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Treatment of macular telangiectasia type 2. A review of recent research findings and a case report

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Abstract: **Introduction:** Macular telangiectasia type 2 (MacTel 2) is a chronic degenerative retinal disease. In everyday ophthalmic practice, both its early and advanced stages pose significant diagnostic and therapeutic challenges.
The aim of this paper is to discuss novel therapeutic options reported in the literature and to present a proposed diagnostic and therapeutic management strategy for patients with advanced disease complicated by secondary pathologies.
Case report: A 57-year-old otherwise healthy woman presented with deterioration of both near and distance vision despite using her current spectacles. Multimodal retinal imaging confirmed the diagnosis of macular telangiectasia type 2. Due to the presence of active neovascularization, the patient was scheduled for intravitreal anti-vascular endothelial growth factor (anti-VEGF) therapy. Following the injection, best-corrected visual acuity initially improved and has remained stable thereafter.
Conclusions: MacTel 2 is a condition encountered in routine clinical practice. Since 2025, the first gene therapy – revakinagene taroretcel (Encelto) – has been approved in the United States for the treatment of the non-proliferative form of the disease. In cases complicated by macular neovascularization, intravitreal administration of anti-vascular endothelial growth factor agents is required.

Key words: macular telangiectasia, retinal degeneration, optical coherence tomography, macular neovascularization.

Introduction

Idiopathic macular telangiectasia type 2 (MacTel 2) is a neurodegenerative disease that can progress unnoticed by the patient for many years; its early symptoms are very difficult to detect during a routine ophthalmic examination.

MacTel 2 affects approximately 0.0045% [1] to 0.1% of the population [2]. It is typically diagnosed in patients around 60 years of age, and no clear sex predominance can be identified [1–3]. To date, the genetic basis of MacTel 2 has not been definitively established. However, the apparent genetic penetrance in observational studies is 0.35 [4]. According to Charbel et al. [5], there are strong indications of a genetic basis for MacTel 2, such as the bilateral presentation of symptoms and the occurrence of the disease in monozygotic twins, parents, and siblings. Nevertheless, studies conducted so far have not conclusively identified the genetic defect responsible for the condition.

The suggested pathomechanism of MacTel 2 remains unclear. Current research suggests that the disease is neurodegenerative in nature and is associated with Müller cell dysfunction. Müller cells are glial cells that provide structural support for retinal neurons and perform several essential functions, including maintaining ionic homeostasis, nourishing neurons, and regulating water balance. The loss of Müller cells consequently leads to thinning of the outer retinal layers, retinal hypoxia, and – through the resulting oxygen deprivation – to the release of vascular endothelial growth factor (VEGF). This, in turn, is responsible for secondary neovascularization [6].

Histopathological studies appear to support this mechanism. The results of one of the most recent studies [7] confirmed the presence of macular depigmentation and the loss of Müller cells in histopathological specimens, with a characteristic involvement of the parafoveal region.

In basic ophthalmic examination, several disease manifestations can be observed, sometimes overlapping. However, additional diagnostic tests are required to establish a definitive diagnosis. Accor-

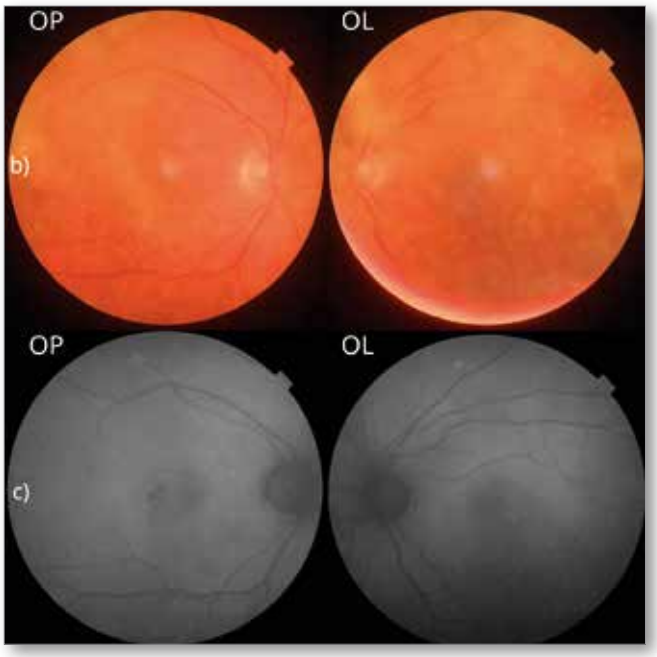

Fig. 1. Stable MacTel 2 with low dynamics. A. OCT B-scan – Grade 1 disease with a low risk of dynamic ellipsoid zone (EZ) loss in the following years. Visible covering of the defect in the inner retinal layers by the ILM. Symmetrical changes in both eyes. B. Fundus photograph – a small area of temporal macular depigmentation in both eyes. C. Discrete hyperautofluorescence with a central area of hypofluorescence in the OD.

