

**Patient/Family Lay Summary:** FON Case Review Conference July 2022

**Coordinated by:** Children’s Hospital Los Angeles

**Topic:** Cyanosis after Fontan

**The Problem:**

Cyanosis (blue discoloration of the skin due to a low oxygen level in the blood) can be a sign of a problem in the Fontan circulation. One possible cause of cyanosis is pulmonary arteriovenous malformations [AVMs], which are abnormal connections between blood vessels in the lung that allow blood to pass through the lungs without picking up oxygen. Pulmonary AVMs are more likely to be a problem in patients with heterotaxy syndrome and can be difficult to treat.

**Clinical Course of the Cases Presented:**

Our patient was born with complex single ventricle heart disease including heterotaxy syndrome with unbalanced right dominant atrioventricular canal, double outlet right ventricle, D-malposed great arteries, pulmonary atresia with discontinuous branch pulmonary arteries, bilateral superior venae cavae, interrupted inferior vena cava with hemiazygos continuation to the left superior vena cava. She underwent stage 1 surgery and subsequent bilateral bidirectional Glenn without complications. After the Glenn surgery her oxygen level was in the expected range at first, but then gradually decreased to much lower than expected over the next several months due to pulmonary AVMs. Since pulmonary AVMs can be treated by directing blood flow from the liver to the lungs, she had the Fontan procedure (conduit from the liver to the right lung) early at 15 months. After the Fontan procedure, her oxygen level remained very low due to worsened AVMs in the left lung. Therefore, she had an additional smaller Fontan conduit placed between the liver and left lung. Unfortunately, her oxygen level did not improve, and she developed worsening ability to exercise and poor appetite. Next, she underwent cardiac catheterization, where the larger right-sided Fontan conduit was blocked to redirect all blood flow from the liver to the left lung to try to decrease the AVMs. This also did not improve her oxygen level and symptoms, so she was then listed for heart transplant.

She underwent a successful heart transplant, but afterwards she still had low oxygen level due to the pulmonary AVMs. Therefore, she had another cardiac catheterization to enlarge a stent to increase blood flow to the healthy right lung. She was then able to leave the hospital with extra oxygen and tadalafil, a medication to improve blood flow to the portions of her lungs that did not have AVMs. By 16 months after transplant, both her oxygen level and ability to exercise improved significantly to the point where she no longer needed extra oxygen or tadalafil.

**Important Points, Lessons Learned, and Potential Solutions:**

* Patients who have cyanosis that does not resolve after the Fontan procedure have an increased risk of poor long-term outcomes.
* Patients with heterotaxy syndrome often have blood vessel differences that increase their risk of developing pulmonary arteriovenous malformations (AVMs). Pulmonary AVMs can cause cyanosis that is difficult to treat.
* Treatment of pulmonary AVMs may require redoing the Fontan surgery differently, using stents to change the direction of blood flow, or rarely heart transplantation.