Truncus arteriosus (pronounced TRUNG-kus ahr-teer-e-O-sus) is a birth defect of the heart. In a normal heart there are two blood vessels that come out of the heart, the aorta and the pulmonary artery. The aorta carries oxygen-rich blood to the body, and the pulmonary artery carries oxygen-poor blood to the lungs. When a baby has truncus arteriosus, there is only one common blood vessel that comes out of the heart instead of two. This occurs when the blood vessel coming out of the heart fails to separate into two blood vessels during pregnancy. Because there are not two blood vessels, both oxygen-poor blood and oxygen-rich blood are mixed together as blood flows to the lungs and body. Too much blood goes to the lungs, and the heart works harder to pump blood to the rest of the body. Normally, there are two valves controlling the flow of blood to the lungs and the heart. The aortic valve controls the flow of blood into the aorta and the rest of the body. The pulmonary valve controls the flow of blood to the pulmonary artery and the lungs. A baby with truncus arteriosus has only one valve controlling the blood flow to the lungs and body. This valve, called the truncal valve, is usually thick and narrow. This can block blood as it leaves the heart. An opening between the lower chambers of the heart called a ventricular septal defect may also be seen with truncus arteriosus.

A baby with truncus arteriosus will need surgery or other procedures soon after birth, so truncus arteriosus is a critical congenital heart defect. Congenital means present at birth, and critical congenital heart defects can cause serious health problems or even death if left untreated.
Less than 1 in every 10,000 babies will be born with truncus arteriosus. There are usually about 300 babies a year in the United States born with it.

The cause of truncus arteriosus for most babies is unknown. There may be many factors that cause truncus arteriosus, but more research is needed to understand the exact cause.

Truncus arteriosus can be diagnosed during pregnancy or after. Screenings are done to check for birth defects. After the baby is born, a screening test called a pulse oximetry screen is performed to check for critical congenital heart defects. A pulse oximeter is a tool that detects oxygen levels in blood. Low levels of oxygen in the blood could mean there is a heart defect. A baby with blue colored skin and lips (cyanosis) also may be a sign of low levels of oxygen in the blood. In addition, the baby might have trouble breathing, a pounding heart, a weak pulse, poor feeding, and extreme tiredness. If a baby fails the screening, then the doctor should perform a diagnostic test called an echocardiogram to check for defects in the heart.

Treatment for a truncus arteriosus usually requires surgery. This is often done during the first few months of life. The goal is to create a separate flow of oxygen-poor blood to the lungs and oxygen-rich blood to the body. The opening between the two chambers of the heart is usually closed with a patch, and the single blood vessel is used to create a new aorta to carry oxygen-rich blood to the body. An artificial tube with an artificial valve is used to connect the right lower chamber to the blood vessels going to the lungs, allowing oxygen-poor blood to get to the lungs. Medicine may be needed to strengthen the heart muscle, lower blood pressure, and get rid of extra fluid from the body. Your child’s doctor should discuss treatment options with you. Regular visits to a cardiologist, a doctor who specializes in the heart, will be necessary to avoid problems and watch for any other health conditions.

For more information:
American Heart Association
http://www.heart.org/HEARTORG/Conditions/CongenitalHeartDefects/AboutCongenitalHeartDefects/About-Congenital-Heart-Defects_UCM_001217_Article.jsp#.Wv2YtPnwbcS

Centers for Disease Control and Prevention
https://www.cdc.gov/ncbddd/heartdefects/truncusarteriosus.html

National Heart, Lung, and Blood Institute
https://www.nhlbi.nih.gov/health-topics/congenital-heart-defects