Ophthalmology Research: An International Journal

Volume 19, Issue 1, Page 26-30, 2024; Article no.OR.112406
ISSN: 2321-7227

Butterfly-shaped Pattern Dystrophy : Findings of Retinal Imaging

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

Article Information

DOI: 10.9734/OR/2024/v19i1411

Open Peer Review History:
This journal follows the Advanced Open Peer Review policy. Identity of the Reviewers, Editor(s) and additional Reviewers, peer review comments, different versions of the manuscript, comments of the editors, etc are available here: https://www.sdiarticle5.com/review-history/112406

Case Study

Received: 25/11/2023
Accepted: 30/01/2024
Published: 12/02/2024

ABSTRACT

Butterfly shaped pattern dystophy (BPD) is a inherited macular disease which is characterized by the accumulation of pigment/lipofuscin in the retinal pigment epithelium, it might be misdiagnosed as age-related macular degeneration (AMD). Retinal imaging is a useful tool for the differential diagnosis of pattern dystrophy. In this report, we describe a 64 year old man presented metamorphopsia and reduced visual acuity in both eyes. Fundus examination showed an area of depigmentation delimited resembling a butterfly. The OCT revealed a subfoveal hyperreflective deposit above the retinal pigment epithelium (RPE) while fundus autofluorescence (FAF) shows hypoautofluorescent areas outlined by a lipofuscin deposits as hyperautofluorescent Finally, fluoroscein angiography (FFA) revealed early macular hyperfluorescence.

Keywords: Butterfly shaped pattern dystophy; macular dytrophy; retinal imaging; AMD.

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1. INTRODUCTION

Butterfly shaped pattern dystophy (BPD) is a inherited dominant macular disease which is characterized by the accumulation of pigment/lipofuscin in the retinal pigment epithelium [1].

Patients generally tend to report a decline in visual acuity around the fifth decade of life [2].

The central lesion is well highlighted by retinal imaging, which distinguishes this condition from other macular dystrophies [3].

Pattern dystrophies (PD) might be misdiagnosed as age-related macular degeneration (AMD). Both Optical Coherence Tomography (OCT) and fundus autofluorescence (FAF) are useful tools for the differential diagnosis of PD.

2. CASE PRESENTATION

A 64 - year - old man presented to our department complaining of metamorphopsia and reduced visual acuity in both eyes, which has progressively deteriorated over the past year. His past ocular history or family history were unremarkable.

On the initial examination, best corrected visual acuity (BCVA) was 04/10 in the right eye (OD) and 05/10 in the left eye (OS).

Slit lamp biomicroscopy revealed a normal anterior segment . Intraocular pressure (IOP) was 14 mmHg in both eyes.

Fundus examination showed an area of depigmentation delimited by a deposit of yellowish pigment resembling a butterfly. (Fig. 1a, b).

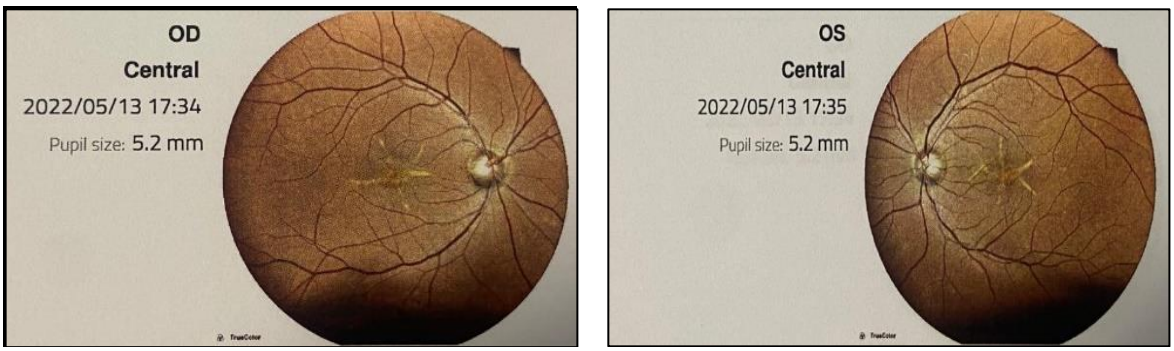

Fig. 1(a,b). Color fundus photograph of the right (a) and the left eye (b) showing butterfly pattern dystrophy. The yellowish white lipofuscin deposits are seen radiating in the form of ‘‘wings’’ from the central lesion

