

Hormone resistance and hypersensitivity: From genetics to clinical management, M. Maghnie, S. Loche, M. Cappa, L. Ghizzoni, R. Lorini, editors (Karger, Basel, Switzerland) 2013. 160 pages. Price: US\$ 198.00 / CHF 168.00 / EUR 140.00
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This book is the 24th volume of the ongoing series 'Endocrine Development', edited by P. -E. Mullis. This book comprises 14 articles contributed by different authors and is a compilation of presentations during the workshop on 'Hormone Resistance and Hypersensitivity: From Genetics to Clinical Management' held in Genoa, Italy, in May 2012. In the preface, the editors have rightly emphasized that the meeting provided a unique opportunity for an updated and perspective view of this exciting topic.

Though disorders of end organ resistance and hypersensitivity to hormones are rare, but availability of powerful genetic/ epigenetic technologies has catalysed translational research in the field of pathogenesis of these disorders. The clinical application of their findings is the development of highly sensitive and specific molecular diagnostic tests for these disorders. This book is an excellent compilation of genotype-phenotype correlations of these paediatric endocrinology disorders.

Each of the articles in this book is on a specific hormone disorder. Each article describes the pathophysiology and clinical phenotype first, followed by compilation of available evidences of molecular genetic defects underlying the hormone resistance syndrome and lastly genotype-phenotype correlations and their therapeutic implications are discussed.

The first article is on thyroid hormone transporters and resistance. Physiology of thyroid hormone action, transportation specifically by MCT8 and MCT10 and their mutations are discussed. A brief overview of the thyroid hormone receptor is also given in this article. The second article is about the pseudohypoparathyroidism types-I and II and genetics and epigenetics of *GNAS* complex and *PRKAR* locus. The pathophysiologic role of their mutations and altered expression due to epigenetic changes is described. The third and fourth chapters are respectively on gonadotropin and androgen insensitivity and outline the clinical and genetic correlations between their receptor mutations and various reproductive hormonal disorders.

The next three chapters are respectively on the molecular mechanism of glucocorticoid receptor action from sensitivity to resistance, ACTH resistance and its defects and generalized glucocorticoid resistance (Crousos syndrome) and sensitivity.

The eighth chapter is on pseudohypoaldosteronism (PHA) and describes its three types and renal sodium channel, and *NR3C2* mutations underlying PHA1 and PHA2, respectively. The subsequent five chapters are on growth hormone releasing hormone (GHRH), growth hormone (GH) and growth hormone–insulin like growth factor (IGF-I) axis resistance disorders. Mutations in genes coding GHRH, growth hormone receptor, GH signal transduction pathway like STAT5b pathway, IGF-1 and its receptor are described. The associated clinical phenotype and therapeutic implications with human growth hormone and IGF are discussed.

A limitation of this book is that there is only one article on insulin resistance and that is on loss of function mutation *PLIN1* gene and its association with severe partial lipodystrophy. Though this book contains an overview of specific hormone resistance syndrome genetics and phenotype, it does not include any article on the general aspects of the molecular basis of hormonal action and genetic/genomic techniques used.

Despite the shortcomings, the book is a good attempt to present information on clinical genetics of hormone resistance and sensitivity disorders. This information is usually not available in most of the clinical endocrinology books. This book is primarily recommended for clinical endocrinologists and postgraduates in this superspeciality, however, it is also recommended for experts interested in planning research in this area.

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