

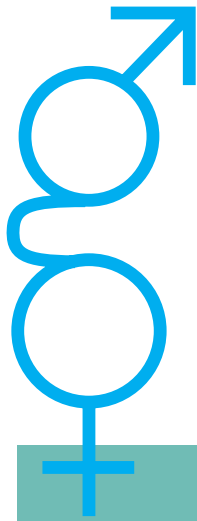
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**GENETIC
INTEREST
GROUP**

*Working to benefit
all people affected
by genetic disorders*

Confidentiality Guidelines



Confidentiality & Medical Genetics

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Preface: the scope of this report

Spectacular advances in human genetics have occurred in recent years. The growth of clinical genetics, and in particular of genetic testing, has generated much discussion on issues related to individual privacy and confidentiality. Important questions have been raised about third-party access to genetic information in a number of distinct contexts.

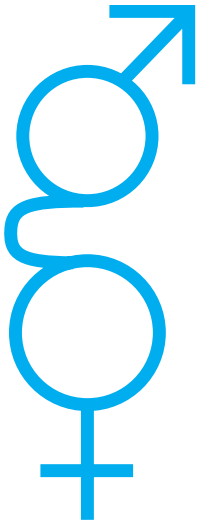
In the UK, questions of access to such information by insurers and employers have been considered by the Human Genetics Advisory Commission. In America, many states have drafted genetic privacy bills, and federal legislation is proposed which may outlaw or restrict third-party access to genetic information.

Another set of issues concerns human tissues used for genetics research and the confidentiality, ownership and use of genetic information so generated.¹

Yet another context is clinical genetics practice and the sharing of genetic information within families and between professional clinical geneticists. This document exclusively concerns this latter debate on individual confidentiality in medical genetics.

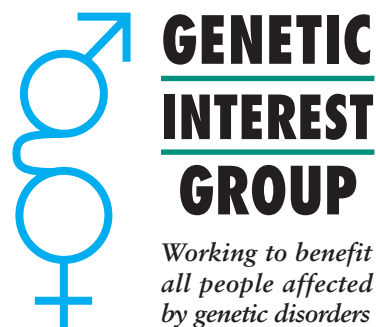
‘Genetic information’ is taken to mean any data of clinical relevance to the genetic status of an affected or at risk individual. No distinction is made between, for example, family information arising from a counselling session, phenotypic observations made during clinical evaluation or laboratory test results.

We restrict our attention to information held within, or available to, medical genetics departments, but we recognise that medical information on an individual that is not explicitly genetic in character, held on record in a medical department other than a genetics department, may have implications for other family members. As we learn more about the genetics of common diseases, the probability of this will increase. It is our hope that the approach taken in this report can lay the basis for an examination of similar issues as they arise in these broader contexts.



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The Genetic Interest Group is a national alliance of organisations with a membership of over 100 charities which support children, families and individuals affected by genetic disorders. Its primary goal is to promote the awareness and understanding of genetic disorders in order that high quality services for people affected may be developed and sustained

The Working Party

This document is the outcome of a Working Party convened by the Genetic Interest Group. The members of the Working Party were:

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1 Introduction

1.1 Family information

Unlike most medical information, genetic information is inherently more than a matter for the individual – it is of relevance to families. In order to give accurate clinical information to an individual at risk for a genetic disorder, it is sometimes necessary to use medical data obtained from one or more members of that individual's family. Because of this, exchange of family information is inherent in medical genetics. The family connection can also mean that the detection of a genetic mutation in one individual has direct implications for other family members who may not be aware of their risk status.

1.2 Questions of principle

Confidentiality is a central tenet of medicine, but in clinical genetics the family connection raises important questions of principle and practice. Is the duty of confidentiality owed to the individual who seeks medical information, or is it owed to the larger family? Should professionals feel duty-bound to inform other family members, even if they are unaware of any problem? In its report on genetic screening, the Nuffield Council on Bioethics argued that it may be appropriate to define the family as a 'unit' in relation to the use of genetic information. However, it also recognised that disclosure of genetic data within the family 'raises some of the most serious issues'.²

1.3 The professional community

In the UK, medical geneticists form a small, close-knit community of practitioners. Within this community, many professionals have seen little problem in sharing relevant items of family information when required for the benefit of their patients.

However, medical genetics is in a growth phase. The volume of genetic testing, (particularly DNA-based) has grown explosively in recent years as the technologies and discoveries of the Human Genome Mapping Programme have been transferred to the clinical diag-

nostic laboratory. This growth will continue as DNA testing for predisposition to common disorders such as coronary heart disease, cancer and diabetes becomes possible.

As the medical genetics community grows, so the personal networks become diluted and a more formal mechanism for sharing family information is required.

1.4 Public scrutiny and the threat of regulation

The activities of geneticists, both in the hospital clinic and the research laboratory, are under increasing scrutiny by the public, the media and by regulatory bodies. Documents recently published in Europe and the USA place more emphasis than was previously the case on individual confidentiality.³ Professional attitudes are also shifting.⁴ In Britain and elsewhere, while there is a recognition that a family approach is needed, there is a trend towards emphasising the duty to the individual over the duty to the wider family.

Such concerns might conceivably lead to restrictions on the exchange of medical-genetic information within families. A regulatory framework is needed, but one that is not too restrictive. It is important to protect confidentiality but it is also important that information can be shared within genetic services when this is necessary and appropriate for the benefit of family members.

1.5 Aims of this report

This report aims to:

- describe current practice in medical genetics in the UK with reference to individual confidentiality
- discuss ethical issues relating to the shared use of individual genetic information within families
- propose a framework to guide professionals which:
 - formalises existing practice
 - suggests a mechanism for resolving 'difficult' situations

Regulatory and legal issues

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2 Creating a space for professional discretion

Although there is no doubt that the obligation of confidentiality is recognised in both law and ethics, there remains scope for considerable discussion over both its basis and its extent.⁵ There is no comprehensive statute dealing with confidentiality in health care, and few cases have come before the courts for decision.⁶ Guidance has been issued by the Department of Health⁷ and by professional bodies⁸ but it does not neatly resolve the problems with which this report is concerned. This section explains the legal context and proposes a framework for the regulation of decisions about disclosure of confidential genetic information in the context of professional and family practice.

A central argument of this section is that established approaches to the question of confidentiality in healthcare cannot resolve all of the difficulties that arise in the field of clinical genetics. Elements of a new approach are outlined at the end, after the limitations of existing frameworks have been discussed.

Two other points about this section should be kept in mind: (i) It does not seek to set out the full range of ethical approaches to confidentiality, rather to illuminate the current legal position by showing how it is related to strands of ethical thinking. (ii) It does not seek to resolve difficult questions about the broader ethical obligations of patients to their families that might outweigh claims to privacy. The primary focus here is on the legal regime, and later discussion will show how it may be important to recognise some moral imperatives that are nevertheless not to be enforced as legal principles.

2.1 Two approaches to confidentiality

Two significant strands of thinking have developed on why confidentiality is important. The first emphasises the individual rights of patients to privacy – not to have personal information made publicly available without agreement. This approach places the interests of the person with whom the information is primarily concerned at the centre of decision making.

The second approach looks at the problems from the perspective of health care providers. It emphasises the collective good that confidentiality provides. It notes that health services are valuable to society, that health professionals cannot provide an effective service unless they have full and accurate information about their patients' problems and needs, and that this information will not (in practice) be shared with them unless guarantees of confidentiality are offered. In this second approach, confidentiality is important because it is instrumental to providing good health services. In the first, confidentiality is a moral requirement in its own right.

The difference between these two understandings of confidentiality is crucial in our context. When the collective approach is dominant it can readily be argued that information should be shared by professionals and within families whenever it would be useful to do so. Occasional breaches of confidence would be justified by the collective interest in maximising the potential for providing advice and other services.

