CASE REPORT

An unusual relapse in acute myeloid leukaemia

Feras Abu Saadeh,¹ Vicki Collins,¹ Maimuna Al-Saadi,² Noreen Gleeson¹¹Department of Gynaecology, St. James's Hospital, Dublin, Ireland²Department of Pathology, St. James's Hospital, Dublin, Ireland

Correspondence to

Dr Feras Abu Saadeh, ferasabusaadeh@dubgyn.org

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SUMMARY

A 40-year-old nulliparous woman, with a history of acute myeloid leukaemia (AML), presented at a gynaecological clinic with an incidental finding of a 5 cm pelvic mass on ultrasound during workup for subfertility. Biopsies confirmed a myeloid sarcoma. The patient underwent a total abdominal hysterectomy and bilateral salpingo-oophorectomy, followed by chemotherapy and radiotherapy. She recovered well from her surgery, 21 months postsurgery with no evidence of recurrence.

BACKGROUND

Myeloid sarcomas (MS) of the gynaecological tract are rare, but should be considered as a differential diagnosis of mass lesions found in a patient with a history of acute myeloid leukaemia (AML) or other myelodysplastic syndrome, even after many years following their primary therapy.

CASE PRESENTATION

A 40-year-old, nulliparous woman presented to our service early in 2013 with an incidental finding of a 5 cm pelvic mass on ultrasound, while undergoing investigations for subfertility. She had a history of AML, diagnosed in 2002, and was treated with chemotherapy followed by a matched sibling bone marrow transplant in the same year. She was known to our service having previously undergone mid-vaginal and lower vaginal adhesiolysis in 2011 and was currently taking a high-dose oestrogen as part of the assisted reproduction programme. She had been keeping well from 2003 until now. The patient was asymptomatic with respect to this mass. Examination was unremarkable.

INVESTIGATIONS

Full blood count, inflammatory markers and CA-125 were normal. MRI of the pelvis revealed a bulky left-sided cervical mass ~4 cm in diameter that appeared homogenous (figure 1). Positron-emission tomography CT showed a fluorescent deoxyglucose (FDG)-avid left lateral cervical mass with SUVmax 5.8, no FDG-avid pelvic/retroperitoneal lymphadenopathy and no FDG-avid distal metastases (figure 2). Examination under anaesthetic, cystoscopy, hysteroscopy, dilatation & curettage were performed. This showed a normal bladder. The cervix rim was just visible at the narrowed vaginal vault (previous vaginolysis). Within the uterine cavity was a solid tumour pushing to the left parametria. Biopsies showed myeloid sarcoma and thus represented an extramedullary relapse of AML.

DIFFERENTIAL DIAGNOSIS

Differential diagnosis included carcinoma, epithelioid sarcomas, lymphoma and inflammatory lesions.

TREATMENT

She underwent total abdominal hysterectomy, bilateral salpingo-oophorectomy with left partial parametrectomy and sampling pelvic lymphadenectomy to the level of the common iliacs (figure 3).

OUTCOME AND FOLLOW-UP

Postoperative histology confirmed extramedullary AML (myeloid sarcoma), involving the cervix and extending into the lower uterine segment (figure 4). Flow cytometry demonstrates a population (35%) of blast cells, which express CD117, CD13, CD45,

Figure 1 MRI of the pelvis (A) sagittal view and (B) transverse view, showing large haemogenous pelvic mass around 5 cm in diameter.

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Figure 2 Positron-emission tomography CT scan (A) coronal view and (B) sagittal view, fluorescent deoxyglucose-avid cervical mass with SUVmax 5.8, no distant metastatic disease.

human leucocyte antigen-DR, TdT, CD34, CD33 and CD38. These features are consistent with extramedullary AML. Cytogenetics analysis with interphase fluorescent in situ hybridisation analysis with an MLL (11q23) probe set detected no evidence of either rearrangement or numerical aberration involving neoplastic cells. The myometrium, fallopian tubes and ovaries were not involved. Four lymph nodes were positive. Following discussions at the multidisciplinary meeting, she was referred back to haematology and has since undergone chemotherapy, radiotherapy and donor lymphocyte infusions. As of August 2014, she completely recovered from this treatment and is seen

regularly in the haematology day ward with no evidence of recurrence.

DISCUSSION

MS are extramedullary lesions composed of myeloblasts or immature myeloid cells. Most often they present in association with acute leukaemias, most notably AML.¹ They may be detected simultaneously with first presentation of disease, during the course of disease or at relapse (as in our case). They can occur at any anatomical site, but most commonly involve the skin and soft tissues, lymph nodes and gastrointestinal tract.

Figure 3 The pathological specimen after surgical excision. The cervix is expanded with the tumour.

Figure 4 Histopathological diagnosis. (A) H&E stain showing atypical medium-sized blast cells. Scant residual normal cervical tissue present. (B) Immunohistochemistry with CD117, CD34 and CD45.

MS involving the female reproductive tract are rarely encountered.^{2 3} Diagnosis of MS in the presence of haematological

malignancy is relatively straight forward, but that of primary MS can present a challenge for the pathologist. It is most frequently misdiagnosed as large B-cell lymphoma.⁴ When the diagnosis of MS is considered, cytochemical and immunohistochemical studies can reliably make the diagnosis in the majority of cases.⁴ Optimal treatment of these patients is not clear due to the relatively low number of cases and a lack of large prospective studies. Chemotherapy regimens, similar to those used in AML remission induction, are often employed $\pm$ surgery or radiotherapy, on a case-by-case basis.¹

Learning points

- Myeloid sarcomas should be considered in the differential of patients presenting with a pelvic mass, with a history of myeloproliferative disorder.
- Myeloid sarcomas can present at any stage of disease, from the first presentation to relapse many years after primary therapy.
- Optimal treatment is unclear, but may include chemotherapy $\pm$ surgery $\pm$ radiotherapy.

Competing interests None.

Patient consent Obtained.

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