Incidental asymptomatic artery dissection revealing vascular Ehlers-Danlos syndrome in a 52-year-old patient

Anica Milinković[1]*,
Mia Dubravčić Došen[1],
Petra Grubić Rotkvić[1],
Tomislav Krčmar[1,2,3],
Ivana Jurca[1],
Sanda Huljev Frković[1,4],
Majda Vrkić Kirhmajer[1,4]

[1]University Hospital Centre Zagreb, Zagreb, Croatia
[2]University Hospital Centre Rijeka, Rijeka, Croatia
[3]University of Rijeka, Faculty of Medicine, Rijeka, Croatia
[4]University of Zagreb, School of Medicine, Zagreb, Croatia

RECEIVED:
September 19, 2025
ACCEPTED:
October 6, 2025

KEYWORDS: arterial dissection, vascular Ehlers-Danlos syndrome, heritable connective tissue disorder.

CITATION: Cardiol Croat. 2025;20(9-10):248-9. | https://doi.org/10.15836/ccar2025.248

***ADDRESS FOR CORRESPONDENCE:** Anica Milinković, Klinički bolnički centar Zagreb, Kišpatićeva 12, HR-10000 Zagreb, Croatia. / Phone: +385-98-925-3461 / E-mail: anica.milinkovic@outlook.com

ORCID: Anica Milinković, https://orcid.org/0000-0002-3456-9540 • Mia Dubravčić-Došen, https://orcid.org/0000-0003-0441-4772
Petra Grubić Rotkvić, https://orcid.org/0000-0002-2587-1932 • Tomislav Krčmar, https://orcid.org/0000-0003-4689-1673
Ivana Jurca, https://orcid.org/0000-0002-0607-3361 • Sanda Huljev Frković, https://orcid.org/0000-0003-1513-2965
Majda Vrkić Kirhmajer, https://orcid.org/0000-0002-1340-1917

Introduction: Vascular Ehlers-Danlos syndrome (vEDS) is a rare genetic connective tissue disorder caused by pathogenic variants in COL3A1, a gene encoding type III collagen. Consequent tissue fragility manifests in a specific clinical phenotype, as well as possible life-threatening complications such as spontaneous arterial dissection or rupture, bowel perforation, and uterine rupture. Initial presentation typically occurs in early adulthood, whereas median life expectancy is estimated at 51 years[1]. While diagnosis may be guided by clinical criteria, molecular confirmation is required[2].

Case report: A 52-year-old man with arterial hypertension and bilateral renal cysts was incidentally diagnosed with a left external iliac artery dissection during a routine computed tomography (CT) scan (**Figure 1**). He reported no inguinal pain and showed no signs of limb ischemia. Further assessment revealed a family history of vascular events: his father died at 55 from a ruptured aortic aneurysm, and his paternal grandfather experienced sudden death at 50. Physical examination showed subtle features suggestive of vEDS, including micrognathia, keloids, varicose veins, and flat feet with piezogenic papules. A subsequent CT scan revealed bilateral saccular aneurysms at the renal artery bifurcations (**Figure 2**), in addition to the previously identified iliac artery dissection. Next-generation sequencing identified a heterozygous COL3A1 missense mutation, p.Gly237Arg. This variant was classified as pathogenic, confirming a diagnosis of vEDS. The patient was started on celiprolol and irbesartan, as these medications have been shown to reduce the incidence of major arterial events in patients with vEDS[3]. Lifestyle modifications were advised, and cascade genetic testing was recommended for relatives.

Conclusion: Although most vEDS patients develop major arterial complications by the age of 40, disease onset is variable and may present later, as observed in our patient. Despite appropriate management, these patients remain at high risk of morbidity and mortality. This case emphasizes the importance of considering vEDS in asymptomatic adults with incidental vascular findings, subtle connective tissue signs, and a relevant family history, in order to enhance clinical awareness and diagnostic accuracy.

