Butterfly-shaped pattern dystrophy : Findings of retinalimaging

Abstract :

Butterfly shaped pattern dystophy (BPD) is a inheritedmaculardiseasewhichischaracterized by the accumulation of pigment/lipofuscin in the retinal pigment epithelium ,itmightbemisdiagnosed as age-relatedmaculardegeneration (AMD). Retinalimagingis a usefultool for the differentialdiagnosis of pattern dystrophy .

In thisreport ,wedescribe a 64 yearold man presentedmetamorphopsia and reducedvisualacuity in botheyes. Fundus examinationshowed an area of depigmentationdelimitedresembling a butterflyThe OCT revealed a subfovealhyperreflectivedepositabove the retinal pigment epithelium (RPE) while fundus autofluorescence (FAF) shows hypoautofluorescent areas outlined by a lipofuscindeposits as hyperautofluorescentFinally, fluorosceinangiography (FFA) revealedearlymacularhyperfluorescence

KeyWords : Butterfly shaped pattern dystophy, maculardytrophy ,retinalimaging ,AMD

Introduction :

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

Patients generally tend to report a decline in visualacuityaround the fifthdecade of life.[2]

The central lesioniswellhighlighted by retinalimaging, whichdistinguishesthis condition fromothermacular dystrophies. [3]

Pattern dystrophies (PD) mightbemisdiagnosed as age-relatedmaculardegeneration (AMD). BothOptical CoherenceTomography (OCT) andfundus autofluorescence (FAF) are usefultools for the differentialdiagnosis of PD

Case presentation :

A 64 - year -oldman presented to ourdepartmentcomplaining of metamorphopsia and reducedvisualacuity in botheyes, which has progressivelydeteriorated over the pastyear. Hispastocularhistory or familyhistorywereunremarkable.

On the initial examination, best correctedvisualacuity (BCVA) was 04/10 in the right eye(OD) and 05/10 in the lefteye (OS).

Slitlampbiomicroscopyrevealeda normalanteriorsegment .Intraocular pressure (IOP) was 14 mmHg in botheyes .

Fundus examinationshowed an area of depigmentationdelimited by a deposit of yellowish pigment resembling a butterfly.. (Fig. 1a, b).

The retinalvasculatureappeared normal.

The macular Optical CoherenceTomography (OCT) revealed a subfovealhyperreflectivedepositabove the level of the retinal pigment epithelium (RPE) (Fig. 4a, b) whilefundus autofluorescence (FAF) shows hypoautofluorescent areas outlined by alipofuscindeposits as hyperautofluorescent (Fig.2).

Finally, fluorosceinangiography (FFA)revealedearlymacularhyperfluorescencewithout diffusion (Fig. 3a,b).

The patient wasevaluatedevery 6 months in ourdepartment for possible deterioration. His BCVA stayed stable for the nextmonths,with no significant change in OCT findings.

Discussion :

Butterfly shaped pattern dystrophy (BPD) is a dominantlyinheritedmaculardiseasecharacterized by an accumulation of pigment/lipofuscin in the RPE due to photoreceptordegeneration[4].

It was first described by Deutman et al[5] , in a white familywhohad a peculiarbilateralbutterfly pigmentation in the macularregion at the level of the RPE .

Previousstudies,have shownthat mutations in the RDS /peripheringeneis the causative factor, whichislocated on chromosome 6p21.2. This gene encodes a maintenance glycoprotein of photoreceptor segmentation discs. Whendisrupted, itinterfereswith the integrity of the photoreceptor membrane [3,6–8] .

Recently, Saksenset al [9]implicated mutations in the CTNNA1 gene as a cause of butterfly pigment dystrophy. Involvement of CTNNA1, a central component of adhesionjunctions, suggeststhat components of the cadherin-basedintercellularadhesionmechanismmayalsobeinvolved in causingmaculardegenerativediseasessuch as BPD [9]

Patients are usuallyasymptomaticwhendiagnosedwith borderline disorder in their second or thirddecade and maintainrelatively normal visualacuity for most of theirlives. However, the disease can progresswithage, and olderindividualsmay have atrophic,

depigmentedlesionsextendinginto the peripapillaryregionwithmarkedlyreducedvisualacuity. [10]

In butterflyshappeddystrophy, the central lesioniseasilydemonstrated by FA, whichdifferentiatesthis condition fromothermacular dystrophies. FA usually shows a large, hypofluorescent, butterfly-shapedmacularlesion. Yellow spots seen in the fundus in the posterior pole block fluorescence. [1,11]

