This bibliography is a selective compilation of books and articles which focus on the psychological and social issues of applied human genetics. It is centered in particular around problems, issues, and discussions of genetic counseling, the primary mechanism by which human genetics has been applied to date. It includes those entries which, on the basis of information conveyed in the title, appear to be primarily concerned with social and psychological issues of applied human genetics in general, and genetic counseling in particular. The articles and books are arranged according to subject matter, usually on the basis of information given in the title, occasionally on the basis of bibliographical source subject-heading. Division of subjects was determined primarily by the number of articles that seemed to have a common area of concern. This bibliography should be especially useful to social and behavioral scientists conducting research in this area, to social workers, to genetic counselors interested in the problems and perspectives of practicing the art and science of genetic counseling, and to other allied health professionals engaged in applying human genetics. The publication of this bibliography was sponsored by the Fogarty International Center of the National Institutes of Health because of its interest in the social and ethical aspects of research in human genetics and in the application of these research advances to medical care. (Author/JM)
FOGARTY INTERNATIONAL CENTER
SOCIAL AND PSYCHOLOGICAL ASPECTS OF APPLIED HUMAN GENETICS:
A BIBLIOGRAPHY
Stone House—nucleus of international health activities
Fogarty International Center
National Institutes of Health
Bethesda, Maryland
FOGARTY INTERNATIONAL CENTER

SOCIAL AND PSYCHOLOGICAL ASPECTS
OF APPLIED HUMAN GENETICS:
A BIBLIOGRAPHY

by
Professor James R. Sorenson
Princeton University

DHEW Publication No. (NIH) 73-412
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INTRODUCTION

Within the past few decades, discoveries in basic science and developments in medical technology have significantly increased the role of human genetics in medical practice and daily life. Discovery of the genetic basis of an increasing array of bodily and mental disorders, as well as the development of a technology permitting in vitro analysis of certain aspects of fetal bio-genetic health, are rapidly altering man's classically passive orientation to his genetic endowment. The development of mass screening for carriers of heritable derangements enables genetic and chromosomal health considerations to become important factors in mate selection and determination of the reproductive future of a couple. Even more frequently, procedures such as amniocentesis are permitting families to consciously determine, for the first time, the outcome of pregnancy on the basis of fetal, genetic and chromosomal characteristics.

Such developments raise an array of psychological and social issues for both the professionals administering these advances as well as for individuals with genetic problems. Our understanding of the meaning and significance of genetic disease for individuals and society and the problems encountered in the professional delivery of such knowledge is very meager compared to the battery of scientific and technical knowledge permitting increased control of genetic disease. For the wisest and most humane utilization of human genetics, it seems mandatory that we begin to explore the social and psychological aspects of applied human genetics as a means to develop an understanding of these issues commensurate with our understanding of the scientific-technical aspects of applied genetics.

For these reasons we have prepared this bibliography on the social aspects of applied human genetics. It should be especially useful to social and behavioral scientists conducting research in this area, to social workers, to genetic counselors interested in the problems and perspectives of practicing the art and science of genetic counseling, and to other allied health professionals engaged in applying human genetics.

This bibliography is a selective compilation of books and articles which focus on the psychological and social issues of applied human genetics. It is centered in particular around problems, issues, and discussions of genetic counseling, the primary mechanism by which human genetics has been applied to date. The bibliography is not exhaustive in its coverage, as it does not try to include those books and articles which deal with the ethical or prescriptive areas of human genetics. Rather, it includes those entries which, on the basis of information conveyed in the title, appear to be primarily concerned with social and
psychological issues of applied human genetics in general, and genetic counseling in particular.

Entries observe the following format: within a subject area, articles are listed first, alphabetically; then books are listed alphabetically. The title of a journal is followed by the number of the volume and the page numbers of its location. The number of the issue is generally omitted.

The articles and books are arranged according to subject matter, usually on the basis of information given in the title, occasionally on the basis of bibliographical source subject-heading. Division of subjects was determined primarily by the number of articles that seemed to have a common area of concern.

Bibliographical reference works searched for this bibliography include the following:

- American Book Publishing Record through 1971
- Books in Print through 1971
- British Books through 1971
- Cumulative Book Index through 1971
- Excerpta Medica Section 22 (Human Genetics) through 1971
- Forthcoming Books through 1971
- Genetic Abstracts through 1971, except for June-October, 1971
- Index Medicus through 1971
- Kerker, Ann E., Biological and Biomedical Resource Literature, Lafayette, Ind., Purdue Univ. Press, 1968.
- Library of Congress Catalogue through 1971
- National Union Catalogue through 1971
- Reader's Guide to Periodical Literature through 1971
- Whitaker Cumulative Book List through May, 1970
- Princeton University Library Catalogue through March, 1972

This bibliography was compiled as part of a research project at Princeton University on the social aspects of applied human genetics and was directed by Professor James R. Sorenson. The funding for the bibliographic work was provided by the Russell Sage Foundation, 230 Park Avenue, New York City, New York. Assistance in the compilation was provided by Katherine Courant, Nancy Sorenson, and Camille Yinger.

