Clinical applications of genetic information about mental disorders: ethical and legal issues
Introduction

5.1 In this chapter we consider the range of ethical and legal concerns, outlined in Chapter 1, that may arise for individuals, their families and their physicians when genetic information about mental disorders is sought or used in a clinical context. We begin with a discussion of genetic counselling, since this is often the prelude to, and a component of, any genetic investigation.

Genetic counselling

5.2 Genetic counselling is defined in Chapter 4 (paragraph 4.11). It may be undertaken when individuals are seeking information about a condition which may be inherited or about methods of risk reduction where this is possible: when they are considering having genetic tests; when they are being treated for genetic disorders; or when they are making reproductive decisions. Genetic information creates difficulties in two senses. It may be technically difficult for many people to understand and its implications for an individual's own future and for a family's future may be emotionally difficult to accommodate.

5.3 There are already accepted ethical standards which genetic counselling must meet.1 Those who provide it have responsibilities:

- to ensure that genetic counselling is voluntarily undertaken;
- to provide accessible and accurate information both about patterns of inheritance and about the condition;
- to ensure confidentiality and to explain to those receiving counselling if there are good reasons for them to share the information with other relatives;
- to emphasise at each stage of counselling that consent to counselling or to a genetic test (if available) does not constitute consent to take any advice that is offered, to take any reproductive decision or to terminate a pregnancy.

5.4 For a few conditions, such as Huntington's disease, it is possible to give very precise figures about the risk of occurrence on the basis of a family history (Box 3.2). For complex disorders such as schizophrenia, all that can be offered by genetic counsellors is an estimate of average risk based on studies of families with the condition. Because the common mental disorders involve a variety of genetic and non-genetic causative factors, and are likely to involve variation in several (or many) genes, the contribution to risk of any one susceptibility gene may be small. While counsellors must convey risks accurately they must also make clear the limitations of current scientific knowledge, in particular about the interaction of different environmental and genetic factors.

5.5 Accuracy in genetic counselling is profoundly important where mental disorders are concerned because anyone left with a misleading view of their risk may suffer additional trauma to their personal integrity and additional fear of stigma. Those providing genetic information about mental disorders must bear in mind that many “people with psychiatric problems have low self-esteem and they may conclude that the results of a genetic test confirm their low opinion of themselves.”2 In particular, before embarking on counselling, they must judge carefully whether

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2 Response by Dr Katherine Rimes and Dr Paul Salkovskis, University of Oxford, Department of Psychiatry, to the Working Party's consultation.
providing information might not add to patients’ difficulties. An exaggerated perception of the degree to which genetic influences determine an individual’s health and future is widespread. For individuals with psychiatric problems, as for those with other conditions, “an increased risk result may cause fatalistic attitudes towards their current problems and decrease their motivation to try to resolve their difficulties.” For these reasons counselling about genetic factors making a slight contribution to risk should never be urged on individuals who do not clearly want it.

5.6 In view of these points it might be thought that there is little to be gained from genetic counselling for those in families with complex disorders. However, it seems that some people value the opportunity to learn about and discuss their risks. Since there is often a tendency to perceive risks as higher than the evidence confirms, genetic counselling can be reassuring in some cases. Moreover, a complaint not infrequently made by members of families, where individuals suffer from mental disorders, is that they find it difficult to get clear and accurate information about the possible inheritance of the disorder, and that this information is often not available from general practitioners.

5.7 The impact of genetic counselling on people who are not themselves ill but are in a family with a history of mental illness must also be considered. Genetic counselling has the potential to affect family dynamics adversely and to trigger anxiety and even illness if it involves giving information about an individual’s risk. Stress may also arise if counselling cannot predict a precise level of risk, leaving individuals in a state of uncertainty. There is as yet little precise evidence about the effects of genetic counselling for mental disorders; caution is indicated. The Working Party recommends that research is undertaken to clarify the appropriate aims and outcomes of genetic counselling for mental disorders and to assess the response of individuals and families to counselling. Such research should investigate the expertise and training needed by those undertaking counselling for various conditions and purposes.

5.8 Counsellors must be aware that, in consenting to counselling, individuals and families have not consented to any subsequent course of action. Although it may be impossible to provide wholly non-directive counselling, the aim should be to enable those counselled to make their own decisions at each stage of the process. This may be particularly demanding for genetic counselling involving mental disorders because, as noted, genetic information is cognitively and emotionally demanding and mental disorders are distressing to patients and their families. The mental health charity and service provider MIND, for example, was concerned that genetic counsellors “may have little experience of mental health problems, and see a life with, say, manic depression as necessarily tragic and a ‘burden’.”

Provision of genetic counselling

5.9 If genetic counselling is to be conducted in an ethically acceptable manner, thought needs to be given to its provision. At present, very few centres provide genetic counselling for mental disorders in the UK. This reflects the fact that mental disorders due to mutations in single genes are extremely rare and that, as yet, there is little information about the susceptibility genes associated with the common mental disorders. The future demand and need for genetic information and counselling is difficult to predict but, as more knowledge about genetics becomes available, demand may well increase.
5.10 Analogies with genetic counselling for other disorders may be helpful. Some breast cancer clinics are developing a useful method for coping with increased demand for genetic information and assessing whether specialist counselling is needed. Those referred to a clinic are interviewed over the telephone by a genetic nurse. The individual’s risk is estimated according to guidelines developed at consensus meetings. For most referrals, specialist counselling is not appropriate and a letter is sent to the GP containing the information needed to inform and reassure the patient.

