MEDICAL PRACTICE

Clinical Topics

Prospective study of genetic counselling

ALAN E H EMERY, JOHN A RAEBURN, ROSALIND SKINNER, SUSAN HOLLOWAY, PEGGY LEWIS

British Medical Journal, 1979, **1**, 1253-1256

Summary and conclusions

A prospective study was carried out on 200 consecutive subjects seen for counselling (consultands) for serious genetic disorders. Educational and social background of consultands and their knowledge and understanding of their particular problem were assessed before counselling, and their response was determined immediately afterwards and three months and two years later by an independent observer not concerned in the genetic counselling. The husband's educational background was particularly important in influencing a couple's comprehension of counselling. X-linked recessive and chromosomal disorders presented the most difficulties in comprehension. The counsellors' assessment of comprehension was a good guide to the consultands' comprehension as assessed at subsequent follow-up. The proportion deterred from having children increased with time and over a third had been sterilised within two years of counselling.

It is suggested that follow-up after counselling should be routine, especially when the counsellor suspects that comprehension has not been good, in X-linked recessive and chromosomal disorders, and when the risks of having an affected child are considered to be high.

University Department of Human Genetics, Western General Hospital, Edinburgh EH4 2XU
ALAN E H EMERY, DSC, FRCPED, professor
JOHN A RAEBURN, PHD, FRCPED, senior lecturer
ROSALIND SKINNER, MD, BS, lecturer
SUSAN HOLLOWAY, PHD, research fellow
PEGGY LEWIS, SRN, field worker

Introduction

Genetic counselling is an established part of clinical practice. Over the past few years several studies have been designed to assess the value and effectiveness of counselling, and these have been the subject of extensive reviews, in which reference to previous work will be found.[1-4] Unfortunately, many of these studies were retrospective. Among the inherent weaknesses of such studies, perhaps the most important is that counsellors may not recollect exactly what information was given to a particular individual several years previously. Furthermore, these provide no information on changes in attitudes and knowledge before and after counselling, and the effects of counselling have usually been assessed by those who actually gave the counselling, which might well bias the results.

We aimed at overcoming some of these difficulties in undertaking a prospective investigation. We assessed the educational and social background of individuals seen for counselling and their knowledge and understanding of their particular problem before counselling, and their response immediately after counselling and three months and two years later was determined by an independent observer.

Subjects and methods

The study population comprised subjects seen for genetic counselling (consultands) in the genetic clinic at the Western General Hospital, Edinburgh, between July 1973 and August 1976. Only those at risk of transmitting a serious genetic disorder were included. The nature of the study was carefully explained to each consultand and seven out of 207 consecutive individuals approached declined to take part. The results were therefore based on the remaining 200. Each consultand was interviewed before counselling, and various personal, social, and educational data were obtained. At the time of counselling the counsellors (AEHE, JAR, and RS) recorded the diagnosis, the advice given, and their impression of the consultands' understanding and likely course of action. The consultands' understanding and

response to counselling were independently assessed by PL immediately after the counselling session and three months and two years later. All recorded information was kept strictly confidential.

Results

SOCIAL AND EDUCATIONAL BACKGROUND

Age and marital state—Most (176) were married, and 148 were aged under 30 when counselled. Those who attended the clinic in later life were often concerned about the risk of Down's syndrome.

Social class—the distribution of consultands according to socioeconomic class (table I) was significantly ($P < 0.01$) different from the general population, because of an excess of consultands from professional and managerial occupations. The distribution of socioeconomic classes among this group of consultands seen for serious genetic disorders also differed (though not significantly) from that of consultands seen in the clinic for general genetic disorders, which was similar to that of the general population.

TABLE I—*Distribution (% in each class) of socioeconomic classes in consultands and general population of Edinburgh region**

	Social class: I	I	III	IV	V
Consultands	12	21	54	10	3
General population	6	16	52	17	9

*Figures for general population derived from 1971 Census (Scotland).[9]

Educational background—The educational background of the consultands was not significantly different from that of the general population, but 52% had at least O level passes and 8% were university graduates (or equivalent), compared with 41% and 3% respectively in the general adult population (table II).

TABLE II—*Distribution (% in each class) of educational attainment among consultands and general adult population**

	No O levels	O levels only	H levels	Higher education
Consultands	48	20	24	8
General population	59	16	22	3

*Figures for O and H levels derived from Scottish Educational Statistics (1974)[10]; those for higher education estimated from 1971 Census (Scotland).[11]

Religion—The proportion of Roman Catholics among consultands did not differ significantly from that of the general population (14% and 16%[5] respectively).

