


## Case Report

# A patient with beta thalassaemia major presenting with cauda equina syndrome due to extramedullary haematopoiesis

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## Introduction

Beta thalassaemia major is an autosomal recessive haemoglobinopathy in which there is minimal to no beta globin chain synthesis, leading to profound transfusion-dependent anaemia [1]. Extramedullary haematopoiesis in thalassemia is usually secondary to undertreated severe anaemia.

Extramedullary haematopoiesis can occur in any organ that participates in haematopoiesis during fetal development. Paraspinal involvement leading to cauda equina syndrome has been reported in a few case reports [2].

## Case presentation

A 16-year-old female with beta thalassaemia major on monthly blood transfusions with a history of splenectomy at the age of six years, was admitted with insidious onset bilateral lower limb weakness for seven days, worsening over three days. On admission, she was unable to walk even with support. There was no upper limb involvement. She had concomitant urinary retention with faecal incontinence. There was no history of a fall or trauma to the back.

On examination, she was moderately pale and had mild hepatomegaly. Neurological examination revealed bilateral asymmetrical lower limb flaccid paraparesis with right side being more affected (right side power 2/5, left side power 3/5). Both ankle and knee jerks were absent, bilaterally. The plantar reflexes were flexor. There was sensory impairment for both fine touch and pain in the lumbar (L)5 and sacral (S)1-3 areas in both limbs with right side being more affected. Proprioception was intact. Saddle anaesthesia (L5- S2) was

noted with reduced anal tone. Upper limb examination was normal and there was no spinal tenderness.

A diagnosis of cauda equina syndrome was made and she was started on intravenous (IV) dexamethasone 8mg 8 hourly.

The full blood count revealed a hypochromic microcytic anaemia (haemoglobin- 6.2 g/dl). Inflammatory markers were normal. Urgent magnetic resonance imaging (MRI) of the lumbar sacral region was arranged which revealed an extradural lesion causing cauda equina compression at L2/ L3 levels most likely due to extramedullary haematopoiesis [Figure 1].

