

COMPREHENSIVE PEDIATRICS

ROBERT L. SUMMITT

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PREFACE

In no field of medicine are the exciting discoveries in basic human biology having a greater impact on professional practice than in pediatrics. Today's student of pediatrics, whether a medical student, a resident in pediatrics or a related discipline, a practicing physician, or another health professional, is confronted and challenged by a rapidly growing and changing fund of information. Discoveries in the realm of molecular biology have made a tremendous impact on our ability to diagnose infectious diseases, acquired disorders of the immune system, and a broad spectrum of genetically determined conditions. The impact on genetic disease is being felt not only in the diagnosis of the affected patient, but also in the detection of late-manifesting conditions such as Huntington disease in the young asymptomatic heterozygote, in the in utero diagnosis of such conditions as cystic fibrosis and phenylketonuria, and in the detection of prospective parents who are heterozygous for recessive mutant genes and are thus at risk for affected offspring.

Similar discoveries in molecular biology have significantly advanced methods of treatment of such conditions as type I diabetes mellitus and malignant neoplasms. Advances in drug therapy of infectious diseases, cancer, and epilepsy are dramatically improving the prognosis for those conditions.

The above-mentioned advances have not only enhanced our ability to understand disease processes, to make accurate and exact diagnoses, and to undertake progressive therapeutic approaches; they also have significant implications for prevention. Historically pediatrics has contributed heavily to the principles and practice of preventive medicine. If we are to be responsible health care providers and educators for the twenty-first century, prevention will require greater emphasis than ever before. Prevention begins with advice and counsel given to couples prior to conception of offspring, extends

throughout pregnancy, and is of paramount importance in infancy and childhood. Professionals who provide health care to children and adolescents must assume greater responsibility for monitoring the nutrition of their patients because of its importance in growth and development. However, equally important is nutritional monitoring and modification in the prevention of adult-onset conditions that result from genetically determined variations in the metabolism of such nutrients as cholesterol.

Societal aspects of prevention require special emphasis if we are to stop the spread of such diseases as hepatitis and acquired immune deficiency syndrome (AIDS). We must be prepared to deal with such important issues as accidents, drug abuse, malnutrition, child physical and sexual abuse, adolescent pregnancy, and homicide. Further, we must be knowledgeable in and capable of managing children and adolescents at risk for the effects of the increasing stresses imposed by our society that lead to suicide. This emphasis on health promotion and disease prevention must extend to the time when the pediatrician relinquishes responsibility for the care of his or her patients to adult-oriented physicians. Increasingly the advances in molecular biology, biochemistry, and pharmacology will play a role in preventive pediatrics.

This book attempts to place an organized and appropriate focus on the pathophysiologic, preventive, diagnostic, and therapeutic aspects of child health. *Comprehensive Pediatrics* is built on the challenging legacy of six editions of the *Synopsis of Pediatrics* edited by James G. Hughes, M.D., Professor and Chairman Emeritus of the Department of Pediatrics, College of Medicine, University of Tennessee, Memphis, and in its sixth and last edition coedited by John F. Griffith, M.D., Professor and Chairman of the same department from 1976 to 1986. The challenge that the editor of and contributors to

this volume have undertaken is to build on that legacy by producing a book that addresses in a comprehensive manner those aspects of child and adolescent health described earlier. Many contributors to the *Synopsis of Pediatrics* have had major roles in the development of *Comprehensive Pediatrics*. Fresh approaches have been

taken on previously addressed subjects, and new topics are included. New contributors have added to the comprehensive approach that we have taken. We believe that we have brought to fruition an approach that truly represents *Comprehensive Pediatrics*.

Robert L. Summitt

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The Scope of Pediatrics

JAMES G. HUGHES

Pediatrics is the study of growth and development of the child from the genetic background and moment of conception through adolescence. It is the science and art of prevention, diagnosis, and treatment of the diseases of children from birth through adolescence, whether these disturbances be physical, mental, or emotional. Pediatrics is a profound concern with, and an abiding interest in, everything that contributes to the final product—healthy adults ready to take their places in life.

Pediatrics is the knowledge of genetics, for this governs the seed. It is the knowledge of the events of prenatal life, for this is the soil in which the seed grows. It is the knowledge of the newborn, the infant, and the young child, for this is the early growth period. And it is the continued scientific and empathetic supervision of the child until he achieves maturity.

The single most characteristic feature of pediatrics is that it deals with the growth and development of the child, comprising all those changes in size and form and in complexity of function that constitute growing up. It is this fact that distinguishes it as a specialty—one of the broadest and one of the most interesting.

Physicians dealing with children must be aware, for example, of the differences in physiologic maturity of newborn and young infants as compared with the older infant and younger child and to an even greater extent the older child or adolescent. There are differences for these various age levels in (1) function of various organ systems, (2) degree of immunity to diseases, (3) response to the effects of disease, (4) drug dosages and tolerance to drugs, (5) mental and motor ability, and (6) pattern of emotional response.