REFERRAL: REASONS AND MODE

The main reason for referral in most cases (198) was for genetic counselling. The two remaining consultands were referred because they had requested more information about prognosis. Altogether 109 (55%) were referred by family doctors, 84 (42%) by hospital consultants, and 7 (4%) by relatives or paramedical workers. Nevertheless, 79 (40%) had initiated the request for advice themselves, usually because they had read an article on medical genetics or seen a television documentary programme. Interestingly, this proportion increased during the study from 20% of those first seen in 1973 to 60% of those first seen in 1976. The proportion of self-referrals seemed to be higher among this group of consultands seen for serious genetic disorders than in those seen for general genetic disorders.[1]

KNOWLEDGE OF DISORDER

Most of the consultands (194) had an intimate knowledge of the particular genetic disorder for which they requested counselling, either because they were affected themselves or had an affected spouse or first-degree relative. Concerning particular diseases, 172 (86%) knew the diagnosis, 160 (80%) knew the prognosis, and 18 (9%) knew that the disorder was genetic. Nevertheless, on close questioning only 14 (7%) knew anything about the genetic mechanism concerned and none was clear about the risks to their children.

Table III shows the mode of inheritance of genetic disorders in consultands and their families. The most common disorders were:

TABLE III—*Mode of inheritance of genetic disorders in consultands and their families*

Mode of inheritance	Number (%) of subjects
Unifactorial:	
Autosomal dominant	30 (15·0)
Autosomal recessive	41 (20·5)
X-linked recessive	11 (5·5)
Chromosomal	26 (13·0)
Multifactorial	86 (43·0)
Not known	6 (3·0)

among the autosomal dominants, Huntington's chorea (6); among the autosomal recessives, fibrocystic disease (6) and profound childhood deafness (6); and among the X-linked recessives, Duchenne muscular dystrophy (6). The most common chromosomal disorder was Down's syndrome (20), and the most common multifactorial disorder was spina bifida/anencephaly (49). The six cases of unknown aetiology comprised leukaemia in three and one case each of de Lange's syndrome, extroversion of the bladder, and multiple congenital abnormalities not associated with any recognised syndrome. In 67 of the consultands (34%) the risk of transmitting a disorder to their offspring was considered to be high (>10%), 133 (66%) being given a low risk (<10%).

PHYSICIANS' CONCLUSIONS AFTER COUNSELLING

At counselling it was considered that only 13 of those given a low risk (10%) but 29 of those given a high risk (43%) would be deterred from having children, and that nearly one-third of the high-risk group who were undeterred would make use of antenatal diagnosis. The consultands' comprehension of the advice given was estimated to be good or excellent in 163 (82%) and fair or poor in the remaining 37 (19%). The counsellors' assessment of comprehension at counselling agreed well with the results at two-year follow-up. The proportion of those who remembered the risks correctly was 66% among those thought to have had an excellent comprehension at the time of counselling, 54% among those who had a good comprehension, but only 32% among those assessed as having a fair or poor understanding at counselling.

RESPONSE TO COUNSELLING

All 200 consultands were interviewed immediately after counselling, 191 were seen three months later, and 170 two years later. Only two people declined to be seen again. The others who were not followed up had either moved and left no forwarding address or did not reply to letters requesting a further interview.

Recollection of recurrence risk

To estimate the consultands' comprehension of the advice that they had been given, the proportions who remembered their risk of transmitting the disease were calculated. Recollection of risks was chosen because it can be easily measured and studied, though we realise that this is only a rough guide to full comprehension of counselling. Immediately after the counselling session 149 (77%) remembered the exact risk correctly, but at the three-month and two-year follow-up the numbers had fallen to 113 (61%) and 87 (53%) respectively. Factors that we thought might influence the consultand's recollection of the risk were socioeconomic class, age, general education, knowledge of biology (as assessed from formal qualifications), and prior knowledge of the disease. In the case of married couples the wife was always considered to be the consultand (176 out of the total 200), and of all these various factors only the husband's educational level was found to have a significant effect (χ^2 test). Two years after counselling the percentage of consultands who remembered the risk correctly was greater the higher the educational level of their husbands ($P < 0.05$). Only 45% of those whose husbands had no O levels remembered the

risk correctly, whereas 74% of those whose husband had a university degree remembered the risk correctly.

