EHLERS-DANLOS SYNDROME TYPE IV

– Anesthetic Considerations –

– Case Report –

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Abstract

This report describes the anesthetic management of a patient with Ehlers-Danlos syndrome type IV. This is one of the rare genetic disorder which can present both in emergency and as a scheduled surgical case.

Key words

Anesthetic management, Ehlers-Danlos syndrome type IV, Vascular EDS.

Introduction

Ehlers-Danlos syndrome is a group of inherited connective tissue disorders characterized by skin extensibility, joint hypermobility and tissue fragility. There are six major types, with a prevalence 1:560000 to 1:5000[1]. Type IV EDS (also known as Vascular EDS), has a high incidence of vascular damage. The arteries are extremely fragile, can have

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multiple aneurysmal formations, spontaneous rupture and dissection. There also high risk of intestinal and uterine ruptures[2].

Case report

A thirty-six year old man presented with acute swelling and severe pain of his left arm 18 hours after playing golf. After evaluation with ultrasound of his left upper extremity vasculature, a diagnosis of compartment syndrome was made. He was brought to the operating room for an emergency fasciatomy. Patient was in severe distress due to pain in his left arm. He admitted to easy bruising and slow healing.

On physical examination, he had a long facial contour, long prominent nose and translucent skins. He had long fingers, but with little skin hyperextensibility. His vital signs were within normal limits. His past history was significant that he was diagnosed with Ehlers-Danlos syndrome type IV after a spontaneous rupture of his azygos vein leading to a hemothorax. He had splenectomy done following splenic artery rupture after pushing a car. He had a hepatic artery aneurysm which was treated by embolization. He also gave a history of spontaneous dissection of distal superior mesenteric artery which was treated conservatively.

At age 32, the diagnosis of Ehlers-Danlos syndrome type IV was established after skin biopsy and fibroblast culture, showing deficiency of collagen III. He had a positive family history of Ehlers-Danlos syndrome type IV. His father died at age 36 years with intra-abdominal hemorrhage of unknown etiology, and his sister died at age of 32 from complications of EDS with a positive history of EDS-IV.

Patient was taken to the operating room. General anesthesia was induced with rapid sequence method using propofol, succinylcholine and fentanyl. Under direct view, his trachea was intubated gently with a 7 mm ID endotracheal tube. Anesthesia was maintained with sevoflurane, oxygen and nitrous oxide. Intra-operative management was also focused on controlling the blood pressure as he had a known pre-existing aneurysm of superior mesenteric artery. Intra and postoperative course was uneventful, and he was discharged home the following day.

Discussion

Ehlers-Danlos syndrome is a group of inherited connective tissue disorders characterized by skin extensibility, joint hypermobility and tissue fragility. There are six major types, with prevalence 1:560000 to 1:5000[1]. Type IV EDS, also known as vascular EDS, has a high incidence of vascular damage[2]. The mutations within the triple-helical coding region of COL 3 A1 (collagen III gene) leading to glycine substitutions and exon skips are the most common. Sometimes deletions of collagen III gene can be seen as well[3]. The arteries of these patients are extremely fragile. They can have multiple aneurysm formation and spontaneous rupture and dissection[4], with the high risk of bowel perforation and uterine rupture[5].

Patient with Ehlers-Danlos syndrome type IV can (1) have distinctive features. They have thin, long thin nose, sunken checks and bulging or protruding eyes and thin lips. (2) Very thin and translucent skin. (3) Tendency to bruise easily, delay in wound healing and (4) rupture of vessels, especially the middle sized arteries, and rupture of viscera such as intestine and uterine. Spontaneous rupture of arteries is the most common presenting symptom. Most patients develop these complications before age of 40. Median age in these patients is 48 years[2].

Diagnosis of this syndrome is based mainly on above four clinical features, but definite diagnosis is made on skin biopsy and culture of skin fibroblast, which show mutations within the triple-helical coding region of COL 3 A1 (collagen III gene). Glycine substitutions and exon skips are the most common mutations and some times deletions can be seen as well[3]. It is a monogenic disease, transmitted as an autosomal dominant trait. Affected individuals have a 50% chance of passing EDS on the each child[6].

The choice of anesthesia in emergency situation can be general or regional, depending on the site of injury. Even though epidural analgesia has been reported to be used successfully in a patient for labor and deliverly[7], bleeding is always a danger because of the fragility of the tissue and difficulty to obtain hemostasis. Often times these patients are presented with a full stomach in an emergency situation with internal

bleeding from a dissecting aneurysm. In these situations, general anesthesia is recommended. Since general anesthesia exposes the patient to the risk of aspiration, laryngeal airway mask is less favorable. LMA can put more pressure on the fragile soft tissue in the pharynx which can cause bleeding form the fragile vessels. Intra-operative management was focused on stabilizing patient's blood pressure, as these patient always carried great risk of spontaneous visceral or arterial aneurysm rupture[8,9]. Prophylactic use of DDAVP under consultation of a hematologist has been reported[10]. In that case report, patient had an increased bleeding tendency with subconjunctival bleeding even after sneezing. Our patient bruised easily, but his coagulation panel was normal, we did not give DDAVP.

In summary, we have described the clinical features and anesthetic management of a patient with Ehlers-Danlos syndrome type IV with multiple episodes of past arterial aneurysm ruptural and/or dissections. Patient was managed with general anesthesia with no peri-operative complications.

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