Genetica Psichiatrica

Psychiatric Genetics

SESSIONE DI CONFERENZE LECTURE SESSION

11 Settembre 1961

- F. J. KALLMANN: Introductory remarks.
- L. Hurst: Classification of Psychotic Disorders from a Genetic Point of View.
- E. Strömgren: Genetic Implications of Alcoholism.
- D. A. PRICE-EVANS: Pharmacogenetics.

The Hybrid Specialty of Psychiatric Genetics

(Introductory remarks)

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No matter what our everyday activity in the mental health field, we can say with conviction that the function of a new superspecialty such as *Psychiatric Genetics*, the subject of this latemorning session, is profoundly influenced by contemporary cultural, social and political conditions as well as by the never-ending challenge of scientific conquests (7). Evidently, therefore, any attempt to formulate a concise and generally acceptable definition of such a modern hybrid discipline is no less difficult than it is to work in it. Actually, since the two parental sciences deal with equally absorbing, intricate and still rather speculative aspects of human existence and health, it would make little difference here if the given definition were developed within the framework of *psychiatry* or that of *human genetics*. What is certain only is the empirical observation that there are very few workers in this particular field of superspecialization, who are not prematurely grey or otherwise debilitated.

In order to facilitate a fruitful discussion despite these considerable difficulties, it may be advisable to use a description of psychiatry and its various schools of psychotherapy, which is plain in its formulation and is focused on appropriate *medical* concepts and techniques from a professional standpoint. This selective procedure is justified, since in many recent conferences on the functions of clinical psychiatry, increasing emphasis has been placed on two general professional requirements (10). One of them is that psychiatry should never be "a field apart from other areas of scientific discourse" (11). The other criterion is that a psychiatrist be "a good physician first and always... capable of acting as the trustworthy friend, the wise counsellor, and the discreet and skillful servant" of people in distress (2, 12).

Hence, a qualified psychiatrist — however profound his knowledge of psychodynamics or however great his mastery of psychotherapy — should be able to use, in the words of Felix (2), "a variety of basit medical skills which any patient has a right to expect of any physician regardless of his specialty".

Even if this were not an assembly of geneticists with a special interest in mental health problems, one would have to specify here that these stipulated skills should include a definite familiarity with modern concepts and objectives of *medical genetics*. Since psychiatrists are known everywhere to represent to trusting and unknowing people one of the unquestioned authorities on human genetics (15), they are frequently consulted in relation to a great variety of difficult family problems.

In line with these general professional considerations, a simple definition of psychiatry would be either that of "a medical specialty concerned with the prevention, diagnosis and treatment of mental and emotional disorders" (2) or that of "a science which deals with the psycho-

pathological aspects of human biology " (14). Accordingly, we may define psychiatric genetics as a sub-discipline within the medical and biological sciences, which is concerned with the study, management and prevention of genetic family and population problems associated with mental disorders, intellectual defects and various other types of emotional maladjustment. In connection with the explanatory or manipulative forms of genetic counseling programs it may be mentioned, however, that current definitions of psychotherapy per se tend to be on the more comprehensive side. In accordance with some of the definitions offered at the recent Congress of Psychotherapy in Vienna, psychotherapy is "a form of treatment for problems of an emotional nature in which a trained person deliberately establishes a professional relationship with the patient, with the object of removing, modifying or reducing existing symptoms, of mediating disturbed patterns of behavior, and of promoting positive growth and development " (5).

