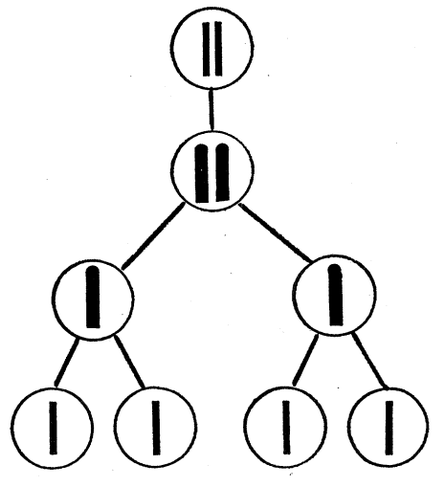
Advances in Human Genetics and Their Impact on Society



AAAS Symposium

28 December 1970

Chicago

Although Mendel's papers were published in the 1860's, Mendelian genetics can be called a 20th-century science. Even though some classics on human genetics were published earlier, it can be safely said that several of the outstanding developments in the field have occurred just in the last few decades. For example, the chromosomal basis of a disorder described clinically in 1866 (Down's syndrome) was established only in 1959; procedures for detection of certain genetic defects in the unborn child are only a couple of years old (still not in general use); and the clinical delineation of several syndromes are based on sound data pertaining to the pattern of inheritance.

In light of the above and other recent advances in human genetics, several professionals have been invited to address some of the complex and vexing issues relating to these developments and to provide guidance and suggestions from the perspectives of a number of disciplines. Although most questions are directed mainly toward family or individual situations, the basic issues and their resolution have far-reaching implications and consequences for society. The nature of the aforementioned scientific advances and their impact on social policy and programs requires that society at large be better informed about these matters and be involved in the decision-making process.

In view of the recency of the aforementioned advances the decisions on some critical questions pertaining to research needs, diagnostic procedures, and ethical issues have been made primarily by the scientists and professionals involved. Understandably applications of innovative scientific and technologic procedures create new contexts for the balancing of conflicting societal and individual interests. The law's historic concern with the balancing of competing and at times conflicting interests is very much relevant to the aforementioned developments. Indeed, the resolution and handling of these complex and pressing social issues should involve the meeting of both scientific and legal thought.

In view of recent publicity in the news media about certain genetic findings, and, in some instances, the premature and erroneous aspects of such publicity, there appear to be pressures aimed at formulating social policy before clear and sound scientific data are available. For example, the use of genetic abnormalities such as 47,XXY and 47,XYY chromosome constitutions as the basis of an insanity defense against criminal charges, and genetic counseling for the parents when their unborn child happens to be a 45 chromosome D/D translocation carrier.

Genetic counseling is offered mainly by stating the risk figures in statistical terms to individual(s) who seek guidance because of their family background, or prior relationship (consanguinity) with the spouse, or an affected child already in the family. Recently, prenatal diagnoses of certain genetic conditions has been made possible by developments in somatic cell genetics and amniocentesis. It would appear that one of the primary concerns in seeking prenatal diagnosis pertains to the consideration of therapeutic abortion in the event of a genetically abnormal fetus. However, lacking adequate and meaningful information, the genetic counselor's own biases may well become important factors influencing the family's decision.

There are situations where individuals who are "carriers" of certain genetic defects wish to withhold such information from their family—sometimes even their spouse. Should such an individual later have an affected offspring, might the investigator (or physician) having prior knowledge of such risk, but maintaining confidentiality, be vulnerable to legal action brought by

Speakers and Topics

Digamber S. Borgaonkar (Johns Hopkins Medical School), *Recent Developments in Human Genetics—Their Usefulness and Impact on Society.*

Kurt Hirschhorn (Mount Sinai Medical School), *Recent Advances in Human Genetics and Their Significance in Clinical Practice.*

Discussant: Neil Macintyre (Case Western Reserve).

