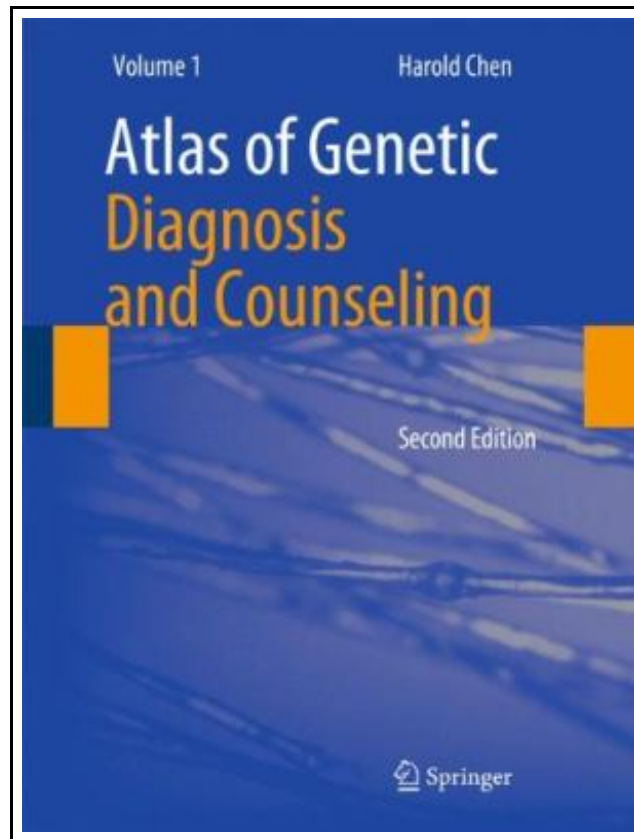


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ATLAS OF GENETIC DIAGNOSIS AND COUNSELING. VOL.1



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, adhesions, mutilations (ADAM) syndrome Androgen insensitivity syndrome Angelman syndrome Apert syndrome Aplasia cutis congenita Arthrogryposis multiplex congenita Asphyxiating thoracic dystrophy Ataxia telangiectasia Atelosteogenesis Autism Bannayan-Riley-Ruvalcaba syndrome Beckwith-Wiedemann syndrome Behcet disease Biotinidase deficiency Bladder exstrophy Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) Body stalk anomaly Brachydactyly Branchial cleft cyst Calcinosis cutis Campomelic dysplasia Carpenter syndrome Cat-eye syndrome Celiac disease Cerebral palsy Cerebro-costo-mandibular syndrome Charcot-Marie-Tooth disease CHARGE association Cherubism Chiari malformation Chondrodysplasia punctata Chromosome abnormalities in pediatric solid tumors Cleft lip/palate Cleidocranial dysplasia Cloacal exstrophy Clubfoot (talipes equinovarus) Collodion baby Congenital adrenal hyperplasia Congenital cutis laxa Congenital cytomegalovirus infection Congenital generalized lipodystrophy...



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