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Melanotic Neuroectodermal Tumor of Infancy: A Case Report with Review of Literature

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Abstract

Melanotic neuroectodermal tumors of infancy are rare pigmented neuroectodermal locally aggressive tumors that usually occur within 1 year of life. Head and neck areas are frequently involved with maxilla being the most common site. Expansile growth and high recurrence rate are characteristics of this cancer. Though rare, radiologists and clinicians must be aware and should consider this entity when diagnosing pediatric head and neck masses. Computed tomography can be used for diagnosing and looking at the extent of the disease and predicting the operative outcome. Surgery with wide resection margin is the mainstay of treatment for these masses. Surgery followed by adjuvant chemotherapy and radiation is reserved for aggressive malignant lesions. Since it has a high recurrence rate, follow-up is done with imaging and clinical examination. Early detection and treatment of recurrence have a favorable outcome for the patients.

Keywords
- melanotic neuroectodermal tumors of infancy
- melanotic progonoma
- imaging
- CT

Introduction

Melanotic neuroectodermal tumors of infancy (MNTIs) are rare tumors that typically affect infants under the age of one and are capable of mimicking other benign and malignant skull tumors in infants.[1] The maxilla and cranial vault are the two areas where tumors are most frequently found.[2] Complete resection with negative margins is not always feasible due to close proximity to critical structures in the head and neck so it has considerable recurrence risk, reported to be as high as 45%.[3] MNTIs are often confused with benign lesions such as dermoid or epidermoid cysts, vascular lesions and sometimes, malignant lesions like small round cell tumors, and must be distinguished by the recognition of their diagnostic features.

Case History

A female child of 6 months born out of nonconsanguineous marriage presented with symptoms of gradually increasing mass in the right cheek for the last 2 months. The child was breathing from the mouth for 10 to 15 days. There was no history of fever or bleeding from any site or the mass. On examination, there was a mass involving the right cheek. It had a smooth surface with few vessels on the surface. The mass was protruding through the oral cavity. The soft palate was pushed down with obliteration of the right nasal cavity. Contrast-enhanced computed tomography (CT) as shown in ►**Fig. 1** showed an expansile sclerotic mass involving bilateral maxillary bones with nasal extension. The soft tissue component showed homogeneous post-contrast enhancement. It had sinonasal extension and bone erosion. No intraorbital extension or pathological neck nodes were observed. Urinary vanillylmandelic acid levels were done outside our institute and were normal. In the given clinical setting and after reviewing the radiological features, several differential diagnoses were considered including rhabdomyosarcoma, Ewing sarcoma, and MNTI. Absence of cervical

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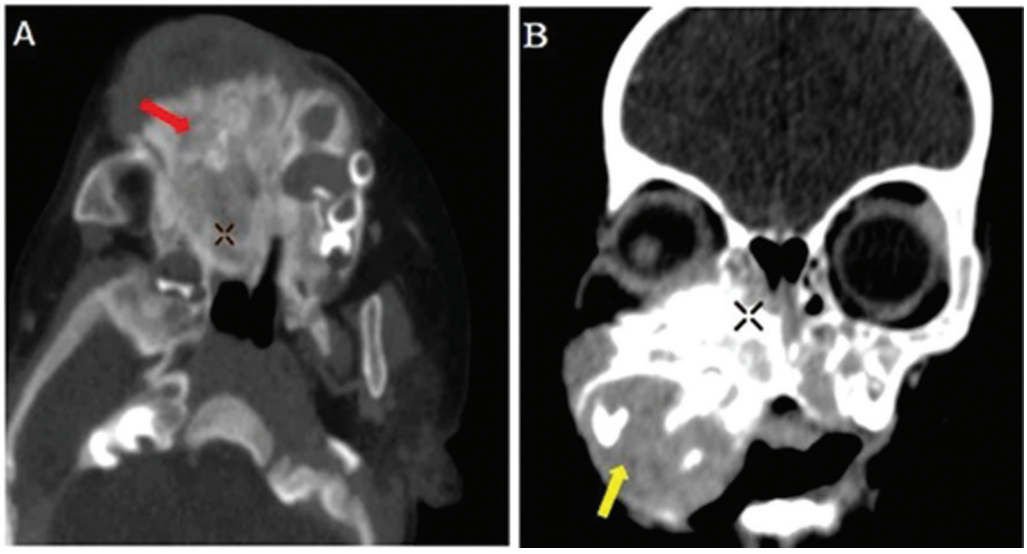

Fig. 1 (**A**) Axial computed tomography (CT; bone window) shows expansile sclerotic mass (red arrow) involving bilateral maxillary bones with nasal extension. (**B**) Coronal reformatted CT image shows expansile mass involving the maxillary bone right > left, with accompanying exophytic enhancing soft tissue (yellow arrow).

nodes made the diagnosis of rhabdomyosarcoma less likely. Ewing sarcoma presents as a permeative lytic lesion with aggressive periosteal reaction, which was absent in this case.

