
Response to: What counts as success in genetic counselling?

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Author's abstract

Clinical genetics encompasses a wider range of activities than discussion of reproductive risks and options. Hence, it is possible for a clinical geneticist to reduce suffering associated with genetic disease without aiming to reduce the birth incidence of such diseases. Simple cost-benefit analyses of genetic-screening programmes are unacceptable; more sophisticated analyses of this type have been devised but entail internal inconsistencies and do not seem to result in changed clinical practice. The secondary effects of screening programmes must be assessed before they can be properly evaluated, including the inadvertent diagnosis of unsought conditions, and the wider social effects of the programmes on those with mental handicap. This has implications for the range of prenatal tests that should be made available. While autonomy must be fully respected, it cannot itself constitute a goal of clinical genetics. The evaluation of these services requires interdepartmental comparisons of workload, and quality judgements of clients and peers.

Audit is a necessity for any system of medical care, and that includes medical genetics. But what measure(s) of outcome can we employ to evaluate medical genetics services? What counts as success in genetic counselling?

As discussed by Ruth Chadwick in her accompanying paper, I have drawn attention to some of the difficulties that would arise if health service management decided to adopt the numbers of terminations of affected pregnancies as the most appropriate measure of outcome (1). I am grateful for her support of my view that this is an unacceptable policy, but would like to comment on some of her other remarks.

Notwithstanding Chadwick's statement that her description of genetic counselling is not intended to be exhaustive, she does focus very much on questions of risks and options in reproduction. Furthermore, she claims that the activity of genetic counselling necessarily entails a concern with the

incidence of genetic disorders in the population. This view arises because a reduction in the birth incidence of individuals affected by genetic disorders is thought to be the only means of reducing the suffering associated with such conditions. Inevitably, this suggests that genetic counselling will encompass only a very narrow range of activities. In fact, this picture of genetic counselling is seriously misleading.

There are several ways in which genetic counselling can operate to reduce the suffering caused by genetic disorders. Genetic counselling can entail any of the following activities:

- 1) The achievement of an early, precise diagnosis of the condition causing concern, where possible, and hence an increase in knowledge and understanding. Even if unsuccessful, the attempt to achieve a precise diagnosis of the cause of a child's handicap can itself be therapeutic for the family.
- 2) The screening for complications of genetic disease, thereby assisting in the management of affected individuals. This already plays an important part in medical genetics work, and this role is likely to increase.
- 3) The provision of social and practical support for those individuals and families with genetic disease: the affected, those who might develop the disorder in the future, and those whose children have been, already are or might in the future be, affected.
- 4) The development and application of specific therapies for genetic disorders (including gene therapies).
- 5) The reduction of 'handicap', itself a social construct, by working to minimise the stigma associated with disability and handicap, hoping to develop the self-esteem of affected individuals. This may not seem to lie fully within the ambit of our professional activities, but we should certainly aim to avoid any actions that might indirectly damage the social status or self-esteem of those with mental handicap, and we may even be able to achieve some progress in this area by helping to influence public attitudes towards mental handicap.

Key words

Genetic counselling; medical genetics; audit; cost-benefit analysis; autonomy; termination of pregnancy.

In addition to:

6) The provision of information about future reproductive risks and options in particular family situations.

It can be seen that the question of reproduction is but one part of medical genetics/genetic counselling, and there are many other ways in which genetic counselling can diminish the burden of inherited disease. It is therefore perfectly feasible for a clinical geneticist to adopt accepted professional goals, and yet not be seeking directly to reduce the birth incidence of specific inherited disorders. A well known example would be the screening of newborn infants for metabolic disorders such as phenylketonuria, permitting the early treatment of these children with a diet that will permit them to develop intellectually as normal. Geneticists are eager to identify cases of such conditions, but are not generally aiming to reduce their birth incidence. In addition, like many of my clinical genetics colleagues, I spend many more of my working hours in attempting to establish the diagnoses of children I see with mental handicap, unusual physical features or specific disabilities, than in discussing reproductive risks and options with families, and I would not want our professional goals to be restricted to that area.

Another point worthy of discussion is Chadwick's brief dismissal of cost-benefit analysis as applied to medical genetics. The simple, monetary analysis of costs and benefits is clearly inappropriate when applied to prenatal screening and the termination of pregnancies, even from the perspectives of public health medicine and health economics (2). However, more sophisticated cost-benefit analyses of genetic-screening programmes have been produced, such as that of Modell and Kuliev (3). In this model, the main benefit is taken to be the making of informed choices by couples at risk of having a child affected by thalassaemia. The birth of an affected child to a couple who have declined prenatal diagnosis or termination of pregnancy is described as a benefit, not a cost of the programme. Termination of pregnancies is described as the main cost of the programme, not its core benefit. The overall effect of this apparent re-valuation of costs and benefits is to persuade one that some completely new, different genetic-screening programme must be on offer; in practice, however, the programme is functionally the same as any other, conventional carrier-screening and prenatal-diagnosis programme, only the packaging has been redesigned.

