EAS Journal of Radiology and Imaging Technology
Abbreviated Key Title: EAS J Radiol Imaging Technol
ISSN: 2663-1008 (Print) & ISSN: 2663-7340 (Online)
Published By East African Scholars Publisher, Kenya

OPEN ACCESS

Volume-7 | Issue-4 | Jul-Aug-2025 | DOI: https://doi.org/10.36349/easjrit.2025.v07i04.005

Case Report

Radiological Features of Extramedullary Hematopoiesis in a Young Male with Beta-Thalassemia: A Case Report

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Article History
Received: 03.06.2025
Accepted: 11.07.2025
Published: 14.07.2025

Journal homepage:
https://www.easpublisher.com

Quick Response Code

Abstract: Hematopoiesis refers to the generation and maturation of blood cells. In adults, this process predominantly takes place within the bone marrow of long bones, vertebrae, and ribs. During fetal development, however, hematopoiesis primarily occurs in the yolk sac, liver, and spleen. When hematopoiesis shifts from the bone marrow to other sites in the body, it is termed extramedullary hematopoiesis (EMH). This phenomenon often arises in individuals with hematologic disorders such as thalassemia and sickle cell anemia, where normal bone marrow function is impaired. Here, we describe a case of a young adult male with beta-thalassemia who presented with complaints of shortness of breath and palpitations persisting for one month. This report highlights the imaging features of extramedullary hematopoiesis identified in this patient.

Keywords: Hematopoiesis, Extramedullary hematopoiesis (EMH), Thalassemia, Bone marrow, Hemoglobinopathies.

INTRODUCTION

Hematopoiesis is the physiological process responsible for the production and maturation of blood cells [1]. In adults, this function is primarily confined to the bone marrow of long bones, ribs, and vertebral bodies, while in fetal life, the yolk sac, liver, and spleen serve as the main hematopoietic organs. When these primary marrow sites fail, as seen in conditions like myelofibrosis (which may result from genetic mutations, chemical exposures such as toluene, or radiation) and hemoglobinopathies like sickle cell disease and thalassemia, extramedullary tissues take over the role of blood cell production [1].

Thalassemia is a genetic disorder inherited in an autosomal recessive manner, characterized by chronic anemia resulting from defective hemoglobin synthesis [2]. Chronic hemolysis in such disorders triggers compensatory mechanisms, with extramedullary hematopoiesis (EMH) being one of the most common responses [3]. EMH refers to the development of hematopoietic tissue outside the bone marrow, most often occurring in conditions like myelofibrosis, leukemia, lymphoma, and thalassemia.

The liver, spleen, and thoracic paraspinal regions are the most frequently involved sites of EMH, though it can manifest in virtually any tissue or organ. When present in unusual locations, EMH can mimic tumors, causing mass-related symptoms. Correctly identifying the imaging characteristics of EMH is critical, as biopsy and histopathology can confirm the diagnosis and help avoid unnecessary treatments intended for malignancy [1].

In this report, we describe a young adult male with transfusion-dependent beta-thalassemia, who presented with complaints of breathlessness and palpitations for one month.

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CASE PRESENTATION

A 28-year-old male with a known history of beta-thalassemia major, dependent on periodic blood transfusions and maintained on daily folic acid supplementation, presented to our institution with complaints of chest pain and palpitations. His symptoms, which had begun a month prior, were initially intermittent and relieved by rest but had progressively worsened over the last 10 days.

The patient had undergone splenectomy eight years ago. He also reported irregular compliance with blood transfusion schedules. On admission, laboratory investigations revealed a red blood cell count of 3.42 x 10^{12}/L (normal range: 4.3–5.8 x 10^{12}/L) and a hemoglobin concentration of 8.3 g/dL (normal range: 14–18 g/dL). Peripheral smear examination demonstrated elevated eosinophil counts.

