

# Endocardial Fibroelastosis Mimicking Dilated Cardiomyopathy in a Neonate: A Case Report


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## Abstract

We report a case of a 20-day-old female child who was full-term born by emergency Caesarean section due to foetal distress. The baby had intermittent dyspnoea and was pronounced dead with unsuccessful resuscitation efforts in her admission to the hospital. Post-mortem examinations revealed dilated cardiomyopathy with endocardial fibroelastosis. Histologic examination of the heart showed mild endocardial fibroelastosis and sub-endocardial and interstitial increased elastin and collagen fibre deposition. Molecular testing was heterozygous for ELAC2 (pathogenic) and PRDM16 (variant of uncertain significance [VUS] genes. Other ancillary test results are non-contributory. Based on the autopsy findings, ancillary test results, and clinicopathological correlation, the cause of death is neonatal dilated cardiomyopathy (DCM).

**Keywords:** Neonate; dilated cardiomyopathy; respiratory failure; endocardial fibroelastosis.

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## Introduction

We report a case of a 20-day-old female child who was full-term born by emergency Caesarean section due to fetal distress. The baby had intermittent dyspnoea and was pronounced dead with unsuccessful resuscitation efforts in her admission to hospital. Postmortem examinations revealed dilated cardiomyopathy with endocardial fibroelastosis. Histologic examination of the heart showed mild endocardial fibroelastosis and sub-endocardial and interstitial increased elastin and collagen fibre deposition. Molecular testing was heterozygous for ELAC2 (pathogenic) and PRDM16 (variant of uncertain significance [VUS] genes. Other ancillary test results are non-contributory. Based on the autopsy findings, ancillary test results, and clinicopathological correlation, the cause of death is neonatal dilated cardiomyopathy (DCM).

## Case report

A 20-day-old female was full-term born by emergency Caesarean section due to foetal distress. She had respiratory distress and required oxygen and positive-pressure ventilation. A few days later, she again developed respiratory difficulties and was admitted to the neonatal intensive care unit, where she was treated for sepsis and improved over a few days. A few days later, she was admitted to the hospital with retching and respiratory difficulties,

and resuscitative efforts were tried, which were ultimately unsuccessful.

A post-mortem examination of the infant was conducted. The growth parameters are appropriate for the age. Pre-autopsy radiology shows cardiomegaly but is devoid of any structural skeletal anomalies. (Figure 01).

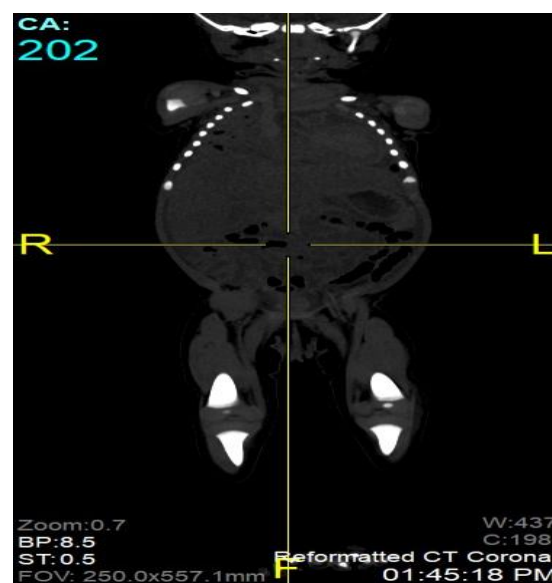


Figure 01. CT Scan with cardiomegaly

There was no evidence of trauma or dysmorphic features. The internal examination revealed an enlarged heart with a globular left ventricle, patent foramen ovale, and large anatomically patent arterial duct, tricuspid and mitral valve leaflet thickening, and dilatation of the main pulmonary artery and ascending aorta. (Figures 02,03,04).

