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Melanotic Neuroectodermal Tumor of Infancy: A Rare Case Report

Authors
Dr Karuna Gupta[1], Dr Priyanka Sharma[2], Dr Deepika Hemrajani[3], Dr Krishna Sireesha Sundaragiri[4], Dr Ajay Yadav[5]
[1]Professor, Pathology, SMS Medical College, Jaipur
[2]Senior Resident, Pathology, SMS Medical College, Jaipur
[3]Associate Professor SMS Medical College, Jaipur
[4]Assistant Professor at RUHS Dental College of Dental Sciences, Govt. Dental College of Jaipur
[5]Professor and Head, Department of Pathology, SMS Medical College, Jaipur, India

Introduction

Melanotic Neuroectodermal Tumor Of Infancy (MNTI) is a rare, benign, pigmented neoplasm arising from neural crest.[1,2] Clinical presentation is often painless, locally aggressive, rapidly expansile growth causing facial disfigurement.[2] In the literature, neoplasm has been known by various synonyms like congenital melanocarcinoma, retinal analage tumor, pigmented congenital epulis and melanotic progonoma.[1,3] This lesion is found mainly in children below 1 year of age. Rare cases have been reported in older children and adults also.[2,3] Gender predilection is not seen through literature.[4] 93% of MNTI occurred in head and neck region with unique predilection for anterior maxilla, although cases including brain, skull, epididymis have also been reported.[5] MNTI has a recurrence rate of 15% and show malignant differentiation in 6.6% cases, hence long term follow up after surgical excision is required.[6] Plain radiographs show expansile areas of translucency with poor circumscription probably as result of rapid tumor growth and local aggressiveness.[5] The present case report describes a case of MNTI in a 1.5 month old male child along with brief review of literature and pathological differential diagnosis.

Case Report

1.5 month old male baby presented with 15 days history of firm, painless, rapidly growing left maxillary swelling. There was history of normal pregnancy and delivery and no medication during pregnancy. On intraoral examination, blue-pink coloured firm swelling was seen measuring approximately 3x3 cm in left side maxilla. Dilated veins were seen over the swelling. Upper lip was stretched and lifts up. Provisional diagnosis of dentigerous cyst, vascular malformation or hemangioma was made. Computed tomography scan shows homogenous translucent lesion in the maxillary bone on left side. Surgical excision was done under general anaesthesia.

Clinical differential diagnosis of jaw swelling in infant are usually benign odontogenic cysts and congenital epulis although neoplasm with rapid growth includes embryonal rhabdomyosarcoma, Ewings sarcoma, neuroblastoma, melanoma, burkits lymphoma and teratoma.

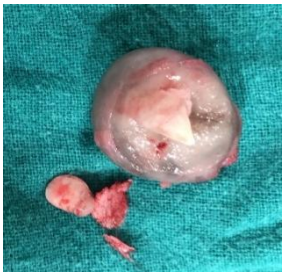
Urinary Vanillylmandelic Acidwas normal (2.8 mg/24 hours).

The gross examination of the specimen revealed 3x3 cm, firm, slight blue coloured tumor with single developing tooth attached. Cut surface of the tumor was blue-black due to pigmentation.

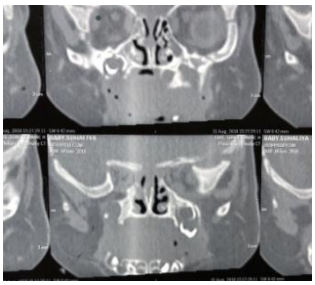
Microscopic examination shows a biphasic population of cells arranged in nests and clusters, separated by fibrovascular stroma. The two population comprised of large melanotic (pigmented) cells and small round (non-pigmented) cell with scant cytoplasm. Large epithelial cells contain melanin pigment. The small round cells have features of primitive neuroepithelial cells. Based upon these findings tumor was dignosed as MNTI. Immunohistochemical marker study (IHC) was done for confirmation.

IHC revealed positive staining for HMB-45 in the melanin containing large epithelial cells and negative staining in small round cells. Expression of synaptophysin was predominantly in small neuroblastic cells but large epithelial cells also show focal positivity. S-100 was negative which helps to rule out melanoma. Cytokeratin was positive.

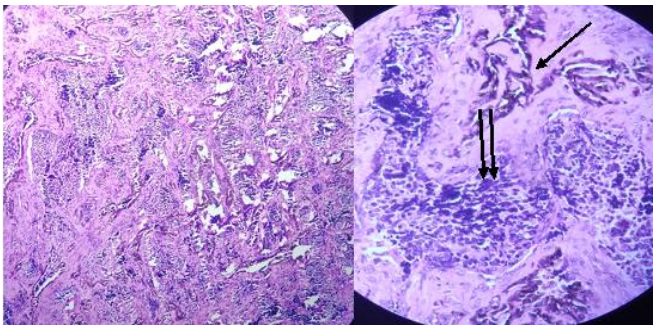
(a) Clinical photograph showing swelling in left maxillary region.



