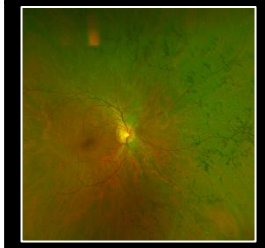
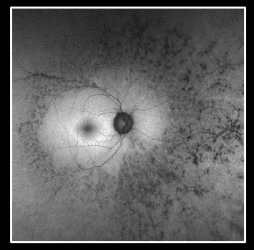
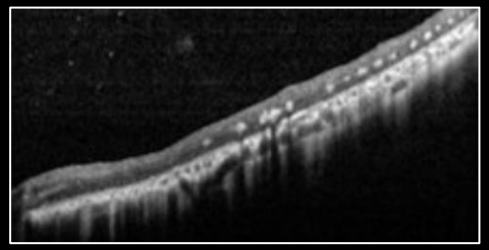
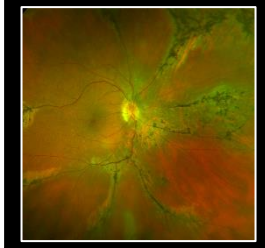
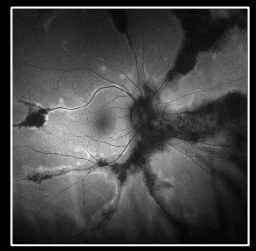
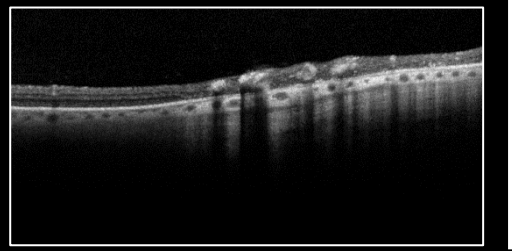
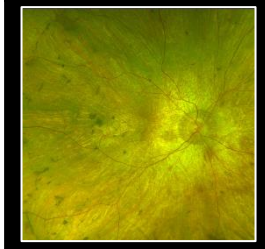
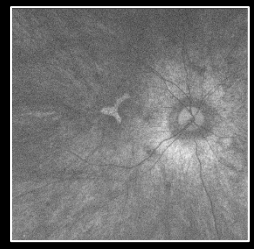
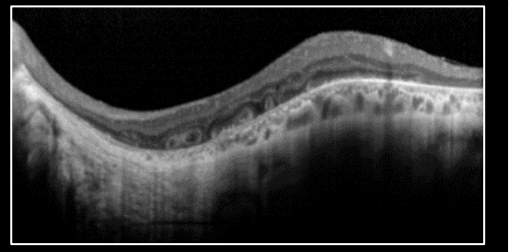
Centre for Eye Health

CHAIR-SIDE REFERENCE: 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. A common symptom of these conditions is nyctalopia, and associated visual field constriction.

Widefield image	Fundus Autofluorescence	Optical coherence tomography (OCT)	Description
Retinitis pigmentosa 			***Fundus***: Bone-spicule pigmentation in mid-peripheral retina, attenuation of retinal vessels, waxy pallor of the optic nerve head. ***FAF***: Hypo-AF in areas of pigmentation, hyper-AF ring around the macula ***OCT***: Disorganisation of outer retina layers (EZ, ELM, ONL, RPE) ***Other***: ERM, macular oedema, posterior subcapsular cataract
Pigmented paravenous chorioretinal atrophy 			***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.
Choroideraemia 			***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. ***Other:*** Central vision preserved until late stages when the macula is affected.

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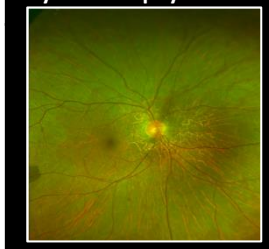
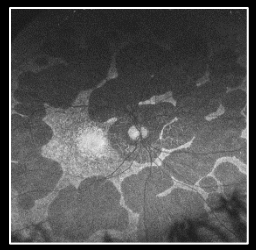
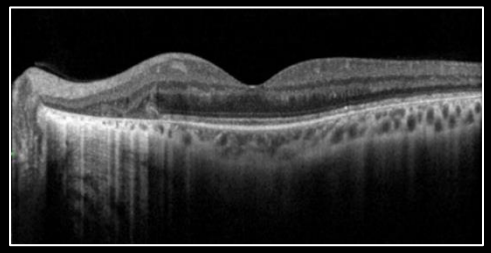
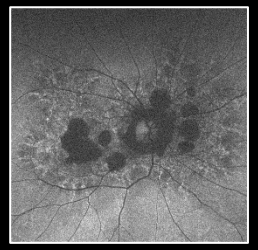
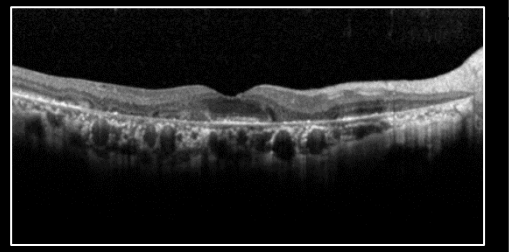
Widefield image	Fundus Autofluorescence	Optical coherence tomography (OCT)	Description
Gyrate Atrophy			***Fundus:*** Large peripheral areas of chorioretinal atrophy that coalesce over time forming a scalloped border peripherally**.** ***FAF:*** Ares of chorioretinal atrophy are hypo-AF ***OCT:*** Atrophy of the outer retina and RPE. Intraretinal cystoid oedema and/or outer retinal tubulations may be present. ***Other****:* Nyctalopia occurs in the first decade of life.
Bietti Crystalline Corneoretinal Dystrophy			***Fundus:*** 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.
Leber's Congenital Amaurosis			***Fundus***: Normal / flecked / varying degrees of intraretinal pigmentation (gene dependent). Macula spared early with maculopathy developing later. Peripheral bone spicule pigmentation may develop. Optic disc drusen, disc oedema or pseudopapilloedema may also be associated. ***FAF****:* Depends on retinal changes present but may include hypo-AF at the macula (maculopathy) surrounded by hyper-AF demarcation line. ***OCT***: Disruption of the EZ initially, sparing the macula. Thinning of the outer retina and RPE over time. ***Other***: Presents early childhood - severe vision loss, congenital nystagmus, sluggish pupillary light reflex and the oculodigital sign (eye poking).
Sorsby Pseudoinflammatory Fundus Dystrophy			**Fundus**: Numerous drusen-like yellowish deposits at the posterior pole with disciform macular atrophy and development of choroidal neovascularisation (CNV). Late stages – peripheral retina becomes involved, fibrotic posterior pole lesions and pigment clumping develop. ***FAF***: Hypo-AF in areas of chorioretinal atrophy. ***OCT****:* Sub-retinal (drusen-like) deposits on BM and sub-retinal CNV. Atrophy of choroid and outer retina (reduced choroidal thickness). ***Other***: Onset second to fourth decade of life. Nyctalopia and/or difficulty with light adaptation may be noted before onset of macular atrophy.

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