The analysis of Modell and Kuliev attempts to respect the autonomy of clients as much as possible, but tries to do so within the context of disease prevention as the desired goal (3). Prevention, however, is excluded from the formal, flowchart analysis of costs and benefits: it cannot be included as a goal

of the programme without changing the status of an affected child born after the parents declined prenatal testing, from a benefit to a wasted opportunity or a cost. Such analyses are more sophisticated than the traditional, purely monetary cost-benefit approaches; however, there are still contradictions in this approach to the prevention of genetic disease by the termination of pregnancy, especially in relation to autonomy and to our making judgements as to what counts as a worthwhile life. Furthermore, additional problems will arise if (as the authors intend) this methodology is applied to conditions causing mental handicap, major external malformation or specific physical disabilities. These conditions are intimately related to the personhood, the sense of identity, of affected individuals. Any programme aimed at the prenatal diagnosis and termination of affected pregnancies is liable to have secondary consequences for living affected individuals: it may affect the way that society values and provides for those with special needs, and how they value themselves. These issues are not raised in such a powerful manner by testing for disorders that do not threaten personhood. One can be a 'normal' person and have thalassaemia or cystic fibrosis: it is much harder to be accepted as essentially 'normal' when you are physically healthy but happen to have Down's syndrome, for example.

I would agree with Chadwick that autonomy is not a suitable goal for genetic counselling; rather it must be respected in the pursuit of whatever goals are identified. Indeed, it could be argued that autonomy in the field of prenatal diagnosis or screening, and the termination of affected pregnancies, should be curtailed. I have argued above and elsewhere (4) that we must consider the effects on living affected individuals of the existence of prenatal-diagnosis programmes aimed at the detection and termination of Down's syndrome or neural tube defect pregnancies: how does this affect the self-esteem of those with these conditions? Their social status? The extent of public provision for their special needs? Can we continue to adopt the consumer-choice model of prenatal diagnosis, as technology permits testing for progressively less serious disorders? As a profession, are we medical geneticists willing to be associated with terminations of pregnancy for largely cosmetic reasons? Arguing from the question of fetal sex determination, as also raised by Chadwick, I contend that society must determine what types of disorder are sufficiently severe to warrant prenatal-screening programmes with the termination of 'affected' pregnancies (4). There is a strong case for the claim that screening for non-progressive mental handicap of mild-moderate degree (for example Down's syndrome) should not qualify, although this argument does not apply so strongly to prenatal diagnosis offered to specific, high-risk couples: the secondary effects of these two activities are quite different. It is clear to me that such judgements are not simply

clinical, medical matters, and certainly not matters to be decided by cost-benefit accounting; it is important that they are acknowledged as social and political questions that warrant wide public debate.

Two other issues raised by the existence of widespread prenatal-diagnosis programmes are 1) Is valid informed consent obtained before screening tests are carried out? 2) Is adequate support provided for families who terminate affected fetuses? If the answer to either question is no, how should this be remedied? By the provision of more pre-test information and counselling, and improved post-termination support, or by the curtailment of the screening programmes? Once the prolonged grieving and remorse following terminations has been considered, have the prenatal-testing programmes achieved much net benefit? When the costs resulting from the inadvertent diagnosis of other conditions are considered (for example for Turner syndrome discovered at amniocentesis, which is usually fully compatible with good physical and mental health), what is the overall benefit? Such pregnancies are often terminated, but who benefits from that? Who benefits when a neural tube defect incompatible with life after birth is discovered and the pregnancy terminated? The death of the fetus is hastened at the cost of additional guilt and remorse. The infant mortality rate is massaged into looking better, but no real improvement in outcome has occurred.

Given the concern to respect autonomy, and given the existence and availability of prenatal screening and diagnostic tests, it is important that women who are offered prenatal testing arrive at their reproductive decisions unconstrained by external influence, and certainly not subject to systematic social pressures. The very existence of a prenatal-screening test carries the implicit recommendation of the local health authority, that to be tested is worthwhile and is the action of a responsible citizen. In this context, I would therefore suggest that Chadwick's decision to inform genetic counselling clients of the interest society has in their terminating an affected pregnancy, would probably be unhelpful. I fear that the effect in practice would be to make it even harder for the clients to arrive at their own decisions. This type of additional influence could easily be experienced as coercive.

In conclusion, then, what measure can we employ as an assessment of the work of a medical genetics unit? My suggestions of workload audit, and of the satisfaction of our client-group and of the referring agencies as expressed on a questionnaire or at interview, are likely to be the best measures available to us. Measures of our diagnostic work could be included, and of our provision of information and support to families, rather than a mere enumeration of reproductive outcomes that we may have influenced. Such a systematic audit of clinical genetics activities permits comparison between units: how much support was offered by different departments to the parents of infants dying of congenital malformations, or to the women/couples who had undergone termination of pregnancy for fetal abnormality or genetic disease? Such analyses could allow the profession to generate national standards of practice which reflected our concern for the individuals involved in these decisions rather than just the possible benefits accruing to society. Review of case records, and interviews with clients, can both contribute to an audit of quality in conjunction with the audit of workload data. To restrict the audit of genetic counselling to the analysis of reproductive decisions and our influence on them, would be tantamount to declaring the rest of our work (the larger part of our work, at that) as being unimportant: that is the message that some public health geneticists may wish to hear, but which we must oppose.

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