Even without superspecialized applications of this kind, however, it is unquestionable that the area of scientific investigation and family guidance work, covered by psychiatric genetics in relation to etiologic, therapeutic and preventive problems, is a wide, promising and particularly important one, psychiatrically as well as genetically. The area extends from maturational deficiencies arising from gross chromosomal disarrangements to that multitude of psychiatric disorders resulting from a quantitative or qualitative change in the production of an enzyme, triggered by some structural defect in a DNA molecule (8); from the earliest forms of a schizophrenic psychosis to the decrepitudes of premature aging and other afflictions of the senium; from excessive drinking patterns to some of the most destructive forms of criminal behavior. No one denies any more in scientific terms that the deleterious effects of most mutant genes "are diluted and postponed by an improved environment, rather than obliterated " (1); or that, as has recently been re-emphasized by Crow (1) and others (4, 13), selection can be under individual human control and be employed with foresight, calling for "birth selection, not death selection... for criteria of health, intelligence, or happiness, not just survival and fertility ". In other words, the real issue is not whether man is influencing his own evolution, but in what direction he should do it.

Within the province of psychiatric-genetic counseling services, it is certain that one of the most demanding family management tasks encountered is that of providing scientifically valid and psychologically unharming guidance in connection with problems of marriage and parenthood in the presence of a pathological condition in one or both of the families concerned (6, 10). Since there are no more fateful decisions in man's life than the one to marry and the one to assume parenthood, it is obvious that whenever beset by uncertainty or doubt — whether well- or ill-founded — any family in an enlightened society should have access to competent advice. In this kind of activity, a conscientious and adequately trained counselor will be guided by the general objective of the geneticist to encourage a person's feeling of responsibility for his own self, without violating the all important concept that a well-planned family is indispensable as a biological, social and cultural unit from the society's standpoint, and as a source of pride and stability for the individual. When confronted with a calculable health risk in a predictably unstable home, he will be sufficiently detached to consider that in this age of growing concern for the welfare of children, every child born "should be given a fair chance in life" (16).

It is also understood that in order to be of constructive help to unknowing or perplexed people in this kind of counseling situation, a good counselor is not only expected to elicit and discreetly evaluate the essential facts bearing on a family's decision, but he will also attempt to understand the given persons' fears and hopes, defenses and rationalizations. Since the majority of people come to such a clinic session with both "reasonable and unreasonable expectations" (3), they should not be assumed to be capable of making important decisions in family matters without clear instruction and reassuring guidance. It is a common observation that even highly intelligent people, when frightened either by some factual morbidity risk information supposed to dispel their state of perplexity and technical unknowingness or by the need of being evaluated in their roles as actual or prospective mates and potential parents, may regress to immature levels of emotion and thought.

It follows, therefore, that information of this nature should neither be given in an impersonal way — for instance, in the form of bare statistics or through third persons — nor without sufficient understanding of the motives and capacities of the persons who come for help. Although the required interviewing sessions need not be categorized as a formal program of psychotherapy, they will greatly gain in effectiveness if conducted according to the rules for short-term psychotherapy aimed at reducing anxiety and tension.

Another requirement of genetic counseling is that when the stage is reached where a program of action can be formulated without psychological harm to the person or persons involved, each step should be suited to their individual needs and resources, both in its contents and in its manner of presentation. In many cases, of course, the decision to marry and that to have children will have to be dealt with as two separate problems. If one or the other problem raises serious doubt, it will be advisable to act in accordance with the truism that healthy children—healthy in terms of genetic endowment, developmental circumstances, and emotional climate in the home—are both the product and the source of health in the parents.

Whenever a preference for a couple's childnessness is indicated on genetic grounds, a frank discussion of the modes of operation and potential risks of certain *adoption procedures* will be most helpful. Obviously, the risk connected with the unknowing adoption of the natural child of two schizophrenics more than equals those of harelip or diabetes in one's own child. Justifiable fears of this kind cannot be effectively dispelled by the glib assertion that the effect of heredity in man is negligible.

Following this brief and rather incomplete description of the *general functions* of psychiatric genetics, we shall have the pleasure to hear several interesting research reports on *more specific problems* of our hybrid- or super-specialty. I am sure that you are looking forward as eagerly to this well-assorted set of reports as I do. My only regret is that Dr. Slater's report on "Genetic Factors in Neurosis and Psychopathy" has already been presented in another session.

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