Outcomes and process in genetic counselling


Although it may be simple to evaluate some elements of clinical genetics, it is difficult to evaluate genetic counselling. We review previous studies of the outcomes of genetic counselling; although the methods used may be valid in research studies, there are practical and ethical difficulties in applying them to the measurement of clinical effectiveness in standard practice. No simple measures of outcomes would be suitable. Research evidence will be helpful in deciding what services it is appropriate to offer, and the quality of a service can then be assured by assessing the quality of the clinical process in three ways: 1) adherence to agreed protocols and standards of care; 2) peer review and audit of clinical activity; and 3) ongoing review of the satisfaction of clients and referring physicians with the service. The assessment of client satisfaction will need to be a sophisticated form of retrospective satisfaction with the service provided, and such a scheme has yet to be fully developed.

The evaluation of medical services on the basis of the outcomes of clinical practice is recognised as essential (Delamothe 1994) as a research activity and as audit of practice. For many branches of medicine where clinical practice is essentially therapeutic and diagnostic only in so far as is necessary for therapy, the measurement of outcomes will be straightforward – in principle, if not always in practice. Evaluation of the physical symptoms suffered by the patient will be the essential means of evaluating the outcomes of the service provided.

Clinical genetic practice does make use of accurate diagnosis, where one is available, but is not generally involved with therapy. In fact, our "patients" are generally healthy. Rather than treatments, our service provides information, explanation and options for reproduction, genetic prediction and health surveillance. Apart from the surveillance for complications of genetic disease, these activities would not be expected to alter the physical condition of our clients. We dispense words, not tablets.

How, then, should the evaluation of genetic counselling be conducted? Should we define the measure of our success by how well our clients recall the information that we have passed on to them? Or with how genetic counselling has altered their reproductive plans? Or how they have in practice chosen to act on the basis of such information?

In this paper, we consider how to evaluate the genetic counselling of individuals and families; we do not examine the rather different contexts of population genetic screening or of the surveillance for treatable complications of genetic disease.

Information as outcome measure

Studies of the recall of information given in genetic counselling generally indicate that the information is provided in an effective manner. Thus, Somer et al. report that 80% of 791 families counselled in Finland had an adequate knowledge of the mode of inheritance, and 74% of the recurrence risk (Somer et al. 1988). Kessler has reviewed other studies in which pre-counselling and post-counselling knowledge of reproductive risk and of diagnosis are compared; genetic counselling appears to be effective at increasing knowledge (Kessler 1989, Kessler 1992). The major study by Sorenson et al., however, has shown that there is a great potential for improving the effectiveness of information giving (Sorenson et al. 1981a). Knowledge may not be retained so well after genetic testing carried out in a population carrier screening programme, such as for cystic fibrosis, in which there may be less...
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emphasize upon individual counselling (Bekker et al. 1994). High levels of long-term recall of information were found, however, after an antenatal carrier screening programme for the haemoglobinopathies. This may result from the provision of pre-test information in a novel or more stimulating form (by video) or because the greater anxiety present in an antenatal context reinforced the memory (Loader et al. 1991a, Loader et al. 1991b).

Although counsellors do recall many items of information correctly after counselling, it would be difficult to use this as a measure of the effectiveness of the service. This would entail pre-judging the purpose of genetic counselling as understood by the clients, who may not value specific items of information as highly as professionals. Furthermore, it has long been recognized that risk information is interpreted by counsellors rather than simply being recalled as neutral facts (Pearson 1973, Evers-Kiebooms & van den Bergh 1979). Clients tend to view risks in binary form — as either destined to happen or not to happen — and process the numerical information in ways that are heavily influenced by the anticipated social consequences of their reproductive decisions (Lippman-Hand & Fraser 1979a, Lippman-Hand & Fraser 1979b). Similarly, carriers of a serious X-linked disorder experience this fact in different ways at different life stages, depending upon the relevance of the information at the time; what is retained is the personal meaning of the information they have been given (Parsons & Atkinson 1992). It is also known that the way in which risks are presented (framed) can influence the interpretation of recurrence risk figures (Shiloh & Sagi 1989). The evaluation of a risk as “high”, “moderate” or “low” may also differ greatly between counsellor and counsellor, depending in part upon the numerical risk of recurrence and in part upon other factors such as the perceived burden of the disorder (Emery et al. 1979). Clients often interpret risk figures more optimistically (as lower) than do counsellors, but a variety of individual and family factors modify these interpretations (Wertz et al. 1986a). There are simply too many subjective and variable factors involved for a service to be evaluated by the recall of risk figures or other items of information by its clients. There may well be occasions on which such recall could be assessed as part of a research study, but not as a measure of the effectiveness of a regular service.

