

Case report

Bilateral rhegmatogenous retinal detachments from giant retinal tears in an infant with abusive head trauma and Stickler syndrome

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ABSTRACT

Purpose: To present a rare presentation of abusive head trauma (AHT) in an infant with a hereditary vitreoretinopathy.

Observations: A two-month-old infant female victim of AHT presented with bilateral rhegmatogenous retinal detachments from giant retinal tears. She had rib fractures, a subdural hematoma, and hyphemas bilaterally. Retinal hemorrhages were not observed. The left eye was repaired by vitrectomy with intermediate-term perfluorocarbon liquid tamponade. Genetic testing demonstrated a pathogenic COL2A1 mutation confirming Stickler syndrome.

Conclusions and Importance: Ophthalmic complications of AHT classically manifest as retinal hemorrhages in multiple layers. Instead, bilateral RRDs from GRTs were observed in this infant with Stickler syndrome.

1. Introduction

Pediatric abusive head trauma (AHT) is defined by the CDC as an injury to the skull or intracranial contents of a child less than 5 years of age secondary to intentional blunt impact and/or violent shaking.¹ AHT is a common subtype of nonaccidental trauma. The most common and classic ophthalmic manifestation of AHT is retinal hemorrhages in multiple retinal layers.² In addition, a wide variety of other retinal features including retinoschisis, retinal folds, epiretinal membrane, macular hole, macular pseudo-hole, and choroidal rupture have been reported in cases of AHT.^{3–7} We report a case of bilateral rhegmatogenous retinal detachments from giant retinal tears in an infant subsequently diagnosed with a hereditary vitreoretinopathy who presented as a victim of AHT without retinal hemorrhages.

2. Case report

A two-month-old female victim of AHT presented to a Pediatric Retina subspecialty clinic following hospitalization for AHT. There was a documented mechanism of AHT involving shaking of the patient by a family member. Documented injuries sustained from her nonaccidental trauma included chronic cortical contusions, a subdural hematoma, and rib fractures in multiple stages of healing.

On ophthalmic examination, the patient was found to have bilateral

rhegmatogenous retinal detachments associated with giant retinal tears. Additionally, periorbital ecchymoses, vitreous hemorrhages, and hyphemas were noted bilaterally. However, retinal hemorrhages were not observed. The closed-funnel total retinal detachment OD was deemed inoperable (Fig. 1). The near-total retinal detachment OS was repaired with lens-sparing pars plana vitrectomy (PPV), membrane peeling of subretinal proliferative vitreoretinopathy, encircling endolaser retinopexy, and intermediate-term perfluorocarbon liquid (PFCL) (Fig. 2A and B). The hyaloid face was not detached. In order to prevent infolding of the giant retinal tear, the hyaloid face was mechanically separated with pick and forceps before stabilizing the macula with PCFL. Topical prednisolone acetate 1% and moxifloxacin were used four times a day post-operatively. Two weeks post-operatively the PFCL was removed by PPV with fluid-air exchange (Fig. 2C). Three months post-operatively macular pigmentary alterations and a moderate epiretinal membrane were observed OS (Fig. 2D). On most recent follow-up, two years post-operatively, the retina OS remained attached. The patient had 20/190 Teller acuity vision and normal contrast discrimination (1.25% by Hiding Heidi) OS and no light perception OD with myopic astigmatism OS (−8.00 + 2.00 x 075). Visual field OS was limited by nasal hemianopia from cortical visual impairment.

Subsequent genetic testing confirmed Stickler syndrome with COL2A1 mutation known to be associated with anterior polar cataracts, hearing loss, micrognathia, and congenital hip dysplasia.⁸

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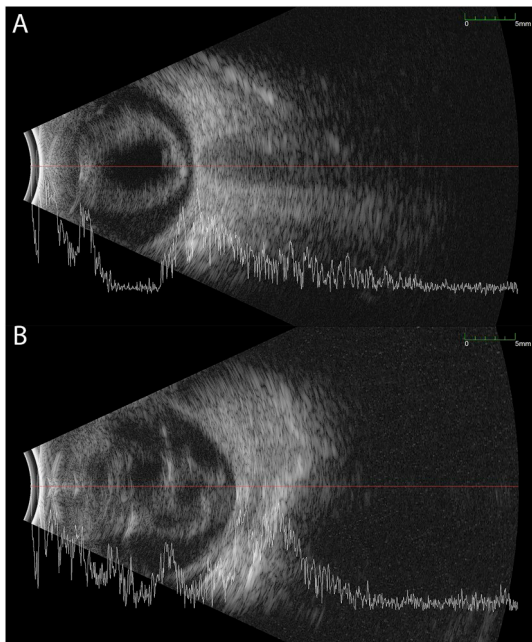


Fig. 1. (A) B-scan ultrasonography image sagittally near the vertical midline of the right eye (OD) demonstrating total retinal detachment. (B) Peripheral view showed no attachment of the peripheral retina to the vitreous base.

