Macular Telangiectasia

Disease

Macular telangiectasia (Mac Tel) leads to abnormalities of capillaries of the fovea or perifoveal region associated with loss outer nuclear layers and ellipsoid zone that can progress to cystic cavitation-like changes in all retinal layers, or development of full-thickness macular hole or subretinal neovascularization in advanced stages. First described by Gass in 1968 as a different entity from Coats disease.

Mac tel is usually divided into 3 main groups:

- **Type 1**: congenital and unilateral. Thought to be similar to and/or possibly a variant of Coats disease. Uncommon.
- **Type 2**: acquired and bilateral. The most common form of the three types. Usually found in middle-aged or older patients. According to the Beaver Dam Eye Study which graded only color fundus images, Mac Tel has a prevalence of 0.1% with an average age of 63 years. Although not initially described by Gass, Yannuzzi found a slight female preponderance of 58%. Of note, on common usage, the term 'Mac Tel' is often used to refer to Mac Tel type 2. This entry will largely focus on type 2 as it has the most clinical relevance.
- **Type 3**: an poorly understood primarily occlusive phenomena which is quite rare.

Etiology

The Mac Tel Project found a prevalence of diabetes mellitus (DM) of 28% and hypertension (HTN) of 52% in Mac Tel type 2. Such high prevalence of DM, HTN and the age of onset of the disease can point toward a long-term vascular stress as the etiology for this entity. These vascular changes are associated with disruption of the outer nuclear layer, ellipsoid zone and lead to pseudolamellar macular holes. RPE hyperplasia and migration into the inner retinal layers is also seen with time.

Risk Factors

Risk factors include age as it is mainly encountered in middle-aged and older patients. Higher prevalence of hypertension and diabetes mellitus have been found and could play a part in association with neurodegenerative processes.

General Pathology

Gass postulated that Muller cells and parafoveal neural abnormalities or degeneration may be the primary sites initially affected. Histopathology has shown disruption of pericytes, ectatic vessels, with dilated venules diving at a right angle into deeper retinal layers with evidence of RPE hyperplasia along them. Vascular occlusion, inflammatory changes and other neurodegenerative processes lead to loss of the outer nuclear layer, ellipsoid zone with later formation of cysts that can encompass all retinal layers.

Pathophysiology

Chronic neurodegenerative processes, vascular inflammation, occlusion and ectatic capillary changes lead to loss of outer retinal layers and formation of pseudolamellar macular holes with cysts or cavitations that can also be in inner retinal layers. Patients can develop subretinal neovascularization temporal to the fovea and can lead to exudates, hemorrhages and in late stages a disciform scar.

Primary prevention

No prevention has been shown to be beneficial, however most clinicians would recommend control of hypertension and diabetes mellitus if present as these two diseases have been shown to be more prevalent in mac tel.

Diagnosis

The diagnosis of Mac Tel is made by ophthalmic clinical exam and ophthalmic imaging. Symptoms and history can help in diagnosis. No labratory test are helpful in diagnosis.

History

If asked, most patients with Mac Tel type 2 will complain of metamorphopsia or scotoma. With progression of the disease, an increased scotoma can be perceived with decreased visual acuity as this entity affects the fovea and perifoveal area. However visual acuity rarely reaches legal blindness, with 50% keeping 20/32 or better vision according to the Mac Tel Project. Vision can decrease if a macular hole develops. Vision can be severely diminished in Mac Tel type 1 and 3.

Physical examination

In Mac Tel type 2, fundoscopic exam is initially significant for a decrease of foveal pit, with subsequent foveal or perifoveal ectatic vessels with possible presence of venules diving at a right angle into deeper retinal layers. Grayish discoloration of temporal perifoveal area is the earliest sign present. Ectatic vessels with evidence of venules diving at a right angle into deeper retinal layers. Of note, these findings may be subtle on routine retinal exam. Subretinal neovascularization temporal to fovea may be seen in this disease and can be associated with exudates or hemorrhage. RPE hyperplasia can also be seen at the fovea or temporal perifovea. Also, in later stages, pseudolamellar holes can develop into full-thickness macular holes.

Imaging

Fluorescein angiography (FA) shows temporal foveal telangiectatic vessels in Mac Tel type 2 that leak in later stages. Evidence of venules diving at a right angle into deeper retinal layers can also be seen. Subretinal neovascularization is evidenced to be retinal in origin as FA shows a feeding arteriole and a draining venule.

Optical coherence tomography (OCT) shows temporal foveal pit enlargement secondary to loss of outer nuclear layer and ellipsoid zone that can progress into large cysts (often called 'cavitation') that can encompass all retinal layers. Often only the internal limiting membrane is left in place over these areas, lending to the term of an "ILM drape." Hyperreflective areas on OCT correlate to areas of RPE hyperplasia and migration.

Fundus autofluorescence (FAF) shows decreased foveal hypofluorescence secondary to loss of foveal luteopigment. Areas of RPE hyperplasia will appear as hypofluorescent spots on FAF.

Differential diagnosis

Differential diagnosis of retinal capillary telangiectasia include branch retinal vein occlusion, diabetic retinopathy, radiation retinopathy. In cases of neovascularization, age-related macular degeneration should be ruled out.

Management

There has been no randomized clinical study for this entity. In non-neovascular cases, laser, intravitreal anti-VEGF or steroid has not been shown to be effective in controlling the disease. For neovascular cases, intravitreal anti-VEGF are the mainstay of treatment, although transpupillary therapy and photodynamic therapy have been used in the past. Neuroprotective agents are currently being investigated. A Phase II trial studying intraocular delivery of ciliary neurotrophic factor (CNTF) in eyes with Mac Tel found decreased ellipsoid zone loss, increased macular thickness, and stable reading speed when compared to controls. Phase III trial is currently underway. Associated full thickness macular holes may be treated with pars plana vitrectomy surgery, membrane peeling and gas tamponade, but are likely to have a lower-than-average closure rate.

Prognosis

According to the Mac Tel Project, 50% of type 2 patients with have a visual acuity of 20/32 or better, with a mean visual acuity of 20/40. Mac Tel can rarely lead to 20/200 or worse vision when associated with full-thickness macular hole or subretinal neaovascularization that can lead to a disciform scar in very advanced stages.

Additional Resources

- Boyd K, Vemulakonda GA. Macular Telangiectasia. American Academy of Ophthalmology. EyeSmart® Eye health. https://www.aao.org/eye-health/diseases/macular-telangiectasia-list. Accessed March 18, 2019.

References

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2. Wu L, Evans T, Arevalo JF. Idiopathic macular telangiectasia type 2 (idiopathic juxtafoveolar retinal telangiectasis type 2A, Mac Tel 2). Surv Ophthalmol. 2013 Nov-Dec;58(6):536-59.

3. Charbel Issa P, Gillies MC, Chew EY, Bird AC, Heeren TF, Peto T, Holz FG, Scholl HP. Macular telangiectasia type 2. Prog Retin Eye Res. 2013 May;34:49-77.

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