Psychological Aspects of Genetic Counselling

Edited by

ALAN E. H. EMERY IAN M. PULLEN

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Preface

How it comes – let doctors tell

Duncan Gray: Robert Burns

With improvements in living standards and advances in medicine and surgery, infectious diseases and nutritional deficiencies are becoming less common causes of morbidity and mortality. As a result these environmental diseases are gradually being replaced by others which are largely, or even entirely, genetic in causation. The genetic counsellor's role has largely been seen as educational, with the counsellor determining the risks of a particular disorder occurring in a family, and then discussing with the individual couple these risks as well as the various options available to them, should they decide the risks are unacceptably high. However, though this is important, there has been increasing interest recently in the psychological aspects of genetic counselling. Here the emphasis is more on fully appreciating the psychological impact of genetic disease on the individual couple, recognizing the various stages of the coping process, and tailoring counselling accordingly. Only by taking into account the couple's personal background can the various options available be discussed in the most sensitive manner. Thus we are moving away from entirely factually oriented counselling to what Kessler (1979) has called *person-oriented* counselling. In this broader scenario, the demands on the counsellor are much greater but it is only in this way that counselling is likely to be really meaningful and couples helped to make decisions which are the right ones for themselves. There is no place for directive counselling.

The particular qualities which make for a really sensitive and effective counsellor may be largely a reflection of personality and at least partly inherent. However, having said this many of the basic skills of genetic counselling can be acquired by formal training through precept and example (Emery, 1982). The purpose of this book is to provide a guide to some of the more important psychological problems in genetic counselling. It is intended for all those involved in this field including geneticists, genetic associates, paediatricians, obstetricians, hospital physicians, psychiatrists, psychologists, family doctors, social workers and nurses. We hope it might also be useful to medical students and others who wish to know more about current practices in genetic counselling.

After a brief review of the general principles of genetic counselling, there follow discussions of some of the important techniques of counselling particularly in regard to the sequence of responses encountered in emotional crises. Since the occurrence of genetic disease in a family may have serious effects on marital relationships a chapter is devoted specifically to marital pathology. The psychological problems particular to early infant loss, mental and physical handicap, late onset genetic disorders, infertility, sterilization, artificial insemination and prenatal diagnosis are all considered in detail. A final chapter describes how communication skills may be acquired and improved.

Like many other specialities, psychology has its own technical terms which are often somewhat bewildering to the uninitiated. We have attempted to avoid these as far as possible for the benefit of those in other fields. We have also tried to present an essentially practical and common sense approach to the psychological problems of genetic counselling. We do not necessarily agree with everything that each contributor has written, neither do we expect the reader to. However we hope that the book will stimulate thought and discussion about what is a rapidly changing and important subject.

December 1983 *A. E. H. Emery*
I. Pullen

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Contents

1 *Introduction – the Principles of Genetic Counselling*

ALAN E. H. EMERY

This chapter presents a general introduction to genetic counselling and emphasizes some of the psychological problems involved which are discussed more fully in subsequent chapters. Details of genetic principles as they relate to genetic counselling and risks of recurrence can be found in several excellent texts which deal specifically with these matters (Murphy and Chase, 1975; Stevenson and Davison, 1976; Fuhrmann and Vogel, 1976; Harper, 1981).

Nature of genetic disease

It is useful to consider all disease as being on a spectrum. At one end are those diseases which are very largely environmental in causation and include nutritional deficiencies and infectious diseases. At the other end of the spectrum are the genetic diseases in which environmental factors play no part in aetiology though they may modify the expression of a disorder. These genetic diseases include single gene or unifactorial disorders, which may be inherited as autosomal dominant, autosomal recessive, X-linked dominant or X-linked recessive traits, and cytogenetic disorders in which there is an inherent abnormality of chromosome number or structure. So far some 3000 unifactorial disorders and over 50 cytogenetic disorders have been recognized for which clinically useful catalogues exist (McKusick, 1983; De Grouchy and Turleau, 1977).

Between these extremes of the spectrum lie a group of disorders in which both environmental and genetic factors are believed to be involved in aetiology. These so-called multifactorial disorders include many of the commoner congenital malformations, certain diseases of modern society (coronary artery disease, essential hypertension, diabetes mellitus) and vari-

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ous psychiatric disorders such as schizophrenia. However, recent research has lead increasingly to the recognition of heterogeneity in many of these disorders and that certain forms may well have a unifactorial basis, for example non-insulin dependent diabetes mellitus of adult onset. The role of genetic factors in multifactorial disorders varies considerably and is much higher in, for example, schizophrenia than in neural tube defects (anencephaly and spina bifida). The greater the role of genetic factors in aetiology, the higher the risks of recurrence in relatives (Skinner, 1983).