5.11 For the common mental disorders, susceptibility genes are unlikely to increase an individual’s risk to a degree which would merit specialist counselling, at least for the purpose of discussing genetic testing. So, beyond the small number of people with rare, single gene disorders, the need for specialist counselling should be low. If this turns out to be the case, it will be important to balance any inappropriate demand for specialist genetic counselling against other healthcare priorities. There is, however, an ethical obligation to identify the few who genuinely need specialist genetic counselling and to provide any useful information to those who do not. Such information will be most needed by primary healthcare teams which undertake 90% of the care of those with mental disorders. These teams, however, cannot, and should not, be expected to provide specialist counselling or advice. Psychiatric nurses trained in genetic counselling would be well placed to provide a link between primary care teams and genetic clinics offering specialist counselling. For those who do provide specialist counselling, a multidisciplinary approach will be needed, drawing both on clinical geneticists’ expertise in interpreting complex genetic information and counselling for the rare single gene disorders, and on psychiatrists’ experience in diagnosis and care of those with mental disorders. It has been suggested that the basics of genetic counselling could usefully be covered in general professional psychiatric training.

5.12 It is important to take a considered view of the resources available, in genetics, in mental health and in primary care. In 1991, the Royal College of Physicians recommended that there should be two consultant clinical geneticists per million population. Although the number of geneticists has been rising, this target has yet to be met in any centre, despite the steadily increasing demand for genetic counselling services. In 1997, the Royal College of Psychiatrists warned of “the current crisis in mental health services.” There is evidence that some psychiatric patients do not receive basic information even about contraception, or counselling about relationships. In such circumstances, it is unlikely that they will receive information or counselling about genetics, even if it might be of benefit. Finally, a general practitioner (GP) who responded to the consultation considered that GPs lack appropriate premises, equipment and staff and, as such, “general practice is ill-equipped to deal with the challenges of the present day, let alone those that will come with the completion of the human genome project.” It was of concern to many who responded to the consultation that genetic research and services might divert resources from the provision of more immediate help and support for those with mental disorders. Provision for genetic counselling and related services for psychiatric patients should be proportional to the urgency with which they are needed. The Working Party recommends that the British Society for Human Genetics and the Royal Colleges of General Practitioners, Nursing, Psychiatrists and Physicians consider arrangements for the education, training and support both of primary health care teams providing genetic information about mental disorders and of those providing specialist genetic counselling.

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6 The results of the Confidential Enquiry into Counselling for Genetic Disorders will be published in late 1998.
9 Personal response by Dr Robert Lefever to the Working Party’s consultation.
10 For example, responses to the Working Party’s consultation from a Mental Health User Consultant and the Christian Medical Fellowship.
Genetic testing

5.13 One outcome of initial clinical consultation or of genetic counselling, may be that a patient is offered, and chooses, genetic testing. At present genetic tests have been developed only for a small number of diseases. Where genetic tests are available patients may ask a number of questions, which the physician, or where appropriate the genetic counsellor, must seek to answer while keeping to the standards outlined in paragraph 5.3. These include:

- How serious is the disorder in question? How variable is it in its effects? What are the therapeutic options?
- If the test result is adverse, how likely are they to suffer from the relevant disorder? If they do suffer, how severe is it likely to be?
- If the gene mutation or variant is inherited, how likely are their children to suffer from the disorder?
- How reliable is the test?
- How will they be told about test results, and what will be done with the samples after the test?
- Might genetic test results reveal unexpected or embarrassing information, for example about paternity?
- What are the current requirements for disclosure of information to insurers and employers?

5.14 As with counselling, there are two broad categories of mental disorder for which testing may have to take quite different approaches. We shall contrast rare single gene disorders, using Huntington’s disease and early onset Alzheimer’s disease as examples, with conditions for which one gene variant is likely to alter risk only slightly, using late onset Alzheimer’s disease as an example.

5.15 There is now almost a decade of experience of predictive genetic testing for Huntington’s disease, first by linkage and since 1993 by direct testing.11 The number of people seeking testing for Huntington’s disease is far lower than was initially predicted. Studies prior to the identification of the disease gene suggested that about three-quarters of those at risk of inheriting the Huntington’s disease mutation from a parent would seek testing. It was widely believed that the apparent advantages of resolving uncertainty and having a clearer basis for planning lives would make testing the usual choice of family members. In the event probably less than 10% of those with a parent with Huntington’s disease have decided to have counselling about the possibility of a test, the majority apparently preferring the hope that uncertainty preserves. Of those considering testing who are counselled, about two-thirds opt to be tested.12

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11 Craufurd D and Tyler A (1992) Predictive testing for Huntington’s disease: protocol of the UK Huntington’s Prediction Consortium, Journal of Medical Genetics, 29:915–18. The first session covers such matters as Huntington’s disease and its inheritance, reasons for requesting testing and present and future ways of coping with Huntington’s disease. The second session which is held after an interval of several weeks for reflection and perhaps discussion with friends or family members, covers questions arising from the first session, reviews support networks and the practical arrangements for giving results. In addition to a clinical geneticist, a second person, sometimes a genetic counsellor or a psychiatrist with special skills in the area, is present at one or both of the sessions. Assuming the individual decides to proceed with testing there is a briefer session at which results are given and four further follow ups – a telephone contact a week later, a home visit by a counsellor at one month, a further telephone contact at three months, if required, and a clinic visit for relion carriers after a year. (Madigan J. (1996) in Marteau T and Richards M (eds) The Troubled Helix, Cambridge University Press, Cambridge, pp 7–22, provides a description of the process from the perspective of someone seeking testing.)

5.16 For the small self-selecting group who are tested, the benefits of knowing their genetic status seem to outweigh the drawbacks. However, adverse reactions, including periods of depression, have been reported for some of those who received either favourable or unfavourable results. The adverse reactions reported for some of those who are found not to have the mutation have been explained as a kind of survivors' guilt, or as a need to adjust their sense of identity after a long period spent living in the shadow of the disorder.

5.17 Similar points can be made about the rare early onset form of Alzheimer's disease which often develops when people are in their early 50s. Mutations in three different genes have so far been identified in families with this form of Alzheimer's disease. These are dominant and so have a 50% chance of being passed on (Box 3.3). It is likely that other such genes will be identified in the future so that most, if not all, of the very small number of families who carry this early onset form of Alzheimer’s disease can be offered genetic testing. For these families the situation is very similar to that for families with Huntington’s disease. The mutations are highly penetrant so that most of those who carry them will develop Alzheimer’s disease. While, as with Huntington’s disease, there are currently no proven measures for the prevention of Alzheimer’s disease, three licensed drugs are available which may be of benefit in the early stages of Alzheimer’s disease. There are also various life planning steps that individuals may wish to take. Early indications are that very few members of families that carry early onset Alzheimer’s disease wish to have a genetic predictive test.

