

## Chapter 8

# Genetic screening and public policy

- 8.1** By their nature, most genetic screening programmes involve large numbers of people. This is so even for programmes limited to defined groups of the population which may be at risk of developing a serious disease or transmitting it to the next generation. We have therefore attempted to assess the present level of understanding of the science of genetics and its significance for the health of individuals, as well as the potential for improving public health.
- 8.2** Such evidence as there is suggests that there is widespread misunderstanding of the mechanism of inheritance, in particular of the importance of recessive genes and carrier status. Much ignorance about how genetic diseases are transmitted is mixed up with notions about the inheritance of physical characteristics, such as height, eye colour etc. Genetics can be confused with eugenics (see paragraph 8.16), and there are concerns about possible stigmatisation which may or may not have a basis in fact.
- 8.3** A broad public understanding of the scientific basis of medical genetics is essential if informed public policy decisions are to be taken about the introduction of genetic screening programmes. Such programmes, as we have emphasised throughout our report, have both an individual and a public dimension.

### Public understanding of genetics

- 8.4** If an individual is to be well enough informed to be able to give consent to genetic screening, he or she needs to have some general understanding of genetics. This means that the public as a whole needs to have a greater knowledge and awareness of the genetic processes that can affect us all.

**8.5** Some relevant teaching about human genetics is included in the National Curriculum for 14 to 16 year olds under **Life and Living Processes**:-

“[Pupils] should consider the interaction of genetic and environmental factors (including radiation) in variation. They should be introduced to the gene as a section of a DNA molecule and study how DNA is able to replicate itself and control protein synthesis by means of a base code. Using the concept of the gene, they should explore the basic principles of inheritance of how sex is determined in human beings and how some diseases can be inherited.”

**8.6** Teaching of the principal modes of inheritance is particularly important. This knowledge is needed to understand most genetic disorders; and, since it conveys the message that all healthy people can carry genes which are abnormal but which only rarely result in disease, it may help to prevent misinformation and prejudice. It may be helpful for people to know that most healthy carriers of genes for recessively inherited disorders will have no family history of that disease. If there are to be screening programmes for recessively inherited conditions, the public must have some knowledge of the recessive mode of inheritance and, most particularly, the meaning of being a ‘healthy carrier’.

**8.7** Accurate information on genetics and genetic disease should be available both for the public and for health workers. This should include general information about reproductive risks and specific material for individual disorders appropriate for particular screening programmes. A wide range of educational aids about genetic screening is required. We hope that the Department of Health will take the lead in addressing the different sections of the community, enlisting the media to help with the task. Appropriate voluntary bodies can also help; we have noted that in the Department of Health’s recently (June 1993) issued outline guide **Population Needs and Genetic Services**<sup>1</sup> it is stated that:-

“voluntary bodies, by virtue of the special experiences and knowledge of their members, and the sources of expert advice available to them, have important roles in providing information and giving support to individuals and their families.”

