

Chapter 5

The results of genetic screening and confidentiality

Introduction

5.1 In this chapter we deal with questions relating to the handling of the results of genetic screening and the safeguarding of the information obtained. These questions are particularly complex in the area of genetic screening, because (as noted in paragraph 4.2) screening may reveal information not only about those who have given their consent to screening, but about members of their families who have not. Genetic screening has to take account not only of the way in which difficult information is to be disclosed to individuals who have been screened, and ways in which the confidentiality of data is to be secured, but also of the interests of family members who have not been screened. Family members may have a strong interest in disclosure of information that is closely relevant to their own genetic make-up, and also in such information being disclosed promptly and in sensitive and effective ways.

Disclosure to the individual

5.2 After undergoing genetic screening, or indeed any other form of testing, an individual should **normally** be fully informed of the results, both positive (ie abnormal) for the disorder being screened for, or negative (ie no defect is found).

5.3 Difficulties can arise when the screening process yields results which are unexpected, unwanted, and have not been covered by consent. For example, a sex chromosome abnormality may be revealed when carrying out prenatal testing for Down's syndrome, or a different inherited disease may show up on a test designed for another purpose. To fail to disclose a serious disease accidentally discovered by testing for which consent had not been explicitly given raises ethical problems. To reveal findings affecting an individual which will not have any clinical implications and may provoke anxiety requires careful individual consideration. Sometimes information may cause distress to the family, although

future decisions about having children could be seriously affected if information is concealed. Unexpected information can present ethical dilemmas for which there are no easy answers, or indeed any correct answers.

- 5.4** Even when the result has been obtained, the sample may be preserved for a number of reasons : to check the results, for future diagnostic needs, including those of the family, and for research purposes. Both the individual and the family may benefit if samples are kept in case a genetic defect could be identified at a later date. Testing of samples as a research procedure may help to improve our understanding of genetic variation and the prevalence of other genetic diseases in the population. Authorisation for such uses should have been obtained when initial consent for screening was given, even when the samples are to be used anonymously, and special care is needed to ensure the confidentiality and security of stored samples.

Disclosure to family members

- 5.5** Our main concern, however, is not with disclosure to the individual, but where the interests of others are concerned. This raises some of the most serious issues in this report. The perceived interests of members of the same family sometimes clash. Such clashes can usually be resolved in careful discussion with experienced professionals. But we need to consider the problems that unhappily do not reach such a resolution. These problems are much more acute for X-linked and autosomal dominant diseases, as illustrated by the example in Fig E.

Fig E

A man diagnosed with a mild form of adrenoleukodystrophy (ALD), an X-linked condition that can be carried by healthy females, did not wish his diagnosis or the genetic implications to be discussed with his family. Seven years later, his niece gave birth to two successive boys who have a more severe form of ALD. The illness only came to light in them when the elder boy started to display symptoms. The mother's sister, the man's other niece, has also given birth to a son subsequently diagnosed with ALD. Both families are bitterly resentful that the medical services did not warn them of their genetic risk.

- 5.6** We have reviewed existing case law, professional guidelines and current academic writing on applying the principle of confidentiality to the special circumstances of information arising from genetic screening that may be vital to the well-being or future of other family members. **In such clearly defined contexts** it may be appropriate to treat those family members as a 'unit' and to place less emphasis on individual patient autonomy. This may not always be feasible, for example where blood relations have lost contact with each other, but even in such cases the individuals being screened should be made aware of the implications for their relations.
- 5.7** We have based our approach on the following general principles:-
- (i) the accepted standards of the confidentiality of medical information should be followed as far as possible;
 - (ii) where the application of such standards might result in grave damage to the interests of other family members, then the health professionals should seek to persuade the individual, if persuasion should be necessary, to allow the disclosure of the genetic information. That task would be eased if it were accepted, and as we have recommended (paragraph 4.29) that genetic screening programmes should include in the information leaflets and the counselling a clear indication that the consequences to the family of genetic information may in some cases make it unfair to confine the information gained solely to the individual who has been screened;
 - (iii) in exceptional circumstances, health professionals might be justified in disclosing genetic information to other family members, despite an individual's desire for confidentiality. For example, confidentiality might justifiably be broken if an individual refused to disclose information which might avoid grave damage to other family members.
- 5.8** We begin by examining the issue of confidentiality, considering in particular why it is important and how it is currently protected by the law and by other means. We then ask whether there are circumstances in which the confidentiality of genetic information ought properly to be overridden to permit disclosure to those interested third parties who, it is sometimes said, have a 'right to know.'

