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Case Report

Paediatrics Section

Acute Myeloid Leukaemia Unmasked by Paraplegia: A Case Report

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ABSTRACT

Acute Myeloid Leukaemia (AML) is a rapidly progressive myeloid neoplasm that is characterised by clonal expansion of immature myeloid lineage cells, known as blasts in the bone marrow and peripheral blood. It is known to present with pallor, bleeding tendencies and infections. However, Extra Medullary (EM) presentations involving the central nervous system are rare and can delay clinical diagnosis. Hereby, the authors report a case of 14-year-old male child who had a unique presentation of acute myeloid leukaemia. His initial symptoms were indicative of a central nervous system disease, and later the child was diagnosed to have acute myeloid leukaemia with spinal metastasis. A case of AML which does not routinely present with lower limb weakness and bowel or bladder dysfunction can be isolated with this case report and a crucial diagnosis can be caught early on. The present case underscores the critical need for maintaining high suspicion for haematological malignancies in children presenting with neurological deficits. It also highlights the different dimensions of the disease pathology and the disease's rapid progression to metastasis. The key role of diagnostic imaging to isolate metastasis has also been focused on.

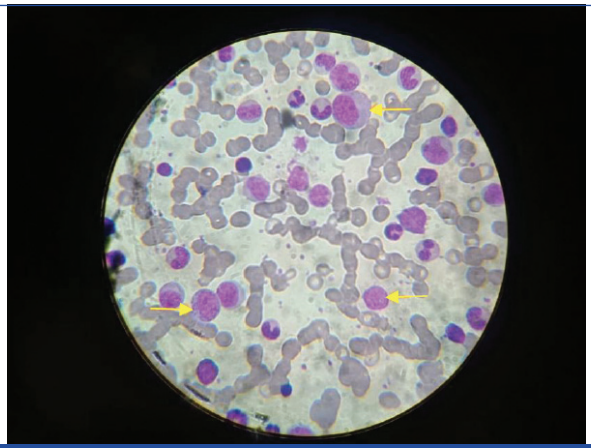
Keywords: Bone marrow, Metastasis, Myeloid neoplasm, Myeloid Sarcomas, Neurological sequelae, Spinal metastases

CASE REPORT

A 14-year-old male child was admitted to the Department of Paediatrics with complaints of pain and progressive weakness of bilateral lower limbs for one week, with burning micturition and dribbling of urine and occasional bowel and bladder incontinence for four days. There was no history of sensory involvement and no familial history of similar complaints in a developmentally normal child. The child had a history of on-and-off bowel and bladder incontinence in the last 7-8 months, 1-2 episodes every month, lasting for around 3-4 days, for which no medical advice was sought. These complaints worsened in the preceding week, and the child was unable to pass urine or stool for two days, accompanied by dull, central abdominal pain. He also had complaints of bilateral lower limb weakness with difficulty and pain during walking a short distance. The mother also reported visible weight loss, as evidenced by the loosening of clothes over a 6-month period, with a history of night sweats present.

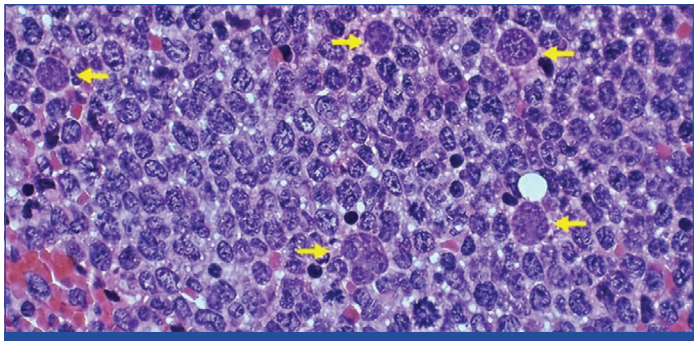
On admission, the child was vitally and haemodynamically stable with a Body Mass Index (BMI) of 12.2 kg/m^2, underweight, Glasgow Coma Scale (GCS) score 15/15, with vitals within normal limits. Evidence of pallor was present but there was no enlarged lymph node or mass. On abdominal examination, inspection did not reveal any abnormality however, on palpation, the bladder was palpable well above the pubic symphysis and was tense. There was no splenomegaly or hepatomegaly noted, percussion did not reveal any abnormality. On neurological examination, higher mental functions and cranial nerve examination normal, however motor examination was suggestive of increased tone in all four limbs with reduced power and exaggerated reflexes in lower limbs with plantar extensors, no cerebellar signs or meningeal signs. The child was catheterised with a Foley's catheter and 400 mL of urine was drained immediately on insertion. Cardiovascular, respiratory and other organ systems were not involved. Considering the neurological findings, child was suspected to have some central nervous system involvement, probably a mass or lesion leading to upper motor neuron lesions. The child was started on antibiotics-ceftriaxone (100 mg/kg/day for seven days) and symptomatic treatment like fever management and pain management was done