Fundus autofluorescencemay show increased or decreasedautofluorescence, corresponding to changes in RPE lipofuscin in the lesion[4]

Conclusion :

Pattern dystrophies are consideredas heterogeneous group of retinaldisordersthat are characterised by a bilateralsymmetric visuel loss[11]

Because PD usuallymanifests in later life, itmaybemisdiagnosed as AMD [12]

Retinalimaging, including FAF, OCT, isuseful in differentiating PD from AMD to ensurethe perfectmanagement.[13]

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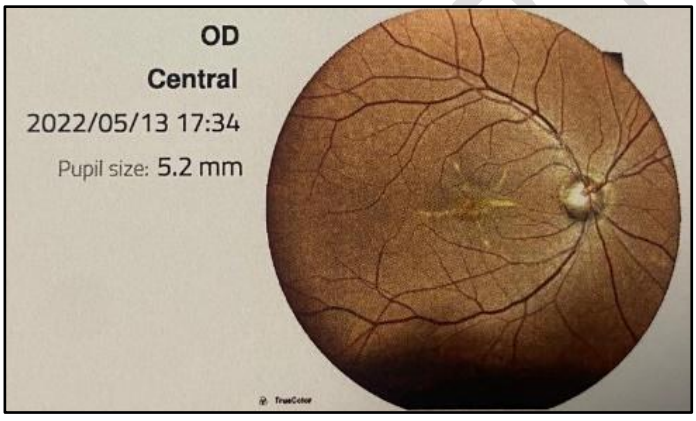
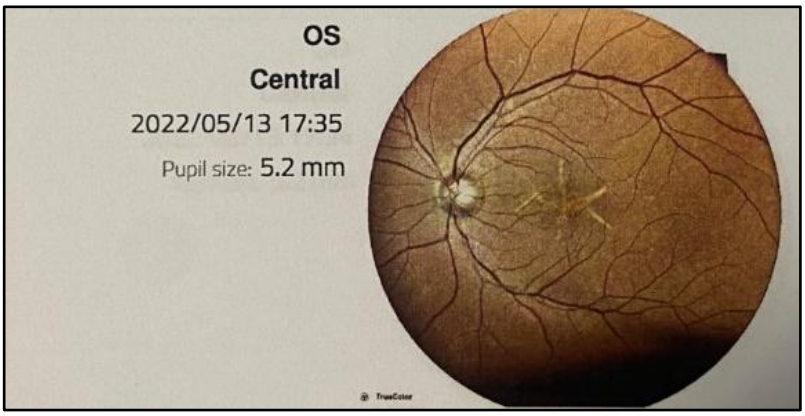
Images :



(a)



(b)

Figure 1 (a, b) :Color fundusphotograph of the right(a) and the lefteye(b) showingbutterfly patterndystrophy. The yellowishwhite lipofuscindeposits areseenradiating in the form of‘‘wings’’ from the centrallesion.

