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Current issues in the practice of genetic counselling

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Ten problems of genetic counselling based on Hungarian experiences are discussed, *viz.* (1) the competency of counsellees; (2) the high rate of self-referrals; (3) the low rate of most competent couples; (4) 20.2% of probands without definite nosological diagnosis; (5) advantages and disadvantages of the information-guidance type of counselling; (6) the unpleasant role of random risk; (7) 12.9% pregnant consultands; (8) the high rate of counsellees with two or more affected children; (9) combinations of independent disorders in the probands; (10) the so-called mixed pedigrees

The Genetic Counselling Service has been part of the National Mother and Child Care System in Hungary since 1976, thus the genetic counselling clinics are an integral part of the Hungarian health care system. Hungary has 10.7 million inhabitants and there are 14 genetic counselling clinics, 5 in Budapest, and 9 in the provinces, all having an adequate cytogenetic and clinical background. Most of them are in hospitals. Each genetic counselling clinic supplies a defined territory. The number of new counsellees showed an increasing trend until 1976 (Table I). Since that year a nearly constant number of about 3700–3800 couples or persons is registered yearly. In 1976–78, this meant a rate of 2.1–2.2 per 100 livebirths in Hungary, and our genetic counselling clinic sees about 20% of all counsellees.

The purpose of this paper is to summarize the current issues of genetic counselling on the basis of Hungarian experience.

1. The counsellees were divided into 15 categories according to their reason for seeking advice (Table II) (3). This classification was based first of all on the disorder of probands. The percentage values of these categories seem to be interesting as they show that in Hungary the genetic counselling service is an important basis of qualitative family planning in general. The first and third largest categories are sterility and fetal death, disorders rarely mentioned in the textbooks of genetic counselling. In a minority of these cases, genetic examinations and advice may be useful, but for the majority the genetic clinic has little to offer. At present this point causes a number of problems. It is a well-known opinion that "ultimately genetic counselling should become available to everyone who needs it" (6). Still, the

TABLE I

Annual number of new counsellees (couples or persons) in 14 genetic counselling clinics in Hungary, 1973–1978

		1973	1974	1975	1976	1977	1978	Total
Other 13 genetic counselling clinics	No.	1 013	1 386	2 020	3 152	3 071	2 624	13 266
	per cent	94.9	81.3	75.1	80.2	78.6	70.0	77.8
Own genetic counselling (NIH)	No.	54	318	668	780	834	1 124	3 778
	per cent	5.1	18.7	24.9	19.8	21.4	30.0	20.8
Total	No.	1 067	1 704	2 688	3 932	3 905	3 748	17 044
	per cent	100.0	100.0	100.0	100.0	100.0	100.0	100.0
No. of livebirths in Hungary		156 224	186 288	194 240	185 405	177 574	168 096	1 067 827
$\frac{\text{No. of new counsellees}}{\text{No. of livebirths}} \cdot 100$		0.68	0.91	1.38	2.12	2.20	2.23	1.60

TABLE II

Distribution of nosological categories in the genetic counselling material, 1973–1978

Nosological categories	No.	per cent
Nosological diagnosis		
Genetic		
Mendelian conditions	582	15.4
Chromosomal aberrations	254	6.7
Multifactorial-congenital abnormalities	473	12.5
Multifactorial-common diseases	154	4.1
Mutagenic noxae	59	1.6
Consanguinity	44	1.2
Enviromental		
Congenital abnormalities caused by teratogenic or maternal factors	102	2.7
Enviromental factors during pregnancy	266	7.0
Clinical diagnosis		
Sterility	760	20.1
Fetal death	498	13.2
Early death	177	4.7
Defects (mental subnormality, etc.)	104	2.7
Congenital abnormalities of unknown origin	163	4.3
Diseases of non-genetic origin	78	2.1
General information	64	1.7
Total	3778	100.0

cost and benefit are an important argument against unsuitable referrals.

2. The appropriate persons to refer counsellees to genetic counselling clinics would be general practitioners or specialists of the Mother and Child Care System. To our genetic counselling clinic, however, 67.9% of the counsellees came on their own initiative. One cause of this fact may be the effect of two TV programme series in connection with genetic counselling and family planning which directly influenced the number of counsellees (Fig. 1). Physicians referred only 26.2% and the rest was called to report in connection with some special preventive programme such as the prevention of recurrence of neural tube defects, etc., or they were sent by other geneticists.