The publication of this bibliography was sponsored by the Fogarty International Center of the National Institutes of Health because of its interest in the social and ethical aspects of research in human genetics and in the application of these research advances to medical care.
1. Genetic Counseling and its Delivery

a. Genetic Counseling


Cameron, R.G., "Genetic Counselling," Bulletin of the Post-Graduate Committee in Medicine, University of Sydney, 19: 165-70, September, 1963.


Fraser, F. Clarke, "Genetic Counseling," Hospital Practice, 49-56, January, 1971.


Lamy, M., "Special Considerations in Regard to Genetic Consultations," Monatsschrift fur Kinderheilkunde, 119: 157-8, 1971 (German).


Mohr, Jan, "Genetic Counseling," Proceedings of the Third International Congress of Human Genetics, 37-43.


b. Psychological Aspects of Counseling


2. The Technical Knowledge Basic to Genetic Counseling

   a. Antenatal Diagnosis


American Society of Human Genetics, Committee on Social Issues, Symposium on Intrauterine Diagnosis, National Foundation, 1971.


b. Carrier Detection (Heterozygosity)


c. Cytogenetics


d. Diagnosis


Diagnostic Tests for Genetic Diseases Available Through the Genetic
Counseling and Treatment Network, New York: National Genetics
Foundation, Inc.

Goodman, R.M., et al., The Face in Genetic Disorders, St. Louis:

Harris, Maureen, ed., Early Diagnosis of Human Genetic Defects:
Scientific and Ethical Considerations, Bethesda, Md.: National
Institutes of Health, 1970.

Priest, J.H., Human Cell Culture in Diagnosis of Disease, Springfield,
Ill.: C C Thomas, 1971.

Stevenson, Ian, The Diagnostic Interview (2cd edition), New York:
Medical Department, Harper and Row, 1971.

e. Inheritance Patterns (and Pedigree Determination)

Falconer, D.S., "The Inheritance of Liability to Certain Diseases
Estimated from the Incidence Among Relatives," Annals of Human
Genetics, 29: 51-76.

"Family Pedigree--Will Be Used to Help Congenitally Disabled?", Journal

Fraser, F.C., "Taking the Family History," American Journal of Medicine

Haberlandt, W.F., "The Genetic Questionnaire: Familial Suicide and
Genetic Counseling," Journal de Genetique Humaine, 17: 179-84,
May, 1969.

Hecht, F., "Family Chromosome Studies," Journal of Pediatrics, 78:
735-6, April, 1971.

Jolly, E., et al., "Experiences of Public Health Nurses in Obraining

Motulsky, A.G., et.al., "Consanguinity and Marriage," Practitioner,

Mayo, O., "The Use of Linkage in Genetic Counseling," Human Heredity,

Roberts, D.F., "Consanguineous Marriages and Calculation of the

Roberts, J.A.F., "Multifactorial inheritance in Relation to Normal and


f. Risk Determination


g. Therapeutic Abortion


Sanders, M.K., "Right Not to Be Born: Refusal to Grant Therapeutic Abortion in Case of Rubella Baby," Harpers, 240: 92-9, April, 1970.


Desnick, R.J., et al., Medical and Genetic Indications for Therapeutic Abortion, Minneapolis: University of Minnesota, 1969.


h. Therapy (Treatment, Training, Diet, etc.)


i. Twin Studies


3. Genetic Diseases and Disorders

a. General Articles


Biass-Ducroux, F., et al., Glossary of Genetics in English, French, Spanish, Italian, German and Russian, Amsterdam; Elsevier, 1970.


Montagu, Ashley, Genetic Mechanism in Human Disease, Springfield, Ill.: C C Thomas, 1970.


b. Congenital Anomalies


c. Chromosomal Aberrations


d. Prenatal Influences (Mutagenic Drugs and Chemicals, Infection, etc.)


e. Blood Disorders


f. Down's Syndrome


Braga, H., "Mongolism (Trisomy 21 or 22) an Infectious Disease?", Pediatra Pratica (S. Paulo), 39: 541-4, 1968.


Pernod, J., et al., "Genetic Counseling in Mongolism (Trisomy 21)," Lyon Medical, 225: 41-8, January 10, 1971, (French)


8. Hearing Disorders


h. Heart Disease


1. Huntington's Disease


j. Mental Retardation


Lisker, R., "Genetic Counseling in Mental Retardation," Gaceta Medica de Mexico, 101: 453-6, April, 1971, (Spanish).


k. Metabolic Disorders


1. Neurological Disorders


m. **Phenylketonuria**


n. **Schizophrenia**


### Sickle-Cell Anemia


### Vision Disorders


q. Other Disorders


4. Medical Genetics and Medical Specialties

a. Medical Genetics


Barigozzi, C., "General Genetics, Human Genetics, and Medical Genetics," Minerva Medica.


b. Genetics and Obstetrics/Gynecology


Gomez, F., "Genetic Factors Involving Human Growth and Development," Gaceta Medica de Mexico, 100: 325-6, April, 1970 (Spanish).


c. Genetics and Pediatrics


d. Genetics and Psychology


e. Genetics and Public Health

Anderson, Odin, "The Utilization of Health Services," *Handbook of Medical Sociology*, 349-68.


