Chapter 4

Providing information and obtaining consent

Introduction

4.1 This chapter considers first the information that people invited for screening need in order to make up their minds whether or not to accept, and the most appropriate way of providing this information. It then discusses the meaning and implications of 'informed consent' in the context of screening. Consent to screening differs in several respects from the consent of an individual undergoing treatment, in particular in the way that families are involved. Informed consent to screening implies that those being screened have thought through the consequences of an abnormal result. These may include an effective therapy, which may however be costly (to the family and/or to the health services), unpleasant, and difficult to sustain over a long period. Where no effective therapy is possible, decisions may be involved about terminating a pregnancy or having children in the future.

4.2 'Family' needs to be understood as covering an extended set of relatives linked by blood ties or by marriage or by both. Members of families may or may not be in close touch. They may live far apart, may be registered with different medical systems, and may sometimes be unaware of the relationship. Nevertheless they may share important genetic traits. Genetic screening may discover information about persons who have neither been screened nor consented to be screened. These points will be particularly important in considering issues of consent, confidentiality and data protection.

4.3 In this chapter the focus is on the ethical aspects of providing screening for genetic defects in day-to-day medical practice. Health services, whether in the general practitioner's surgery or a hospital clinic, are constrained by time and resource limitations that do not apply to most research projects to the same degree. Many of the genetic screening programmes described in Chapter 3 are pilot projects and therefore in the research stage. Research teams may be able to build into their projects ample time for counselling, and to call on the assistance of nurses trained in genetics and other counsellors. Furthermore, only

relatively small numbers of people are involved in most research projects and a high level of support can be offered to such individuals and close members of their families. This may not always be feasible in a normal clinical setting.

4.4 In recent years a number of projects have examined the problems of population screening in a variety of clinical settings for carriers for cystic fibrosis, a serious genetic disease that every year affects about 300 babies born in the UK to parents of Northern European descent. We have drawn on published and unpublished material generated by this work in the discussion which follows.

4.5 We have already drawn attention to some of the differences between a research programme and general clinical practice and we fully appreciate that what is learnt in a research setting is not always easily transferrable into clinical practice. It is also clear that some established programmes have not always followed the ethical principles that we outline, and we have learnt from their difficulties. Our aim in this chapter is to emphasise how screening should be done in the future rather than to dwell on deficiencies in the past.

Information

4.6 The Department of Health's 1990 circular, **A Guide to Consent for Examination or Treatment** is a useful starting point.[1]

> "Patients are entitled to receive sufficient information in a way that they can understand about the proposed treatments, the possible alternatives and any substantial risks, so that they can make a balanced judgement. Patients must be allowed to decide whether they will agree to the treatment, and they may refuse or withdraw consent at any time."

This statement makes four important points relevant to screening:-

(i) those being screened are entitled to receive sufficient information in a way that they can understand about what is proposed;

(ii) they must be made aware of any substantial risks;

(iii) they must be given time to decide whether or not to agree to what is proposed; and

(iv) they must be free to withdraw at any time.

4.7 Screening programmes differ from traditional medical practice in that the process is usually initiated by the health care providers contacting people who are well : these people are being offered the possibility of avoiding detriment to their future health or that of their children. As we have already emphasised, what particularly marks out **genetic** screening are the potential implications for the **family**; in addition, a test result will give the individual tested no certain prediction but rather a range of possibilities that may be quite wide.

4.8 The kinds of information and procedures that people need to help them decide whether or not to be screened for a genetic disorder may be summarised as follows:-

(i) the condition to which the genetic disorder may give rise: how serious is it? how variable is it in its effects? what are the therapeutic options?

(ii) the way in which the disorder is transmitted, ie dominant, recessive and sex-linked mechanisms, and the significance of carrier status;

(iii) the reliability of the screening test, ie the typical rate of false positives and false negatives, and the probability of the development of a serious genetic disease;

(iv) the procedures for informing individuals of the results, negatives (normal) as well as positives (abnormal), and what will be done with the samples;

(v) information about the implications of screening positive (abnormal) for their future and existing children, and for other family members; and

(vi) a warning for pregnant women that genetic screening may reveal unexpected and awkward information, for example about paternity.

