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EXTRAMEDULLARY PARAVERTEBRAL MASS: AN UNUSUAL PRESENTATION OF HAIRY CELL LEUKAEMIA

Samanidou Valentini[1*], Panteli Aikaterini[1*], Liontos Angelos[1], Biros Dimitrios[1], Diamantina Lymperatou[1], Haralampos Milionis[1], Kapsali Eleni[2]

[1] 1st Division of Internal Medicine and Infectious Diseases' Unit, University General Hospital of Ioannina, Ioannina, Greece
[2] Department of Haematology, University General Hospital of Ioannina, Ioannina, Greece
* Contributed equally

Corresponding author's e-mail: valentinasmnsmn@gmail.com

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ABSTRACT

Background: Hairy cell leukaemia (HCL) is an uncommon, indolent, B-cell, lymphoproliferative disorder typically involving peripheral blood, spleen and bone marrow. It is commonly presenting with pancytopenia, monocytopenia and massive splenomegaly, while accounting for 2% of lymphoid leukaemias. Cases of extranodal lesions caused by HCL are rare, although these have been reported. Here, we report a case of HCL presenting as a paravertebral mass without systemic involvement.

Case description: A 58-year-old man was admitted to our hospital due to progressive difficulty walking for a month, without any other symptoms. Blood examination noted mild anaemia with Hb=12.6 g/dl and mild thrombocytopenia of 140,000/μl. Magnetic resonance imaging (MRI) and computed tomography (CT) imaging demonstrated a T6 posterior paravertebral mass lesion, extending into the spinal canal with metastatic bone lesions along the thoracic and lumbar spine. Further imaging study with CT indicated mild splenomegaly (13.4 cm) and an enlarged abdominal lymph node (3.5 cm) near celiac trifurcation.

Conclusion: A core-needle biopsy from the paravertebral mass was performed. Results showed small-sized cells with round or oval nuclei, and pale cytoplasm with immunophenotype: B-cell origination with CD20+, Cyclin D1+, DBA.44+, Annexin+ and BRAF+, indicative of HCL.

KEYWORDS

Hairy cell leukaemia, paravertebral mass, extramedullary

LEARNING POINTS

- Hairy cell leukaemia (HCL) is relatively uncommon, accounting for 2% of all leukaemia cases.
- Extramedullary and skeletal involvement in HCL is rare and shares morphological characteristics with other peripheral small B-cell lymphoma neoplasms.
- Spine biopsy is essential for diagnosis in these cases, since it is always helpful to narrow the differential diagnosis.
- Correct diagnosis is essential for treatment of HCL and leads most patients to clinical remission and sometimes long-term cures.

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INTRODUCTION

Hairy cell leukaemia (HCL) is an indolent B-cell lymphoproliferative disorder. It is an uncommon disease of small mature B cells, accounting for about 2% of lymphoid leukaemias[1]. HCL is characterised by the presence of typical hairy cells in the peripheral blood and bone marrow and cytopenias, including monocytopenia and a variable degree of splenomegaly. It more commonly affects middle-aged men with a median age at diagnosis of 58 years. Patients may be asymptomatic or complain about weakness, fatigue, abdominal discomfort related to splenomegaly, easy bruising, bleeding, and symptoms or signs due to infections. Extramedullary disease caused by HCL is extremely rare and reported as isolated observations. Such unusual sites of involvement include bones, central nervous system, abdominal and – more rarely – peripheral lymph nodes, serosa, skin, kidneys, eye and pancreas[2]. According to the literature, the incidence of skeletal involvement in HCL is approximately 3%. These lesions are most commonly osteolytic, have a predilection for the proximal femur and are usually associated with extensive bone marrow infiltration by hairy cells[3]. Tumour infiltration of vertebrae and compression of the spinal cord has been extremely rarely reported[4]. We present this case of an unusual first presentation of HCL and discuss the challenges diagnosing an uncommon presentation of a rare disease.

CASE DESCRIPTION

A 58-year-old man was referred to the emergency department of our hospital due to progressive weakness in his lower extremities over a one-month period. He did not report any other systemic symptoms. No significant medical history was noted aside from cigarette smoking of 50 pack years, and he was not receiving any medication or using illicit drugs. On further questioning, there was no family history of known malignancies or autoimmune diseases. A neurological examination revealed reduced muscle strength and exaggerated deep tendon reflexes in both lower limbs.

