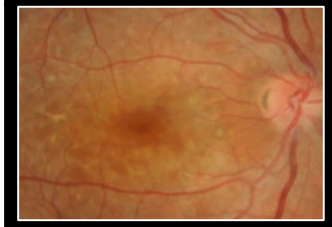
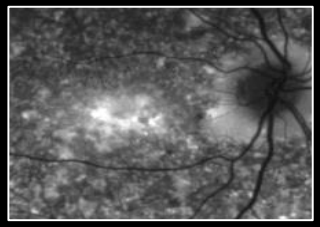
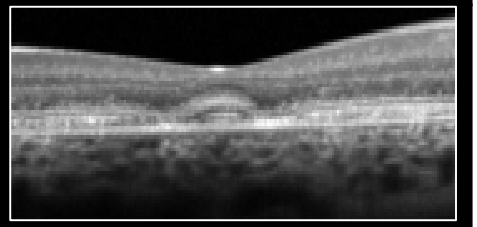
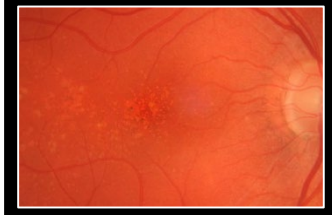
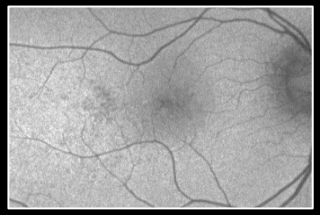
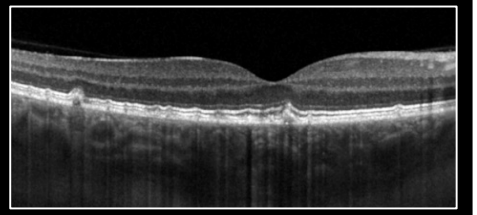
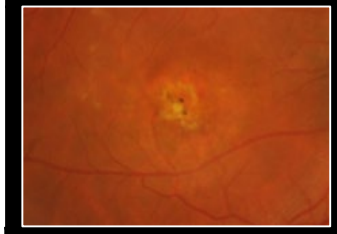
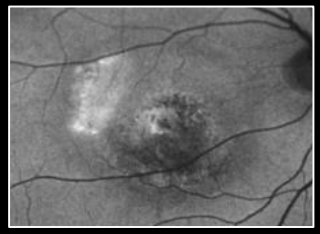
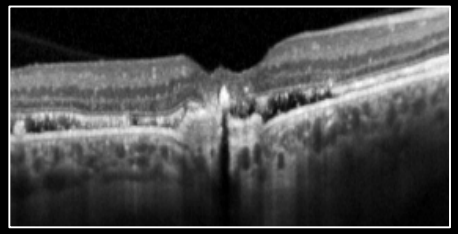
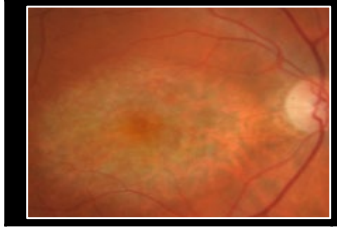
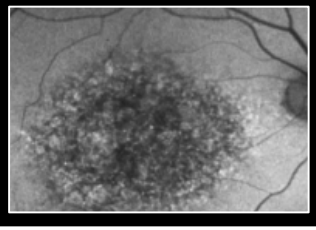
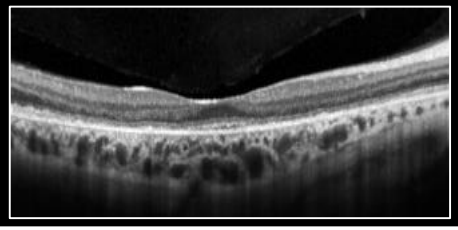
Centre for Eye Health

CHAIR-SIDE REFERENCE: MACULA DYSTROPHIES

MACULA DYSTROPHIES

* The specific diagnosis of dystrophies may be uncertain in the absence of electrophysiology results and genetic testing

