CASE REPORT

A Case of Myeloid Sarcoma Masquerading as Neuroblastoma

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ABSTRACT

Myeloid sarcoma (MS) is an uncommon type of malignancy, and its diagnosis is comparable to acute myeloid leukaemia (AML). In the rare circumstances in which MS does present without AML, it is known as MS de novo. We report a case of a 10-month old child who presented with bilateral proptosis and a pelvic mass due to synchronous primary MS de novo. She was initially misdiagnosed with neuroblastoma, which has this typical presentation. The histopathological result from the biopsy of the orbital mass also showed a small blue round cell tumour (SBRCT) as seen in cases of neuroblastoma. However, the diagnosis of MS was confirmed using immunohistochemistry (IHC) from the orbital biopsy specimen, which usually plays a major role in the diagnosis of orbital tumours and as a prognostic indicator. Our patient remains in clinical remission two years after antileukemic treatment, with no relapse or progression to AML.

Keywords: Proptosis, Neuroblastoma, Myeloid sarcoma, Immunohistochemistry

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INTRODUCTION

Bilateral proptosis is a very uncommon presentation in children, and it is highly indicative of a systemic malignancy. Once noted in a child, cancers such as metastatic neuroblastoma, myeloid sarcoma, leukaemia and lymphoma need to be ruled out urgently. Neuroblastoma is the most common extra-cranial solid tumour seen in children (1). Patients typically present with fever, weight loss, bilateral proptosis and an enlarged adrenal gland mass in the abdomen. Occasionally, this mass may be seen in the chest, pelvis or neck. Many patients also present with "racoon eyes" due to periorbital ecchymosis (2).

Myeloid sarcoma (MS) is also another cause of bilateral proptosis. It is also known as granulocytic sarcoma or chloroma, owing to its green colour from the enzyme myeloperoxidase. It consists of extramedullary proliferation of blasts from the myeloid lineage. Therefore, the diagnosis of MS is comparable to acute myeloid leukaemia (AML). MS is seen in only 2-8% of cases of AML, with nearly half the cases of MS appearing after the diagnosis of AML, with another 15-35% seen concurrently. However, in a small number of patients, MS may precede AML by several months. When MS presents without AML, it is known as MS de novo. Owing to the rarity of MS de novo, it has only been described in the literature in the form of case series or reports. We therefore describe one such case.

CASE REPORT

In August 2016, a 10-month-old girl presented to the ophthalmology clinic with a history of bilateral proptosis of seven days duration. The mother of the child also reported of drooping of right eyelid and increasing severity of the proptosis in the left eye over the last two days (Figure 1).

On general inspection, the child was lethargic, and a bloodstained fluid oozed from her left nostril. On eye examination, there was no periorbital inflammation or ecchymosis seen. There was complete ptosis of the right eye, with hypoglobus and limitation in upgaze,

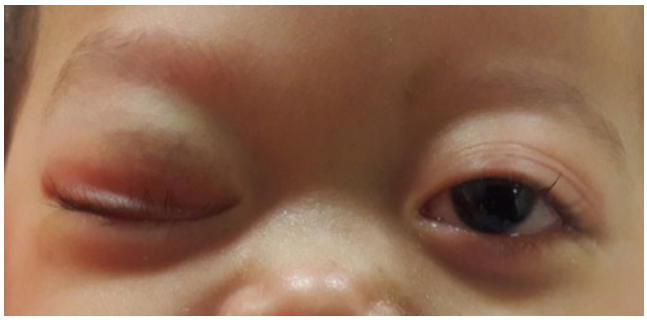

Figure 1: External photograph, pre-chemotherapy: showing bilateral proptosis with complete ptosis of the right eye

originating from a poorly defined mass which was palpable in the superomedial part of the right upper lid extending into the orbit. The proptosis in the left eye was more severe and associated with restrictions in all direction of gaze. The pupillary reflexes in the left eye was impaired, and the intraocular pressure in both eyes was elevated at 28 mmHg. Bilateral fundus examination was unremarkable, with no signs of optic disc swelling or choroidal striae seen. On systemic examination, there was no lymphadenopathy, organomegaly or a palpable mass in the abdomen. Laboratory workups, which included complete blood count, peripheral smear, coagulation screening, renal and liver function tests, were within the normal reference range. The fungal smear and culture from the nasal cavity were also negative.

