Case Report

Myeloid Sarcoma Presenting with Proptosis, Ruptured Globe, and Facial Swelling

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Abstract

Myeloid sarcoma (MS) is a rare malignancy of immature myeloid cells and/or monocytes that occur in an extramedullary site. It is frequently mistaken for small-round-blue cell tumors, in the absence of immunohistochemistry. A case report of an unusual presentation of MS with a ruptured globe at a tertiary setting of North-western Nigeria is presented. The patient was a 12-year-old girl with progressive bilateral proptosis and spontaneous rupture of the left eye. A histologic diagnosis of embryonal rhabdomyosarcoma was made, but she had a minimal improvement in her clinical condition following chemotherapy. Further review showed bone marrow myeloblasts >98%. The previous tissue biopsy was subjected to immunohistochemistry and found to be CD117 – strongly positive and CD34 – patchy positive. The conclusion was that of myeloid leukemic infiltration of orbital tissue MS. The patient abandoned the treatment due to caregiver fatigue and financial exhaustion.

Keywords: Facial swelling, leukemia, myeloid sarcoma, proptosis, ruptured globe

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Introduction

Myeloid sarcoma (MS) is a rare malignancy comprising immature myeloid cells and/or monocytes that occur in an extramedullary site. It is sometimes referred to as granulocytic sarcoma, myeloblastoma, or extramedullary myeloid cell tumor.[1-3] MS may present *de novo* as the initial manifestation of acute myeloid leukemia (AML) or appear concomitantly. In other cases, it presents as a relapse of AML. MS can present at any age, with reported ages being between 1 and 81 years.[2,4] It can affect a variety of tissues, with the skin, bones from any site, lymph nodes, genitalia, orbit, breasts, and gingivae reported to be affected.[2,5] Other sites include the pleura, retroperitoneum, oral mucosa, and gastrointestinal tract.[1] The skin and the orbit are the most common sites involved in children.[3] Commonly, MS manifests with compression symptoms, pain, and abnormal bleeding, depending on the location and size of the tumor.[1] The incidence of MS in adults with AML is about 2%–5%, but in children with AML, it reaches to about 40%.[3]

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MS is also termed chloroma, due to the greenish color imparted by the presence of myeloperoxidase (MPO).[3] Histologically, MS displays one of the several patterns. The immature granulocytic sarcoma is characterized by numerous blasts, the cytoplasm of which is agranular, with varying degrees of basophilia. These are positive for CD34, CD43, CD117, and lysozyme on immunohistochemistry.[5] Differentiated granulocytic sarcomas have more mature neutrophils, and they show reactivity for CD43, MPO, CD15, lysozyme, and CD117. Monoblastic sarcomas have large populations of monoblasts, with eosinophilic cytoplasm and strongly positive for CD43, lysozyme, CD68, and CD163; weakly for CD4; and negative for CD34. On the other hand, monoblastic sarcomas have marked nuclear lobulation and are positive for CD43, lysozyme, CD68, CD163, and stain variably with MPO. Myelomonocytic

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sarcomas show both neutrophilic and monocytic precursors, with the corresponding immunohistochemical markers, as well as increased numbers of eosinophils.[5] The monoblastic sarcoma was observed in 100% of *de novo* MS in one study.[5] Many immunohistochemistry panels include CD43 and lysozyme, but MPO is considered nonspecific. CD117 signifies myeloid differentiation, while CD68 and CD163 are expressed on monocytic cells.[3]

When MS presents *de novo*, it has been reported to take up to a year before it is detected in the bone marrow.[3] It is frequently mistaken for small-round-blue-cell tumors, and the diagnosis is missed in half of the cases where immunohistochemistry is not utilized.[3,5] MS is treated with conventional AML-type chemotherapeutic regimens, and it has similar cure rates and prognostic factors.[3] However, in cases initially misdiagnosed as lymphomas or the likes, other chemotherapy regimens had been given before the diagnosis of MS was made.[2] Overall, the presence of MS gives a poorer outcome, although other factors such as cytogenetics do influence the prognosis.[3]

An unusual presentation of MS is presented here. Written informed consent was obtained from the parents, and assent was given by the patient.

