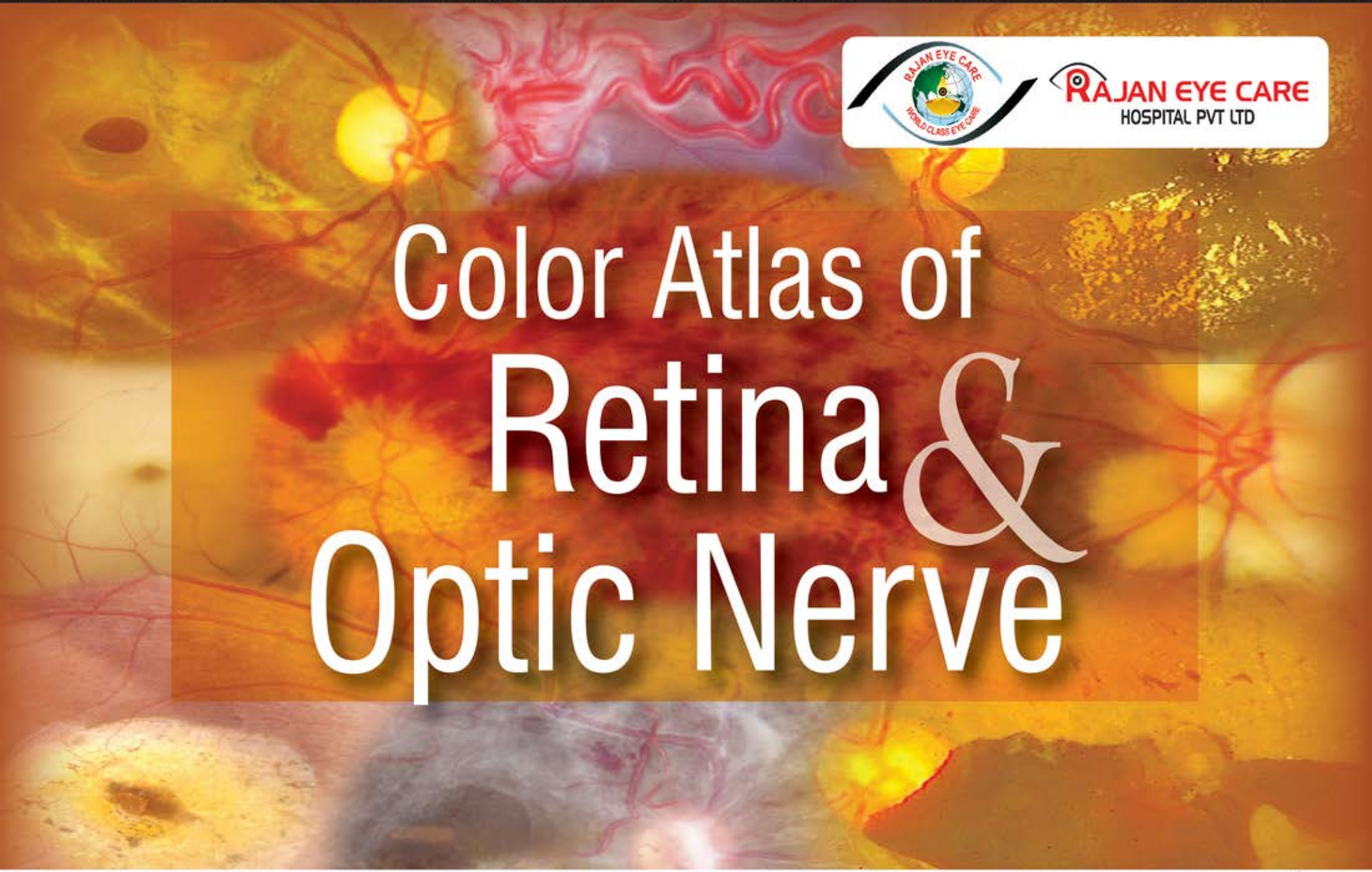

RAJAN EYE CARE HOSPITAL PVT LTD

Color Atlas of Retina & Optic Nerve

Editors
Mohan Rajan
Nicey Roy Thomas

Foreword
