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Unusual presentation of tractional retinal detachment in beta thalassemia minor

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ABSTRACT

Purpose: To report a case of proliferative retinopathy with tractional retinal detachment associated with beta thalassemia minor in a 27-year-old female.

Observations: A young lady having beta thalassemia minor presented with decreased vision in both eyes, the effect being more severe in her right eye. The patient's other systemic history, including ophthalmic history, was unremarkable. The fundus examination revealed peripheral retinal ischemia in both eyes and tractional retinal detachment in the right eye.

Conclusion: Beta thalassemia minor is not associated with striking retinal pathology, nevertheless proliferative retinopathy. However, in this case the patient developed tractional retinal detachment that required surgery. This indicates that proliferative changes may develop in patients with beta thalassemia, and routine fundus examinations could be recommended for these patients.

1. Introduction

Hemoglobinopathies are a group of inherited blood disorders that are primarily caused by a single gene mutation, and they can affect hemoglobin either qualitatively or quantitatively.[1] The worldwide prevalence of hemoglobinopathies, particularly in developing nations, is a significant concern. According to a study, approximately 22,989 new cases of beta thalassemia major are thought to occur each year.[2] In Saudi Arabia, the National Premarital Screening Program conducted a cross-sectional study involving 488,315 individuals and found that the prevalence of the thalassemia trait was 3.22 %, while the thalassemia disease was 0.7 %.[3] Additionally, data from the same program showed that 1.8 % of the 2.7 million participants were thalassemia carriers.[4]

Proliferative sickle retinopathy (PSR), along with other sickle cell phenotypes, has been widely documented in the literature. Previous studies have shown that sickle cell disease, sickle cell anemia, and sickle cell thalassemia patients have an elevated risk of developing PSR.[5–8]

Beta thalassemia-related retinal problems have been linked to desferrioxamine and deferiprone, iron chelating medications, and the disease itself.[9]

Beta-thalassemia minor patients may develop pseudoxanthoma elasticum (PXE) which causes abnormalities in the retina that appear like peau d'orange, angioid streaks, alterations that resemble pattern dystrophy, and drusen in the optic disc.[9] Furthermore, these patients may have non-pseudoxanthoma elasticum-like retinal anomalies, such as retinal venous tortuosity. Only one study reported two beta-thalassemia minor patients who developed proliferative retinopathy.[10] In this report, we present a young lady with beta thalassemia minor who presented with proliferative retinopathy in both eyes, as well as tractional retinal detachment (TRD) in her right eye.

2. Case report

A 27-year-old woman presented at the emergency department complaining of decreased vision in both eyes that began four months earlier. She stated that her vision was progressively worsening, with her right eye more severely affected than the left. However, she did not complain about headache; nausea; vomiting, diplopia; tinnitus; or other vitreoretinal symptoms such as floaters, flashes, or curtain-like loss of vision.

The patient had a significant past medical history of beta thalassemia minor, which was shared by her mother, while two of her siblings were carriers. There was no significant history of ophthalmic conditions. The Hemoglobin electrophoresis result showed the following data for the

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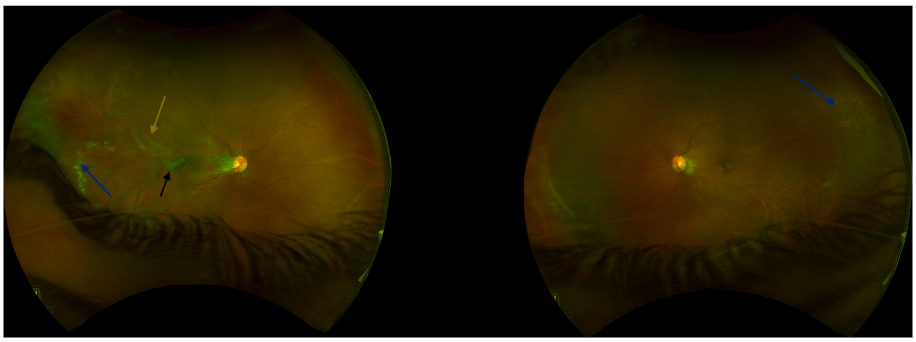

Fig. 1. wide-field Pseudo-color fundus photos of both eyes at the first presentation (Right eye: temporal macular dragging with fibrous membrane (yellow arrow), oval shape hyperpigmented lesion temporal to the fovea and pointed to it corresponding to torpedo maculopathy (black arrow), peripheral telangiectatic and sclerosed vessels (blue arrow). Left eye: peripheral ischemic retina with telangiectatic and sclerosed vessels (blue arrow). (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)

