PRADER-WILLI SYNDROME

Edited by
Vanja A. Holm,
Stephen J. Sulzbacher,
and **Peggy L. Pipes**

Technical Editor
Michael J. Steffes

University Park Press
Baltimore

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Foreword

This report of a 1979 conference on Prader-Willi syndrome represents communication among a heterogeneous band of researchers with a joint interest in a puzzling and unusual disorder. The sophistication of the investigations within their several disciplines is a result of the dedication with which the authors have pursued the joint goals of understanding and intervention. To some extent they have succeeded; yet, the puzzle remains unsolved.

This volume presents up-to-date and comprehensive sets of findings that suggest that Prader-Willi syndrome can and should be identified very early in a child's life, and that with sufficient vigor and resolve its management can yield substantially positive results. The crippling effects of two cardinal symptoms, obesity and behavioral disorder, can be modified; life can be lengthened; cognitive deficits may even be reduced. The studies reported here can serve as guidelines for other clinical researchers who are willing to accept the demanding, unrelenting challenge of treating a most difficult and intractable disorder. I have seen the monthly clinic at the University of Washington's Child Development and Mental Retardation Center, and it is obvious that the professionals involved conduct both research and service there with the highest standards of the clinician-scientist, and with clarity, wisdom, patience, firmness, and good humor. To the authors of these papers, both the University of Washington group and their guests, my hat is off.

Nancy M. Robinson, Ph.D.

Preface

Prader-Willi (P-W) syndrome is a disorder characterized by poor muscle tone and feeding problems in infancy, and by obesity, short stature, disorders in sexual development, and behavioral and cognitive disabilities throughout the remainder of life. These medical and behavioral problems result in very complex needs that require the services of professionals from many fields, including medicine, nutrition, psychology, education, social work, occupational and physical therapy, and speech pathology. Unfortunately, the information available to help professionals in planning for and providing these services has in most cases been scattered and limited in extent. This book summarizes research and clinical experience from all these fields. All the chapters contain new information, in some cases making important revisions in the picture of this syndrome and in the needs it creates. Knowledge about this syndrome continues to grow rapidly. Some current research indicates that a high proportion of cases of P-W syndrome might be caused by the absence of minute portions of chromosome 15. If these exciting findings are confirmed, only parts of the discussions of etiology and diagnosis might be of historical interest by the time this book is in print. However, the great majority of the information is still applicable. Since previous publications addressed themselves for the most part to medical or nutritional problems, readers will find the information on behavioral problems especially pertinent.

The bulk of the book is divided into four sections dealing with etiology and diagnosis, obesity management, behavioral and social aspects, and medical aspects of the syndrome. One of the functions of the first chapter in each section is to provide an introduction to the issues dealt with in that section. Chapter 1, which deals with the historical perspective, contains background information relevant to the whole book. An annotated bibliography is included to aid those who wish to read further.

Although this book is intended primarily for professionals, we have made an attempt to reach a wider audience, especially parents of P-W syndrome children and caregivers in group homes and other community facilities. We think that they will find much of the information useful.

Most of the chapters were presented at a conference on P-W syndrome sponsored by the University of Washington and funded by the U.S. Health Services Administration, Bureau of Community Health Services, Office of Maternal and Child Health (Grant no. MCT-000986-01-0). The conference and, to an even greater extent, this book represent a synthesis of work relating to P-W syndrome by clinical researchers from throughout the United States and Canada.

Although each of the editors has been involved in all parts of the book, Vanja Holm has been primarily responsible for the sections on diagnosis and medical management, Stephen Sulzbacher for the behavioral section, and Peggy Pipes for the obesity management section.

We are grateful to the Office of Maternal and Child Health for providing the funding for the conference out of which this book grew and for many of the expenses of editing and producing it as well. Their support was essential.

Credit is also due the Child Development and Mental Retardation Center of the University of Washington and its director, Dr. Irvin Emanuel, for providing the clinical facilities and academic working atmosphere for much of the pioneering work reported here. We also appreciate the efforts of the personnel, particularly Jean Reeves, involved in running the Prader-Willi Syndrome Clinic at the center.

We would like to thank the contributors of the individual chapters, who often took time from very busy clinic schedules to complete them. For help and encour-

agement with this project in its initial stages we are grateful to Dr. Gerald LaVeck. Susan Chapman made a major contribution to the success of the conference out of which this book grew. Finally, we would like to express our appreciation to Ken Westby for doing an excellent job of typing the manuscript.

We hope that our efforts and those of the contributors lead to further improvements in the diagnosis and treatment of Prader-Willi syndrome. This is certainly our paramount wish in producing this book.

This book is dedicated to the memory of
Daniel Neason (1966–1980),
one of our earliest patients
for whom we had high hopes,
and
Paula Carman (1945–1980),
a beloved colleague whom we miss.