Routine complete blood count revealed haemoglobin 8.3 g/dL and total leukocyte count 30,000 cells/mm^3. Differential leukocyte count showed myeloblasts 35%, promyelocytes 5%, myelocytes 5%, mature polymorphs 25%, lymphocytes 20%, monocytes 7%, eosinophils 2%, and basophils 1%. Platelet count was reduced (66,000/mm^3). Premature blast cells were noted on peripheral smear, suggestive of acute leukaemia [Table/Fig-1], and hence the child was taken up for bone marrow biopsy and aspiration, due to a strong suspicion of leukaemia. Bone marrow aspiration and biopsy revealed a hypercellular marrow with marked myeloid predominance. Differential count showed monoblasts 7%, myeloblasts 45%, promyelocytes 5%, myelocytes 15%, metamyelocytes 10%, band forms 5%, lymphocytes 5%, normoblasts 6%, plasma cells 1%, and eosinophils 1%. The blast percentage (52%) with evidence of granulocytic maturation beyond the promyelocyte stage is suggestive of AML with maturation (French-American-British (FAB) classification: AML-M2 subtype) [Table/Fig-2].



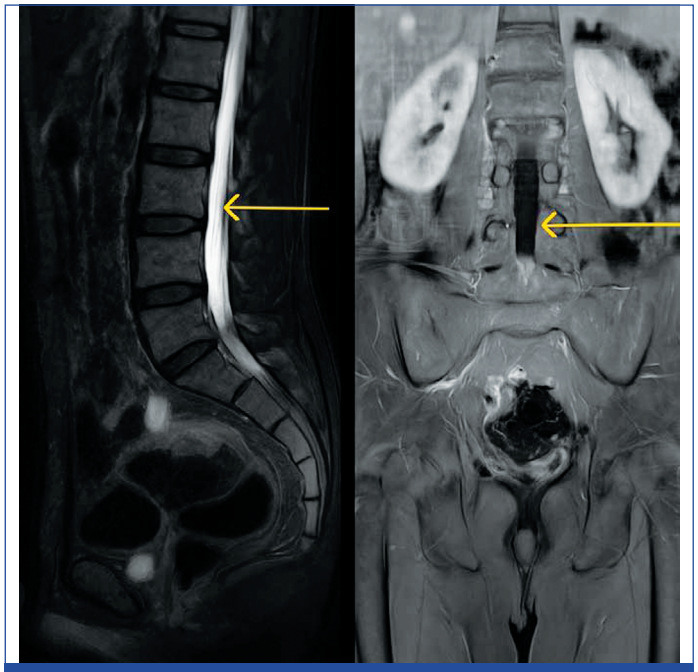
[Table/Fig-1]: Peripheral smear showing circulating myeloblasts with high nuclear-cytoplasmic ratio (yellow arrow) Leishman stain, 1000× oil immersion.

Flow cytometry was sent for further specifications, and the results were positive for Cluster of Differentiation (CD)13, CD33, Myeloperoxidase (MPO), CD56, CD38, Human Leukocyte Antigen – DR isotype (HLA-DR), CD34, and CD117. The child had no improvement in neurological symptoms with persistent retention of urine and stools, and paraesthesia in both lower limbs and was hence planned for chemotherapy.



[Table/Fig-2]: Bone marrow aspirate smear, showing myeloblasts and granulocytic precursors suggestive of AML-M2 (yellow arrow), Leishman stain, 1000× oil immersion magnification.

MRI of the spine with contrast was done, which was suggestive of an intradural Extra Medullary (EM) lesion in the spinal canal at the level of L5-S1 of size 1.4×1.8×3 cm {Anteroposterior×Transverse ×Craniocaudal (AP×TV×CC)}, causing mass effect on the thecal sac in the form of displacement of the nerve roots towards the right with obliteration of the anterior and posterior thecal sac. Presacral mass lesion of size of approximately 6.1×2.2×8.2cm (TV×AP×CC) with extension along the sacral foramina and post elements. The features were suggestive of lymphoid infiltrative masses [Table/Fig-3]. The child was started on intravenous hydration as per daily requirements, oral allopurinol at 10 mg/kg/day, Sulphamethoxazole and trimethoprim at 160 mg per day and fluconazole at 12 mg/kg/day. Initial suspicion was of a mass compressing the cord as child had complaints of lower limb weakness. Since there was no ascending progression of the weakness and also similar history in the past, Guillain-Barré syndrome was excluded.



[Table/Fig-3]: Magnetic Resonance Imaging (MRI) lumbosacral spine, T2-weighted (T2W) and contrast-enhanced scan showing intradural extra medullary lesion in the spinal canal at level of L5-S1, lymphoid infiltrative mass (yellow arrow).

