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Nota di contenuto	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 -- Congenital hemihyperplasia (congenital hemihypertrophy) -- Congenital hydrocephalus -- Congenital hypothyroidism -- Congenital muscular dystrophy -- Congenital toxoplasmosis -- Conjoined twins -- Corpus-callosum agenesis/dysgenesis -- Craniometaphyseal dysplasia -- Cri-du-chat syndrome -- Crouzon syndrome -- 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 (X-linked lymphoproliferative disease) -- Dyschondrosteosis/Langer mesomelic dysplasia -- Dysmelia (limb deficiency/reduction) -- Dysplasia epiphysealis hemimelica -- Dystonia -- Dystrophinopathies -- EEC syndrome -- Ehlers-Danlos syndrome -- Ellis-van Creveld syndrome -- Enchondromatosis -- Epidermolysis bullosa -- Epidermolytic palmoplantar keratoderma -- Faciogenital (Aarskog) syndrome -- Facioscapulohumeral muscular dystrophy (FSH) -- Familial adenomatous polyposis syndrome -- Familial hyperlysinemia -- Familial Mediterranean fever -- Fanconi anemia -- Femoral hypoplasia-unusual facies syndrome -- Fetal akinesia syndrome -- Fetal alcohol syndrome -- Fetal hydantoin syndrome -- Fibrodysplasia ossificans progressiva -- Finlay-Marks syndrome -- Floppy infant -- Fragile X syndrome -- Fraser syndrome -- Freeman-Sheldon (whistling face) syndrome -- Friedreich ataxia -- Frontonasal dysplasia -- Galactosemia -- Gastroschisis -- Gaucher disease -- Generalized arterial calcification -- Genitopatellar syndrome -- Giant congenital melanocytic nevi (giant congenital nevi) -- Glucose-6-phosphate dehydrogenase deficiency -- Glycogen storage disease, type II (Pompe) -- Goldenhar syndrome -- Gorlin (nevoid basal cell carcinoma) syndrome -- Greig cephalopolysyndactyly syndrome -- Hallermann-Streiff syndrome -- Harlequin fetus -- Hemophilia A -- Hereditary hearing loss -- Hereditary hemochromatosis -- Hereditary multiple exostosis -- Herlyn-Werner-Wunderlich syndrome -- Holoprosencephaly -- Holt-Oram syndrome -- Huntington disease -- Hydrolethalus syndrome -- Hydrops fetalis -- Hyper-IgE syndrome -- Hypochondroplasia -- Hypoglossia-hypodactyly (oromandibular limb hypogenesis) syndrome -- Hypohidrotic ectodermal dysplasia -- Hypomelanosis of Ito -- Hypophosphatasia -- Hypopituitarism -- I(1p), I(1q) syndrome -- Isodic(Yq) syndrome -- Incontinentia pigmenti -- Infantile myofibromatosis -- Ivemark syndrome -- Jarcho-Levin syndrome -- Joubert syndrome -- Kabuki syndrome -- Kassback-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 -- Marfan syndrome -- McCune-Albright syndrome -- Meckel-Gruber syndrome -- Megalencephalic leukoencephalopathy with subcortical cysts (van der Knaap disease) -- Menkes disease -- Metachromatic leukodystrophy -- Miller-Dieker syndrome -- Mitochondrial Leber hereditary optic neuropathy -- Mobius syndrome -- Mowat-Wilson Disease -- Mucolipidosis II (I-cell disease) -- Mucolipidosis III (pseudo-Hurler Polydystrophy) -- MPS I (Hurler syndrome) -- MPS II (Hunter syndrome) -- MPS III (Sanfilippo syndrome) -- MPS IV (Morquio syndrome) -- MPS VI (Maroteaux-Lamy syndrome) -- Multiple endocrine neoplasia Syndrome -- Multiple epiphyseal dysplasia -- Multiple pterygium syndrome -- Myotonic dystrophy --

Nail-Patella Syndrome (hereditary Osteo-onychodysplasia) -- Neonatal Herpes simplex infection -- Nephrogenic diabetes insipidus -- Netherton syndrome -- Neu-Laxova syndrome -- Neural tube defects -- Neurofibromatosis 1 -- Neurofibromatosis 2 -- Noonan syndrome -- Oblique facial cleft syndrome -- Oligohydramnios sequence -- Omphalocele -- Oro-Facial-Digital Syndrome -- Osteogenesis imperfecta -- Osteopetrosis -- Osteopoikilosis -- Otopalatodigital spectrum disorders -- Pachyonychia congenita -- Pallister-Killian syndrome -- Phenylketonuria (PKU) -- Pierre Robin sequence -- Polycystic kidney disease, AD form -- Polycystic kidney disease, AR form -- Popliteal pterygium syndrome -- Prader-Willi syndrome -- Progeria -- Prune belly syndrome -- Pseudoachondroplasia -- R(18) syndrome -- Retinoid embryopathy -- Rett syndrome -- Rickets -- Rigid spine syndrome -- Roberts syndrome -- Robinow syndrome -- Rubinstein-Taybi syndrome -- Saethre-Chotzen syndrome -- Sagittal synostosis associated with chromosome abnormalities -- Schizencephaly -- Schmid metaphyseal chondrodystrophy -- Seckel syndrome -- Severe combined immune deficiency -- Short rib polydactyly syndromes (SRPS) -- Sickle cell disease -- Silver-Russell syndrome -- Sirenomelia -- Smith-Lemli-Optiz syndrome -- Smith-Magenis syndrome -- Sotos syndrome -- Spinal muscular atrophy -- Spondyloepiphyseal dysplasia -- Stickler syndrome -- Sturge-Weber syndrome -- Tay-Sachs disease -- Tetrasomy 9p syndrome -- Thalassemia -- Thanatophoric dysplasia -- Thrombocytopenia-absent radius (TAR) syndrome -- Treacher-Collins syndrome -- Trimethylaminuria -- Triploidy -- Trismus pseudocamptodactyly (Hecht syndrome) -- Trisomy 8 mosaicism (Warkany) syndrome -- Trisomy 13 syndrome -- Trisomy 18 syndrome -- Tuberous sclerosis -- Turner syndrome -- Twin-twin transfusion syndrome -- Ulnar-mammary syndrome -- Urofacial (Ochoa) Syndrome -- VATER (VACTERL) association -- Von Hippel-Lindau disease -- Waardenburg syndrome -- Weill-Marchesani syndrome -- Williams syndrome -- Wolf-Hirschhorn syndrome -- X-linked agammaglobulinemia (Bruton type) -- X-linked ichthyosis -- XX Male -- XXX syndrome -- XXXXX syndrome -- XXXXY syndrome -- XY female.-XYY syndrome.

Sommario/riassunto

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.