GENETIC DISEASE CONTROL

A Social Psychological Approach

FRED MASSARIK
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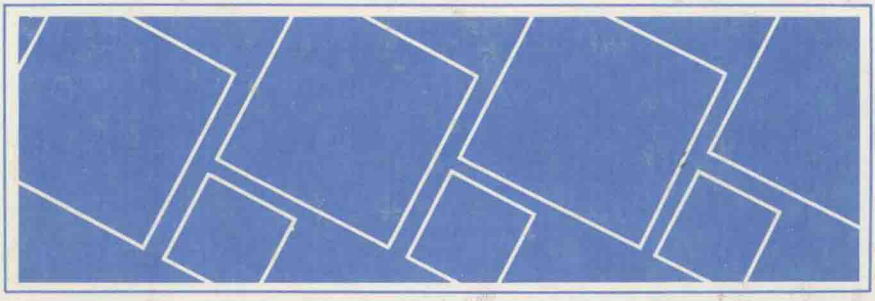

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Volume 116
SAGE LIBRARY OF
SOCIAL RESEARCH

SAGE PUBLICATIONS Beverly Hills London

For information address:

SAGE Publications, Inc.
275 South Beverly Drive
Beverly Hills, California 90212

SAGE Publications Ltd
28 Banner Street
London EC1Y 8QE, England

Printed in the United States of America

Library of Congress Cataloging in Publication Data

Massarik, Fred.
Genetic disease control.

(Sage library of social research)
Bibliography: p.
Includes index.
1. Genetic counseling–Social aspects.
2. Genetic counseling–Psychological aspects.
I. Kaback, Michael M., joint author. II. Title.
[DNLM: 1. Hereditary diseases–Prevention and control. 2. Genetics, Medical. QZ 50 M414g]
RB155.M35 362.1'96042 80-28219

ISBN 0-8039-1054-1
ISBN 0-8039-1055-X (pbk.)

FIRST PRINTING

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PREFACE

Not every Man or Woman was design'd
To propogate and multiply their kind;

Forbid we rightly the Deform's and Foul
To clothe with ill-shap'd Limbs the heav'nly Soul?

Has not the Poet's Song divinely told
Of Births detested in the days of old?

—Callipaedia, or The Art of
Getting Beautiful Children,
Claudius Quillet (N. Rowe, trans.)
London: A. Bell et al., 1720, p. 21.

Since ancient days, folkways and myths, as well as prescriptive medical lore, have provided cautionary guides to the process of conception and birth. Rooted in the world views current in a given place and age, intertwined with prevailing social values no less than with knowledge (actual or presumed) of medical reality, homilies like the following have had their day:

- A child conceived on the night of the new moon may well come to life grossly deformed.
- Offspring of marriages of Black and white cannot find grace in the eye of God.
- The bleeding disease (hemophilia) is strictly a disease of royalty.

Whether "old wives' tales," quasi-religious injunctions, or commonly held "folk wisdom," some notions of genetic process have been part and parcel of virtually every culture and era of social thought. Revealed systematically in the works of anthropology or discussed informally among pregnant women or marriage partners, what is indeed known about genetics—on the basis of systematic research—and what is commonly assumed to be factual are often dramatically different matters. Even among the presumably well-educated, lack of information and misinformation abound concerning matters of heredity and its consequences.

This is not a "medical" book, at least not in a narrow sense. Rather, it addresses some issues relating to genetics and medicine in the perspectives of the social and behavioral sciences. It provides viewpoints, especially bearing on impact of new genetic disease control methods and research data, as prototype, reporting psychosocial research conducted in connection with the California Tay-Sachs Disease Prevention Program.

In this context, we consider how a specified population perceives genetic disease and its consequences, levels of genetics knowledge, and attitudes toward the diagnosis, treatment, cure, and prevention of genetic illness. Additionally, we examine what these people (primarily Jewish, married, and of reproductive age) think ought to be done in terms of genetics testing and screening, and what they are prepared to do, by way of specific intervention such as abortion, in instances in which a genetic disorder is diagnosed *in utero.* Finally, we consider the hypothesis of "stigma," by studying the attitudes and feelings of couples in which one member has been identified as a carrier of Tay-Sachs disease.

The principal empirical data reported derive primarily from a series of sociopsychological research projects conducted by the University of California, Los Angeles, at its Division of Medical Genetics (Harbor-UCLA Medical Center), by UCLA Behavioral and Organizational Science Faculty

(Graduate School of Management), and by an independent consulting group (Behavioral Science Research Associates), 1975-1980. These empirical data on primarily Jewish populations are presented as both substantive and illustrative, with implications for comparable needed research in other populations and in other settings.

