# GENETIC ISSUES IN PEDIATRIC AND OBSTETRIC PRACTICE

KABACK, Editor

# Genetic Issues in Pediatric and Obstetric Practice

*Edited by*

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YEAR BOOK MEDICAL PUBLISHERS, INC.
CHICAGO • LONDON

**Library of Congress Cataloging in Publication Data**
Main entry under title: Genetic issues in pediatric and obstetric practice.
Includes index.
1. Children——Diseases——Genetic aspects.
2. Abnormalities, Human——Genetic aspects.
3. Medical genetics. I. Kaback, Michael M.
[DNLM: 1. Hereditary diseases. 2. Abnormalities.
OZ 50 G325]
RJ47.3.G46 616′ .042 81-4943
ISBN 0-8151-4952-2 AACR2

# GENETIC ISSUES in PEDIATRIC and OBSTETRIC PRACTICE

THIS BOOK IS BASED ON A CONFERENCE SPONSORED BY
THE DEPARTMENT OF PEDIATRICS
UNIVERSITY OF CALIFORNIA AT LOS ANGELES (UCLA)
HARBOR-UCLA MEDICAL CENTER
and
THE INSTITUTE FOR PEDIATRIC SERVICE
OF THE JOHNSON & JOHNSON BABY PRODUCTS COMPANY
and
held at San Francisco, California

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*Deceased

# Dedication

To David W. Smith, M.D., our teacher, colleague, and friend. He was truly one of the pioneers in bringing genetic issues into pediatric and obstetric practice.

# Preface

A decade ago in the United States, fewer than 25% of the medical schools had any defined curriculum in human genetics. For this reason, most physicians in practice today have had little, if any, formal exposure to the precepts of medical genetics. This is particularly problematic since perhaps no other area of biomedical science has witnessed such an explosion of new technology over the past two decades. Dramatic advances in cytologic, biochemical, and molecular methodologies have led to vast new knowledge of the intermediary metabolism in man and, in turn, to the elucidation of the role of genetic factors in human disease. Moreover, these discoveries have resulted in the development and promulgation of a variety of new "genetic health" programs aimed at a wide spectrum of individuals. One need look no further than to the dramatic advances which have been made in newborn screening for inborn metabolic errors and thyroid deficiency—the dramatic reduction in rhesus hemolytic disease of the newborn achieved through blood typing of pregnant women and the appropriate use of rhesus immunoglobulin, the exponential growth of prenatal diagnosis through amniocentesis—and to the development of carrier detection programs for the prevention of such hereditary disorders as Tay-Sachs disease, sickle cell anemia, and β-thalassemia.

We are still at the beginning of the development and application of much of this new knowledge. Not only have there been dramatic advances in technology but, with the extensive media and press coverage given these developments, an explosion in public awareness has also resulted. Concomitantly, the consumer is assuming a more active role in the decision-making process with regard to many health ser-

vices. Dramatic shifts in public attitudes toward family size, life quality, birth control, and abortion have created important and dynamic interfaces with these scientific advances. As such, new and critical social, ethical, and legal questions for physicians and for society at large are being recognized.

Optimal implementation of much of this new technology will require the active participation of the primary care provider. The family practitioner, the pediatrician, and the obstetrician-gynecologist are expected to become progressively more involved in the delivery of medical genetic services. Thus, these symposium proceedings provide the physician with a critical update on many new and important topics relevant to an expanded general practice.

The present text is not meant to be a comprehensive volume on medical genetics. Rather, it is a compendium of selected topics, each addressed by a recognized authority, with emphasis on areas of particular interest to the family physician, pediatrician, and obstetric specialist. The volume is divided into five sections. Section I (chaps. 1 through 7) examines basic genetic principles in the light of hereditary disease and congenital defects. Section II (chaps. 8 through 17) addresses etiologic factors and genetic considerations in the care of the abnormal or sick newborn, infant, and young child. The third section (chaps. 18 through 22) deals primarily with the most recent developments in genetic medicine. Chapters 23 through 28 (section IV) examine important treatment and management strategies which much of our recent knowledge has led to. Last, chapter 29 (section V) addresses selected medicolegal, ethical, and social issues associated with these dramatic advances in medical science.

I wish to express my deep appreciation to all of my colleagues for their lucid and stimulating presentations. A special word of gratitude is extended to Steven Sawchuck, M.D., his staff, and the Board of the Institute for Pediatric Service. Their encouragement, support, and hard work were instrumental in enabling us to convene the symposium and produce this volume.

Michael M. Kaback, M.D.

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Section I

# Hereditary Diseases and Congenital Defects: Genetic Perspectives

# 1

## Genetic Factors in Human Disease

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Although medical texts often exceed 2,000 pages in length, few devote even a few lines to a definition of disease. This may be because the authors recognize the philosophical traps that lurk in such exercises, or it may be that they simply assume that the definition is self-evident. In fact, history reports two concepts of disease that have been argued since antiquity.[1-4] The first, which has been called ontological* or Platonic, accords disease the status of specific entity. It is something visited at random upon a healthy person, a result of a well-defined, usually external, cause, and is expressed in a more or less constant form. Each case is exemplary of the universal, which exists apart from the persons who have the disease. What we generally call "genetic" diseases fit very well into this mold since the "deleterious" genes or chromosomes are perceived to be agents of disease and to differ from bacteria, say, only in that they are internal rather than external.

Proponents of the second concept, commonly called physiologic or Hippocratic, proclaim that there are no diseases, only sick people. Disease is perceived not as entity, but as quantitative variation, a statistical deviation from normal.[3] The causes of illness are not themselves harmful but become so in the presence of "limitations in the inventory of adaptive resources" of particular individuals or of the whole species.[4] The expressions of illnesses, then, are dictated by the

*Ontology is that branch of metaphysics that deals with the nature of reality.



0-8151-4952-2/81/000003-14$3.75