5.18 An important conclusion from research so far is that reactions to the availability of genetic testing are specific to particular conditions. Different uptake rates for testing and outcomes have been reported for a number of adult onset conditions. These may depend on the perception of the disease and of the distress it may cause, on the age and certainty of onset, on the options for prevention and treatment, and finally on the implications for health care and life insurance. Uptake may also depend on the way testing is offered, for example by letter or in person. This variability shows that as further genetic tests for mental disorders become available, research will be needed into the response of individuals and families to genetic testing for mental disorders.

Genetic testing for susceptibility genes

5.19 Most of the mental disorders considered in this report do not follow the simple Mendelian pattern of inheritance seen if a single gene mutation is associated with a disease. Late onset Alzheimer’s disease (Box 3.3) illustrates the ethical issues which arise. Within populations, the slightly increased average risk of Alzheimer’s disease associated with one, or even two, copies of the apoE4 allele is of limited value for diagnosis or prediction of individual risk for two reasons. First, the alteration in risk is small and second, it is calculated for the whole population, and does not take into account individual genetic and environmental variation. Testing for such genes would produce false positives and negatives and might unnecessarily burden NHS services. Given the very low predictive power of apoE4 tests the Working Party concurs with others that there is no case for testing for apoE4 alleles to provide predictive or diagnostic information for Alzheimer’s disease. It recommends that genetic testing for susceptibility genes providing predictive
or diagnostic input of certainty comparable to, or lower than, that offered by apoE tests for Alzheimer’s disease should be discouraged unless and until the information can be put to effective preventive or therapeutic use. It must be borne in mind that, for many of the common mental disorders, evidence even of weak associations between gene variants and the population occurrence of a disorder has yet to be confirmed. While there may well be public health reasons for determining the frequencies of some genetic variants in populations, such screening should be carried out on an anonymous basis.18

5.20 ApoE testing was first undertaken in the context of the diagnosis and treatment of heart disease since different apoE variants are associated with variation in levels of lipoprotein. Even in the context of heart disease, apoE testing is less useful than more direct biochemical measurement of cholesterol and lipoprotein levels. Nevertheless, the question has been raised whether patients who have undergone apoE testing in connection with the treatment of heart disease, or who may do in the future, should be informed about any slight increase in their risk of Alzheimer’s disease. As susceptibility genes are identified which may influence a number of conditions, it is more likely that genetic testing will reveal additional medical information about a patient (paragraphs 3.11–3.15). The possibility that additional information will be revealed should be discussed with the patient before the test is undertaken. The Working Party recommends that the duty of physicians to discuss and disclose any possible increase in risk revealed by genetic tests for conditions other than that under investigation be considered equivalent to the duty to do so for other, non-genetic, types of information.

Direct marketing of genetic tests

5.21 It has been predicted that the range of genetic test kits marketed directly to the public will increase rapidly over the next five years. These self-test kits, sometimes known as over the counter (OTC) tests, may be marketed by mail order or over the Internet. It has been suggested that while “susceptibility screening may be bad science, it is likely to be excellent business. Screening tests applicable to the general population will hold out promise of enormous profits for those corporations that can develop and patent tests and techniques ahead of their competitors.” 19 These commercial pressures might lead to promotion of susceptibility testing even where this would not be advisable or appropriate. In the UK, the Advisory Committee on Genetic Testing has introduced a voluntary code of practice for directly marketed tests and has recommended that the development of such tests be restricted to those which determine carrier status for inherited recessive disorders where such status carries no significant direct health implications for the carrier individual.20 However, tests for apoE status are already commercially available to the public in the United States and others will follow. The Working Party endorses the position of the Advisory Committee on Genetic Testing, but considers that the present voluntary system of approval is likely to prove unworkable. The Working Party recommends that the Advisory Committee on Genetic Testing monitors the uptake of directly marketed tests and the consequences of their use. If, in the light of such monitoring, adverse consequences become apparent, it recommends that the UK government seeks stronger national or international regulation of directly marketed tests.

Consent and impaired capacity

5.22 As set out in Chapter 1, an important principle in ethics is respect for human beings, their autonomy and dignity. This ethical principle underlies the legal requirement to seek consent prior to any genetic counselling or testing of adults; any invasive procedure undertaken without consent will be illegal. These consent requirements apply to individuals already suffering from a mental disorder as well as to those who may seek counselling or testing to discover if they are at increased risk. In most cases neither current mental disorder nor risk of future mental disorder will impair capacities; even when there is impairment it is often no more than intermittent. The Law Commission has recommended that statutory force should be given to the existing common law presumption that an adult has full legal capacity unless it is shown that he or she does not.21

5.23 Because of the significance attached to consent, the ethical principles have been developed in some detail in law. The law requires that, in determining if a patient has the necessary capacity to decide whether or not to consent to a procedure, the psychiatrist or other responsible medical officer must be satisfied that the patient:

- possesses the capacity to make a choice;
- understands what the procedure is, that somebody has said that he, or she, should have it and why it is being proposed;
- understands in broad terms the nature of the procedure;
- understands the principal benefits and risks of the procedure;
- understands the consequences of not receiving the procedure.22

5.24 Different decisions require different levels of understanding and an individual may be capable of making one decision but not another. This will depend partly on the relative complexity of the issues involved. It has already been argued that decisions about genetics are particularly complex, because of the extensive family involvement and the difficulty of interpreting the implications of findings in a field where genetic influence is generally only one of many (Chapter 1). The level of understanding required for any procedure might, in practice, be expected also to depend in part on its risks and benefits. The greater the potential benefits and the less the risk of harm, the more flexibility might be allowed in relation to an individual’s consent; the less the benefit and the greater the risk, the more stringent the requirements should be.

