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OFFICE OF THE DIRECTOR



STATE OF CALIFORNIA
Department of Public Health

2151 BERKELEY WAY
BERKELEY 4, CALIFORNIA

January 15, 1963

Joshua Lederberg, Ph.D.
Professor of Genetics
Department of Genetics
Stanford University School of Medicine
300 Pasteur Drive
Palo Alto, California

Dear Dr. Lederberg:

At its meeting December 11, the Advisory Committee on Maternal and Child Health accepted the report of the Subcommittee on Human Genetics and recommended that the Subcommittee be dissolved.

I want to take this opportunity to thank you for the assistance given this department as a member of the Subcommittee on Human Genetics during 1962. Your help and counsel have been most helpful.

Very sincerely yours,

A handwritten signature in cursive script that reads "Malcolm H. Merrill".

Malcolm H. Merrill, M.D.
Executive Officer
State Board of Public Health

California

California State Department of Public Health
Bureau of Maternal and Child Health

Subcommittee on Human Genetics
of the
Advisory Committee on Maternal and Child Health

Minutes of meeting on June 4, 1962, at Stanford Medical School, Palo Alto

Members Present:

Herbert Bauer, M.D., Health Officer, Yolo County Health Department, Woodland
George N. Donnell, M.D., Professor of Pediatrics, University of Southern
California School of Medicine, Los Angeles
Alfred G. Knudson, Jr., M.D., Department of Pediatrics, City of Hope Medical
Center, Duarte
Norman Kretchmer, M.D., Professor and Chairman, Department of Pediatrics,
Stanford University School of Medicine, Palo Alto (Subcommittee Chairman)
Joshua Lederberg, Ph.D., Professor of Genetics, Department of Genetics,
Stanford University School of Medicine, Palo Alto
Curt Stern, Ph.D., Professor of Zoology and Genetics, University of California,
Berkeley
Richard Straw, Ph.D., Associate Professor of Biology, Los Angeles State College
Los Angeles
Stanley Wright, M.D., Associate Professor of Pediatrics, UCLA School of Medicine,
Los Angeles

Guests Present:

Robert Goodlin, M.D., Assistant Professor of Obstetrics, Stanford University
School of Medicine, Palo Alto
Professor Conrad Kerpel-Fronius, Visiting Professor (at Stanford) from the
University of Pecs, Hungary

State Department of Public Health Staff Present:

Allan M. Butler, M.D., Special Consultant, Bureau of Maternal and Child Health
William A. Clark, M.D., Assistant to the Chief, Division of Research
David S. Kleinman, Assistant Chief (Administrative), Bureau of Maternal and Child Health
B. J. Milmore, M.D., Chief, Bureau of Chronic Diseases (arrived at 10:45 a.m.)
Theo. A. Montgomery, M.D., Acting Chief, Bureau of Maternal and Child Health

Members Absent:

Charles R. Graham, M.D., Director, Berkeley State Mental Hygiene Clinic,
State Department of Mental Hygiene, Berkeley
Ernest W. Page, M.D., Professor and Chairman, Department of Obstetrics and
Gynecology, University of California School of Medicine, San Francisco

California

The Chairman opened the meeting by requesting the participants to outline the significant questions in human genetics and their medical application today.

It was stated that, although DNA coding is an important and basic area in genetics, it would not be appropriate to the purposes of this meeting which relate to areas of more immediate concern to the State Department of Public Health.

The following were presented by members of the Committee as important issues of concern to persons interested in human genetics and disease:

1. Statistics regarding:
 - a. Frequency of diseases involving genetic factors and characteristics of distribution of such diseases in the population;
 - b. Rates of consanguineous marriages;
 - c. Mutation rates in human population, including effects of radiation;
2. Genetic counseling services, including resources for information and consultation which could be used in genetic counseling;
3. Chromosome laboratories which could provide services for 2. above and for research.

Counseling Services

Several members noted:

1. The need for genetic counseling services for couples recently married who had questions as to their being carriers of genetic disease and for parents who have had a child with a genetically determined anomaly and are concerned regarding future children;

2. Until physicians are trained to be competent in human genetics and even thereafter, centers will be required to provide information and consultation to physicians on genetic aspects of specific disease entities, as well as, perhaps, to provide counseling directly to individuals; and
3. Reservations about the counseling of patients by anyone other than their physicians, since genetic counseling should be an integral part of medical practice, including concern with social, personal and psychological characteristics of patient, as well as family history and diagnoses involved.

It was pointed out that:

1. Frequently the diagnosis of disease involving genetic factors is difficult although in some pathologies such as PKU, galactosemia, Mongolism, the mechanics are relatively better known but there seemed to be little consensus that these entities were significant enough to warrant special centers are present beyond the facilities now providing counseling services;
2. There was concern from one of the Committee members at least as to whether he could provide consultation with confidence in this area unless he could be confident of the diagnosis which as was noted was difficult;
3. There was some question as to how well the general physician untrained in genetics would be able to use the consultation and information provided by a counseling and information center; and
4. Specialists are generally familiar with the genetic factors in the areas in which they are expert, such as in cardiology, etc.

However, it was also noted that despite indications that counseling should be part of medical practice, most physicians are not qualified; and until the medical school curricula and interest catch up with the need, some supportive centers would be required.

The discussion turned to the needs and demands for such services.

The City of Hope genetic counseling service is part of a total medical care program, the demand for services is not heavy and there are problems with getting sufficient information from referring physicians.

