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CLINICAL STUDY

Macular full-thickness and lamellar holes in association with type 2 idiopathic macular telangiectasia

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Abstract

Purpose **To describe patients with full-thickness macular holes (FTMHs) and lamellar macular holes (LMHs) in association with type 2 idiopathic macular telangiectasia (type 2 IMT).**
Methods **Six patients with either FTMH or LMH and type 2 IMT were evaluated by means of optical coherence tomography (OCT) imaging, funduscopy, and fluorescein angiography.**
Results **The age of the examined patients ranged from 57 to 70 years (mean 62.5 ± 5.2), and best-corrected visual acuity of the affected eyes ranged from 20/50 to 20/200 (mean 20/100). All eyes showed macular abnormalities typical for nonproliferative type 2 IMT except for one eye with a proliferative disease stage. Three patients had an FTMH, one presenting with bilateral FTMH, and three had an LMH on OCT. In all cases of FTMH, the macular holes did not have elevated margins. Surgery was performed in two patients with a FTHM without subsequent functional improvement.**
Conclusions **The altered foveal anatomy with progressive atrophic changes within the neurosensory retina in type 2 IMT may predispose to the development of FTMH and LMH. Type 2 IMT should be considered in the differential diagnosis in patients presenting with macular holes. The association between the two may reflect alternative pathogenetic mechanisms in the development of macular holes.**
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Keywords: idiopathic macular telangiectasia; idiopathic parafoveolar telangiectasis; full-thickness macular hole; lamellar macular hole; optical coherence tomography

Introduction

Type 2 idiopathic macular telangiectasia (type 2 IMT) is a rare condition which typically presents with a slow decrease in visual acuity and/or metamorphopsia in the 5th–7th decade.[1–4] Angiographically, the parafoveolar capillaries mainly temporal to the fovea appear to be dilated, and a diffuse parafoveal hyperfluorescence is seen in the late phase of fluorescein angiography. With disease progression, parafoveal scotomas may develop,[5] and advanced stages are characterized by intraretinal pigment clumping and neovascular membranes.[2–4]

Common findings in optical coherence tomography (OCT) include a disruption within the foveal photoreceptor layer and intraretinal hyporeflective spaces.[6–9] However, full-thickness macular holes (FTMHs)[1,10,11] and lamellar[12] macular holes (LMHs) complicating type 2 IMT have been reported in only few patients.

We report six patients with type 2 IMT who developed a full-thickness or an LMH, respectively.

Methods

All patients underwent a complete ophthalmic examination, including best-corrected visual acuity determination, slit-lamp examination, indirect ophthalmoscopy, fluorescein angiography by digital photography or by confocal scanning laser ophthalmoscopy (cSLO,

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HRA2, Heidelberg Engineering, Heidelberg, Germany), fundus photography, and time-domain OCT imaging (Stratus OCT, Carl Zeiss Meditec, Jena, Germany). Diagnosis was based on typical clinical findings, fluorescein angiography, and OCT imaging.

Results

The typical features for type 2 IMT on fluorescein angiography and on OCT imaging were present in all patients including parafoveal fluorescein leakage from telangiectatic vessels and foveolar hyporeflective spaces. Additional funduscopic findings were loss of retinal transparency, right-angled venules, superficial retinal crystalline deposits, and pigment clumping. Only one patient (case 6) presented with a proliferative disease stage in one eye, exhibiting a vascular membrane originating from retinal vessels. Table 1 summarizes the clinical findings in all patients.

Full-thickness macular holes

Patient 1

A 61-year-old woman was first examined elsewhere for impaired vision in OS. Funduscopy showed bilateral greying of the retina in the temporal part of the macula with a small dilated, blunted venule in OD. Telangiectatic capillaries, mainly located in the temporal part of the fovea, were visible angiographically (Figure 1a and b). The left eye showed a large FTMHs and a fine epiretinal membrane.

