

Ethical aspects of genetic counselling

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Editor's note

The author, a scientist working in clinical genetics, outlines the functions of the genetic counsellor and discusses some of the ethical issues involved. The paper is based on one given at a London Medical Group conference.

Procreation is error-prone

'Is my baby all right?' is one of the first questions a mother asks after she has given birth to a child. For most people, it is of more importance than the sex of their child. This question is asked, not without reason, for a whole range of abnormalities affecting all systems of the body is known to afflict Man, and around 1 in 30 children born have an abnormality, to a greater or a lesser extent, which requires medical attention.

However, those children who are born abnormal are merely the tip of the iceberg – they are the survivors, for many more abnormal babies are conceived but have already died prenatally.

There is a very high pregnancy wastage in Man. Gathering together all the relevant data it is possible to estimate (1, 2) that of 100 eggs which are ovulated and surrounded by sperm following coitus, 15 are lost because they are not properly fertilised, 10-15 fail to develop before implantation in the uterus, 12-33 die in the early post-implantation stage, 9-13 are aborted spontaneously as recognised miscarriages and 1 is lost as a perinatal death. Thus, of 100 eggs which are potentially fertilised, only 28-48 survive to be born.

This very high prenatal mortality is largely associated with abnormalities in the conceptus. Surveys of spontaneous abortions (miscarriages) show that a high proportion of the abortuses are abnormal (3, 4, 5) and the earlier the abortion occurs, the more likely it is to be abnormal. At 22 weeks gestation, approximately 5 per cent of miscarriages are chromosomally abnormal, at 16 weeks 30 per cent, while at 8-12 weeks the figure is around 60 per cent. In addition to chromosomal aberrations, developmental abnormalities are common and the incidence of, for example, neural tube defects, amongst spontaneous abortions is ten times higher than amongst neonates (6).

Key words

Genetic counselling; genetic diseases; prenatal diagnosis.

Thus, human procreation is associated with a rather high degree of error in that which is created, but the mistakes are largely eliminated by the natural means of failure of development and spontaneous abortion, and their live-birth is avoided. A small proportion, however, do somehow survive, and these form the congenitally abnormal group of the birth statistics, the 1 in 30 newborn who have some defect. It is not unreasonable for parents to hope that their children will be born healthy and normal, so what has medicine to offer to prevent this abnormal residuum?

Genetic cause of abnormality

While some congenital anomalies are sporadic and are unlikely to occur again in the same sibship, many do have a tendency to recur in families. This is because there is a genetic element in their aetiology. The likelihood of recurrence can be measured, the actual magnitude depending upon the mode of inheritance of the condition (the strength of the genetic element in the aetiology of the condition). Some disorders have a strong genetic element in their aetiology, whereupon the chance of recurrence is high (1 in 2 or 1 in 4), while in others the genetic contribution is less and the recurrence risk is correspondingly lower (for example, 1 in 25 or 1 in 40). It is one of the functions of medical genetics to determine the inheritance of genetic disorders and to define the recurrence risks for each one. It is one of the tasks of the genetic counsellor to transmit this information in a comprehensible way to families which bear such inherited conditions.

Role of genetic counsellor

Usually, genetic disorders cannot be cured; often they are grave and ultimately lethal conditions associated with much suffering for both the patient and his family. A genetic counsellor cannot ensure that a couple *will* have a normal child, for the reassortment of genes in procreation is a chance phenomenon, but the genetic counsellor can ensure that the information which comes out of genetic studies of disease is given to those primarily concerned, that is, those at risk for the disease – to whom it is of immediate practical importance so that they can plan their futures in an informed

way. Genetic counselling is an unusual form of medicine in that it cares for its patients not by the more conventional remedial practice, but by imparting information.

Before the facts are given, it is essential that the counsellor first establishes the correct diagnosis of the condition affecting the family which is consulting him or her. While this may be straightforward in some cases, it is more complicated in others. For instance, there are several forms of both diabetes and epilepsy, some of which have a negligible recurrence, others of which have a somewhat higher risk. Thus it is important that a precise diagnosis is made, so that information pertaining to the form of the disease diagnosed and not to another form of the particular disease, is given. Genetic counselling is worthless, and even harmful, if it gives advice on the wrong condition.

Similarly, there is little to be said in favour of genetic advice if it is incomplete. Consequently, before any information is given, it is essential to discover if there are further genetic conditions in the family or other predisposing factors which are likely to create problems in the future, so the taking of a detailed family history is another essential preliminary.

