Case Report

Stargardt’s disease presenting with bilateral central ring scotoma

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Abstract

Ring scotoma is an annular field defect centered on fixation. Age-related macular degeneration in the elderly and hydroxychloroquine toxicity in younger patients are usual causes of central ring scotoma. We report bilateral ring scotoma as the presentation of adult onset Stargardt’s disease. Central ring scotoma has a precise localizing value, to a lesion involving macula.

Keywords: Stargardt’s disease, Ring scotoma, ABCA4 gene

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Introduction

Macular pathology usually presents with loss of visual acuity and/or central scotoma. However, central scotoma may not involve the fixation in some cases and the central visual acuity remains intact. The ring may be midperipheral, peri-central or central.1 Central ring scotoma involves 10 degrees of visual field. Central ring scotoma has been reported in several conditions, which include geographical atrophy in age-related macular degeneration, mitochondrial retinal dystrophy, central areolar choroidal sclerosis, acute macular neuroretinopathy, trauma, coffee and doughnut maculopathy1 as well as hydroxychloroquine toxicity. We report Stargardt’s disease presenting with bilateral central ring scotoma.

Case report

A 37-year man presented with slowly progressive blurred vision in the right eye of four weeks duration. He denied any history of photophobia or worsening of vision in bright light. He was unable to recall any history of genetic ocular diseases in other family members. His blood pressure was under adequate control with regular medications for past two years. He had undergone LASIK ten years ago. He did not smoke or consume alcohol and denied use of recreational drugs, phosphodiesterase5 inhibitors or hydroxychloroquine.

His visual acuity was 20/20 in both eyes. He was reading nine and eleven, out of thirteen Ishihara color plates, with the right and left eye respectively. Intraocular pressure was normal. Both pupils reacted to light and no afferent pupillary defect was observed. On slitlamp biomicroscopy anterior segment was normal. Fundus examination revealed mottling in both maculae more obvious in the right eye. Both discs were pink and well defined. He could see central dot but four corners of Amsler grid were not visible with either eye. Automated perimetry (24-2) showed bilateral central scotoma. Humphrey visual field (10-2) revealed 360-degree central ring scotoma with foveal sensitivity of 31 and 36 db in right and left eye respectively (Fig. 1). High resolution, line scan, Optical Coherence Tomography (OCT) to evaluate mottling of maculae (Fig. 2A), showed hyper reflective flecks in the inner aspect of retinal pigment epithelium and outer nuclear layers in both eyes (Fig. 2B). Fundus autofluorescence (FAF) revealed mottled area of hypo autofluorescence surrounded by a ring of hyper autofluorescence localized to the both
maculae (Fig. 2 C). Fundus fluorescein angiography (FFA) showed relatively dark choroid in both eyes. Right eye showed parafoveal hypofluorescent spots corresponding to flecks and hyperfluorescent areas due to retinal pigment epithelium atrophy (Fig. 2D). Macula 3D retinal thickness scan showed generalized thinning of maculae but the thinning was most pronounced in inner perifoveal ring. Full field electroretinogram (ERG) showed mild reduction of both rod and cone response with delayed flicker in both eyes. Latency of P100 was normal but amplitude was mildly reduced on pattern reversal visual evoked potential. The pattern of distribution of hyperreflective flecks on OCT, relatively dark choroid on FFA and fundus autofluorescence features was typical of Stargardt’s disease. Repeat HVF at nine months follow up examination showed mild progression of ring scotoma but visual acuity remained unchanged (Fig. 3).

Discussion

Stargardt’s disease (STGD) is an autosomal recessive macular dystrophy, which presents with progressive loss of central vision, including dyschromatopsia and central scotoma during first two decades of life. In adults it may present with metamorphopsia or oscillopsia without any decrease in visual acuity. The hallmark of STGD is a premature accumulation of lipofuscin in RPE cells and the sub-retinal space visible as yellow white flecks at posterior pole on fundus examination.

STGD is a phenotypically heterogeneous disease. Allikmets et al linked Stargardt’s disease to the mutation in ABCA4 gene. ATP-binding cassette (ABC) is an ATP dependent membrane bound transport protein encoded by ABC genes. Photoreceptor specific ABC, subfamily A-member 4 is also known as ABCA4. Protein encoded by ABCA4 gene is believed to clear a byproduct of the retinoid cycle of vision from photoreceptor cells. Mutation in ABCA4 can cause a spectrum of phenotypes ranging from mild macular dystrophy to severe pan retinal dystrophy. Macular involvement is the most consistent feature of ABCA4 related retina degeneration.

The age of onset relates to the severity of the underlying ABCA4 variants. The childhood onset is often associated with more deleterious effects while the adult onset STGD is often milder. The median age of presentation of foveal sparing adult onset phenotype is fourth decade, much later compared to foveal atrophy type.

Selective foveal sparing can be unilateral or bilateral. Presence of foveal atrophy or foveal sparing in the Stargardt’s disease suggests that there may be more than one disease...
mechanism in ABCA4 retinopathy. Although the mechanism of foveal sparing remains unknown, partial ABCA4 activity attributable to mild missense alleles, G1961E allele in either homozygosity or heterozygosity and higher prevalence of the variant p.Arg2030Gln have been reported in foveal sparing STGD.

Fig. 2. A: Color fundus photographs showing macular mottling. B: Macular OCT, line scan, showing hyperreflective flecks in the inner aspect of RPE and outer nuclear layer of macula. C: Fundus autofluorescence of the right and left eyes. D: Fundus fluorescein angiography showing parafoveal hyper fluorescent areas due to RPE atrophy (window defect). Hypofluorescent spots are due to flecks.
Our case illustrates that a subset of patients suffering from STGD can have normal visual acuity and the extent of photoreceptor dysfunction may not be evident from the clinically detectable fundus changes. STGD presenting with central scotoma, slight dyschromatopsia, decrease in amplitude of P100 with subtle foveal RPE changes can be erroneously diagnosed as atypical optic neuropathy. Compressive, demyelinating, and vascular disorder of optic nerve involves the fixation. Central scotoma sparing fixation indicates macular pathology.

Conflict of interest

The authors declared that there is no conflict of interest.

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