The patient was seen 6 years later for a gradual loss of vision in her right eye. Fundus examination and red-free photographs (Figure 1c and d) now also showed a FTMH with an epiretinal membrane in OD, and some pigment clumping had occurred temporal to the fovea. OCT confirmed bilateral FTMHs (Figure 1e and f). Subsequently, routine pars plana vitrectomy with peeling of the internal limiting membrane, air/gas-tamponade, and post-operative posturing were performed in OD, which did not result in closure of the FTMH or improvement in visual acuity (20/400). Complete posterior vitreous detachment was present before surgery.

Patient 2

A 57-year-old female patient was referred for surgical treatment of an FTMH. She had no history of ocular trauma or surgery. OCT showed an FTMH in OS and a small disruption in the foveolar photoreceptor layer in the asymptomatic OD (Figure 2, top row). Fluorescein angiography confirmed the suspected diagnosis of type 2

Table 1 Summary of patients with type 2 IMT and associated full thickness macular holes (FTHM; 1–3) or lamellar macular holes (LMH; 4–6), respectively

Patient	*Age*	*Gender*	*VA OD/OS*	*Index eye*	*Fellow eye*	*Procedures performed and outcome in index eye*
Full thickness macular holes						
1	61	F	20/100 20/200	Bilateral full thickness macular hole, loss of parafoveal transparency, dilated blunted venules, intraretinal pigment proliferation		OD: vitreoretinal surgery with air/gas-tamponade, no anatomic or functional improvement
2	57	F	20/32 20/100	Intraretinal gliotic changes temporal to the fovea	Foveal fine hypopigmentations	Vitreoretinal surgery with silicon oil. Increase in VA to 20/63, but no complete closure of the MH
3	70	F	20/160 20/60	Loss of parafoveal transparency	Loss of parafoveal transparency, dilated blunted venule, intraretinal pigment proliferation	Intravitreal triamcinolone No functional improvement
Lamellar macular holes						
4	64	M	20/32 20/63	Epiretinal membrane with LMH, intraretinal pigment proliferation, dilated blunted venules, superficial crystalline deposits	Intraretinal pigment proliferation, dilated blunted venules, superficial crystalline deposits	Acetazolamide No improvement
5	67	F	20/200 20/25	Intraretinal pigment proliferation, dilated blunted venules, superficial crystalline deposits	Dilated blunted venule and loss of retinal transparency temporal to fovea	No treatment
6	57	M	20/50	Intraretinal pigment proliferation and neovascularization, dilated blunted venules	No obvious pathology	OD: vitrectomy for synchisis scintillans

OD, right eye; OS, left eye; VA, visual acuity.

