Infantile Vitreous Hemorrhage as the Initial Presentation of X-linked Juvenile Retinoschisis

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The authors report two cases of X-linked juvenile retinoschisis (XLRS) manifested as bilateral vitreous hemorrhage as early as in an 1-month-old infant and in a 3-month-old infant. The one-month-old male infant showed massive bilateral vitreous hemorrhage. During vitrectomy, thin membrane representing an inner part of schisis cavity was excised and intraschisis hemorrhage was evacuated. As intraschisis cavities were cleared, the stump of inner layer appeared as the demarcation line between the outer layer of the schisis retina and non-schisis retina. The other three-month-old male infant presenting with esodeviation also showed bilateral vitreous hemorrhage. Typical bilateral retinoschisis involving maculae could be seen through vitreous hemorrhage in both eyes on fundus examination. Spontaneous absorption of hemorrhage was observed on regular follow-up. XLRS could be manifested as massive hemorrhage inside or outside of the schisis cavity early in infancy.

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Fig. 1. Preoperative ultrasonography (USG) and intraoperative fundus findings of the right eye in case 1. USG shows vitreous hemorrhage (arrow) and suggests retinoschisis (arrowhead) (A). Massive vitreous hemorrhage is being removed after lensectomy (B). Thin membranous structure (arrow) is detected during careful vitrectomy (C). After excision of thin membranous inner layer, nonschisis retina (N) can be shown (D). The stump (arrowheads) of inner layer can be shown with adherent intraschisis blood clot (E). As intraschisis blood is being removed, the outer layer (star) of the schisis retina (S) appears (F).

Fig. 2. Fundus findings observed at the age of 32 months in case 2. Bilateral retinoschisis involving posterior pole can be shown (A: right eye, B: left eye). Fundus findings were stationary without progression with partially remnant hemorrhage in the right eye at the age of 32 months.

tractional membrane. At 2 months after the last vitrectomy, eleetroretinogram (ERG) was flat in both eyes. All six exons and flanking regions in XLRS1 gene were directly sequenced, but known gene mutations were not detected.5

Case 2

One 3-month-old male infant was referred with esodevi-

viation of his left eye. He had no perinatal problems and was
full-term delivery. There was no family history of bleeding tendency or retinal disorder. His fix and follow was poor to moderate in both eyes. Fundus examination under general anesthesia revealed bilateral retinoschisis involving maculae with vitreous and intraschisis hemorrhage obscuring retina. The schisis involved the temporal retina in his right eye and nearly total retina except the nasal part in his left eye. The absorption of initial vitreous hemorrhage was observed during regular follow-up but recurred vitreous hemorrhage was detected at the age of 38 months. Fundus findings were stationary without progression with partially remnant hemorrhage at the age of 42 months on the last follow-up (Fig. 2). The ERG showed typical “negative ERG” of retinoschisis in both eyes. XLRS-related gene (RS1) mutation (c.544C>T, p.Arg182Cys) was detected in the infant and his mother.5

Discussion

Vitreous hemorrhage is uncommon in the first year of life, but could be seen in ROP, shaken baby syndrome, intracranial hemorrhage, thrombophilic diseases, or retinal dysplasia.6 Intraschisis or vitreous hemorrhage in XLRS results from rupture of unsupported retinal vessels or rarely from neovascularization. It usually occurs later in childhood and clears spontaneously.1 Herein, we provided two infantile XLRS with vitreous hemorrhage and different clinical courses. In case 1, although the genetic mutation associated with XLRS was not detected, clinical history and intraoperative findings of retinoschisis with vitreous hemorrhage strongly suggest XLRS, and exclude the possibility of other systemic and ocular causes except DIC, which could be the cause of diffuse subretinal hemorrhage.4 Among the clinically diagnosed XLRS cases, the gene mutation cannot be detected in about 9%.5 Case 2 showed typical fundus findings in XLRS and the gene mutation. We described two cases of XLRS in infants exhibiting the wider spectrum of disease expression. Our report shows that XLRS could be demonstrated as massive vitreous hemorrhage even in early infants.

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