Mysterious Infantile Cyanosis: An Imaging Case Series

Sudeep Sunthankar, MD, Nhue L. Do, MD, David Parra, MD, Kimberly Vera, MD, MSCI, and Jonathan H. Soslow, MD, MSCI, Nashville, Tennessee

INTRODUCTION

In the setting of cyanosis in the neonatal period with no evidence of a cyanotic congenital heart defect, a cardiologist should consider a diagnosis of cor triatriatum dexter (CTD). We present four cases of CTD, a rare form of congenital heart disease. Our objectives are to (1) describe the presentation of this disease, (2) demonstrate the common echocardiographic features, and (3) discuss the management of these patients.

CASE 1 PRESENTATION

A 6-week-old with cyanosis presented as a transfer from an outside hospital with small right-sided structures with a plan for Blalock-Taussig-Thomas shunt to augment pulmonary blood flow. Prenatally, he was followed by a fetal cardiologist for a diagnosis of hypoplastic right ventricle (RV). Following delivery, he was observed in the neonatal intensive care unit (NICU), where he was able to maintain oxygen saturations in the 80s. He was discharged after 2 days of observation. Over the course of the following 6 weeks, his oxygen saturations began to drift lower into the 70s. At 5 weeks old, he began to have difficulty with feeds and was found to have episodes of oxygen saturations in the 50s. The patient was transferred to our institution for surgical management. Upon arrival, an echocardiogram demonstrated a normal sized right atrium and an underfilled and mildly hypoplastic RV. The tricuspid valve annulus measured normal, but the pulmonary valve annulus (Z score, –2.4) and branch pulmonary arteries (left pulmonary artery Z score, –2.9; and right pulmonary artery Z score, –3.0) were mildly hypoplastic. A patent foramen ovale (PFO) was present with predominantly right-to-left shunting explaining the child’s cyanosis. A hypermobile structure was seen in the right atrium that at times overlayed the tricuspid valve. Color Doppler demonstrated limited inflow across the tricuspid valve due to the presence of this membrane (Figure 1A–D, Videos 1 and 2). An agitated saline contrast injection study confirmed the right-to-left atrial-level shunt (Video 3). The following day, the patient was taken to the operating room for membrane resection. A membrane with attachments to the superior vena cava (SVC), Eustachian valve, and tricuspid valve annulus was resected, consistent with a diagnosis of CTD. A single hole was noted in the specimen (Figure 2), corroborating images obtained with intraoperative transesophageal echocardiography (TEE; Figure 1E, Video 4). Following membrane resection, while still in the operating room, the child’s oxygen saturations were 99%. In addition, there was improvement in filling of the RV. He recovered in the hospital and was discharged home on postoperative day 4.

CASE 2 PRESENTATION

A term newborn, delivered at an outside hospital, was found to be cyanotic with choking episodes and emesis on the first day of life. At 8 days of age, she was found to be persistently hypoxic. Three different echocardiograms were obtained in the first week of life, all of which showed bidirectional atrial-level communication with elevated pulmonary pressures, but otherwise a structurally normal heart. With concern for pulmonary hypertension, the patient was placed on 100% FiO2 and 20 ppm inhaled nitric oxide. Due to concern for low cardiac output and patient lability, dobutamine, dopamine, and milrinone drips were started. The patient was transferred to our institution for consideration for extracorporeal membrane oxygenation support. Repeat echocardiogram performed at our institution upon arrival to the NICU demonstrated a large membrane that obstructed the tricuspid valve in diastole (Figure 3, Video 5). It appeared to originate from the Eustachian valve and diverted the inferior vena cava return across the PFO to the left atrium. There was a space between the membrane and the tricuspid valve annulus through which superior vena cava return could be seen circumventing the membrane and traversing the tricuspid valve to supply pulmonary blood flow. An agitated saline contrast injection study performed via the upper extremity demonstrated a significant portion of venous return entering the LV (Video 6). The right-sided structures were normal in size. With a diagnosis of CTD, the child’s oxygen saturation goal was lowered. The inotropic and pulmonary vasodilator support was gradually removed, and the patient was able to maintain saturations >75%. Patient was discharged home with saturations above 95%. Serial outpatient follow-up demonstrated oxygen saturations >97% with steady weight gain. Despite continued progress and normal saturations, the family elected to...
Serial clinic visits demonstrated oxygen saturations > 93%, mean tricuspid inflow gradients between 3 and 6 mm Hg (Figure 4D, Video 9), and normal RV size and function. She continued to develop and grow well. She is now 7 years old and has not required surgical intervention.