Reproductive plans as outcome measure

It is possible to examine the reproductive intentions of genetic counselling clients before and after counselling. In one study by Wertz and Sorenson (1986b), 43.5% of 628 counselees indicated that their reproductive plans were influenced by the genetic counselling, although 52.7% of those whose plans were reported to have been influenced by counselling held the same plans as they held before the counselling. Those who had come to counselling to get information for making a decision, those who discussed their decision in depth with the counsellor, and those with higher education were more likely to report that the counselling influenced their plans. It is not entirely clear, however, in what sense clients were influenced by the counselling if their reproductive plans did not alter, and reproductive uncertainty will often not be resolved by genetic counselling because so many of the factors that contribute to a decision are social rather than clinical (Wertz et al. 1984).

One important factor that certainly does influence reproductive plans is the availability of prenatal diagnosis. In the absence of prenatal diagnosis for a disorder, reproductive plans correlate with the perceived burden of the condition rather than the statistical risk of recurrence (Sorenson et al. 1987). In counselling couples at risk of neural tube defect, the advent of prenatal diagnosis had a major impact, increasing the number willing to embark upon a pregnancy (Morris & Lawrence 1976). More recent studies confirm that the availability of prenatal diagnosis is important in helping families at high risk (≥15%) of a child with a severe disorder in deciding to have further children (Frets et al. 1990). Couples who already had an affected child were less likely to try again in the absence of prenatal diagnosis. Other important factors were the desire to have children and personal experience of a close relative affected by the disorder.

Could the influence on reproductive plans be used as an outcome measure in genetic counselling? There would be many problems in using it in this way. While this may be a valid topic for research, it would not be helpful as a measure of outcome for a clinical service. First, as with recall of information, to use reproductive plans as an outcome measure for genetic counselling is to pre-judge the service that is wanted by client families. Some of them will want information that will inform their reproductive planning, but there may be many other reasons for seeking a referral to a clinical geneticist. Second, there are far too many confounding factors that influence reproductive plans for this to be a measure of the effectiveness of a clinical service. Third, the choice of time-frame in which to assess the impact on reproductive plans would be arbitrary. Finally, reproductive plans are essentially hypothetical, and would be a poor
proxy for reproductive behaviour. Caution is clearly required in the interpretation of stated reproductive intentions, given the findings of studies comparing the hypothetical with the actual uptake of genetic tests in two different contexts – predictive testing for Huntington’s disease and the rate of utilisation of prenatal diagnosis for cystic fibrosis. In both cases, the actual uptake of testing was much lower than the uptake predicted from hypothetical attitude surveys: 10-15% vs 60% for Huntington’s disease (Tyler et al. 1992, Harper et al. 1993); 17% vs 51% for cystic fibrosis (Jedlicka-Kohler et al. 1994).

Reproductive behaviour as outcome measure

Some attempts have been made to report the effects of genetic counselling on long-term reproductive behaviour. Genetic counselling can be followed by an increase in pregnancies, irrespective of reproductive risk, even when prenatal diagnosis is not available (Sorenson et al. 1987). Reproductive risk and the availability of prenatal diagnosis have the expected effects (Sommer et al. 1988). Controlled studies of reproductive behaviour after genetic counselling, however, are almost totally lacking, and there are no reports of randomised trials examining post-counselling reproductive behaviour. One controlled study of 46 families with a child with Down’s syndrome showed no influence of genetic counselling on reproductive behaviour over at least 18 months from the birth of the affected child: 9 in each group of 23 families initiated at least one further pregnancy, and only 3 of the 18 couples utilised prenatal diagnosis (two in the counselled group, one non-counseled) (Oetting & Steele 1982).

