Truncus arteriosus is a rare type of heart disease that is present at birth (congenital) in which there is a single main blood vessel, rather than the normal two, carrying blood away from the heart. Instead of having a separate pulmonary artery, to carry blood to the lungs, and aorta, to carry blood to the rest of the body, a baby with truncus arteriosus has just one blood vessel leaving the heart which then branches into other blood vessels. Blood from both ventricles of the heart is mixed, resulting in a situation in which some oxygen-rich blood travels needlessly back to the lungs and some oxygen-poor blood travels to the rest of the body. Babies with this condition may have a bluish tint (cyanosis) to their skin, lips, and fingernails. In most cases, truncus arteriosus occurs in conjunction with a missing upper portion of the wall between the ventricles of the heart (ventricular septal defect). The exact cause of truncus arteriosus is not known. It has been suggested that some cases may develop due to the interaction of many genetic and environmental factors (multifactorial inheritance). The malformation is the result of an error in embryonic development. Approximately 35 percent of children with this disorder also have DiGeorge syndrome, which is a severe congenital immune deficiency disorder involving the thymus and parathyroid glands. (For more information on DiGeorge Syndrome, see the Related Disorders Section of this report.) The diagnosis of truncus arteriosus is confirmed by clinical evaluation, electrocardiogram (EKG), and specialized imaging techniques that allow the physician to view the structure of the heart. These may include an echocardiogram, angiocardiogram, and/or cardiac catheterization.