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β Thalassemia with systemic lupus erythematosus: A case and review

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ABSTRACT

β Thalassemia is a hereditary blood disorder and very common in India[1]. It is characterized by decrease in synthesis of β-chain of haemoglobin resulting in variable phenotypes. The association of Systemic Lupus Erythematosus (SLE) with β Thalassemia traithas rarely reported[2]. Moreover, there is data lacking from India[3,4]. We are reporting a β thalassemic, 8 year old male child whodiagnosed for SLE. Pediatricians should alert for this association so the diagnosis and treatment cannot be delayed.

KEYWORDS

Beta-Thalassemia;
Systemic Lupus Erythematosus (SLE).

CASE REPORT

We report an 8year old male child, a diagnosed case ofβthalassemia since 9 month of age. He received multiple blood transfusions since early infancy. However, he was never received iron chelation. He presented with complaints of fever, arthralgia, bone pain, skin rash from last 6weeks with history of increased need for blood transfusion over last 5-6 months. Fever was intermittent, low to high grade, was not associated with chills and rigors. On physical examination we foundhemolytic facies Figure 1, pallor, high body temperature (101.8p F) and tachycardia (HR=120/min), otherwise hemodynamically stable. He had short stature (height < 3rd centile) and was severely undernourished. He had multiple petechial spots over chest and abdomen. He has bony tenderness, which was attributed to haemolytic anaemia. Abdominal examination revealed massive splenomegaly (15 cm below left costal margin) with ascites Figure- 2. Rest of the systemic examination was unremarkable. Features like pallor, bony tenderness, growth failure, weakness were explained by beta thalassemia alone. But other features like fever, skin rash, petechial rashes and increased need for blood transfusion werenot explained. So possibility of infection, malaria and collagen vasculardisorders were kept. Child worked up accordingly. Haematological tests revealedhemoglobin of 4.8 gm/dl, WBC 7200/cumm andDLC includedneutrophils 51%, and lymphocytes 43%, platelets were 11,000/cumm. Blood indices included MCV of 78.7, MCH of 27.3, and MCHC of 34.7. Peripheral blood smear showed normocytic normochromic anaemia with few hypochromic microcytic cells. Few pencil cells and tear drop cells also seen. Reticulocyte count was 4.0 % and ESR was 98 mm in first hour. Malarial parasite test, widal, blood culture and tubercular work up was negative. Child received blood transfusion on hemoglobin of 4.0 gm/

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dl. Blood samples were preserved before transfusion for work up. After transfusion, the hemoglobin level increased to 6 gm% but within 5 days again came down to 4.2 gm/dl. The renal function test was normal other than mild proteinuria. Stool analysis was normal. Liver function test revealed bilirubin was 1.5 mg/dl with mild derangement of transaminases. Viral markers like hepatitis A, B, C and HIV were negative. USG abdomen showed enlarged liver and spleen with normal echo texture. Further investigations revealed a strongly positive direct coombs test. Anti-Nuclear factor and anti-dsDNA were positive. Serum complement level (C3) was also reduced (69 mg/dl). The qualitative tests for anti-SS A and anti Sm were also positive. Child has skin rash, anemia, thrombocytopenia, proteinuria, arthritis/ arthralgia and positive serology for SLE thus fulfilled the diagnostic criteria for it. Child received methyl prednisolone, followed by kept on oral steroids. On follow up the need for blood transfusion reduced and hemoglobin start to increase.

DISCUSSION

This case reportdescribed an 8 year old child, who was a diagnosed case of β thalassemia trait since 9 months of age. At this admission he diagnosed to have SLE. The association has remained under diagnosed because of overlapping of clinical features of β thalassemia and SLE. The association remained uncommon as limited number of patients hasbeen reported so far. Cutaneous manifestations are not so frequently mentioned. Most of the patient presented with fever and increased need of blood transfusion. There are many causes for increased need for blood transfusion in B thalassemia patients including formation of alloantibodies, infections and collagen vascular disorders[5]. High clinical suspicion is neededto diagnose SLE in a thalassemic patient who hashigh unexplained blood transfusion requirements. The underlying mechanism for the association of β thalassemia with SLE is partially explained. It has been found that hemoglobin β gene is in close proximity to other genes for immune regulation like STIM1, CD151and TC21/RRAS22. These genes are involved in anti-inflammatory activity. These anti-inflammatory genes may also get deleted with the globin gene β[6]. This may account for the association of beta thalassemia with some autoimmune disorders like SLE. *Castellino G et* al published a series of 177 patients with SLE, out of