CASE 4 PRESENTATION

A term neonate with a prenatal diagnosis of pulmonary atresia and intact ventricular septum delivered at an outside hospital where prostaglandin infusion was started and then transferred to our institution. Initial echocardiography (Figure 5A–C) demonstrated a membrane superior to the hypoplastic tricuspid valve (Z score, −2.6) that directed venous return across the atrial septum consistent with CTD (Figure 5B and Video 10) in addition to his known pulmonary atresia and intact ventricular septum anatomy. On day of life 5, a cardiac catheterization demonstrated RV-driven coronaries and a patent ductus arteriosus stent was placed. The remainder of his initial hospitalization was uneventful, and he was discharged home at 3 weeks old. He returned at 5 months old for atrial septectomy, Glenn palliation, pulmonary artery augmentation, ductal stent removal, and membrane resection. Transesophageal echocardiography (Figure 5D) demonstrated two fenestrations within the membrane allowing flow across the tricuspid valve. He has since proceeded to Fontan palliation. His tricuspid valve has remained mildly hypoplastic with Z scores of −2.2 to −2.6 despite membrane resection.

DISCUSSION

Cor triatriatum dexter is an exceedingly rare finding, comprising <0.025% of cases of congenital heart disease. The presenting scenario can be variable. A neonatal diagnosis may be made in the setting of a hypoxic newborn, as described above. Older patients may be diagnosed incidentally by echocardiogram during a cardiology evaluation for other reasons. The membrane of CTD is believed to be maldevelopment of the right valve of the sinus venosus, which forms the Thebesian and Eustachian valves. Prominence of these structures is thought to comprise the membrane in CTD. While both Chiari network and CTD are formed by prominent remnants of the right valve of the sinus venosus, CTD typically has none or only a few perforations, while Chiari network has many large fenestrations. While CTD is a rare condition, it should be considered in a patient with right-to-left atrial shunt and normal cardiac segmental relationships. Missed diagnosis of CTD could lead to unwarranted surgical interventions and thus should be ruled out in the absence of more common cyanotic congenital heart lesions. While easily overlooked due to limitations of transthoracic echocardiography, a diagnosis of CTD can be made by giving special attention to the surrounding structures of the tricuspid valve during the assessment of a hypoxic neonate with right-to-left atrial shunt. The apical four-chamber and subcostal sagittal views from our experience are the optimal views to profile this membrane and its obstruction of the tricuspid valve.

Our first case highlights an interesting and challenging presentation of CTD as the right-to-left atrial-level shunt was initially thought to be secondary to inadequate right-sided structures based on fetal and initial postnatal echocardiograms. In fact, the presence of CTD, directing systemic venous return across the atrial septum causing the right-to-left shunt, was most likely the precursor to the mildly hypoplastic right-sided structures. This case demonstrates the importance of ruling out CTD as this patient was referred to our center for Blalock-Taussig-Thomas shunt to intervene on what was believed to be a case of
hypoplastic right heart. In the setting of normal cardiac segmental relationships, but hypoplastic tricuspid and pulmonary valves, attention should be given to secondary causes of obstruction (i.e., muscle bundles, membranes, clot, or mass).

