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Case Report

Peripheral retinal ischemia in a young Indian woman with neurofibromatosis type 1

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Abstract

Neurofibromatosis type 1 (NF-1) is an autosomal dominantly inherited disease characterized by café-au-lait spots, neurofibromas, axillary freckling, Lisch nodules of iris, gliomas and various systemic vascular ischemic manifestations mainly in the aorta, brain and kidney. Retinal vascular manifestations in patients with NF-1 are usually representative of retinal capillary hemangiomatosis. Few cases of NF-1 with retinal vascular occlusive disease have been described. We describe a young Indian woman with NF-1 with unilateral peripheral retinal ischemia but no vascular abnormality at the posterior pole.

Keywords: Peripheral retinal ischemia, Neurofibromatosis-1, Café-au-lait spots

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Introduction

Neurofibromatosis type 1 (NF-1) is an autosomal dominantly inherited disease characterized by café-au-lait spots, neurofibromas, axillary freckling, Lisch nodules of iris, gliomas and various systemic vascular ischemic manifestations mainly in the aorta, brain and kidney.[1,2] The gene responsible for this condition, NF1, has been isolated to chromosomal region 17q11.2 and is a tumor-suppressor gene. Its gene product, neurofibromin has a domain homologous to the GTPase activating protein (GAP) family, and downregulates RAS activity and consequently controls cell proliferation. The loss of neurofibromin expression in NF-1 leads to increased RAS activity, which initiates the process of tumor formation in various parts of the body.[3]

Ocular manifestations in NF-1 are plexiform neurofibromas of the eyelids, Lisch nodules of the iris, congenital glaucoma, optic glioma, retinal astrocytic hamartomas, retinal capillary hemangiomas and microvascular retinal abnormalities.[4,5] Retinal vascular manifestations in patients with NF-1 are usually representative of retinal capillary hemangiomatosis.[4] Few cases with retinal vascular occlusive disease have been described.[6–9] Two cases with peripheral retinal ischemia and neovascular glaucoma have also been recently described.[10,11] We describe a young Indian woman with NF-1 with unilateral peripheral retinal ischemia and an unremarkable posterior pole.

Case report

A 26 years old female, a known case of neurofibromatosis type 1, presented with floaters in both eyes for one month and pain in central abdomen for 2 weeks. She had history of use of myopic glasses in both eyes (−10 DS) since 15 years and history of retinal detachment surgery in left eye 10 years back. The diagnosis of NF-1 was made in childhood, on the basis of physical findings such as multiple café-au-lait spots and neurofibromas throughout the body, axillary freckling and three first-degree relatives with NF-1. She was born at full term with a birth weight of 2500 g.

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Other than the findings of NF-1 (Fig. 1a–c), the general physical examination of the patient was unremarkable. Her best corrected visual acuity (BCVA) in the right eye was 20/20 and the left eye was 20/100. The anterior chamber was within normal limits in both eyes and the iris did not show any Lisch nodules or neovascularization. Her intraocular pressure (IOP) by Goldmann applanation tonometry was 18 mm of mercury in both eyes. On fundus examination in the right eye, the disk and macula were normal. There was mild venous dilatation at the posterior pole and multiple sclerosed vessels in the periphery (Fig. 2a). The left eye fundus showed an attached retina with 360° encirclage indent with dispersed pigmentation throughout, with subretinal fibrosis passing through the macula, probably suggestive of resolved retinal detachment (Fig. 2a). Both eyes did not have any peripheral treatable lesions.

The fundus fluorescein angiography of both eyes showed a normal arm-retina time with normal filling of all vessels. The right eye showed peripheral capillary non-perfusion (CNP) areas temporally and inferiorly without any macular leakage or neovascularization of the disk or elsewhere (NVD/NVE) (Fig. 3a). The FFA of the left eye did not show any capillary non-perfusion areas (Fig. 3b). Spectral-domain optical coherence tomography (SD-OCT) showed normal fovea in the right eye (Fig. 4a) and foveal thinning in the left eye (Fig. 4b).

The patient had a normal blood pressure and all other laboratory investigations such as blood sugar, complete blood count, coagulation profile, chest X-ray, Mantoux test, rheumatoid factor and anti-nuclear antibody done to rule out other causes of vascular occlusive disorder were found to be within normal limits. Non-invasive Doppler flow study of carotid arteries did not show any stenosis.