The patient was planned for biopsy. Histopathological examination of the maxillary mass showed malignant round cell tumor infiltrating bone and muscle. Scattered large cells with more abundant cytoplasm were noted. Some cells showed melanin pigment. On immunohistochemistry, scattered larger cells were positive for AE1/AE3, and tumor was diffusely positive for synaptophysin while being focally positive for HMB45, MIC2 (dot-like). Tumor was negative for FLI1, LCA, desmin, CD43, TdT, CD10, MYOD1, and melanA. In view of its solitary nature and absence of distance metastases, the patient underwent right total and left partial maxillectomy with anterolateral thigh flap. Currently, she is on follow-up with no evidence of the disease.

Discussion

A rare subtype of locally aggressive pigmented neoplasms, the MNTI occurs in children under the age of 6 months. Ninety-five percent cases occur at less than 1 year of age. However, some have been reported in adults too.[4] MNTI was first described by Krompecher in 1918. There is no sex predilection. Though it has been found in the testis, epididymis, soft tissue, mediastinum, uterus, ovary, thigh, and central nervous system, the majority occurred in the head and neck region (92.8%).[5] Among the most commonly involved sites, the order of frequency is the maxilla, followed by skull, mandible, and brain.[6] Neuroectodermal origin of the tumor is supported by elevated urinary levels of vanillylmandelic acid in some of the MNTIs. MNTI has a high recurrence rate (10–15%).[4] Recurrence usually occurs within 1 or 2 months after primary surgical excision. Recurrences are caused by incomplete removal, surgical dissemination of neoplastic cells, and multicentricity. Also referred to as melanotic progonomas, these can show malignant conversion rarely. Metastases have been reported in the lymph nodes, bone marrow, pleura, liver, pelvis, and adrenal glands.[7] Lesions arising from cerebellum are more aggressive and can show metastases to meninges, brain, and spinal cord. Congenital melanocarcinoma, retinal anlage tumor, pigmented congenital epulis, tumor, and melanotic progonoma are some of its synonyms.[2]

The tumor grows rapidly. It infiltrates the bone marrow without formation of a capsule. Clinically, the lesion is usually painless, solitary, firm, sessile, lightly pigmented (blue or black), nontender, and nonulcerated tumor. They are typically asymptomatic, but parents may detect them because of an increase in middle-face bulk and asymmetry. In some instances, increased vanillyl mandelic acid levels in the urine are noted, supporting neuroectodermal origin.[6]

Though melanin granules are a feature of many neoplasms, the ultrastructure of MNTI under electron microscope helps in its characterization.[8] Microscopically, these are biphasic tumors, characterized by smaller neuroblast-like round cells and larger polygonal epithelioid cells with melanin deposits that resemble melanocytes.[6] The differential diagnosis includes rhabdomyosarcoma, neuroblastoma, primitive neuroectodermal tumor, Ewing sarcoma, malignant melanoma, and lymphoma. Radiograph shows a lytic lesion with or without irregular margins.

On CT scans, they present as expansile tumors that expand and destroy the cortical bone, sometimes in conjunction with spiculated/sunburst periosteal reaction and may show floating teeth. The soft tissue component may appear hyperdense on noncontrast scan, due to melanin. However, hypodense lesions are also observed. In our case, the expansile mass showed maxillary erosion with sinonasal extension, along with homogenous post-contrast enhancement.

On T1- and T2-weighted magnetic resonance imaging (MRI) sequences, MNTIs appear isointense or hypointense

with homogenous post-contrast enhancement. Hyperintensity on T1-weighted images may be seen owing to the large amount of melanin. T1 and T2 hypointense areas may be seen due to calcifications and hyperostosis. MRI was not done in our case, as CT evaluation was enough to provide us with a differential, and biopsy done later helped provide us with a definitive diagnosis. Hyperintense T1 soft tissue mass would have potentially favored a melanotic component of disease that would have helped narrow our differential, but since biopsy provided us with a definitive diagnosis, additional MRI could be avoided.

Conclusion

MNTIs are uncommon neuroectodermal neoplasms with unknown behavior, but they exhibit a pattern that can be used to differentiate them from other head and neck space-occupying lesions in pediatric patients. A competent pathologist is essential for a timely diagnosis that allows for the excision of the tumor, preventing complications and local recurrence. Hence, it needs to be emphasized that despite its low incidence, the clinicians and radiologists need to keep this differential in the back of their mind, to administer treatment at the earliest. Owing to its risk of recurrence, proper treatment and close clinicoradiological follow-up are of paramount importance.

Patient Consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for images and other clinical information to be reported in the journal. The patient understands that name and initials will not be published and due efforts will be made to conceal the identity, but anonymity cannot be guaranteed.

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Conflict of Interest

None declared.

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