A normal electrooculogram in a patient with a typical vitelliform macular lesion

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The diagnosis of vitelliform macular dystrophy can be made from fundus examination alone when the classical egg-yolk lesion is intact. After the lesion has scrambled, however, other information is needed to be certain of the diagnosis. The electrooculogram (EOG) has been reported abnormal in vitelliform macular dystrophy and has been called “an indispensable diagnostic element.” This paper, however, presents a patient with the classical sunny-side-up egg-yolk lesion of vitelliform macular dystrophy and an entirely normal EOG and slightly abnormal electroretinogram (ERG). To our knowledge, this is the first report of a patient with these findings.

The sunny-side-up egg-yolk lesion of classical vitelliform macular dystrophy cannot be mistaken for any other entity (Fig. 1). When the egg yolk is intact, a diagnosis can be made from fundus examination alone, and the concomitant findings of a positive family history, fairly good vision, an abnormal electrooculogram (EOG), and a normal electroretinogram (ERG) are helpful, but totally unnecessary.

In more advanced stages of vitelliform macular dystrophy, after the egg yolk becomes scrambled, it is difficult, if not impossible, to distinguish this macular lesion from other lesions such as central chorioretinitis or other more common macular dystrophies. In these cases, additional information such as a positive family history with an intact sunny-side-up lesion in a family member and electrical studies can help distinguish this disease from other macular problems.

The status of electrical measures, particularly the ERG seems controversial among authors. Krill; Krill, Morse, and Potts; and Francois report little or no ERG abnormality in patients with vitelliform macular lesions, while Rudemann observed consistent changes in the photopic components of the ERG. Rudemann concludes that failure to find ERG changes in macular degenerations originates in poor instrumentation, inadequate procedures, or inadequate interpretation of data.

There is general agreement that the EOG is consistently subnormal where vitelliform lesions are found. Moreover, Deutman found reduced EOG’s in carriers...
of the disease who displayed no other symptoms. Similar changes were noted by Francois who also saw large EOG changes in both eyes of children where ophthalmoscopic signs of the disease were monocular. The apparent reproducibility of the EOG in macular disease, and especially where vitelliform lesions are seen, was described in a brief review by Dorne who called the EOG “an indispensable diagnostic element.” Francois, DeRouck, and Fernandez-Sasso and Deutman have concluded that “electrooculography is essential for the diagnosis of vitelliform dystrophy of the fovea.”

The case history which follows, however, is of a patient with the classical sunny-side-up egg-yolk lesion of vitelliform macular dystrophy and an entirely normal EOG.

Methods

The technique and standards for EOG have been described by Krill and Arden, Barrada, and Kelsey. Records (Fig. 2) are shown at 8 and 12 minutes after onset of dark adaptation and beginning each fourth minute after exposure to the adapting light. The adapting light was turned on at the arrow. A peak in the EOG may be seen in the period between 8 and 16 minutes after the adapting light was turned on. Recovery per cent was calculated from the average maximum response in the light divided by the average response amplitude at the end of dark adaptation.

ERG measurements were made by the methods described by Rudemann, with minor changes.

“Stressing” ERG’s were also done (Fig. 3). Stress is imposed by the first flash of a pair. As the second flash is moved temporally toward flash one, it is assumed that the retina is under increasing stress from flash one. The second response is a measure of response recovery from flash one. The duration between stimulus pairs is 15 seconds. Elenius and Hope and Dawson have suggested that inhibitory interaction between the two flashes may produce unmasking of a relatively pure cone response under high-stress conditions.

Case report

The patient, a 48-year-old white male, was first examined at the University of Florida Eye Clinic, in November, 1970. At that time he complained of decreased vision of two months’ duration. He was otherwise in good health and his family history revealed no evidence of any unusual ocular disease. His visual acuity was 20/40 (O.D.) with a +2.00 spherical correction and 20/60 (O.S.) with a +2.50 spherical correction. Intraocular tensions and external examination were normal (O.U.). Funduscopic examination revealed a typical sunny-side-up egg-yolk lesion of each macula (Fig. 1).