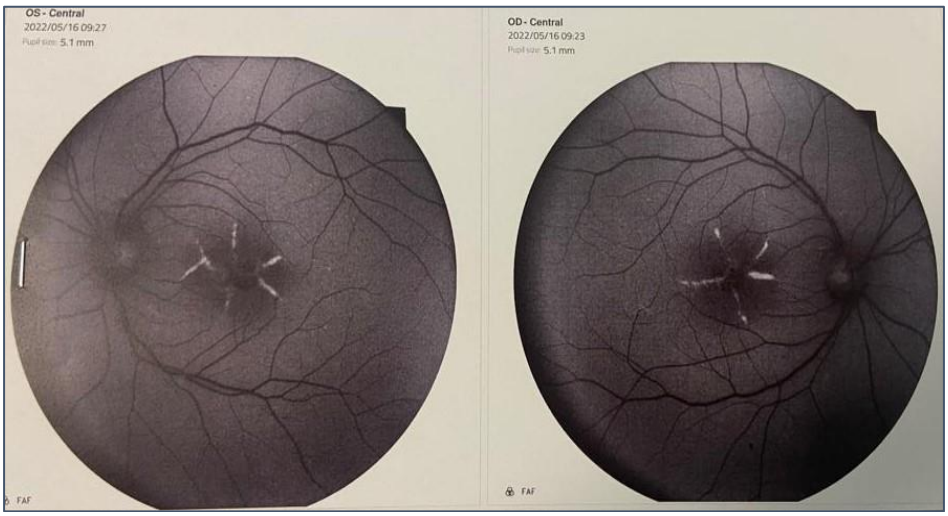

Fig. 2. Fundus autofluorescence of both the eyes shows hypoautofluorescent areas outlined by a lipofuscin deposits as hyperautofluorescent

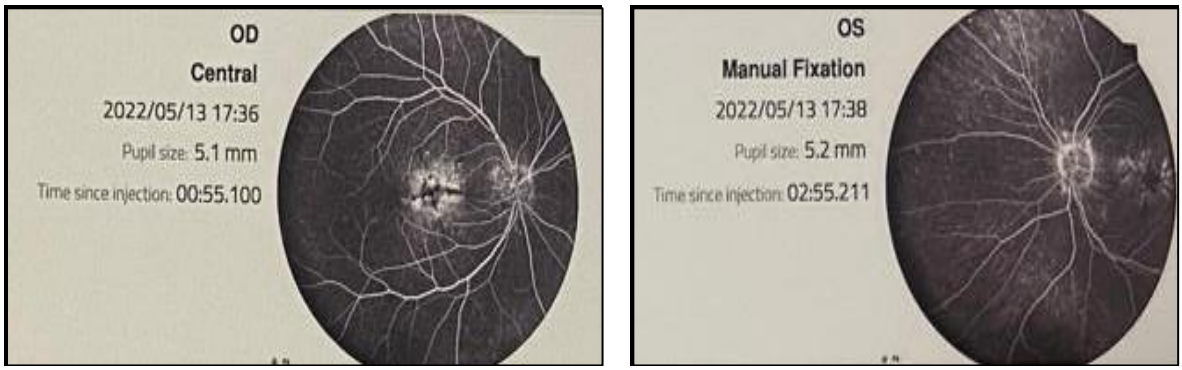

Fig. 3(a,b). Fluorescein angiograms of the right and the left eyes reveal an early macular hyperfluorescence without diffusion

