Endocardial Fibroelastosis

A Comprehensive Review

Maryam Barkhordarian, MD, Atefeh Ghorbanzadeh, MD,† William H. Frishman, MD,‡ and Wilbert S. Aronow, MD§*

Abstract: Endocardial fibroelastosis emerged as a challenging clinical phenomenon in the 1940s. It is characterized by an atypical proliferation of fibrous and elastic tissue within the heart and is primarily observed in childhood, occasionally displaying familial inheritance. While the precise cause remains elusive, various factors, including genetic, infectious, metabolic, autoimmune, oncologic, and medication-related influences, appear to play a role in its pathogenesis. The coexistence of endocardial fibroelastosis with multiple cardiac structural abnormalities manifests in symptoms of congestive heart failure and rhythm abnormalities. Despite its challenging diagnosis, various findings from ECG and imaging have proven beneficial in further evaluation of this condition. Finally, the treatment approach to endocardial fibroelastosis became complex due to addressing its concurrent cardiac abnormalities. Strategies for managing and preventing this condition are still under investigation. In this review, we intend to highlight the existing knowledge and illuminate future considerations regarding the etiology, diagnosis, and management of this disease.

Key Words: endocardial fibroelastosis, congestive heart failure, cardiac abnormalities, childhood heart conditions

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Endocardial fibroelastosis (EFE) is a cardiac condition characterized by an atypical proliferation of fibrous and elastic tissues, resulting in a distinct left ventricular endocardium thickening. First identified in the early 1940s by Weinberg et al.,[1] EFE was primarily observed in children presenting with unexplained heart failure, signifying a pivotal development in the field of cardiac pathology.[1,2] Over the years, research and clinical experiences have propelled the understanding and recognition of EFE, shaping our comprehension of its etiology, pathogenesis, clinical manifestations, diagnostic methods, and potential management strategies. Subsequently, EFE has been documented in various other heart-related conditions, including cardiomyopathies,[3] infectious diseases,[4] immunological disorders,[5] and congenital heart diseases such as hypoplastic left heart syndrome (HLHS).[6]

This review aims to provide a comprehensive exploration of the historical context, anatomy, etiology, clinical manifestations, diagnostic approaches, and management strategies associated with EFE, aiming to contribute to a deeper understanding of this cardiac condition and its implications for patient care.

EPIDEMIOLOGY-GEOGRAPHIC DISTRIBUTION

EFE is a rare and potentially life-threatening cardiac condition primarily affecting infants and young children. While the exact geographical distribution of primary EFE remains uncertain, it has been reported in diverse locations worldwide. While no definitive evidence establishes a causal link between this condition and maternal viral infections, the presence of a positive skin reaction to the mumps antigen in affected individuals suggests a potential association, however, with unclear mechanistic underpinnings.[4,7]

Historical data from the United States, dating back to 1964, reported an incidence rate of 1 per 5000 live births,[8] yet over the years, the prevalence of EFE has significantly declined, with almost no new cases reported in recent times. This decline is hypothesized to be related to factors such as the decreasing prevalence of mumps,[9,10] though the precise contributing factors remain elusive. On the international stage, EFE once accounted for 1–2% of all congenital heart diseases but has now dwindled to near-zero cases. Equally affecting both sexes, EFE predominantly manifests within the first 3–6 months of life, with the typical age at diagnosis ranging from 2 to 12 months. Adolescents and adults rarely present with EFE, underscoring its primary occurrence in infancy. Furthermore, EFE's the potential of EFE involvement in nonimmune hydrops fetalis underscores its clinical significance and the need for ongoing research to better comprehend this enigmatic cardiac condition.

In a study by Angelov et al,[11] which examined 38 autopsies of individuals with EFE, it was found that 82% of cases involved infants under 1 year old. Of these cases, 55% were classified as primary EFE, while the remaining 44% were associated with congenital heart malformations (secondary EFE). Additionally, 24% of cases exhibited pathological conditions during pregnancy.