One may ask, "Is there any reason why the topic of genetic disease control is any more relevant now than it had been in the past?" The first chapter addresses this issue.

Chapter 1

NEW VISTAS IN GENETIC DISEASE CONTROL

The once Inevitable no longer is inevitable.

—Anonymous

In times past, beyond adherence to popular prescription for simplistic home remedy, there seemed to be little that people could—or were prepared to—do about their hereditary legacy. Even fairly systematic understanding of certain genetic regularities (such as Mendelian laws, rudimentary knowledge of genes and chromosomes) widely appeared as irrelevant curiosity or as foreordained circumstance, but hardly as something that could be affected in direct manner. "Having that deformed child is a burden that just must be borne." "There will always be those who are mentally retarded—this simply is an affliction that the parents, and society, must endure." And at one time, more drastic views of eugenics, the tragic views of "pure-bred" Aryan master races, the "brave new worlds," these and other ideologies illustrate reactions at the other extreme: the proposed creation of vast systems of centralized effort—in the vein of some kind of giant "societal engineering"—that would alter a people's genetic character, and lead to some imagined never-never land of total genetic health.

At another point in the spectrum, there were those who looked beyond the issue of genetics and denied all relevance of any inherent predisposition. Most articulately, the behaviorists in the mode of J. B. Watson, placed all their bets—at least as far as personality and its social correlates is concerned—with the impact of "environment." These theoreticians and their grass-roots adherents gave no consideration to the realm of heredity, often simply by not addressing the specific problem (e.g., mental retardation), or by seeking explanations which would derive *totally* from external causes. In some sense, psychoanalysis, in spite of its medical roots, further supported this line of reasoning. It did so by the preponderant emphasis on the impact of the childhood environment, and by its stress on the functional development of psychodynamic health or malfunction. All this, we may note parenthetically, in spite of the evidence of Freud's reluctance to his dying day to abandon his view of himself as a "neurologist," as a physician concerned with the specific functioning of the nervous system's physical reality.

We thus find a strangely mottled background of psychological thought, variously recast in the popular psyche, against which quite recent medical developments bearing on the potential control of genetic disease have made their appearance, including an often-confused acceptance of the presumably inevitable, the idealized and often misguided adulation of "hypothetically pure" and "uniformly healthy" artifactual populations, and the broad deemphasis of genetic cause, based on behavioristic or psychoanalytic doctrine. Together with simple lack of concern, the popular perspective on genetics is not encouraging.

In spite of this melancholy background of popularly held belief, significant change may be in the offing. A rather different contribution to popular thought relating to the role of genetics has recently emerged from fundamental advances in the basic theory of genetics itself. The discovery and unfolding knowledge of deoxyribonucleic acid (DNA), the

chemical substance of genetic material as drawn to wide public attention by books such as *The Double Helix,* the notion that forms of life may be created or genetically manipulated, as by experimentation with recombinant DNA, and the present-day folklore relating to the possibilities of cloning (the exact reproduction of identical "persons" or other life forms) have brought renewed interest to, and confusion in, people's ideas about genetics and its uses. Genetics has been publicly "rediscovered," but once again scientific progress has come to be reflected in the minds of many by a melange of hope and anxiety, knowledge and misunderstanding.

At any rate, there are some indications that the interplay of medical advances and changes in personal and social values may be setting the stage for a realistic application of new medical technology for far-ranging human benefit. The medical technology alone, of course, cannot do the job unless the relevant people beyond the confines of the laboratory, are prepared to avail themselves of the new and rapidly evolving methods and their potential advantages.

Every human being carries between five and eight deleterious genes. These recessive genes have no negative consequences for the health of the carrier. However, being a carrier predisposes, under certain conditions, to development of genetic anomaly at certain risk levels in the carrier's fetus and child.

In this context, we must now turn to a more explicit description of certain major medical technologies that have become available in the past decade, in the purposeful control of genetic disease.

Two interrelated lines of development are observed: (a) an enhanced large-scale capability for screening—e.g., for determination of carrier status; (b) a vastly improved technology for the prenatal diagnosis of genetic anomalies, making possible early in fetal development consideration of alternative options, such as therapeutic abortion.