This would have the effect of removing control from the person to whom the information primarily relates and placing the power to decide when to disclose information, together with the duty to exercise that power responsibly, on to the health professionals.

Although such an argument may be superficially attractive to professionals who are seeking to provide the best care to those who seek their help, we believe that it pays insufficient regard to the rights of the individuals involved. It might also prove to be counter-productive, because people might become wary of consulting genetics services for fear of personal information being broadcast. Instead, we believe that it is important to develop a framework that recognises the rights of the individuals concerned, but encourages them to consider the interests of others who might benefit from information being shared.

2.2 Confidentiality and the law

Rights to privacy are recognised in many legal jurisdictions, but there has traditionally been no such general recognition in English law. Here confidentiality is usually said to arise because of the circumstances in which the information is confided or generated. The courts have regarded information as confidential provided that it is of a confidential nature – and it is accepted that personal information comes into this category – and that it is imparted, or created, in circumstances of confidence.

There can be no doubt that the relationship between patients and health professionals is a confidential one, and satisfies the second stage in this test. This second point also emphasises that the rules are intended to ensure that patients' expectations of professional confidentiality are honoured. For litigation purposes, the courts may also require that any breach of confidentiality causes 'damage' to the person suing, but it has been recognised that an invasion of personal privacy by disclosing sensitive information will suffice, although a more substantial impact may need to be proved before significant damages are awarded.

Although the English courts have not developed a specific right of privacy, one is set out in the European Convention on Human Rights. The United Kingdom is a signatory to that Convention, and therefore the Government is obliged to ensure that citizens' rights under it are protected. Further, it will soon be incor-

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Regulatory and legal issues

porated into English law and become binding upon public authorities such as the NHS. Under Article 8 of the European Convention: 'everyone has the right to respect for his private and family life'. This right is not absolute, and is subject to limitations in accordance with the law, and necessary for a range of purposes including the protection of health or for the protection of the rights of others. Thus, even within a rights based approach there is scope for legitimate breaches of confidence in some circumstances.

2.3 Consent to disclosure of confidential information

The most straightforward and widely accepted justification for disclosing confidential information is the consent of the patient. If confidentiality exists primarily to protect the rights of patients, then patients should be entitled to waive the requirement of confidentiality. They may do this in relation to their own position, such as when they are satisfied that disclosure does not compromise their interests, or because they believe that their responsibilities to others require them to do so. From the legal perspective, it is the patient's consent that justifies disclosure. The patient's motivation may raise ethical issues, on which it may be important to express views, but these do not affect the legality of disclosure – in law the consent is the justification for disclosure.

In some cases, it will be possible and appropriate to seek consent from the patient for each disclosure of confidential information and this would be the usual course of action in relation to controversial or difficult decisions. The starting point when considering the importance of sharing genetic information will thus usually be to seek the consent of the patient (see section 7 for more on this). This preserves the autonomy of the patient, and allows professionals who believe that it would be right in the circumstances to share information to explain to the patient why they think so. In many cases, consent will be given. If consent is refused, and the professionals believe that it would be best if the information was disclosed, then consideration would have to be given to other recognised limitations on the obligation of confidence.

2.4 Sharing information within the professional team

The first of these is the 'need to know' principle. The nature of modern health care, delivered by teams of professionals working together in the interests of patients, is such that it is impracticable to seek specific consents every time a nurse communicates with a doctor about a patient's care, or when laboratory tests are discussed with clinical staff. The 'need to know' exception to the obligation of confidence has been developed to explain why such disclosures are acceptable.

The sharing of information in this way is part of the process of care that patients have sought, so it constitutes a performance of the professionals' duties towards their patients not a breach of them. The precise boundaries and basis of the need to know principle are unclear, but there are two reasons for thinking that it has limited relevance to the question of disclosure of genetic information within families.

First, it is probable that the principle should be interpreted as being based on the implied consent of patients. Provided that patients understand the way in which information is used by the health services, they can be taken to have consented to those uses when they agree to receive the services.

Thus, the Department of Health's guidance on confidentiality offers a draft of an information sheet explaining to patients the way in which information may be shared within the NHS.⁹ Similar advice for patients could be provided by specialist services to indicate particular needs to share information in the area of work involved. This could be used to alert patients to the possibility that information may be shared with other members of the family.

But if the basis of the need to know exception is consent, then patients will always be entitled to opt out of sharing information by withdrawing any implied consent that can be assumed on the basis of such general information. In the context of sharing genetic information with other family members it may be undesirable to risk proceeding on the basis of an implied consent when an explicit consent could be sought (see below). Thus, the 'need to know' principle will rarely be used in this context, although it might be important in relation to surveillance mechanisms such as genetic registers.

The second reason why the 'need to know' exception to confidentiality is likely to have a limited role to play in our context is that it relates to the care of the patient from whom the information came. Where the suggestion is that information needs to be disclosed to other members of the family, the 'need' that is in issue is not the need of the index patient, but that of the other family members.

Unless the need to know exception is based on the broader public interest rather than implied consent, it will not therefore address the question with which we are concerned. There is some basis for asserting such a public interest basis, but it is of doubtful authority.¹⁰ For reasons outlined above, we do not believe that it is sufficient merely to assert a collective interest in efficient genetic medicine as a justification for overriding confidentiality. Where the basis for disclosing information is the interests of someone other than the patient, then those interests should be explicitly considered as competing with the rights of the patient.

Regulatory and legal issues

2.5 Competing interests: current practice

There are a number of cases in which the interests of others are recognised as justifying disclosure of otherwise confidential information. In some cases the law requires disclosure, such as the statutory notification by doctors of patients with some infectious diseases.¹¹ Here the law provides a duty to disclose the information to the appropriate authority. However, this is the exception, rather than the norm.

It is widely accepted that English law does not impose on health professionals a general duty to warn people in danger from their patients.¹² The more common position is that there is a legal power to disclose, but no legal duty to do so. In such circumstances, it becomes a matter for professional discretion and professional ethics whether information should be disclosed. Here, the law makes neither disclosure nor the maintenance of confidentiality obligatory, regarding the issues as too variable to be answered in advance by a uniform rule. An example of this type of situation is child abuse, where there is no legal obligation for health professionals to disclose information but professional guidance indicates that their ethical obligation is to do so.¹³

These two examples illustrate that the law permitting disclosure on the basis of the interests of others is usually concerned with cases where there is an identifiable threat of harm presented by the patient. These circumstances will rarely be present in the context of the sharing of genetic information. The patient whose confidentiality may be breached will not himself or herself be presenting a threat to the other members of the family. Rather, those other members of the family are faced with a threat of ill health from an independent source (i.e. their genetic make up) about which the index patient might be able to warn them. This is likely to be seen by the English courts as a question of whether the patient should be required to go to the assistance of their relatives, not as a matter of a threat presented by the patient.

2.6 English law and the obligation to warn

As a general principle, English law does not require anyone to go to the assistance of people in distress. It is only where some special circumstance exists that such an obligation to rescue is accepted. This is sometimes illustrated by the scenario of a toddler drowning in a shallow pool. The law would say that a passer by could simply walk past, without helping, because there is no duty to act as a 'Good Samaritan' rescuing strangers.

There would, however, be some people who would be obliged to help the toddler. Those with parental responsibility would have to save the toddler because part of their responsibilities is to take reasonable steps to protect it from harm – an obligation that is imposed and enforced by law. This obligation would not exist for other family members unless they had undertaken to look after the toddler, but if they had done so the law would require them to keep their promise.¹⁴

Those who were employed to save people in the pool, most obviously the lifeguard responsible for safety in it, would also be expected to rescue the toddler. Finally, if the toddler had been pushed into the pool, the person who pushed it would be expected to go to its rescue, because he or she was responsible for the danger. There is a precedent for this last approach in relation to confidentiality in that it may be relevant whether there is a degree of culpability on the part of the patient. This has been explained in terms of there being no confidentiality in relation to fraud, criminal conduct and iniquity.¹⁵ Someone who had lied to a prospective spouse about a genetic condition might forfeit his or her right to confidentiality on this basis.