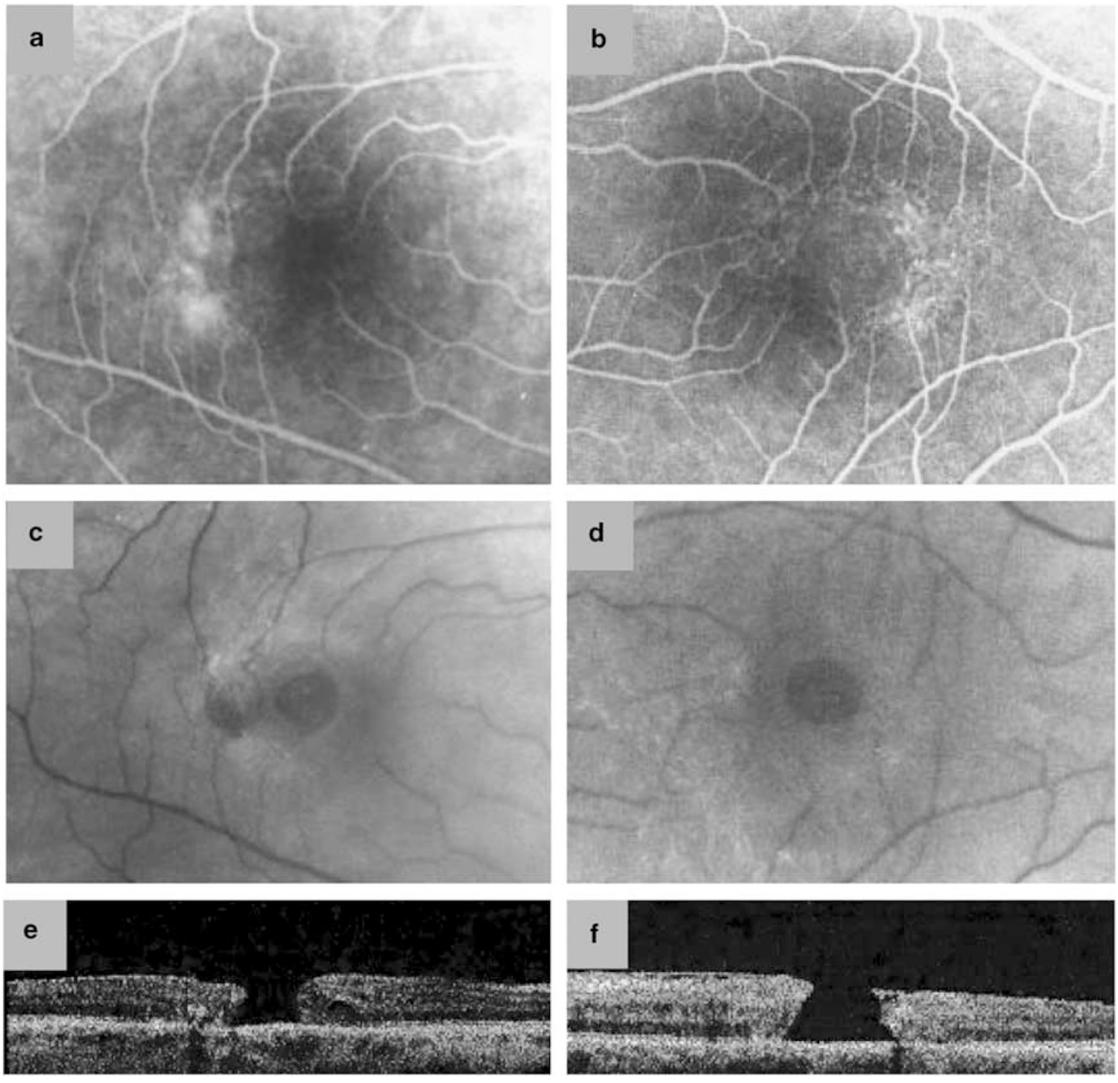

Figure 1 Bilateral full-thickness macular holes (FTMHs) in presence of type 2 IMT. (a and b) Fluorescein angiography of OD (a) and OS (b) showing telangiectatic vessels temporal to the fovea. (c–f) FTMH shown in red-free photographs (c and d) and OCT imaging (e and f). OCT illustrates an epiretinal membrane mainly in OS.

IMT. Intraoperatively, the posterior vitreous appeared to be entirely adherent to the inner retinal interface. The patient opted for silicone oil tamponade as she was unable to posture. Consecutive OCT imaging during the following year revealed increasing foveal intraretinal hyporeflective spaces in OD. The FTHM in OS became smaller but did not close entirely. After a combined silicone oil removal with cataract operation, the adjacent retina showed an atrophic foveal aspect, and only little functional gain from initially 20/100 to 20/63 was noted.

Patient 3

A 70-year-old female patient was diagnosed with type 2 IMT, 7years prior to initial presentation. By that time visual acuity was 20/30 in both eyes. She complained about a gradual decrease in visual acuity in her right eye. Intravitreal triamcinolone injection was performed elsewhere 2 years before presentation in OD without any functional improvement. OCT now showed an FTMH in OS (Figure 3). The defect was more pronounced within the deep foveal layers, and small intraretinal cystic changes were present in proximity to the FTMH. In OD, an atrophic foveal aspect with a foveal hyporeflective space within the photoreceptor layer and an adjacent intraretinal hyper-reflective structure was found in OCT imaging. The patient opted against vitreoretinal surgery.

