

EUROSCREEN: ethical and philosophical issues of genetic screening in Europe

EUROSCREEN is one of the Concerted Actions supported by the Biomedical and Health Research Programme of the European Union (Galton DJ. *Medical Research in Europe. J R Coll Physicians Lond* 1995;29:378). It is a three year project (1994–1996) to investigate the ethical and philosophical issues raised by developments in genetic screening techniques, with special reference to multifactorial diseases. The project is coordinated by Dr Ruth Chadwick from a centralised facility in the University of Central Lancashire, with specific tasks allocated to several groups.

The objectives of EUROSCREEN are to ascertain the extent of development of screening programmes in different European countries, to analyse the public policy responses to these developments and to make recommendations. The study is multidisciplinary, with participants drawn from public health medicine, genetics, anthropology, law, history of medicine and sociology as well as philosophy. There are 25 participants from 14 European countries.

Asking for informed consent when screening children for genetic diseases raises the question of *whose* consent is to be sought—the child's or the parents'. The issue can be portrayed as one of ownership:

- What are the parents' rights over, and duties towards, their child?
- How far can a child be regarded as a morally autonomous person?
- Does the child have the right to know and *not* to know?
- Is obligatory pre-test counselling compatible with the prevailing ethics of counselling?

A group, coordinated by Angus Clarke (Cardiff) has recognised the importance of gathering empirical data and has designed and sent out a questionnaire to geneticists, lawyers and ethicists in different European countries.

A group on concepts of health and disease is coordinated by Ingmar Porn (Finland) and is concerned with the adverse effects of genetic screening on people's self-image. Should these people regard themselves as healthy or not? Genetics have an impact on our very concepts of health and disease. Some reject the whole notion of *genetic* disease, either claiming that it is incoherent because every disease has an environmental component, or on the grounds of the implica-

tions for policy, since focusing on genetic disease is seen as an undesirable 'geneticisation' of problems, and in particular of health problems.

The concern of the anthropological group, led by Brunetto Chiarelli (Florence), is that while there are new biotechnological possibilities for the individual, contemporary humankind is creating an artificial environment that can cause damage and undesirable genetic modification.

Genetics as a service is often vulnerable to criticism because of historical precedents of abuse. The historical group, coordinated by Urban Wiesing (Munster), is considering the question of how historical evidence should be used; how stories of historical events can justifiably influence moral decisions; whether one can, in fact, make a moral decision without knowing the relevant past; what were the circumstances that led to previous undesirable uses of genetics and screening techniques and how can they be avoided.

Screening programmes

Objectives—There were varying views on the extent of individual autonomy but a consensus that the purpose of prenatal testing is to detect *serious* disease and that there is no right to test for everything. There is, of course, the problem of defining what constitutes a serious disease, whether there should be a formal, legal definition and if so, who should make this definition. It is assumed to be desirable to eliminate non-genetic disease to alleviate suffering. Suffering from genetic disease should also be alleviated.

When looking at the motives for introducing screening programmes, the 1993 Danish Council of Ethics report suggested that there is an obligation to help the weak which will be best exemplified when screening results in the curing of a serious disease. In genetics, the duty to help has a special intention—to offer information that will facilitate autonomy. There may, nevertheless, be pressures from health authorities, relatives of people with a genetic disorder, the community and politicians, to publish results. The fundamental aims of screening programmes may be neglected as the development of techniques by scientists leads to pilot programmes which create demand. If insufficient people make use of the services it is assumed that a lack of knowledge is the reason, and education will begin.

Geographic variations—Developments in genetic screening and the social response to them vary widely within Europe. Finland has a different pattern of genetic disease from the rest of Europe due to its isolated and scattered population; selective abortion following testing is not legally acceptable in Ireland; and in

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Germany any discussion of genetics and biotechnology is itself controversial. None of the population screening programmes is common to all countries represented in the EU project although neonatal screening for phenylketonuria (PKU) is routine in all countries except Finland and prenatal screening of older women (usually over 35) is offered routinely for Down's syndrome in all countries except the Republic of Ireland.

Some of the diseases which have been particularly suitable for genetic screening in other western countries are unsuitable in Finland because they are extremely uncommon or totally absent; for example phenylketonuria (PKU) and cystic fibrosis. Some of the hereditary diseases that occur in Finland are almost unknown in the rest of Europe; eg Salla disease and AGU_{Fin} which both cause mental retardation, HOGA disease (causes blindness at the age of 20–40) and congenital nephrotic syndrome. Current screening programmes in Finland include the screening of newborns for genetic susceptibility to diabetes, due to be expanded to the whole country by the end of 1996, screening of selected risk families for insulin dependent diabetes mellitus; familial colon cancer; fragile X and AGU_{Fin}.

In the Republic of Ireland the legal prohibition on abortion has influenced the availability of individual genetic testing, especially antenatal testing, since such testing could be construed as part of the process of decision-making that could lead to abortion. The most common inherited disease in Ireland is cystic fibrosis and researchers are working on a way of bypassing the genetic defect for cystic fibrosis as the selective abortion of affected fetuses following testing is not legally acceptable. A Supreme Court judgment does allow for abortion if the woman's life is in danger, physically or psychologically, but there has been no written law. Irish medical consultants have used services in Belfast and London and over 6,000 women travel to England each year for abortions.

Social responses

Counselling and informed consent—Although all participants agree on the importance of counselling before informed consent, they recognise that the increasing use of genetic tests will render the provision of pre-test counselling impracticable. It is likely that, due to the cost, counselling will only be given where the test result is positive, for example in testing for Down's syndrome. Commercial companies already offer testing for cystic fibrosis carrier status by post without any pretest counselling.