Case Report

A 12-year-old girl was admitted to the pediatric hematology–oncology unit of a tertiary hospital in Northwestern Nigeria, with a 10-week history of progressive bilateral periorbital swellings and eyeball protrusions. The left eyeball spontaneously ruptured after 4 weeks of the onset of the protrusion. She also had reddening and excessive tearing of the right eye though vision was preserved. There was an 8-week history of painful swelling of the left temple and preauricular area. She had lost some weight, otherwise, nil other systemic symptoms. She was treated with traditional herbs and concoctions at home before hospital care was sought [Figure 1].

Examination revealed a tender and firm left eye fleshy mass, measuring 4 cm × 5 cm, and a diffuse left preauricular swelling, 9 cm × 6 cm [Figure 1]. The right eye was proptosed, with conjunctival injection. There was a generalized facial edema, with nasal obstruction and mouth breathing. No masses were noticed in the oral or nasal cavities. There were no lymphadenopathy and no splenic or hepatic enlargements.

A clinical diagnosis of head-and-neck rhabdomyosarcoma was entertained, and she had an incisional biopsy from the left temporal swelling done. Baseline investigations were within the normal limits, and infection screens were negative for tuberculosis and the human immunodeficiency virus. The histology result was in keeping with round-blue-cell tumor and favored a diagnosis of embryonal rhabdomyosarcoma [Figure 2].

She received four cycles of chemotherapy for rhabdomyosarcoma, with vincristine, cyclophosphamide, and actinomycin pulses. There was a marginal improvement in her clinical condition. She developed progressive anemia and recurrent epistaxis during chemotherapy, which necessitated multiple blood transfusions. On re-assessment of her condition, she was found to have a raised white blood cell count on the full blood count, and a peripheral blood film showed myeloblasts.

A re-assessment of her bone marrow aspirate subsequently showed markedly increased myelopoiesis with increased myeloblasts >98% of bone marrow nucleated cells; erythropoiesis and lymphopoiesis were markedly reduced, while megakaryopoiesis was absent. A conclusion of AML FAB M5 was made [Figure 3].

The previous tissue biopsy was reviewed and subjected to immunohistochemistry, which showed CD117 – strongly positive [Figure 4] and CD34 – patchy positive, [Figure 5]. Therefore, a conclusion of MS was made.

Figure 1: Proptosis and chemosis of the left eye, with a ruptured globe and corneal laceration. There is a swollen left ear and temple

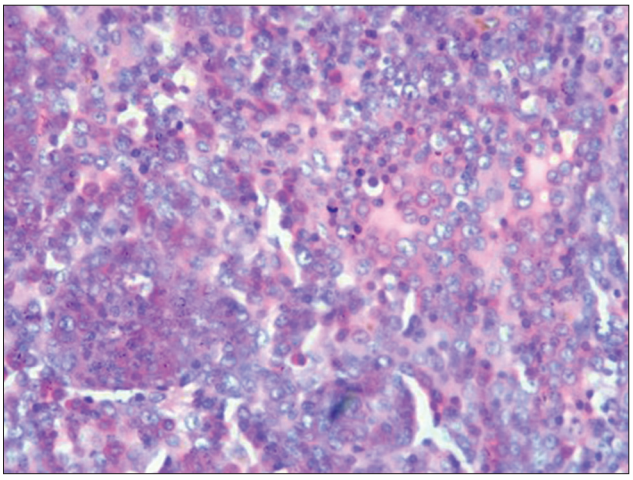

Figure 2: Micrograph showing sheets of undifferentiated cells, a few of which show nuclear indentation. H and E, ×200