The risks of recurrence in relatives are usually high (greater than 1 in 10) in unifactorial disorders, but low (less than 1 in 10) in multifactorial disorders. The risks in unifactorial disorders are determined in a straightforward manner from basic genetic principles. However, in multifactorial disorders such risks are derived empirically (and therefore referred to as *empiric risks*) by actually determining the frequency of occurrence of a disorder among relatives of affected individuals. In cytogenetic disorders the risks depend on the nature of the disorder. In Down's syndrome (mongolism), which is the commonest cytogenetic disorder, most cases are due to an extra chromosome 21 (trisomy-21) and the risks depend on mother's age at conception. A small proportion of Down's syndrome are due to a translocation, where material from chromosome 21 is transferred to another chromosome, and the risks depend on the type of translocation involved and whether it is carried by the mother or the father. Risk figures for unifactorial and some multifactorial disorders as well as for Down's syndrome are given in the Appendix.

Genetic counselling

Genetic counselling is a process of communication between the genetic counsellor and those who seek genetic counselling. The information to be communicated falls roughly into two main areas. Firstly, information about the nature of the disorder: its severity and prognosis and whether or not there is any effective therapy, what the genetic mechanism is that caused the disease and what are the risks of its occurring in relatives. Secondly, information on the available options open to a couple who are found to be at risk of transmitting a genetic disorder. This latter may include discussions of methods of contraception, adoption, prenatal diagnosis and abortion and artificial insemination by donor (AID).

Though essentially an educational process, to be really successful it is important that the counsellor recognize the problems of attempting to communicate information of a personal and delicate nature in a situation where the parents may be grieving over the loss of a child, and which may also be emotionally charged with feelings of guilt and recrimination fre-

quently coupled with a loss of self esteem. The effective communicator in this field is the one who not only recognizes these problems, but can empathize and help a couple transcend them.

Changing patterns in genetic counselling

Over the last twenty years there has been a gradual change in both the mode of referral of individuals for counselling as well as in those seeking genetic counselling (Emery, 1977). Whereas at one time most individuals were referred by hospital physicians, nowadays more are being referred by family doctors, the impetus often coming from couples themselves. Whereas in the past most individuals who sought counselling were from the professional classes and were married, nowadays the social class distribution is the same as in the general population and more individuals now seek counselling before marriage. These changes probably reflect an increasing awareness of the importance of genetic counselling by the medical profession as well as the general population at large. This increasing awareness is no doubt the result of articles in the press, coverage by the news media and more emphasis on genetics in school. These are changes to be encouraged though much still remains to be done in educating both the profession and public about such matters (Childs, 1974).

Coupled with this increasing awareness of genetic disease has also been an increase in what is expected from genetic counselling. All too often in the past some members of the medical profession have tended to view genetic counselling either with nihilism (there is nothing that can be done so why bother with genetic counselling) or with a frankly eugenic philosophy (couples at risk should be *advised* not to have children). However as genetic counselling becomes more informative and helpful and as counsellors are seen to be sensitive to the much wider issues involved in this field, its value is more likely to be recognized and those seeking advice more likely to have their expectations satisfied.

Changing emphasis in genetic counselling

Two or three decades ago genetic counselling was often the province of the scientist, usually as an extension of interests in plant and animal genetics. Understandably this resulted in the emphasis being on genetic mechanisms and risks. Then, largely through the interests of individuals like McKusick, Motulsky, and Clarke Fraser in North America and Carter, Klein, Francois, Böök and Becker in Europe, there was a gradual shift in emphasis to the more

medical aspects of genetic disease. This resulted in the recognition of the widespread occurrence of genetic heterogeneity: that clinically similar disorders could be inherited differently and have different prognoses. Subsequent developments led to the redefining of recurrence risks in many genetic disorders.

Recently the emphasis has begun to change again from what Kessler (1979) has referred to as *content-oriented* counselling to *person-oriented* counselling with more emphasis on the psychological aspects of genetic counselling. This change of emphasis has resulted from the recognition of a number of important factors. Firstly, that information about genetic disease is rarely emotionally neutral and often has profound psychological effects. Secondly, these effects may have long-term consequences and extend throughout the family to other relatives. Thirdly, and perhaps most importantly, has been the realization that couples given genetic counselling may opt for a course of action which may be at variance with what the counsellor might have regarded as reasonable. There is often a gap between the counsellor's expectations and the actual consequences of counselling. For example in a two-year follow-up study of 200 consecutive couples seen in a genetic counselling clinic, over a third of those who were told they were at high risk of having a child with a serious genetic disease were undeterred and actually planned further pregnancies (Emery *et al.*, 1979). In the past such behaviour has often been regarded as 'irresponsible', a failure on the part of the counsellor and an indictment of counselling in general. But when the couples in this study were carefully questioned their reasons for planning further children were often very understandable.

