Natural History and Optical Coherence Tomography (OCT) Characteristics of Macular Telangiectasia Type 2

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Abstract

Macular telangiectasia type 2 (MacTel type 2) is a bilateral retinal degeneration that is usually diagnosed in older adults presenting with symptoms of decreased vision and metamorphopsia. Clinical retinal examination can often be unrevealing. However, optical coherence tomography (OCT) reveals characteristic hyporeflective cavities temporal to the foveal center without retinal thickening. This case presents a relatively stable natural course of MacTel type 2 presented over an 8-year time period.

Case History

- **73 year old Caucasian male**
- **Chief complaint(CC):** Blurred vision OD greater than OS over several years
- **Ocular history:** Nuclear cataracts and dry eye OU
- **Family ocular history:** Unremarkable
- **Medical history:** All well controlled: diabetes mellitus type 2 for 30 years, hypertension, hyperlipidemia, hypothyroidism, asthma
- **Medications**: glipizide, metformin, lisinopril, atorvastatin, levothyroxine, albuterol

Pertinent Findings

Date	Visual Complaint	Best corrected visual acuity	OCT findings
08/30/10	Right eye vision is distorted	Right: 20/30 Left: 20/25	• Outer retinal cysts temporal to foveal center, both eyes (Fig 1a, b) • Loss of foveal contour, both eyes (Fig 1 a, b) • Break in photoreceptor inner and outer segment (IS/OS) junction temporal to foveal center, right eye (Fig 1a)
07/02/12 to 09/14/15 (6 visits)	Patient reports stable vision	Right: 20/30- Left: 20/25+ (stable)	• Break in IS/OS junction temporal to foveal center, left eye (Fig 2)
03/30/15 to 03/16/18 (5 visits)	Patient reports stable vision	Right: 20/40+ Left: 20/20 (stable)	• Collapse of outer plexiform layer (OPL) and diminished retinal cyst size, right eye (Fig 3)
07/13/18	Blurry vision with current glasses	Right: 20/40+ Left: 20/20-	• Atrophic outer retina, right eye (Fig 4)

Differential Diagnosis

- Primary: MacTel type 2
- Leading:
  - Neovascular ARMD
  - Retinal vein occlusion with macular edema
  - Diabetic maculopathy with macular edema
  - Lamellar macular hole

MacTel2 Stages and OCT Findings[1]

Stage	OCT Findings
1 (Fig 1a)	• Disruption in IS/OS junction line temporal to foveal center • Inner and outer retinal cysts temporal to foveal center
2 (Fig 3)	• Larger break in IS/OS reaching foveal center • Inner and outer cysts developing nasal to foveal center • "Collapsing layers" of OPL and other internal layers extending toward choroid • Hyperreflective intraretinal lesion
3 (Fig 4)	• Break in IS/OS reaches nasal area near foveal center • Minor retinal cysts • Extensive "collapsing layers" • Hyperreflective intraretinal lesion
4	• Signs of neovascularization located in or external to outer neurosensory retinal layers associated with subretinal or intraretinal fluid

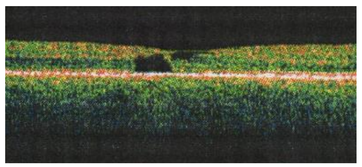

Figure 1a. OD

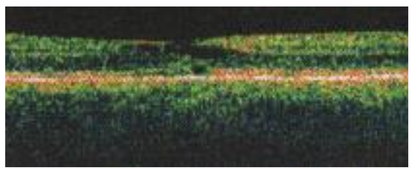

Figure 1b. OS

Patient's retinal appearance at baseline. OD was more advanced at initial exam due to presence of the break in the IS/OS junction

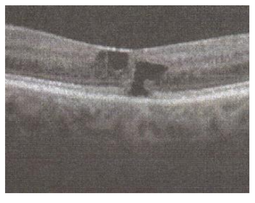

Figure 2. OS shows progression with development of IS/OS junction breaks 3 years after initial visit

