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Research Article

Frequency of Cardiomyopathy in Beta-Thalassemia Major Patients Using Echocardiography

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ABSTRACT
Introduction: Despite the advances in therapeutic management of thalassemia major and the resulting substantial improvement of patients' survival, heart disease still remains the primary cause of mortality and morbidity. Heart disease accounts for over half of the deaths in patients with thalassemia major and may present as heart failure, cardiomyopathy, pulmonary hypertension, arrhythmias, pericarditis and myocarditis. Thalassemia –related cardiomyopathy may manifest as dilated left ventricular, cardiomyopathy or restrictive left ventricular filling.
Objective: To determine the frequency of cardiomyopathy in beta-thalassemia major patients using echocardiography.
Study Design: Descriptive, cross-sectional.
Study duration: 15th February 2026 to 15th May 2026
Settings: Department of Pediatrics, Fauji Foundation Hospital, Rawalpindi.
Materials & Methods: A total of 142 patients with beta-thalassemia major aged 2-12 years, of either gender, were included. Patients with hemoglobinopathies other than beta thalassemia major were excluded. After taking history of the disease and treatment, echocardiography was done in every patient to confirm cardiomyopthy.
Results: Most patients 123 (86.67%) were aged 8-12 years, with a mean age of 9.62 ± 1.62 years. Among the 142 patients, 86 (60.56%), were male and 56 (39.44%) were females. cardiomyopathy was diagnosed in 62 (43.66%) patients. The estimated frequency of cardiomyopathy in beta-thalassemia major patients in this population was at least 36% with p value 0.035.
Conclusion: We concluded that cardiomyopathies is a prevalent complication among β-thalassemia major patients, occurring in 36% of the cases. Early screening and management are recommended to mitigate cardiac complications.

Keywords: Beta-Thalassemia Major (BTM), Cardiomyopathies, Iron overload, Severe Anemia, Transfusion therapy.

INTRODUCTION

Thalassemia is is characterized by the defects in the hemoglobin production chain[1]. Alpha-thalassemia (AT) and beta-thalassemia (BT) are their two recognized types. BT is widely prevalent in Pakistan. It is categorized into three forms: thalassemia major, thalassemia intermedia and thalassemia minor. Patients who suffer from BT major (B-TM) often develop severe anemia, redundant growth, and abnormalities in skeletal development within the first two years of life with severe anemia, poor growth, and skeletal abnormalities[2]. Regular transfusions of blood

throughout the life of the patient are required. Beta-thalassemia intermedia (B-TI) is less severe and may only require episodic blood transfusions[3].

Pneumonia, elevated levels of iron, deformation of bones, and cardiovascular disorders are some of the complications faced by a thalassemia patient[4]. Among these, Iron overload cardiomyopathy is one of the most common causes of death in patients with thalassemia. Iron overload occurs due to chronic blood transfusion and results in the deposition of iron in vascular and cardiac smooth muscles, leading to complications[5]. While current treatments such as regular blood transfusions and iron chelation therapy can improve survival rates, iron overload cardiomyopathy is still the most common cause of death[6].

Though much advancement have been introduced in the treatment techniques and management strategies of such patients and many efficient results have been achieved regarding their survival but cardiac disorders are still one of the major causes of mortality and morbidity[7]. Thalassemia cardiomyopathy may manifest as heart failure, pulmonary hypertension, arrhythmias, pericarditis and myocarditis dilated left ventricular cardiomyopathy or restrictive left ventricular filling[7,8]. This study is aimed to determine the frequency of cardiomyopathy in patients with B-TM on echocardiography in our population.

MATERIALS & METHODS

It's a descriptive, cross-sectional study conducted at the department of Pediatrics, Fauji Foundation Hospital, Islamabad, from 15 February 2026 to 15th May 2026. The sample size of 142 patients was calculated using WHO sample size calculator 1.1, with an expected prevalence of cardiomyopathy 10.3% in beta thalassemia major patients in children[6], a 95 % confidence level, and a 5% precision. Inclusion criteria: Patients aged between 2-12 years with beta thalassemia major of both genders, receiving blood transfusions for at least 1 year were included in the study.

Exclusion criteria: Patients with hemoglobinopathies other than beta thalassemia major, with preexisting congenital cardiac diseases, rheumatic fever, or infective endocarditis, were not enrolled in this study.

Ethical approval was obtained from the hospital ethical review committee. Patients fulfilling the inclusion criteria were enrolled with parental/guardian consent. The patients were selected through consecutive non-random sampling technique.