## What are the dangers of stigmatisation?

- 8.8** Stigmatisation has been defined as 'branding, marking, or discrediting because of a particular characteristic.' It has been suggested that genetic screening could lead to stigmatisation of carriers.
- 8.9** Concern has been expressed that routine screening for carriers of a genetic disorder might be viewed as a tacit requirement that the birth of children with handicapping genetic conditions should be avoided. Stigmatisation of carriers is likely to focus on beliefs that it is irresponsible and immoral for people to have children who could transmit disability to them.
- 8.10** There have always been some negative social reactions to disability in all its forms. These social reactions can be related to conflicting feelings, for example not knowing how to talk to parents or people with a specific problem, fear of creating offence by being healthy, a consciousness of good fortune because one does not have a similar problem and has no idea how one would cope if one had. There is a fear that a known genetic cause of handicap could add to social isolation, because, due to prevailing ignorance of genetics, people are inclined to feel that inherited disorders affect only a few families, and fortunately 'this could **not** affect me'.
- 8.11** It has been argued that the availability of prenatal screening and diagnosis, together with the termination of seriously affected pregnancies, both reflect and reinforce the negative attitudes of our society towards those with disabilities. Indeed medical genetics may add a new dimension if genetic disorder came to be seen as a matter of choice rather than of fate. On the one hand, there is an effort to create an environment in which people with a disability are accepted into society and seen as having a worthwhile life; for example, integration into mainstream schooling and changes in the language used to describe people with disabilities. At the same time as encouraging a more positive environment for people with severe disabilities, resources are spent on preventing their births. Given the option of prenatal diagnosis and abortion of affected fetuses, some parents may feel that to produce a child with a potentially diagnosable disability is to be blameworthy for that child's birth.
- 8.12** It has been further suggested that an emphasis on genetic differences between ethnic groups could increase social differences and discrimination. Ethnic groups with a high prevalence of genetic disorders might be additionally stigmatised. Members of an ethnic group may feel stigmatised, although other communities do not in fact attach stigma to that group.

## Evidence on stigmatisation as a result of genetic screening

**8.13** We have noted the unhappy consequences of the introduction of sickle cell screening programmes in the USA during the 1970s. A study by the Office of Technology Assessment of the US Congress reported :

“Some who participated in screening programs and were found to be carriers of sickle cell trait experienced discrimination at work and from insurance companies that raised their premiums. Apparently, discrimination in the workplace sometimes occurred because it was believed that those with sickle cell trait could experience the painful episodes characteristic of sickle cell disease (which occur when sickle-shaped red blood cells occlude the normal flow of blood). The result for some job applicants was denial of employment based on their carrier status and removal for some who were already employed. In some cases, life insurance companies either raised premiums for carriers or denied coverage for applicants with sickle cell trait. At that time, laws were enacted in Florida, Louisiana, and North Carolina that prohibited such discrimination. Since the mid-1970s, many of the State laws requiring mandatory sickle cell testing have been repealed.”

**8.14** The lessons of that episode have been learnt by those responsible for current screening programmes. Such evidence as exists suggests that current genetic screening programmes need not result in any significant stigmatisation. A study of over 3,000 individuals in Hertfordshire looking at the psychological and social consequences of community carrier screening programme for cystic fibrosis reported that fears of possible social costs of screening may be ill-founded.<sup>2</sup> Carriers and non-carriers uniformly approved of screening and were glad to have been tested. Carriers told partners, siblings, relatives, and friends of their result and did not seem to feel stigmatised. A large majority (89%) told their partners. “It is very encouraging that being screened has increased awareness of CF and recessive inheritance even for those testing negative, and it is very unlikely that carrier testing will stigmatise or cause lasting psychological damage to those testing positive.” Other studies have concluded that, with a few exceptions, individuals taking part in genetic screening programmes did not feel stigmatised. It should be noted that a high standard of information and counselling was provided in all of these studies. The Fragile X Society stated in its submission to us that “no family has said that it has experienced this (stigmatisation) as a problem; on the contrary, many have found that they and others have found their children

easier to accept and deal with once they have an explanation for their problem.”

### **How can stigmatisation be avoided?**

- 8.15** The dangers of stigmatisation have sometimes been outlined, for the most part, in general and **hypothetical** terms. Proper educational programmes should reduce those dangers. At the same time the quality and extent of education and counselling provided should have a major effect on the extent of stigmatisation perceived by individuals. A well-informed individual is less likely to feel stigmatised than a poorly-informed individual who has received little or no counselling. Indeed, it could be argued that, if we all found out our genetic variations, then there would be less concern about stigma. It is estimated that we all carry mutations for at least one serious recessive disorder and when “everyone realises that he or she is a carrier there can be no stigma.”<sup>3</sup>

### **Limiting the improper use of genetic screening : the legacy of the eugenics movement**

- 8.16** Eugenics is the doctrine which claims that it is possible and desirable, through selective breeding and the elimination of undesirable individuals, to alter the hereditary qualities of a race or population. It thus aims to improve the qualities of the species rather than of an individual. Some societies and governments have attempted to apply this doctrine in practice. The most notable example was provided by the Nazi party in Germany, which supported human geneticists in their eugenic research in return for practical support for the party’s race policies.