Radiological evaluation began with a skull X-ray, which showed a widened diploic space and the classic "hair-on-end" appearance, indicating active marrow hyperplasia. The outer and inner tables of the calvarium were notably thinned (Figure 1). A chest X-ray revealed expansion of multiple ribs, particularly at the costovertebral junctions and the anterior aspects of the ribs, suggestive of marrow hyperplasia secondary to chronic anemia (Figure 2).

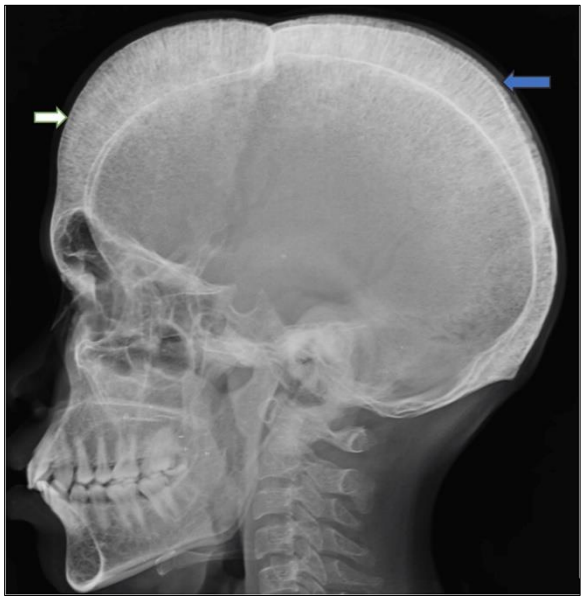

Figure 1: Skull X-ray lateral view showing widened diploic space of frontal (white arrow) and parietal (blue arrow) bones giving a hair-on-end appearance

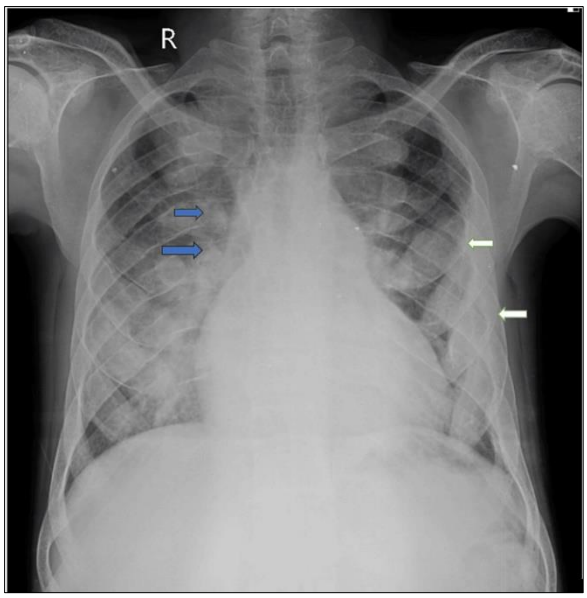

Figure 2: Chest X-ray posteroanterior view showing the expansion of multiple ribs at the costo vertebral junctions (blue arrows) and anterior aspect of the ribs (white arrows)

A CT scan demonstrated diffuse expansion of the vertebrae, ribs, and sternum (Figure 3). Additionally, multiple well-circumscribed, smooth, lobulated soft tissue density masses were identified in the bilateral paravertebral and posterior mediastinal regions, subpleural areas, and near the anterior ends of the ribs, findings suggestive of thoracic extramedullary hematopoiesis (EMH). The largest soft tissue mass, measuring 3.2 x 1.8 cm, was located in the anterior subpleural region adjacent to the right first rib (Figure 4). The lungs appeared diffusely reduced in volume, and cardiomegaly was also observed (Figure 5).