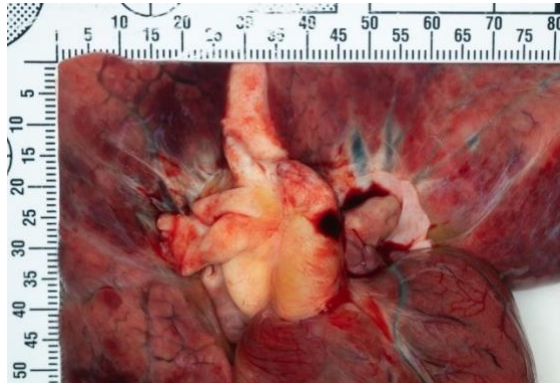


Figure 02. Dilatation of the main pulmonary artery and ascending aorta.

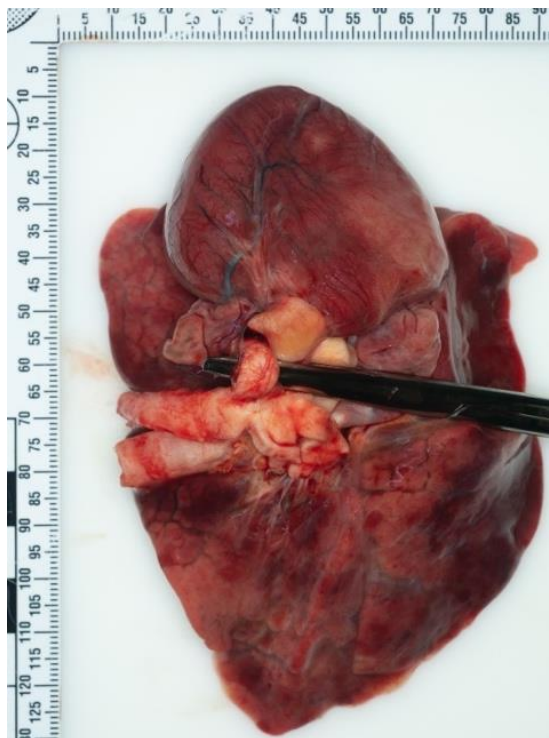


Figure 04. Enlarged heart with a globular left ventricle

Histologic examination of the heart showed myocardial disarray (Figure 05) mild endocardial fibroelastosis and sub-endocardial and interstitial increased elastin and collagen fibre deposition. (Figures 06,07)

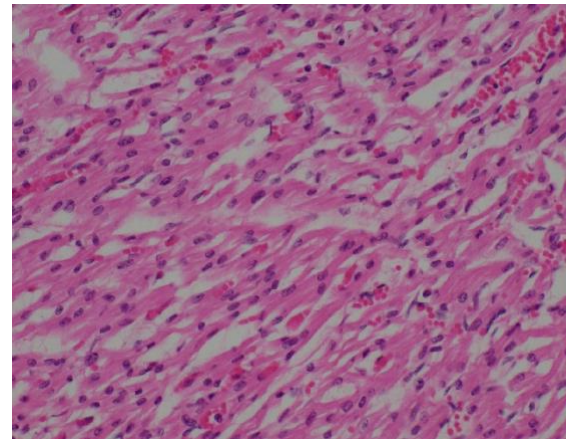


Figure 05. Photomicrograph of Heart (20x10 Hematoxylin and Eosin) – Myocardial disarray

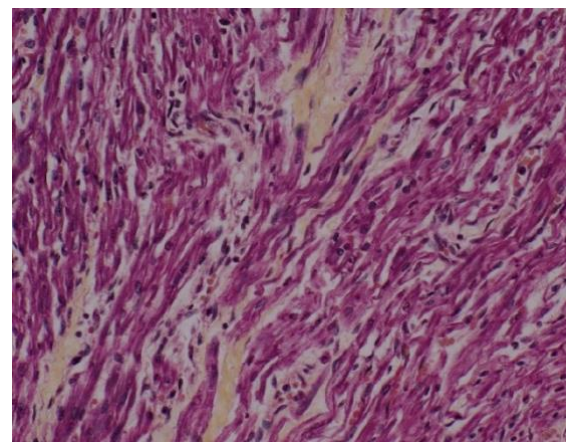


Figure 06. Photomicrograph of Heart (20x10 Movat pentachrome) - interstitial and perivascular fibrosis

