

Classic congenital adrenal hyperplasia



CYP21A1 gene mutation causes 21- α -hydroxylase deficiency



As a result, the adrenal glands can't make enough cortisol and aldosterone



Pituitary gland releases excess ACTH to try and stimulate cortisol production

Excess ACTH release stimulates diffuse enlargement of the adrenal glands



Greater risk of adrenal tumors



Adrenal glands have a 'cerebriform' appearance on CT

Enlarged adrenals produce excess androgens leading to hormonal effects on phenotype



Chen

Adrenal Hyperplasia

Robert Holmes Greene, Harlow Brooks

Adrenal Hyperplasia:

Congenital Adrenal Hyperplasia Peter C. Hindmarsh, Kathy Geertsma, 2017-04-19 Congenital Adrenal Hyperplasia A Comprehensive Guide addresses how hydrocortisone works what can go wrong and how to correct it also explaining why the timing of doses and measurement of cortisol from a dose is extremely important The book provides an in depth analysis of this disorder for pediatric endocrinologists and primary care providers allowing them to help patients with an updated model of care and appropriate treatment Patients and family members will benefit from the trend forward information that will empower them to approach their healthcare providers with the expectation of receiving individualized care and treatment for this disorder Outlines the basics of congenital adrenal hyperplasia and its interrelation with hormones and bodily functions Presents the known cognitive and emotional aspects of the disease Reviews multidisciplinary management as well as post treatment management of the disease *Primary Care of the Child With a Chronic Condition E-Book* Patricia Jackson Allen, Judith A. Vessey, Naomi Schapiro, 2009-06-25 Written by nurse practitioners for nurse practitioners this one of a kind resource provides the expert guidance you need to provide comprehensive primary care to children with special needs and their families It addresses specific conditions that require alterations in standard primary care and offers practical advice on managing the major issues common to children with chronic conditions A consistent format makes it easy to locate essential information on each condition Plus valuable resources help you manage the issues and gaps in health care coverage that may hinder quality care This is the only book authored by Nurse Practitioners that focuses on managing the primary health care needs of children with chronic conditions More than 60 expert contributors provide the most current information available on specific conditions Comprehensive summary boxes at the end of all chronic conditions chapters provide at a glance access to key information Resource lists at the end of each chronic condition chapter direct you to helpful websites national organizations and additional sources of information that you can share with parents and families Updated references ensure you have access to the most current evidence based coverage of the latest research findings and management protocols Four new chapters Celiac Disease Eating Disorders Muscular Dystrophy and Obesity keep you up to date with the latest developments in treating these conditions Autism content is updated with the latest research on autism spectrum disorders including current methods of evaluation identification and management Coverage of systems of care features new information on how to help families obtain high quality and cost effective coordinated services within our complex health care system Easy to find boxes in the chronic conditions chapters summarize important information on treatment associated problems clinical manifestations and differential diagnosis *Encyclopedia of Endocrine Diseases*, 2018-09-12 Encyclopedia of Endocrine Diseases Second Edition Five Volume Set comprehensively reviews the extensive spectrum of diseases and disorders that can occur within the endocrine system It serves as a useful and comprehensive source of information spanning the many and varied aspects of the endocrine end metabolic system Students will find a concise description of the physiology

and pathophysiology of endocrine and metabolic functions as well as their diseases Each article provides a comprehensive overview of the selected topic to inform a broad spectrum of readers from advanced undergraduate students to research professionals Chapters explore the latest advances and hot topics that have emerged in recent years such as the molecular basis of endocrine and metabolic diseases mutations epigenetics signaling the pathogenesis and therapy of common endocrine diseases e g diabetes and endocrine malignancies new technologies in endocrine research new methods of treatment and endocrine toxicology disruptors Covers all aspects of endocrinology and metabolism Incorporates perspectives from experts working within the domains of biomedicine e g physiology pharmacology and toxicology immunology genetics and clinical sciences to provide readers with reputable multi disciplinary content from domain experts Provides a one stop resource for access to information as written by world leading scholars in the field with easy cross referencing of related articles to promote understanding and further research

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