

Evaluation of Cardiomyopathies and Echocardiographic Study of Different Cardiomyopathies in Children

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ABSTRACT

BACKGROUND

An understanding of cardiomyopathy (CMP) is very important, as it is a common cause of heart failure in children and the most common indication for heart transplantation in children older than 1 year. We wanted to evaluate the cardiomyopathies in children clinically and conduct an echocardiographic study of different types of cardiomyopathies.

METHODS

This is a hospital based descriptive study conducted in Katuri Medical College and Hospital, Guntur, among 32 children with cardiomyopathy who attended the departments of paediatrics and cardiology over a period of 2 years.

RESULTS

Dilated cardiomyopathy, hypertrophic cardiomyopathy and restrictive cardiomyopathy were the commonest cardiomyopathies seen in children. Dilated cardiomyopathy was the commonest myopathy observed in this series followed by HOCM and RCM. The incidence of cardiomyopathy was more in male children. The sex ratio is 1.28:1 (M:F). More number of cases were observed in children of <5 years than in children of >5 years with age ratio of 4.3:1. Majority of the patients were having the ejection fraction less than 40% particularly with dilated cardiomyopathy. Majority of the patients were having cardiomegaly on x-ray chest. ECG showed tachycardia, low voltage complexes, ST segment / T wave changes and arrhythmias.

CONCLUSIONS

Cardiomyopathies are heterogenous and rare diseases but not uncommon in children. They require complete evaluation to know the exact cause, pathophysiology, clinical pattern and natural history to prevent and treat appropriately because heart transplantation is the only treatment that can be given after exhausting all other treatment options.

KEYWORDS

Pediatric Cardiomyopathy, Echo, Various Types

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BACKGROUND

An understanding of cardiomyopathy (CMP) is very important, as it is a common cause of heart failure in children and the most common indication for heart transplantation in children older than 1 year.¹ Cardiomyopathy is a heterogeneous disease caused by a functional abnormality of the cardiac muscle. The World Health Organization (WHO) defines CMP as "a disease of the myocardium associated with cardiac dysfunction" and classifies it as dilated, hypertrophic, restrictive, arrhythmogenic right ventricular, or unclassified.² Secondary cardiomyopathy is caused by extrinsic factors, including infection, ischemia, hypertension, and metabolic disorders.³ Primary cardiomyopathy is diagnosed when the extrinsic factors of secondary cardiomyopathy are absent. Primary cardiomyopathy is rare. Dilated cardiomyopathy is the most common form of cardiomyopathy worldwide and has many causes. In 30% to 48% of patients, dilated cardiomyopathy is genetically inherited.⁴ Moreover, inflammatory disorders such as myocarditis, or toxic agents such as medications and alcohol can result in Dilated cardiomyopathy. Of all the Dilated cardiomyopathy cases, 20% to 48% have a family history of the disease.⁵ The aetiology of hypertrophic cardiomyopathy in the paediatric population is heterogeneous, including Inborn metabolism errors, neuromuscular disorders, and malformation syndromes. However, most cases of apparently idiopathic hypertrophic cardiomyopathy (HCM) in childhood are caused by mutations of cardiac sarcomere protein genes. In the paediatric population, risk stratification is necessary because most cases of HCM have a family history of the disease. Evaluation of first-degree relatives and any at-risk family members should be a routine component of clinical management.⁶ Restrictive cardiomyopathy (RCM) is a rare form of heart muscle disease that is characterized by restrictive filling of the ventricles. It appears to affect girls somewhat more often than boys. There is a family history of cardiomyopathy in approximately 30% of cases.⁷ In most cases the cause of the disease is unknown (idiopathic), although a genetic cause is suspected in most cases.

There are other forms of cardiomyopathy which comprise only a very small percentage of the total (~2–3%) number of cardiomyopathies in children. These cardiomyopathies may have overlapping features with any of the previous types described and include Arrhythmogenic Right Ventricular Dysplasia (ARVD).

We wanted to evaluate the cardiomyopathies in children clinically and conduct an echocardiographic study of different types of cardiomyopathies.

METHODS

This is a hospital based descriptive study conducted in Katuri Medical College and Hospital, Guntur, among 32 children diagnosed with cardiomyopathy based on the clinical, x-ray,

electrocardiographic, echocardiographic evaluation who attended the departments of paediatrics and cardiology over a period of 2 years. Benefits to the study participants was early diagnosis of cardiomyopathy, which led to early intervention which in turn decreased the morbidity and increases the lifespan.

Inclusion Criteria

1. Neonates with cardiomyopathy both inborn and referred.
2. Children presenting clinically with signs and symptoms Suggestive of cardiomyopathy.
3. Babies with genetic syndromes having cardiomyopathy.