Based on the bone marrow biopsy and aspiration with flow cytometry reports along with the neuroimaging, child was diagnosed with AML, M2 type with spinal metastasis. Child was planned for the azacitidine and Venetoclax protocol or for the 7+3 protocol-Cytarabine and anthracycline. Parents were advised for karyotyping and molecular marker testing of peripheral blood and then further conduction of chemotherapy; however, parents declined any further treatments and were then lost to follow-up.

DISCUSSION

Haematological malignancies can infiltrate EM soft tissue masses that occur at sites other than the bone marrow and present with varying symptoms. Extra Medullary Leukaemia (EM AML), also known as Myeloid Sarcoma (MS), is an uncommon as a presenting features and occur in 2-9% of newly diagnosed cases [1]. As seen in the case, the child was approached with a Central Nervous System (CNS) differential in mind, which later unearthed the diagnosis of AML. A similar case was noted in a 15-year-old male child who had difficulty in walking and was later diagnosed with AML with space-occupying lesions causing severe cord compression in thoracic spinal canal [2]. EM tumour masses consisted of malignant primitive myeloid cells that infiltrate and efface the underlying tissue structures, most commonly the lymph nodes, skin, soft tissues, bone, and peritoneum. Because of its rarity, a misdiagnosis rate of 25-47% has been reported [3]. EM AML is most commonly labelled as malignant lymphoproliferative disorder or large-cell lymphoma. The clinical presentation is often dictated by the mass effect and location of the tumour [3]. As seen in the presenting case, the features that the child presented with, like lower limb paresis, dictated a workup for neurological pathology, which eventually led to the diagnosis of AML.

Considering the wide variety of sites that EM AML can occur, proper imaging is crucial for diagnosis [4]. As MS presents as a soft tissue mass, CT imaging is the most suitable diagnostic modality [5]. For more effective screening in patients with bone-marrow AML, 18F-Fluorodeoxyglucose-Positron Emission Tomography (18FDG-PET) imaging is highly recommended [4]. For cases with suspicion of brain, spine or orbit involvement, MRI with or without gadolinium contrast is considered [6].

Despite a CNS-focused presentation, confirmation of the diagnosis of MS is done by tissue sampling, typically by fine-needle aspiration. MS typically consists of a diffuse and infiltrative population of myeloblasts and granulocytic cell components, with the malignant cells appearing large with abundant cytoplasm and large nuclei. Samples are then further analysed by flow cytometry, fluorescence in situ hybridisation and molecular analysis. Immunohistochemistry (IHC) markers contribute to lineage classification, of which CD68-KP1 is the most commonly expressed marker, followed by MPO [6].

It is important to rule out Non Hodgkin's lymphoma, lymphoblastic leukaemia, melanoma, plasmacytoid dendritic cell neoplasm, and Ewing's sarcoma while considering the diagnosis of extra medullary metastasis of AML in atypical presenting features [6,7].

EM AML with concurrent medullary involvement has an overall poor prognosis, with a 5-year survival rate between 20-30% [3]. The presence of cytogenetic alterations has a conflicting role in the prognosis of the disease, with t(8;21) having a relatively better prognosis [8] whereas those involving chromosome 8 had a worse outcome [7].

Treatment modalities of AML with EM disease are governed by the extent of involvement; isolated involvement calls for intensive AML chemotherapy with radiotherapy as consolidation, and when marrow and EM involvement is seen, intensive chemotherapy with Haematopoietic Cell Transplantation (HCT) and radiotherapy, if EM is persistent after induction chemotherapy [9,10]. Chemotherapy in younger individuals includes a two-drug combination-cytarabine plus an anthracycline (daunorubicin or idarubicin), the 7+3 regimen, which was planned for the child [11]. The consolidation phase usually involves long-term administration of a single drug-cytarabine. Another option for individuals in whom intensive therapy is unsuitable or not tolerated is azacitidine and venetoclax [10].

Newer modalities in management of AML with metastasis introduced by the US Food and Drug Administration (FDA) include targeted therapies including midostaurin (FLT3 inhibitor), gemtuzumab ozogamicin (CD33-directed antibody-drug conjugate), and enasidenib (IDH2 inhibitor). [11]. These biologics have demonstrated an improved response in target sub populations; however, further studies are still required to determine their efficacy.

The present case underlines the importance of maintaining a high index of suspicion for haematological malignancies in children presenting with predominant neurological symptoms. It contributes to improving the overall survival outcome of such atypical cases.

CONCLUSION(S)

Nervous system symptoms like lower limb weakness and bladder dysfunction can rarely be the initial presentation of AML. Moreover, diagnosis of the same requires a high degree of suspicion due to varied unusual modes of presentation as seen in the above case. Early catch facilitates prompt intervention that improves the prognosis of the disease.

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