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Case Report

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A Multidisciplinary Approach to Anaemic Retinopathy in a Young Female with Thalassemia Major

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ABSTRACT

Anaemic retinopathy is often managed solely from an ophthalmic perspective overlooking the underlying systemic pathology. In our report we present a case of Thalassemia Major with secondary Anaemic Retinopathy, who was referred to us with a complaint of diminution of vision. Instead of going forward with any invasive ophthalmic procedure we explored conservative treatment options with a focus on managing the systemic pathology. This collaborative approach between different medical specialities helped in achieving near complete resolution of patient's ocular condition and regaining the lost vision over a period of six months.

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Introduction

Anaemic Retinopathy (AR) is the retinal manifestation of systemic anaemia. It has an incidence of around 28% when the haemoglobin level is below 8g/dL. The reduced haemoglobin levels cause retinal hypoxia leading to vascular changes. The key clinical features of AR include retinal haemorrhages which occur in various layers of retina including flame-shaped, dot-blot, and preretinal haemorrhages. Roth spots and cotton wool spots are also frequently observed along with optic disc edema and sub-hyaloid haemorrhages. In this report the patient presented with AR secondary to thalassemia major along with hypersplenism and Vitamin B12 deficiency. We intend to emphasize the importance of a multidisciplinary approach in such cases to achieve favourable results- as documented in sequential follow ups and imaging.

Case Description

A 17-year-old female was referred to Choithram Netralaya by her haemato-oncologist following a complaint of diminution of vision. The patient is a known case of thalassemia major, and developed the ophthalmic complaint after her third unit of blood transfusion creating an alarm amongst her parents.

On initial examination the patient's BCVA was 2/60 in both eyes, while her IOP was 13 mmHg in the right eye and 14 mmHg in the left eye. On Slit Lamp examination, the Anterior Segment was within Normal limits in both eyes.

On Posterior Segment examination multiple multi-layered retinal haemorrhages were seen on the posterior pole as well as scattered dot-blot haemorrhages were seen in the periphery of both eyes (Fig 1A). On further examination, OCT of both eyes correlated well with the fundus findings of retinal haemorrhages (Fig 1C). The FFA performed to rule out sickle cell retinopathy, showed central blocked fluorescence and no peripheral neovascularization (Fig 1B).

The patient's haemoglobin at presentation was critically low at 4.5 g/dL along with a body weight of 39 kgs. The investigations and clinical findings helped to ascertain the diagnosis as Anaemic Retinopathy secondary to thalassemia major.

Instead of planning any surgical intervention we proceeded with a conservative systemic management of the condition. Collaborating with her haemato-oncologist we focussed on correcting the haemoglobin levels through regular blood transfusions. Additionally, the patient was started on oral folic acid tablets and Vitamin K and B12 injections. The patient was also prescribed hydroxyurea oral capsules along with multivitamin tablets.

The parents were counselled to be vigilant about patient's haemoglobin levels and maintain them above 10 g/dL. The patient was advised regular follow-ups with ophthalmologists and haemato-oncologists. Over the next three months the patient's systemic condition improved under haematological care. The haemoglobin level rose to about 9.5 g/dL with continued blood transfusions and supportive therapy.

At 6-month follow up the patient's investigations revealed a reassuring reversal of symptoms. The BCVA was 6/9 in both eyes. Similarly encouraging findings were noted in fundus examination as well where there was complete resolution of retinal haemorrhages (Fig 2A). OCT confirmed the effectiveness of treatment with normalization of the retinal layers, consistent with clinical recovery (Fig 2B).

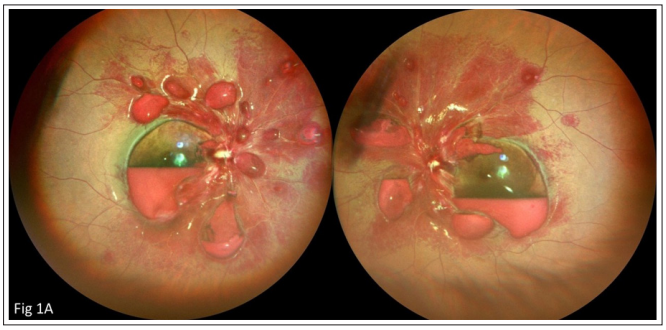

Figure 1A: Right and Left eye Fundus Photograph Showing Multiple Multi-Layered Retinal Haemorrhages

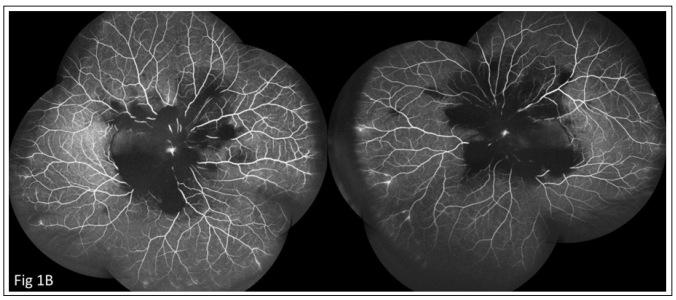

Figure 1B: Right and Left eye Fundus Fluorescein Angiography Showing Central Blocked Fluorescence and No Peripheral Neovascularisation

