6/30/68

The main conclusion that we've reached so far is that the Society for Human Genetics ought to press the National Academy of Sciences to set up working groups that will deal directly with ~~the~~ a number of issues of genetic interest. The Society could provide the impetus for taking interest in a number of specific questions, and could also suggest some of the people who might ~~serve~~ serve as experts as consultants on such committees. In very many cases these will be necessarily interdisciplinary, and for that reason the Academy is a very much better vehicle than the Society would be for dealing with them. For example, if the present group were to function as this consultative committee, it might be in a position to formulate the following specific recommendation:

We propose to the Academy that it set up a working group on large scale screening programs that will take into account the current controversy over the obligatory screening for PKU that operates in a number of states, and help to clarify some of the confusion that has arisen out of the criticism of this program. On the one hand, it is very apparent that there have been many shortcomings in the management of that has followed upon the discovery of examples of hypophenylalaninaemia. On the other hand it seems likely that the programs have already uncovered uncovered enough cases of the disease to have justified the continued existence of the program. Such a committee ought to survey the way in whcih current practice for screening is implemented, the effectiveness of the organization of such programs in various states, make specific technical recommendations about the way in which samples should be collected, forwarded to the central laboratories, specific criteria for the analysis of the samples, and the way in which these results are reported. It should also survey the manggement techniques that have come into being for the treatment of proposed cases of hypophenylaneninaemia, with partifular reference to the differentiation

between those likely to lead to mental retardation if untreated,and otherwise. It should also make some recomendation with regard to the cost effectiveness of screening programs as compared to other investments in medical metabolic disease. It is also important to bring in advice from the Pediatric Society and from the World Health Organization, which have interested themselves deeply in this matter, and of course, a very critical consideration is the availability of trained manpower to run programs of this kind. All of this presuppposed that a committee will have been preselected to represent the societies con-concerns for changes in public policy. The title of the committee might be "Advisory Committee on Public Issues in Human Genetics". One of the principal by-laws of the committee should be that its principal function is to press the National Academy of Sciences to set up appropriate machinery for specific studies on the topics that are suggested by our advisory committee and those of other interested societies. There ought to be specific provision in an amendment to the by-laws of the society to provide for the composition of this comittee. It should be a permanent committee with a membership rather like ten than five, however able to act with a quorum of as few as four or five members in order to take account of the difficulties of meeting that such a committee would have. Since the committee will never act in a conclusive fashion with respect to the policies of the society, this ought to be quite acceptable. We would have to have more rigorous requirements for its member-ships if it were to make direct public statements that reflected the considered judgment of the society as a whole. The committee presumably would be appointed by the President, and should be arranged so that it rotates one-fifth of its membership every year, which would presuppose a term of five years for its members. A reasonably long tenure would seem to be important in order to develop familiarity with the problems and with the political procedures that are effective, and all the rest of it that goes into the work of such a committee.

Note: There ought to be some informal communication with Henry David and others at the Academy about how they would respond to such an activity. There is no point in pursuing it if they would be extremely reluctant to have this kind of pressure from outside. We're supposin g that they will not be reluctant. We might even try to get the Academy to help provide the financial base for the cooperation of the Society with it, that is, to help provide travel expendes for this committee, etc. The way we now view it is that the standing committee would make recommendations to the Academy about the substantive issues that require further study, and some suggestions about experts in the field of human genetics would be able to contriubte technical expertise. In almost every case there will be considerations other than human genetics in relation to public policy, and we would leave it to the Academy to attempt to provide very broad coverage for these considerations and of course would provide a stamp of prestige of the Academy on its deliberations. A specific liaison between the Board on Medicne and the Committee on Life Sciences and Public Policy, since they reflect two different divisions of the Academy, will have to be created and the workinggroups ought to be developed out of that.

