# Autopsy Findings of Endocardial Fibroelastosis in an Adult

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**Abstract**

**Background:** Endocardial fibroelastosis (EFE) is commonly presented with diffuse endocardial thickening due to deposition of collagen and elastin, usually affecting the left ventricle of the heart. **Case presentation:** A 59-year-old gentleman who had no known medical illness had collapsed while performing house chores at home. Autopsy examination showed cyanosed lips and nail beds. The heart weighed 360 gm with distinct presence of whitish, firm and thickened endocardium of the left ventricle. The anterior descending coronary artery showed an almost complete occlusion by atheroma, in keeping with coronary artery disease. Mild pulmonary oedema was present. Histological examination revealed fibroelastosis forming plaque-like covering of the inner ventricle, with patchy myocardial fibrosis. Numerous 'heart failure' cells were present in the intraalveolar spaces. **Conclusions:** This case showed remarkable autopsy findings of EFE in adult with evidence of congestive heart failure episodes in his lifetime.

**Keywords**: Endocardial fibroelastosis; Cardiomyopathy; Sudden cardiac death; Autopsy

## Background

Endocardial fibroelastosis (EFE) is a rare heart disease which usually presents with diffuse endocardial thickening due to deposition of collagen and elastin, most commonly affecting the left ventricle.[1,2] EFE can be categorized into primary and secondary. Primary EFE refers to an absence of any causative factor, therefore it is hypothesized to be caused by genetic factors, viral infections or transplacental crossing of maternal antibodies.[1,3] Secondary EFE occurs as a result of structural heart diseases, particularly related to the left ventricular outflow tract obstructions.[1] An underlying condition includes congenital cardiac malformation, aortic stenosis, coarctation of the aorta, hypoplastic left heart syndrome and ventricular septal defect.[1,4] This pathology is commonly found in infants and children and its progressive nature usually leads to heart failure and death.[5] EFE in adults is rarely diagnosed and reported. Therefore, we report a case of an adult man who had collapsed and pronounced dead at home, and EFE was diagnosed at autopsy.

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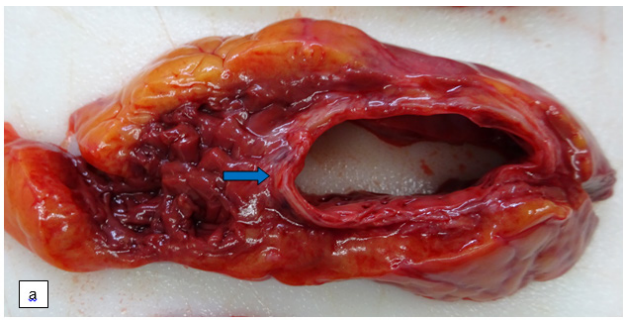
## Case Presentation

A 59-year-old male subject was brought to the forensic department for a medico-legal autopsy examination. He was pronounced dead at home by a paramedic who responded to an ambulance call made by his wife, as he collapsed in the living room while performing some house chores. There was minimum history which could be obtained. He was an immigrant with no medical record and no history of illness known to his spouse.

Autopsy examination revealed a male subject, measuring 169 cm in length and 90 kg in weight. Post-mortem changes such as lividity and rigor mortis were present. Slight bluish discolouration of the lips and nail beds was present, indicating cyanosis. Internal examination of the skull and brain showed intact skull with foci of ischaemic infarction. The thoracic cavity was clear of effusion or adhesions. The lungs were relatively heavy, weighing 400 gm and 450 gm on the right and left sides respectively. On cut surfaces, the lungs were slightly firm, however, there was no apparent consolidation seen. Mild pulmonary oedema was present. The heart weighed 360 gm. On cut surfaces, the endocardium of the left ventricle was whitish, firm and distinctly thickened, with some areas appeared almost detached from the myocardium (Fig. 1a). The right ventricle was normal. The coronary arteries showed an almost complete occlusion of the left anterior descending coronary artery located at the proximal third of the vessel, 3.5 cm in length. There was no evidence of acute myocardial infarction seen. Examination of the other internal organs such as the liver, spleen, kidneys, and intestines generally showed congestion, with no gross pathology observed. Representative tissue samples from the heart, lungs and other internal organs were obtained for histology examination.