It follows from the reluctance of English law to accept the 'Good Samaritan' principle that compulsory disclosure of confidential genetic information is unlikely to be justified under the present law, save possibly in the case of the parents of children while they are under 18, where the duties of parents might be extended. It is possible that an initial agreement between genetic services and patients that they would voluntarily accept a responsibility to assist other members of their families would be interpreted as sufficient to require them to do so. In effect, to provide a consent to disclosure that they would not be permitted to revoke. However, this would be unprecedented and unnecessarily coercive.

2.7 An ethical imperative to share information

It would probably be more appropriate to concentrate on the ethical principles pointing towards disclosure rather than the more heavy-handed approach of the law. People would then be encouraged to act ethically, but not forced to do so against their will. This focus on acting ethically would be relevant to both health professionals and patients, although only the professionals would be called to account, by their professional bodies, if they did not act ethically. It is well established that health professionals are expected to seek to do good for their patients and not merely refrain from harming them. This is generally described as the principle of beneficence.

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Regulatory and legal issues

Beauchamp and Childress have argued that this principle can also apply to all individuals, including patients, in certain circumstances. They outline these as being as follows:

- That the person to be helped is at significant risk of harm.
- That help from the person faced with the choice is needed to prevent that risk materialising.
- That there is a high probability that their help will prevent the harm.
- That helping would not present significant risks, costs or burdens to the person asked to help.
- That the benefit for the person to be helped outweighs the costs or burdens to the person asked to help.¹⁶

These conditions might well exist in many cases where patients are encouraged to permit information to be shared with their families and it would be appropriate for health professionals to draw their attention to the argument that they have moral responsibilities to help them. Many patients may decide, after considering these arguments that it is appropriate to consent to the disclosure. Indeed, practical experience and academic study¹⁷ tells us that most patients do consent to disclosure and are otherwise keen to help. However, there will be some who may not want to. It is therefore necessary to consider whether there is a case for professionals sometimes disclosing confidential information to other family members even without the consent of patients. It is unlikely that such disclosure would fall within the existing exceptions recognised by the law.

2.8 The relevance of a professional consensus on disclosure

A first step in this consideration would be to identify current professional practice. This may have an indirect relevance to the legal framework of confidentiality as the courts have shown themselves willing to look favourably on the terms of professional guidance when interpreting the scope of medical confidentiality. In at least one case, they have considered that evidence that a doctor's disclosure was within the scope of the exceptions accepted by the General Medical Council was also evidence that it was lawful to pass on confidential information in those circumstances.¹⁸

If geneticists were to develop a consensus on the approach to be taken to cases where consent was withheld, the courts might be prepared to accept that it served to define the limitations of confidentiality. This would be particularly likely if it was adopted by services across the country and made known to patients. As explained above, the legal foundations of confidentiality include an assessment of the context in which the information became available. Such a consensus could serve to define that context, with explicit indications of how information would be used, so that disclosure in accordance with the protocol would become the honouring of the promise of confidentiality rather than its breach. To see whether such a consensus can be identified, it is necessary to describe and appraise current practice and the values on which it is based.

Current practice

3.1 The medical genetics service

Medical genetics is an integrated service comprising clinical genetics and laboratory genetics (molecular genetics, cytogenetics and biochemical genetics). Medical genetics overlaps with most other specialities since mutant genes and chromosomal abnormalities may have pathogenic effects in any organ system and at any age. A high level of communication is therefore essential within the medical genetics community and between geneticists and all other groups of health-care professionals.

Medical genetics is a young speciality and is still an imperfect art. Most of the 100,000 human genes have yet to be characterised and while a small proportion of genetic illness can be treated or managed with benefit, the majority cannot yet be cured. Routine gene therapy will not be available for some years. In many cases, the best medical genetics currently can offer is (sometimes) prevention of disease development, amelioration of symptoms or complications associated with particular disorders, or informed reproductive decision making.

3.2 Presumptions and expectations

A framework of presumptions and expectations underpins current practice in medical genetics.

3.2.1 'Professional' presumptions

The primary presumption that underpins professional practice can be simply stated:

It is better for individuals to be informed of their genetic risk than not to be so informed.

This presumption is based on the fact that knowledge can be used to inform decision-making and clinical management of disease, as discussed below. Based on this presumption, professionals encourage patients to inform their relatives that they are at high risk of carrying gene mutations or chromosome abnormalities.

This presumption is based on a wealth of collective professional experience and anecdotal evidence. Our recommendation is that practice should continue to be guided by this, but we also strongly recommend that more formal research should be undertaken to test it.

3.2.2 Patient expectations

Most patients are referred for genetic counselling by other healthcare professionals as a result of a condition experienced by themselves or their close relatives. For some conditions, especially in certain high-risk target populations, referral may follow a positive test in a general screening programme.

Patients seek counselling when they want to know their genetic status. In particular, population screening programmes specifically select those most motivated to learn of their genetic risk. If individuals do not wish to know, they cancel appointments or do not seek information.

The decision to seek genetic counselling is informed presumably by the expectation that obtaining genetic information is likely to be more beneficial than not, although decisions may be made without the advantage of full information. Some prior consideration of the issues suggests some awareness of the possible psychological or emotional costs. This awareness itself may minimise the impact of any eventual costs – 'expecting the worst'.

3.2.3 Costs as well as benefits

Professional assumptions and patient expectations largely hold true when the various medical, social, psychological and emotional benefits gained from the acquisition of genetic information outweigh the costs to the individual or family. However, for some patients the costs of learning about their genetic risk outweigh the benefits. Such situations may occur for some individuals when the disease in question is, for example, serious, late onset and untreatable (such as Huntington's disease).

One objective of genetic counselling is to provide genetic information and support in such a way as to minimise the psychological costs of genetic investigations.

3.2.4 Confidentiality and the right not to know

Presumptions, expectations, and the balance of costs and benefits all have a bearing on issues related to the sharing of genetic information within families. Dilemmas occur in two distinct contexts relating to the privacy of individual genetic information.

The first of these concerns the conflict between maintenance of individual confidentiality and provision of accurate information to other family members. The second concerns the conflict between the rights of an individual who is ignorant of his or her genetic risk to be told and the right not to know. Discussion of these issues forms sections 4 and 5 respectively of this report.

3.3 How genetic information is used

One of the central goals of the genetics service is to acquire and communicate information and the imperative is for that information to be as accurate as possible. Genetic information is used primarily to provide accurate diagnosis and to estimate carrier risks.

3

Current practice

3.3.1 Diagnosis

Accurate diagnosis of genetic diseases is clearly beneficial for individual patients when options exist for clinical management. For example, accurate early diagnosis of cystic fibrosis in a newborn child who is failing to thrive can lead to prompt prophylactic antibiotic treatment and pancreatic enzyme supplements, both of which can improve prognosis.

Even when there is little medical treatment to offer (for example, in the case of fragile-X syndrome, Prader-Willi syndrome or tuberous sclerosis), many families take a degree of comfort from having a diagnosis. This allows them to contact patient support groups; coping strategies can be developed and families have better access to special educational facilities and social services.

3.3.2 Carrier testing

Determining that individuals carry recessive gene mutations or balanced chromosome rearrangements allows couples to make informed reproductive choices.

