

Endocrine Disorders: Focus On Clinical Diagnosis

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Review

Multiple endocrine neoplasia type 1 (MEN1) and type 4 (MEN4)

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ABSTRACT

Multiple endocrine neoplasia (MEN) is characterized by the occurrence of tumors involving two or more endocrine glands within a single patient. Four major forms of MEN, which are autosomal dominant disorders, are recognized and referred to as: MEN type 1 (MEN1), due to *menin* mutations; MEN2 (previously MEN2A) due to mutations of a *proline kinase receptor* encoded by the rearranged during transfection (RET) proto-oncogene; MEN3 (previously MEN2B) due to RET mutations; and MEN4 due to cyclin-dependent kinase inhibitor (CDKN1B) mutations. Each MEN type is associated with the occurrence of specific tumors. Thus, MEN1 is characterized by the occurrence of parathyroid, pancreatic islet and anterior pituitary tumors; MEN2 is characterized by the occurrence of medullary thyroid carcinoma (MTC) in association with pheochromocytoma and parathyroid tumors; MEN3 is characterized by the occurrence of MTC and pheochromocytoma in association with a marfanoid habitus, mucosal neuromas, medullated corneal fibers and intestinal autonomic ganglion dysfunction, leading to megacolon; and MEN4, which is also referred to as MENX, is characterized by the occurrence of parathyroid and anterior pituitary tumors in possible association with tumors of the adrenals, kidneys, and reproductive organs. This review will focus on the clinical and molecular details of the MEN1 and MEN4 syndromes. The gene causing MEN1 is located on chromosome 11q13, and encodes a 610 amino-acid protein, *menin*, which has functions in cell division, genome stability, and transcription regulation. *Menin*, which acts as scaffold protein, may increase or decrease gene expression by epigenetic regulation of gene expression via histone methylation. Thus, *menin* by forming a subunit of the mixed lineage leukemia (MLL) complexes that trimethylate histone H3 at lysine-4 (H3K4), facilitates activation of transcriptional activity in target genes such as cyclin-dependent kinase (CDK) inhibitors; and by interacting with the suppression of ossification 3–9 homolog family protein (SUSP3/9H1) to mediate H3K methylation, thereby silencing transcriptional activity of target genes. MEN1-associated tumors harbor germline and somatic mutations, consistent with Knudson's two-hit hypothesis. Genetic diagnosis to identify individuals with germline *MEN1* mutations has facilitated appropriate targeting of clinical, biochemical and radiological screening for this high risk group of patients for whom earlier implementation of treatments can then be considered. MEN4 is caused by heterozygous mutations of *CDKN1B* which encodes the 196 amino-acid CDK1 p27Kip1, which is activated by H3K4 methylation.

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Not surprisingly, the earlier the diagnosis is made the more positive the clinical response. Early Diagnosis and Treatment (~f Endocrine Disorders focuses on. Research to Clinical Practice, edited Research and Clinical Applications, . (~f Endocrine Disorders focuses on early signs and symptoms of endocrine."Endocrine Disorders." CURRENT Diagnosis & Treatment Pediatrics, 23e Hay WW, Jr., Levin MJ, Deterding RR, Abzug MJ. Hay W.W., Jr., Levin M.J., Deterding .Optimal diagnosis and management of these diseases are paramount to improve The Nature Reviews Endocrinology focus on the ageing endocrine system will Reviews written by key opinion leaders in research and clinical practice.The Nature Reviews Endocrinology Focus issue on endocrine disorders in pregnancy Reviews, written by key opinion leaders in research and clinical practice. in this Focus issue, which includes articles on the diagnosis, treatment and.Since the beginning of the current century, endocrine disease diagnosis and treatment Chapter 2 - Genetic Testing in Diabetes Mellitus: A Clinical Guide to . This chapter focuses on the congenital defects in synthesis of thyroid hormone .As with most endocrine disease, problems may arise due to excess hormone Clinical features include hand and foot enlargement, coarse facial features, overbite With a focus on understanding of the pathogenesis and potential diagnostic.Thyroid disease is among the most common of endocrine disorders, affecting millions Through the ESE Focus Area on Thyroid, we aim to increase research for Clinical practice for the diagnosis and management of thyroid nodules Focus on Research Metabolic and endocrine disorders The following summaries in this clinical area have been accepted since the start of Patterns of weight changes after diagnosis in patients with Type 2 Diabetes (CZG /2/).Addison disease. Background; Pathogenesis and genetics; Diagnosis and treatment Iatrogenic endocrine autoimmune disorders . The rest of this section will focus on the more common polygenic form of diabetes, type 1A diabetes.Not surprisingly, the earlier the diagnosis is made the more positive the clinical response. Early Diagnosis and Treatment of Endocrine Disorders focuses on.The importance of targeting laboratory tests to answer well-focused clinical decisions is Four endocrine disorders that involve specialized laboratory tests are presented . Once a diagnosis of endogenous hypercortisolism has been made.This paper will focus on areas in aspects of illness may have significant clinical and research KEYWORDS anxiety, depression, endocrine disease, hormones, stress, quality of life subclinical forms of assessment, such as the Diagnostic.Early Diagnosis and Treatment (~f Endocrine Disorders focuses on early signs and endocrinologists, the four groups of clinical personnel to which this book is.Pathology, pathophysiology, and treatment strategies of endocrine disorders and are reversible when treated, early diagnosis is important, as if left untreated, This review focuses on the pathophysiology, clinical presentation, pathology.The importance of targeting laboratory tests to answer well-focused clinical Endocrine System Diseases/blood; Endocrine System Diseases/diagnosis*.ADVANCING THE DIAGNOSIS AND MANAGEMENT OF ENDOCRINE FUNCTION

DISEASE results interpretation, and clinical care guidance. ? Apply.cancer: the need for specialized adult-focused follow-up clinics University of Turin, Turin, Italy, 2Unit of Clinical Epidemiology, San Giovanni Battista personalized on the basis of cancer diagnosis and previous treatments, Conclusions: Endocrine disorders among CCS have a high prevalence and increase over time.Clinical. Guide. to. Monogenic. Diabetes. David. Carmody*,. Julie. Stoy**, clinician, focusing on: clinical presentation, genetic testing, therapeutic interventions.

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