Screening for fetal and genetic abnormality: social and ethical issues*

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SUMMARY In answer to questions raised by practitioners, an ethics of genetic screening is located in a tension between liberty and responsibility in three respects: (1) to nature and biological processes; (2) to the disposal of human life; and (3) to the relation of persons to society. Under (1), the obligation to pursue research, fundamental as well as applied, is affirmed, offering the benefit of economy with fetal life, but requiring discrimination between the beneficial, the trivial, and the bizarre. Under (2) the abortion question, when relevant to diagnosed abnormality, is discussed, not in the language of conflicting rights, but of the relation of duties to interests. Under (3) the familial and social dimensions of screening raise questions of the disclosure of information and the keeping or extending of confidences. Last comes the value placed on truth in two related areas of developing practice. In infertility treatment, the donors of gametes are required to remain anonymous. Gene tracing through families requires for its effectiveness some correspondence between assumed identity and genetic identity. This conflict of social policies should be resolved.

Definitions and intent

By 'screening' in the context of this discussion, I understand "any procedures undertaken in order to detect presymptomatic disease in a population".¹ Genetic screening I treat as a means to furthering "the main purpose of medical genetics (which) is to apply the principles of genetics to the practice of medicine so that the burden of genetic disorders can be reduced in the community".² I observe the common agreement here to limit the discussion of screening to patients for whom there are already indications of risk.

The relevant moral reasoning requires the holding in tension of a cluster of principles, each exerting moral claims. I reduce these to three. I shall then relate cases to them.

A.1. The tension between human liberty and responsibility within nature and biological processes

In the glib language of today's pseudo-piety, medical scientists are alleged to 'play God'. If this

is meant to imply that only God, as the Creator of nature, may manipulate it, the slogan is absurd. It has no foundation either in theology or in human experience. Theology, with its language of man made in the image of this Creator God, attributes responsibility to man: a limited dominion, within the created order. Millenia of human experience, from animal husbandry and agriculture to medical practice, has recognised human intervention as a timeless and universal human activity proper to the nature of man. God is neither jealous nor mocked.

Nature is neither divine, perfect, nor immutable. In the realms of human biology its products from time to time are marred. A combination of scientific disciplines now yields, first, some understanding of how these mishaps occur; secondly, means of detecting some of them prenatally, at ever earlier stages of development; and, thirdly, means to remedy or palliate some few of those detected. It would be hard to establish a reasoned moral case for forbidding the exercise, in this field, of normal human faculties to protect human kind from the random adversities of nature.

B.1. The tension between human liberty and responsibility with human life

Concomitant with genetic screening goes the option of terminating the life of a defective conceptus.

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Technical progress offers that option both more extensively and ever earlier: from fetal scanning and amniocentesis at mid-pregnancy, to chorion villus sampling at the transition from embryo to fetus, back to cellular biopsy and analysis during pre-embryonic cleavage, with DNA technology adding remarkably to precision. A defective pre-embryo need not be implanted; a defective embryo or fetus may be aborted: the choice lies with the mother, clinically advised.

All responsible participants in this debate hold human life to be sacred in the sense that it is of paramount worth, and that it has claims to care and protection, and to service in its vital interests. Some go further and interpret 'sacred' to mean inviolable. They would prohibit as a moral wrong the arrested development of any pre-embryo and any termination of embryonic or fetal life. To kill is an inhuman avoidance of the duty of loving care for the handicapped, with the consequent loss of a reciprocated warm human relationship with them. Others, without weakening in resolve to care for the handicapped if born, or in appreciation of the depth of affectional relationship possible with them, would still maintain the option of their not being born, partly in their own assumed interest, partly out of an estimation of the capacity or incapacity of their parents or of society to give them that optimal care. They would regard the prospect of severe congenital handicap as an indication strong enough to rebut the accepted presumption in favour of life.

