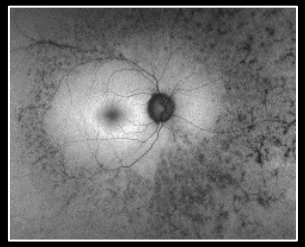
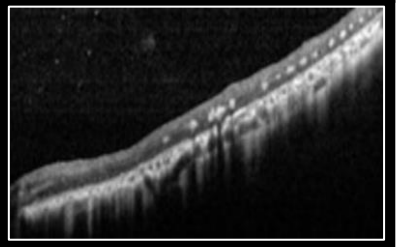
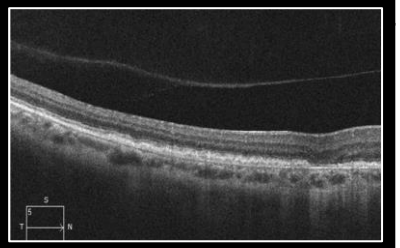
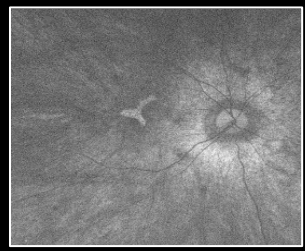
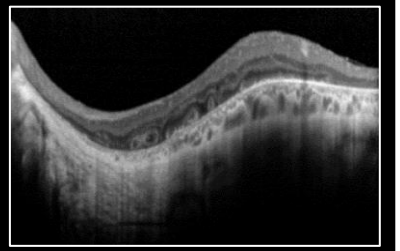
Centre for Eye Health

CHAIR-SIDE REFERENCE: PANRETINAL PIGMENTARY INHERITED RETINAL DEGENERATIONS

INHERITED RETINAL DEGENERATIONS (IRD)

Inherited retinal degenerations (IRDs) encompass a large group of clinically and genetically heterogeneous diseases that can lead to progressive vision loss. As such, there are a number of different methods utilised to group them into categories. This chairside reference is specific to panretinal dystrophies which are associated with pigmentary changes. A common symptom of these conditions is nyctalopia and associated visual field constriction.

DIFFERENTIAL DIAGNOSIS OF PAN-RETINAL INHERITED DYSTROPHIES

Retinal image	Fundus Autofluorescence	OCT	Description
Retinitis pigmentosa (Rod-Cone Dystrophy)			• The most common inherited retinal disorder characterised by diffuse progressive photoreceptor degeneration (predominantly rods followed by cones) and RPE. • **Symptoms**: nyctalopia with variable timing of onset • **Fundus appearance**: Bone-spicule pigmentation in mid-peripheral retina, attenuation of retinal vessels, waxy pallor of the optic nerve head. Retinal changes are evident in established cases with changes often subtle in early disease. • **FAF**: Hypo-AF in areas of pigmentation, hyper-AF ring around the macula • **OCT**: Disorganisation of outer retina layers (EZ, ELM, ONL) and RPE. Outer retinal tubulations (hyper-reflective round structures with hypo-reflective lumen) may be seen in late-stage disease. • **Othe**r: ERM, macular oedema, posterior subcapsular cataract
Fundus Albipunctatus	*Image not available*		• A form of congenital stationary night blindness • **Symptoms**: night blindness, restricted peripheral visual field • **Fundus appearance:** numerous small whitish-yellow spots scattered in the midperiphery and perifovea. • **FAF:** grainer apperanace than do normal retinas, no hyper-FAF spots correlated to the albipunctate spots funduscopically • **OCT**: hyperreflective lesions at the level of RPE extending into EZ and ELM with focal thinning of ONL • **Prognosis**: generally stable or minimally progressive
Choroideraemia			• A choroiretinal dystrophy characterised by the diffuse, progressive degeneration of the photoreceptors, RPE and choriocapillaris. • **Symptoms:** nyctalopia in the first decade of life • **Fundus:** RPE degeneration leads increased visibility of choroidal vessels and sclera as well as pigment clumping. • **FAF:** Reduced background autofluorescence (AF) with isolated “islands” of relatively normal AF • **OCT:** Pigment clumps visible as hyper-reflective deposits. Outer retinal tubulations in advanced disease. Thinning and loss of outer retina, choriocapillaris and choroid.

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CHAIR-SIDE REFERENCE: PANRETINAL PIGMENTARY INHERITED RETINAL DEGENERATIONS

Retinal image	Fundus Autofluorescence	OCT	Description
Gyrate Atrophy			• A rare retinal dystrophy characterised by progressive chorioretinal degeneration, early cataract formation and myopia • **Symptoms***:* Nyctalopia occurs in the first decade of life. • **Fundus appearance:** Large peripheral areas of chorioretinal atrophy that coalesce over time forming a scalloped border peripherally. • **FAF***:* Ares of chorioretinal atrophy are hypo-AF. FAF imaging shows the extent of chorioretinal atrophy much more clearly than retinal imaging. • **OCT:** Atrophy of the outer retina and RPE. Intraretinal cystoid oedema and/or outer retinal tubulations may be present.
Bietti Crystalline Corneoretinal Dystrophy			• A rare dystrophy characterised by presence of crystalline deposits, RPE degeneration and choroidal vessel sclerosis • **Symptoms**: reduced vision, night blindness, visual field loss typically from 2nd decade of life • **Fundus appearance**: Multiple, refractive crystals in the retina with associated RPE and choriocapillaris atrophy • **FAF**: Ares of chorioretinal atrophy are hypo-AF. • **OCT**: The crystalline deposits appear as hyperreflective dots within the retinal layers. Retinal and choroidal architecture is disrupted. Outer retinal tubulation often present. • **Other**: Crystalline deposits in the peripheral cornea. CNV and/or macular oedema are complications of this condition.
DIFFERENTIAL DIAGNOSIS			
Pigmented paravenous chorioretinal atrophy			• A rare disorder of unknow aetiology, characterised by pigment accumulation along the distribution of retinal veins. Typically non-progressive and has minimal effect on vision. • **Fundus**: Pigmentation and associated chorioretinal atrophy along retinal veins. • **FAF**: Linear areas of hypo-AF along the large retinal veins, often surrounded by an area of hyper-AF • **OCT**: Thinning of the outer retina layers and disorganisation of the RPE-choriocapillaris complex. Hyper-reflective intra-retinal plaques with posterior shadowing, corresponding to pigmentary changes.

CFEH TELEHEALTH
SUPPORTING OPTOMETRISTS

This reference is based on the current literature and evidence at the time of writing. This reference is designed a guide to aid diagnosis however individual cases must be assessed in the context of all available clinical data. If you need additional advice or guidance, consider making a free telehealth appointment with a senior CFEH optometrist.

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