It should be made clear precisely what is being screened for at each stage of the screening process. A clear statement of what will be done with the results and with the sample (blood or other bodily fluid) should be provided, and individuals should be able to stipulate that their samples should not be kept.

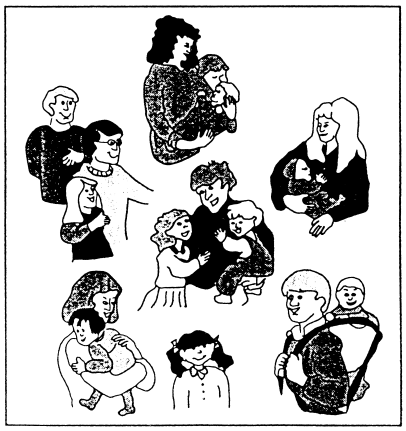
4.9 This information can be provided during a personal consultation, by means of a leaflet, or through some combination of the two. The evidence of the cystic fibrosis screening pilot projects suggests that a combination is desirable. It is important that both written and oral information is in a language appropriate to the individual.[2]

4.10 A number of pilot screening projects for cystic fibrosis recently carried out have addressed with great thoroughness many of the problems of obtaining informed consent. We have therefore drawn extensively on their experience. The leaflet used in approaching people about screening in a London general practice (Fig B)[3] provides answers to the following questions: what is the nature of the disease? what is a carrier? what are the chances that I will be a carrier? what are the chances that I will have a child with the disease? is it important for me to tell my partner? The question "What does it mean to be a carrier?" is explained in another leaflet given to those whose test is positive.

Fig B

Leaflet issued in a London general practice

Have you heard of.....



Cystic Fibrosis Screening ?

This pamphlet contains information about a disease called Cystic Fibrosis.

• In Britain, about 300 babies a year are born with this disease. About one in every twenty people carry the gene for cystic fibrosis. These people do not have cystic fibrosis but are known as *carriers*. If their partner also has the cystic fibrosis gene, the couple might pass on this serious disease to their children.

• There is now a test to detect those people most likely to pass on cystic fibrosis to their children. If you choose to be tested we can tell you your chance of having a baby with cystic fibrosis. We are offering the test at this health centre as part of a research project.

The test is being offered to both men and women aged between 18 and 45.

• This pamphlet is to help you to decide whether you want the test or not. The result of the test will not affect your health at all. The result could prevent you having a baby with cystic fibrosis.

• Your test result will be treated as confidential.

Please read on.....

4.11 A question and answer sheet used in Manchester (Fig C)[4] goes into the issue of informing other family members. It also raises a question about insurance : the comforting answer, though it may be correct for cystic fibrosis and perhaps other recessively inherited disorders, does not apply to all areas of genetic screening. (The ethical issues relating to insurance are discussed in Chapter 7.)

Fig C

Leaflet issued in Manchester and
other North-West general practices

WHAT DOES IT MEAN TO BE A CARRIER OF CYSTIC FIBROSIS?

1.

Question. Will it affect my health now or in the future?

Answer. No. Because you carry two genes - one from each of your parents the normal gene protects you completely from the effects of the gene carrying cystic fibrosis. It is only when both genes carry cystic fibrosis that you could have the symptoms of the disease.

2.

Question. Are my children at risk in any way?

Answer. They would only be at risk if they had inherited the cystic fibrosis gene from both their parents. If your partner is not a carrier then they have a 50/50 chance of being a carrier like you and a 50/50 chance of not being a carrier at all. Children are not routinely tested for carrier status. Probably by the time they are adults and having children themselves the test will be generally available.

3.

Question. Should I tell any other members of my family that I am a carrier?

Answer. This is entirely up to you. It is likely that if your brothers and sisters are having families themselves they might like to have this test done too.

4.

Question. Will it affect my chances of obtaining insurance in the future?

