Centre for Eye Health

Condition Spotlight: Macular telangiectasia type 2

Optometry NEW SOUTH WALES AUSTRALIAN CAPITAL TERRITORY

Case report by Christina Ly

A 50 year old female was referred to the Centre for macular assessment due to reduced and distorted vision OS. She had strabismus surgery as a child; ocular history was otherwise unremarkable. Family ocular history and general health were unremarkable.

	OD	**OS**
Entering aided acuities	6/6-1	6/7.5-2
Amsler grid	Unremarkable	Generalised metamorphopsia
Contrast sensitivity – MARS test (normal Range 1.72 – 1.92)	1.72 units	1.68 units

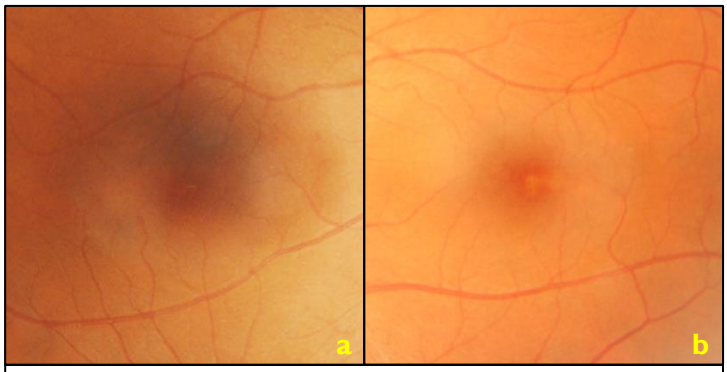

Figure 1. Fundus photography showed a loss of retinal transparency temporal to the fovea OD (a) and hypopigmentary changes temporal to the fovea OS (b).

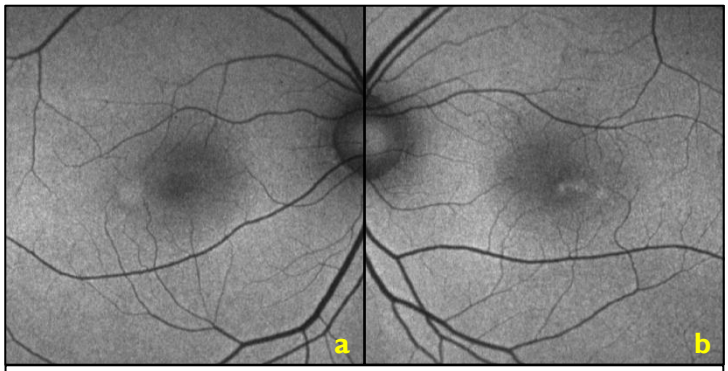

Figure 2. Fundus autofluorescence showed relative hyperfluorescence temporally OU (a, b).

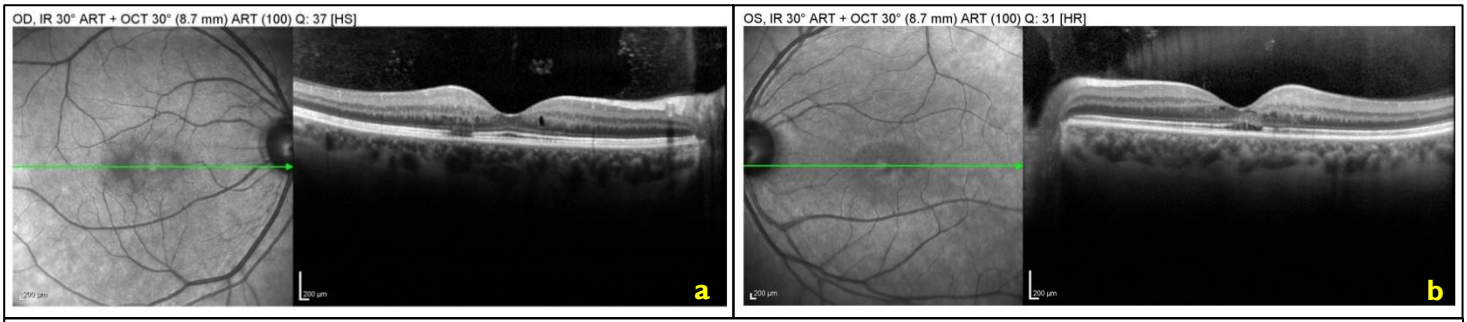

Figure 3. Optical coherence tomography showed a hyporeflective cavity nasally, ellipsoid zone disruption temporally and disorganization of the Henle fibre layer surrounding the foveal pit OD (a). There was a hyporeflective cavity nasally, ellipsoid zone disruption underlying the foveal pit, and disorganization of the Henle fibre layer predominantly temporally OS (b).

Case Summary

The findings were consistent with macular telangiectasia type 2 (MacTel type 2).

Management

Referral to a retinal specialist was recommended.

Clinical Implications

- ***MacTel type 2 displays a spectrum of funduscopic changes over the disease course***

MacTel 2 is a bilateral condition of unknown aetiology characterized by alterations of the macular vasculature and atrophy of the neurosensory retina.[1] Funduscopically, there may be a loss of the foveal reflex and reduced transparency of the retina (Figure 1a) in the early stages. As the disease progresses, there may be crystalline deposits in the inner limiting membrane[2], pigment hyperplasia, and the presence of vitelliform-like yellow spots.[1] Vascular changes include ectatic capillaries, blunted venules, haemorrhages and neovascularisation.[1] These changes occur temporally initially and spread circumferentially around the fovea as the disease progresses.[1] Funduscopic changes can be asymmetric and early changes can difficult to visualize funduscopically.[3]

- ***Advanced imaging modalities may assist in the diagnosis of MacTel type 2***

The prevalence of MacTel type 2 has been found to be 0.004-0.022% in the Melbourne Collaborative Cohort Study[4] and 0.1% in the Beaver Dam Eye Study[5]. These figures are based on assessment of colour fundus images alone, and are therefore likely to underestimate the true disease prevalence, particularly in early cases. Advanced imaging modalities have been shown to be more sensitive in the diagnosis of early MacTel type 2.[6]

Fluorescein angiography (FA) shows hyperfluorescence and leakage from the temporal capillaries in the early stages, which spreads around the fovea as the disease progresses.[7] While FA is considered the gold standard for the diagnosis of MacTel type 2[1], it has been shown to be limited in assessing retinal changes[8]. Fundus auto-fluorescence (FAF) has been proposed as an additional diagnostic tool to detect early retinal changes prior to FA due to its ability to detect sub-clinical changes in lipofuscin distribution in the retinal pigment epithelium.[9] In MacTel type 2, there is relative hyper-autofluorescence due to changes in macular pigment and subsequent lack of foveal masking[1] (Figure 2a and 2b).

Optical coherence tomography (OCT) enables visualization of changes to the retinal architecture. In early disease, there may be temporal enlargement of the foveal pit and eventual development of hyporeflective cavities (Figure 3a and 3b).[1] These cavities do not pool with FA as with exudative cysts[10], and therefore suggests an alternate, possibly atrophic, cause[1]. As the disease progresses, there is disruption of the ellipsoid zone (Figure 3a and 3b) and eventual atrophy of the photoreceptors.[1] In addition to standard OCT, OCT angiography enables visualization of vascular flow using changes in motion contrast created by the movement of erythrocytes without intravenous dye injection.[11] It has been shown to effectively detect areas of microvascular change corresponding to FA leakage in people with MacTel type 2.[12] In addition, there was improved visualization of the vasculature as compared to FA as the vasculature was not obscured by leakage.[12]

References

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