CASE REPORT

Macular telangiectasia type 2 (MacTel) in a 34-year-old patient

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SUMMARY

We report macular telangiectasia type 2 (MacTel) in a 34-year-old man, the youngest patient so far published with MacTel type 2. The patient presented with metamorphopsia and impaired reading ability. Diagnosis was based on bilateral abnormal macular autofluorescence, perifoveal telangiectasia with fluorescein angiographic hyperfluorescence without cystoid oedema, a small foveal avascular zone, asymmetric configuration of the foveal pit, disruptions in the inner segment/outer segment layer and hyper-reflective haze and spots in the outer nuclear layer. Although MacTel usually manifests with a slowly progressive decrease in visual acuity in the fifth to seventh decades of life, younger patients may occasionally be diagnosed with the disease. Awareness of subtle signs of the condition is essential for early diagnosis.

BACKGROUND

Macular telangiectasia type 2 (MacTel) is a relatively rare macular disease that usually manifests with a slowly progressive decrease in visual acuity in the fifth to seventh decades of life.^{1–4} Visual acuity may decline as a result of degenerative changes or neovascularisation and scar formation.⁵ The advanced disease resembles age-related macular degeneration. In addition to the diagnostic gold standard of fluorescein angiography,^{5–7} several non-invasive imaging techniques, such as fundus autofluorescence and optical coherence tomography (OCT)^{3, 8} can contribute to the diagnosis and are required for early diagnosis of the disease. Familial screening of symptomatic patients has demonstrated familial occurrence with both symptomatic and asymptomatic cases.^{9, 10} An underlying dominantly inherited genetic abnormality of variable penetrance and expressivity is suspected.¹⁰ Clinicopathological studies¹¹ and image analysis^{2, 3, 8} have demonstrated characteristic abnormalities in this condition, including macular pigment depletion and loss of both Müller's cells and photoreceptors. We report MacTel type 2 in a 34-year-old man who is, to the authors' knowledge, the youngest patient described in the literature to date.

CASE PRESENTATION

An otherwise healthy 34-year-old man experienced metamorphopsia, impaired reading ability and difficulties with computer work. Best-corrected visual acuity was 20/20 in both eyes with refractive correction of −2.75D right eye (RE) and −3.50D left

eye (LE). Mild metamorphopsia OU was demonstrated on the Amsler grid. Biomicroscopy of the anterior segments was normal. A myopia-associated temporal atrophic conus was observed on both optic discs. The macular reflex was absent bilaterally.

INVESTIGATIONS

Fundus autofluorescence showed bilateral loss of the normal central attenuation (figure 1A). Fluorescein angiography demonstrated telangiectatic capillaries invading the central macular area. This produced a very small avascular zone (figure 1B) and over 360° of diffuse perifoveolar late hyperfluorescence without cystoid macular oedema (figure 1C). The area of late diffuse hyperfluorescence corresponded with the area of increased autofluorescence, which suggested the diagnosis of MacTel type 2. Spectral domain OCT (SD-OCT) using axial scans showed bilaterally symmetric quantitative and qualitative macular abnormalities compatible with the presumed diagnosis. The macular thickness in the anatomic foveal centre was only 222 µm in the RE and 212 µm in the LE (figure 2). These values are on the lower border of values expected in a 34-year-old man with only 3D of myopia.¹² In addition, the foveal pit was asymmetric, with the thinnest retinal region located slightly temporal to the anatomic foveal centre. Here, the thickness was reduced to 176 µm in the RE and 180 µm in the LE. The outer nuclear layer had an irregular thickness, with both thickened and thinned areas, and demonstrated a diffuse hyper-reflective haze and several hyper-reflective spots. A focal attenuation of the hyper-reflective signals and a small break could be identified in the hyper-reflective inner segment/outer segment (IS/OS) layer that is assumed to correspond with the ellipsoids of the photoreceptor inner segments. Neither cysts nor inner retinal abnormalities were noted.

First-degree relatives of the patient were screened. No signs of maculopathy were identified in his brother and mother. However, his father has type 2 diabetes mellitus and demonstrated a hyper-reflective haze and spots in the outer nuclear layer temporal to the foveal centre on SD-OCT.

DIFFERENTIAL DIAGNOSIS

This case report supports the opinion that the early changes seen in MacTel type 2 can be difficult to detect, and that the diagnosis can be missed or delayed. Identification of cases, or of asymptomatic family members, does not necessarily require fluorescein angiography, but can be accomplished using

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Figure 1 Left eye of the index patient. (A) Autofluorescence imaging shows loss of the normal central attenuation. (B) The early phase fluorescein angiogram showing telangiectatic capillaries invading the central macular area with very small avascular zone. (C) The 10 min fluorescein angiogram showing perfoveal diffuse hyperfluorescence corresponding to telangiectatic capillaries and increased autofluorescence.

the non-invasive techniques of SD-OCT and autofluorescence imaging.

