International Journal of Biomedicine 14(2) (2024) 345-347
http://dx.doi.org/10.21103/Article14(2)_CR3

CASE REPORT

INTERNATIONAL JOURNAL OF BIOMEDICINE

Extramedullary Hematopoiesis in a Patient with Beta Thalassemia: A Rare Case Report

Mohammed Saad Alqahtani*

Internal Medicine Department, College of Medicine, Prince Sattam bin Abdulaziz University, Al-Kharj 11942, KSA

Abstract

Extramedullary hematopoiesis (EMH) is a rare disorder, defined as the appearance of hematopoietic elements outside the bone marrow or peripheral blood due to ineffective erythropoiesis or inadequate bone marrow activity in a variety of hematological diseases. EMH often manifests as hemopoietic masses in a variety of normal and abnormal bodily sites. We present a 21-year-old man with a medical history of beta thalassemia since he was nine months old. The primary clinical symptom was mild abdominal pain. In this case, we describe a rare instance of small bowel obstruction due to EMH and portal hypertension. Surgery solved the clinical problems, and the patient was discharged home. **(International Journal of Biomedicine. 2024;14(2):345-347.)**

Keywords: thalassemia • small bowel obstruction • extramedullary hematopoiesis

For citation: Alqahtani MS. Extramedullary Hematopoiesis in a Patient with Beta Thalassemia: A Rare Case Report. International Journal of Biomedicine. 2024;14(2):345-347. doi:10.21103/Article14(2)_CR3

Introduction

Beta thalassemia is caused by an inherited mutation of the beta-globin gene and by the reduced or absent synthesis of the beta-globin chains of the hemoglobin tetramer. Three clinical and hematological conditions of increasing severity are recognized: the beta thalassemia carrier state, thalassemia intermedia, and thalassemia major.[1] Another classification of thalassemia defines it as two categories: transfusion-dependent thalassemia (TDT) and non-transfusion-dependent thalassemia (NTDT). TDT is defined as a condition where patients cannot produce adequate hemoglobin to survive without blood transfusion. NTDT is a descriptive term for patients who do not require regular lifelong transfusions. The majority of the complications of TDT are related to iron overload and bone-deforming marrow expansion with EMH.[2-8] EMH is a compensatory response to poor bone marrow function, which can result in the production of ectopic hematopoietic components outside of the bone marrow and peripheral circulation.[9]

Case Presentation

A 21-year-old man with a medical history of beta thalassemia since he was nine months old arrived at the Medical Services Center complaining of mild abdominal pain. His physical assessment was normal, and plain abdominal radiography showed incomplete small intestinal obstruction (Figure 1). After that, he was brought to the Department of Hematology for a computed tomography (CT) scan, which showed hepatomegaly and small paraspinal lobulated masses (Figure 2). The patient's abdominal pains subsided, and he was released after a five-day fast. The patient came back to the hospital complaining mainly of vomiting, distension, pain in the abdomen, and flatus for a week. His vital signs were normal upon physical examination, but there were small, palpable abdominal lumps, hyperactive bowel noises, and pallor. Hepatomegaly was also seen. After a repeat abdominal CT scan, the results showed hepatomegaly, massive ascites, and a thicker ileum wall with clear arterial phase enhancement that was obstructing flow. It was challenging to differentiate the nature of the intestinal lesion. The patient was given a nasogastric tube, blood transfusions, intravenous proton pump inhibitors, and antibiotics throughout the first ten days of hospitalization. After that, the patient was referred to the Department of Gastrointestinal Surgery, where a laparotomy and a partial enterectomy were carried out. Significant proliferation of vascular endothelial cells and blood vessel hyperplasia in the deep layers of the intestine were confirmed by a microscopic examination. Histopathological analysis

showed many myeloid and erythroid cells proliferating together with multiple megakaryocytes (Figure 3). Several inflammatory cells had invaded the intestinal wall, and there were also a lot of megakaryocytes around the serosal region, coupled with a buildup of immature myeloid and erythroid cells. Surgery solved the clinical problems, and the patient was discharged home.