RESULTS

From the above analysis of 32 cases of cardiomyopathies, it was observed that DCM, HOCM, RCM and ARVC comprising of 26, 4, 2, 0 respectively. DCM was the commonest cardiomyopathy seen in the present series comprising of 81.25%. The Other cardiomyopathies like HOCM and RCM were recorded 12.5% and 6.25% respectively. No case of ARVC was recorded in this series. So, DCM was the commonest among the myopathies, seen in four-fifths of cases of cardiomyopathies.

Gender wise evaluation of cardiomyopathies, it was observed that the incidence in male and female children comprising of 18 (56.25%) and 14 (43.25%) cases respectively. From the above analysis, that male preponderance was seen in this series. The Sex ratio from male: female is 1.28:1 From the above analysis of gender wise distribution of DCM that male preponderance was seen in 16 cases of DCM as against 10 cases in females comprising of 61.5% and 38.4% respectively. From the above analysis of gender wise distribution of HOCM it was observed that sex incidence was equal. Since it was observed in a small number of children it is very difficult to estimate the sex incidence. Evaluation of RCM cases, the 2 cases were seen in females only. Since it was observed in a small number of children it is very difficult to estimate the sex incidence.

| Age Group | Frequency | % |
|-------------|-----------|--------|
| 0-2 Y | 16 | 50% |
| >2 Y – 5 Y | 10 | 31.25% |
| >5 Y – 10 Y | 4 | 12.5% |
| >10 Y | 2 | 6.25% |
| Total | 32 | 100% |

Table 1. Age Wise Distribution of Cardiomyopathy

Age wise incidence of cardiomyopathies in children, it was observed that maximum number of cases were seen below 2 years of age comprising of 50% (16/32). Another increase of incidence was recorded in the age group of 2-5 years comprising of the incidence 31.25% (10/32). Small number of cases were recorded between 5-10 years and >10 years comprising of 4 (12.5%) and 2 (6.25%). As seen from the above table 26 cases were seen below 5 years and 6 cases after 5 years. So, the incidence of cardiomyopathies was more in children less than 5 years.

| Symptom | Present | Absent | Total | % |
|---------------------------------|---------|--------|-------|--------|
| Dyspnoea / Cough | 30 | 2 | 32 | 93.75% |
| Syncope | 11 | 21 | 32 | 34.3% |
| Oedema / Hepatomegaly / Ascites | 2 | 30 | 32 | 6.25% |

Table 2. Analysis of Symptomatology in Cardiomyopathy

Analysis of symptoms of cardiomyopathies, it was observed that dyspnea/cough, syncope and edema/hepatomegaly/ ascites were seen in 30, 11 and 2 cases respectively with the percentage of 93.75%, 34.3% and 6.25% respectively. Dyspnea / cough was seen in almost all cases except in 2. Syncope was the next common symptom of the cardiomyopathy. Even though 32 cases of cardiomyopathies were seen only 2 cases were having pedal edema/hepatomegaly/ascites. Evaluation of the above data it was observed that only 7 cases (21.8%) were having pulmonary edema and the rest of 25 cases (78.1%) were not having pulmonary edema. So, not all cases of cardiomyopathies are associated with pulmonary edema. From the above data on incidence of pulmonary edema in DCM among the 26 cases, it was observed that 15.38% (4) of cases developed pulmonary edema at presentation and 84.62% (22) of cases showed no signs of pulmonary edema. From the above data on incidence of pulmonary edema in HOCM among the 4 cases, it was observed that 25% (1) of cases developed pulmonary edema at presentation and 75% (3) of cases showed no signs of pulmonary edema. From the above data on incidence of pulmonary edema in RCM among the 2 cases, it was observed that 100% (2) of cases developed pulmonary edema at presentation.

| Grade | Frequency | % |
|----------|-----------|--------|
| Absent | 11 | 34.3% |
| Mild | 13 | 40.62% |
| Moderate | 7 | 21.8% |
| Severe | 1 | 3.12% |
| Total | 32 | 100% |

Table 3. Analysis of Pulmonary Arterial Hypertension in Cardiomyopathy

| Cardiomegaly | Frequency | % |
|--------------|-----------|--------|
| Present | 27 | 83.33% |
| Absent | 5 | 16.67% |
| Total | 32 | 100% |

