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Genetics of Endocrine Diseases and Syndromes

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Preface

This book is intended for both clinicians and researchers to help in understanding the complexity and relevance of genetics in endocrinology.

Genetic factors are implicated in the disturbances of hormone homeostasis and diseases of hormone-producing endocrine organs. Some of them are primarily genetic diseases, such as monogenic diseases. Several but rare monogenic diseases are known which affect the function of the hormones and their receptors, e.g., hormone resistance syndromes. An important group of monogenic endocrine diseases increase the susceptibility to tumors. Several hereditary tumor syndromes are known, in which an individual suffers from multiple tumors in different organs. Genetic alterations in several endocrine tumors are known to contribute to disease manifestations. Apart from monogenic diseases, the most common diseases with endocrine relevance are polygenic, e.g., obesity, and there are chromosome alterations with endocrinological relevance, as well.

In this book, we present a synopsis of the most important diseases with endocrine relevance. The book comprises 20 chapters divided into five parts. In the first part, basic concepts of genetics, inheritance patterns, issues of family screening and genetic counseling, and the molecular methodology in genetics are discussed. The following three parts discuss monogenic diseases: in Part II, hormone resistance syndromes; in the most extensive Part III, monogenic diseases predisposing to tumor formation; and in Part IV, monogenic diseases predisposing to hormone deficiency and infertility are presented. In the fifth part, a prototype of polygenic inheritance, the genetics of obesity is discussed along with chromosomal aberrations. The list of authors includes leading international experts on these topics.

The book includes 70 figures and 34 tables to facilitate understanding.

The chapters discuss both molecular genetics and clinical issues, highlighting genetic counseling, and thereby aim to present a complex picture of these disease entities.

The editors are indebted to the late Professor Károly Rácz, the former director of the 2nd Department of Internal Medicine of Semmelweis University, who had the original idea to compile this book, but he unfortunately passed away in 2017.

We hope that the reader will find this book interesting, and the book will help to gain insights into this fascinating field of research and clinical medicine.

Budapest, Hungary

Peter Igaz
Attila Patócs

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List of the Most Commonly Used Abbreviations

11-OHD	11β-Hydroxylase deficiency
17-OHD	17α-Hydroxylase deficiency
17-OHP	17-Hydroxyprogesterone
21-OHD	21-Hydroxylase deficiency
ACC	Adrenocortical carcinoma
aCGH	Array comparative genomic hybridization
ACTH	Adrenocorticotropic hormone
ADH	Antidiuretic hormone
AHR	Aryl hydrocarbon receptor
AIP	Aryl hydrocarbon receptor-interacting protein
AIS	Androgen insensitivity syndrome
AL	Anterior lobes of pituitary
AMH	Anti-Müllerian hormone
ANP	Atrial natriuretic peptide
APA	Aldosterone-producing adenoma
APC	Adenomatous polyposis coli
AQP	Aquaporin water channels
ARR	Aldosterone-to-renin ratio
AS	Angelman syndrome
AVP	Arginine vasopressin
BAH	Bilateral adrenal hyperplasia
BBB	Blood–brain barrier
BMI	Body mass index
BWS	Beckwith–Wiedemann syndrome
cAMP	Cyclic adenosine monophosphate
CAH	Congenital adrenal hyperplasia
CaSR	Calcium sensor
CAVD	Congenital absence of vas deferens
CDKN1C	Cyclin-dependent kinase inhibitor 1C
cDNA	Complementary DNA
CEL	Carboxyl ester lipase