The importance of confidentiality

- 5.9** Article 8(1) of the European Convention on Human Rights provides that “Everyone has the right to respect for his private and family life, his home and his correspondence.” The right to private life, or to privacy, clearly includes the right to be protected from the unwanted publication or disclosure of intimate personal information. Although there is disagreement in this country about the extent to which personal privacy should be protected by law, there appears nevertheless to be widespread acceptance that, to the extent that there is such a thing as a right to privacy, it includes at least “privacy of information, that is the right to determine for oneself how and to what extent information about oneself is communicated to others.”¹
- 5.10** These general principles are particularly important in medicine. Respect for privacy is vital to the doctor/patient relationship. The relationship is one which must be built on trust and confidence if patients are to reveal information essential to the proper diagnosis and treatment of their condition. Yet trust and confidence would soon be shattered if doctors were to fail to respect the confidentiality of intimate personal information. Indeed it has been suggested that this would have unhelpful implications for both public as well as private health: in a High Court decision it was stated that: “In the long run, preservation of confidentiality is the only way of securing public health; otherwise doctors will be discredited as a source of education, for future patients will not come forward if doctors are going to squeal on them.”²
- 5.11** The case for confidentiality in medicine must apply with equal force in the specific area of genetic screening. Individuals agreeing to be screened need to be confident that no personal information about the results will be made available to anyone other than themselves and their medical advisers without their explicit consent. Otherwise people may be reluctant to participate, with damaging implications possibly for themselves, their families, and potentially other third parties. If doctors were to break the confidence relating to genetic information, this would have adverse implications for other areas relating to the care and treatment of the patient. And how could the patient be confident that other medical information might not also be disclosed to a third party?
- 5.12** But, although the right to privacy generally and the confidentiality of personal medical information in particular are of the greatest importance, it does not necessarily follow that both should be wholly unqualified. Article 8(2) of the European Convention on Human Rights provides, for example, that the individual’s right to personal privacy may be overridden by requirements prescribed by law introduced to protect health or morals, or the rights and

freedoms of others. This acknowledgement may be particularly important in the area of genetic screening. Information gained in the course of genetic screening will have implications for other family members which could clearly affect the future conduct of their lives. (The information might also be deemed to be relevant by employers and insurers; these issues are dealt with in Chapters 6 and 7 respectively.)

- 5.13** Here we are concerned particularly with family members who may claim to have a legitimate interest in being informed about the results of genetic screening. The claims may vary in strength. An individual may have an interest in knowing whether a partner or prospective partner is likely to suffer from, for instance, familial colon or breast cancer, or Huntington's disease in the future. But such an interest, while understandable, falls far short of any right to claim knowledge. The emphasis is somewhat different if children with a particular partner are contemplated. For example, a pregnant woman may legitimately want to know the result of the screening test on the father of her child if she herself has had a positive test for the cystic fibrosis or Tay-Sachs gene. A different type of problem may arise with blood relatives where non-disclosure of information might lead to an unnecessary termination, or where a relative, not informed of a high genetic risk, might unknowingly become the parent of a child with a serious genetic disorder (see Fig E on page 42).

Legal protection of genetic information

- 5.14** The confidentiality of medical information is protected by law, first by the common law principles, and secondly by the Data Protection Act 1984. (In the limited circumstances of infertility treatment confidentiality is further protected by the Human Fertilisation and Embryology Act 1990 as amended by the Human Fertilisation and Embryology (Disclosure of Information) Act 1992.)