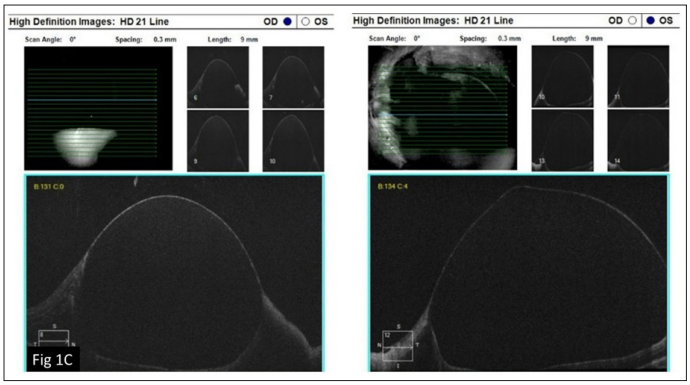

Figure 1C: OCT Image of Both Eyes Showing Retinal Haemorrhages

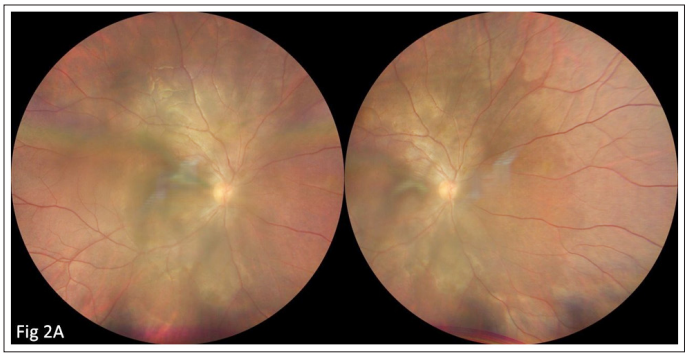

Figure 2A: Six Month Follow Up Fundus Photograph of Both Eyes Showing Resolution of Retinal Haemorrhages along with Healing Vitreous Haemorrhages

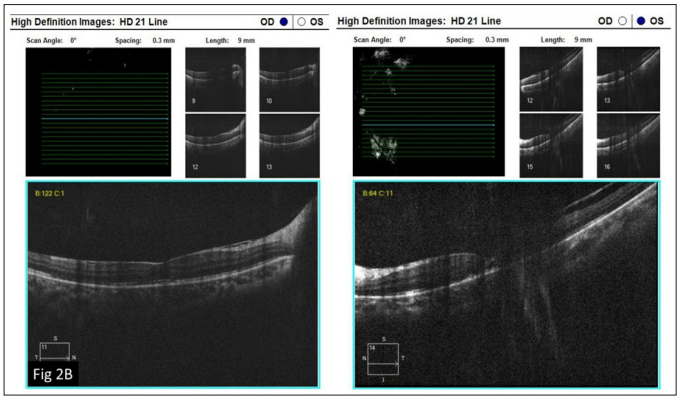

Figure 2B: Six Months Follow Up OCT of Both Eyes Showing Resolved Retinal Haemorrhages along with Slightly Raised Internal Limiting Membrane (ILM) in Left Eye

Discussion

Anaemic retinopathy is characterized by the presence of numerous symptoms and fundus findings caused by the underlying retinal hypoxia and vascular compromise. The diagnosis is hence complex and requires a combination of thorough systemic and ophthalmic evaluation. Clinical signs of AR may include retinal haemorrhages (flame-shaped or dot-blot), Roth spots, cotton wool spots and optic disc edema or pallor. These should be evaluated in conjunction with the OCT and FFA findings. A systemic evaluation using a CBC, Hb and haematocrit, and peripheral blood smear investigations is required to confirm the diagnosis.

The underlying pathophysiology of AR is a decrease in the blood's oxygen carrying capacity, which results in retinal hypoxia. It causes compensatory vascular dilatation along with increased capillary permeability and microtrauma to the vessel walls in retina. These vascular changes render the retina vulnerable to edema and haemorrhages. The eventual nerve fibre layer infarction manifests as cotton wool spots.

With proper interventions to augment the haemoglobin levels, there is a subsequent improvement in the retinal oxygenation. Correcting the hypoxia also aids in stabilising the retinal vasculature which ultimately leads to the resorption of retinal haemorrhages and exudates. Hence, timely treatment of anaemia can not only stop the progression of retinal damage, but it can also reverse it- helping bring the visual acuity back to baseline levels.

The case also highlights the crucial importance of a multidisciplinary approach in the management of AR. No surgical intervention was required in the patient despite alarming fundus findings. Ocular improvement could be achieved by systemic optimization, demonstrating that retinal haemorrhages in AR can be self-resolving if the underlying anaemia is addressed. The case also emphasizes the necessity of FFA in similar cases to rule out neo-vascularisation and peripheral ischemic changes, especially when Sickle cell disease is in the differential [1-3].

Conclusion

In young patients with severe retinopathy due to anaemia, particularly secondary to diseases such as thalassemia major, systemic correction often results in complete visual and structural recovery. This case illustrates how close coordination between haematologists and ophthalmologists can help avoid unnecessary procedures and result in successful conservative management.

Conflicts of Interest: None

Financial Support: Nil

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