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LETTER TO THE EDITOR (BY INVITATION)

Reply: natural course of the vitelliform stage in best vitelliform macular dystrophy: a five-year follow-up study

Maurizio Battaglia Parodi[1] · Alessandro Arrigo[1] · Francesco Bandello[1]

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Dear editor,

We are grateful to our colleagues for the opportunity to offer more details about our investigation on best vitelliform macular dystrophy [1].

Fundus autofluorescence and fluorescein angiography are useful tests in order to achieve the diagnosis, especially for the differential diagnosis, but do not provide prognostic information regarding the functional and anatomic outcomes over the follow-up. On the other hand, indocyanine green angiography, showing a hyperfluorescent lesion due to the binding of the dye to the vitelliform material [2], may result misleading, requiring a differential diagnosis with respect to type 1 macular neovascularization secondary to age-related macular degeneration and central serous chorioretinopathy, which can be solved just by means of optical coherence tomography angiography [3, 4].

All our patients belonged to Caucasian families affected by genetically confirmed Best 1 vitelliform macular dystrophy. Genotype-phenotype correlation proved to be extremely difficult in best vitelliform macular dystrophy [3–7] with the exception of p.Ala243Val *Best1* mutation, resulting in a pattern dystrophy-like phenotype [8].

Uncertain cases in our case series were judged by a third ophthalmologist, and statistical analyses were carried out using student *t* test and ANOVA.

We respectfully disagree regarding the binocular vision assessment, because in a variable figure reaching 20% of cases [3–7], the two eyes of the same patient revealed different stages.

Best vitelliform macular dystrophy is a complex and multifaceted disease, and the identification of biomarkers, like hyperreflective foci [7], related to the progression of the disease would be essential in view of the upcoming gene therapy.

Our data showed that usually the vitelliform lesion tends to progressively enlarge up to its collapse, followed by the vitelliform material reabsorption and the fluid formation, correlated to the passage towards the vitelliruptive stage. Even though the above-mentioned evolution probably reflects the most frequent course of the disease, some exceptions can be found, like the case depicted in Fig. 1.

Overall, further longitudinal studies based on larger samples and longer-term follow-up are warranted to understand the natural course of best vitelliform macular dystrophy, separating specific subgroups characterized by different clinical manifestations.

✉ Alessandro Arrigo
alessandro.arrigo@hotmail.com

[1] Department of Ophthalmology, IRCCS San Raffaele Hospital, University Vita-Salute, via Olgettina 60, 20132 Milan, Italy

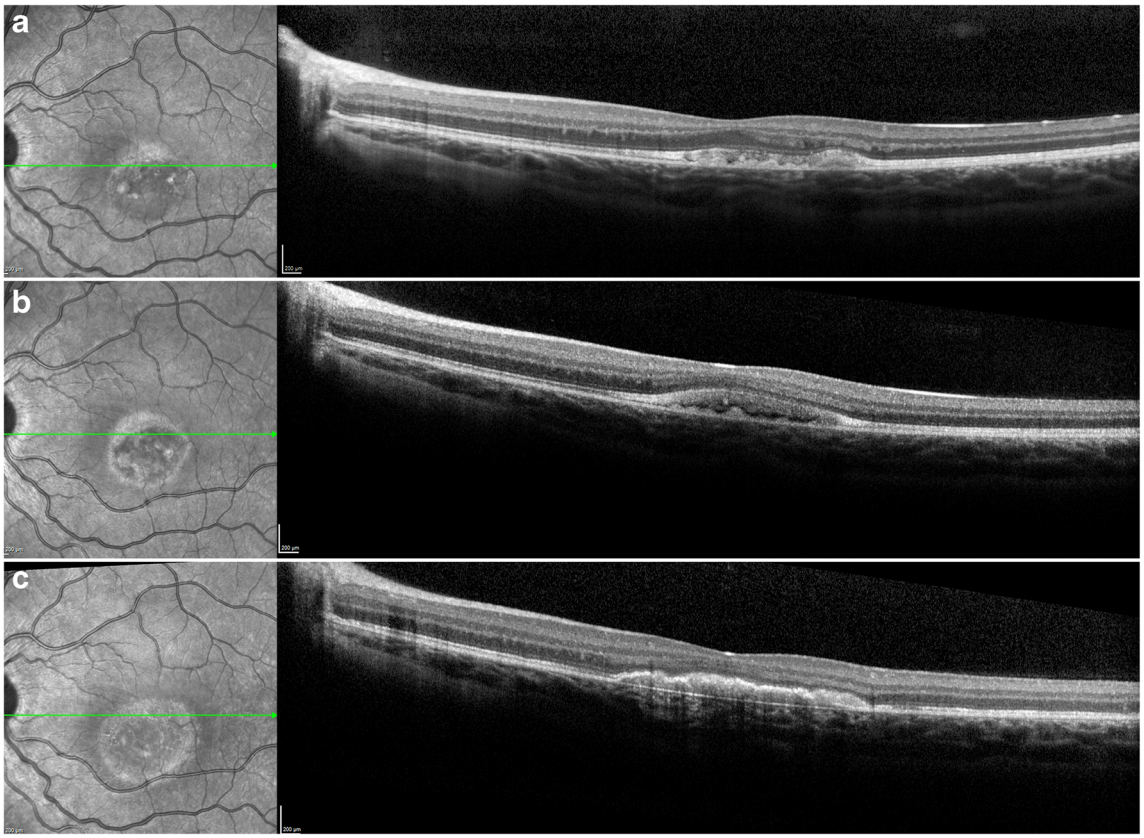

Fig. 1 Best vitelliform macular dystrophy in a 3-year-old girl characterized by a small vitelliform lesion on optical coherence tomography, with solid aspect (**a**). Over a 6-month follow-up (**b**), a progressive enlargement with partial reabsorption of the vitelliform material is evident along with fluid formation. Twenty-four months later (**c**), the vitelliform lesion shows a further expansion with solid aspect

Compliance with ethical standards

Conflict of interest None.

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