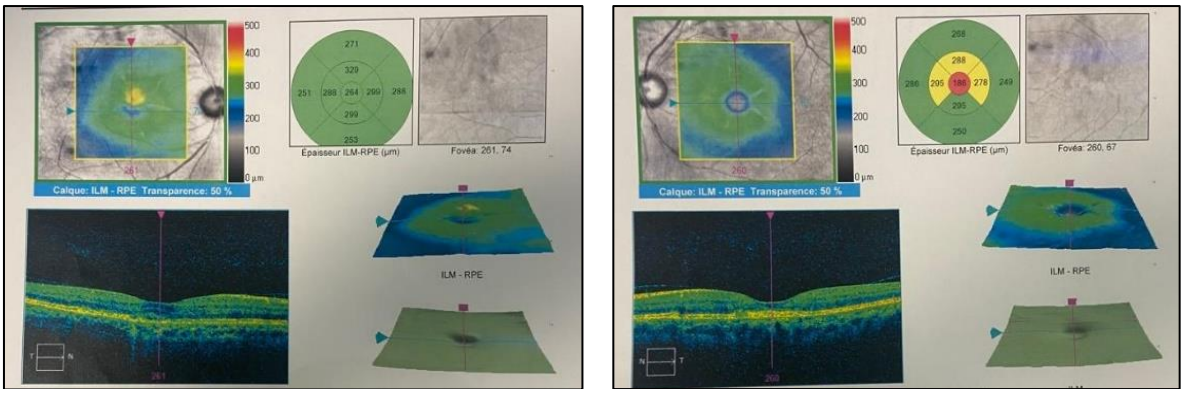

Fig. 4. Macular OCT (a,b) : Subfoveal hyperreflective deposit above the level of the RPE

The retinal vasculature appeared normal.

The macular Optical Coherence Tomography (OCT) revealed a subfoveal hyperreflective deposit above the level of the retinal pigment epithelium (RPE) (Fig. 4 a,b) while fundus autofluorescence (FAF) shows hypoautofluorescent areas outlined by a lipofuscin deposits as hyperautofluorescent (Fig.2).

Finally, fluoroscein angiography (FFA) revealed early macular hyperfluorescence without diffusion (Fig. 3 a,b).

The patient was evaluated every 6 months in our department for possible deterioration. His BCVA stayed stable for the next months , with no significant change in OCT findings.

3. DISCUSSION

"Butterfly shaped pattern dystrophy (BPD) is a dominantly inherited macular disease characterized by an accumulation of pigment/lipofuscin in the RPE due to photoreceptor degeneration" [4].

It was first described by Deutman et al [5], "in a white family who had a peculiar bilateral butterfly pigmentation in the macular region at the level of the RPE".

"Previous studies , have shown that mutations in the RDS /peripherin gene is the causative factor, which is located on chromosome 6p21.2. This gene encodes a maintenance glycoprotein of photoreceptor segmentation discs. When disrupted, it interferes with the integrity of the photoreceptor membrane" [3,6–8].

Recently, Saksens et al [9] implicated "mutations in the CTNNA1 gene as a cause of butterfly pigment dystrophy. Involvement of CTNNA1, a central component of adhesion junctions, suggests that components of the cadherin-based intercellular adhesion mechanism may also be involved in causing macular degenerative diseases such as BPD" [9].

"Patients are usually asymptomatic when diagnosed with borderline disorder in their

second or third decade and maintain relatively normal visual acuity for most of their lives. However, the disease can progress with age, and older individuals may have atrophic, depigmented lesions extending into the peripapillary region with markedly reduced visual acuity" [10].

"In butterfly shapped dystrophy, the central lesion is easily demonstrated by FA, which differentiates this condition from other macular dystrophies. FA usually shows a large, hypofluorescent, butterfly-shaped macular lesion. Yellow spots seen in the fundus in the posterior pole block fluorescence" [1,11].

"Fundus autofluorescence may show increased or decreased autofluorescence, corresponding to changes in RPE lipofuscin in the lesion" [4].

4. CONCLUSION

Pattern dystrophies are considered as heterogeneous group of retinal disorders that are characterised by a bilateral symmetric visuel loss [11]. Because PD usually manifests in later life, it may be misdiagnosed as AMD [12]. Retinal imaging, including FAF, OCT, is useful in differentiating PD from AMD to ensure the perfect management [13].

CONSENT

As per international standards or university standards, patient(s) written consent has been collected and preserved by the author(s).

ETHICAL APPROVAL

As per international standards or university standards written ethical approval has been collected and preserved by the author(s).

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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Peer-review history:
The peer review history for this paper can be accessed here:
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