Table 4. Analysis of Chest X-Ray in Cardiomyopathy

| Finding | Present | Absent | Total | % |
|-----------------------------|---------|--------|-------|--------|
| Tachycardia | 32 | 0 | 32 | 100% |
| ST segment / T wave changes | 21 | 11 | 32 | 65.6% |
| Low voltage complexes | 7 | 25 | 32 | 21.8% |
| Arrhythmias | 6 | 26 | 32 | 18.75% |

Table 5. Analysis of ECG in Cardiomyopathy

| LVEF | 0-20% | 21-40% | >40% |
|-------|-------|--------|-------|
| DCM | 2 | 22 | 2 |
| HOCM | 0 | 0 | 4 |
| RCM | 0 | 0 | 2 |
| Total | 2 | 22 | 8 |
| % | 6.25% | 68.75% | 25.0% |

Table 6. Analysis of LVEF (%) in Cardiomyopathy

Evaluation of PAH in cardiomyopathies 21 patients were found to have PAH ranging from mild to severe comprising of 65.7% and absent in 34.3% patients. X-ray evaluation of the patients of cardiomyopathies 83.33% patients were found to have cardiomegaly as against 16.67% of patients without cardiomegaly. Evaluation of the patients of

cardiomyopathies revealed that all the patients were found to have tachycardia comprising of 100%. ST segment / T wave changes, low voltage complexes and arrhythmias were seen in 65.6%, 21.8% and 18.75% respectively.

Evaluation of the patients of various cardiomyopathies by Doppler echo regarding the functional capacity of the heart in the form of ejection fraction, it was revealed that 6.25%, 68.75% and 25% of the patients with ejection fraction of <20%, 21-40% and >40% were found in patients of DCM, HOCM and RCM respectively. Majority of patients of DCM were having low ejection fraction of <40% comprising of 75%. So small number of patients of HOCM and RCM were having ejection fraction >40%. It revealed that majority of the patients of DCM were associated with low ejection fraction because of global hypokinesia.

DISCUSSION

Cardiomyopathies are the heterogenous group of diseases without the history of rheumatic fever, hypertension and congenital heart diseases. Cardiomyopathies are mainly diagnosed on exclusion basis after considering all the possibilities of cardiac involvement. Even though these disorders are rare in children, but cases are reported in children of less than 15 years of age. All the cases that were attended the pediatric OP, Cardiology OP and also admitted in these 2 wards were evaluated thoroughly by taking detailed history, thorough physical examination and relevant investigations like chest x-ray, ECG and 2D Echocardiography were done. A total number of 32 cases were studied in the Department of Pediatrics, over a period of 2 years. All the cases were evaluated thoroughly by taking proper history, detailed physical examination and all the cases were subjected for routine investigations like chest x-ray and ECG. All the cases were evaluated by 2D Echocardiography to detect the anatomical, functional capacity in the form of ejection fraction and also to identify complications like thrombosis, arrhythmias etc. The results were analysed to know the magnitude of these diseases in children.

Prevalence of Cardiomyopathy

Evaluation of 32 cases of cardiomyopathies such as dilated cardiomyopathy, hypertrophic cardiomyopathy and restrictive cardiomyopathy it was found that the incidence was 81.25%, 12.5% and 6.25% respectively. Dilated cardiomyopathy was the commonest cardiomyopathies found in the present study. HOCM and RCM were found in 19% of the patients.

Gender Wise Distribution

Gender wise evaluation of cardiomyopathies, it was observed that the incidence of cardiomyopathies was more in male children comprising of 56.25% (18/32) whereas 14 cases were seen in female children comprising of 43.75% (14/32). It showed that the incidence was more in male children with sex ratio from male to female was 1.28:1.

Gender Wise Distribution in DCM

Among the sex wise incidence of Dilated Cardiomyopathy 16 cases were seen in male children comprising of 61.5% whereas 10 cases were seen in females comprising of 38.4%. so, the incidence of dilated cardiomyopathy was also more in male

Gender Wise Distribution in HOCM

Sex wise evaluation of hypertrophic cardiomyopathy showed that the incidence was equal in both sexes comprising of 2 each (50%). Since it is a small study sex incidence cannot be determined.

Gender Wise Distribution in RCM

Evaluation of sex incidence of RCM showed that both the cases were seen only in females. Since it is a small study sex incidence cannot be determined.

Age Wise Distribution of Cardiomyopathy

Age wise distribution of cardiomyopathies it showed maximum number of cases were observed below 5 years of age comprising of 26 cases (81.25). 6 cases were observed after 5 years of age in children. Maximum number of cases are seen below 2 years of age comprising 50%. From the above analysis it showed that the incidence of cardiomyopathies was decreasing with increase of age. The incidence was inversely proportional to the age of the child. The age incidence ratio before 5 and after 5 years was 4.3:1.