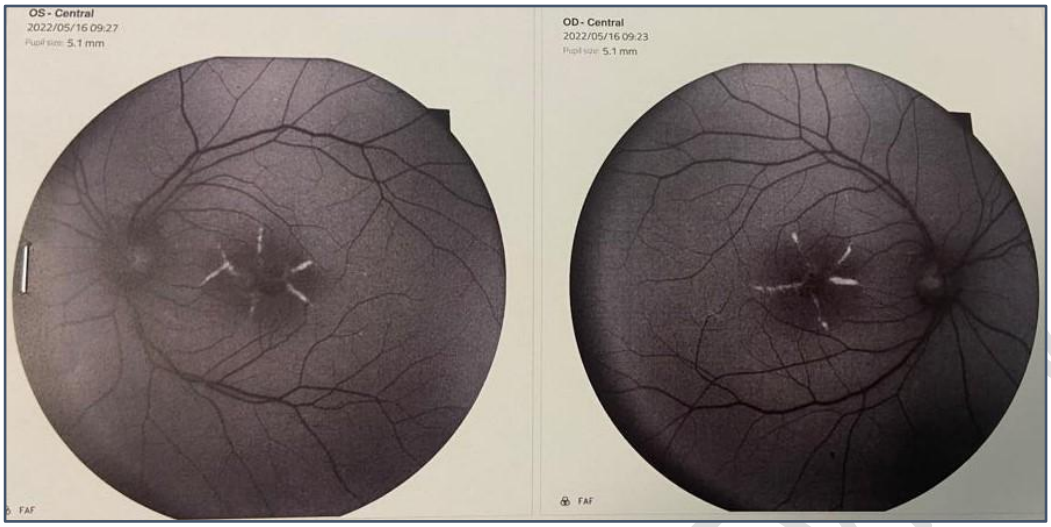
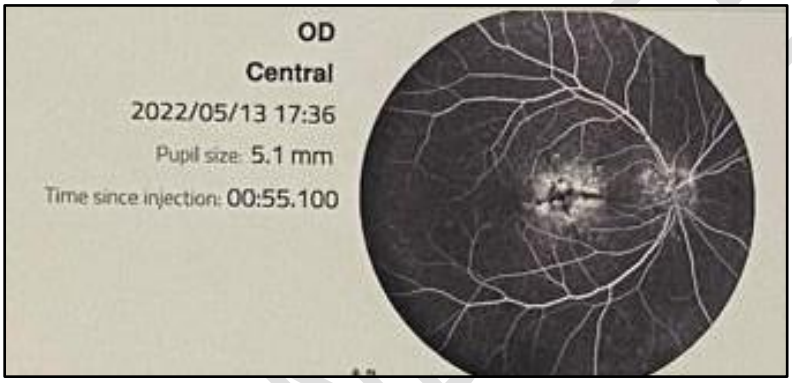
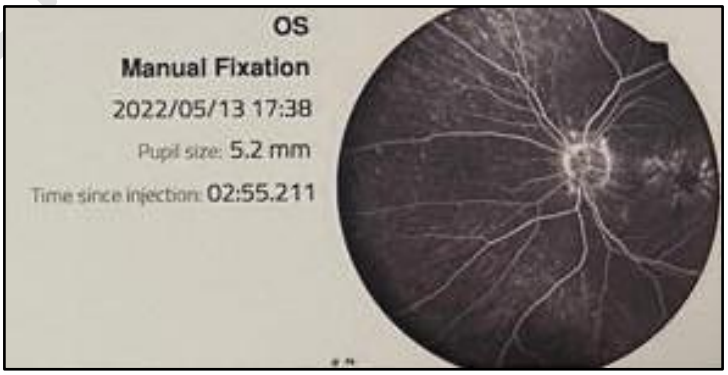


Figure 2 : Fundusautofluorescence of both theeyesshows hypoautofluorescent areas outlined by alipofuscindeposits as hyperautofluorescent



. (a)



(b)

Figure 3 (a,b) : Fluoresceinangiograms of the right and the lefteyesreveal an earlymacularhyperfluorescencewithout diffusion

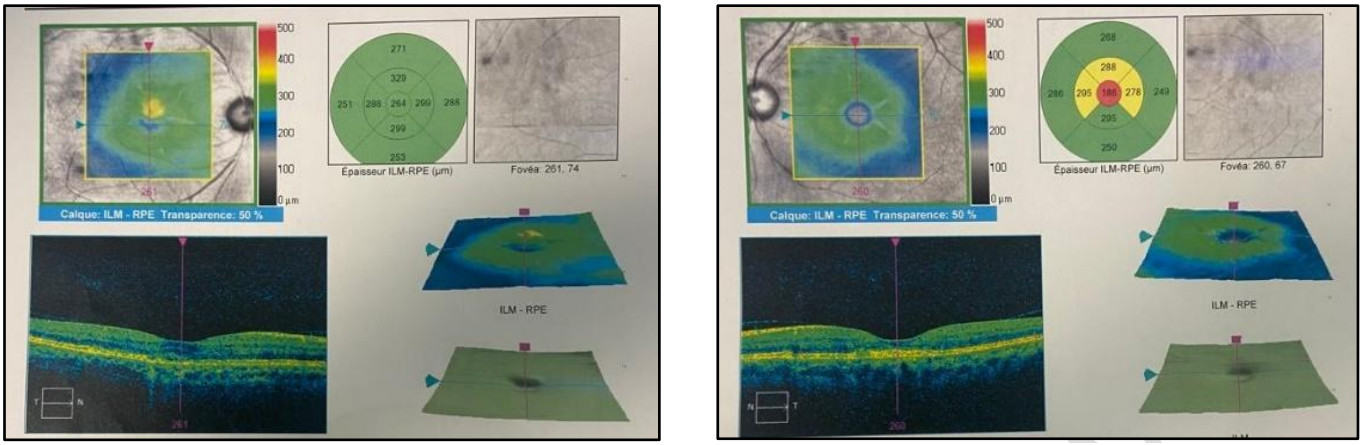


Figure 4 :Macular OCT

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