Optomap/Retinal Photo	Fundus Autofluorescence	Optical coherence tomography (OCT)	Description
Stargardt Disease/Fundus flavimaculatus*			• Foveal atrophy often surrounded by discrete yellowish round or pisciform flecks scattered throughout the fundus with intense hyper-autofluorescence • Juvenile onset • Gradual and progressive visual decline ranging from 6/15-6/60 • Predominantly autosomal recessive inheritance • Hyper-reflective thickening of the retinal pigment epithelium (RPE) and thinning of the ellipsoid zone (EZ). • May be foveal sparing with regular EZ profile in some cases
			
Autosomal dominant drusen /Malattia Leventinese/Doyne honeycomb retinal dystrophy			• Small, discrete drusen radiate in streaks or lines from the centre of the fovea in the early stage • Drusen progressively become confluent, leading to the honeycomb appearance • Onset in the 3rd to 4th decade of life • Usually asymptomatic before the age of 40, then more rapid progressive central vision loss occurs • Autosomal dominant inheritance • Risk of geographic atrophy and/or choroidal neovascularization in later stage • A hyper-reflective thickening of the retinal pigment epithelium-Bruch's membrane complex, associated with localised dome-shaped elevations
			
Best Vitelliform Macular Dystrophy*			• Initially presents with a yellow, yolk like macular lesion • Progresses to atrophy and/or neovascularisation in later stages • Variable age of onset ranging from 1st to 6th decade • Usually symptomatic before the age of 40 • Autosomal dominant inheritance • Early lesions found between the RPE and sensory retina • Later stage may involve sub-retinal fluid, subretinal fibrosis and oedema • Variable hyper-fluorescence corresponding to vitelliform material , hypo-autofluorescence in atrophic areas
			
Central Areolar Choroidal Dystrophy*			• Initially parafoveal pigmentary RPE changes progressing to enlarged RPE atrophy and eventually confluent chorioretinal atrophy • VA deteriorates at age 30-50 years but may be asymptomatic until later. Usually causes profound vision loss • Occasionally photophobia associated • Autosomal dominant inheritance • Reduced retinal thickness with disruption of the EZ and outer retina • Remaining retinal layers are intact • Speckled pattern of hyper- and hypo-fluorescence confined to the macula region in a round / oval shape
			

This reference is based on the current literature and evidence at the time of writing. This reference is designed a guide to aid diagnosis and management decisions however individual cases must be assessed in the context of all available clinical data.

