INTRODUCTION

Cardiomyopathy is a myocardial disorder in which the heart muscle is structurally and functionally abnormal in the absence of haemodynamically significant structural heart disease as primary structural, congenital, or valvular heart disease significant enough to cause the observed myocardial dysfunction **(Lipshultz** *et al.,* **2003).**

The types of cardiomyopathy is grouped according to ventricular morphology and function into: **(Thiene** *et al.,* **2004).**

- Dilated cardiomyopathy (DCM).
- Hypertrophy cardiomyopathy (HCM).
- Restrictive and infilterative cardiomyopathy (RCM).
- Arrhythmogenic right ventricular cardiomyopathy (ARVC).
- Unclassified.

Although rare, the importance of cardiomyopathies lies in the fact that around one-third of cardiomyopathies will progress to death or require cardiac transplantation. Cardiomyopathies are the most common reason for cardiac transplant in childhood (**Boucek** *et al.,* **2005).**

Although major advances have been made in the management of childhood heart disease, there has been only minimal change in outcome over the past 30 years for those with cardiomyopathies. Cardiomyopathy may be the first manifestation of both an inborn error of metabolism or neuromuscular disorder with attendant genetic implications. Other associations with metabolic disease, dysmorphic syndromes, and neuromuscular disease are important to establish, particularly in pediatric

patients, Survival in children with dilated cardiomyopathy depends on accurate diagnosis and aggressive therapy (**Pereira** *et al.*, **1999***).*

Metabolic cardiomyopathy in pediateric accounts for approximately 15% of all cardiomyopathies presenting in pediatric age group. Several groups of disorders associated with cardiac hypertrophy, such as the mucopolysaccharidoses (e.g., Hurler syndrome), are easily diagnosed because of evident extracardiac manifestations. Cardiomyopathies due to a defect in mitochondrial oxidative metabolism are usually associated with multi-system abnormalities (e.g., central nervous system deterioration, skeletal, and ocular complaints). On the other hand, the diagnosis of mitochondrial fatty-acid oxidation defects is often delayed because patients are typically asymptomatic until they manifest acutely with often-catastrophic metabolic decompensations. Timely establishment of the correct etiology of cardiomyopathy is therefore important for initiation of treatment and/or prediction of a patient's long-term outcome **(Nugent** *et al.*, **2003).**

The clinical presentation of patients with metabolic cardiomyopathies is highly variable; however, a thorough history and physical examination can point to a possible diagnosis (**Andrews** *et al.*, **2008).**

There are many metabolic cardiomyopathies which present with a phenotype of ventricular hypertrophy and reduced systolic function. They do not clearly fit into the categories of hypertrophic or dilated cardiomyopathies. A history of chronic failure to thrive, repeated hospitalizations, and exaggerated viral illnesses can also provide clues. At presentation, the history may include an apparent viral infection, respiratory or gastrointestinal complaints. Presenting signs and symptoms

may be attributable to sepsis, respiratory infections, and/or gastrointestinal disorders. Tachypnea, poor feeding, pallor, lethargy, and vomiting may constitute symptoms of heart failure. Chronic symptoms may include a heart murmur, excessive fatigue, and poor weight gain. Either acutely or chronically, primary metabolic cardiomyopathies may mimic ischemic heart disease **(Laschi** *et al.*, **1986).**