Simultaneous surgical treatment of an aortic root aneurysm and pectus excavatum in a child with Loeys-Dietz syndrome

Michał Buczyński[1], Karolina Szymczak[1], Jacek Kuźma[1], Maciej Karolczak[1], Michał Zawadzki[1], Paulina Kopacz[1], Mateusz Safijan[1], Mariusz Kuśmierczyk[2]

[1]Department of Cardiac and General Pediatric Surgery, Medical University of Warsaw, Warszawa, Poland
[2]Department of Cardiac Surgery, Medical University of Warsaw, Warszawa, Poland

Correspondence to:
Karolina Szymczak, MD, Department of Cardiac and General Pediatric Surgery, Medical University of Warsaw, Żwirki i Wigury 63A, 02-091 Warszawa, Poland, phone: +48 698 448 688
e-mail: karolina.szymczakk@gmail.com

DOI: 10.33963/KP.a2022.0156

Received:
May 9, 2022

Accepted:
June 21, 2022

Early publication date:
June 23, 2022

Loeys-Dietz syndrome is a rare genetic connective tissue condition, inherited in an autosomal dominant manner, caused by mutations in genes encoding components of the transforming growth factor β (TGF-β) signaling pathway [1].

It is characterized by multisystemic involvement [2] with vascular findings (mostly aortic aneurysms and/or dissections), skeletal manifestations (scoliosis, pectus excavatum, joint laxity, arachnodactyly), hypertelorism, bifid uvula or cleft palate, and also cutaneous abnormalities (easy bruising, translucent skin) [3, 4].

A 2-year-old male with Loeys-Dietz syndrome was admitted to the hospital due to a progressive aortic root aneurysm, patent arterial duct, and pectus excavatum. In medical history, the patient presented decreased muscle tone, problems with feeding, varus foot, and inguinal hernia. Diagnosis of the Loeys-Dietz syndrome was established at the age of 8 months after multidisciplinary consultations, while the aortic arch dilation was diagnosed on prenatal ultrasonography. Computed tomography angiography (CTA) showed the heart displaced into the left hemithorax, aortic valve (AV) diameter: 20.5 mm (z-score: +5.3), aortic root (AR): 37.5 mm (z-score: +7.5), and chest-wall deformity with a Haller index: 8.2. The minimal distance from the sternum to the vertebral column was 22.4 mm.

Transthoracic echocardiography (TTE) confirmed CTA findings and showed mild aortic valve regurgitation (grade I), narrow patent arterial duct with left-to-right shunt, and pressure gradient of 43 mm Hg.

The patient was qualified for simultaneous valve-sparing aortic root replacement (the Yacoub remodeling technique) [5] and correction of pectus excavatum with the Nuss procedure. Extracorporeal circulation was established via the brachiocephalic trunk and the right atrium. The excision of the coronary ostia and the dissection of the aortic root was performed. The cusps of the aortic valve were corrected, and the resected aortic root was replaced by a 20 mm Dacron tubular graft (FlowWeave BIOSEAL, JOTEC GmbH, Lotzenäcker, Germany). The prosthesis was trimmed to create three longitudinal extensions, replacing Valsalva sinuses. The coronary ostia were attached to the graft. At the end of the cardiac procedure, transesophageal echocardiography (TEE) confirmed a satisfactory hemodynamic effect. Thereafter, the Nuss procedure was used to correct the pectus excavatum. Two lateral incisions on either side of the chest on the midaxillary line were introduced, and an adequately curved titanium CHM 205 mm steel bar was inserted transversally before being rotated 180 degrees around the origin, thus elevating the sternum. Due to the small size of the patient, the steel bar was fixed with one stabilizer attached to the rib. Postoperative TTE showed normal aortic valve and prosthesis function with laminar flow.

To our knowledge, a combined correction of aortic root aneurysm and pectus excavatum in Loeys-Dietz syndrome has not been reported before. Surgical strategy presented here lowers the risk of perioperative complications when compared to two separate procedures, also reducing the total length of hospitalization. In 6-month follow-up, the patient was in good condition, on antihypertensive irbesartan. TTE showed normal function of the reconstructed aortic root and prosthesis.

