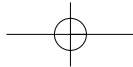


Consent and confidentiality in clinical genetic practice: Guidance on genetic testing and sharing genetic information

A report of the Joint Committee
on Medical Genetics

2nd edition September 2011



This report, *Consent and confidentiality in clinical genetic practice*, is the second such report from the Joint Committee on Medical Genetics. The first report was published in 2006. It was substantially rewritten in 2010/11 by Anneke Lucassen (CELS, University of Southampton) and Alison Hall (PHG Foundation).

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The exceptions to the above are the flowcharts on pp 32–4, which may be photocopied for clinical use.

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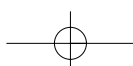
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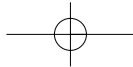
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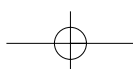
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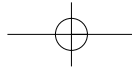
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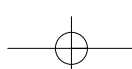


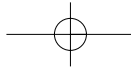
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Foreword

The distinctive nature of consent in genetic practice

Reliable information is the keystone to good practice in healthcare. Patients are content to undergo tests and to entrust confidential information to professionals in order to ensure that they get sound advice and treatment. The distinguishing feature in clinical genetics is that much of this information affects not only the individual patient but also other family members. The principles of confidentiality and data protection underpin health services and give patients the reassurance that their privacy is properly protected. However, these are not so straightforward in the genetics context where the result of a genetic test provides information not just about the patient but also about others.

As the scope of genetic testing increases, the management of that information may generate new challenges for both the patient and the healthcare professional. Patients generally expect professionals to be able to access their health information and rely on their expertise and experience to interpret it so as to advise them wisely. Such information may be personal and private and intrusion into privacy is usually justified by the fact that the patient has authorised it, possibly explicitly but sometimes implicitly within a broader request for advice or treatment. However, the information obtained may enable inferences to be drawn about other family members, whose views may not be known. The information has been generated in circumstances of confidence to one person, but is also of significance to another. Clinical genetic services will often want to use that information to assist the other person but may be unsure whether this is acceptable within the rules of data protection and confidentiality.

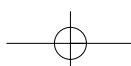
It is open to patients to authorise the use of their confidential information for other purposes, including research and the treatment of others, and where a person agrees to the information generated being used in this way, there is little cause for concern. This form of altruism or 'genetic solidarity' is likely to be beneficial to others, and it may be appropriate to take steps to encourage patients to give such consent even if they are initially hesitant. Much clarity can be achieved by proactive seeking of consent on these issues.

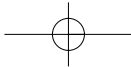
In some circumstances, patients may be reluctant to give such consent, or clinicians involved may think it inappropriate to ask for such consent which may be many years after a health professional's contact with the patient. Consideration needs to be given to whether it might be legitimate to use the information in question even without consent. Such use may not involve disclosure of the information in question to a third party, but merely the reliance on it by the health professional, when contacting another person involved in the care of the patient, or in assessing their risk of disease.

The privacy protection established by the European Convention on Human Rights, Article 8, recognises that the value of privacy needs to be balanced against the rights and freedom of others. It may be acceptable to interfere with such privacy provided that the interference is proportional to the protection afforded to others. Such balancing acts may be difficult and healthcare professionals may be uncertain in which circumstances certain disclosure actions are legitimate. This report can only provide guidance, but aims to indicate what current acceptable practice might be. The introduction of case scenarios throughout the report provides helpful illustrations of such practice.

September 2011

Professor Jonathan Montgomery
Professor in Health Care Law, University of Southampton



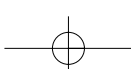


Introduction to the revised version

This report aims to provide up-to-date guidance on issues of consent and confidentiality arising in clinical genetic* practice. The first version of this guidance was aimed specifically at professionals within the specialty of clinical genetics and was partly prompted by proposed legislative changes such as those governing the use of human tissue. This new report recognises that many of these issues are becoming increasingly relevant for all medical specialties as clinical genetic practice branches out into mainstream medicine, and outlines relevant changes in legislation and professional guidelines.

The main section of the report introduces general principles of consent and confidentiality in clinical genetic practice and expands on these using clinical case scenarios as illustrations. This section explores the issues involved in taking a family history and in the giving and sharing of genetic information and samples. The appendices outline and examine relevant recent changes in legislation, in particular the implementation and impact of two major pieces of legislation, the Human Tissue Act 2004¹ and the Mental Capacity Act 2005,² which came into force after the first version of this document was published, as well as relevant aspects of the Data Protection Act 1998.³ The guidance also takes account of the revised General Medical Council (GMC) guidance on consent⁴ and confidentiality⁵ and the House of Lords Science and Technology Committee Inquiry on Genomic Medicine (2009).⁶ Throughout this document, the underlying ethical and legal principles are illustrated using hypothetical (but based on real) clinical cases to highlight key points. Each case should be judged on its own facts, as the clinical scenarios are intended to guide rather than dictate practice.

*We refer to genetic practice throughout this document. However, we acknowledge that there is considerable overlap, and consequently no clear distinction from, genomic practice.

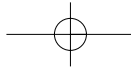


Summary of recommendations

The Joint Committee on Medical Genetics makes the following recommendations for good clinical practice.

- 1 As with other types of medical information, genetic information will vary in how sensitive it is: information should not necessarily be viewed as being sensitive, just because it is genetic. Some genetic information is of potential interest not only to the patient, but to others (such as family members, health services or even employers and insurers) and issues of consent and confidentiality should therefore be tailored to the particular situation.
- 2 The following issues should usually be discussed as part of a consent process during clinical consultations where medical and family history information is sought and/or where genetic investigations are initiated:
 - (i) knowledge of a family history of a condition or disease, or genetic test results, has a potential benefit to other family members
 - (ii) the fact that communication of certain aspects of information to family members may therefore be recommended
 - (iii) the means of contacting those at-risk family members where relevant
 - (iv) the fact that a summary of relevant clinical and genetic information will usually be sent to other appropriate health professionals
 - (v) the likely timescales for availability of test results
 - (vi) the possibility of unexpected or incidental findings from genetic testing and how these might be managed
 - (vii) the predictive nature of certain genetic tests (for example, indicating risks many years in the future rather than current risks)
 - (viii) the routine practice of long-term storage of samples for possible future analysis and the patient's preferences regarding further testing if it becomes available
 - (ix) the routine practice of using stored samples from one family member as quality assurance for clinical testing in another family member.
- 3 Individuals seeking genetic advice are often motivated to do so, at least in part, by feelings of altruism and solidarity towards family members. We support the good practice of facilitating appropriate (and where possible anonymised) use of one person's genetic information to benefit the clinical management of family members. It is helpful to document agreement to such sharing in the patient's notes so that their wishes are clear should other family members be seen.
- 4 Where family members are identified as being at risk of developing a genetic condition, attempts should be made to communicate this information, either via the consultand, or where possible and appropriate, via their general practitioner. Such communication can usually be done in a stepwise fashion with first degree relatives being the initial point of contact and subsequent dissemination to appropriate relatives. Details should be provided of how those at risk can seek a genetics referral for themselves.
- 5 Family history and clinical information should be shared with other health professionals (regardless of their geographical location) if the sharing of confidential information is necessary for the purposes of healthcare, and disclosure is between healthcare professionals who share in their duty of confidence.*

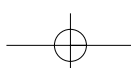
*Data Protection Act 1998, Schedule 3.

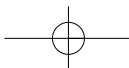
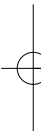
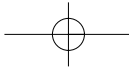


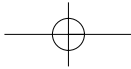
- 6 Where genetic testing involves a person who is unable to consent, a decision about testing should be made considering why testing would be in the patient's best interest. This should be documented in the notes.
- 7 Access to medical information about deceased relatives can be very helpful in determining or refining risks of disease in persons seeking genetic advice. We recommend that the medical record departments of healthcare facilities should help in accessing medical information about a deceased person for such purposes.
- 8 In order to develop better evidence-based care and manage services, we support the good practice of sending relevant information to national or international disease or mutation registers. Patients should be informed in general terms that their information may be used in this way.
- 9 Health professionals within clinical genetic services should take the lead in disseminating good practice regarding the predictive and familial issues which arise in relation to the testing and storage of genetic samples.
- 10 NHS IT strategy should take into account the necessity for long-term storage of diagnostic information for the potential benefit of relatives.

Definitions

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. **Service user** is used in some situations to reflect the fact that not all those who attend a genetic service can legitimately be regarded as **patients**. A **pedigree** is a pictorial summary of a person's family describing relationships and diseases within the family.







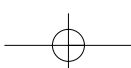
1 Fundamentals of clinical genetic practice

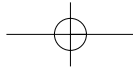
This guidance is intended to aid all health professionals involved at any stage in the diagnosis, referral or testing of patients or their relatives for diseases with a genetic component. As such it is hoped that it will be of assistance to a wide range of health professionals, from the GP who may be the patient's first contact with the health service, through to hospital clinical specialists who discover a genetic condition through investigation of particular signs or symptoms. It may also assist those making decisions about storage of samples for future possible genetic testing and help guide who should access the results of tests on one person when these are potentially beneficial to another.

Historically, genetic tests in the UK have been performed largely by regional genetic services – a series of NHS-funded clinics to which individuals or families can be referred. The genetics professional usually starts by taking a detailed family history, identifying details about the relationship and health of members of three or four generations in that family. This allows conclusions to be drawn about likely inheritance patterns and may help direct genetic testing. It will also identify named individuals who might benefit from the information gleaned through this process. Rather than holding individual records, genetic services often hold family records which group all the relevant genetic information into one set of case notes. Over recent years, genetic technologies have become faster and cheaper, and the evidence basis for diagnostic or predictive information arising from such tests, greater. Genetic testing is now being integrated rapidly into mainstream medical specialties. The issues surrounding consent, confidentiality and sharing of genetic information are therefore now pertinent to most branches of medicine. See Box 1 for some practical aspects of clinical genetic practice.

Whilst genetic information is relevant to an individual, as noted, it may also be relevant to that person's family because much genetic information will be common to both. Indeed, genetic testing may only be requested because of wider knowledge about a condition within a family. The traditional medical approach which focuses on the individual patient to the exclusion of others may be difficult to apply to the use of genetic information. For example, testing one person can reveal information about the chances of a condition occurring in their close relatives and providing the tested person with a right of veto over such risk information in all situations may be legally and ethically unsound. At the same time, respecting confidential information is an important aspect of clinical practice and is vital in securing public trust and confidence in healthcare.

Once a genetic diagnosis has been made, health professionals will usually ask an individual to share their genetic results with the relatives to whom it may be relevant. Discussion of the family history and the pattern of inheritance will facilitate identification of those to whom such information might be relevant. Clinical genetic services often offer help with this process of cascading information, and may provide letters or information leaflets to be passed on. There is considerable variation in practice in such communication, and the degree to which patients directly contact their relatives. Some research suggests that direct contact of relatives by professionals is more effective in identifying those at risk than awaiting intrafamilial communication⁷ whilst others are concerned about invading the privacy of relatives who have not asked for contact.⁸ Notwithstanding ethical objections, there are significant practical hurdles in contacting a range of relatives who may be difficult to identify and locate. For these reasons, current UK genetic practice largely leaves the onus of communication with the individual first diagnosed. Such communication may take place effectively but at times, perhaps where family members are not in close contact, or because of concerns to protect people from distressing information, or through fear of blame or stigmatisation, relatives may remain in ignorance. Health professionals may not know whether information has been passed on and may meet individuals from a family to whom they could provide more accurate information, but concern about breaching the confidentiality of another prevents them from doing so.





