THE ANATOLIAN JOURNAL OF CARDIOLOGY

Official journal of the TURKISH SOCIETY OF CARDIOLOGY

Long-Term Follow-Up of a Patient with Arterial Tortuosity Syndrome After Bilateral Pulmonary Artery Stent Implantation

CASE REPORT

INTRODUCTION

Arterial tortuosity syndrome (ATS) is a rare autosomal recessive connective tissue disorder characterized by widespread tortuosity and elongation of the large and mid-sized arteries as well as phenotypic manifestations such as hyperextensible skin, joint hypermobility, inguinal hernia, micrognathia, convex nasal ridge, and downslanting palpebral fissures.[1]

Even though the main feature of ATS is diffuse arterial tortuosity, cardiovascular system involvement varies between different cohorts. The prevalence of pulmonary artery stenosis ranges between 28% and 60% among different cohort studies and may be accompanied by pulmonary hypertension due to peripheral pulmonary artery stenoses.[2,3] Treatment options for these stenoses include surgery, transcatheter pulmonary stenting, and hybrid procedures.

Most of the patients are diagnosed in childhood, and reconstruction surgery of pulmonary arteries during childhood is the most commonly performed treatment option among those patients requiring pulmonary intervention.[4] In this article, an adult patient with ATS and long-term follow-up results of bilateral pulmonary artery stenting are presented.

CASE REPORT

A 27-year-old patient with ATS was referred to the clinic 8 years ago for clinical follow-up due to pulmonary hypertension. The patient was diagnosed with ATS due to typical phenotypic features and radiological findings at the age of 11 years. The patient had typical dysmorphic features for ATS such as long face, beaked nose, downslanting palpebral fissures, high palate, micrognathia, hyperextensible skin, and hypermobile joints. The patient was born from a third-degree consanguineous marriage. Family screening with computed tomography (CT) angiography also showed the presence of arterial tortuosity in the patient's father and sibling. The family's diagnosis of ATS was later reestablished with genetic testing during the patient's pediatric follow-up.

Patient was not under any medication at the time of admission to the clinic, and he had shortness of breath during ordinary daily activities. He was reevaluated with echocardiography and pulmonary CT angiography. Echocardiography revealed dilated right heart chambers with severe tricuspid regurgitation. Pulmonary CT angiography revealed dilated main pulmonary artery and bilateral peripheral pulmonary artery tortuosity with focal stenoses (Figure 1).

Patient then underwent right heart catheterization (RHC) and selective pulmonary angiography for stenoses of the left pulmonary artery in May 2018. Right heart catheterization was consistent with precapillary pulmonary hypertension. Mean pulmonary artery pressure (PAP) was 80 mm Hg and pulmonary vascular resistance (PVR) was 20.9 woods unit (WU) with pulmonary capillary wedge pressure (PCWP) of 8 mm Hg (Table 1). Pulmonary angiography revealed critical ostial stenoses in the segments A3, A4, A5, and A6 (Figure 2, Videos 1 and 2). Lesions

Çağan Yıldırım
Halil Ataş

Department of Cardiology, Marmara University Faculty of Medicine, İstanbul, Türkiye

Corresponding author:
Çağan Yıldırım
caganyildirim94@hotmail.com

Available Online Date: August 11, 2026

Cite this article as: Yıldırım Ç, Ataş H. Long-term follow-up of a patient with arterial tortuosity syndrome after bilateral pulmonary artery stent implantation. *Anatol J Cardiol.* 2026;XX(X):1-7.

