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BILATERAL RETINAL DETACHMENT IN A PEDIATRIC PATIENT

Caleb Cadis, MD, Alex Wang, BS, Maneesha Julakanti, BS, and Andrew Juergens, MD

Scott & White Medical Center – Temple, Temple, Texas

Reprint Address: Caleb Cadis, MD, Scott & White Medical Center – Temple, 2401 S. 31st St., MS-11-AG062, Temple, TX 76508

☐ **Abstract—Background: Pediatric retinal detachments occur rarely, and thus may be easily missed. Without treatment, this condition leads to permanent vision loss. Patients with Stickler syndrome, an inherited disorder of collagen synthesis, are more likely to have retinal detachments than the general population. Case Report: We present a case of a 9-year-old boy who presented to the Emergency Department with blurry vision, and who was subsequently diagnosed with bilateral retinal detachments. The patient underwent successful operative intervention. He was eventually determined to have Stickler syndrome. Why Should an Emergency Physician Be Aware of This?: It is important for emergency physicians to recognize pediatric visual problems such as retinal detachment, as their presentations may be unusual, and delay of definitive care could result in lifelong visual impairment.** © **2019 Elsevier Inc. All rights reserved.**

☐ **Keywords—emergency medicine; ophthalmology; retinal detachment; pediatric; vision; ocular**

INTRODUCTION

We present a case of a 9-year-old boy who presented to the Emergency Department (ED) with acute bilateral vision loss. He was ultimately diagnosed with bilateral retinal detachment due to the genetic connective tissue disorder Stickler syndrome type I.

CASE REPORT

A 9-year-old boy presented to the ED with sudden-onset bilateral blurry vision that occurred 2 nights prior to presentation while he was playing video games. While at school the next day, he reported difficulty completing assignments. The next morning, he complained of the same problem to his mother, who noted that the patient seemed unsteady without support and "very wobbly." She brought him to the ED to be evaluated for these symptoms.

On initial history, he reported that he could only see to the side and could not see straight ahead. He denied specific trauma to the eyes, pain, infectious symptoms, contact lens use, eyelid abnormalities, headache, nausea, or vomiting. On physical examination, pupils were equal, round, and reactive, extraocular movements were intact, and there was no nystagmus. External examination revealed normal-appearing anterior segments with no hypopyon or hyphema. A slit-lamp examination was not performed. Funduscopic examination was attempted unsuccessfully. The physician noted that the patient deviated both eyes down and to the right to focus on objects situated directly in front of him. The patient also demonstrated abnormal coordination and gait, with some stumbling while walking, concerning for cerebellar ataxia. Due to the unusual nature of his symptoms and concern for a possible posterior fossa tumor, he was admitted to the pediatric service.

During his stay, magnetic resonance imaging revealed a funnel retinal detachment of the right eye and an

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inferior retinal detachment of the left eye (Figure 1). There were no other clinically significant abnormalities on imaging. Ophthalmology was consulted, and he underwent surgical repair. Subsequent genetic testing revealed that he had Stickler syndrome—a rare autosomal dominant connective tissue disorder known to be associated with retinal detachment.

DISCUSSION

Rhegmatogenous retinal detachment (RRD) is a condition in which the neurosensory retina is detached from the underlying retinal pigment epithelium due to a retinal tear, permitting vitreous leakage through the break in the retina. Pediatric RRD occurs at an annual incidence of 0.38–0.69 per 100,000 (1). Stickler syndrome, a rare cluster of genetic disorders involving mutations in genes for collagen synthesis, is the most common inherited cause of pediatric RRD (2–4). The mean and median ages for pediatric retinal detachment range between 9 and 13 years, with 70–80% occurring in males. Only 40–70% of diagnosed patients report experiencing classic retinal detachment symptoms, including perception of floaters, light flashes, or a "dark curtain" (1,5). Due to a combination of gradual disease progression and the difficulty pediatric patients can have explaining their symptoms, the diagnosis tends to occur at a later stage when permanent vision loss has already occurred. Thus, these cases tend to fare more poorly than their adult counterparts (1).

