Clinical Challenge | RADIOLOGY

Progressive Left Periorbital Swelling

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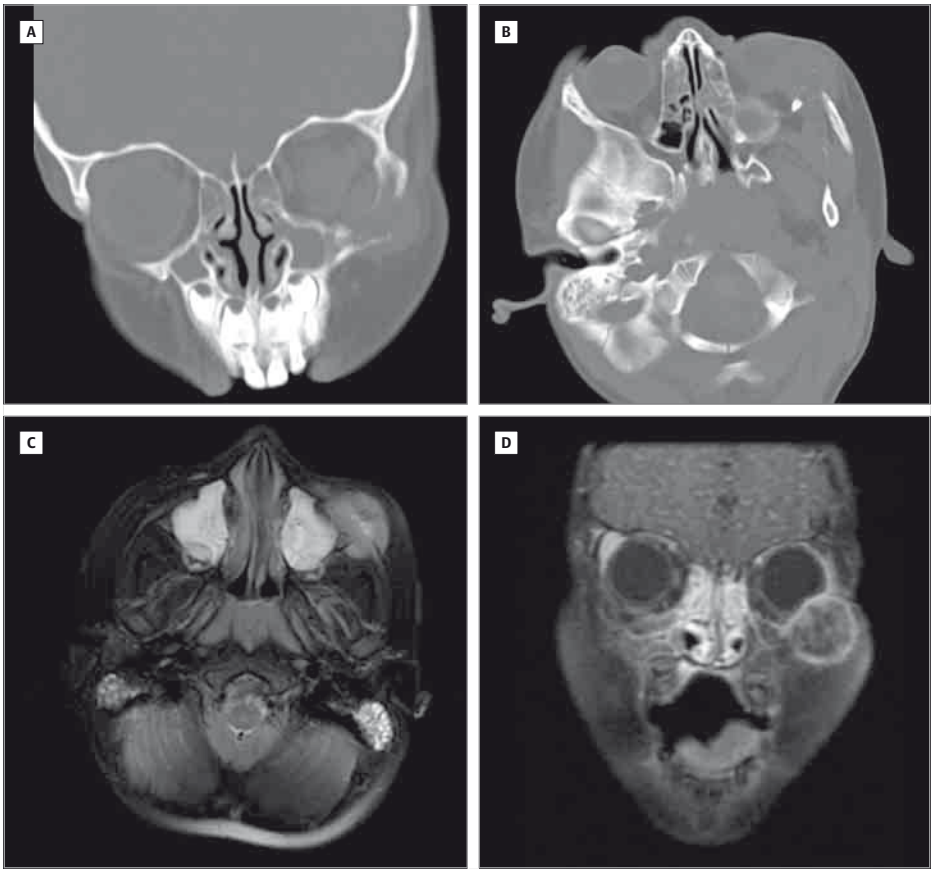

Figure. A and B, Computed tomographic images of the maxillofacial region showing a round, soft-tissue mass. A, Coronal view. B, Axial view. C and D, Magnetic resonance imaging (MRI) scans of the maxillofacial region; C, T2-weighted MRI scan, axial view. D, Postcontrast T1-weighted MRI scan, coronal view.

A young boy with mild developmental delay and autism presented with a 1-week history of progressive left periorbital swelling. His parents reported that he complained of left orbital pain just before they noticed the swelling, and he was treated with cold compresses with only slight improvement. He had never experienced these symptoms before. The patient had not experienced diplopia, change in visual acuity, nasal obstruction, change in oral intake, weight loss, recent upper respiratory infection, sick contacts, fevers, or chills. At the time of presentation, the patient was breathing comfortably and was afebrile. Findings from his head and neck examination were significant for a firm mass over the left zygoma and lateral infraorbital rim, 2 × 3 cm in diameter. The mass was nontender, nonerythematous, and fixed to the underlying zygoma and lateral infraorbital rim. There was no cervical lymphadenopathy. Ophthalmology examination revealed intact extraocular movements, bilateral visual acuity of 20/50, and no evidence of afferent papillary defect. Computed tomographic (CT) images showed a round, soft-tissue mass with osseous destruction and erosion into the zygoma, infratemporal fossa, inferolateral orbit, and maxillary sinus (**Figure**, A and B). Concern for orbital involvement prompted magnetic resonance imaging (MRI) of the maxillofacial region (Figure, C and D). He was taken to the operating room the following day for open biopsy.