Lingam Gopal

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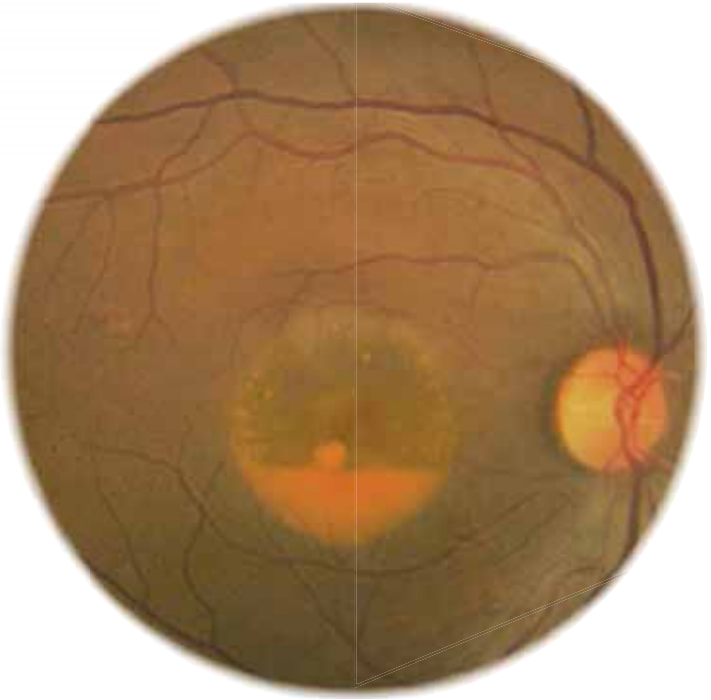
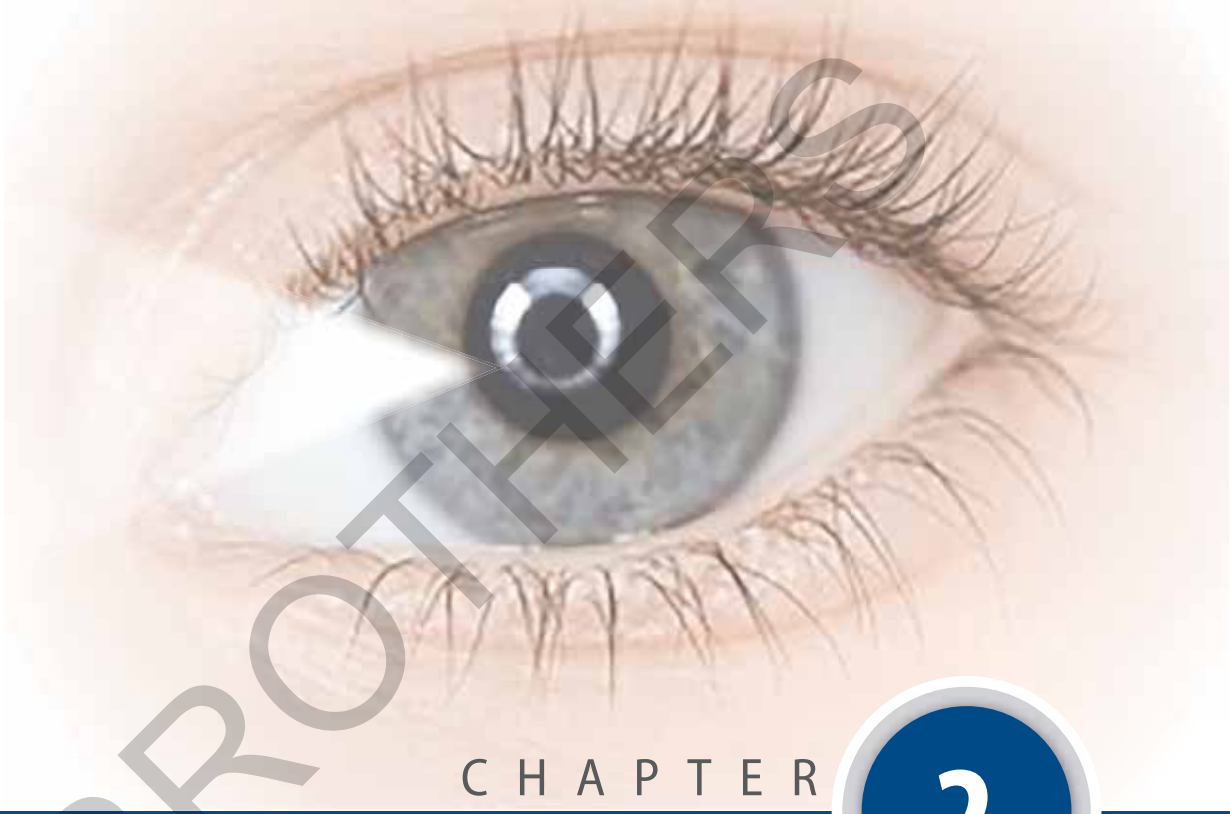