The Human Genetics Commission has recommended that health professionals should recognise the value of familial solidarity and altruism by encouraging and facilitating the appropriate sharing of relevant information with relatives.⁹ This approach reflects the fact that most individuals seeking advice from genetic services will do so with at least a partial aim of helping family members. This should be made explicit during consultations and any objections or difficulties that patients have with this approach should be explored and documented. Where such wishes have not been addressed and/or documented, careful consideration will need to be given as to whether the assumption of altruism is appropriate.

Outlining the circumstances in which disclosures might be ethically and legally acceptable and providing guidance to practitioners on how to manage complex cases is therefore one of the aims of this document. An important question will be how these issues can be addressed in the mainstream medical specialties outside clinical genetic services.

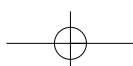
A proportionate response to these issues is required. Genetic ‘information’ covers a wide range of different types of information.¹⁰ On the one hand it will include variations in the genetic code that are simply part of normal human variation and that carry no known health consequences. On the other it includes, for example, the inferences about a person’s genetic code that can be made from their appearance or from a particular family history of a condition. Where genetic information is diagnostic or highly predictive of an illness or relationship,* it might carry greater significance than in other situations. Genetic information should not be viewed as being sensitive and requiring special protection per se.

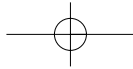
Box 1: Some practical aspects of clinical genetic practice

- *A need to recognise the limitation of approaches which focus on an individual patient:* although a genetic test result may arise from one individual, it may be relevant to other family members. Many clinical genetics services hold family rather than individual case records, so that a clinical consultation with one person may inevitably highlight relevant information for or from another.
- *A need to recognise and encourage expressions of altruism and solidarity:* an individual seeking advice in the context of a clinical genetic service will generally wish their tests to be used for the benefit of other family members.
- *Complexity:* genetic information is complex – both technically and in terms of the implications for family members. Consultations should be tailored to the individual circumstances of the patient and/or family, and multidisciplinary case discussions may be helpful.
- *The distinction between diagnostic and predictive genetic testing:* although some tests will have both diagnostic and predictive elements, there are different implications for understanding and management of genetic conditions depending on whether a test is diagnostic or predictive of ill health in the future, and these differences may need highlighting to other health professionals involved in a patient’s care.
- *The importance of professional judgement:* those seeking to use or share genetic information may be involved in making finely balanced judgements requiring different and sometimes competing interests to be balanced against each other. For example, this might include balancing the rights of confidentiality of an individual against disclosure of that information for the benefit of at-risk relatives.
- *The clinical/research continuum:* the speed of technological change means that there is a greater degree of overlap between clinical and research practices in genetics than in many other specialties. A research laboratory may be utilised to offer a genetic test not yet available on the NHS and the

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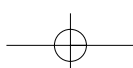
* Genetic testing in a family can reveal misattributed paternity or unsuspected adoption, for example.



**Box 1: Some practical aspects of clinical genetic practice – continued**

risks associated with particular genetic changes may have not yet been thoroughly evaluated. This may have implications for the manner in which results are cascaded out to other family members.

- *A potential wish not to know:* although the emphasis of this guidance is upon the appropriate sharing of samples and information disclosure, sometimes individuals may wish not to know their risks of developing a genetic condition or the detail of any genetic changes. Such wishes need also to be afforded appropriate respect. ■
- ▶ We support the good practice of encouraging appropriate communication of relevant genetic information or results within families and explaining that facilitation of such communication is a key role of clinical genetic services.



2 Issues of confidentiality in clinical genetic practice

One of the fundamental ethical obligations owed by all health professionals to their patients is that of confidentiality. This duty is also recognised in law, but the health professional's ethical and legal duty of confidentiality is not absolute since it is balanced by a duty of disclosure under certain circumstances, eg under compulsion of law and/or in the public interest. An assessment of what might be a public-interest reason to break a confidence will often involve complex decisions and finely balanced judgements.

The tension between confidentiality and disclosure is heightened in clinical genetics because of the shared nature of genetic information. On the one hand, genetic or genomic information is medical information that should generally be kept confidential. On the other hand, a result in one person may identify others who carry the same genetic trait and would benefit from regular surveillance and/or prophylactic options. If such information is relayed to other family members without appropriate consent, the trust between the patient and health professional may be undermined. Conversely, if the information is withheld from other family members, their interests may be placed in jeopardy.

Health professionals involved in such cases may feel a three-way tension between respecting an individual's confidence, communicating potentially beneficial information to other family members, and respecting the possibility that those family members may not want to receive such information.

Ideally, the confidentiality/disclosure dilemma is pre-empted by ensuring that a patient consents to the disclosure of any relevant and potentially beneficial information to other family members, but sometimes gaining such consent can breach the confidence of another and at other times the original patient has been seen many years previously, or by a different health professional and it is not clear what consent has been given.

Where such consent has not been explicitly sought, practitioners may be left uncertain as to what they can disclose when they see other family members.

It is important to recognise (a) that most people attending a clinical genetic service do so with at least a partial aim of also providing relevant information to their relatives, and (b) relevant information can sometimes be transmitted to relatives in an anonymised way, thus not breaching any confidence.

The assumption that confidentiality is always paramount is as inappropriate as the assumption that disclosure is always permissible, and the decision will need to be tailored to the individual circumstances of the case.

3 Issues of consent in clinical genetic practice

The process of seeking consent ensures that a person understands the nature and purpose of a procedure and the practice has its origins in the ethical principle that a person has a right to self-determination. In legal terms, consent is valid only if three questions are satisfied:

- Is the patient competent?
- Was the person giving consent appropriately informed beforehand?
- Was the consent voluntarily given?

Consent is valid only if the implications of a procedure are disclosed and understood, but it is the complexity of these implications that poses a particular challenge in clinical genetic practice. Can such consent ever be fully informed? What constitutes sufficient information? How much can a patient be expected to weigh up the emotional consequences of a particular result, to the extent that they can be said to fully understand its implications? Genetic information can be upsetting to individuals and may have collateral consequences, for example, fear of discrimination or stigmatisation. In addition, results of genetic tests are frequently of significance to the patient's relatives, but the patient may not want others to know about their genetic make-up.

Given these complexities, and the fact that competence is decision-specific, what is the bar for determining competence? Is it realistic to expect patients to weigh up all these issues before consenting to genetic testing? If insufficient consideration is given to such implications, then the validity of their consent becomes questionable. This tightrope must be carefully navigated based on the individual circumstances of the case.

In addition to consent for the test itself, it may be appropriate to include discussion of the following as part of the consent process, particularly since these additional aspects are often not routine for other types of tests or procedures:

- consent to disclosure of relevant genetic information to relatives
- consent to research
- consent to the storage and future use of the sample, and the information derived from it.

We suggest that a form such as that suggested in Appendix C2 may be a useful record of the fact that such discussions have taken place.*

There are some issues which are particularly relevant to the clinical practice of genetics, and which the Joint Committee believes need special consideration and attention during the consent process. They are summarised in Box 2.

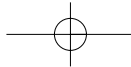
Box 2: Issues to be discussed during the consent process

The following issues should be discussed:

- **knowledge of a family history of a condition or disease, or genetic test results, has a potential benefit to other family members**
- **the fact that communication of certain aspects of information to family members may therefore be recommended**
- **the means of contacting those at-risk family members where relevant**

continued

* This form has been developed in response to concerns that the standard consent form based upon a template from the Department of Health, and used by many NHS trusts around the UK, does not adequately cover familial and genetic aspects.



Box 2: Issues to be discussed during the consent process – *continued*

- the fact that a summary of relevant clinical and genetic information will usually be sent to other appropriate health professionals
- the likely timescales for availability of test results
- the possibility of unexpected or incidental findings from genetic testing and how these might be managed
- the predictive nature of certain genetic tests (for example, indicating risks many years in the future rather than current risks)
- the routine practice of long-term storage of samples for possible future analysis and the patient's preferences regarding further testing if it becomes available
- the routine practice of using stored samples from one family member as quality assurance for clinical testing in another family member. ■

- We recommend that these points are highlighted in a general leaflet for genetic service users, and included as appropriate in pre- and post-clinic summary leaflets and/or letters.

3.1 Enabling clinicians from a range of backgrounds to take consent

In order for the issues summarised in Box 2 to be adequately addressed, those taking consent need to be suitably trained and qualified. Existing guidelines from the GMC and Department of Health^{4,5,11} place the onus for gaining consent upon the clinician requesting the investigation. The request for consent may be delegated, but the clinician is responsible for ensuring that any substitute is suitably trained and qualified, and that they can convey the reason for a request for information or investigation, and any risks involved.

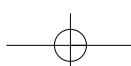
Clinical scenario 1

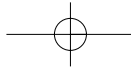
Elizabeth (aged 35) wants to know whether she is at increased risk of breast cancer since she has a strong family history of the disease. Dr Jones therefore suggests BRCA testing in her affected relative, Rose, since if positive, this would improve the predictive value of testing Elizabeth. Elizabeth provides the name and contact details of Rose, who is currently an inpatient in a hospital in another city. As Dr Jones wants to delegate the taking of the sample to another clinician, she should provide enough information to that clinician so that they may discuss the possible implications of the test for Rose, as well as for Rose's relatives. ■

Since the onus to obtain consent lies with the clinician, it is generally appropriate for laboratory staff to presume that a clinician who has sent a sample has obtained valid consent. The laboratory is not required to confirm and document this.

For certain tests, the UK Genetic Testing Network (UKGTN) has published additional criteria which need to be satisfied in order for a test to proceed.* In some circumstances where genetic laboratories receive samples from medical specialties who may be less familiar with the potential predictive and familial aspects of testing, it may be reasonable for laboratories to query whether the requirements of the UKGTN are being met.

* See www.ukgt.nhs.uk/gtn/Information/Services/Testing_Criteria





Clinical scenario 2

A genetics laboratory receives a request to perform Huntington's gene testing 'for reassurance' in 22-year-old Katy who has a family history of dementia. The laboratory has no record of a Huntington's gene test in family members, nor any indication that Katy has been counselled on the pros and cons of predictive testing. Since the majority of individuals who come forward for predictive testing for Huntington's disease decide not to have a test once they have had an opportunity to consider the implications in detail,¹³ it is appropriate for the laboratory to check with the referring clinician that specific consent has been given and to suggest that a clinical genetic referral may be useful. ■

Box 3: Laboratory request forms

Some laboratories have included specific statements on their laboratory request forms to prompt consideration of familial aspects during consent to blood tests taken outside genetic services.

For example:

In submitting this sample the clinician confirms that consent has been obtained for genetic testing; that the implications of such testing have been discussed; that routine long-term storage of the DNA has been discussed and that agreement has been obtained 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 deemed appropriate for the communication of risk to others.

Although this may clarify that consent has been obtained, it may also generate uncertainty if the relevant box has not been completed. As electronic ordering of tests becomes more routine, such prompts may be difficult to include in a meaningful way. ■

- ▶ **The Joint Committee recommends that health professionals in clinical genetic services should take the lead in disseminating good practice regarding the predictive and familial issues which arise in relation to the testing and storage of genetic samples.**

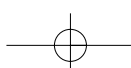
3.2 Ensuring that colleagues involved in the care of the patient are kept informed

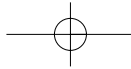
It is important that the primary healthcare team and other relevant health professionals are kept informed about a genetic diagnosis, the implications for management and the risk implications for others. However, we acknowledge that there are rare circumstances where a patient may not want genetic test results communicated to other health professionals. The reasons why the usual process is not followed should be carefully documented in the clinical notes.

