1. Record Nr. UNINA9910254482103321

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Titolo	Atlas of Genetic Diagnosis and Counseling [[electronic resource] /] / by Harold Chen
Pubbl/distr/stampa	New York, NY : , : Springer New York : , : Imprint : Springer, , 2017
ISBN	1-4939-2401-X
Edizione	[3rd ed. 2017.]
Descrizione fisica	1 online resource (1242 illus., 1050 illus. in color. eReference.)
Disciplina	616.042
Soggetti	Pathology Pediatrics Neurology Neurology
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Nota di contenuto	Acardia -- Achondrogenesis -- Achondroplasia -- Adams-Oliver Syndrome -- Agnathia -- Aicardi Syndrome -- Alagille Syndrome -- Albinism -- Alpha-Thalassemia X-linked Mental Retardation Syndrome -- Ambiguous Genitalia -- Amniotic Deformity, Adhesions, Mutilations (ADAM) Complex -- 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 -- Body Stalk Anomaly -- Brachydactyly -- Branchial Cleft Anomalies -- Calcinosis Cutis -- Campomelic Dysplasia -- Carpenter Syndrome -- Cat Eye Syndrome -- Celiac Disease -- Cerebral Palsy -- Cerebro-Costo-Mandibular Syndrome -- Charcot-Marie-Tooth Disease -- CHARGE Syndrome -- Cherubism -- Chiari Malformation -- Chondrodysplasia Punctata -- Chromosome Abnormalities in Pediatric Solid Tumors -- Cleft Lip and/or Cleft Palate -- Cleidocranial Dysplasia -- Cloacal Exstrophy -- Clubfoot -- Collodion Baby -- Congenital Adrenal Hyperplasia -- Congenital Cutis Laxa -- Congenital Cytomegalovirus Infection -- Congenital Generalized Lipodystrophy --

Congenital Hemihyperplasia -- Congenital Hydrocephalus -- Congenital Hypothyroidism -- Congenital Muscular Dystrophy -- Congenital Toxoplasmosis -- Conjoined Twins -- Corpus Callosum Agenesis/Dysgenesis -- Craniometaphyseal Dysplasia -- Cri-Du-Chat Syndrome -- Crouzon Syndrome -- Cutaneous Vasculitis -- Cutis Marmorata Telangiectatica Congenita -- Cystic Fibrosis -- Dandy-Walker Malformation -- De Lange Syndrome -- Del(18p) Syndrome -- Del(22q11.2) Syndrome -- Del(Yq) Syndrome -- Diabetic Embryopathy -- Down Syndrome -- Duncan Syndrome -- Dyschondrosteosis -- Dysmelia -- Dysplasia Epiphysealis Hemimelica -- Dystonia -- Dystrophinopathies -- Ectrodactyly-Ectodermal Dysplasia-Clefting (EEC) Syndrome -- Ehlers-Danlos Syndrome -- Ellis-van Creveld Syndrome -- Enchondromatosis -- Epidermolysis Bullosa -- Epidermolytic Palmoplantar Keratoderma -- Faciogenital (Faciodigitogenital) Dysplasia -- Facioscapulohumeral Muscular Dystrophy -- Familial Adenomatous Polyposis -- Familial Hyperlysinemia -- Familial Mediterranean Fever -- Familial Patella Instability -- Familial Spastic Paraplegia -- Fanconi Anemia -- Feingold Syndrome -- Femoral Hypoplasia - Unusual Facies Syndrome -- Fetal Akinesia Deformation Sequence -- Fetal Alcohol Spectrum Disorders -- Fetal Hydantoin Syndrome -- Fibrodysplasia Ossificans Progressiva -- Fibular Hemimelia -- Finlay-Marks Syndrome -- Floppy Infant -- Fragile X Syndrome -- Fraser Syndrome -- Freeman-Sheldon Syndrome -- Friedreich Ataxia -- Frontonasal Dysplasia -- Galactosemia -- Gastroschisis -- Gaucher Disease -- Generalized Arterial Calcification of Infancy -- Genitopatellar Syndrome -- Giant Congenital Melanocytic Nevi -- Gilbert Syndrome -- Glucose-6-Phosphate Dehydrogenase Deficiency -- Glycogen Storage Disease, Type 2 -- Goldenhar Syndrome -- Gorlin Syndrome -- Greig Cephalopolysyndactyly Syndrome -- Growth Hormone Deficiency -- Hallermann-Streiff Syndrome -- Harlequin Ichthyosis -- Hemangiomas of Infancy -- Hemophilia A -- Hereditary Hearing Loss -- Hereditary -- Hemochromatosis -- Hereditary Multiple Exostoses -- Hereditary Sensory and Autonomic Neuropathies -- Herlyn-Werner-Wunderlich Syndrome -- Holoprosencephaly -- Holt-Oram Syndrome -- Huntington Disease -- Hydranencephaly -- Hydrolethalus Syndrome -- Hydrops Fetalis -- Hyper-IgE Syndrome -- Hypertrophic Cardiomyopathy (HCM) -- Hypochondroplasia -- Hypoglossia-Hypodactylia Syndrome -- Hypohidrotic Ectodermal Dysplasia -- Hypomelanosis of Ito -- Hypophosphatasia -- Hypopituitarism -- I(1p), I(1q) Syndrome -- Idic(Yq) Syndrome -- Incontinentia Pigmenti -- Infantile Myofibromatosis -- Ivemark Syndrome -- Jarcho-Levin Syndrome -- Joubert Syndrome -- Kabuki Syndrome -- Kasabach-Merritt Syndrome -- KID Syndrome -- Klinefelter Syndrome -- Klippel-Feil Syndrome -- Klippel-Trenaunay Syndrome -- Kniest Dysplasia -- Larsen Syndrome -- LEOPARD Syndrome -- Lesch-Nyhan Syndrome -- Lethal Multiple Pterygium Syndrome -- Loeys-Dietz Syndrome -- Lowe Syndrome -- Lymphangiomas and Lymphangiomatosis -- M#xf6;bius Syndrome -- Macrodactyly -- Marfan Syndrome -- McCune-Albright Syndrome -- Meckel-Gruber Syndrome -- Megalencephalic Leukoencephalopathy with Subcortical Cysts -- Menkes Disease -- Metachromatic Leukodystrophy -- Miller-Dieker Syndrome -- Mitochondrial Leber Hereditary Optic Neuropathy -- Mitochondrial Myopathy -- Mowat-Wilson Syndrome -- Mucolipidosis 2 -- Mucolipidosis 3 -- Mucopolysaccharidosis 1 (MPS 1) -- Mucopolysaccharidosis 2 -- Mucopolysaccharidosis 3 -- Mucopolysaccharidosis 4 -- Mucopolysaccharidosis 6 -- Multiple Endocrine Neoplasia Syndromes -- Multiple Epiphyseal Dysplasia --