Answer. Categorically no. All insurance companies take advice at the highest medical level. They accept that being a carrier of cystic fibrosis will not affect your health or your life expectancy in any way.

5.

Question. Why do 1 in 20 people carry the cystic fibrosis gene?

Answer. We don't really know the answer to this. We think that it protected us in some way from other serious illnesses or malnutrition in the past.

4.12 Two of the cystic fibrosis screening leaflets we have seen (Edinburgh and Manchester) outline the treatment options, including prenatal diagnosis of the fetus. The Edinburgh leaflet[5] advises women who are unsure about who is the father of their baby to refuse the test. The reason for this is presumably because, as the fetus can only have cystic fibrosis if the father is also a carrier, antenatal diagnosis would not be contemplated without knowing this fact and the mother might be made unduly anxious.

4.13 The voluntary nature of genetic screening is of particular importance. In connection with the Edinburgh trial of screening for cystic fibrosis a follow-up study of the reasons given by the women who declined testing, after having been informed of the nature of the disease and significance of the test, is of interest. Out of the 2207 women who were invited to participate in the trial, 260 refused. They gave a variety of reasons, including a lack of interest in the result, a wish to avoid anxiety about the result and, in just over half the instances, opposition in principle to the termination of pregnancy, either specifically for cystic fibrosis or in any event.[6] This study underlines the importance of avoiding any hint of coercion in genetic screening programmes; it also illustrates the difficult and delicate task of providing adequate information in a non-directional way. A further illustration is provided in the invented examples in Fig D below:

Fig. D

Jane B and her partner decide to be screened for the cystic fibrosis gene. They have read the leaflets and talked with their family doctor. If they are both carriers they have decided to have prenatal diagnosis and terminate the pregnancy if the fetus has cystic fibrosis. They are found to be carriers and the baby is affected. Jane B is now very doubtful about termination and both parents are excited by reports in the press that gene therapy for the disease is being tried. They are glad to have been screened so that their baby can be treated as early as possible after birth, and may be able to benefit from advances in treatment.

Mary S and her partner are in a similar situation but have gone ahead with termination which is done the day before the press reports about gene therapy. They feel cheated and wish they had never consented to being screened.

These invented examples illustrate also the high level of information that may need to be given to couples at risk. Gene therapy is still in the experimental stage and counselling needs to be readily available when 'advances' or 'breakthroughs' are announced.

4.14 Equally critical ethical issues arise from the identification of late onset diseases such as Huntington's disease or Alzheimer's disease. For Huntington's disease the genetic mechanism causing the disease was discovered in March 1993 and a specific test is now available. Is termination of pregnancy an acceptable option when screening for late onset diseases? Future screening programmes will have to provide information on such matters as the definition of what constitutes 'late onset'; the accuracy and the predictive power of the test for a disease-related gene or genes; whether the causal relationship between genetic and environmental factors is well established; and what treatment options are likely to become available.

4.15 We have so far concentrated on information given in programmes in which the defective gene is detected directly and have pointed out that such programmes are at present research based. But, as described in Chapter 3, many programmes of screening for genetic disease by indirect methods have been in operation for a considerable number of years. The way in which information is given and consent obtained for programmes that are part of normal medical care (for example, phenylketonuria, congenital hypothyroidism, Down's syndrome) seem to be very variable (paragraphs 3.14 and 3.15). The Health Education Authority's **Pregnancy Handbook**, available free to all women pregnant for the first time, now has a section which describes the various methods of prenatal diagnosis and some of the conditions that can be detected, and comments on how couples can make use of the information.

4.16 A review of routine screening for Down's syndrome in antenatal care indicates that the information provided is often not adequate and that women are not always sure of what tests they have undergone or what the results mean.[7] In a recent small study of the psychological consequences of screening for Down's syndrome some of the difficulties surrounding the giving of information are highlighted:[8]

> "One woman did not read the information sheet ... assuming it to be about screening for spina bifida."
>
> "One woman believed she was informed when she had the test but when the news came that she had a 1 in 20 chance of having a Down's syndrome baby, she realised she knew nothing."

