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Isolated Macular Cherry-Red Spot Without Systemic Disease

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Abstract:
Introduction: **Macular cherry-red spot can be a sign of different disorders. It is mostly a sign of retinal artery occlusion in adults, and sphingolipid storage diseases in infants. In the presenting case, parafoveal ganglion cell hypertrophy in an otherwise healthy man presented.**
Case presentation: **A 52 years old man presented with nearsightness. He had no significant past medical history except myopia. Best corrected visual acuity was 10/10 in both eyes. Dilated fundus examination showed bilateral abnormal parafoveal yellow-white light reflex resembling cherry-red spot. Midperipheral retina had tiny pigmentary mottling and pigment epithelium atrophy areas in both eyes. OCT demonstrated bilateral hyperreflective parafoveal thickened ganglion cell layer. Bilateral mottled hyperfluorescent areas of midperipheral retina was seen in fundus fluorescein angiography. Hexosaminidase A, β-galactosidase, neuraminidase, and acidsphingmyelinaze activity were normal. Abdominal ultrasonography and central nervous system imaging had no pathological findings.**
Conclusions: **This case seems a rare, sporadic, isolated retinal disorder with no systemic or ophthalmic associations.**

Key Words: Cherry-red spot, ganglion cell layer, sphingolipidoses

INTRODUCTION

Macular cherry-red spot can be a sign of different disorders. It is a consequence of ganglion cell layer thickening and blockage of choroidal red hue except foveola which is devoid of ganglion cell layer. In consequence, a central red area surrounded by a white halo exists. It is mostly a sign of retinal artery occlusion in adults, and sphingolipid storage diseases in childhood. In the presenting case, an isolated parafoveal ganglion cell layer thickening in an otherwise healthy man presented.

CASE PRESENTATION

A 52 years old man presented with nearsightness. He had no significant past medical history except myopia. Best corrected visual acuity was 10/10 in both eyes. Refractive error was -3,0-1,0*75 in right, and -2,25-0,50*91 diopters in left eye. Intraocular pressure was 15 mmHg in right, and 16 mmHg in left eye. Slit-lamp examination revealed normal anterior segment findings. Dilated fundus examination showed bilateral abnormal parafoveal yellow-white light reflex resembling cherry-red spot (Fig 1). Midperipheral retina had tiny pigmentary mottling and pigment epithelium atrophy areas in both eyes. OCT demonstrated bilateral hyperreflective parafoveal thickened ganglion cell layer (Fig 2). Bilateral mottled hyperfluorescent areas of midperipheral retina was seen in fundus fluorescein angiography Fig 3). Hexosaminidase A, β-galactosidase, neuraminidase, and acid sphingomyelinase activity were normal. Abdominal ultrasonography and central nervous system imaging had no pathological findings.

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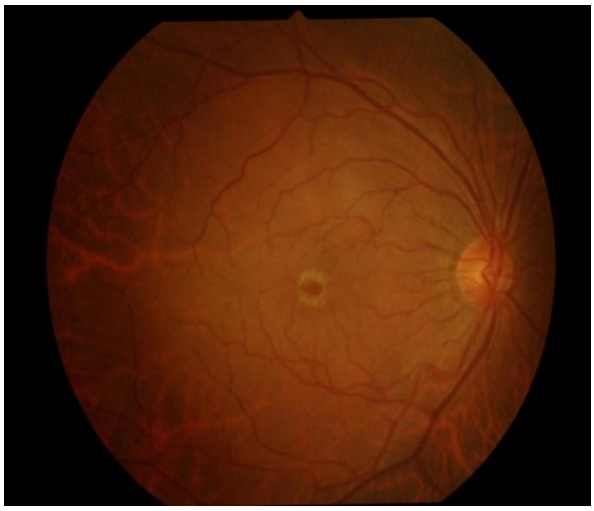

