

Type 2 Ideopathic Macular Telangiectasia

BY FRANK G. HOLZ, MD

Our goal as doctors is to prevent visual loss via earlier identification of disease, application of prophylactic medicine, and where necessary, earlier treatment. Using fundus autofluorescence (FAF) imaging along with spectral-domain optical coherence tomography (SD-OCT) with the Spectralis HRA+OCT (Heidelberg Engineering, Heidelberg, Germany), we have been able to detect early pathologies that are unable to be seen using other imaging methods. In this article, we present two pairs of siblings with type 2 idiopathic macular telangiectasia (IMT). There is an underlying genetic component to this disease, and with the SPECTRALIS, we are able to isolate several factors that clue us in to the presence of asymptomatic type 2 IMT.

CASE 1

A 42-year-old asymptomatic woman presented to us with 20/20 vision. We classified the fluorescein angiogram (FA) as normal; the blue laser fundus autofluorescence (FAF) and SD-OCT also appeared normal when viewed separately. When the FAF was compared to the corresponding SD-OCT scan, however, an anomaly was recognized (Figure 1). Usually the foveal dip appears immediately after the area that corresponds to the hyperreflective band (the little bulge in the fovea seen in Figure 1). For our patient, however, this was not the case. The thinnest part of the fovea, rather, was temporal to where the anatomic fovea sits. After running a volume scan we confirmed that the deepest point of the fovea was not properly located (Figure 4).

So why were we taking FA, FAF, and OCTs for a patient with no apparent pathology? The patient's sister had a classic case of type 2 IMT. Although the patient was asymptomatic with 20/20 vision, we suspected that she had type 2 IMT without leakage due to the thinning temporal to the fovea. We found the Spectralis BluePeak blue laser autofluorescence (Heidelberg Engineering) as seen in Figure 2 to be helpful in making this diagnosis in the patient's sister. The confocal red-free imaging provides a markedly increased signal in a noninvasive manner that picks up any area of blue reflectance. FAF shows a maldistribution because of a depletion of luteal pigment, which is pathognomonic to type 2 IMT in the later stages.

CASE 2

In another set of siblings, we had similar findings. In Figure 3, there is again displacement of the thinnest part of the fovea, so there must be some atrophy, loss of cells, or cytoplasmic volume in the neurosensory retina. Had we put this automatic volume scan over the deepest pit we would not have picked this up. With the Spectralis, however, the location of the fovea can be moved to where it makes the most sense. The other angiographic features of early type 2 IMT are seen in Figure 4; CBR shows that there is abnormality. In the twin, the angiographic findings show more advanced disease (Figure 5).

DISCUSSION

It is important to diagnose this disease as early as possible.

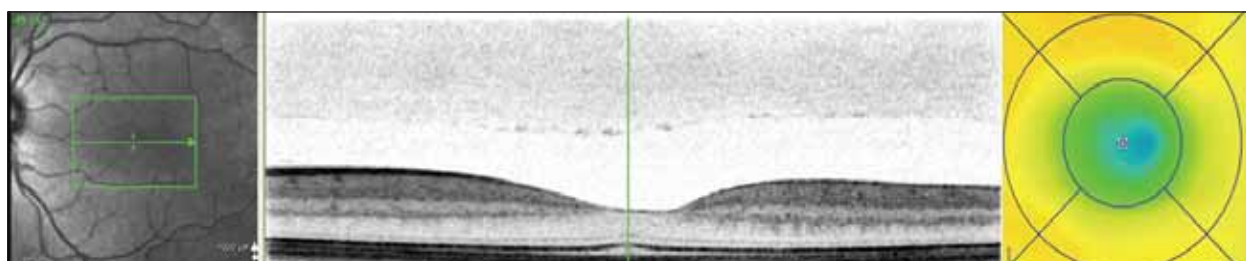


Figure 1. The thinnest part of the fovea was temporal to where the anatomic fovea sits. After running a volume scan we confirmed that the deepest point of the fovea was not properly located.