The Standing Committee on Social Issues of the American Society of Human Genetics coulc nominate some people for the study on screening, namely, Scriver, Charles (Montreal), Barton Childs, Norman Kretchmer. Perhaps the pediatricians ought to be nominees of the Pediatrics Society rather than the American Society of Human Genetics.

There ought to be some activity to elicit grassroots interest in a number of other societies who might be encouraged to make representations very similar to those that we are suggesting for the Society of Human Genetics. For example, the APHA, the American Anthropological Society, the Population Association of America, the American Eugenics Society, and the Pediatrics Society of America.

There is no specific point in restricting this to concerns about genetics, and almost any one of the other medical professional societies who don't have their own large lobbies in Washington at the present time might very well want to second members to this kind of consultation with the Academy. More names might be furnished a little later on, but we're trying to get together a model recommendation that we might make so that we have some idea of what the functions of such a group might be later on. Possibly there ought to be a little more text outlining the nature of the problem, that is to say, some preamble that says why this ought to be studied and what kind of confusion has arisen - some suggested instructions and guidelines for the task force that is supposed to go into it.

Recent progress in biochemical genetics has revealed the existence of many detectible biochemical errors which may lead to serious disease in infants and children, In addition to phenylketonuria, diseases like galactosemia, maple syrup urine disease and a variety of others can be detected. The possibility of treating these diseases makes it mandatory to sponsor programs in which the best ways of screening and best ways of treating these diseases can be elucidated. Work in biochemical genetics has also shown that it is likely that in each of these conditions considerable heterogeneity can be found. It is possible, therefore, that conditions showing as possible disease on initial screening turn out to be innocuous. Particular attention will have to be devoted to the separation of these entities from real disease-causing biochemical errors. The question also has been raised whether the cost of extensive screening programs justifies detection of very rare diseases, such as the order of magnitude 1 in 40,000 or so. It should be recognized, however, that the availability of autoanalyzers may make the addition of additional test of even rare diseases possible without much additional cost. Although laws for phenylketonuria screening have been passed in most states of the United States, there is still no agreement whether this kind of activity might be better done without passing laws at this time. The complexities of some of these programs also raise the question whether enough trained personnel is available to execute a high standard scientific and practicable program at this time all over the country. An initial activity of the committee might be a careful evaluation of the phenylketonuria program which has been passed in all but very few states in this country. The committee also might pay particular attention to the feasibility and advisability of screenigg for much more common biochemical errors which potentially lead to disease. An example of this sort of the various hypolipoproteinemias which can be detetted at birth, which presumably lead to early ? arterio athrosclerosis. Treatment methods for these diseases

are becoming increasingly available and are already being tested in a variety of studies. Attention also might be paid to the feasibility of storing specimens for later testing of new traits or diseases which might occur outwardly which could be better by the existence of having specimens taken at a much earlier time. It has been pointed out that the placenta is a very rich source of fetal tissue that usually is discarded. The possibility of doing studies on placentas and saving placentas in some manner might be considered. As a point of information we need to know whether phenylkentonuria can be diagnosed on the basis of placental specimens. The point was raised as well whether the new cystic fibrosis tests could be done with placental cells. It was mentioned that some official body of obstetrician/gynecologists would need to be brought in if we are seriously talking about saving placental tissues. The problem of cystic fibrosis was mentioned in that screening at birth may be possible by methods other than blood screening done heretofore. Placental screening or tissue cell screening are the possibilities.

AIMS OF THE SEMINAR

1. To identify some important social problems (and to understand the processes for identifying these problems) raised as a consequence of applying biological discoveries.
   a. Introduction and exposure to the current state of science and technology in relation to the problem (including deficiencies in current knowledge).
   b. Examination of the possible consequences of applying these discoveries.

2. To study alternatives available to society for dealing with the problems and the likely consequences of these alternatives.

3. To determine what additional studies need to be done to reduce the uncertainties about likely consequences.

4. To develop criteria for deciding which alternatives should be chosen.