Carrier couples may decide not to have children, they may seek a child by artificial insemination by donor, or they may choose natural pregnancy and prenatal diagnosis. The birth of an affected child can be prevented by termination of an affected pregnancy. Alternatively parents may choose to continue an affected pregnancy using the information gained to plan for the future.

3.4 How genetic information is obtained

Professionals obtain data from a number of sources. This includes clinical evaluation, genetic data about other family members and laboratory investigations.

3.4.1 The family tree

The primary tool for gathering genetic information in families is the pedigree diagram or family tree. The very act of drawing up a pedigree involves discussion of the disease status of all members of the immediate and extended family.

Knowledge about the 'index case' – a key affected individual in the family and usually the first family member seen in the genetics clinic – is used for the benefit of other family members.

The degree of genetic risk for each relative is determined, in part, by his or her relationship to the index case. Estimations of genetic risk from pedigree structure alone ('prior' risks) are often unhelpful – especially for more distant relatives of the index case. Prior risks may be modified by laboratory test results or other information in order to provide more accurate information to relatives.

3.4.2 Laboratory testing

Most UK medical genetics centres include laboratories performing clinical cytogenetic (chromosome) and clinical molecular genetic (DNA) analysis. All centres have access to Regional clinical chemistry laboratories specialising in inborn errors of metabolism.

3.4.2.1 Clinical molecular genetics

DNA tests fall into two types – linked-marker analysis (indirect testing) or mutation analysis (direct testing).

Linked-marker analysis

Linked-marker analysis is used when the disease gene in a family cannot be characterised and always requires simultaneous investigation of several family members.

Linked markers lie physically close on the chromosome to the disease gene under investigation and typically are stretches of non-functional DNA which vary in length between individuals. Because of the stable inheritance of DNA length variants ('alleles') from parent to offspring and the availability of laboratory techniques to score individuals by the alleles they carry, linked-markers are used to track disease genes through families. A particular allele *per se* does not cause the disease. Rather, it marks the chromosome that bears the mutant gene in a family.

Individuals are identified as gene carriers if they inherit the same pattern of marker alleles as affected or carrier relatives. Linked-marker analysis cannot be applied to an individual in isolation from other family members; a consequence is that the likely disease status of individuals other than the person seeking the test may be revealed.

Mutation analysis

Mutation analysis is used when the particular causative mutation is known to segregate in a family – or is likely to do so. These are direct tests that can often be applied to individuals without using information about other family members.

For example, a single type of readily identifiable mutation almost always causes fragile-X syndrome. If the mutation is present in a developmentally delayed child, the diagnosis of fragile-X syndrome is confirmed.

However, for other disorders, despite the direct test, information about an affected relative is essential to support accurate interpretation of results. This is exemplified by cystic fibrosis (CF). Although over 500 different pathogenic CF mutations have been reported, viable genetic testing is only possible for a handful of the most prevalent. CF is recessive and is relatively common in Caucasians – one person in 25 is a carrier.

Genetic testing for CF families is routinely available at most genetics centres and information, where known, about the mutations carried by the affected child can be useful in interpreting a relative's test results as is shown by the following example.

Current practice

The uncle of an affected child and his partner seek testing prior to starting a family. The uncle's partner tests positive but the uncle himself tests negative. This result reduces the uncle's carrier risk from 1 in 2 to about 1 in 7. However, since his partner tested positive, there is still an increased risk (1 in 28) of this couple giving birth to an affected child. The uncle's carrier risk could be modified further with information about the mutations carried by the affected child. If the affected child carries any of the common mutations detected by the DNA test, the uncle's carrier risk would be further reduced to about 1 in 500, and the risk of an affected pregnancy to about 1 in 2000.

Information that the affected child carries a particular rare mutation would clearly be important in selecting further tests for the uncle.

In the absence of a testable mutation, the uncle's carrier risk could be clarified by indirect linked marker analysis, but as discussed above, this would entail analysis of several family members.

3.4.2.2 Clinical cytogenetics

Patients are often referred for chromosome analysis after detection of an abnormality in another family member. In these situations it is extremely important to check that the information supplied relating to the index case is accurate before undertaking analysis of relatives. This may mean requests for information generated by other Regional cytogenetics laboratories

Balanced rearrangements

Rearrangements of chromosomal material occur in some individuals. If a rearrangement is balanced – there is no missing or extra chromosomal material – it will usually have no implications for the individual's health, but could predispose him or her to having a child with a serious mental and physical impairment. Other members of the individual's family may also carry the rearrangement and therefore may also be at risk of having seriously disabled children.

In cases such as this, when family members may have been tested by several different laboratories, it may be necessary to share information regarding the precise nature of the rearrangement. This is particularly true if the rearrangement is very subtle because, in such cases, cytogeneticists need to direct their attention to the relevant regions of the chromosomes. In addition, in calculating the risks of miscarriage and/or the birth of affected children for individuals with balanced chromosome rearrangements, it is often helpful to take into account the clinical and chromosome correlations in other family members.

Fluorescence in-situ hybridisation (FISH)

Fluorescence in-situ hybridisation can be used to detect sub-microscopic chromosome abnormalities. In cases where, for example, a deletion has been detect-

ed in an individual, it may be appropriate to offer testing to other family members. In these cases it is extremely important that information regarding the particular FISH probes used in the initial analysis is available to those performing subsequent testing.

3.5 Genetic 'information'

As outlined in the Preface, in discussions on confidentiality, 'genetic information' is taken to mean any data of clinical relevance to the genetic status of an affected or at risk individual. No distinction is made between, for example, family information arising from a counselling session, and phenotypic observations made during clinical evaluation or laboratory test results. However, as far as consent to sharing information is concerned, individuals may feel differently about the sharing of different types of information. This issue is discussed in section 7.

The guidelines outlined in this document are intended for implementation in genetics centres. As also mentioned in the Preface, once accepted and fine-tuned within clinical genetics, principles could and should be extended to other medical professionals – haematologists for example – who also deal with medical information having family implications.

3.6 The professional community

The relationship between a clinician and his or her patient is based on confidentiality. However, this confidentiality is not absolute. One obvious way in which confidentiality is modified arises from the fact that patient care requires input from a multidisciplinary team of healthcare professionals rather than a single clinician. This is evident in genetics for, as stated above, genetic disorders cut across all medical specialities.

The need for teams of professionals to access patient information is compounded in genetics by the fact that many families are large and geographically dispersed. Different branches of a family may live in different locations and be seen at different regional genetics centres.

If it is accepted that individual patient information may be used for the benefit of other family members, there should be no restriction on the passage of information from professionals in one centre to colleagues in another centre, so long as both parties are confident that the request for information is legitimate. The relationship between two geneticists in different centres is substantially the same as the relationship between a geneticist and his or her colleagues at their own genetics centre.

The restriction on the flow of relevant information from one centre to another should be based only on security of passage; special procedures may need to be put in place if information is transmitted by potentially insecure methods such as fax or e-mail.

3

4

Confidentiality

4.1 The easy situations

Most individuals volunteer to share relevant information for the benefit of their relatives or agree to do so if consent to disclose is sought at the most appropriate point. In other cases, individuals can be encouraged to share information by considerate and patient discussion of the issues.

If consent is given, it is worthwhile using any information that increases the accuracy of diagnosis or risk estimation.

4.2 The difficult situations

In some cases, individuals are less willing to share genetic information with family members. This may be for any number of reasons and usually this wish should be respected.

Dilemmas occur when there is a high likelihood of serious harm to the relative from not knowing this information.

4.3 How conflict may occur

Issues of confidentiality can occur in two distinct ways depending on which individual in an affected family seeks to maintain privacy.

Consider an hypothetical family in which the index case ('A') is a man at risk of developing a serious, late onset condition such as familial adenomatous polyposis coli (FAP – a dominantly inherited colon cancer syndrome) and A's niece ('B') seeks genetic counselling.