DOI: 10.14744/AnatolJCardiol.2026.6686

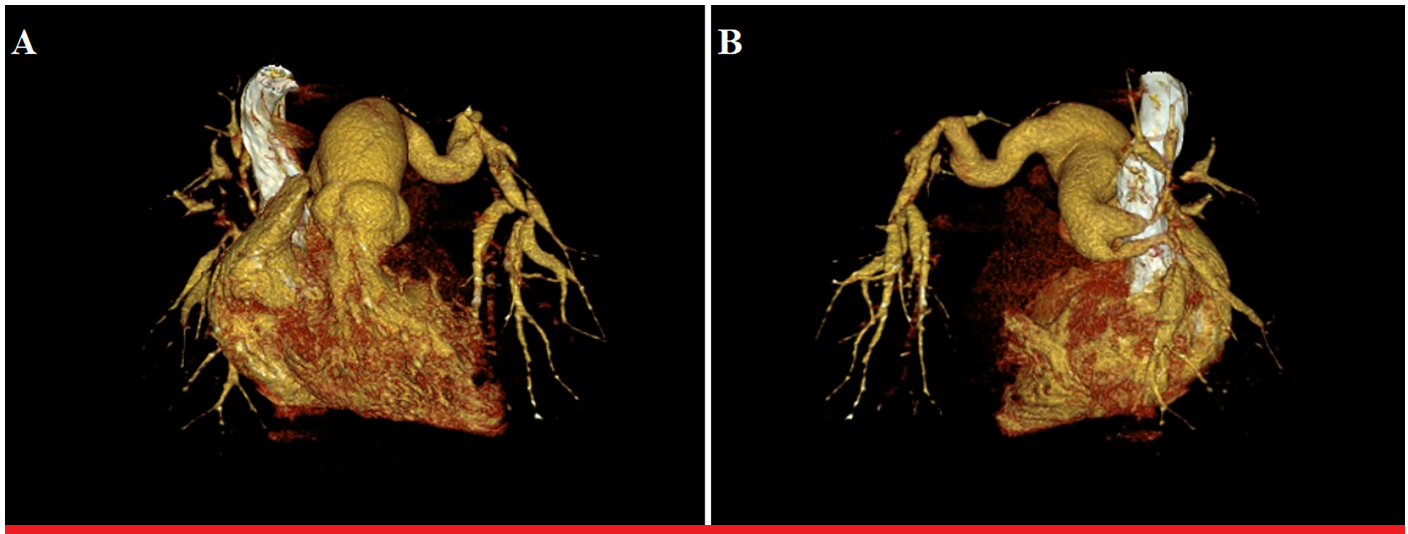

Figure 1. (A) Anterior view of pulmonary artery with computed tomography angiography with three-dimensional (3D) volume rendering technique. (B) Posterior view of pulmonary artery with computed tomography angiography with 3D volume rendering technique.

were evaluated with fractional flow reserve which showed significant obstruction, and percutaneous pulmonary stent implantation was planned.

Patient underwent percutaneous pulmonary stent implantation for left-sided lesions in July 2018, and total revascularization was achieved with 4 different renal stents (Abbott's RX Herculink Elite (®) Renal Stent, Chicago, IL, USA) (Figure 3, Video 3). Dual antiplatelet therapy with aspirin and clopidogrel was established. Patient developed lung edema and fever accompanied by hemoptysis during follow-up after the procedure. Control chest CT showed consolidation in the left lung, which was considered a reperfusion injury (Figure 4). Patient was treated with furosemide and antibiotics. Symptoms resolved after 2 days, and the patient was discharged.

Control RHC and selective pulmonary angiography for right-sided stenoses were planned after completion of the aforementioned first procedure. Pulmonary angiography revealed stenoses of 80%-90% in both the right upper and right lower lobar arteries (Figure 5, Video 4). The patient underwent percutaneous pulmonary stent implantation for right-sided lesions in January of 2019, and total revascularization was achieved with the deployment of self-expanding stents (Abbott's Supera Peripheral Stent, Santa Clara, CA, USA) in each lobar artery (Figure 6, Video 5). Follow-up treatment was established with dual antiplatelet therapy as in the previous procedure, with the addition of sildenafil, and the patient was discharged without any adverse event.

Patient was mostly asymptomatic during follow-up, with occasional mild exertional dyspnea. Control RHC at the first year showed a substantial decrease in pulmonary pressures, with a mean PAP of 37 mm Hg and corresponding PVR of 4.4 WU (Table 1). Most recent RHC was performed

Table 1. Serial Right Heart Catheterization Measurements During Follow-Up

	May 2018: Before First Procedure	Sep. 2018: After First Procedure	Jan. 2019: Before Second Procedure	Sep. 2019: After Second Procedure	March 2022: Long-Term Follow-up
Systolic PAP (mm Hg)	119	101	97	74	82
Diastolic PAP (mm Hg)	48	40	25	19	29
Mean PAP (mm Hg)	84	58	60	37	49
RAP (mm Hg)	12	8	12	8	4
PCWP (mm Hg)	8	8	12	10	7
PVR (WU)	20.39	9	7.45	4.4	6.75
SVR (WU)	21.3	24	16.15	17.69	19.08
PVR/SVR	0.95	0.37	0.46	0.24	0.35
CO (L/min)	5.1	5.71	6.44	5.54	6.08
CI (L/min/m^2)	2.22	2.58	2.76	2.37	2.61
6MWT (m)	390	520	515	500	450

6MWT, six-minute walking test; CI, cardiac index; CO, cardiac output; m, meters; PAP, pulmonary artery pressure; PCWP, pulmonary capillary wedge pressure; PVR, pulmonary vascular resistance; RAP, right atrial pressure; SVR, systemic vascular resistance; WU, Woods unit.