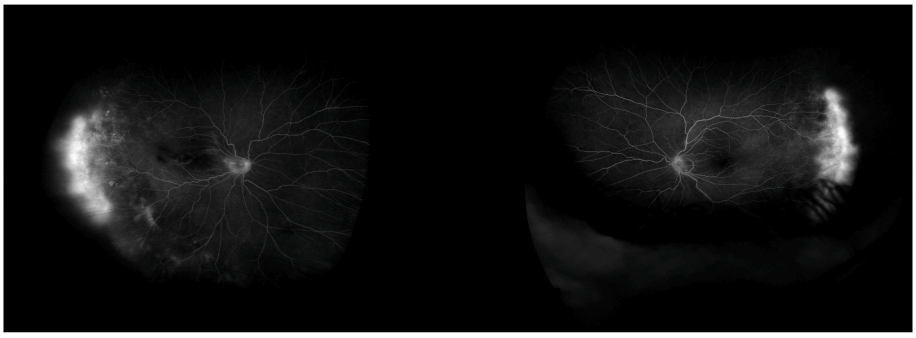

Fig. 2. Fundus Fluorescein Angiography (FFA) photos of the right and left eye at the first presentation. (Both eyes: temporal area of capillary non-perfusion with active leakage of the telangiectatic vessels at the junction between perfused and non-perfused retina. Right eye optic disc: oval-shaped disc with staining in the late frames, straightening of the blood vessel emanating from the disc. Left eye optic disc: staining in the late frames with area of peripapillary atrophy).

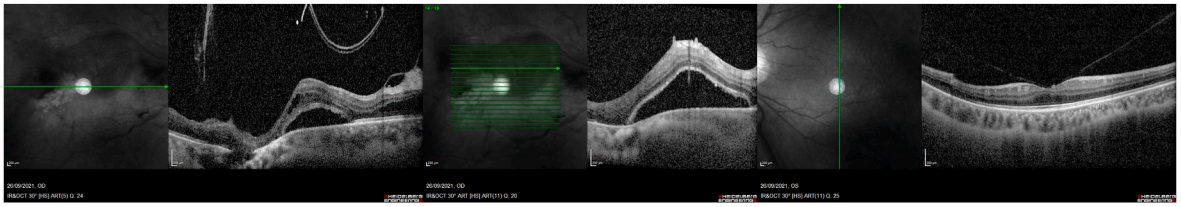

Fig. 3. Optical coherence tomography (OCT) image of the right and left eye at the first presentation. Right eye: tractional retinal detachment partially involving the macula with subretinal fluids. As well as loss of the outer retinal layers including the ellipsoid zone with outer retina cavitation temporal to fovea which corresponds to torpedo maculopathy. Left eye: vitreomacular adhesion (VMA).

patient: HbA1: 94 %, HbA2 5.3 %, and Hb F 0.7 %.

On examination, the patient's visual acuity was 20/50 in the right eye and 20/25 in the left eye. In the dilated fundus exam, the right eye showed temporal macular dragging with a fibrous membrane, an oval-shaped hyperpigmented lesion temporal to the fovea that pointed to it, peripheral telangiectatic and sclerosed vessels with inferior

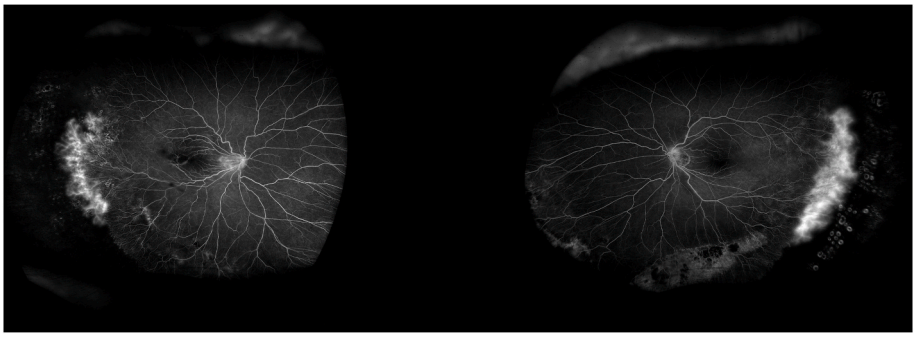

Fig. 4. FFA of both eyes after sectoral laser photocoagulation (Both eyes: Lasers scars to the avascular area temporally).