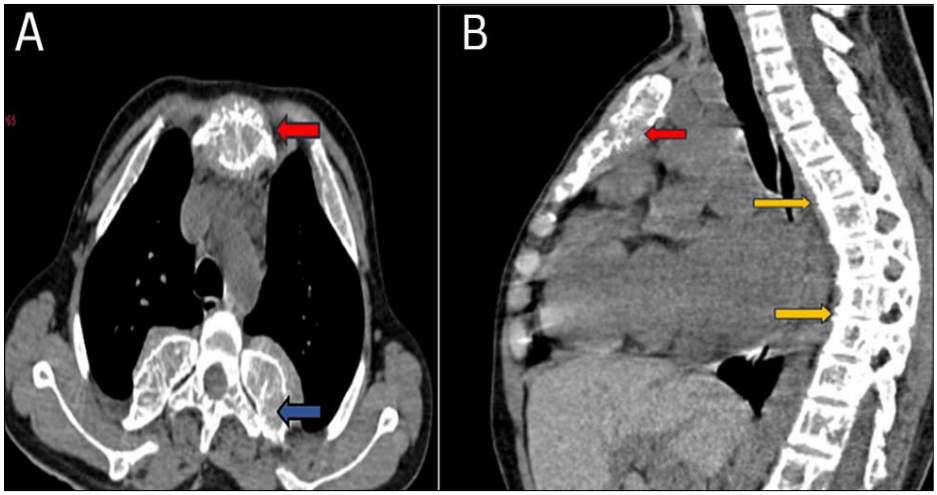

Figure 3: CT thorax - (A) axial section showing expanded sternum (red arrow), ribs (blue arrows) and (B) kyphotic deformity of the spine with reduced intervertebral disc spaces (yellow arrows)

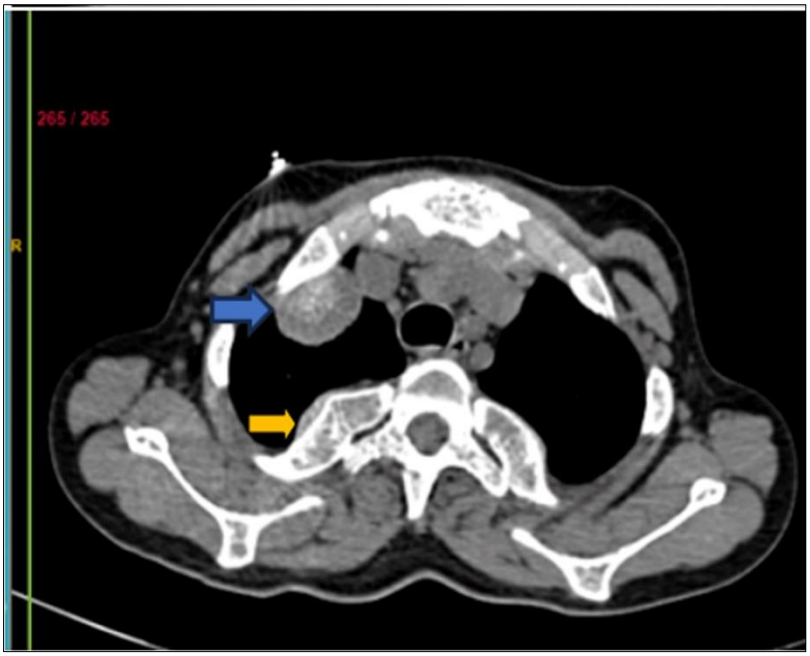

Figure 4: CT thorax axial section revealed diffusely expanded vertebrae (yellow arrow) and soft tissue mass (blue arrow) along the anterior end of the ribs

