Bone Dysplasias: An Atlas of Genetic Disorders of Skeletal Development

Jurgen W. Spranger, Paula W. Brill, Andrew K. Poznanski

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Many advances have been made in understanding skeletal dysplasias since the first edition of this classic text appeared in 1974. The diagnostic process has been refined, many new disorders have been recognized, and the molecular aspects in many cases have been elucidated. The second edition has been completely renovated, with the help of two new co-authors, to incorporate these advances. The book's format is similar to the original but the number of conditions covered has almost doubled and molecular information has been added wherever available. The number of figures has been increased to the limit of economic wisdom. As in the first edition, the illustrations have been selected and sequenced to illustrate both the degree of variability of a given disorder and its changs with age.

This book is designed for physicians involved in the evaluation and treatment of patients with skeletal dysplasias, including radiologists, medical geneticists, pediatricians, and orthopedic surgeons. Its main goal is to assist in the diagnosis of specific conditions and the care of affected individuals. Though mutations of specific genes can produce dysplasias with very different phenotypes and prognoses, the primarily clinical aim of this book dictated a phenotypic classification in general, with compromises on etiologic grounds where necessary. Since the attempt to diagnose a skeletal dysplasia from single signs, alone or in cominations, is fraught with errors, the authors focus on basic patterns of skeletal abnormalities.

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