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Hypothalamic-Pituitary Development
Hypothalamic-Pituitary Development
Genetic and Clinical Aspects

Volume Editors

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34 figures, 2 in color, 22 tables, 2001
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The fourth volume in the series *Endocrine Development* continues the pattern established in earlier volumes of combining discussions on fundamental scientific and clinical advances. Professor Raphaël Rappaport and Professor Serge Amselem, themselves experts in clinical and molecular endocrinology respectively, have assembled an impressive list of world authorities to contribute to this volume. Hypothalamic-pituitary development is at the origin of many essential endocrine systems and defects in this developmental process cause a wide spectrum of clinical defects relevant to both paediatric and adult endocrinology.

The genetic basis of many of these defects has been established in the past 20 years. The book includes descriptions of normal and abnormal neuroendocrine development and function. Anatomical and genetic derangements are described and linked to their clinical syndromes.

Overall, this book continues the long-term aim of the series, which is to explain, instruct and stimulate scientists and clinicians to increase their knowledge of important and clinically relevant endocrine disorders.

London, March 2001

*Martin O. Savage*
Preface

Over recent years, dramatic progress has been made in understanding pituitary insufficiency in the human by the combination of experimental studies, molecular genetics and clinical research. The description of pituitary gland development has opened a new and exciting field of research and provided clues to the ill-defined group of idiopathic isolated growth hormone deficiency or multiple pituitary hormone deficiency.

The unique opportunity to provide an updated and prospective view of this field was given to us when it was proposed by Professor M. Savage to edit a book covering this rapidly expanding knowledge. All authors have accepted the challenge to integrate complex knowledge from various scientific and medical fields providing updated and didactic information with new approaches in developmental endocrinology.

Anterior and hypothalamic organogenesis is described in animal models with particular reference to the human. Whenever possible, emphasis is put on the combination of pituitary defects with a multitude of facial and cranial abnormalities, some of which are part of well-described syndromes like septo-optic dysplasia. An effort is made to correlate genotypic and phenotypic data in order to support innovative practice in pediatrics and endocrinology.

Fine imaging of the brain and the pituitary, using magnetic resonance, contributes to define new organic pituitary defects reducing the component of ‘idiopathic’ conditions with growth hormone deficiency. The pituitary stalk interruption syndrome with its various presentations remains unexplained although suggesting strong interactions between hypothalamus and anterior pituitary during embryogenesis.

Exciting clinical data are also reported, indicating the role of a number of homeodomain transcription factors regulating pituitary cell specification and thereby various combinations of hormonal defects. Several months or years after the initial reports, some of which were made by contributors to this book, follow-up of patients and reports of new cases have added valuable new information for the clinician and the genetist. In addition, various aspects of posterior pituitary development and dysfunction are described with particular emphasis on recent imaging data.
It was the intention of the editors and the contributors to help clinicians and
to stimulate their interest in unique models of genetic and developmental defects
of the pituitary which bear ultimate consequences on metabolic control, growth
and reproduction. It is our wish that this effort might have contributed to provide
an easy entry to modern ‘molecular medicine’ as well as to ‘developmental endo-
crinology’.

Raphaël Rappaport
Serge Amselem