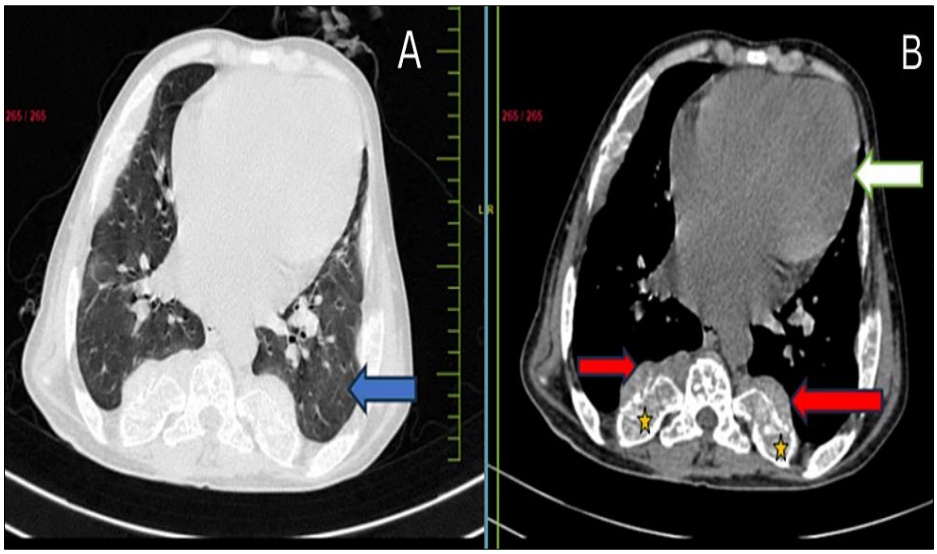

Figure 5: CT scan axial section - (A) Lung window: Lungs show diffusely decreased lung volume (blue arrow); (B) Mediastinal window showing enlarged heart suggesting cardiomegaly (white arrow) and expanded ribs (star) with paraspinal soft-tissue masses adjacent to them

Kyphotic deformity of the dorsal spine with loss of bony trabeculae and reduced intervertebral disc spaces were noted. The visualized part of the liver shows diffuse hyperattenuation, suggesting secondary hemochromatosis. The spleen was not visualized in the left hypochondrium, which is consistent with post-splenectomy status (Figure *6*).

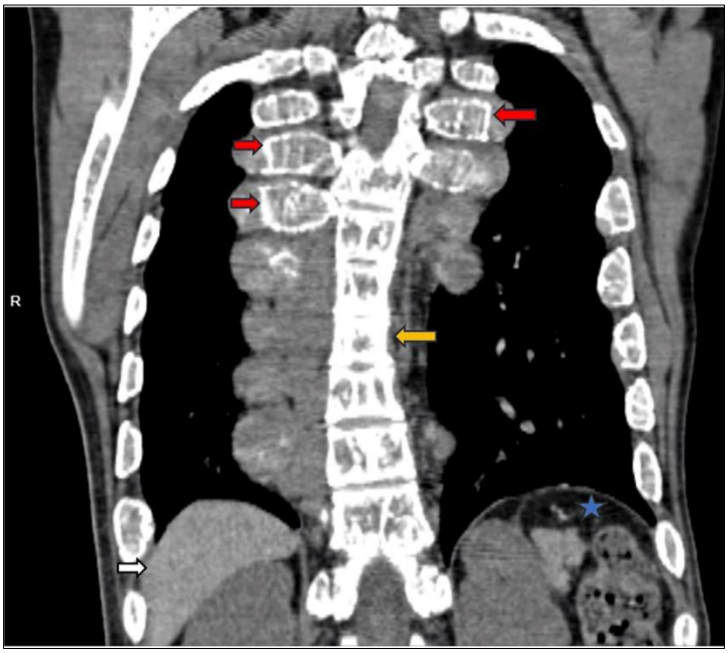

Figure 6: CT coronal section showing kyphotic deformity of the dorsal spine with loss of bony trabeculae (yellow arrow) and reduced intervertebral disc spaces. Bilateral ribs appear expanded (red arrows). The visualized part of the liver shows diffuse hyperattenuation (white arrow). Spleen was not visualized in the left hypochondrium, which is consistent with post spleenectomy (star)

The patient has undergone one blood transfusion because of low hemoglobin, following which the hemoglobin level rose to 9.2 mg/dl. The patient underwent high-resolution computed tomography (HRCT) temporal bone as advised in view of pain in the right ear. The scan suggested otitis media of the right ear and showed a widened diploic space (Figure *7A*). The bilateral maxillary sinuses showed hyperplasia and consequent dilatation with decreased wall thickness (Figure *7B*).