A contrast-enhanced computed tomography scan (CT scan) of the orbits was performed, which showed bilateral hyperdense extraconal masses with calcification within (Figure 2). The extraconal mass in the left orbit was larger than that of the right orbit; it involved the orbital apex and extended into the maxillary sinus and posterior nasal cavity. Also notable, was the fact that the extraconal mass in the right orbit compressed the globe inferiorly (Figure 3). These findings were suggestive of systemic metastases, leukaemia, or lymphoma. Subsequently, a staging CT scan was done which revealed a large, lobulated, heterogeneous enhancing mass in the right pelvis. It extended from the fifth lumbar spine to the pelvic floor, with no calcification or necrosis within (Figure 4). There was no involvement of the para-aortic

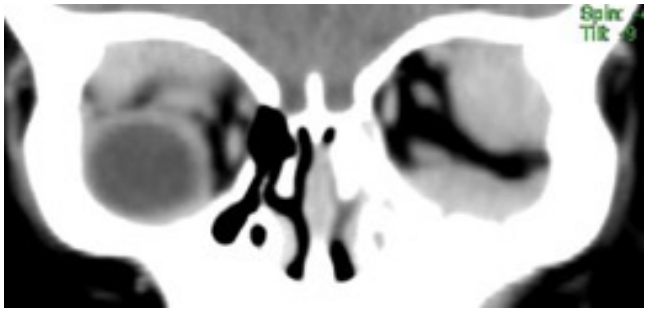

Figure 2: Coronal CT scan of the obit demonstrates significant proptosis in the left eye due to large contrast enhancing extraconal masses invading the orbital apex infiltrating the posterior nasal cavity. In the right orbit, the extraconal mass compresses the globe inferiorly.

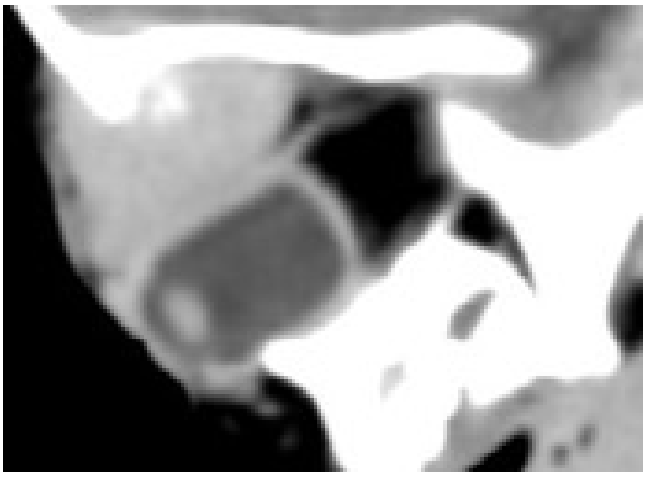

Figure 3: Sagittal CT scan of the right orbit showing an extraconal enhancing mass with calcification within causing compression of the globe inferiorly

lymph nodes, liver, spleen, or kidneys seen. A provisional diagnosis of neuroblastoma stage 4 was made with the primary tumour originating in the pelvis and causing metastasis to both orbits. A biopsy of the mass from the right upper lid was done under general anaesthesia, for which the histopathological results pointed to a small blue round cell tumour (SBRCT), which supported the diagnosis of neuroblastoma. Immediately, one course of chemotherapy was commenced to salvage the remnant vision in the left eye. Within one week of starting the chemotherapy, the bilateral proptosis reduced significantly, and the pupillary reflexes of the left eye returned to normal.

