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Original Research Article

Childhood cardiomyopathies: a study in tertiary care hospital of Kashmir

Abid Ahmad Shairgojri[1]*, Khalid Mohiuddin[2], Amber Bashir[1], Muzafar Jan[1]

[1]Department of Pediatrics, [2]Department of Cardiology, Government Medical College, Srinagar, Jammu and Kashmir, India

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***Correspondence:**
Dr. Abid Ahmad Shairgojri,
E-mail: abidshaira88@gmail.com

ABSTRACT

Background: Cardiomyopathy constitutes a group of diseases that directly affect the structural or functional ability of myocardium. They are the most common form of heart diseases that are inherited in children and responsible for sudden deaths in healthy young adults. The Aim of our study was to provide a detailed description of clinical profile, epidemiology and etiology of cardiomyopathies in children.
Methods: This was a hospital based prospective observational study, conducted over a period of two and half years from September 2017 to March 2020 in the Post Graduate Department of Pediatrics Government Medical College Srinagar. Study group included all patients aged between 1 month and 18 years diagnosed with cardiomyopathy. They were subjected to a detailed clinical history and physical examination. All the patients underwent echocardiography.
Results: During the study period 37 patients were diagnosed with different types of cardiomyopathies. Dilated cardiomyopathy was commonest seen in 19 (51.35%), followed by hypertrophic cardiomyopathy 13 (35%) cases. Fast breathing was most common presenting complaint in dilated cardiomyopathy while most of cases with hypertrophic cardiomyopathy were asymptomatic. There was one case of distinct form of cardiomyopathy isolated left ventricular hypoplasia. Males were more involved 21 (56.76%) cases. Majority of patients 18 (48.64%) were less than one year at time of diagnosis. Parental consanguity was seen in 6 (16.22%) cases. Underlying cause was identified in 10 (27.02%) cases with myocarditis being the commonest cause.
Conclusions: Pediatric cardiomyopathy represents a considerable percentage of children with cardiac disorders. Dilated cardiomyopathy is the most common type usually presented with congestive heart failure, majority of cases of hypertrophic CMP were asymptomatic. Male preponderance was seen in dilated and hypertrophic cardiomyopathy. Parental consanguinity was seen in significant number of patients. Myocarditis and inborn error of metabolism was seen in significant number of patients.

Keywords: Cardiomyopathies, Children, Heart, Myocarditis

INTRODUCTION

Cardiomyopathy (CMP) is defined by the World Health Organization (WHO) as a disease of myocardium associated with cardiac dysfunction.[1] It is a common cause of heart failure in children and the most common indication for heart transplantation in children older than 1 year.[2] In multiple population-based studies, the incidence of primary cardiomyopathies has been estimated to be 1 in 100,000 persons per year in children <20 years of age, ranging between 0.7 in 100,000 (Finnish population) and 1.24 in 100,000 (Australian population).[3-5] Dilated cardiomyopathy has the highest reported incidence, accounting for approximately 50% of all pediatric cardiomyopathies.[4,6,7] Hypertrophic cardiomyopathy accounts for 35-50% of the cases and

restrictive cardiomyopathy for less than 5% of the cases.[3-5] Left ventricular non-compaction cardiomyopathy accounts for about 5% of all cases.[8] Although cardiomyopathies have multifactorial etiology in both children and adults, genetic defects play an important role in infants and children,[9] whereas environmental factors become increasingly important towards adulthood.[9]

The demographics and underlying causes of pediatric cardiomyopathies are not well characterized, particularly in our region. This is the first study on epidemiology of pediatric cardiomyopathy from our region which has high rate of consangious marriages. A better understanding of the epidemiology, etiology and outcome of the disease would facilitate planning and provision of medical services.

The aim of the study was to provide a detailed description of clinical profile, epidemiology and etiology of cardiomyopathies in children presenting in tertiary care hospital.

METHODS

Study design

This study was a hospital based prospective observational study, conducted in the Department of Pediatrics Government medical college Srinagar (GMC). The study was conducted over a period of two and half years from September 2017 to March 2020. Cardiomyopathies were categorized according to current WHO cardiomyopathy classification.[1] Classification system based on AHA was later incorporated.[10]

Inclusion criteria

Any child older than one month and ≤18 years of age presenting with clinical signs and symptoms suggestive of underlying cardiomyopathy and those which were incidentally documented on echocardiography performed for other reasons.