PATHOPHYSIOLOGY OF ENDOCARDIAL FIBROELASTOSIS

EFE is characterized by the deposition of acellular fibrocartilagenous tissue within the subendothelial layer of the endocardium, particularly affecting the inflow tracts and apices of the ventricles. This condition leads to diffuse endocardial thickening and myocardial dysfunction.[12] The thickening of the endocardium is believed to result from persistent and increased wall tension in the ventricles, potentially linked to myocardial damage, mitral regurgitation, or a combination of these factors. Notably, these changes in EFE tend to progress with age, so rarely can be seen in adults.[13] While the disease is often sporadic, familial cases have been reported, although infrequently. Various observations suggest a possible viral etiology, including clinical similarities to chronic myocarditis, the presence of myocarditis or myocardial fibrosis in affected individuals, a higher incidence following coxsackievirus B epidemics, detection of persistent viral infection through molecular studies, and experimental induction of the disease in animal models through viral myocardial

From the *Department of Internal Medicine, Hackensack Meridian Health- Palisades Medical Center, North Bergen, NJ; †Department of Cardiovascular Disease, Division of Vascular Medicine, Mayo Clinic, Rochester, MN; ‡Department of Medicine, New York Medical College, Valhalla, NY; and §Departments of Medicine and Cardiology, Westchester Medical Center and New York Medical College, Valhalla, NY.

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Correspondence: Wilbert S. Aronow, MD, FACC, FAHA, Departments of Cardiology and Medicine, Westchester Medical Center and New York Medical College, 100 Woods Road, Macy Pavilion, Room 141, Valhalla, NY 10595. E-mail: wsaronow@aol.com.

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infections. Additionally, EFE shares phenotypic similarities with dilated cardiomyopathy. Recent research suggests that the fibroblasts responsible for EFE may originate from epicardium-derived mesenchymal cells. Evaluation of myocardium in patients with EFE, either due to dilated cardiomyopathy or structural heart disease showed that fewer B and T cell lymphocytes were involved in the myocardium.[14]

GENETIC CONSIDERATION

Familial instances of EFE underscore the presence of a genetic underpinning in certain cases. Notably, EFE can manifest through diverse inheritance patterns, encompassing X-linked recessive, autosomal dominant, and autosomal recessive modes. In familial cases, genetic mutations have been pinpointed as key drivers of the condition.

X-Linked Recessive Endocardial Fibroelastosis

A subset of EFE cases exhibits X-linked recessive inheritance, notably linked to the G4.5 gene, also known as tafazzin.[15] Mutations within the G4.5 gene have been ascribed to familial X-linked EFE and Barth syndrome. These genetic alterations are known to evoke structural alterations within the fetal heart, detectable as early as the 18th week of gestation.[16]

Autosomal Dominant and Autosomal Recessive Endocardial Fibroelastosis

In addition to X-linked inheritance, autosomal dominant and autosomal recessive inheritance patterns have also been observed in EFE cases. The specific genetic determinants responsible for these forms of EFE are currently under active exploration.[17]

Alternate Causal Mechanisms

While genetic factors constitute a prominent facet of EFE etiology in select cases, the origins of the condition extend beyond genetics. Several alternative mechanistic hypotheses and contributing factors have been postulated: Endothelial-Mesenchymal Transition: Some studies have advanced the notion that the transformation of endothelial cells into mesenchymal cells may play a role in the pathogenesis of EFE.[6]

Association with Concomitant Conditions

Figure 1 shows common secondary causes of EFE. While EFE can manifest independently, it is noteworthy that it is occasionally co-occurrent with other congenital heart anomalies, such as hypoplastic left heart syndrome, aortic stenosis, and atresia. Approximately 25–50% of EFE cases have been observed in conjunction with these related genetic conditions. In addition, loss of nexilin function leads to recessively inherited lethal cardiomyopathy with EFE.[18]

CLINICAL VARIATIONS OF ENDOCARDIAL FIBROELASTOSIS

There are 2 primary clinical forms of EFE—dilated and contracted. Dilated EFE is characterized by a significantly enlarged and globular heart, primarily affecting the left ventricle (LV) and left atrium (LA). The LV endocardium becomes milky white and thickened, especially in the outflow tract. The papillary muscles become shortened and thickened, leading to abnormalities in mitral leaflet movement and increased valvular deformities. Valvular involvement, characterized by thickening and adhesion of valvular leaflets or shortened papillary muscles, often results in valvar regurgitation or stenosis. In contrast, contracted EFE

