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Case Report

Melanotic neuroectodermal tumour of infancy in maxilla of a 5-month-old infant: A case report

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Abstract

Melanotic neuroectodermal tumour of infancy (MNTI) is a rare biphasic tumour composed of small neuroblast-like cells and larger melanin-producing epithelial cells. Most tumours (>90%) occur in the craniofacial bones where maxilla is the most common site (> 60%) as was in our case, followed by skull (~15%) and mandible (~8%); soft tissue tumours are rare. Most common presentation is as a sessile, painless, rapidly enlarging mass in the upper alveolus which causes facial deformity and feeding disruption. We are presenting an interesting case of MNTI which came to our institute.

Keywords: Melanotic neuroectodermal tumour of infancy, Biphasic tumour, Maxilla.

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1. Introduction

Melanotic neuroectodermal tumour of infancy (MNTI) is a rare biphasic tumour composed of small neuroblast-like cells and larger melanin-producing epithelial cells.[1] It was first described by Krompecher in 1918 who named it as congenital melanocarcinoma.[2] Most of the tumours (>90%) involve the craniofacial bones where maxilla is the most common site (>60%) as was in our case, followed by skull (~15%) and mandible (~8%). It rarely involves the soft tissue. Very rare sites are testis and epididymis, ovary, uterus, mediastinum, scapula and the bones and soft tissues of the extremities. There is a slight male predilection. The typical manifestation is a sessile, painless, rapidly enlarging mass in the upper alveolus, causing facial deformity and feeding disruption. The mass usually has a bluish-black appearance due to its melanin content. Some tumours produce vanillylmandelic acid (VMA) (34%).[1] Surgical resection with wide margin is the treatment of choice with or without adjuvant chemotherapy.[3] Its main differential diagnoses are neuroblastoma and alveolar rhabdomyosarcoma.[4]

2. Case Details

A 5 months old male infant was brought with the complaint of mass in maxilla, feeding difficulty and facial deformity. An MRI and trucut biopsy were performed.

Imaging MRI showed an ill-defined lobulated altered signal intensity mass lesion involving body of maxilla and maxillary sinus on left side. The size of the lesion was approximately 4.2 x 4.1 x 6.2 cm (AP x TR x CC). Anteriorly the lesion was found extending into premaxillary soft tissue, but overlying skin was free. Inferiorly the lesion extended to left half of superior alveolar process of maxilla and adjacent hard palate with exophytic component in the oral cavity. Superiorly, the lesion extended through floor of orbit, while superomedially, there was extension into adjacent ethmoidal air cells and nasal cavity. A neoplastic etiology was suggested with differentials including small round cell tumour and rhabdomyosarcoma.

Microscopic examination of trucut biopsy showed cores of fibrocollagenous tissue and bony spicules with infiltration by nests of biphasic tumour cells. One population was of

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small round cells with hyperchromatic nuclei and scant cytoplasm. Second population was of larger epithelial cells having round nuclei, vesicular chromatin and containing melanin pigment [**Figure 1**]. Diagnosis was given as Melanotic neuroectodermal tumour of infancy (MNTI). Immunohistochemistry (IHC) was done and we found that both small and large cells were positive for Synaptophysin, large cells were positive for AE1 and HMB45, while small cells showed positivity for CD56. GFAP came positive, suggesting glial differentiation. Other markers like NKX2.2 were negative. Hence a final diagnosis of Melanotic neuroectodermal tumour of infancy (MNTI) was rendered.