Clinical scenario 3

Mary (aged 38) has a family history of Huntington's disease and a predictive genetic test shows she has inherited the familial mutation. Although Mary realises she will need to inform her GP at some point, at present she wishes this result to be known by as few people as possible. She is worried about being stigmatised as well as possible adverse insurance consequences since she is about to apply for a new mortgage.* Mary is well and has no signs or symptoms of the condition. The clinician documents Mary's decision in the genetics notes and plans to review this decision with Mary at a later date. ■

* The Association of British Insurers (ABI) and UK government have a long-standing concordat and moratorium on the use of predictive genetic tests recently extended to 2017. The moratorium applies to life assurance and critical illness insurance for applications less than £500,000 and £300,000 respectively.





3.3 Consent for continuing and further investigations

The Department of Health has emphasised that consent should be viewed as a process and not simply a one-off event. Consent for genetic testing can be assumed to remain valid if a further investigation remains within the scope of the original consent and if the patient has been informed that further investigation might take place.

Molecular, biochemical and cytogenetic laboratory investigations are often technologically demanding and hence by their nature prolonged. Where appropriate, it is good practice to point out to patients that results may take some time, or that further tests could be done in the future when more sensitive technologies are adopted by the laboratory.

Clinical scenario 4

Alan has consented to genetic testing to determine whether a gene mutation explains his own and his family history of bowel cancer. Initial mismatch repair gene testing reveals no mutations. Six months later the laboratory develops a new technique to look at promoter mutations, which a recent research publication has determined to be a possible cause of Alan's type of family history. Since Alan has consented to the genetic investigation of the cause of his family history, and is aware that such testing may take some time, it is assumed that his consent covers this new test.

A discussion about possible time intervals should be had during the initial consent process with Alan. If many years have passed then it may be less appropriate to assume that his consent still holds. ■

Good liaison between laboratory staff and clinical staff is required so that clinicians can consider whether further testing falls within the scope of original consent, or whether further consent ought to be obtained. We suggest one way to determine preferences is to ask at the time of initial consent (see Box 4 and Appendix C2).

Box 4: Future tests on stored samples

A possible summary on the consent form is suggested below and on the model consent form in Appendix C2:

I would like to be contacted before further relevant tests are done on the stored sample if new tests become available.

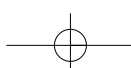
OR

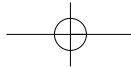
I am happy for further diagnostic tests on the stored sample to be undertaken without being contacted. ■

Rapid advances in genetics mean that novel or more comprehensive investigations may have the potential to reveal new or more accurate diagnoses for patients or families who have been seen in the past. Although it is important to mention the possibility of future contact, a genetic service may not be able to give an assurance that this will happen 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.

3.4 Consent for samples taken for storage only

Samples may be taken for storage for future analysis (usually as extracted DNA rather than tissue) should a test become available. Although such practice is declining as genetic tests are becoming more





widely available, where it still takes place careful consideration should be given to if and how the patient will be re-contacted when testing becomes available.

3.5 Consent for samples tested in research laboratories

Many aspects of clinical and laboratory genetics remain at the interface between service and research. For some genes there may not yet be a clinical testing service. Using research laboratories as an aid to diagnosis is acceptable as long as the patient understands that such results may not have the same standards of quality assurance. It is considered best practice to confirm any potentially informative results in an NHS laboratory if possible. No local research governance procedures need to be implemented in such cases since the research laboratories are used as an aid to clinical diagnosis.

On other occasions patients may be told about research studies that are likely to yield general information about their inherited disease rather than provide specific information relating to their own case, and that participating in the research by providing a sample is unlikely to directly aid their diagnosis or management. In such cases, the research ethics committee which authorised the research will have stipulated the form of consent which is required and other regulation and governance issues may apply.*

3.6 Covering the possibility of death in the consent process

Sometimes the donor of a sample will have died before a genetic diagnosis is made. Whilst death may be anticipated in the near future in some consultations, it will not be in others and routinely mentioning this during a consent process may be inappropriate. Ensuring that consent encompasses a broadly worded statement of intention to benefit family members may avoid potential uncertainties about who can be informed about a deceased person's results.

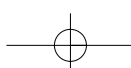
Mentioning the possibility of unanticipated death may be insensitive or not appropriate during many consent processes.

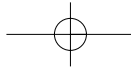
Clinical scenario 5

A UK genetic service had developed a consent form which asked for names and contact details of relatives to be contacted if that individual were to die or become incapacitated. Whilst such practice may be very helpful in indicating consent for using results for the benefit of other family members, unanticipated problems arose when relatives, not originally named on the form, were identified as being potentially at risk through genetic tests on the sample after the donor's death.

We consider that a broad consent to allow communication of potentially beneficial relevant information to any relative is therefore more appropriate. ■

* We note that there are advantages and disadvantages to both routes. Using a research laboratory to perform clinical testing (albeit with the subsequent step of confirmation in an NHS laboratory) is likely to be relatively quick. In contrast, the bureaucracy involved in gaining local ethical and research and development approval is likely to significantly increase the time before the sample can be studied, but this approach means that such research activity within a clinical setting is appropriately recognised (eg in comprehensive local research network figures).





3.7 Incidental or unexpected information

Some genetic tests have the potential to reveal incidental or unexpected information. Where there is a significant potential for incidental findings to arise, that possibility should be included in the initial consent process. This may involve sensitive discussions, for example, indicating to individuals that their results may show that genetic and social relationships within a family are not what they had thought.*

The potential for discovering clinically relevant incidental information is increasing rapidly as genetic technologies allow faster and cheaper testing of large sections of a person's genome rather than targeted analyses of specific genes. It is anticipated that the speed and cost of sequencing an entire genome will be the same as current analysis of just one gene within the next few years. Technologies such as array comparative genome hybridisation (aCGH) can already identify inherited abnormalities not expected on the basis of clinical or family history information. Whilst many medical tests have the possibility of discovering clinically significant incidental findings, and this is therefore not new to genetic practice, the likelihood will increase dramatically over the next few years. A major challenge is that many of these incidental findings will be predictive of future rather than current disease risks, and may also be relevant to other family members. More research is needed as to how this can best be addressed in both consent and disclosure practices, as well as how to address the implications for other family members.

Clinical scenario 6

Chloe (aged 5) has mild developmental delay and subtle dysmorphic features. She is investigated with array CGH and this reveals a large deletion which is likely to explain her clinical signs and symptoms. The deletion encompasses a known tumour suppressor gene which indicates a high likelihood of adult onset cancer. Further investigations reveal that her father (aged 28) has also got this deletion. There are several surveillance recommendations for adults who have abnormalities in this tumour suppressor gene. Whilst Chloe's immediate management is not affected by this discovery, her father's is.

Although it may be difficult to be specific about the type of information that might be discovered, the possibility of incidental findings should be raised in the consent process. ■

3.8 Use of samples as laboratory clinical controls, quality assurance, audit, education and training

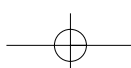
A sample may be stored and/or used for quality assurance of laboratory tests for other patients. The Department of Health's model consent policy suggests that tissue samples may be used for quality assurance purposes without specific patient consent provided that NHS bodies have an active policy of informing patients of such use and could include the potential for samples to be used for education and training.¹¹ A sample may also be used for clinical audit, education and training without consent. Consent is also not required for such purposes under the provisions of the Human Tissue Act 2004 for samples from living patients.

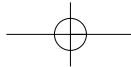
Clinical scenario 7

Karen has requested predictive testing for BRCA2, since a mutation has been found in her paternal aunt, Millie, in another part of the country. Karen has been given a copy of the report by a relative, so the exact details of the mutation are known, but the laboratory would like a small amount of DNA from Millie to act as a positive control in her predictive test (in case the family has a private

continued

* Including the potential discovery that paternity has been misattributed.





Clinical scenario 7 – continued

polymorphism that affects the accuracy of the test). Millie had been tested several years ago and the laboratory requests reassurance from Millie's clinician that she provided consent for use of her sample in this way. Millie's clinician does not recall the consultation and does not think it appropriate to contact her about this, because such testing is of no clinical significance to Millie.

Using Millie's stored DNA as quality assurance for another family member does not require further consent; it will not reveal any new information about Millie's sample and will not breach her confidence. ■

Box 5: Quality assurance

An example of wording to include in consent or information leaflets might be:

After testing, part of your sample might be used to monitor the quality of laboratory results. ■

3.9 Counselling as a part of the consent procedure for a genetic test

The term counselling has many different definitions and genetic service users may have differing interpretations of the term. Some will understand it to mean a psychotherapeutic interaction whilst others will see it more as an opportunity for relevant information to be exchanged. The latter is the more usual meaning of the term in genetic counselling.

In some cases, for example, predictive testing of adult onset conditions such as Huntington's disease where there are no effective interventions, consensus would hold that a considered decision over time is important before genetic testing is performed, not least because most people given this opportunity decide not to have a genetic test.¹³

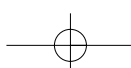
However, in other cases, it will not be appropriate to insist that a patient has to have several appointments before a genetic test is performed. The key is to ensure that patients have the relevant information to make an informed choice.

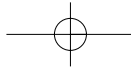
The Joint Committee agrees with the Genetic Alliance UK (formerly the Genetic Interest Group (GIG)) that the consent process should be tailored to the patient and the condition being considered.¹⁴ This is in keeping with the GMC's recommendation that when providing information, health professionals must do their best to take account of individual patient circumstances by finding out about patients' needs, wishes and priorities.⁴

Whilst good practice is always to encourage the individual to consider the issues involved, an insistence on imparting unwanted information is inappropriate.¹⁵ For instance, some family members may be willing to give a blood sample for the benefit of their relatives, but not wish to receive counselling or detailed information themselves. This is legitimate providing that those individuals understand the consequences of their decision not to receive information. A note of these wishes should be made in the medical records.

3.10 How should consent be recorded?

GMC guidelines confirm that patients may indicate their consent either orally or in writing. A record in a patient's notes of the issues discussed and the agreement reached, signed by the clinician, is adequate for



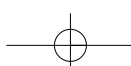


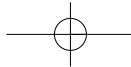
most purposes. The extent of the written record will vary according to the nature and implications of the genetic information or test results to be generated. GMC advice states that written consent is important if there are significant consequences for the patient's employment, social or personal life or where providing clinical care is not the primary purpose of the test, but the judgement of what constitutes significant should be made on a case by case basis.⁴ We note that many clinical genetics professionals do not routinely request written consent for genetic investigations* but suggest that forms such as those in Appendix C2 can be a useful record of discussions. In particular such documentation can clarify the level of consent to familial sharing should a different clinician see a relative of the person tested.