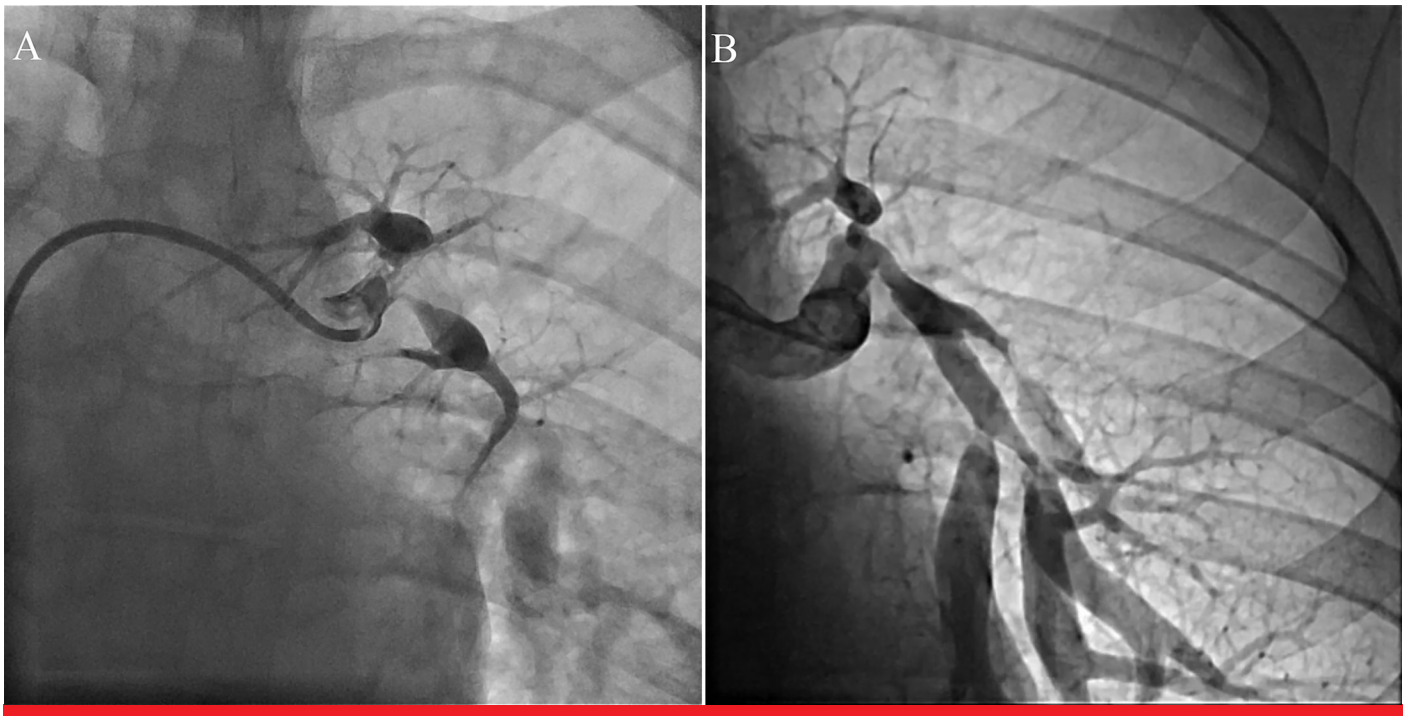

Figure 2. (A) Pulmonary artery stenoses in left A3 and A4 segments shown with selective pulmonary angiography. (B) Pulmonary artery stenoses in left A5 and A6 segments shown with selective pulmonary angiography.

in March 2022. RHC measurements showed slight increases in both pulmonary pressures and PVR compared to first-year control. Follow-up RHC measurements are shown in Table 1. Control selective pulmonary angiography revealed in-stent restenosis at left-sided renal stents (Figure 7, Video 6) while right-sided stents were found to be retaining their patency (Figure 7, Video 7). Medical treatment was rearranged with riociguat monotherapy in addition to single antiplatelet treatment with aspirin. Periodic outpatient controls have been carried out for the last 8 years without any rehospitalization.

