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How bad can vision get in beta-thalassemia? A case report and review of the literature

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Abstract

Background Review various causes of optic disc-related decreased vision and present a rare anterior ischemic optic neuropathy (AION) case in a patient with beta-thalassemia.

Case presentation A 38-year-old man with a known case of major beta-thalassemia presented with a chief complaint of acute decrease in central and peripheral vision in the right eye. The patient was on deferoxamine and had previous splenectomy surgery. Fundus examination revealed visible vascular obstruction, more prominent superior disc swelling, hyperemia, and retinal nerve fiber layer (RNFL) edema in the right eye. After 6 months, the visual acuity improved in the follow-up examination, and the fundus examination revealed subtle right disc pallor.

Conclusion AION with visible vascular occlusion can be a rare retinal cause of acute decreased vision in beta-thalassemia patients.

Keywords Optic ischemic neuropathy, Anterior ischemic optic neuropathy, Beta thalassemia, Beta-thalassemia major

Background

Beta thalassemia is a genetic condition that disturbs the hemoglobin synthesis. Ocular involvement is relatively common in beta-thalassemia and may cause serious complications [1]. Ocular manifestations in beta-thalassemia may relate to the anemia caused by disease, overload of iron, or the chelating agents used.

Nonarteritic anterior ischemic optic neuropathy (NAION) is one of the causes of disc swelling in adults, especially in old age [2]. Optic neuropathy is a rare complication of beta-thalassemia. Anemia can cause a statistically significant reduction in the optical disc area due to changes in the optic nerve head's blood flow and oxygenation compared to the average population [3]. In response to this decrease in blood flow and oxygen, the ganglion cells are damaged, which declines the best corrected visual acuity (BCVA) defect in the visual field and causes the relative afferent pupillary defect (RAPD).

To date, no reported cases of AION have been associated with nerve damage in the major beta-thalassemia cases. This paper highlights the rare occurrence of AION in a beta-thalassemia patient and underscores the importance of regular fundus examination in these patients.

Case presentation

A 38-year-old man with a known case of major beta-thalassemia under the follow-up of a hematologist was referred to the ophthalmology clinic with a chief complaint of acute decrease in the right eye central vision 3 days ago. The patient was a non-smoker and had no other risk factors, such as hypertension, hypercholesteremia, or diabetes mellitus. The patient had no history

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of headaches, intermittent jaw claudication, or recent weight loss. Drug history includes receiving blood transfusions every 4 weeks, taking an iron-chelating agent (deferoxamine), and undergoing splenectomy at the age of 8 years. The vital signs and his physical examination were without any specific finding.

In the ophthalmology examination, BCVA was 20/200 on the right (O.D.) and 20/20 on the left (O.S.). Intraocular pressure with Goldmann applanation tonometry (GAT) was within standard limit in both eyes. There was 3 + RAPD in the right eye. The slit lamp examination was unremarkable. Fundus examination revealed transparent media with mild venous tortuosity in both eyes, visible vascular obstruction, and more prominent superior disc swelling, hyperemia, and retinal nerve fiber layer (RNFL) edema in the right eye. (Fig. 1, A, B) Optical coherence tomography also showed disc edema and decreased ganglion cell layer thickness in the inferior portion of the macula, besides the increased RNFL thickness of the optic nerve head. (Fig. 1, C, and D).

The patient had significant hemochromatosis, evidenced by an elevated serum ferritin level of 3200 ng/ml. Erythrocyte sedimentation rate (ESR) and serum C reactive protein (CRP) were checked, and they were within the standard limit. Complete blood count (CBC) revealed anemia (Hb: 9) and a rise in the platelet count (800×1000) that could be due to the previous splenectomy. The coagulation test revealed a PT of 16.2 s, PTT of 32.5 s an INR of 1.2. liver function tests and thyroid function tests were within normal ranges, and viral markers for HBV, HCV, and HIV were negative.