5.25 Even for individuals able to give consent, fully informed consent is an unattainable ideal. “The ethically significant requirement is not that consent be complete but that it be genuine.”23 Obtaining genuine consent requires health care professionals to do their best to communicate accurately and in an understandable and appropriate way the purposes and implications of the procedure as well as its risks. They should respect the limits of individuals’ understanding and ability to deal with difficult information, and allow time for them to ask questions. It may be helpful for consent to be sought in the presence of another person - perhaps, in the case of

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21 The Law Commission (1995) Mental Incapacity, Law Com No 231, HMSO, London. As regards children (those under 18) it may be that the legal presumption is somewhat weaker for those aged 16–18, even though, where medical treatment is involved, they are to be regarded as if they are adults (LR) Family Law Reform Act 1969) and there are other contexts in which they may be regarded as having the necessary capacity to give valid consent. As regards children under 16, though they may have the necessary capacity to consent (see Gillick v West Norfolk and Wisbech Area Health Authority (1986) 3 All ER 402) the presumption should, perhaps, be reversed.


someone with a mental disorder, their key worker – so that the individual feels supported and any questions or concerns that arise can more easily be addressed. It can be helpful to offer leaflets or other written information presented in a clear, balanced and non-technical way with translations and interpreters available where English is not the first language.

5.26 For an adult person deemed mentally incompetent to make his or her own treatment decisions, a doctor must act in that patient’s ‘best interests’. There are difficulties, however, in translating from the general principle to the specific case. ‘Best interests’ may vary according to the nature and degree of certainty of the information, the person’s capacity for understanding and acting on the information and his or her wishes, social and family context and needs. Often ‘best interests’ can only be determined after prolonged consultation, and even then a certain amount of subjective judgement may be involved. However, unless it is necessary for there to be an application to the Courts, ‘best interests’ are a matter for the judgement of the appropriate doctor or other responsible health authority.

5.27 The Mental Health Act 1983 does not refer to genetic testing with or without consent. It follows, therefore, that genetic testing of patients without consent who are subject to that Act is only permissible if the testing forms a part of, or is itself, therapeutic treatment authorised by the patient’s responsible medical officer (see also paragraph 7.13).

The genetic testing of children

5.28 By section 8(1) of the Family Law Reform Act a child between the ages of 16 and 18 may give valid consent to treatment as if he or she were an adult, provided, of course, that he or she is otherwise competent. Furthermore, a child below the age of 16 may also give valid consent to medical treatment if able fully to understand what is involved in the proposed medical treatment or procedure. The emphasis in both situations is on treatment, thus the issues are comparable to those raised by the genetic testing of adults in circumstances in which testing contributes to treatment. It is probable that only diagnostic testing and perhaps, very rarely, carrier detection would be so regarded.

5.29 Different issues arise when the testing of children is proposed for purposes other than diagnosis or treatment. There are difficult lines to be drawn where children are concerned, particularly where the child is considered to be competent to make a range of decisions but the wishes of the child and parent/guardian do not coincide. For example, parents may want the child to be tested to resolve uncertainty, although they know that there is no treatment (paragraph 5.31). The child, although competent, may disagree, or not even be consulted. Older children may wish to be tested on their own initiative, for similar reasons, but their parents may object. It has been said the parental right yields to the child’s right to make his or her own decisions when he or she reaches a sufficient understanding and intelligence to be capable of making up his or her own

24 In the case of an incompetent child, the doctor can ordinarily look to the parents to establish the child’s best interests. Once again, these ethical principles find more detailed expression in law. See, for example, Re F (mental patient: sterilisation) [1990] 2 AC 1; Airedale National Health Service Trust v. Bland (1993) 1 All ER 821 (HL) and L v. Bournewood NHS Trust [HL], The Times, 30 June 1998.

25 For recent guidelines laid down by the Court of Appeal, see St George’s Healthcare NHS Trust v. S (No. 2), The Times, 3 August 1998.

26 Following the decision of the House of Lords in Gillick v. West Norfolk and Wisbech Area Health Authority (1981) 3 All ER 402.

mind on the matter requiring decision. However, testing of the kind under discussion here may fall in a novel category raising such complex issues of benefit and possible harm that additional caution should be exercised before leaving the decision solely to the child (particularly if the child is below the age of 16). On the present state of the authorities it is unclear whether, in the case of children under the age of 16, they would be regarded as capable of giving reliable, valid, consent to testing which is of no diagnostic benefit and cannot be categorised as treatment.

Diagnostic testing

5.30 The law permits the testing of children unable to consent only when it is in the child’s best interests. When effective interventions are available, the issues raised by genetic testing are not, in principle, different from those related to any kind of medical test or treatment which involve issues of consent and understanding. With rare exceptions such as phenylketonuria (Box 3.1) effective interventions for mental disorders in children (most commonly mental retardation) are not available. Nevertheless, the use of genetic tests to help establish a diagnosis may be viewed as being in the child’s best interests since a firm diagnosis will enable a clearer prognosis and management plan for the child. It may also benefit the parents (but not the child) by relieving uncertainty and providing information which they can use in deciding whether to have further children and in some situations the child’s interests might best be served by permitting testing to benefit the family as a whole.

5.31 In the context of screening, the ethical arguments are more finely balanced (see also paragraphs 6.32–6.35). For example, diagnostic screening for fragile X syndrome in children with mental retardation is feasible. But a positive diagnosis may have limited management implications for the child, whilst the genetic implications for the family will not necessarily be welcome if the information is unexpected. We would emphasise the importance of obtaining fully informed consent from the family unit, if consent from the child is not possible, before diagnostic testing occurs.