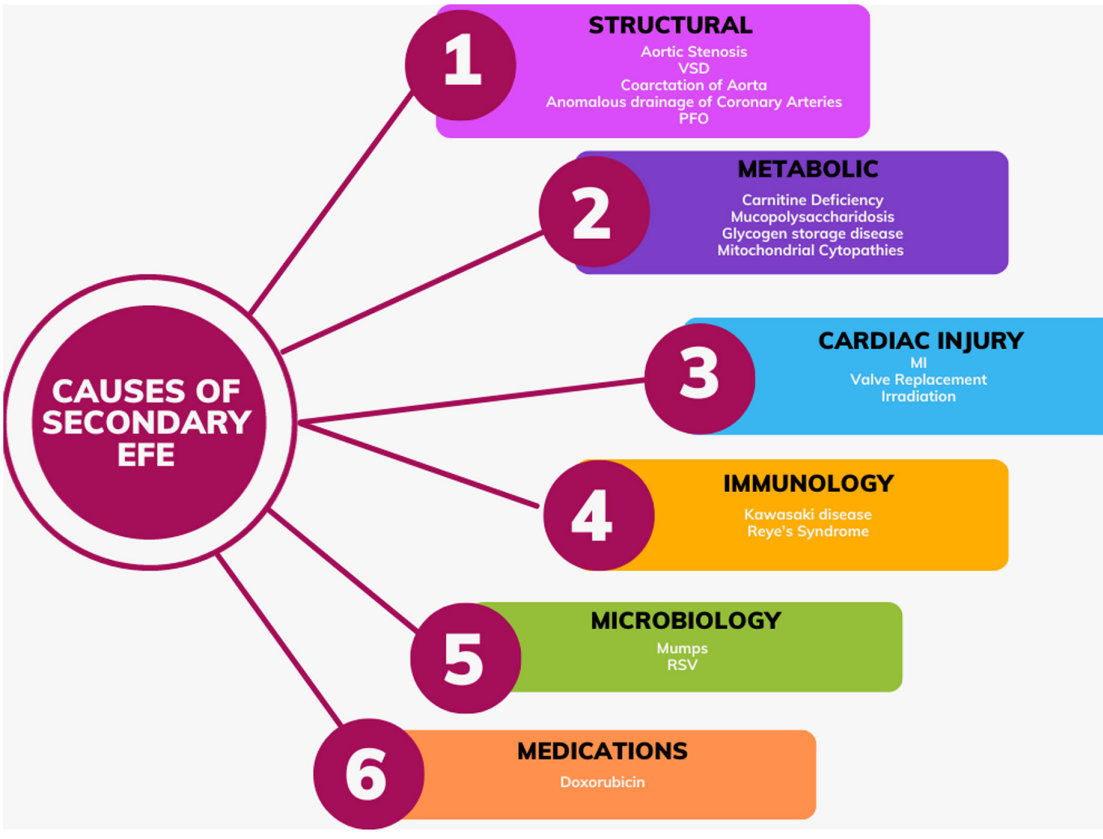

FIGURE 1. Common causes of secondary EFE. EFE indicates endocardial fibroelastosis.

is associated with relatively normal LV size, but significant hypertrophy and enlargement of the right atrium, right ventricle, and both atria are observed, with minimal endocardial sclerosis. This contracted form may evolve from the dilated type during fetal development, suggesting a continuum of the disease.[19] Additionally, secondary EFE, often associated with cardiac malformations, is attributed to cardiac hypertrophy and resultant oxygen supply-demand imbalances, resulting in focal and less severe fibroelastotic thickening of the endocardium. While endocardial thickening is prominent, myocardial thickness remains within the normal range. This condition can affect the mitral and aortic valves in approximately 50% of patients, leading to valvular deformities and regurgitation or stenosis.