Breach of confidence

- 5.15** At common law personal information held by health professionals about genetic screening is almost always held in confidence. This means that as a general rule there is no right to disclose the information to a third party without the consent of the person to whom the information relates. There is an exception which allows disclosure of information without the individual's consent where the disclosure is in the public interest. The courts recognise that, while "there is a public interest that confidences should be preserved and protected by the law", in some cases "public

interest may be outweighed by some other countervailing public interest which favours disclosure.”³

- 5.16** In determining whether confidential information may lawfully be disclosed, a court thus has to balance competing considerations, some which will argue in favour of confidentiality and others which will argue in favour of disclosure. It is difficult to know in advance how this balance will be struck in any particular case, but it has been argued that the law “is necessarily vague to take account of the many different situations which might arise.”⁴ In a case where a doctor wished to disclose confidential genetic information to a member of the patient’s family against the individual’s expressed wish, the court would have to balance the public interest in confidentiality against the public interest in enabling individuals to make informed decisions about their health and reproduction.

Data Protection Act 1984

- 5.17** In addition to the common law duty not to disclose confidential information, there is also statutory protection in the Data Protection Act 1984 which applies to genetic information stored on a computer. The Act applies to “information recorded in a form in which it can be processed by equipment operating automatically in response to instructions given for that purpose” (s 1(2)). It seeks to control the storage and use of ‘personal data’, a term defined to mean data relating to a living individual who can be identified from the information (or from that and other information in the possession of the data user) (s 1(3)).
- 5.18** The Act establishes a number of data protection principles and creates a system for the registration and supervision of data users. It also makes provision for the improper disclosure of information. When a data user registers, the entry must include a description of any person or persons to whom it is intended to disclose data. Once registered, the data user must not disclose to anyone who is not described in the entry. These restrictions are qualified in the sense that the information may also be disclosed to the data subject or to another person authorised by the data subject. Disclosure is also permitted if it is “urgently required for preventing injury or other damage to the health of any person or persons.” (s 34(8)).

- 5.19** It is a criminal offence for a data user knowingly or recklessly to disclose information to a third party. The aggrieved individual could also refer the matter to the Data Protection Registrar who is empowered to issue an enforcement notice to the data user directing him or her to comply with the data protection principles. The Registrar may ultimately issue a de-registration notice to a data user who violates the data protection principles. There is no provision in the Act for compensating a data subject who is a victim of the unauthorised disclosure of information by the data user. There may, however, be a right to damages for any loss suffered, arising under the general law of tort. A data subject is, in contrast, expressly entitled to compensation for damage or distress caused by the disclosure of the data without the authority of the data user.

Professional codes of conduct

- 5.20** Apart from the common law and statute, the confidentiality of medical records in general and genetic information in particular is protected by professional rules of conduct governing at least some health professionals. The General Medical Council's guidance states that:

“Patients are entitled to expect that the information about themselves or others which a doctor learns during the course of a medical consultation, investigation or treatment, will remain confidential.”

The General Medical Council's guidance also states, however, that a doctor's duty of confidentiality is not absolute and may be overridden in the public interest. This appears to reflect (but may not be identical with) the legal obligations already considered.

- 5.21** Concern has been expressed that professional obligations of this nature do not govern everyone employed in the health service who may come into contact with confidential genetic information. This concern is misplaced. There are professional codes, similar to those laid down for doctors, for disciplines such as nursing, from whom the majority of genetic counsellors are drawn. Other health service staff are likely to be employed under a contract of service which either expressly or by implication prohibits the disclosure to unauthorised persons of confidential medical (including genetic) information. Breach of any such condition could lead to the dismissal of the employee responsible.

- 5.22** Do additional measures need to be taken to deal specifically with the unauthorised disclosure of genetic information by health service employees? Does genetic information raise any questions of confidentiality which are radically different from those which apply to other sensitive personal medical information? Why should the confidentiality of genetic information be singled out for special treatment beyond that accorded to other medical information about individual patients? We appreciate that there are concerns about confidentiality generally, recently expressed for example in the Report of a Working Group on the Access to Named Data by Management and Administration under the chairmanship of Professor Roy Weir in 1991.⁵ In our view the confidentiality of genetic information is best seen as an aspect of the problem of confidentiality generally. Nevertheless it is a serious issue in the context of genetic screening and, before programmes are set up, the mechanisms for ensuring confidentiality should be defined and secured.

The disclosure of genetic information

- 5.23** Thus the confidentiality of genetic information is protected in a number of ways, involving the common law, statute, professional codes of practice and contracts of employment. But the duty of confidentiality is not absolute. At common law confidential information may be disclosed where it is in the public interest to do so (paragraph 5.16). Under the Data Protection Act 1984, protected information may be disclosed where it is urgently required for preventing injury or damage to the health of any person or persons (paragraph 5.18). And under the General Medical Council's Guidance to doctors confidentiality may be overridden in the public interest (paragraph 5.20).