Non-directive counselling

When imparting the sometimes rather complicated scientific information, it must be done in a manner appropriate to the intellectual level of the patients, being explained several times over if necessary to ensure as complete a comprehension as possible, for factors such as 'risk' and 'probability' may well be entirely new concepts. Then other topics are introduced, such as the availability of prenatal diagnosis, if it exists for the disorder in question, together with other possible options open to the couple, such as artificial insemination by donor or perhaps sterilisation. During this information-giving process, many important issues are therefore touched upon, such as future reproductive plans, birth control and abortion, about which personal decisions must be made. The counsellor is bound to have his own opinion on these matters, but it is vital he keeps them from those consulting him, for it is their lives and futures which are involved. Thus the counselling should be non-directive. The counsellor's job is to inform people of their risk of having a child with a particular condition, of their chances of having normal children, and of what other options are available to them, so that they are in a position to judge and to decide for themselves whether to embark on a pregnancy or not. For they alone are aware of their hopes and aspirations, their religious and cultural background, and the many factors other than the genetic ones which impinge on their lives and which will affect their ability to deal with the future should there be a handicapped child to care for.

Impartiality on the part of the counsellor is essential because however much he may think he knows the aspirations of his patients, he may not always do so.

This is strikingly demonstrated by a couple, both of whom had achondroplasia (a particular form of severe dwarfism) who sought genetic advice (R Harris, personal communication). It is usual to assume that couples who consult are hoping that they will have normal children. However, it transpired that what was important to this particular couple was that they should have children similarly affected to themselves. They saw their condition as a positive advantage, for they had secure and well-paid employment as circus clowns and they hoped that this profession would be perpetuated in their family.

This variability in people's attitudes, which cannot be predicted, is also seen in the case of two adult female patients with spina bifida. One was in a wheelchair because she was totally paralysed from the waist down and she also had an ileal bladder. She said she would take the risk of possibly having a similarly affected child, because it was no problem being only 'mildly affected' as she was. The other patient was able to walk, albeit with a caliper on one leg, and had no other defects. But by contrast, she was adamant that she would have no children because she didn't want a child of hers 'to suffer as she had'. To the observer, this patient had few problems compared with the former. In reality, the experienced problems of these individuals were the reverse. Thus decisions must not be made by the genetic counsellor on behalf of the patient, for he knows only the medical and scientific facts about the individual.

Information giver or adviser?

Thus genetic counselling provides basic information to equip people to make decisions for themselves, about themselves, and avoids supplying a specific course of action for people. However, many come to a consultation wanting, or expecting, the latter. When this happens, the counsellor should be supportive and encouraging, amplifying the discussion so that the patients may see where their own thoughts and feelings are leading them. The line is fine, but it is possible to guide without exerting influence.

Do patients make a choice?

There is evidence that people do use the information given them in genetic clinics to make decisions and modify their future reproductive patterns. The degree to which they do this seems to be related to the level of risk of the disorder involved. Three different surveys (7, 8, 9) have shown the same trends: of couples who were at 'high' risk (a greater than 1 in 10 chance of having an abnormal child), 64 per cent, 82 per cent and 55 per cent decided not to have any more children, while of couples in the 'low' risk category (chance of having an affected child less than 1 in 10), only 24 per cent, 39 per cent and 26 per cent decided against another pregnancy. Another follow-up study (10) has shown that almost all patients considered extremely

seriously whether or not to proceed with another pregnancy, and those who had no further children did so because of the risks or potential burden, or both, of having an affected child, rather than because they had completed their families. Over one-third of couples who decided against more children had been sterilised within two years of counselling.

The option of abortion

It is only within the last decade that the option of abortion has entered this field. For certain genetic disorders, there is the possibility of prenatal diagnosis in the mid-trimester, and if desired, the termination of affected pregnancies. This has expanded and altered the genetic counsellor's role, for before, he had to stop at probabilistic counselling; now he has, in some cases, something more to offer. If parents can bear the idea of undergoing diagnostic tests and possibly an abortion, then they can be largely helped to have only normal children. Thus, in the past, if a couple wanted reliably to prevent a genetic disease occurring in their family, they had to avoid having children of their own. Now, the facility for reproduction is restored to such couples if they find it possible to accept the option of abortion. Genetic disease can now be prevented in some cases by eliminating affected fetuses.

