Chapter 1

# Introduction

## Introduction

1.1 Mental disorders place a heavy burden on individual sufferers, on those who care for them and on society at large. This report examines the ethical issues that may arise not only in the course of genetic research into mental disorders but in application of that research in clinical and other settings. Research into the genetics of mental disorders may lead to a range of potential benefits. It may add to our understanding of their underlying causes; improve diagnosis; enable the development of new drug-based or other treatments; and allow treatment to be tailored more accurately to individuals.

1.2 However, genetics and mental health are both areas which raise significant and sometimes distinctive ethical, social and legal concerns. This report examines the issues that arise when these fields come together. As with other diseases, the development and course of most mental disorders are affected by a complex mixture of biological, psychological and social factors. The focus of the report on the genetics of mental disorders is not intended to imply that genetic research is the only, or even the most important, approach for understanding and treating mental disorders, or that it is the only one to raise ethical issues. The extent of current research into the genetics of mental disorders, however, suggests that it is timely to try and anticipate its consequences.

1.3 The report focuses on schizophrenia, a range of affective disorders (manic depression and depression), dementias (Alzheimer's disease), neurotic disorders (anxiety) and personality disorders. Many of these have complex causes, and any genetic influences are not well understood. So the report also draws on experience of relatively simple single gene disorders, such as phenylketonuria, Huntington's disease and fragile X syndrome. While some of the ethical considerations relevant to these disorders are also relevant to genetically more complex disorders, there are also important ethical differences. Indeed, we should be cautious in regarding these single gene disorders as good models for understanding genetic influences in other more complex conditions.

## The whole person and 'geneticisation'

1.4 Some current thinking on genetics suggests that any additional ethical perspective is redundant, as if we could view genetics as basic not only to human biology but also to ethics. We accept, of course, that human behaviour can be viewed from a variety of theoretical perspectives. For example, extreme reductionists think that it is in principle possible to provide an entirely mechanistic explanation of human behaviour, and that given sufficient scientific progress this will eventually be practically possible also. However, even if this were the case, there is no reason to think that these scientific approaches incorporate an adequate ethical perspective. Indeed, there are those, including some respondents to the Working Party's consultation, who are opposed to any research into the genetics of mental disorders on the grounds that it is anti-humanistic. The Working Party does not take that view, though it maintains that the proper object of ethical attention is the condition of the whole human being viewed as a person, that is, as a unified subject of experience, thought and action. This report is primarily concerned with whole persons and not simply with their genes. Present day interest in genetics should not let the molecular complexity revealed by current science distract from the fact that the subjects of study are human beings and their values. Were this inquiry a purely scientific one, our concerns might be otherwise, but it is not and nor should they be. The need for the ethical perspective that focuses on the whole person is inescapable.

1.5 This broad ethical and humanistic perspective may be contrasted with an approach which has been labelled 'geneticisation'.[1] This ungainly term is used to mean an increasing tendency on the part of some, but by no means all, researchers to view human beings essentially as gene carriers, and to characterise issues of nature and functioning, of health and disease, solely in the language of genetics. This tendency has a number of consequences. By giving priority to the study (and manipulation of) genetic structures and effects, other kinds of explanation – such as those which refer to social and physical environments and economic conditions – are given less attention. Genetics may attract a disproportionate share of resources available for research and policy; too little attention and too few resources may be devoted to social research and policy. These issues were of concern to many of those who responded to the Working Party's consultation.[2]

1.6 Paradoxically 'geneticisation', although it focuses on characteristics human beings share, is often linked to an individualistic view of the appropriate treatment for health problems. The genetic defects associated with diseases are carried by individuals, and responsibility for health may then also be ascribed to individuals rather than being seen as something shared by society. 'Geneticisation' is also often associated with a change in attitudes to parenthood and reproduction. The traditional emphasis on the importance of good parenting may become less important, and genetic 'quality' in reproduction more so. People with genetic defects may be made to feel that they are flawed persons; parents may feel that they are expected to have genetically 'perfect' children, if necessary by using prenatal diagnosis and abortion.

