Obstruction of the Anomalous Pulmonary Venous Connection in Scimitar Syndrome: Progression to Spontaneous Anomalous Pulmonary Vein Atresia during Infancy without Intervention

Rachel E. Henning, MD, David E. Segar, MD, and Peter C. Frommelt, MD, *Milwaukee, Wisconsin*

INTRODUCTION

Scimitar, or pulmonary venolobar, syndrome is a rare form of congenital heart disease that is characterized by total or partial anomalous pulmonary venous return from the right lung into the inferior vena cava (IVC) with frequently associated right pulmonary hypoplasia. The term "scimitar syndrome" was first proposed in 1960 by Neill *et al.*[1] due to the frequently appreciated radiological shadow caused by the anomalous pulmonary vein on chest x-ray that resembles a Middle Eastern scimitar sword. Scimitar syndrome is also characterized by cardiac dextroposition (due to right lung hypoplasia), anomalous systemic arterial blood supply to the right lower lung, and bronchial abnormalities affecting the right lung.[2] We present a case of a neonate with scimitar syndrome and anomalous right pulmonary venous stenosis at the confluence as it drained into the IVC that progressed to spontaneous atresia during infancy without intervention.

CASE PRESENTATION

A 6-day-old female infant born at 37 weeks presented to the pediatric cardiology clinic at a large academic pediatric institution for evaluation of a heart murmur. At time of presentation she was asymptomatic, but her exam was notable for a 2/6 blowing long systolic murmur heard at the left upper sternal border with radiation to the left axilla and back. An echocardiogram was obtained and demonstrated a small restrictive anterior muscular ventricular septal defect (VSD) with low-velocity left-to-right shunting. The right pulmonary artery (RPA) was mildly hypoplastic (RPA measured 3.1 mm; *Z* score of −2.18) with diminished flow that was characterized by marked early systolic deceleration and retrograde diastolic flow (Video 1, Figure 1). Continuous turbulent anterograde flow to a good size left pulmonary artery (LPA) was appreciated by Doppler without imaging evidence of pathologic stenosis (LPA dimension of 5.9 mm; *Z* score of 1.05). Two left pulmonary veins drained normally into the left atrium (LA), but right pulmonary vein flow into the LA was not demonstrated, with turbulent continuous flow from an anomalous vessel draining into the right atrium (RA) at the IVC-RA junction (Figure 2, Video 2), suggestive of a scimitar vein that has restrictive flow as it enters the RA. There was a small atrial septal defect with bidirectional atrial-level shunting, although systemic oxygen saturations were normal. Due to the concerning RPA flow pattern and inability to clearly see normal right pulmonary venous drainage with the suggestion of a scimitar vein, a chest x-ray (Figure 3) was obtained that showed a hyperinflated left lung and a diffusely opacified right hemithorax with the cardiothymic silhouette shifted into the right chest, concerning for right lung hypoplasia.

Because of concerns about possible scimitar syndrome, a cardiac computed tomography was obtained at 10 days of life, which showed that all the right lung pulmonary veins drained anomalously into the posterior aspect of the IVC at the IVC-RA junction, with stenosis of the anomalous pulmonary venous confluence at its entrance (Figure 4). In addition, there was an RPA hypoplasia and a small anomalous abdominal aortopulmonary collateral arising from celiac truck and coursing to the inferior lobe of right lung. No bronchial anomalies were identified, but there were scattered areas of atelectasis in the right lung, while the left lung was clear, with no evidence of parenchymal lung disease. Subsequent cardiac catheterization at 20 days of life found a 10 mm Hg gradient from the anomalous venous confluence to the IVC with normal pulmonary artery (PA) pressures (mean, 17 mm Hg). The aortopulmonary collateral was very small, so it was not felt to need intervention. Given the patient was asymptomatic with estimated normal pulmonary pressures, the decision was made to follow conservatively without proceeding to high-risk and difficult anomalous vein baffling of the small atretic segment to the LA at such a young age. The likelihood of long-term baffle patency was felt to be low.

