

# Adult foveomacular vitelliform dystrophy

## *Distrofia viteliforme foveomacular do adulto*

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### ABSTRACT

*Adult foveomacular vitelliform dystrophy is a rare pathology. Less than 1% of the reported cases display perifoveal capillary permeability. The three-year follow-up period of the case revealed a rare form, which had not yet been documented. The patient was a 40-year-old female with normal visual acuity, and a minor complaint of metamorphopsia on the left eye. Retinography showed a perifoveal yellowish subretinal area OS. Angiography showed perifoveal leakage OS. Follow up showed that, over 3 years, capillary incompetence disappeared and the yellow area underwent alterations, becoming atrophic OS. Angiography also showed hyperfluorescence (windows defect). Towards the end, it resembled the appearance of late stage of Best's Disease.*

**Keywords:** Macula; Macular degeneration; Macular edema; Telangiectasis; Case reports

### RESUMO

Distrofia viteliforme foveomacular do adulto é uma patologia rara. Menos de 1% dos casos relatados mostram permeabilidade capilar perifoveal. O acompanhamento de 3 anos deste paciente revelou uma forma rara, ainda pouco documentada. O paciente era uma mulher de 40 anos de idade com acuidade visual normal, mas com queixa de metamorfopsia leve no olho esquerdo. A retinografia mostrou uma área sub-retiniana amarelada, perifoveal, com hiperfluorescência na angiofluoresceinografia. O seguimento mostrou, em 3 anos, que a incompetência capilar desapareceu e que a área, antes amarelada, tornou-se atrófica, com hiperfluorescência na angiografia, lembrando o aspecto tardio da doença de Best.

**Keywords:** Mácula; Degeneração macular; Edema macular; Telangiectasia; Relatos de casos

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## INTRODUCTION

The differential diagnosis of macular pathologies in adults includes a myriad of conditions, both acquired and inherited. In general, age related macular degeneration is the prime suspect in patients over 50 years of age, whereas hereditary conditions are the most frequent in the first two decades of life<sup>(1)</sup>.

Adult foveomacular vitelliform dystrophy (AFVD) was first described by Gass in 1974<sup>(2)</sup>. The onset typically occurs between the ages of 30 and 50. Patients report mildly blurred vision or metamorphosis.

Macular telangiectasia (MacTel) type 2 consists of a retinal vascular anomaly exclusive of the capillaries of the juxtafoveal region, which may result in retinal edema and progressive vision loss<sup>(3)</sup>.

The typical AFVD angiogram shows the yellow area as hypofluorescent, surrounded by a hyperfluorescent ring<sup>(1,4)</sup>. The angiographic aspect of perifoveal capillary permeability is described in only 0,83% of registered AFVD cases. Angiography evolution in cases of the disease which initially manifest as the three cases described by Dubovy has never been registered before, as well as the follow up<sup>(5)</sup>.

The diagnosis is confirmed by angiographic changes in a three years follow up.

## CASE REPORT

A 40-year-old female consulted an ophthalmologist in 2003 after realizing that she had a scotoma in the central field along with minor complaint of metamorphosis in the left eye (OS).

Visual acuity without correction was 20/20 and J1, photomotor pupillary reflexes were normal, as well as external ocular motility, and biomicroscopy of the anterior segment, with no signs of inflammation, transparent lens, vitreous without cells or Tyndall. Goldman tonometry was 12 mm Hg and chromatic vision, according to HRR-AO test, was also normal. Amsler test showed mild pericentral metamorphosis in the OS.

Binocular indirect ophthalmoscopy and retinal biomicroscopy showed, in the OS, a yellowish subretinal lesion in the temporal and upper foveal region.

The complementary tests ordered were hemogram, immunofluorescence for toxoplasmosis and lues, fasting glucose - all with normal results. Full-field ERG was normal, as was EOG, with an Arden index of 2 in the right eye (OD) and 2,1 on the OS.

Retinography revealed a normal OD (figure 1A) and a perifoveal subretinal yellowish area OS (figure 1B).

Angiography of the OD was normal, with only a small area of alteration in the perifoveal capillary ramification (figure 1C). Fluorescent angiography of the OS showed a slightly hypofluorescent area in the region corresponding to the lesion and permeability of juxtafoveal capillaries, which became more noticeable during the exam, creating a final scenario of diffuse macular edema with no cystoids characteristics (figure 1D-F).

In 2004, exams were repeated. Retinography of the OS showed signs of epithelium atrophy in the upper edge of the yellow lesion, with a decrease in the vitelliform area (figure 1G). Angiography, of the same eye shows a significant decrease in the retinal capillary permeability, in the area of altered pigment epithelium, where an early transmitted hyperfluorescent point appears, in more advanced phases there is a moderate staining of the surrounding area, which suggests that the scar tissue was