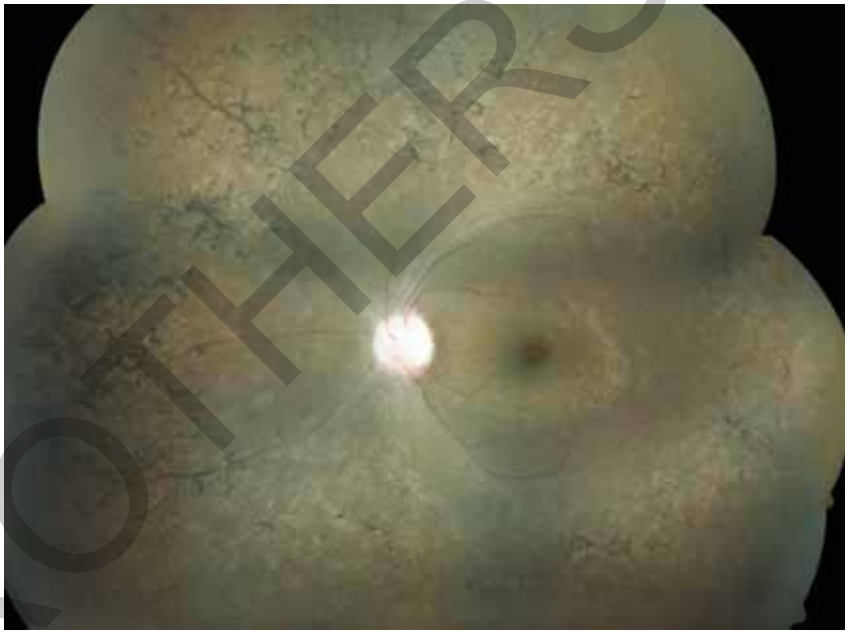
CHAPTER 2

Retinal Degenerations and Dystrophies

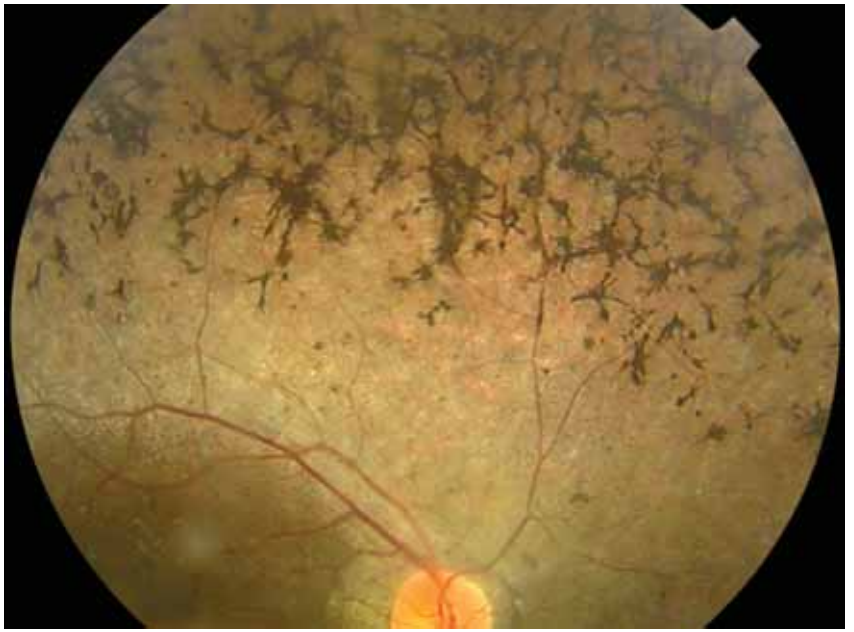
RETINITIS PIGMENTOSA AND ALLIED DISORDERS

Typical Retinitis Pigmentosa

Retinitis pigmentosa (RP) refers to a group of hereditary retinal degenerations that is characterized by progressive damage to the photoreceptor–pigment epithelial complex. RP may be seen in isolation (typical RP) or in association with systemic disease (syndromic RP). The two hallmark symptoms of typical RP are night blindness and progressive loss of peripheral visual field, usually beginning in the midperiphery and then extending into the far periphery. An annular scotoma may progress to "tunnel vision" late in the course of the disorder. RP has various inheritance patterns, which include an autosomal dominant pattern (30–40%), an autosomal recessive pattern (50–60%), and an X-linked pattern (10–15%) with the autosomal dominant forms often conferring a less severe phenotype than the X-linked and recessive forms. The classically described fundus appearance of RP includes attenuated retinal vessels, mottling and granularity of the retinal pigment epithelium (RPE), bone spicule intraretinal pigmentation, and optic nerve head pallor. Associated vitreous and retinal complications include vitreous veils and pigmentary cells, cystoid macular edema, epiretinal membrane formation, and a Coats-like retinal vascular response.



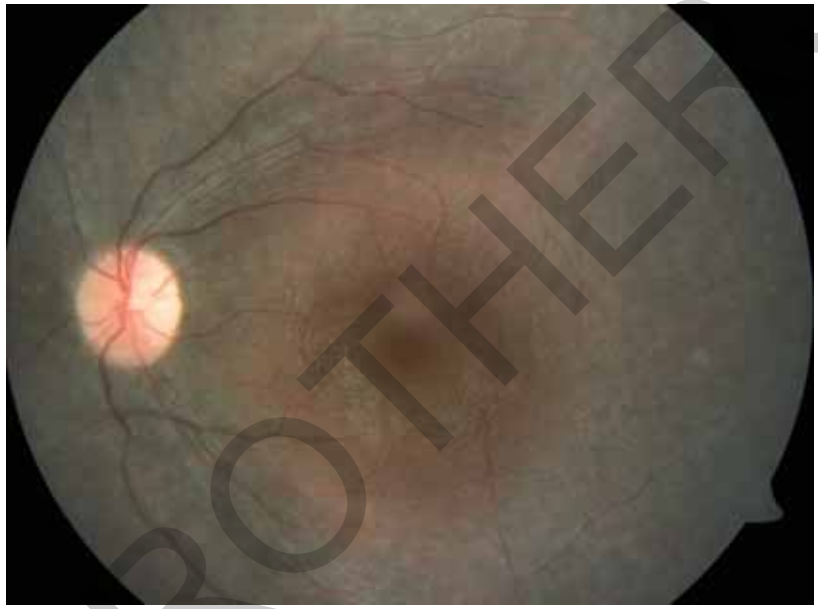
Fundus photograph of a patient with typical RP showing attenuated retinal vessels, mottling and granularity of the RPE, bone spicule intraretinal pigmentation, and optic nerve head pallor.



Intraretinal, bone spicule pigment formations represent migration of pigment into the retina from disintegration of RPE cells with accumulation in the interstitial spaces surrounding retinal vessels. This process occurs most prominently at the junctions of vessels producing perivascular pigmentary cuffing and spicule-shaped deposits.

Retinitis Pigmentosa Sine Pigmento

Almost all forms of RP go through a stage where the retina appears either normal or nearly normal. Patients who have very early RP without fundus pigmentary abnormalities are often diagnosed as having RP sine pigmento or paucipigmentary RP. The sine pigmento stage may exist for decades before typical RP signs appear.

