SPECIAL ARTICLES

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GENETIC COUNSELLING AND EUGENICS *

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Genetic counselling is the application of genetics in preventive medicine. The present-day genetic knowledge is so extensive and significant that it cannot be ignored by any doctor if he wants to give the best service to his patients. Furthermore all those who work in the field of public health must realize the place of genetics in the control of disease and promotion of health in a population. All of us must face the responsibility which rests upon us in relation to the protection of the genetic material which we have received from our ancestors and which we are handing down to our descendants. The present generation may be considered as the shells of the material on which the future of mankind has to be built.

The primary aim of genetic counselling has for a long time been the prevention of the occurrence of inherited abnormalities and disorders. The growing relative importance of genetically determined diseases has made the need for genetic counselling more and more pronounced. Genetic counselling has mainly been practised on the individual or family level, but many of the problems which now meet public health officers should also be referred to genetic counsellors for more complete elucidation.

The meaning of the word counsel is advice and/or recommendation. Most counsellors are, however, aware of the fact that genetic counselling must always be preceeded by collection of medical and genetic information and careful evaluation of the material obtained in each individual case. This makes it possible to explain the genetic situation and, following this, advice is in place. When the question is raised about which actions are to be taken as a consequence

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of the advice, the counsellor should remember that many aspects other than the purely genetic ones often enter the picture. Action can only rarely be determined by an isolated consideration of the genetic aspects of the problems; help may be needed from other medical specialists, e.g. psychiatrists.

The question which meets the counsellor is most often concerned with the risk of recurrence of some specific anomaly in a given family. This problem is quite naturally not raised until some abnormality has been observed at least once in the family and often not until the same type of event has occurred two or even more times in the same family. This latter situation provokes the more general question whether physicians should always give genetic advice or refer the persons involved to counselling clinics as soon as they have observed a possibly hereditary anomaly in a family.

The main principles of genetic counselling will appear quite clearly from a consideration of a typical case. Two parents, both seemingly quite healthy, have had a child with some abnormality of a serious nature and want to know if this may happen again. The counsellor should never give an immediate answer to their question; counselling can never be carried out on the basis of general statements or simple tabulations. The counsellor must know the specific background in each individual case before he is able to solve the problem. His first task is to collect all medical and genetic facts about the child. It is evident that the complete medical history of the child has to be obtained and, if the child is alive, it is not unusual that supplementary examinations are desirable; more extensive knowledge is quite often required for counselling purposes than for the establishment of a clinical diagnosis. The aim is to get the most exact and complete picture of the child in question and to disclose all disturbances of a biochemical, a cytological or any other nature, even if they seem to be quite trivial. The full elucidation of each individual case is indispensable if the most precise genetic information is wanted. But this is only the first step in the preparations for counselling. Even if it looks as if the abnormality of the child is quite clear and simple, the next step should *never* be omitted; this aims at the collection of information about both parents and their families. The minimum requirement includes a search for any significant deviations from the normal in the parents, the sibs, and quite often in the sibs of the parents and the grandparents of the child, and in some cases

even this is not enough. Contacts with general practitioners, hospitals, and other medical services are often established in order to supplement or verify diagnoses of disorders or anomalies observed among the family members. Furthermore, special examinations of the parents or other family members may be needed in order to supplement the genetic picture of the family. These investigations often call for collaboration and conferences with other specialists who are thus made members of the counselling team. In Denmark much valuable information is obtained through a registry of hereditary or possibly hereditary disorders kept by the Institute of Human Genetics. This includes all cases of mental retardation diagnosed in Denmark during the last 25 years, almost all cases of psychoses from the same period, quite a high proportion of known cases of neurological disorders and, in addition, many congenital malformations—harelip and cleft palate being the most complete group. It is mainly the responsibility of the medical geneticist who finally puts all the pieces of information obtained together to make sure that all possible sources of information have been exhausted. It is only too obvious that the task of the genetic counsellor is getting more and more difficult as he ought to have a good knowledge of all major studies relevant to the case in question. Two individuals may present what seem to be identical types of defects, but even very minute anomalies in one of the individuals, or the pattern of the family distribution of the same abnormality, may disclose that quite different genetic mechanisms are at work, leading to quite different genetic risks in the two cases. A few spots in the cornea, a displacement of a tiny bit of only one chromosome, a minimal change in the amount of just one of the hundreds of the constituents of the organism may be the decisive guide to an exact answer to the questions asked by the family. After having tried to realize the genetic situation in the family under consideration it is the task of the counsellor to find the risk figures to be used in the evaluation of the genetic prognosis; this states the risk of recurrence of the same or a related type of abnormality in a relative of an individual with a given hereditary anomaly. The *exact* or theoretical figures are applicable if the mechanism behind the anomaly in question is genetic and fully understood, and if there is no reason for suspecting that the laws deduced from previous studies of this type of anomaly do not apply in this particular instance. When the etiological background of a given anomaly is obscure and the possible influence of genetic factors cannot be excluded, *empirical* risk figures must be used—if they exist. They are also based on research