ding to the MacTel Study [8], clinical signs visible during fundus examination include a "dull macula", telangiectasias, dilated and blunted retinal venules, crystalline deposits, and pigment clumps.

In optical coherence tomography (OCT), patients with MacTel 2 frequently exhibit characteristic abnormalities such as hyporeflective spaces within the inner and outer layers of the neurosensory retina, disruption of the inner and outer photoreceptor segments, increased reflectivity of the inner retinal layers, hyperreflective subretinal and intraretinal changes, and neurosensory retinal atrophy in the late stages of the disease. A characteristic sign accompanying the "collapse" of cavities in the outer retinal layers is their coverage by the internal limiting membrane (ILM) (Fig. 1). Disruption of the ILM may lead to the formation of a lamellar retinal hole [5–9].

The results of a recent study, published in November 2025, correlated morphological changes in the ellipsoid zone (EZ) defect visualized on OCT scans with the rate of EZ atrophy. The absence of an EZ defect, or a sharply demarcated hyporeflective space in the outer retinal layers with preserved layer architecture, was associated with a slower rate of EZ loss over subsequent years [10].

The characteristic changes seen in macular telangiectasia are not limited to OCT imaging. Fundus autofluorescence (FAF) is also useful in diagnosing these abnormalities. According to Report No. 9 of the MacTel Study [11], FAF reveals several characteristic findings, including hyperautofluorescence temporal to the fovea – associated with macular pigment loss – which is observed in the early stages of the disease (Fig. 1). Hyperautofluorescent areas are also associated with foveal cavities, photoreceptor atrophy, and retinal holes – even in the phase prior to the disruption of the ILM.

FAF imaging can also reveal hypoautofluorescent changes, which typically appear at a later stage of the disease and are associated with pigment clumps and the presence of a neovascular membrane (mixed hypo- and hyperautofluorescent changes) [5, 11].

Currently, the most widely used classification for MacTel 2 progression is the Gass & Blodi classification, which divides the disease into five primary stages based on fluorescein angiography (FA) findings [12].

Stage 1. No obvious abnormality. Early fluorescein shows minimal leakage and mild staining temporally.

Stage 2. Graying of perifoveal retina, minimal or no telangiectatic vessels: early fluorescein shows outer capillary network temporally.

Stage 3. Blunted and right-angle veins: fluorescein shows unusual capillary dilation and permeability in outer retina.

Stage 4. Pigmentation: often associated within the blunted tips of right-angle veins.

Stage 5. Subretinal neovascular: biomicroscopic and fluorescein evidence of neovascularization.

In 2023, as part of the MacTel Project, a new classification based on multimodal imaging using OCT was proposed [13]. It is presented in Tab. I.

Grade	Description
0	No EZ break; no pigmentation; no OCT HR
1	Non-central EZ break; no pigment; no OCT HR
2	Central EZ break; no pigment; no OCT HR
3	Non-central pigment; no, non-central, or central EZ; no OCT HR
4	OCT HR; EZ break (either central or non-central); no pigment
5	Central pigment; no exudative neovascularization; EZ present or not gradable
6	Neovascularization (exudative) ± central pigment

Non-central means that the central subfield of the retina remains unaffected. EZ – ellipsoid zone; HR – hyper-reflectivity. Source: Chew et al., 2023.

Tab. I. Classification of macular telangiectasia type 2 (MacTel classification)

Case report

A 57-year-old female patient presented with a complaint of deteriorating vision in her current spectacles. She denied any chronic diseases or allergies. Her BMI was within the normal range.

On ophthalmic examination, visual acuity was 0.63 cc with +0.5/-1.0 ax 100 in the right eye (OD) and 0.3 cc with -0.75 ax 95 in the left eye (OS). Intraocular pressure (IOP) was within normal limits.