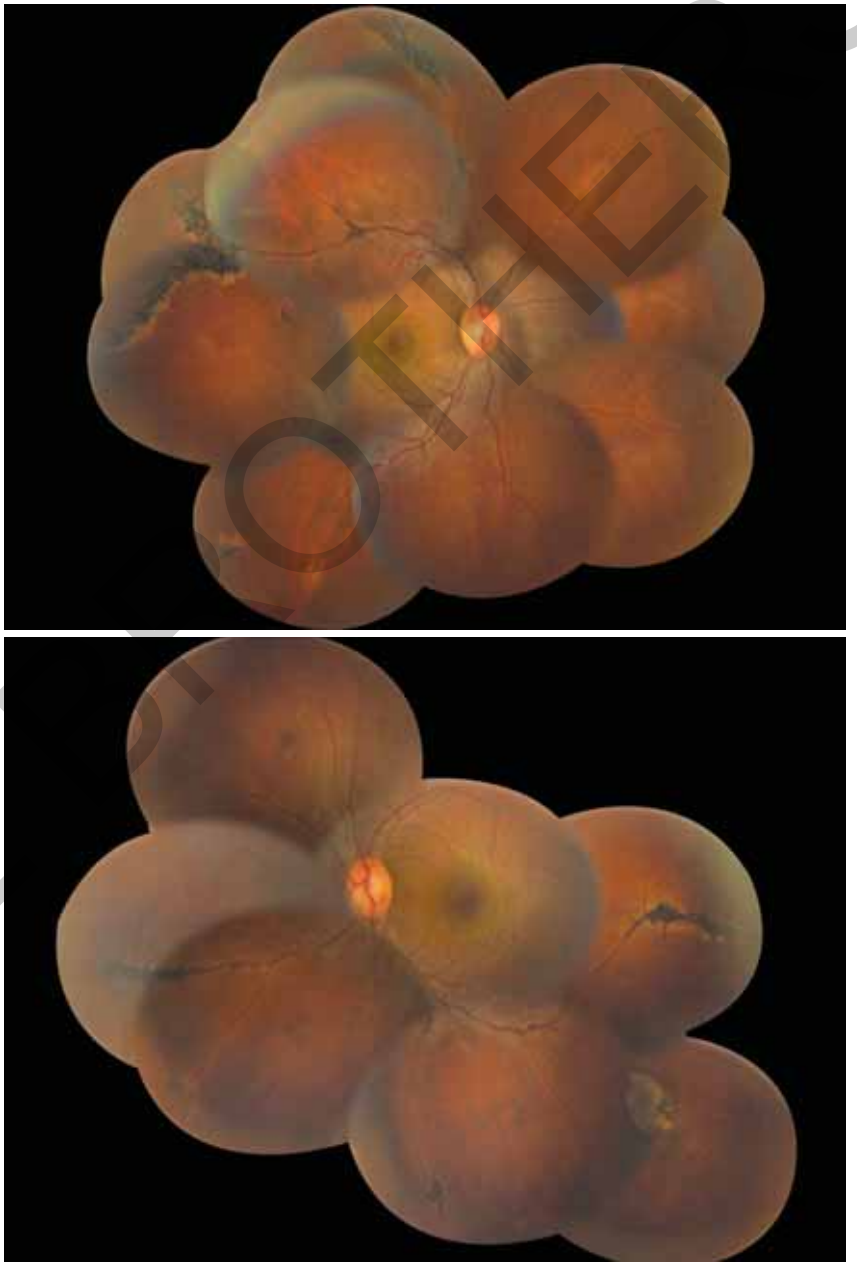


Retinitis Pigmentosa Inversa

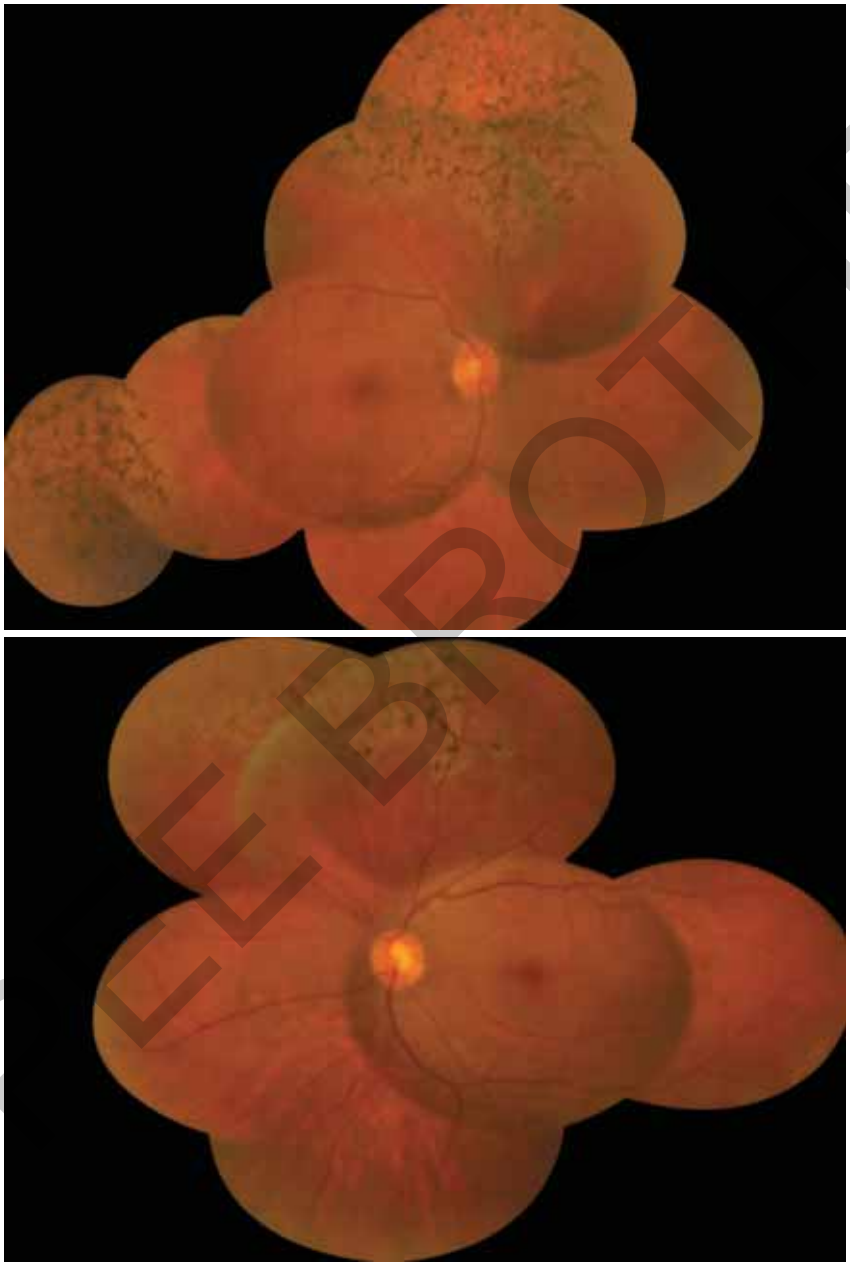
Retinitis pigmentosa inversa or inverse RP is a rare variant of this disorder characterized by waxy pallor of the optic disk, attenuation in retinal arterioles, areas of choroidal degeneration with pigment migration, and bony spicule formation in the macular area. In contrast to more typical forms of RP, this anomaly destroys central vision, leaving peripheral vision intact.