Symptomatology in Cardiomyopathy

The common presenting symptoms in the present series of cardiomyopathies were dyspnea/cough, syncope and edema/hepatomegaly/ascites were seen in 30, 11 and 2 cases respectively with the percentage of 93.75%, 34.3% and 6.25% respectively. Dyspnea / cough was seen in almost all cases except in 2. Syncope was the next common symptom. Even though 32 cases of cardiomyopathies were studied, pedal edema /hepatomegaly /ascites were seen only in 2 cases. Even though pulmonary edema and pulmonary artery hypertension were seen in 7 and 21 cases, but pedal edema /hepatomegaly/ ascites were seen only in 3 cases because of the compensatory mechanisms of the right side of the heart.

Incidence of Pulmonary Oedema in Cardiomyopathy

Evaluation of the present series regarding the congestion of the lungs in the form of pulmonary edema it was seen only in 7 cases comprising of (21.8%). from the above analysis, it revealed pulmonary congestion was less in cardiomyopathies but most of the times heart failure will be seen in dilated cardiomyopathy. In the present series 2 out of 2 of RCM and 1 out of 4 HOCM cases were having congestion of lungs.

Incidence of Pulmonary Arterial Hypertension in Cardiomyopathy

Of the 32 cases of cardiomyopathies pulmonary artery hypertension was found in 21 cases ranging from mild to

severe comprising of 65.7%. Rest of the 11 cases comprising of 34.3% were normal. In any heart disease, examination of the pulmonary area for pulmonary artery hypertension is very important because majority of patients of heart diseases are associated with pulmonary artery hypertension particularly with acyanotic heart diseases and acquired heart diseases as a measure of severity.

Evaluation by X-Ray Chest in Cardiomyopathy

Evaluation of the patients by X - ray chest 27 cases out of 32 were associated with cardiomegaly comprising of 83.33%. Rest of the 5 patients were having normal sized heart.

Evaluation by ECG in Cardiomyopathy

Evaluation of the patients of cardiomyopathies revealed that all the patients were found to have tachycardia comprising of 100%. ST segment / T wave changes, low voltage complexes and arrhythmias were seen in 65.6%, 21.8% and 18.75% respectively.

LVEF (%) in Cardiomyopathy

Evaluation of the patients of various cardiomyopathies by doppler echo regarding the functional capacity of the heart in the form of ejection fraction, it was revealed that 6.25%, 68.75% and 25% of the patients with ejection fraction of <20%, 21-40% and >40% were found in patients of DCM, HOCM and RCM respectively. Majority of patients of DCM were having low ejection fraction of <40% comprising of 75%. Small number of patients of HOCM and RCM were having ejection fraction >40%. It revealed that majority of the patients of DCM were associated with low ejection fraction because of global hypokinesia. Normal ejection fraction ranges from 55-65%. If the ejection fraction is <40%, it can be considered as low ejection fraction.

Similar Studies

Shyam S Kothari, et al.⁸ from All India Institute of Medical Sciences studied the clinical course and prognosis of dilated cardiomyopathy in Indian children. They retrospectively reviewed the records of 82 children with DCM from 1992 to 2001. Of the 82 children 50 were males. Family history of DCM was present in 3.5% of cases. Cardiomegaly was present in all cases with mean CTR 66%. They found that mortality was high in infants diagnosed with DCM.

Piers E. F. Daubeney, et al⁹ conducted population-based study on clinical features and outcomes of childhood dilated cardiomyopathy in Australia from 1987 to 1996. There were 184 subjects with DCM. Childhood dilated cardiomyopathy is most common during the first year of life and is associated with significant morbidity and mortality. At presentation, 90% of cases had signs and symptoms of congestive heart failure, and sudden death was the presenting symptom in 4%. The median age of the 9 cases whose initial symptom was sudden death was 2 months (range 8 days to 11 months). Familial cardiomyopathy was identified in 14.7% of subjects, a metabolic or mitochondrial disease in 8.9%. A potential viral contribution (lymphocytic myocarditis or

positive viral identification) was identified in 68.2% of case subjects (most commonly coxsackievirus or adenovirus). Early mortality is high in childhood dilated cardiomyopathy, but the clinical status of long-term survivors is good was the conclusion given.

Anita Khalil, et al¹⁰ from Maulana Azad Medical College studied the clinical profile, treatment and outcome of DCM in children. 25 children were evaluated for their clinical profile and comparative efficacy of beta blockers and ACE-inhibitors in children with dilated cardiomyopathy. All of them presented with congestive failure. Electrocardiograms revealed tachycardia in all. There was increase in LVDD and LVDs (left ventricular diameters in diastole and systole) and depression in fractional shortening (FS) in all the patients.

