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Extramedullary myeloid cell tumour: presentation as anterior chest wall mass during AML relapse

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Summary

Acute myeloid leukaemia is an uncommon but an important cause of soft tissue swellings. Such extrameningeal, extramedullary leukaemic infiltrates are called extramedullary myeloid cell tumours. Despite their large size they may respond well to chemotherapy and local radiotherapy, as is demonstrated in this case.

Keywords: myeloid cell tumour; acute myeloid leukaemia

therapy, he presented in November 1997, with a progressively increasing painless lump over the anterior chest wall in the midline, of 3 weeks duration. Examination revealed a performance status of I (ECOG), and a hard mass measuring 10 × 14 cm overlying the anterior chest wall (figure 1). It was fixed, non-tender and non-pulsatile with bluish discoloration of overlying skin. Hepatomegaly (3 cm) below the right costal margin and splenomegaly (4 cm) below the left costal margin were also noted.

Extramedullary myeloid cell tumour (EMT), also known as granulocytic sarcoma, is known to occur in 3-7% of patients with acute myeloid leukemia (AML).^{1,2} It can occur at initial presentation or at relapse. We report a case of AML in relapse presenting with a large anterior chest wall mass which was confirmed to be EMT and which responded completely to a combination of chemotherapy and radiotherapy.

Case report

A 49-year-old man was diagnosed as a case of AML-M1 in May 1996. He failed to achieve remission after induction chemotherapy with idarubicin and cytosine arabinoside. Complete remission was achieved after re-induction chemotherapy with daunorubicin and cytarabine given for 3 and 7 days, respectively, in the standard dosages. He subsequently completed consolidation chemotherapy (total of three cycles). The first and third cycle of consolidation therapy were similar to the re-induction chemotherapy regimen. The second cycle consisted of intermediate dose cytosar (1 g/m² bid for 5 days). He subsequently received alpha-interferon as part of research protocol to study the efficacy of alpha-interferon during consolidation and maintenance. While on interferon

Figure 1 Lateral view showing a large mass over the anterior chest wall (reproduced with the patient's permission)

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Figure 2 FNAC of the chest wall mass

Investigations revealed haemoglobin 12 g/dl, total leucocyte count $26 \times 10^9/l$ and platelet count $338 \times 10^9/l$. Peripheral smear revealed 40% blasts, 2% myelocytes, 20% neutrophils and 38% lymphocytes. Fine needle aspiration cytology (FNAC) of the chest wall mass revealed a cellular smear showing haemopoietic cells including 18% blasts, 6% myelocytes, 29% neutrophils, 12% monocytes, and 35% lymphocytes (figure 2), confirming the diagnosis of EMT. On cytochemistry, blasts were positive for myeloperoxidase. Immunocytochemistry (APAAP) revealed the blasts to be positive for CD34, CD13 and CD33 and negative for other T and B cell markers. X-Rays of sternum and the bone scan were normal. Thus, a diagnosis of AML with relapse and EMT of the anterior chest wall, was made.

The patient received chemotherapy including daunorubicin and cytarabine for 3 and 7 days, respectively. He also received local radiotherapy to the mass using 12 Mev electrons (dose = 8 Gy/single fraction). Following this therapy, he achieved complete remission and the chest wall mass disappeared, leaving behind only the skin discolouration. The patient has now completed consolidation chemotherapy using high-dose cytosar, and was doing well on maintenance therapy with cytarabine and 6-mercaptopurine at last follow-up in September 1998.

Discussion

The term EMT refers to a solid tumour of any organ recognisable microscopically as a sarcoma, composed of leukaemic cells of the myeloid series. The term EMT encompasses all terms of extramedullary, non-medullary

leukaemic infiltrates.² Skin is one of the commonest sites of involvement. An autopsy study revealed that observable tumour nodules were most frequent in the orbit and in the chest wall.³ EMT may appear as an isolated disease, during the course of the illness, or may antedate it by a few days to months. In this patient soft tissue swelling diagnosed as EMT developed during the course of illness. This patient presented with EMT of skin of anterior chest wall and was also found to be in medullary relapse. Underlying bony involvement (of sternum/ribs) was excluded by a normal bone scan.