Pigmented Paravenous Retinochoroidal Atrophy

Pigmented paravenous retinochoroidal atrophy (PPRCA) is a pigmentary retinopathy that is currently poorly understood, but probably represents an acquired response pattern to an infectious or inflammatory disease. It has been reported in association with meningoencephalitis, tuberculosis, syphilis, and rubeola. In most, the fundus appearance is first noticed on a routine examination. The pigmentary changes are closely associated in distribution with retinal veins. Most cases are relatively stable over time, although progression has been reported. Electroretinographic responses are only mildly to moderately abnormal, if at all, but the electro-oculogram (EOG) is usually affected, often significantly.



A typical case of PPRCA shows pigment clumping and spicules in a predominantly paravenous distribution with variable amounts of retinochoroidal atrophy along the same distribution.

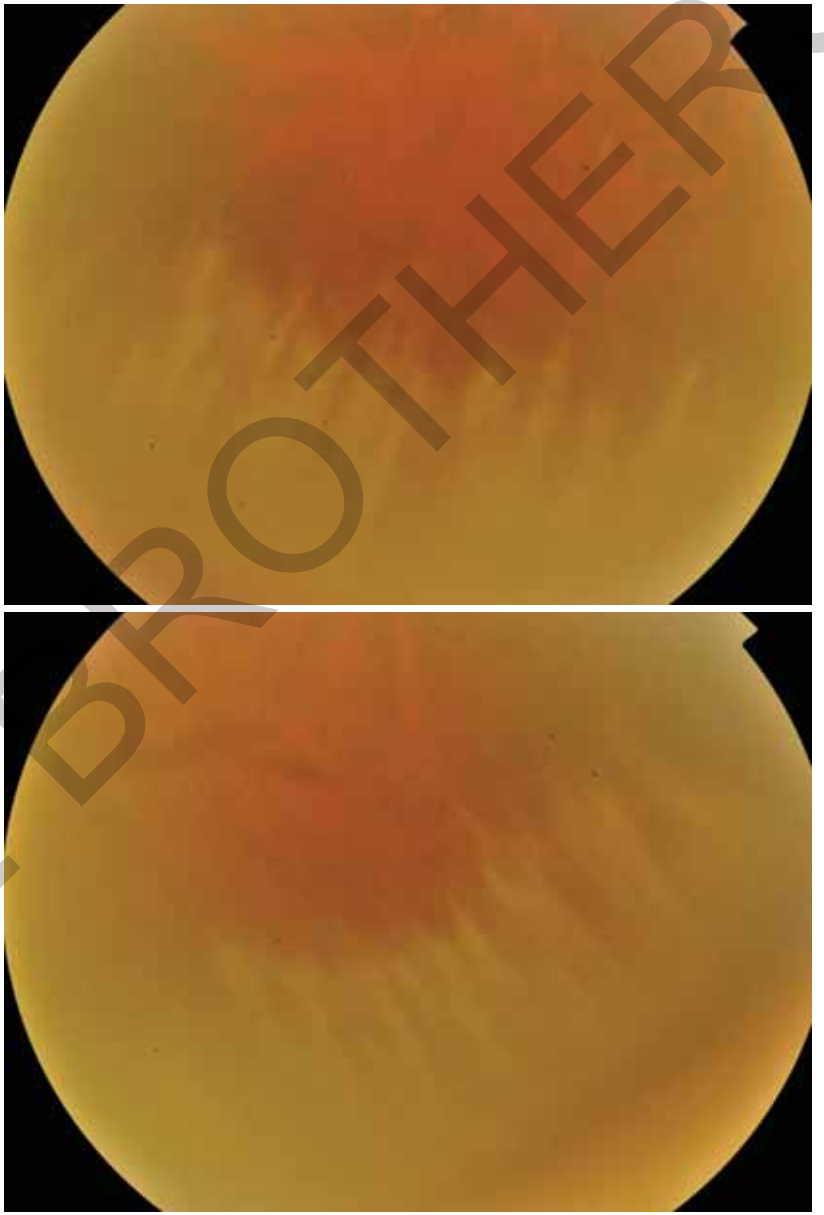


Fundus photographs of a patient with PPRCA. The optic disk, macula, and retinal vessels are typically normal.

