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Nota di contenuto	Section I: Basic Knowledge. Retinal Histology and Anatomical Landmarks -- Fluorescein Angiography -- Optical Coherence Tomography -- Fundus Autofluorescence -- Electroretinography -- Electrooculography -- Glossary of Relevant Genetic and Molecular/Cell Biology -- Section II: X-linked Forms -- X-Linked Retinitis Pigmentosa -- X-Linked Choroideremia -- X-Linked Juvenile Retinoschisis -- X-Linked Ocular Albinism -- Progressive Cone Dystrophy and Cone-Rod Dystrophy -- Congenital Stationary Night Blindness -- Blue Cone Monochromatism -- Section III: Autosomal Dominant Forms -- Autosomal Dominant Retinitis Pigmentosa -- Best Vitelliform Macular Dystrophy -- Pattern Dystrophy -- Doyne Honeycomb Retinal Dystrophy (Malattia Leventinese, Autosomal Dominant Drusen) -- Occult Macular Dystrophy -- Sorsby Pseudoinflammatory Fundus Dystrophy -- North Carolina Macular Dystrophy -- Pigmented Paravenous Chorioretinal Atrophy (PPCRA) -- Late-Onset Retinal Degeneration -- Section IV: Autosomal Recessive Form -- Rod

Monochromatism (Achromatopsia) -- Retinitis Pigmentosa (Non-syndromic) -- Leber Congenital Amaurosis -- Stargardt Disease -- Enhanced S-Cone Syndrome (Goldmann-Favre Syndrome) -- Best Vitelliform Macular Dystrophy -- Section V: Systemic Disorders -- Mitochondrial Disorder: Kearns-Sayre Syndrome -- Mitochondrial Disorder: Maternally Inherited Diabetes and Deafness -- Ciliopathy: Usher Syndrome -- Ciliopathy: Bardet-Biedl Syndrome -- Ciliopathy: Senior-Løken Syndrome -- Ciliopathy: Alström Syndrome -- Ciliopathy: Sjögren-Larsson Syndrome -- Inborn Errors of Metabolism: Gyrate Atrophy -- Inborn Errors of Metabolism: Pseudoxanthoma Elasticum -- Inborn Errors of Metabolism: Refsum Disease -- Inborn Errors of Metabolism: Bietti Crystalline Dystrophy -- Extracellular Matrix: Alport Syndrome -- Section VI: Phakomatoses -- Von Hippel-Lindau Disease -- Tuberous Sclerosis -- Neurofibromatosis -- Section VII: Phenocopies -- Rubella Retinopathy -- Syphilis -- Autoimmune Retinopathy -- Drug-Induced Retinal Toxicity -- Acute Zonal Occult Outer Retinopathy (AZOOR) and Related Diseases -- Diffuse Unilateral Subacute Neuroretinitis (DUSN) -- Section VIII: Managing IRDs in Clinics -- A Practical Approach to Retinal Dystrophies -- Genetic Testing for Inherited Retinal Dystrophy: Basic Understanding.

Sommario/riassunto

This Atlas of Inherited Retinal Disorders provides a thorough overview of various inherited retinal dystrophies with emphasis on phenotype characteristics and how they are related to the most frequently encountered genes. It will also meet the hitherto unmet need of PhD students who would benefit from seeing the phenotypes of the genes they work on and study. Further, because it would help geneticists use and familiarize themselves with the candidate gene approach to test patients' genomes, enabling them to test more efficiently and cost-efficiently (as the cost of genetic testing is quite high and spiralling higher). This invaluable atlas is organized into eight sections starting with the basic knowledge on retinal imaging as an introduction to the subject matter, then diseases are listed according to their inheritance pattern while disorders with extraocular manifestations are grouped by their defining features. This structure will be intuitive to clinicians and students studying IRDs.