Saleem A. Shah (National Institute of Mental Health), *Some Biological Influences on Behavior and Their Implications for Problems of Social Deviance.*

Nathan Hershey (University of Pittsburgh), *Legal and Social Policy Issues Pertaining to Recent Developments in Genetics.*

Discussant: Elyce Zenoff Ferster (George Washington University).

other family members? This issue provides yet another example of the complex problems involved in careful balancing of individual and societal interests. It is evident therefore, that major stress needs to be placed on the careful obtaining of sound and sufficient information prior to making rather crucial personal as well as social decisions.

This symposium is designed to deal with issues pertaining to recent developments in human genetics from the standpoint of a geneticist, a medical geneticist, a behavioral scientist, and a legal scholar, in order to highlight a number of social implications. Some of the specific topics to be discussed by the speakers will include availability of techniques to detect "carrier individuals" of "abnormal" genetic material in liveborn and fetal populations; mass genetic screening to obtain "sound" incidence and prevalence data; the importance and implications of genetic screening as a diagnostic tool; genetic factors associated with socially deviant behavior and the influence of societal norms and values in the definition, labeling, and handling of social deviance; administrative, legal, and social policy considerations in conducting genetic research involving human subjects (issues of "informed consent" and confidentiality); and the need for balancing individual rights and concerns with those pertaining to broad societal interests and public benefits in the formulation of social policy.

The symposium is being sponsored by the American Genetic Association, one of the oldest genetics organizations in this country and will be held under Section N-Medical Sciences.

DIGAMBER S. BORGAONKAR
Division of Medical Genetics, Johns Hopkins University School of Medicine, Baltimore, Maryland 21205

SALEEM A. SHAH
Center for Studies of Crime and Delinquency, National Institute of Mental Health, Chevy Chase, Maryland 20014

Genetic Diseases and the Quality of Life

AAAS Symposium

29 December 1970

Chicago

Arranged by Albert A. Dietz (Biochemist, Veterans Administration Hospital, Hines, Illinois. Associate Professor of Biochemistry, Loyola University, Stritch School of Medicine, Haywood, Illinois) and Donald T. Forman (Director of Biochemistry, Evanston Hospital, Evanston, Illinois. Associate Professor of Biochemistry, Northwestern University Medical School, Chicago, Illinois).

The study of the biochemical basis of genetic disease received its initial impetus from the papers of Garrod, published some 60 years ago. Subsequent work led to the identification of many types of biochemical abnormalities, their mode of inheritance, and many of the specific reactions involved. Recent work has placed more emphasis on the prediction of the development of genetic diseases with their eradication as a goal. Since not all genetic disease can be completely controlled, a better understanding of the nature of the defects and how they may be influenced will lead to techniques for the improvement in the quality of the lives of the persons who may be affected.

Topics and Speakers

Tissue Culture for Identification of Hereditary Defects, David Y. Hsia (Professor and Chairman, Department of Pediatrics, and Professor of Biochemistry, Loyola University, Stritch School of Medicine, Haywood, Illinois).

Hereditary Variants of Serum Cholinesterase, Herbert M. Rubinstein (Chief of Rheumatology, Veterans Administration Hospital, Hines, Illinois, and Professor of Medicine, Loyola University, Stritch School of Medicine, Haywood, Illinois).

Genetic Abnormalities of the Mucopolysaccharides, Albert Dorfman (Professor and Chairman, Department of Pediatrics, and Director, Joseph P. Kennedy Mental Retardation Center, University of Chicago, Chicago, Illinois).

Genetic Abnormalities of the Red Cell Enzymes, Henri Frischer (Assistant Professor of Medicine, University of Chicago, Chicago, Illinois).

Program Notes about the 1970 AAAS Annual Meeting appear in the 25 September issue of Science. *Reports on symposia appear in the following issues: 28 August, "Human Behavior and Its Control"; 4 September, "Land-Use Problems in Illinois"; 11 September, "Aleutian Ecosystem"; 18 September, "Reducing the Environmental Impact of Population Growth"; 2 October, "Critical Issues in Research Related to Disadvantaged Children; and 9 October, "Women in Science." Forms for housing and registration appear in the 25 September issue; tour information appears in the 2 October issue.*