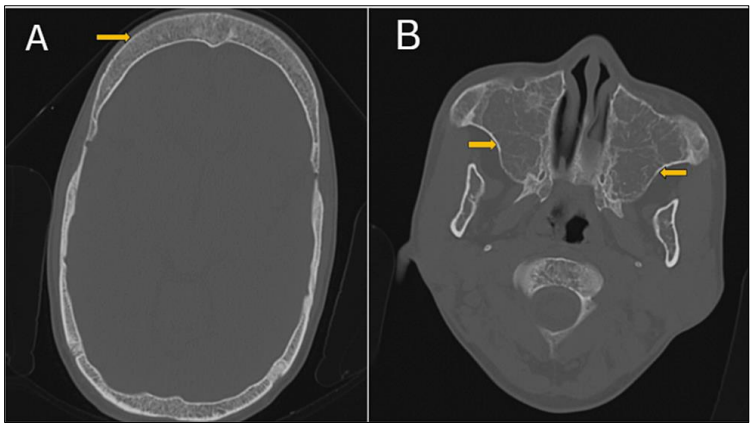

Figure 7: CT Temporal bone axial section showing (A) widened diploic space of frontal bone (arrow) and (B) Hyperplasia and consequent dilatation of bilateral maxillary sinuses with decreased wall thickness (arrows)

Following this, the patient underwent a bone marrow biopsy, which revealed eosinophilia with hemolytic features, erythroid hyperplasia, with early megaloblastic change. The findings were compatible with the diagnosis of extramedullary hematopoiesis. The patient has been advised of blood transfusions, steroids, and radiation therapy. The patient refused further stay and hence was discharged against medical advice with a prescription of medications.

DISCUSSION

Hematopoiesis refers to the generation and maturation of blood cells, a process that primarily takes place within the bone marrow of long bones, ribs, and vertebrae in adults, while in fetal life, the liver, spleen, and yolk sac serve as the main sites for this function [1]. Normally, a small population of hematopoietic stem cells (HSCs) circulates in the spleen and peripheral blood [4]. When primary hematopoietic sites in adults become ineffective, such as in certain hematologic disorders or when regular blood transfusions are lacking, the body compensates by intensifying marrow activity. This compensatory hyperactivity can lead to bone deformities and osteoporosis [1,4]. As a result, erythroid precursor cells migrate from the marrow to other tissues, giving rise to extramedullary hematopoiesis (EMH) — the formation of blood cells outside the bone marrow. This process commonly occurs in the liver, spleen, and thoracic paraspinal regions, although virtually any organ or tissue can be involved. In some cases, EMH manifests as a soft-tissue mass that may mimic a tumor [1].

Paraspinal EMH is often discovered incidentally, remaining asymptomatic in over 80% of cases. However, when symptomatic, it can cause significant discomfort due to the compression of adjacent neural structures [5]. In thalassemia, EMH frequently appears as bilateral, smooth-surfaced paraspinal soft tissue masses on imaging, often showing fat attenuation on computed tomography (CT) [6].

On CT imaging, EMH typically presents as hypovascular, heterogeneous soft tissue masses interspersed with fat density areas [1]. On ultrasound, EMH may appear as solid lesions with detectable internal blood flow. Magnetic resonance imaging (MRI) usually shows heterogeneous masses with variable signal intensity on T1- and T2-weighted images, reflecting the presence of fatty components and varying degrees of enhancement [6]. In hemoglobinopathies, paraspinal EMH may result from extrusion of marrow elements through thinned vertebral cortices. Thoracic involvement is more common than abdominal or pelvic manifestations [6]. Rib expansion, particularly pronounced in thalassemia patients, is another notable thoracic finding [1].

