A Case Report of Extramedullary Hematopoiesis (EMH) in a Retroperitoneal Mass in a Non Transfusion-Dependent Beta-Thalassemia Patient

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HIGHLIGHTS

- Rare case of retroperitoneal extramedullary hematopoiesis (EMH) in a patient with non–transfusion-dependent thalassemia (NTDT)
- Extramedullary hematopoiesis (EMH) can mimic retroperitoneal malignancy
- Diagnosis confirmed with biopsy after inconclusive imaging
- Managed with embolization and supportive care
- Early recognition avoids unnecessary surgery and complications

BACKGROUND

We report an unusual case of extramedullary hematopoiesis (EMH) in a patient with non-transfusion-dependent β-thalassemia (NTDT) who presented with a retroperitoneal mass, underscoring the need to include EMH in the differential diagnosis of retroperitoneal lesions, particularly in individuals with underlying hematologic disorders.

CASE PRESENTATION

- A middle-aged, unmarried, nulliparous Bahraini female, known to have osteoporosis and NTDT.
- Upon focused examination, the patient was vitally stable, alert, conscious, and oriented to time, place, and person. On general inspection, she was pale-looking with no signs of jaundice.
- On abdominal examination, marked hepatosplenomegaly was noted.
- The rest of the examination was unremarkable.
- MRI revealed a large, solid retroperitoneal mass as an incidental finding.
- Tumor marker blood analysis, which showed a raised CA 15-3 level.
- Lieomyosarcoma was suspected
- Excision of the mass along with total abdominal hysterectomy (TAH) with bilateral salpingo-oophorectomy (BSO) was the plan.
- Intraoperatively, the uterus, ovaries, and fallopian tubes appeared normal, but a significantly bleeding retroperitoneal cystic mass, resembling an ovarian cyst, was discovered.
- Upon this finding, the procedure was abandoned, a biopsy was taken, and hemostasis was achieved.
- Subsequently, the procedure was followed by embolization of the mass.
- Biopsy led to EMH diagnosis.
- Six months later, the patient presented to our emergency department with fatigue and poor concentration.
- Blood hemoglobin level was 5.4 mg/dl.
- She was admitted under hematology for three months during which continuous investigation, frequent blood transfusions and various medications took place to stop the decline in hemoglobin levels.
- Splenic artery embolization under ultrasound guidance was used as an alternative for surgery to control the most probable cause which is splenomegaly.

INVESTIGATIONS

- Initial non-contrast MRI: fat-containing retroperitoneal mass (16.5 × 11 × 15.5 cm) with iliac/presacral nodules.
- Labs: persistent anemia, elevated CA 15-3, B12 deficiency; other values normal.
- Pelvic USG: large heterogeneous mass with cystic areas and peripheral vascularity (Figure 1).
- Intraoperative biopsy: EMH with trilineage maturation; IHC CD20/CD3/CD68/FVII+, CD34–; no malignancy → leiomyosarcoma, ovarian origin, and other anemia etiologies ruled out.
- Post-op CT: residual hemorrhagic mass (10.8 × 9.9 × 11 cm).
- At 6 months: pelvic mass stable, but new enlarging paravertebral, retroperitoneal, and presacral nodules; hepatic hyperdensity indicating progressive EMH with hemosiderosis.

