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RETINOPATHY IN A CASE OF FAMILIAL HYPERCHOLESTEROLEMIA WITH DIABETES- A RARE CASE REPORT.

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ABSTRACT

Background

Familial hyperlipidemia is a genetic disorder characterized by high levels of LDL cholesterol and is associated with an increased risk of sudden cardiac death. However, the majority of cases of familial hypercholesterolemia remain undiagnosed or diagnosed at a later stage. Hyperlipidemia per se plays a major role in the pathophysiology of diabetes and its complications. Lipid-lowering agents have a protective effect on the progression of diabetic retinopathy and reduce the risk of diabetic macular oedema. Hence in cases of familial hypercholesterolemia with diabetic retinopathy a combined and aggressive approach is vital for better visual outcome.

Case presentation

In this report, an unusual case of 54 years old male presented to the ophthalmology OPD of Kalinga Institute of Medical Sciences, Bhubaneswar with diabetic retinopathy which was unresponsive to conventional treatment. Upon thorough evaluation, he was diagnosed with a case of familial hypercholesterolemia, and lipid-lowering therapy was initiated along with intravitreal anti-VEGF injections to which he responded better and there was a significant improvement in visual acuity.

Conclusion

This case report emphasizes that all cases of diabetic retinopathy unresponsive to treatment should be thoroughly evaluated. Familial hypercholesterolemia is a grave disease with serious implications. Early diagnosis and timely intervention play a crucial role in improving visual prognosis and life expectancy in such patients.

Keywords: *Familial hypercholesterolemia, Xanthomas, Anti-vascular endothelial growth factor, Statins, Diabetic retinopathy*
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INTRODUCTION

Familial hypercholesterolemia (FH) is a genetic disorder associated with high levels of low-density lipoprotein cholesterol (LDL-C). Most cases of FH are autosomal dominant, however, rarely it can be autosomal recessive. FH can occur in homozygous or heterozygous forms. Mutations in APOB, LDLR, and PCSK9 genes are responsible in most of these cases **(1).** FH is a relatively common genetic disorder affecting 20 million people worldwide but 90% of cases remain undiagnosed. FH is characterized by elevated levels of LDL-C cholesterol. Clinical findings in FH include increased risk of premature cardiovascular events such as angina and myocardial infarction, skin and tendon xanthomas (cholesterol deposits in tendons), Xanthelasmas (yellowish, waxy deposits on eyelids), and corneal arcus at an early age **(2)**. Diabetic retinopathy is one of the leading causes of blindness worldwide and hyperlipidemia has a significant role in the progression of diabetic retinopathy and the development of macular oedema. As per recent studies, some lipoproteins are closely linked with the pathogenesis of diabetic retinopathy and lipid-lowering drugs have been found to alleviate diabetic retinopathy **(3)**.

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CASE PRESENTATION

A case of 54 years old male presented to the ophthalmology OPD of Kalinga Institute of Medical Sciences, Bhubaneswar, with chief complaints of diminution of vision in both eyes for 8 months. The patient was alright 8 months back. To start with he developed a diminution of vision in both eyes which was painless, progressive not associated with pain, photophobia, and watering. He was diagnosed with a case of diabetic retinopathy and treated for the same at other centers but there was no significant improvement in vision for which he was referred to this hospital for further management. The patient was diagnosed with type 2 DM 9 years back and hypertension 1 year back. The patient was on oral hypoglycemics, antihypertensives, and topical NSAID eye drops. He had been treated elsewhere with 2 doses of intravitreal anti-VEGF injections 2 months back. The patient had no previous history of coronary artery disease, renal, hepato-biliary, or thyroid disease. However, there was a positive family history of hypercholesterolemia in his 25 years elder son since 1 year and there was the presence of xanthelasmas and skin, and tendon xanthomas (Fig:1,2) in his 21 years younger son who had accompanied him.

Fig 1: The presence of xanthelasmas in the younger son

Fig 2: The presence of xanthomas in the younger son

On general examination, the patient was conscious, cooperative, and well-oriented to time, place, and person, vitals were stable. On ocular examination, BCVA was CF 3 meters in both eyes and slit lamp examination showed

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immature cortical cataractous changes with nuclear sclerosis grade 1 in both eyes. Fundus examination with indirect ophthalmoscope showed multiple hemorrhages, extensive hard exudates throughout the fundus, and the presence of new vessels in both eyes (Fig 3, 4). FFA showed leakage of dye in the superotemporal quadrant in the right eye and the inferotemporal quadrant in the left eye. OCT findings were the presence of diffuse macular oedema in the right eye and macular oedema with posterior hyaloid traction in the left eye.

