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Acute Thoracic Compressive Myelopathy as a Rare Manifestation of Thalassemia: A Case Report

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Abstract: ***Acute thoracic compressive myelopathydue to extra medullaryhaematopoiesis and wedge compression fracture can occur as a rare complication in beta thalassemia. Here we present a 22 year thalassemic male presented with acute thoracic compressive myelopathy. On examination, power of upper limb 5/5, power of both lower limbs 0/5, definite sensory level at T10, UMN type bladder. MRI shows acute wedge compression fracture D7 vertebra and several paravertebral masses with expansion of osseous medullary spaces suggestive of extra medullary haematopoiesis. The patient did not improve on medical management, and the patient party refused for surgical treatment.***

Keywords: Thalassemia, Thoracic compressive myelopathy, Extra medullary haematopoiesis, Paravertebral mass, Bone mineral density.

1. Introduction

Thalassemia is an autosomal recessive disorder, also known as Cooley's disease, characterized by insufficient or absent globin chain production leading to ineffective erythropoiesis causing the expansion of extrahemotopoietic tissues. Extra medullaryhematopoiesis (EMH) in paravertebral areas can lead to compression of spinal cord causing various neurological manifestations.

Here we report a rare case of spinal EMH with dorsal cordcompression and myelopathy in a 22-year-oldthalassemia male with who presented with paraparesis and acute sphincter disturbance.

2. Case Report

A 22 year old male presented with upper back pain since 3 days, weakness of both the lower limb since 3 days, loss of sensation below the level of umbilicus since 3 days, not able to pass stool and urine voluntarily since 3 days. Patient is a known case of beta thalassemia major for which he is transfusion dependent since childhood. Patient underwent splenectomy at the age of 3 years. No history of trauma or fever and no history of previous similar condition. Higher mental function and cranial nerve examinations were within normal limits. Bulk and tone of muscles of both lower limb as well as both upper limb was normal. Power of muscles around shoulder, elbow and wrist joints was 5/5 bilaterally. Power of muscles around hip, knee and ankle joint was 0/5 bilaterally. Knee jerk and ankle jerk was brisk bilaterally. Biceps, triceps and supinator jerk was normal bilaterally. Plantar response was extensor bilaterally, with absent abdominal as well as cremasteric reflexes. All modalities of sensation like fine touch, vibration, proprioception, pain and temperature were absent below the level of T10. Spinal tenderness was present at thoracic region from T2 to T 10 without any kyphosis, scoliosis or gibbus. Laboratory investigation shows haemoglobin of 6.9 g/dl; LFT, RFT and electrolytes were within normal limits. HPLC shows HbA_2-3.8%, HB F-91.7%. MRI thoracic spine shows acute wedge compression fracture D7 vertebra with severe loss of vertebral body height (50%) with adjacent marrow edema and retropulsion of fracture fragment, causing severe stenosis of spinal canal and cord compression. $D_{4\text{-}10}$ levels shows reduced disc space, with posterior bulge causing indentation on thecal sac and mild stenosis of spinal cord (7mm), several paravertebral masses with expansion of osseous medullary spaces suggestive of extra-medullary haematopoiesis. Patient was treated conservatively, and the neurological condition did not improve on medical management.

