

Endocardial fibroelastosis (EFE) is a rare heart disorder that affects infants and children. It is characterized by a thickening within the muscular lining of the heart chambers due to an increase in the amount of supporting connective tissue (inelastic collagen) and elastic fibers. The normal heart has four chambers. Two chambers, known as atria, are separated from each other by a partition called the atrial septum. The other two chambers, known as ventricles, are also separated by a septum. Valves connect the atria (left and right) to their respective ventricles. Endocardial fibroelastosis is a rare disorder that affects males and females in equal numbers. Fewer than 1 percent of infants and children with congenital heart disease are diagnosed with this disorder. A 1964 study suggested an incidence of 1 in 5,000 live births. However, in the United States there has been a marked reduction in incidence since then for reasons that are not known. The disorder is extremely rare. The diagnosis of endocardial fibroelastosis is confirmed by a thorough clinical evaluation, including a physical examination that may reveal signs of respiratory distress (i.e., moist rales) and galloping heart rhythms. Radiographic studies (x-ray) of the chest typically reveal abnormal enlargement of the heart, especially the left ventricle (ventricular hypertrophy). Damage to the heart may be demonstrated by measuring the electrical activity of the heart (i.e., electrocardiogram [EKG]). This test may show subtle changes (i.e., S-T segment and T-wave changes) that strongly suggest damage to the heart that is characteristic of EFE. Repeated electrocardiograms may be required to monitor changes in heart function.