Examination of the anterior segment revealed no pathologies, except for early corticonuclear opacities.

Fundus examination revealed the following findings. OD: The optic disc was round and flat, with well-defined margins; the retinal

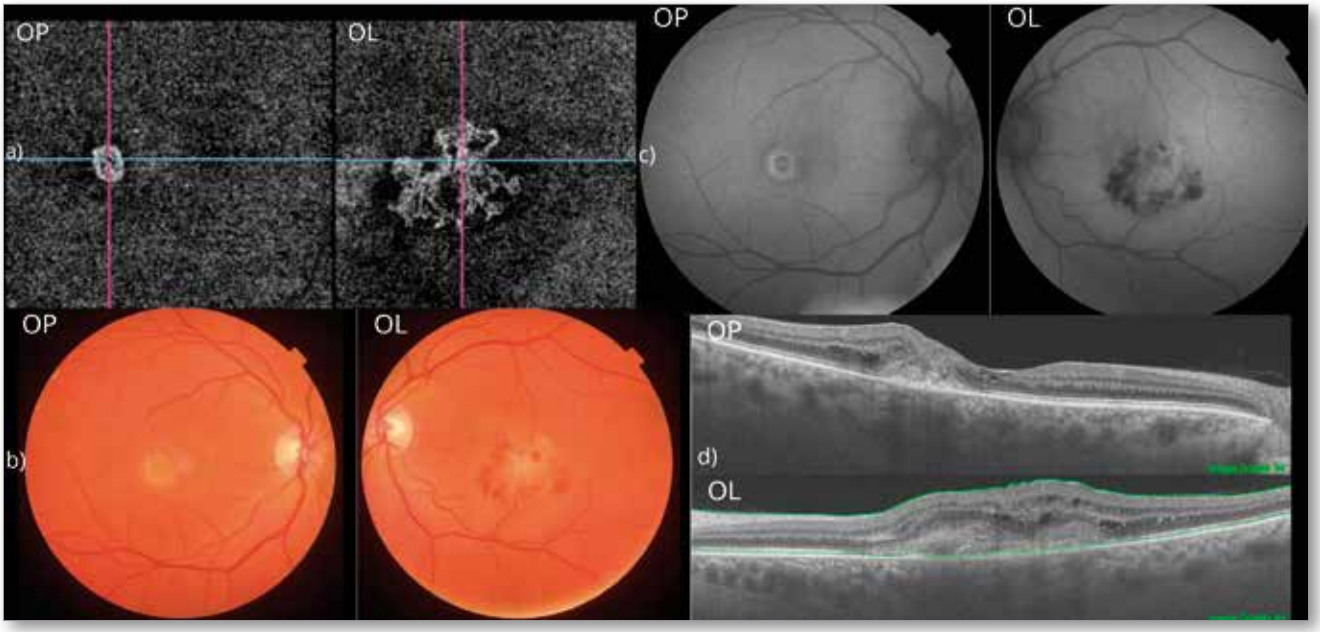

Fig. 2. MacTel 2, Grade 6. Status at the time of admission. A. OCTA – pathological flow visible in the outer retinal layers, corresponding to MNV in both eyes. B. Fundus – photograph showing an area of depigmentation in the OD and intraretinal hemorrhages in the OS. C. FAF – areas of hypo- and hyperautofluorescence. D. OCT B-scan – visible loss of the EZ and features of active neovascularization in the OD and OS.

vessels showed a normal course. An area of depigmentation was present in the macula. OS: The optic disc was round and flat, with well-defined margins; the retinal vessels showed a normal course. In the macula, a pronounced area of depigmentation was observed, with surrounding perifoveal hemorrhages/pathological vessels.

To establish the diagnosis, FAF, OCT, and OCT angiography (Angio-OCT, OCTA) were performed. FAF revealed areas of hyperautofluorescence and hypoautofluorescence, while OCT demonstrated the characteristic appearance of the ILM covering the site of loss of inner retinal layers, as well as elevation of the retinal pigment epithelium (RPE) corresponding to a neovascular membrane with features of activity (subretinal fluid) in the OS. OCTA confirmed the presence of pathological flow typical of macular neovascularization (MNV) in both eyes (Fig. 2).