Patient Consent

A written informed consent was obtained from the patient. The patient consented to publish case details and imaging data without personal images.

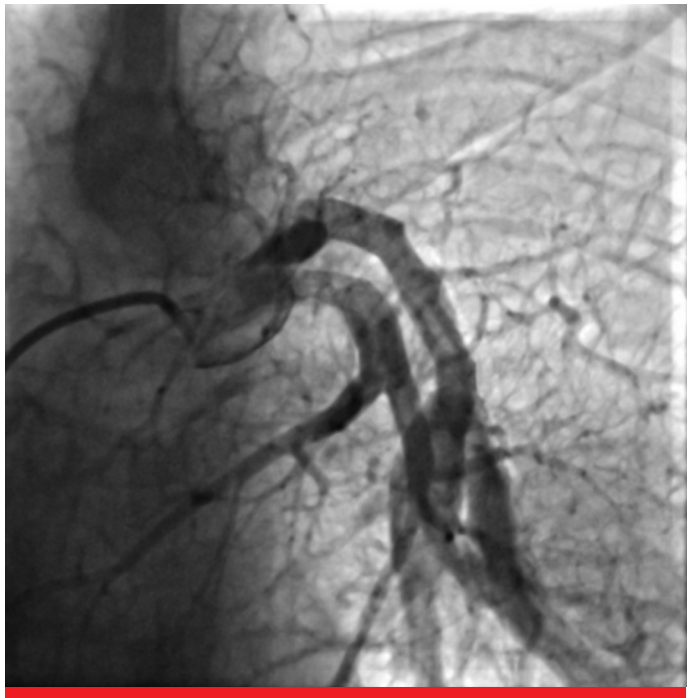

Figure 3. Selective pulmonary angiography of left pulmonary artery after stent implantation.

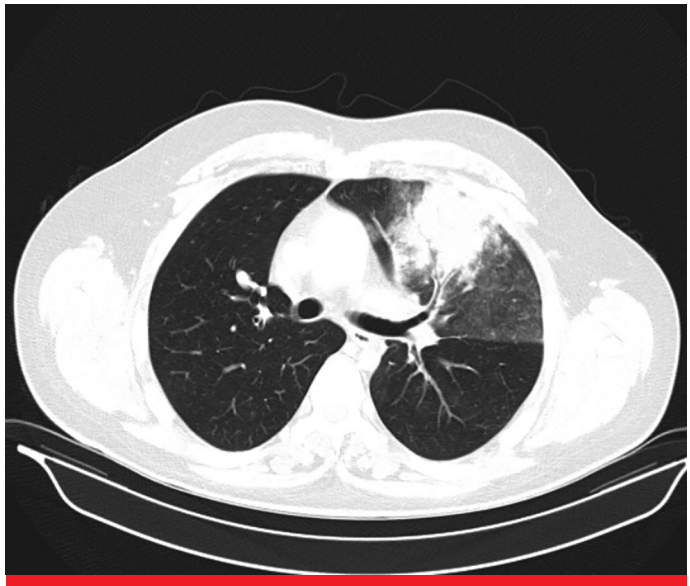

Figure 4. Computed tomography image of reperfusion injury in left lung after pulmonary stent implantation.

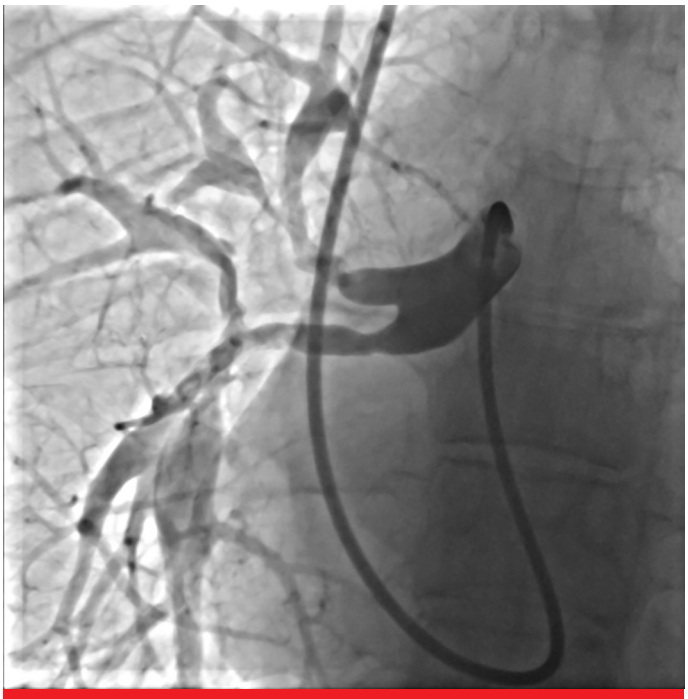

Figure 5. Pulmonary artery stenoses in right upper and right lower lobar arteries shown with selective pulmonary angiography.

