I. CASE HISTORY
A 62 year-old Caucasian male presented to the Salt Lake City Department of Veterans Affairs (VA) with a complaint of gradual vision loss and a central scotoma manifest as missing letters when reading. Symptoms had been gradually worsening over the past five years, and he had been told by his previous optometrist that he had macular degeneration. His systemic health was significant for acid reflux, for which he was taking omeprazole; he otherwise reported to be in good health with no other systemic conditions, including diabetes and hypertension. He reported a positive family history of macular degeneration (mother).

II. PERTINENT FINDINGS
Entering acuities were 20/50+ in each eye with no improvement by pinhole. Pupils and extraocular muscle movements were normal both eyes. Anterior segment findings were significant for mild meibomian gland inspissation and grade 2 nuclear sclerosis in both eyes. Intraocular pressures were 17 and 15 mmHg.

Dilated fundus examination revealed parafoveal retinal graying with an absence of the foveal reflex in both eyes, with a faint yellow appearance at the fovea OS.

Optical coherence tomography (Heidelberg Spectralis) revealed distinct perifoveal cystic spaces at the level of the inner nuclear layer with accompanying loss of the inner segment/outer segment junction (IS/OS, also referred to the ellipsoid zone). No drusen or epiretinal membrane were evident.

Fundus autofluorescence (Heidelberg Spectralis, 488nm) revealed a lack of normal central foveal hypofluorescence.

Fluorescein angiography revealed early hyperfluorescence of perifoveal vessels in a telangiectatic pattern with late diffuse hyperfluorescence and leakage in both eyes.

III. DIFFERENTIAL DIAGNOSIS
Primary
Idiopathic juxtafoveal macular telangiectasia (MacTel) Type II
Other
Diabetic retinopathy
Retinal vein occlusion
Retinal angiomatous proliferation
Age related macular degeneration

IV. DIAGNOSIS AND DISCUSSION
• MacTel Type 2 is a bilateral disease with hallmark features of distinctive alterations of the perifoveal capillaries, inner and outer nuclear cystic changes, and neurosensory atrophy with photoreceptor loss.
• First signs most often occur temporal to the fovea; disease does not respect the horizontal midline
• Onset typically in the fifth or sixth decade of life.
• Bilateral but may be asymmetric
• Often a positive family history

• Nomenclature
  o Should be distinguished from less common MacTel type I (aneurysmal) and MacTel Type III (occlusive)
• **Characteristic findings**
  
  *Biomicroscopy:*
  - Blunting or lack of the foveal reflex
  - Reduced retinal transparency
  - Crystalline deposits
  - Mildly ectatic capillaries and blunted, dilated venules
  - Foveal atrophy
  - Retinal pigment plaques
  - A round yellow spot
  - Neovascularization (late stages)

  *Fluorescein angiography:*
  - Telangiectatic capillaries (often temporal) in the early phase, with diffuse hyperfluorescence/leakage in the late phase.

  *Optical coherence tomography:*
  - Hyporeflective inner and outer retinal cystic spaces
  - Disruption of IS/OS junction
  - Hyperreflective intra or subretinal lesions due to pigment migration or neovascular membranes (late stage)
  - Outer neurosensory atrophy

  *Fundus Autofluorescence*
  - Increased foveal autofluorescence, or decreased normal hypo-autofluorescence

V. TREATMENT, MANAGEMENT

• Historically therapeutic approaches have had limited or no efficacy.
  - Lutein and zeaxanthin oral supplements: therapeutic trials showed that depleted areas of macular pigment do not re-accumulate.
  - Anti-VEGF: clinical trials have not shown significant efficacy for the nonneovascular disease stages.

• Currently a phase 1 clinical trial is being conducted testing the impact of implantable ciliary neurotophic factor (CNTF). Consists of encapsulated human retinal pigment cells genetically modified to secrete therapeutic doses of CNTF into the retina, with the goal of regenerating neurosensory cells.

• Ongoing research is being done as part of The Macular Telangiectasia Project (“MacTel Project”). Since 2005, it is a privately funded multicenter project which aims to develop a better understanding of pathogenesis, disease process, role of genetics, and develop new treatments for MacTel.

VI. CONCLUSION

MacTel may commonly be misdiagnosed as age-related macular degeneration or conditions such as diabetic retinopathy, but should be distinguished. With ongoing developments in treatment, it is important to identify MacTel and appropriately address this unique disease process. Awareness among optometrists and understanding the key clinical features will ensure accuracy and prevent the delay of the diagnosis.
REFERENCES


