



Unilateral Best macular dystrophy: a case report

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INTRODUCTION

Best disease (Vitelliform Macular Dystrophy - VMD) is an early onset macular dystrophy typically characterized by bilateral accumulation of subretinal deposit resulting from heterozygous mutations in the BEST1 gene (OMIM 153700)¹. It has two clinical variants: Best's (VMD2) and adult onset vitelliform macular dystrophy (AOVMD). Both are characterised by classic "egg-yolk" lesions seen in the macula, and affected eyes may demonstrate various clinical stages, ranging from the previtelliform stage to choroidal neovascularization^{2,3}. Visual function generally remains good despite ophthalmoscopically visible lesion until the disease process progresses to cause structural alterations in the outer retinal layers³.

METHODS

Medical records review.

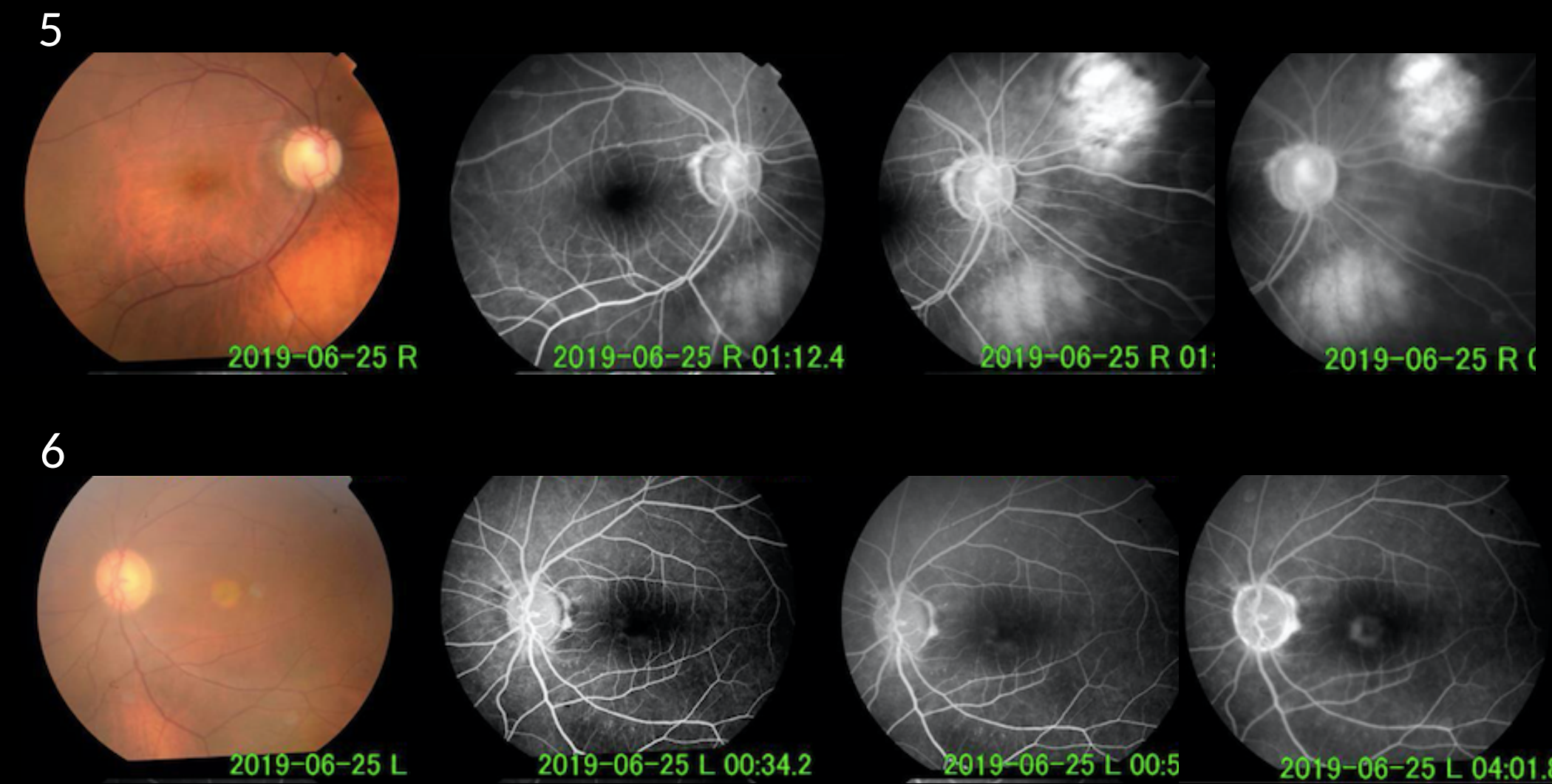
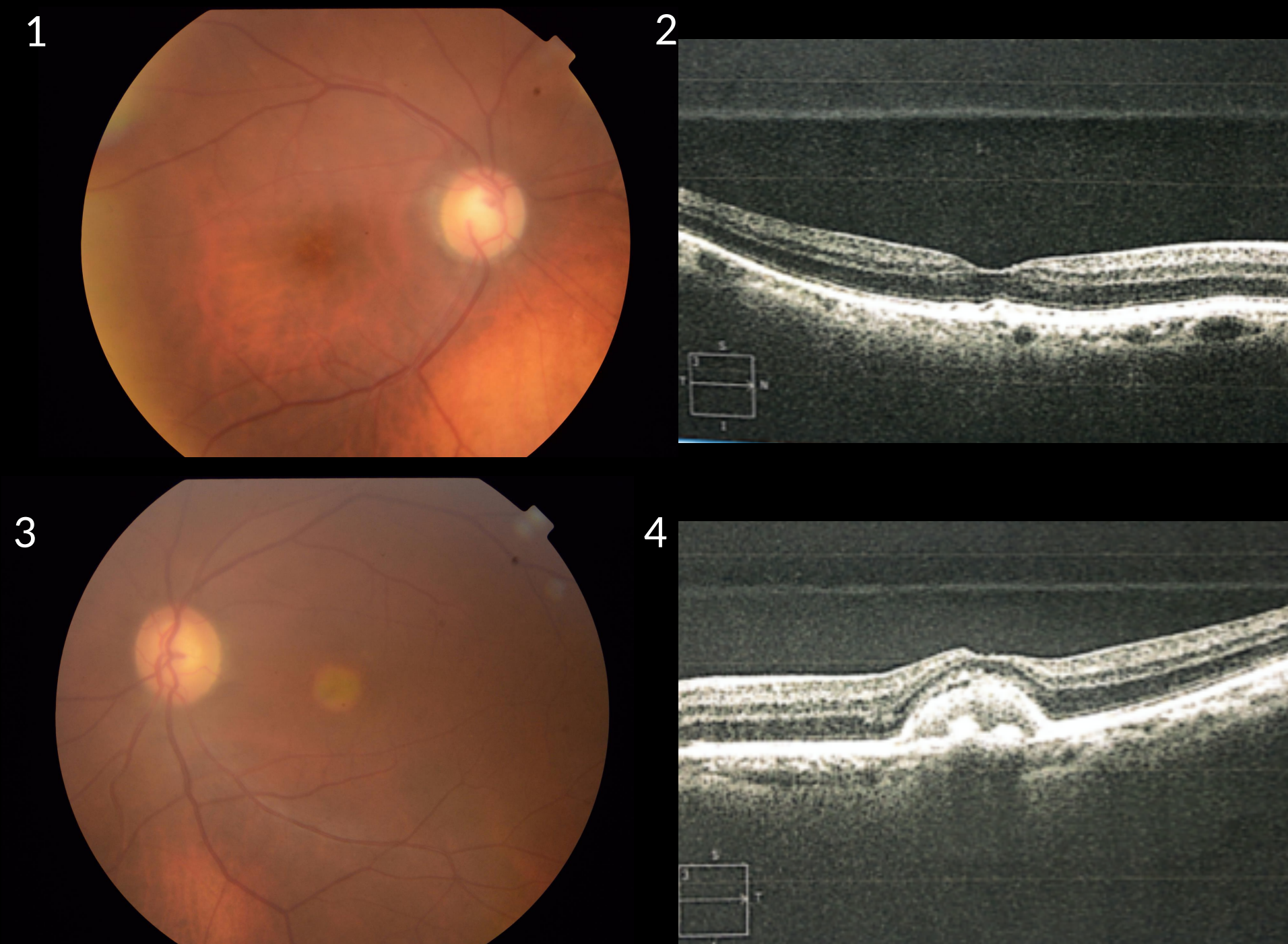
RESULTS