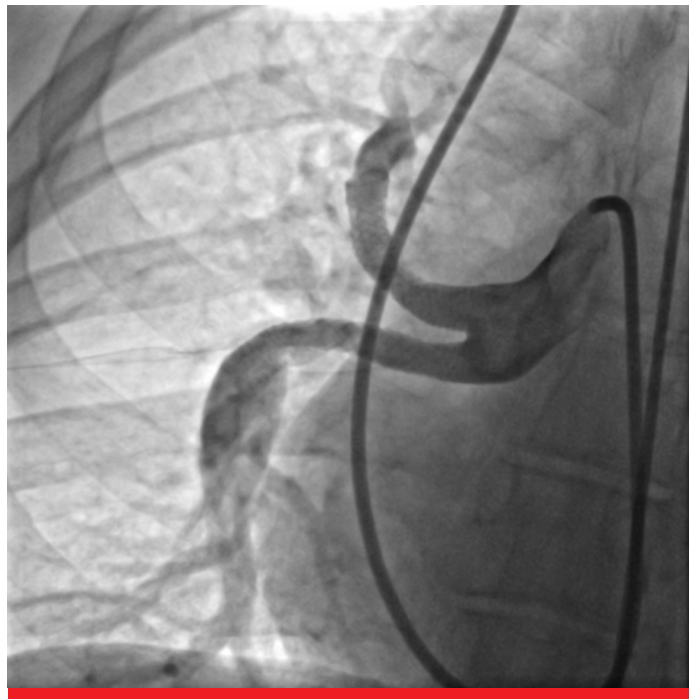

Figure 6. Selective pulmonary angiography of right pulmonary artery after stent implantation.

DISCUSSION

First case of diffuse tortuosity of the arteries as a congenital defect was reported by Ertugrul in 1967.[5] Arterial tortuosity syndrome got recognized as a different disorder over the years with genetic testing in addition to characteristic imaging findings. The diagnosis is established with molecular genetic testing by showing biallelic pathogenic variants in the solute carrier family 2 member 10 (*SLC2A10*) gene in addition to generalized arterial tortuosity.[3] Although most patients get diagnosed at childhood, adult presentations have also been reported.[6,7]