She thrived during serial follow-up exams throughout infancy with no clinical symptoms, excellent growth (weight increased from the 19th percentile to the 32nd percentile at 20 months of age; height remained consistent around the 17th percentile), and normal systemic oxygen saturations. Sedated echocardiograms at 2 and 6 months of age demonstrated continued patency of the anomalous vein with mean Doppler gradients of 5-10 mm Hg with normalized RPA flow patterns by spectral Doppler without diastolic flow reversal. Right ventricular (RV) size and function were also normal. There was no systolic septal flattening or notched PA Doppler patterns to suggest PA hypertension. There was only trivial tricuspid insufficiency, so a more accurate estimate of RV and PA pressures could not be made. However, her sedated echocardiogram at 12 months did not demonstrate anomalous pulmonary venous flow entering the IVC, and the RPA remained small (measuring 4.1 mm; *Z* score, −2.42) with recurrence of retrograde diastolic flow while the LPA continued to enlarge

From the Department of Pediatrics (R.E.H.) and Department of Pediatric Cardiology (D.E.S., P.C.F.), Medical College of Wisconsin, Milwaukee, Wisconsin.

Keywords: Scimitar, Echocardiography, Infant, Pulmonary vein obstruction

Conflicts of Interest: None.

Correspondence: Peter C. Frommelt, MD, Children's Wisconsin, 9000 West Wisconsin, Mail station 713, Milwaukee, WI 53226 (E-mail: *pfrommelt@chw.org*).

2468-6441

https://doi.org/10.1016/j.case.2022.02.002

VIDEO HIGHLIGHTS

Video 1: Two-dimensional transthoracic echocardiogram, parasternal short-axis view, color flow Doppler display from the initial evaluation demonstrated diastolic reversal (*blue flow signal*) in the RPA and continuous anterograde turbulent flow in the LPA.

Video 2: Two-dimensional transthoracic echocardiogram, subcostal coronal window, color flow Doppler display from the initial evaluation demonstrated turbulent anomalous pulmonary venous flow draining into the posterior aspect of the IVC.

Video 3: Percutaneous femoral approach, right upper PA wedge angiography, at 1 year of age demonstrated markedly hypoplastic right pulmonary veins filling and coalescing into a single confluence with long-segment atresia.

View the video content online at www.cvcasejournal.com.

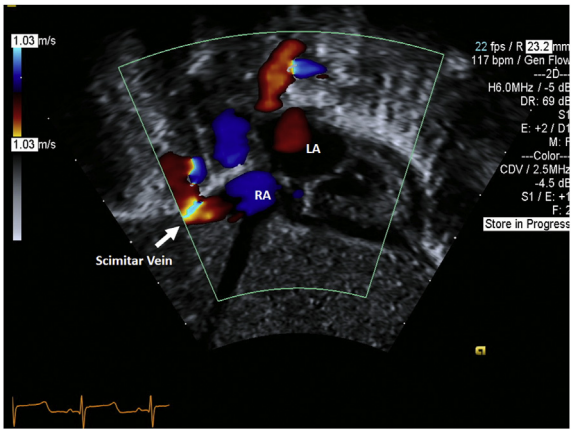

Figure 2 Two-dimensional transthoracic echocardiogram, subcostal coronal window, color flow Doppler display from the initial evaluation demonstrated the anomalous right pulmonary venous confluence draining into the RA at the IVC -RA junction with turbulent flow by color Doppler, suggestive of narrowing of the scimitar vein as it enters the RA (*arrow*).

(measuring 7 mm; *Z* score, 0.9). Again, there was good RV function without imaging evidence of pulmonary hypertension. Repeat cardiac catheterization was performed because of the echo changes, and it confirmed spontaneous anomalous pulmonary vein atresia. Angiography confirmed the hypoplastic right pulmonary veins coalesced into a single confluence, which previously drained to the IVC but now shows long-segment atresia (Figure 5, Video 3). Right PA angiography demonstrated pruning of the distal arterial branches with delayed emptying and stagnant contrast as well as retrograde emptying of the RPA into the LPA. The PA collateral remained small, with no evidence of significant flow. There was no increase in saturations through the right heart and PA, so no important left-to-right shunt was identified. The RV systolic pressure was 26 mm Hg, which matched peak systolic PA pressures. The pulmonary vascular resistance was estimated at 1.9 Woods units/m^2 with a transpulmonary gradient of 8 and Qp of 4.2 L/minute/m^2, all consistent with normal PA pressures through the functionally single left lung. Because of the long-segment atresia/striking anomalous pulmonary venous hypoplasia, normal PA pressures, and absence of clinical symptoms, no surgical intervention was attempted.

