AHA 2012 - Laennec Young Clinician Submission – Case Study

BLUE AGAIN:
Recurrent Cyanosis In A 30-year-old Man With Surgically Palliated Cyanotic Congenital Heart Disease

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A 30-year-old man with a history of congenital heart disease presented with dyspnea and blue lips. He had been diagnosed at birth with tricuspid atresia in conjunction with atrial and ventricular septal defects, and a persistent left superior vena cava. A right-sided Glenn shunt was performed at age 13 months, followed by an atriopulmonary Fontan procedure at age 8 years. From 8 to 21 years of age, the patient was able keep up with his peers without difficulty. At age 21, he developed atrial fibrillation, which evolved from paroxysmal to persistent. Thrombosis of the Fontan circuit prompted revision with an unfenestrated lateral tunnel Fontan, left bidirectional Glenn cavopulmonary anastomosis, biatrial MAZE procedure and prophylactic implantation of an epicardial dual-chamber pacemaker at age 26 years. A laminar thrombus was noted within the lateral tunnel shortly after surgery; evaluation for a hypercoagulable state was unrevealing. After Fontan revision and MAZE procedure, the patient achieved freedom from atrial fibrillation, continued on anticoagulation, and maintained normal systemic ventricular function, 92% oxygen saturation on room air, and subjectively unlimited exercise tolerance.

Despite recurrence of paroxysmal atrial fibrillation 18 months later, the patient remained asymptomatic until age 29, when he sought medical attention for dyspnea and mild oxygen desaturation to 90% on room air. Evaluation revealed volume overload in the setting of atrial fibrillation with rapid ventricular response. Transthoracic echocardiography (TTE) disclosed persistent thrombus in the lateral tunnel and moderately depressed systemic ventricular systolic function. Amiodarone, beta-blocker uptitration, and diuretic therapy restored his baseline functional capacity and arterial oxygenation.

He remained well until 2 weeks prior to the current presentation, when he developed myalgia and cough, without fever. Although his flu-like symptoms resolved after one week, he noted progressive dyspnea on exertion. He became dyspneic at rest and unable to sustain a conversation. For the first time in his memory, he noticed that his lips were blue. Examination demonstrated heart rate of 82 bpm in sinus rhythm, blood pressure of 112/80 mmHg, and respiratory rate of 24 times per minute. Oxygen saturation on room air was 81% and did not change with escalating oxygen support. Weight was 5 pounds above baseline. The patient was small in stature. Lips had a bluish hue. There was no appreciable jugular venous distention. Breath sounds were minimally decreased at left lung base. The point of maximal impulse was mildly displaced to the left. A 1/6 holosystolic murmur was heard at left lower sternal border. There were well-healed sternal and abdominal scars. The pacemaker generator was palpable in the upper abdomen. There was mild epigastric and right upper quadrant tenderness. The extremities were lukewarm without edema. Nails showed mild clubbing with bluish discoloration of nailbed with delayed capillary refill.

Pacemaker interrogation revealed no episodes of rapid rates during the two weeks prior to presentation. Complete blood counts revealed a hemoglobin level of 17.9 g/dL, increased from a baseline of 16.3 g/dL; white blood cell and platelet counts were normal. Serum chemistries demonstrated lactic acidosis, acute kidney injury, transaminitis, elevated conjugated bilirubin, and normal albumin. TTE on admission (Figure 1) revealed enlargement of previously noted thrombus in the lateral tunnel, mild mitral regurgitation, severely reduced left ventricular systolic function, Doppler findings suggestive of elevated left atrial pressure, and low-velocity bidirectional flow through the known septal defects.

Formulation of a physiology-based differential diagnosis for recurrent cyanosis in this case required consideration of the patient's clinical presentation in the context of his reconstructed anatomy. Possible explanations included obstruction of the Fontan circuit by progressive thrombosis with increased right-to-left shunting via venous communications, thromboembolism to the pulmonary

arterial system, and increased shunting through pulmonary arteriovenous malformations or venous collaterals due to increased pulmonary vascular resistance in the setting of the recent flu-like illness. Progressive systolic dysfunction of the systemic ventricle could further contribute to hypoxemia and hypoperfusion due to elevation of left atrial pressure and decrease in the transpulmonary pressure gradient, resulting in diminished Fontan flow.