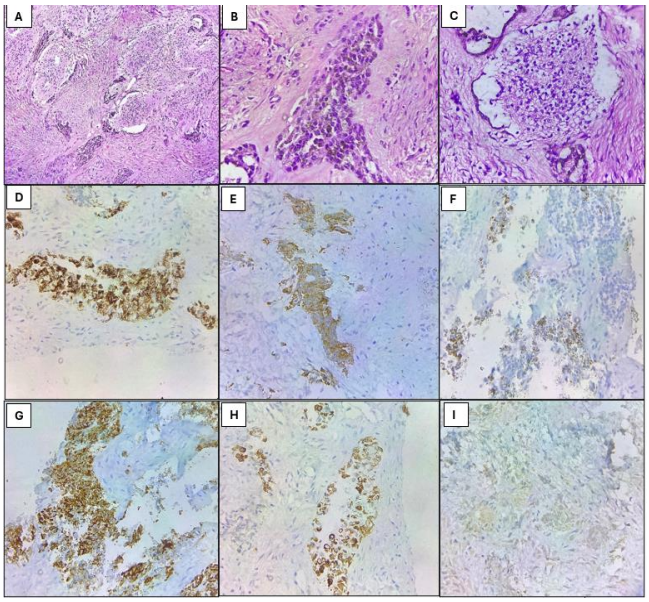

Figure 1: A: Biphasic population comprising of small neuroblast like cells and larger melanin-producing epithelial cells in fibrocollagenous stroma (H&E, ×100); **B:** Larger pigmented cells (H&E, ×400); **C:** Small round neuroblast-like cells with neurofibrillary matrix (H&E, ×400); **D:** HMB45 positive larger epithelial cells (×400); **E:** Synaptophysin positive larger epithelial cells (×400); **F:** GFAP positive area showing glial differentiation (×400); **G:** CD56 positive small cells (×400); **H:** AE1 positive larger cells (×400); (I) NKX2.2 negative in tumour cells (×400)

Subsequently a total maxillectomy of left side was performed and the specimen was received in the Department of Oncopathology, GCRI. Grossly, it was a firm to hard well circumscribed mass with a tooth impacted measuring approximately 5.8 x 4 x 4 cm. On cut section, the tumour had firm consistency and showed blue black pigmentation [**Figure 2**]. Necrosis was not seen and overlying skin was grossly unremarkable. The relevant margins and tumour bits were submitted. Microscopic examination showed a biphasic population comprising of small neuroblast like cells and larger melanin-producing epithelial cells arranged in alveolar nests, cords and trabeculae and infiltrating a dense, vascularized, fibrocollagenous stroma. The melanotic epithelial cells typically surrounded the small cells forming tubuloglandular structures at places. Mitoses and necrosis were absent.

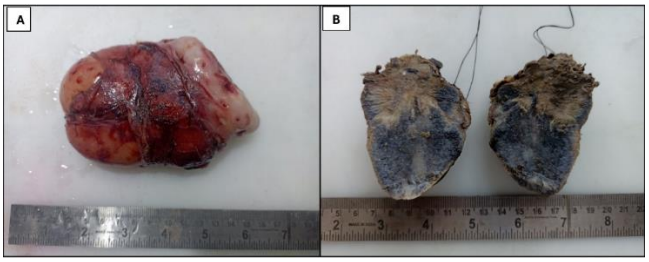

Figure 2: A: Resected specimen of the tumour; **B:** Cut section showing blue black pigmentation

3. Discussion

MNTI was first described by Krompecher in 1918, who named it congenital melanocarcinoma.[2] Terminologies previously used for this tumour like Melanotic Progonoma or Retinal anlage tumour are not recommended by WHO.[1] In 1966, MNTI was proposed to be of neural crest origin, because of the discovery that many cases are associated with an increase of urinary vanillylmandelic acid excretion. This finding also helped to explain the biphasic population of melanocytic and primitive neuroectodermal cells, both of which are derived from the neural crest.[5]

MNTI is a rare clinically benign tumor in infants but it is notorious for being locally aggressive and also known to recur.[6] There is a slight male predilection and our case was also a male infant.[7]

MNTI needs to be differentiated from other malignant childhood tumours like neuroblastoma and alveolar rhabdomyosarcoma,[4] as were given as probable diagnoses on MRI in our case, hence microscopic examination along with IHC is mandatory for confirmatory diagnosis.[8]

So far only around 450 to 500 cases have been reported, making it a rare tumour, but owing to its locally destructive nature and tendency to recur, it is important to identify this entity.

4. Conclusion

Melanotic neuroectodermal tumour of infancy is a rare entity. Its diagnosis is very important because its albeit benign, yet notorious for its locally destructive and recurrent nature.

5. Source of Funding

None.

6. Conflict of Interest

There is no conflict of interest.

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