Hormone Resistance and Hypersensitivity
From Genetics to Clinical Management

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Contents

VII Preface
Maghnie, M. (Genoa); Loche, S. (Cagliari); Cappa, M. (Rome); Ghizzoni, L. (Turin); Lorini, R. (Genoa)

1 Thyroid Hormone Transporters and Resistance
Visser, T.J. (Rotterdam)

11 Genetics and Epigenetics of Parathyroid Hormone Resistance
Bastepe, M. (Boston, Mass.)

25 Gonadotropin Resistance
Latronico, A.C.; Arnhold, I.J.P. (São Paulo)

33 Clinical and Molecular Aspects of Androgen Insensitivity
Hiort, O. (Lübeck)

41 Exploring the Molecular Mechanisms of Glucocorticoid Receptor Action from Sensitivity to Resistance
Ramamoorthy, S.; Cidlowski, J.A. (Research Triangle Park, N.C.)

57 ACTH Resistance: Genes and Mechanisms

67 Primary Generalized Familial and Sporadic Glucocorticoid Resistance (Chrousos Syndrome) and Hypersensitivity

86 Pseudohypoaldosteronism
Riepe, F.G. (Kiel)

96 New Aspects of the Physiology of the GH-IGF-1 Axis
Vottero, A. (Parma); Guzzetti, C.; Loche, S. (Cagliari)

106 Molecular and Clinical Aspects of GHRH Receptor Mutations
Corazzini, V.; Salvatori, R. (Baltimore, Md.)

118 Current Issues on Molecular Diagnosis of GH Signaling Defects
Feigerlova, E.; Hwa, V.; Derr, M.A.; Rosenfeld, R.G. (Portland, Oreg.)

128 Molecular IGF-1 and IGF-1 Receptor Defects: From Genetics to Clinical Management
Walenkamp, M.J.E. (Amsterdam); Losekoot, M.; Wit, J.M. (Leiden)
Phenotypes, Investigation and Treatment of Primary IGF-1 Deficiency
Savage, M.O. (London)

Human Congenital Perilipin Deficiency and Insulin Resistance

Author Index
Subject Index
Preface

Over recent years, a tremendous progress has been made in the field of hormone resistance. The meeting on Hormone Resistance and Hypersensitivity – From Genetics to Clinical Management held in Genoa, Italy, on May 13–15, 2012, provided a unique opportunity for an updated and prospective view of this exciting topic.

The scientific program was designed to focus on the most recent advances related to the various aspects of hormone resistance affecting a number of endocrine or target organs. The impressive advances in genetic/epigenetic technology have greatly improved our understanding of the pathogenesis of pediatric endocrine diseases due to hormone resistance or hypersensitivity, as well as our diagnostic skills. Careful characterization of the phenotype of patients with hormone resistance together with decades of efforts in translational research have led to relevant improvements in the care of affected patients.

Hormone resistance is in fact a condition caused by a reduced or absent end-organ responsiveness to a biologically active hormone, which may be due to a hormone receptor defect (e.g. for glucocorticoids, androgens, estrogens, vitamin D derivatives, thyroid hormones, thyroid-stimulating hormone, parathyroid hormone, antidiuretic hormone, insulin) or a post-receptor defect. This book introduces clinical and genetic aspects of hormone resistance through a number of breakthrough developments that illustrate how molecular defects at various steps in hormone production, signaling, or responsiveness can cause disease in humans.

The volume contains 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. We have broadened our understanding of the diseases affecting ACTH, glucocorticoid and aldosterone receptors. 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 are comprehensively addressed.

A key chapter on metabolic insights into insulin resistance is also included.
Overall, this volume provides information directly useful to the clinician, and stimulates thought and future research opportunities with cutting-edge scientific results in the broad and important field of hormone resistance.

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