We decided to pursue cardiac catheterization following transesophageal echocardiographic (TEE) imaging of the Fontan thrombus, to assess Fontan pressures, right-to-left shunt, pulmonary vascular resistance and cardiac output. Identification of vascular malformations or collaterals responsible for significant right-to-left shunting could permit percutaneous intervention to reduce right-to-left shunt and improve systemic arterial saturation. Hemodynamic measurements would also provide important data for heart or heart-lung transplantation evaluation—a necessary consideration, as Fontan revision is unsuitable with systemic ventricular failure.

TEE (Figures 2 and 3) revealed poor ventricular function as well as extensive acute and chronic thrombosis, with extension to the anastomosis of the Fontan conduit to the right pulmonary artery, and near-obliteration of the lateral tunnel. Significant spontaneous echo contrast was seen in the right atrium. There was no evidence of baffle leak. The extent of thrombosis was surprising and substantially worse than noted on admission TTE. Strategy was changed to pursue contrast-enhanced computed tomography (CT) to obtain a three-dimensional assessment of clot burden and potential thromboembolism of the pulmonary vasculature, followed by thrombolytic therapy and immediate referral for transplantation. CT was protocolled carefully, with planning for delayed image acquisition to allow for adequate contrast mixing and avoidance of known flow void artifacts due to streaming of poorly mixed contrast from persistent left superior vena cava and passive filling of Fontan circuit. During CT scanning, the patient suffered a respiratory arrest with brief stabilization after intubation and mechanical ventilation with recovery of baseline blood pressure, followed by a cardiac arrest thought most likely related to embolization or extension of the Fontan thrombus into the pulmonary vasculature. Spontaneous circulation could not be restored. The family declined autopsy.

Tricuspid atresia is a congenital cyanotic defect with single-ventricle physiology that can be associated with additional congenital anomalies. With advances in pediatric cardiac surgery, more children with congenital heart diseases are surviving into adulthood, and may be encountered in adult cardiology practice. This case underscores the importance of understanding the anatomy and physiology of congenital defects and associated surgical repairs as well as the potential limitations of bedside examination and imaging studies. Among patients with Fontan circulation, thromboembolism, arrhythmia, and eventual systemic ventricular failure are known complications. Cyanosis can be an ominous sign. Concurrent thrombosis of the Fontan circuit and failure of the systemic ventricle carries a particularly poor prognosis, as systemic ventricular failure contributes to the risk of recurrent Fontan thrombosis, and precludes isolated Fontan revision due to the adverse impact of increased left ventricular end-diastolic pressure on passive venous flow in Fontan physiology. In affected individuals, transplantation may be the only option. As such, recognition of systemic ventricular failure should prompt close follow-up, additional imaging to further assess Fontan circuit, and early referral for initiation of transplant evaluation at a center with expertise in adult congenital heart disease and transplantation.

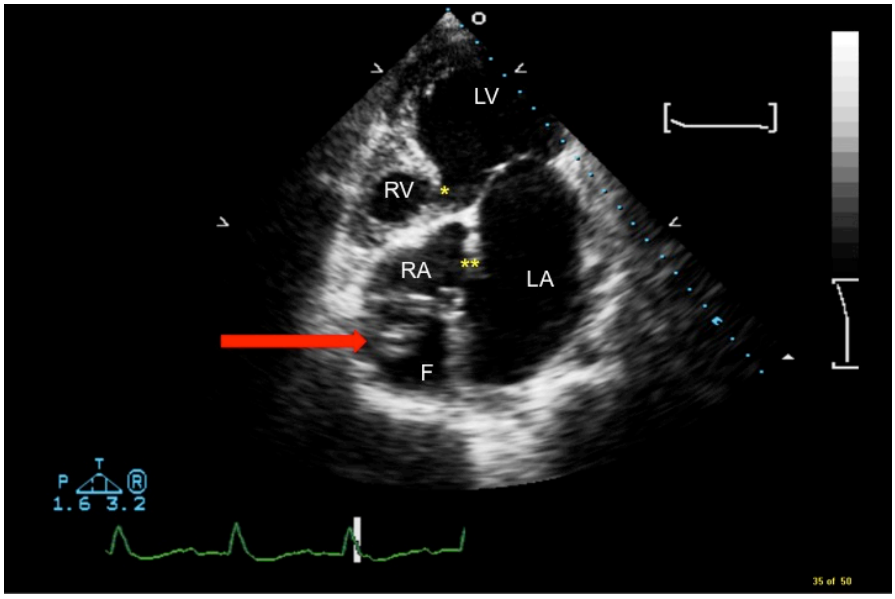

Figure 1. Transthoracic echocardiography on admission, showing the right atrium (RA), left atrium (LA), right ventricle (RV), left ventricle (LV), atrial septal defect (**), ventricular septal defect (*), Fontan lateral tunnel (F), and mural thrombus in lateral tunnel (red arrow).