Counselling and consent

4.17 In most of the research programmes and pilot projects we have considered, written information has been supplemented by counselling. This has been done either in conjunction with giving out a leaflet or by emphasising the availability of a trained counsellor to answer questions and talk through the problems. In two trials of screening for cystic fibrosis in primary care, through general practices in inner and outer London, the take-up of an invitation by letter to be screened, without any counselling or discussion with a doctor, was low, around 10% of the sample.[9] Those approached (both sexes aged 16-44 in one trial and aged 18-45 in the other) were neither pregnant nor known to be contemplating having a child. It may therefore have been lack of interest, rather than informed refusal, that led to the low take-up.

4.18 Follow-up studies in the Edinburgh[5] and Manchester[3] programmes indicate that the implications of the test were well understood by a majority of the participants. They included the recessive character of the defective gene, the fact that the test would not identify all carriers, and the probability of a child of two carriers being born with cystic fibrosis. These are complex matters requiring an understanding of the basic patterns of inheritance and disease transmission, and of risk analysis, and it is encouraging to note that they can be explained, and the information retained for some time, by means of written material plus a brief discussion. The general practitioners in the Manchester trial estimated that cystic fibrosis counselling added about 10 minutes to a normal prenatal consultation.

4.19 The results of the two London trials illustrate one of the problems associated with introducing screening for a genetic disease in a population which has no direct experience of the disease; namely how to convey adequate information to people who do not perceive a need for the knowledge that the test would supply. It is not clear what meaning can be attached to providing information and obtaining informed consent in such circumstances. The take-up among patients approached 'opportunistically' by a member of the trial team when visiting the surgery was much higher, around 70% in the participating practices. The take-up among women and couples approached in family planning clinics was even higher than this at 87%.

4.20 The evidence suggests that written information needs to be supplemented with a face-to-face discussion about the facts and the choices and moral issues that may arise from a positive test result. This is not necessarily so very different from the kind of discussion that a patient may have with his or her general practitioner (or hospital consultant) before agreeing to diagnostic

tests for any potentially serious disease. Screening large numbers will put pressure on staff resources, but the successful introduction of screening for high risk of heart disease and strokes in general practice, using trained practice nurses as counsellors, suggests that the task is manageable. Health care staff would require training in the basic principles of genetics, in the particular characteristics of the disease for which screening is being introduced, and in handling the moral issues, especially in respect of conception and pregnancy. Artificial insemination by donor or egg donation may be an option in certain circumstances, and termination is an option that couples may need to consider carefully if screening takes place during pregnancy. Health care staff may also need training in the best ways of exploring the familial implications of a positive test result. Individuals or couples with positive test results will need further counselling and support, perhaps over a considerable period.

4.21 There is general agreement that counselling at each stage of the screening process should be 'non-directive', as far as possible. In practice a dialogue that helps an individual to explore the facts and issues in the context of his or her particular social and moral background is unlikely to remain completely neutral : experience of genetic counselling suggests that a completely neutral stance can seem cold and unhelpful to some. The key ethical principles of genetic counselling should be:-

(i) the voluntary nature of genetic screening, and the freedom and responsibility of the individual or couple to decide;

(ii) the importance of ensuring that the individual or couple offered screening understand the purpose of the test and the significance of a positive result;

(iii) an assurance of confidentiality in the handling of the results, coupled with an emphasis on the responsibility of individuals with a positive (abnormal) result to inform partners and family members; and

(iv) an appropriate emphasis, at each stage in the screening process, on the fact that consent to screening, or to a subsequent confirmatory test, does not imply consent to any specific treatment, or to the termination of a pregnancy.

It is important that interpreters are available when seeking informed consent from individuals who do not speak English.[10]

4.22 Although the training and supporting of professional staff is clearly essential, there is little empirically based work to guide practice. There is a need to evaluate the effectiveness of different approaches to the provision of information and the obtaining of consent. Such evaluation should be built in to all screening programmes.