Predictive testing

5.32 Where genetic tests offer some degree of predictive certainty, professional opinion amongst clinical geneticists has been against the testing of children for adult onset conditions, on the grounds that this has no benefit for the individual during childhood, denies him or her the chance of making their own choice as an adult, and could lead to discrimination within the family. Some parents and patient groups have argued, to the contrary, that parents have a right to know about their children’s genetic make-up and, in the case of Huntington’s disease, that they would

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28 See Lord Scarman in Gillick v. West Norfolk and Wisbech Area Health Authority [1985] 3 All ER 402, and contrast, for example, Lord Donaldson MR in In re R [1982] Fam. 11.
29 See the views of the Law Commission referred to at footnote 21 above. If a child, whether or not below 16, is considered as having a right to know his or her own genetic make-up, then the answer to the problem may be simpler – that, if the child is otherwise competent and is capable of understanding the information, it should not, perhaps cannot, be prevented from agreeing to be tested.
rather know than have to live with the uncertainty of not knowing if their children had inherited the disorder.\(^3\) It has also been suggested that parents may want to use testing to ensure that they have at least some children who are free of the disease. Given that the great majority of adults at risk of Huntington’s disease choose not to be tested, however, it is hard from an ethical point of view to justify parents’ requests to have their children tested. It would be even more difficult to do so for tests for the common mental disorders which are likely to offer less predictive certainty. Moreover, whatever the ethical arguments, such testing if not carried out explicitly to serve the best interests of the child, would not be permissible in law. The Working Party recommends that, for children unable to give consent, predictive genetic testing should be strongly discouraged unless there are implications for clinical intervention in childhood. This would include situations in which a child is currently asymptomatic for a disorder which may begin in childhood and for which there may be a family history.

### Carrier detection

5.33 Carrier detection tests for young children are sometimes proposed when an affected sibling is diagnosed. An example is genetic testing of healthy girls who have siblings with fragile X syndrome. It is sometimes suggested that early carrier tests are helpful to children who can then have the implications explained progressively and as appropriate through childhood so that they are well prepared before they need to make any choices about partners or reproduction; for some children this may be earlier than 16. However, the Working Party considers that, as with childhood predictive testing, this denies children the possibility of making their own decisions at a later date. The Working Party recommends that children should not be tested for carrier status for mental, or indeed other, disorders until they are competent to make their own decisions.

### Directly marketed tests

5.34 It may be very difficult to ensure that children are being tested out of concern for their best interests if genetic tests are marketed directly to the public. Guidance from the Advisory Committee on Genetic Testing suggests that testing should not be offered to those under the age of 16 and that persons under the age of 16 should not be tested presymptomatically for adult-onset conditions for which there are no clinical treatments.\(^4\) It is not clear, however, how a company would determine whether a sample had in fact come from a child. This difficulty adds to the reasons for monitoring the uptake of directly marketed tests (paragraph 5.21).

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Adoption

5.35 Genetic testing of children might also be considered during adoption. Placing children born to parents with mental disorders for adoption is not uncommon since severe mental disorders may be a reason for a parent to give up a child for adoption voluntarily or as a result of a Court Order. Requests to perform genetic tests before children are adopted have already been encountered in other contexts: the Tuberous Sclerosis Association has received inquiries about genetic tests from adoption agencies with the purpose of informing prospective parents if there is a possibility that a baby available for adoption will inherit tuberous sclerosis.\textsuperscript{35} The stigma associated with mental disorders might encourage prospective parents to insist on testing.

5.36 Adoption regulations require that all children have medical examinations before they are adopted: the issue is what those examinations are for and what, therefore, they should contain. They were originally designed to ensure that the child was ‘adoptable’: prospective adoptive parents were assumed to want healthy babies with no recognisable liability to illness. Nowadays, all children are regarded as potentially adoptable. The law would once again insist that a test may only be carried out on a child incapable of giving consent if it can be shown to be in the child’s best interests to do so. But it is not in a child’s best interests to be adopted if there is a risk that he or she will later be rejected because the adoptive parents had an incomplete understanding of the child they were adopting. Most good adoption agencies would probably want to address the issue of mental illness in the birth family, just as they would want to address issues of physical disability, HIV status, sexual abuse or any other matter which might impact upon the full integration of that child into the adoptive family. In very rare situations, this might involve genetic testing of a child. Indeed, agencies may now run the risk of being sued by the adoptive parents if they do not properly inform them about the child they are adopting. The Working Party recommends that, given the importance and complexity of the issues, the Health Departments, in consultation with the appropriate professional bodies, provide guidance on the pre-adoption use of genetic testing.

5.37 It is also worth considering whether an adoptive child should have access to information about possible family histories of disease so that, from early adulthood, they may make informed decisions about seeking genetic counselling or testing or other forms of investigation or treatment. It would seem unfair to deprive adoptive parents and adopted children of information about family histories of disease which would be available to birth parents and their children. At 18 years of age, adopted children may ask to know the identity of their birth parents and this might be an appropriate time at which to provide this kind of additional information.

Genetic information and reproductive decisions

5.38 One of the most important uses of genetic information is to inform reproductive choices. Many people with mental health problems, or a family history of them, have chosen to have children and have not encountered any difficulties. But reproductive choices may be complicated even when there is no relevant genetic information. The consultation responses indicated that, for some people, decisions about whether to marry or have children had been influenced by what they knew of their family history. One woman, who had helped to care for her brother since he was diagnosed as having schizophrenia forty years ago wrote that "I have led a reasonably

\textsuperscript{35} Response by the Tuberous Sclerosis Association to the Working Party’s consultation. Tuberous sclerosis is a rare single gene disorder which results in abnormal tissue growth. The symptoms, and their severity, vary but can include autism, seizures, learning difficulties and early death.
healthy, normal (but not married and no children - mental health considerations may have entered into this) and successful life." For others the decision had either not been easy or had been one which they felt they had to justify. Some had been subject to pressure from others about the choice they should make. One respondent to the consultation wrote "I am personally very glad that accurate genetic counselling was not available when I was pregnant with any of my three children. Medical reasoning at the time seemed to be along the lines of 'Well, you could be carrying another schizophrenic: and, even if you aren't, you'll never be able to be a fit mother anyway." Many respondents referred to the history of eugenic abuse and expressed concern about the possible use of genetic information for eugenic purposes.

