MULTIFOCAL VITELLIFORM RETINAL LESION

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SUMMARY
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The authors present retrospective follow up of patient with bilateral multifocal vitelliform retinal lesion during the 18 years period. At this time, spontaneous improvement of objective picture on retina and subjective visual troubles was observed. It is probable, that this case is a part of the same symptom complex as a variant of Best´s hereditary disease. This conclusion was based on initial stadium of phenotypical expressivity and additional evaluations. The course and outcomes of visual functions were different. The hereditary transmission was not confirmed.

Key words: multifocal vitelliform retinal lesion, electrophysiology of retina, the fluorescence angiography, optical coherence tomography

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INTRODUCTION

This rare vitelliform unit was presented by Littann (10) in 1965 to a diagnostic consilium over 67 days at the German Ophthalmological Society, where the pathology gained its name and nosological classification among retinal disorders. Subsequently Denden (3) described two patients in middle age with a bilateral finding of a micro and macro lesion without evidence of hereditary transmission. In a further joint observation (4), Littann and Denden recorded developmental phases of vitelliform lesions from the earlier pseudocystic stage to the final process and scarring with pigmentations. The finding of a macular deposit, in its scope and appearance, was very similar to juvenile form of Best's solitary macular lesion, whilst the satellites were always in a smaller scope and variable in number. At this time questions arose as to how this related to Best's disease, or whether this pathology represented a variant thereof within the framework of a closed symptomatic complex.

CASE REPORT

Within the framework of a consultation from another ophthalmological workplace, we were able to observe this pathology in a 38 year old woman with a typical bilateral finding (fig. 1A, b). We have other data on the further course from the workplace, where the patient underwent the first examination and a number of further follow-up checks. Centrally 4-5 PD a large deposit was filled in the lower part with a compact vitelline substance, around which, especially in the course of the temporal vascular arcades and papilla, there was a small, circular, sharply bordered ¼ to ½ PD large yellowish-white deposit. Visual acuity was slightly reduced, with a feeling of fogging of vision, especially upon close -up vision. After correction of hypermetropia with astigmatism, the subjective complaints receded, but with a surprising finding on the ocular fundus. Through an evaluation of the initial conclusions, differential diagnostics determined multifocal serous detachment of the retinal pigment epithelium (RPE), multifocal choroiditis, white dot syndrome and acute multifocal placoid pigment epitheliopathy.

Fluorescence angiography (FAG) from the first period of examination detected blocked hypofluorescence within the scope of the central deposit, densest in the lower part, which corresponded to an accumulation of not yet reabsorbed vitelline material. The satellite deposits in the surrounding area of the superotemporal vascular arcades and around the disc of the optic nerve formed either a blockade from the present vitelline material or transmitting hyperfluorescence from a lesion of the RPE after applications of argon laser coagulation (ALC). The vascular system of the retina was regular throughout the entire scope and in all phases (fig. 2A, b). The therapy consisted of infusions of mannitol, corticosteroids and ALC in a number of 5 in each eye, but this did not influence the finding on the fundus.

Eventually, after numerous checks and consultations at other workplaces, a diagnosis of multifocal vitelliform retinal lesion was determined, very close to juvenile Best's disease as a possible variant thereof. This hypothesis was supported also by an eletrophysiological examination. Under photopic and scotopic conditions ERG was within the norm, EOG pathological, Arden quotient bilaterally 110. The patient's two children of adult age had regular vision and ophthalmoscopic finding, her siblings and family anamnesis had no ocular pathologies.