In patients with non-hepatosplenic EMH, about 63% present with site-specific symptoms, while the remainder are either asymptomatic or exhibit non-specific complaints such as fatigue. Apart from hepatosplenomegaly, the most characteristic imaging finding is bilateral paravertebral masses containing fat and appearing heterogeneous on scans. In such cases, biopsy is usually unnecessary [6].

EMH is rare in transfusion-dependent thalassemia major, with a reported incidence of just 1% [7]. This rarity is attributed to the suppressive effect of regular transfusions, which reduce the need for compensatory extramedullary hematopoiesis [3].

In beta-thalassemia, common radiological findings include expansion and trabecular thickening of the ribs, resulting from marrow hyperplasia affecting the entire rib. This may produce the classic “rib within a rib” appearance, particularly in the anterior and middle segments. This pattern reflects the longitudinal extension of subperiosteal red marrow within the cortical bone, leading to rib expansion as a compensatory mechanism for ineffective hematopoiesis [8, 9]. Small, well-defined medullary lucencies (1-2 mm) may also be observed. Early initiation of a hypertransfusion regimen in childhood can prevent these bony changes [8].

Patients with intermediate beta-thalassemia typically present with features between those of carriers and individuals with thalassemia major. They have moderate anemia, often requiring occasional transfusions at wider intervals compared to major thalassemia patients. These individuals may also experience complications from hemolysis and ineffective erythropoiesis, such as pain, pulmonary hypertension, and peptic ulcers [8].

This report presents a case of intermediate beta-thalassemia, where the patient demonstrated diploic space widening, rib expansion, and thoracic paraspinal masses, locations where EMH is relatively uncommon.

Radiological imaging plays a pivotal role in diagnosing EMH, although clinical history and physical examination provide important contextual information that narrows the differential diagnosis. On MRI, lesions may display significant iron deposition, especially in patients receiving regular transfusions. Inactive EMH lesions tend to accumulate iron and fat, whereas active lesions are marked by prominent neovascularization [10].

Management of EMH includes measures to suppress erythropoietin production, such as repeated blood transfusions and radiotherapy, to curb excessive hematopoietic proliferation. Surgical decompression is another therapeutic option, especially for symptomatic cases. However, the overall benefits of these treatments remain unclear due to the rarity of this condition. In thalassemia patients, maintaining adequate transfusion therapy is essential to reduce inefficient hematopoiesis and prevent extensive EMH. Careful monitoring is required to minimize complications from hypertransfusion therapy, which aims to maintain hemoglobin levels above 10 g/dL [10].

CONCLUSIONS

Although uncommon, extramedullary hematopoiesis (EMH) can develop in individuals with transfusion-dependent thalassemia (TDT), potentially resulting in notable clinical complications. In TDT patients, EMH typically manifests at an older age compared to those with non-transfusion-dependent thalassemia, with the spine being the most frequently affected site, although other organs may also be involved. Management strategies for EMH in TDT include both invasive and conservative treatments, yet comparative data on the effectiveness of these approaches remain limited. With a rising number of EMH cases reported in TDT, there is a growing need for broader investigations into optimal management practices. Clinicians should strive to maintain pre-transfusion hemoglobin (Hb) levels above 9 g/dL to reduce the risk of EMH development. Additionally, careful surveillance for EMH is essential in TDT patients, especially in those who fail to achieve the target Hb levels, to enable early diagnosis and tailored treatment plans, thereby improving clinical outcomes.

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Cite This Article: Rishabh Barman, Sachin K Shetty, Piyoosh Priyadarshee (2025). Radiological Features of Extramedullary Hematopoiesis in a Young Male with Beta-Thalassemia: A Case Report. *EAS J Radiol Imaging Technol, 7*(4), 85-90.