For example, further pregnancies were planned in some cases because, after seeing the effects of a disorder in a previous child or in one of the parents, it was not considered sufficiently serious (congenital cataract, congenital deafness, peroneal muscular atrophy), or prenatal diagnosis was available (Sandhoff's disease, X-linked mental retardation) and yet in other cases the parents planned further pregnancies because if a subsequent child were affected it would not survive (renal agenesis) or if it survived it would succumb within a year or so (Werdnig–Hoffmann disease). There was also a small but lamentable group of couples who had no living children and dearly wanted a family at whatever cost.

Thus a course of action which might seem irresponsible to one person may be eminently reasonable to another. In a free society this choice is the individual's prerogative provided it is made in the full knowledge of all the facts and appreciation of the possible consequences. Since the counsellor's role is to help couples reach decisions which are the best for themselves genetic counselling should always be non-directive (Anon, 1982).

The genetic counsellor's role

With this change of emphasis the genetic counsellor is now seen to have two major roles: not only to communicate factual information but also, armed with an appreciation of the psychological aspects of the problem, to help couples in their decision making. It is no longer sufficient to be conversant merely with the genetic and medical aspects of a problem. It is also important to be fully aware and appreciative of the psychological effects on the individual. Only in this way can the counsellor empathize and thereby be a more effective communicator. These latter skills are much more difficult to acquire. Formal training (Emery, 1982) through precept and example is important but the qualities which make for a really sensitive and effective counsellor may be largely a reflection of personality and at least partly inherent.

Nevertheless the genetic and medical aspects should not be underestimated. It is a *sine qua non* that the first step in genetic counselling is the establishment of a precise diagnosis especially because of the possibility of genetic heterogeneity. The counsellor also needs to be as conversant, as is reasonably possible, with all the important medical aspects of a genetic problem if counselling is to be at all meaningful.

Problems in counselling

Because of the intense emotional stress often engendered by genetic disease, genetic counselling should only be given in a relaxed and quiet atmosphere where couples can be encouraged to ask questions and express their feelings. Only in this way is it possible to explore a couple's attitudes to a genetic disorder and their interpretation of its implications.

As a prelude to genetic counselling it is important to divine a couple's educational and social background, their religious attitudes and, if possible, something of their marital relationships if information is to be presented most effectively and sensitively.

Having initially established a precise diagnosis, the genetics of the condition and family details, how much further the counsellor should proceed depends on how far a couple will welcome and benefit from more information at the time. Immediately after the loss of an affected infant or immediately after the diagnosis of a serious genetic disorder has been made is not the time for detailed and extensive discussions because the parents are unlikely to be either receptive or comprehending. To know precisely when and how much information to impart during the grieving period requires considerable skill

and understanding. The various stages of the so-called 'coping process' have to be recognized and subsequent counselling tailored accordingly (Falek, 1977).

Having decided the time is right to proceed further with counselling, the relevant medical and genetic details of the disorder and the risks of recurrence can be presented. With regard to the medical details, information should be available on such matters as age at onset, progression, complications, the ultimate prognosis and possibilities for effective treatment. Much has been written on the concept of 'burden' in genetic disease by which is meant the psychological, social and, to a lesser degree, financial problems associated with such diseases. A disorder in which the burden is great but short lived (for example a child born with a lethal congenital malformation) may be far more acceptable, even if the risk of recurrence is high, than a disorder where the burden is moderate but protracted (such as a child with slowly progressive and disabling muscular dystrophy).

Unlike the clinician concerned with management and who must therefore maintain a positive and optimistic approach throughout, the genetic counsellor has to present an accurate picture even if depressing and disturbing if the parents are to make a reasoned decision about future children. In this regard the counsellor is therefore often seen as the 'harbinger of woe'. It is questionable whether it is right to encourage parents in the hope that any future affected children might be treatable when at present there is little likelihood of this. Such discussions of treatment in any future affected children require considerable sensitivity when the parents already have an affected child. In fact it is not uncommon for the genetic counsellor to find himself in this dilemma and therefore to have to temper his remarks very carefully.

