A presacral mass in a patient with thalassemia intermedia: A case report and review of the literature

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A presacral mass in a patient with thalassemia intermedia: A case report and review of the literature

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ABSTRACT

Extramedullary hematopoiesis (EMH) is defined as the production of the myeloid and erythroid elements outside the bone marrow. It is usually a compensatory mechanism of the myeloid and erythroid production due to increased breakdown or diminished production of erythrocytes. Presacral EMH is an extremely rare condition, and there is a limited number of case reports published in the literature. A 39-year-old female patient as a known case of thalassemia intermedia presented with lower abdominal pain. A computerized tomography scan showed a large presacral mass, associated with bone destruction. The patient was admitted for exploratory laparotomy with suspicion to malignant lesions, but the final pathological diagnosis was EMH. It should be considered in differential diagnosis of mass-like lesions in the presacral area in patients with predisposing factors such as thalassemia, although there were malignant features such as bone destruction in imaging studies. Preoperative diagnostic tools such as fine needle aspiration and biopsy could help us to render the definite diagnosis and prevent unnecessary operation.

Keywords: Extramedullary hematopoiesis, presacral, thalassemia intermedia

Introduction

Extramedullary hematopoiesis (EMH) is defined as an abnormal growth of hematopoietic tissue outside the bone marrow. It is usually a compensatory mechanism of the myeloid and erythroid production due to increased breakdown or diminished production of the erythrocytes, such as in myelofibrosis, thalassemia, and sickle cell anemia. It has been reported in almost all organs in the body and may also manifest as a tumor-like mass. Presacral location of EMH is extremely rare, and only a limited number of cases have been described in the literature.[1-5] We report one such rare case in a patient with thalassemia intermedia which was misdiagnosed as malignant lesion in imaging studies, but the final pathological diagnosis was EMH after surgery.

Case Report

A 39-year-old female patient, a known case of thalassemia intermedia, presented at the outpatient department with abdominal pain. Her previous medical history included cholecystectomy due to symptomatic gallstone, traumatic splenectomy, and two times cesarean sections. In physical examination, a mass was identified in the pelvis that could be palpated vaginally. The laboratory findings were within normal limits except for hemoglobin (Hb) 8.1 g/deciliter and MCV 67 femtoliters. Abdominal and pelvic ultrasonography showed a large presacral mass with bone destruction. The patient was admitted for exploratory laparotomy with suspicion to malignant lesions, but the final pathological diagnosis was EMH after surgery.

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revealed a 70 mm × 50 mm pelvic mass. A computerized tomography (CT) scan showed a large presacral soft tissue mass lesion (70 mm × 65 mm × 60 mm), associated with the bone destruction and causing anterior displacement of the uterus [Figure 1]. According to the radiologic image, it was assumed that the mass could be a malignant lesion, such as plasmacytoma or lymphoma, and no preoperative tissue diagnosis was performed. Exploratory laparotomy and debulking of the presacral mass was performed. The mass was lobulated with the involvement of the underlying bone. The mass was removed completely and sent for further histological examination. Grossly, it consisted of an ill-defined red brown, friable mass resembling an organized hematoma [Figure 2]. Microscopic studies revealed hypercellular sections containing a polymorphic population of mature hematopoietic cells (myeloid, lymphoid, erythroid, and megakaryocytic lines). There is no mitosis or necrosis. Scattered hemosiderin laden macrophages are present [Figure 3]. The final pathological diagnosis was EMH. After operation, she received no medical therapy or blood transfusion, and she is well with no recurrence 2 years postoperatively.