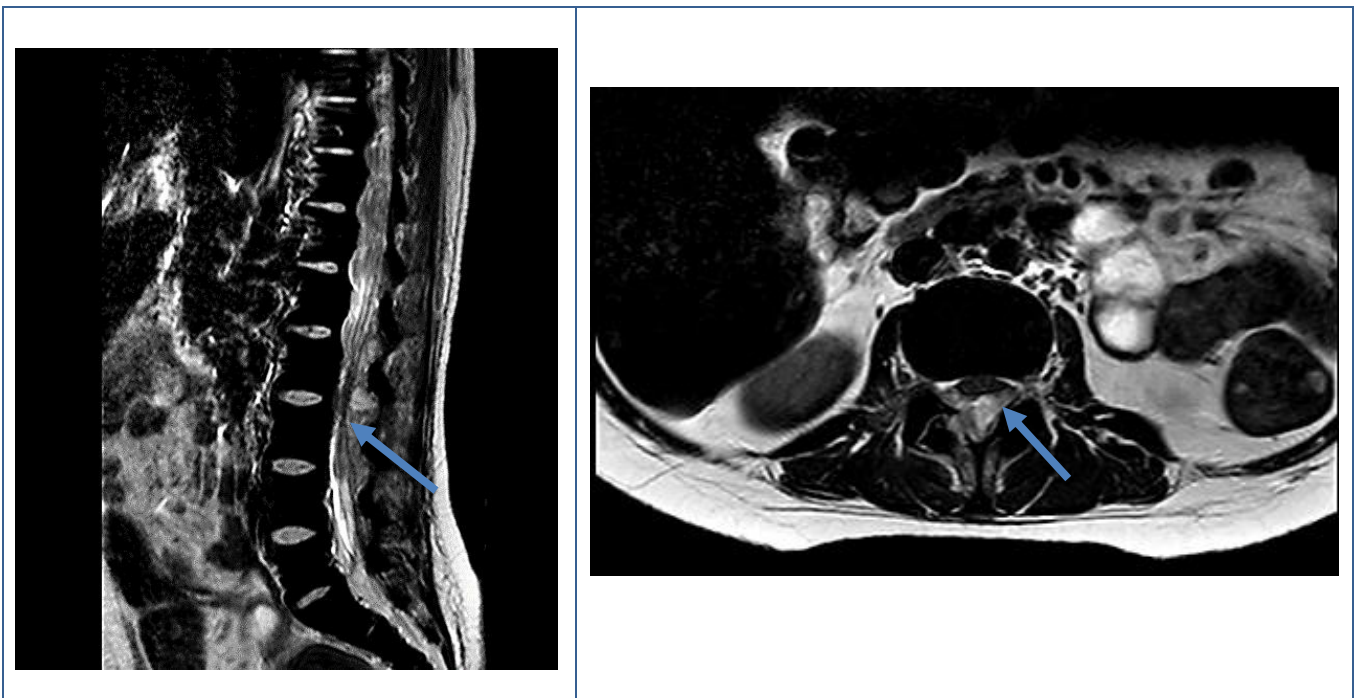


Figure 1: Sagittal view and axial view of MRI of lumbar spine showing canal stenosis with cauda equina compression at L2/L3 level caused by the extra-dural lesion at L2/L3 levels posteriorly

Urgent laminectomy and spinal canal decompression was performed by the neurosurgical team. She was discharged three weeks after the surgery and was able to walk with support by then. With continued physiotherapy she was able to walk independently eight weeks after surgery.

### Discussion

Extramedullary haematopoiesis is a well-recognized complication of conditions with ineffective haematopoiesis which results in production of blood elements outside the bone marrow.

Paraspinal involvement manifests with debilitating clinical consequences secondary to nerve compression. Various clinical presentations have been reported including back pain, paraesthesia, paraplegia and bladder and bowel involvement [3]. Early diagnosis of

paraspinal involvement is important in reducing irreversible neurologic damage that may occur with prolonged undiagnosed cord compression. Currently, MRI is considered the method of choice for diagnosis [3].

According to published data, both transfusion and hydroxyurea therapy have a role in the prevention and management of paraspinal involvement with extramedullary haematopoiesis [4]. Surgical decompression also plays a role depending on the clinical severity [5].

A tentative diagnosis of cauda equina syndrome was made in our patient on admission and she was started on steroids in view of reducing the extent of permanent nerve damage. She was referred to the neurosurgical unit after confirming the diagnosis with MRI and laminectomy was performed which resulted in a dramatic response. This case highlights the importance of early diagnosis and treatment of paraspinal involvement due to extramedullary haematopoiesis in patients with beta thalassaemia.

## References

1. Galanello R, Origa R. Beta-thalassemia. *Orphanet journal of rare diseases*. 2010 Dec;5(1):1-5. <https://doi.org/10.1186/1750-1172-5-11>
2. La VT, Diatte M, Gaston J, Dick D, Sweiss R, Pakbaz Z. Spinal cord compression due to extramedullary hematopoiesis in a patient with E-beta-thalassemia managed without radiation or surgery. *Journal of community hospital internal medicine perspectives*. 2018 Jul 4;8(4):246-9. <https://doi.org/10.1080/20009666.2018.1490141>
3. Haidar R, Mhaidli H, Taher AT. Paraspinal extramedullary hematopoiesis in patients with thalassemia intermedia. *European Spine Journal*. 2010 Jun;19(6):871-8. <https://doi.org/10.1007/s00586-010-1357-2>
4. Lim JL, Chan LL, Lim JL, Lin YL. Spinal stenosis secondary to extramedullary hematopoiesis in beta-thalassaemia. *QJM: An International Journal of Medicine*. 2018 Sep 1;111(9):659-60., <https://doi.org/10.1093/qjmed/hcy055>
5. Soman S, Tharadara GD, Chhatrala N, Jain S. A case report of extramedullary haematopoeisis in lumbosacral region presenting as cauda equina syndrome. *International journal of spine surgery*. 2016 Jan 1;10. <https://doi.org/10.14444/3025>