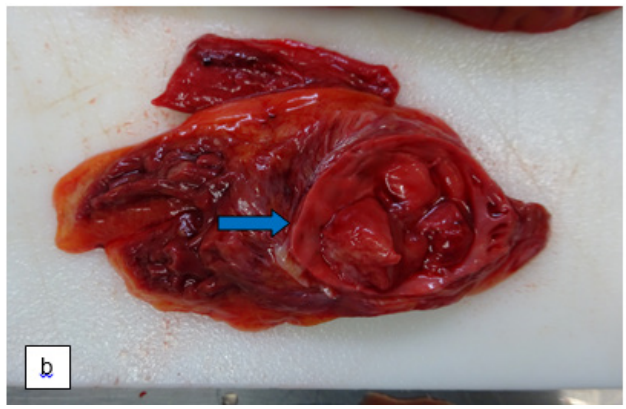
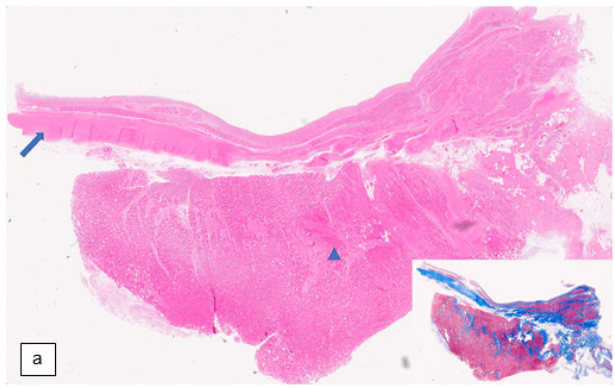




**Fig. 1 (a): Cross section of the heart at the mid-ventricular level shows a diffuse thickening of the endocardium of the left ventricle. The fibrotic endocardium appears slightly detached from the myocardium (blue arrow). (b): The apex shows a firm layer of fibrous tissue lining the endocardium of the left ventricle (blue arrow).**

### *Microscopic findings*

The endocardial layer was markedly thickened by fibroelastosis, involving the entire left ventricular wall, forming a plaque-like covering of the inner ventricle (Fig. 2a). Patchy myocardial fibrosis is also seen in the subendocardial myocardium, confirmed by Masson Trichrome stain (Fig. 2a, inset). Eosinophils were inconspicuous and there were no microscopic features of an acute myocardial infarction seen. The lungs show vascular congestion with numerous intraalveolar haemosiderin-laden macrophages, consistent with heart failure (Fig. 2b). Representative sections from other organs were non-contributory. In view of the gross autopsy and microscopic findings, the cause of death was concluded as 'coronary artery disease compounded by endocardial fibroelastosis'.



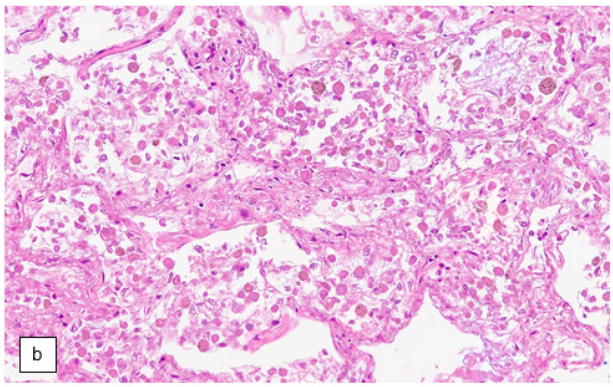



**Fig. 2 (a). The endocardial fibrosis forms a plaque-like covering (arrow). The fibrosis merges with the underlying myocardium. Patchy myocardial fibrosis is also seen (arrowhead). H&E and Masson Trichrome (inset). (b) The intraalveolar spaces contain numerous haemosiderin-laden macrophages. H&E 20x.**