Acute monocytic and myelomonocytic leukaemias have a higher frequency of overt infiltrative tissue lesions than other forms of AML. Previous studies have shown that EMT is most commonly associated with AML-M4.² This patient had been diagnosed as AML-M1 and later progressed to develop EMT.

The present case report highlights certain important findings. Meticulous FNAC of the swelling should be an essential component of the initial diagnostic work-up of patients with haematological malignancy who develop soft tissue swelling. FNAC smears stained with Jenner-Giemsa (Romanowsky stain) give a diagnostic picture showing the presence of myeloid and erythroid precursors, azurophilic granules, and Auer rods, making the diagnosis quick and easy. Therefore, for the diagnosis of EMT, morphological examination of a Jenner-Giemsa stained FNAC smear is a must and may be accompanied by cytochemical (MPO, NSE) and immunocytochemical (CD13, CD14, CD33, CD43, anti-MPO) stains, as done in our case.

Patients who present with EMT at AML relapse have always been treated with systemic re-induction chemotherapy. The tumour response to local external radiotherapy in combination with chemotherapy has been well documented.⁴⁻⁶ This patient also showed complete response to chemotherapy with daunorubicin and cytosine arabinoside and local electron beam therapy. He achieved haematological remission and there is no evidence of local tumour. Prognosis for patients with EMT is related to the course of the underlying disease process. Survival in such patients has been reported to range from one to 30 months after the onset of EMT.³

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Hereditary hypophosphataemic rickets: report of a family from the Indian subcontinent

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Summary

Nutritional deficiency continues to be an important cause of rickets in the underdeveloped and developing parts of the world. In the western hemisphere, predominantly non-nutritional forms of rickets and osteomalacia are now seen. In this report we discuss a family with X-linked hypophosphataemic rickets from the Kashmir region of the Indian subcontinent (which is an uncommon entity here).

Keywords: hypophosphataemia; rickets

Nutritional deficiency is no longer an important cause of rickets in the western world, although it still is in India.¹ Rickets and osteomalacia can occur in association with a variety of disorders of proximal renal tubular function; these disorders have in common increased renal clearance of inorganic phosphates and hypophosphataemia with a normal glomerular filtration rate. Primary hypophosphataemic rickets is characterised by severe short stature and rickets in association with hypophosphataemia and hyperphosphaturia and usually has an X-linked mode of inheritance.² In this report we describe a family with hypophosphataemic rickets where we believe the mode of transmission is X-linked dominant. A Medline search for the last 30 years revealed only one such family described from India so far.³

Case report

Index case

A 3-year-old girl, 5th in birth order was born of a full term normal delivery at home. She had failed to thrive since the age of 2 years. There was no suggestion of any chronic systemic disease. Examination revealed a well-nourished girl, height 79 cm, weight 10 kg (expected 50th percentiles for her height and weight: 94 cm and 10.5 kg, respectively). Upper segment lower segment ratio was 1.23. She had frontal bossing, widening of epiphyseal ends of long bones with knock knees, rachitic rosary, Harrison's sulcus and pigeon chest deformity. Family history was significant (figures 1 and 2). Of her four siblings, the three males (E-G) were normal while the other girl (B) was affected. Her father (C) and paternal uncle (D) were affected while her mother was normal. Her paternal grandmother, who had died 10 years

earlier, was also reported to have had short stature and leg deformities.

Other affected cases

Elder sister (B): Although she had a chronological age of 16 years, there had also been a failure of linear growth since the age of 3 years, and she had gradually developed deformities of the lower limbs. She had attained menarche at age 14 years and was having regular cycles. Examination revealed a height of 134 cm and weight of 35 kg (expected 50th percentiles were 157 cm and 47.5 kg, respectively). She had mild pallor, knock knees, mild anterior bowing of legs, widening of epiphyses and pigeon chest deformity. Systemic examination was normal.

Father (C): Aged 48 years, he had developed progressive deformity of upper and lower limbs during his childhood. He was 140 cm tall (corresponding to a height age of 11 years) and weighed 42 kg (consistent with his present height). He had multiple bony deformities including bowing of legs, bending of forearms, lumbar kyphoscoliosis. Systemic examination was normal.

Figure 1 Family photograph showing short stature and anterior bowing of legs in affected individuals (reproduced with their permission)

Figure 2 Family pedigree, identifying individuals affected

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