We also thought that comprehension of counselling might depend on the mode of inheritance of the disease. Table IV shows the numbers who remembered the risk according to the mode of inheritance of the disease. Chromosomal and X-linked recessive (XR) disorders presented the most difficulties in comprehension and the recollection of risks. The XR group comprised only 11 subjects, but it seemed that although most could remember whether the risk was high or low they were sometimes confused about the exact recurrence risk.

TABLE IV—*Number of consultands seen at various times after counselling and proportions who remembered the recurrence risk correctly, according to mode of inheritance of disease. Table excludes six cases in which mode of inheritance was unknown*

Mode of inheritance:	Unifactorial			Chromosomal	Multifactorial
	AD	AR	XR		
*Immediately after counselling**					
Total seen	30	41	11	26	86
No (%) who remembered risk correctly	27 (90)	32 (78)	6 (55)	12 (46)	72 (84)
Three months after counselling					
Total seen	28	38	10	26	83
No (%) who remembered risk correctly	20 (71)	25 (66)	3 (30)	11 (42)	54 (65)
*Two years after counselling***					
Total seen	27	33	8	21	75
No (%) who remembered risk correctly	15 (56)	22 (67)	2 (25)	5 (24)	43 (58)

AD = Autosomal dominant. AR = Autosomal recessive. XR = X-linked recessive.
*$P < 0.001$.
**$P < 0.05$.

Estimate of importance of risk

We regarded a risk of 10% or greater as high and a risk of less than 10% as low. Nevertheless, 25 of those given a low risk by the physician (19%) considered the risk as high, and 12 of those given a high risk (18%) considered the risk as low when interviewed immediately after counselling. A more detailed investigation of those consultands who regarded a low risk as high showed that most of them either had a risk of between 5% and 9% or had a family history of a disease with a poor prognosis. The results of the investigation were less clear in those who regarded a high risk as low. We found no evidence that these subjects had risks nearer to 10% than others with a high risk, and no indication that the diseases in their families were necessarily those with a good prognosis, for they included Huntington's chorea, cystic fibrosis, and tuberous sclerosis.

Decision about further children

At each of the three interviews after counselling consultands were asked whether they were deterred from having children. In a few cases the question was not asked because the consultand was widowed or unmarried with no immediate prospect of marriage. In the "low-risk" group the percentage who were undeterred was constant over the two-year period and the percentage who were deterred increased from 11% to 18% because the number who were uncertain at the beginning had decreased (table V). In the "high-risk" group the percentage who were deterred also increased (table V), not only because those who were initially uncertain decided against further children but also because those who were initially undeterred subsequently changed their minds. Sometimes they had had a child in the interim and did not wish to take the risk again, but usually they had considered the consequences of having an affected child more carefully.

TABLE V—*Decision about further children in consultands given a low (<10%) or high (>10%) risk by the counsellor, according to time of subsequent interview. Figures are numbers (%) of consultands*

Decision	Immediately after counselling	3 months after counselling	2 years after counselling
Consultands given a low risk			
Deterred	14 (11)	19 (15)	19 (18)
Undeterred	107 (80)	103 (81)	88 (82)
Uncertain	12 (9)	5 (4)	0
Total	133 (100)	127 (100)	107 (100)
Consultands given a high risk			
Deterred	22 (33)	26 (41)	31 (53)
Undeterred	36 (54)	35 (56)	28 (47)
Uncertain	9 (13)	2 (3)	0
Total	67 (100)	63 (100)	59 (100)

REASONS FOR DECISIONS MADE AFTER COUNSELLING

Consultands deterred from having children

Three months after counselling 45 consultands were deterred from having children. Most (39; 87%) had been deterred because of the risk or burden (or both) of having an affected child, and the remaining 6 (13%) had completed their families. Two years after counselling 51 consultands were deterred, of whom 39 (78%) were deterred by the risk or burden (or both) of having an affected child and 11 (22%) because they had completed their families.

The consultands were asked which method of contraception they used (table VI). Oral contraception was by far the most common method. In over a third the consultand or spouse had been sterilised within two years of counselling.