The patient remains under regular follow up with hematologist for ongoing management of his beta thalassemia. Additionally, he has routine follow up of an ophthalmologist to monitor the ocular health and detect any complications. After 6 months, in the follow-up examination, the visual acuity of the right improved to 20/100 with 1 + RAPD. Despite the improvement in the visual acuity, fundus examination revealed subtle right disc pallor (Fig. 2, A, B), a generalized decrease in the macular ganglion cell layer thickness and optic nerve head RNFL thickness (Fig. 2, C and D), and OCT angiography revealed generalized decreased vessel density of superficial vascular complex (SVC) (Fig. 3). Humphrey visual field analyzer C-24-2 revealed generalized depression. (Fig. 4) Visual field examination with an automated perimeter was impossible due to severely impaired central vision at the presentation time.

The patient gave written consent for the description of all data.

Discussion

This paper takes a new look at AION as a cause of decreased visual acuity and impaired visual field in beta-thalassemia patients. Although chronic hypoxemia and iron overload contribute to optic ischemia, there is no previous report of AION with a visible clot in the fundus in the case of beta-thalassemia.

Deferoxamine is a chelating agent widely used in hematological diseases with iron overload. What we know about deferoxamine and its ability to cause neuroophthalmological complications in beta-thalassemia patients is based on previous studies. In an earlier investigation, the prevalence of ocular toxicity associated with Deferoxamine was reported to be 1.2%. Among the observed ocular complications mainly involved the RPE layer, there were central blurriness and alterations in retinal pigmentation and Macular RPE mottling [4–6]. We cannot ignore the possible role of this drug in causing AION in our patients. Earlier findings showed that deferoxamine alters the electrophysiological retinal tests and may not affect vision clinically; this patient presented to the ophthalmology clinic with a chief complaint of reduced vision [5, 6].

Besides the iron chelating drugs effect, splenectomy can also cause microvascular abnormality in the retinal and choroidal vessels. Koctekin. et al. noted that coriocapillary flow decreased in the case of beta thalassemia with prior splenectomy surgery [7]. This effect may contribute to the optic nerve's vulnerability to ischemia.

Contrary to the expectation of permanent visual field defect of AION, our patient's visual acuity and visual field improved after 3 months. A decrease in the inner retinal layer thickness map on optical coherence tomography (OCT) despite the improvement in the visual parameter can be related to the beta-thalassemia disease effect on microvascular abnormality and reduced macular thickness, as mentioned by Haghpansh et al. [8]

Occurring optic nerve vascular occlusion in our patient, besides the previous report of central retinal vein and artery in case of beta thalassemia, draws attention to the possibility of any vascular obstruction as the leading cause of impaired vision in these patients and highlights the importance of prompt fundus examination, early diagnosis and treatment of underlying systemic disorders in all cases of beta-thalassemia [9].

