## MACULA DYSTROPHIES

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

<table>
<thead>
<tr>
<th>Optomap/Retinal Photo</th>
<th>Fundus Autofluorescence</th>
<th>Optical coherence tomography (OCT)</th>
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</table>
| **Stargardt Disease/Fundus flavimaculatus*** | ![Image](image1.png) | ![Image](image2.png) | - 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** | ![Image](image3.png) | ![Image](image4.png) | - 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*** | ![Image](image5.png) | ![Image](image6.png) | - 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 asymptomatic 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*** | ![Image](image7.png) | ![Image](image8.png) | - 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.
# Pattern Dystrophies

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

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| **Adult-Onset Foveomacular Vitelliform Dystrophy** | ![Image](image1) | ![Image](image2) | - 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** | ![Image](image3) | ![Image](image4) | - 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** | ![Image](image5) | ![Image](image6) | - 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** | ![Image](image7) | ![Image](image8) | - 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.