3.11 Consent to clinical photography and video recording

As well as being part of the medical record (as a way of delineating and understanding the natural history of dysmorphic syndromes), clinical photographs and video recordings are important for teaching, audit and research. The purpose and possible future use of the photographs and film must be clearly explained to the person (or parent), as part of the consent process. Where possible, photographs/film should be anonymised. A verbal explanation may suffice and the patient's consent should be recorded in their medical notes. Consent in writing should be sought if the images are to be used for secondary purposes. A specimen consent form is provided in Appendix C3. (See GMC's *Making and using visual and audio recordings of patients* (April 2011).¹⁶)

* The Great Debate: British Society of Human Genetics annual conference 2003.





4 Consent by others

Adults with capacity should provide consent for any investigation they wish to have. In this context, ‘capacity’ means that the person is able to make a decision about a particular matter and be able to understand relevant information, retain that information, use and weigh it up as part of the process of making and communicating their decision. Where individuals lack capacity to decide, a decision to proceed is made on the basis of what will be in their best interests (or benefit) and a proxy decision-maker may be needed.

4.1 Children

Young people aged 16 or 17 are presumed to have capacity to consent. Under the age of 16, the presumption is reversed, but rebuttable if an assessment shows the child does have the capacity to make a specific decision (‘Gillick competence’).*

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 British Society of Human Genetics report, *Genetic testing of children* (2010), which highlights practical and ethical issues in more detail, and particularly that the child’s best interests should be the main driver.¹⁷ Another useful resource is the GMC publication *0–18 years: guidance for all doctors*.¹⁸

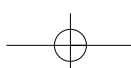
Where treatment or surveillance will be instituted or altered, genetic testing of children is likely to be appropriate. However, where the test will not be of medical benefit to the child for some time, the recommendations suggest a presumption of delay until a child is old enough to choose for him or herself, unless there are compelling reasons to test earlier.

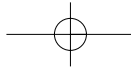
In some cases, genetic testing is undertaken in childhood to diagnose a condition requiring immediate intervention or treatment (for example, the detection of cystic fibrosis (CF) as part of a neonatal screening programme). Those found not to have CF, or other recessive conditions, may however be ‘carriers’ of the condition. The consequences of detecting that carrier status may hold few, if any, immediate clinical implications for that child. We consider this an incidental finding of a diagnostic test and therefore distinct from the request for carrier status of a healthy child to determine future reproductive risks.

Clinical scenario 8

Richard and Jane have recently had a baby who was tested through a newborn screening programme for CF and found to be a carrier. The couple now request carrier testing of their two older children aged six and eight, who are healthy. Since newborn screening aims to identify affected children, and discovery of carrier status is an incidental finding in a small proportion of families, this should be explained to the parents. Testing their older children has no medical benefit and is not indicated until they are of reproductive age. ■

* *Gillick v West Norfolk and Wisbech Area Health Authority* [1985] 3 All ER 402 (HL).





Clinical scenario 9

Chris was diagnosed with an inherited eye tumour syndrome, retinoblastoma (RB), as a child and a mutation in the RB gene was (eventually) found. He and his partner Mandy have recently had a daughter, Claudia, who has a 50% chance to have inherited RB from him. The couple do not want Claudia to have a genetic test as they worry this will stigmatise her and increase the chances of discrimination against her. However, because Claudia has such a high risk, she will need regular eye examinations for the first few years of her life, and these will need to be done under general anaesthetic – with its attendant risks and costs. A genetic test would have a 50% chance of showing she had not inherited the RB gene mutation and that such examinations would not be necessary. A strong argument can be made therefore that it is in Claudia's best interests to have a genetic test despite her parents' anxiety about this. ■

4.2 Adults lacking capacity

In the case of an adult lacking capacity to consent to a specific test, the test may still be undertaken if it is considered to be in their best interests, for instance, to make a diagnosis. The Mental Capacity Act 2005 (MCA)² (see sections 4–6) establishes the principles for making a best interests judgement and requires the caregiver to question whether the same outcome can be as effectively achieved in a way that is less restrictive of the person's rights or freedom of action. In carrying out genetic testing as part of treatment or care, there is also a requirement to consult (if practicable and appropriate) certain named individuals including a carer, or person interested in the person's welfare whether in an official or unofficial capacity.

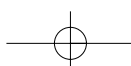
4.2.1 What are 'best interests'?

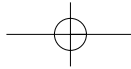
The House of Lords has suggested that action taken 'to preserve the life, health or well-being' of a patient will usually be in their best interests and subsequent court judgements have emphasised that a patient's best interests include broad welfare considerations. These can include indirect benefit: 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 MCA includes a list of factors which must be taken into account in determining 'best interests' which include the requirement to consult and take into account the person's past and present wishes and feelings. Views must also be sought (if practicable and appropriate) from the range of individuals described above.

Clinical scenario 10

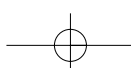
Michael (aged 35) has learning difficulties and lives in sheltered accommodation. His sister has recently been found to have attenuated familial polyposis and a mutation has been identified. Michael says he does not mind giving a blood sample, but the health professionals involved have assessed that he does not have the capacity to consent to a genetic test. A multiprofessional team meeting considers that it is in Michael's best interests to have a genetic test; if he does not have the mutation he can be spared future screening interventions such as colonoscopies and surgery. ■

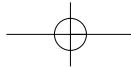
If a direct medical benefit is not likely (for example, taking part in family studies), there should be careful consideration whether a person lacking capacity should provide a sample. However, the Genetic Alliance UK has argued that it should not be presumed that an adult with incapacity would be less altruistic than a competent adult in wishing to assist other family members in genetic investigations.¹⁴ Department of Health guidelines¹¹ also note that care should be taken not to underestimate the capacity of a patient with a learning difficulty or cognitive decline.



**Clinical scenario 11**

Delia (aged 44) has severe learning difficulties and lives in a care home. She was diagnosed with a dilated cardiomyopathy (DCM) in her 30s and is on a cocktail of medication. Her sister Daphne recently attended the genetic service as she was concerned about her own risks. There is a strong family history of cardiac-related deaths at young ages and her father also had DCM based on his post-mortem details. In order to facilitate accurate risk assessment in Daphne, a diagnostic test in a living affected relative is required. Without this, the negative predictive value of any test in Daphne would be poor. Testing Delia might establish a genetic diagnosis in her, but this is unlikely to affect her management in any way. Delia is deemed not to have the capacity to consent to such testing. A narrow view of best interests might conclude that testing Delia would not serve these, since her management would not be altered. A wider view would acknowledge that Delia's interests may be served by her helping her sister through a genetic test. The discomfort of the test and any distress it might cause would have to be factored in. It should not be assumed that just because Delia does not have the capacity to consent, that testing for the benefit of her sister is not permitted, but multidisciplinary team discussions are recommended in such circumstances.¹⁴ ■





5 The use of familial information in clinical genetic practice

5.1 The family history as an aid to genetic diagnosis

One of the first things a genetics professional does in their initial consultation is to take a detailed family history of a disease, condition or symptom. A three- or four-generation history identifying names, ages, deaths, illnesses and so forth is taken and recorded in the form of a pedigree, supported by information about the patient and known family members.

Such details are usually supplied by the first person to be seen in that family and the accuracy of the information is therefore dependent on the extent of communication within the family. The information may subsequently be amended by details provided by other family members or by hospital records of others. The composite or 'master' pedigree is kept in a family folder with identifying details of several family members, some of whom may not be aware of this process. Family records are held in confidence by regional genetics centres and it is good practice to explain to individuals that their records are kept as family records and that the information gleaned from a pedigree may be used to advise other family members of their risks.*

Clinical scenario 12

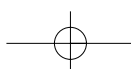
John has a family history of sudden cardiac death and investigations show that he has Long QT syndrome and a pathogenic mutation in the LQT1 gene. Two years later John's cousin, Gavin, is referred to the genetic service because of a family history of death thought to be caused by epilepsy. Gavin provides details which show that his family is related to John, and that what was thought to be a family history of epilepsy is in fact one of sudden cardiac death due to Long QT syndrome.

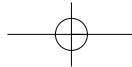
At the time of John's consultation it was explained that investigations in his branch of the family might be helpful to other relatives. John made no objection to this use of information provided by him and his agreement was documented in his medical notes. The genetic service was able to advise Gavin that information from another branch of the family suggested the cause of death in his relatives was cardiac in origin and that a genetic test may clarify Gavin's chances of developing Long QT syndrome. Of course, the difficulty with this general approach is that in some families, depending on their structure, it will be very clear in which individual a result has been found. ■

Clinical scenario 13

David has been found to be a carrier of the BRCA1 gene. This is unlikely to carry any health implications for him, but he had the test to see whether his daughters might be at increased risk in the future. They are only two and five years old at present, and testing at this age is not generally regarded as appropriate for this adult onset condition. David's GP is uncertain as to how this information can be recorded appropriately. She considers that David's result should only be documented in his records because she is worried that the daughters may be somehow disadvantaged if the information were to be added in their records. After consultation with the local genetic service, she decides to note the following in the children's records: 'History of inherited breast/ovarian cancer in paternal family. Genetic testing to determine risks may be appropriate in adulthood.' ■

* For a detailed description of clinical genetics pathway, see *Do once and share: clinical genetics*.
[www.bshg.org.uk/documents/official_docs/DOAS_final_printed_report\[1\].pdf](http://www.bshg.org.uk/documents/official_docs/DOAS_final_printed_report[1].pdf)





5.2 Use of medical records to confirm diagnosis

A family history of a condition may be unclear or uncertain and health professionals may attempt to confirm key diagnoses through hospital and other records in order to more accurately ascertain risks and direct genetic testing. The disclosure of this secondary information will usually require the consent of the person to whom it relates. Consent forms such as the example in Appendix C1 may be sent to relatives from whom confirmation is sought via the person attending the clinic or through an address supplied by them.

Health professionals should usually respect the confidences of patients who have subsequently died, but since it is no longer possible to gain their consent to access their records, the GMC has recommended that in the absence of specific instructions,* disclosure may be justified if the potential benefits to relatives outweigh the risks from breaching confidence.¹⁹ The statutory basis for obtaining access to health records in hospitals, general practices and other healthcare settings of deceased patients is governed by the Access to Health Records Act 1990, rather than the Data Protection Act, and is at the discretion of the person holding them, unless the deceased refused permission.²⁰

It has come to the Joint Committee's attention that some record holders allow access to such records only with consent from the spouse (or 'closest' relative), but the information contained in the records may be relevant to the medical management of a blood relative, a possibility not considered in the Act.

Clinical scenario 14

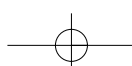
George is seeking advice about his risk of cancer. Several relatives have died of cancer at a young age. His half-brother and cousin both died in their 30s and he remembers being told about them having had polyps. The genetics department is refused access to histology information from these two relatives because they have 'not demonstrated that written consent to disclosing these results had been obtained from the deceased's next of kin.'

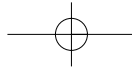
Whilst such a reaction from a pathology department may be understandable, the consent from the next of kin is not required to authorise the disclosure of confidential information. Seeking such consent could be unethical because:

- (a) it could breach George's confidentiality (if the reason for the request is inferred by the 'next of kin')
- (b) seeking signatures may be based upon a misunderstanding of what the law requires. In such circumstances, the public interest in disclosure may justify breaching the confidentiality of George's deceased relatives in the interests of preventing harm to living relatives
- (c) the signatures alone are not verifiable by the pathology departments and therefore have limited usefulness, creating an additional administrative burden.²¹ ■

- ▶ **Access to medical information about deceased relatives can be very helpful in determining or refining risks of disease in persons seeking genetic advice. We recommend that the medical records department of healthcare facilities facilitate access to medical information about a deceased person for such purposes. This is legitimate practice under the Access to Health Records Act 1990, is consistent with disclosures made by cancer registries and should not require the signature of any relative to do so.**

* and most people will not have left specific instructions regarding access of their family members to their medical records.





