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Management of Patients with Vascular Ehlers-Danlos Syndrome and Acute Coronary Syndrome: a Case Report

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Case Report

Abstract

BACKGROUND: Ehlers-Danlos syndrome (EDS) is a hereditary collagen vascular disorder characterized by joint hypermobility, skin hyperextensibility, and tissue fragility. Vascular EDS (vEDS) is a subtype of EDS which is characterized by vascular fragility.

CASE PRESENTATION: This is a case report of a young man with vEDS hospitalized for myocardial infarction. He was presented with a coronary dissection and developed aortic dissection, coronary rupture, and cardiac tamponade until death.

CONCLUSION: This case report highlights how patients with vEDS and acute coronary syndrome show a higher risk of vascular complications compared with other patients, and their admission to the institution with a cardiac surgery room could be helpful and safe for better management of the complications. Non-invasive methods could be useful to exclude other vascular diseases, before the emergency coronary intervention.

Keywords: Vascular Ehlers-Danlos syndrome; Acute coronary syndrome; Coronary dissection; Aortic dissection; Coronary rupture; Cardiac tamponade

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Introduction

Ehlers-Danlos syndrome (EDS) refers to a group of autosomal dominant genetic disorders affecting connective tissues, which consists of more than 13 subtypes based on the new EDS classification [1]. The vascular EDS (vEDS) subtype is a dominantly inherited disorder secondary to pathogenic variants within the COL3A1gene encoding type III collagen chains [2]. Patients with vEDS are predisposed to medial degeneration of arteries, often leading to aneurysm formation and arterial dissection or rupture. The median survival age in patients with vEDS is 51 years and vascular complication is the most common cause of death [3]. The present case report presents a man with vEDS and acute coronary syndrome who developed fatal cardiovascular complications. There are limited data on the best therapeutic strategy in the literature.

Case Report

A 43-year-old Italian man was admitted to the emergency department for new-onset resting chest pain.

The patient was known to have hypertension in optimal drug therapy and vEDS diagnosed by molecular testing. His father had died of sudden death; his brother was also affected by vEDS and ischemic heart disease and survived after splenic artery rupture.

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The 12-lead electrocardiography recorded sinus rhythm and ST-segment elevation in anterolateral leads (Figure 1). His blood pressure was 100/ 60 mmHg and he had weak femoral artery pulse. Echocardiography showed reduced left ventricular ejection fraction (LVEF=45%) and hypokinesia of the apex, anterior and anterolateral walls. The RV size was normal with no pericardial effusion. The aortic root and arch were also explored which were normal. Ticagrelor 180 mg, Flectadol 250 mg, and LMWH 5000 UI were administered.

The patient was transferred to our Hospital, a medical institution without a cardiac surgery room. The differential diagnosis for this patient included other plausible causes of chest pain, including myocarditis, valvular disease, and vascular disease.

Management

The patient with severe chest pain and persistent ST elevation was urgently transferred to the cardiac catheterization laboratory where he underwent coronary angiography, through radial artery access using 5 and 6 French