Further staging investigation was undertaken to establish a treatment strategy. However, when the MIBG scan did not show any tracer uptake in the orbit, adrenal glands nor soft tissue density in the pelvis, this presented as a diagnostic dilemma. Other investigations including bone marrow trephine biopsy also did not show any abnormality. After two weeks, the immunohistochemistry (IHC) results returned, which was negative for neuroblastoma but positive for myeloperoxidase and lysozyme biomarkers which was strongly indicative of MS. Based on the IHC results, the

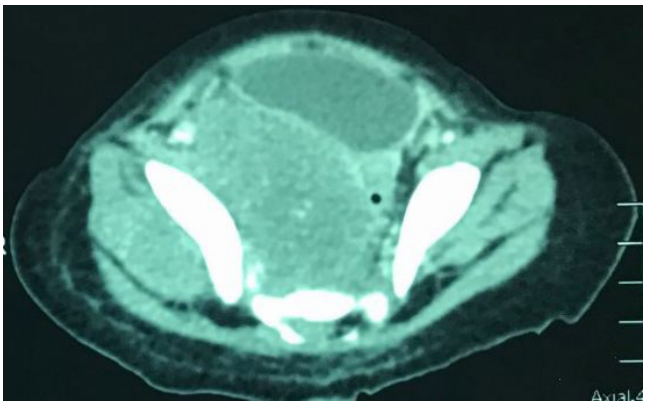

Figure 4: Axial CT scan abdomen: demonstrating a large lobulated heterogeneously enhancing mass pushing the urinary bladder anterosuperiorly

case was re-diagnosed as synchronous MS de novo of the orbit and pelvis.

The patient was subsequently treated with one cycle of chemotherapy for AML. The post chemotherapy CT scan which was conducted four months later, did not show any residual mass in the orbit or pelvis. She remains in clinical remission two years after treatment, with no relapse or progression to AML. Her vision also remains normal bilaterally.

DISCUSSION

Children with MS commonly present with skin manifestation (54%) followed by the orbital involvement (3). Ocular signs in orbital MS include acute pain, proptosis, and abnormal bleeding due to the tumour mass infiltrating the extraocular muscles and the surrounding structures, such as the ethmoid and maxillary sinus.

In general, the investigations for orbital tumours include radiological imaging and histopathological examination of a biopsy specimen. The usefulness of magnetic resonance imaging or CT scan in the diagnosis of orbital MS is limited, as it is not sensitive enough to distinguish between this granulocytic neoplasm from other tumours. However, it may be indicative of MS when a contrast-enhancing solid mass is seen on the CT scan at different sites or at different points in time (4).

In general, histological examination of orbital tumours is the only way to accurately establish the diagnosis of most orbital tumours. MS falls under the group of SBRCT, which is characterised by cells with a predominant nuclei and scanty cytoplasm on histology. Ophthalmologically-related SBRCT includes neuroblastoma, myeloid sarcoma, non-Hodgkin's lymphoma, rhabdomyosarcoma, and Wilms' tumour (5). IHC examination is essential to distinguish between the various SBRCT, and can diagnose MS with almost 100% accuracy. However, the pathologist must have a high index of suspicion to run the specific biomarker panel for this rare malignancy. The correct diagnosis of MS de novo is crucial as aggressive systemic chemotherapy has been proven to reduce the risk of progression to AML, which usually occurs within the first 12 months of diagnosis (3).

CONCLUSION

It is essential for clinicians to know that bilateral orbital lesions in children strongly suggest the presence of a metastatic malignant lesion, and that prompt systemic workup is needed to identify the primary tumour. This case highlights the importance of considering MS as a differential diagnosis of multifocal tumours involving the orbit. As most metastatic orbital lesions consist of SBRCT, IHC examination plays a crucial role, not only for diagnostic purposes, but also as a prognostic indicator and identifying the potential target for therapy.

ACKNOWLEDGEMENT

The authors like to thank the patient and her parents for the kind permission in the publishing of this case report.

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