5.39 Genetic information is used by those making reproductive choices in three main ways: before they marry or enter long-term relationships (prenuptially), before they have children ('preconceptionally') or during pregnancy (prenatally). An example of prenuptial decision-making is the requirement that Cypriot couples take carrier tests for thalassaemia before they can marry in the Orthodox church. Another example is found in some Jewish communities in which the results of carrier tests for Tay–Sachs disease are available only to matchmakers. This allows young carriers to avoid entering relationships with other carriers and, at the same time, to avoid the stigma and damaged self-esteem that can be associated with knowledge of carrier status. Although this consideration may be particularly important in mental disorders where stigma is especially great, the circumstances under which pre-nuptial testing might be employed appear to be very limited.

5.40 Prenatal genetic testing may lead to information which bears on a decision to seek abortion. The starting point for consideration of the option of abortion must be the Abortion Act 1967, including S.1(1)(d) which provides that an abortion may be carried out where there is a substantial risk that if the child were born it would suffer from such physical and mental abnormalities as to be seriously handicapped. Experience to date of prenatal genetic testing indicates that decisions to abort for these reasons are not made lightly. Where a test can reveal with some certainty the presence of a severe, early onset disorder for which no treatment is known, genetic testing and abortion may be accepted by many parents. In principle, this would apply to mental, as well as physical, disorders. However, when even one of these factors is missing, the relevance and acceptability of prenatal genetic testing is lower. In effect, this means that, for the common mental disorders, prenatal genetic testing and termination will be less likely to meet the criteria of the Abortion Act as well as being less acceptable and accepted by parents. Table 5.1 provides details of some common single gene conditions for which prenatal testing may be offered. Most of the mental disorders considered in this report are not associated with mutations of a single gene, but with more weakly predictive susceptibility genes. For the reasons already given, genetic testing for predictive reasons for these conditions is unhelpful at present and such testing is not offered prenatally (paragraph 5.19).

36 An individual’s response to the Working Party’s consultation.
37 An individual’s response to the Working Party’s consultation.
38 Including the Christian Medical Fellowship, Oxford Hearing Voices Group and several individual respondents to the Working Party’s consultation.
Table 5.1: Factors affecting the uptake of prenatal testing for different single gene conditions

<table>
<thead>
<tr>
<th>Disease</th>
<th>Age of onset</th>
<th>Severity</th>
<th>Treatable?</th>
<th>Predictive certainty of test (% of people developing condition with adverse result)</th>
<th>Uptake of test</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tay-Sachs</td>
<td>From birth</td>
<td>Fatal</td>
<td>No</td>
<td>100%</td>
<td>High levels of uptake in counselled couples in certain communities; many do not seek counseling.</td>
</tr>
<tr>
<td>Huntington’s</td>
<td>Middle age</td>
<td>Fatal</td>
<td>No</td>
<td>100% but some variation in age of onset (see Box 3.2)</td>
<td>18% uptake in counselled couples; vary low overall.</td>
</tr>
<tr>
<td>Phenylketonuria</td>
<td>From birth</td>
<td>Severe</td>
<td>No</td>
<td>100% (see Box 3.1)</td>
<td>All testing is neonatal; National screening programme.</td>
</tr>
<tr>
<td>Neurofibromatosis</td>
<td>Childhood</td>
<td>Variable</td>
<td>Some</td>
<td>100% for NF1 but severity varies</td>
<td>1/80 familial</td>
</tr>
<tr>
<td>Fragile X</td>
<td>From birth</td>
<td>Variable</td>
<td>No</td>
<td>Prognosis may depend on the mutation present</td>
<td>Some at least but epidemiological data unavailable.</td>
</tr>
<tr>
<td>Early onset Alzheimer’s disease</td>
<td>Middle age</td>
<td>Severe</td>
<td>Potential treatments currently being explored.</td>
<td>100% for the APP or presenilin 1 or 2 genes (see Box 3.3)</td>
<td>Low uptake of predictive testing for early onset forms.</td>
</tr>
</tbody>
</table>

5.41 Genetic information will not be particularly helpful in making reproductive decisions for many with a family history of a common mental disorder. The reasons underlying reproductive decisions are always varied and personal. Some people are reluctant to terminate fetuses because they may share the condition that affects a parent or other members of the family. This may be one reason for the very limited use of prenatal diagnosis and abortion in Huntington’s disease. One respondent to the consultation wrote: “Schizophrenia has become a part of me – it defines who I am. . . I cannot divide my experiences into illness and health – they all feel to be part of me. Therefore when I hear of moves to try to eradicate schizophrenia it feels like an attack on my status as a full human being.” While prenatal testing for schizophrenia or any of the other common mental disorders is not possible at present, or likely in the near future, this comment captures a common view. Optimism about future treatments or cures may also contribute to a

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39 Predictive certainty depends both on the amount of risk increases, and on variability in prognosis.
43 Personal communication. Professor Daniel Pollen, University of Massachusetts Medical Centre.
44 An individual’s response to the Working Party’s consultation.
reluctance to undergo prenatal testing and abortion. Yet some people are anxious that their children should not have to suffer a disease that they, or other family members, have suffered and several studies have found that parents are more likely to countenance prenatal testing and abortion for mental, rather than physical, disability. For those reasons the Working Party recommends that people making reproductive decisions in the light of a family history of a mental disorder should have access to genetic counselling.