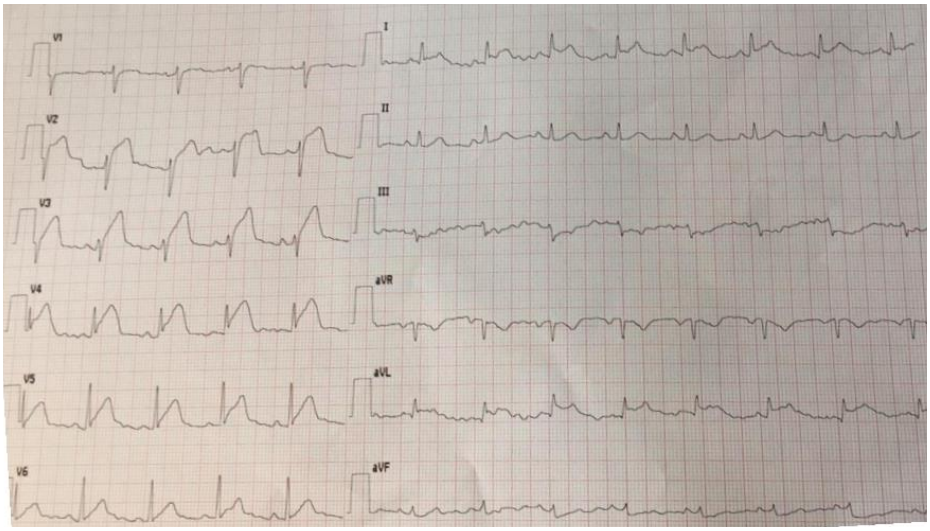

Figure 1. The 12-lead electrocardiography shows ST-segment elevation in the anterolateral leads

catheters.

Invasive coronary angiography (ICA) revealed right coronary (RC) without significant stenosis (Video 1) and type 1 spontaneous coronary artery dissection (SCAD) of left main (LM) and left anterior descending (LAD), (Video 2). Stenting of LM and LAD was performed with 3.5/22 mm drug-eluting stent (Resolute Onyx), (Video 3). After PCI, the angiogram showed proximal LCx's dissection which was treated with placement of stent 3.0/15 mm (Resolute Onyx, tap technique), (Video 4-5). The next angiogram showed well-apposed stents but likely coronary dissection of LAD and the first diagonal branch (Video 6). A drug-eluting stent (2.75/15 mm-Resolute Onyx) was implanted in mid-LAD (Video 7) causing immediate rupture of the coronary artery (Video 8). The patient showed psychomotor agitation due to strong chest pain during stent implantation and we accidentally implanted the stent in mid-distal LAD rather than in mid-proximal LAD. Immediately after the coronary rupture, the cannulation of the left coronary artery was lost.

The patient quickly developed unstable hemodynamic status with echocardiographic

signs of cardiac tamponade and he underwent emergency orotracheal intubation. In the meanwhile, pericardiocentesis (PCC) was performed. Next, we attempted unsuccessful cannulation of the left coronary artery to implant a covered stent. Immediately, aortic angiography showed aortic dissection (Video 9). Furthermore, echocardiography revealed a severe reduction of left ventricular ejection fraction (LVEF=35%) and a pericardial effusion which was circumferential with RV collapse in early diastole (Video 10). Pending transfer to a hospital with cardiac surgery room, PCC was continued but the patient died of cardiac arrest. The autopsy was not performed after the patient expired.

Discussion

Ehlers-Danlos syndrome (EDS) is an inherited genetic disorder of the connective tissues. The vascular subtype is characterized by vascular fragility causing arterial aneurysm and dissection [1-4]. In particular, SCAD results from the development of a false lumen, generally in the outer third of tunica media [4,5]. It is a rare cause of myocardial infarction (MI) and most frequently involves LAD [6] and dissection of the proximal vessel is rare [7]. Acute management depends on several factors; a conservative approach should be considered in hemodynamically stable patients with maintained distal flow in the culprit coronary and without demonstrable ongoing ischemia since coronary revascularization is associated with an increased risk of complications and adverse outcomes [8]. The interventional approach should be considered in all other cases, such as our case, where revascularization is necessary, though further research is required to clarify the optimal strategy. The present clinical case concerns the management of patients with acute coronary syndrome (ACS) and concomitant vEDS. Management is either surgical or endovascular. The literature review shows how surgery and endovascular interventions are both associated with an increased risk of complications [9]. We opted for an endovascular intervention for three reasons. First, the patient showed ACS-STEMI which is an emergency condition; second, aortic dissection had been excluded, although only by ultrasound; third, our hospital did not have a cardiac surgery room. The present clinical case is novel; myocardial infarction occurred due to coronary dissection, yet the patient also developed other vascular complications. The aortic dissection could be either the result of coronary dissection or an iatrogenic event (manipulation of catheters during ICA in a patient with vascular fragility), though it might have been present at the beginning. However, aortic dissection was excluded by 2D echocardiography (the pericardial effusion was absent and the aortic root and arch were regular in size). Before ICA, computed tomography (CT) scan could allow us to understand the order of events. Since the patient had been transferred from Spoke Center, which is about 1 hour away from our institution, and for his clinical presentation (patient with severe chest pain and persistent ST elevation), a CT scan was not performed due to the priority of myocardial revascularization. The limitations of this clinical case include the choice not to perform a baseline CT scan, and the impossibility of performing OCT or/and IVUS (patient with hemodynamically unstable with rapid clinical deterioration).

