

Management of Genetic Syndromes



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Although genetic disorders and syndromes are usually considered rare, their aggregate frequency makes them an inescapable part of medicine. It is crucial that affected individuals, their families, and their primary and specialty care physicians have an accurate and reliable resource so that the best care possible can be provided. Furthermore, with the remarkable surge of information that has become available due to networking of researchers and families, the development of support groups, and the availability of research reports and data, it is essential for one reference to contain all of this material organized in an easy-to-use manner. Management of Genetic Syndromes fills this vital need. Through an in-depth understanding of a specific disorder, physicians and parents can help patients reach their full potential, benefit from useful therapies, and avoid complications. Management of Genetic Syndromes is written by expert authors and offers a practical approach to the evaluation and management of the 30 most common genetic syndromes from birth to adulthood. It provides information about the spectrum of variation that can occur, risk of recurrence in the affected individual's siblings and offspring, and whether prenatal diagnosis and diagnostic testing are available. Management of Genetic Syndromes incorporates a wealth of knowledge and experience with each chapter containing detailed coverage of: Incidence Etiology and pathogenesis Natural history Diagnostic criteria Diagnostic testing Differential diagnosis Evaluation Management Reference and resources This essential book will offer medical geneticists, clinical geneticists, human geneticists, pediatricians, and other health care professionals insight into treatment and management, as well as the development of new interventions and therapies.

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