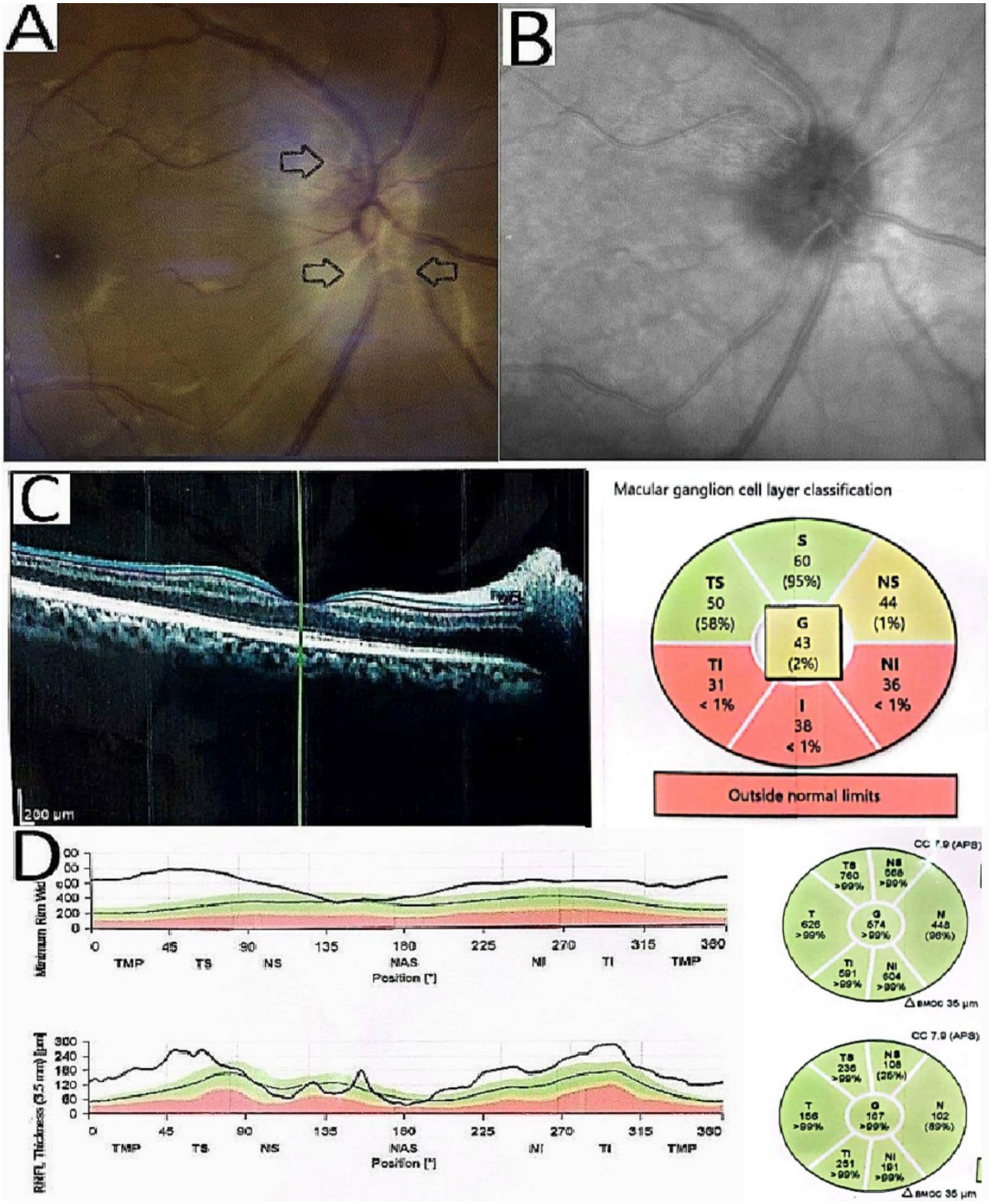

Fig. 1 Right fundus photograph (**A**) and Right Infrared fundus image (**B**) Shows the optic disc edema with RNFL opacification and blurred margins that are more severe in the superior besides the visible vascular occlusion in superior and inferior (arrows). Macular ganglion cell OCT thickness map. Visible RNFL and inner retinal layer edema (**C**). Optic nerve head OCT shows the increase optic nerve head RNFL thickness (**D**)

