurance purposes as and when required.

In "Human Bodies, Human Choices", the government has proposed making the handling, use and storage of human tissues taken without the appropriate informed consent a criminal act. This will include not only those primarily responsible for obtaining the consent, but also the Scientists processing the tissues. It is particularly important, therefore, that genetic issues are considered when the proposed Parliamentary Bill is presented.

## **4 THE DATA PROTECTION ACT AND THE PROCESSING OF MEDICAL GENETIC INFORMATION**

### **4.1 Disclosure of information from family pedigrees**

Our interpretation of detailed advice from the Information Officer is that information on a family pedigree can be passed between health professionals (under Schedule 3) 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 (clinic and laboratory) if necessary for the care of family members.

### **4.2 Fair processing and disclosure of results of genetic tests**

We asked the Information Officer 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 twenty 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 a case by case 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, he or she 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 Officer 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 he or she 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 E on the record of consultation form in Appendix 2.

## REFERENCES AND FURTHER READING

(URLS were accessed 14 November 2003)

1. Human Genetics Commission. *Inside Information: Balancing interests in the use of personal genetic data* 2002  
[www.hgc.gov.uk/insideinformation/index.htm](http://www.hgc.gov.uk/insideinformation/index.htm)
2. Department of Health. *Reference Guide to Consent for Examination or Treatment* 2001  
[www.doh.gov.uk/consent/guidance.htm](http://www.doh.gov.uk/consent/guidance.htm)
3. General Medical Council. *Seeking patients' consent: the ethical considerations* (1998) GMC: London [www.gmc-uk.org/standards/consent.htm](http://www.gmc-uk.org/standards/consent.htm)
4. The Genetic Testing of Children. *Report of a Working Party of the Clinical Genetics Society (UK)*  
J Med Genet 1994;31: 785-797 [www.bshg.org-uk/official%20docs/testchil.htm](http://www.bshg.org-uk/official%20docs/testchil.htm)
5. Draft Mental Incapacity Bill. *Presented to Parliament June 2003 / Mental Incapacity Act 2003.*
6. Adults with Incapacity (Scotland) Act 2000
7. Genetic Interest Group *Confidentiality Guidelines* 1998 [www.gig.org.uk/docs/gig-confidentiality.pdf](http://www.gig.org.uk/docs/gig-confidentiality.pdf)
8. Human Rights Act 1998
9. Data Protection Act 1998
10. Access to Health Records Act 1990
11. General Medical Council, *Confidentiality: Protecting and providing information* GMC: London [www.gmc-uk.org/standards/secret.htm](http://www.gmc-uk.org/standards/secret.htm)
12. Human Tissue Act 1961. September 2000
13. Nuffield Council on bioethics. *Mental disorders and genetics: the ethical context (1998)*  
[www.nuffieldbioethics.org/filelibrary/pdf/mentaldisorders2.pdf](http://www.nuffieldbioethics.org/filelibrary/pdf/mentaldisorders2.pdf)

## Further Reading

Advisory Committee on Genetic testing: *Report on genetic testing for late onset disorders* 1998

Anne Marie Slowther, Tony Hope. *Clinical ethics committees*. British Medical Journal 7262 649-650 2000

Department of Health. The Caldicott Committee: *Report on the review of patient-identifiable information*. 1997

Department of Health. *Code of Practice and guidance on genetic paternity testing services* Department of Health March 2001

Medical Research Council. *Guidelines for good clinical practice in clinical trials* 1998

Medical Research Council. *Guidelines: Personal Information in Medical Research* 2000

The Royal Liverpool Children's enquiry: Summary and recommendations Jan 01

Royal College of Pathologists. *Guidelines for the retention of tissues and organs at post mortem examinations* March 2000.

Royal College of Pathologists. *Transitional guidelines to facilitate changes in procedures for handling "surplus" and archival material from human biological samples*. Royal College of Pathologists 2001.

UNESCO. Universal Declaration on the Human Genome and Human Rights.

United Kingdom Central Council for Nursing, Midwifery and Health Visiting. *Guidelines for professional practice* 1996

United Kingdom Central Council for Nursing, Midwifery and Health Visiting. *Guidelines for records and record keeping* 1998

World Health Organisation. *Guidelines on Ethical Issues in Medical Genetics and the Provision of Genetics Services* WHO 1995

**Legal Documents of interest**

Health and Social Care Act 2001

Department of Health, Health service guideline (96) 18

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

**APPENDIX 1:**

## **Consent**

12 key points on consent: the law in England

<http://www.doh.gov.uk/consent/twelvekeypts.htm>

### **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 health care 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, some- one 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 detail, consult the *Reference guide to consent for examination or treatment* , available from the NHS Response Line 08701 555 455 and at [www.doh.gov.uk/consent](http://www.doh.gov.uk/consent)

APPENDIX 2

Record of Consultation

Genetics Record Number:

Name:

Date of birth:

Address:

During this consultation we have discussed the following issues, and I believe that you have agreed to the uses indicated. (Please put ticks or crosses in the boxes)

<u>Issue</u>	<u>Discussed</u>	<u>Agreed</u>
A I agree to analysis of the sample for .....	<input type="checkbox"/>	<input type="checkbox"/>
B I agree to the sample being stored in case future checks or tests are needed	<input type="checkbox"/>	<input type="checkbox"/>
C I would like to be contacted if new tests become available <u>before</u> further tests are done on the stored sample	<input type="checkbox"/>	<input type="checkbox"/>
OR		
D I am happy for further diagnostic tests on the stored sample to be undertaken without being contacted	<input type="checkbox"/>	<input type="checkbox"/>
E I agree that information and test results may be shared to help other family members	<input type="checkbox"/>	<input type="checkbox"/>

Signed .....  
(Clinician)

Date ... ..

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

Signed .....  
(Patient/Parent)

So that our laboratories can continually monitor the quality of their results, they may anonymise a sample after it has been tested for use in quality assurance.

Copy Records  
Patient/Parent

APPENDIX 3

Agreement to a photographic record

Date: \_\_\_\_\_ Consultant: \_\_\_\_\_  
 First Names: \_\_\_\_\_  
 Surname: \_\_\_\_\_ : \_\_\_\_\_  
 Date Of Birth: \_\_\_\_\_ Record Number: \_\_\_\_\_

I agree that the photographic images of

.....  
 .....  
 .....  
 .....  
 .....

can be stored

- |  | <u>Discussed</u>         | <u>Agreed</u>            |
|--|--------------------------|--------------------------|
| a) as a confidential medical record and used as an aid to diagnosis.   | <input type="checkbox"/> | <input type="checkbox"/> |
| b) for the purpose of medical teaching and research may be shown to the appropriate health professionals.  | <input type="checkbox"/> | <input type="checkbox"/> |
| c) may be published in a journal, textbook, as part of a display or information leaflet or on an open access web site, which may be seen by the general public as well as medical professionals. | <input type="checkbox"/> | <input type="checkbox"/> |

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

If in the future, you wish to withdraw this consent, you have the right to do so at any time by writing to the Clinical Genetics Unit  
 The choice of level of consent will not affect your treatment in any way.