Others, however, did feel some heavy demand but it may have been only apparently heavy because consultation was extra work for them, not part of their regular assignments and it was difficult to estimate what demand there would be for an organized and well-publicized (to the medical profession) center.

Chromosome Laboratory Facilities

The next item discussed was the need for chromosome laboratory facilities.

The City of Hope operates a chromosome laboratory but the work is largely research oriented, very little resources being available for direct services. It was noted that cytogenetics is not well enough developed and progress is very rapid in this area so that a strictly service laboratory would not be feasible although some service might be provided by research laboratories if financial assistance were available from the State Department of Public Health or others. Some of the service provided might pay for itself if financed by the State. For instance, identification of translocation in Mongoloid births to younger mothers might, through counseling, prevent some of the future mongoloid births with consequent savings of long-term institutional care costs.

A question was raised regarding the State Department of Public Health research program and presentation outlining the program was made by Dr. Clark.

Data Collection and Statistical Information

As information is required regarding the frequency of genetic diseases, suggested diseases were noted which might be reported to a State registry, thus providing information on incidence and a source of data for research and other purposes. These were:

Achondroplasia
Amaurotic idiocy
Cerebro-ocular renal disease
Cleft palate and lip
Coagulation defects
Cretinism
Cystic fibrosis
Dislocation of hip
Drug sensitivities
Erythroblastosis
Galactosemia
Huntington's Chorea
Mongolism
Muscular dystrophy
Nephrogenic diabetes insipidus
Phenylketonuria
Pyloric stenosis
Sickle cell anemia
Thalassemia

It was emphasized that birth certificates may not be valid sources of data for reporting and that a hospital reporting system would be more valid. It was also suggested that study of a high risk population for incidence might be more fruitful than total population registry. It was agreed that information is very much lacking and even a minimum nose count and identification would be useful; prospective studies might be made on surgically treated congenital heart disease, possibly with follow-up of offspring.

The Chairman introduced the afternoon session with the directive that the principal consideration of this session should be the role of the State Department of Public Health.

It was the consensus of the Committee that the State Health Department should be concerned with hereditary diseases and does have a role in this area.

It was suggested that the State Department of Public Health employ a human or medical geneticist to coordinate its responsibilities in this area. While it was recognized that there may be difficulty in recruiting a highly qualified person for such a position, there was agreement that responsibility for the State's involvement in human genetics be a specific assignment with appropriate staff to carry it out with assistance from a small expert advisory and consultant group.

Discussion regarding the responsibilities involved were concerned with:

1. Data collection and investigation, including registries to determine incidence of genetic diseases, incidence of consanguineous marriages, twinning and related data. It was felt that the State Department of Public Health was in the best position to perform this service. This would assist in determining to what extent genetic disease is handicapping, the number of people affected and at risk, remediability of conditions and as resource for further research interest.
2. Genetic Counseling Services: Five sources were identified as providing consultation on genetic counseling but, by and large, this is a "side line" to their other principal functions and with present resources, it appears questionable that their activities in counseling and consultation could be expanded. These sources are:

City of Hope Medical Center.....	Dr. Knudson
Los Angeles Childrens Hospital.....	Dr. Donnell
Stanford Medical School.....	Dr. Kretchmer
UCLA Medical School.....	Dr. Wright
University of California.....	Dr. Stern

It was suggested that the State Department of Public Health list these centers as approved resources and circularize this information to appropriate persons and agencies, but most of the resources mentioned are not prepared to accept the additional work which may be entailed. Also, there was some question as to whether the adequacy of information and knowledge available and personnel available to provide an acceptable service would warrant expansion of services beyond what is presently being provided. There was a consensus that the State should not operate any counseling services and there was mixed response to the idea that the State should promote the establishment of counseling centers outside of supporting (including fiscal support for expansion) the present work of medical schools and medical centers where emphasis would be in developing medical competence in this area. No resolution was reached in this area; most of the discussants questioned the advisability of promoting counseling centers outside of medical school centers and related facilities.

The Subcommittee considered the role of the State Department of Public Health in training in human genetics. It was the consensus that the State Department of Public Health should stimulate, be involved in and support symposia and similar meetings conducted by medical schools and appropriate professional organizations.

In summary, the Subcommittee at this meeting indicated:

1. The State Department of Public Health should not be involved in promoting genetic counseling or chromosome laboratories except through support, including fiscal resources, of present and future medical school and medical center services and related facilities where competence is assured.
2. The State Department of Public Health should encourage, support and participate in symposia, and other training programs in human and medical genetics.
3. The State Department of Public Health is the agency best suited for collecting data, including establishment of registries and conducting related studies, to determine incidence and significance with regard to lethal, remediable, preventive and handicapping characteristics, and to provide data for research; that certain diseases may be made reportable.
4. The State Department of Public Health should assign a position to be responsible for work in human genetics. Committee members felt that additional staff should be employed. The consensus was that the field and its implications are of sufficient significance to warrant a specific and definitive program with staff to carry it out.
5. A small expert advisory group should be appointed to provide continuing consultation in this area as needed.

The Chairman thanked the Committee for its assistance, noted that it would probably not need to meet again as a Committee, that he would recommend its dissolution to the Advisory Committee on Maternal and Child Health, and that he hoped the members of the Subcommittee would be prepared to act as expert consultants in assisting the State Department of Public Health to organize and conduct its activities in this area.

DSK:mc
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