WHAT IS YOUR DIAGNOSIS?

A. Metastatic neuroblastoma

B. Rhabdomyosarcoma

C. Lymphoma

D. Lipoma

Diagnosis

B. Rhabdomyosarcoma

Discussion

Rhabdomyosarcomas are rapidly growing, aggressive neoplasms in children, representing 4% to 8% of all malignant neoplasms in children younger than 15 years. Rhabdomyosarcomas are more common in white males, with a peak incidence at ages 2 to 6 years.[1,2] Rhabdomyosarcomas originate from the embryonic mesenchymal precursor of striated muscle, and the diagnosis is largely based on immunohistochemical staining.[3] Histologically, rhabdomyosarcomas are divided into 5 groups: embryonic (58%), alveolar (31%), botryoid (6%), pleomorphic (4%), and undifferentiated (1%).[4]

In our patient, computed tomographic images showed a 2.7 × 2.1 × 2.2-cm round, soft-tissue mass with bony erosion of the left zygoma and orbit. The exact site of origin was unclear. It was difficult to determine if the lesion was a bony metastatic lesion or a soft-tissue lesion with surrounding bony erosion. The mass displaced the globe medially, but there was an intact soft-tissue plane between the mass and the globe (Figure, A and B). Owing to the extensive destruction of the inferior and lateral orbit, the patient also underwent MRI. On T2-weighted axial MRI, the mass showed intermediate signal intensity (Figure, C). On postcontrast T1-weighted coronal MRI, the mass was hypointense with irregular enhancement (Figure, D).

Rhabdomyosarcoma has variable reported imaging findings. Owing to the rarity of head and neck rhabdomyosarcoma, most CT and MRI findings are derived from small case series. Furthermore, different subtypes may have different radiographic findings.[5] They are commonly isodense or slightly hypodense on precontrast CT images with homogeneous enhancement on postcontrast CT images. They are isointense or variably hyperintense on T1-weighted MRI. On T2-weighted MRI, they are moderate to markedly hyperintense with homogeneous and heterogeneous enhancement.[5]

The differential diagnosis also included metastatic neuroblastoma, Langerhans cell histiocytosis, Ewing sarcoma, lymphoma, and osteosarcoma. Neuroblastoma is the third most common pediatric malignant neoplasm, with 25% of patients presenting with metastases to the orbit with bony destruction.[6] Both Langerhans cell histiocytosis and metastatic neuroblastoma characteristically involve the posterolateral part of the orbit.[6] This patient's tumor was not in this characteristic location, although neuroblastoma metastases to the maxilla have been reported.[7] Ewing sarcoma of the orbit is rare.[8] On CT, Ewing sarcoma appears as a diffuse, unevenly enhancing mass associated with mottled bone destruction, making the exact site of origin difficult to determine.[5] Lymphoma of the orbit is hyperdense on CT, hypointense on T1-weighted MRI, and hypointense to hyperintense on T2-weighted MRI with homogeneous enhancement. Typically, lymphoma molds to rather than destroys the orbital rim.[9] Any portion of the orbit may be affected.[9]

Among these differential diagnoses, there are no pathognomonic imaging findings. Further imaging with ultrasound or positron emission tomography/CT has not proven to significantly contribute to diagnosing these tumors. Biopsy is necessary for diagnosis. This patient underwent a sublabial approach for open biopsy. The frozen specimen revealed a small round blue cell tumor. The tumor stained strongly for muscle markers desmin, myogenin, and myoD1. Myogenin and MyoD1 identify nuclear proteins and are sensitive and specific for rhabdomyosarcoma.[4] It was negative for NB84 and chromogranin, markers for neuroblastoma. The final pathologic diagnosis was embryonal rhabdomyosarcoma.

Treatment of rhabdomyosarcoma has evolved over the past 25 years to multidisciplinary protocols consisting of surgery, radiation, and various chemotherapy regimens. In this case, extensive orbital erosion prevented primary surgical resection owing to concern for significant functional morbidity and incomplete resection. The use of risk-adapted multidrug chemotherapy combined with radiotherapy and surgical excision when possible has improved the overall 5-year survival rate for pediatric head and neck rhabdomyosarcoma from 25% in 1970 to 74% in 1997.[10] The patient was referred for chemotherapy and proton beam radiation.

ARTICLE INFORMATION

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