which may be even more extensive than in cases of anomalies with a simple mode of inheritance, but which have not permitted the formulation of any valid hypothesis and, consequently, exact risk figures cannot be deduced from any theory. If, however, such studies conform to a number of specifiable requirements, fully reliable risk figures may be calculated on the basis of these materials; as these figures do not rest on any theory regarding etiology, but directly on the findings, they are labelled empirical figures. Although they may be applied safely in a high number of cases, their natural limitations should always be kept in mind. Empirical risk figures are based on collections of cases which undoubtedly often have various genetic as well as non-genetic causes. Although such figures are valid on the average, they may be too low or too high in a given situation. The specific case encountered may in reality have been caused by purely non-genetic factors which implies that the average empirical risk figure is too high as the true risk of recurrence is negligible in this special situation. On the other hand, the case under consideration may happen to be one of extremely few caused by a rare gene, and the true risk figure may here be higher than the average value. It seems preferable to try to explain these limitations to the families seeking advice. This will also make it easier for them to understand why the estimate of the genetic risk may be greatly changed when for instance a new case of the same or some analogous anomaly appears in the family. This latter fact forms one of the reasons for asking the family to come for renewed consultations when the etiology of the disorder in question is not quite clear at present and an exact figure cannot be given. The same applies if the counsellor must admit that it is impossible to give any reasonably valid estimate of the risk in the situation which is being presented to him; here it seems permissible to suggest that a consultation some years later may be rewarding considering the speed at which new facts about human abnormalities are brought to light.

One of the most difficult problems is the isolated appearance in a family of an abnormality known to be caused sometimes by genes, perhaps with different modes of transmission, and sometimes by purely non-genetic factors which are difficult or impossible to identify. Such an abnormality may for instance have appeared in just one son of healthy parents with fully elucidated, but perfectly healthy families. Previous research is only very rarely extensive enough to permit an estimate of the relative proportion of what are usually called sporadic cases, due to mutation or to non-genetic factors, which would imply practically no increased

risk of recurrence in the same sibship, and it may be impossible to evaluate the chance of the case being due to a recessive or a sex-linked gene.

As far as the practical performance of genetic counselling is concerned it has been customary in Denmark to accept people only if they have been referred by a physician, quite often the family doctor. We usually ask the individual to come to the Institute for personal interview if they live in Copenhagen; if they live outside, information and advice are usually given by the family doctor. In all cases the doctor who has referred the person would get a full report about the results of all our investigations and about our estimate of the genetic risk.

What is to be expected from genetic counselling? The immediate results are easier to see than the more remote effects. It is hoped that the family gets an understanding of the genetic risk when the situation has been carefully explained to them. It is not unusual that a new interview after some weeks is rewarding; this allows the family to absorb and digest the information they have received and to bring forward any problems which may have occurred to them in the meantime. Furthermore, a second interview gives the counsellor an impression of whether the family has fully understood the explanations; this is a field where misunderstandings may have very serious consequences. A feeling of relief is pronounced not too rarely when the situation has been carefully exposed. It is not uncommon that people have unrealistic and very exaggerated ideas about the risks involved. If a couple after full explanation of the situation decides to run the risk, the genetic counsellor may contribute to early diagnosis and treatment if the inherited abnormality should turn up again by informing the family doctor about the chances. The importance of this type of prophylactic work is clearly evidenced by galactosemia which is caused by a recessive gene.

