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Case Report

Paraparesis in Hb D-Hb E Heterozygous Thalassemia- a Case Report

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Abstract:

Thalassemia is a group of autosomal recessive hemoglobinopathies where exramedullary hemopoisis is frequently observed in the liver, lymph nodes, spleen and also around spinal cord which may cause spinal cord compression producing different clinical manifestations including para paresis or paraplegia. A case report of thoracic spinal cord compression in a 20-year-old male with Hb D-Hb E heterozygous thalassemia is reported from medicine department of Bangabandhu Sheikh Mujib Medical College, Faridpur. Bangladesh. This patient presented with progressive weakness of both lower limbs and difficulties in walking for 2 months. He also had history of fluctuating jaundice since childhood. His hemoglobin electrophoresis was suggestive of Hb D-Hb E heterozygous and MRI of dorsal spine with screening whole spine reveals extramedullary hemopoiesis at D2 to D12 causing spinal cord compression and low signal intensity of the bone marrow of the vertebrae. After blood transfusion he underwent referred to higher specialized center (National Institute of Neurosciences and Hospital, Dhaka) for further better management.

Key words: Hemoglobinopathy, Paraplegia.

Introduction:

Thalassemia is a group of autosomal recessive hemoglobinopathies affecting the production of normal alpha- or beta-globin chains that comprise hemoglobin. Thalassemia prevalence is highest in Africa, India, the Mediterranean, the Middle East, and Southeast Asia[1]. The compensatory mechanisms in thalassemia lead to diffuse marrow hyperplasia, yellow to red marrow reconversion, osteopenia, and pathologic fractures. Furthermore, excess iron in blood is a natural byproduct of frequent blood transfusions and hemolysis. Exramedullary hemopoisis (EMH) is frequently observed in the liver, lymph nodes, spleen and in 11 to 15 percent of cases have a para spinal site involvement where in paravertebral area produces pseudo tumors. Depending on the site of the spinal cord is involved, the various neurological symptoms can develop[2]. Thalassemia can cause neuropathy, myopathy, and evoked potential (sensory, auditory, and visual). Additionally, this increases the risk of a hypercoagulable state that results in stroke[3]. Paraspinal extramedullary hematopoietic pseudotumors may cause a variety of neurological symptoms due to spinal compression although more than 80% of cases may remain asymptomatic and neurologic symptoms mostly found in third and fourth decades of life[4,5]. Most frequently observed spinal cord compression seen in the thoracic spine. Multiple minor compression fractures may eventually result in biconcave abnormalities of the vertebral bodies, also referred to as "fish-type" vertebrae, and weakening of the subchondral bone

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plates[6]. Lobulated intraspinal epidural extramedullary masses and widespread low signal intensity of the bone marrow of the thoracic and lumbar vertebrae may be seen in MRI (Sagittal T2 Weighted images)[7].

Our patient is 20 years old unmarried male presented with progressive weakness of both lower limbs and difficulties in walking for 2 month. He also had history of fluctuating jaundice since childhood. He had no history of any blood transfusion and none of his family member was affected with such a disease. He was moderately anaemic with moderate splenomegaly. Neurologically found spastic paraparesis with motor 2/5 and impaired all modalities of sensation up to T10 dermatome from below. His bladder and bowel habit was intact. His Hb was 7 gm/dl, MCV 70 fl, and PBF was microcytic hypochromic. Haemoglobin Electrophoresis shows suggestive of Hb D-Hb E heterozygous (HbA 14.5% (N= 95-98%), HBF 40.2% (N=0-0.9%), HbD 5.2%. His MRI of dorsal spine with screening whole spine reveals extramedullary hemopoiesis at D2 to D12 causing spinal cord compression and low signal intensity of the bone marrow of the vertebrae.

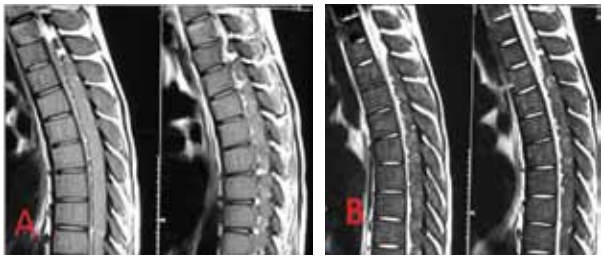

Figure A: MRI dorsal spine T1WI sagittal sections show an isointense epidural lesion posterior to the cordextending from D2 to D12

Figure B: MRI dorsal spine T2WI sagittal sections show a hyperintense epidural lesion posterior to the cord and low signal intensity of the bone marrow of the vertebrae extending from D2 to D12

Discussion:

Gatto first described spinal cord compression from the extra medullary overgrowth of the hematopoietic tissue[8]. Savini et al described a paraparesis case from spinal cord compression at T6-T7 levels in a compound heterozygote for haemoglobin E and for β-thalassemia. The patient underwent a full recovery after the mass excision and an emergency T3-T9 laminectomy[9]. Taher A et al reported a 25-year-old male with moderate phenotypic hemoglobin E/β-thalassemia not being able to walk and who had never received a transfusion[10]. Ghieda U et al. reported a 21-year-old man with β-thalassemia intermedia with progressive low back pain, worsening para paresis and sphincter disturbance. MRI demonstrated spinal cord compression by EMH epidural mass lesions extending from T3 to T10 levels[11]. SA Salehi et al found a 34-year-old male with a 12-month history of pain in the mid-thoracic region, paresthesia in his feet, progressive difficulty with ambulation and increased frequency of urination with urgency[12].