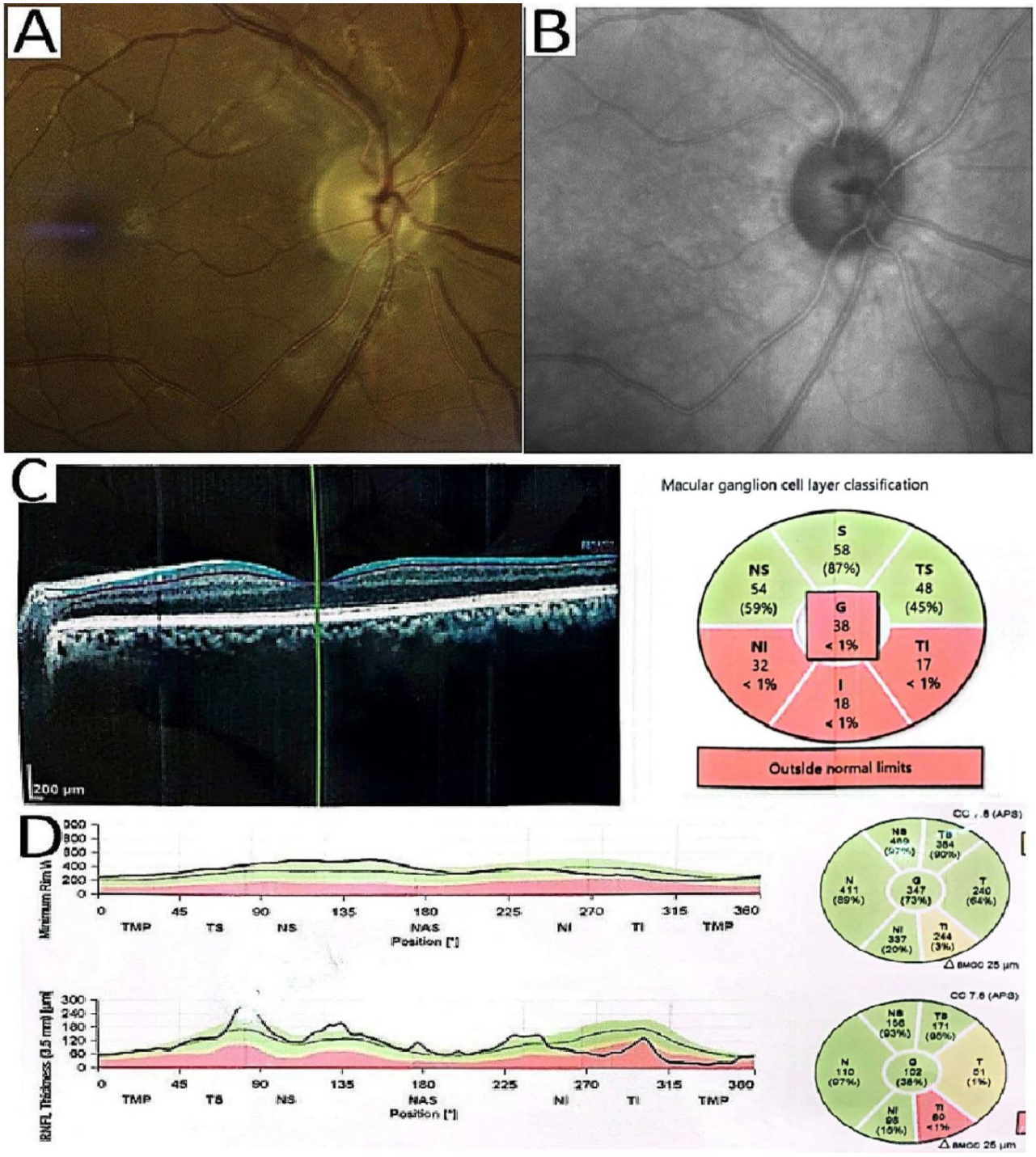

Fig. 2 Right fundus photograph (**A**) and Right Infrared fundus image (**B**) the subtle generalized disc pallor after 6 months. Macular ganglion cell OCT thickness map (**C**) generalized RNFL and inner retinal layer decrease thickness. Optic nerve head OCT (**D**) the RNFL edema resolved after 6 months with a mild decrease in superior and inferior thickness