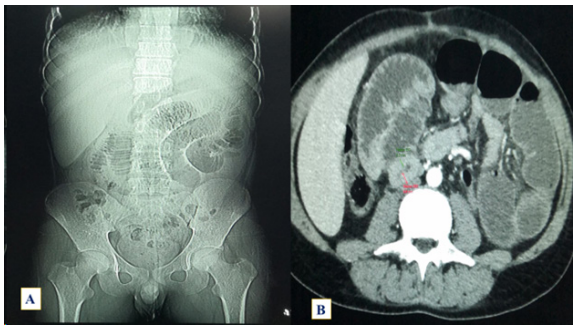

Fig. 1. *A) Abdominal radiography showing a dilated small bowel. B) CT scan: axial post IV contrasted abdominal study; a picture of the enhanced jejunal wall, with focal wall thickening and signs of pseudo intestinal obstruction.*

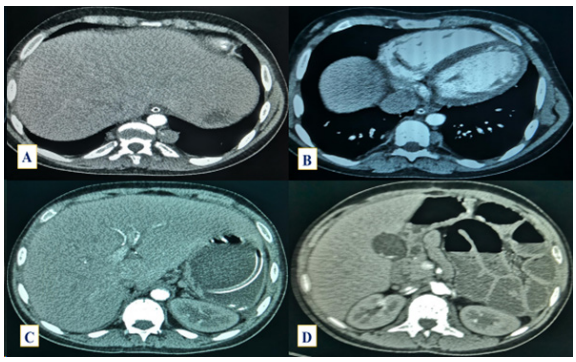

Fig. 2. *A) Axial CT abdomen shows huge hepatomegaly, left lobe focal low attenuation left paraspinal soft tissue masses.*
B) Axial contrast-enhanced lower chest mediastinal CT scan, arterial phase, nasogastric tube noted. No pericardial and pleural effusions; left paraspinal lobulated masses.
C) Abdomen CT: hepatomegaly, dilated stomach, and nasogastric tube. Normal enhanced left kidney and PV.
D) Axial contrast-enhanced CT scan, venous phase. Dilated bowel, pseudo-obstruction, GB stones, and nasogastric tube tip.

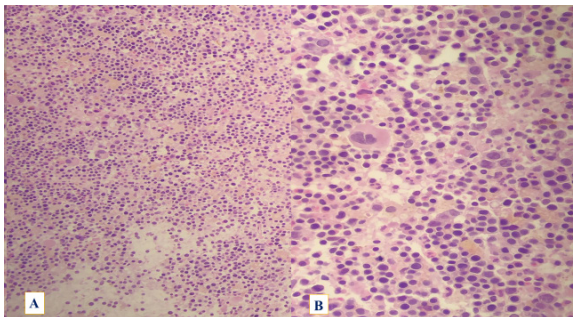

Fig. 3. *Histopathological study: the presence of EMH mass. A (X100), B (X200).*

Discussion

The clinical signs of EMH might vary widely because it can happen anywhere. There have been reports of EMH imitating acute appendicitis, intestinal obstruction, rectal stenosis, gastric outlet obstruction, and bladder outlet obstruction.[10] The reticuloendothelial system is the organ most frequently affected by EMH, but it can also affect the pleura, lungs, gastrointestinal tract, breast, skin, brain, kidneys, paraspinal tissue, and adrenal glands.[11] During the fetal stage, these areas are assumed to be involved in active hemopoiesis. Though this pathway ordinarily ends at birth, in cases of chronically inadequate erythropoiesis, the extramedullary hematopoietic vascular connective tissues continue to be able to synthesize red blood cells.[12]

