Fundus Autofluorescence and Fluorescein Angiogram Findings: Stargardt's Disease

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Abstract: Fundus Autofluorescence is a new imaging modality to study the pathology of the retinal pigment epithelial cells. A young patient with suspected diagnosis of bilateral dry macular degeneration was later-on diagnosed to have Stargardt's disease with the help of new ancillary test such as fundus autofluorescence in addition to other tests such as fluorescein angiography (FA) and optical coherence tomography (OCT).

Keywords: Fundus Autofluorescence, Stargardt's Disease.

CASE DESCRIPTION

A 28-year-old male presented with a two-year decrease in vision in both eyes and occasional floaters. He was diagnosed with macular degeneration at age 23 and is being observed for it annually. However, he noted decreasing central vision and difficulty in night driving. Amsler chart showed central distortion and metamorphopsia. The visual acuity was 20/50 in right eye and 20/100 in left eye. Intraocular pressures were within normal limits in both eyes. Anterior segment examination of both eyes was unremarkable. Fundus examination of both eyes showed similar findings. There were multiple yellowish spots in the posterior pole region with retinal pigment epithelial atrophy surrounded by peppering of the pigment in the center of the macula (Figure **1A**, **B**). The peripheral examination showed abnormal retinal pathology. Fundus Autofluorescence (FAF) demonstrated oval shaped hypofluorescence in the fovea suggestive of retinal pigment epithelial (RPE) atrophy, surrounded by areas of hyperfluorescence suggestive of lipofuscin deposition in the RPE (Figure **1C**, **D**). Heidelberg-Spectralis optical coherence tomography showed disruption of the RPE layer and abnormal deposits. There were multiple hyperautofluorescent areas in the posterior pole region of both eyes. Fluorescein angiogram showed dark choroid, a central hypofluorescence surrounding an area of hyperfluorescence (Figure **1E**, **F**) and few hyperfluorescent areas in the posterior pole region. Counseling was done regarding the visual prognosis and he was advised to use vitamin A.

DISCUSSION

Stargardt's disease is a common inherited macular degeneration characterized by a significant loss in central vision in the first or second decade of life [1]. Autosomal recessive Stargardt's disease, the most common macular dystrophy is caused by mutations in the gene encoding ABCA4, which encodes an ATP-binding cassette (ABC) transporter protein that is expressed by rod outer segments [2]. Patients present with bilateral loss of central vision over a period of several months with almost near normal peripheral vision. It is characterized by depigmentation and atrophy of the macular retinal pigmentary epithelium (RPE). Stargardts'disease is easy to recognize by the presence of discrete yellowish round or pisciform flecks at the level of the RPE in the macula (hyperfluorescent macular lesions-bull's eye). If the yellow specks spread, the entire fundus it is known as fundus flavimaculatus [1]. Fluorescein angiogram demonstrates "dark choroid" which was thought to be due to the masking of choroidal fluorescence by an accumulation of lipofuscin-like pigment throughout the RPE. FAF is a new non-invasive imaging modality to study the pathology of the RPE [3]. Damage to the RPE leads to excess accumulation of lipofuscin pigment, which was demonstrated by hyper-autofluorescence in the FAF images [3-5]. Whereas the dead or absent RPE were demonstrated by the hypo-autofluorescence on the FAF images [3-5]. The pisciform lesions in Stargardt's disease can be easily evaluated by the FAF images as shown in our patient's central macula (Figure **1C**, **D**). Vision loss is slowly progressive, in later stages is accompanied by central scotoma. Currently, there is no definitive treatment for Stargardt's disease. Low vision aids are helpful in these patients along with protection from the bright lights. Autofluorescence imaging was a best non-