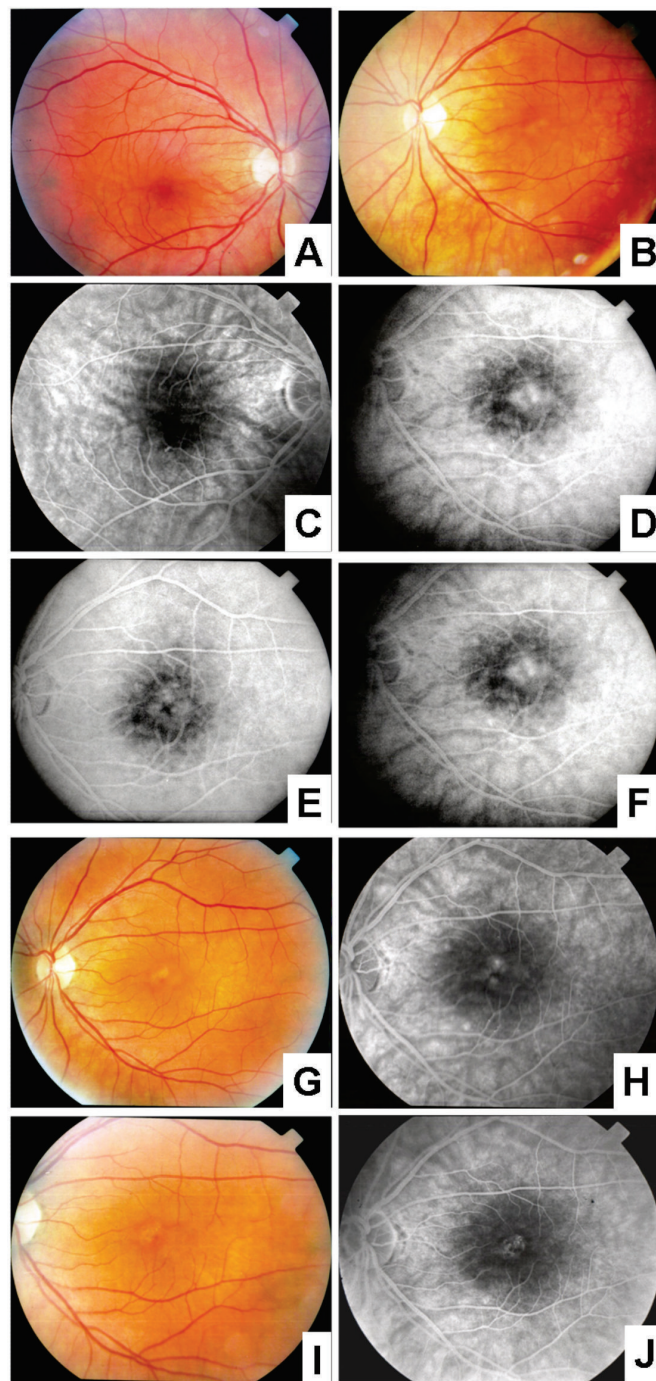


Figure 1: Adult foveomacular vitelliform dystrophy retinography, 2003 (OD fig.1A and OS fig.1B); angiography (OD fig.1C and OS fig.1D/1F); retinography, 2004, OS (fig.1G); angiography, OS (fig.1H); retinography, 2006, OS (fig.1I); angiography, OS (fig.1J).

impregnated by the dye. There was also a hypofluorescent area, which was smaller than the one previously found (figure 1H).

In 2006, exams were repeated. In the OS there was an increase in area of atrophy of the epithelium in the upper edge of the yellow lesion, with a concurrent decrease in the vitelliform aspect figure 1L). Angiography showed, in early stages, growth of the hyperfluorescent area transmitted on the upper edge of the lesion, and disappearance of retinal capillary permeability.

In later stages, the hypofluorescent area showed a decrease in size, with liquid level; the upper area showed impregnation of scarring tissue (figure 1J). Visual acuity of the OS in this occasion had decreased to 20/25.

## DISCUSSION

Diagnosis of AFVD was based on normal visual acuity, normal EOG and ERG, minor distortion of Amsler grid, the presence of a single subretinal perifoveal round yellowish lesion, in the OS, and also on the bibliographic research which confirmed the similarity with uncommon angiographic aspect of perifoveal capillary permeability as described by Fishman<sup>(9)</sup>.

Dubovy registered three cases of macular capillary permeability, with subretinal yellowish lesion, and named this pathology pseudovitelliform macular degeneration (PMD), whose angiographic findings are very similar to the ones described in this case we were able to describe its follow up<sup>(4)</sup>.

Even though this disease usually presents bilaterally, unilateral forms, as well as development of bilateral conditions after unilateral onsets, have been reported. Visual prognosis is usually good<sup>(5,6)</sup>. Conditions with subretinal macular yellow deposits include: foveomacular vitelliform dystrophy, dominant slowly progressive macular dystrophy, butterfly-shaped and pattern dystrophy with yellow plaques. All these hereditary conditions have common characteristics. Whether they present phenotypic expressions regardless of age, or are multiple pathological dysfunctions of foveal EPR caused by different genetic defects, remains to be established.

Epstein and Rabb state, for the cases they studied, that as the vitelliform lesion progresses, there is an increase in retinal exudation, and that this would be the cause of retinal leakage in later stages, probably from choir-capillaries<sup>(7)</sup>. In the case at hand, such evolution has not been observed. The disappearance of retinal capillary permeability occurred before retinographic and angiographic alterations of the vitelliform lesion took place. In the angiography, perifoveal capillary permeability disappears at the same time that atrophic areas of pigment epithelium appear in the vitelliform lesion area. And, finally, alterations which

resemble the pseudohypopyon phase of Best's disease appear the yellow area.

At present, new diagnostic methods enable a better characterization of AFVD. Optical coherence tomography (OCT) shows a hyper-reflective structure in the region of the vitelliform lesion, located between the pigment epithelium and the photoreceptor layers. The pigment epithelium remains without elevation, while the neurosensory retina over the hyper-reflective structure is elevated due to the presence of vitelliform material, but with diminished thickness<sup>(7)</sup>.

In the future, similar cases that may present will benefit from these new diagnostic and characterization methods.

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