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CASE REPORT

Benign Familial Fleck Retina

Rohini Sangoram[1*] | V. H. Karambelkar[2] | Gaurav Paranjpe[3]

[1]Senior Resident, Department of Ophthalmology, Krishna Institute of Medical Sciences, Karad, Maharashtra, INDIA.

[2]Professor and HOD, Department of Ophthalmology, Krishna Institute of Medical Sciences, Karad, Maharashtra, INDIA.

[3]Assistant Professor, Department of Ophthalmology, Krishna Institute of Medical Sciences, Karad, Maharashtra, INDIA.

Abstract

Familial fleck retina is a rare inherited retinal disease. Sabel Aish & Dajani (1980) first reported ocular findings in seven of 10 siblings in one Arab family. It is an autosomal-recessive condition associated with a distinctive retinal appearance and no apparent visual or electrophysiological deficits . Affected individuals are asymptomatic with a large number of yellow–white flecks of variable size and shape in the midperipheral to far peripheral retina, but did not have any ocular complaints such as loss of visual acuity (VA), impaired visual fields and dark adaptation disturbances. Fluorescein angiograms documented an irregular and spotty hyperfluorescence throughout the retina (sparing the macula). This report discusses a case of 22 year old female of both eye Benign familial fleck retina.

1 | INTRODUCTION:

Benign fleck retina refers to an autosomal-recessive condition associated with a distinctive retinal appearance and no apparent visual or electrophysiological deficits. (1) Affected individuals are asymptomatic, but fundus examination reveals a striking pattern of diffuse, yellow-white, fleck-like lesions extending to the far periphery of the retina but sparing the foveal region. (2–5) The phenotype associated with benign fleck retina was first described in 1980 in seven affected siblings born to consanguineous parents. (2) A similar clinical appearance was subsequently reported in three unrelated individuals originating from diverse ethnic backgrounds. (3–5) Elucidating the genetic basis of human ocular phenotypes such as that of benign fleck retina remains a major goal because it will provide important insights into the complex biochemistry and cellular physiology of the human eye.

A 22-year-old female who presented with headaches. She gave no history of ophthalmic or systemic disease. On examination her visual acuities were (RE) 6/6 and (LE) 6/6(p) corrected to 6/6 with -0.50 DC 140 in Left eye. The anterior segments of both eyes were normal, and the intraocular pressures were within normal limits. Central and peripheral visual fields did not show any abnormalities, and dark adaptometry was normal. Fundoscopy showed that in both eyes the fundi were invaded by an enormous number of bright white and sometimes yellowish white flecks of variable size and configuration, arranged in a concentric pattern around the posterior fundus and sparing the optic disc, macula, papillomacular area as well as 1-2 disc diameters circular region surrounding the disc and macula. They spread everywhere in the equator and mid and extreme periphery of fundus. The more centrally located flecks appeared sparse, small, round, and dot-like. Others of larger size in the equatorial and peripheral areas

varied in shape, being, round, elongate, pisciform, star-shaped, and sometimes circular. The flecks were always discrete, well defined, with almost sinuous margins and a flat or prominent surface. They appeared to be solid and well behind the retinal vascular tree. Throughout the fundus they showed the same mosaic appearance. They spared no area in the periphery. No pigmentary disturbances, calcification or conglomeration was observed, nor were choroidal vessels observed.

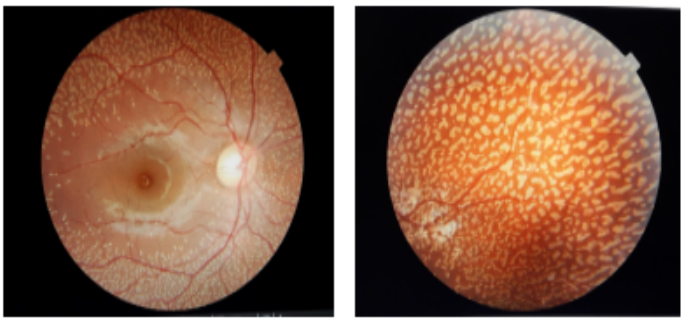

FIGURE 1: Righteye fundus

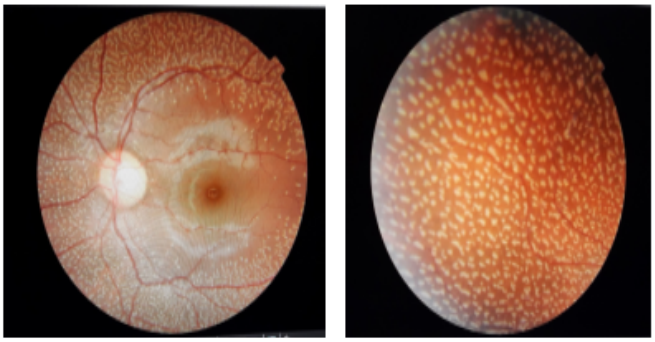

FIGURE 2: Left eye fundus

[Fluorescein angiography carried out at this time showed that apart from the macula the whole fundus was hyperfluorescent, including the central areas which appeared free of flecks on fundoscopy and white light photography. This indicated there was no close correlation between the site of the flecks and the hyperfluorescent areas, particularly in the central part of the fundus]

Supplementary information The online version of this article (https://doi.org/10.15520/jcmro.v3i10.354) contains supplementary material, which is available to authorized users.

