

A fatal red eye: Ehlers Danlos syndrome

Valerie Purvin, Aki Kawasaki

CASE REPORT

A healthy 24-year-old archeology student reported headache and a red eye. In addition, he noted double vision. Examination revealed dilated conjunctival vessels without evidence of ocular infection. There was limited movement in abduction of the left eye and the diagnosis was made as a left sixth nerve palsy. The redness of the left eye was attributed to local dryness and irritation (Figure 1). Evaluation including serologies for inflammatory disorders, head MRI and lumbar puncture was unrevealing for a specific etiology. One month later, the patient complained of increasing redness of the left eye as well as pulsatile tinnitus. Subconjunctival hemorrhages and dilated, tortuous episcleral veins having a “corkscrew” formation were visible. In addition, a mild proptosis of the left eye was observed (Figure 2). A bruit was audible over his left brow, and a clinical diagnosis of carotid cavernous sinus fistula was made.

Additional questioning revealed a history of sigmoid volvulus and a family history of skin fragility including easy bruisability and scar formation. The patient demonstrated joint laxity and hyperextensible skin (Figure 3 and 4). The vascular subtype of Ehlers Danlos syndrome (EDS) was suspected to be the basis of all the patient’s clinical manifestations, including the spontaneous development of a carotid cavernous sinus

fistula. Angiography confirmed a fistula on the left side and revealed the presence of bilateral fusiform aneurysms of the cervical carotid artery. A skin biopsy was performed and confirmed both a qualitative and quantitative defect of collagen. Genetic testing was not available at the time.

The fistula was successfully treated with balloon occlusion. The patient was well until one year later



Figure 1: The left eye shows mild conjunctival hyperemia and limited movement to left gaze.



Figure 2: One month later, compared to figure 1, subconjunctival hemorrhages and dilated “corkscrew” episcleral veins are now present and best seen in the superior sclera of the left eye.

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Figure 3: Hand joints demonstrate excessive flexibility and laxity.



Figure 4: The patient's skin stretches easily to gentle pulling (hyperextensibility).

when he developed an acute severe headache and then died. He was presumed to have suffered an intracranial hemorrhage; an autopsy was not performed.

DISCUSSION

An isolated, non-traumatic sixth nerve palsy can be the first sign of central nervous system pathology such as tumor, inflammation, infection and even aneurysm, particularly in younger patients [1]. We describe a young

man whose sixth nerve palsy was the heralding sign of a spontaneous carotid cavernous sinus fistula. A carotid cavernous sinus fistula is an abnormal communication between the carotid arterial system and the venous cavernous sinus [2]. In the non-traumatic setting, a carotid cavernous sinus fistula may develop in older persons in association with atherosclerosis or in pregnant women during or after delivery. It is a decidedly rare occurrence in otherwise healthy young persons except in the setting of underlying collagen vascular disease.

Ehlers-Danlos syndrome (EDS) comprises a heterogeneous group of inherited disorders of collagen and connective tissue synthesis caused by mutations in the *COL3A1* gene which codes for procollagen. Clinical features of tissue fragility include skin translucence and hyperextensibility, joint hypermobility, easy bruisability and hematomas. The collagen dysfunction leads to weakening of the walls of the blood vessels, intestines and uterus with subsequent rupture of vascular and digestive structures in 80% of patients by age of 40 years [2]. Patients with vascular EDS require special precautions during surgical procedures and referral to a high-risk obstetrics clinic in the event of pregnancy. Invasive procedures are generally contraindicated. Aneurysm formation, arterial rupture, dissection and fistulas are the defining features and major cause of mortality of EDS type IV, more commonly known as vascular EDS. The mean age of a spontaneous major vascular complication is 25 to 28 years, and the average age of death is between 45 to 50 years. EDS type IV is inherited as an autosomal dominant trait that is caused by mutations in the *COL3A1* gene coding for type III procollagen [2, 3].

This case highlights the importance to keep spontaneous carotid cavernous sinus fistula in the differential diagnosis of an acute red eye. Furthermore, if such a fistula occurs spontaneously in a young person, consider Ehlers Danlos syndrome. Clinicians should be aware of the special precautions required in the care of persons with vascular Ehlers Danlos syndrome and the possible vascular complications which lead to early mortality.

CONCLUSION

Patients with vascular EDS have increased risk of arterial, digestive and obstetrical complications related to rupture of weak tissue walls. Invasive procedures are generally contraindicated or approached with special caution.

Keywords: Carotid cavernous sinus fistula, Collagen disorder, Ehlers Danlos syndrome

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Author Contributions

Valerie Purvin – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Aki Kawasaki – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor of Submission

The corresponding author is the guarantor of submission.

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Conflict of Interest

Authors declare no conflict of interest.

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