Lamellar macular holes

Patient 4

A 64-year-old male patient presented with increasing bilateral blurred vision and discrete metamorphopsia for

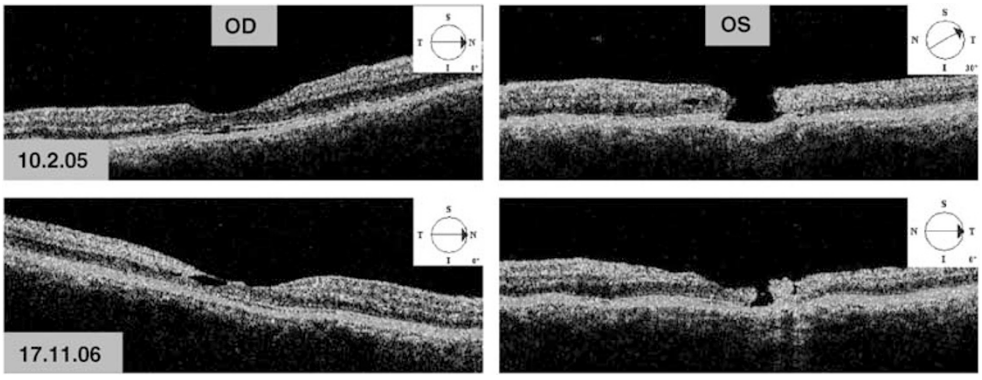

Figure 2 Full-thickness macular hole (FTMH) in OS of patient 2. Bilateral OCT horizontal scans before (upper row) and after (21 months later; lower row) vitreoretinal surgery in OS. OD initially shows a small disruption in the photoreceptor layer and a larger hyporeflective space in the inner neurosensory retina 21 months later. The FTMH (top right) with cystoid structures in proximity to the hole did not close after vitreoretinal surgery with temporary silicon oil tamponade. At follow-up, the FTHM was smaller and the adjacent retina showed an atrophic aspect.

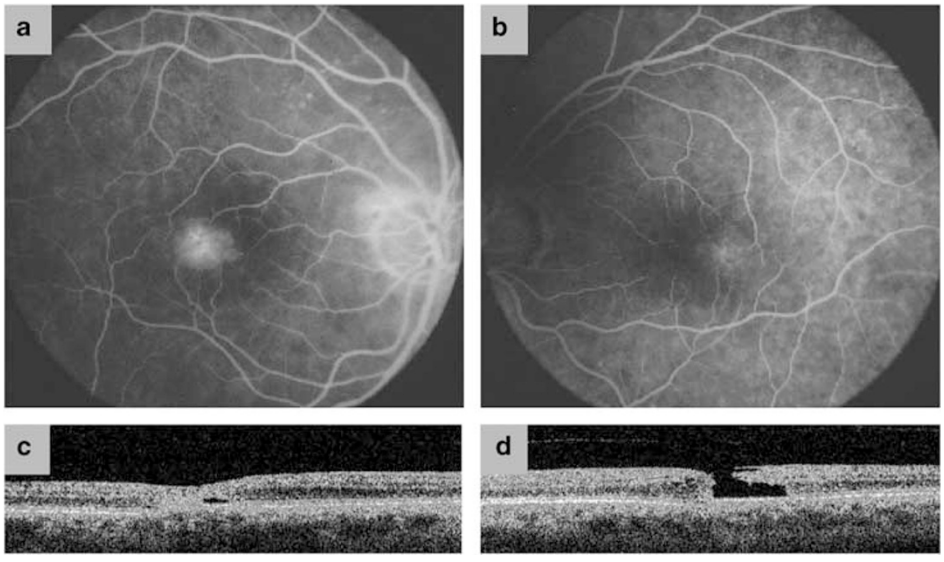

Figure 3 (a and b) Late-phase fluorescein angiography shows diffuse hyperfluorescence temporal to the fovea. (c and d) OCT imaging of OS (d) showing a full-thickness macular hole and an epiretinal membrane or drape-like structure. (c) In OD, atrophic, hypo- and hyper-reflective foveal changes are revealed.

5 years. Type 2 IMT was diagnosed based on the typical angiographic appearance (Figure 4a, b). OCT showed a foveolar thinning and intraretinal hyporeflective spaces in OD and confirmed the diagnosis of an epiretinal membrane or disrupted drape like structure with an LMH in OS (Figure 4c, d). Visual acuity and OCT findings remained unchanged within a follow-up time of 19 months.

