

Vascular Ehlers-Danlos syndrome – case report

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KEYWORDS: Ehlers-Danlos syndrome, celiprolol, collagen.

CITATION: *Cardiol Croat.* 2021;16(1-2):74. | <https://doi.org/10.15836/ccar2021.74>

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Introduction: Vascular Ehlers-Danlos syndrome (vEDS) is considered the most severe form of EDS because of its typical life-threatening complications in young adults: spontaneous rupture of arteries, uterus or intestine¹. The prevalence of vEDS is at least 1:200.000. It is associated with autosomal dominant mutation in COL3A1 gene, which encodes pro- α 1 chains of type III collagen. The median life expectancy is 48 to 51 years. The beneficial role of celiprolol in reduction of arterial complications has been described, and the mechanisms may be related to reduction of hemodynamic stress and by upregulation of collagen synthesis via transforming growth factor- β ^{2,3}.

Case report: A 26-year-old man was referred with long history of serious vascular complication. From early childhood he was prone to spontaneous bruising. At the age of 7, after minor trauma, he had a duodenal hematoma. Hemophilia was ruled out. In 2014, urgent nephrectomy was done because of spontaneous rupture of right renal artery. Calf varicose veins were treated with foam sclerotherapy in 2016. In 2017, urgent femoro-femoral bypass was performed due to spontaneous rupture of the left common iliac artery (CIA) and failure to repair extremely vulnerable CIA wall. Several months later hybrid vascular procedure was done due to spontaneous dissection of right external iliac artery. Early postoperative course was complicated by spontaneous pneumothorax. His physical appearance was also suggestive to vEDS: thin skin, characteristic facial appearance (thin lips, small chin, thin nose, prominent eyes with dark circles), small joints hypermobility. Molecular genetic testing in 2019 confirmed vEDS: our patient is heterozygous for COL3A1c.1149+2_1149+51del. His current medical therapy includes vitamin C and tolerable dose of celiprolol. For the last three years he is without new adverse vascular events.

Conclusion: Accurate diagnosis, genetic consulting, avoiding high risk activities and procedures are crucial in patients with vEDS. Endovascular or surgical intervention are mainly reserved for urgent complications of arterial or organ rupture. Celiprolol, a beta blocker with a unique pharmacologic profile, demonstrated a promising role in reduction of vascular complication in vEDS.

RECEIVED:
November 25, 2020

ACCEPTED:
December 18, 2020



LITERATURE

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