Compromising A's confidentiality

B seeks genetic counselling. B's geneticist needs information about A to give B the most accurate information. B is open in her request to A (or his geneticist) but A does not wish the information about his genetic status to be made known.

OR

Compromising B's confidentiality

A has attended genetic counselling and would be happy to have his information shared within his family. B needs this information but does not want A to know of her need (because she is pregnant and does not want him to know of this).

4.4 Consent to share information

If A's consent to share information within his family was obtained at the initial counselling session, the second of the two scenarios above, compromising B's confidentiality, would not occur.

4.4.1 Implied consent

During genetic counselling sessions, the risk to the individual is discussed in the context of the family history of the disorder. It should become apparent to the individual that information about other family members is useful in interpreting his or her own genetic risk. Equally, it should become apparent to the individual

that his or her own genetic information could be important to other members of the family.

Such discussions are based on the implication that sharing genetic information within the family is the 'right' thing to do – in a sense, an implied consent to disclose.

4.4.2 Explicit consent

However, sharing genetic information on the basis of implied consent is unsatisfactory, open to misunderstanding and the individual whose information is being shared may feel a loss of control. Time should be made in the counselling session to explain the issues openly and, wherever possible, to seek explicit and informed consent to share genetic information. This should be recorded in medical notes.

4.4.3 A consent form

One of the recommendations of this report is that consent forms are introduced as a means of formalising and recording explicit consent to share family information. A consent form should be used as a matter of routine when samples are taken and perhaps eventually when an individual is counselled (see 7.4).

Consent forms would formalise current practices of sharing necessary genetic information between professionals in different centres when the family is geographically dispersed.

These issues are discussed in some detail in section 7 of this report, where questions of timing are also considered.

4.5 Refusal to consent

A consequence of explicitly seeking consent is that some individuals will refuse. Such circumstances can raise difficult questions and dilemmas.

4.5.1 Rights and duties

The two most straightforward approaches to resolving these dilemmas can be summarised in conflicting statements of principle, based either on the absolute rights of the individual (the sacrosanct privacy of his or her information) or the absolute rights of the at-risk family members (to be informed of medically relevant genetic information).

Is the professional's obligation owed to the individual or to the wider family? This dilemma is acute when one clinician is in contact with both the index case and the relative. The clinician has to resolve the conflict between duty to the index case (protection of confidentiality) and duty to the relative (not withholding important information).

4.5.2 A middle road – the balance of benefit and harm

Many such dilemmas can be resolved if professionals have the flexibility to tread a middle road between these two approaches. Professionals should have the

Confidentiality

discretion to breach confidentiality according to the following principles:

The right to confidentiality of the index case should be protected when the potential harm caused to the index case by breaching confidentiality outweighs the potential benefits to the relative of being informed.

Conversely, disclosure without the index case's permission could perhaps be justified if the potential harm to the relative of not being informed (and the benefits of being informed), outweighs the potential harm to the index case of confidentiality being broken.

4.6 Breaking confidentiality

In deciding to breach confidentiality, professionals will need to make judgements about the significance of the information – to both patients – and in reaching such judgements, a number of factors will need to be considered. Some of these factors are discussed below, although this is not intended to be a comprehensive list, and some examples are listed in a later section (5.4.2), to illustrate how information can be of use to others. It is worth emphasising once again that the decision to breach confidentiality should not be taken lightly. Wherever possible, professionals should make another effort to convince the patient of the importance of sharing information for the benefit of others.

4.6.1 Factors to consider

1. Knowledge of the individuals' character, disposition, psychiatric health and ethical views

for example, it may be best to maintain confidentiality if there is reason to believe that disclosure would cause the index case a high degree of emotional stress.

2. The situation by which the information was originally obtained

for example, if breaching confidentiality would reveal a teenage pregnancy, non-paternity or other such sensitive circumstances

3. In general, professionals may be influenced towards a decision to breach confidentiality when the knowledge is of significant potential benefit to the relative because:

- the condition is 'serious'
this is subjective but most people would include conditions which result in reduced life-expectancy, serious developmental delay, chronic pain or discomfort or severe behavioural disturbances
- treatment or clinical management of the condition is possible
an example might be inherited colon cancer. Diagnosis by endoscopy and/or DNA testing may be followed by prophylactic colectomy.
- the prior likelihood of being a carrier/affected is high
it might be more appropriate to breach confidentiality in order to inform the sibling of the index case than the cousin or second cousin

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- there is high risk of the birth of a seriously handicapped child

4. In general, professionals may be influenced against a decision to breach confidentiality when the knowledge is of little potential benefit to the relative because:

- no treatment or clinical management is possible
- the condition is mild
- the likelihood of being a carrier/affected is low
- there is a high chance that an affected pregnancy will end with early miscarriage
- the anxiety caused to the relative is likely to outweigh the benefits of being told

4.6.2 Harm to the service

In addition to potential harm either to the index case or the relative, consideration must be given to the damage done to the counsellor's relationship with the index case, that person's trust in other doctors in the future, and public perception of the trustworthiness of geneticists.

4.6.3 Who decides

The decision to break confidentiality will ultimately have to be made by an individual, usually the consultant in charge of the patient's care. But this will often follow discussion between a number of healthcare professionals including genetic counsellors. Occasionally it may be useful to contact GPs. However, such discussions and contacts risk breaching confidentiality in themselves – accordingly, others may need to be consulted on the process rather than the specifics of the case.

If a decision is made to breach confidentiality, a written record must be kept of who was involved in making the decision, and the grounds on which the decision was made.

4.6.4 The opportunity to reconsider

When all the issues have been considered, a decision to breach confidentiality might be agreed amongst a group of suitably qualified professionals. In such circumstances and before the at-risk relative is informed, it may be appropriate to contact the index case to ask them to reconsider.

This would need to be done carefully if the privacy of the relative seeking information is to be protected (as discussed in section 4.3). If the patient still refuses to consent, they should be made aware that it is possible that their information may be divulged against their wishes.

4.6.5 General Medical Council guidelines

Breaking of individual confidentiality in a general sense is permitted within the GMC guidelines, outlined in the booklet *Confidentiality*. Paragraph One includes the following statement: "You must respect requests by patients that information should not be disclosed to third parties, save in exceptional circumstances (for example, where the health or safety of others would otherwise be at serious risk)."

5

The right not to know

The 'right not to know' (unsolicited disclosure) comprises a separate but overlapping set of issues to those of 'confidentiality'. Dilemmas occur in different ways but approaches to resolving conflicts arising from the right not to know are not dissimilar to those suggested above to resolve issues of confidentiality.

5.1 Context – degrees of not knowing

There are two contexts in which a discussion of a 'right not to know' can be discussed. The first is where an individual knows that they have an increased prior risk but reserves the right not to seek extra information. The second is where an individual has no knowledge of prior risk.

These contexts can be illustrated by an example. Consider the adult daughter of a man who died with clinically diagnosed Huntington's disease

The right not to seek additional information

The daughter knows she has a prior risk of 1 in 2 of also developing the disease. She reserves the right not to take a definitive DNA test which will either reduce her risk to zero or predict with absolute certainty that she will develop the disease.

Torleiv Austad argues against an absolute right not to know in such cases since 'A person who says 'no' to important genetic information, and at the same time opposes passing on this information to his/her relatives, also makes a decision concerning them.'¹⁹

However, few would seriously question the daughter's right not to know in the example given above. Indeed this is the basis upon which the genetics services operate.

The right not to be told

The daughter has no knowledge of her father's disease status although her widowed mother does, and this information becomes available to the geneticist. Should the geneticist seek to inform the daughter of her family history?

This context – unsolicited disclosure to someone ignorant of their risk status – forms the basis for discussion in the remainder of this section of the report.

