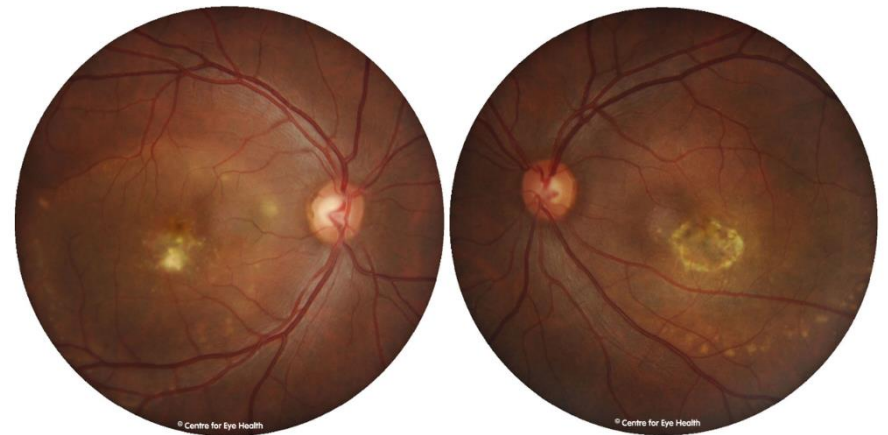
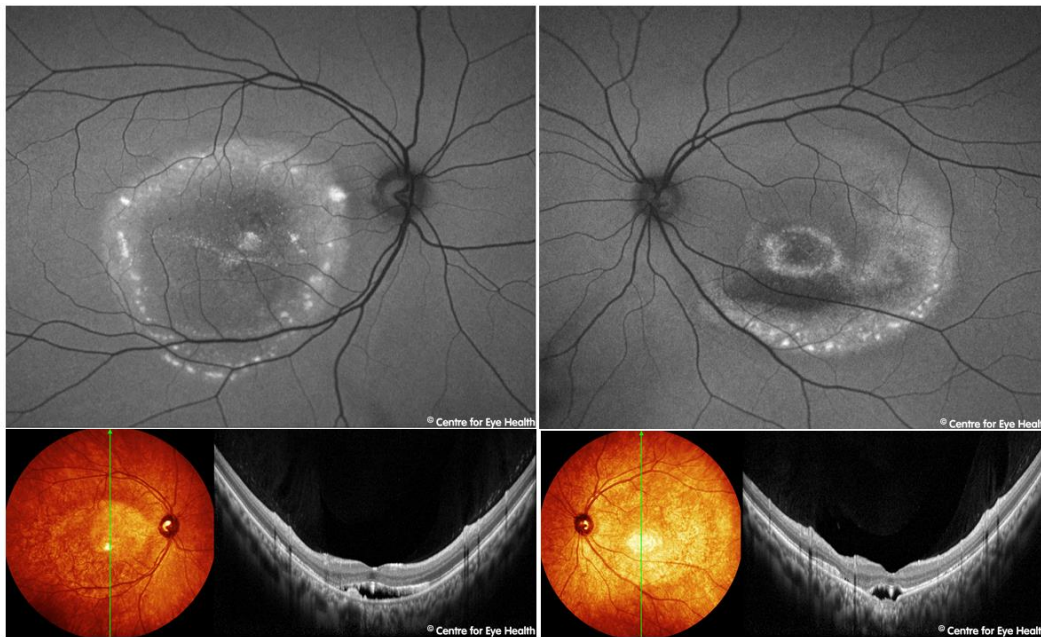




## CFEH Facebook Case #120

A 37 year old male presented for a macular assessment. His visual acuity was 6/6- in the right eye and 6/7.5- in the left. What macular condition does this patient likely to have and what is the prognosis of this condition?



# ANSWER

Optomap and Spectralis OCT images showed bilateral subretinal fluid, loss of photoreceptors and subretinal deposits with associated hyperautofluorescence. This presentation and early onset suggests Best's disease (Best Vitelliform Macular Dystrophy), to be confirmed by genetic testing.

Best's disease is an autosomal dominantly inherited macular dystrophy characterized by bilateral multiple well-circumscribed yellow-orange macular sub-retinal lesions (vitelliform lesions). The disease onset is young (3-15 years of age) but diagnosis is typically delayed until the atrophic stage (after age 40) due to good visual acuity prior to this.

Best's disease usually diagnosed progresses through several stages: Progression through these stages is slow and variable, and may not correspond directly to visual acuity.

Previtelliform stage: Normal macula/early RPE pigment changes. Acuity 6/6.

Vitelliform stage: Well-defined, elevated, round lesion with an egg yolk appearance at the fovea. The rest of the fundus is normal and VA 6/6-6/15.

Pseudohypopyon stage: Yellow material and fluid accumulates in the sub-retinal space. Can occur anywhere in the range of 8-38 years. Acuity is 6/6 – 6/15.

Vitelloruptive stage: Vitelliform lesion breaks up causing a "Scrambled egg" appearance. Pigment changes and atrophy may be seen and there is a moderate reduction in acuity to 6/6 – 6/30.

Atrophic stage: Re-absorption of the yellow material and atrophy of the RPE. Similar appearance to macular degeneration. Significant reduction in visual acuity to less than 6/60.

CNVM/cicatrical stage: Choroidal neovascularisation may develop in response to retinal atrophy.