Patient 5

A 66-year-old woman complained about blurred vision in her right eye for about 2 years. Type 2 IMT, more pronounced in OD, was diagnosed angiographically. OCT showed an LMH in OD with the posterior vitreous attached to the upper half of the rim of the LMH. In OS, there were superficial foveal hyporeflective spaces covered by a drape-like structure that had a little central defect. No obvious vitreoretinal adhesion could be detected in the left eye. Since visual acuity had been stable for the last 2 years, no treatment was recommended (Figure 5).

Patient 6

A patient with known type 2 IMT underwent combined cataract surgery and vitrectomy for severe synchisis scintillans in OD. At 3-month review, OCT revealed an

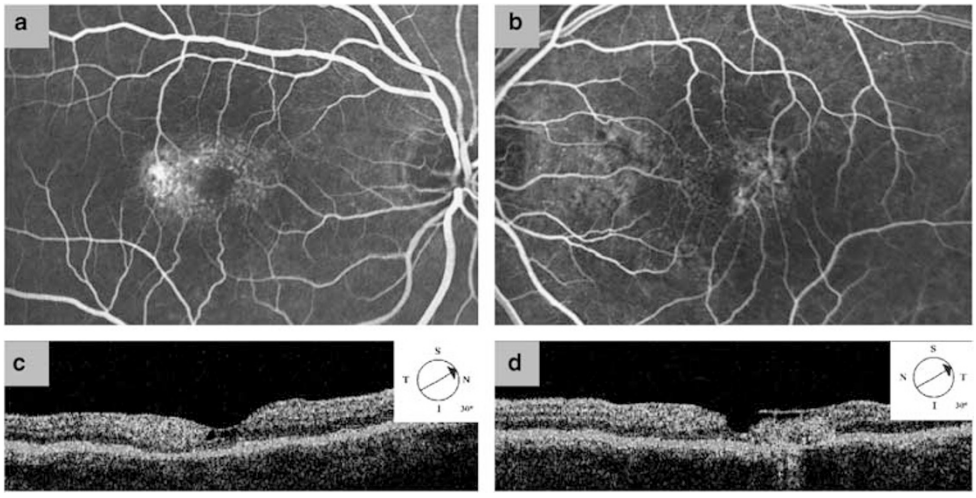

Figure 4 (a and b) Fluorescein angiography shows temporally accentuated juxtafoveal telangiectasia in both eyes. (c and d) OCT imaging of OD shows foveal thinning and hypo-reflective spaces covered by a thin drape. In OS, a lamellar macular hole and an epiretinal membrane or disrupted drape-like structure are visible. There is intraretinal hyper-reflectivity in an area of pigment clumping.

