Summary

Pediatric metabolic cardiomyopathy is a rare but serious and often life-threatening condition. In children, cardiomyopathy is often a part of multisystem disorder which requires the attention of multiple sub specialists. Observational epidemiology has a notable success in identifying many modifiable exposures increasing or decreasing the diseases risk.

This retrospective descriptive study was carried out in the Pediatric cardiomyopathy clinic, Abo-Resh Hospital, Cairo University, to review the files of patients diagnosed with metabolic cardiomyopathy during the, in the period from January 2003 to November 2010.

The aim of this study was to describe the most common demographic features as regard (age, sex…) of children diagnosed with metabolic cardiomyopathy in the clinic during that period to perform metabolic screening for the selected patients in a trial to identify reversible causes of metabolic causes of cardiomyopathy .

The cases diagnosed with metabolic cardiomyopathy during that period were 94 cases. The files of all of these patients were reviewed for the following data: file number, age, sex, presenting symptoms, clinical manifestations, consanguinity, other sibling affected, previous viral infection, investigation done and treatment.

The 94 patient files included 39 cases with DCM, 48 cases with HCM, 4 cases with RCM of total cases followed up at the clinic.

The age of the patients ranged from one month to 13 years with a mean age at presentation of 3.1 years. The most common age group affected by this disorder were from 1 to 5 years old (35.4%), (32.9%) of our patients were younger than 6 months, (9.5%) were seven to twelve months, and (14.8%) were six to 14 years old, with a male to female ratio of 1.6: 1.

The most common clinical presentation was cardiac symptoms (77.6%) and seizures (12.5%).

In our study consanguinity was positive in (26.6%) of cases of metabolic cardiomyopathy. A history of similar condition was positive in 8 cases (8.5%).

In our overviewed study, among patients diagnosed as metabolic cardiomyopathy, 13.8% (13/94) had lysosomal storage disorders (MPS), 13.8% (13/94) had a disorder of fatty-acid beta-oxidation (L-carnitine deficiency), 3.2% (3/94) had glycogen storage diseases (GSD) and 2.2% (2/94) had mitochondrial disorder.

Sixty-three patients of them were having a history strongly suggestive for metabolic cardiomyopathy (positive consanguinity, extra cardiac manifestations as seizures, mental retardation, psychomotor regression, hepatomegaly and retinitis pigmentosa) and were classified in our study as query metabolic.

As regard results of laboratory investigations done as screening of metabolic cardiomyopathy, serum L-carnitine was done for 38 patients of 94 patients who were diagnosed as metabolic cardiomyopathy thirteen patients had L-carnitine

deficiency representing (13.8%) with missing 56 cases not reported in the files.

The medications received were mainly for treatment of heart failure as (57.2%) of patients were on Diuretics, (36.4%) were on Digoxin, (55.2%) were on ACE inhibitors and (18%) of patients were on L-carnitine therapy.

Inborn errors of metabolism (IEM) account for only 5% of all pediatric cardiomyopathy and 15% of those with known causes, but they are of particular interest to clinicians because many have disease-specific treatment. Hence, it is important that clinicians should be familiar with the evaluation, diagnostic approach, and management of those often lethal pediatric cases.