DISCUSSION

We present an unusual case of an infant with scimitar syndrome and stenosis of the anomalous right pulmonary vein as it enters the IVC who developed spontaneous atresia of that vein. Most infants diagnosed with scimitar syndrome have patent pulmonary venous drainage at the time of diagnosis. Pulmonary venous obstruction is a known potential surgical complication, but spontaneous atresia appears very rare.[3-5] What makes this case even more unusual is the lack of significant symptoms and absence of PA hypertension, despite the unilateral pulmonary venous obstruction. The presence of RPA diastolic flow reversal by echo with continuous and augmented LPA flow was a key finding that led to the suspected anomaly at presentation, and this recurred with

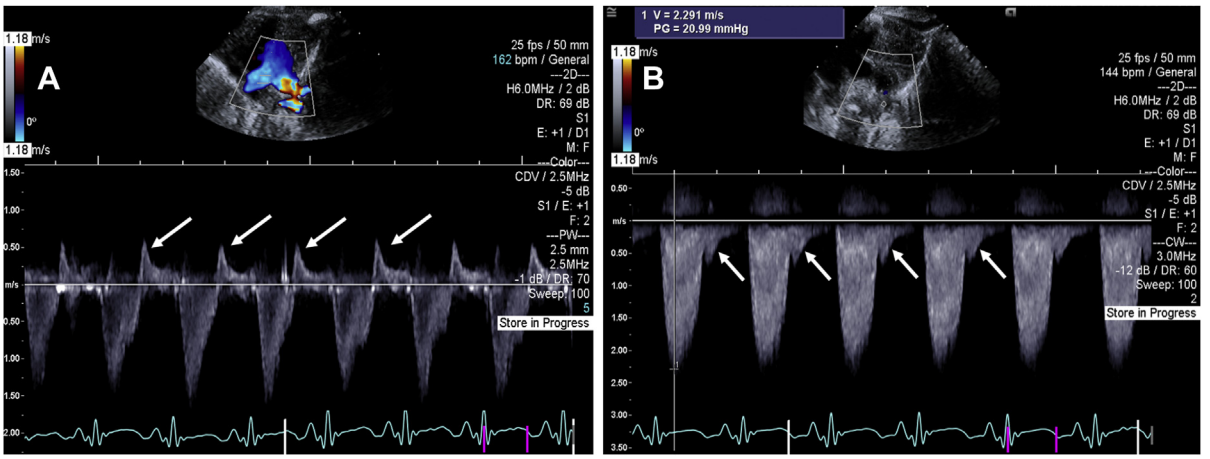

Figure 1 Two-dimensional transthoracic echocardiogram from the parasternal short-axis guided pulsed Doppler integration of the RPA **(A)** and LPA **(B)** from the initial evaluation. Flow in the RPA was characterized by retrograde diastolic flow (*arrows*), which was strikingly different from the turbulent anterograde flow seen in the LPA that continues into diastole (*arrows*).

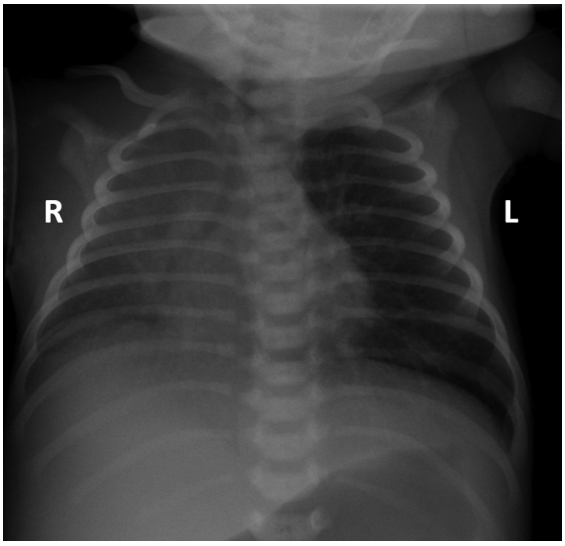

Figure 3 Chest x-ray, posterior-anterior orientation, from initial evaluation showed a hyperinflated left (L) lung and a diffusely opacified right (R) hemithorax with the cardiothymic silhouette shifted into the right chest.

development of the spontaneous atresia. We suspect that the RPA flow reversal was related to the pulmonary venous obstruction, so that right lung resistance is increased with resultant limited flow into the RPA, with some of the flow being decompressed into the LPA. Although the finding of unilateral PA flow reversal has not been described in patients with scimitar syndrome as a marker of anomalous vein obstruction, a recent series found echocardiographic color Doppler identification of branch PA flow reversal in children with single-ventricle physiology to be a high-risk indicator for PA/pulmonary venous abnormalities and adverse outcomes.[6]