## Discussion

EFE is characterized by a thickened fibrotic or elastic layer in the endocardium, resulting in reduced ventricular distensibility and impaired diastolic filling. EFE may be primary (idiopathic) or secondary to an existing cardiac anomaly causing severe left ventricular outflow tract obstruction such as aortic stenosis or atresia, mitral incompetence, coarctation of aorta and patent ductus arteriosus.[2,6] Primary EFE is often diagnosed in infancy or childhood, with reports illustrating potential associations with foetal hydrops, inherited conditions such as X-linked recessive cardiomyopathy and mutations involving the MYBPC3 gene.[1,5,7] Only rare cases present in adulthood.[2,8,9]

Until now, the diagnosis of EFE is difficult to be established due to its non-specific presenting clinical symptoms and echocardiographic findings.[2] As this disease is more commonly seen in infants, the typical age of diagnosis is 2 to 12 months. Infants in this age group may present with unexplained acute heart failure and cardiogenic shock, which may lead to sudden infant death.[5,6] Similarly, EFE in adults may have presenting symptoms such as cyanosis and progressive dyspnea, indicating congestive heart failure of unknown cause.[6] Sudden death without any apparent clinical symptoms also occurs in adult.[8]

Our case is an example of EFE diagnosed in adulthood, discovered during an autopsy examination. While cases in infancy and childhood tend to present with rapidly progressive clinical course, cases diagnosed in adulthood may have relatively mild symptoms or progress at a slower rate in comparison.[2,10] There were no overt structural cardiac anomalies present in our case to ascribe as a potential cause for secondary EFE. The fact that the deceased was an immigrant in the country, a good medical history and clinical investigations record were impossible to be obtained. Even the actual nature of his final clinical presentations could not be established, therefore, the authors had to rely completely on the macroscopic and microscopic findings at autopsy to hypothesize the possible clinical symptoms the deceased had in his lifetime, attributed by the EFE.

It is plausible that the EFE had contributed to some degree of ventricular dysfunction, which, in concert with severe coronary artery luminal occlusion, resulted in an acute ischaemic event leading to his sudden demise. It is worth to note that patchy myocardial fibrosis which was present throughout the underlying myocardium of the left ventricular wall may be explained by reduced diffusion capacity from the ventricular lumen, as a direct result of the subendocardial thickening. The presence of numerous intraalveolar haemosiderin-laden macrophages ('heart failure' cells) is highly suggestive of past episodes of congestive heart failure, however, there was no sign of acute heart failure discovered at the time of death.

## Conclusions

In conclusion, the findings of this case are consistent with EFE, with accompanying patchy myocardial fibrosis. As the heart also showed presence of coronary artery disease, in the absence of gross and microscopic findings of acute myocardial infarction, we hypothesized that a very acute ischaemic event could have been the immediate cause of death. While the EFE certainly did not cause the patient to be in failure at the time of death, the presence of numerous 'heart failure' cells in the lungs were the tell-tale sign that he probably had episodes of congestive heart failure with a fairly good recovery in his lifetime.

**Conflict of interest:** The authors declare that they have no competing interests.

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**Ethics approval and consent to participate:** We seek for waiver of ethical review and approval since the data were not directly indicative of the individual subject, observatory in nature and the research involved no risk to the deceased subject.

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**Authors' contributions:** RR and NAMA performed the autopsy examination. They were involved in the conceptual design of the study and major contributor in writing the manuscript. MAA performed the histological examination and wrote the relevant part in the Microscopy findings and Discussion section. All authors read and approved the final manuscript.

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**List of abbreviations**

**EFE:** Endocardial fibroelastosis

**H&E**: Hematoxylin and eosin

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