Disease symptoms and treatment history were taken from the patient's file and through parent/guardian interview. Along with clinical examination, Echocardiography (ECHO) was performed on all patients to assess for cardiomyopathy. Cardiomyopathy was decided on the basis of ejection fraction (EF<55%). Dilated cardiomyopathy was considered if the EF≤40% or on the evidence on the increased ventricular dimension, thin ventricular wall, mitral regurgitation or reduced wall motion across all segments. Left ventricular hypotrophy was considered on the basis increase wall thickness >13mm or decrease left ventricular cavity size, impaired diastolic function assessed through doppler image, and on the basis of mass index (calculated to quantify hypertrophy). Ventricular stenosis was declared on the basis of increased doppler velocity, reduced valve area, thickened or calcified tissue, and post-stenotic dilation. Valvular regurgitation was declared on the basis of regurgitant jet using color Doppler image, increased regurgitant volume and fraction, dilated chambers, and on valve abnormalities. Wall motion abnormalities were decided on the basis of visual assessment of each segment of the left ventricular wall.

Data was analyzed through SPSS 25.0. Mean and standard deviation were calculated for quantitative variables such as age, frequency of blood transfusion, duration of transfusion and ejection fraction. Frequency and percentage were calculated for qualitative variables including gender and clinical symptoms. Effect modifiers like gender, bone deformities, enlarged spleen and liver, delayed growth, were controlled through stratification and post-stratification chi square was applied. A p-value of <0.05 was considered as significant.

RESULTS

Out of the 142 patients, 86 (60.56%) were male and 56 (39.44%) were females, with male-to-female ratio of 1.5:1. The patient's ages range from 6-12 years, with a mean age of 9.62 ± 1.62 years. The mean disease duration was 6.10 ± 1.53 years, and mean frequency of blood transfusion was 12.70 ± 3.65. The clinical characteristics found in the patients were mentioned in table I.

Table I: Characteristics of Beta Thalassemia Patients

Characteristics	Categories	Frequency	Percentage
Bone deformities	Yes	65	45.8
	No	77	54.2
Enlarge Spleen and Liver	Yes	73	51.4
	No	69	48.6
Delayed Growth	Yes	106	74.6
	No	36	25.4
Jaundice	Yes	53	37.3
	No	89	62.7
Severe Anemia	Yes	66	46.5
	No	76	53.5
Iron Overload	Yes	59	41.5
	No	83	58.5

Cardiomyopathy was diagnosed in 62 (43.66%) patients. The mean ejection fraction was 54.44 ± 11.23%. The result showed that the estimated proportion of cardiomyopathy in children affected from beta thalassemia major, in our population, is at least 0.36 with p value 0.035. The results of echocardiography is mentioned in figure 1.

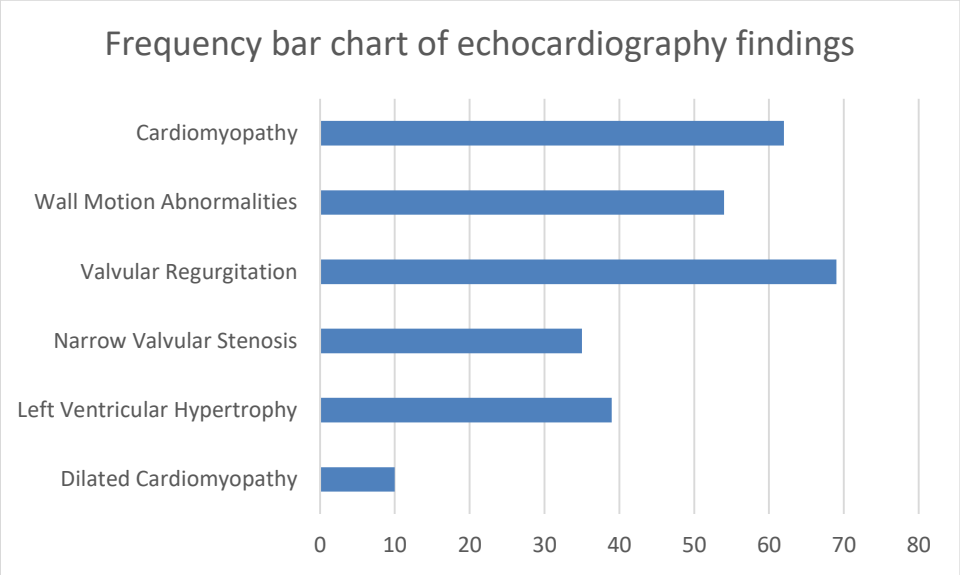

Figure 1: Echocardiography findings of the children affected from Beta thalassemia major

Stratification of the result was done through clinical characteristics of the patients. Post-Stratified results are mentioned in table II.