TYPES: PRIMARY VERUS SECONDARY

EFE encompasses a spectrum of cardiac conditions that can be classified into 2 broad categories based on their underlying causes and characteristics. EFE primarily affects neonates and children, with rare occurrences in adults. The majority of cases occur in children under 1 year of age, with 55% classified as primary EFE and 44% as secondary, associated with congenital cardiac malformations[11]

The primary category, often referred to as "idiopathic" or "primary" EFE, is characterized by diffuse and widespread involvement of the endocardium. In primary EFE, the condition appears to originate within the endocardium itself, and the exact cause remains elusive. This form typically presents as marked hyperplasia of elastic tissue in the endocardium, particularly affecting the inflow tracts and apices of one or both ventricles.[3,20]

In addition, secondary EFE, also known as "focal" EFE, arises as a result of other underlying myocardial diseases or external factors. This form of EFE is typically focal in nature and is secondary to various conditions, including viral infections, myocarditis (inflammation of the heart muscle), metabolic disorders, inherited genetic diseases, and congenital malformations of the heart. In secondary EFE, the fibroelastosis changes in the endocardium are often a consequence of these underlying conditions, and the presentation can vary based on the specific etiology.[21,22] Among autoimmune diseases, Behcet's disease is also accompanied by EFE and right ventricular fibroelastic mass originating from the endocardium.[23]

It is important to note that primary EFE is relatively rare and primarily occurs without a clear association with other cardiac or noncardiac conditions. In contrast, secondary EFE is more commonly linked to other pathological processes affecting the heart. The classification into primary and secondary forms helps clinicians better understand the underlying mechanisms and contributing factors when diagnosing and managing individuals with EFE. However, the exact cause of primary EFE remains a subject of ongoing research, and further studies are needed to elucidate its origins.

Enzyme Deficiency Implications

Secondary EFE was also associated with disease involving enzyme deficiencies. Secondary EFE also has been linked with glycogen storage disease II (Pompe disease) accompanied by multicyclic dysplastic kidney[24] and Neimann-Pick disease. An infant case presenting with the common characteristics of Neimann-Pick disease, accompanied by endocardial fibroelastosis without myocardial infiltration of mononuclear cells and involving of aorta and mitral valve, was reported.[25] Its association with carnitine deficiency in liver, heart, and skeletal muscle, led to the classical presentation of EFE.[26] EFE also manifested as one of the initial signs of dilated cardiomyopathy in mucopolysaccharidosis type VI.[27] Its correlation with other types of mucopolysaccharidosis, such as Hurler syndrome (type 1) was found in twins with alpha-L-iduronidase deficiency.[28] EFE was reported as a possibility of X-linked inheritance in a familial case involving multiple male deaths within 6 months of age, secondary to mitochondrial cytopathies.[15]

Infectious and Viral Etiology

Infections and viral agents have been proposed as potential triggers for EFE. Investigations have explored the role of infectious pathogens or viral infections in the condition's development.[6,23] Intrauterine mump infection has been associated with the postnatal development of EFE.[29] The presence of positive cutaneous delayed hypersensitivity to the mumps virus in EFE patients with a history of gestational exposure to the virus supports this hypothesis. However, seronegativity might arise from minimal intrauterine exposure to the mumps virus.[30]

Autoimmune Implications

Evidence suggests that autoimmune factors may influence EFE. The presence of anti-Ro and anti-La antibodies has been documented in certain cases. Administrating corticosteroids to expectant mothers with these antibodies has shown the regression of EFE in fetuses, supporting the autoimmune hypothesis.[6,23] EFE is involved along with abnormalities in the conduction system, myocardial fibrosis, and coagulation necrosis in the stage IV presentation of Kawasaki disease.[31] Additionally, EFE has also been described as one of the findings of Reye's syndrome in the context of respiratory syncytial syndrome, alongside fatty changes of the liver and kidney.[32] Behcet's disease, another autoimmune disease, has also been observed to accompany EFE along with right ventricular fibroelastic mass originating from the endocardium.[23]

Concomitant Structural Heart Malformation Can Also Lead to Endocardial Fibroelastosis

Some structural abnormalities include the right coronary artery originating from the pulmonary artery,[33] coarctation of aorta, patent ductus arteriosus, bicuspid aortic valve,[34] and severe aortic stenosis with the origin of the right pulmonary artery from the ascending aorta.[35,36] Fibroelastosis of the foramen oval valve, due to increased mural stress, can contribute to the development of EFE.[37]