Fig 1. Perifoveal white reflex

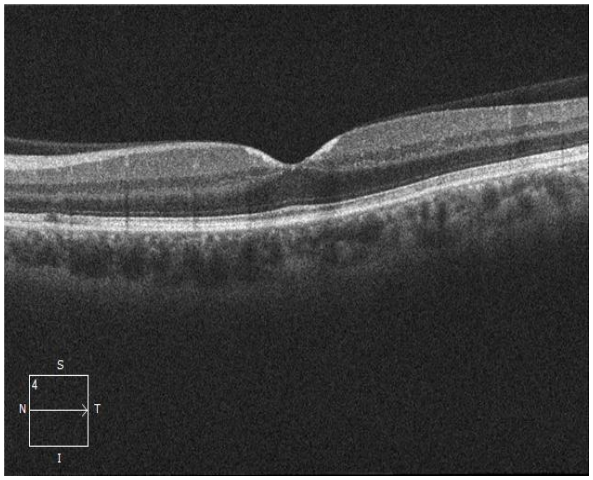

Fig 2: Parafoveal thickened ganglion cell layer

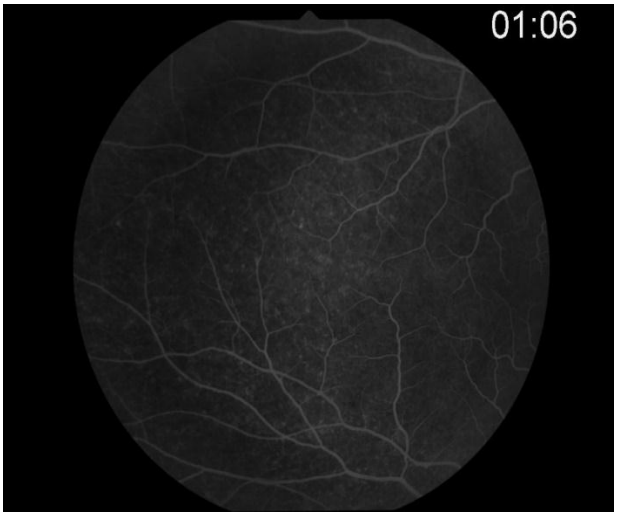

Fig 3: Midperipheral mottled hyperfluorescence

DISCUSSION

Macular cherry-red spot occurs in the sphingolipid storage diseases, which comprise a group of rare inherited metabolic diseases. Certain glycolipids and phospholipids accumulate in excessive quantities in various tissues, including the retina. The lipids accumulate in the ganglion cell layer of the retina, giving the retina a white appearance. As ganglion cells are absent at the foveola, this area retains relative transparency and contrasts with the surrounding opaque retina. Ganglion cells die progressively and the spot becomes less evident during the advanced stages. Finally, degeneration of the retinal nerve fiber layer and consecutive optic atrophy occurs. GM1 and GM2 (Tay-Sachs disease) gangliosidosis, sialidosis, Sandoff, Niemann-Pick and Farber diseases can cause accumulation of lipids in the ganglion cell layer. Most of these disorders are associated with multisystem involvement with mental retardation, myoclonus, seizures, hepatosplenomegaly, bone marrow involvement, skin lesions and cause death in childhood. Type E Niemann-Pick (adult type Niemann-Pick) patients can reach adulthood, but only type A and B have involvement of retina. Late onset sialidosis patients can reach adulthood but, associated with neurological disorders like myoclonus and seizures [1]

The presenting case shows parafoveolar ganglion cell layer involvement with macular cherry-red spot and pigmentary mottling in midperipheral retina pointing out a congenital hereditary disorder mentioned above. Nevertheless, the interesting thing is that systemic investigations revealed normal findings. Also, siblings of the patient didn't show a retinal or systemic disorder. Previously, cherry-red spot in adulthood is reported in a few papers. Esmer et al reported a familial disorder characterized with isolated cherry red spot and lens opacities [2]. Rudich et al reported three adult cases of Niemann-Pick disease with OCT findings similar to our case [3]. Kim et al reported adult onset familial cherry-red spot-myoklonus syndrome [4]. Nevertheless, cases reported in these papers were either familial or associated with systemic manifestations. In conclusion, this case seems a rare, sporadic, isolated retinal disorder with no systemic or ophthalmic symptoms.

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