3. This point is connected with the two former ones. The necessity of genetic counselling has a wide spectrum within the potential consultands. It is well known that only a low percentage of those who would really need it, seek such advice (4). Experience in Hungary has shown that the severity of manifestation immediately after birth, the notoriety of the disease and its genetic background,

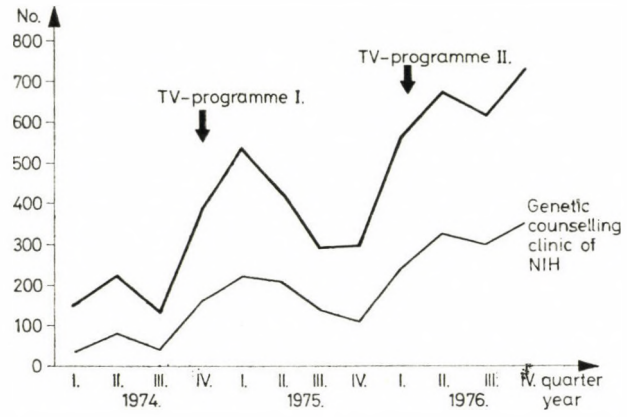

FIG. 1

and the educational level of the couples are the most important factors (3). Mendelian conditions manifesting in adult life have been rare in our material, e.g. hyperlipoproteinaemia type II has not occurred at all. The greatest problem is the absence of couples with low educational and socioeconomic conditions, e.g. the number of gipsy consultands was 6 compared with the figure 150 expected, based on their proportion in the population.

4. According to a well-known rule, the reliability and efficacy of genetic counselling depends on the accuracy of the clinical, and mainly of the nosological diagnosis. However, in a number of couples referred by physicians and mainly in the self-referral counsellees there was only a clinical diagnosis (e.g. cataract, muscular dystrophy, multiple malformation, sterility). Thus, the most difficult task of the genetic counsellor is to try to establish an exact nosological diagnosis. That is why we have a network of specialists. The previous diagnosis was sufficient for genetic counselling in 57.0% of the probands, and the diagnosis had to be changed in 22.8%. Unfortunately, in 20.2% of the consultands genetic counselling had to be done without a definite nosological diagnosis. Of course, in these cases we had to rely on empirical risk figures or subjective estimations. Another important rule of genetic counselling is that "any medical advice is better than no advice". As Fraser wrote: "Useful advice can still be given even in the absence of certainty" (7). Our data have proved the importance of an aetiological diagnosis. A recurrence occurred in 8.9% of the consultands who had previously had a baby with multiple malformation of unknown aetiology. In contrast, this rate was only 1.9% in the Mendelian conditions excluding cases when pregnancy had not been recommended (2).

5. The method of genetic counselling is an important theoretical prob-

lem. The classical English opinion is to give information without any advice (7). Carter (1) developed the method for consultands with low and moderate risk, encouraging the counsellor to say "In your place I would not take a risk of this kind too seriously". Our counsellees, however, needed, as they said, "more help"; they were not satisfied with the explanation of the risk and the perspectives, they usually wanted to hear a definite advice from the counsellor. This demand was the basis of our information-guidance type of counselling. Thus, knowing the history and the pedigree of the proband and having established the diagnosis, our counselling session involves three steps, viz. (i) information and explanation concerning the perspectives; (ii) a summary of the counsellor's advice; (iii) checking whether the explanation and advice had been understood. The information concerning the perspectives of family planning has been summarized in 8 points (Table III): (1) the severity of the expected disorders in the offspring; the possibilities of (2) treatment and (3) prenatal diagnosis; (4) the grade of the specific genetic (or teratogenic) risk; the possible environmental—"familial" hazard regarding the development of the offspring connected with (5) diseases of the parents, (6) the socio-economic status of the family, and (7) the familial structure: the number of healthy and affected sibs; and, finally (8) the potential maternal risk during pregnancy. Although genetic counselling is always individual, the advice given by the genetic counsellor may be considered to fall into five categories:

a) No significant problem (e.g. the specific risk is under 2 and sometimes 5% or the expected disorder is mild or treatable), pregnancy is suggested.

b) Pregnancy is suggested after some preparations, e.g. because the high risk is preventable by some treatment or the risk involves "only" fetal death, as in habitual abortion.

c) Pregnancy is suggested with prenatal examination, provided the latter is feasible and the risk is at least 2%.

d) Family planning deserves consideration for some specific reason, e.g. (i) the specific risk is between 5–19% and the expected disease is severe and there is no effective treatment; (ii) the disease (e.g. alcohol addiction, schizophrenia, mental subnormality) of the parents, mainly of the mother, may affect the offspring's development; (iii) the pregnancy increases the maternal hazard.

e) Pregnancy is not suggested, as the specific risk exceeds 20% in a severe untreatable disorder in the offspring or the pregnancy threatens the life of the mother.

Of course, the decision is the right and responsibility of the consultand, and the counsellor has to help its realization independently of his own previous advice. It is another point that a considerable part of our counsellees expect a directive for their family planning as they prefer to

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TABLE III

The main factors having an influence on genetic counselling (with an increasing hazard from left to right)

I. Specific genetic (or teratogenic) hazard							
1. Severity ("burden")	Minor	Mild	At disadvantage	Semilethal	Semilethal + at a disadvantage	Lethal	Serious long-term handicap
2. Treatment	Very effective	Effective	Low effectivity	No	"Severe"		
3. Prenatal diagnosis	Yes, reliable	Yes, not very reliable	None				
4. Specific risk	Minimal (<2%)	Low (2–4%)	Moderate (5–9%)	High I (10–19%)	High II (20–50%)	High III (>50%)	
II. Environmental-familial factors							
5. Dangerous disease of parents	Father	Mother	Father and mother				
6. Socio-economic status	Low	Very low	Dangerous				
7. Number of sibs	Only affected sib(s)	No sib	Healthy sib(s)				
III. Maternal hazard							
8. Hazard during pregnancy and/or delivery	Low risk	Moderate risk	High risk				

accept the counsellor's decision. This, however, is not permissible.