Surgical Alterations Could Also Result in Secondary Endocardial Fibroelastosis

Postoperative mitral valve replacement has been found to cause endocardial thickening and fibrosis due to turbulent flow in the left ventricle induced by the caged ball mitral valve within it.[38] Additionally, following a myocardial infarction, autopsies have revealed that more expanded infarcts correlate with increased occurrence of endocardial thrombosis and endocardial fibroelastosis.[39]

Secondary EFE has been linked to many noncardiac abnormalities in the fetus, including micrognathia, hypoplastic lungs, dysplastic kidneys, clubfoot, and central nervous system periventricular heterotopias, attributed to mutations in the genome.[40] Sclerema edematosum, a diffuse edematous skin condition, has been associated with EFE, accompanied by fibrosis of subcutaneous fat tissue and an increase in fibrotic tissue in various visceral organs like the gastrointestinal tract.[41] Other familial malformations linked to EFE include autosomal dominant macrocephaly, cryptorchidism, and an unusual facial appearance.[42] Reports also indicate that EFE is associated with multiple orthopedic abnormalities such as ulnar agenesis and oligodactyly.[43]

Malignancy by itself, as well as certain chemotherapeutic medications, have been linked to EFE. Secondary EFE may occur in the context of leukemic infiltration of the myocardium in chronic lymphocytic leukemia when no other cause is identified.[44] Additionally, EFE can result from adverse medication effects, such as doxorubicin, known for causing cardiotoxicity and is reported to be

associated with EFE.[45] Common causes of secondary EFE are illustrated in Figure 1.

CLINICAL COURSE

Acute congestive heart failure is the most common presentation, often leading to progressive heart failure and death within the first 6 months of life.[46] Additionally, perinatal echocardiography showed primary EFE as a cause of dilated heart failure.[47] Other presentations were reported, including pulmonary hypertension secondary to EFE in a pregnant woman.[48]

DIAGNOSIS CONSIDERATIONS OF ENDOCARDIAL FIBROELASTOSIS

The diagnosis of EFE is often challenging due to its rarity and the absence of definitive diagnostic criteria. It is most frequently established in infants, typically between 2 and 12 months of age, who may present with unexplained acute heart failure and cardiogenic shock, occasionally resulting in sudden infant death.[19,20]

ELECTROCARDIOGRAM MANIFESTATIONS

The assessment extends to the heart's electrical activity through electrocardiograms (ECGs), which may detect subtle changes like S-T segment and T-wave alterations, characteristic of EFE-related cardiac damage. In EFE fetuses with positive maternal anti-RO and anti-LA antibodies, atrioventricular block was the most common heart rhythm finding.[49]

Additionally, primary EFE with dilated cardiomyopathy was accompanied by premature ventricular contractions and nonsustained ventricular tachycardia.[3] Other ECG findings included left ventricular hypertrophy, T-wave inversion in leads V5-V6, a prolonged PR interval, partial right bundle branch block, complete left bundle branch block, and T-wave inversion in leads I and II.[2,50] Monitoring these ECGs over time may be necessary to track changes in heart function. Importantly, the diagnosis of EFE often involves a process of exclusion, as there are no definitive criteria for its identification. Clinicians must consider and rule out other potential causes of heart dysfunction, making it a diagnosis of exclusion[51].

FURTHER DIAGNOSTIC TOOLS

Clinical evaluation plays a pivotal role in confirming the diagnosis. Physical examinations may reveal signs of respiratory distress, such as moist rales and galloping heart rhythms. Chest radiographs frequently indicate abnormal heart enlargement, particularly in the left ventricle, suggesting ventricular hypertrophy.[51] The initial imaging studies on EFE were done using a 2-dimensional echocardiogram, which primarily showed an abnormally thickened endocardium alongside concomitant left ventricular pathologies.[52] Some anatomical parts involved in the echocardiography of fetuses with EFE are the proximal great arteries, mitral and tricuspid valve chordae, the atrial septum, and the pulmonary outflow tract.[53] However, certain studies indicate that 2-dimensional contrast echocardiography cannot reliably predict the presence of EFE.[54]

