Endocardial fibroelastosis in the current era: associated congenital heart disease and outcome

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Introduction: Endocardial fibroelastosis (EFE) occurs primarily in infants during the first year of life, and is often associated with congenital heart disease (CHD), especially stenosis within the left ventricular outflow tract (LVOT) and hypoplastic left ventricle syndrome (HLHS). Outcome of infants with EFE in the current era has not been well delineated.

Methods: The echocardiography database from 1997 until December 2012 was searched for all children and infants (pt) with echocardiographic signs of EFE (dense echoes along the endocardial surface of the left (and sometimes right) ventricle (LV)). Associated CHD, echocardiographic findings, age at diagnosis and survival were analyzed.

Results: There were 35 children (27 males) with EFE. HLHS was present in 18 (51%), bicuspid aortic valve in 14 (40%), and/or other CHD in 22 pt (63%). Age at last echocardiographic examination was 0.9±2 years. At least moderate mitral mitral regurgitation was present in 9 pt (26%). A dysplastic mitral valve was seen in 15 pt (43%). Decreased LV function was present in 57%.

Survival was severely diminished, 5 year survival was 64% but could not predicted by the presence of hypoplastic left heart syndrome, the ejection fraction, gender, age or associated congenital heart disease (p=NS). Cardiac interventions were necessary in 15 pt including hybrid operations, balloon valvuloplasty, Ross procedure.

Conclusions: The outcome of cardiac disease associated with endocardial fibroelastosis in the current era is still severely limited. Death cannot be predicted by the underlying etiology of EFE. Detection of EFE by echocardiography identifies a high risk population.