Ibrahim Kocak, J Clin Exp Ophthalmol 2016, 7:3 (Suppl)
http://dx.doi.org/10.4172/2155-9570.C1.034

Isolated macular cherry-red spot without systemic disease

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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 was presented. A 52 year old man was presented with nearsightedness. 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 area of midperipheral retina was seen in fundus fluoresce in angiography. Hexosaminidase A, β-galactosidase, neuraminidase, and acidsphingmyelinaze activity were normal. Abdominal ultrasonography and central nervous system imaging had no pathological findings. Macular cherry-red spot occurs in the sphingolipid storage diseases, which comprise a group of rare inherited metabolic diseases. 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. The presenting case showed 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 did not show a retinal or systemic disorder. Previously, cherry-red spot in adulthood is reported in a few papers. 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 associations.

Biography

Ibrahim Kocak has completed his PhD from Istanbul University and Post-doctoral studies from Istanbul Training Hospital and Medipol University School of Medicine. He is an Assistant Professor in Ophthalmology Department of Medipol University, mostly associated with vitreoretinal surgery. He has published almost 20 papers in medical journals.

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