1.7 These trends may also be expressed in deterministic or fatalistic attitudes which claim that ‘it is all in the genes’, and similar attitudes which risk undermining both moral responsibility and social solidarity. The latter is a somewhat neglected, but very important, notion. It involves distributing benefits and losses across society as a whole, so recognising the reality of social existence, deepening a sense of community and expressing equality of respect for persons. Instead of thinking of human life as something to be shaped by individual and social choice, 'geneticising' thinking regards circumstances as given, and classifies individuals and groups according to their genetic potential.

## Limiting suffering and having respect for persons

1.8 We take the view that the search for, and availability of, genetic information about mental disorders raises ethical concerns which cannot be answered simply by further genetic inquiry. In exploring such concerns, many approaches regard two ethical requirements as basic. These are the limitation of harm and suffering to humans (even to all sentient animals) and the maintenance of respect for human beings and human dignity.

1.9 Limiting harm and suffering is shown by seeking to cure, to care and not to injure, and so to establish and maintain medical systems that deliver effective, affordable and timely treatment. Respect for persons is shown by treating others as persons who can make their own decisions and lead their own lives; it is expressed in action and procedures that give due weight to personal autonomy and integrity, to human (including patients') rights, and to the obligation of doctors and researchers to seek informed consent, to preserve confidentiality, to respect privacy and to communicate effectively with patients.

1 A term coined by Abby Lippman. See for example, Lippman A (1992) Led (astray) by genetic maps: the cartography of the human genome and healthcare, **Social Science and Medicine** 35:1469–76.

2 For example, the Ethics and Genetic Engineering Network established by the Luton and Leighton Monthly Meeting of the Religious Society of Friends (Quakers), Leeds MIND, Theresa Marteau, Mencap and The Mental Health Foundation.

1.10 Whereas we all have an intuitive idea of suffering, and of the importance of limiting it, the idea of respect for persons is more complex. Persons think and act; they are the agents and recipients of all behaviour, including moral and immoral behaviour. It is only to persons that we ascribe both duties and rights (although some people ascribe rights without duties to other animals). Failure to respect persons is wrong because it threatens or undermines the very sources and possibility of any moral action; this is why respect for persons is ethically fundamental.

1.11 There is widespread agreement that the limitation of suffering and respect for persons are both of fundamental ethical importance. Some philosophical positions place greater weight on one or other of these principles, but this report does not take a stand on this fundamental issue.[3] The practical need is to identify, construct and support institutions, regulatory systems, professional codes and informal practices and ways of life that uphold both principles as thoroughly as possible.

1.12 It is often difficult, however, to establish exactly what the principles of limiting suffering and respect for persons require in practice. Sometimes dilemmas arise when promoting or protecting one value is not fully compatible with promoting or protecting the other; we have not offered ways of resolving all such dilemmas. Even so, it is necessary to recognise that the attempt to achieve ethically acceptable solutions requires that one take account both of the duties of limiting harm and of according respect to persons. There will be few issues that can be resolved in an ethically acceptable way unless both limiting harm, and respect for persons, are taken seriously.

1.13 The genetics of mental disorders raises distinctive issues both for the limitation of (human) suffering and for maintaining respect for persons (and for human dignity). Some of these distinctive ethical issues arise because the concern is with genetic conditions; some arise because (with some exceptions) the concern is with genetic predispositions rather than with gene mutations that have a more predictable effect; some arise because the concern is specifically with the genetics of mental disorders. We shall set out very briefly why these three aspects of our topic raise distinctive ethical concerns.

## Genetics and ethics: general issues

1.14 Genetic disorders are distinctive because they affect not merely individuals (as do all diseases), or groups of unrelated persons (as with epidemics), but groups of related individuals. Genetic information about one individual may reveal either certain, or more commonly probabilistic, information about their relatives, including any future children. Yet genetic information can be obtained by testing or treating a single individual. Both individuals and their doctors will then have to decide whether sharing information with relatives to whom it pertains, or its non-disclosure, is the better course of action.

