Endocardial Fibroelastosis

A rare condition characterised by pronounced, diffuse, thickening of the ventricular endocardium. It appears as unexplained heart failure in infants and children.\(^1\)

Pathogenesis

The disease can be primary or secondary, although some argue it is only ever secondary.\(^2\)

The two forms of primary endocardial fibroelastosis (EFE) are dilated (most common), and contracted.\(^3\) It has been suggested that one form may progress to the other.\(^4\)

- Primary dilated EFE occurs when the heart is otherwise normal and there is no other cause of unexplained heart failure.
- Secondary dilated EFE is associated with aortic stenosis or atresia.
- Secondary contracted EFE is associated with hypoplastic left heart syndrome.

Epidemiology

- It is rare - only 1-2% of all congenital heart diseases. The number of cases has fallen dramatically in recent years, possibly secondary to better antenatal scanning.
- It may be familial (10%) with a predominantly X-linked pattern.\(^3\)
- It affects both sexes equally, usually presenting during the first 3-6 months of life in 80% of cases.
- Typical age of diagnosis is 2-12 months. It rarely is reported in adolescents and adults.
- It is an important cause of non-immune hydrops fetalis.\(^5\)

Aetiology

Possible factors include:

- Intrauterine viral infection (mumps,\(^6\) Coxsackievirus B).
- Neonatal lupus syndrome.\(^7\)
- Subendocardial ischaemia.
- Impaired lymphatic drainage of the heart.
- Systemic carnitine deficiency.
- Familial.

Presentation

It presents with typical signs of congestive heart failure (CHF) in previously healthy children less than 6 months old.

Symptoms
Breathlessness
Cough
Wheezeing
Feeding difficulty
Excessive sweating
Failure to thrive
Recurrent chest infections

In children, remember that severe abdominal pain may indicate coronary insufficiency.

**Signs**

Onset may be acute and result in cardiogenic shock or sudden death.\(^9\)

- Respiratory distress during feeding - tachypnoea, grunting, subcostal or intercostal recession.
- Fever.
- Pallor (anaemia).
- Peripheral cyanosis.
- Cardiomegaly - normal or quiet first and second heart sounds, a gallop rhythm with an audible third heart sound.
- Apical pansystolic murmur.
- Hepatomegaly.
- Oedema.
- Rash.
- Leukocytosis.

There is also an increased risk of thromboembolic episodes.

It may present as part of Barth's syndrome, which comprises dilated cardiomyopathy ± endocardial fibroelastosis.\(^10\)

**Investigations**

- **Blood tests:**
  - Blood urea and creatinine.
  - FBC.
  - Autoantibody profile (including anti-Ro and anti-La).\(^{11}\)
  - Blood culture tests indicated for management of acute episodes.

- **CXR:**
  - Cardi thoracic (CT) ratio exceeds 0.65.
  - Left lower lobe atelectasis may be seen.
  - The cardiac silhouette is often globular.
  - Pulmonary venous congestion is common.

- **ECG:**
  - Tall R waves.
  - Deep Q waves.
  - T-wave inversion or flattening in the left precordial or inferior lead.
  - Findings depict left ventricular (LV) hypertrophy.
  - Right axis deviation and isolated right ventricular (RV) hypertrophy (more common in the first few weeks of life).
  - Left, right (or both) atrial enlargement.
  - Wolff-Parkinson-White syndrome, left bundle branch block, supraventricular and ventricular arrhythmias and varying degrees of atrioventricular block.
  - The early and terminal stages of heart failure show low-voltage tracings.
Echocardiography:\[12\]
- Increase in left atrium and LV dimensions.
- Reduced ejection fraction.
- Abnormal mitral valve motion.
- Dense echogenicity along the endocardium of the LV.
- A varying degree of mitral regurgitation is common.

Fetal echocardiography:
- Valuable tool for early identification, particularly of the secondary type.
- Doppler studies may be useful if the morphology is unclear.\[13,14\]

Electron beam CT can demonstrate calcification and fibrosis of the ventricles (particularly at apex).\[15\]
Cardiac MRI.\[16\]

Management
This is the same as chronic cardiac failure:

- Acute exacerbations are often precipitated by respiratory infections.
- Early and prolonged treatment with digoxin is suggested. This may result in clinical improvement and reversion of the cardiac enlargement to normal. Therapy should be continued for several years after the symptoms disappear, as stopping the drug may result in acute cardiac failure.
- Steroid therapy (dexamethasone) has been shown to cure fetal endocardial fibroelastosis (EFE) and AV conduction delays associated with maternal anti-Ro and anti-La antibodies.\[17\]
- Supportive measures for acute failure and exacerbations may be required, eg treatment of infection and anaemia.
- Thromboembolic complications may require anticoagulation.

Surgical
Cardiac transplantation may be recommended for those with end-stage disease.

Prognosis
The prognosis for primary endocardial fibroelastosis (EFE) is relatively poor, although the condition is not universally fatal. There is a 4-year survival rate of 77%.\[8\]

- Infants presenting with acute failure almost always die from the acute episode, unless they receive a transplant.\[18\]
- Those with a chronic presentation have a 30-40% mortality rate from resistant heart failure.
- Acute congestive heart failure (CHF) becomes progressive CHF. Death occurs within weeks, usually in the first six months of life.
- One third of the patients survive and they can experience persistent symptoms, show residual ECG abnormalities or show evidence of cardiomegaly.
- Episodes of CHF recurring more than six months after initial onset of symptoms also indicate a poor prognosis.

Further reading & references
3. Endocardial Fibroelastosis, Online Mendelian Inheritance in Man (OMIM)
8. Venugopalan P; Endocardial Fibroelastosis, Medscape, Jun 2010

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