Thyroid Gland Development and Function
Endocrine Development

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Thyroid Gland
Development and Function

Volume Editors

Guy Van Vliet  Montreal, Que.
Michel Polak  Paris

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Guy Van Vliet, MD
Endocrinology Service and Research Center
Department of Pediatrics
Sainte-Justine Hospital
University of Montreal
Montreal, Quebec, Canada

Michel Polak, MD, PhD
Endocrinologie pédiatrique
INSERM U845
Hôpital Necker Enfants Malades
Paris, France

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Contents

VII  Foreword
    Savage, M.O. (London)

IX  Preface
    Van Vliet, G. (Montreal, Que.); Polak, M. (Paris)

Disorders of Thyroid Gland Development

1  Murine Models for the Study of Thyroid Gland Development
    De Felice, M.; Di Lauro, R. (Naples/Ariano Irpino)

15  Familial Forms of Thyroid Dysgenesis
    Castanet, M.; Polak, M.; Léger, J. (Paris)

29  Possible Non-Mendelian Mechanisms of Thyroid Dysgenesis
    Deladoëy, J. (Montreal, Que.); Vassart, G. (Brussels); Van Vliet, G. (Montreal, Que.)

43  Thyroid Imaging in Children
    Garel, C.; Léger, J. (Paris)

Disorders of Thyroid Function

62  Clinical and Biological Consequences of Iodine Deficiency during Pregnancy
    Glinoer, D. (Brussels)
86  Ontogenesis of Thyroid Function and Interactions with Maternal Function
Obregon, M.J.; Calvo, R.M.; Escobar del Rey, F.; Morreale de Escobar, G. (Madrid)

99  New Phenotypes in Thyroid Dysshormonogenesis: Hypothyroidism due to DUOX2 Mutations
Moreno, J.C.; Visser, T.J. (Rotterdam)

Disorders of Thyroid Hormone Metabolism

118 Thyroid Hormone Transporter Defects
Grüters, A. (Berlin)

127 Novel Biological and Clinical Aspects of Thyroid Hormone Metabolism
Dumitrescu, A.M.; Refetoff, S. (Chicago, Ill.)

Pediatric Thyroid Tumors

140 Papillary and Follicular Thyroid Cancers in Children
Vasko, V. (Bethesda, Md.); Bauer, A.J. (Bethesda, Md./Washington, D.C.); Tuttle, R.M. (New York, N.Y.); Francis, G.L. (Richmond, Va.)

173 Hereditary Medullary Thyroid Carcinoma: How Molecular Genetics Made Multiple Endocrine Neoplasia Type 2 a Paediatric Disease

188 Author Index

189 Subject Index
This volume in the Endocrine Development series entitled Thyroid Gland Development and Function fits perfectly into the primary aim of the series, which is to discuss the physiology and clinically relevant pathophysiology of key endocrine systems. Scientific and clinical interests are given prominence in this volume. Professor Polak and Professor Van Vliet are highly experienced, both from experimental and clinical standpoints, to edit this issue. They have chosen subjects of major interest and contributors of very high quality.

Human thyroid development and its defects are described with the help of genetic studies in mouse models. The metabolic aspects of thyroid hormone action are also discussed. Genetic defects of thyroid hormone synthesis are covered and their clinical relevance debated. The important field of thyroid cancer in the context of spontaneous occurrence and as part of familial neoplasia syndromes is described in detail. Finally the important problem of environmental iodine deficiency which has emerged as a global public health concern is rightly included.

Overall, this excellent volume will inform scientists and clinicians of key areas in the field of thyroid disorders. I enthusiastically welcome this latest addition to the series.

Martin O. Savage, London
Preface

In 1985, Karger published a book entitled Pediatric Thyroidology which had been edited by F. Delange, D.A. Fisher and P. Malvaux. Since then, tremendous advances have taken place in developmental and molecular biology. These advances have had a major impact on all fields of medicine, and pediatric thyroidology is no exception. Consistent with its publication in the Endocrine Development series edited by Martin Savage, this book starts with chapters focusing on developmental abnormalities of the thyroid gland in genetically engineered mice. Studies are described on the possible mendelian and non-mendelian mechanisms involved in abnormalities of thyroid development in humans and on their proper classification by imaging. Ironically, the commonest developmental abnormality, a defect in migration resulting in thyroid ectopy, remains an enigma in the field.

The recent advances in our understanding of thyroid hormone metabolism and transport into the cells, which have been revealed by astute observations of ‘experiments of nature’ observed in children, followed by sophisticated molecular investigations, are reviewed next.

What makes the thyroid rather unique in the field of endocrinology is its critical dependence on an environmental factor, iodine. Pregnant women are particularly sensitive to a low nutritional supply of iodine. Within the thyroid, iodine needs to be oxidized, a process which requires H$_2$O$_2$; genetic lesions resulting in decreased function of the protein involved in the generation of H$_2$O$_2$ lead to a form of hypothyroidism that may be exacerbated during pregnancy and the newborn period. The intricate relationships between maternal and fetal thyroid function may result in major consequences of maternal hypothyroidism on
the psychomotor development of the offspring. These aspects are reviewed in the next section.

The biology of tumors arising from thyroid follicular cells in childhood differs from those arising from the same cells later in life. Tumors arising from the parafollicular or C cells represent the first example of the major impact that DNA-based diagnosis has had on the practice of pediatric endocrinology; in this area, we stand on the verge of ‘codon-specific’ medicine. These pediatric thyroid tumors are reviewed in the last section of this book.

We are well aware that our choice of topics may seem rather arbitrary. It was not our aim to produce a complete overview of pediatric thyroid diseases and their consequences, but rather to focus on selected topics which fell under the general umbrella of Endocrine Development and in which we felt that major advances had recently been made, usually through a combination of clinical observations and patient-oriented basic biological investigations. We thank all the authors for their outstanding contributions and sincerely hope the readers will learn from perusing this book as much as we have from editing it.

Guy Van Vliet, Montreal, Que.
Michel Polak, Paris