The ethical dilemmas

- 5.24** We discuss first the responsibility of the individual in resolving the dilemmas and next the role and responsibility of the doctor or other professional adviser. The main ethical dilemma arises from a conflict between the right of the individual to personal privacy on the one hand and the interest of family members to be made fully aware of available information which would play a part in making important life decisions on the other. A balance needs to be struck between the two. A further complicating factor is that some family members may not wish to be presented with the

information. We note that this would become a much more serious problem if widespread screening were introduced for X-linked or autosomal dominant diseases.

The individual's responsibility

- 5.25** The question of responsibility has at least two dimensions in this area. The first is the responsibility of the individual to pass on relevant information to other family members, and the second is the responsibility of the other family members to receive the information. As a starting point, we adopt the view that a person acting responsibly would normally wish to communicate important genetic information to other family members who may have an interest in that information, and that a responsible person would normally wish to receive that information, particularly where it may have a bearing on decisions which he or she may be called upon to take in the future. We are also of the view that the primary responsibility for communicating genetic information to a family member or other third party lies with the individual and not with the doctor who may, however, do this at the request of the person concerned.
- 5.26** The situation regarding family members who may not wish to know can be more difficult. If family members were unaware that a relative had been screened, they would be unable to know whether or not they would wish to be informed about the result. In these circumstances the individual who had been tested would have to take care about the manner in which other family members were informed.
- 5.27** Evidence submitted to us suggests that in practice the withholding of genetic information obtained by a screening procedure from those who may need to know is not a common occurrence, although it does happen from time to time. Some submissions to us raised the possibility of creating a legally enforceable duty on the part of the individual to communicate genetically relevant information to interested family members. Although serious problems can arise as a result of non-disclosure, and certain family members may clearly have a legitimate interest in the information, we do not consider that this should always supersede the individual's right to privacy, whatever the circumstances. We have difficulty in contemplating how any such legal obligation would work and how any legal right of family members (assuming that they could always be identified) could be enforced. In any event, in certain circumstances there may be perfectly good reasons why an individual would not wish to inform family members about the result of a genetic test. For example, a woman who has discovered she is a carrier for Duchenne

muscular dystrophy may not wish at that time to tell her sister who is seven months pregnant.

- 5.28** The best way of ensuring that genetic information is appropriately shared with family members (and occasionally with other third parties) is through the information and counselling procedures that we have discussed in Chapter 4. Although the desirability of sharing information with family members can be emphasised, disclosure ought not to be made a condition of participation in a screening programme. Inevitably some individuals will refuse to allow disclosure and this can present the doctor or health professional with an ethical dilemma.

The doctor's dilemma

- 5.29** Just as we have rejected the suggestion that there should be a legally enforceable duty placed on people who have been screened to inform family members or other third parties about the results, so too we reject the idea that doctors could be placed under a legal duty to reveal information against the wishes of the individual concerned. No such duty is acknowledged by law in this country, though the position may be different elsewhere. The furthest the law appears to go is to recognise that in exceptional and ill-defined cases the doctor may have discretion to disclose genetic information to third parties. This is as far as we believe the law ought to go, although even here we are reluctant to suggest that the wishes of the individual should readily be overruled.
- 5.30** But while we are firmly of the view that privacy and confidentiality should be respected and maintained, we also accept that there may be exceptional circumstances where these might properly be overridden by the doctor. We have in mind here, for example, a case submitted to us in evidence where the information was withheld out of malice. We do not suggest that the wishes of the individual should be overridden only in this type of case. But it does illustrate how exceptional is the type of situation where it may be appropriate and reasonable to subordinate the individual's privacy to the interests of others.
- 5.31** It is impossible to prescribe in advance all the circumstances in which a doctor might properly disclose confidential information to family members. Although it may be helpful to develop guidelines to help the doctor in taking decisions, and to seek clarification of the legal position to ensure that disclosure within the framework of such guidelines can be made within the requirements of the law, the actual decision to disclose can only be made case by case. This imposes a heavy burden of responsibility on the health