The availability of technology to provide often exceedingly reliable and valid information regarding the genetic health or dysfunction of the fetus makes it worthwhile to ascertain the potential parents' possible carrier status, as it may place a fetus at risk with respect to a specific genetic ailment. The issue of what is "worthwhile" may be addressed variously in statistical and in human terms–that is, in terms of the probabilities that being or not being a carrier may contribute to birth of defective offspring, and in terms of the benefits and/or liabilities (including possible stigma or anxiety) may affect persons identified as carriers.

Beyond these considerations, there is the issue of "knowing the worst"–the instance in which little or nothing can be done in specific medical terms, except perhaps by mild psychological or medical amelioration, to avoid ultimate tragedy. An instance of the latter appears, for example, under conditions of positive diagnosis of a fatal genetic illness *in utero* in which the parents choose not to, or cannot, avail themselves of therapeutic abortion; here, based on present medical knowledge and experience, the afflicted child, may well die at an early age, as in Tay-Sachs disease, though various temporary and supportive measures can be the only feasible interventions in the face of an unavoidable and tragic reality.

As relates to screening for carrier status and prenatal diagnosis, a basic schema of alternatives appears in Figure 1. Schematically, we may consider a chain of events which begins with the procedures identified in (a), above, denoted as screening, proceeding toward a variety of possible subsequent interventions–e.g. (b), denoted as prenatal diagnosis–and subsequent potential remedial actions.

The schema represents an oversimplification; the needed rectification is indeed our central concern. One must examine the *psychosocial context* in which the chain of events shown in Figure 1 unfolds. This context includes the attitude base motivating or demotivating compliance with suggested

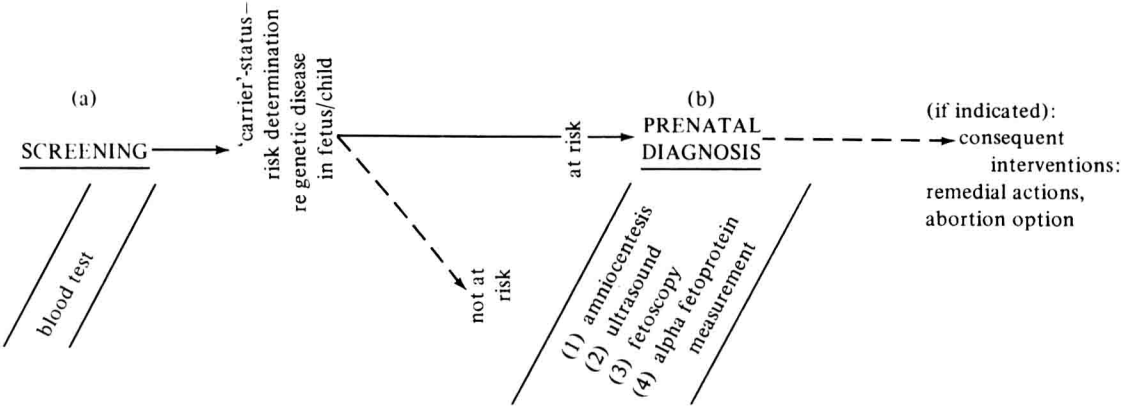

Figure 1: Schema: The Genetic Disease Control Process

screening for various genetic disorders, responses to the screening process itself and to its outcomes (i.e., carrier/non-carrier status), attitudes relating to mating and conception under various conditions of risk, views of prenatal diagnosis, response to positive or negative diagnostic findings, and eventually readiness to seek or to reject therapeutic abortion.

It is evident that a complex decision tree confronts the potential user of the evolving medical technologies, all imbedded in a complex framework of human response. This framework determines the ultimate personal and social effectiveness of the available technical means for genetic disease control.

This psychosocial context poses the emergent question, "How will this new medical/genetic technology be received by the public(s) that it is intended to benefit?" In light of correct and incorrect perceptions of the nature of genetics and its consequences, and within a pattern of changing social values, what is the probable impact of the varied linkage "screening⟶prenatal diagnosis⟶consequent intervention" on control of genetically caused illness, or well-being?

SCREENING

A substantial literature is available describing the concept and process of screening. For present purpose, a brief summary of typical screening efforts must suffice. In most instances, we shall report particularly on the California Tay-Sachs Disease Prevention Program as an example of screening for carrier status among adults. Other genetic screening programs seek to determine possible presence of a genetic illness in the person affected, especially among the newborn. Perhaps best known are screening programs for phenylketonuria (PKU) and sickle cell anemia.

Some definitions relating to these programs may be briefly cited (Committee for the Study of Inborn Errors of Metabolism, 1975: 9).