Multiple Pterygium Syndrome -- Myotonic Dystrophy Type 1 -- Nager Acrofacial Dysostosis -- Nail-Patella Syndrome -- Nasal Obstruction in Neonates and Children -- Neonatal Herpes Simplex Infection -- Nephrogenic Diabetes Insipidus -- Netherton Syndrome -- Neu-Laxova Syndrome -- Neural Tube Defects -- Neurofibromatosis 1 -- Neurofibromatosis 2 -- Niemann-Pick Disease -- Noonan Syndrome -- Oblique Facial Cleft Syndrome -- Oligohydramnios Sequence -- Omphalocele -- Opitz Trigonocephaly (C) Syndrome -- Oral-Facial-Digital Syndrome -- Osteogenesis Imperfecta -- Osteogenesis Imperfecta Ehlers-Danlos Overlap Syndrome -- Osteopetrosis -- Osteopoikilosis -- Otopalatodigital Spectrum Disorders -- Pachyonychia Congenita -- Pallister-Killian Syndrome -- Peutz-Jeghers Syndrome -- Phenylketonuria -- Pierre Robin Sequence -- Polycystic Kidney Disease, Autosomal Dominant Type -- Polycystic Kidney Disease: Autosomal Recessive Type -- Popliteal Pterygium Syndrome -- Prader-Willi Syndrome -- Primary Microcephaly -- Progeria -- Prune Belly Syndrome -- Pseudoachondroplasia -- R(18) Syndrome -- Radioulnar Synostosis -- Retinoid Embryopathy -- Rett Syndrome -- Rickets -- Rigid Spine Syndrome -- Roberts Syndrome -- Robinow Syndrome -- Rubinstein-Taybi Syndrome -- Saethre-Chotzen Syndrome -- Sagittal Craniosynostosis Associated with Chromosome Abnormalities with a Brief Review on Craniosynostosis -- Schizencephaly Schmid Metaphyseal Chondrodysplasia -- Seckel Syndrome -- Severe Combined Immune Deficiency -- Short Rib-Polydactyly Syndromes -- Sickle Cell Disease -- Silver-Russell Syndrome -- Sirenomelia -- Smith-Lemli-Opitz Syndrome -- Smith-Magenis Syndrome -- Sotos Syndrome -- Spinal Muscular Atrophy -- Spondyloepiphyseal Dysplasia -- Stickler Syndrome -- Sturge-Webber Syndrome -- Symphalangism -- Tay-Sachs Disease -- Tetrasomy 9p Syndrome -- Thalassemia -- Thanatophoric Dysplasia -- Thrombocytopenia-Absent Radius Syndrome -- Treacher-Collins Syndrome -- Trimethylaminuria -- Triploidy -- Trismus-Pseudocamptodactyly Syndrome -- Trisomy 13 Syndrome -- Trisomy 18 Syndrome -- Trisomy 8 Mosaicism Syndrome -- Tuberous Sclerosis -- Turner Syndrome -- Twin-Twin Transfusion Syndrome -- Tyrosinemia -- Ulnar-Mammary Syndrome -- Urofacial Syndrome -- VATER (VACTERL) Association -- Von Hippel-Lindau Disease -- Waardenburg Syndrome -- Weill-Marchesani Syndrome -- Williams Syndrome -- Winchester syndrome -- Wolf-Hirschhorn Syndrome -- X-Linked Agammaglobulinemia -- X-Linked Ichthyosis -- XX Male -- XXX Syndrome -- XXXXX Syndrome -- XXXXY Syndrome -- XY Female -- XYY Syndrome.

Sommario/riassunto

Dr. Harold Chen shares his almost 50 years of clinical genetics practice in this new edition of a comprehensive pictorial atlas, featuring almost 290 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. Since the publication of the previous edition in 2012, the atlas has been widely accepted and used in light of rapid progress in genetic and gnomic information. In this new edition, additional genetic disorders are added, as well as extensive updates to the previous disorders with

new illustrations, supplemented by case and family history, clinical features, and laboratory data, especially molecular confirmation if available. The atlas is written in outline format for ease of use. Atlas of Genetic Diagnosis and Counseling, Third Edition is of great value to medical geneticists, genetic counselors, pediatricians, neonatologists, developmental pediatricians, perinatologists, obstetricians, neurologists, pathologists, and any physicians and health care professionals caring for handicapped children such as craniofacial surgeons, plastic surgeons, otolaryngologists, and orthopedists. It is the definitive volume for helping all physicians to understand and recognize genetic diseases and malformation syndrom es and better evaluate, counsel, and manage affected patients.