The patient underwent sectoral pan-retinal photocoagulation in the right eye and was subsequently referred to the department of surgery for abdominal pain, where an ultrasonography of abdomen was done that showed a right sided paravertebral lesion, 10 cm × 6 cm in size, abutting the liver and causing mass effect. Following this, an MRI (brain and spine) was done that revealed a dumb-bell shaped mass lesion in right paraspinal region at D10 level suggestive of a Nerve-Sheath tumor/neurofibroma (Fig. 1d).

At 1 year follow-up, the patient has maintained the same BCVA and does not exhibit any fibrovascular proliferation in the right eye fundus.

Discussion

Systemic vascular occlusion disease has been previously reported in NF-1 to affect aortic, cerebral, renal, celiac and mesenteric vessels.[2] The occlusive manifestations include stenosis of small vessels, progressive capillary ischemia and compensatory collateral circulation.

The pathogenesis of vascular occlusion is related to the growth of Schwann cells and dysplasia of arteriolar smooth muscle cells, which leads to a progressive narrowing of the vessel and consequent occlusion with secondary fibrous changes.[2] Ozerdem described NF-1 gene mutations as a cause of increased proliferation of pericytes and endothelial

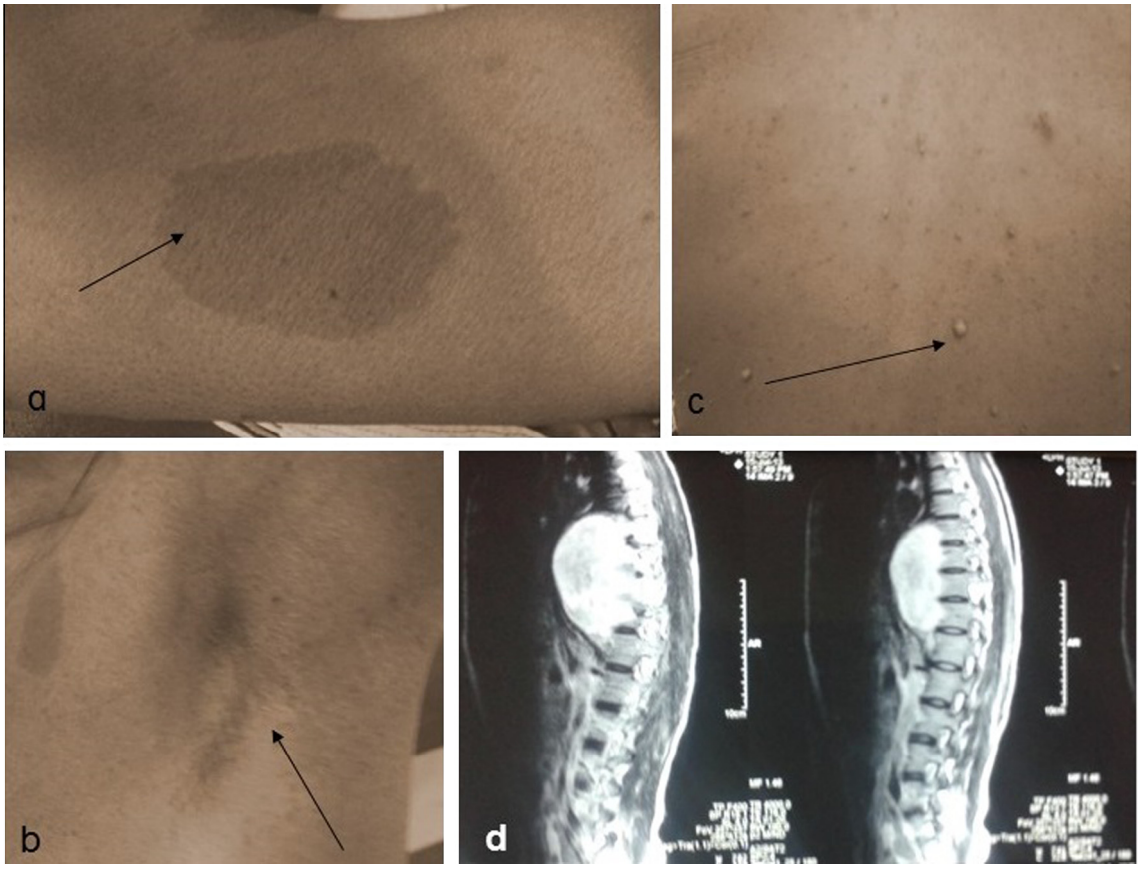

Figure 1. Showing (a) cafe au lait spots, (b) axillary freckling, (c) neurofibromas, and (d) MRI spine showing plexiform neurofibroma at the level of D10.