1.15 Even in cases where it is relatively clear whether disclosure or non-disclosure would better limit suffering, it is often difficult to decide which would better express respect for persons. Would withholding knowledge from relatives about the possibility that they too might have a genetic mutation that could lead to a disorder be acceptable? Relatives might use that information in

3 Utilitarians, who maintain that an action is right to the extent that it promotes happiness, have argued that respecting persons (special cases of which are respecting patients' rights to privacy and confidentiality) is just another aspect of limiting harm and suffering. Other approaches, such as versions of Kantian thinking (which emphasise principled action), religiously inspired views (which view ethics as based on divine command or on the sacredness of life), and rights-based approaches, all argue that limiting suffering is just one aspect of respect for persons, and not invariably the most important. Finally, there are other philosophical positions which view the limiting of harm and suffering and showing respect as distinct and mutually independent goals.

making decisions about their lifestyle or their medical treatment, or about whether to have children. Would withholding information be a form of paternalism that denied them the possibility of making their own well-informed decisions? In that case, would respect for others require relatives or doctors to communicate what they knew about the results of genetic tests, or other genetic information? Or should genetic test results be treated as confidential to individuals, like other medical information? Can we think of a 'right to know' – or of a 'right not to know'– in purely individualistic terms in the case of genetic knowledge? Or does genetic knowledge challenge the basis of our usual individualistic understanding of medical confidentiality? Does respect for others require doctors or researchers to seek consent for genetic investigation from all who might be affected? Even if these questions can be resolved, and they are legal as well as ethical questions, showing proper respect for persons may make complex demands in seeking consent for investigation and treatment of genetic conditions.

## Genetics and ethics: single gene disorders and predispositions

1.16 Experience of genetic counselling and testing so far has been mainly concerned with single gene disorders, where a genetic test result may offer a high degree of certainty as to whether an individual will or will not develop a certain condition.[4] For example, prior to testing or screening, individuals who know that the Huntington's disease mutation is present in their family can often be given quite clear information about their own risk and relatives' risk of suffering from the disorder. If they then choose to have a genetic test, the result will allow a very confident prediction of who will and who will not suffer from the condition. For gene mutations of this sort information can be clearly established and communicated, and the difficult ethical questions are about whether and when to seek such information and whether, when and how to communicate it.

1.17 These issues are much more complex for gene variants which are associated with relatively slight predispositions to a disorder, rather than those which impose a near certainty of suffering from a disorder.[5] For example, a variant of the apoE gene (called the apoE4 allele) is associated with a predisposition to Alzheimer's disease, but a genetic test result can indicate no more than a somewhat increased susceptibility. Knowing that one has a gene variant associated with a predisposition to a disorder might nevertheless be useful (for example, if some medical or lifestyle change could reduce the susceptibility or the severity of the disorder) or alternatively harmful (for example, if it become a source of anxiety and there were no known way of reducing the susceptibility). It is often difficult to decide whether having information of this sort or lacking it might be more likely to cause suffering.

1.18 Showing respect for others may also make complex demands in the case of genetically-based predispositions. In particular, doctors have to determine whether and how to offer information about genetic tests for predispositions, whether to advise patients to take tests, how to disclose the results of any tests that are taken and how to explain the degree of risk to those tested (and to any relatives) without causing undue alarm. Those who choose to be tested and learn their own test results (adverse or otherwise) have in their turn to decide whether and how to inform their relatives.

4 Nuffield Council on Bioethics (1993) **Genetic Screening: Ethical Issues**, Nuffield Council on Bioethics, London.
5 Where a variant (or allele) of a gene is associated with only a slight predisposition we have used the term **susceptibility gene**.

