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Perfect for residents, pediatricians, practitioners, or parents seeking further information, Smith's Recognizable Patterns of Human Deformation provides evidence-based management for a range of common pediatric problems affecting the limbs and craniofacial region. The only source devoted to the diagnoses and management of birth defects resulting from mechanical forces, this reference supplies ...

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Smith's Recognizable Patterns of Human Malformation, 5th ed, K.L. Jones, Philadelphia, Pa, Saunders, 1997, hard cover, 357 pp, \$79. Wayne Stuberger, PhD, PT, PCS, Munroe-Meyer Institute, University of Nebraska Medical Center. Smith's Recognizable Patterns of Human Malformation provides a comprehensive overview of malformation syndromes. Many malformations have a genetic etiology so this text is a reference for genetic disorders in addition to malformation syndromes that to date have not been ...

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Perfect for residents, pediatricians, practitioners, or parents seeking further information, Smith's Recognizable Patterns of Human Deformation provides

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evidence-based management for a range of common pediatric problems affecting the limbs and craniofacial region. The only source devoted to the diagnoses and management of birth defects resulting from mechanical forces, this reference supplies the essential guidance needed for timely intervention and effective treatment. Examines the initial clinical approach to suspected deformation problems, and then walks you through pathogenesis, diagnostic features, management, prognosis, and counseling for each condition. Addresses a full range of lower extremity deformations; joint dislocations; nerve palsies; chest and spinal deformations; head and neck deformations; craniosynostosis and cranial bone variations; problems associated with abnormal birth presentation, birth palsies, and procedure-related defects; infant head shape variations; and torticollis. Distinguish deformations from malformations for appropriate management. Each chapter utilizes four consistent sections - Genesis, Features, Management and Prognosis, and Differential Diagnosis - to provide concise yet comprehensive information on 50 common pediatric conditions. These chapters are available for individual purchase or download to serve as educational guides for parents regarding evidence-based management of these conditions. Diagnosis and management of common pediatric orthopedic conditions is covered in detail. Updated discussion of Sudden Infant Death Syndrome brings a new focus to the important topic of infant sleeping environments. New before-and-after illustrations and detailed discussions focus on cranial-orthotic molding helmets and the surgical correction of craniosynostosis. Provides evidence-based management recommendations on common fetal complications such as oligohydramnios, pulmonary hypoplasia, and uterine structural abnormalities, and discusses current management techniques for each. Selected references at the end of each chapter provide further recent information regarding each of these topics. Offers essential information to a range of professionals, including neonatologists, pediatricians, family practitioners, nurses, physical and occupational therapists, rehabilitative specialists, pediatric nurse practitioners, and residents in all fields. Expert Consult eBook version included with purchase. This enhanced eBook experience allows you to search all of the text, figures, and references in the book on a variety of devices.

2014 BMA Medical Book Awards 1st Prize Award Winner in Illustrated Book category and Highly Commended in Paediatrics category! Smith's Recognizable Patterns of Human Malformation has long been known as the source to consult on multiple malformation syndromes of environmental and genetic etiology as well as recognizable disorders of unknown cause. This esteemed medical reference book provides you with complete and authoritative, yet accessible guidance to help accurately diagnose these human disorders, establish prognoses, and provide appropriate management and genetic counseling. Recognize the visual signs of each environmental and genetic abnormality by consulting more than 1,500 full-color photographs and illustrations, many from the personal collections of Drs. Smith and Jones. Find all the answers you need about normal and abnormal morphogenesis, minor anomalies and their relevance, clinical approaches to specific diagnoses, and normal standards of measurement for the entire spectrum of human malformation syndromes. Efficiently identify genetic disorders in your patients with the inclusion of nearly 20 recently recognized entities/syndromes, as well as new chapters on Microdeletions and Microduplication Syndromes. Available digitally for the first time! You can now access Smith's as an eBook or online at Expert Consult.