DISCUSSION

Multifocal vitelliform retinal lesion has been described a number of times since the first descriptions by Littann and Denden. Remka and Kölbl (15) referred to 7 cases in 1971, and Lisch et al. (9) referred to 30 patients in 1989. The number of such cases continues to increase with variable expressivity of phenotype and various results of examinations of the visual functions of the retina. In the majority these are isolated cases (2, 6, 8, 9, 19), less commonly in family re-

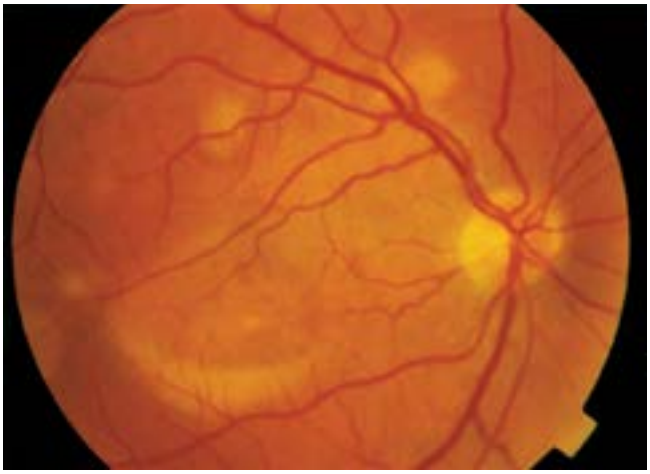

Fig. 1a Macular and extramacular vitellform deposits. Right eye, 1996

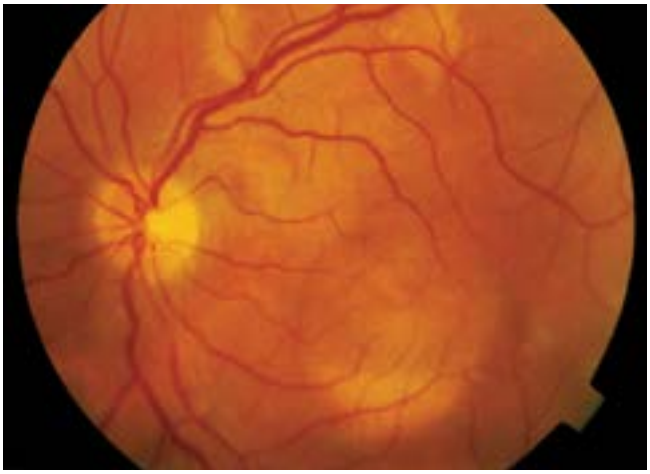

Fig. 1b Macular and extramacular vitelliform deposits. Left eye, 1996

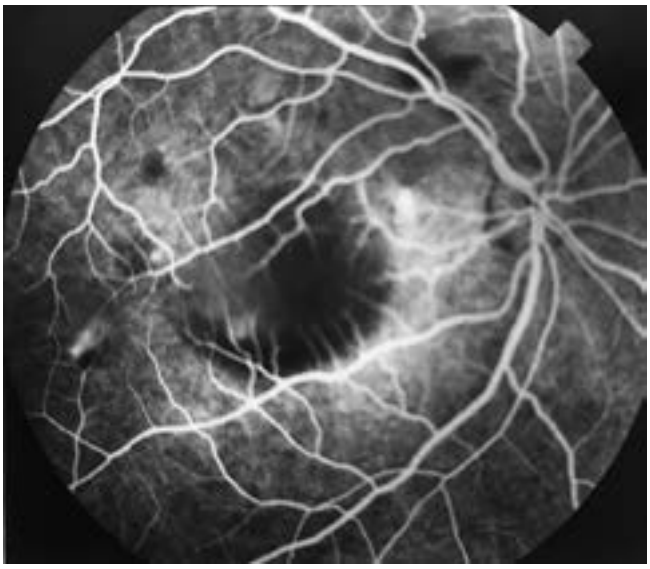

Fig. 2a FAG Right eye, 1996

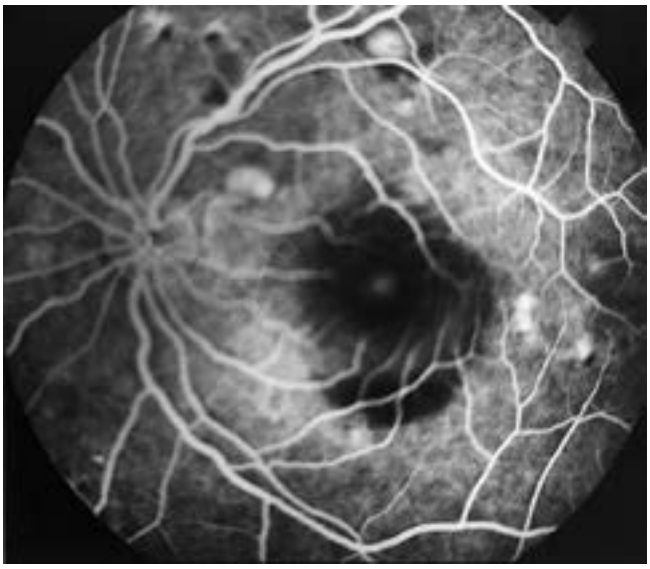

Fig. 2b FAG Left eye, 1996

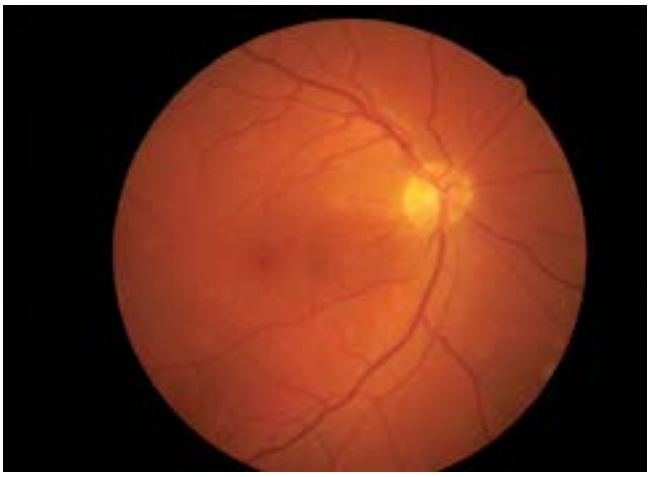

Fig. 3a FAG Right eye, 2014

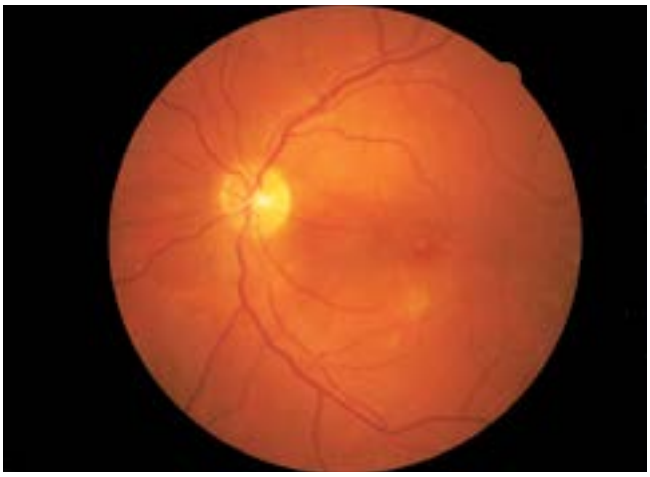

Fig. 3b FAG Left eye, 2014

lations (5, 7, 11, 12). The average age of detection of this pathology is the third to fourth decade of life, which applied also in our case, but in rare cases it also appears in the sixth (9, 17) and seventh decades (13). There is large variability in the appearance, size and number of vitelliform deposits of both central and satellite localisation. Some deposits may merge (9, 17) and block choroidal fluorescence through non -reabsorbed vitelline material upon FAG. The pathological substrate responsible for the vitelline substance is an accumulation of lipofuscin in the RPE and its closest surrounding area. According to histological studies, this pigment of ageing is located throughout the entire RPE (18) and can be detected and displayed by autofluorescence of the fundus (6). It is derived from fats of the outer segments of the photoreceptors which were digested by the RPE cells during the process of renewal of the outer segments of the sensory epithelium. To date it has not been reliably investigated as to why lipofuscin is most present in the macular and extramacular retina in the case of solitary and multifocal localisation. This connection is being sought in a dysfunction of the protein substance of the VMD2 gene bestrophin, which regulates the metabolism of the sensory epithelium of the retina via the RPE as its external barrier.