5.42 In practice, there is evidence that some decisions about abortion may be made without adequate and impartial information. Pregnant women may be led to think that they would not be offered a test unless there was a clear and proven benefit; they may not have appreciated at the time of testing that abortion would be the only intervention available; they may take tests in order to gain reassurance and not have been adequately prepared for an adverse result. If an adverse test result is received, they may feel that abortion is expected and the only course of action. Indeed, in one study, over one third of a sample of obstetricians said that they generally require a woman to agree to termination of an affected pregnancy before offering prenatal diagnosis. There is certainly a widespread impression among many of those who counsel women that, in choosing to abort a fetus that has been diagnosed with an abnormality amounting to a serious handicap, a parent is making the obvious choice. It is essential that access to genetic tests is not tied to conditions which might prejudice individuals' abilities to accept or refuse tests, such as willingness to consider abortion as a condition of prenatal genetic testing. That said, pregnant women should certainly be alerted to the risks associated with fetal genetic tests which they may prefer to avoid if they do not wish to consider abortion in the light of an adverse test result and no other intervention is available. They should also be aware that, if they do have such a test, any child born will not have the option of deciding not to be tested.

5.43 In their consultation response, MIND argued that there may be social pressure on parents not to burden themselves or society with 'affected' children and emphasised that real reproductive choice will "necessitate political and social commitment to providing opportunities (like the chance to work), and support where necessary for people who do develop mental health problems." In this context, government measures to integrate health and social service initiatives in tackling mental health are welcome but, even if social provision were to improve far beyond what is currently available, parents and their affected children will still face problems that cannot be alleviated by social means. In the face of these considerations, it has been argued that some parents may not be in a position to make independent reproductive choices.

5.44 The ideal of 'non-directiveness' in genetic counselling has been widely endorsed and the failure to meet this ideal equally lamented. The Working Party questioned the clarity and feasibility of 'non-directiveness' as a universal aim and noted the importance of enabling individuals to make their own informed decisions at each stage of the process. These important issues will need to be included in the consideration of education, training and support for those providing genetic information and counselling recommended in paragraph 5.12. The adequacy of genetic counselling also has to be judged against the realities of situations in which termination decisions

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have to be made quickly, in the light of difficult information and emotionally fraught circumstances. The available evidence indicates that, in any case, reproductive intentions are seldom changed by genetic counselling and the main outcome seems to be that couples feel confirmed in whatever they planned to do beforehand. The Working Party notes the need for further debate about the appropriateness of non-directiveness in genetic counselling and recommends that further research to establish appropriate aims and outcomes for genetic counselling is undertaken.

Eugenic programmes

5.45 There have been concerns that the growing deployment of new genetic technologies will lead to a ‘new eugenics’. Professor Peter Harper, for example, has pointed out that the existence of genetic predictive tests and the feasibility of keeping computerised genetic registers could provide the information required for serious abuse and has argued that at “a time when psychiatric and behavioural genetics are again entering controversial areas, everyone involved should be fully aware of the long shadow that is still cast by the abuse of genetics in these disorders.”

5.46 In the first three decades of this century eugenic programmes were set up in many industrialised countries. In both Britain and the United States programmes were targeted at those with mental handicaps and criminal behaviour; they also covered many with the mental disorders that are the subject of this report. In Britain there was legislation allowing the confining of individuals in institutions for reasons that some would regard as eugenic, while in the United States and elsewhere there were extensive programmes of compulsory sterilisation.

5.47 In Nazi Germany, Huntington’s disease was specifically listed as one of the nine categories of disorder suitable for compulsory sterilisation under the German law of 1933. It has been suggested that there could have been 3,000-3,500 sterilisations of those from families with Huntington’s disease. Later there were countless killings of persons with mental handicaps and psychiatric conditions including schizophrenia.

5.48 Eugenic programmes were originally designed to change the genetic characteristics of a population either by preventing or discouraging those with the (inherited) characteristics held to be undesirable from having children or by encouraging those with characteristics held to be desirable to have more children. Subsequently, eugenic programmes in many countries had compulsory elements and a degree of coercion or the restriction of individual choices.

5.49 A number of distinct issues underlie concerns about the possibility of a new eugenics. There is a possible cause for concern over the development of genetic registers and research studies of the general population where DNA samples are collected. Genetic registers have been set up to collect information about individuals and families who carry particular genetic disorders both for

54 Kevles D (1985), In the Name of Eugenics, Knopf, New York.
57 Meyer J (1988), The fate of the mentally ill in Germany during the Third Reich, Psychological Medicine 18:3108-14.
research purposes and to target specialist genetic services. Such registers, where they are kept on a computer, are regulated in Britain by the Data Protection Act which requires their registration and allows access by individuals to their recorded information. Clearly, release of such information to third parties could be very damaging to the individual concerned. Other problems may arise when an individual who has set up and maintained a register moves to a new position and there may be no one to take over responsibility for it. Clear guidelines are required for the establishment and maintenance of registers whether or not they are kept on computers. These could usefully draw on the principles set out in the recent Report on the Review of Patient-Identifiable Information.58 The Working Party recommends that the British Society for Human Genetics explores mechanisms for the development of guidelines for the establishment and maintenance of genetic registers in the new NHS.

5.50 Concerns have been raised about the existence of genetic testing, and the provision of genetic services more generally, for conditions that some regard as differences rather than disabilities. In the case of achondroplasia (an inherited condition with short stature) for example, disabled rights groups have argued that the existence of genetic testing is intrinsically eugenic and medicalises a socially constructed disability. Clearly, such issues could potentially arise in the field of mental disorders, though this seems relatively unlikely given the limited role for genetic testing in this field.

5.51 Individual choices about having children can alter gene frequencies in future populations. If those who carry mutations related to a dominantly inherited late onset condition such as Huntington’s disease restrict their own reproduction or use techniques (prenatal diagnosis and abortion or preimplantation diagnosis) in order to avoid producing children with the mutation, those mutations will become rarer in future generations. We see no reason not to welcome such reductions.

5.52 For recessively inherited conditions, current practice may limit the numbers of affected children born, but not of those who are carriers. Hence, such practice ensures the continuation of the current frequencies of carriers in populations. Indeed, there may be benefits from continued genetic diversity in the population for conditions where carriers have selective advantage in certain situations. For example, resistance to malaria is found for carriers of some inherited blood disorders. In the case of common polymorphisms such as the susceptibility genes for some complex disorders, there may well be population advantages for the existing genetic diversity. Assessment of any potential testing programme for such susceptibility genes should include public health considerations.