There are very good reasons why this area has hardly been investigated. There are both practical and ethical problems in using reproductive behaviour as a measure of the effectiveness of genetic counselling. While it may be possible to gather outcome data for families seen because of concern during an ongoing pregnancy, there are severe practical difficulties in using reproductive outcomes in other contexts. These include the duration of follow-up required, and the inability to gather these data as a part of routine service operation. Also, it is clear that the relationship between reproductive risk and reproductive behaviour is complex, and mediated by many different social factors in addition to the medical ‘facts’ (Parsons & Atkinson 1993).

There are also serious ethical difficulties in any use of reproductive outcomes as a measure of the effectiveness of genetic counselling. While it is perfectly legitimate research to assess the impact of genetic counselling on reproductive plans and behaviour, it is altogether different to monitor the effectiveness of the service in this way. That would entail imposing a value judgement on particular behaviours, declaring the birth of an affected child as undesirable, and the uptake of prenatal diagnosis and the termination of an affected fetus – or the childlessness of the at-risk couple – as desirable. Such judgements are unacceptable, and must be left to individuals. As Harper states: “It is also important that those giving genetic counselling do not judge ‘success’ or ‘failure’ in terms of a particular outcome, and that they give support to families whatever their decision may be” (Harper 1988). For clinicians to know that their work would be evaluated in this frankly eugenic manner would impose intolerable pressures on them. Clinicians would feel anxious that their clients make the “correct” reproductive decisions. Health service purchasers would doubtless regard the correct decisions as those that imposed least monetary cost on the health service, and so clinicians (obstetricians, perhaps, as well as geneticists) could be regarded as failing in their duties if too many of their patients had infants affected by disorders that could have been diagnosed prenatally. This would lead to a conflict between the clinicians’ duties to their patients and to their employers, and would rapidly bring their service into well-deserved disrepute. Followed to the logical extreme, this policy would lead to a recommendation that pregnancy be terminated for any fetus likely to cost more in medical care and social benefits than he or she would pay in taxes over his or her lifetime. But why should genetics services be expected to generate financial savings in this way, when the provision of all other aspects of clinical medicine are accepted as costing money? These issues have been discussed elsewhere (Clarke 1990, Clarke 1991, Modell & Kuliev 1993).

A more sophisticated means of evaluating genetics services relating to reproduction might be to count the “informed reproductive choices made”, regarding informed choice as itself a benefit (Modell & Kuliev 1993). It would be very difficult to count the number of reproductive choices made by those who choose to have no more children, or who choose to proceed with pregnancies without prenatal diagnosis or without further discussion with genetics services, so that it might be difficult in practice to distinguish this approach from the previous one. Furthermore, it is very difficult to regard choice per se as the goal of a clinical activity (Chadwick 1993, Clarke 1993) – this would lead to a proliferation of progressively less useful tests. The numbers of births of healthy children who would not have been born without prenatal diag-
nastic testing could be another measure of reproductive outcome (Beech et al. 1992), although it would be difficult to obtain suitable control data to validate the approach. Such studies may be possible as research projects, but not as a routine means for the evaluation of clinical genetics services.

We have to accept that crude, numerical measures of success are simply not applicable in the field of reproductive genetics. We need instead to adopt measures of outcome that accord with the goals of genetic counselling. The prime goal could be formulated as the clarification of the client's reproductive risks and options, where this was wanted by the client – or more generally, the promotion of the client's understanding of their genetic situation. How does this relate to the goals of genetic counselling in general?

What then is genetic counselling?