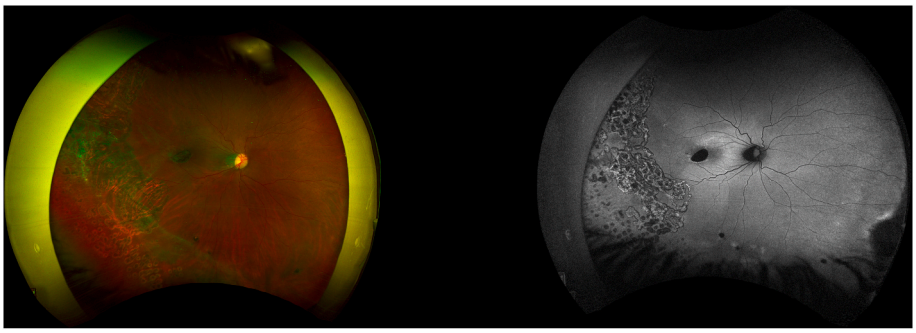

Fig. 5. color fundus photo and fundus autofluorescence of the right eye in the first postoperative visit (Right eye: Color fundus photo shows resolution of the fibrous tissue postoperatively with scatter laser in periphery. Autofluorescence: shows hypoautofluorescence temporal to the fovea corresponds to the torpedo maculopathy). (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)

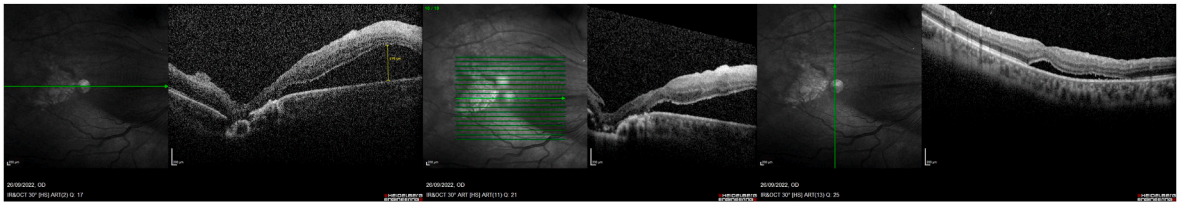

Fig. 6. OCT images of the right eye in the first post-operative visit (Vertical and horizontal cuts showed less subretinal fluid compared to preoperative OCT).

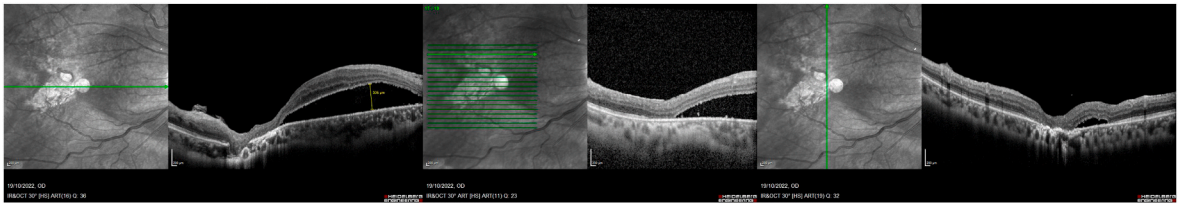

Fig. 7. OCT images of the right eye in the second post-operative visit (Vertical and horizontal cuts showed less subretinal fluid compared to 1st Post-operative OCT).

retinoschisis. The left eye was normal apart from a peripheral ischemic retina with telangiectatic and sclerosed vessels (see Fig. 1).

Fundus fluorescein angiography (FFA) of both eyes revealed a temporal area of capillary non-perfusion with active leakage of the telangiectatic vessels at the junction between the perfused and non-perfused retina, staining of the optic discs in the late frames of both eyes with area of peripapillary atrophy was observed in the left eye (see Fig. 2).

An optical coherence tomography (OCT) of the right eye revealed tractional retinal detachment partially involving the macula with subretinal fluids, as well as loss of the outer retinal layers, including the ellipsoid zone with outer retina cavitation temporal to the fovea, corresponding to torpedo maculopathy. The optical coherence tomography of the left eye revealed vitreomacular adhesion (see Fig. 3).

The patient was initially managed by FFA-guided sectoral laser photocoagulation of the ischemic retina in both eyes (see Fig. 4). After the neovascularization regressed, the patient underwent surgery in the form of a chandelier-assisted pars plana vitrectomy, with membrane peeling, endolaser, and air-fluid exchange for the right eye (see the attached surgical video in a 3D forum).