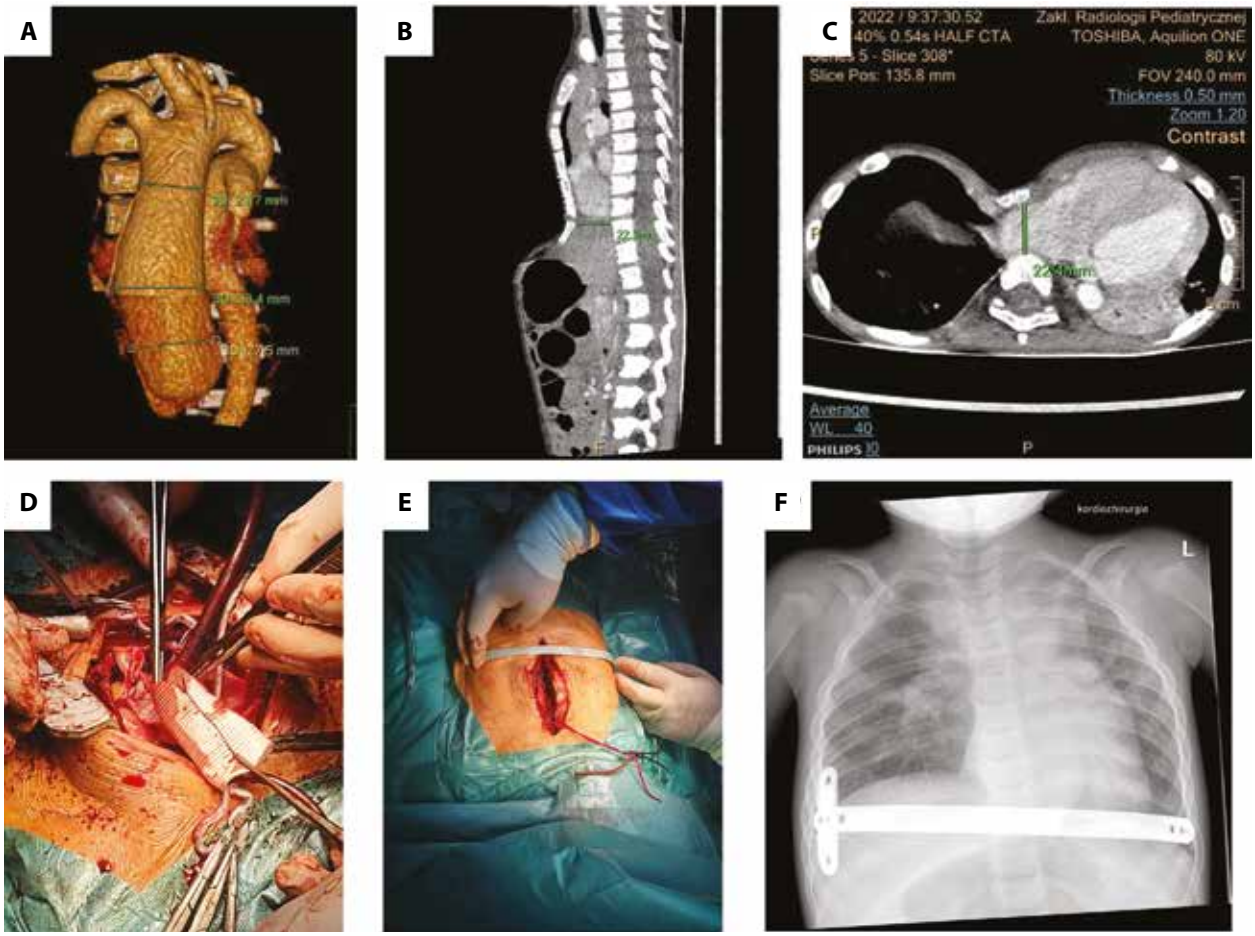

Figure 1. A. 3D imaging of the aorta with aortic root diameter. **B.** Computed tomography (CT) imaging of chest deformity — sagittal plane. **C.** CT imaging of chest deformity — the transverse plane. **D.** Dacron tubular graft **E.** A stainless steel bar adapted to deformation **F.** X-ray imaging after surgery showing the attached stainless steel bar

Article information

Conflict of interest: None declared.

Funding: None.

REFERENCES

1. Van Laer L, Dietz H, Loeys B. Loeys-Dietz syndrome. Adv Exp Med Biol. 2014; 802: 95–105, doi: 10.1007/978-94-007-7893-1_7, indexed in Pubmed: 24443023.
2. Erkula G, Sponseller PD, Paulsen LC, et al. Musculoskeletal findings of Loeys-Dietz syndrome. J Bone Joint Surg Am. 2010; 92(9): 1876–1883, doi: 10.2106/JBJS.I.01140, indexed in Pubmed: 20686062.
3. Loeys BL, Dietz HC, et al. Loeys-Dietz syndrome. In: GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2022. 2008 Feb 28 [updated 2018 Mar 1], indexed in Pubmed: 20301312.
4. Loeys BL, Schwarze U, Holm T, et al. Aneurysm syndromes caused by mutations in the TGF-beta receptor. N Engl J Med. 2006; 355(8): 788–798, doi: 10.1056/NEJMoa055695, indexed in Pubmed: 16928994.
5. Gocoł R, Bis J, Malinowski M, et al. Outcome comparison of different approaches to aortic root aneurysm. Kardiol Pol. 2022; 80(4): 436–444, doi: 10.33963/KP.a2022.0045, indexed in Pubmed: 35152397.