C.1. The tension between human liberty and responsibility in relation to persons in society

Medical genetics shares with most medical practice the primary obligation to each patient, one by one: to a person, in strict terms the bearer of rights, and more generally the embodiment of interests to be enhanced and enjoyed. The patient is also a member of a body corporate, a human society in its various cellular structures: blood group, family, kindred, nation, race, and so on. As a social morality grows out of a tension between personal interests and corporate or social interests, so an ethics of medical genetics has to take account of the social consequences of accumulated clinical decisions made primarily in the interest of individual patients. In an ethically harmonious society, and when scientific development is slow enough to carry within it a general understanding and acceptance, that tension between the personal and the corporate wills resolves itself in conventions, norms of conduct, professionally and socially accepted. When these two conditions, ethical homogeneity and a manageable tempo, are lacking, as they are today, conflict is

inevitable. Resort is had to law, either through the Courts or in Parliament, as those fearful for the common good, or for their own absolutist standards of morality, seek to restrain the liberty of those who attach wider options to their professional responsibility. There is also the other fear, that governments, or the tyranny of television or other media pressure, will override personal liberty and impose screening and termination on those who would not choose it.

Do I err greatly in seeking to locate our ethical discussion within these three fields of choice or tension? If not, let us see into which field or fields some of the empirical features of practice fall.

A.2

Our understanding of man's place and limited dominion within nature entails an obligation to pursue scientific enquiry and the beneficial application of its results. Disciplined curiosity and the liberty to pursue fundamental research are to be affirmed. There is ample encouragement to go forward, evidenced especially by DNA technology. For example, affected male haemophiliacs can be identified by this means from chorionic villus samples or later, so that no normal male need be terminated, a notable advance on the old 50:50 gamble. Similarly with Duchenne muscular dystrophy: with the multiplication of gene probes only 1 to 2% of families are not amenable to identification. Carrier and non-carrier females can be distinguished. Work on single embryonic cells from pre-implantation mouse embryos indicates the possibility of identifying in human pre-embryos the HGPRT deficiency for couples at risk of having children with Lesch-Nyhan syndrome.³ These are examples of research which yields an ethically valuable *economy* in the sparing of fetal life, in reducing the trauma of late abortion, and—if pre-implantation diagnosis, despite its higher cost, can go forward—a pointing of the way to obviate, for certain diseases, termination of pregnancy altogether.

But modesty of expectation must yet prevail. There is no prospect, either theoretical or practical, of ultimate certainty in these matters; it is not simply a question of diagnostic error. Random events in evolution must elude control. Enough is known to rule out the possibility of those spectral powers attributed to geneticists of fabricating men after their chosen image. It is ethically necessary to explode false expectations of this sort. I learn from my friend the Chief Rabbi that Jewish Orthodoxy would support what he would call 'repair' genetics, to eliminate when possible the expression of deleterious genes in human bodies. What Jewish Ortho-

doxy dreads is the spectre of the superman or super race, genetically fashioned. If that is, as I believe, beyond possibility as well as intent, let it be said.

Equally the power inherent in medical genetics must be preserved from abuse in trivial causes. People speak of 'the camel's nose'. The threat is not only that our concept of the tolerable in handicap might diminish to the point where our humanity is diminished with it, it is also that the human will might intrude too far, to aborting a child of the unwanted sex, for instance, without the justification of a sex linked disorder. The responsibility of genetic counsellors is serious here. The price of liberty must be self-restraint from the frivolous and the bizarre.

B.2.

What has just been said about the use of our liberty within the natural process has sharper definition in relation to our responsibility for human life. This is not an occasion to rehearse the old abortion debate. We work, anyhow, on the fringe of that problem: only a small percentage of terminations are registered on the ground of fetal handicap.

The leading fact is that nature itself discards spontaneously some of its defective products. Unfortunately, being as uncertain in its calculations as we are, if not more so, it does not discard them all. Neither is there, beyond a certain point, any exemplary scale in what it discards: some of those which it spares are among the most gravely handicapped. Furthermore (except in conditions fatal before puberty) nature seems not to check the descent of defective genes from generation to generation, which is one of the goals (with recognised limitations) of medical genetics. The question then follows: if genetic screening with selective abortion is in fact a rationalised adjunct to natural processes, are there ethical controls to guide it?

