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Original Research Paper

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IRON OVERLOAD CARDIOMYOPATHY IN THALASSEMIA MAJOR :A CASE REPORT

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ABSTRACT

BACKGROUND: Cardiomyopathy is a major cause of morbidity and mortality in patients with thalassemia .Repeated blood transfusions leads to iron overload culminating in heart failure & death

CASE PRESENTATION: A 16 year old boy known case of β-thalassemia major on multiple blood transfusions and iron chelation therapy presented to emergency with features of congestive cardiac failure, predominantly of right heart failure with pedal edema and ascites .on evaluation Transthoracic echocardiography showed dilated cardiomyopathy and raised cardiac enzymes .patient was started on guideline directed management for heart failure along with iron chelation therapy

CONCLUSION: This is a case of β- thalassemia major with severe iron overload leading to dilated cardiomyopathy presenting with congestive cardiac failure.

KEYWORDS :

INTRODUCTION:

β thalassemia is an inherited disorder where there is reduced or no production of β globin chains.β thalassemia is classified into thalassemia major & thalassemia intermedia clinically.

In β-thalassemia major there is no synthesis of b globin chains leading to excess of α chains and finally hemolysis of RBCs.

This severe hemolytic anemia demands repeated blood transfusions for survival. Increased serum iron levels from hemolysis and transfusions lead to iron deposition in reticuloendothelial system and organs including heart Deposition of iron in myocardium results in cardiomyopathy and finally in heart failure

Case Presentation:

A 16 year old boy presented to emergency with complaints of dyspnea, pedal edema, abdominal distension since 15days which were gradually progressive in nature.

The patient is a known case of β- thalassemia major diagnosed at 6 months of age by HPLC analysis [hemoglobin (Hb)F: 98%; (Hb)A2: 1.2%,]. Molecular analysis revealed **homozygous mutation for IVS 1-5(G-C)** of β **globin gene .** The patient is on regular blood transfusion since 10yrs and iron chelation therapy with **Deferasirox 250mg**

Past history:

Patient underwent a total of 164 whole blood transfusions . spleenectomy was done at the age of 13years following meningococcal and pneumococcal vaccination.

Physical examination:

On examination patient had typical thalassemic facies, pallor and icterus, distended abdomen and pedal edema. Patient had respiratory distress with RR of 38cpm and saturation of 93% on Room air. His blood pressure was 100/70mmHg. Patient had raised visible jugular venous pulsations. On auscultation heart sounds heard (no murmurs), crepitations were heard in basal segments of lungs on both sides. on Abdominal examination a tender hepatomegaly was appreciated.on percussion of abdomen dullness noted in all areas.

Laboratory findings :

Complete blood picture was suggestive of hemolytic anemia with Hb of 6.2g/dl, mean corposcular volume of 84fl, platelet count of 4.3lakhs/cumm , WBC: 12000cells/cumm.peripheral smear suggestive of erythropenia,anisopoikilocytosis ,pencil forms.

Renal Function Test:

serum creatinine: 0.6mg/dl, blood urea:15mg/dl

Iron Profile:

serum iron: 340 mcg/dl serum ferritin 3600 ng/ml,total iron binding capacity(TIBC) 250mg/dl,

Liver function test:

serum glutamic oxaloacetic transaminase(SGOT)34 IU/L, serum glutamic pyruvic transaminase(SGPT)38 IU/L, serum alkaline phosphatase(ALP) 67 IU/L

Serum calcium:

8.6mg/dl, C reactive protein: negative, electrolytes: sodium: 130 potassium: 4.0 chlorides: 100 mmol/L

NT PRO BNP:

560 pg/ml

Chest X-Ray:

increased cardiothoracic ratio which is suggestive of cardiomegaly.

Echocardiography:

dilated all chambers with global hypokinesia with severe lv dysfunction (Ef 30%) moderate tricuspid regurgitation (TR).

Electrocardiogram:

left axis deviation.

Pt was started on treatment with torsemide, spironolactone, carvedilol, enalapril, deferasirox.

DISCUSSION :

Cardiomyopathy in β thalassemia presents in two different types: a dilated cardiomyopathy which leads to ventricular dilation and hence decreased ejection fraction and heart failure, a restrictive cardiomyopathy which leads to impaired left ventricular filling, pulmonary hypertension,right heart failure. Cardiomyopathy occurs due to multiple mechanisms.

Major mechanism involves the deposition of iron in myocardial cells. Inflammatory mechanisms also play a role in cardiomyopathy. Inadequate or noncompliance to chelation therapy expedites heart failure.

In a setting of inadequate transfusions there is high output cardiac failure secondary to chronic anemia which leads to tissue hypoxia, increased bone marrow,spleenic hyperplasia.

Iron overload in patients with thalassemia may occur due to ineffective erythropoiesis which leads to hemolysis, increased absorption of iron from gut. Major amount of iron deposition is by repeated blood transfusions.

Iron in the blood is bound to transferrin.when transferrin gets saturated, free iron circulates in the blood. Oxygen free radicals are produced by series of reactions which occur in the presence of free iron. Moreover this free iron gets deposited in organs such as liver,pancreas,heart.

Deposition of iron in myocardium leads to free radical mediated injury thereby apoptosis of cardiac myocytes occur and cardiac dysfunction starts in.

In Restrictive cardiomyopathy there is restrictive left ventricular filling leading to pulmonary hypertension,right ventricular dilation causing right heart failure. The left ventricular function remains normal till the end stage of the disease. Patients with right heart failure have higher iron and ferritin levels.

In Dilated cardiomyopathy the ejection fraction is decreased due to ventriclular dilation and impaired ventricular systolic function. It leads to left heart failure. It is seen that dilated cardiomyopathy occurs in younger patients with abrupt onset of symptoms. Majority of the heart failure in thalassemia is attributed to dilated cardiomyopathy Myocarditis in patients with thalassemia may also contribute for the development of cardiomyopathy.

As the patients are exposed to a high viral load by repeated blood transfusions, infections by cardiotropic viruses like adenovirus, human herpes virus, enterovirus, parvovirus B19 can lead to myocarditis. Myocardial injury occurs by cytotoxicity, cytoskeleton and extracellular matrix damage. In the course of the disease ventricular dysfunction sets in the form of dilated cardiomyopathy Immunological and inflammatory mechanisms also play a role in development of dilated cardiomyopathy In the course of progressive heart failure arrhythmias occur depending upon the stage of the disease. Intially first degree heart block, bradycardias, ST – T segment changes, occasional ventricular premature complexes occur. As the cardiomyopathy progress second degree heart block, complete heart block, ventriclular arrhythmias may occur which may lead to sudden cardiac death.

CONCLUSION:

Cardiomyopathy stands as the major cause of morbidity and mortality in patients with thalassemia.

Myocardial iron deposition which results in ventricular diastolic dysfunction , pulmonary hypertension leads to restrictive cardiomyopathy Iron overload combined with other mechanisms such as high output state, immunological and inflammatory factors lead to dilated cardiomyopathy.

Management of β-thalassemia major includes regular blood transfusions combined with adequate iron chelation therapy.

As the survival rate of β thalassemia patients beyond second decade of life is very low in India Every attempt should be made to prolong the life span with improvement of quality of life Regular monitoring of patients with Sequential echocardiograhies,laboratory indices which determine the survival should be performed.

Thus it is crucial to implement a systematic approach in managing the patients with β-thalassemia major with regular blood transfusions, adequate iron chelation therapy combined with prompt treatment for heart failure.

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