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REVIEW ARTICLE

# Neonatal dilated cardiomyopathy

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**KEYWORDS**
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Management

**Abstract** Cardiomyopathies are rare diseases of the heart muscle, of multiple causes, that manifest with various structural and functional phenotypes but are invariably associated with cardiac dysfunction. Dilated cardiomyopathy is the commonest cardiomyopathy in children, and the majority present before one year of age. Its etiology may be acquired or genetic. Myocarditis is an important cause and is responsible for the majority of acquired cases. Inherited (familial) forms of dilated cardiomyopathy may occur in 25-50% of patients. Echocardiographic and tissue Doppler studies are the basis for diagnosis of dilated cardiomyopathy in most patients. Marked dilatation of the left ventricle with global hypokinesis is the hallmark of the disease. This review will cover the classification, epidemiology and management of newborns with dilated cardiomyopathy. In particular, a comprehensive and up-to-date review of the genetic study of dilated cardiomyopathy and of detailed echocardiographic assessment of these patients will be presented.


**PALAVRAS-CHAVE**
Cardiomiopatia dilatada;
Neonatal;
Estudo genético;
Avaliação ecocardiográfica;
Abordagem

## Cardiomiopatia dilatada com apresentação neonatal

**Resumo** As cardiomiopatias são doenças raras do músculo cardíaco, de múltiplas causas, que se manifestam com vários fenótipos estruturais e funcionais, mas invariavelmente associadas a disfunção cardíaca. A cardiomiopatia dilatada é a forma mais comum em crianças, apresentando-se maioritariamente antes do ano de idade. A sua etiologia pode ser adquirida ou genética. A miocardite é uma importante causa, responsável pela maioria dos casos adquiridos. As formas hereditárias (familiares) de cardiomiopatia dilatada podem ocorrer em 25-50%

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dos pacientes. Os estudos ecocardiográficos e Doppler são a base para o seu diagnóstico, caracterizando-se por uma acentuada dilatação do ventrículo esquerdo com hipocinesia global. Neste artigo serão abordadas a classificação, a epidemiologia e a abordagem dos recém-nascidos com cardiomiopatia dilatada. Em particular, será apresentada uma revisão abrangente e atualizada da avaliação genética desta entidade, bem como aspetos detalhados da ecocardiografia nestes pacientes.


**List of abbreviations**

| | |
|---|---|
| ACE | angiotensin-converting enzyme |
| AD | autosomal dominant |
| AHA | American Heart Association |
| AR | autosomal recessive |
| ARVC/D | arrhythmogenic right ventricular cardiomyopathy/dysplasia |
| ASE | American Society of Echocardiography |
| BNP | B-type natriuretic peptide |
| CM | cardiomyopathy |
| CMR | cardiovascular magnetic resonance |
| CO | cardiac output |
| DCM | dilated cardiomyopathy |
| DD | diastolic dysfunction |
| EF | ejection fraction |
| ESC | European Society of Cardiology |
| HCM | hypertrophic cardiomyopathy |
| HF | heart failure |
| IEM | inborn errors of metabolism |
| ISFC | International Society and Federation of Cardiology |
| LV | left ventricular |
| LVEF | left ventricular ejection fraction |
| MR | mitral regurgitation |
| RCM | restrictive cardiomyopathy |
| WHO | World Health Organization |

## Introduction

Cardiomyopathies (CMs) are a group of diseases of the myocardium associated with mechanical and/or electrical dysfunction that usually (but not invariably) exhibit inappropriate ventricular hypertrophy or dilatation and are due to a variety of causes that frequently are genetic. CMs are either confined to the heart or are a part of generalized systemic disorders, often leading to cardiovascular death or progressive heart failure (HF)-related disability.[1]

Different definitions and nomenclature have been proposed for this important and heterogeneous group of diseases. Neonatal CMs are diseases of the heart muscle of the neonate in which the myocardium is affected without primary abnormalities of the valves, great vessels or septum.[1] They include a variety of myocardial disorders that manifest with various structural and functional phenotypes. Neonatal CMs account for about 1% of childhood cardiac disease, with an estimated incidence of 10:100 000 live births, and are responsible for 10% of all pediatric cardiac deaths.[2]

In this review we focus on clinical aspects, management, prognosis and follow-up in dilated cardiomyopathy (DCM) of the newborn.

## Definition and classification

CMs are rare diseases of the heart muscle, of multiple causes, that manifest with various structural and functional phenotypes but are invariably associated with cardiac dysfunction.

At present there is no consensus on how to classify CMs on the basis of etiology, physiology or treatment, as their origin and pathophysiology are not well understood.

The first classification to take anatomical presentation into consideration was the 1995 World Health Organization (WHO)/International Society and Federation of Cardiology (ISFC) report, which divided myocardial disease into DCM, hypertrophic cardiomyopathy (HCM), restrictive cardiomyopathy (RCM), arrhythmogenic right ventricular cardiomyopathy/dysplasia (ARVC/D), and unclassified cardiomyopathies.[3]

The American Heart Association (AHA) and the European Society of Cardiology (ESC) published classifications based on etiology and pathophysiology.[4,5] In the AHA classification, CMs are categorized into two groups: primary (predominantly involving the heart) and secondary (accompanied by systemic involvement of other organs). Primary CMs are subdivided into genetic, mixed (predominantly non-genetic; less commonly genetic), or acquired. Genetic CMs include hypertrophic cardiomyopathy, ARVC/D, left ventricular noncompaction, glycogen storage diseases, conduction defects, mitochondrial myopathies, and ion channel disorders. Mixed CMs include DCM and RCM. Acquired CMs include myocarditis, stress-induced (Takotsubo), peripartum and tachycardia-induced CM, and CM in infants of insulin-dependent diabetic mothers.[6]

The AHA and ESC classification systems differ from the earlier WHO/ISFC classification in emphasizing the distinction between familial/genetic and non-familial/non-genetic