The advantage of this type of counselling is a high social efficacy as only 4.7% and 9.6% of the consultands did not want to have a further child after an advice of types *a–b* and *c*, respectively (2), as well as the fulfilment of the counsellee's demand. The disadvantages may be an offence against human rights and the hazard to the counsellor. The former point may be negligible because a summary of the counsellor's advice is asked for by the counsellees. Obviously, this kind of genetic counselling increases the counsellor's responsibility and may raise some legal problems. The random risk is a disturbing factor as the outcome of pregnancy may be unfavourable, even after adequate advice. Therefore, it is an obligatory task of the counsellor to draw attention to the unpredictable random risk. This problem is somewhat similar to the activity of general practitioners, and in Hungary the medical geneticists obtain protection against the random risk. From both the practical (efficacy) and the ethical (the interest of counsellees is more important than the interest of counsellors) point of view the information-guidance model of counselling is better than the non-directive model, i.e. information and explanation, because by such an approach we can give more help to our consultands leaving to them the right to make their own decisions. We have not experienced the alleged psychological drawbacks of our more direct approach (5).

6. In spite of the legal protection, the greatest practical problem is the random risk. Genetic counsellors could tell about several such cases. Let us mention here three cases only. In the first one, a woman whose first child was born with anencephaly, amniotic AFP-examination was suggested during her second pregnancy. The value was normal, the baby, however, was born with a complex heart defect. The other example was a male consultand whose two wives had one spontaneous abortion and three livebirths: these children were born with three different congenital abnormalities. The third case was a pregnant woman who allegedly had had rubella-virus exposition during this pregnancy. Because it had been excluded serologically, she was dissuaded from having her pregnancy interrupted. She agreed happily and later she delivered a baby with Down syndrome.

The 1973–1978 material of our genetic clinic included 3778 cases had 76 "clusters" of independent serious congenital abnormalities in sibs. Obviously this 2% frequency fits well with the random risk (2) but in practice it caused much distress.

7. Of the counsultands, 20.4% were pregnant when they reported at the clinic. The number of pregnant women seeking genetic counsel for teratogenic or maternal noxae, as well as of the women referred for prenatal diagnosis has to be deducted from this value. Still, the remaining 12.9% seems to be high. On the one hand, the time is too short to make the

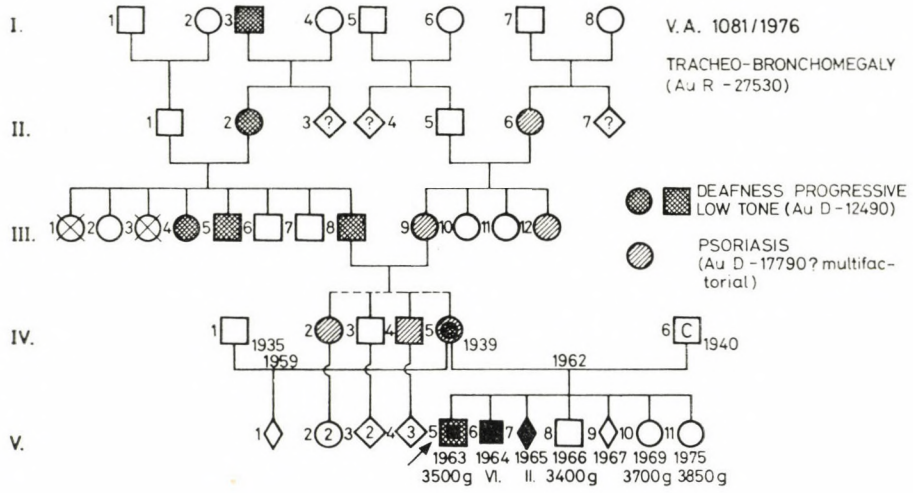

FIG. 2

necessary examinations and, on the other, the range of possible actions is limited.

8. The proportion of consultands who seek counselling after one or more affected children is extremely high. Obviously, fresh mutations and most recessive and multifactorial disorders are recognized only after the affected child was born. Still, 75 consultands, 5.1% of our material of genetic origin came to our genetic clinic after two affected children. The fact that there were 7 new counsellees with three seriously affected children each, is somewhat surprising.

9. The combinations of independent disorders in probands may sometimes raise a problem, e.g. cleft palate associated with coeliac disease; adrenal hyperplasia with myopia and congenital dislocation of the hip; phenylketonuria with spina bifida cystica. The question is whether these are different entities with different recurrence risks (i.e. their risk are additive), or they can modify each other's risk, or else, they represent a special nosological entity with one definite risk.

10. The so-called "mixed" pedigree (Fig. 2), when two or three independent inherited diseases appear in the same family, may also cause problems. Is there only a simple additive risk $(a+b+c-abc)$ or can they modify each other? It would be important to establish the principles of genetic counselling in such cases.

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