In the absence, as yet, of any reasonable means of primary prevention or cure of genetic disorders, the evidence is that except where there are religious objections, mid-trimester abortion is an acceptable form of management of genetic disease for most people. Of course opinion is vigorously divided about the moral permissibility of abortion and clearly there is usually no point in offering amniocentesis and prenatal diagnosis of genetic abnormality to women who believe that abortion is morally unacceptable. Equally clearly genetic counsellors would not be working in the field if they themselves believed that abortion was always morally unacceptable. They tend to believe, as their patients usually do, that it is preferable to giving birth at term to another affected child (11). For many couples a therapeutic abortion has enabled them to have normal children which they otherwise might not have had. The positive effects of prenatal diagnosis on one such group is clearly seen in a survey of couples in Wales at risk of producing children with spina bifida (12). Before prenatal diagnosis was available, only 5 per cent of couples embarked upon another pregnancy within a year of genetic counselling and 50 per cent had decided to have no more children. After prenatal diagnosis was available, 50 per cent became pregnant within the first year and only 25 per cent decided to have no more children.

Individual's right to choose

Prenatal diagnosis has been widely used to detect chromosome abnormalities in the offspring of elderly mothers. There is a significant correlation of Down's syndrome, in particular, with advanced maternal age.

In discussing the availability and implications of these diagnostic tests, the genetic counsellor must ensure that the patient herself freely makes the choice whether to be tested or not, and, should an abnormal fetus be diagnosed, whether to have a termination or not. Many, but by no means all, women over the age of 38 years accept prenatal diagnosis if it is offered to them, and most, but not all, of those in whom a Down's fetus is detected ask for a termination.

Patient's autonomy versus doctor's responsibility not to harm

A by-product of these chromosome studies is that certain information, other than that being directly sought, is also obtained, namely, the sex of the child and the existence of chromosome abnormalities other than Down's syndrome. In principle, a mother should be given all available information concerning her unborn child and originally she was told the sex. However, as time went on, it was found that around half of the mothers tested in this way did not want to know the sex, they preferred to wait until the moment of delivery. Consequently, it now seems better if this information is given only to those who actually request it.

There are other reasons for withholding such information for, in some cases, it has even been found to be harmful. This happens more often in the older woman who perhaps has several children already, and who often has a definite preference as to the sex of the next baby. Also sometimes this late pregnancy is unplanned and she may be ambivalent about it. If the sex of the new child turns out to be contrary to the wishes of the mother, this disappointment is usually largely mitigated once the baby is in her arms. If, however, news of the undesired sex is received four months before there is actually a baby present to preoccupy and stimulate her, then it can upset her during the remainder of the pregnancy and be detrimental to the subsequent mother-child bonding. It seems that it is incumbent upon a genetic counsellor to assess carefully the possible consequences upon a patient of divulging certain information which he has at his disposal.

It seems best to tell parents if chromosome abnormalities other than Down's syndrome are discovered. One reason why the information might be withheld is that sometimes the conditions are virtually inconsequential, and if parents know that their child does not have a wholly normal chromosome constitution, they may 'brand' him as 'abnormal' or 'deviant' every time he is naughty, which may then actually encourage behavioural problems. However, many chromosome abnormalities other than Down's syndrome are associated with pathological states and what must accompany news of the chromosome findings in these cases is what phenotypic abnormalities are associated with the particular condition, and how often they occur; again so that the parents can decide what to do in an informed way. Although most people know about Down's syndrome and its implications, rarely will they know about

other autosomal trisomies, sex chromosome aberrations and chromosome translocations, deletions and duplications.

The question of fetal sex arises in another context. In the case of severe sex-linked disorders such as Duchenne muscular dystrophy, fetal sex determination is the primary indication for prenatal diagnosis. A male fetus of a carrier mother stands a 50 per cent chance of having the disorder and in the absence of a test which can identify those which are affected, termination of all male fetuses is offered to such women. However, some women who are not at risk from a sex-linked disease demand prenatal diagnosis to determine the sex of their fetus, with a view to terminating the pregnancy simply if the fetus is the 'wrong' sex. On the grounds of the patient's autonomy, including her right to determine her own future, it might be argued that such measures should be allowed. However, prenatal diagnosis is never even contemplated in such cases, for simply being the 'wrong' sex does not constitute legal grounds for abortion.

This paper has considered only one aspect of genetic counselling, namely that concerned with parents or individuals who wish to reproduce but who have a known chance of having an abnormal child. However, this comprises most of a genetic counsellor's work.

That genetic clinics continue to have full appointment books suggests they are seen as serving a useful purpose. Properly conducted genetic counselling makes a welcome contribution to medical practice. However, it must be performed in specially designated clinics by trained personnel. A casual remark on chances of recurrence by someone in another clinic can be harmful, for there should be a full explanation and the opportunity for discussion. There is much to be said in favour of good genetic counselling for those who request it; there is much

to be said against poorly given genetic advice, especially if patients do not really want it.

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