Paediatric Thyroid Disorders: New Insights

Guest Editors
Michel Polak, Paris
Gabor Szinnai, Basel
Paediatric Thyroidology

Editor
Gabor Szinnai

This book presents a comprehensive overview of paediatric thyroid diseases and thus provides a useful tool for clinical problem solving. Opinion leaders in the field present reviews on all relevant diseases of the hypothalamic-pituitary-thyroid axis. Sixteen chapters cover topics ranging from foetal thyroidology, congenital hypothyroidism, central hypothyroidism, inherited defects of thyroid hormone action, cell transport and metabolism to iodine deficiency, autoimmune thyroid disease and thyroid tumours. Written by clinicians, the chapters provide in-depth information and current guidelines for clinical problems encountered in paediatric thyroidology. As a unique feature, a case seminar collection for each chapter presents typical patient histories providing key learning points and key references for clinical problem solving in family medicine, paediatric endocrinology and medical genetics.

Providing a succinct update on clinical paediatric thyroidology, this book is an essential tool for paediatric and adult endocrinologists, as well as for general practitioners, paediatricians and medical geneticists.

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Following technical and methodological improvements of the last decade, the underlying genes of a number of syndromes involving severe intellectual disability resembling Angelman and Rett syndromes have been identified. In order to keep track with these new entities, this special issue of Molecular Syndromology provides comprehensive reviews of the state of clinical and genetic knowledge about the ‘old’ entities of Angelman and Rett syndromes, as well as the newer syndromes related to MECP2 duplication or defects in the CDKL5, FOXG1, MEF2C, TCF4, NRXN1, CNTNAP2, SHANK3, EHMT1 and FOXP1 genes. Furthermore the special challenge that presents itself when seeking to establish a diagnosis in adult patients is discussed. All articles are authored by experts specializing in these particular syndromes. This publication should therefore provide a unique source of knowledge about these relatively common syndromes and should be an asset to all clinical geneticists, neuropediatricians, and researchers in the field of neurodevelopmental disorders. Since this collection of articles is presented within a single issue, it facilitates comparison between the different syndromes and seems to be destined to become a desk book that anyone involved in the field of medical genetics dare not overlook.

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The European Thyroid Journal publishes papers reporting original research in basic, translational and clinical thyroidology. Original contributions cover all aspects of the field, from molecular and cellular biology to immunology and biochemistry, from physiology to pathology, and from pediatric to adult thyroid diseases with a special focus on thyroid cancer. Readers also benefit from reviews by noted experts, which highlight especially active areas of current research. The journal will further publish formal guidelines in the field, produced and endorsed by the European Thyroid Association.

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• 2013 European Thyroid Association Guidelines for Cervical Ultrasound Scan and Ultrasound-Guided Techniques in the Postoperative Management of Patients with Thyroid Cancer: Leenhardt, L. (Paris); Erdogan, M.F. (Ankara); Hegedus, L. (Odense); Mandel, S.J. (Philadelphia, Pa); Paschke, R. (Leipzig); Rago, T. (Pisa); Russ, G. (Paris)
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Thyroid FNA: Challenges and Opportunities

Editors
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Over recent years, impressive advances in genetic/epigenetic technology have greatly improved the understanding of the pathogenesis of pediatric endocrine diseases due to hormone resistance and hypersensitivity. This book presents reviews of thyroid hormone and thyroid hormone receptor resistance, and genetics and epigenetics of parathyroid hormone resistance. Abnormalities of the pituitary-gonadal axis affecting puberty as well as androgen receptor are covered. Novel insights into the diseases affecting ACTH, glucocorticoid and aldosterone receptors are discussed. Further chapters address new aspects of the physiology of the GH and IGF-1 axis as well as the diseases related to GH-IGF-1 receptor and post-receptor signaling defects. A key chapter on metabolic insights into insulin resistance is also included.

Covering clinical and genetic aspects of hormone resistance and hypersensitivity, this book will be a useful tool in the hands of scientists, physicians and other healthcare professionals who wish to be up to date with novel research findings in this area.
Paediatric Thyroid Disorders: New Insights

New data concerning screening, diagnosis and management is important for a constant broadening of our knowledge and expertise. This special topic issue of Hormone Research in Paediatrics focuses on new developments in the field of thyroid disease in the paediatric age group. Topics covered include epidemiology, screening, radiologic approaches, and new molecular data. Reviews of specific diseases associated with thyroid disorders such as Down syndrome, Rett syndrome, and pseudohypoparathyroidism add new insights into clinically relevant questions. Providing a selection of papers on clinical and molecular aspects of congenital hypothyroidism, this special topic issue offers an update on fetal, neonatal, and childhood thyroidology in patients suffering from congenital hypothyroidism, namely thyroid disease associated with genetic syndromes.