Persons requiring special safeguards

4.23 In some cases, it may not be possible to obtain properly informed consent. The testing of the following categories of persons should be subject to special safeguards:-

(i) minors;

(ii) the mentally ill and those with severe learning difficulties.

Minors

4.24 The Family Law Reform Act 1969 permits a person between the ages of 16 and 18 to give consent to medical treatment which is carried out for his or her benefit. Following the decision of the House of Lords in *Gillick v DHSS*[11] a child below the age of 16 may also give valid consent to medical treatment if he or she has the ability fully to understand what is involved in the medical procedure or treatment in question. Otherwise consent must be given by the parents of the child, and may only be given to promote the interests of the child. Under the Children Act 1989 consent may be given by one and is not required to be given by both parents.

4.25 Genetic screening of children which is not of immediate benefit to them should normally be deferred until they can give valid consent. An exception may be where testing of the child is essential for the diagnosis of a family member, though it may be difficult to argue that such testing is always undertaken to promote the interests of the child. The genetic testing of children for late onset/adult diseases raises particularly difficult issues of informed consent and is likely to become increasingly important as a growing number of genes which predispose to disease in later life are being discovered. The child, when adult, may not wish to know the increased risk of developing a disorder. This again is an area where there is no ready answer or right procedure, even for testing within families for a particular disorder. So particular

caution is needed when any wider screening of children for later onset genetic disorders is to be considered.

Mentally-ill and those with severe learning difficulties

4.26 So far as genetic testing of the mentally ill is concerned, the legal position has been held by some to be governed by the House of Lords decision in *F v West Berkshire Health Authority*[12], a case which concerned the sterilisation of a 36-year old woman. If strictly followed, it is not clear that genetic testing could ever be properly conducted on someone who is mentally disabled when the purpose of the test is to benefit a family member or someone other than the person being tested. It is a matter for consideration whether genetic tests on mentally ill individuals or those with severe learning difficulties should be permitted in situations where the information gained would be of clear benefit to other family members. For example, should men with severe learning difficulties be tested for the gene for fragile X syndrome in order to find out whether sisters or nieces might be carriers, and therefore at risk of having boys with the disorder? The benefit to the family could be great and the risk of harm to the individual being tested negligible.

Other vulnerable individuals

4.27 Special care is always needed when consent is being sought from vulnerable individuals, such as prisoners, student volunteers, and (as noted in paragraph 4.21) from individuals who do not speak English.

Conclusions and recommendations

4.28 We do not agree with some recent commentators who have taken the view that so much information is necessary for individuals or couples invited to accept screening for a genetic disease that it is not practicable to obtain truly informed consent at all. Provided that the aim is to provide adequate information, with opportunities for reflection, questioning and further explanation before consent is given, it should be possible to obtain consent in a normal clinical setting. The communication of information is at present likely to be easiest, and best understood, in the context of having children, including preconception and antenatal stages. It should, however, become established outside this framework. We discuss the importance of education in human genetics in Chapter 8 on Public Policy.

4.29 **We recommend that adequately informed consent should be a requirement for all genetic screening programmes.** The voluntary nature of the screening process must be emphasised. Adequate information must be provided for all those being invited to enter a genetic screening programme and should include information about the implications for other family members. Information for all genetic screening programmes is best delivered in both written and oral form.

4.30 **We recommend that counselling should be readily available for those being genetically screened, as well as for those being tested on account of a family history of a genetic disorder.** Counselling should be available at all stages of the screening process. This will require the diffusion of an understanding of genetics (at present mainly confined to genetic counsellors) in particular among those engaged in primary health care. The resource implications, including the need to train large numbers of practice nurses and health visitors in the subject matter and the basic principles of counselling, need to be assessed within the broader context of the expansion and extension of primary care.

4.31 Screening of individuals who are unable to give properly informed consent (minors, the mentally ill and those with severe learning difficulties) require special safeguards (paragraphs 4.24 - 4.26).