Exclusion criteria

Neonates, death within 24 hours of hospital admission and patients with any hemodynamically significant congenital heart defect or valvular defects.

All patients were examined by a single pediatric cardiologist, and the final diagnosis was made either by echocardiography or cardiac MRI. The echocardiographic criteria for various cardiomypathies was used as per scientific statement given by American Heart Association.

Data included complete medical history of the patients with special attention to cardiac symptoms particularly those which are in favor for diagnosis of CMP, such as symptoms of heart failure (dyspnea, palpitation, exercise intolerance, interrupted feeding, failure to thrive and recurrent chest infections) and syncope. All the patients were subjected to detailed physical examination, blood chemical analysis and hematologic measurements, electrocardiography, chest radiography, echocardiography and cardiac MRI wherever needed. Some patients on clinical suspicion were subjected to other specific investigations like troponin T, serum calcium levels, creatinine phosphokinase levels, thyroid function test, serum lactate and ammonia levels, metabolic screening, enzyme assay levels. Skeletal muscle biopsy was carried out only in selected patients.

Statistical analysis

Both clinical, laboratory and radiological data were analyzed in a spreadsheet of Microsoft Excel. Continuous variables were expressed as mean, whereas categorical variables were summarized as frequencies and percentages.

RESULTS

During the 2.5 year study period, a total of 37 new cases of primary cardiomyopathies were identified. 21 (56.76%) patients were males. Majority of patients were less than 1 year at time of presentation 18 (48.64%).

Table 1: Baseline characteristics of patients enrolled for study.

	Dilated CMP n = 19 (51.35%)		Hypertrophic CMP n =13 (35.13%)		Restrictive CMP n = 2 (5.4%)		Other n = 3 (8.1%)		Total n= 37
	M; n=10 (52.65%)	F; n=9 (47.6%)	M; n=8 (61.5%)	F; n=5 (38.4%)	M; n=0	F; n=2 (100%)	M; n=3 (100%)	F; n=0	
Age at presentation									
<1 year	5	4	4	4	-	-	1	-	18 (48.64%)
1-6 year	3	2	2	1	-	1	2	-	11 (29.72%)
6-12 year	1	2	1	-	-	1	-	-	05 (13.51%)
12-18 year	1	1	1	-	-	-	-	-	03 (8.10%)
Consangunity									
Consangious	2	1	1	-	-	1	1	-	06 (16.22%)
Non consangious	8	8	7	5	-	1	2	-	31 (83.78%)

Consanguinity between parents of studied children was present in 6 (16.22%) patients. Dilated cardiomyopathy was seen in 19 (51.35%) of cases, hypertrophic cardiomyopathy in 13 (35.13%) cases and restrictive cardiomyopathy in 2 (5.4%) of cases. Remaining 3 cases included one case each of non-compacted cardiomyopathy, tachycardia induced cardiomyopathy and one case of distinct form of cardiomyopathy called isolated left ventricular hypoplasia. Table 1 shows the baseline characteristics of the 37 patients who were enrolled in our study.

Table 2: Clinical symptoms in various cardiomyopathies.

	Dilated CMP n=19	Hypertrophic CMP n=13	Restrictive CMP n=2	Others n=3
Fast breathing	13 (68.42%)	03 (23.08%)	1 (50%)	2 (66.67%)
Cough	04 (21.05%)	01 (7.69%)	1 (50%)	1 (33.3%)
Feeding difficulty	03 (15.79%)	01 (7.69%)	-	1 (33.3%)
Fatigue	03 (15.79%)	-	1 (50%)	-
Sweating while feeding/crying	02 (10.53%)	03 (23.08%)	-	-
Swelling of feet	02 (10.53%)	-	2 (100%)	-
Chest pain	02 (10.53%)	-	-	-
Palpitations	-	01 (7.6%)	-	1 (33.3%)
Poor weight gain	01 (5.26%)	01 (7.69%)	-	1 (33.3%)
Fever	01 (5.26%)	01 (7.69%)	-	-
Abdominal distension	-	-	2 (100%)	-
Asymptomatic	-	05 (38.46%)	-	-

Table 3: Clinical signs in various cardiomyopathies.

