

Butterfly-shaped pattern dystrophy : Findings of retinal imaging

Abstract :

Butterfly shaped pattern dystrophy (BPD) is an 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 deposit as hyperautofluorescent. Finally, fluorescein angiography (FFA) revealed early macular hyperfluorescence.

Key Words : Butterfly shaped pattern dystrophy, macular dystrophy, retinal imaging, AMD

Introduction :

Butterfly shaped pattern dystrophy (BPD) is an 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.

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).

Slitlamp 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).

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. 4a, b) while fundus autofluorescence (FAF) shows hypoautofluorescent areas outlined by lipofuscin deposits as hyperautofluorescent (Fig. 2).

Finally, fluorescein angiography (FA) revealed early macular hyperfluorescence without diffusion (Fig. 3a, 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.

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 shaped 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]

Conclusion :

Pattern dystrophies are considered as a heterogeneous group of retinal disorders that are characterised by a bilateral symmetric visual 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]

Ethical Approval:

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

Consent

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

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Images :

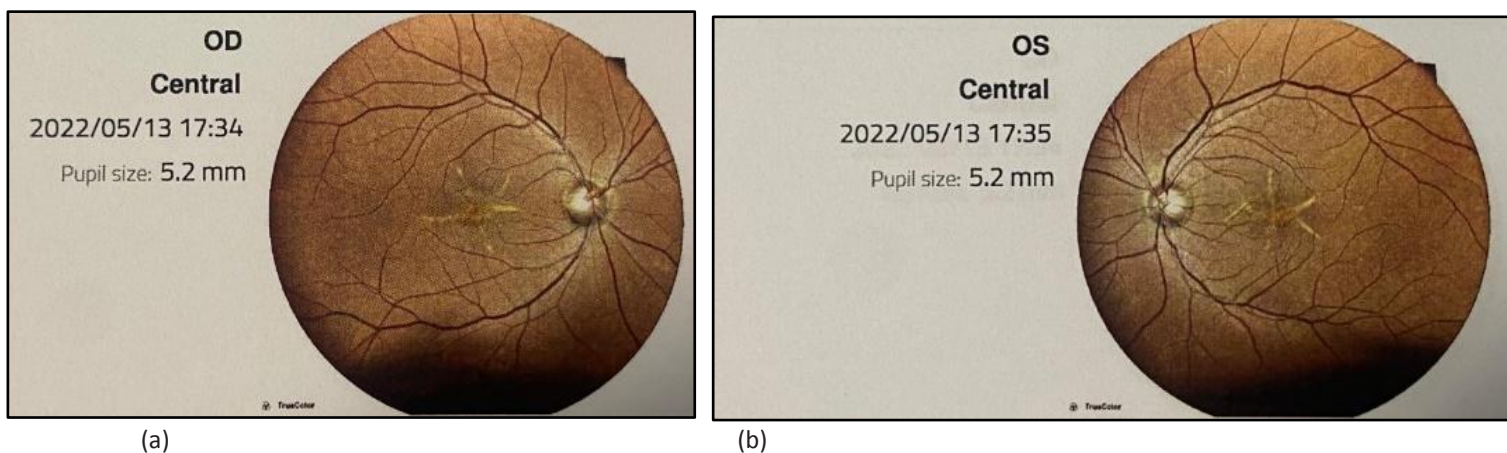


Figure 1 (a, b) :Color fundusphotograph of the right(a) and the lefteye(b) showingbutterfly pattern dystrophy. The yellowishwhite lipofuscin deposits are seen radiating in the form of “wings” from the centrallesion.