Sanders, Irwin, "Public Health in the Community," Handbook of Medical Sociology, 369-96.


f. Medical Education


Berg, K., "How is Education in Medical Genetics Pursued today in the Scandinavian Countries?", *Nordisk Medicin*, 85: 531-5, April 29, 1971 (Norwegian).


De La Chapelle, A., "Genetics in Medical Studies. The Situation in
Finland," Nordisk Medicin, 85: 528-9, April 29, 1971 (Swedish).

Gang, S., et al., "Social Worker and General Practitioner. An Experi-

cation of the Private Practitioner," New York State Journal of
Medicine, 61: 3779-82, November 15, 1961.

Gustavsson, K.H., "Education in Medical Genetics in Sweden," Nordisk
Medicin, 85: 535, April 29, 1971 (Swedish).

Harvald, B., "Volume and Development of Clinical Education in Genetics,"
Nordisk Medicin, 85: 529-31, April 29, 1971 (Danish).

Herndon, C.N., "Genetics in Medical Education," Journal of Medical

Herndon, C.N., "Genetics in the Medical School Curriculum," American

Herndon, C.N., "Human Genetics and Medical Education," Journal of

Houle, C.O., "To Learn the Future," Medical Clinics of North America,

Lindsten, J., "Genetics in Medical Studies," Nordisk Medicin, 85:
527-8, April 29, 1971.

McEwen, C., "The Importance of Human Genetics in Medical Education,"

Macklin, M.T., "Should the Teaching of Genetics as Applied to Medicine
Have a Place in the Medical Curriculum?", Journal of the Association
of American Colleges, 7: 368-373.

Macklin, M.T., "Teaching Medical Genetics; Essential Part of Medical
Curriculum from Standpoint of Prevention," Journal of the Association
of American Medical Colleges, 8: 291-304, September, 1933.

of the Association of American Medical Colleges, 21: 351-3.

Stetten, DeWitt, Jr., "Projected Changes in the Medical School

Therkelsen, A.J., "Teaching and Special Education in Medical Genetics,"

Tobias, P.V., "Genetics in Medical Education," South African Medical


8. Genetics and Other Specialties


Reed, W.B., "Genetics in Dermatology," Hautarzt, 21: 8-16, 1970 (German).


5. Genetics and Other Subjects

a. Eugenics


Gyorgy, L., "Contrasting Interests of the Individual and the Population, the Solution of these Problems in Genetic Counseling," Orvosi Hetilap, 111: 3063-6, December 27, 1970 (Hungarian).


Seitz, L., "How Physicians and Gynecologists can Aid in Preventing Birth of Children with Hereditary Taints and Encouraging Birth of those with Normal Heredity as Provided in New German Law on Sterilization," *Deutsche Medizinische Wochenschrift*, 60: 546-9, April, 1934 (German).


b. Genetics and the Law


Cropwood Round-Table Conference, Institute of Criminology, 1969, Criminological Implications of Chromosome Abnormalities, University of Cambridge Institute of Criminology, 1969.


Simpson, C.K., Forensic Medicine, Baltimore, Maryland: Williams and Wilkins, 1969.


West, Donald James, ed., Criminological Implications of Chromosome Abnormalities, University of Cambridge Institute of Criminology, 1969.

c. Genetics and Philosophy


"Cardiac Replacement - Medical, Ethical, Psychological, and Economic Implications," report by Ad Hoc Task Force, National Heart Institute, October, 1969.


d. Genetics and Sociology


Bustamante, M.E., "Perspectives on Social Medicine II. Medicine and Sociology in its Interdisciplinary Aspects," Gaceta Medica de Mexico, 98: 576-9, May, 1968 (Spanish).


e. Genetic Engineering


Gustafson, James M., "Genetic Engineering and the Normative View of the Human," a paper, Yale University.


Handler, P., "Can Man Shape His Future?" Perspectives in Biology and Medicine, 14: 207-27, Winter, 1971.


Lederberg, J., "What Controls for Genetic Engineering?" Current, 121: 48-51, September, 190.


f. Human Genetics


*Genetic Control of Differentiation*, Upton, New York: Brookhaven National Laboratory, 1965.


g. Population Genetics


Eckland, Bruce K., "Trends in the Direction and Intensity of Natural Selection, With Special Reference to the Effects of Destratification and Equal Opportunity upon Current Birth Rates," a paper, University of North Carolina Department of Sociology.


Association for the Aid of Crippled Children, Genetics of Migrant and Isolate Populations, Baltimore: Williams and Wilkins, 1963.