The natural course of development of this pathology is highly variable in terms of time, mostly parallel amongst the central deposits by extramacular satellites. Upon FAG examination, which displays the developmental stages, hypofluorescence of the deposits is always a sign of an early stage. Upon reabsorption of the vitelline substance and atrophy with scarring on the level of the RPE, hyperfluorescence of the central and extramacular deposits proliferates. This is then corresponded to by a reduction of visual functions, linked to the afflicted areas. The merging of areas is a prognostically adverse symptom of the disease. However, not all cases need necessarily pass through stages of the central deposit, as in the case of solitary disc in Best's disease. Vitelline substances, which are a characteristic of the image on the retina, may be absorbed with minimum damage to the RPE. This remains functional, mainly within the scope of the central deposit. This could also apply to the case of our patient (fig. 3A, b). Optical coherence tomography (OCT-conventional TD) from the recent period of observation detected a bilateral normally configured neuroretina and complex BM -RPE. In the right eye there is a finely diffuse constricted area of the central fovea (fig. 4).

The type of relationship and connection between multifocal vitelliform retina lesion and Best's disease as a variant thereof is being sought on the principle of evidence of the gene VMD2 and pathological EOG. In the case of autosomally dominant hereditary solitary vitelliform Best's disease, the gene VMD2 and reduced EOG are almost always demonstrable. In the case of the multifocal form of this pathology this is not always the rule (1, 16). Both values need not be present, which applies for the independent incidence of the disease as well as for the rarer familiar cases. Newer and accessible genetic examinations enable us to focus more closely on this connection in our observation.

CONCLUSION

Our long-term observation of vitelliform lesions of the retina detected a completely different model of the course and resulting condition of the functions of the central region and its satellite deposits. The finding on the retina was bilaterally symmetrical, functionally auspicious, not lin-

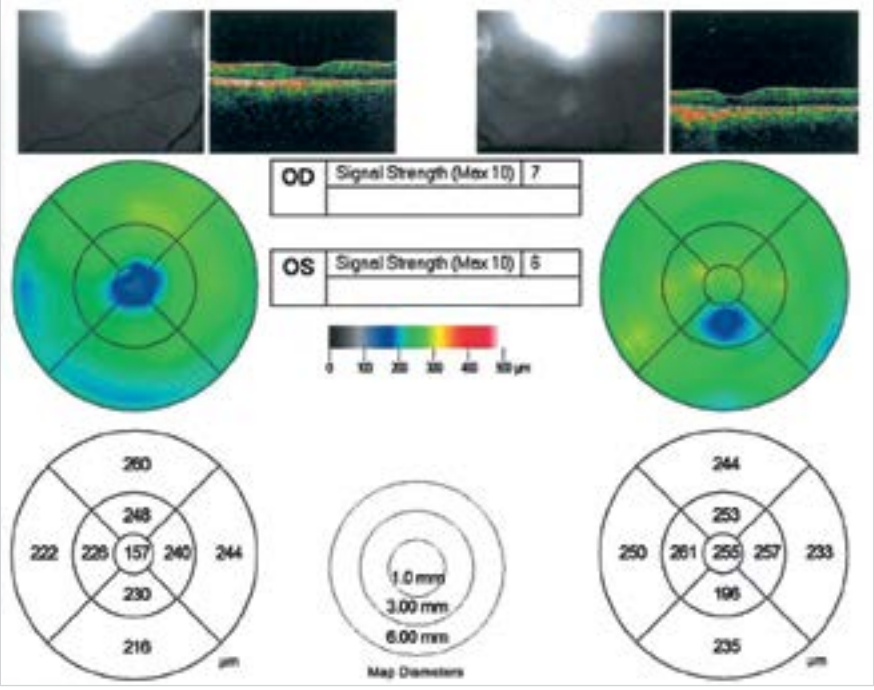

Fig. 4 OCT of right and left eye, 2014

ked to any ocular systemic disorders or hitherto hereditary transmission. We have not recorded this pathology in such a unique condition to date in the literature.

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