A complete blood count indicated mild anaemia with haemoglobin of 12.6 g/dl (normal range 13–18 g/dl), a white blood count of 5.4 × 10^9 (normal range 4–11 × 10^9) with a differential count of 72% neutrophils, 24% lymphocytes, 2% monocytes, 1.3% eosinophils and mild thrombocytopenia of 140 × 10^9/l (normal range 150–440 × 10^9/l). Moreover, biochemistry including calcium and lactate dehydrogenase levels, and protein electrophoresis were normal, except for kidney function with creatinine 1.3 mg/dl and estimated glomerular filtration rate of 56.7 ml/min/1.73m^2. Peripheral blood flow cytometry did not detect any monoclonal B-cell population. Further examination with MRI imaging demonstrated a T6 posterior paravertebral mass lesion, extending into the spinal canal with metastatic bone lesions along thoracic and lumbar spine *(Fig. 1)*. Additional imaging study with CT of head, chest and abdomen indicated mild splenomegaly (13.4 cm) and an enlarged abdominal lymph node (3.5 cm) near celiac trifurcation. An initial differential diagnosis of plasma cell disorder particularly plasmacytoma, primary bone tumour or atypical presentation of lymphoproliferative disorder was made.

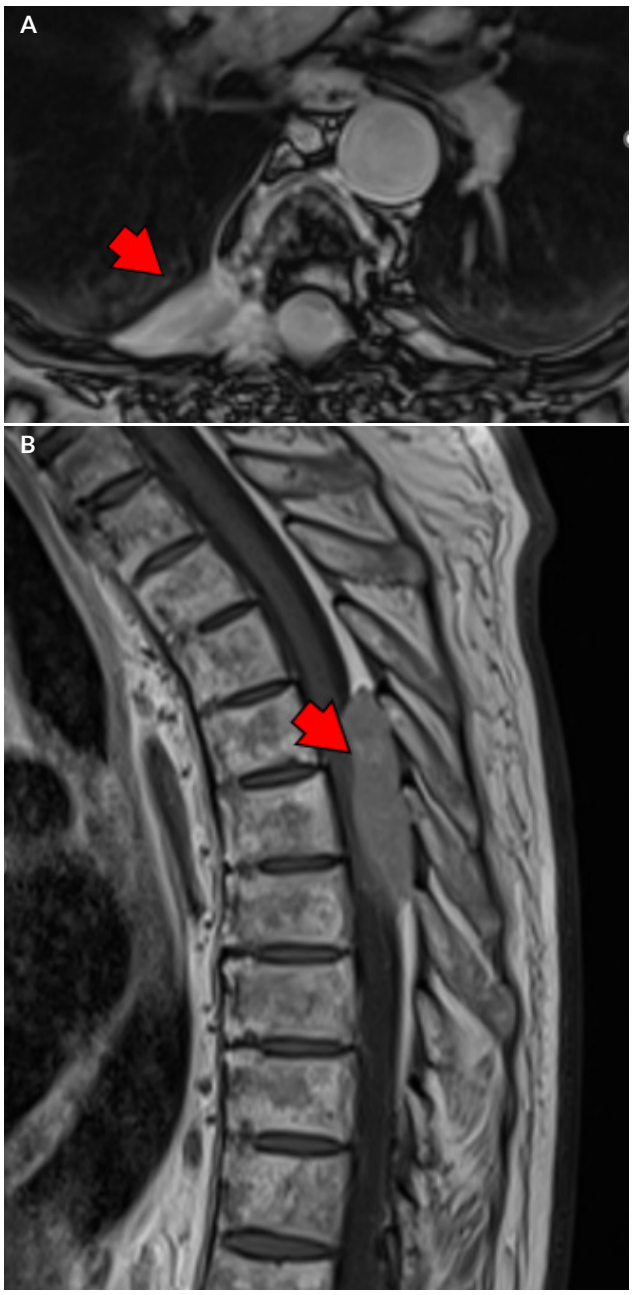

Figure 1. MRI scan of head and spinal cord revealed T6 posterior paravertebral mass extending into the spinal canal.

A vertebral mass biopsy was performed, and results showed small-sized cells with round or oval nuclei, and pale cytoplasm with immunophenotype: B-cell origination with CD20+, Cyclin D1+, DBA.44+, Annexin A1+ and BRAF+, indicative of HCL. Bone marrow aspiration indicated normocytic bone marrow with 30% lymphocytes while a bone marrow biopsy indicated 80% infiltration from HCL. The patient was moved to the special haematology department of our hospital and treatment with pentostatin was initiated. A follow-up scan with MRI four months after chemotherapy *(Fig. 2)*, as well as a bone marrow biopsy six months after chemotherapy, showed an excessive response to treatment. The patient was completely asymptomatic.