5.2.1 Destruction of medical records

Valuable information about specific diagnoses (for instance, confirmation of the types of cancer or cardiac disease occurring in families) may be lost when paper records are destroyed.

- ▶ **The Joint Committee recommends that the NHS IT strategy takes into account the necessity for long-term storage of such diagnostic information for the potential benefit of relatives.**

5.3 The use of existing genetic information to facilitate accurate genetic testing

For some genetic disorders, a detailed family history is required to make or confirm a precise diagnosis or to determine the most likely mode of inheritance. The information available to the clinician is usually based on what a person has observed or been told by their family members. Some information may be known by many, including people outside the family, or be evident from a person's appearance. Other information will be known by a limited number of people. As suggested above, a master pedigree may contain information from a number of sources, not all of which should necessarily be regarded as confidential information to which the common law of confidence would apply.

Clinical scenario 15

Dr John has taken a detailed family history of a particular (dominantly inherited) neurological condition. He has noted the names, dates of birth (and death) of approximately 20 relatives and whether or not they have the condition. He is worried that he should not record such identifying information without the consent of the individuals concerned because they are not his patients.

The information stored at this stage is 'hearsay' information. It is based on that provided by one person about others. In this sense it is not confidential information between the doctor and the individuals concerned. However, genetic records are kept in confidence, just like any other medical records and Dr John should make sure details are not inadvertently released to others who have no need to know the information. ■

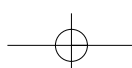
If information is recorded about one family member in another person's records, the Data Protection Act³ gives specific rights for that relative to obtain copies of the information that is held about them (provided that disclosure does not breach the confidentiality of another) if they request this. Although there may be a requirement to release such information upon request, there is no requirement to seek specific consent from each relative in a family history before that information can be shared for medical purposes.

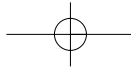
Clinical scenario 16

Stephanie, who has unexpectedly found herself to be eight weeks pregnant, attends the genetics clinic to enquire about prenatal testing for achondroplasia. Her (former) partner Darren has the condition and she is aware that any child the couple conceive together has a 50% chance of also inheriting achondroplasia. She knows that Darren was tested by another genetics centre some years ago. She does not, however, want to ask him for permission to access this result as she does not want him to know about the pregnancy.

Although the diagnosis in this case is highly likely to be achondroplasia (because of the level of detail Stephanie can provide), knowing the exact mutation in the gene and comparing the fetal

continued





Clinical scenario 16 – continued

sample with that of a stored sample of Darren's DNA, will provide a more accurate fetal test than without this information.

The laboratory should be able to confirm the exact mutation details and provide a positive control (as quality assurance for the fetal test) without Darren's consent on the basis that this is not a breach of Darren's confidence and that when Darren consented to testing he was aware, and did not object to, his test results being used to benefit appropriate medical care of others. ■

When a person requests a genetic testing it is often because they already know that a family member has been tested. Confirmation of the exact mutation in that relative allows targeted and more accurate testing of the consultand so the communication of result details between health services can be important. It is appropriate for good clinical care that technical information and laboratory reports should be shared between the different laboratories undertaking the testing, and be available to clinical staff in genetics units. It is not appropriate to require new consent from the sample donor every time such activities take place, but as discussed in previous sections, this possibility should be covered in the initial consent process.

The Joint Committee considers it inappropriate to circumvent such communication, for example, by performing a general screen of a consultand's sample. There are two main reasons for this:

- (1) If no anomaly were to be identified, this could be because the person had not inherited the disorder, or it could be due to differences in the sensitivities of the techniques employed by the laboratories testing each of the samples (for example, use of different primers).
- (2) Targeted testing is usually more cost effective than generalised screening, although this situation may change over the next few years.

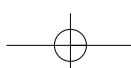
Clinical scenario 17

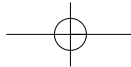
Clare (aged 22) has been referred to the genetics department because of a recent diagnosis of Duchenne muscular dystrophy (DMD) in her nephew. Her sister (Natalie) is found to be a carrier for the condition and has told Clare to 'get herself tested' for DMD before she has children. Clare's genetic service writes to the laboratory where Natalie and her son were tested (in another part of the country) and ask for details of the exact mutation. The laboratory refuses to release this information until they have evidence that Natalie has consented to this.

Whilst it is possible to re-contact Natalie and obtain her signature, good genetic practice would ensure that such uses of Natalie's sample and genetic information would have been discussed with her at the time of testing, her agreement documented, and that any dissent on her part would have been both recorded and notified to the laboratory. If this is the case, then the laboratory would not need to request further evidence of consent and the delays that would have been imposed by the additional bureaucracy can be avoided. By speaking to her sister, Natalie has clearly indicated that she wishes her sister to be aware of her condition, and it would seem disproportionate to expect Natalie to explicitly consent to the details of her mutation to be released. ■

5.4 Retention and storage of pathology and genetic samples in laboratories

The Royal College of Pathologists has published guidance setting out optimal periods of storage and retention of all pathology samples, ie diagnostic specimens are retained for 30 years if needed for 'family studies' and indefinitely if used in research.²² It has been common practice amongst DNA laboratories in the UK to regard isolated diagnostic DNA samples as being relevant to 'family studies' and for that





reason many laboratories routinely provide long-term storage for these samples, which can be considered an important part of a medical record.

- ▶ **The Joint Committee has become aware of such storage facilities requiring payment for release of samples for further studies. We consider such charges contrary to NHS standing financial instructions and recommend access is permitted without charge where clinically appropriate.**

5.5 National or international disease or mutation registers

In order to develop better evidence-based care and manage services, the Joint Committee supports the good practice of sending relevant information to national or international disease or mutation registers. Such registers should be anonymised in the sense that patient names or addresses should not be used, but with certain types of genetic or genomic information, it will be difficult to completely anonymise such entries. Patients should be informed in general terms that their information may be used in this way and that this is an important part of service evaluation and gathering evidence for their future healthcare.

5.5.1 Cancer registries

Cancer registries are valuable sources of information and can help to confirm a family history of disease. If the person is alive, consent should be sought and documented, before the release of information is requested. UK cancer registries will release information about deceased relatives to clinical genetics units under the mutual understanding of respect for confidentiality.

- ▶ **The Joint Committee strongly supports this practice for good clinical care.**

5.6 Disclosure of information to others

5.6.1 Where the basis of consent for releasing information is unclear

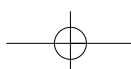
Sometimes the scope of consent sought in the past is not clear. This may be because the information which patients and clinicians discussed during the consent process is out of date, following advances in genetic knowledge or technologies. Individuals may have been seen many years previously by different clinicians.

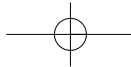
Health professionals will need to explore carefully whether any consent exists, whether it is sufficient, or whether a new consent should be sought. Such decisions will depend on the clinical situation and on the sensitivity of the information potentially released by the proposed action. Obtaining new consent may not be possible, if contact with an individual has been lost, or may not be appropriate, if in doing so the confidentiality of another person may be compromised.

Clinical scenario 18

Caroline is seeking prenatal diagnosis for an X-linked condition known to be present in her family. She does not want anyone to know about her pregnancy until the test results are available. In order to provide her with an accurate genetic test, the health professional will need to access the mutation details of an affected relative of Caroline. Caroline does not know which of her relatives

continued





Clinical scenario 18 – continued

have been tested, and they do not know that she is interested in a predictive test. Asking their consent to disclose their result to a relative may compromise Caroline's privacy. The public interest in keeping Caroline's pregnancy confidential (and in maintaining trust between patient and physician) may be more important than the requirement for consent to the disclosure of a test result from a family member, if seeking consent from that family member may reveal the pregnancy and thus breach Caroline's confidentiality. However, there is also a clear public interest in maintaining a confidential health service in which individuals are not deterred from testing by concerns that their confidential information might be disclosed. Ideally, possible use of a result for the benefit of a relative would have been discussed and documented at the time of testing, but this may not be clear to the clinicians who are seeing Caroline.

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 test result from a relative, than would result to another member through the use of their result without confirmation that consent had been granted. ■

- ▶ **The Joint Committee recommends that as far as possible the information from the sample remains confidential but is used to inform the appropriate care of the consultant.**

It may be sufficient to acknowledge that a familial mutation exists and provide its details without naming the person in whom it was found. If such a statement inevitably points to one particular person, then the harms and benefits of proceeding without further consent need to be carefully assessed.

Clinical scenario 19

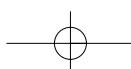
Helen is referred to a genetic service by her general practitioner because she is concerned about her family history of breast cancer. She is not aware of any family members who have sought genetic advice or had genetic testing. However, through details of her family history, it is apparent to genetics professionals that a pathogenic mutation in BRCA1 has been identified in another member of her family, Angela, even though this information has not been disseminated to Helen. Predictive genetic testing would be more informative if the nature of the mutation could be disclosed to the laboratory performing Helen's test. Using the genetic mutation from Angela to facilitate the test will not necessarily point to her. However, because the information is the result of Angela's test, the health professionals involved believe the information is confidential and that they need to obtain consent from Angela to use the result for the benefit of Helen. Without consent they do not feel able to tell Helen about the familial mutation.²³

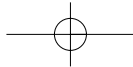
In such cases, the disclosure of Angela's mutation details may be justifiable (especially if they can be de-identified) if they will substantially inform the choice of test to be offered to Helen, or modify her treatment, and if there is no evidence that Angela has refused or objected to her test results being shared. ■

Much genetic information, although originating from an individual, does not necessarily identify one particular individual and could be considered to be used for the benefit of family members unless there are very good reasons to do otherwise.

5.6.2 Where consent to release information has been refused

Outright refusal to share relevant genetic information with relatives is probably rare, although difficult to measure accurately, but where it does occur it is important to weigh up the harms of breaching





confidentiality with the potential benefits to relatives of doing so. This includes weighing up the public interest in maintaining a confidential health service against the public interest in disclosure. Guidance from the Human Genetics Commission,⁹ the Nuffield Council on Bioethics²⁴ and the GMC⁴ reaffirms that the rule of confidentiality is not absolute. In special circumstances, it may be justified to break confidence where in doing so a serious harm can be avoided.

For an illustration of a variety of approaches that might be used in a clinical case where consent to disclose genetic test results had been actively refused, see reference 25.

Before breaching any confidence in circumstances in which the patient/consultand has refused consent to release relevant information, a practitioner should generally:

- (a) attempt to obtain consent to disclosure from the patient in question
- (b) have discussed the case with experienced professional colleagues
- (c) tell the patient that they intend to breach this confidence and why
- (d) keep any disclosure to that which is strictly necessary for the communication of risk
- (e) document discussions with the reasons for disclosure without consent.

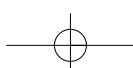
5.6.3 General Medical Council guidance on confidentiality

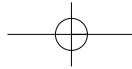
Additional guidance is available from the GMC which in its revised guidance on confidentiality⁵ describes the distinctive nature of some types of genetic information as 'shared information':

Genetic and some other information about your patient might at the same time also be information about others the patient shares genetic or other links with.