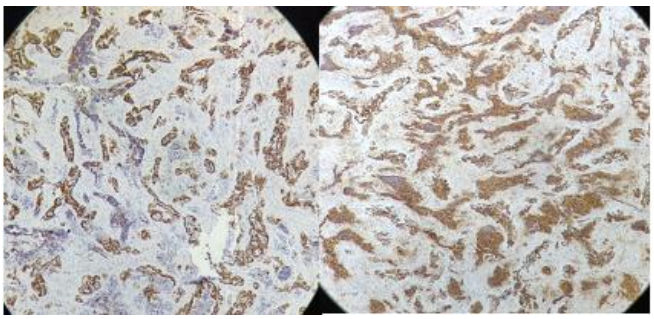
(b) Image showing excised specimen with single deciduous incisior.



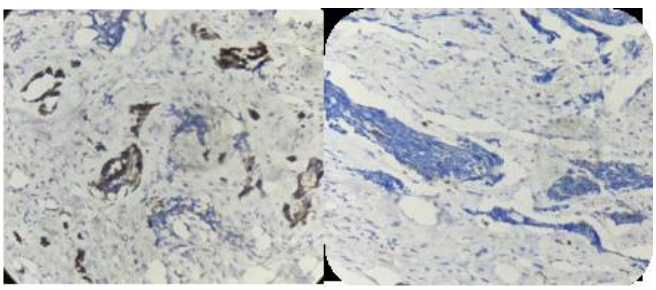
(c) Enhanced CT scan show homogenous mass in the left maxilla.



(d) & (e) H&E stain shows dual population of cells. Large melanin containing pigmented cells (single arrow) and small round monomorphic neuroblastic cells (double arrow).



(f) HMB-45 positivity in large polygonal melanin containing cells. (g) Positive synaptophysin staining in small neuroblastic cells.



(h) & (i) Focal cytokeratin positive staining and negative S-100 staining.

Discussion

MNTI is a rare neoplasm affecting maxilla followed by calvaria and mandible.[1] The origin of tumor is controversial. Rarely it can arise in the skull, mandible, brain, meninges, retina, mediastinum, bone, soft tissue, testies and epididymis.[1-4] Krompecher in 1918 reported a case of pigmented tumor of maxilla associated with developing tooth in 2 month old baby and reported as congenital melanocarcinoma.[2,8] other designations for the same neoplasm was retinal analage tumor, pigmented ameloblastoma, pigmented epulis and melanotic progonoma.[1,9,10] Borello and Gorlin in 1966 first coined the term Melanotic Neuroectodermal Of Infancy based on IHC, ultrastructure, electron microscopic studies and a high urinary VMA levels.[11] High urinary VMA level suggest neural crest origin of tumor. Many case reports with normal urinary VMA levels also reported in literature.[3] The IHC and ultrastructural features of MNTI cells make them very similar to neural crest cell origin. Diagnosis before surgical excision and distinction from other paediatric small round cell tumor (rhabdomyosarcoma, neuroblastoma, melanoma and lymphoma) is mandatory to plan the complete surgical resection and reducing the possibilities of tumor recurrence.[12] classic clinical presentation is rapidly growing, painless, expansile, non ulcerated, unencapsulated partly pigmented mass, typically in the maxillary region. It has tendency for local invasion therefore aggressive surgical approach consisting of complete surgical excision and long term follow up is recommended. Microscopically MNTIs are biphasic tumors with one cell population consisting of cuboidal epithelial cells with open vesicular nuclei arranged in tubular and alveolar pattern. These cells have abundant melanin pigment. Another cell type is small dark round cells with hyperchromatic nucleus and minimal cytoplasm, arranged in nests within the fibrovascular stroma. It has appearance of neuroblast.

IHC markers are helpful in differentiating MNTI from embryonal rhabdomyosarcoma (desmin and myoglobin positive), Melanoma (s-100 positive, while negative in MNTI), EWING sarcoma (CD99 and FLI 1 positive), lymphomas (LCA positive).

MNTIs are reactive for HMB-45 and synaptophysin both indicate melanocytic and neuroblastic differentiation of the cells (biphasic).

Conclusion

MNTI is rare benign paediatric neoplasm with tendency for local invasion and recurrence. Exact diagnosis by histomorphology combining with IHC is recommended. Complete surgical excision and long term follow up is required.

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