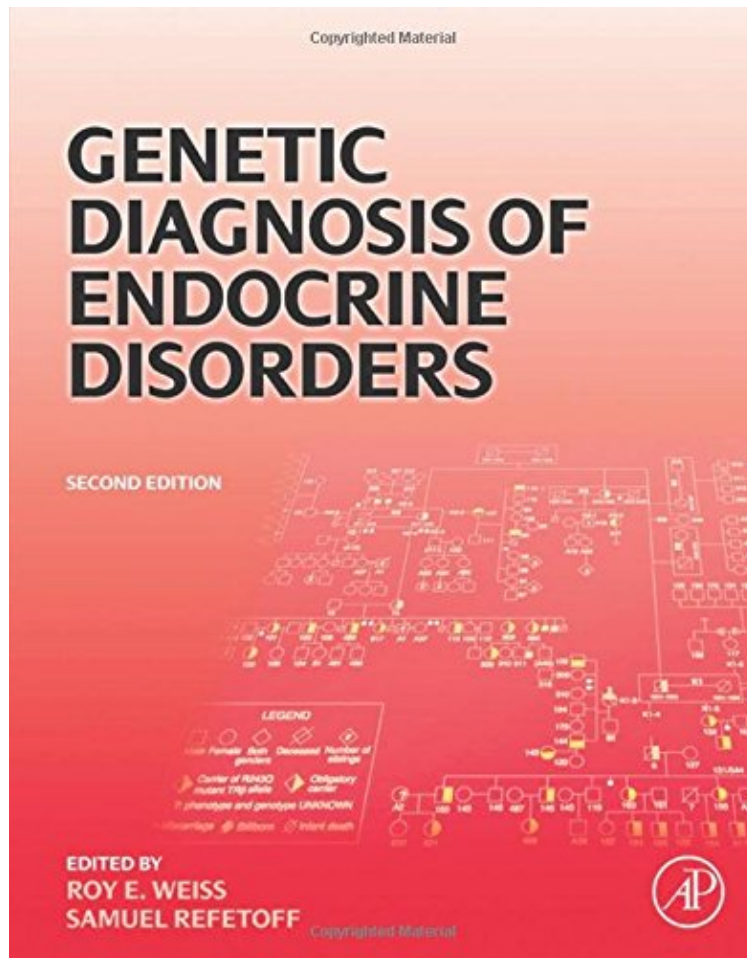


[Download pdf ebook] Genetic Diagnosis of Endocrine Disorders, Second Edition

Genetic Diagnosis of Endocrine Disorders, Second Edition

From Academic Press

*ebooks | Download PDF | *ePub | DOC | audiobook*



DOWNLOAD



READ ONLINE

#2667586 in Books 2015-11-06Original language:EnglishPDF # 1 10.90 x 1.10 x 8.50l, 3.65 #File Name: 012800892X472 pages | File size: 64.Mb

From Academic Press : Genetic Diagnosis of Endocrine Disorders, Second Edition before purchasing it in order to gage whether or not it would be worth my time, and all praised Genetic Diagnosis of Endocrine Disorders, Second Edition:

Genetic Diagnosis of Endocrine Disorders, Second Edition provides users with a comprehensive reference that is organized by endocrine grouping (i.e., thyroid, pancreas, parathyroid, pituitary, adrenal, and reproductive and bone), discussing the genetic and molecular basis for the diagnosis of various disorders. The book emphasizes the practical nature of diagnosing a disease, including which tests should be done for the diagnosis of diabetes mellitus in adults and children, which genes should be evaluated for subjects with congenital hypothyroidism, which genetic tests should be ordered in obese patients or for those with parathyroid carcinoma, and the rationale behind testing for multiple endocrine neoplasias.Offers a clear presentations of pharmacogenetics and the actual assays used in detecting

endocrine diseases Teaches the essentials of the genetic basis of disease in each major endocrine organ system Offers expert advice from genetic counselors on how to use genetic information in counseling patients Includes new chapters on the genetics of lipid disorders and glycogen storage diseases, genetics of hypoglycemia, and whole genome/exome sequencing

About the Author Dr. Roy Weiss joined the University of Miami Leonard M. Miller School of Medicine as Chairman and the Kathleen and Stanley Glaser Distinguished Chair in Medicine in June 2014. He is also the Rabbi Morris I. Esformes Professor Emeritus at the University of Chicago. Dr. Weiss is an expert in diseases of the thyroid and has described several genetic diseases of the thyroid along with Dr. Samuel Refetoff. Together, they have one of the largest referral centers for genetic thyroid disease in the world. His research centers on the mechanisms of thyroid hormone action at the molecular, physiological, and psychological levels. Dr. Weiss has examined the molecular basis for the syndrome of resistance to thyroid hormone (RTH) as well as other diseases of thyroid hormone action. He has published over 175 papers. He is a fellow in the American College of Physicians, The American College of Endocrinology, and the American Academy of Pediatrics. His research has been supported by the NIH and has received numerous teaching awards. He is the current President of the Central Society for Clinical and Translational Research. Samuel Refetoff is known for his discovery (1967) of resistance to thyroid hormone (Refetoff Syndrome) and elucidation of its genetic and molecular basis (1989/92). Devoted to the study of inherited thyroid disorders, his laboratory was first to identify mutations in: serum thyroid hormone transport proteins [TBG (1989) and albumin (1994)]; the TSH receptor producing resistance to TSH (1995); and two syndromic thyroid defects combining neuropsychological and thyroid abnormalities caused by mutations in the TTF1 (2002) and the MCT8 (2004) genes. In 2005 his laboratory identified a defect of TH metabolism caused by mutations in the SBP2 gene, which is involved in the synthesis of selenoproteins. A graduate of McGill University, Dr. Refetoff is professor of medicine, pediatrics and genetics at the University of Chicago. He is author of over 500 publications and recipient of numerous national and international prizes, two NIH MERIT awards (1989/2006) and three honorary doctorate degrees.