Thus pediatrics encompasses a fascinating and almost endless variation that distinguishes it from other specialties that focus only on certain systems or regions of the body or that are related entirely to adults, who are more or less of similar

size and maturity of functions. These differences between children and adults have been summed up in the statement, "The child is *not* a little man."

GENETICS

The pediatrician must maintain a lively interest in genetics. Although the mechanisms of heredity are working long before the pediatrician sees the newborn child, he or she is the first to observe deviations caused by defective genes. Thus congenital malformations fall within the province of the pediatrician, who may call on various other specialists to help in the management of such children.

Heredity also determines those subtle enzymatic abnormalities known as inborn errors of metabolism. These, too, are usually first detected by the pediatrician, who must know how to diagnose them and how to manage them in the global sense.

Finally, a sound knowledge of genetics is important in regard to the effects it has on general growth and development—knowledge that may explain body build, facial appearance, intellectual level, and many other facets of the child.

Interest in genetics is quickened by virtue of remarkable discoveries regarding chromosomes and their molecular structure. Phenomenal progress has occurred over the last two decades through the development of techniques that allow detailed study of chromosomes and their abnormalities, mapping of the human genome, the application of recombinant DNA techniques to the diagnosis of genetic disease and the application of knowledge of genetic engineering to the treatment of those diseases. In utero diagnosis of an increasing number of genetic defects by chromosome analysis of cells of fetal origin, by measurement of the contents of amniotic fluid, and by direct and indirect DNA analysis on fetal

cells provides knowledge and choices to parents formerly not possible. Some of these same techniques can identify those at risk for producing affected offspring well before a first pregnancy. They can also identify early in life those who will develop a genetic disease that has its first clinical manifestations in adulthood. Knowledge of genetic diseases and their causes has led to exciting approaches to curative treatment through gene replacement therapy. To what extent these explorations toward the earliest beginnings of life will someday afford a better understanding and possible control of various diseases constitutes an exciting challenge.

PRENATAL EVENTS

It is said that the Chinese believe a child to be 1 year of age at birth. Of course, it is not a year, but the major portion of a year of intrauterine growth and development that precedes the infant's birth. If heredity and intrauterine environment are normal, a normal baby may be born. However, just as heredity may determine abnormality, so also may adverse prenatal influences.

Some adverse prenatal factors that may distort intrauterine growth and development to cause death, deformity, premature delivery, or subsequent disability are (1) poor prenatal supervision, (2) maternal malnutrition, (3) acute or chronic maternal diseases (systemic or specifically affecting the reproductive organs), (4) maternal infections that permit organisms to cross the placenta and infect the fetus (usually viral agents such as that of rubella and less often bacterial or protozoal agents), (5) faulty implantation and hormonal imbalance, often leading to threatened abortion or miscarriage, (6) pelvic irradiation, (7) deleterious effects of certain drugs or toxic substances such as excessive alcohol intake or excessive smoking during pregnancy, and (8) rarely, trauma.

Knowledge of such factors is of great aid in evaluation of the sick newborn infant or the child who later shows evidence of brain damage that may have resulted from adverse prenatal influences.

Furthermore, if such factors act within the early months of gestation, growth and development may be so distorted that the resulting abnormalities may closely resemble or even be

clinically indistinguishable from genetically determined malformations.

LABOR AND DELIVERY

Since the infant may be profoundly affected by the vicissitudes of labor and delivery, knowledge of what transpired as the child was born is of great significance. Proper interpretation necessitates that the pediatrician know as many of the circumstances as possible, including the following:

1. Whether any one or combinations of the above high-risk factors occurred
2. Whether the infant was born in the home, in the hospital, or on the way to the hospital
3. The mother's condition (fever, foul vaginal discharge, etc.)
4. Whether the mother is primiparous or multiparous
5. Type of delivery (spontaneous, induced, or operative—including cesarean section or use of forceps)
6. Fetal presentation
7. Duration of the stages of labor
8. Relative intensity of labor
9. Whether cephalopelvic disproportion existed
10. Time interval between rupture of the membranes (spontaneous or induced) and birth of the infant
11. Drugs used for analgesia or anesthesia, doses, routes of administration, and timing with respect to delivery
12. Whether labor was induced and managed by an intravenous oxytocic agent
13. Whether any untoward event occurred, such as maternal hemorrhage or severe hypotension
14. The infant's birth weight, gestational age, and condition at the moment of birth, including Apgar scores
15. Whether resuscitation was required and, if so, methods employed
16. Condition of the infant when sent from the delivery room to the nursery

It is apparent that teamwork between the obstetrician, in recording pertinent information, and the pediatrician, in interpreting this information, is of great importance.

NEWBORN PERIOD

Patterned by genes, nurtured in the womb, and launched by labor and delivery, the infant