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Titolo	Inherited Chorioretinal Dystrophies : A Textbook and Atlas / / edited by Bernard Puech, Jean-Jacques De Laey, Graham E. Holder
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ISBN	3-540-69466-8
Edizione	[1st ed. 2014.]
Descrizione fisica	1 online resource (477 p.)
Disciplina	599935 610 611.01816 616.07
Soggetti	Ophthalmology Medical genetics Pathology Medical Genetics
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and index at the end of each chapters.
Nota di contenuto	Investigations -- Introduction to molecular genetics and genetic testing for retinal dystrophies- Electrophysiological testing -- Dark adaptation -- Fluorescein angiography -- ICG angiography -- Fundus autofluorescence in retinal dystrophies -- Spectral-domain optical coherence tomography in hereditary retinal dystrophies. Inherited Retinal Dystrophies -- Inherited stationary disorders of the retina -- Retinitis pigmentosa and allied disorders -- Leber congenital amaurosis -- Retinitis punctata albescens -- Usher Syndromes -- Cone and Cone-rod dystrophies -- Enhanced S-Cone syndrome -- Chorioretinopathies: Choroideremia and gyrate atrophy -- Late onset retinal degeneration -- Stargardt Disease -- Bestrophinopathies -- Retinal dystrophies associated with the PRPH2 gene -- Alström syndrome -- Bardet-Biedl syndrome -- Cohen syndrome -- Juvenile neuronal ceroid lipofuscinosis (JNCL) -- Adult Refsum disease -- Abetalipoproteinemia -- LCHAD deficiency -- Jalili syndrome --

Spinocerebellar ataxia -- Dominant cystoid macular dystrophy -- Autosomal dominant Stargardt-like macular dystrophy (ELOVL4). - Spastic paraplegia and retinal degeneration. Kjellin syndrome -- Autosomal dominant drusen -- Cuticular drusen -- Extensive macular atrophy with pseudodrusen-like appearance (EMAP) -- Congenital hypotrichosis with juvenile macular dystrophy -- Mitochondrial retinopathies -- Sorsby fundus dystrophy -- Bietti crystalline corneoretinal dystrophy -- Cystinosis -- Oxalosis -- Alport syndrome -- X-linked retinoschisis -- Paramacular choriocapillaris atrophy -- Exudative vitreoretinopathy -- Stickler syndrome -- Wagner syndrome -- Incontinentia pigmenti Type II (IP2) -- Ganglion cell disease -- Pseudoxanthoma elasticum -- Aicardi Syndrome -- Chorioretinal dysplasia-microcephaly-mental retardation syndrome (Azial-Dufier syndrome) -- Alagille syndrome -- Future therapies for retinitis pigmentosa.

Sommario/riassunto

This lavishly illustrated atlas provides indispensable information to clinicians, geneticists and visual scientists working with inherited retinal diseases. It is filled with high-quality images, up-to-date genetic information and comprehensive electrophysiology. The data for each individual disorder have been summarised in an accessible, reader-friendly format for easy reference. The illustrations include colour fundus photographs, fluorescein angiograms, OCT scans, electrophysiological studies and pedigrees. The editors and authors are well-known experts in the field and have drawn upon their extensive experience to produce this unique atlas.