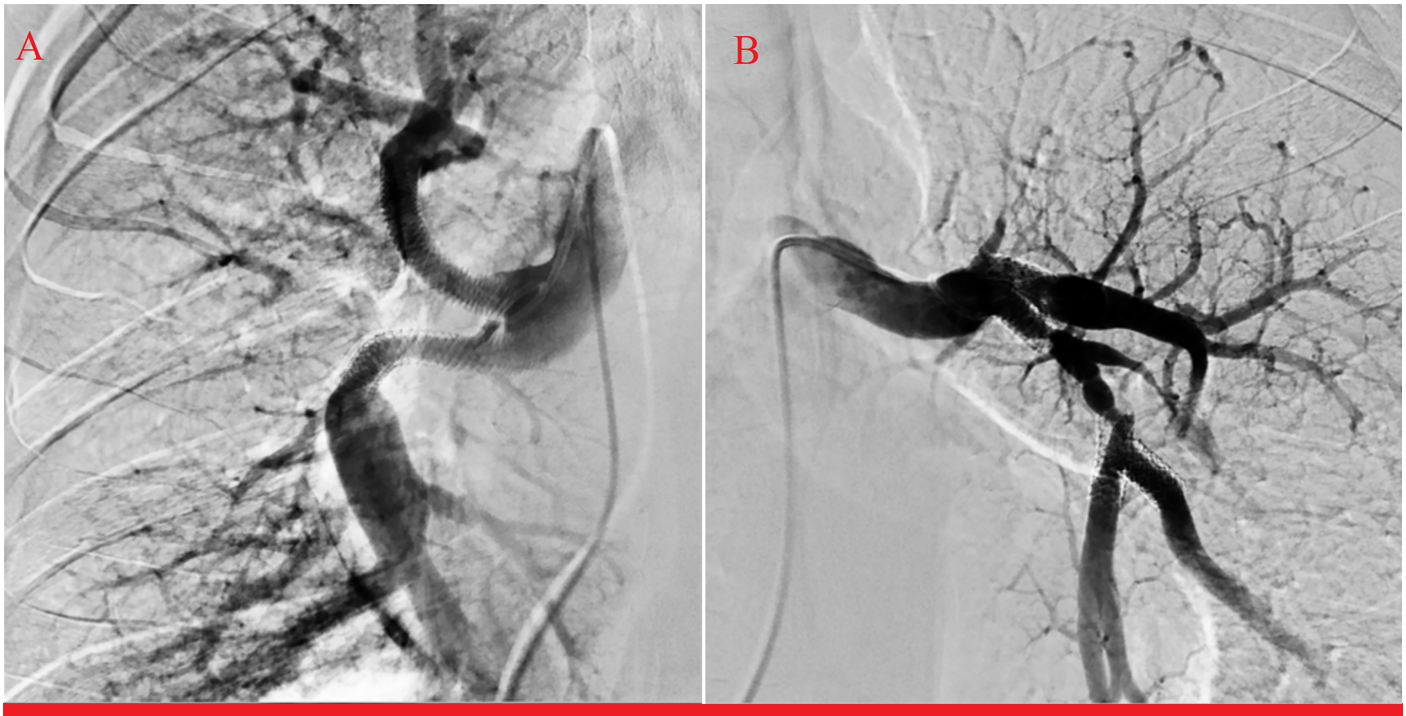

Figure 7. (A) Digital subtraction angiography showing patent stents in right pulmonary artery at 4th year control. (B) Digital subtraction angiography showing in-stent restenosis in left pulmonary artery stents at 4th year control.

Underlying mechanism was suggested as fragmentation of the inner elastic membrane and elastic fibers of the tunica media of large arteries due to loss of function of the *SLC2A10* gene that encodes the facilitative glucose transporter GLUT 10.[1,8] Consequently, GLUT 10 deficiency disrupts the transforming growth factor-beta pathway, leading to vessel wall impairment.[6]

Cardiovascular system involvement is the main source of morbidity and mortality among ATS patients.[9] Arterial tortuosity impairs blood circulation and increases vascular shear stress that may lead to stenosis, atherosclerosis, or dissection in affected arterial territories, as well as ventricular failure due to increased afterload.[4] Earlier studies have shown poor prognosis in childhood, with a mortality rate of 40%.[2] However, more recent studies have shown more favorable outcomes.[1]

Most common cardiovascular findings are tortuosity of the large arteries including aorta and pulmonary arteries. Arterial tortuosity syndrome is also a rare cause of pulmonary hypertension among connective tissue disorders.[10] Kinks and loops of the pulmonary artery cause pulmonary artery stenosis leading to pulmonary hypertension. Computed tomography signs include a V-shaped sign on the coronal plane and an inverted V-shaped sign on the axial plane, which was also present in the case.[6,11]

Stent implantation has been shown to be a successful choice of intervention for the treatment of pulmonary artery stenosis.[12-14] However, the patient population for the given intervention is heterogeneous and includes various complex defects with different anatomies accompanied by different physiologies such as single or biventricular circulation. Ing et al[15] have shown the immediate success rate of stent implantation to be 98% in patients with biventricular physiologies. Additionally, adult patients make up only a small fraction of pulmonary intervention populations with prevalence ranging from 11.8% to 13%.[13,14]

Clinically significant pulmonary artery stenoses of ATS patients are mostly treated with reconstruction surgery during childhood.[4,16] However, there is not any study regarding percutaneous pulmonary artery stenting in ATS patients. Lack of study on this topic can be potentially attributed to preexisting concerns such as future requirement of stent redilation to accommodate somatic growth of children and risk of pushing tortuosity distally via stenting due to redundancy of pulmonary artery. Aliramezany et al[17] have reported a case of pulmonary balloon angioplasty that decreased pulmonary artery pressure from 120 mm Hg to 60 mm Hg in an adult ATS patient who was deemed unsuitable for stenting due to vascular sizing. Santoro et al[18] have reported a hybrid transcatheter-surgical strategy in which stents were implanted through a stab incision of the main pulmonary artery via sternotomy followed by reductive surgical reconstruction to eliminate redundancy of pulmonary artery. Hereby, the long-term outcome of percutaneous pulmonary artery stenting in an adult ATS patient is presented.