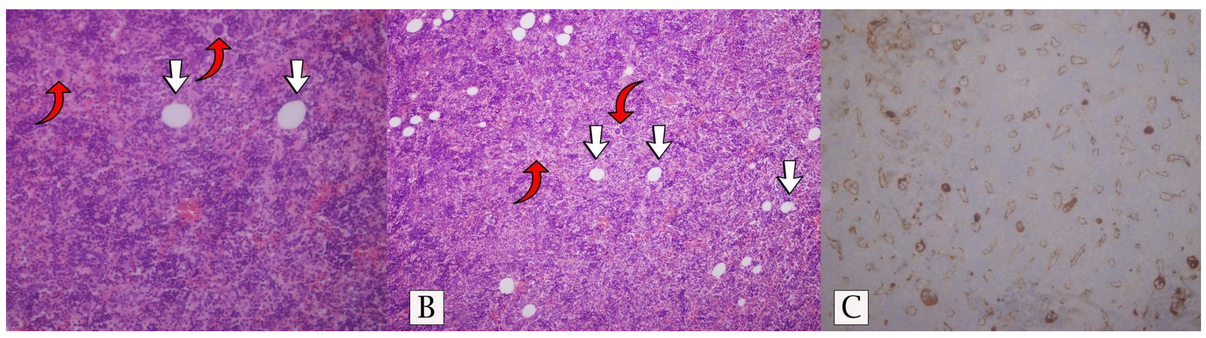

Figure 2: A and B- H&E-stained slides (200 and 400X) showing a mixture of extramedullary hematopoietic cells with full maturation and with many megakaryocytes and scattered mature adipocytes. C- Factor-VII stained megakaryocytes.

TREATMENT

- The mass was embolized and an intraoperative biopsy was taken.
- Post-operative anemia management at ICU with regular transfusions and medical therapy.
- Splenic artery embolization was performed as a non-surgical alternative to control hypersplenism.

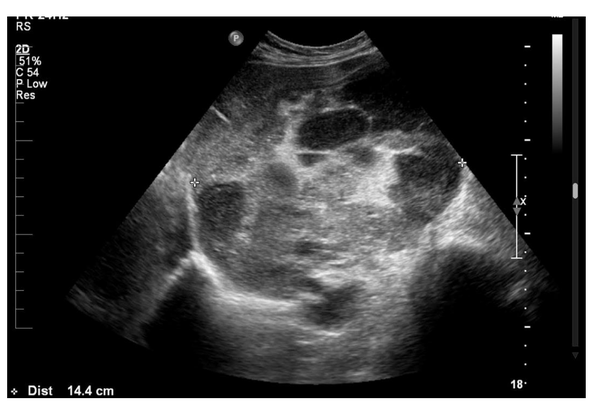

FIGURE 1: Pelvic sonography performed in May 2023 revealing a large hyperechoic heterogenous mass measuring 14.1 x 11.2 x 14.4 cm occupying the mid pelvic-abdominal region with multiple areas of cystic changes and peripheral vascularity.

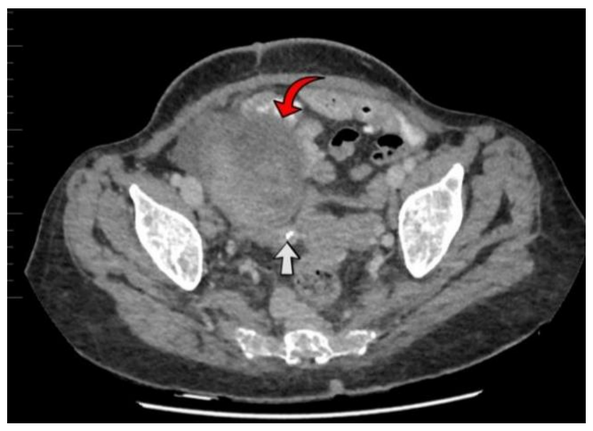

FIGURE 2: CT scan of the abdomen and pelvis performed postoperatively shows pelvis retroperitoneal mass related to EMH. It is seen in the midland towards the right side of the pelvis. It is predominantly hyperdense with areas of fat density and focus of calcification. It measures 10 x 9 x 11 cm (AP x ML x CC dimension)

DISCUSSION

Extramedullary hematopoiesis (EMH) is more common in NTDT, with incidence up to 20% (1,2) and is often incidentally discovered because patients are usually asymptomatic (3). Diagnosis can be approached with multiple modalities, but biopsy is the confirmatory test (4); imaging findings are nonspecific yet supportive for EMH (2,5). Clear treatment guidelines remain limited (4,5). Splenomegaly is a common complication in NTDT. Because EMH masses help maintain hemoglobin, surgical resection may precipitate clinical decompensation, making accurate preoperative diagnosis essential to avoid unnecessary surgery and improve outcomes (6,7).

CONCLUSION

Retroperitoneal EMH is rare and can mimic malignancy, so clinicians should keep a high index of suspicion in patients with hemoglobinopathies. Biopsy confirms the diagnosis, and early recognition helps avoid unnecessary surgery and improves outcomes.

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