At this point, we must consider what takes place in a genetic counselling clinic. Genetic counselling can be defined as: “An educational process that seeks to assist affected and/or at-risk individuals to understand the nature of the genetic disorder, its transmission and the options open to them in management and family planning” (Kelly 1986). This definition makes it clear that genetic counselling is a process centred on the clients and their need to understand the condition in their family. Because of this focus on understanding and education, it is essential for the professionals to listen to their clients, to explore their present understanding and their questions and concerns. Reports of genetic counselling in practice show that this process of communication has often broken down, so that counsellors are frequently unaware of the issues that the clients want to discuss, and these issues are therefore often not addressed (Sorenson et al. 1981b, Wertz et al. 1988a). It is interesting that the concerns of female clients attending genetic counselling on their own were more likely to be addressed by female than by male counsellors (Zare et al. 1984).

The ethos of genetic counselling, then, is for the clients to set the agenda, and the first element of genetic counselling must therefore be listening (Clarke 1994). The clients’ questions may focus on the diagnosis or prognosis of the condition being considered, or on reproductive risks and options, or on other issues. The structure of a genetic counselling service is clearly important in deciding whether or not the client’s concerns will be heard, in turn deciding whether or how a client’s concerns will be addressed.

The next element is the process of clinical assess-

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ment that allows the questions to be answered, followed by communicating the relevant information to the family. Then, for some clients, there is the facilitation of decisions – scenario-based decision counselling – that helps clients to consider the practical and emotional consequences for each of the different possible outcomes of any reproductive or testing decisions that confront them. Finally, there is the provision of ongoing support where this is appropriate.

This model of genetic counselling operates through the process of clinical service that we provide. After a referral is received, our first point of contact with the client is usually a visit to the family home by the local genetics nurse. At this visit, there is a two-way exchange of information. The nurse asks about the family’s questions and concerns, and also gathers information about the condition or event in the family that has prompted the referral, and more generally about the family history. Who in the family has been affected? Are previous medical records available on other relevant family members? Will it be possible to obtain their consent for these records to be reviewed? Gathering this information in advance often reduces the length and number of clinic appointments required by the family. At the same time, the genetics nurse is able to explain the ethos of the service, often reassuring the family that we will not be telling them what course of action to take, and she will also explain the limitations of the service – for example, that we may not be able to arrive at a firm diagnosis to account for a child’s developmental delay, or that the prognosis for an infant with a particular congenital anomaly may be uncertain.

The next point of contact is usually a clinic visit (or a few visits) to see the nurse and clinical geneticist. The family’s questions and concerns are elicited once more in the clinic – they may have changed since the preliminary discussion with the nurse. The clinician asks the necessary clinical questions, and performs the appropriate physical examination and investigations, so that the client’s questions can be answered as far as possible. The clinician’s conclusions are explained to the clients and discussed with them. If the clients are confronting important decisions – perhaps in relation to predictive testing, carrier status testing, or testing of an ongoing pregnancy – then the various options will be discussed, and the implications for the clients and their families of the possible test results, or of not having the test, will be explored. Finally, arrangements will be made for future contact where appropriate. This may involve contact with other members of the client’s family, a planned prenatal diagnosis, the provision of sup-
port after a termination of pregnancy or for those living at risk of serious disease, or a review for diagnostic reassessment or for surveillance for early signs of complications of the disorder.

There are other aspects of clinical genetics, but they do not fit so well within this description of genetic counselling. Thus, clinical genetics can be taken to include the coordination of surveillance for early signs of potentially treatable complications of such conditions as familial cancers and Marfan syndrome. The evaluation of these aspects of clinical genetics fits much more readily into standard assessments of outcome, because they relate to physical health. The provision of population screening for some genetic conditions, for example, newborn screening for phenylketonuria and hypothyroidism, antenatal screening for Down's syndrome, or carrier screening for recessive disorders, is also not a part of family-centred genetic counselling although such screening programmes are a part of clinical genetics in the broader sense.