They also acknowledge the tensions that might sometimes arise for health professionals, advising that:

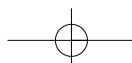
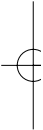
If a patient refuses consent to disclosure, you will need to balance your duty to make the care of your patient your first concern against your duty to help protect the other person from serious harm.

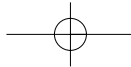




6 Support available

Although local or multidisciplinary team discussions can often help in management of complex clinical cases, other avenues may be utilised in cases where local management has not resulted in consensus. For example, the Genetics Club or local hospital ethics committees may have a role in reconciling conflicts or uncertainties.²⁶





Appendix A: Key legal issues

A1 Human Tissue Act 2004

A1.1 Overview

The 'ownership' of human tissue is illustrated by the common law doctrine that there is 'no property' in a body; thus, there is no legal owner of a dead human body or, by inference, of 'dead' body parts. Instead of conferring property value, the law has focused on the definitions of 'legal possession' of the body, which is a concept describing a transient 'guardianship', in contrast to ownership. A person may have legitimate 'possession' of a body or body part until such time as a person with a greater claim arrives to take possession of the body for burial or appropriate disposal. Alternatively, a practitioner might be viewed by the law as having legal possession of a tissue if that tissue had been removed with consent. Following the revelation that several English hospitals had retained patients' body parts without the consent of their families, it became apparent that existing law made no provision to proscribe such behaviour, which was considered to be unethical by those who investigated the facts, describing the views of the hospitals involved as 'institutional paternalism'. The chief medical officer responded with advice and a government consultation formed the basis of the Human Tissue Act 2004 (HTA).¹

The important thing to understand about this rather impenetrable piece of legislation is that it impinges very little on the everyday diagnosis, investigation and treatment of patients. These activities are governed by the rules that govern obtaining a patient's consent, as provided by the common law and the Mental Capacity Act 2005 (MCA), and laid out as guidance by, amongst others, the GMC.⁴ However, some clinical activities outside the immediate realm of diagnosis and treatment are affected by this Act. The Act also introduces an offence of 'DNA theft', something that was thought to be particularly relevant to clinical genetic practice though, as we will discuss below, the final wording of this part of the legislation meant that DNA theft was limited to very circumscribed circumstances.

The HTA was implemented on 1 September 2006 and provides a legal framework in England, Wales and Northern Ireland for regulating the storage and use of 'relevant material' from the living, and removal, storage and use of tissue from the deceased for 'scheduled purposes', underpinned by consent from the 'appropriate' person.

'Relevant material' 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, which are all excluded from the Act. Chromosome preparations in fixative, dried blood spots, and unfixed tissues fall within the HTA, but cell lines (including lymphoblastoid) and extracted nucleic acid (DNA) are excluded, as is any other human material created outside the human body.

Box 6: Scheduled purposes

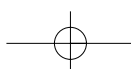
Scheduled purposes are defined as those purposes that generally require consent under the Act.

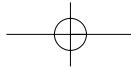
They are as follows.

Part 1 Purposes requiring consent: both living and deceased persons

- 1 Anatomical examination
- 2 Determining the cause of death
- 3 Establishing after a person's death the efficacy of any drug or other treatment
- 4 Obtaining scientific or medical information about a living or deceased person which may be relevant to any other person (including a future person)

continued





Box 6: Scheduled purposes – *continued*

5 Public display

6 Research in connection with disorders or the functioning of the human body (*but see below*)

7 Transplantation

Part 2 Purposes requiring consent: deceased persons only

8 Clinical audit

9 Health-related education or training

10 Performance assessment

11 Public health monitoring

12 Quality assurance

The Act distinguishes between samples taken before (ante-mortem) and after death; the ante-mortem rules apply even if the donor subsequently dies. Thus the death of a patient does not change the purposes for which it can be used (such as clinical audit or research involving anonymised samples). ■

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. This is because these activities are considered integral to good medical care.

A1.1.1 Consent for analysis of cellular material

In general, a living competent adult or child must give consent to the removal, use and analysis of his or her own tissue. After death, only those in a ranked relationship can provide consent. Consent should be sought from those at the top of the list. If it is not reasonably practicable for this person to give consent, (for example, because their contact details are unknown, or they do not wish to make a decision) a person from the next level down the hierarchy may be contacted.²⁸ Where two or more people have equal ranking it is sufficient to obtain the consent of one of them. Consent to the removal of tissue from the living person is not regulated by the HTA and remains a matter for the common law.

A1.1.2 Research

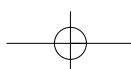
It is legal to perform health-related research on anonymised material derived from living people without consent as long as it is approved by a research ethics committee. 'Anonymised' in this context means that the person carrying out the research is not able to identify the donor, and is not likely 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.

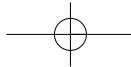
A1.1.3 Samples taken after death including at post-mortem examination

The HTA has made the retention of samples from post-mortem more transparent, and strengthened the role of relatives in that process.²⁹ Previously, samples could be retained for 'therapeutic, educational and research purposes' if there was no reason to believe that the deceased or surviving spouse or relatives would object. Written consent is now required. Model consent forms for post-mortem examination provide that tissue samples may also be taken for genetic testing.³⁰

A1.1.4 Existing holdings: cellular material held before 1 September 2006

Where material held before 1 September 2006 is to be analysed for scheduled purposes (or if anonymised, for any purpose) then the consent requirements of the Act do not apply. This means that this material can be analysed for the benefit of family members without consent.





A1.1.5 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 beyond the screening programme must be obtained.

A1.2 Requirements of the Human Tissue Act relevant to genetic medicine

A1.2.1 Consent for analysis of DNA in cellular tissue

Special rules apply where cellular tissue is held for the purpose of analysing the DNA within it. The Act lists those who can give 'qualifying consent' for analysis of DNA in cellular tissue (consent in the rest of the Act is called 'appropriate consent'). In general, a living competent adult or child must give consent to the analysis of his or her own DNA in cellular material for any of the purposes covered by the Act. The gaining of '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 DNA analysis*²⁸ of posthumous cellular samples is regulated by the wishes of the donor immediately before s/he died, or if no such decision was made, by the consent of a representative appointed by or having a 'qualifying relationship' with the donor.

Those who can give qualifying consent:

- 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.

This list is not ranked; anyone on it can provide consent. Compare with appropriate consent (see section A1.2.4) where the list is ranked.

A1.2.2 Consent for analysis of extracted DNA

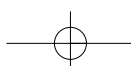
The legal requirements of the Act regarding consent, storage and use do not apply to nucleic acids already extracted from, or cell lines derived from, the cellular material governed by the HTA. This is an important distinction from section A1.2.1 since the extracted DNA stored in NHS diagnostic laboratories is therefore not governed by the HTA. Common law and professional guidance determine the consent requirements for analysis of such samples.

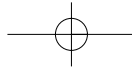
A1.2.3 Non-consensual analysis of DNA held in cellular material ('DNA theft')

A majority of genetic tests are undertaken as part of the medical diagnosis or treatment of the person whose body manufactured the DNA. Where cellular material is held with the intention of extracting DNA and using it for such purposes, it falls outside the HTA.

However, the Act does establish that it is a criminal offence to hold bodily material with the intention of analysing human DNA from it *without* consent for other specific purposes, including research, and where the purpose includes testing for the benefit of a family member (for example, paternity testing). These

* In this context, the term 'DNA analysis' is interpreted quite broadly and includes any process intended to provide information about the DNA in the bodily material such as DNA sequencing, generation of DNA markers, or chromosome visualisation. Methods of deducing information about the DNA from RNA, protein and metabolites are also included (Human Tissue Authority. Code of practice 1: consent (2009), paragraph 152).





provisions were prompted by the Human Genetics Commission which recommended that the theft of DNA and its use for malicious or prurient reasons be unlawful.

Analysing extracted DNA (which has been separated from its parent tissue at some previous time point) does not constitute an offence under the Act; it is only the holding of bodily material with the intention of analysing the DNA within it that does.

A1.2.4 Consent requirement for analysis of cellular material to assist in the care of other relatives

Specific consent under the Act is required to use cellular material where the primary purpose is to assist in the diagnosis and treatment of other relatives. As noted above, this does not apply to extracted DNA.

For an adult with capacity, testing of their stored cellular sample should generally only be undertaken with their consent, since any result may have implications for their diagnosis and/or management. Contact might be re-established through the family member who is seeking advice from the genetics clinic, and who may stand to benefit from the results of the testing.

A person may make a decision about what happens to their tissue after death, or appoint a nominated representative to make a decision on their behalf, but if s/he has not provided a valid consent during their lifetime, then after death, a consent from someone 'appropriate' must be sought. For analysis of cellular tissue the list of those that can provide consent is ranked, whilst 'qualifying consent' (an unranked list) is required if the intention is to analyse the DNA within the cellular material (see section A1.2.1).

Clinical scenario 20

In order to determine the likelihood of Daniel having an inherited predisposition to bowel cancer, a clinical genetics department seeks to obtain tumour blocks from his deceased relatives who had developed bowel cancer. The clinical team are unsure how to proceed. They have heard that they must gain consent from a relative to release the tumour blocks and that a strict hierarchy exists to determine who should be approached.

The exact consent required in this situation depends on what the genetic service wants to do with the tumour blocks:

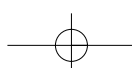
- (a) If they wish to obtain information about histological details recorded in the deceased person's record, then this does not fall under the HTA. Clinicians should be able to access such information if it is to be used for the benefit of a living relative who has consulted them (see section 5.2 for more detail).
- (b) If they wish to perform further tests on the cellular tissue then the clinician will need to obtain consent from the ranked list of appropriate people.
- (c) If they intend to analyse the DNA within the tumour sample then they will also need to obtain consent, but this time the list is unranked. ■

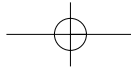
If the primary purpose of testing a tumour block after death is to clarify the diagnosis of the deceased then this testing falls outside the HTA. If however, the analysis is done primarily for the benefit of a relative then consent must generally be obtained from a qualifying relative, as discussed above.

A1.2.5 Analysis of cellular material to assist in the care of relatives where consent has been refused

If a living person with capacity 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 HTA prevent the sample from being analysed.

A distinction should be drawn between the use of existing information to benefit a relative (see section 5) and the new testing of stored DNA to benefit a relative. The balance of harm between the two scenarios is





likely to differ as testing the stored sample may also have implications for the donor (whilst the existing information will already have been disclosed to the donor).

Clinical scenario 21

Rodney was diagnosed with bowel cancer three years ago at the age of 40. Several of his relatives have died from bowel cancer. Although a sample of blood is sent to the genetics laboratory, Rodney decides that he does not want genetic testing. He is concerned this may affect his insurance status in the future (see footnote, page 7) and furthermore does not believe that knowing his genetic status would be helpful. When his sister, Anne, attends the clinical genetic service she is told that accurate predictive testing is only possible where a known familial mutation can be identified. Rodney is the only living affected relative and still declines testing even after being approached by Anne. If Rodney had already been tested and informed of his result, then using this result to benefit Anne (even without Rodney's explicit consent) may be permissible (see section 5.3). However, testing Rodney without his consent, for the benefit of Anne, has potential to generate new and clinically relevant information about Rodney and is therefore a different situation. ■

A1.2.6 Deemed consent where a person cannot be traced

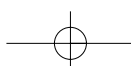
Where it is in the interests of another person (even a future person) that the analysis of cellular material be undertaken to provide scientific or medical information about the donor, the HTA states that where a donor cannot be traced and there is no reason to believe that s/he has died, has refused consent or is incompetent, then the analysis can be carried out following application to the Human Tissue Authority²⁸ 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.