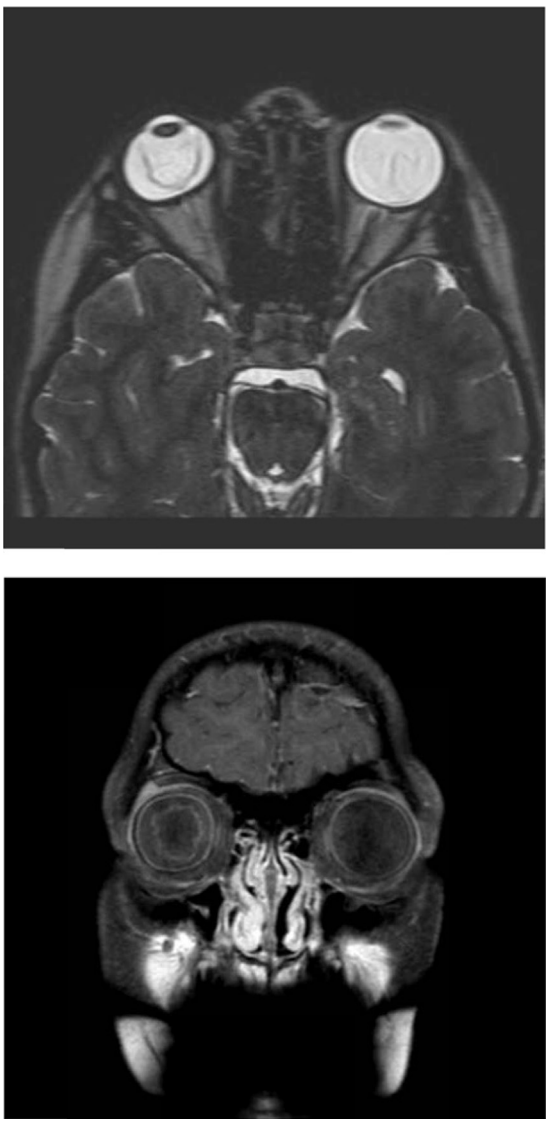

Figure 1. Axial and coronal magnetic resonance image depicting bilateral retinal detachment.

In this case, the diagnosis was complicated by the unusual simultaneous occurrence of bilateral retinal detachments. The vision loss resulted in difficulty ambulating, which mimicked other neurologic pathologies. In addition to retinal detachment, the differential included posterior vitreous hemorrhage, vitreous hemorrhage, endophthalmitis, and dry eye, as well as migraine with aura, intracranial mass, increased plasma viscosity secondary to lymphoma/leukemia, psychosomatic etiology, and many other possible causes. The astute clinician should conduct a thorough eye examination in patients with neurologic symptoms. Fundoscopy should be performed but can be difficult in uncooperative or young patients. If retinal detachment is suspected, ocular ultrasound can have a high sensitivity and specificity, but has not been studied in children (6). Urgent ophthalmology consult is indicated, given the time-sensitive nature of this disease.

WHY SHOULD AN EMERGENCY PHYSICIAN BE AWARE OF THIS?

Retinal detachment is not an exclusively adult disease process. Although rare, pediatric retinal detachments require prompt recognition and treatment. The emergency physician should maintain a high level of suspicion for retinal detachment when there is any complaint of visual symptoms, especially when the child describes classic symptoms (e.g., floaters, light flashes, a "dark curtain"). Because children may have difficulty accurately describing symptoms, this diagnosis is often missed. Additionally, a direct fundoscopic examination may be difficult to obtain, and ocular ultrasound should be considered. If concern remains, consider obtaining specialty consultation with Ophthalmology or possible admission for further evaluation. Failure to diagnosis this condition can lead to permanent loss of vision.

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