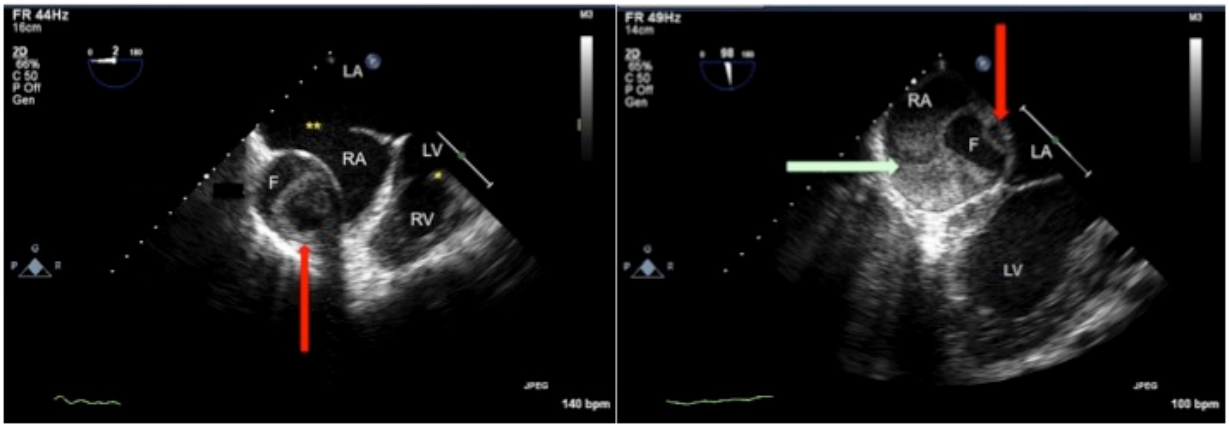

Figure 2. Transesophageal echocardiography, showing the right atrium (RA), left atrium (LA), right ventricle (RV), left ventricle (LV), atrial septal defect (**), ventricular septal defect (*), Fontan lateral tunnel (F), mural thrombus in lateral tunnel (red arrow), and stasis in right atrium (green arrow).

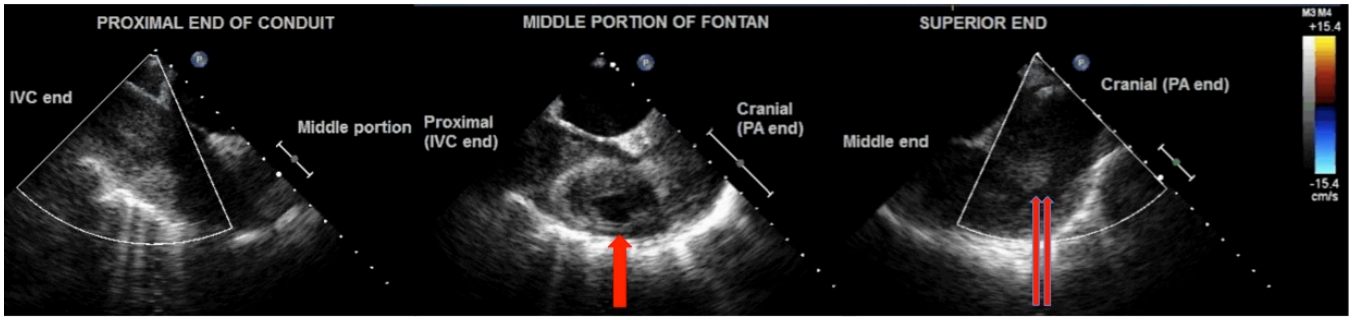

Figure 3. Transesophageal echocardiography showing extensive thrombosis and stasis throughout the Fontan conduit, with acute thrombosis with mobile elements (double red arrows) and large chronic thrombosis with central necrosis and liquefaction (red arrow).