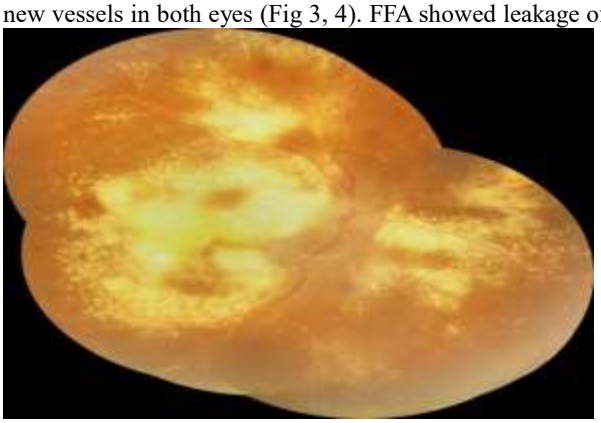

Fig 3: Fundus photo of RE pre-treatment

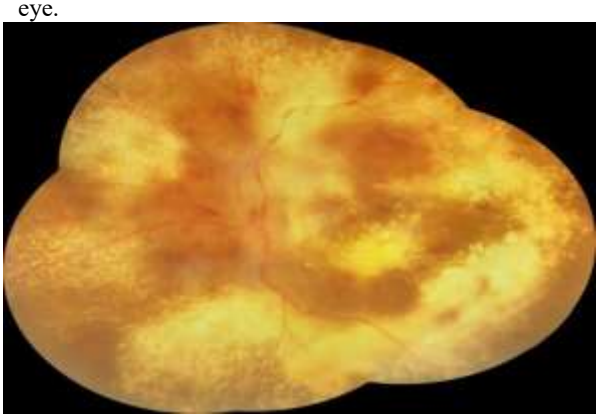

Fig 4: Fundus photo of LE pre-treatment

The presence of extensive hard exudates arouses suspicion of some other associated disease. So, on careful questioning patient gave a history of hypercholesterolemia in his elder son diagnosed elsewhere 1 year back. Likewise, an examination of his younger son, who had accompanied the patient revealed the presence of eyelid xanthelasma and xanthomas of extremities (fig3,4). However physical findings of our patient were normal. Serological investigations are as follows- FBS-248 mg/dl, PPBS- 308 mg/dl, HA1C-9.8%, serum cholesterol-245 mg/dl, serum triglyceride- 490 mg/dl, LDL-182 mg/dl, VLDL-29mg/dl, HDL-34mg/dl. ECG AND ECHO were normal. Based on these findings the patient was diagnosed as a case of familial hypercholesterolemia according to Dutch Lipid Clinic Network Clinical criteria **(4)** with proliferative diabetic retinopathy and macular oedema. Therapy was initiated with an Intravitreal injection of Bevacizumab (Anti VEGF) 1.25mg/0.05ml in both eyes under topical anesthesia with the help of a 30G needle and the patient was started on lipid-lowering agents- Fenofibrate (Lipicard)- 200mg OD at night, Rosuvastatin- 20mg OD after dinner. Oral hypoglycemics and anti-hypertensives were continued. On follow up repeat intravitreal injections were given every month for 7 months and serial fundus photo and OCT were taken at each visit. Serial monthly monitoring of lipid and blood sugar levels was done. On subsequent follow-ups, there was- a decrease in cholesterol levels, control of blood sugar, resolution of macular oedema, decrease in macular thickness, clearing of cholesterol deposits (Fig 5,6), and significant improvement in visual acuity from CF 3mts to 6/60 BES over 7 months.

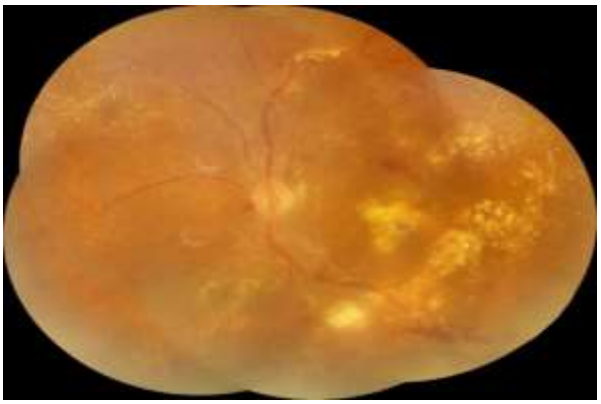

Fig 5: Fundus photo of RE posttreatment

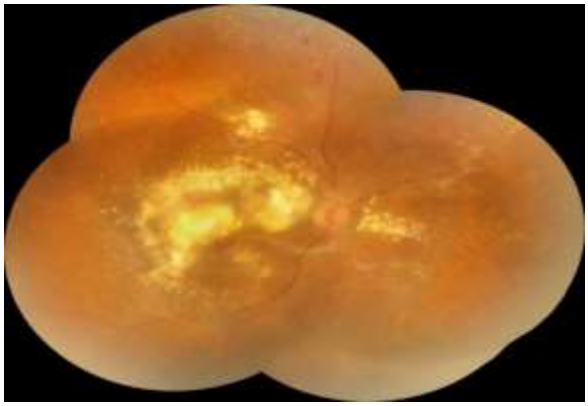

Fig 6: Fundus photo of LE post-treatment

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DISCUSSION

FH is a grave disease with onset in early childhood. But most cases are diagnosed late and many cases have a family history of sudden cardiac deaths. Although genetic testing is available most diagnosis is based on clinical features and history. Physical findings include corneal arcus, and xanthomas which if present at an early age should raise strong suspicion and such patients should be advised of lipid profile and careful evaluation. All cases of FH should undergo mandatory family screening **(4)**. Treatment of diabetic retinopathy in cases of FH poses a challenge. Apart from the treatment of macular oedema with anti-VEGFS, all cases should be referred to a physician for control of lipid levels. Lipid-lowering drugs such as statins, bile acid sequestrants, ezetimibe, and newer drugs like mipomersen, and lomitapide play a major role in such cases in determining the prognosis and final visual outcome **(5)**. In this case, there was a strong family history of hypercholesterolemia in both the sons of the patient and failure to respond to conventional therapy. All these led to further evaluation and appropriate diagnosis and management. Hence, in all such cases with diabetic retinopathy where there is a failure of response to conventional therapy and the presence of signs of significant hyperlipidemia, it is essential to take a thorough family history and early referral to a physician for screening and treatment of associated underlying disorders which can aggravate diabetic retinopathy and its related complications.

CONCLUSION

As per recent studies, both lipid-lowering drugs have shown beneficial roles in preventing the progression of diabetic retinopathy, and aggressive therapy for diabetic patients with hyperlipidemia is required for positive outcomes. Hence a combined approach involving physicians and ophthalmologists is required in such cases of familial hypercholesterolemia for better outcomes.

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