Table. II: Association of Occurrence of Cardiomyopathy with Patient's Characteristics

Signs	Categories	Cardiomyopathy		p value
		No n (%)	Yes n (%)	
Bone deformities	No	54 (70.1)	23 (29.9)	<0.001
	Yes	26 (40)	39 (60)	
Enlarge Spleen and Liver	No	43 (62.3)	26 (37.7)	0.162
	Yes	37 (50.7)	36 (49.3)	
Delayed Growth	No	36 (100)	0	<0.001
	Yes	44 (41.5)	62 (58.5)	
Jaundice	No	55 (61.8)	34 (38.2)	0.089
	Yes	25 (47.2)	28 (52.8)	

Severe Anemia	No	60 (78.9)	16 (21.1)	<0.001
	Yes	20 (30.3)	46 (69.7)	
Iron Overload	No	55 (66.3)	28 (33.7)	0.005
	Yes	25 (42.4)	34 (57.6)	

The post-stratification results showed that bone deformities, delayed growth, severe anemia and iron overload is associated with the occurrence of cardiomyopathy with p value <0.001, <0.001, <0.001 and 0.005, respectively.

DISCUSSION

Lee et al. analyzed a 13 years prevalence trend of thalassemia on the data of Korean population. They revealed that the prevalence of thalassemia increased over time and almost doubled in last two years. They found that only 27.7% patients of thalassemia were transfusion dependent, but the risk of incidence of comorbidities i.e. diabetes, hypertension, dyslipidemia, atrial fibrillation, myocardial infarction, end stage renal disease, stroke and congestive heart failure was more associated with blood transfusion.[9]

Dilated cardiomyopathy is the leading risk factor for heart transplantation. Ciarambino et al. reviewed the clinical trials available on 'Pubmed' till March 2021 related to cardiomyopathy. They revealed that Hypertrophic cardiomyopathy (HCM) is common in healthy subjects with the ratio of 1:500, whereas the prevalence of dilated cardiomyopathy is about 1:2500.[10] In our study, out of 62 (43.66%) cases of cardiomyopathy there were 10 (7%) cases of dilated cardiomyopathy. Non-invasive methods to monitor iron loading and unloading in the liver, heart and pancreas, can increase the patient's life expectanacy.[11,12]

Darwish et al, included the children less than 18 years of age and studied the cardiac functional changes with beta thalassemia major. They included 20 patients of BTM and 20 healthy patients and compared their Echocardiography reports. The results showed that mean ejection fraction (%) was not significantly different in BTM as compared to healthy children with p value 0.734. Echocardiography results showed that posterior wall thickness, of the left ventricle, mass of the left ventricle, mass index of the left ventricle and the thickness of the intraventricular septum was significantly in BTM patients, at 5% level of significance.[13]

The similar results were reported by Najimi etal. They included 50 adults aged between 8-37 years with BTM and compared them with 50 healthy subjects included as control. There was no significant difference between two groups with respect of BMI, heart rate, systolic and diastolic blood pressure at 5% level of significance. The results showed that echocardiography results were not significantly different between two groups.[14]

In an Indian study, the researchers included 56 adult thalassemia patients with mean duration of the disease of 12.9 ± 3.05 years. There were 21 (37.5%) patients receiving chelating therapy for against iron overload. The resulst of echocardiography showed that only 8 (14.2%) patients had systolic dysfunction.[15] In our study, the mean duration of the disease was 6.10 ± 1.53 years. The incidence of iron overload in our study was a little higher 59 (41.5%) and the prevalence of cardiomyopathy was also observed in 62 (43.66%) cases, which is quite high from the reference study.

Elfaituri et al. discussed that iron overload in BTM patients is a prognostic factor for the prevalence of cardiomyopathy.[16] Nomani et al. discussed the cardiac complication with BTM. Iron overload is associated with cardiac complications in BTM.[17] Prevalence of atrial fibrillation is associated with BTM. The researchers also concluded that the available risk score for prediction of AF is not working in BTM cases.[18,19]

In our study, there is significant association found between presence of iron overload and presence of cardiomyopathy as cardiomyopathy developed in 34 out of 59 (57.6%) cases having iron overload whereas in other BTM patients with normal iron the presence of cardiomyopathy was significantly low i.e. 28 out of 83 (33.7%), with p value 0.000.

In thalassemia major, patient completely dependent on blood transfusion to control the prognosis of the disease concurrently leading to iron overload, transplantation of bone marrow, management of iron overload, hematopoietic stem cell transplantation, stimulation of fetal hemoglobin production and gene therapy is also required.[20] We concluded that along with the management of BTM a routine (once in a 06 months) screening of cardiomyopathy helps the clinicians for early

detection and proper management of cardiac complications.

CONCLUSION: We concluded that estimated frequency of cardiomyopathies in the population of the thalassemia major patients is 0.36 with p value 0.035. It is also concluded that cardiomyopathy is associated with the symptoms of prognosis of the disease and when any of the symptom appears the patient must go for screening of cardiomyopathy.

DISCLAIMER: The current manuscript has not been presented or published in a conference or abstract book or a part of thesis/project.

CONFLICT OF INTEREST: There is no conflict of interest with regards to honorarium, funding, credits.

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