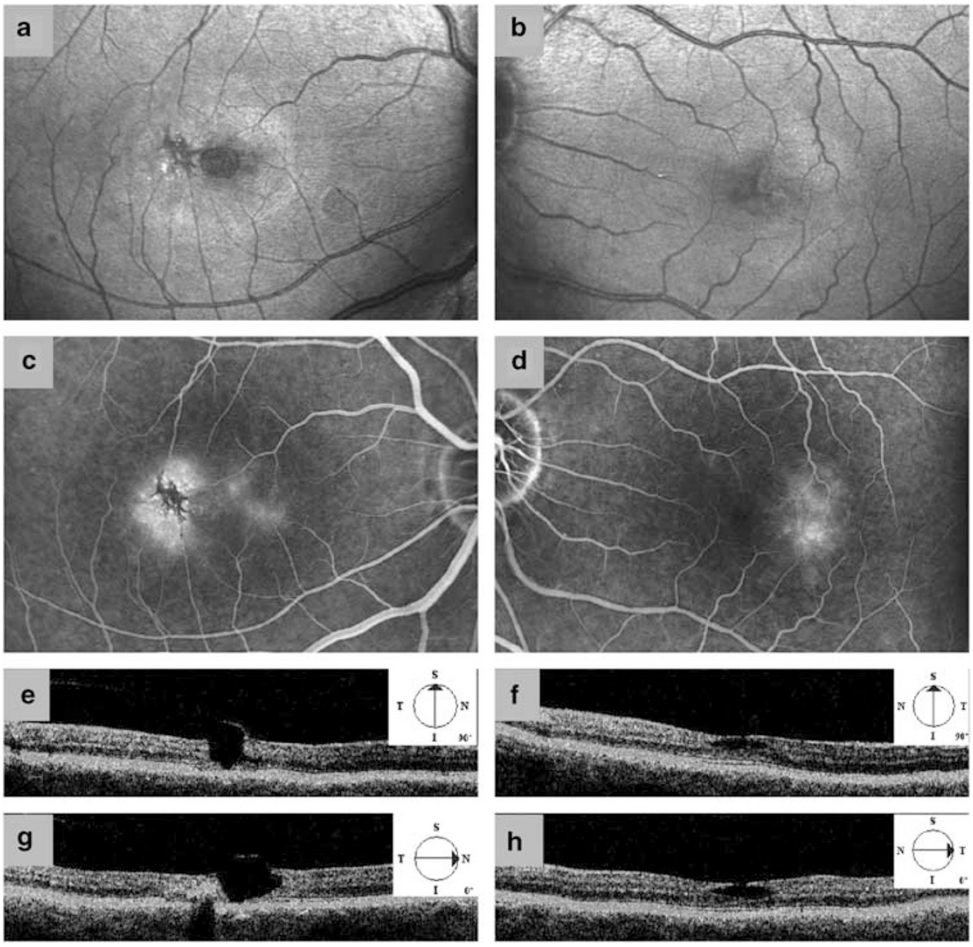

Figure 5 (a and b) Confocal blue reflectance imaging showing a macular hole and pigment clumping temporal to the fovea in OD (a). (c and d) Late-phase fluorescein angiography showing diffuse parafoveal hyperfluorescence predominantly temporally to the fovea. (e–h) OCT imaging showing a vitreoretinal adhesion and an LMH in OD (e and g). OS with a superficial hyporeflective space and a central defect in the covering drape-like structure (f and h).

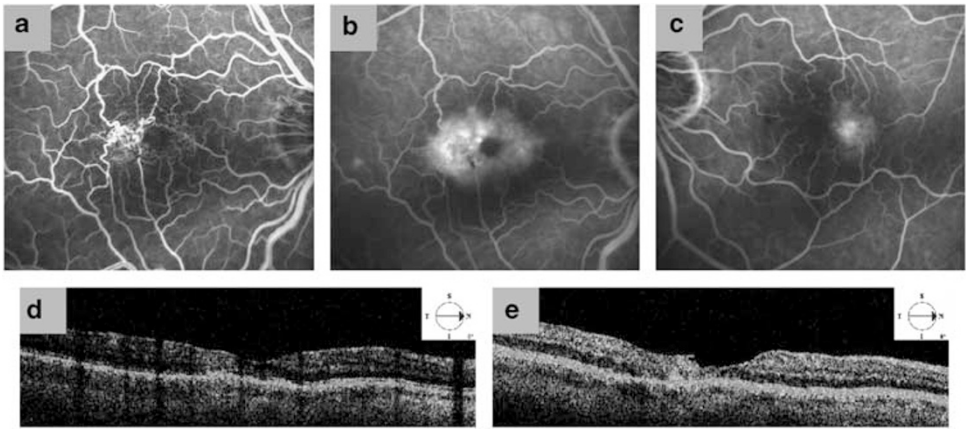

Figure 6 Early- (a) and late- (b) phase angiography of OD and late-phase angiography of OS (c). (a) Shows a retinal neovascularization temporal to the fovea. (b) and (c) show late diffuse hyperfluorescence mainly temporal to the fovea. (d and e) OCT imaging of the right eye before (d) and after (e) vitrectomy with a lamellar macular hole in (e). (d) OCT with artefacts due to marked synchisis scintillans.

LMH in OD which was not present preoperatively. A neovascularization with retino-retinal anastomosis temporal to the fovea in the same eye did not change within the observation period (Figure 6).

Discussion

Current concepts for the pathogenesis of idiopathic FTMHs and LMHs include vitreoretinal antero-posterior traction, tangential vitreous traction on the macula, tangential traction caused by an epiretinal membrane or un-roofing of foveal cysts in macular oedema.[13–15] The distinguished foveal anatomy with attenuation of the internal limiting membrane, the oblique orientation of the Henle nerve fibre layer and Müller cells may render the macula vulnerable to dehiscence by mechanical forces.