TABLE VI—*Methods of contraception used by consultands deterred from having children. Figures are numbers (%) of consultands*

Method used	3 months after counselling	2 years after counselling
Oral contraceptive	17 (38)	19 (38)
Tubal ligation	7 (16)	10 (20)
Vasectomy	6 (13)	7 (14)
Sheath	4 (9)	7 (14)
Intrauterine device	7 (16)	4 (8)
Diaphragm	2 (4)	2 (4)
Abstinence	1 (2)	0
Unknown	1 (2)	1 (2)
Total	45 (100)	50 (100)

Consultands undeterred from having children

Altogether 138 consultands were undeterred from having further children three months after counselling, and 49 of these (36%) planned to make use of antenatal diagnosis. Thirty-five were pregnant at this time, but only seven had become pregnant since being counselled. Two years after counselling 116 consultands were undeterred, of whom 81 (70%) had already had children or an abortion (three therapeutic, eight spontaneous), 15 were pregnant (three in their second pregnancy), 21 had not managed to conceive, and two were still unmarried. Those who were undeterred despite a high risk of having an affected child were studied in detail. They gave the following reasons (diseases in the consultands' families are given in parentheses). In nine the prognosis was regarded as good (aniridia; congenital cataract; peroneal muscular atrophy; Wiedemann-Beckwith syndrome; telangiectasia; deafness (two cases); and non-specific epilepsy (two cases)). In eight antenatal diagnosis was available (anencephaly (three cases); spina bifida (two cases); autosomal translocation; Sandhoff's disease; and X-linked mental retardation). Five thought that an affected child was unlikely to survive long (congenital muscular dystrophy; endocardial fibroelastosis; renal agenesis (two cases); and Werdnig-Hoffmann's disease). Consultands giving these reasons formed the largest group. Four others did not regard the risk as high (Huntington's chorea; Marfan's syndrome; myotonic dystrophy; and tuberous sclerosis); three desperately wanted children (cystic fibrosis; renal hypoplasia; and multiple congenital abnormalities); two refused to believe they were affected (myoclonic epilepsy and facioscapulohumeral muscular dystrophy); and one (sickle-cell anaemia) would choose a marriage partner who was not a carrier.

OUTCOME OF PREGNANCIES

During the two years after counselling 85 individuals had had a child or an abortion (or both), seven having had two pregnancies and 39 (46%) having had antenatal diagnosis. Twenty-five of these consultands (30%) had a high risk of having an affected child. Out of a total of 81 live births, one infant had a balanced autosomal transloca-

tion, and seven had a high risk of developing a genetic disorder in the future (tuberous sclerosis, myotonic dystrophy, facioscapulohumeral muscular dystrophy, deafness, porphyria, and peroneal muscular atrophy).

Discussion

Most of those who sought counselling were young married couples referred specifically for this. Results of an extensive study of genetic counselling in Britain[6] showed an excess of couples from the upper socioeconomic classes, which was also the case, though to a lesser degree, in our study, while their educational background (based on formal school and college qualifications) was also somewhat better than average. Nevertheless, we see no reason to believe that those seen for counselling in Edinburgh are different from those seen in other centres in the United Kingdom. Over the past few years the proportion of couples from the lower socioeconomic classes has been gradually increasing, as has the proportion of self-referrals who initiated the request for advice themselves.[1] The proportion of self-referrals in our study increased from 20% in 1973 to 60% in 1976, often because the subjects had read an article on medical genetics or had seen a television documentary programme. This emphasises the importance of health education in its broadest sense.[7] We found that general educational background, particularly that of the husband, was an important factor affecting understanding of genetic counselling. We suggest that both husband and wife should be counselled together whenever possible. Our experience also suggests that particular attention should be given to explaining the inheritance of X-linked and chromosomal disorders, since these appeared to be least understood and remembered. Familiarity with the disorder is another important factor likely to influence response to counselling. In the present study 97% of consultands had an intimate personal knowledge of the disorder, either because they were affected themselves or had an affected spouse or first-degree relative.

The counsellors' assessment of the consultands' comprehension at the time of counselling seemed to agree well with the consultands' actual comprehension as determined at follow-up. Nevertheless, the counsellors' view of the gravity of the risks may not always correspond to that of the consultands. Roughly a fifth of those given a "low risk" (<10%) considered it to be high, and a similar proportion of those given a "high risk" (>10%) considered it to be low. Actual risks given to those in the "low-risk" group were usually between 5 and 9% and sometimes the particular disorder had a poor prognosis, so the subject might have been influenced as much by the "burden" as the risks of recurrence. Those who considered a "high risk" to be low included families with Huntington's chorea, tuberous sclerosis, and cystic fibrosis. Perhaps their response was a reflection of a subconscious wish and an inability to face the realities.

