Supplementary material

Konieczyńska M, Wypasek E, Karpiński M, et al. Vascular Ehlers-Danlos syndrome in 2 Polish patients: identification of 2 novel COL3A1 gene mutations. Kardiol Pol. 2019; 77: 1070-1073. doi:10.33963/KP.15005

Table S1. Current diagnostic criteria for vEDS and clinical findings in the affected individuals

	C1	C2
• Inheritance	+	+
• Major criteria		
Family history of vEDS with documented causative variant in COL3A1		
Arterial rupture at young age	+	
Spontaneous sigmoid colon perforation in the absence of known diverticular disease or other bowel pathology	+	
Uterine rupture during the third trimester in the absence of previous C-section and/or severe peripartum perineum tears		
CCSF formation in the absence of trauma		
• Minor criteria		
Bruising unrelated to identified trauma and/or in unusual sites such as cheeks and back	+	+
Thin, translucent skin with increased venous visibility	+	+
Characteristic facial appearance	+	
Spontaneous pneumothorax		
Acrogeria		
Talipes equinovarus	+	
Congenital hip dislocation		
Hypermobility of small joints	+	+
Tendon and muscle rupture		
Keratoconus		
Gingival recession and gingival fragility	+	
Early onset varicose veins (under age 30 and nulliparous if female)		

Abbreviations: vEDS, vascular Ehlers-Danlos syndrome; CCSF, carotid-cavernous sinus Fistula