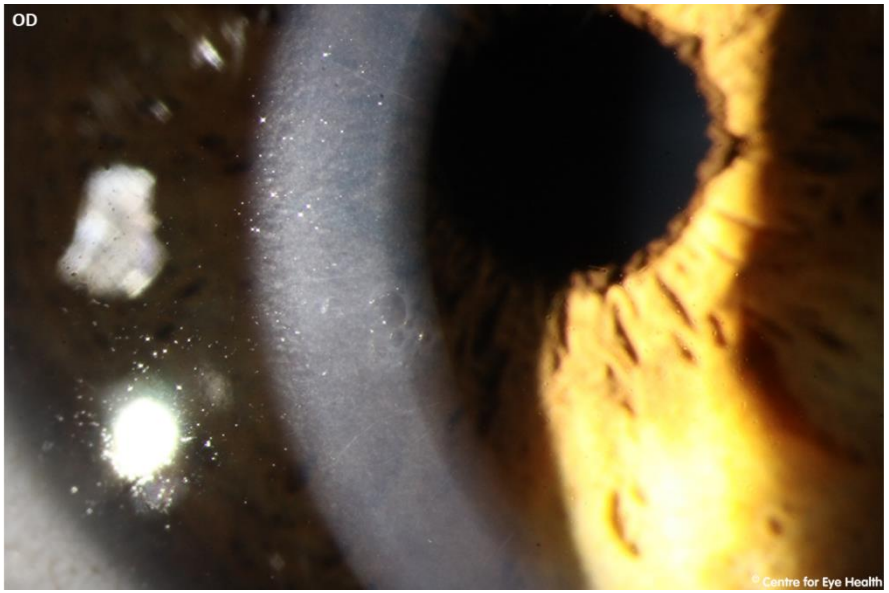
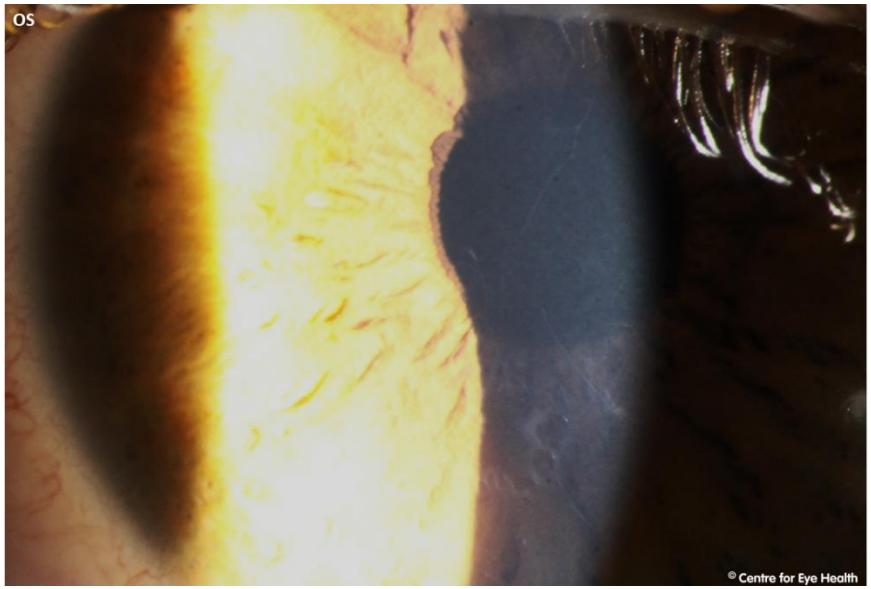
CFEH facebook Cases

CFEH Facebook Case #123

A 12 year old boy was referred for corneal assessment. His vision was 6/6 in each eye uncorrected, he was good general health and asymptomatic. An endothelial cell count was conducted and found 2239 cells/mm2 in the right eye and 1540 cells/mm2 in the left. Corneal photos are below. What is the likely diagnosis and prognosis for this patient?





ANSWER

This presentation appears consistent with asymmetric posterior polymorphous dystrophy with the left eye more advanced than right.

Posterior polymorphous dystrophy (PPMD) is a rare, autosomally dominant inherited condition with variable clinical expression between those affected.

The characteristic signs of PPMD in this patient are the corneal vesicles (circular or oval transparent cysts with a gray halo) at the level of Descemet's membrane. Other features of this condition can include band lesions or diffuse opacities. Band lesions range from 2 to 10 mm long and have parallel scalloped edges. The lesions may show shallow trenches and ridges on specular microscopy when there are a large number of confluent vesicles.

Progression is variable in PPMD and patients may remain asymptomatic or the condition can progress causing corneal oedema and/or irido-corneal adhesions and peripheral anterior synechiae. These developments can cause visual disturbance and increased intraocular pressure, both of which require treatment.