The patient was scheduled for injection of anti-vascular endothelial growth factor (anti-VEGF) agents in the OS. An intravitreal injection of aflibercept was administered to the OS. Given the very good therapeutic response and the emergence of signs of activity in the OD, the patient was scheduled for anti-VEGF injections in the OD. To date, the patient has received four injections of aflibercept in the OS and two injections in the OD.

During the follow-up period, the patient's visual acuity improved. Five weeks after the initial injection, acuity reached 0.5 in the OS. Six months after initiating treatment and following two intravitreal aflibercept injections, visual acuity was 0.7 in the OS. At the most recent follow-up, the best-corrected visual acuity (BCVA) remained stable at 0.7 in both eyes. Significant improvement in macular morphology was also observed, as shown in Fig. 3.

Fig. 3. MacTel 2, Grade 6. Status post-intravitreal anti-VEGF injections. A. OCT B-scans of both eyes show a reduction in retinal edema and the formation of a subfoveal scar. B. Color fundus photographs show depigmentation of the temporal macula in both eyes and resolution of hemorrhages.

Conclusions

To date, various treatment approaches have been proposed for MacTel 2, including photodynamic therapy, intravitreal injections of steroids and anti-VEGF agents, laser therapy, and pars plana vitrectomy with ILM peeling. In a meta-analysis published in 2026 [14], an improvement in visual acuity was observed in 58% of patients in the non-proliferative phase (i.e., without the presence of MNV) who received anti-VEGF injections. In contrast, among patients who underwent active observation only – without any medical intervention – visual acuity improved in approximately 31% of cases. This finding, together with the meta-analysis results contradicting clinical trial data on anti-VEGF agents, does not allow for a definitive determination of the role of anti-VEGF therapy in the non-proliferative stage of the disease – an issue emphasized by the authors themselves. At the same time, the above-mentioned meta-analysis clearly demonstrated the ineffectiveness of laser photocoagulation. The efficacy of photodynamic therapy (PDT) also remains undetermined. In a study evaluating the efficacy of PDT that included three patients with MacTel 2, no improvement in visual acuity or central retinal thickness was demonstrated [15].

In cases where macular neovascularization (MNV) develops, intravitreal anti-VEGF injections are used [5, 14–16]. In the meta-analysis cited above, visual improvement occurred in 60% of patients with MNV treated with anti-VEGF agents, whereas an observational strategy resulted in visual deterioration in 49% of cases (with improvement in only 7%).

The meta-analysis demonstrated the efficacy of vitrectomy using the inverted flap technique compared to ILM peeling in cases involving the formation of a full-thickness macular hole. Use of the inverted flap technique resulted in a higher macular hole closure rate – 95% vs. 44% in patients who underwent ILM peeling alone – and led to visual acuity improvement in 60% of patients (vs. 45%).

The SAFE clinical trial is currently ongoing, and the SEErine study is in preparation, aiming to evaluate the efficacy of oral L-serine (in combination with fenofibrate in the SAFE trial) in the treatment of MacTel 2.

In 2025, the first gene therapy for MacTel 2 was approved for use in the United States. This therapy is recommended during the non-proliferative phase of the disease. Revakinagene taroretcel (Encelto) is an implant containing allogeneic retinal pigment epithelial cells engineered to secrete recombinant human ciliary neurotrophic factor (rhCNTF). This factor is naturally produced by Müller cells. It is responsible for activating the Janus kinase pathway and the transcription factor involved in the activation of genes that facilitate neuroprotection and enhance photoreceptor survival [17]. The efficacy of the therapy was demonstrated in two phase 3 clinical trials (NCT03319849 and NCT03316300), in which treatment resulted in a slower rate of EZ loss compared with the placebo group. It should be noted, however, that these studies found no differences in visual acuity or vision quality questionnaire scores between the treatment group and the group receiving a non-drug-releasing (sham) implant [18]. This therapy involves delivering the medication via an implant inserted into the eye through the pars plana [19].

In summary, the most recent therapeutic option for the non-proliferative form of macular telangiectasia type 2, with efficacy confirmed in clinical trials, is an implant containing allogeneic retinal pigment epithelial cells – revakinagene taroretcel (Encelto). Unfortunately, this treatment is not available in Europe. Consequently, our patients can only be monitored, and in the event of MNV, intravitreal anti-VEGF therapy should be initiated.

Disclosure
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