5.2 Rights, prohibitions and obligations

Issues relating to unsolicited disclosure may be addressed from either of two opposing principles: *the right not to know; or the right to be told.*

5.2.1 A right not to know

Is there a right not to know that should limit unsolicited disclosure to an at-risk individual, irrespective of the wishes of other family members?

If there is a right not to know, should professionals who are aware of an individual's genetic risk be prohibited from informing them?

Graham T. Laurie argues that '...an appeal to the concept of privacy can justify the existence of a right not to know one's genetic constitution or, at least, can serve as the basis for making a strong claim to recognise such a right.'²⁰ Based upon this idea, he suggests that the unsolicited disclosure of genetic information is rarely justified. Drafts for an American Privacy Act make a similar point.

5.2.2 A right to be told

If there is no general right not to know, are professionals under an obligation to disclose relevant information?

5.2.3 A conflict of rights

Professionals need to resolve the conflict between the right not to know and the right to be told. By disclosing, the individual has had *the right not to know* taken away from them. Conversely, by making a decision not to disclose, the individual has lost *the right to be told.*

5.2.4 A middle road, discretion to disclose

Professionals should be permitted the discretion to disclose, but without a duty to do so being imposed upon them. Flexibility is required for professionals to tread a middle road. As with issues of confidentiality, discretion to disclose should be based on the principle of maximising benefit and minimising harm.²¹

5.3 Costs and benefits

Some of the general costs and benefits of acquiring medical genetic information have been discussed in section 3 of this report. The same kinds of potential costs and benefits are likely to be derived following unsolicited disclosure.

However, when an individual who is ignorant of a high prior risk is informed 'out of the blue' by a geneticist, there are additional potential costs unique to the situation in which the information was obtained.

5.3.1 Anxiety

The most obvious potential cost is the creation of a great deal of anxiety. This anxiety may be short lived – particularly if the information disclosed leads to a course of action that avoids serious harm to the individual. However, at least in the early period following disclosure, some anxiety is virtually unavoidable.

5.3.2 Knowledge is permanent

Once information has been disclosed, that knowledge cannot be taken back (what has been told cannot be 'untold').

In some cases, the decision to disclose can be delayed until 'the time is right'. Circumstances may change so that the information ceases to be important to the individual and dilemmas regarding whether or not to disclose information are no longer pertinent.

The right not to know

5.4 The decision to disclose

A judgement on the circumstances in which unsolicited disclosure might be appropriate needs to be based on professional consideration of the condition in question and the likely ‘costs’ and ‘benefits’ to the individual of suddenly being made aware of the information.

Professionals, following the presumptions outlined in section 3.2.1, and in accord with the professional practice of encouraging patients to inform relatives of their risk, start from the presumption that disclosure may be appropriate in many cases. Some specific ‘factors to consider’ are listed below. However, inevitably, information on the individual will be limited, and the decision will often be something of a ‘leap in the dark’. Decisions will be based on experience in previous similar cases, but in all cases the burden of proof will lie with the professionals who decide whether or not to make such a disclosure.

5.4.1 Factors to consider

Some of the specific factors which professionals should consider when making a decision whether or not to disclose information to an at-risk individual are essentially similar to those factors which should be considered when making a decision whether or not to breach confidentiality. These are listed in section 4.6.1 (above), but are re-iterated here, as the emphasis may be different for some situations.

1. Knowledge of the individual’s character, disposition, psychiatric health and ethical views
unsolicited disclosure of this nature will almost inevitably cause some distress, therefore this factor must be considered very carefully
2. The situation by which the information was originally obtained (for example, teenage pregnancy, non-paternity etc.)
3. Professionals may be influenced in the direction of disclosing when the knowledge is of significant potential benefit to the individual because:
 - the condition is ‘serious’ and treatment or clinical management is possible
 - the likelihood of being a carrier/affected is high
 - there is high risk of the birth of a seriously handicapped child
 - the benefits are likely to outweigh the anxiety of knowing
4. Professionals may be influenced in the direction of not disclosing (withholding information) when the knowledge is of no significant potential benefit to the individual or may even be harmful because:
 - the condition is mild
 - no treatment or clinical management is possible
 - the likelihood of being a carrier/affected is low
 - there is high chance that an affected pregnancy will end with early miscarriage
 - the anxiety caused is likely to outweigh the benefits of knowing

5.4.2 Examples

Examples can illustrate the judgements that are made in practice; and areas that could fruitfully be researched further.

Balanced translocations

Professionals may consider it appropriate to initiate contact with the relative of a balanced translocation carrier, if the relative were considering starting a family. This might be particularly likely if there were a strong possibility that potential unbalanced translocation products could lead to the birth of a live-born baby with serious disabilities.

Huntington’s disease

Professionals, in general, encourage individuals with a positive test for Huntington’s, or who are at risk for Huntington’s, to inform their relatives that they are also at risk. In part this is based on the broad professional presumption that it is better to know one’s risk status than not to know it. However, another calculation is that in a family context information is likely to come out at some point, so it is better that it is done as soon as possible. Where a relative is out of contact with their family, this second consideration is no longer relevant, and it falls to the professional to determine whether the balance of benefit and harm favours unsolicited disclosure. Opinion is divided on this. We recommend that more formal research be conducted to determine (i) current professional practice, and (ii) the views of individuals who have had information about risk status disclosed to them in the past.

5.4.3 Harm to the service

As with issues of confidentiality, when considering unsolicited disclosure, the potential damage to individual and public perceptions of the genetics service must not be forgotten. The professional may be blamed for inflicting harm by causing stress and anxiety even though they believe they are acting in the best interests of the patient.

5.4.4 Who decides?

As in the case of breaking confidentiality, the decision to disclose will ultimately have to be made by an individual, usually the consultant in charge of the patient’s care. However, in our view, the decision is one of such importance that other professionals involved with the patient’s care should be consulted including, if possible, someone who has had a good deal of contact with the individual concerned.

5.5 Further research

We should always consider such issues as availability of early treatment, other uses to which information can be put – regarding reproductive choices, career choices, etc. – and the amount and type of harm that might be done if information is given and if it is not given. As already suggested, further research is needed to investigate the long term psychosocial impact on individuals who have been given unsolicited information about their genetic risk.²²

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6

Professionals and families working together

Professionals often look to families and patient support groups to spread information, to play a pro-active role where this is needed in the family context. In particular, professionals will want to work with families to address the issues raised in sections 4 and 5.

Problems can arise in families that do not communicate very well. In addition, recent research has shown that the lay concept of ‘family’ and the genetic one are not always the same.²³

Within a flexible framework of discretion to disclose, families and professionals can work together to ensure that best practice prevails. In some circumstances the difficulties and dilemmas are manifold and professionals may need to take the initiative in the best interests of the individual and the family.

The consent form

As outlined in section 4 of this report, explicit consent to the sharing of relevant information within families should be obtained whenever possible. Our recommendation is that UK clinical genetics departments formalise this by adopting the use of consent forms. Such forms are already being used in some Centres.

We recognise that this represents a substantial change in working practice in many Centres. Consent forms would need to be introduced in such a way that the extra burden of information giving and counselling is minimised but at the same time the opportunities to persuade patients to agree to give consent are maximised.

One means of investigating some of the problems which may be encountered and of defining the optimum mechanism for applying consent forms to genetic counselling sessions would be for various centres to undertake pilot studies. Such pilots would be designed to assess patient take-up rates, barriers to consent and other factors. Some of the factors that might be investigated are discussed below.

We recognise that many of the specifics will need to be fine tuned, and we therefore recommend that no changes are made to existing practice until the results of the consent form pilots are evaluated. We also recognise that other issues will need to be considered, or considered in more detail, including the handling of genetic information on minors and the legal situation in relation to patients suffering from dementia.