**Discussion**

EMH is defined as the production of the myeloid and erythroid elements outside the bone marrow. It is usually a mechanism to compensate for hemolytic anemia, such as spherocytosis or thalassemia, or as a reaction to abnormal bone marrow function seen in disorders, such as myelofibrosis or leukemia. Normally, EMH tends to be microscopic, but it sometimes manifests as organomegaly involving the spleen, the liver or lymph nodes. It has been reported in almost all organs in the body and may also present as a tumor-like mass. The paravertebral region is an unusual site for EMH, with the possible exception of sporadically reported intrathoracic presentations. However, EMH in the presacral area is rare, with about 25 cases reported in the English literature, to the best of our knowledge; few of them were presented in thalassemic patients as in our case. Extrusion of proliferating marrow through the weakened bone cortex, because of stimulated erythropoietin production, illustrates its presence. One hypothesis by Forster and Schob that an EMH in the presacral region may be caused by previously sacral trauma that causes hematopoietic cells to plant in the presacral area. An alternative hypothesis by Macki et al. recently suggests that manipulation of the uterine tissue liberates the mesenchymal stem cells, and these cells differentiate into the stromal tissue that interacts with multipotent hematopoietic stem cells to form an EMH mass. It is usually asymptomatic, and the majority of the reported cases were discovered incidentally. The rest had variable presentations including pelvic pain, pelvic mass, constipation, symptoms of anemia, and rare symptoms of cord compression.
CT scan and magnetic resonance imaging (MRI) are preferred as the common initial investigations. EMH usually appears as a heterogeneous lobulated solid mass with smooth margins. Its density is similar to that of the soft tissue, slightly denser than fluid. According to CT imaging, a differential diagnosis is lymphoma, which tends to produce a pattern of retroperitoneal involvement of the lymph nodes surrounding the great vessels which are suspected in our case. An MRI of presacral EMH often shows a characteristic well encapsulated tumor with somewhat higher signal intensity than the muscle on T1 and T2-weighted imaging, consistent with fatty tissue. The mass gets uniformly enhanced after gadolinium injection.[1,3,4]

A tissue sample is usually considered mandatory to proper diagnosis with any mass. The role of preoperative fine-needle aspiration or biopsy is challenging. The presacral location presents a technical problem to obtaining such a sample. Potential complications associated with an aspiration or biopsy of this region contains infection and bleeding, particularly resulting from injury to the middle sacral artery. [1,4] Therefore, Wolpert et al. advised that preoperative biopsy be done only in solid lesions with signs of malignancy, such as sacral invasion.[6] Examination of an aspiration biopsy consistent with EMH reveals hematopoietic cells with polymorphous infiltrates including megakaryocytes and lymphocytes, which Kumar et al. are describing completely in their paper.[10] Grossly, EMH often present as soft red-brown mass-like lesion resembling hematoma on the sections which were cut. Histologically, it is composed of the polymorphic population of normal hematopoietic cells in all lineages and different maturation stages.[5] Tissue biopsy samples may be misdiagnosed when atypical megakaryocytes are misinterpreted as malignant cells, which occurred in the Denies et al.’s case.[2] Misdiagnosis can occur even more often when EMH is not considered in the differential diagnosis due to its rare occurrence. Our patient like some other cases on the record did not undergo a biopsy and diagnosis was made by histopathological studies on the tissue submitted postoperatively.

The management of EMH depends on symptoms and underlying disease. In asymptomatic patients with underlying diseases such as thalassemia, myelofibrosis, and sickle cell anemia, combined treatment with a blood transfusion and iron chelation is an ideal treatment because it relieves anemia and suppresses EMH.[4,11] Furthermore, the use of drugs such as hydroxyurea which increases Hb F production could be helpful.[11] In symptomatic patients, radiation therapy and surgery are recommended according to the severity of symptoms. Hematopoietic tissue is highly sensitive to radiation, so radiotherapy can be used for relieving of mild neurological symptoms in 3-7 days. For immediate relief of spinal cord compression, surgery is advised. The complications of surgery include mass bleeding from the surgical site and incomplete excision.[4,7,11] In our case, the management was influenced by the symptoms and unusual site of growth in the presacral space, and the malignant features of the mass including bone destruction in imaging studies.

**Conclusion**

Presacral EMH is very rare, and it is a difficult diagnostic challenge in this situation. We should consider EMH in differential diagnosis of mass-like lesions in the paravertebral area (presacral, retroperitoneum and posterior mediastinum) in patients with predisposing factors as mentioned earlier, although there were malignant features such as bone destruction in imaging studies. Preoperative diagnostic tools such as fine-needle aspiration and biopsy could help us to render the definite diagnosis and prevent unnecessary operation.

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**Conflicts of interest**

There are no conflicts of interest.

**References**