A rare case of benign flecked retina - A case report

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Abstract
The term Familial Dominant Drusen is used to describe a group of visual conditions in which radiating drusens and macular degeneration occurs in young patients. Drusens are described as yellowish deposits that lie between basement membrane of Retinal Pigment Epithelium and inner collagenous zone of Bruch's membrane. They consist of neutral fat, phospholipids, glycoconjugates, fibrogen, vitronectin, factor x, apolipoprotein E, thrombospondin, 7ketocholestrol etc. Clinically they are classified as hard drusen, granular soft drusen, membranous soft drusen, reticular drusens and cuticular variant. Drusens commonly present in age group of 20 to 30 years. We describe a case of young male aged 35 years presenting with chief complaints of diminution of vision in both eyes, insidious in onset, gradually progressive since childhood, worsened since last one year and more during night time. On evaluation of family history, mother was found to have similar complaints. Fundus examination revealed numerous yellowish white polymorphous lesions extending from mid periphery to peripheral retina.

Key words: Familial Dominant Drusen, radiating drusen, Benign flecked retina syndrome.

Introduction
Drusens are subretinal pigment deposits. The term Familial Dominant Drusen describes a group of visual conditions in which radiating drusens and macular degeneration occurs in young patients. The term flecked retina was introduced by Krill and Klien to describe fundus condition characterised by multiple yellowish-white lesions of various size and configurations sparing the optic nerve head and background retinal vasculature. The inheritance has been reported to be a mutant gene, FBLN1, which is also called by name EFEMP1 (epidermal growth factor containing fibrillin like extracellular matrix protein) on 6q14. This gene is responsible for the protein fibulin 3, mutant of that protein causes proteoglycan accumulation, reduction of Matrix metalloprotease -2 and Matrix metalloprotease -9, but increase of tissue inhibitor of metalloprotease -3, and is also known to enhance diffusion across Bruch's membrane resulting in deposition of drusens.[¹²]

The pathognomonic feature of Familial Dominant Drusen is presence of yellowish-white deposits which are thought to be metabolic breakdown deposits from retinal pigment epithelium within the inner collagenous layer of bruch's membrane.[³]

Case report
A 35 year old male presented with chief complaints of diminution of vision in both eyes, insidious in onset, gradually progressive since childhood increased since last one year, more during night time. He had no specific medical history, On evaluation of family history, mother (first degree relative) was found to have
similar complaints. Visual acuity in right eye was 6/18 on Snellen's visual acuity chart, improving to 6/6 with -1.50 Dioptre sphere and in left eye it was 6/36 improving to 6/6 with -1.75 Dioptre Sphere. Fundus examination with 20 Dioptre lens on indirect Ophthalmoscopy revealed numerous yellowish white polymorphous lesions which were of varied size and shape extending from the posterior pole to the Ora Serrata. The lesions increased in size and also appeared to be merging into each other from the posterior pole towards the periphery. (Figure1 & 2)

On Evaluation with 90 Dioptre lens on slit lamp biomicroscopy the lesion size appeared to be greater than a venular breadth. The interspaced normal retina was lesser than a venular breadth. Since the vascular architecture was completely seen, the lesions seems to be arising at the deeper layers of the retina. These lesions were non refractile in appearance, suggesting the origin to be sub-retinal pigment epithelium level. This was confirmed by interposing a red free filter on slit lamp biomicroscopy. The appearance qualified them to be classified as a cuticular variant of Drusen.

![Figure 1 and 2](image)

**Figure 1 and 2**: Drusens present in radiating fashion from mid periphery to peripheral retina.

Fluorescein angiography with 2% Fluorescein Sodium revealed multiple sharply defined hyper fluorescent lesions, showing window defect in choroidal phase which doesn't increase in either size or intensity in further phases of Fluorescein angiography. Larger drusens showed auto fluorescence. (Figure 3)

![Figure 3](image)

**Figure 3**: Autofluorescence of the drusens

Optical coherence tomography shows thickening of retinal pigment epithelium-bruch's membrane complex with preservation of the neurosensory retina. (Figure 4)

![Figure 4](image)

**Figure 4**: Optical coherence tomography showing thickening of Retinal pigment epithelium-Bruch's membrane complex. The retinal pigment epithelium complex showed Irregularities.

Electroretinogram: showed reduction in amplitude of b-wave. [3] Electroocculogram was found to be within normal limits. [4]

**Discussion**

Familial Dominant Drusen, also known as Malattia Leventinese, was first described in patients living in Leventine valley in Canton Ticino of Southern
Switzerland. Emma E Tarttelin et al, concluded that, it is a Autosomal dominant disease with mutation of EFEMP, on 6q14 is a rare presentation of drusen cases as against EFEMP promotor sequence mutation located on 2p16 which is responsible for manifestation of drusen in majority of the cases. It's a autosomal dominant type with variable penetrance, with radiating confluent drusens extending from posterior pole to periphery. This is said to be the most pathognomonic finding of Familial Dominant Drusens.

Kasmann B et al. concluded that Familial dominant drusen shows a complete penetrance and variable expression. It's a metabolic defect of Retinal pigment epithelium with accumulation of metabolic breakdown products from retina within inner collagen layer of Bruch's membrane.\(^{(1)}\)

Elliott H Sohn et al concluded that morphological constituents of drusens present in Familial Dominant Drusen share similar phenotype with the deposits seen in age related macular degeneration, except for the presence of collagen type IV as in drusens of Familial Dominant Drusen.\(^{(6)}\)

No treatment is required, as it does not involve the posterior pole, neither it is known to cause peripheral visual constriction.

**Conclusion**

On the basis of above finding we conclude the case to have familial dominant drusen, which is a variant of benign flecked retina syndrome, which is non vision threatening and non progressive in nature.

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**Reference**


