Proptosis and ovarian masses: extramedullary manifestations of acute myeloid leukaemia

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DESCRIPTION

A 33-year-old woman presented with back and pelvic pain of 3-week duration. Associated symptoms included diplopia and fatigue. Physical examination revealed proptosis, gum hypertrophy and abdominal tenderness. Her white blood cell count was $61 \times 10^9/L$ with 52% blasts, haemoglobin 102g/L, and platelets $178 \times 10^9/L$. CT of abdomen and pelvis revealed bilateral solid adnexal masses measuring up to 10 cm. MRI of the pelvis showed rectal thickening, the solid ovarian masses and pathologic perineal and inguinal adenopathy. MRI orbits noted thickening and infiltration of orbital muscles (figure 1).

Her bone marrow revealed 90% involvement by acute myeloid leukaemia (AML) with monocytic differentiation (figure 2).

Biopsy of inguinal lymph node was also consistent with AML. Ovarian masses, inguinal lymph, nodes rectal thickening and orbital muscle infiltration were deemed to be extramedullary manifestations of AML. The patient received induction chemotherapy with cytarabine and daunorubicin with resolution of proptosis and clearance of leukaemic blasts in peripheral smear in 2 days. A follow-up CT of the abdomen and pelvis revealed a decrease in size of the ovarian masses.

AML is a haematopoietic neoplasm that results in clonal proliferation of myeloid precursors.¹ These cells proliferate with a reduced capacity to differentiate and replace normal bone marrow elements causing pancytopenia.² AML is the most common adult acute leukaemia with median age of diagnosis between 62 and 71 years.¹ The clinical presentation

Figure 2 Bone marrow biopsy, H&E stain, revealing >90% involvement by blasts.

usually involves the complications related to pancytopenia and includes weakness, fatigability, infections and haemorrhagic findings such as gingival bleeding, ecchymosis, epistaxis or menorrhagia.² Patients with extramedullary disease (EMD) may also have symptoms related to organ dysfunction due to tumour involvement.³

EMD can occur in the presence or absence of bone marrow involvement. The two predominant forms of EMD are myeloid sarcoma (MS) and leukaemia cutis. Leukaemia cutis describes leukaemic invasion of the integument, which often results in subcutaneous nodules. MS, which was present in this patient, most commonly involves bone, periosteum, soft tissues and lymph nodes.⁴ Rarely, with data being limited to case series, ocular and ovarian disease involvement by MS has been reported. The present case is unique from these due to clinical AML involvement of the multiple sites including the ovaries, rectum, gingiva and ocular muscles.^{5,6}

MS involves 2.5%–9.1% of patients with AML and its diagnosis requires a high index of suspicion. Even in patients with an established diagnosis of AML, a core biopsy should be considered unless obtaining a tissue sample would be too high risk. Up to 27% of patients with MS will have isolated tumours.³ The present patient had initially been referred for evaluation of ovarian masses and pelvic lymphadenopathy and the diagnosis was only made with the constellation of symptoms and leukaemic blasts on peripheral smear.

Initial treatment of EMD, including isolated disease without bone marrow involvement, involves systemic induction chemotherapy. Local therapy with radiation, surgery or both can be considered for palliation of symptoms from mass effect such as nerve compression. However, this must be in

Figure 1 (A) T2 axial MRI orbits showing severe mass-like thickening of bilateral extraocular muscles, most severely involving the right medial and lateral rectus muscles, and left medial rectus muscle. (B) T2 coronal view pelvis MRI showing bilateral ovarian masses. The largest on the right measures >10 cm. Multiple soft tissues and intramuscular masses, and inguinal lymphadenopathy are also present.

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conjunction with systemic chemotherapy as nearly all cases of even isolated MS progress to systemic disease.^{3 7}

EMD such as MS can have a variety of presentations, depending on the tissues and organs involved, and requires a high index of suspicion for diagnosis.

Learning points

- ▶ Extramedullary acute myeloid leukaemia (AML), such as myeloid sarcoma, can have a variety of presenting symptoms depending on which organs and tissues are involved.
- ▶ The mainstay of treatment of extramedullary AML is systemic chemotherapy. Even isolated extramedullary disease has an extremely high rate of progression to systemic disease when treated with surgery or radiotherapy alone.

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