## Mental disorders: integrity, reproduction and stigma

1.19 The range of ethical issues raised by genetic information expands when the information concerns mental disorders. Some of these additional issues cluster around the notion of personal well-being, of how one views oneself and is viewed by others; others concern reproductive decisions and some arise from the fact that mental disorders are often stigmatised.

1.20 A wide range of cognitive and emotional capacities are relevant to a person's identity, integrity and rationality; their absence may impair abilities to function as a person, may reduce personal well-being and may even lead to severe dysfunction. Genetic information which might be used to diagnose, or suggest susceptibility to a mental disorder, might raise questions about an individual's ability to function as a whole person and about their personal relationships. It might also undermine or weaken a person's sense of integrity and well-being, even when they are not suffering from any manifest difficulty or disorder. In the most vulnerable cases, acquiring genetic information about a predisposition to some mental disorders might cause great anxiety and even precipitate the feared condition.

1.21 A second area in which information about genetic susceptibility to mental disorder might raise difficult questions is that of reproductive choice. Even in the absence of genetic information, reproductive decisions can be hard for people with mental disorders. Some respondents to the Working Party's consultation who had suffered from mental disorders described the difficult considerations they had faced in deciding whether to marry or to have children; many had in fact done both successfully. At the same time there will be some individuals for whom such information may be helpful both as relevant and as a reinforcement of a decision already arrived at. For a few, rare single gene disorders, prenatal testing may provide definite information about the fetus; if information is both certain and adverse and the law permits it, termination is possible, and sometimes chosen. However, where genetic influences are slight (as for many mental disorders) and prenatal tests cannot provide accurate predictions, the relevance of genetic tests to reproductive decisions may also be slight.

1.22 Nevertheless, concerns have been expressed that new genetic technologies could be used for eugenic purposes. The concerns are often linked to the fact that in the past some eugenic abuse was directed at people whose behaviour was considered socially unacceptable, including those with mental disorders. The possibility that genetic information relevant to mental disorders might be misused to influence reproductive choices, or for other forms of genetic abuse, cannot be simply dismissed.

1.23 A third distinctive group of ethical problems raised by mental disorders is that those afflicted often have to suffer not only their disease, but also the associated stigma. Relatives caring for a patient with mental disorder may also have to cope with the stigma of having an afflicted relative. Stigma is a distinctive form of suffering in which a person experiences shame, and is the object of blame, often for matters which were in no way avoidable. There is little shame suffered, or blame apportioned, for most physical injuries and illnesses. Broken legs, measles and heart attacks will attract sympathy and concern; patients are not usually blamed for their sufferings; their relatives are not usually stigmatised. Many mental disorders, however, are a matter of shame for those affected and for their relatives and, far from attracting sympathy, are a source of avoidance, criticism or even of blame by others. It is important, therefore, to consider whether the availability of genetic information will increase or decrease the stigma associated with mental disorders, and whether fear of stigma will affect reproductive decisions.

1.24 The requirement to limit suffering makes similar demands on those treating and caring for persons with mental and with physical disorders. The requirement to respect persons is more complex in the case of mental disorders, however, and makes distinct and difficult demands. Mental disorders frequently disrupt cognitive processes and capacities for social interaction; sufferers may have difficulty in making decisions, in giving consent to the investigation or treatment of their conditions, in assimilating the implications of genetic counselling and in communicating relevant information to relatives. If the limitation of suffering were the sole ethical requirement, then a purely paternalistic perspective, directed solely towards promoting the welfare of those with mental disorders, would suffice. If people with a mental disorder are to be respected as persons, however, their autonomy must be supported (even when it is greatly reduced), they must be given accurate information (even when they are having difficulty in following it), their concerns must be treated with due confidentiality, they must be offered privacy like other patients and, above all, informed consent must be sought if they are to be subject to investigation and treatment. All of these can prove demanding, not least because of the need to observe legal as well as ethical duties.[6]

