1. Record Nr. UNINA9910845499603321 Autore Mundlos Stefan Titolo Limb Malformations [[electronic resource]]: An Atlas of Genetic Disorders of Limb Development / / by Stefan Mundlos, Denise Horn Pubbl/distr/stampa Berlin, Heidelberg: .: Springer Berlin Heidelberg: .: Imprint: Springer. , 2014 **ISBN** 3-540-95928-9 Descrizione fisica 1 online resource (274 p.) Disciplina 599935 610 611.01816 616.0757 Soggetti Human genetics Orthopedic surgery **Pediatrics** Radiology, Medical **Human Genetics** Surgical Orthopedics Imaging / Radiology Lingua di pubblicazione Inglese **Formato** Materiale a stampa Livello bibliografico Monografia Note generali Description based upon print version of record. Nota di bibliografia Includes bibliographical references at the end of each chapters and index. Nota di contenuto ""Foreword""; ""Preface""; ""Acknowledgements""; ""Table of Contents""; ""I Introduction""; ""1 Development of the Limbs""; ""2 Clinical Assessment and Anthropometry of the Limbs""; ""3 Radiology of the Limb ""; ""4 Surgical Management Principles for Congenital Upper Limb Malformations""; ""II Disorders: Polydactyly Isolated""; ""5 Preaxial Polydactyly""; ""6 Postaxial Polydactyly ""; ""7 Greig Cephalopolysyndactyly Syndrome ""; ""8 Triphalangeal Thumb-Polydactyly Syndrome ""; ""8 Synpolydactyly ""; ""10 Laurina€?Sandrow Syndrome""; ""III Disorders: Polydactyly Associated"" ""11 Ellisa€?Van Creveld Syndrome"""12 Short Rib-Polydactyly

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Sommario/riassunto

One aim of this atlas is to present a comprehensive overview of limb malformation phenotypes in order to provide the clinician with a tool that facilitates the diagnostic process. With the enormous advances in molecular and developmental biology, the genetic basis of many limb malformations and their relationship to each other has been elucidated. Thus, a further aim of this atlas is to provide the reader with a basic understanding of the molecular pathology of these conditions. The book is extensively illustrated with clinical photos and radiographs of conditions or groups of related conditions. In addition, a concise description of the conditions is provided featuring structured information on "Synonyms", "Major clinical findings", "Genetic transmission", "Differential diagnosis", "Molecular Pathology", and references to Mendelian Inheritance in Man (OMIM). The book is designed for medical geneticists, radiologists, pediatricians, hand surgeons, orthopedic surgeons, as well as medical personnel and other physicians involved in the evaluation and treatment of patients with abnormal limbs.