Pediatric Endocrinology

Sally Radovick Madhusmita Misra Editors

Pediatric Endocrinology

A Practical Clinical Guide

Third Edition



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This textbook is dedicated to Margaret MacGillivray (Aug. 30, 1930–Sept. 17, 2016), for her vision in editing the first edition and with thanks for asking me to join her. I knew Margaret as a colleague, a mentor, and a friend. For those of us that were lucky enough to know her, she will be remembered as an outstanding clinician, investigator, mentor, and leader. Her passion for pediatric endocrinology, her compassion for others, and her upbeat spirit made her not only a most respected colleague but also a deeply valued friend.

Margaret was a towering figure in pediatric endocrinology making groundbreaking contributions to it. A true professional and role model, Margaret contributed to almost every aspect of pediatric endocrinology: thyroid disease, disorders of growth and puberty, and diabetes. In the mid-1960s, she published a seminal study on growth hormone secretion, defining for the first time short stature in children commonly seen with delayed puberty. She was a true physician scientist, investigating the factors that regulate growth hormone in these affected children. Her legacy to our field will be the pioneering use of growth hormone treatment for children with dwarfism. With this impact, Margaret is alive today and for generations to come.

I met Dr. MacGillivray in 1995 when she was president of the Pediatric Endocrine Society. I believe she never sought to be a leader, but became one naturally through her brilliance, compassion, patience, and selflessness. Her presidential address to the society was inspirational as I was beginning my career. I got to know Margaret well as a member of a prestigious grant review panel and little did I know that she had recommended my membership. Her guidance was critical as I was beginning to develop my academic reputation. In her gentle well-meaning, but somewhat blunt, way, she asked me if I had considered the insecurity associated with my academic position and whether the benefits were sufficient (which I had only cursorily considered). My salary was being funded entirely by NIH grants, which were subject to the vagaries of federal funding; I had 2 children and was married to a physician scientist. It was this discussion that changed my career course. She asked me to consider "replacing her" (imagine that) in Buffalo as she was thinking about stepping down as division director. Unfortunately, this did not work out, but her "reality check" stayed with me as I made my future career decisions. Twenty years later, I followed in her footsteps and was elected president of the Pediatric Endocrine Society.

On several occasions, we discussed the need for a pediatric endocrinology textbook focused on the knowledge required by clinicians that was comprehensive, organized,

and relevant. Agreeing in principal, she gained the support of Humana Press and asked me to co-edit the book with her. This was again an example of her mentorship, allowing me to share her academic stature. Her main goal, reflected in the preface, was to encourage the senior author of each chapter to include "a junior coauthor" as an opportunity to learn, to be mentored, and to give the next generation recognition in the field. With this third edition, we continue her tradition of a junior colleague as coauthor.

My relationship with Margaret has taught me most about the importance of mentorship. She taught mentorship by example and never demanded of her mentees what she would not expect of herself. She brilliantly mentored a generation of doctors with her characteristic compassion, grace, wisdom, and clever sense of humor. She was and still is an inspiration to women who pursue a career in medicine – very seldom looking backward to difficulties she had to endure as a woman, rather looking always forward. Some women would be very angry and bitter, but she always looked back on that as a challenge, and she overcame it. There were no role models or mentors at the time. She broke the glass ceiling and became the role model. Although she was dedicated to her roles as professor, clinician, and researcher, she was passionate about her role as wife, mother, and grandmother.

She taught me that hard work, determination, refusal to give up when the going gets rough, and, above all, sticking to one's ideals make for a successful career and a contented life. Margaret was a star. She didn't just shine; she blazed.

In this spirit, I welcome Madhu Misra as a co-editor of the third edition. Dr. Misra is the Fritz Bradley Talbot and Nathan Bill Talbot professor of pediatrics, Harvard Medical School, and division chief of pediatric endocrinology at the Massachusetts General Hospital. Her clinical interests include disorders of the pituitary gland and bone. Her research interests include the neuroendocrine and bone consequence of conditions that span the nutritional spectrum from anorexia nervosa to exercise-induced amenorrhea to obesity and conditions such as autism spectrum disorder and major depressive disorders.

Additionally, Dr. Misra is known for her successful mentorship of the next generation of pediatric endocrinologists and her service to the field as exemplified by her distinguished service to the Pediatric Endocrine Society.

Sally Radovick, MD

Preface

We welcome you to the third edition of *Pediatric Endocrinology: A Practical Clinical Guide*. The aim of this edition remains similar to the previous: to provide practical detailed and concise guidelines for the clinical management of pediatric endocrine diseases and disorders. Thus, the audience includes pediatric endocrinologists, pediatricians, and primary care physicians who provide medical care for children and adolescents.

The scope of the text continues to include the most common and the most challenging diseases and disorders seen by both primary care physicians and pediatric endocrinologists. We have encouraged the involvement of a junior coauthor to give recognition to our young investigators in the field. We believe we have assembled a state-of-the-art, comprehensive text on the practice of pediatric endocrinology.

Although the main focus of this text is on diagnosis and treatment, each author has included a brief discussion on pathophysiology and molecular mechanisms. The chapters have been organized in such a way as to present the following elements in synchrony: (1) a table of contents and key points; (2) an introductory discussion with background information; (3) a brief over-

view of recent progress on the mechanism involved; (4) a discussion of the etiology and clinical features that characterize each condition; (5) a delineation of the criteria used to establish a diagnosis; (6) a therapy section which comprehensively reviews the options available and the risks and benefits of each approach corroborated by clinical trial and outcome data, includes information on the long-term safety and efficacy of the treatment modality, and cites guidelines when available; (7) where relevant, a discussion of psychosocial and quality-of-life issues; and (8) finally a new section in this edition which includes related case studies and relevant questions.

Due to the dynamic clinical practice of pediatric endocrinology, extensive revisions and significant changes have been made to reflect current knowledge and practice. We have added chapters and expanded chapter content on care of gender nonconforming/transgender youth, diagnosis and management of osteoporosis, mineralocorticoid disorders and hypertension, and delayed puberty and hypogonadism.

We are most thankful for the generous contributions of our author colleagues. We hope you find the textbook helpful, and we are, of course, open to your comments.

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