Long known as the source to consult for guidance on diagnosis, prognosis, plan management, and genetic counseling, this easy-to-use reference focuses on patterns of human defects caused by inborn errors in morphogenesis as opposed to defects caused by mechanical problems. Each chapter includes a description of the specific abnormality--including occasional associated abnormalities--natural history, etiology, and references. Full color.

The most recent update to one of the most essential references on medical genetics Cassidy and Allanson's Management of Genetic Syndromes, 4th Edition is the latest version of a classic text in medical genetics. With newly covered disorders and cutting-edge, up-to-date information, this resource remains the most crucial reference on the management of genetic syndromes for students, clinicians, and researchers in the field of medical genetics. The 4th edition includes current information on the identification of genetic syndromes (including newly developed diagnostic criteria), the genetic basis (including diagnostic testing), and the routine care and management for more than 60 genetic disorders. Each, "expert authored", chapter includes sections on: Incidence Diagnostic criteria Etiology, pathogenesis and genetics Diagnostic testing Differential diagnosis Manifestations and Management (by system) The book focuses on genetic syndromes, primarily those involving developmental disabilities and congenital defects. The chapter sections dealing with Manifestations and Management represents the centerpiece of each entry and is unmatched by other genetic syndrome references. Management of Genetic Syndromes is perfect for medical geneticists, genetic counselors, primary care physicians and all health care professionals seeking to stay current on the routine care and management of individuals with genetic disorders.

Often, information in review books can raise as many questions as it answers. This interferes with the study process, because the learner must either look up additional information or skip ahead without truly comprehending what he or she has read. As an alternative, Pediatric Genetics and Inborn Errors of Metabolism: A Practically Painless Review presents bite-size chunks of information that can be read and processed rapidly, helping learners to stay active while studying and to pick up new information the first time they read it. This book's question and answer format allows for self-testing or

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study with a partner or a group. The format also facilitates dipping into the book during a few minutes of downtime at the hospital or office. *Pediatric Genetics and Inborn Errors of Metabolism: A Practically Painless Review* is a quick and easy way to master these tricky topics and is suitable for those studying for the pediatric board exam, practicing physicians brushing up their skills and any busy clinician who wants to learn more about these topics while on the go.

In a field where even experts may find that years have elapsed since they last encountered a child with a given disorder, it is essential for the clinician to have a comprehensive source of practical and highly illustrated information covering the whole spectrum of metabolic disease to refer to. The content is divided into sections of related disorders, including disorders of amino acid metabolism, lipid storage disorders, and mitochondrial diseases for ease of reference, with an introductory outline where appropriate summarizing the biochemical features and general management issues. Within the sections, each chapter deals with an individual disease, opening with a useful summary of major phenotypic expression including clear and helpful biochemical pathways, identifying for the reader exactly where the defect occurs. Throughout the book, plentiful photographs, often showing extremely rare disorders, are an invaluable aid to diagnosis. Key Features • Fully updated to incorporate all new developments in the field • Brand new chapters cover methylmalonic aciduria of ACSF3 deficiency, branched chain keto acid dehydrogenase deficiency, serine deficiencies, purine nucleoside phosphorylase deficiency, antequitin deficiency, and others • Excellent and detailed clinical descriptions, with numerous valuable hints and suggestions for management • Helpful explanatory algorithms and decision trees, and high-quality illustrative material including biochemical pathways and an unrivaled photographic collection, which enhance clinical applicability The fourth edition of this highly regarded book, authored by two of the foremost authorities in pediatric metabolic medicine, continues to provide incomparable insight into the problems associated with metabolic diseases and remains invaluable to pediatricians, geneticists, and general clinicians worldwide.

A highly illustrated field guide to dysmorphology, a key area of clinical genetics and a vital competency for all clinicians. Oxford Genetics is a comprehensive, cross-searchable collection of resources offering quick and easy access to Oxford University Press's prestigious genetics texts. Joining Oxford Medicine Online these resources offer students, specialists and clinical researchers the best quality content in an easy-to-access format. Online only benefits include downloading images and figures to PowerPoint and downloading chapters to PDF.

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