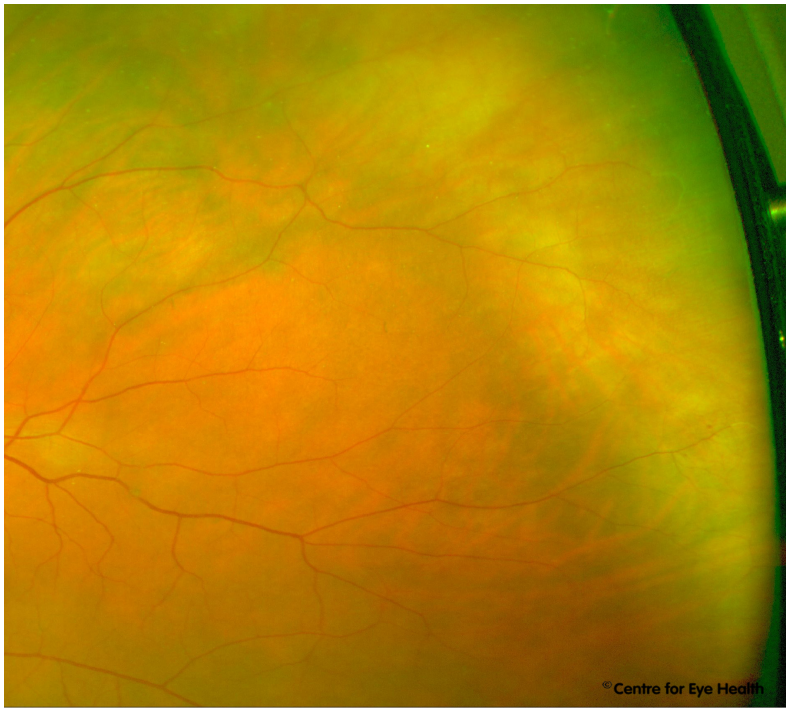
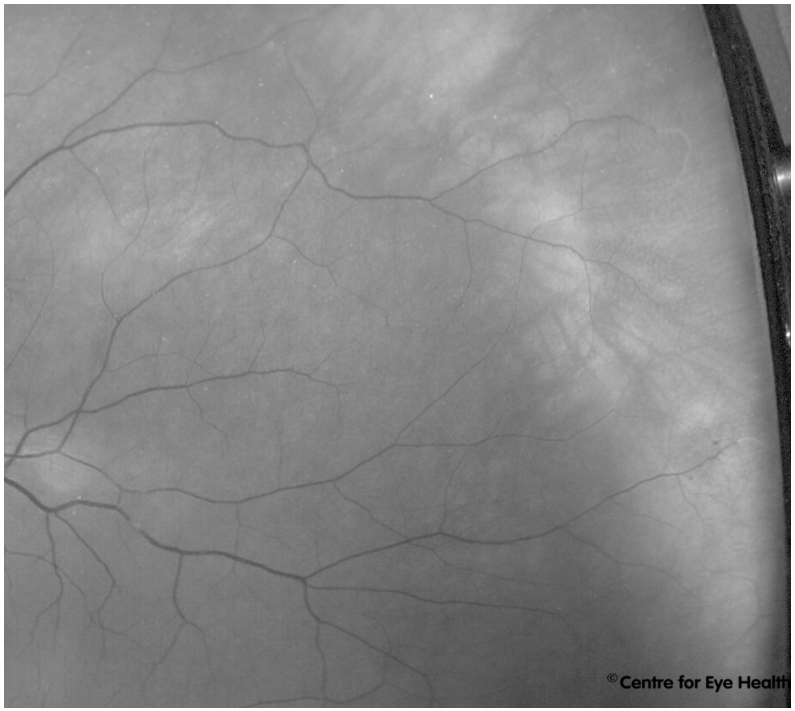
CFEH facebook Cases

CFEH Facebook Case #70

A 61 year old Caucasian male was referred for a peripheral retinal assessment. Aside from a positive history of AMD (mother), his ocular and medical histories are unremarkable. The examining optometrist noted abnormal vasculature in the peripheries of both eyes. What are these findings and what systemic condition is it associated with?





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ANSWER

Hereditary haemorrhagic telangiectasia (HHT).

The Optomap imaging shows regions of retinal vessel telangiectasia, microaneurysms and ghost vessels in the periphery. Upon further questioning, the patient revealed he had already been previously diagnosed with HHT and was under the care of medical practitioner.

HHT is also known as Osler-Weber-Rendu disease and is a rare autosomal dominant condition that results in abnormal blood vessel formation affecting the whole body. These arteriovenous malformations (AVM) occur between arteries and veins, bypassing the capillaries and allowing blood to flow directly into the veins. AVMs are usually fragile and can rupture, resulting in bleeding. This condition can affect the skin, mucous membranes, and organs such as the lungs, liver and the brain.

Medical intervention for this condition aims to decrease the degree of haemorrhages and to prevent complications by AVMs. These patients require long term monitoring as lesions can progress, recur or newly manifest as a result of this condition.