HEREDITARY VITREORETINAL DEGENERATIONS

Snowflake Vitreoretinal Degeneration

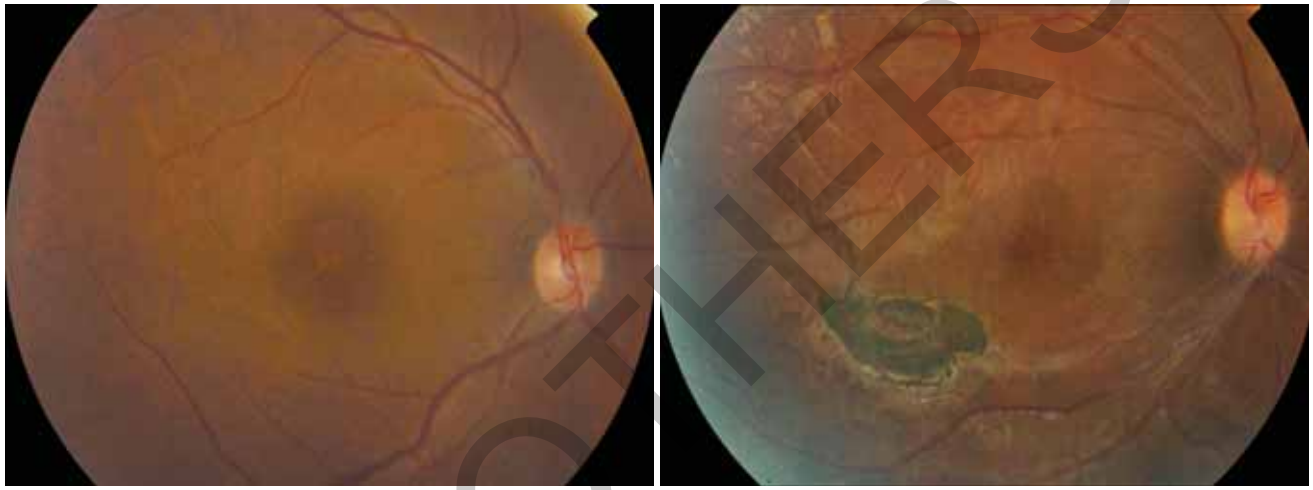
Snowflake vitreoretinal degeneration is an autosomal dominant, distinct vitreoretinopathy caused by mutation in the *KCNJ13* gene, which encodes a potassium channel expressed predominantly in the retina. The condition received its name from the minute crystalline-like deposits (snowflakes) in the peripheral retina. Other distinguishing clinical features include optic nerve head dysmorphism with fibrillar degeneration of the vitreous, corneal guttae, and peripheral retinal degeneration. Rhegmatogenous retinal detachment occurs in approximately 20% of affected individuals.



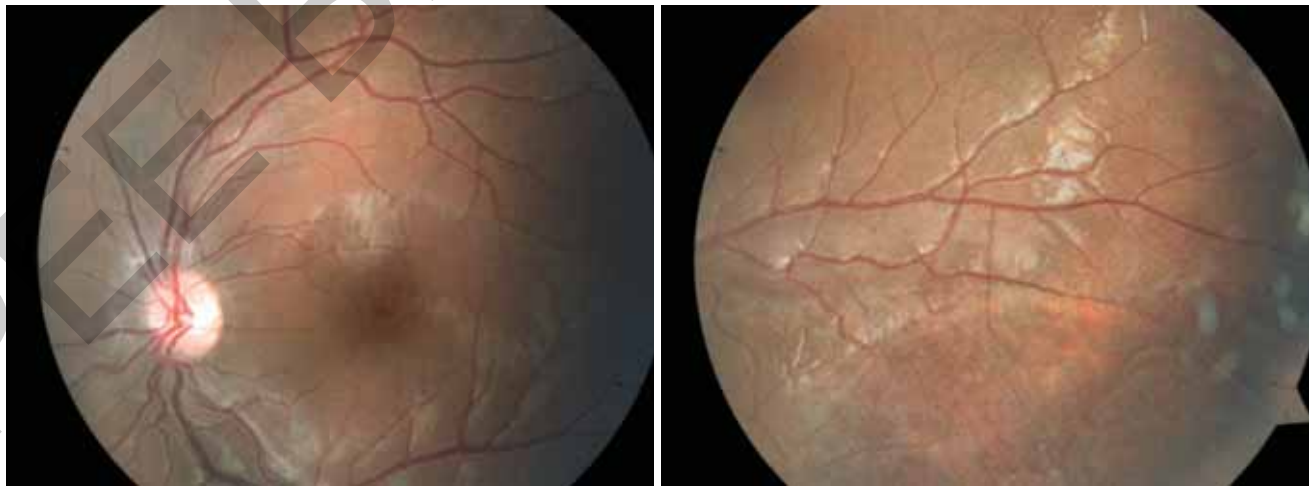
This patient with snowflake hereditary degeneration showed white or yellow-white granular-like deposits of the peripheral retina. They can be rather evenly distributed about the entire circumference of the eye, but sometimes they may show a predilection for the inferior quadrants.