Sildenafil and riociguat have been shown to improve exercise capacity, symptoms, and hemodynamics in pulmonary arterial hypertension (PAH) patients.[19,20] Additionally, switching from sildenafil to riociguat was found to further improve exercise capacity in intermediate-risk PAH patients.[21] Furthermore, PAH drugs have been shown to improve hemodynamics in peripheral pulmonary stenosis patients, whether or not additional percutaneous pulmonary angioplasty was performed. On the other hand, combining both interventional treatment and medical therapy was found to be associated with greater hemodynamic improvement.[22] Pulmonary stenting was considered to be the main factor for the clinical improvement of the patient, given the severity of lesions and pulmonary hypertension. However, differentiating the benefits of interventional and medical therapies is not possible because both treatment options were implemented in the patient.

Pulmonary reperfusion syndrome due to increased flow after pulmonary stent implantation has also been described as in the case.[23] Treatment of this condition includes diuresis, keeping hemoglobin at normal levels with close observation for pulmonary hemorrhage and mechanical ventilation with positive end-expiratory pressure, as well as extracorporeal membrane oxygenation in refractory cases.[4,16]

Neointimal proliferation and restenosis were found between 1.8% and 4% of pulmonary artery stent patients at repeat catheterization during follow-up.[24,25] Gonzalez et al[26] have reported different rates of neointimal proliferation and restenosis between different stent types after pulmonary artery stenting. Although lesion and stent lengths differ between the stent types, more significant restenosis was found in Abbott's RX Herculink Elite renal stents compared to Abbott's Supera peripheral stents after pulmonary artery stenting.

CONCLUSION

Stent implantation is a safe and successful choice of intervention for the treatment of pulmonary artery stenosis in adult ATS patients with preserved stent patency and maintained hemodynamic status at long-term follow-up. Dedicated stents for pulmonary artery may prevent neointimal proliferation and restenosis, hence recurrent interventions.

Informed Consent: Written informed consent was obtained from the patient.

Declaration of Interests: The authors have no conflicts of interest to declare.

Funding: The authors declare that this study received no financial support.

Video 1: Pulmonary artery stenoses in left A3 and A4 segments shown with selective pulmonary angiography.

Video 2: Pulmonary artery stenoses in left A5 and A6 segments shown with selective pulmonary angiography.

Video 3: Selective pulmonary angiography of left pulmonary artery after stent implantation.

Video 4: Pulmonary artery stenoses in right upper and right lower lobar arteries shown with selective pulmonary angiography.

Video 5: Selective pulmonary angiography of right pulmonary artery after stent implantation.

Video 6: Digital subtraction angiography showing patent stents in right pulmonary artery at 4th year control.

Video 7: Digital subtraction angiography showing in-stent restenosis in left pulmonary artery stents at 4th year control.

