Familial Exudative Vitreoretinopathy: Presentation in the First Week of Life

To the Editors:

A male infant born at 36 weeks of gestation with a birth weight of 2,500 grams presented to our department at 37 weeks post-conceptional age. The infant was not admitted in any neonatal intensive care unit, had not received oxygen, and did not have a history of neonatal sepsis. He was otherwise healthy.

The infant was brought for a retinal check-up because his elder sibling was bilaterally blind since early infancy. The elder child was not brought but the records showed that the elder child had been investigated for retinoblastoma due to presence of leukocoria. The results were negative and the elder child was blind due to presence of bilateral retinal detachment.

The anterior segments of both eyes of our patient were normal. The fundus of the left eye showed dilated tortuous veins with subretinal exudation along the superotemporal arcade and in the temporal retina and preretinal hemorrhages with straightening of retinal vessels temporally. Avascular retina was seen in zone II with a shallow ridge/demarcation between vascular and avascular retina (Figure 1). The right eye showed large avascular retina encroaching onto zone I. There was a tractional detachment with falciform fold formation and dragging of the disc and retina nasally (Figure 1).

An ear-nose-throat evaluation and hearing test were normal. Considering absence of risk factors of retinopathy of prematurity (ROP), presentation at week 1 of life, and history of blindness in the sibling due to retinal cause, we made a diagnosis of familial exudative vitreoretinopathy (FEVR). Both eyes underwent confluent laser photocoagulation of the avascular retina. The left eye did not show any signs of worsening and was stable at 1 month of follow-up. The detachment in the right eye persisted.

ROP is one of the important differential diagnoses. Many clinical features associated with FEVR overlap with those found in ROP. Differentiating these two entities may be challenging in infants. Gollogorsky et al. reported a case of FEVR in a premature infant.1 Ranchod et al. reported cases of FEVR in children with a mean birth weight of 2.80 kg (range: 740 g to 4.76 kg), mean gestational age of 37.8 weeks (range: 25 to 42 weeks), and mean age at presentation of 6 years (range: < 1 month to 49 weeks).

Figure 1. (A) Left eye fundus image showing extensive avascular retina in zone II with straightening of vessels temporally. Note presence of preretinal hemorrhages and exudates in temporal retina. Few exudates are also noted along the superior arcade. (B) Right eye fundus showing extensive avascular retina encroaching on zone I and a tractional retinal fold with dragging of disc and retina nasally.
Because our patient was born at 36 weeks, with birth weight of 2,500 grams and an uneventful first week of life, ROP was unlikely. Norrie’s disease, another differential diagnosis of FEVR, was less likely because the child had normal auditory function. A genetic analysis could not be done due to cost being a limiting factor.

The presence of detachment in one eye at the age of 7 days implies that the proliferative retinal changes would have started in the late intrauterine period itself. We want to highlight the fact that FEVR may present early in life and timely detection and treatment might salvage vision in these eyes. We also recommend early retinal screening of infants whose siblings have been known to have lost vision due to retinal pathology early in life.

REFERENCES


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