Presacral extramedullary haematopoiesis: A diagnostic update and case report of a late diagnosis

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ABSTRACT

INTRODUCTION: We report a rare case of presacral extramedullary haematopoiesis, which manifested as a tumoural mass on a routine ultrasonography in a patient presenting with symptoms of cholecystitis. Since Ask-Upmark in 1945 reported 3 cases of pelvic extramedullary haematopoiesis, we are aware of only published 14 additional cases.

PRESENTATION OF CASE: A 73-year-old female patient presented with abdominal pain in the right hypochondrium. An abdominal ultrasonography revealed cholecystitis with cholecystolithiasis and a coincidental hyperreflective mass of 9.5 cm was visualised behind the bladder. A clinical examination identified a mass in the pelvis that could be palpated vaginally. A computerised tomography scan showed a large presacral, inhomogeneous, multilobular and nodular tumour. The patient was admitted for laparoscopic resection of the gall bladder and laparoscopic exploration of the presacral mass. An anatomopathological examination of the tissue revealed the presence of extramedullary haematopoietic tissue. A postoperative haematological investigation indicated that the extramedullary haematopoiesis was idiopathic.

DISCUSSION: Presacral EMH may occasionally present with symptoms of nerve compression. Symptoms of haematologic disorders may accompany EMH.

Barium enema, abdominal ultrasound, CT scan, MRI and radionuclide bone marrow imaging have all been used by previous authors in establishing the diagnosis. Tissue samples may be misdiagnosed when atypical megakaryocytes are misinterpreted as malignant cells, which occurred in this case. Misdiagnosis can occur even more often when EMH is not considered in the differential diagnosis due to its rare occurrence. In this case, the final diagnosis was made tissue sampling after surgery.

Treatment of EMH is only necessary when it is symptomatic.

CONCLUSION: This case report shows that extramedullary haematopoiesis is very rare and that it is a difficult diagnostic challenge when its location is unusual and when it is not associated with a haematologic disorder. Together with this case report, we present an update of the available diagnostic methods.

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1. Introduction

Extramedullary haematopoiesis (EMH) consists of the presence of ectopic haematopoietic elements outside the bone marrow and peripheral blood. It is a compensatory mechanism in several haematologic diseases when erythrocyte production is compromised or destruction is accelerated. Normal, EMH tends to be microscopic, but it occasionally manifests as organomegaly involving the spleen, the liver or lymph nodes. It may also manifest as a tumour-like mass. The paravertebral area is an uncommon site for EMH, with the possible exception of sporadically reported intrathoracic manifestations. Presacral presentation is extremely rare. Extrusion of proliferating marrow through the weakened bone cortex, due to stimulated erythropoietin production, explains its presence. One hypothesis by Forster and Schob suggests that an EMH in the presacral region may be caused by earlier sacral trauma that induces haematopoietic cells to seed the presacral space. Since the ASK-Upmark reported 3 cases of pelvic EMH in 1945, we are aware of 14 published cases.

2. Case report

A 73-year-old female patient presented at the emergency department with abdominal pain.

Her previous medical history included arterial hypertension, lower back surgery and a vaginal hysterectomy with anterior and posterior colphoraphia for a prolapse. An abdominal ultrasonography (US) revealed cholecystitis with cholecystolithiasis, and a coincidental 9.5 cm hyperreflective mass was visualised behind the bladder. A clinical examination identified a mass in the pelvis that could be palpated vaginally. The laboratory findings were

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unremarkable with a CA125 tumour marker concentrate of 4 kU/L (normal value <24 kU/L). Computerised tomography scan (CT) was performed to assess the mass. This revealed a large presacral, inhomogeneous, multilobular and nodular tumour (Fig. 1). The patient was admitted for laparoscopic resection of the gall bladder and laparoscopic exploration of the presacral mass.

It was assumed that the mass was a liposarcoma, and no pre-operative tissue diagnosis was performed because the laboratory findings were normal and a cholecystectomy was indicated. As such, a tissue sample could be taken during surgery. After the cholecystectomy, it became obvious that the mass was retroperitoneal. The peritoneum was opened, and the tumour was visualised. Whilst manipulating the tumour, it showed a great tendency to bleed at the presacral plexus. As such the procedure was converted to an open laparotomy. A tissue sample was sent for intraoperative histological cryosection examination and was diagnosed as a malignant tumour because it showed atypical megakaryocytes. The mass was removed incompletely and sent for further histological examination. This revealed myelopoietic cells surrounded by fatty tissue, consistent with EMH (Fig. 2). A postoperative haematological investigation showed that the EMH was idiopathic. Thalassemia, leukaemia, myeloproliferative diseases and chronic haemolytic diseases were not discovered.