**Satisfaction with genetic counselling**

Could satisfaction with the process of genetic counselling, as outlined above, be used as an outcome measure? Counsellor satisfaction with counselling sessions has been reported to be very high (Wertz et al. 1988b), and appears to depend upon the perception that the counsellor has communicated knowledge successfully—preferably to better-educated clients—rather than to any meeting of the clients' needs. For satisfaction to be considered as an outcome of genetic counselling, it will be necessary to focus upon the satisfaction of clients, not counsellors. Few attempts to measure client satisfaction with genetic counselling have been reported, however, and a considerable problem could be dissatisfaction associated with the lack of available information or with unwelcome information provided in the course of genetic counselling (Shiloh et al. 1990, Mushlin et al. 1994) blaming the messenger. Simple measures of client mood or contentment after the counselling will obviously be inadequate. Such measures as a short form of the Spielberger State-Trait Anxiety Inventory and the General Health Questionnaire have been used to assess the psychological impact of population screening programmes, and have generally shown that subjects are stressed when given unwelcome information (for example, that they are a carrier of cystic fibrosis) but that this distress is only moderate and is not sustained (Bekker et al. 1993, Mennie et al. 1993). Even in that context, the "psychological consequences [of population carrier testing] are unclear" (Martea 1994). Such measures are simply not appropriate in the much more varied contexts of generic genetic counselling. The sharing of information in genetic counselling may often cause sustained and thoroughly appropriate distress, as when an adult, child or fetus is diagnosed as having a serious, degenerative disorder. What measure of satisfaction, or emotional well-being, could possibly be appropriate to the varying circumstances of a genetics clinic?

The possibility of using a sophisticated form of Quality of Life (QOL) evaluation for genetics services has been raised (Johal 1995), but would involve such a simplification of complex outcomes that it would inevitably lead "to unwise or unfair decisions" (Spiegelhalter et al. 1992). Multidimensional measures of QOL would certainly be more suitable than any single, global measure of QOL (Jenkins 1992), but will still be inadequate to permit valid comparisons of health gains resulting from genetic counselling with those resulting from other areas of clinical medicine.

Shiloh and colleagues have devised a scale for the evaluation of instrumental, affective and procedural aspects of genetic counselling (Shiloh et al. 1990), but this evaluates the process of genetic counselling rather than clear end-points or outcomes. Such measures will need to be refined, but hardly answer the question of what is achieved in genetic counselling. If no diagnosis is achieved for a child with serious disabilities, for example, the clinician may not be at fault (there may be no diagnosis to make) but the parents may well feel dissatisfied (Abramovsky et al. 1980).

It is clear from a consideration of patient satisfaction surveys in general, not just in genetics, that attempts to assess global levels of satisfaction with a service will not be helpful in identifying either the strengths or the weaknesses of a service (Williams 1994). Instead, attention must be focused on specific aspects of performance wherever possible (Bruster et al. 1994). Few such studies have been conducted in genetic counselling. It has been shown that a routine visit by a genetics nurse to the family home after genetic counselling does not improve the client's recall of information (Curtis et al. 1988). It has also been shown that the use of structured scenarios in genetic counselling can be very helpful (Arnold & WInsor 1984), and also a potentially powerful way of shaping decisions (Huys et al. 1992).

A patient satisfaction survey carried out in our unit through the Hospital Audit Office has also indicated a very high degree of satisfaction with the role of the genetics nurse allocated to each family referred for genetic counselling in Wales. Questionnaires were posted out to two cohorts of patients, and 59% of 98 replied. Referral was in-
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Beyond the simplistic, quantitative measures of client satisfaction with health care rightly criticised by Carr-Hill (1992) and Williams (1994). This could generate a much more valid measure of clinical worth than a client satisfaction questionnaire administered or posted to clients soon after a clinic appointment.

The way forward through looking back?

The principal tool for evaluating genetic counselling, therefore, may have to be the confrontation of clients with three questions:
1. Were you satisfied with the service provided at the time? Was the service efficient and convenient to use? – incorporating the dimensions of satisfaction of Shiloh et al. (1990);
2. What changes have occurred in your expectations of the genetic counselling service since your referral? – a measure of the clients' education about genetic counselling and of their adjustment to their genetic situation;
3. With hindsight, how satisfied are you with your experience of the genetic counselling service? If you went through a diagnostic process, was this of any use or value, even if no diagnostic certainty was achieved? Did the service meet the expectations that you now think you "should" have had at the time of your referral?