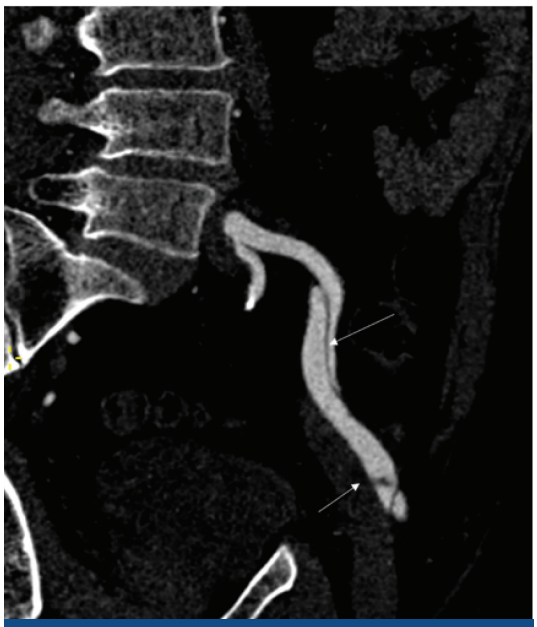

FIGURE 1. Contrast-enhanced CT angiography (maximum intensity projection reconstruction, oblique coronal plane) showing a dissected left external iliac artery with an intimal flap (white arrows).

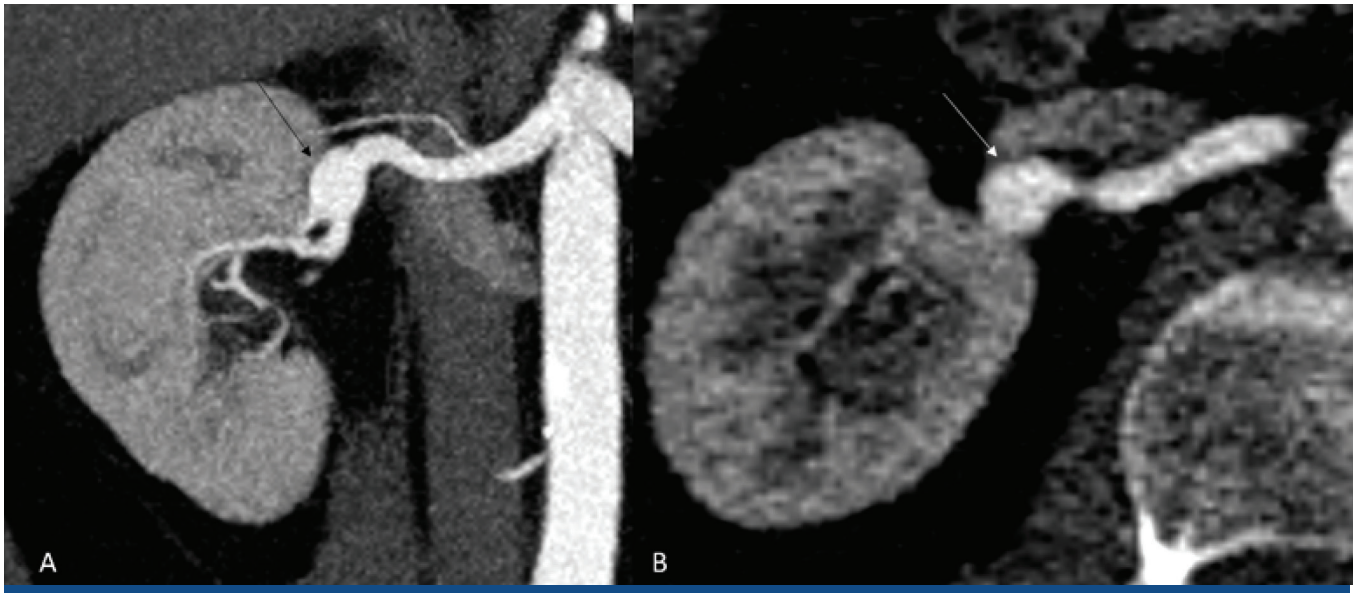

FIGURE 2. Contrast-enhanced CT angiography showing a right renal artery aneurysm. A) maximum intensity projection reconstruction (oblique coronal), black arrow. B) axial plane, white arrow.

LITERATURE

1. Pepin MG, Schwarze U, Rice KM, Liu M, Leistritz D, Byers PH. Survival is affected by mutation type and molecular mechanism in vascular Ehlers-Danlos syndrome (EDS type IV). Genet Med. 2014 Dec;16(12):881-8. https://doi.org/10.1038/gim.2014.72
2. 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 Mar;175(1):8-26. https://doi.org/10.1002/ajmg.c.31552
3. Jeunemaitre X, Mousseaux E, Frank M, Adham S, Pitocco F, Billon C, et al. Efficacy of Irbesartan in Celiprolol-Treated Patients With Vascular Ehlers-Danlos Syndrome. Circulation. 2025 Mar 11;151(10):686-695. https://doi.org/10.1161/CIRCULATIONAHA.124.072849