TREATMENT

There is currently no treatment available for the early and atrophic stages of MacTel. However, neovascularisation associated with MacTel can be treated with intravitreal antivascular endothelial growth factor injections, which target neovascularisation¹³ and reduce the risk of large atrophic scars resulting from neovascularisation. CNTF (ciliary neurotrophic factor) therapy, which employs a vitreous implant containing genetically modified human cells that secrete neurotrophic factor, is under investigation.

DISCUSSION

MacTel type 2 is a rare and potentially blinding macular disease usually diagnosed during the fifth to seventh decades of life.¹⁻⁴ The maculopathy manifests as a slowly progressive decrease in visual acuity, which may occur when degenerative changes or neovascularisation and subsequent scar formation reach the foveal centre.⁵ Biomicroscopic findings may include reduced retinal transparency, crystalline deposits, mildly ectatic capillaries and, in more advanced disease, blunted venules, retinal pigment plaques, foveal atrophy and neovascular complexes.^{3, 5} The characteristic early-phase fluorescein angiographic findings are parafoveal telangiectatic capillaries predominantly temporal to the fovea; late-phase angiography demonstrates diffuse hyperfluorescence without cystoid oedema.³ Moreover, the telangiectatic capillaries may invade the central macular area and reduce size of the avascular zone.^{6, 7}

Diagnostic capabilities have become more advanced. In the past, the diagnosis was based on the identification of characteristic abnormalities by biomicroscopic examination and fluorescein angiography.⁵⁻⁷ However, OCT has become the gold standard for confirmation and for early diagnosis of the disease.^{3, 8, 9} In advanced stages, SD-OCT may reveal large defects in the IS/OS layer, hyporeflective cavities in the inner or outer retina, neovascular complexes and retinal atrophy. Early retinal alterations include attenuation of the hyper-reflective signals in the IS/OS layer, outer retinal hyper-reflective spots and a hyper-reflective haze, and an asymmetric configuration of the foveal pit, which is the thinnest temporal of the foveal centre.^{2, 4, 8, 9} Interestingly, eyes with MacTel type 2 have a unique local depletion of the macular pigment,¹⁴ and depleted areas cannot reaccumulate lutein and zeaxanthin, even after oral supplementation.³ Loss of macular pigment can be evaluated via analysis of macular pigment density, but can also be demonstrated in a simple, non-invasive manner with blue (488 nm) autofluorescence imaging.³

Postmortem histology from a patient with MacTel type 2 showed correlations between clinical and histological data. This includes clinically recorded macular pigment depletion that closely matched the loss of Müller's cells on histological examination and a good correlation between IS/OS layer loss as seen on OCT and histological rod depletion.¹¹ The authors speculate that Müller's cell dysfunction is an early and possibly causative feature of MacTel type 2.¹¹

Although the disease is bilateral, it can manifest asymmetrically, with the fellow eye seemingly unaffected and showing only very subtle anomalies on SD-OCT, autofluorescence and fluorescein angiography. Familial occurrence is not exceptional and reports of vertical transmission in families suggest a dominantly

Figure 2 Left eye of the index patient. Horizontal spectral domain optical coherence tomography scan 100 μ superior to the foveal centre showing a relatively flat and asymmetric slope with a thickness of 212 μ superior to the fovea and 180 μ in the thinnest area temporal to the fovea. The outer retina is irregular, with thinned and thickened areas, and showing a diffuse hyper-reflective haze. The inner segment/outer segment line is focally attenuated.

inherited genetic abnormality of variable penetrance and expressivity.^{3 8 9}

The MacTel project is an international observational clinical study designed to evaluate the structural and functional changes associated with MacTel.¹ In addition, the pathobiology of the disease will be assessed in order to improve the understanding of its pathogenesis and potential treatment. In 2008, a total of 310 participants were enrolled.¹ To date, over 600 participants are included in the study.^{15 16} Of these patients, the mean age at the baseline examination was 61±9 years (range 36–83 years) and the mean duration between diagnosis and baseline examination was 3 years (range 0–25 years). The medical characteristics of patients with MacTel were analysed and the prevalence of various systemic conditions was compared with age-matched and sex-matched controls. Patients with MacTel had significantly increased prevalence of diabetes mellitus.¹⁵ The current study demonstrates that MacTel can occur in patients of much younger age than usually expected, and that the clinician must

be aware of this possibility to avoid a delayed or missed diagnosis.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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Learning points

- ▶ Although macular telangiectasia type 2 (MacTel) usually manifests with a slowly progressive decrease in visual acuity in the fifth to seventh decades of life, occasionally younger patients are diagnosed with the disease.
- ▶ The diagnosis of MacTel can be delayed or missed. In early disease visual acuity is hardly affected and the ocular anomalies are often subtle and difficult to detect. Advanced disease resembles age-related macular degeneration.
- ▶ Identification of cases or affected family members does not necessarily require fluorescein angiography, but can be done using the non-invasive techniques of spectral domain optical coherence tomography and autofluorescence imaging.
- ▶ Diabetes mellitus is prevalent in MacTel cohorts. Screening for diabetes mellitus should be part of the investigations in patients with MacTel type 2. Moreover, a family history of diabetes mellitus, as described in this report, can be a hint for identification of affected family members.

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