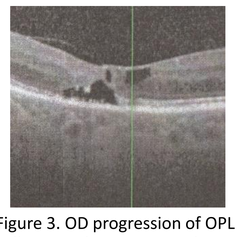

Figure 3. OD progression of OPL collapse with a corresponding decrease in VA

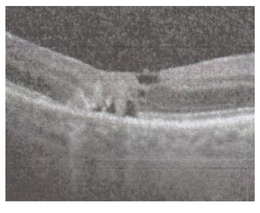

Figure 4. OD progression with atrophy of the outer retina. The VA remained stable

Discussion

- Diagnosis of macular telangiectasia type 2 was supported by:
  - Fluorescein angiography findings from retinal consultation
  - Bilaterality
  - Reduced retinal transparency "retinal graying" in parafoveolar area
  - Blunted, dilated venules along with right angle venules
  - OCT findings throughout the patient's exam visits that represent complications of MacTel2 progression include:
    - OD: progressive foveal atrophy
    - OS: development of full thickness macular hole – intraretinal cyst temporal to foveal center with intact internal limiting membrane (ILM).

Other Imaging Techniques

Imaging techniques for MacTel type2 in other patients:

- Fluorescein angiography (FA):
  - Telangiectatic capillaries (Fig 5)
  - Hyperfluorescence (Fig 6)
- Fundus autofluorescence: Loss of foveal masking (Fig 7)
- OCT angiography (Fig 8):
  - Early stage: mild dilation of superficial capillary plexus and deep capillary plexus temporal to fovea
  - Late stage: Dilated venules over entire macula
- Confocal blue reflectance: Increased parafoveolar reflectance (Fig 9)

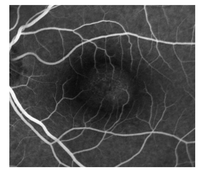

Figure 5. Representative photo of early stage (FA)[1]

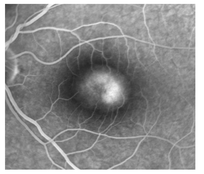

Figure 6. Representative photo of late stage (FA)[1]

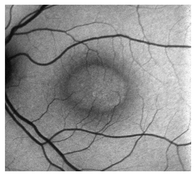

Figure 7. Representative photo of fundus autofluorescence[1]

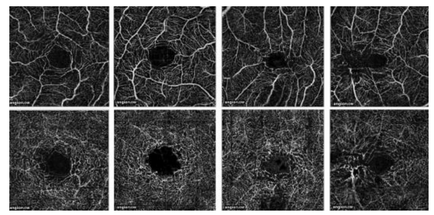

Figure 8. Representative photo of early to late stage OCT angiography findings of the superficial capillary plexus (top) and deep capillary plexus (bottom)[1]

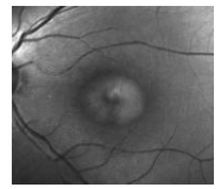

Figure 9. Representative photo of confocal blue reflectance[1]

Treatment

- Due to low prevalence and few successful treatment studies, current management includes observation. Intervention is necessary with disease sequelae (choroidal neovascularization and macular hole)[2]
- The patient will continue to be monitored every 6 months

Conclusion

- Although FA is the gold standard for diagnosis, OCT may be used for management and monitoring.
- True prevalence is likely underestimated due to lack of funduscopic findings[2,3]
- This case is unique in that the patient elected to be monitored without intervention providing insight into the natural course of the condition.
- MacTel2 is likely genetic due to bilaterality and dominant inheritance due to vertical transmission[2]
- Potential systemic association: diabetes[4]

References

1. Toto, Lisa, *et al.* Multimodal Imaging of Macular Telangiectasia Type 2: Focus on Vascular Changes Using Optical Coherence Tomography Angiography. *Invest Opthalmol Vis Sci.* 2016; 57(9): 268-276.
2. Issa, Peter Charbel, *et al*. Macular Telangiectasia Type 2. *Prog Retin Eye Res*. 2013; 57: 49–77.
3. Wu, Lihteh. When Is Macular Edema Not Macular Edema? An Update on Macular Telangiectasia Type 2. *Taiwan J. Ophthalmol.* 2015; 5(4): 149–155.
4. Jhingan, Mahima, *et al*. Idiopathic Macular Telangiectasis Type 2 and Co-Existent Diabetic Retinopathy. *International Journal of Retina and Vitreous*. 2017; 3(1): 50