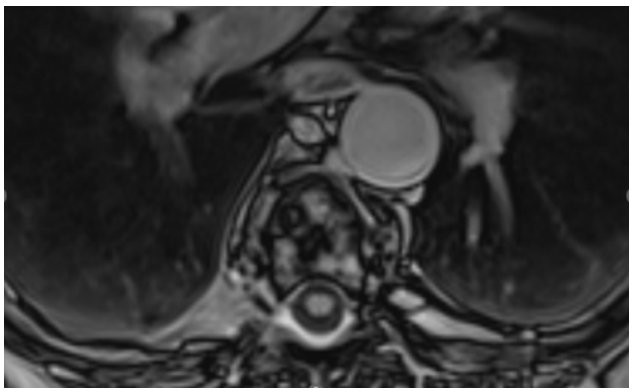

Figure 2. MRI of head and spinal cord four months after chemotherapy.

DISCUSSION

HCL, a rare B-cell lymphoproliferative disorder, is characterised by the presence of lymphoid cells with typical morphological and phenotypic features accumulating in the peripheral blood, bone marrow and spleen[5]. Leukaemic cells have central nuclei, abundant cytoplasm with filamentous surface micro-projections and express CD11c, CD25, CD103, CD123, CD20, CD22, CD52 and mild to moderate expression of Cyclin D1. Classical HCL must be differentiated from the hairy cell variant, splenic diffuse red pulp lymphoma and splenic marginal zone lymphoma. Expression of at least three of the CD11c, CD25, CD103 and CD123 markers is required for the diagnosis of classical HCL[6]. Furthermore, all patients present a BRAF V600E mutation, which is considered a disease-defining genetic event. Cytopenias, monocytopenia and splenomegaly are the most common features of the disease. Treatment with purine analogues alone or in combination with an anti-CD20 monoclonal antibody results in durable, complete responses[7]. Among unusual manifestations of HCL is skeletal involvement, which is rarely observed at the presentation of the disease, complicating the course of the HCL in about 3% of patients[3]. The lesions are mainly osteolytic, painful, and have a preference for the axial skeleton and proximal long bones with the hip being the predominant site[8]. There are also reports of cases of diffuse osteosclerosis, mixed sclerotic and lytic pattern and osteoporosis. Arthralgia with or without a localised effusion may occur rarely. Areas of aseptic necrosis can also appear in the case of high infiltration with HCL. Bone involvement is associated with high tumour burden and extensive bone marrow infiltration by hairy cells. Systemic treatment with purine analogues should be given in these patients[3]. Bone lesions with minimal or absent marrow infiltration have been also rarely been described[9]. Osteolytic lesions respond well to local radiotherapy in solitary bone involvement without bone marrow infiltration[3]. Because of more sensitive imaging techniques (MRI, PET/CT scans), bone involvement will be discovered more often[10].

Our patient did not experience any bone pain associated with his osteolytic bone lesions along the thoracic and lumbar spine. His main complaint that led to diagnosis was neurological, suggesting a central or peripheral nervous system disease. Central nervous system involvement of the brain parenchyma or meninges has been reported as anecdotal cases in HCL. Very rare extranodal HCL with invasion of vertebrae, radicular infiltration and spinal cord compression results in manifestations of the peripheral nervous system[2]. Mayo clinic has reviewed 108 patients with HCL and found that only 8 patients had neurological disease, and one person had symptoms from leukaemic compression of neural structures[11]. Recently, Claves et al. described a case of HCL with meningeal localisation treated with cladribine and rituximab. The authors also reviewed the literature. Nine other cases with central nervous system manifestations were identified. A complete response was achieved in the patients who were treated with cladribine alone or in combination with rituximab[12]. Extranodal HCL presenting in the spine has also been described in a case report by Rosen et al. Unlike our patient, this patient experienced back pain[4].

In addition, typical findings of peripheral cytopenias and massive splenomegaly in HCL were not seen in our case. Although splenomegaly is the most common clinical finding in HCL, the incidence of HCL without splenomegaly in various series ranged from 0 to 40%[13]. This suggests that absence of an enlarged spleen does not exclude the diagnosis of HCL. It is also estimated that the incidence of true pancytopenia is about 40%. In a recent review involving more than 500 patients, 75–84% of them had anaemia, 57–79% had a platelets count below 100,000 /mm^3, and 60% of patients had leukopenia at presentation[14].

The atypical clinical manifestation with bone lesions and a paravertebral mass, as well as the lack of indicative laboratory data, initially led to a differential diagnosis which did not include HCL. Obtaining a core biopsy of the mass was crucial for the diagnosis and appropriate treatment of the patient.

CONCLUSIONS

HCL may have atypical presentations including symptoms from spinal canal compression. When HCL presents as an isolated extramedullary mass, the diagnosis can be challenging. An appropriate biopsy specimen is critical for distinguishing HCL from other more common lymphoproliferative, plasma cell disorders or primary bone tumours.

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