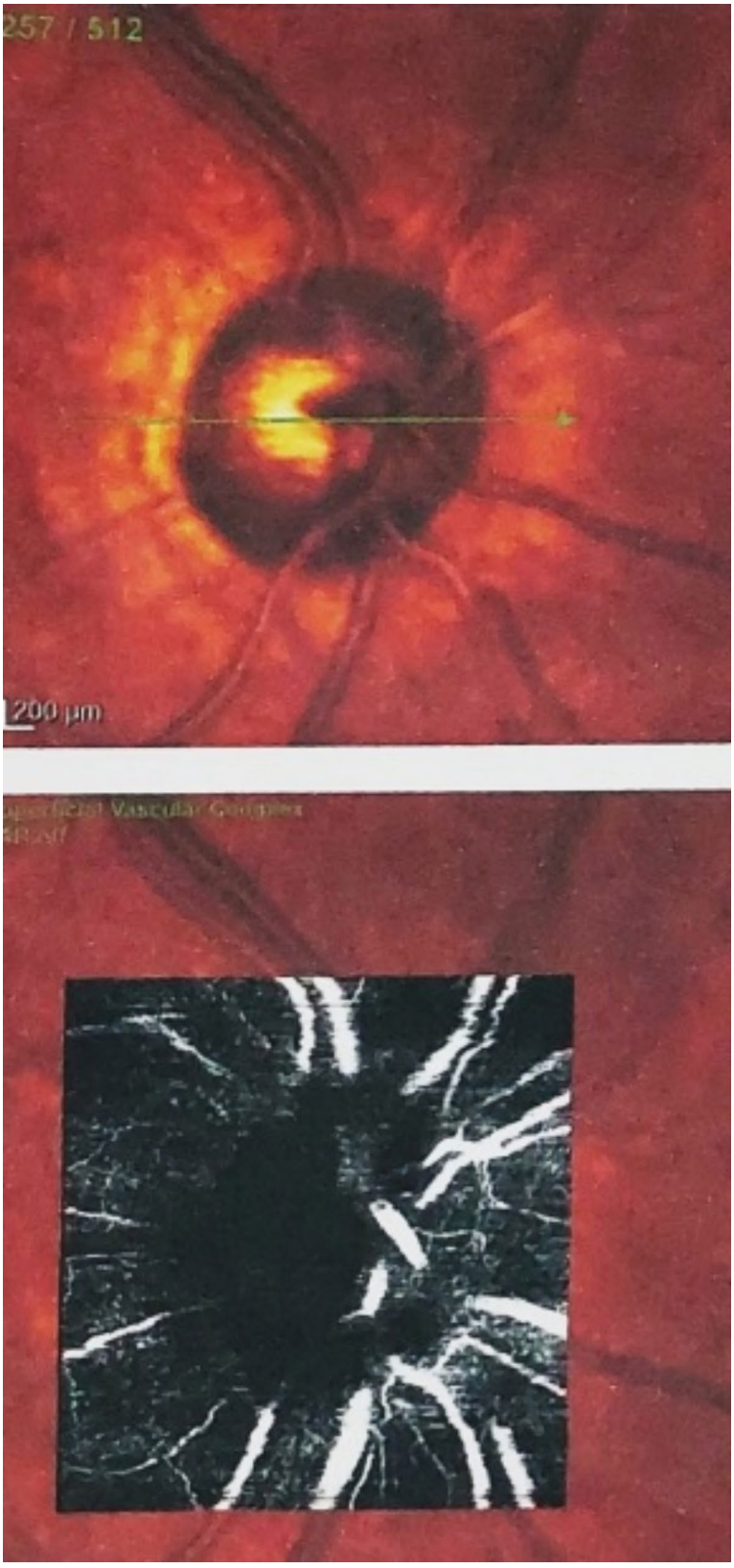

Fig. 3 Rt eye optic disc OCTA. Revealing generalized decreased vessel density of superficial vascular complex (SVC) in follow up

