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ADVANCES IN PEDIATRICS

Conduction Defects/ Cardiomyopathies

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Keywords

• Cardiomyopathies • Conduction disorders • Cardiac arrhythmias
• Inherited diseases • Sudden death

Key points

- Congenital heart block is frequently associated with maternal lupus.
- Carnitine arrhythmias may result in sudden death.
- Histiocytoid cardiomyopathy frequently results in death by the first 2 years of life.
- Barth syndrome is X-linked and is a mutation of Xq28-linked *G4.5 (TAZ)*.

INTRODUCTION

Conduction disorders result in cardiac arrhythmias that may be fatal and are inherited with arrhythmias and sudden death that are prominent features and include histiocytoid cardiomyopathy, arrhythmogenic right ventricular dysplasia, isolated noncompaction of the left ventricular myocardium, long QT syndrome, Brugada syndrome, congenital short QT syndrome, catecholaminergic polymorphic ventricular tachycardia, and carnitine deficiency. Although the histologic appearance of some of these disorders may be diagnostic, molecular analysis is necessary to clearly define the particular type of cardiomyopathy.

CONGENITAL HEART BLOCK

Congenital heart block (CHB) may be present in infants with both anatomically normal and malformed heart [1]. Most cases are sporadic; CHB occurs in 1 in 20,000 live births [2]. It may be a manifestation of neonatal lupus erythematosus (NLE).

The 2 most common neonatal manifestations of NLE are CHB and cutaneous lesions; approximately 50% of affected patients present with one or

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the other system involvement. The skin lesions most commonly appear on the infant's face, scalp, and upper trunk, resembling the lesions of discoid lupus erythematosus.

The manifestations of NLE result from the transplacental passage of maternal autoantibodies to the fetus. In more than 95% of the patients, the autoantibodies found in the mother and infant are anti-Ro, also known as Sjögren syndrome A antibodies (SSA) or anti-Ro/SSA, anti-La (SSB) antibodies, and antiribonucleoprotein (RNP) antibodies. Most mothers who have infants with this syndrome have no symptoms of collagen vascular disease; most infants of mothers with anti-Ro/SSA, anti-La, or anti-RNP antibodies do not develop NLE syndrome. The risk of a mother having a second child with NLE is about 25%.

CHB may be present in 50% of infants with neonatal systemic lupus erythematosus (SLE) [3]. Endocardial fibroelastosis (EFE) is commonly present; the Atrioventricular node is often absent or scarred, and the sino-atrial (SA) node and ventricular components may be calcified and fibrotic [4,5]. Complete heart block and other conduction defects with a high association with Ro antibodies implicate transplacental transfer of the antibody [6]. CHB develops in utero, usually during the second trimester. The heart block, unlike the other manifestations, is permanent. About one-quarter of infants with CHB have associated structural congenital cardiac anomalies, including atrial septal defect or ventricular septal defect, transposition of great arteries, and anomalous pulmonary venous drainage. Rarely, affected infants may have pericarditis, myocarditis, or a cardiomyopathy.

Because of the strong association of CHB with maternal lupus, every infant with CHB should undergo laboratory studies for anti-Ro/SSA, anti-SSB, and anti-RNP antibodies. Mothers of infants with CHB may have a detectable soluble tissue RNP antigen, anti-Ro/SSA, in their serum [7]. Immunoglobulins (Ig) IgG and IgA may be detected in the SA node, epicardium, and nerves [8].

Most infants with neonatal SLE have characteristic skin lesions and cardiac problems [9]. The characteristic annular erythematous macules, papules, or plaques of neonatal SLE may be present at birth, but they usually appear on the scalp and elsewhere by 2 months of age and disappear by 6 months of age.

LONG QT SYNDROME

Several genes that have been identified (Table 1) encode for cardiac ion channels for potassium ion channels or sodium ion channels. Mutations in these genes cause disturbed function of these channels which are called *channelopathies*. In each case, the altered ion channel function produces prolongation of the action potential and propensity to *torsade de pointes* ventricular tachycardia. Characteristic findings are prolongation of the QT interval and T-wave abnormalities on the electrocardiogram (EKG). However, the QT interval at presentation is normal about 10% of the time and just borderline prolonged another 30%, so diagnosis may be difficult. The symptoms are syncope and sudden death, typically occurring during exercise or emotional upset. The