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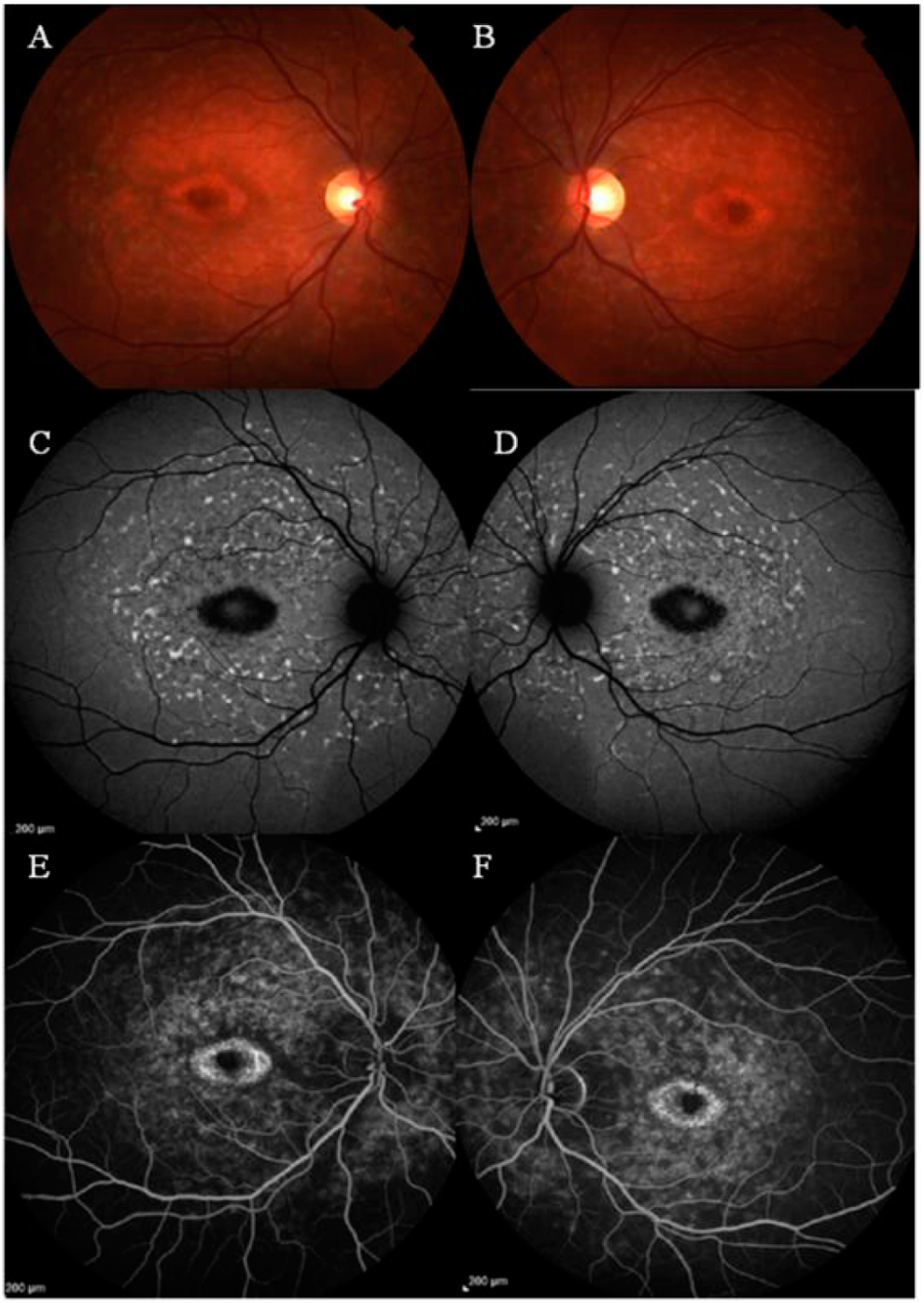

Figure 1: Color fundus photographs of the right and left eye showing pigment peppering with loss of pigment and multiple yellowish-white spots resembling drusen in the macula (Figure **1A**, **B**). Fundus Autofluorescence reveals an oval shaped hypoautofluorescence area in the fovea suggestive of RPE atrophy surrounded by areas of hyperautofluorescence suggestive of lipofuscin deposition in the RPE (Figure **1C**, **D**). Fluorescein angiogram demonstrates dark choroid, a central hypofluorescence surrounded area of hyperfluorescence (Figure **1E**, **F**) and few hyperfluorescent areas in the posterior pole region.

invasive tool for evaluation of changes on the RPE level typical for Stargardt's disease.

CONFLICT OF INTEREST

Authors have no interest in the devices or medication used in this paper.

REFERENCES

[1] Armstrong JD, Meyer D, Xu S, Elfervig JL. Long-term follow-up of Stargardt's disease and fundus flavimaculatus. Ophthalmology 1998; 105: 448-58. http://dx.doi.org/10.1016/S0161-6420(98)93026-3

[2] Rudolph G, Kalpadakis P, Haritoglou C, Rivera A, Weber BH. [Mutations in the ABCA4 gene in a family with Stargardt's

disease and retinitis pigmentosa (STGD1/RP19)]. Klin Monbl Augenheilkd 2002; 219: 590-6. http://dx.doi.org/10.1055/s-2002-34425

[3] Chen Y, Roorda A, Duncan JL. Advances in imaging of Stargardt disease. Adv Exp Med Biol 2010; 664: 333-40. http://dx.doi.org/10.1007/978-1-4419-1399-9_38

[4] Nandakumar N, Buzney S, Weiter JJ. Lipofuscin and the principles of fundus autofluorescence: a review. Semin Ophthalmol 2012; 27: 197-201. http://dx.doi.org/10.3109/08820538.2012.711415

[5] Cukras CA, Wong WT, Caruso R, Cunningham D, Zein W, Sieving PA. Centrifugal expansion of fundus autofluorescence patterns in Stargardt disease over time. Arch Ophthalmol 2012; 1 30: 171-9. http://dx.doi.org/10.1001/archophthalmol.2011.332

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