	Dilated CMP n=19	Hypertrophic CMP n=13	Restrictive CMP n=2	Others n=3
Tachycardia	16 (84.21%)	5 (38.46%)	2 (100%)	2 (66.67%)
Hepatomegaly	15 (78.9%)	5 (38.46%)	1 (50%)	2 (66.67%)
Pedal edema	02 (10.52%)	-	2 (100%)	-
Increased JVP	02 (10.53%)	-	1 (50%)	1 (33.3%)
Murmur	01 (5.26%)	9 (69.23%)	-	2 (66.67%)
Developmental delay with hypotonia	01 (5.26%)	3 (23.08%)	-	-

Table 3: Etiology of CMP.

	Dilated CMP n=19	Hypertrophic CMP n=13	Restrictive CMP n=2	Others n=3
Inborn error of metabolism				
Disorder of glycogen metabolism	-	2	-	-
Carnitine deficiency	1	-	-	-
Neuromuscular disorder				
Duchenne Becker dystrophy	1	-	-	-
Myocarditis				
probable (cardiac troponin T positive)	3	-	-	-
Malformation syndrome				
Noonas syndrome	-	1	-	-
Infiltrative disorder				
Hypereosinophilic syndrome	-	-	1	-
Tachycardia induced cardiomyopathy	-	-	-	1

Fast breathing was the most common presenting complaint in dilated cardiomyopathy seen in 13 (68.42%) cases. Tachycardia and hepatomegaly were the most common clinical signs in dilated cardiomyopathy seen in 16 (84.21%) and 15 (78.2%) patients respectively. Children with hypertrophic cardiomyopathy were mostly asymptomatic 5 (38.46%) cases and were incidentally diagnosed on cardiac auscultation. 3 (23.08%) patients

with hypertrophic cardiomyopathy had developmental delay with hypotonia. Both patients with restrictive cardiomyopathy had swelling of feet and abdominal distension on presentation. Table 2 and 3 depict the cardinal clinical symptoms and signs in various cardiomyopathies.

We have detected the etiology of CMP in 10 (27.02%) patients only, while in remaining 72.98% the cause couldn't be established and were labeled as idiopathic. Suspected viral myocarditis was the most common identified etiological agent responsible for three cases with dilated cardiomyopathy, while one case each was secondary to neuromuscular dystrophy and carnitine deficiency. Two patients with hypertrophic cardiomyopathy had deficiency of acid alfaglucosidase and one child had Noonan's syndrome. One child with restrictive cardiomyopathy had hypereosinophilic syndrome. Incessant focal atrial tachycardia was present in child diagnosed with tachycardia induced cardiomyopathy (Table 4). One infant with abnormally thin interventricular septum underwent cardiac MRI and was later found to have a rare form of cardiomyopathy viz. isolated left ventricular hypoplasia.

DISCUSSION

Contrary to the adult population, there is a huge knowledge gap regarding the epidemiological characteristics of cardiomyopathies in the pediatric population. To our knowledge, few if any, studies have been conducted to assess the epidemiological characteristics of cardiomyopathies in our region. This is to mention that our region has high percentage of second and third degree consangious marriages which have bearing on genetic disorders. Among 34328 children that attended our institution during the study period, we identified 37 new cases of primary cardiomyopathy. Even though the design of our study did not allow us to calculate the incidence or prevalence of these cardiac conditions, our findings clearly surpassed the expected number of cases. In multiple population-based studies, the incidence of primary cardiomyopathies was estimated to be 1 in 100,000 persons in children <20 years of age, which demonstrates a higher-than-expected number of cases in our institution.[3-5] The main reason for this could be the fact that our institution represents one of the main pediatric referral center. The male preponderance and characteristics of cardiomyopathies in our population are in line with global trends and reported studies, with dilated cardiomyopathy being the most common type seen in more than half of the cases, followed by hypertrophic cardiomyopathy.[3,4,10]

In our study, majority of children (48.54%) presented were less than 1 year of age. In dilated cardiomyopathy 47.37% were younger than one year while in hypertrophic cardiomyopathy 61.54% children presented in infancy. Similar results with majority presenting in infancy has been shown in several epidemiological studies.[3-5]

Parental consanguity was present in 16.21% cases of cardiomyopathy. It was 15.79% in dilated cardiomyopathy whereas it was 7.69% in hypertrophic cardiomyopathy. Our values are more than study by Nugent et al, were rate of consanguity was 6.70%.[4] This higher value can be due to high rate of consangious marriage in our region.