In most cases it is doubtful if genetic mechanisms and recurrence risks need be discussed in much detail. The actual interpretation of risk is in any event very subjective (Pearn, 1973). Certainly a pedantic obsession with risk figures by the counsellor may completely obscure the real issues and detract from effective communication on more important matters. Nevertheless risks form a useful basis for further discussion and in general terms can be a significant factor in influencing parents' decision making (Carter *et al.*, 1971).

Turning to the options available to a couple who decide they could not accept an affected child, prenatal diagnosis has been a major development and has removed much of the uncertainty in genetic counselling (Table 1.1.). Here developments are taking place so rapidly that it would not be unreasonable to hold out this option in some cases where at present this is not yet possible (Galjaard, 1980). Again however care is necessary for it would seem unreasonable to have a couple delay planning a pregnancy in the hope of a reliable prenatal diagnostic test becoming available in the near future when at present there seems little likelihood of this. In the waiting period there is also

Table 1.1 Main indications for prenatal diagnosis

(1) Cytogenetic abnormalities
. mainly Down's syndrome with a previously affected child or maternal age >35–40
(2) Inborn errors of metabolism
. over 60 can now be diagnosed *in utero*
. most are rare recessive disorders
(3) Fetal sexing
. X-linked disorders which cannot yet be diagnosed *in utero* (e.g. Duchenne muscular dystrophy)
(4) Congenital abnormalities
. mainly neural tube defects with a previously affected child or raised maternal serum alphafetoprotein
. some others can be diagnosed by fetoscopy or ultrasonography

always the danger of an unplanned pregnancy, and in any event an exaggerated concern with this possibility could well lead to serious marital problems. Both these eventualities could be prevented by careful and detailed discussions of contraceptive methods at an early stage in counselling.

When parents decide that they could not accept having an affected child and prenatal diagnosis is either not possible or, for whatever reason not acceptable, then other options have to be considered. Contraception in this context requires expert advice because the results of failure will be far more devastating when there is the risk of a severely affected child than when it is practised for social or economic reasons. Further, the very deep fear of having an affected child may well generate serious psycho-sexual problems which can often be prevented only by resort to definitive contraceptive methods such as sterilization. However abstinence may be the only acceptable alternative to some couples and these cases require sympathetic understanding and perhaps a discussion of other forms of sexual relief. The latter may be particularly important in any event in those genetic disorders which lead to physical disability in one of the parents (e.g. adult forms of muscular dystrophy). However not all counsellors will feel competent or confident to discuss such very personal matters in which case they should refer a couple to someone who is.

At one time adoption was often an option which could be encouraged by the genetic counsellor but in the last few years this has become increasingly difficult. This is partly because some adoption agencies have become reluctant to place children in families where a parent may have a disabling disorder, but mainly because fewer children are now available for adoption. Before even

raising this option it is therefore advisable for the counsellor to determine beforehand whether, in a particular case, this is likely to be a realistic possibility. Otherwise there can be considerable frustration and disappointment.

Finally AID might be offered where it is known that both parents carry the same *rare* recessive gene or the father has an autosomal dominant disorder or carries a chromosome translocation which in the unbalanced state is likely to result in an affected child, e.g. Down's syndrome. However, the success rate, even in expert hands, is not always high and several inseminations may be necessary before success is achieved (Richardson, 1975). Again considerable frustration can result unless a couple are warned about these problems beforehand. Understandably many couples also find this an unacceptable option for personal and aesthetic reasons and if the counsellor senses this the subject should of course not be pursued.

Conclusions

There are essentially three aspects to genetic counselling. The scientific aspect is concerned with genetic mechanisms and risks of recurrence. It is perhaps the least important. The medical aspect is concerned with diagnosis and the resolution of genetic heterogeneity and is an essential first step in counselling but it should be viewed only as a preliminary to a consideration of the wider issues. The psychological aspect of genetic counselling is concerned with understanding and appreciating the psychological effects of genetic disease so that more effective communication is possible. Couples are thereby helped to make decisions which are the best ones for themselves in their particular circumstances and which may not necessarily be those which the counsellor would have made. However, having witnessed so much suffering and unhappiness caused by genetic disease, it is understandable that many genetic counsellors, despite themselves, may harbour the deep-seated wish that when faced with the dilemma some couples would be wise to exercise caution . . .

> Past sorrows, let us moderately lament them;
> For those to come, seek wisely to prevent them.
> John Webster's *The Duchess of Malfi*, 1616

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