In the postoperative period, the patient had a visual acuity of 20/40 on the Snellen chart, with residual subretinal fluid noted on OCT (See Figs. 5 and 6). However, the fluid decreased during subsequent visits, and six months post-surgery, the patient's visual acuity was 20/25 (see Fig. 7).

3. Discussion

While hemoglobinopathies are associated with proliferative retinopathy, such occurrences are relatively uncommon in cases of thalassemia minor. Numerous anomalies of the posterior segment have been linked to beta thalassemia major and intermedia.[9] Barteselli et al. described PXE-like features in patients with beta thalassemia major and intermedia. About 27 % of the study population (n = 255) showed PXE-like features. These features were more common in patients who had thalassemia intermedia, who were older, and who had history of splenectomy. The study concluded that patients with risk factors for developing posterior segment pathology may warrant regular follow-up for fundus examinations.

Various studies have noted ocular changes or anomalies linked with beta-thalassemia minor; however, these conditions are not identical to those linked with beta-thalassemia major or intermedia. Magli et al. described color vision and visual field alterations in patients with thalassemia minor.[11] Rudd et al. described retinal detachment in a patient with thalassemia minor.[12] Several studies have investigated the posterior segment pathologies associated with thalassemia minor through multi-modal imaging of the retina and choroid. Cennamo et al. described optical coherence tomography angiography (OCT-A) changes in the different phenotypes of beta-thalassemia.[13] The vascular density did not differ between the beta thalassemia minor group and the control group but was statistically significantly reduced between the control groups and the transfusion-dependent beta thalassemia group as well as the non-transfusion-dependent beta thalassemia group. The study also

examined retinal nerve fiber layer (RNFL) thickness and the foveal avascular zone (FAZ), RNFL thickness is significantly decreased in beta -thalassemia major compared to control group, while no difference exists between beta-thalassemia minor and control group. The same behavior observed with FAZ, where FAZ is significantly decreased in transfusion-dependent beta-thalassemia but no difference observed between beta-thalassemia minor and control group.

Another study employed spectral-domain optical coherence tomography (SD- OCT) to study structural differences between patients with beta-thalassemia minor and healthy individuals. It found statistically significant differences in choroidal thickness and paracentral macular thickness between healthy individuals and patients with thalassemia minor. Nasal, temporal, and subfoveal choroidal thickness values were thinner in patients with thalassemia minor. In patients with beta-thalassemia minor, the macular thickness of their superior, inferior, temporal, and nasal quadrants was observed to be thinner than those in healthy controls, although the central macular thickness was similar in both groups.[14] The changes in the optic nerve topography and beta thalassemia minor were studied, and differences found only in the mean disc area, which was less in the thalassemia group. No differences observed in other parameters.[15]

The studies mentioned above make it clear that beta thalassemia minor is not associated with striking retinal pathology, even on multimodal imaging, and that no differences exist in most parameters between patients with beta thalassemia minor and healthy individuals. In 2018, Stultz and his group described two patients with beta-thalassemia minor who presented with vitreous hemorrhage and proliferative retinopathy in two case reports. In this case report, the patient was proven to have beta thalassemia minor and presented with bilateral proliferative retinopathy, TRD in the right eye, and torpedo maculopathy. The patient had no other underlying etiology for proliferative retinopathy. Systemically, the patient was under the care of an internist, and she was free of any systemic disease except beta-thalassemia minor, and this includes diabetes mellitus. She did not have any features of uveitic entity, as no vasculitis, vascular sheathing or cuffing demonstrated on clinical examination or FFA. She was a full-term infant with no postnatal complications and no diagnosis of retinopathy of prematurity or other ophthalmic diseases. One of the main differential diagnoses in this case was familial exudative vitreoretinopathy (FEVR), but the patient has no family history of FEVR. The patient's sibling was brought to us, and she underwent FFA of both eyes. The FFA was completely normal with normal complete vascularization, no vascular leakage in the periphery, and no areas of capillary dropout.

4. Conclusion

Proliferative retinopathy is a rare sequela to thalassemia minor. The incidence of proliferative retinopathy associated with beta-thalassemia minor warrants further investigations, as does the requirement for patients with beta thalassemia to undergo routine check-ups to monitor the development of proliferative retinopathy.

Patient's consent

Written consent was obtained from the patient to publish this case report.

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.ajoc.2023.101946.

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