Cases 2 and 3 highlight patients who were able to grow into early childhood without intervention. These cases demonstrate that permissive hypoxia in the short term may delay or even prevent the need for surgical intervention. While case 2 decided to undergo surgery at another institution (instead of observation, as recommended by our institution given the normal saturations), case 3 will require continued intermittent follow-up, but as long as she remains clinically asymptomatic, we do not anticipate surgical intervention. Case 4 demonstrates a more severe presentation. We hypothesize that the membrane led to a decrease in antegrade flow, which contributed to the hypoplastic tricuspid valve and the atretic pulmonary valve. This patient required neonatal intervention with a patent ductus arteriosus stent for adequate pulmonary blood flow. As the membrane obstructed RV inflow but not right-to-left shunting across the atrial septum, membrane resection was delayed until the time of atrial septectomy with Glenn palliation.

With these cases in mind, we advocate for an individualized plan for each case of CTD. Over the first 2–3 months of life, right-to-left atrial-level shunt in CTD may decrease as seen in cases 2 and 3. This is likely due to a combination of somatic growth reducing the effective obstruction of the membrane over time and decreasing pulmonary artery pressures. Thus, observing patients through this transitional period is encouraged if the patient is not having persistent saturations <80% or episodic significant desaturations, as seen in case 1. A key feature in determining the utility of an observation period is the number of fenestrations and tricuspid valve inflow. While evaluating the adequacy of fenestrations can be challenging with transthoracic echocardiography, color flow mapping may be
Figure 3 Parasternal short-axis view showing the membrane and color mapping with flow circumventing the membrane (A). Parasternal long axis showing membrane (*) obstructing the tricuspid valve (B). PFO with right-to-left shunting (C). Apical four-chamber view (D) showing obstruction of the tricuspid valve by the membrane.

Figure 4 Parasternal short-axis (A) and parasternal long-axis (B) views showing aliasing of flow across the membrane (*) and obstruction of the tricuspid valve. Apical four-chamber view at diagnosis (C) and at 5 years old (D) showing increased flow across the tricuspid valve with less aliasing on follow-up study.
useful to identify defects within the membrane or space by which the membrane can be circumvented. Bubble contrast studies, as performed in cases 1 and 2, can also be beneficial. In addition, pulse-wave Doppler tracings of flow across the tricuspid valve are helpful to detect tricuspid valve obstruction. Finally, use of TEE may allow for better visualization of defects. Regardless of the assumed fenestration size, the treatment should be based on clinical findings, with saturations as the primary outcome measure. Resection of the membrane is warranted in the setting of persistent hypoxia that does not improve with age or in the setting of hemodynamically significant right-to-left shunt causing hypoplasia of the right-sided structures and/or cyanotic spells.

CONCLUSION

In patients with small right-sided structures and no abnormality of the pulmonary or tricuspid valve, a diagnosis of CTD should be excluded. This can be a challenging diagnosis to make; therefore, special attention must be paid to exclude a membrane overlaying the tricuspid valve. We advocate for conservative management of neonates with mild hypoxia as we anticipate this to improve with somatic growth. In patients with more severe hypoxia, membrane resection is warranted and should serve as a definitive repair.

SUPPLEMENTARY DATA

Supplementary data related to this article can be found at https://doi.org/10.1016/j.case.2021.07.013.

REFERENCES

1. Moral S, Ballesteros E, Huguet M, Panaro A, Palet J, Evangelista A. Differential diagnosis and clinical implications of remnants of the right valve of the sinus venosus. J Am Soc Echocardiogr 2016;29:183-94.
2. Gold BM, Parekh DR, Kearney DL, Silva GV, Fish RD, Stainback RF. Forme fruste cor triatriatum dexter by transesophageal echocardiography and its impact on percutaneous heart procedures: a case series. CASE 2019;3:189-99.

3. Zoltowska D, Kalavakunta JK. Cor triatriatum dexter. Clin Case Rep 2018;6:1189-90.

4. Ducharme A, Tardif JC, Mercier LA, Burelle D, Rodrigues A, Petitclerc R, et al. Remnants of the right valve of the sinus venosus presenting as a right atrial mass on transthoracic echocardiography. Can J Cardiol 1997;13:573-6.

5. Hansing CE, Young WP, Rowe GG. Cor triatriatum dexter. Persistent right sinus venosus valve. Am J Cardiol 1972;30:559-64.