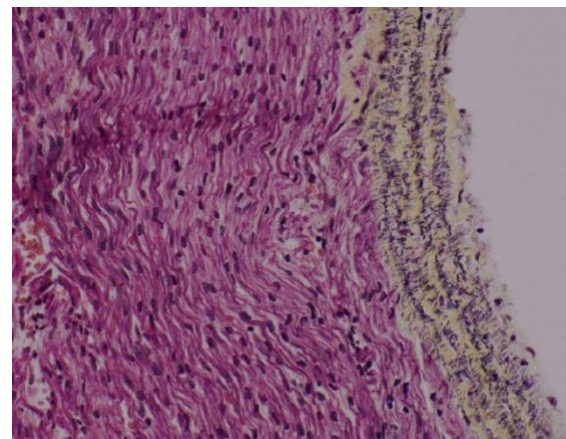


Figure 07. Photomicrograph of Heart (20x10 Movat pentachrome) – Endocardial Fibroelastosis (EFE)

Microbiology, Toxicology, biochemical, and metabolic analysis were negative or non-contributory. Molecular testing was requested, and the genetic panel included Arrhythmia, Cardiomyopathy and Congenital Heart Disease panels. The test results were heterozygous for ELAC2 (pathogenic) and PRDM16 (variant of uncertain significance [VUS] genes. (Table 01)

Table 01. Genetic Panel

Gene	Variant	Zygosity	Variant Classification
ELAC2	c.2342G>A (p.Arg781His)	Heterozygous	Pathogenic
PRDM16	c.3106C>T (p.Pro1036Ser)	Heterozygous	Uncertain Significance

### Discussion

Our case presented with induced dilated cardiomyopathy secondary to endocardial fibroelastosis. Pediatric cardiomyopathy is a rare heart condition that affects infants and children.

Several different types of cardiomyopathies exist, and the specific symptoms vary from person to person. In some affected individuals, this can be asymptomatic. In many children, cardiomyopathy is a progressive condition that may result in an impaired ability of the heart to pump blood, fatigue, heart block, irregular or rapid heartbeats, and, potentially, heart failure and sudden cardiac death.

Cardiomyopathy may also be termed primary or secondary. The primary is genetic defect [6,7], viral or bacterial infections, or exposure to certain toxins. Approximately 79 percent of pediatric cardiomyopathy is idiopathic. Some disorders affect the heart and other organ systems called secondary cardiomyopathy.

Often fetal endocardial fibroelastosis can be caused by several factors, such as intrauterine viral myocarditis (mumps, coxsackie virus B), autoimmune reaction, and mucopolysaccharidosis. Fetal Endocardial Fibroelastosis could also be secondary to aortic stenosis, dilated cardiomyopathy, or left ventricular noncompaction [8].

Dilated Cardiomyopathy (DCM) is characterized by dilatation and impaired contraction of the left ventricle or both ventricles and commonly results in congestive heart failure.

Endocardial fibroelastosis is a distinct pathologic finding in some cases of DCM characterized by the presence of glistening white fibroblastic tissue (consisting of collagen and elastic hyperplasia) over the endocardium with some subendocardial invasion, usually involving left ventricle and left atrium, and rarely, the right ventricle.

### Conclusions

This case report suggests that Endocardial fibroelastosis-induced pediatric heart failure may present in neonates with or without any apparent precipitants. The associated clinical presentations are macroscopically like that of pediatric Dilated Cardiomyopathy. Based on the microscopy, clinicopathologic correlation and genetic studies, the cause of death, in this case, is neonatal dilated cardiomyopathy.

### Disclosure statement

**Conflicts of interest:** The author declares that she has no conflicts of interest.

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