5.53 Cost-benefit arguments are widely used in the evaluation of medical interventions. In the case of genetic screening programmes, the cost of the programmes may be set against the potential economic savings in the reductions of births of individuals who may require extensive medical and social services. In a rationed health care system there are likely to be pressures to deploy screening programmes which may result in cost savings for services. But as we have seen, the scope for genetic screening programmes related to mental disorders is limited. However, such a programme has been advocated for fragile X syndrome. Whilst a programme designed to provide choice for parents might, through their collective actions, reduce the number of children born with fragile X, in itself such a programme would not be eugenic as its aim would be to provide choice.
5.54 Again, with regard to pregnancy screening for Down's syndrome, the intention is to provide the possibility of choice for parents. If parents exercise the choice not to proceed with a pregnancy when a fetus with Down's syndrome is detected, costs may be saved, but it is important not to conflate arguments about financial savings with eugenic intentions. One study has shown, however, that a small minority (13%) of a sample of obstetricians agreed with the statement, 'The state should not be expected to pay for the specialised care of a child with a severe handicap where the parents had declined the offer of prenatal diagnosis of the handicap.' It is not uncommon for the argument to be put that the cost of a genetic screening programme would be covered by the savings resulting from the prevention of affected births. It has been further suggested, though we are unaware of evidence to support the point, that services available for those with a disability may be reduced if that condition is seen to be preventable through the use of prenatal diagnosis and abortion. Clearly, the reductions of such services might provide indirect pressures for parents to choose testing and selective abortion and such pressures should be resisted as unethical. We believe that parents should make their own decisions whether or not to proceed with a pregnancy, if a fetus is diagnosed as having Down's syndrome, and should be supported in whichever choice they make. In conclusion, the Working Party considers that the present use of genetic testing for reproductive choice in the UK cannot be regarded as eugenic. It takes the view that the best safeguard against any new eugenic pressures is properly informed, freely given consent.

**Confidentiality and disclosure**

5.55 An area of great concern is the use that might be made of genetic information about an individual's mental health. The use of information for non-medical purposes is discussed in Chapter 6. But one of the unique aspects of genetic information is that it is likely to be common to, and therefore relevant to, other family members. This raises distinctive issues about confidentiality: who should have access to genetic information derived from one individual if it is of relevance to another family member, yet the individual tested does not wish it to be disclosed?

5.56 In brief, of prime importance is the doctor or hospital's obligation of confidentiality. If that is not assured, individuals may not agree to provide information or to be tested, exposing themselves to the unexpected and unchecked development of mental disorder. It must be a matter for the individual concerned to agree to the disclosure of information about his or her genetic make-up, unless there are strong public interest justifications for disclosure.

5.57 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. It must be accepted, however, that there will be situations in which the public interest in disclosure will override the public, and private, interest in confidentiality. There is an obvious analogy with information about a person's HIV status. In relation to serious communicable diseases, the General Medical Council has advised that disclosure is justified "in order to protect a person from risk of death or serious harm." Thus the duty of confidentiality is not absolute. In the Council's

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61 General Medical Council (1997) Serious Communicable Diseases, London, General Medical Council, p 9, paragraph 32; and see also W v. Egdell
report Genetic Screening: Ethical and Legal Issues it was argued that “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 . . . 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.” 61 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, though there are those who hold a contrary view.22 If a risk is real and serious a doctor has discretion, but not a duty, to inform others exposed to that risk and this disclosure will not be regarded as a culpable breach of confidentiality.

5.58 Problems of non-disclosure, already rare, are likely to be even less pressing where the common mental disorders are concerned, since genetic information is unlikely to lead to such significant modification of risk that non-disclosure would have serious consequences. Many mental disorders are relatively late onset and some are treatable. Any genetic information may well indicate increased susceptibility rather than any degree of certainty that a particular disorder will develop.

The right not to know

5.59 A further complication is that some family members may wish not to be presented with information. There are three possible scenarios:

- Relatives are aware of a family history and have the opportunity to participate or not in genetic counselling.
- Relatives do not wish to participate in genetic counselling but genetic testing of another family member would reveal information about them.
- Relatives are not aware of a family history. Should they be informed and asked if they want counselling, or does this action deprive them of the possibility not to know?

5.60 Arguments about disclosure to other family members who may be aware of inherited risks are finely balanced. Some would see it as a duty for the doctor or genetic counsellor to break confidentiality and provide the information for a family member in situations where the members who have the information cannot be persuaded to pass it on. Others would argue that an individual has a right to have confidential information kept secret whatever his or her reason. Breaking confidentiality in such situations may serve to further undermine the already fragile concept of medical confidentiality and bring genetic counselling into disrepute. It may be argued that the disinclination of a family member to pass on information should be respected since they are likely to be better informed about their own family than an outsider. We should be wary of breaking confidentiality in a context where professionals are much keener to provide services than many family members are to use them. The Working Party recommends that the confidential nature of genetic information should be maintained. It can conceive of exceptional circumstances in which, in the absence of the consent of the individual, disclosure to close family members might be justified, if there are serious implications for them. Such decisions should be judged on a case by case basis.

5.61 Although doctors owe an obligation of confidence to competent adult patients, there is some
doubt whether there is a similar legal obligation owed to those who lack mental competence to
form a confidential relationship with doctors. The unsatisfactory outcome (if the doubt is valid),
could be that the confidentiality of those who are most vulnerable is not subject to any legal
protection. In practice this is unlikely to be a significant problem. Where the lack of mental
competence is temporary, the issue of disclosure is more straightforward than in, say, infective
conditions. The decision about whether to disclose information can and must be deferred until
the individual has regained sufficient competence for the matter to be discussed. Where the lack
of mental competence is likely to be permanent and they are unable to consent to the disclosure
of information to other family members, we assume that the requirement to act in their best
interests would extend to disclosure of information to others.