Inas Abdull Sattarsaad¹¹ from Cairo University, in his study on: natural history and predictors of prognosis idiopathic dilated cardiomyopathy in children has reported a gender distribution of 65.5% female and 35.5% male among 55 children with DCM. Cardiomegaly 3 was noted on chest radiography in 90% of our patients, but only 56.3% had increased lung vascularity. ST segment and T wave changes were seen on electrocardiogram in 69% of cases, 13% had low voltage, and 14.5% had arrhythmias. Younger age of presentation, higher z-score of inter-ventricular septum and left ventricle posterior wall dimension in diastole are predictors for favorable outcomes, and left ventricular end diastolic dimension of high z-score is related to unfavorable outcomes. Miranda JO et al¹² in their study conducted from 2005 to 2012 on: "Paediatric dilated cardiomyopathy: clinical profile and outcome. The experience of a tertiary centre for paediatric cardiology." A retrospective review of 61 patients (37 female; 24 male) diagnosed with dilated cardiomyopathy from January, 2005 to June, 2012 at a single institution was performed. The median age at diagnosis was 15 months. Heart failure was present in 83.6% of patients and 44.3% required intensive care.

Jaiyesimi O¹³ in his study in 2007 on: "Clinical profiles and outcomes for Omani children with dilated cardiomyopathy seen in a regional referral hospital" analysing the data from 32 DCM children who received care in their unit between January, 1999, and August, 2007. His study consists of 17 male children in the age group of 5 weeks to 8 years at presentation, with a median age of 7 months with cardiac failure in almost four-fifths of cases which was the most frequent presenting feature. Correspondingly, increased cardiothoracic ratio was seen in 68% in 20 infants and 65% in older children of 8 years, and the depressed left ventricular ejection fraction in 41% of cases.

| Sl. No. | Gender | Shyam S Kothari, et al ⁸ | Jaiyesimi O ¹³ | Present Study |
|---------|--------|-------------------------------------|---------------------------|---------------|
| 1. | Male | 60.97% | 53.12% | 61.5% |
| 2. | Female | 39.02% | 46.88% | 38.4% |

Table 7. Comparison of Gender Wise Distribution of Asthma among Different Studies

From the above analysis, the incidence of cardiomyopathy is more in male children when compared to female children. The present study was in correlation with the studies of other authors.

| Parameter | Inas Abdull Sattar Saad ¹¹ | Present Study |
|---------------------|---------------------------------------|---------------|
| Cardiomegaly | 90% | 83.33 |
| ST – T Wave Changes | 69% | 65.6 |
| Low Voltage | 13% | 21.8% |
| Complexes | | |
| Arrhythmias | 14.5% | 18.75% |

Table 8. Comparison of X-Ray Chest and ECG Changes with another Study

From the above comparison it is evident that, cardiomegaly is present in majority of cases and among the ECG changes ST / T wave changes are common followed by low voltage complexes in present study. The present study was in correlation with other study.

CONCLUSIONS

Dilated cardiomyopathy, hypertrophic cardiomyopathy and restrictive cardiomyopathy were the commonest cardiomyopathies seen in children. Dilated cardiomyopathy was the commonest myopathy seen in this series followed by HOCM and RCM. The incidence of cardiomyopathy was more in Male children. the sex ratio from male: female is 1.28:1. More number of cases were observed in children of <5 years than in children of >5 years with age ratio of 4.3:1. It was also the observation the incidence of cardiomyopathies was decreasing with increasing age. So, more number of cases were also seen below 2 years of age which requires thorough evaluation regarding the vulnerability of children. Majority of the patients were having the ejection fraction less than 40% particularly with dilated cardiomyopathy which comprise the major part of cardiomyopathies. Echo cardiography is the investigation of choice for evaluation and identification of various types of cardiomyopathies. Surprisingly, even though some of the patients have pulmonary edema and pulmonary hypertension, only small number of patients were having pedal edema/ascites/hepatomegaly.

Majority of the patients were having cardiomegaly on X ray chest. ECG showed Tachycardia, low voltage complexes, ST segment / T wave changes and arrhythmias. Even though chest X ray and ECG may not identify the specific type of cardiomyopathies, these are the preliminary investigations of cardiac diseases and complimentary to the echocardiography. Cardiomyopathies are heterogenous and rare diseases but not uncommon in children. They require complete evaluation to know the exact cause, pathophysiology, clinical pattern and natural history to prevent and treat appropriately because heart transplantation is the only treatment that can be given after exhausting all other treatment options.

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