The majority of EMHs are unintentionally found. The mass effect-related symptoms are specific to the afflicted spot.[13] There have been reports of EMH-related intestinal blockage, rectal stenosis, gastric outlet obstruction, and bladder outlet obstruction. The hematopoietic mass in our patient was adhered to nearby structures, which was a significant contributing factor to the symptoms. Due to an extramedullary hematopoietic mass adhering to the adjacent intestinal wall and intestinal stenosis, the patient experienced a closed-loop intestinal obstruction. Prior to surgery, the lesion was discovered via CT scanning. CT scanning can be a valuable technique for identifying gastrointestinal lesions, in addition to gastrointestinal endoscopic examinations. Masses with dense, soft sections that are typically homogeneous and have features comparable to those described by conventional radiologists can be observed on CT scans. These masses may or may not be highlighted after contrast material is administered.

The pathogenesis of this outside-bone marrow hematopoiesis is not clear. It may originate from the extension of hyperplastic marrow through the thin cortex of ribs and vertebral bodies; the capsule of the mass is formed by the periosteum.[14] Another explanation is that EMH results from transforming embryonal rests of osteogenic tissue into hematopoietic tissue under stress conditions to maintain sufficient red cell production.[15-17]

In conclusion, EMH in the small intestine is uncommon in patients with thalassemia. It is a sign of the severity of the disease and a poor prognostic factor because of small bowel obstruction. Surgery can solve the clinical problems that have arisen with EMH.

Acknowledgments

This publication was supported by the Deanship of Scientific Research at Prince Sattam bin Abdulaziz University, Al-Kharj, Saudi Arabia. We also thank all research participants.

Competing Interests

The authors declare that they have no competing interests.