Contents

Contributors viii
Foreword
Nancy M. Robinson xiii
Preface xv

chapter 1
The Prader-Willi Syndrome Historical Perspective
Michael J. Steffes, Vanja A. Holm, and Stephen Sulzbacher 1

chapter 2
Federal Programs for the Developmentally Disabled
Gerald D. LaVeck 17

part I **ETIOLOGY AND DIAGNOSIS**

chapter 3
The Diagnosis of Prader-Willi Syndrome
Vanja A. Holm 27

chapter 4
A View of the Etiology and Pathogenesis of Prader-Willi Syndrome
James W. Hanson 45

chapter 5
Diagnosis and Therapy in the First Phase of Prader-Willi Syndrome
Hans Zellweger 55

chapter 6
Clinical Experience with 23 Cases of Prader-Willi Syndrome
Henry G. Dunn, W. Jun Tze, Robert M. Alisharan, and Michael Schulzer 69

part II **OBESITY MANAGEMENT**

chapter 7
Nutritional Management of Children with Prader-Willi Syndrome
Peggy L. Pipes 91

chapter 8
Nutrition, Metabolism, Body Composition, and Response to the Ketogenic Diet in Prader-Willi Syndrome
Ralph A. Nelson, Diane M. Huse, Ralph T. Holman, Barbara O. Kimbrough, Heinz W. Wahner, C. Wayne Callaway, and Aldine B. Hayles 105

chapter 9
Surgical Treatment of Morbid Obesity in Prader-Willi Syndrome
Robert T. Soper, Edward E. Mason, Kenneth J. Printen, and Hans Zellweger 121

chapter 10
Elevated Adipose Tissue Lipoprotein Lipase in the Pathogenesis of Obesity in Prader-Willi Syndrome
Robert S. Schwartz, John D. Brunzell, and Edwin L. Bierman ... 137

part III **BEHAVIORAL AND SOCIAL ASPECTS**

chapter 11
Behavioral and Cognitive Disabilities in Prader-Willi Syndrome
Stephen Sulzbacher, Keith A. Crnic, and Jeff Snow 147

chapter 12
Cognitive Processing in Children with Prader-Willi Syndrome
Judith L. Warren and Earl Hunt 161

chapter 13
Speech and Language Characteristics of Children with Prader-Willi Syndrome
Cynthia Branson 179

chapter 14
A Behavioral Approach to Treatment of Prader-Willi Syndrome
Barringer D. Marshall, Jr., Charles J. Wallace, John Elder, Karen Burke, Tim Oliver, and Richard Blackmon 185

chapter 15
Maintenance of Programmed Weight Loss in Patients with Prader-Willi Syndrome
Glenn Hirsch and Karl Altman 201

chapter 16
A Retrospective Study of the Behavior of Prader-Willi Syndrome versus Other Institutionalized Retarded Persons
Ruth Turner and Rogelio H. A. Ruvalcaba 215

chapter 17
Relaxation Training with Youngsters with Prader-Willi Syndrome
Stevan L. Nielsen and Stephen Sulzbacher 219

chapter 18
Implications of Prader-Willi Syndrome for the Individual and Family
Jurgen Herrmann 229

chapter 19
Social Work Intervention Strategies for Families with Children with Prader-Willi Syndrome
Judith M. Leconte 245

part IV **MEDICAL ASPECTS**

chapter 20
Medical Management of Prader-Willi Syndrome
Vanja A. Holm 261

chapter 21
Physical Growth in Prader-Willi Syndrome
Jane K. Nugent and Vanja A. Holm 269

chapter 22
Endocrine Profiles and Metabolic Aspects of Prader-Willi Syndrome
W. Jun Tze, Henry G. Dunn, and Raphe L. Rothstein 281

chapter 23
Scoliosis in Prader-Willi Syndrome
Edwin L. Laurnen 293

chapter 24
Physical Exercise for Children and Adults with Prader-Willi Syndrome
Paula M. Carman 299

chapter 25
The Prader-Willi Syndrome Directions in Future Research
Stephen Sulzbacher, Vanja A. Holm, and Peggy L. Pipes 313

The Prader-Willi Syndrome An Annotated Bibliography 317

Index 339

chapter 1

THE PRADER-WILLI SYNDROME
Historical Perspective

Michael J. Steffes, Vanja A. Holm, and Stephen Sulzbacher

In this chapter the historical background of the Prader-Willi syndrome is discussed, including two possible cases from the 17th and 19th centuries. —*Eds.*

We can assume that Prader-Willi (P-W) syndrome individuals existed long before the syndrome was identified in 1956, and they were given a specific diagnostic label. Since the discovery was so recent, it is especially interesting to see whether indications of the syndrome can be found earlier. Humankind's long-standing interest in obesity and hyperphagia can be seen in art, mythology, folklore, and literature. Characters such as Falstaff, Mr. Pickwick, or, for that matter, Santa Claus, still appeal to the imagination. However, one would not expect a search in these areas to reveal individuals who could be specifically identified as having P-W syndrome because they have not been concerned primarily with the kind of precise physical description necessary for medical or scientific purposes.

This makes it all the more surprising that in two portraits of a 17th century Spanish girl, the clinical features of P-W syndrome are clearly in evidence (Figures 1.1 and 1.2). The subject of the paintings was 6-year-old Eugenia Martínez Vallejo, known as "La Monstrua" because of her great obesity. The paintings were done by Juan Carreño de Miranda, painter to the Spanish court, at the order of King Charles II about 1680. Eugenia had been summoned to the court by the king, who was noted for his fondness for human oddities, when news of her unusual appearance reached him (Beruete y Moret, 1909). At the time, she weighed about 120 pounds (Moreno Villa, 1939). The excessive obesity at such an early age, in addition to the small triangular mouth and the small hands and feet, strongly suggests P-W syndrome. The distribution of adipose tissue is typical for the syndrome. Naturally one cannot make a precise clinical diagnosis on the basis of two paintings and a few scraps of historical information, but there is no likely alternate explanation for this girl's appearance. We can, of course, assume that many other cases of P-W syndrome existed