McElhinney and associates devised an echocardiographic grading for EFE in the fetus's left ventricle. The severity was graded as none, mild (characterized by scattered echogenic spots within the LV and papillary muscles), moderate (noncontiguous echogenic patches in the LV), and severe (contiguous echogenic lining of LV).[55] The fast-spin echocardiogram has shown an inability to distinguish between EFE and normal myocardium. In this context, myocardial delayed enhancement in cardiac magnetic resonance imaging (MRI) is useful.[56] A more accurate, noninvasive diagnosis of EFE can be achieved through electron beam computed tomography, revealing the extent of calcification and fibrotic tissue in the myocardium.[57]

Cardiac MRI in EFE reveals cardiac fibrosis and other heart failure-related findings such as ventricular function, regional contractility, and wall thickness.[58] The use of MRI with perfusion and myocardial delayed enhancement can be diagnostic in EFE. EFE may appear as a hypointense signal in perfusion sequences at the endocardial surface and a hyperintense signal in myocardial delayed enhanced sequences.[59] Angiocardiography has reported dilatation and hypertrophy, along with mobility and distensibility of the left ventricle.[60]

The definitive diagnosis is made by endocardial biopsy and histopathology. However, for noninvasive features, echocardiogram and MRI are also used for initial evaluation.[61] As a last diagnostic tool, endomyocardial biopsy may be helpful in diagnosing the underlying cause of myocardial disease.[62]

MANAGEMENT AND PREVENTION

As the changes resulting from EFE appear irreversible once they occur, it is crucial to prevent progression, starting as early as intrauterine development. In-utero aortic valvuloplasty has demonstrated improvements in LV ejection fraction and a decrease in moderate to severe mitral regurgitation.[63] Patients with left heart obstructive disease and EFE have shown improved LV function with primary left heart rehabilitation with resection of EFE and valvuloplasty.[64] Eliminating precipitating factors, such as repairing coarctation of the aorta alongside EFE, can reduce left ventricular workload and improve the patient's condition.[65] Sharp dissection of the thickened endocardium in a fibrotic near-obliterated left ventricle that failed medical therapy, showed postprocedural compliance improvement.[66]

While some individuals may have a more chronic course and respond to heart failure medications, however, others may not respond and might require heart transplantation.[13] Heart transplantation emerged as a successful method of managing EFE, introduced in 1994 by Frazier and associates.[67]

COMPLICATIONS

Thromboembolic events have always been associated with EFE; observations include mural thrombosis, and multiple emboli in the brain, kidney, and lungs.[68] Additionally, it can lead to complications such as myocardial infarction and calcific arterial lesions.[69] In patients who underwent EFE removal, electrical desynchrony, including elongation of the QRS was noticed, However, the duration of QRS was correlated with LV size.[70] An ischemic pattern on the ECG, such as anterolateral or inferior wall infarction, can serve as a negative prognostic factor for survival in patients with EFE and is typically associated with death.[71]

Recurrence of EFE due to flow disturbance is possible in patients with valvar defects.[72] Sudden cardiac death has been observed in patients with EFE, even without any evidence of premortem heart disease. Postmortem examinations have revealed EFE along with other histologic abnormalities of the heart such as dilated cardiomyopathy and hypoplastic coronaries.[73,74]

DIFFERENTIAL DIAGNOSIS OF ENDOCARDIAL FIBROELASTOSIS

In evaluating patients with suspected EFE, clinicians must consider other potential conditions, including congenital heart diseases, particularly those involving the left ventricular outflow tract obstruction and an anomalous left coronary artery from the pulmonary artery. Additionally, other causes of nonimmune hydrops fetalis, neonatal lupus erythematosus, left ventricular noncompaction,

and centronuclear myopathy should be included in the differential diagnosis.[51] Special attention should be given to detecting treatable causes, such as congenital heart disease or systemic carnitine deficiency. In subsequent pregnancies, there is a 3–5% risk of EFE, warranting fetal echocardiography for early diagnosis. Additionally, a list of differential diagnoses is provided to guide a comprehensive evaluation.[75]

CONCLUSIONS

EFE, whether as a primary or secondary manifestation, appears to occur in the context of various cardiac and noncardiac abnormalities. Its origin remains unclear; however, reports have linked it to a wide spectrum of causes including autoimmune, infective, and metabolic conditions. Further investigation is warranted to determine the cause for proper prevention and management of this phenomenon.

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