Other than the PKU testing for newborns, which is routine in most countries, all the screening is offered rather than expected, although the degree to which it can be said to be freely chosen is variable due to a number of general and country specific factors. General factors include a lack of knowledge of genetic

disorders which is seen as a barrier to informed consent and the fact that those giving informed consent may not be the ones who will be directly involved in the consequences of the test: eg parents give consent for a fetus or young child; individuals give consent but their relatives will be affected. A country specific factor was the financial incentive for mothers in Austria to ensure that their children have all the required examinations, tests and inoculations; a mother receives a monetary grant when the child is born and at regular intervals thereafter, provided the mother-child passport shows that she has attended regularly for the required checks until the child's third year.

The stress on individual counselling raises the fundamental question of whether genetic screening should benefit the individual or society and whether these concepts are necessarily in opposition. Currently, decisions are made by the individual or couple, although their decision can be negated by lack of resources and legal restrictions. However, in the area of public health, genetic screening programmes will be seen as a way of cutting costs as well as preventing individual suffering. Putting the onus on the individual can be seen as allowing autonomy or, in a less benign way, shifting the burden of a difficult decision from the public health authorities on to the shoulders of prospective parents and partners.

Normality and abnormality

The importance of the debate on normality and abnormality was illustrated by a participant who gave an example from personal experience of a controversial request to abort a fetus with cleft palate. In Ireland the strong heritage of religious, moral and cultural values questions the value of defining some conditions as normal and others as 'abnormal', especially when those definitions may have implications for the protection and nurturing of human lives. There is an awareness of the dangers of stigmatisation, particularly resulting from psychiatric diagnoses. A similar caution over normality/abnormality was expressed in the German report arising from the memories of Auschwitz. For many Germans, any discussion of genetics or euthanasia is dangerous and has to be suppressed. In Austria there has been little discussion of issues of informed consent, childhood testing or the requirements of insurance companies for genetic testing. There is a strong antipathy to genetic registers, because of memories of their misuse during the national socialist years.

The 'right to abnormality'—Organisations of handicapped people in the Netherlands and Germany have emphasised the 'right to abnormality' and suggested that abnormality can be creative, enriching or a positive challenge. Handicap may be abnormal in the statistical sense but being handicapped is a special kind of health. These views are also represented in the

antibioethics movement in Germany which looks upon bioethics as 'deadly ethics' or the 'ethics of killing'. The movement has an important influence on the debate and disrupts, often successfully, conferences and seminars on bioethics; however, other representatives from Germany felt that it was not very influential on a day-to-day level.

Among the handicapped themselves, fears have been expressed about the long-term harm which could result from screening programmes. The view frequently put forward since the 1970s has been that it is part of the parents' responsibility to see to it that the quality of their children's life is optimal. If abortion is allowed, then unborn life is considered of lesser value than born life, whether handicapped or not. The ability to detect genetic disorders could lead to a decrease in respect for handicapped life and especially handicapped unborn life. Assessment of the outcome of screening programmes has concentrated on the effects on individuals: the dangers of stigmatisation and of anxiety about discrimination, understanding of the results and the effect on behaviour. However, long-term harmful effects on society are possible and difficult to assess.

Human beings are good at making a mess of things and if the prospect of shaping 'man' ever becomes real there is no certainty that we could succeed in doing so reliably. The pessimistic view, from Italy, was that curing genetic disease will contribute to genetic deterioration; for example, the incidence of PKU is increasing as effective treatment enables sufferers to reach adulthood. Modern genetics will change society, so there is a need for a wider discussion of what society should be like. However the majority felt that 'doom and gloom' arguments do not stand up to scrutiny; for example, the desire for boys in some cultures makes unequal sex selection likely but this was felt to be a diminishing problem with increasing sex equality.

Legislation

Debate over the need for legislation to control the use of genetic testing and screening by third parties is likely to be most prominent in those countries with the heaviest reliance on individual private health insurance. Reports in the Netherlands and the United Kingdom have called for a moratorium on requiring disclosure where life insurance policies are proportionate to income or of moderate size (Committee of the Health Council of the Netherlands 1989, Nuffield Council on Bioethics 1993). However, in Belgium a total ban on the use of genetic testing to predict the future health status of applicants for life insurance was laid down in the Law on Insurance Contracts, which came into force in September 1992. There is a complete prohibition without exception on the communication of genetic data to insurers by physicians, insurance takers and insurers. The insurance taker cannot volunteer favourable genetic information to get better

conditions and lower premiums. This is to avoid discrimination between those with genetic 'good luck' and 'bad luck'. In France, genetic testing for insurance or employment is forbidden through laws approved in July 1994. Punishment for testing without consent and testing for other than scientific or medical purposes is one year's imprisonment and a 100,000F penalty. In Ireland, under the Medical Council guidelines (1994), businesses and insurance companies are not entitled to require genetic testing of their employees or clients as a basis for deciding on employment or insurance agreements, nor can they receive genetic information held by a medical practitioner or clinical genetics centre unless the client gives consent.

Despite such attempts to prevent people from becoming uninsurable because of genetic 'bad luck' it was pointed out that in practice there might be no need to obtain test results before deciding not to insure someone. A family might be unable to get insurance for a child because the bank which employs the child's father already knows the genetic history. There are still dangers of the 'sick family syndrome'.

Future work

The EUROSCREEN group plans to continue its work beyond the current three year period under the BIOMED programme, exploring genetic information and insurance; commercialisation and genetic testing; and promoting public awareness.

It is intended to include a demonstration activity as part of the public awareness programme, which will take the form of a 'gene shop' on a city high street in the north-west of England, to provide information on different aspects of genetics and to allow for the presentation of a variety of viewpoints on their implications.

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Further reading

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