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The Twists and Turns of Neurofibromatosis: a case report

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INTRODUCTION

Neurofibromatosis has the most diversified ocular findings among the phakomatoses. This condition can involve the eyelid, conjunctiva, aqueous outflow channels, uveal tract, retina, orbit, and optic nerve.[1] Type 1 neurofibromatosis (NF1) or Von Recklinghausen disease is characterized by cutaneous manifestations and is related to an abnormality on chromosome 17 by an autosomal dominant mutation in the NF1 gene that leads to decreased production of the protein neurofibromin, which has a tumor suppressor function.[1] Occurs at a rate of 1 out of every 3000 persons.[1]

An ocular feature initially described in 2002 is represented by retinal microvascular abnormalities.[2] Three different vascular arrangements are described in literature: simple vascular tortuosity, corkscrew retinal vessels' configuration and the moyamoya-like pattern.[2]

METHOD

This report aims to describe a case of a retinal microvascular abnormality in a patient with neurofibromatosis type 1.

CASE REPORT

A 47-years-old male patient attends a routine check-up. As part of his medical history, he reports having neurofibromatosis type 1. During the examination, he demonstrates a best-corrected visual acuity of 20/20 in both eyes. Slit-lamp biomicroscopy reveals a neurofibroma on the upper eyelid of the right eye, with no other significant findings.

Fundus biomicroscopy reveals a spiral attitude of vessels superior temporal arcade of the right eye, a 'corkscrew' pattern configuration. No alterations were observed in the left eye.

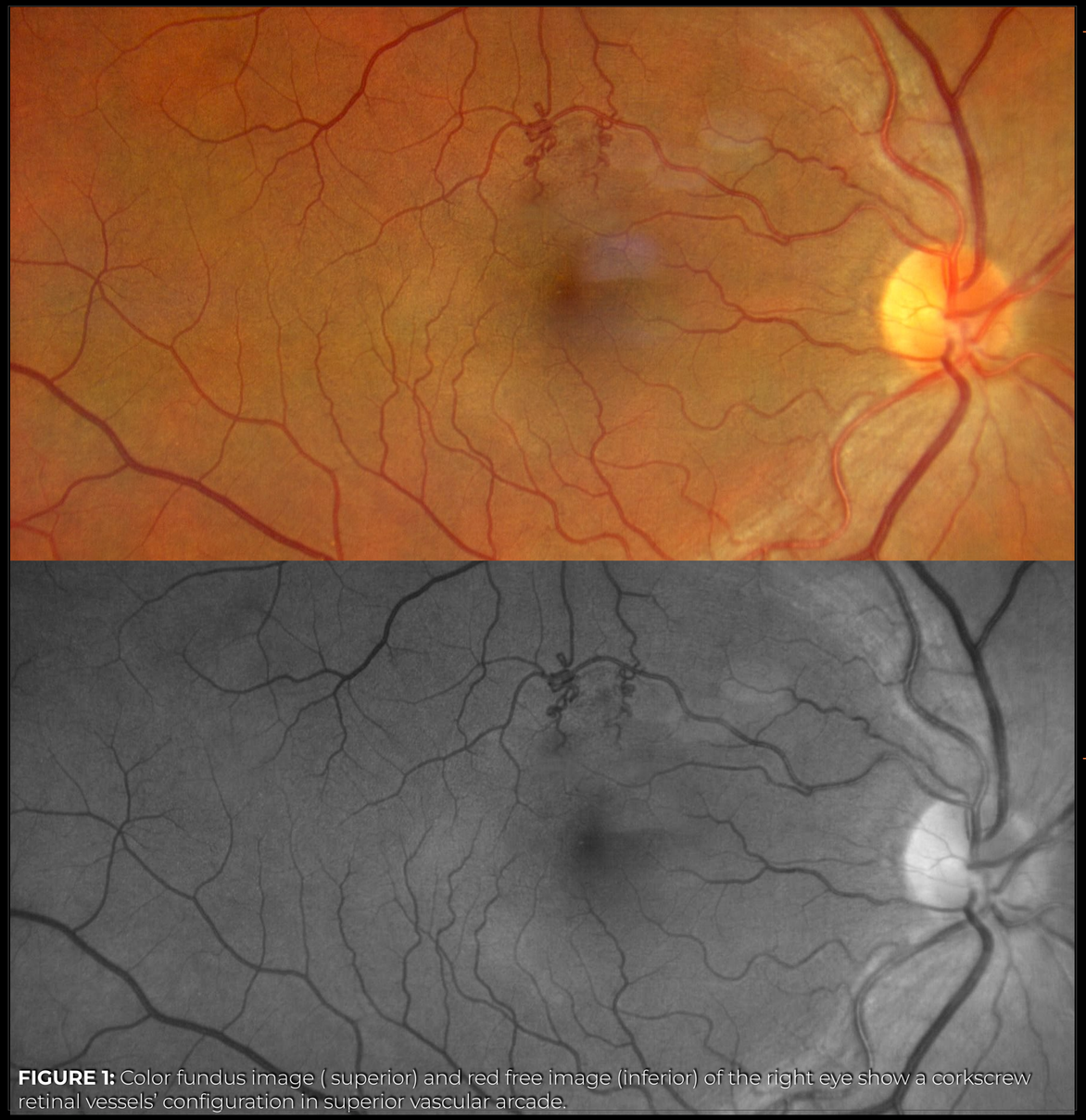

FIGURE 1: Color fundus image (superior) and red free image (inferior) of the right eye show a corkscrew retinal vessels' configuration in superior vascular arcade.

DISCUSSION

Neurofibromin is typically present in the endothelial and smooth muscle cells lining blood vessels. It has been proposed that the vascular issues associated with NF-1 stem from a lack of neurofibromin in these cells.[2]

The diagnostic specificity and positive predictive value of the corkscrew retinal pattern and moyamoya-like type establish a strong correlation with the diagnosis of NF1.[2] The presence of retinal microvascular abnormalities did not correlate with age and sex and not with any specifc features of NF-1, including Lisch nodules and choroidal abnormalities.[3]

In a 2019 study with 334 patients with NF1, microvascular abnormalities were detected in 30% being more prevalent in females and in adulthood (after the age of 16), age found in the described case.[2] Retinal vascular abnormalities have been reported in patients affected by NF1, but their frequency, diagnostic relevance, and pathologic implications still need to be clarified.[4] They are present in a moderate proportion of patients affected by NF1 and can be considered an additional distinctive sign of the disease. [4]

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