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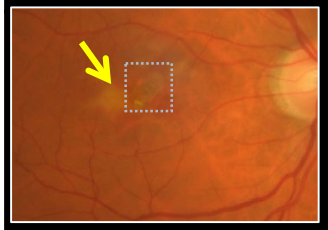
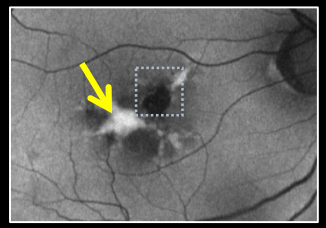
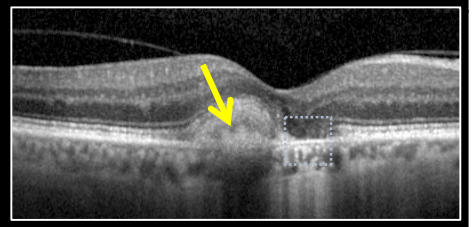
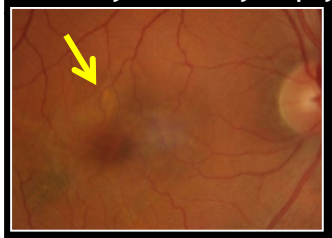
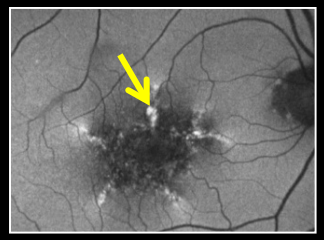
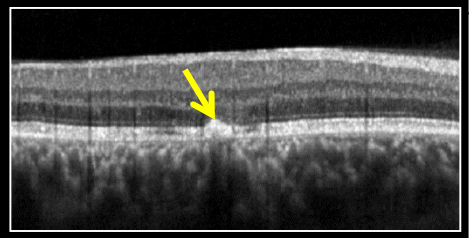
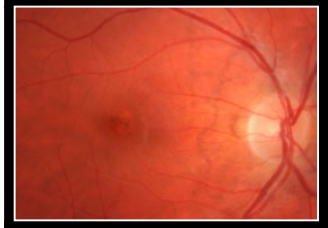
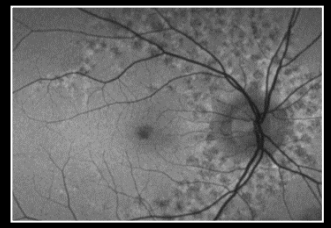
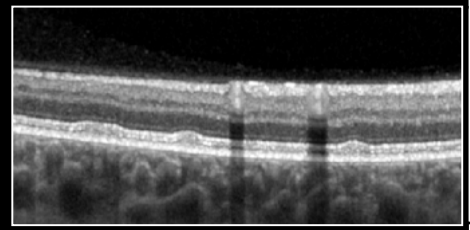
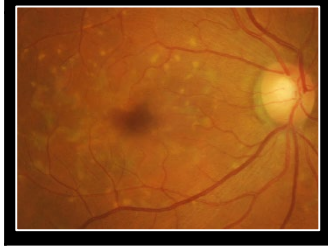
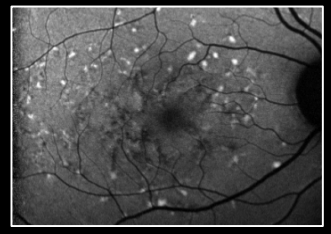
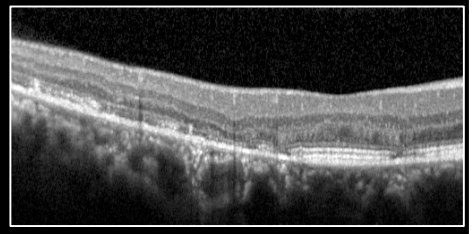
PATTERN DYSTROPHIES

A heterogeneous group of progressive retinal pigment epithelium (RPE) alterations with onset typically in 30-50s

Optomap/Retinal Photo	Fundus Autofluorescence	Optical coherence tomography (OCT)	Description
Adult-Onset Foveomacular Vitelliform Dystrophy			• Bilateral subfoveal yellowish subretinal deposit (yellow arrow) at an average size of 1/3 disc diameter with intense hyper-autofluorescence • Multifocal vitelliform lesions can be present in some cases • With time, lesion can show more pigmentary changes, progressive atrophy (dashed blue square) and/or choroidal neovascularisation with corresponding vision loss • Can be classified into vitelliform, pseudohypopyon, vitelliruptive, and atrophic stages.
			
Butterfly Pattern Dystrophy			• Yellow deposits consist of 3 to 5 linear lines, resembling the wings of a butterfly • The yellow lesions are hyper-autofluorescence and show hyper-reflective changes at the photoreceptor-RPE interface • VA usually stable, but can decline rapidly after the seventh decade by progressive photoreceptor and RPE atrophy in the macula
			
Reticular Dystrophy of the RPE			• Clearly defined network of hyperpigmented lines that resemble a fishnet with knot • OCT shows small RPE elevations • Fundus autofluorescence may show mixed hyper and hypo-autofluorescence • The hyperpigmented areas gradually fade, leaving corresponding areas of RPE atrophy
			
Multifocal pattern dystrophy stimulating Stargardt			• Irregular yellow flecks within the posterior pole that resembles flavimaculatus flecks in Stargardt disease • Flecks are initially hyper-autofluorescent • OCT shows disturbance and abnormality in the photoreceptor outer segment-RPE level • Can be differentiated from Stargardt by: autosomal dominant inheritance, a relatively late age of onset, a comparably good visual acuity and no dark choroid on fluorescence angiography.
			

Please note that another subtype of pattern dystrophies **Fundus Pulverulentus** (Coarse Pigment Mottling of the Macula) is not presented in this reference.