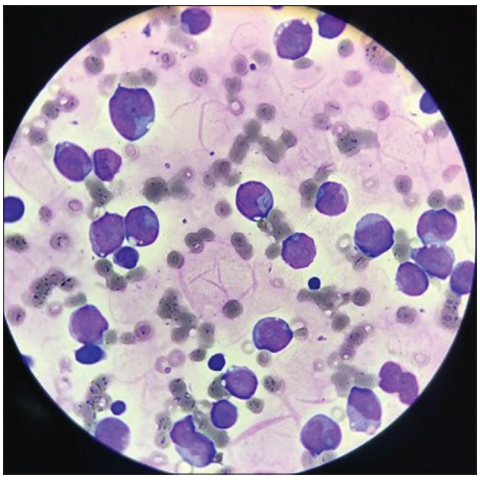

Figure 3: Bone marrow aspirate showing numerous myeloblasts

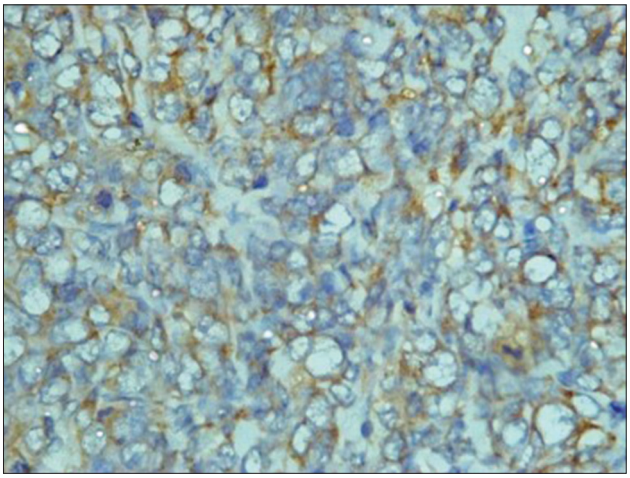

Figure 4: Micrograph showing strong positive membranous staining of tumor cells for c-kit (CD117)

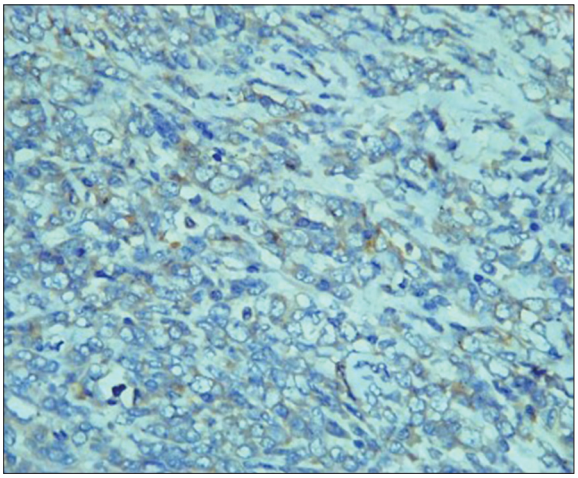

Figure 5: Micrograph showing focal staining of tumor cells for CD34

Unfortunately, the patient abandoned the treatment due to caregiver fatigue and financial exhaustion. She was discharged home on palliative care.

Discussion

This case report shows an unusual presentation of MS with a ruptured globe and facial swelling, as well as a delayed presentation and diagnosis. Late presentation is a common occurrence in childhood malignancies as documented from our environment.[6] This contributed to the advanced nature of the disease at presentation, evident by the rupture of the globe. As demonstrated in this case, such delays in a presentation to the hospital may be due to the patronage of traditional or alternative medicines before orthodox care was approached.

The rarity of MS makes its diagnosis easily missed, especially when it arises *de novo* and in situations where immunohistochemistry panels are not routinely employed.[3,5] Hence, the index case presentation of swelling on the head-and-neck region and the histological hematoxylin and eosin appearance of round blue cells influenced the initial diagnosis as rhabdomyosarcoma. Indeed, literature have shown that MS is frequently diagnosed as a lymphoma or other round blue cell tumors before immunohistochemistry clarifies the diagnosis.[3,5]

The findings in keeping with AML raised the possibility of chloromas. This latter differential was confirmed after the initial histology blocks were subjected to a limited immunohistochemistry panel. The positivity of the myeloid markers such as CD117 and CD34 supported the consideration of a chloroma in the tissues of the facial swelling.[5]

With hindsight, it is obvious that the initial facial swellings and proptosis were due to the chloromas, hence a MS, which subsequently became manifest in the bone marrow. The only question would be whether it was a *de novo* MS that subsequently spilled into the bone marrow or it was a concurrent tissue and bone marrow disease from the beginning.

The immunohistochemistry panel utilized was dictated by availability and cost–benefit. This is a reality in our environment and practice, whereby judicious use of limited resources must be made to avoid heavy costs on patients. Therefore, investigations are rationalized based on clinical judgment. The ideal would have been employed a larger panel of markers to get a more refined diagnosis. Ultimately, a high index of suspicion, as well as improved diagnostic facilities, is vital to sharpen diagnostic accuracy of rare tumors such as MS, in our resource constrained settings.

Sadly, after the diagnosis of MS was reached, caregiver fatigue and financial exhaustion took its toll, and the patient abandoned the treatment. This is an unfortunate outcome in a lot of oncology cases in our environment, which only adds more odds against survival from an already poor prognostic disease.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients

understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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