7.1 Acceptance

In order to be most effective, the entire UK clinical genetics community would need to accept and use consent forms. Raised public, professional and media concerns suggest that these issues be considered with some urgency.

In preparation of this report we considered the consent form developed by South Thames Regional Genetics Centre in relation to the use of blood samples (see Appendix A). Small pilot studies were undertaken in South Thames and in Aberdeen to assess patient views on the appropriateness of securing written consent (see Appendix B).

7.2 Wording and implementation

Without resorting to subterfuge or deceitful wordplay, the phrasing of the consent form is likely to influence whether or not the patient gives consent. The effectiveness of different draft consent forms could be evaluated in pilot studies.

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7.3 Timing

A consent form could be presented to the patient at a number of different points in the counselling process.

7.3.1 Appointment letter

Appointment letters often include information that refers to the 'family' nature of the condition. Including a consent form would be an extension of this.

Seeking a signature at this point may not be appropriate, as this would be only minimally 'informed' consent. Some patients may be put off and refuse to sign the consent or even to attend clinic. However, the patient will be forewarned that a signature for consent will be sought in clinic.

7.3.2 At the start of the counselling session

Presenting the form at the start of the session allows the paperwork to be settled early and the patient is made aware of the issue at the outset.

At this stage, however, full family implications will not yet have been discussed. Patients may be inhibited from open discussion. Again, consent would be only minimally 'informed'.

7.3.3 At the end of the counselling session

At the close of the session all the issues will have been aired and the patient should be informed. The disadvantage is that the patient may feel duped into having divulged information they may not have done if they had realised that it could be made known to relatives. They may withhold consent. Broadly, raising the issue early but seeking consent at the end of the process of consultation would be one option to consider.

Another problem is that the counselling session may have been highly charged emotionally and patients very upset. Techniques need to be developed to 'change gear' in order to establish an appropriate mood in which to present a consent form. Questions of timing could be addressed in further pilot studies.

7.4 Nature of the information

Consideration also needs to be given to the different kinds of information for which consent to sharing is being sought.

The consent form outlined in Appendix A only applies to the results of DNA or other blood tests. Preliminary pilot study data suggest that most patients were happy to consent to this information being used for the benefit of others. This suggests that this aspect of the problem might be relatively straightforward in most cases.

However, during the course of a consultation other family details may be divulged which may have a bearing upon individual risks. This may include

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The consent form

previous investigations or clinical consultations by specialists in fields other than medical genetics. How much of this information is covered by the consent? This aspect of the problem has not been piloted and further consideration of the issues is needed.

7.5 Exclusions

A consent form would formalise existing practice for patients seen in the genetics clinic. However, many referrals for laboratory analysis do not come from clinical geneticists.

7.5.1 adult onset conditions

For adult onset conditions, a specific question on consent to disclosure could be included in the request form. However, request forms are rarely filled-in by geneticists and consent may be only minimally informed, particularly if the patient has dementia and next of kin are unavailable.

It might be better to seek consent after a positive result. Geneticists would need to be involved since they are likely to see other family members.

7.5.2 children with developmental delay

The most common referrals for laboratory analysis are from paediatricians referring children with 'developmental delay'. It would be inappropriate to seek consent to disclose genetic information when samples are obtained from these children. This is because so many of the samples test negative. Again, most referrers are not geneticists.

Consent for disclosure could be discussed with parents when the result is given. However, some may be too upset to take it in. These families should be referred to the genetics clinic, but not all are and of those who are referred not all attend.

7.5.3 banked genetic material

Most genetics centres maintain a DNA bank. Almost without exception, banked DNA samples will have been obtained without disclosure consent. It would be impossible to try to obtain retrospective consent. Samples could remain banked and used in the same way as at present. A long-term decision should await the outcome of further pilot studies.

7.6 Refusal to consent

One consequence of the use of consent forms will be that express refusal to consent to the sharing of genetic information will sometimes arise. One aim of further pilot studies would be to establish what proportion of patients refuse to consent and the reasons for that refusal.

7.7 A time limitation on consent?

In general, there should be no time-limitation on consent. Once given, consent to the sharing of genetic information for the benefit of others should usually be held indefinitely. However, family circumstances may change, and individuals may change their minds. Good professional practice dictates that the professional should revisit the issue; perhaps seek to encourage the individual to reconsider, and if this fails consider a breach of confidentiality in line with the guidelines set out in earlier sections of this report.

7.8 Research and education

Most of these discussions have taken place without a great deal of evidence on attitudes to genetic counselling and testing. We would like to see more research undertaken to find out more about patients' experiences of, and attitudes towards, medical genetics services.

Some of these issues arise because of a lack of awareness of genetics and the fact that genetic information is important to all members of a family. A huge amount of work is required in educating both professionals and the public about genetics and family information. Education is needed at all levels: secondary school; medical and GP training; and especially the media.

7.9 Applications to other Specialities

We have deliberately restricted our discussion to the genetics services and disciplines that come into contact with genetics services. However, we recognise that medical information on an individual that is not explicitly genetic in character, held on record in a medical department other than a genetics department, may have implications for other family members.

Such information may include radiographs held in an X-ray department, medical notes from a previous neurology consultation, for example, or fixed tumour tissue (a potential source of DNA), in a histopathology laboratory.

As we learn more about the genetics of common diseases, this possibility will become increasingly common. It is our hope that the approach taken in this report can lay the basis for an examination of similar issues as they arise in these broader contexts.

Summary and recommendations

Central to this document is the notion that it is often necessary and appropriate to use individual medical-genetic information for the benefit of relatives. Medical genetics in the UK is based on this notion and on professionals using such family information responsibly. This professional practice, although well established, is based on 'common sense' and a collective understanding and consensus view of 'best practice'.

This report aims to support, supplement and formalise existing best practice. We have tried to provide a framework of written guidelines that define both limits to individual confidentiality in the medical genetics context and how issues and conflicts relating to confidential information can best be approached in daily practice.

In attempting to formulate written guidelines, the following recommendations are made (numbers in parentheses refer to numbered passages in the main body of this document).

- In general and wherever possible, individual genetic information should be used for the benefit of family members (section 3).
- In general, individual genetic information should only be used with the explicit consent of the index case (4.4).
- A consent form should be adopted as a means of formalising and recording consent to share family information. These should initially focus on molecular and other laboratory data but eventually might also cover aspects of the counselling session (4.4).
- If consent is given, it is worthwhile using any information that increases the accuracy of diagnosis or risk estimation (3; 4.1).
- Consent forms should facilitate sharing necessary genetic information between professionals in different centres when the family is geographically dispersed (4.4.3).
- Generally, when individuals are unwilling to share genetic information with family members this wish should be respected (4.2).
- Professionals should have the discretion to breach confidentiality according to the following principles (4.5.2; 4.6):

The right to confidentiality of the index case should be protected when the potential harm caused to the index case by breaching confidentiality outweighs the potential benefits to the relative of being informed.

Conversely, disclosure without the index case's permission could perhaps be justified if the potential harm to the relative of not being informed, and the benefits of being informed, outweigh the potential harm to the index case of confidentiality being broken.

- The decision to break confidentiality will ultimately have to be made by an individual, usually the consultant in charge of the patient's care. But this will often follow discussion between a number of healthcare professionals (4.6.3).
- Before breaking confidentiality, it may be appropriate to try to contact the index case to ask them to reconsider their objections (4.6.4).
- Such decisions and the grounds by which they are reached should be documented (4.6.3).
- Professionals should be permitted the discretion to disclose genetic information to an individual who is unaware of their risk status (although this should not be a duty imposed) (5.2.4).
- A decision to do so should be based on the following principle:
The likely benefit of being told outweighs both the potential harm of remaining ignorant and the potential harm and anxiety of being told.
The burden of proof for this lies with the professionals who decide whether or not to make such a disclosure (5.4).
- The decision to disclose will ultimately have to be made by an individual, usually the consultant in charge of the patient's care. However, others should be consulted including, if possible, someone who has had a good deal of contact with the individual concerned (5.4.4).
- Further research is needed on the psychosocial impact on individuals who have been given the unsolicited information about their genetic risk (5.4.2, 5.5).
- A high level of education is needed at all levels: secondary school; medical and GP training; and the media (7.7).