The patients with dilated cardiomyopathy presented with constellation of symptoms, however the predominant presenting symptom was fast breathing seen in 13 (68.42%) patients. The symptoms had gradually developed over weeks or even months. The most common preceding symptoms in infants consisted of feeding difficulties, poor weight gain, and sweating while feeding, whereas older children and adolescents had typically suffered from a gradual loss of weight and chest pain. Most patients of dilated cardiomyopathy had clinical signs of congestive heart failure at presentation tachycardia and hepatomegaly present in 16 (84.21%) and 15 (78.95%) respectively. In contrast to patients with dilated cardiomyopathy, most patients with hypertrophic cardiomyopathy were asymptomatic and were incidentally diagnosed on auscultation of cardiac murmur at presentation seen in (69.3%) patients. Atypical features of developmental delay with hypotonia was present in 3 (23.08%) patients. In large epidemiological study conducted in Finland significant number of children with hypertrophic cardiomyopathy were asymptomatic or had atypical features.[5] This highlights high index of suspicion among pediatricians treating children who present with non-specific symptoms. Two patients with restrictive cardiomyopathy presented with swelling in feet and abdominal distension. In the miscellaneous group the infant with non compacted left ventricle had fast breathing as presenting feature. An infant who was diagnosed to have isolated left ventricle hypoplasia had poor weight gain and one with tachycardiomyopathy had complained of episodic palpitation**.**

We could find the etiology in 10 (27.02%) patients only. In a study by Lipshultz et al 68% of dilated and hypertrophic cardiomyopathy were idiopathic.[3] Serum troponin T was carried out in all 19 cases of dilated cardiomyopathy, and it was elevated in 3 patients who were diagnosed as having probable myocarditis. It is advisable that levels of troponin T be measured in the serum of all patients with dilated cardiomyopathy, as this may provide important clues to its etiology.[11] Cardiac troponin T was elevated in 4.2% cases in study by Bakeet et al.[12] Inborn error of metabolism was found in 3 patients, severe levocarnitine deficiency in one patient with dilated cardiomyopathy and acid alpha glucosidase deficiency in two patients with hypertrophic cardiomyopathy. A significant proportion of cases 11.25% was caused by inherited metabolic diseases in a study by Miranda et al.[13] These data reinforce the

importance of considering inherited metabolic diseases in the differential diagnosis of children presenting with cardiomyopathy, as it is crucial to define prognosis. The ideal time to diagnose inherited metabolic diseases is the neonatal period, before clinical manifestations using newborn screening followed by confirmatory studies. Cardiac MRI was done in 2 patients who had uncertainty of diagnosis on the basis of echocardiographic findings. One such patient was 2 year old child in which echocardiography revealed trabeculated left ventricle and septal bowing towards the right ventricle. Cardiac MRI revealed features suggestive of under reported form of cardiomyopathy called as isolated left ventricular hypoplasia. This form of cardiomyopathy was first reported by Fernandez et al in a case series.[14,15] In another patient with features of restrictive cardiomyopathy cardiac MRI revealed hyperintensity of endomyocardium with patchy enhancement. Complete blood analysis showed peripheral eosinophilia and was labelled as hypereosinophilic syndrome, which is characterized by an initial acute inflammatory phase with pancarditis then a thrombotic phase and finally a fibrotic stage with irreversible endocardial fibrosis.[16]

Limitations were time period for this study was less and cardiac MRI was not done in all patients for correlation.

CONCLUSION

Despite limitations, this study is the first one which describes characterization of pediatric cardiomyopathy in Kashmir valley. We found that pediatric cardiomyopathy represents a considerable percentage of children with cardiac disorders. Dilated cardiomyopathy is the most common type usually presented with congestive heart failure, majority of cases of hypertrophic CMP were asymptomatic. Male preponderance was seen in dilated and hypertrophic cardiomyopathy. Parental consanguinity was seen in significant number of patients. Myocarditis and inborn error of metabolism was seen in significant number of patients. We found some rare form of cardiomyopathy. Our study is limited by design, due to its observational nature and due to the fact that we only considered data from our institution. A comprehensive analysis of data obtained from different observational studies can expand our understanding about the cardiomyopathy in the pediatric population.

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