3. Discussion

Our patient presented with asymptomatic EMH. A review of our patient’s medical history and further haematologic testing revealed no haematologic disorder explaining the development of EMH. Presacral EMH may occasionally present with symptoms of medullary/cauda equina compression and/or sciatic nerve pain.7 Symptoms of the earlier mentioned haematologic disorders, such as signs of anaemia and hepatosplenomegaly, may accompany EMH.

However a plain radiograph is unable to show an EMH. Two published cases in which a barium enema was used revealed a mass effect of the tumour on the rectum.8,9

An abdominal ultrasound revealed a solid soft tissue mass as described by other authors.10

The CT scan of our patient showed an inhomogeneous, multilobular and nodular tumour. Using CT imaging without intravenous contrast, authors report attenuation values of EH between 20 and 55 HU.7,9 In some cases, as in ours, the EMH may have a fatty component with negative Hounsfield values, but calcifications are always absent.6 The fatty components in this case suggested to the radiologist that the tumour was a liposarcoma.

Ruiz Carazo et al. describe that the signal intensity of the lesions was discretely higher than that of muscle on T1-weighted images and hyperintense on T2-weighted images.7 This behaviour is different from the one described by Mesurolle et al. who observed a low signal on T2-weighted images.11 A diffuse enhancement is described when administering gadolinium.7,10,12–14 Al-Aabassi and Murad use MRI as the technique of choice to diagnose EMH.10 However, MRI has been known to misinterpret EMH as a neurogenic tumour.7

The use of radionuclide bone marrow imaging could be helpful in establishing the diagnosis by demonstrating activity in the mass as observed in previously reported cases.9

In histological sampling, all haematopoietic elements found are myeloid cells and erythroid cells at various stages of maturation with the presence of megakaryocytes. Macroscopically, an EMH shows similarity with a haematoma.15 Tissue samples may be misdiagnosed when atypical megakaryocytes are misinterpreted as malignant cells, which occurred in this case. Misdiagnosis can occur even more often when EMH is not considered in the differential
diagnosis due to its rare occurrence.\textsuperscript{10} The differential diagnosis for presacral masses includes cystic or solid lesions, with the majority of solid lesions being neoplasms.

In all but one previous case described by Gupta et al. and in this case, the diagnosis was made using a fine needle aspiration/biopsy or tissue sampling after surgery. Gupta et al. concluded that in the appropriate clinical setting, when the appearance of the mass is typical, a biopsy is not necessary for diagnosis.\textsuperscript{8} Making the diagnosis based solely on the clinical background of the patient and imaging techniques is possible, but one must feel very confident with the diagnosis because malignancy must be excluded. This can be performed with greater accuracy after acquiring tissue samples by biopsy or aspiration.

Treatment of EMH is only necessary when it is symptomatic. EMH responds well to radiotherapy and can be treated with 1500–3000 cGy in a divided dosage. Authors describe rapid response with this regime.\textsuperscript{6,10,13} In patients with spinal cord compression, neurological improvement has been achieved within 3–7 days of treatment initiation.\textsuperscript{7} Surgical treatment is contraindicated in EMH, but it may be useful for immediate symptomatic relief. The complications of surgery include mass bleeding from the surgical site and incomplete excision, which were both observed in this case. EMH surgery often consists of operating on an anaemic patient, which is always a suboptimal condition. Combined treatment with a blood transfusion and iron chelation is an ideal treatment for asymptomatic individuals because it relieves anaemia and suppresses EMH and has been shown to be very effective when used in conjunction with surgery or radiotherapy.\textsuperscript{2–15}

Relieving the anaemic stress by transfusion therapy has been used to cause regression of haematopoietic tissue.\textsuperscript{14} Stimulating foetal haemoglobin production using hydroxyurea alone or in conjunction with transfusion therapy has also been applied with some success.\textsuperscript{2,15}

### 4. Conclusion

This case report shows that extramedullary haemopoiesis is very rare and that it is a difficult diagnostic challenge when its location is unusual and when it is not associated with a haematologic disorder.

### Conflict of interest

The authors declare that they have no competing interests.

### Funding

None.

### Ethical approval

Written informed consent was obtained from the patient for publication of this case report and the accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal upon request.

### Authors’ contributions

ED wrote the case report, analysed and interpreted the patient data regarding haematological disease and reviewed the literature on the subject. VD performed the histological examination of the tissue. PD was a major contributor in guiding the search for literature on the subject and writing the manuscript. All authors read and approved the final manuscript.

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