Conclusion

The management of vEDS patients diagnosed with ACS is very complex. Our clinical case shows a higher risk of vascular complications in these patients; thereby, it is safer to immediately transfer them to a hospital with a cardiac surgery room for better management of any complications. Non-invasive methods, such as a baseline CT scan, could be considered to exclude the dissection of other vessels and possibly change the treatment plan before ICA.

Summary points

- Patients with vascular EDS have a high risk of vascular complications.
- In patients with vascular EDS, myocardial infarction may present as coronary

dissection associated with the dissection of other blood vessels.
- Before coronary angiography, the baseline CT could rule out concomitant vascular dissections.
- Vascular complications could also occur due to endovascular procedures (iatrogenic complications) due to vascular fragility.
- Hospitalization in institutions with a cardiac surgery room is safer for better management of patients with vEDS.

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Disclosures

None. The authors have reported that they have no relationship regarding the contents of this paper to disclose and they have no funding.

Consent

The consent was obtained from the patient's family.

References

1. Malfait F, Francomano C, Byers P, Belmont J, Berglund B, Black J, et al. The 2017 international classification of the Ehlers-Danlos syndromes. Am J Med Genet C Semin Med Genet 2017; 175(1): 8-26.
2. Jorgensen A, Fagerheim T, Rand-Hendriksen S, Lunde PI, Vorren TO, Pepin MG, et al. Vascular Enhlers-Danlors Syndrome in siblings with biallelic COL3A1 sequence variants and marked clinical variability in the extended family. Eur J Hum Genet 2015; 23(6): 796-802.
3. Frank M, Adham S, Seigle S, Legrand A, Mirault T, Henneton P, et al. Vascular Ehlers-Danlors Syndrome. Long term observational Study. J Am Coll Cardiol 2019; 73(15): 1948-57.
4. Pepin M, Schwarze U, Superti-Furga A, Byers PH. A Clinical and genetic features of Ehlers-Danlos syndrome type IV, the vascular type. N Engl J Med 2000; 342(10): 673–80.
5. Adlam D, Alfonso F, Maas A, Vrints C, Committee W. European society of cardiology, acute cardiovascular care association, SCAD study group: a position paper on spontaneous coronary artery dissection. Eur Heart J 2018; 39(36): 3353-68.
6. Tweet MS, Hayes SN, Pitta SR, Simari RD, Lerman A, Lennon RJ, et al. Clinical features, management, and prognosis of spontaneous coronary artery dissection. Circulation 2012; 126(5): 579-88.
7. Hayes SN, Kim ES, Saw J, Adlam D, Arslanian-Engoren C, Economy KE, et al. Spontaneous coronary artery dissection: current state of the science: a scientific statement from the American Heart Association. Circulation 2018; 137(19): e523-57.
8. Tweet MS, Eleid MF, Best PJM, Lennon RJ, Lerman A, Rihal CS, et al. Spontaneous coronary artery dissection: revascularization versus conservative therapy. Circ Cardiovasc Interv 2014; 7(6) :777–86.
9. Bergqvist D, Bjorck M, Wanhainen A. Treatment of vascular Ehlers- Danlos syndrome: a systematic review. Ann Surg 2013; 258(2): 257-26.

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