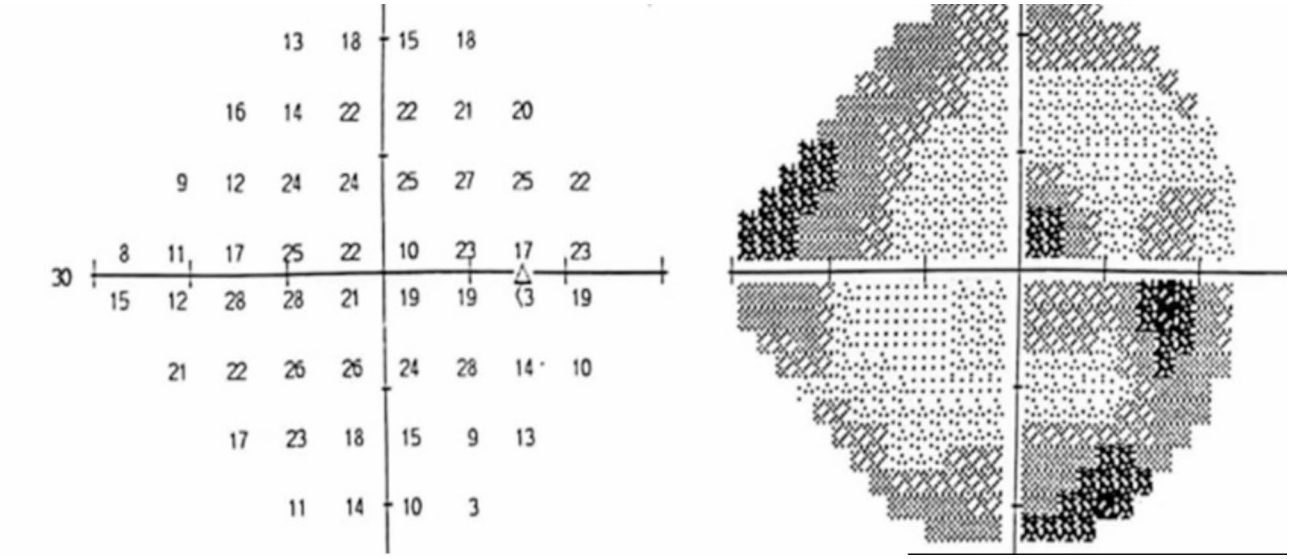

Fig. 4 Rt eye Humphrey automated perimetry. Gray scale and a numerical map showing generalized depression after 6 months

Conclusions

This case report highlighted the importance of regular fundus examination in all patients who suffer from beta thalassemia, especially ones who received deferoxamine and had previous splenectomy surgery and consider optic nerve head vascular obstruction and microvascular abnormality as the cause of various types of acute vision loss.

Abbreviations

AION	Anterior ischemic optic neuropathy
BCVA	Best corrected visual acuity
CBC	Complete blood count
CRP	C reactive protein
ESR	Erythrocyte sedimentation rate
GAT	Goldmann applanation tonometry
NAION	Nonarteritic anterior ischemic optic neuropathy
OCT	Optical coherence tomography
RAPD	Relative afferent pupillary defect
RNFL	Retinal nerve fiber layer
SVC	Superficial vascular complex

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Author contributions

Zahra Zia and Ali Azimi contributed to the design of the study and preparation of data and photographs and write the manuscript draft. All authors read and approved the final manuscript.

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Data availability

No datasets were generated or analysed during the current study.

Declarations

Ethics approval and consent to participate

This study received ethical approval from the Shiraz University of Medical Sciences Ethics Committee. Informed consent was obtained from the participant included in the study. The study was conducted following the ethical standards as outlined in the Declaration of Helsinki.

Consent for publication

Informed consent was obtained from the participate.

Competing interests

The authors declare no competing interests.

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References

1. Liaska A, et al. β-Thalassemia and ocular implications: a systematic review. BMC Ophthalmol. 2016;16(1):1–13.
2. Hattenhauer MG, et al. Incidence of nonarteritic anteripr ischemic optic neuropathy. Am J Ophthalmol. 1997;123(1):103–7.
3. Cetin T, et al. The effect of beta-Thalassemia minor on the optic nerve head analysis. Blood. 2005;106(11):3831.
4. Baath JS, et al. Deferoxamine-related ocular toxicity: incidence and outcome in a pediatric population. Retina. 2008;28(6):894–9.
5. El-Shazly AA, et al. Electroretinography and visual evoked potential changes in relation to chelation modality in Childeren with thalassemia. Retina. 2017;37(6):1168–75.
6. Di Nicola M, et al. Functional and structural abnormalities in deferoxamine retinopathy: a review of the literature. Biomed Res Int. 2015;1:249617.
7. Koctekin B, et al. Optical coherence tomography angiography findings in transfusion-dependent beta-thalassemia patients with and without splenectomy. Photodiagnosis Photodyn Ther. 2023;42:103282.
8. Haghpanah S, et al. Optical coherence tomography findings in patients with transfusion-dependent β-thalassemia. BMC Ophthalmol. 2022;22(1):279.
9. Li H, et al. Combined central retinal vascular occlusion as the presenting feature in β-thalassemia with iron deficiency anemia. Retin Cases Brief Rep; 2022.

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