Corresponding Author: *Rohini Sangoram*
Senior Resident, Department of Ophthalmology, Krishna Institute of Medical Sciences, Karad, Maharashtra, INDIA.
Email: drrohs199@gmail.com

2 | DISCUSSION:

Benign familial fleck retina is inherited as an autosomal recessive condition. The term ”flecked retina” was introduced by Krill and Klien to describe fundus conditions characterized by multiple yellowish-white lesions of various size and configuration, without vascular or optic nerve abnormalities. Originally this group consisted of four diseases: fundus albipunctatus, fundus flavimaculatus, familial drusen and fleck retina of Kandori. However, far more diseases correspond to the rather vague definition, including primary hereditary ocular diseases such as retinitis punctata albescens or Bietti’s crystalline dystrophy, neuro-ophthalmologic syndromes such as Kjellin’s syndrome, secundary retinal flecks due to metabolic disorders such as Alport’s syndrome, cystinosis, oxalosis or membrano-proliferative glomerulonephritis, iatrogenic retinopathy due to Tamoxifen or Canthaxanthin and carential diseases such as vitamin A deficiency. The precise diagnosis is not only based on the close observation of the fundus, but demands often extensive psychophysical examination of the patient and sometimes of his family.

Massive mosaic hyaline excrescences along the cuticular layer of Bruch’s membrane, leading to the appearance of multiple deep yellow to yellowish white lesions of variable size and shape in the fundi have been well known for more than a century. (1–3) Many forms of this condition have been described with different findings, prognosis, and inheritance (4–15) . Kril (16, 17) regarded all these conditions as fleck retina diseases. He then classified them into 4 groups: fundus albipunctatus, fundus flavimaculatus, familial drusens, and fleck retina of Kandori. This classification with subgrouping according to the severity of the condition seemed to bring order into the confusion that had lasted for a century. In this paper we present a family with flecked retina, with typical fundoscopy picture and normal visual findings. We suggest they belong to a fifth group benign familial fleck retina.

Clinically there was no night blindness or any delay in dark adaptation.

FAF reveals multiple hyperautofluorescent lesions corresponding in location with the flecks. On SD-

OCT, discrete deposit accumulation located posterior to the photoreceptor inner/outer segment junction but without disrupting it is observed. Normal full-field and pattern ERGs confirm the diagnosis; (15) multifocal ERG may be slightly subnormal in some isopters. (16)

Recessive mutations in the gene encoding group V phospholipase A_2 (*PLA2G5*) and mildly elevated low-density lipoprotein and total cholesterol levels have been identified in some individuals with benign fleck retina. (17)

Benign fleck retina should not be confused with "fleck retina of Kandori." The latter is associated with large white lesions, possibly atrophic changes, and night-blindness. (18) It is not clear if "fleck retina of Kandori" is a genetic condition or even an independent clinical entity.

The main features of the different types of flecked retina disorders reported in the literature are summarised below Table 1

Benign familial fleck retina is inherited as an autosomal recessive condition. This is supported by the fact that both sexes were involved and that both parents were free of the disease.

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TABLE 1: different types of flecked retina disordersreported

FLECK RETINA DISEASE	CHANGES IN FUNDI	FUNCTIONAL DISTURBANCES	MODE OF INHERITANCE
Fundus albipunctatus	Discrete uniform white dots. Night blindness. Dark Autosomal recessive Distribution: over the whole adaptation: normal	Slow or Autosomal dominant23 fundus greatest density at monofunctional (only cone midperiphery. No macular threshold) involvement. No pigmentary disturbances or secondary calcification	Discrete uniform white dots. Night blindness. Dark Autosomal recessive Distribution: over the whole adaptation: normal
Fundus flavimaculatus	Round, linear, or pisciform lesions. Distribution: limite to the posterior pole, or extends to the equator. Macula is involved. Network atrophy of retinal pigment epithelium. Choroidal vascular atrophy	Central visual loss, colour vision loss, photophobia, paracentral scotoma, slow dark adaptation	Autosomal recessive
Familial drusen"	Round or oval lesions in almost grape-like clusters. Distribution: concentrated in the posterior polar region. Pigmentary disturbances and secondary calcifications. Macula is almost always involved, may appear oedematous or haemorrhagic	Loss of vision during progressive stages, central scotoma slow dark adaptation	Autosomal dominant
Fleck retina of Kandori	Irregular flecks with great variability in size. Distribution: in the equatorial or between the equatorial and macular region with tendency for confluence. No macular lesions. Disturbance of pigment epithelium	Some night blindness. Initially delayed dark adaptation, recovers to normal value after 30-40 minutes in the dark	Autosomal recessive
Benign familial fleck retina	Round, linear, or pisciform. Distribution: in the whole fundus except the disc and macula. No macular lesions. No tendency for confluence. No pigmentary disturbances	No disturbance of visual function (no symptoms)	Autosomal recessive

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