It would be unreasonable to expect that genetic counselling of families as described here would lead to an appreciable improvement of the qualities of the population as a whole which is the main aim of eugenics. This can only be obtained by other more extensive and profound measures. Genetic counselling as outlined here is certainly serving aims which have many points in common with eugenic ideas and thoughts, but the important differences should be kept in mind. Genetic counselling is based solely on the science of human and medical genetics, whereas eugenics

has a much broader base and may be called a science of its own. Complicated value judgements cannot be avoided in the development of eugenic programs, but do not enter genetic counselling. It may be found difficult to reconcile personal liberty, on which genetic counselling always rests, with effective eugenic measures. Furthermore, it is worth pointing out that it was Galton in Great Britain who, more than 80 years ago, coined the term eugenics. This new science was an obvious consequence of Darwin's theory of evolution and especially of its emphasis on the primary importance of natural selection. It seemed not unreasonable to suppose that civilization was interfering with natural selection or at least had very good opportunities of doing so. Galton felt that a new science had to be developed to study these very important problems. Eugenics did *not* grow out of human genetics. Its primary goals and many of its methods were fixed long before genetics existed. Galton emphasized that improvement of the population could rather be obtained by increasing what he called «the best stock» than by preventing reproduction in the worst; the fact that it was completely unknown whether desirable qualities were inherited and whether «the selected stock» would get children of the same high quality was disregarded. These so-called *positive* eugenic measures were, however, never put into practice, and soon after Galton's death in 1911, *negative* eugenics attracted much more general interest; this aimed at the prevention of so-called «unworthy parenthood». In many places radical measures were now introduced, and by 1930, 26 American states and two countries in Europe, a Swiss canton and Denmark, had introduced voluntary sterilization laws. The picture was completely changed by the introduction of negative eugenics in Nazi population policy in 1933. It was here stated that it was not only a right but also a duty to apply the recent genetic knowledge and prevent the procreation of individuals incapacitated by hereditary abnormalities. The ultimate result of this action was not only the death of eugenics in Germany, but also a wide-spread disgust at everything connected with eugenics *and* genetics which were in many places considered as synonymous.

A few countries in Europe and some of the states in North America succeeded in keeping the path of eugenics spotless during this period. Norway, Sweden, Finland and Iceland enacted voluntary sterilization laws between 1934 and 1938, and in 1939 Denmark took a remarkable step by making legal the voluntary interruption of pregnancy on eugenic indications. Similar laws appeared shortly afterwards in Sweden and Finland.

One might think that new demands on eugenics could now be expected as a consequence of the population explosion in many areas, as for instance Japan and India, but such considerations have, however, found no place as yet. When it becomes possible to bring eugenic thinking and ideas to such countries, the lessons of the past should be remembered. The use of fallacious arguments so often presented as established facts by the older eugenics should be avoided. An open discussion of the present *lack* of genetic knowledge is essential in order to prevent the creation of distrust of eugenics and human genetics in general. To mention just one of the controversial points : it should be made quite clear that as yet, no universally valid answer can be given to the question about what happens genetically or eugenically, when the infant mortality rate falls. This is a very characteristic trend in many places all over the world. It is often stated that an increase in the number of weak or sick individuals will result, not only immediately, but also in later generations. The genetic background of this postulated weakness leading to early death is not at all clear, and if genes are unimportant, it cannot straight away be taken for granted that an increased number of weak individuals will appear. It has not been proved that those who previously died in infancy or early childhood were generally weaker. On the contrary, as is well-known, in certain areas deaths from malaria affect a higher proportion of individuals carrying *normal* genes at a given point in their chromosomes than of individuals who at this place have abnormal genes, leading for instance to sickle cell anemia.

Although it is desirable that close contact is kept between eugenics and human genetics, the two disciplines should never be confused. Geneticsts taking an active interest in eugenics must be aware of their great responsibility; eugenic statements without sufficient genetic basis may once again endanger the development of eugenics as well as of human genetics. It is now and then overlooked that *no* single gene is yet known which is invariably good or bad, favorable or harmful. Even if agreement could be reached as to what should be called the human ideal, we would not yet be able to tell to which extent it is built upon a genotype of a specific kind with a fair chance of being represented in the offspring and showing itself equally ideal in the evironment existing 30-40-50 years ahead. Finally it should be kept in mind, that we are *all* of us carriers of potentially detrimental genes.

Genetic counselling is needed and acceptable everywhere as it is one of the tools of preventive medicine. It tends to transform

medicogenetic research and theory into information of practical value as a particular branch of medical practice. Genetic counselling is thus intimately connected with medical and genetic research. The types of study which may be expected to bring results of the greatest immediate significance to genetic counselling are first of all those which try to disclose the heterozygous carriers of autosomal recessive genes or the female carriers of X-linked recessive genes. Furthermore, extensive population studies, especially of diseases of a heterogeneous nature, are wanted as they may give a basis for the separation of etiologically different entities. Every step forward within these fields will alleviate the task of the counsellor and make his contribution to medical practice still more important.