1.25 Because these requirements cannot be met in all cases, separate legal and medical procedures have been established which permit paternalistic investigation and treatment undertaken in the 'best interests' of the patient. This may take place in circumstances where the attending doctor has formed the view that the patient lacks capacity to consent,[7] or under the provisions of the Mental Health Act, on the certification (normally) of two doctors that the person comes within the terms of the Act. Even at its worst, however, mental disorder is rarely a matter of comprehensive incapacity; it is commonly a matter of impaired or intermittently impaired capacities. Most people with a mental disorder can continue, throughout the duration of their disorder, to take all decisions for themselves with no more assistance than a person without mental disorder. Accordingly, no general case can be made for those suffering mental disorders to be exceptions to the usual requirements for consent, or to other aspects of respect for persons. Care and sensitivity are needed if due respect is to be shown to mentally disordered persons, particularly if they are detained and subject to compulsory treatment. In some circumstances it can be very hard to meet these ethical demands; but this is no reason to overlook or deny them.

## Science and ethics: genetic research

1.26 Everybody recognises that ethical considerations must govern the treatment of patients but some think that research itself, including genetic research, does not raise difficult ethical questions. For example, it is sometimes argued that scientific inquiry itself is value neutral, and that any attempt to direct or evaluate basic or applied research by using ethical norms may frustrate the principal goal of research, which is the acquisition of knowledge, and may even lead to the very abuses it was intended to avoid. This argument has several flaws. Science is, by definition, committed to certain values. Systematic investigation of the natural order is an expression of the desire to understand, and involves a commitment to the value of knowledge as such as well as to certain standards of inquiry: honesty in data gathering; accuracy in reporting results; fairness in using others' work and so on.

6 Law Commission Report (1995) **Mental Incapacity**, Law Com No 231, HMSO, London; Nuffield Council on Bioethics (1995) **Human Tissue: Ethical and Legal Issues**, Nuffield Council on Bioethics, London; The Lord Chancellor's Department (1997) **Who Decides? Making Decisions on Behalf of Mentally Incapacitated Adults**. Cm 3803, HMSO, London.

7 See *Re F* [1989] 2 All ER 545 (HL) as regards adults and *Re W* [1992] 4 All ER 627 as regards children.

1.27 Current scientific practices clearly rely on further ethical norms. Science funding is provided by state and private sponsors allocating scarce resources. Their priorities and decisions will rightly reflect various principles and values, and scientists seeking funding will point out the intrinsic worth and the other benefits of particular proposals. The question is not whether values underlie scientific research, but which values are most significant, how conflicts between them are to be weighed, and what they may show about the importance of supporting, of refusing to support or even of restraining certain types of research. Scientific research, like any other activity, is subject to discussion about the best way to proceed in the light of principles and values.

1.28 This conclusion contrasts with the view that once something is scientifically or technically possible, it will invariably be done. It indicates how important it is to distinguish between questions about what can be done, about what will be done and about what should be done. Scientific possibilities do not of themselves determine policies, which may and should reflect ethical, legal, social and economic considerations as well. Doing science is not a way of escaping moral responsibility. There may be no simple rules whose automatic application will yield acceptable solutions and, as respondents to the consultation have pointed out, the "*current paucity of information on genetics and mental disorders suggests that it is too early to try to develop detailed ethical frameworks*." [8] Even so, scientific and medical researchers cannot abrogate moral responsibility for their own investigations, or for the ethically significant issues which their work may create for society. Scientific possibility is one thing, moral permissibility another and moral obligation a third.

1.29 In summary, this report has two main aims: to describe the current possibilities for diagnosis and treatment based on genetic research into mental disorder, and to consider the ethical and legal reasons for supporting, for regulating or for setting aside these possibilities. It is not the purpose of the report to speculate about long-term social trends, or to resolve fundamental philosophical questions about human nature. However, as has been noted, the orientation of this and subsequent chapters is at odds with that of 'geneticisation'.

8 Response by the Royal Society to the Working Party's consultation. The response continues, *"None the less it is important to identify the issues that may arise."*