

Peter Igaz  
Attila Patócs *Editors*

# Genetics of Endocrine Diseases and Syndromes

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Editors

# Genetics of Endocrine Diseases and Syndromes

 Springer

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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 | 11 $\beta$ -Hydroxylase deficiency            |
| 17-OHD | 17 $\alpha$ -Hydroxylase deficiency           |
| 17-OHP | 17-Hydroxyprogesterone                        |
| 21-OHD | 21-Hydroxylase deficiency                     |
| ACC    | Adrenocortical carcinoma                      |
| aCGH   | Array comparative genomic hybridization       |
| ACTH   | Adrenocorticotropin 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                         |