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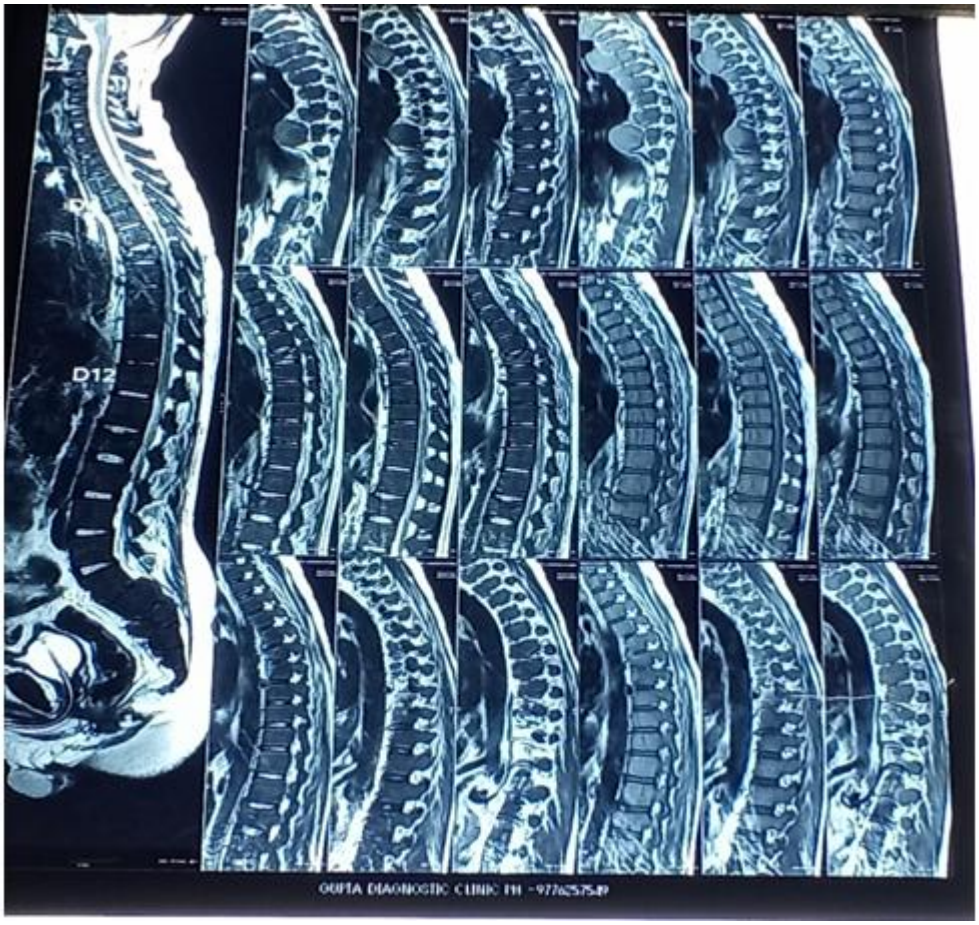

MRI image showing wedge compression fractures and extra medullary hematopoietic masses.

3. Discussion

Thalassemia major is an autosomal recessive disorder with complete inability to produce beta chains. About 1.5% of the worldwide populations are carriers of the gene, from which 60, 000 symptomatic individuals are born annually[1]. Paraspinal extramedullary hematopoiesis (EMH) is more common in males with M: F ratio ranging from 2.5: 1 to 5: 1 and occurs mainly in chronic cases in the third and fourth decades of life[2]. Thalassemia bone diseases (TBD) have become a major cause of skeletal complications. These unusual skeletal changes include decreased bone mineral density (BMD), spontaneous fractures and spinal deformities with compression of the vertebrae and nerves often causing severe pain and discomfort[3].

Extra medullaryhematopoiesis (EMH) occurs as a response to chronic anemia states as a homeostatic mechanism via the production of erythropoietin[4]. Pathologically, the plausible hypothesis suggested for occurrence of paraspinal EMH includes, [5]i) Direct extension of erythrogenesis from adjacent vertebral bone marrow, ii) Common embryonic origin from thoracic hematopoietic tissue masses, iii) Development from branches of the intercostal veins, iv) Arterial embolus.

Patients with TM display an unbalanced bone turnover with an increased resorption phase and decreased formation phase, resulting in severe bone loss[6]. The primary disease causes bone marrow expansion due to ineffective erythropoiesis, leading to mechanical interruption of bone histology, cortical thinning, increased distortion and, finally, bone fragility[7]. Polymorphism at the Sp1 site of COLIA 1 has been associated with severe osteoporosis and pathological fractures of the spine and hip in TM patients in addition to the effects of the vitamin D receptor gene (VDR) polymorphisms[1].

Factors predisposing to TBD[8]:

1) Bone marrow expansion
2) Iron overload
3) Hypogonadism
4) Parathyroid dysfunction
5) Hypothyroidism and hyperthyroidism
6) Growth hormone deficiency
7) Diabetes
8) Liver disease
9) Renal disease
10) Vitamin C, D, K deficiency.

A MRI is the diagnostic imaging of choice for extra medullary hematopoiesis and it shows the presence of a lobulated mass, hyperintense on T1 and T2-weighted images with minimal post-contrast enhancement[9]. Dual Energy X-ray Absorptiometry (DXA), which is a non-invasive technique and can be performed on the hip, lumbar spine, and distal radius, is still the gold standard for the measurement of BMD. Based on DXA findings, osteoporosis is defined as a BMD T-score $\leq$2.5 leading to higher risk of fracture and osteopenia as a BMD T-score

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between -1 and -2.5, with normal values of T-score being ≥1.0.

4. Conclusion

Paraplegia due to compression fracture and extra medullaryhaematopoiesis is an uncommon presentation in thalassemia patients. Early diagnosis and treatment can prevent irreversible neurological damages. Key recommendations includes the following but not limited to i) Regular blood transfusion, ii) Annual check-up of BMD starting at adolescence iii) Annual check-up of bone markers iv) Encouragement of physical activities v) Adequate calcium and vitamin D intakevi) Iron chelation therapy.

Conflict of interest: There was no conflict of interest.

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