Macular holes occurring as a result of vitreoretinal traction may show vitreoretinal attachments, elevated and thickened margins and/or a detached vitreous with an operculum at the vitreous base, dependent on the disease stage.[14] Interestingly, the FTHM of the patients presented in this study did not show these signs on OCT. The LMH in patient no. 5 presented with an apparent vitreoretinal traction at the superior margin of the LMH. The fellow eye with vitreous detachment showed foveal hyporeflective spaces and a covering drape-like structure with a central hole. It could be speculated that the latter was torn open when the vitreous detached.

OCT findings in type 2 IMT include intraretinal cyst-like spaces and photoreceptor layer disruption without retinal thickening.[6,7] Possible aetiologies include chronic leakage from hyperpermeable capillaries, ischaemia, progressive degeneration of the neuronal and glial cellular elements or any combination of the above. It was hypothesized by Olson and Mandava[10] that the atrophic foveal changes seen in type 2 IMT could result in a weakening of the foveal structure and therefore be a predisposing factor for macular hole formation, similar to the occurrence of macular holes in cystic macular oedema. However, the reason why macular holes rarely develop despite the frequent observation of these foveolar atrophic changes in type 2 IMT is unclear. Paunescu *et al*[6] described an 'internal limiting membrane drape' (such as in Figure 4c) in 42% of eyes with type 2 IMT which could contribute to some stabilization of the foveal contour. They also showed an OCT image of a 'full-thickness macular hole', which was still covered by the drape-like structure. To our knowledge, the histopathology of such 'roofs' in type 2 IMT has not been studied to determine if the 'internal limiting membrane drape' actually represents the internal limiting membrane.

In addition to the atrophic neurosensory changes in type 2 IMT, there could be mechanical stress on the fovea, resulting in disruption of the inner drape-like structure. FTMH or LMH would be a possible consequence. Predisposing factors could be additional surgical trauma (as in patient 6) or vitreoretinal traction, which could have been the cause in patients 3–5. However, there was no posterior vitreous detachment in patient 2 as well in the case described by Olson and Mandava.[10] A sequence from formation of an LMH before occurrence of an FTMH also appears to be possible. Since patient no. 1 developed bilateral FTMH, other individual, so far unidentified anatomic or genetic factors could play a role.

To further determine the pathogenesis of macular holes in type 2 IMT, sequential imaging over time in more patients with type 2 IMT appears prudent. The development as well as the spontaneous course of LMHs

should be investigated in future natural history studies. Such longitudinal data are expected from the MacTel-project (www.mactelresearch.org) with over 280 patients recruited so far.

It is possible that the coincidence of macular telangiectasia with macular holes is underestimated due to the possibility of only subtle ophthalmoscopic changes in early stages of macular telangiectasia. This subgroup of macular holes could at least partially be responsible for macular holes that developed without prior macular oedema, vitreomacular, or tangential traction. Usually, fluorescein angiography is not performed as part of the diagnostic evaluation for macular holes, and therefore, type 2 IMT as a causative factor may be missed. Recently, confocal blue-reflectance imaging has been proposed as a non-invasive technique to diagnose type 2 IMT.[16] Since this technique also documents the size of a macular hole, it is suggested to be included as diagnostic tool, especially if the aetiology of the macular hole is questionable (ie no posterior vitreous detachment or epiretinal gliosis) or if there are signs for type 2 IMT in the opposite eye (as perifoveal greying of the retina or atrophic cystic changes in OCT imaging).

In conclusion, based on this observational case series including cross-sectional and longitudinal observations, we suggest that macular hole formation represents a potential complication during the natural course of type 2 IMT. A different pathophysiology compared to 'idiopathic' macular holes is conceivable, since type 2 IMT is typically associated with disruptions in the neurosensory retina. It appears prudent to consider type 2 IMT in patients who initially presented with an LMH or FTMH as co-morbidity. The response to treatment and the functional outcome may differ from other macular hole entities.

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Conflict of interest

None of the authors have any financial interest to disclose.

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