Our case is almost similar to above mentioned cases of different authors in consideration of age and sex but pattern of haemoglobinopathies are different. Their observed Phenotypic distribution were hemoglobin E/β-thalassemia, β-thalassemia intermedia, in contrast we found Hb D-Hb E heterozygous which is an uncommon phenotype. Hemoglobin E (HbE) is an extremely common structural hemoglobin variant in Asian countries and usually causes mild form of β thalassemia. But interactions with different forms of α thalassemia and its coinheritance with β thalassemia, a condition called hemoglobin E β thalassemia, is by far the most common severe form of β thalassemia in Asia and, globally, comprises approximately 50% of the clinically severe β-thalassemia disorders.

Hb D is asymptomatic in the heterozygous state and is marked by mild hemolytic anemia and chronic non progressive splenomegaly. No treatment is required. When Hb D is co-inherited with β^0-thalassemia, patients have mild microcytic anemia, but when inherited with Hb S it usually produces a mild form of sickle cell disease[13]. Our patient is of Hb D-Hb E heterozygous which has produced an advanced level of manifestations not yet mentioned.

Conclusion:

Among neurological manifestations paraparesis is a common one observed in HbE/ beta- thalassemia heterozygous and beta- thalassemia intermedia. But HbE incoherence with Hb D (HbD-HbE hetrerozygous) may also cause paraparesis.

References:

1. Baird DC, Batten SH, Sparks SK. Alpha- and Beta-thalassemia: Rapid Evidence Review. *Am Fam Physician*. 2022 Mar 1;105(3):272-280. PMID: 35289581.

2. Dore F, Cianciulli P, Rovasio S, Oggiano L, Bonfigli S, Murineddu M, et al. Incidence and clinical study of ectopic erythropoiesis in adult patients with thalassemia intermedia. *Ann Ital Med Int*. 1992;7(3):137–40.

3. Shah FH, Idrees J, Shah ST, Khan R, Khan AT, Salman S, et al. Neurological deficits among beta-thalassemia patients and its possible therapeutic intervention: a comprehensive review. *Journal of the Dow University of Health Sciences*. 2020 Aug 30;14(2):83-90.

4. Parsa K, Oreizy A. Nonsurgical approach to paraparesis due to extramedullary hematopoiesis. Report of two cases. *J Neurosurg*. 1995;82(4):657–60.

5. Issaragrisil S, Piankijagum A, Wasi P. Spinal cord compression in thalassemia: report of 12 cases and recommendations for treatment. *Archives of Internal Medicine.* 1981 Jul 1;141(8):1033-6.

6. Adamopoulos SG, Petrocheilou GM. Skeletal Radiological Findings in Thalassemia Major. *JRPMS*. September 2020; 4(3):76-85.

7. Subahi EA, Abdelrazek M, Yassin MA. Spinal cord compression due to extramedullary hematopoiesis in patient with Beta thalassemia major. *Clin Case Rep*. 2020 Dec 15;9(1):405-9. doi: 10.1002/ccr3.3542. PMID: 33489190; PMCID: PMC7812994.

8. Gatto I, Terrana V, Biondi L. Compression of the spinal cord due to proliferation of bone marrow in epidural space in a splenectomized person with Cooley's disease. *Haematologica*. 1954 ;38(1):61-76. PMID: 13151530.

9. Savini P, Lanzi A, Marano G, Moretti CC, Poletti G, Musardo G, et al. Paraparesis induced by extramedullary haematopoiesis. *World J Radiol*. 2011 Mar 28;3(3):82-4. doi: 10.4329/wjr.v3.i3.82. PMID: 21512655; PMCID: PMC3080054.

10. Taher A, Vichinsky E, Musallam K, Cappellini MD, Viprakasit V. Guidelines for the Management of Non Transfusion Dependent Thalassaemia (NTDT). *Thalassaemia International Federation, Nicosia (Cyprus)*; 2013. PMID: 24672826.

11. Ghieda U, Elshimy M, El Beltagi AH. Progressive Spinal Cord Compression Due to Epidural Extramedullary Hematopoiesis in Thalassemia Intermedia: A Case Report and Literature Review. *The Neuroradiology Journal*. 2013; 26(1):111-7. doi:10.1177/197140091302600119.

12. Salehi SA, Koski T, Ondra SL. Spinal cord compression in beta-thalassemia: case report and review of the literature. *Spinal Cord*. 2004 Feb;42(2):117-23. doi: 10.1038/sj.sc.3101544. PMID: 14765145.

13. Randolph TR. Hemoglobinopathies (structural defects in hemoglobin). Rodak's hematology: clinical principles and application. St Louis, MO: Elsevier. 2019 Feb 22:394-423.