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Atlas of Genetic Diagnosis and Counseling. Vol.1

By Chen, Harold

Book Condition: New. Publisher/Verlag: Springer, Berlin | Dr. Chen shares his almost 40 years of clinical genetics practice in a comprehensive pictorial atlas of almost 250 genetic disorders, malformations, and malformation syndromes. The author provides a detailed outline for each disorder, describing its genetics, basic defects, clinical features, diagnostic tests, and counseling issues, including recurrence risk, prenatal diagnosis, and management. Numerous color photographs of prenatal ultrasounds, imagings, cytogenetics, and postmortem findings illustrate the clinical features of patients at different ages, patients with varying degrees of severity, and the optimal diagnostic strategies. The disorders cited are supplemented by case histories and diagnostic confirmation by cytogenetics, biochemical, and molecular techniques, when available. The Atlas of Genetic Diagnosis and Counseling will help all physicians to understand and recognize genetic diseases and malformation syndromes and better evaluate, counsel, and manage affected patients. In this new edition, 47 additional genetic disorders are added, as well as extensive updates made to the previous disorders. New illustrations, as previous edition, will be supplemented by case and family history, clinical features, and laboratory data, especially molecular confirmation. | Acardia Achondrogenesis Achondroplasia Adams-Oliver syndrome Agnathia Aicardi syndrome Alagille syndrome Albinism Alpha thalassemia-mental retardation (ATR-X) syndrome Ambiguous genitalia Amniotic deformity,...



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