One week later, the patient returned with two of his five siblings for further evaluation. His visual fields, dark adaptation, and color vision were found to be entirely normal as were those of both sisters. Funduscopic examination of the two sisters revealed no evidence of any macu-
Fig. 3. ERG's produced by flash at time zero and at 100 msec. Normal eyes, left; eyes with vitelliform lesion, right. Time, 20 msec. per division, left; 50 msec. per division, right. Vertical amplitude, 50 microvolts per division, all records. Note absence of second response in eyes with vitelliform lesion.

lopathy. All three, then, had EOG's and ERG's performed.

The EOG of the patient is presented in Fig. 2A. The value was 270 per cent, well within the normal range which is generally considered to be any value above 185 per cent. An EOG from a carrier of the vitelliform trait is shown in Fig. 2B for comparison. The value of 105 per cent is clearly abnormal.

ERG's were then done and responses to single stimuli were not remarkable. In light adaptation with dim (1.2 log td.) stimuli the amplitudes from the most negative portion of the a-wave to the b-wave peak were smaller than those of our norm group, but were within one standard deviation of the mean. The only major departure from normal occurred during the "stressing" tests. Retinal "stress" ERG's are made under dark-adapted conditions. Fig. 3 (right) shows ERG's from this patient in response to two flashes separated by 100 msec. and response from two normal eyes (left), with flashes also separated by 100 msec. flashes occurred at the arrows. Reduction of response "two" follows a predictable course in normal subjects, and does not become complete until the flash separation is 50 msec. or less. In this patient the response to flash "two" failed to occur whenever the stimuli were separated by 100 msec. or less.

The EOG, ERG, and "stressing" ERG of each sister tested were normal.

The patient was examined again in August, 1971. Clinical findings were unchanged from the previous examination, and funduscopic examination revealed the macular lesions to be intact. The EOG was again normal and similar to the one done nine months earlier. The value was 285 per cent, again, clearly in the normal range.

Subsequent to the original examination, the patient's only son, one brother, and father were examined in other cities and no evidence of maculopathy was found. Unfortunately, electrical studies on these individuals could not be done.

Comment

From the classical fundus findings alone (Fig. 1) we can be quite certain that our patient does have the lesion of vitelliform macular dystrophy. The relatively good vision associated with the large macular lesion would support a diagnosis of vitelliform macular dystrophy. Atypically, a positive family history appears to be lacking. Three possible explanations for this exist, however. First, sporadic cases have been reported. Second, a complete evaluation of all family members could not be carried out. And third, decreased penetrance and the existence of a carrier state complicate the tracing of a genealogy and may mask the genetic pattern in the family. This probably accounts for most apparent "isolated cases" including our patient, currently presented.

Our patient is somewhat old to have an intact vitelliform lesion; most patients show disruption of the lesion by their late
teens, and nearly all by age 40, but intact lesions in older individuals have been reported.14

A most unusual finding is the normal EOG demonstrated by our patient. A current concept in the ophthalmic literature is that the EOG in vitelliform macular dystrophy is generally abnormal.1, 2, 7, 11 In fact, when a suspicious looking macular lesion is found it has been suggested that the EOG can be used to differentiate between late-stage vitelliform lesions and other macular pathology.1, 2 An abnormal EOG would indicate vitelliform, while a normal EOG would be suggestive of another diagnosis.

To our knowledge, this is the first detailed report of a patient with a clear cut vitelliform lesion and a normal EOG.

It is possible that this patient does not have true vitelliform macular dystrophy. This, however, seems highly unlikely in view of the absolutely classic fundus finding. The only other possible explanation is that this patient has some other heretofore undescribed maculopathy with the identical fundus finding of classical vitelliform macular dystrophy. Since our first examination of this patient, we have done EOG's on seven other patients with vitelliform lesions and one on a suspected carrier, all from other families. All of these patients had abnormal EOG's as classically described (Fig. 2B).

We agree with other examiners that the EOG is generally abnormal in vitelliform macular dystrophy. We have shown, however, that this may not always be true. Additionally, while the classical ERG was normal, more sensitive testing showed abnormalities.

REFERENCES