Odds in genetic counselling

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SUMMARY The text that follows is a statement of opinion on the use and meaning of recurrence rates in genetic counselling. It will have served its purpose well if it becomes the starting point of fruitful discussion among counsellors, and if its major points are formally tested.

Historically, the first task of medical geneticists has been to calculate and establish 'recurrence risks'. In their classic volume *Genetic Counselling*, Stevenson and Davison¹ listed and explained how to derive these risks and kept three pages at the end of the book as an afterthought on the more human and psychological aspects of their work. Nine years later, newer books with the same title^{2,3} deal almost exclusively with the psychological dimensions involved in the delivery and interpretation of this information.

But whereas diagnostic procedures and the estimation of recurrence are well defined activities and a part of every counsellor's formal training, the psychodynamics of the counselling process is still under study, and consensus has not yet been reached on exactly what counselling techniques should be used to deliver the information and help the counsellees to reach decisions that suit their own lives.

However, there exists overwhelming evidence from both individual practice and published reports, that the interpretation of genetic odds is essentially subjective and depends in the first place on the highly personalised view each person has of a disease.^{4,5} Individual feelings about a particular probability of a specific disease vary widely, even among well-informed mathematicians or physicians. The matter becomes even more personal when parents must weigh their feelings about a recurrence rate against their desire to have further children, that is, against another subjective consideration.

In this context, it appears illogical for a counsellor to adopt a directive stance in the decision-making process since his own beliefs, or his understanding of a situation, are bound to differ from the only factors that should determine the appropriate decision in each case, namely, the counsellee's own feelings. Accordingly, most of the recent publications on the subject clearly state that counselling should be supportive and non-directive. To achieve this,

several interview techniques are available,⁶ but two basic principles permeate them all. Firstly, the counsellor must speak in terms with which the counsellee is familiar and, secondly, he must do his best to remain an uninvolved catalyst and refrain from expressing his own beliefs and feelings. And this, it appears, most counsellors fail to do when they state the recurrence risks.

First of all, if one really intends to stay neutral and induce the counsellees to express their true feelings freely, one should not deter them by talking of recurrence 'risks' but of recurrence 'rates' instead.⁵ The word 'risk' always brings to mind the idea of danger; it implies that some judgement has been made, and should not be used by the counsellor before it is clear that the counsellee has a good understanding of the problem and definitely sees its recurrence as an unpleasant possibility or a real threat.

Secondly, it is a common observation that most physicians suffer from 'probability dysgnosia',⁷ and it appears that more than half the geneticists in two European samples could not properly calculate a probability of recurrence when it differed from 1/4 or 1/2.⁸ This in itself is evidence that exact rates are not that important, since a substantial proportion of counsellees reach reasonable decisions after having been given the wrong numbers. But most geneticists have been influenced by training or habit into thinking that exact recurrence rates are essential and should be discussed at length, and many seem to impose this opinion on counsellees who do not share it. On the other hand, there are others who think that it hardly makes any difference to the counsellees whether the rates are 20, 35, or 62%. It is the feeling of being at risk that counts rather than the exact mathematical figure.^{9,10}

This is the rule in all spheres of life and there is no reason why human nature should be different when confronted with a genetic problem. No one uses percentages or probabilities in uncertainties that

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depend on others (Will he marry me? Will I get a rise? Will it rain tomorrow?) or even in situations that depend on oneself and should therefore be more exactly predictable (Shall I marry him? Shall I ask for a rise? Shall I go hiking tomorrow?). It would be unreasonable to expect an exact probability to be of crucial importance in a frightening situation where both types of questions are involved (Shall I bear another child? Will it be normal?).

One solution might be simply to inform the counselees that one is not quite sure what the outcome of a pregnancy will be, as meteorologists sometimes do in their weather forecasts. But in keeping with the preferred neutral stand, it appears essential to quote the rates and let the counselees decide for themselves whether they can feel sure about the outcome, one way or the other.

After all, their opinion is as good as any, and it can be noted that geneticists may also be illogical when it comes to such decisions. The delivery of exact rates also has the additional advantage of making the counsellor sound competent.