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Appendix A: Consent form to share genetic information

SOUTH THAMES REGIONAL GENETICS CENTRE (EAST)

CONSENT FOR BLOOD STORAGE AND ANALYSIS

PATIENT'S / GUARDIAN'S CONSENT:

A. I,, give consent for a sample of my blood to be analysed for If no relevant test is currently available, my sample will be stored until such time that an appropriate test is developed.

I understand that in developing and standardising new tests it may be necessary to use part of my sample anonymously. If any commercial benefits arise from such studies, I understand that I personally will have no claim to this.

Signed: Date:

B. I would / would not like to be told the results of my test.

C. Results from my test may enable other family members to benefit from genetic testing. I hereby give consent for genetic information that may be relevant to other family members to be made available to their doctor.

Signed: Date:

PRACTITIONERS SECTION:

I,, have explained to the above patient the purpose of obtaining a blood sample for genetic studies.

Signed: Date:

Appendix B: Consent form for pilot study

The following questions were asked of patients attending genetic counselling in the South Thames Region (n=20) and in Aberdeen (n=37).

1. Do you think that written consent for DNA testing is important?
In South Thames 90% said yes; in Aberdeen 70.3% said yes.
2. Do you think that written consent should be obtained for results of DNA tests to be used for the benefit of other relatives?
In South Thames 90% said yes; in Aberdeen 68% said yes.
3. Would you have liked more time to think about the matter?
In South Thames 0% said yes; in Aberdeen 0% said yes.

John Dean (Aberdeen) commented that the use of a consent form does add a noticeable amount of time to the consultation. It is our feeling that the responses to the questions indicate that this is time well spent.

Notes and references

- ¹ For a discussion of this set of issues see Peter S. Harper, 'Research samples from families with genetic diseases: a proposed code of conduct', *BMJ*, 1993, 306: 1391-4; Ellen Wright Clayton et al, 'Informed Consent for Genetic Research on Stored Tissue Samples', *JAMA*, 1995, 22: 1786-92; Bartha Maria Knoppers and Claude Laberg, 'Research and Stored Tissues: Persons as Sources, Samples as Persons?', *JAMA*, 1995, 22: 1806-7; and Philip R. Reilly et al, 'Ethical issues in genetic research: disclosure and informed consent', *nature genetics*, January 1997: 16-20.
- ² Nuffield Council on Bioethics, *Genetic Screening: ethical issues*, London 1993, p. 42.
- ³ Documents which give great weight to individual confidentiality in relation to genetics include: The Genetic Privacy Act (USA: produced by George Annas et al as a part of the ELSI project); The Council of Europe's Bioethics Convention; and The House of Commons Science and Technology Committee Third Report: *Human Genetics: The Science and its Consequences*, London: HMSO, 1995.
- ⁴ For example, surveying professional attitudes in Canada, Dorothy Wertz points out that over the period 1985-1994, one of the significant changes in professional attitudes was an 'increased willingness to preserve client confidentiality if the client refuses disclosure to relatives at genetic risk.' See Dorothy C. Wertz, 'Professional Perspectives', in Timothy A. Caulfield, Patricia L. James and Gerald B. Robertson (eds), *Health Law Journal*, Health Law Institute, University of Alberta, Volume 3, 1995: 128.
- ⁵ Editorial, 'Medical Confidentiality', *Journal of Medical Ethics*, 1984, 10: 3-4.
- ⁶ For discussion of the relevant law, see J. Montgomery, *Health Care Law*, Oxford: Oxford University Press, 1997, ch 11.
- ⁷ Department of Health, *The Protection and Use of Patient Information*, London: Department of Health, 1996.
- ⁸ See e.g. General Medical Council, *Confidentiality*, London: GMC, 1995, and United Kingdom Central Council for Nursing, Midwifery and Health Visiting, *Guidelines for Professional Practice*, London: UKCC, 1996, pp. 26-30.
- ⁹ Department of Health, *The Protection and Use of Patient Information*, London: Department of Health, 1996, Annex A.
- ¹⁰ See J. Montgomery, *Health Care Law*, Oxford: Oxford University Press, 1997, pp. 255-6.
- ¹¹ Public Health Control of Disease Act 1984; Public Health (infectious Diseases) Regulations 1988, SI 1988 No 1546. See J. Montgomery, op. cit., pp. 28-30.
- ¹² See M. Jones, *Medical Negligence*, London: Sweet & Maxwell 2nd ed, 1996, paras 2.77-2.86; A. Grubb & D. Pearl, *Blood Testing, AIDS and DNA Profiling*, Bristol: Family Law, 1990.
- ¹³ See General Medical Council, *Confidentiality*, London: GMC, 1996, para 11.
- ¹⁴ See e.g. R v Stone [1977] 2 All ER 341.
- ¹⁵ Lion Laboratories v Evans [1984] 2 All ER 417, 431.
- ¹⁶ T.L. Beauchamp & J.F. Childress, *Principles of Biomedical Ethics*, New York: Oxford University Press 4th ed., 1994, chapter 5.
- ¹⁷ See for example Josephine Green et al, 'Family Communication and Genetic Counselling: The Case of Hereditary Breast and Ovarian Cancer', *Journal of Genetic Counselling*, 1997, 6, 1: 45-60.
- ¹⁸ W v Egdell [1990] 1 All ER 835. See also Re C [1996] 1 FCR 605.
- ¹⁹ T. Austad, 'The Right not to Know – Worthy of Preservation any Longer? An Ethical Perspective', *Clinical Genetics*, 1996, 50: 85-88
- ²⁰ Graeme T. Laurie, 'The Most Personal Information of All: An appraisal of Genetic Privacy in the Shadow of the Human Genome Project', *International Journal of Law, Policy and the Family*, 1996, 10: 89.
- ²¹ The *Report on Genetics in Canadian Health Care* (1991) argued for a breach of confidentiality under certain circumstances:
 1. reasonable efforts to elicit voluntary consent have failed;
 2. there is a high probability both that harm will occur if the information is withheld and that the disclosed information will actually be used to avert harm;
 3. the harm that identifiable individuals would suffer would be serious;
 4. appropriate precautions are taken to ensure that only genetic information needed for diagnosis and/or treatment of the disease in question is disclosed. (cited in Timothy A. Caulfield, Patricia L. James and Gerald B. Robertson (eds), *Health Law Journal*, Health Law Institute, University of Alberta, Volume 3, 1995: 9).
- ²² A suggested research project might be to study (using in-depth interviews, questionnaires and psychometric tests) individuals from translocation families who were tested at selected Regional Genetic Centres.
- ²³ See Martin Richards, 'Families, kinship and genetics', in Theresa Marteau and Martin Richards (eds), *The troubled helix: social and psychological implications of the new human genetics*, Cambridge: Cambridge University Press, 1996.

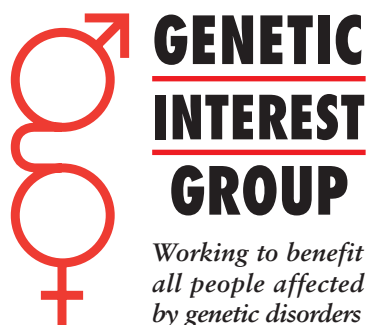
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