

Rarer cardiomyopathies in children and adolescents: the experience of a paediatric pathologist

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Cardiomyopathies (CM) are diseases of the heart muscle. Secondary diseases due to congenital malformations are excluded. Cardiomyopathies are classified according to the dominant pathology or by etiologic factors. Some cardiomyopathies have a genetic basis while others are acquired.

Incidence of paediatric CM is between 1.13 and 1.24 cases per 100,000 children. Highest incidence is in the first year of life and second peak in adolescence. Dilated CM is the most frequent type in children. The two most frequently identified causes are myocarditis and neuromuscular disease. These are some of the CM encountered in our routine autopsy practice:

Duchenne CM: CM is common in NM disorders in children. There is an overlap between mutations in primary skeletal muscle myopathies and primary CM (dystrophin gene in dilated CM in Duchenne muscular dystrophy [DMD]). Cardiac screening recommended in children with NM disease. By 20 years of age 80-90% of DMD have CM. Histology shows replacement by adipose cells and fibrosis.

Histiocytoid CM: rare X-linked disorder of infancy and childhood, predominantly girls below 2 years of age. Manifests as severe cardiac arrhythmias or dilated CM with heart failure. Hamartomatous lesion of Purkinje cells frequently associated with mitochondrial DNA mutations. May present in the fetus. Histologically collections of myocytes with vacuolated cytoplasm resembling histiocytes.

Mitogenic CM: rare form of dilated CM. more ventricular than atrial dilatation and endocardial fibroelastosis. Histology shows hypertrophic nuclear changes and increased mitotic activity including atypical mitoses. Recently described.

Arrhythmogenic right ventricular CM: Rare in children, more in adolescents and young adults. Autosomal dominant. Recessive form associated with skin problems (Naxos disease). Five desmosomal genes identified. Progressive fibro fatty replacement of myocardium usually starts in subepicardial or medial layers and progresses to subendocardial. Inflow, apex and infundibulum most frequently involved. With time may also involve the left ventricle.