We present a case of an 80-year-old patient who complains of low visual acuity in both eyes (20/60) for the past 3 months. The patient had cataract surgery in OD 12 years ago and OE 10 years ago. On funduscopy examination has in the right eye, an optic disc with 0.8x0.8 excavation, decreased macula brightness, exudative upper nasal injury, EPR rarefaction, vascular thinning, applied retina. In the left eye, it presents an optic disc with a 0.8x0.7 excavation, a well-defined yellow circular lesion in the macular area, applied retinal and vascular thinning.

DISCUSSION

This case presents typical and consistent findings from Best except for the unilateral presentation. The OCT shows an accumulation of subretinal hyperreflective material. The oculogram shows a reduction in the values bilaterally. According to the literature, normal appearance in the eye without a fundus finding may precede a clinical manifestation of vitelliform lesions.

IMAGES



Figures 1 and 2: Shows normal exams; Figures 3: Well-defined yellow lesion in the macular area in left eye; Figures 4: OCT showing an accumulation of subretinal hyperreflective material in the left eye. Figures 5 and 6: Fluorescein angiography of the OD was normal, with an area of alteration in the perifoveal capillary ramification. In the OS showed a slightly hypofluorescent area in the region corresponding to the lesion and permeability of juxtafoveolar capillaries, which became more noticeable during the exam, creating a final scenario of diffuse macular edema with no cystoids characteristics

REFERENCES

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2. Talia R, Kaden, MD, Anna C. S. Tan, MD, Leonard Feiner, MD, PhD, J. K. Bailey Freund, MD. UNILATERAL BEST DISEASE: A CASE REPORT
3. Pradnya Kamat, Pratik Doshi, Manasi Prabhudesai, Srishti Prabhudesai Department of Ophthalmology, Goa Medical College, Goa- India. Adult onset Best's macular dystrophy (VMD2) with unilateral presentation