REFERENCES

1. Callewaert BL, Willaert A, Kerstjens-Frederikse WS, et al. Arterial tortuosity syndrome: clinical and molecular findings in 12 newly identified families. *Hum Mutat*. 2008;29(1):150-158. [CrossRef]
2. Wessels MW, Catsman-Berrevoets CE, Mancini GMS, et al. Three new families with arterial tortuosity syndrome. *Am J Med Genet A*. 2004;131(2):134-143. [CrossRef]
3. Beyens A, Albuisson J, Boel A, et al. Arterial tortuosity syndrome: 40 new families and literature review. *Genet Med*. 2018;20(10):1236-1245. [CrossRef]
4. Alshair FM, Alsulami AS, Shihata MS, et al. Total pulmonary arterial reconstruction in a patient with arterial tortuosity syndrome affecting the pulmonary artery: a case report and review of the literature. *J Cardiothorac Surg*. 2024;19(1):432. [CrossRef]
5. Ertugrul A. Diffuse tortuosity and lengthening of the arteries. *Circulation*. 1967;36(3):400-407. [CrossRef]
6. Elnaggar ME, Aly MM, Abduljawad H, et al. A rare case of arterial tortuosity syndrome in an adult. *Radiol Case Rep*. 2022;17(8):2594-2597. [CrossRef]
7. Castori M, Ritelli M, Zoppi N, et al. Adult presentation of arterial tortuosity syndrome in a 51-year-old woman with a novel homozygous c.1411+1G>A mutation in the SLC2A10 gene. *Am J Med Genet A*. 2012;158A(5):1164-1169. [CrossRef]
8. Coucke PJ, Willaert A, Wessels MW, et al. Mutations in the facilitative glucose transporter GLUT10 alter angiogenesis and cause arterial tortuosity syndrome. *Nat Genet*. 2006;38(4):452-457. [CrossRef]
9. Ekhator C, Devi M, Barker C, et al. Arterial tortuosity syndrome: unraveling a rare vascular disorder. *Cureus*. 2023;15(9):e44906. [CrossRef]
10. Sarı A, Temiz Karadağ D, Erdoğan M, et al. Screening for pulmonary hypertension in connective tissue diseases: literature review and multidisciplinary consensus statement. *Anatol J Cardiol*. 2025;29(11):599-607. [CrossRef]
11. Bhat V. Arterial tortuosity syndrome: an approach through imaging perspective. *J Clin Imaging Sci*. 2014;4:44. [CrossRef]
12. Holzer RJ, Gauvreau K, Kreutzer J, et al. Balloon angioplasty and stenting of branch pulmonary arteries: adverse events and procedural characteristics: results of a multi-institutional registry. *Circ Cardiovasc Interv*. 2011;4(3):287-296. [CrossRef]
13. Moore JW, Vincent RN, Beekman RH, 3rd, et al. Procedural results and safety of common interventional procedures in congenital heart disease: initial report from the National cardiovascular Data Registry. *J Am Coll Cardiol*. 2014;64(23):2439-2451. [CrossRef]
14. Lewis MJ, Kennedy KF, Ginns J, et al. Procedural success and adverse events in pulmonary artery stenting: insights from the NCDR. *J Am Coll Cardiol*. 2016;67(11):1327-1335. [CrossRef]
15. Ing FF, Khan A, Kobayashi D, et al. Pulmonary artery stents in the recent era: immediate and intermediate follow-up. *Catheter Cardiovasc Interv*. 2014;84(7):1123-1130. [CrossRef]
16. Al-Khaldi A, Momenah T, Alsahari A, et al. Late outcomes after pulmonary arterial reconstruction in patients with arterial tortuosity syndrome. *Ann Thorac Surg*. 2022;113(5):1569-1574. [CrossRef]
17. Aliramezany M, Firouzi A, Parsaee M, et al. Successful pulmonary artery balloon angioplasty in a Rare case of arterial tortuosity syndrome. *Iran J Cardiovasc Med*. 2020;9(2):52-54.
18. Santoro G, Caianiello G, Rossi G, et al. Hybrid transcatheter-surgical strategy in arterial tortuosity syndrome. *Ann Thorac Surg*. 2008;86(5):1682-1684. [CrossRef]
19. Galiè N, Ghofrani HA, Torbicki A, et al. Sildenafil citrate therapy for pulmonary arterial hypertension. *N Engl J Med*. 2005;353(20):2148-2157. [CrossRef]
20. Ghofrani HA, Galiè N, Grimminger F, et al. Riociguat for the treatment of pulmonary arterial hypertension. *N Engl J Med*. 2013;369(4):330-340. [CrossRef]
21. Hoeper MM, Al-Hiti H, Benza RL, et al. Switching to Riociguat versus maintenance therapy with phosphodiesterase-5 inhibitors in patients with pulmonary arterial hypertension (REPLACE): a multicentre, open-label, randomised controlled trial. *Lancet Respir Med*. 2021;9(6):573-584. [CrossRef]
22. Tamura Y, Tamura Y, Shigeta A, et al. Adult-onset idiopathic peripheral pulmonary artery stenosis. *Eur Respir J*. 2023;62(6):2300763. [CrossRef]
23. Berenguer Potenciano M, Piris Borregas S, Mendoza Soto A, et al. Pulmonary reperfusion syndrome after pulmonary stent implants in a patient with vascular tortuosity syndrome. *An Pediatr (Barc)*. 2015;82(1):e17-e20. [CrossRef]
24. McMahon CJ, El-Said HG, Grifka RG, et al. Redilation of endovascular stents in congenital heart disease: factors implicated in the development of restenosis and neointimal proliferation. *J Am Coll Cardiol*. 2001;38(2):521-526. [CrossRef]
25. Law MA, Shamszad P, Nugent AW, et al. Pulmonary artery stents: long-term follow-up. *Catheter Cardiovasc Interv*. 2010;75(5):757-764. [CrossRef]
26. Gonzalez I, Kenny D, Slyder S, et al. Medium and long-term outcomes after bilateral pulmonary artery stenting in children and adults with congenital heart disease. *Pediatr Cardiol*. 2013;34(1):179-184. [CrossRef]