Infants who are diagnosed with scimitar syndrome often have severe symptoms, which is what frequently leads to their initial evaluation and diagnosis.[7-9] Infantile scimitar syndrome typically has more severe

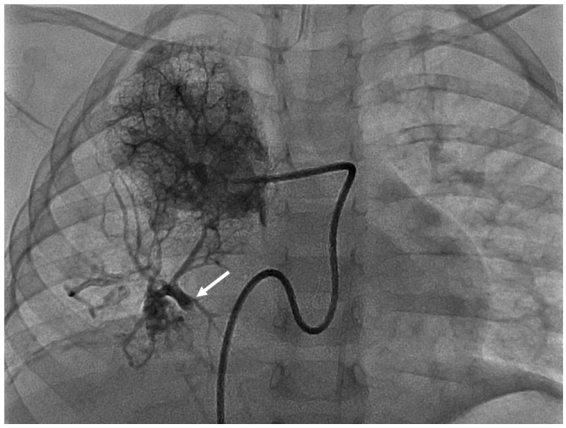

Figure 5 Right upper PA wedge angiography at 1 year of age showed markedly hypoplastic right pulmonary veins filling and coalescing into a single confluence (*arrow*) with long-segment atresia.

disease and higher risk of early mortality (33%-60%) than the "adult onset" scimitar syndrome that presents in later childhood or adulthood.[2,10-12] There are few reports describing infants with scimitar syndrome and stenotic anomalous pulmonary connections, but they suggest a poor outcome. Gao *et al.*[2] described two infants with stenotic anomalous pulmonary veins. One patient underwent stainless steel balloon-expandable stent placement at 28 days of life; however, at 1 year of age the child was noted to have complete occlusion of the scimitar vein and IVC by cardiac catheterization. The other infant died at 44 days of life. Honey[13] found data on three cases with evidence of obstruction of the anomalous vein in older children. However, these three cases all had pulmonary hypertension.

We could find no case reports of asymptomatic infantile scimitar syndrome with primary pulmonary venous obstruction and subsequent spontaneous atresia of the anomalous pulmonary vein. O'Byrne *et al.*[14] reported a 6-year-old boy who was diagnosed with childhood/adult-onset scimitar syndrome with right pulmonary vein atresia.

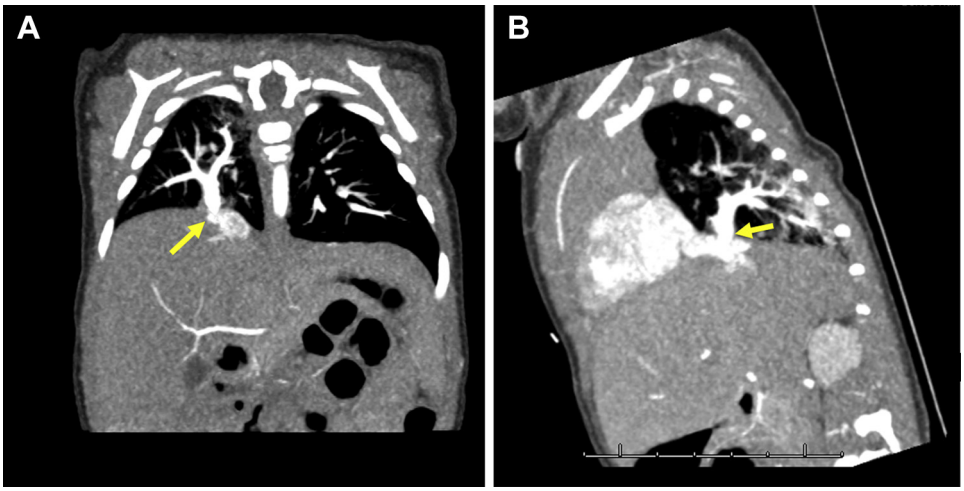

Figure 4 Contrast-enhanced cardiac computed tomography scan, coronal **(A)** and sagittal **(B)** windows, demonstrated all the right lung pulmonary veins draining anomalously into the posterior aspect of the IVC at the IVC-RA junction with stenosis of the anomalous pulmonary venous confluence at its entrance (*arrows*).