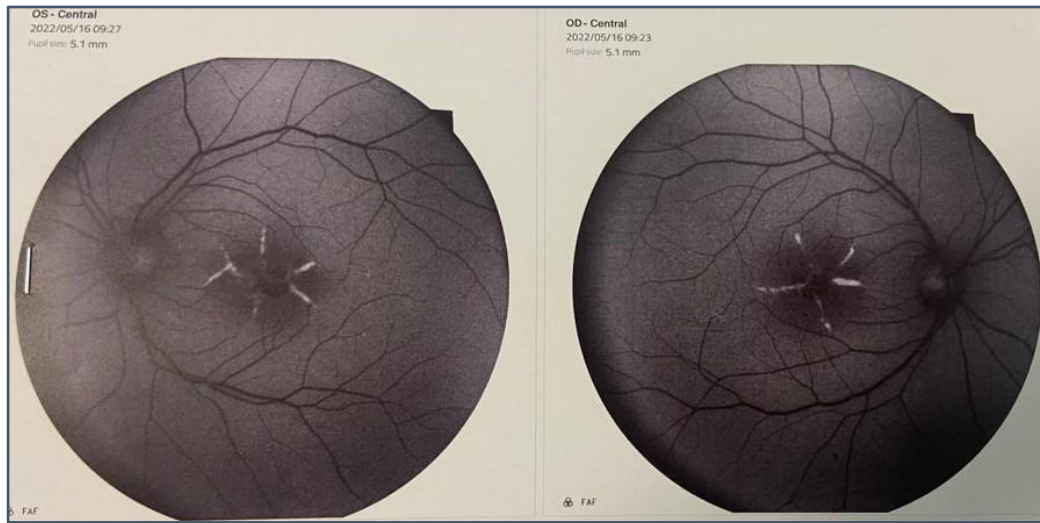
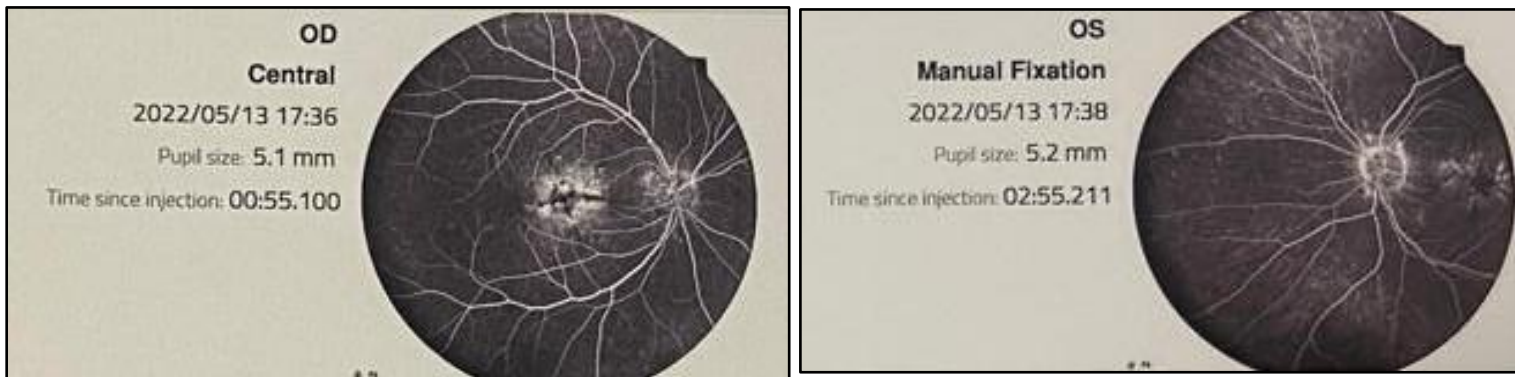


Figure 2 : Fundus autofluorescence of both the eyes shows hypoautofluorescent areas outlined by alipofuscin deposits as hyperautofluorescent



(a)

(b)

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

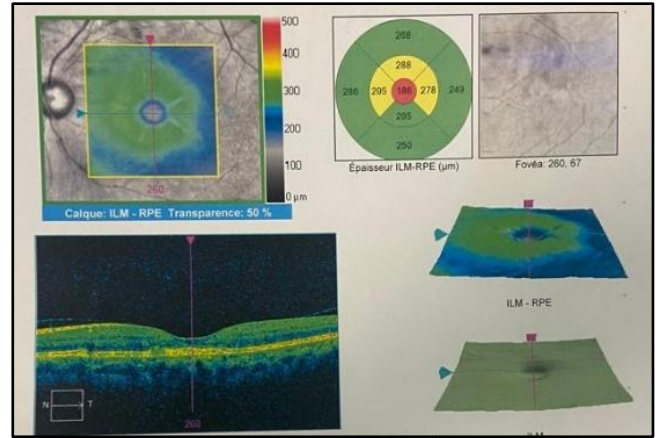
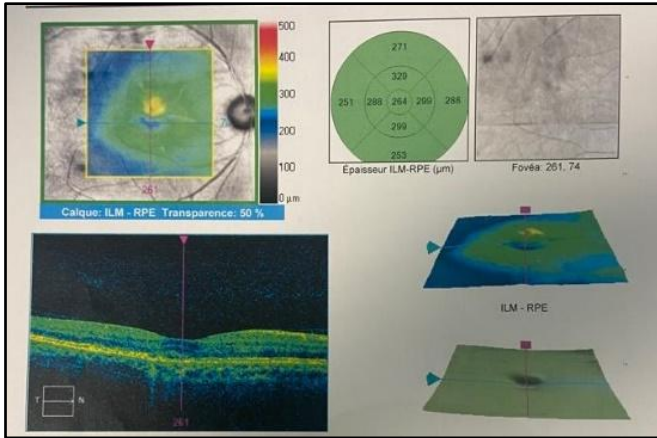


Figure 4 :Macular OCT

(a, b) : Subfoveal hyperreflective deposit above the level of the RPE