References

1. Cao A, Galanello R. Beta-thalassemia. Genet Med. 2010 Feb;12(2):61-76. doi: 10.1097/GIM.0b013e3181cd68ed. PMID: 20098328.
2. Soliman A, Yassin M, Al Yafei F, Al-Naimi L, Almarri N, Sabt A, De Sanctis V. Longitudinal Study on Liver Functions in Patients with Thalassemia Major before and after Deferasirox (DFX) Therapy. Mediterr J Hematol Infect Dis. 2014 Apr 7;6(1):e2014025. doi: 10.4084/MJHID.2014.025. PMID: 24803998; PMCID: PMC4010606.
3. Yassin MA, Soliman AT, De Sanctis V, Abdelrahman MO, Aziz Bedair EM, AbdelGawad M. Effects of the anti-receptor activator of nuclear factor kappa B ligand denusomab on beta thalassemia major-induced osteoporosis. Indian J Endocrinol Metab. 2014 Jul;18(4):546-51. doi: 10.4103/2230-8210.137516. PMID: 25143915; PMCID: PMC4138914.
4. Soliman A, Yasin M, El-Awwa A, Osman M, de Sanctis V. Acute effects of blood transfusion on pituitary gonadal axis and sperm parameters in adolescents and young men with thalassemia major: a pilot study. Fertil Steril. 2012 Sep;98(3):638-43. doi: 10.1016/j.fertnstert.2012.05.047. Epub 2012 Jun 29. PMID: 22749224.
5. Soliman AT, Yassin M, Majuid NM, Sabt A, Abdulrahman MO, De Sanctis V. Cortisol response to low dose versus standard dose (back-to-back) adrenocorticotrophic stimulation tests in children and young adults with thalassemia major. Indian J Endocrinol Metab. 2013 Nov;17(6):1046-52. doi: 10.4103/2230-8210.122620. PMID: 24381882; PMCID: PMC3872683.
6. De Sanctis V, Soliman A, Candini G, Campisi S, Anastasi S, Iassin M. High prevalence of central hypothyroidism in adult patients with β-thalassemia major. Georgian Med News. 2013 Sep;(222):88-94. PMID: 24099820.
7. De Sanctis V, Elsedfy H, Soliman AT, Elhakim IZ, Pepe A, Kattamis C, Soliman NA, Elalaily R, El Kholy M, Yassin M. Acquired Hypogonadotropic Hypogonadism (AHH) in Thalassaemia Major Patients: An Underdiagnosed Condition? Mediterr J Hematol Infect Dis. 2016 Jan 1;8(1):e2016001. doi: 10.4084/MJHID.2016.001. PMID: 26740862; PMCID: PMC4696472.
8. Bukhari SS, Junaid M, Rashid MU. Thalassemia, extramedullary hematopoiesis, and spinal cord compression: A case report. Surg Neurol Int. 2016 Mar 2;7(Suppl 5):S148-52. doi: 10.4103/2152-7806.177891. PMID: 27069747; PMCID: PMC4802988.
9. Chiam QL, Lau KK. Extramedullary haematopoiesis in thalassaemia major causing spinal cord compression. Australas Radiol. 2007 Apr;51(2):168-71. doi: 10.1111/j.1440-1673.2007.01689.x. PMID: 17419864.
10. Elpek GO, Bozova S, Erdoğan G, Temizkan K, Oğüş M. Extramedullary hematopoiesis mimicking acute appendicitis: a rare complication of idiopathic myelofibrosis. Virchows Arch. 2006 Aug;449(2):258-61. doi: 10.1007/s00428-006-0230-5. PMID: 16738896.
11. Zhu G, Wu X, Zhang X, Wu M, Zeng Q, Li X. Clinical and imaging findings in thalassemia patients with extramedullary hematopoiesis. Clin Imaging. 2012 Sep-Oct;36(5):475-82. doi: 10.1016/j.clinimag.2011.11.019. Epub 2012 Jun 8. PMID: 22920349.
12. Haidar R, Mhaidli H, Taher AT. Paraspinal extramedullary hematopoiesis in patients with thalassemia intermedia. Eur Spine J. 2010 Jun;19(6):871-8. doi: 10.1007/s00586-010-1357-2. Epub 2010 Mar 5. PMID: 20204423; PMCID: PMC2899982.
13. Keikhaei B, Zandian K, Rahim F. Existence of cord compression in extramedullary hematopoiesis due to beta thalassemia intermedia. Hematology. 2008 Jun;13(3):183-6. doi: 10.1179/102453308X343347. PMID: 18702878.
14. Abdulla MAJ, Yassin MA, Abdelrazek M, Mudawi D, Ibrahim F, Soliman DS, ElOmri H, Nashwan AJ, Fernyhough LJ, De Sanctis V, Soliman AT. A persistent cough as atypical clinical presentation of intrathoracic extramedullary hematopoiesis (EMH) in a female with thalassemia intermedia. Acta Biomed. 2018 Feb 16;89(2-S):41-46. doi: 10.23750/abm.v89i2-S.7086. PMID: 29451228; PMCID: PMC6179037.
15. Bolaman Z, Polatli M, Cildag O, Kadiköylü G, Culhaci N. Intrathoracic extramedullary hematopoiesis resembling posterior mediastinal tumor. Am J Med. 2002 Jun 15;112(9):739-41. doi: 10.1016/s0002-9343(02)01128-2. PMID: 12079720.
16. Pornsuriyasak P, Suwatanapongched T, Wangsuppasawad N, Ngodngamthaweesuk M, Angchaisuksiri P. Massive hemothorax in a beta-thalassemic patient due to spontaneous rupture of extramedullary hematopoietic masses: diagnosis and successful treatment. Respir Care. 2006 Mar;51(3):272-6. PMID: 16533417.
17. Smith PR, Manjoney DL, Teitcher JB, Choi KN, Braverman AS. Massive hemothorax due to intrathoracic extramedullary hematopoiesis in a patient with thalassemia intermedia. Chest. 1988 Sep;94(3):658-60. doi: 10.1378/chest.94.3.658. PMID: 3409758.

***Correspondence**: Mohammed Saad Alqahtani. E-mail: Ms.alqahtani@psau.edu.sa*