X-linked Juvenile Retinoschisis

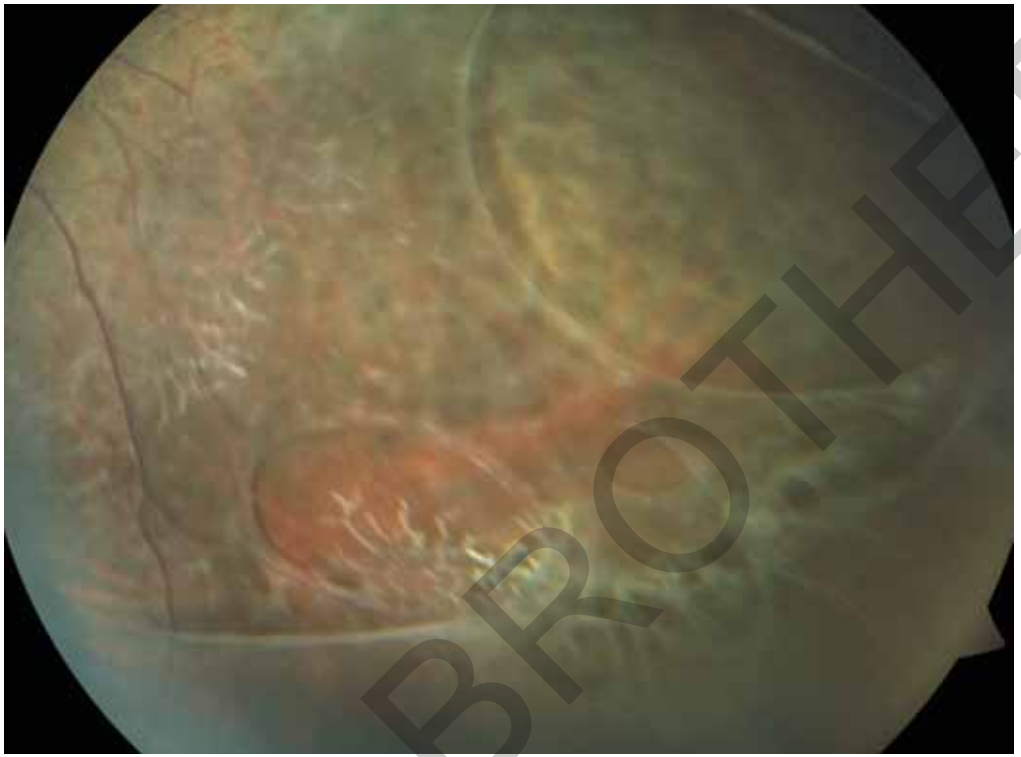
Hereditary X-linked retinoschisis (XLRS) is the most common cause of juvenile macular degeneration in males characterized by splitting of the neural retina. It is an X-linked recessive disorder caused by mutation in the retinoschisin gene (*RS1*) at Xp22, which plays a role in retinal cell adhesion. The condition is usually bilateral. Foveal schisis is the characteristic sign of XLRS and is present in 98–100% of cases. The typical foveal schisis, seen as a spoke wheel pattern of folds radiating out from the fovea, has been found to appear in only about 70% of XLRS patients. Despite a cystic appearance, the macular lesions do not leak on fluorescein angiography. Peripheral retinoschisis, typically inferotemporal, is present in around 50% of patients. These patients may also experience large, inner layer holes associated with "vitreous veils." Sheathed, occluded, and unsupported retinal vessels with vitreous hemorrhage may also occur. About 5–20% of XLRS patients may progress to retinal detachment. Other changes, including subretinal linear fibrosis, pigmentation, white retinal flecks, and vascular attenuation or sheathing, often appear in peripheral retina.



X-linked juvenile retinoschisis is associated with macular schisis in all cases. The schisis can vary from barely detectable to a very prominent cystic change at the fovea with spoke-like radial extension into the paramacular region.



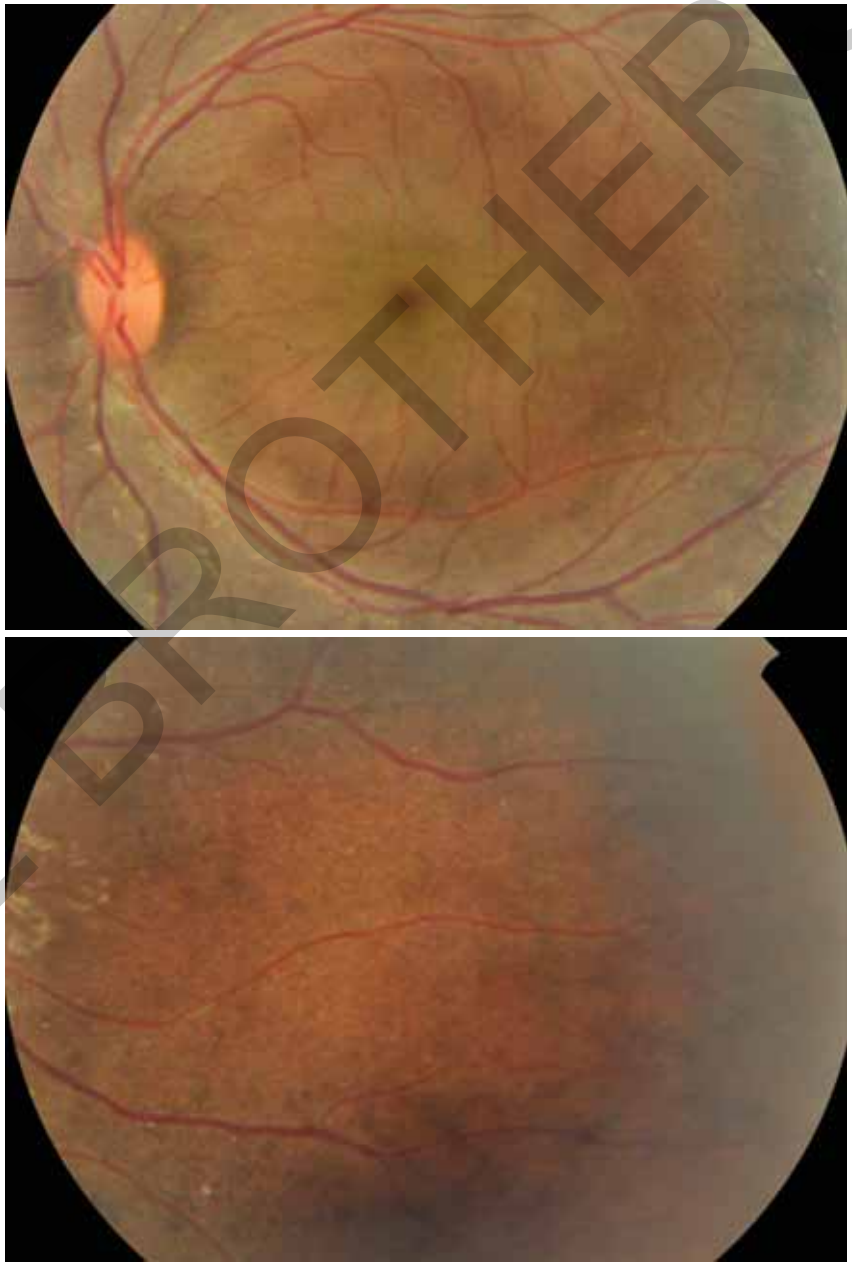
Peripheral retinoschisis is seen in X-linked juvenile retinoschisis in about 50% of cases. This is the fundus photographs of a patient who had both foveal and peripheral retinoschisis.