There is a difficulty here that constitutes a constant source of worry in some genetic centres. The probability value is meant to convey the idea of doubt or certainty as the counselees choose; it is not meant to convey on its own the concept of risk or danger. It is extremely important that the counsellor should not confuse uncertainty and doubt with risk and danger: it must be clearly understood that 'doubt' and 'uncertainty', whether in the presence or absence of rates, only mean that we cannot be sure which single event will be chosen out of two or more. The 'risk' and 'danger' only exist when at least one of these possible events is perceived as being bad, and *whether a condition is bad or not should be discussed when one talks about the condition itself, not its recurrence*. In my experience, when the mathematics are thus deliberately separated from the consequences of the outcome, a careful presentation of exact figures never engenders any problem, especially when the condition and its effects on the family have already been discussed in depth.

One must also appreciate that the decision to be taken by the parents will be based on feelings rather than abstract comprehension. Even Einstein never reconciled himself with the principle of uncertainty in quantum physics because it did not fit with his philosophical approach to nature. Uncertainty is a part of every day life (weather, sports results, death, sex of future children); it is sometimes feared but also widely enjoyed (card games, lotteries, horse races, thrillers) and a large number of people live on their ability to deal with it (stock exchange, business deals, new crops). And although it may appear unreasonable to a visitor from outer space, observation shows

that gut feelings and experience are given much more importance in these activities than exact mathematics.

It follows from this that the best way to introduce a recurrence rate is to try to minimise the importance of arithmetic in the conversation and to use the simplest mathematical terms, devoid of complexity and biased meanings. For this purpose, as is the case for horse races and stock exchange, odds are far preferable to probabilities and percentages.

That odds are more readily intelligible than other probabilistic statements may not be appreciated by everyone, but any instructor in statistics who is fond of likelihoods will have noticed how the very physicians and biologists who cannot make out elementary probability theory easily understand a problem when it is explained to them in terms of odds. The same is true at the genetic clinic where those counselees who are intelligent but illiterate and those of borderline intelligence can only be made to understand in those terms. Geneticists are no exception to the rule: they all know the 9:3:3:1 compound ratio in genetics, but how many of them know the equivalent percentages: 56.25, 18.75, 18.75, and 6.25%?

Odds are easily understood by anyone, they imply the clear formulation of rival hypotheses, and they keep the dichotomy present in mind while conferring the idea of uncertainty.

Obviously, odds cannot be given 'against' or 'in favour of' something without being utterly biased, but they can easily be neutralised if both sides of the dichotomy are given together. For example, one can say: "Each time you produce a child, it is just as if you were choosing by chance one of four children, of whom three are like Peter and one is like Judy". Or: "If 40 families exactly like yours, and with exactly the same history, come to this office and then go home, and each of them produces one more child, 39 will have children with ordinary palates, and one will have a child with a cleft, and no-one can tell which one of these 40 families yours will be". (Note that the words 'normal' and 'affected' are deliberately avoided at this point in the discussion.)

Such statements are neutral, easy to grasp, and best made after a thorough discussion of the abnormal condition. They can be repeated in several variations and compared to the odds for the general population until the idea is clearly understood. Their exact formulation can be adapted to fit the parents' vocabulary and feelings, even with a voluntary loss of neutrality when appropriate, but if the question arises before the counsellor has had time to assess these feelings in depth, he or she can easily stick to a neutral stand. Odds so worded are most easily understood. They are more likely to be remembered

than their analogous percentages and probabilities, especially when the parents are in a state of stress or shock, and even more so in the case of people with borderline intelligence. They satisfy counsellees at all educational levels and are only rarely translated into probabilities or percentages by scientifically oriented counsellees.

The provision of odds in this manner denotes a wholly different understanding and attitude from that of the counsellor who says with the best of intentions that "you've got a 25% risk". It helps the parents leave the office with a clear understanding of *their* family situation rather than the counsellor's percentages, and it clears the atmosphere for friendly follow-up sessions, because at no time was the counsellor perceived to be an antagonist.

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