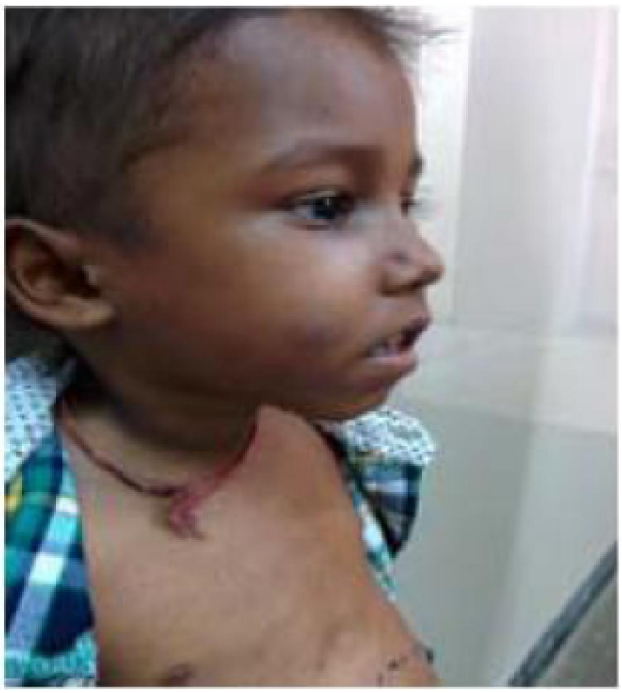

Figure 1

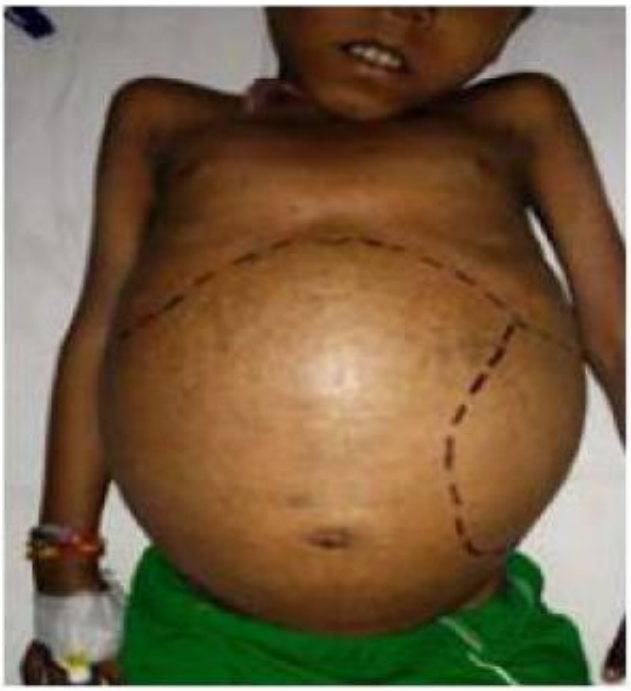

Figure 2

them17 cases had thalassemia[2]. All these17 cases were female. Increased incidence of other autoimmune disorders was also noted. However, our case was a male child but he also hadpositive anti-SS-A but no evidence of serositis. The diagnosis of SLE in a case of thalassemia is difficult in absence of florid clinical features. Auto antibodies used to diagnose SLE may be false positive because of multiple transfusions inthalassemia patients[7]. Beta thalassemia was also associated with other hemoglobinopathies. Sickle cell disease with β thalassemia has reported more commonly than β thalassemia with SLE[8]. The association may aggravate the disease severity. The patients can develop lupus anticoagulant and can cause intracranial hemorrhage. Prednisolone is said to be effective in lupus anticoagulant positive cases[9]. Our patient recovered with pulse methyl prednisolone therapyand is now being maintained on oral steroidsand hydroxychloroquine. Repeat blood tests showed hemoglobin 8.7 gm/dl after 2 week of therapy. The need for increased blood transfusions came down. Child was monitored for other features of SLE during follow ups. Cases that have multi system involvement needed aggressive immunosuppression therapy.

CONCLUSION

In thalassemia cases if there increased need for blood transfusion over period of time SLE should rule out. Early diagnosis and treatment may decrease morbidity and mortality.

CONSENT

Written informed consent was obtained from the parents for publication of this case report and with images.

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