Whatever the risk given, the proportion deterred from having children seems likely to increase with time. Discussions with the consultands at two-year follow-up suggested that this was often because they had considered the consequences of having an affected child more carefully. Most did not wish to have children because of the risks or burden (or both) of having an affected child, and not because they had already completed their families. This has been emphasised in previous studies.[8] Oral contraception was the most common method of family planning adopted by these couples but, interestingly, of those deterred from having children, over a third had been sterilised within two years of counselling. When consultands were undeterred from having children, despite a high risk of having an affected child, this was often because antenatal diagnosis was available, an affected child would not survive long, or the consultand considered that the disorder was not serious, which again seems likely to be due to a subconscious inability to face the realities.

Though additional genetic information was never given at any follow-up interview, since this would have vitiated the results, the interviewer was repeatedly impressed by the value consultands placed on the opportunity to discuss their problem again. We suggest that follow-up should perhaps be routine after counselling, especially when the counsellor suspects that comprehension has not been good, in X-linked recessive and chromosomal disorders, and when the risks of having an affected child are high. It also gives an opportunity to keep consultands up to date with developments in treatment and antenatal diagnosis.

We are particularly grateful to all those who kindly agreed to take part in this study, which was generously supported by a grant from the National Fund for Research into Crippling Diseases.

References

1 Emery, A E H, in *Equalities and Inequalities in Family Life*, ed R Chester and J Peel, p 71. London and New York, Academic Press, 1977.
2 Hsia, Y E, in *Genetic Counseling*, ed H A Lubs and F de la Cruz, p 53. New York, Raven Press, 1977.
3 Shaw, M W, in *Genetic Counseling*, ed H A Lubs and F de la Cruz, p 35. New York, Raven Press, 1977.
4 Childs, B, in *Genetic Issues in Public Health and Medicine*, ed B H Cohen, A M Lilienfeld, and P C Huang, p 329. Springfield, Illinois, C C Thomas, 1978.
5 *Annual Report of the Registrar General for Scotland, 1971*. Edinburgh, HMSO, 1972.
6 Carter, C O, *et al*, *Lancet*, 1971, **1**, 281.
7 Childs, B, *American Journal of Human Genetics*, 1974, **26**, 120.
8 McCrae, W M, *et al*, *Lancet*, 1973, **2**, 141.
9 General Register Office, Edinburgh, *Census 1971. Scotland. Economic Activity, County Tables. Pt 2 (10 percent Sample)*. Edinburgh, HMSO, 1978.
10 *Scottish Educational Statistics 1974*. Edinburgh, HMSO, 1974.
11 General Register Office, Edinburgh, *Census 1971. Scotland. Qualified Manpower Tables (100%). Usual Residence and Birth Place Tables*. Edinburgh, HMSO, 1976.

(*Accepted 16 February 1979*)

Is asthma a contraindication to immunisation against cholera?

Asthma is not a contraindication to immunisation against cholera. As it is a "killed" vaccine, even if the patient was taking steroids I do not think there would be any danger.

Have there been any reports of cataracts being caused by anovulatory pills?

Many different types of eye disorder have been described in oral contraceptive users, but cataract does not appear to be among them.[1] Connell and Kelman reported the results of a careful study of ocular abnormalities in 305 users and 611 non-users of oral contraceptives, including tests for visual acuity and slit-lamp and fundoscopic examinations.[2] No statistically significant differences in the occurrence of abnormalities were found between the two groups.

1 Walsh, F B, *et al*, *Archives of Ophthalmology*, 1965, **74**, 628.
2 Connell, E B, and Kelman, C D, *Fertility and Sterility*, 1969, **20**, 67.

I am told that negative ionisers are being used in the treatment of asthma. Is this so, and are they successful?

For at least 20 years there have been reports that symptoms of asthma may be relieved by exposure to negatively ionised air, but most studies have failed to carry out objective lung function tests or adequate statistical analyses. More recently, a controlled study of seven patients with bronchial asthma showed some improvement in airways obstruction in two patients, who reported subjective improvement, but it was thought necessary for further double-blind studies to be conducted before concluding that negatively ionised air should form part of the routine management of patients with asthma.