It seems to me that once we repudiate (as I must) the absolutist prohibition on abortion, the ethics must be wrought in a continual tension between a general presumption in favour of life and the strength of a claim to rebut it in any particular case. The claim must not be trivial if it is to prevail, and it should have respect to human life in its relational capacity as well as in its biological reality. The solution may well be worked out in a tension between the genetic counsellor and the potential parent, the one sharing something of a professional ethics in the matter, the other something of the wide spectrum of attitudes current in our society.

I suggest further that we only make the task harder if we pose the problem in the language of a conflict of rights. In hard fact, the fetus has no

rights, as the law and hard reasoning understand rights, until it is born alive. And English law does not confer any general right to an abortion: the Statute of 1967 defined conditions in which the termination would not be a criminal act. Progress would look more promising, I believe, if within the concept of a duty owed to the unborn child by both parent and physician—for duties can stand without the supposition of rights—the ethics are sought in a weighing of *interests*, those of the unborn child and those of the mother, and a consideration of how these interests are to be served, whether together or one at the expense of the other. One interest clearly to be served by genetic screening is that of a potential mother in an assurance, if it can be given, that her next child should be normal.

C.2.

We pass finally to the tension between individual persons, whether physicians or patients, and the society of which both are members.

The sciences of medical genetics and gene tracking now carry the study out into the patient's family, whose cooperation has to be won. Here again the language of rights will prove unhelpful. Human *wills* are involved, and they have to be wooed and won to serve the relevant interests. However, the search requires the sharing of knowledge gained in a clinical relationship, and here we meet the old questions: How much must be kept secret? How much may be told? How much should be told? Let us take this step by step.

The House of Lords in *Sidaway* established the duty of a physician or surgeon to give such information as would enable a patient to consent or not to any medical procedure proposed for him. I doubt whether that duty extends to giving a patient *all* information gained in clinical investigation, and particularly when no medical intervention is indicated. It is often asked: Has the doctor a duty to disclose the sex of a fetus prenatally determined? If the context were genetic counselling in relation to a sex linked disorder, the obligation to tell would seem clear: disclosure is relevant to a vital interest of the mother and of the unborn child. But where the interest is trivial or non-existent, the obligation, I would say, diminishes; it must remain discretionary. It would be hard to sustain a right to know, and a duty to disclose, the sex of the fetus simply in order for termination to be sought if it were not of the sex preferred. A multiplication of such terminations is likely to have social consequences in an imbalanced population structure, either absolutely between the sexes or timewise in the order of births. In some cultures that likelihood amounts to a certainty. The

social interest or common good must prevail against personal whim.

Next, a patient has an interest in the non-disclosure of genetic information to third parties, in relation, perhaps, to employment, insurance, social stigma, and the like. Without consent such disclosure would be unethical. But genetic disorder is not strictly a personal matter only. It has ramifications for sibs and consequences for future generations. It touches also the social interest in the health of a population. Without some sharing of information, the genetic tracking of families cannot be undertaken.

The language of rights is useless again. We are left with an obligation, first to establish trust between the patient and the medical geneticist, and confidence in the operation, and then to seek for willed consent to such disclosure as, step by step, will aid the serving of all the interests involved, personal and social, as they emerge.

Fundamental to medical practice, and especially to medical genetics, is what we call truth. The more the study moves out from the individual subject to the family, the more important is the assumption that social identity—who we believe we are—coincides with genetic identity—who genetically we

are. Uncertainty of parentage arising from human waywardness is morally irrelevant. If this be so, is it not time to call into question the insistence on anonymity for donors of gametes in the medical remedying of infertility? And to press for methods of registration which, while tolerant of acceptable fiction for those who want it, will enable fact to be disclosed to those who need it? If we believe in any coherence between rationality in science and moral rationality, not only medical disciplines in their complementarity, but also social policies in their consistency, should be grounded in what science professes to seek: truth.

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