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Case Series on Hemoglobin E-Beta-Thalassemia Major

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ABSTRACT

Aim: The aim of case series is to describe the 3 hemoglobin E ß-thalassemia cases, which are unique and requires special care and attention to diagnose/manage. Its natural history is little known and also the reasons for their clinical diversity or/and its management.

Presentation of case: 3 cases of transfusion dependent hemoglobin E β-thalassemia major were included in the study. The patients reported similar complaints of weakness and delayed milestones. The patients were on regular red blood cell transfusion and iron chelation therapy from the age of 3 years. The beta-globin gene defects were defined in all the cases using similar techniques. Thalassemia mutation analysis by reverse clot blot testing showed a compound heterozygous for IVS 1-5 [G-C] and codon 26 [G-A] beta E mutation in the beta globin gene. Evaluation for iron overload showed severe cardiac iron deposit and severe hepatic iron deposit on MRI T2. During hospital stay, the patients received antibiotics and immune-suppressants in common.

Discussion and conclusion: Patients are treated by lifelong blood transfusion every 15 to 30 days along with iron chelation therapy. Repetitive transfusions cause iron overload, with life-threatening complications, like such as cardiomyopathy, endocrine disorders, liver failures and ultimately, premature death. Awareness, education and screening play the most important part in the prevention of life-threatening complications and control of thalassemia.

Keywords: Hb E/β thalassemia, Transfusion dependent, Hb E mutation, Thalassemia major

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