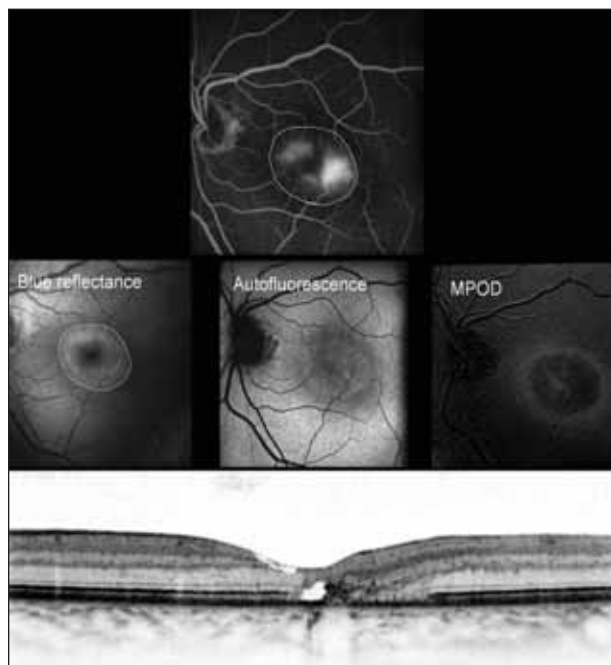


Figure 2. CBR imaging provides an increased signal in a non-invasive manner that picks up any area of blue reflectance. The two-wavelength Heidelberg angiograph has a green light and a blue laser for creating autofluorescence to achieve a quantitative measure of luteal pigment.

Type 2 IMT is much more common than we previously thought. It is probable that with standard imaging methods we missed a large proportion of patients for whom we could achieve better outcomes had we diagnosed them earlier. In the Spectralis we have three components to improve our diagnostics of this disease. SD-OCT allows us to see thinning temporal to the fovea and FAF aids in the detection of the loss of luteal pigment. Red-free imaging captures increased reflectivity. As result, we can identify the disease

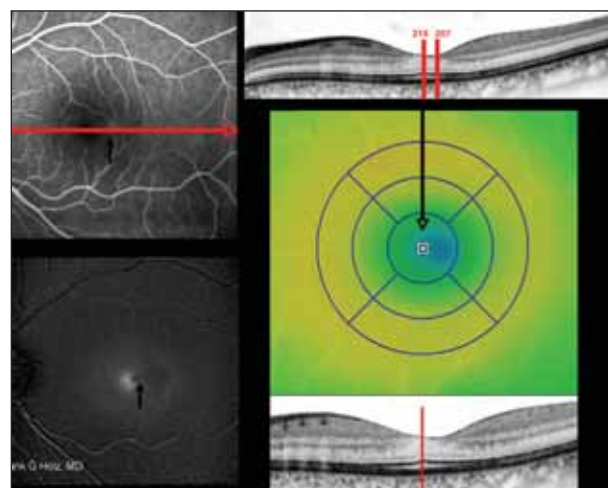


Figure 3. Displacement of the thinnest part of the fovea, suggesting atrophy, loss of cells, or cytoplasmic volume in the neurosensory retina.

earlier before it becomes apparent on FA with the added benefit of avoiding an invasive procedure.

In conclusion, the Spectralis is an important advance in achieving a better understanding of the genetic components type 2 IMT, both for earlier diagnosis and a better understanding of the disease itself. ■

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1. Helb HM, Charbel Issa P, Van Der Veen RL, et al. Abnormal macular pigment distribution in type 2 idiopathic macular telangiectasia. *Retina*. 2008;28(6):808-816.
2. Gillies MD, Zhu M, Chew E, et al. Familial asymptomatic macular telangiectasia type 2. *Ophthalmology*. 2009;116(12): 2422-2429.



Figure 4. Angiographic features, CBR, two and one wavelength autofluorescence.

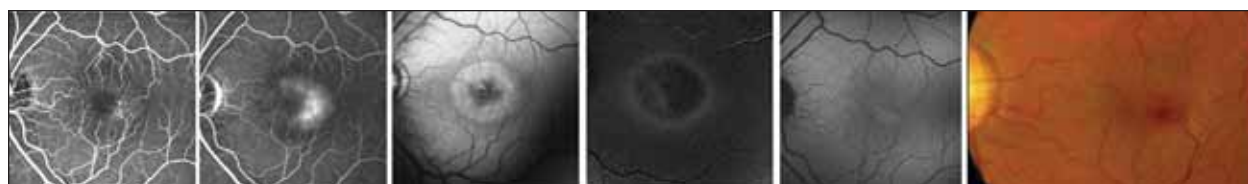


Figure 5. Findings in a more advanced case.