Wang *et al.*[15] described 10 infants with isolated scimitar syndrome with no associated significant congenital heart disease (as we would group our patient, since she had only a tiny muscular VSD). Three of those infants had scimitar vein obstruction at the IVC entrance. Obstruction was more commonly seen in infants with scimitar syndrome and associated congenital heart disease. However, all infants reported in this case series, regardless of whether they had congenital heart disease and/or pulmonary venous obstruction, were symptomatic with heart failure at the time of diagnosis, whereas our patient has been asymptomatic.

CONCLUSION

This is a very rare case describing neonatal anomalous pulmonary venous obstruction that progressed to spontaneous atresia during infancy with scimitar syndrome. Echocardiography is an essential diagnostic tool in the assessment of scimitar syndrome, and evidence of RPA diastolic flow reversal by Doppler can be an indirect sign of anomalous pulmonary venous obstruction. Infantile scimitar syndrome typically carries high mortality no matter the management strategy, with significant risk of late obstruction following surgical anomalous vein baffling; interestingly, our patient has always been asymptomatic and had normal PA pressures by serial cardiac catheterizations, despite the progression from stenosis to atresia.

SUPPLEMENTARY DATA

Supplementary data to this article can be found online at https://doi.org/10.1016/j.case.2022.02.002.

REFERENCES

1. Neill CA, Ferencz C, Sabiston DC, Sheldon H. The familial occurrence of hypoplastic right lung with systemic arterial supply and venous drainage "scimitar syndrome. Bull Johns Hopkins Hosp 1960;107:1-21.
2. Gao YA, Burrows PE, Benson LN, Rabinovitch M, Freedom RM. Scimitar syndrome in infancy. J Am Coll Cardiol 1993;22:873-82.
3. Dusenbery SM, Geva T, Seale A, Valente AM, Zhou J, Sena L, et al. Outcome predictors and implications for management of scimitar syndrome. Am Heart J 2013;165:770-7.
4. Vida VL, Padalino MA, Boccuzzo G, Tarja E, Berggren H, Carrel T, et al. Scimitar syndrome: a European Congenital Heart Surgeons Association (ECHSA) multicentric study. Circulation 2010;122:1159-66.
5. Najm HK, Williams WG, Coles JG, Rebeyka IM, Freedom RM. Scimitar syndrome: twenty years' experience and results of repair. J Thorac Cardiovasc Surg 1996;112:1161-9.
6. Spearman AD, Ginde S, Goot BH, Schaal AM, Feng M, Pan AY, et al. Echocardiographic identification of pulmonary artery flow reversal: an indicator of adverse outcomes in single ventricle physiology. Pediatr Cardiol 2020;41:1632-8.
7. Canter CE, Martin TC, Spray TL, Weldon CS, Strauss AW. Scimitar syndrome in childhood. Am J Cardiol 1986;58:652-4.
8. Haworth SG, Sauer U, Bühlmeyer K. Pulmonary hypertension in scimitar syndrome in infancy. Br Heart J 1983;50:182-9.
9. Gikonyo DK, Tandon R, Lucas RVJ, Edwards JE. Scimitar syndrome in neonates: report of four cases and review of the literature. Pediatr Cardiol 1986;6:193-7.
10. Dupuis C, Charaf LA, Brevière GM, Abou P. "Infantile" form of the scimitar syndrome with pulmonary hypertension. Am J Cardiol 1993;71:1326-30.
11. Huddleston CB, Exil V, Canter CE, Mendeloff EN. Scimitar syndrome presenting in infancy. Ann Thorac Surg 1999;67:154-9. discussion 160.
12. Dupuis C, Charaf LA, Brevière GM, Abou P, Rémy-Jardin M, Helmius G. The "adult" form of the scimitar syndrome. Am J Cardiol 1992;70:502-7.
13. Honey M. Anomalous pulmonary venous drainage of right lung to inferior vena cava ("scimitar syndrome"): clinical spectrum in older patients and role of surgery. Q J Med 1977;46:463-83.
14. O'Byrne ML, Cross RR, Martin GR. Asymptomatic atresia of the anomalous pulmonary vein in a patient with scimitar syndrome presenting in childhood. Cardiol Young 2018;28:329-33.
15. Wang H, Kalfa D, Rosenbaum MS, Ginns JN, Lewis MJ, Glickstein JS, et al. Scimitar syndrome in children and adults: natural history, outcomes, and risk analysis. Ann Thorac Surg 2018;105:592-8.