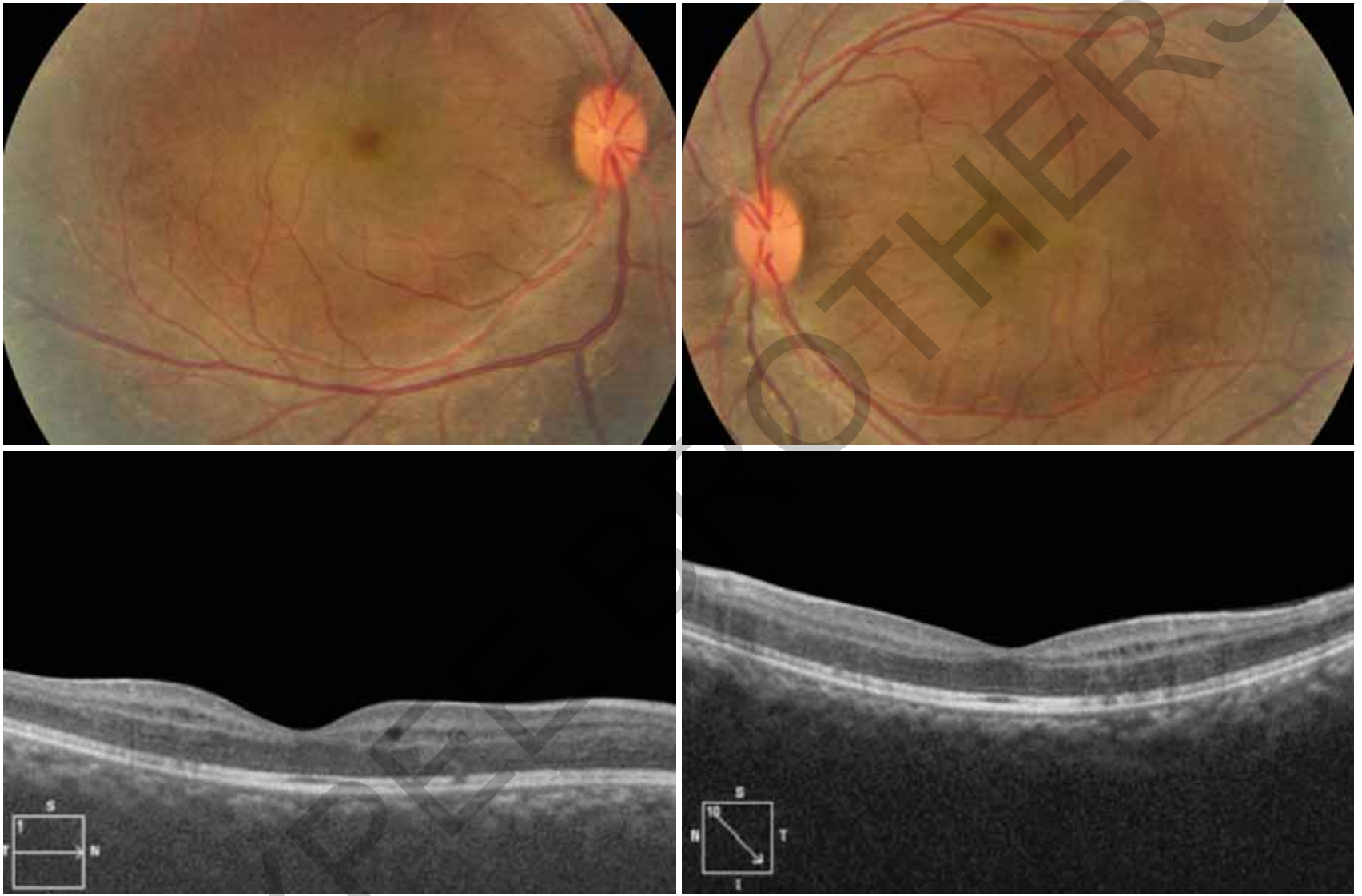
Fundus photograph of a patient with inferotemporal XLRS with large inner retinal breaks. Sheathed, occluded retinal vessels are seen as well as a delicate lacy pattern in the periphery.

Enhanced S-cone Syndrome (Goldmann–Favre Syndrome)

The enhanced S-cone syndrome (ESCS) is an autosomal recessive disorder caused by mutations in the nuclear receptor gene (*NR2E3*) on chromosome 15q23. There is increased differentiation of photoreceptor precursors to S cones and underproduction of rods resulting in altered ratio of S to L (long-wavelength, red) and M (middle-wavelength, green) photoreceptor subtypes. Patients typically present in the first decade with nyctalopia, which may or may not be associated with diminished vision. The fundus findings are highly variable and include the typical nummular pigment deposits at the level of RPE and intraretinal cysts, intraretinal yellow dots, helicoid subretinal fibrosis, and torpedo lesions. Cystoid maculopathy/macular schisis is commonly seen. Other findings that may be noted include an optically empty vitreous with preretinal bands, lattice degeneration, and even retinal detachment. Patients have characteristic electroretinogram (ERG) findings that reflect the near absence of rods and a predominance of S cones. The scotopic response is, therefore, extinguished and the maximal rod–cone response resembles the photopic flash waveform.



Fundus photographs of this patient with ESCS shows midperipheral nummular pigmentary changes.



Spectral domain-optical coherence tomography (SD-OCT) shows early macular cystic changes involving the inner and outer retinal layers.

Color Atlas of
Retina & Optic Nerve

Salient Features

- An atlas of common retinal and optic nerve disorders that provide a visual guide to the diagnosis of the full spectrum of retinal disease, including early and later stages of the disease
- Each disorder has been represented with multiple images associated with key clinical features and signs
- This book is an essential source to all ophthalmologists, especially postgraduates and residents, to review fundus pathologies

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