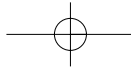
A2 Data Protection Act 1998 and the processing of genetic information

The Data Protection Act 1998 (DPA) came into force in 2000 and replaced and broadened the Data Protection Act 1984. The purpose of the Act is to protect the rights and privacy of individuals, and to ensure that data about them are processed with their knowledge wherever possible. The Act covers *personal data* relating to living individuals.

A2.1 Disclosure of information from family pedigrees

Information shown on a family pedigree can generally be passed between health professionals (under Schedule 3 of the DPA) without the 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). Health professionals should discuss with the person giving a family history that it may be used to determine the mode of inheritance of a disorder, and 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.





The DPA also contains a requirement that data are processed 'fairly and lawfully'. Some suggest that this wording creates a statutory obligation to comply with the wider requirements of the common law on confidentiality. As yet there are no definitive legal judgments resolving this point.²⁷

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; in others this may be necessary in order for another department to link in their relative to the pedigree.

Clinical scenario 22

John, aged 28, is admitted to intensive care with a dissecting abdominal aortic aneurysm. After his acute treatment he is referred to his local genetics unit to see whether he has inherited a gene mutation that predisposes him to aneurysms. He tells the doctor that his cousins have had something similar and were seen in another part of the country. The clinical geneticist asks the relevant genetics unit for further information. An anonymised pedigree is sent because of concerns about breaching the DPA.

In this case, sending names and dates of birth on the pedigree could have helped the healthcare professionals treating John to determine where he was located in the pedigree, and could have been useful in establishing the mode of inheritance as well as identifying whether genetic testing in others could target John's testing. Genetic services (or other clinical departments) should treat such pedigree information in the same way as any other clinical information: sharing such information on a 'need to know' basis is appropriate. ■

Family history and clinical information should be shared with other health professionals (regardless of their geographical location) if the sharing of confidential information is necessary for the purposes of healthcare, and disclosure is between healthcare professionals who share in a duty of confidence.

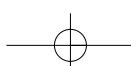
A2.2 Fair processing and disclosure of results of genetic tests

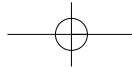
In some instances it may not be possible to determine the extent of consent sought when a sample was taken. Also, it may not be possible, or appropriate, to contact the person who gave the sample, if in so doing confidential details about other family members are revealed.

Advice from the information commissioner about the fair processing of results, particularly those originally generated from samples stored in genetics units during the development of clinical DNA services over the last 20 years, suggests 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 DPA, s/he must be informed of this fact. If re-contact of the person who gave the sample would involve 'disproportionate effort' then an exemption is available. Furthermore, if there is evidence that they would have wanted their relatives to know then it may not be necessary to seek consent.

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, taking into account the nature of the data, and the length of time and the cost involved in contacting the original patient to provide the fair processing information.

If, having considered all the factors, a health professional wishes to share information for clinical care (for instance, scientific information about a mutation) with another health professional under this exemption of the DPA, this should be documented. If this process does not require a specific individual





to be named (if it is possible to say ‘the results on a family member show mutation X’), then the DPA may not need to be invoked.

For new and prospective samples, the requirements of the DPA 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, as in the model consent form in Appendix C1.

A2.3 Payment for copy records

Under the DPA and the Access to Health Records Act, holders of records (hospitals and general practices) are permitted to 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.

A3 Mental Capacity Act 2005

A3.1 Overview

The MCA² provides a statutory framework for care or treatment of individuals who lack capacity to consent in England and Wales. Section 1 of the Act establishes a set of principles which govern how decisions relating to that person should be made. These include an assumption that a person has capacity unless it is proved to the contrary and that, in making that assessment, all practicable steps to help that person make a decision have been taken. Section 1(5) also establishes that an act done or decision made for or on behalf of a person who lacks capacity must be done or made in his best interests. The MCA is supplemented by codes of practice which provide practical advice and examples concerning the interpretation and implementation of the Act.

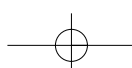
A3.2 Scope of individuals and activities covered by the MCA

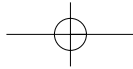
The MCA applies whenever it is reasonable to believe that a person lacks capacity to decide about a particular matter, having taken reasonable steps to make a careful assessment, and involves a determination of best interests (section 5b (i) and (ii)). The focus of the MCA is on those aged 18 and above, although it does allow financial decisions to be made about 16- to 18-year-olds. It does not apply to children. It also provides a framework for those lacking capacity to be involved in research, provided that certain conditions are satisfied.²

A3.3 Providing consent

The MCA 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. In many cases, individuals will have the capacity to consent if time is spent explaining the issues in simple language. A general explanation of the test or procedure, and the risks and benefits of proceeding may suffice.

The MCA states that if a person lacks capacity and a person has been appointed as an independent advocate then the appointed person can give consent on their behalf (as long as they act in the patient’s

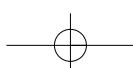




best interests). Those with more informal relationships (such as family members interested in the patient's welfare or carers) may also have a role in advising what might be in the patient's best interests. The MCA also allows individuals with diminishing capacity to grant lasting powers of attorney.

Where genetic testing involves a person who is unable to consent, a consent form should not be signed, but it is good practice to document in the medical records why the action was believed to be in the patient's best interests.

In Scotland, the Adults with Incapacity (Scotland) Act 2000 (as amended) provides a clear legal framework for a range of medical treatment and welfare decisions relating to adults unable to consent. It allows consent to be given on behalf of an adult with incapacity: there is a statutory requirement to consult a nearest relative, carer or court-appointed official but decisions are not necessarily made on a 'best interests' basis (as this is felt to be unduly protective). In theory this means that carers or close relatives may refuse testing even if other family members regard an intervention as indirectly beneficial.



Appendix B: Three flowcharts (Figs 1–3) summarise the main guidance pictorially

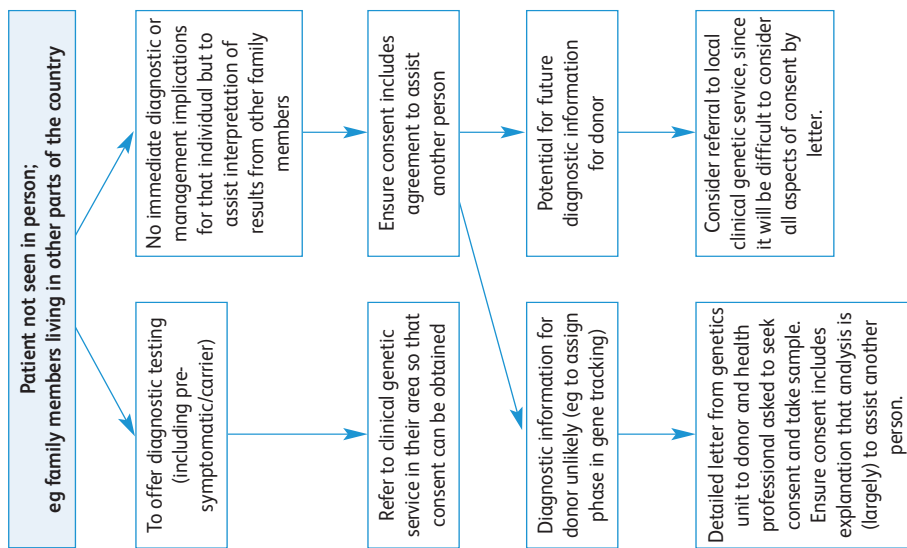
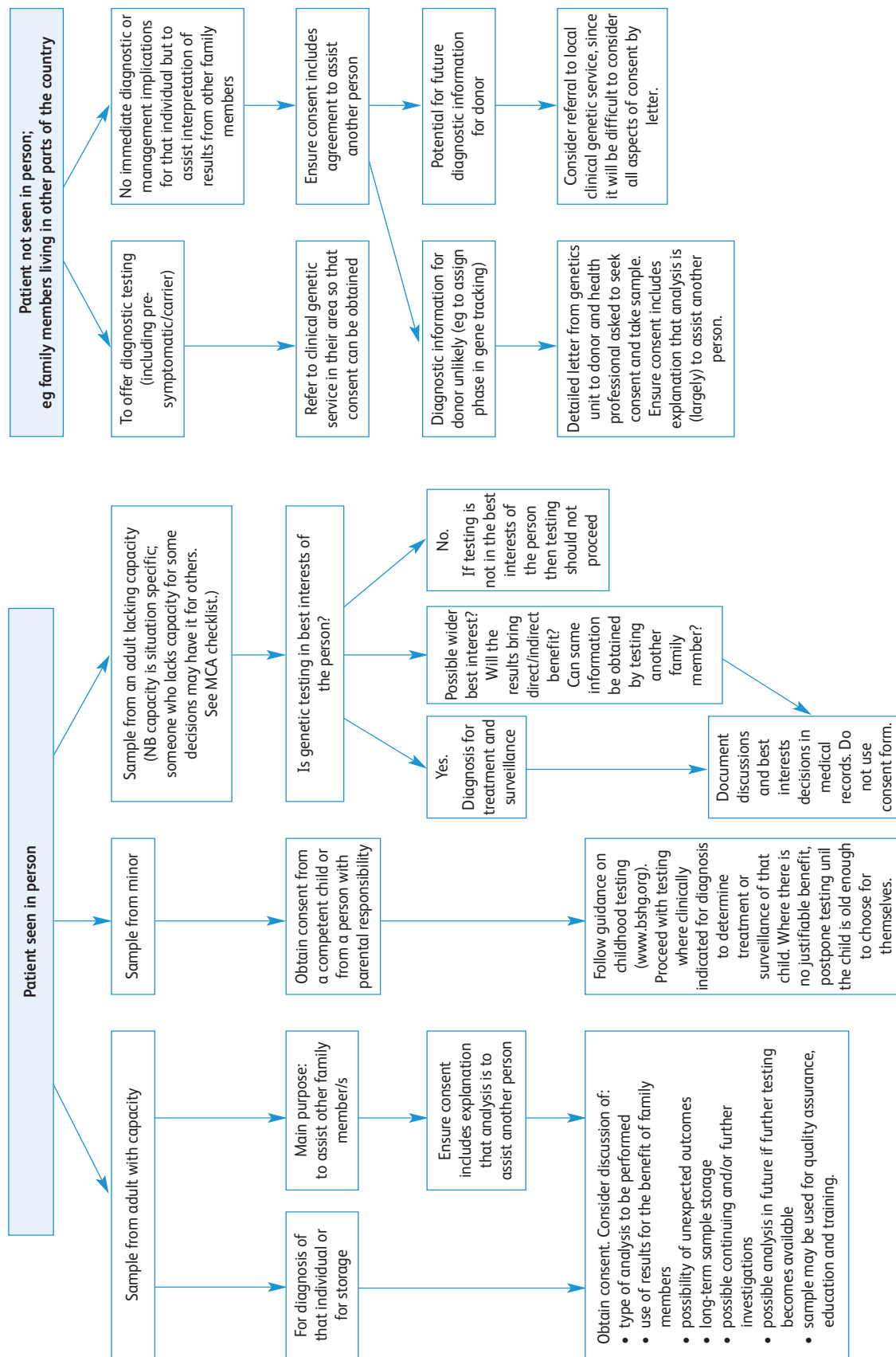


Fig 1 Consent for (new) samples. This flowchart is only a summary and should be used in conjunction with the main report.