Adopting this approach to the evaluation of genetic counselling would enable evaluation to be an ongoing part of service activity, requiring few additional resources. The necessary initial data can be gathered as a routine by the genetics nurse at her first contact with each family, and the retrospective assessment of the service by each client could be incorporated into a standard questionnaire sent to each client at the end of a series of genetic counselling consultations. In addition to this continuing feedback from clients, it would also be necessary to carry out peer review of service quality and clinical audit, as in other units, and to contact the referring doctors to discover how satisfied they have been with the service provided to their patients.

Simpler measures of outcome may be superficially attractive but are neither methodologically nor ethically acceptable. However, while the type of evaluation proposed would provide some subjective outcome data, it must be recognised that it still fails to provide a global outcome measure for the entire process of genetic counselling.

We are claiming that genetic counselling deals with information where other physicians prescribe treatments, and so our services must be evaluated in a distinct fashion. Genetic counselling may be particularly suited to the purchasing of protocols

Quality of process in genetic counselling

Similar considerations apply to the evaluation of counselling and support services in other contexts such as primary health care, where simple measures of well-being are also inadequate as measures of outcome (Hazzard 1993). In such contexts, where no medical interventions are prescribed and where the activity being evaluated is primarily a process of communication, it may be more appropriate to rely upon research evidence to decide what services to offer, and then to assess the quality of the process of the service provided rather than continuing to grope towards elusive measures of outcome.

The inadequacy of relying on statements of satisfaction with a service made by naive clients has been emphasised by Calnan in his development of a framework for the lay evaluation of health care (Calnan 1988). If satisfaction is to be assessed, it would only be appropriate once the client is sufficiently informed to make a reasonable judgement about the quality of the service provided. For genetic services, clients may be in this position once the process of genetic counselling is completed.

This leads us to propose a "retrospective assessment of satisfaction" approach, which moves beyond the simplistic, quantitative measures of client satisfaction with health care rightly criticised by Carr-Hill (1992) and Williams (1994). This could generate a much more valid measure of clinical worth than a client satisfaction questionnaire administered or posted to clients soon after a clinic appointment.
of care rather than of activity as such (Sheldon & Borowitz 1993). We should be judged by our choice of process, and by how well we meet the appropriately specified service standards, and not by attempts to monitor reproductive or other outcomes in this delicate field. This argument is a form of special pleading, but it will be recognised as valid by, for example, those health economists who recognise that monetary valuation of prenatal screening is unsuitable, so that cost-benefit analyses of these services have to be excluded (Phin 1990).

In response to the manifest inadequacy of outcome measures in genetic counselling, Kessler has suggested that we need to focus on the process of genetic counselling itself: "We probably have come almost as far as we can in terms of applying outcome methodology and it may be time now to focus on the counselling process itself ... Perhaps then we can assess where success is being achieved and where remedy is needed" (Kessler 1992).

Research into the process of genetic counselling is required to enable us to describe and evaluate different models of genetic counselling services. Purchasers will then be able to use the research evidence to decide which model of genetic counselling to select. The need for the monitoring of services to focus on quality issues and not outcomes will then be much more acceptable to purchasers, and we will no longer need to plead a special case.

This discussion also touches on a broader debate about the role of qualitative methodologies in public health analyses. Current practice is to focus largely on quantitative methods, assuming that benefits exist only if they can be counted, while qualitative methods are also required if purchasing decisions in the National Health Service are to do justice to the array of competing claims in certain supremely sensitive areas (Williams 1994, Baum 1995). Clinical genetics services comprise one such area of clinical practice in which the need to incorporate qualitative assessments of process into the evaluation of services is pressing because the exclusive adoption of simply quantifiable outcomes will result in great harm to patients and families.

References

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