3. Discussion

In cases of confirmed AHT, retinal hemorrhages have been noted to be present in 78% of patients.² Systematic review has shown that in studies which included the number of retinal hemorrhages present in cases of AHT, 83% of patients had “large numbers” of retinal hemorrhages documented on examination as defined by the systematic review.² However, instead of classic retinal hemorrhages, pediatric

victims of AHT rarely present with rhegmatogenous retinal detachment.⁹ This case highlights the influence of an underlying hereditary vitreoretinopathy on the manifestation of pediatric AHT as bilateral near-total retinal detachments from giant retinal tears.

Stickler syndrome is a hereditary connective tissue disorder resulting from a defect in type II collagen, which is present in cartilage and secondary vitreous. Ophthalmologically, Type I Stickler syndrome (*COL2A1*) is associated with an “optically empty” vitreous with membranous fibrillar vitreous degeneration leading to a high rate of rhegmatogenous retinal detachment, often in the pediatric age group. In a previous study, rhegmatogenous retinal detachments in this age group ranged widely in severity with giant retinal tears accounting for 31.4% of causative breaks on presentation.¹⁰ The age range represented was 3–45 years of age, with a mean age of 11.5 years of age. Therefore, it can be inferred that our patient presented as a result of AHT.

Retinal detachment in the pediatric population is an infrequent finding often occurring in the setting of an inciting event or underlying retinal condition.¹¹ In cases of pediatric rhegmatogenous retinal detachment the macula is often involved and total detachment is not uncommon due to late presentation.¹² The rate of anatomical success of retinal detachment surgery in pediatric cases is etiology-dependent with rhegmatogenous retinal detachments having better outcomes compared to tractional retinal detachments.¹¹ The use of PFCL for short-term postoperative tamponade for retinal detachment in the pediatric population has previously been described in complex retinal detachment cases.¹³ PFCL was advantageous compared to silicone oil or gas tamponade in this infant as it allowed for supine positioning and eliminated the difficulty of prone positioning.

Retinal hemorrhages are the most common retinal finding in AHT. However, the absence of retinal hemorrhages does not exclude AHT. This rare case demonstrates that bilateral rhegmatogenous retinal detachments can be the presenting ophthalmic finding in a victim of AHT with an underlying hereditary vitreoretinopathy. In this case, short-term PFCL with supine positioning facilitated reattachment of this complicated retinal detachment.

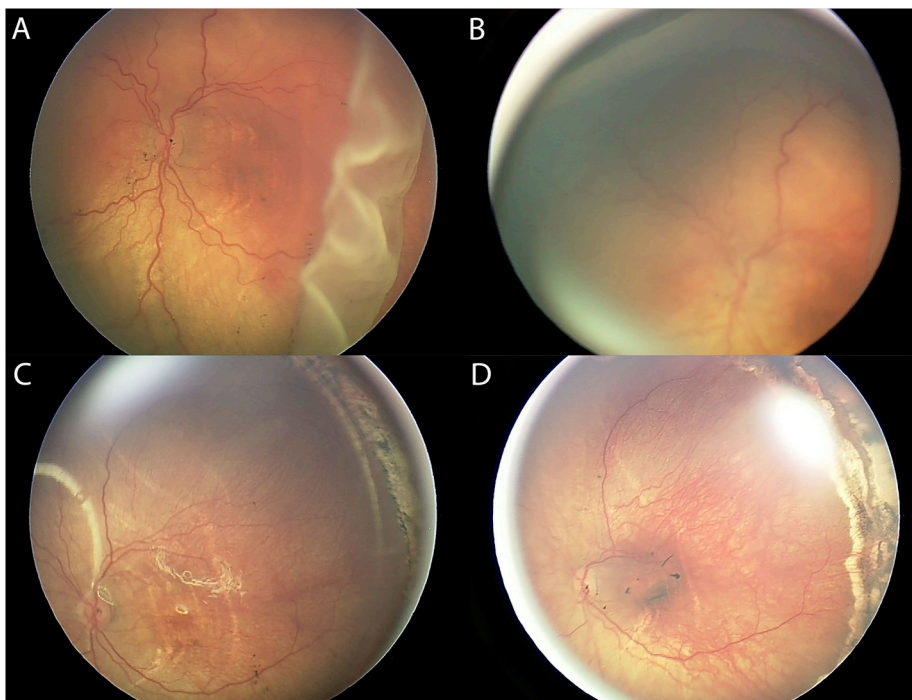


Fig. 2. (A, B) Fundus photography of the left eye (OS) demonstrating macula-involving retinal detachment from a giant retinal tear. Multiple concentric demarcation lines were observed through the macula and posterior retina. (C) Two weeks later, the meniscus of the PFCL bubble is observed at the border of laser retinopexy to the completely reattached retina OS. The PFCL was removed and replaced with air tamponade. (D) Three months later, macular pigmentary alterations and an epiretinal membrane were observed contracting the temporal arcades.

Patient consent

The patient's power of attorney verbally consented to publication of the case.

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Declaration of competing interest

The following authors have no financial disclosures: JJE, VMU, RAS.

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