professional. Two factors stand out as especially relevant. The first is the consequences of the refusal to share information. There would be a stronger case for overriding individuals' objections where the information would influence a decision having potentially damaging rather than merely inconvenient consequences for other family members. The second is the reason for the individual's refusal to give permission. If it can be determined that the reasons are malicious, the decision may be straightforward. However, if the reason were a fear that the information might yield compromising evidence about paternity, the ethical issues would be quite different. If information about non-paternity were not disclosed, a man who incorrectly believed himself to be the father of a child with a particular genetic status might make the wrong decisions about having children. On the other hand, for the health professional to reveal such information might lead to harm to the woman concerned, not only because of the breach of confidentiality itself, but also because of its impact on the woman's relationship with the man involved. For this dilemma there is no easy answer.

Genetic registers

- 5.32** So far, we have considered the consequences of individual results and their disclosure. In the context of genetic screening, where large numbers of tests are being undertaken, this may be recorded in the form of a genetic register or similar database. Special consideration has therefore to be given to the implications for security of these grouped results.
- 5.33** A register can be defined as a systematic collection of relevant information on a group of individuals. Genetic registers record information on individuals with specific genetic disorders, and may include relatives at risk of developing or transmitting the condition. The information may be recorded by hand, or may be held on computer. Genetic registers may be set up for a variety of reasons, including research on the disorder, the effective provision of services to those on the register, and the systematic offering of genetic counselling to family members. The amount and type of information recorded also varies greatly, as does the presence of identifying details.
- 5.34** A number of general ethical issues concerning genetic registers exist. Here we outline those issues relating to genetic screening. They need to be seen against the background of the following points:-

- (i) a genetic register may be the starting point for genetic screening; for example, the systematic testing of relatives of individuals with fragile X syndrome or Duchenne muscular dystrophy;
- (ii) genetic screening may also be based on a register which is not specifically genetic in its basis; for example, registers of specific cancers or of those with severe learning difficulties; and
- (iii) a genetic register may be the result of a genetic screening programme; for example, a register of carriers for cystic fibrosis or sickle cell disease in a population screened for the purpose.

5.35 Consent of individuals on a register to be screened is clearly essential as stated earlier, but it is also important that individuals know that they are on the register.

5.36 Consent of individuals for long term storage of information resulting from genetic screening has also been emphasised earlier; but should this form the foundation of a genetic register, separate and specific consent should be sought for any subsequent tests or other measures.

5.37 While confidentiality of all medical information is essential, this is particularly the case for genetic registers, which may contain highly sensitive and potentially identifiable data on large numbers of individuals with, or at risk for, serious genetic disorders.

5.38 Computer-based genetic registers are subject to the Data Protection Act, but there is need for additional safeguards for all genetic registers, including storage of information in a safe place and manner, restriction of access to those specifically responsible for the register, and the removal of identifying information when data are used for research purposes.

5.39 This is an important area of concern. In our view the Department of Health should consider with health authorities and the appropriate professional bodies effective arrangements for the preservation of confidentiality, particularly in relation to genetic registers, and should issue the necessary guidance.

Conclusions and recommendations

- 5.40 We regard it as axiomatic that:-
- (i) individuals should normally be fully informed of the results of genetic screening, and in particular of the implications of those results for the family; and
 - (ii) the accepted standards of the confidentiality of medical information should be followed as far as possible.
- 5.41 When genetic screening reveals information that may have serious implications for relatives of those who have been screened, health professionals should explain why the information should be communicated to other family members. **We recommend that in such circumstances health professionals should seek to persuade individuals, if persuasion should be necessary, to allow the disclosure of relevant genetic information to other family members.** They should also seek to ensure that treatment, counselling and other appropriate support are made available to those to whom such unsought information is disclosed.
- 5.42 We note that both the law and professional guidelines provide for exceptional circumstances, when an individual cannot be persuaded to inform family members with a legitimate right to know. In such exceptional circumstances the individual's desire for confidentiality may be overridden. The decision can only be made case by case. **We recommend that the appropriate professional bodies prepare guidelines to help with these difficult decisions.**
- 5.43 **We recommend that the Department of Health should consider with health authorities and the appropriate professional bodies effective arrangements for the preservation of confidentiality, particularly in relation to genetic registers, and should issue the necessary guidance.**