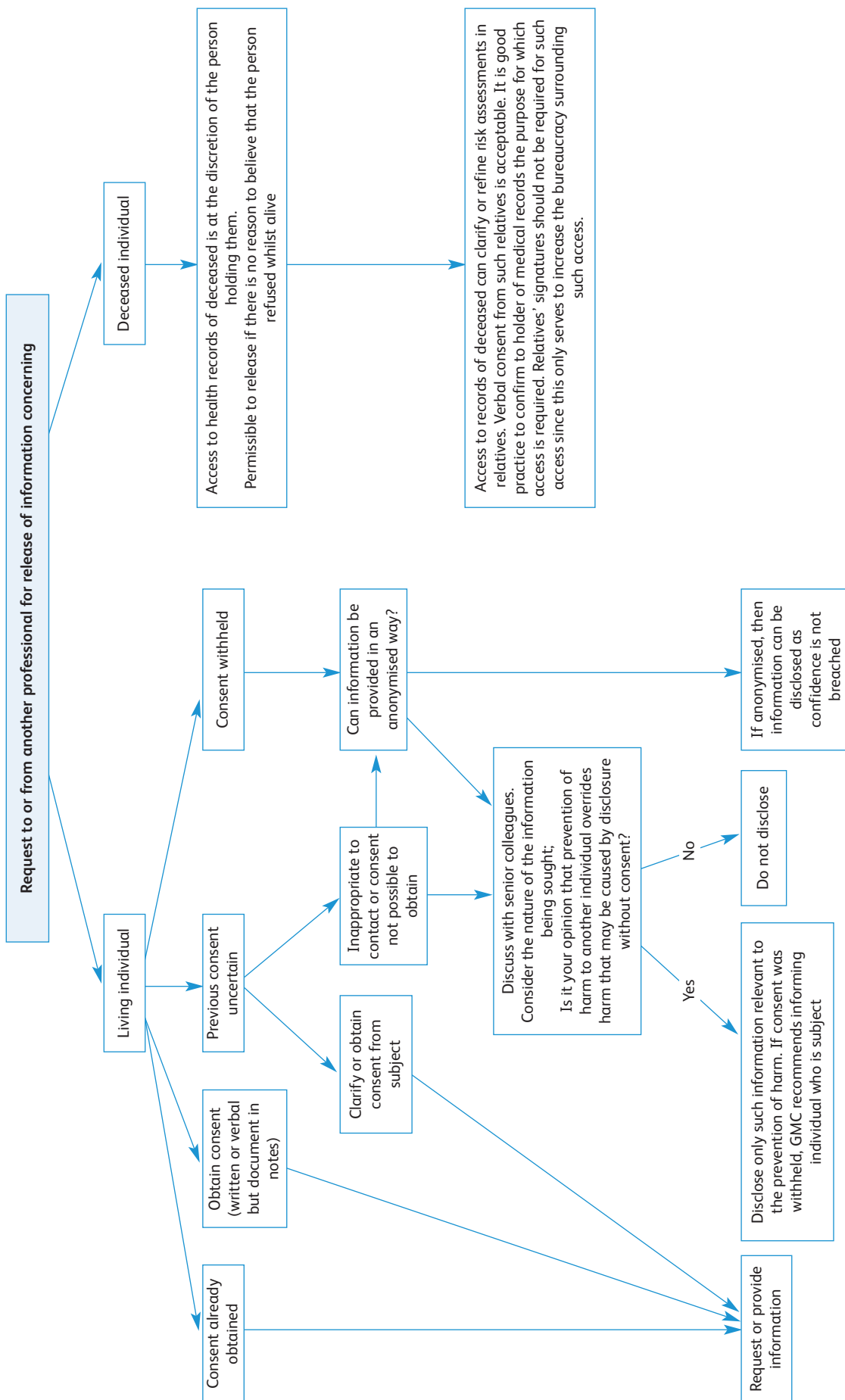


Fig 2 Sharing and disclosing genetic information. This flowchart is only a summary and should be used in conjunction with the main report.

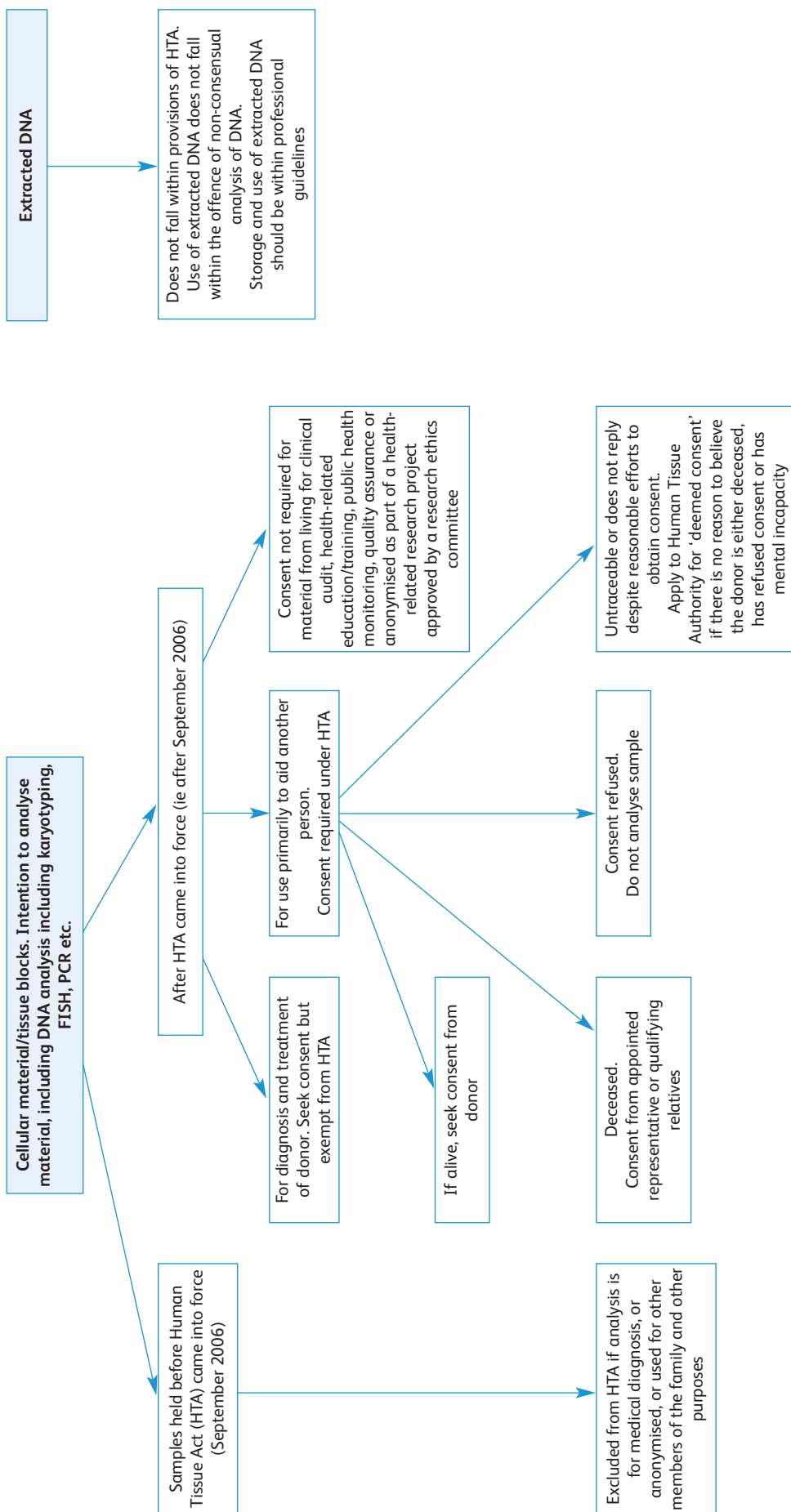
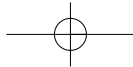


Fig 3 Consent for analysis of stored samples or pathological material. This flowchart is only a summary and should be used in conjunction with the main report.



Appendix C: Suggested consent forms

C1 Consent to access medical records

Dear xxx

You may be aware, that a relative of yours has been referred to the [insert] Clinical Genetic Service to assess their risk of developing [insert details]. It would be helpful to obtain some medical information about you, so that we can carry out this assessment and decide how best to assist your relative.

In order to do this, it would be useful to access your medical records as they contain information about the tests and treatment that you have received. The purpose of this letter is to request your written consent to access your medical records at the hospital where you were treated.

This information will be treated in the strictest confidence and will not be used for any purpose other than making a specific assessment of risk for family members. This information may also be sent to another regional genetic service if requested, if it is relevant to another family member. I would be most grateful if you could complete the enclosed consent form and return it to me at your earliest convenience.

[Signature]

[Contact number of applicant]

.....

[Instructions to addressee: Please complete the attached form and return it to the address below, in the attached SAE]

I, the undersigned, consent to the release of information contained in my medical records.

Name

Date of birth

Address.....

Date of diagnosis

Type of diagnosis.....

Hospital where treated.....

Consultant.....

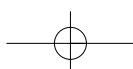
Address at time of diagnosis

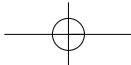
Print name

Signature of authorisation.....

Date..... / / File No.....

Please return this consent form to at the above address.





C2 Record of discussions regarding testing and storage of genetic material

1. The results of a genetic test may have implications both for the person being tested and for other members of that person's family, and I acknowledge that my results may sometimes be used to inform the appropriate healthcare of members of my family.
2. Normal laboratory practice is to store the DNA extracted from a blood sample even after the current testing is complete. This is because in the future (months or years) further/new tests may become available. In such cases I would like:
 - (a) to be contacted before further relevant tests are done on the stored sample if new tests become available; or *Delete as appropriate*
 - (b) further diagnostic tests to be undertaken on the stored sample. I will be told of any informative results. *Delete as appropriate*
3. Occasionally leftover samples may be useful in checking laboratory techniques and my sample might be used as a 'quality control' for other testing, for example, that of family members.
4. Note of other specific issues discussed [*eg research, insurance*]:

.....

.....
5. I have discussed these issues with [*name of clinician*].....

I consent to my sample being tested for:	Date
..... / /
(test to be undertaken) <i>If parent is consenting for a child, please amend as appropriate.</i>	
Patient/parent signature	Consent undertaken by:
.....	(clinician's name and signature)
.....

Affix sticky label or fill in details

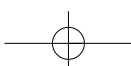
Patient name:

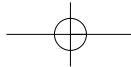
Date of birth: / /

Patient address:

Genetics ref:

1 copy for notes, 1 copy for patient to retain





C3 Record of agreement to a photographic record (still or video)

Date:..... Consultant:.....

First names:.....

Surname:.....

Date of birth:..... Record number:.....

I agree that the photographic images of

.....

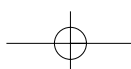
(a) can be stored as part of a confidential medical record and used as an aid to diagnosis
Yes / No / Not applicable

(b) may be shown to appropriate health professionals to aid medical teaching and research
Yes / No / Not applicable

(c) may be published in a journal, textbook or website.
Yes / No / Not applicable

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 contacting or writing to [].



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