Contents

VII Preface
Szinnai, G. (Basel)

Thyroid Hormone Physiology

1 Evolution, Child Development and the Thyroid: A Phylogenetic and Ontogenetic Introduction to Normal Thyroid Function
Krude, H. (Berlin)

17 Human Fetal Thyroid Function
Polak, M. (Paris)

26 The Role of Thyroid Hormones for Brain Development and Cognitive Function
Rovet, J.F. (Toronto, Ont.)

Congenital Hypothyroidism

44 Neonatal Screening for Congenital Hypothyroidism
Torresani, T. (Zurich)

50 Diagnosis, Treatment and Outcome of Congenital Hypothyroidism
Van Vliet, G.; Deladoëy, J. (Montreal, Que.)

60 Clinical Genetics of Congenital Hypothyroidism
Szinnai, G. (Basel)

79 Central Hypothyroidism in Children
García, M.; Fernández, A.; Moreno, J.C. (Madrid)

Impaired Sensitivity to Thyroid Hormones

108 Thyroid Hormone Cell Membrane Transport Defect
Ramos, H.E. (Salvador)

118 Resistance to Thyroid Hormone
Onigata, K. (Izumo); Szinnai, G. (Basel)

Thyroid and Environment

130 Iodine Deficiency in Children
Pearce, E.N. (Boston, Mass.)
Autoimmune Thyroid Disease

139 Thyroid Autoimmunity
Wiersinga, W.M. (Amsterdam)

158 Clinical Aspects of Hashimoto's Thyroiditis
Radetti, G. (Bolzano)

171 Graves' Disease in Children
Léger, J. (Paris)

Thyroid Cancer

183 Thyroid Nodules and Differentiated Thyroid Cancer
Bauer, A.J. (Philadelphia, Pa.)

202 Medullary Thyroid Carcinoma in Children
Viola, D.; Romei, C.; Elisei, R. (Pisa)

Clinical Case Seminar

214 Clinical Case Seminar in Pediatric Thyroid Disease
Szinnai, G. (Basel); Léger, J. (Paris); Bauer, A.J. (Philadelphia, Pa.); Pearce, E.N. (Boston, Mass.); Ramos, H.E. (Salvador); Canalli, M.H. (Florianópolis); Onigata, K. (Izumo); Elisei, R. (Pisa); Radetti, G. (Bolzano); Polak, M. (Paris); Van Vliet, G.; Deladoëy, J. (Montreal, Que.)

245 Author Index

246 Subject Index
Preface

An early Karger publication on thyroid disease in children, *Pediatric Thyroidology*, edited by F. Delange, D.A. Fisher and P. Malvaux, appeared in 1985. It represented an extensive summary of the body of knowledge of all the thyroid diseases encountered by paediatricians and paediatric endocrinologists. Almost 30 years later, it is still a source of fascinating data with detailed clinical and scintigraphic descriptions of thyroid physiology and thyroid diseases.

This up-to-date volume resumes this tradition and summarizes the latest knowledge on all clinically relevant aspects of thyroid diseases – from the neonate to the adolescent. There are 15 clinical chapters, providing reviews of distinguished experts in the field. The chapters cover topics ranging from the developmental role of thyroid hormones, fetal thyroidology, congenital hypothyroidism, central hypothyroidism, inherited defects of thyroid hormone cell membrane transport and thyroid hormone action to iodine deficiency, autoimmune thyroid disease and thyroid tumours. All of the chapters are written to provide detailed clinical and molecular knowledge, and to review the current guidelines in paediatric thyroidology.

To extend the classical textbook format, the final chapter ‘Clinical case seminar in pediatric thyroid disease’ comprises a collection of representative case histories selected by each chapter author. This ‘crystalline’ form of the preceding 15 chapters aims at providing concise key learning points and references to specific problems encountered in clinical practice.

I am grateful to Prof. Primus Mullis, the editor of the *Endocrine Development* series, and Karger Publishers, for inviting me to edit this book. I wish to thank all of the authors and colleagues who participated in this book project with great enthusiasm despite short deadlines to provide an up-to-date volume and illustrated case histories. The chapters reflect their tremendous experience and commitment to children with thyroid diseases. Together with all of the coauthors, I hope the book will be useful for the reader and their patients.

*Gabor Szinzai*, Basel