### **Eugenics and other societies**

- 8.17** Many societies have been influenced by eugenic doctrines. Thus, for example, early in the century certain legislatures in the United States sought to control social characteristics such as degeneracy, drunkenness, unemployment, criminality, prostitution and alcoholism through a targeted sterilisation policy, combined with restrictive immigration laws. In the 1930s, the Canadian provinces of Alberta and British Columbia passed legislation which permitted the sterilisation of mentally ill persons without their consent. The legislation remained in force in both provinces until 1972.

- 8.18** The UK has differed in this respect in that no legislation has ever existed to carry forward the eugenics doctrine, although there has been in the past scientific, political and even ecclesiastical support for the ideas reflected in the doctrine.
- 8.19** Eugenics is often regarded as a subject that belongs to the past, at least in democratic societies, but recent developments in genetic technology have understandably raised fears among the public and professionals that these might be misused for eugenic purposes. It will continue to be important to reassure the public that genetic testing in medicine in the UK is used to help individuals and their families avoid the occurrence of serious inherited disorders or their associated complications. This is also the primary goal of those wider population-based genetic screening programmes that have so far been established.

### **The dangers and safeguards for our society**

- 8.20** Large-scale genetic screening does raise issues relating to population and public health that might conflict with the interests of individuals. Any genetic screening programme set up with the specific aim of reducing the incidence of a particular disorder may come into conflict with those members of that population who do not wish to be screened. The public health definition of 'success' or 'failure' of a programme may be in danger of turning on too narrow a calculation of costs and benefits. Benefits must not be calculated in purely financial terms of preventing the birth of individuals who may have higher than average health care needs and costs. The benefits should be seen as enabling individuals to take account of the information for their own lives and empowering prospective parents to make informed choices about having children.
- 8.21** Genetic screening programmes for recessively-inherited diseases (for example, cystic fibrosis and thalassaemia) will have no significant effect on the frequency of the abnormal gene in the population, even though the frequency of the disease at birth may be greatly reduced; in testing for dominantly-inherited diseases, such as Huntington's disease, the gene frequency would be reduced in line with any reduction in births of those likely to develop the disease. Genetic screening in such situations could only be considered 'eugenic' in nature if the decisions of individuals were subjugated to those aims considered to be of benefit to the population or the state.

- 8.22** As it becomes feasible to test for the genetic basis of many common, usually polygenic or multifactorial disorders, as well as for normal characteristics, the potential for eugenic misuse of genetic testing will clearly increase. The existence of genetic registers (see paragraphs 5.32 - 5.39) requires safeguards against the potential for eugenic misuse. This makes it all the more important for society to keep genetic screening under review and, if necessary, limit misapplications at an early stage. We must ensure that neither specific individuals, nor society as a whole, are harmed by a hasty or ill-considered application of genetic testing.

## **Conclusions and recommendations**

- 8.23** The threat of eugenic abuse of genetic screening requires safeguards. In a democracy, public understanding of human genetics should serve to create awareness of the dangers of eugenics, and of the possible stigmatisation of those carrying or suffering from genetic disorders. **We recommend the need for improving public understanding of human genetics should be borne in mind in any review of the National Curriculum and in the work of all public bodies concerned with the public understanding of science.**
- 8.24** We recognise that there are limits to the effects of educational work, however good. We, therefore, regard as essential to the safeguards against eugenic abuse our recommendations on **adequately informed consent, confidentiality** and the **central coordination and monitoring** of genetic screening programmes.