Case Report

A Rare Case of Melanotic Neuroectodermal Tumor of Infancy: Histopathological and Immunohistochemical Features

Shuchi Ghai[1,*], Tanvi Arora[2], Ajay Malik[2], Arvind Tyagi[3]

[1]Yashoda Hospital and Research Centre, III M, Nehru Nagar, Ghaziabad, India
[2]Department of Pathology, Yashoda Hospital & Research Centre, Ghaziabad, India
[3]Department of Oncosurgery, Yashoda Hospital & Research Centre, Ghaziabad, India

***Correspondence:** drshuchichopra@gmail.com

DOI
10.21276/apalm.3751

Article History
Received: 13-11-2025
Revised: 19-01-2026
Accepted: 11-02-2026
Published: 02-03-2026

How to cite this article
Ghai S, Arora T, Malik A, et al. A Rare Case of Melanotic Neuroectodermal Tumor of Infancy: Histopathological and Immunohistochemical Features. Ann Pathol Lab Med. 2026;13(3):C77-C80.

Abstract

Melanotic neuroectodermal tumor of infancy (MNTI) is a rare, pigmented neoplasm of neural crest origin occurring predominantly in infants under one year of age. Although histologically benign, it exhibits locally aggressive behavior and can mimic malignancy due to rapid growth and bone destruction. Early recognition is essential for appropriate management. Case Report: We present a case of a 3-month-old male infant from Uttar Pradesh who presented with a bluish intraoral swelling involving the left side of the mandible. Computed tomography revealed a well-defined expansile lesion with mild enhancement in the body of left mandible. Serum vanillylmandelic acid (VMA) levels were within normal limits. Histopathological examination showed a biphasic tumor composed of large melanin-containing epithelial-like cells and small neuroblast-like cells arranged in nests and alveolar patterns. Immunohistochemistry demonstrated positivity for HMB-45, CD-99 and synaptophysin whereas S-100, desmin and WT1 were negative. A final diagnosis of *melanotic neuroectodermal tumor of infancy* was rendered. Complete surgical excision was performed, and postoperative recovery was uneventful. The patient remains under follow-up with no recurrence to date. Conclusion: *MNTI* is an uncommon but distinct entity that should be considered in the differential diagnosis of pigmented maxillary swellings in infants. A combination of histopathological and immunohistochemical findings is crucial for accurate diagnosis. Early surgical intervention with adequate margins provides an excellent prognosis.

Keywords: melanotic neuroectodermal tumor of infancy; *mnti*; maxilla; neural crest

Introduction

Melanotic neuroectodermal tumor of infancy (*MNTI*) is a rare, fast-growing, pigmented neoplasm of neural crest origin. It affects the craniofacial skeleton of the infants, maxilla being the most involved site, during the first year of life. Very few cases of these tumors have been reported. The tumor is seen to arise commonly in maxilla (70%) and less commonly in skull and Mandible [1]. On histopathology it is commonly confused with neuroblastoma and alveolar rhabdomyosarcoma. Proper histopathological and immunohistochemical evaluation can help in diagnosing the case accurately [2].This tumor should be included in the differential diagnosis of head and neck neoplasms in infancy. We present here a case of a newborn with *MNTI* involving the mandibular region along with its clinical, pathological, therapeutic, and prognostic aspects. The treatment included left sided hemimandibulectomy. The patient had no recurrence.

Case Report

A 3-month-old male infant from Uttar Pradesh presented with a firm, bluish-tinged intraoral swelling involving the left side of the mandible. The swelling was initially noticed at the age of one month by his mother which progressively increased in size. The child was brought to our hospital. The growth of the neonate was adequate for his age.

On examination it revealed non ulcerated tumor with bluish discoloration measuring 5.5x3x1.5 cms. Mucosa was intact (Fig. 1). On palpation it was firm, non-tender and non-fluctuant. The routine laboratory investigations were found to be normal. Contrast computed tomography (CT) of the skull was done. The CT scan showed an expansile, lytic lesion showing minimal enhancement in the body of left mandible with severe thinning of cortex and solid periosteal reaction. Serum vanillylmandelic acid (VMA) was found to be normal, urinary VMA level could not be done as patient was taken for biopsy and frozen section was planned.

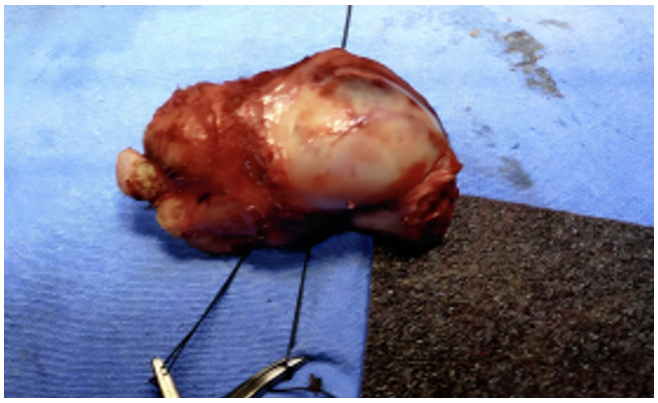

Figure 1: Intraoperative image depicting non ulcerated tumor with bluish discoloration measuring 5.5x3x1.5 cms with intact mucosa.

Frozen section was planned and performed on tissue bits which showed small round blue cells. Further core biopsy was done in which possibilities considered were *MNTI*, Neuroblastoma and Primitive neuro ectodermal tumor. Histopathologic examination of the excised core tissue biopsy revealed biphasic pattern of cell distribution with smaller round cells with scant cytoplasm and hyperchromatic nuclei in a fibrillar & vascularized fibrocollagenous background and cell population consisting of larger cells with vesicular nuclei and eosinophilic cytoplasm containing brownish pigment which clustered in alveolar and pseudoglandular patterns (Fig. 2). The cellular heterogeneity probably arises from the dual mesodermal and ectodermal characteristics expressed by neural crest cells as they progress through various developmental stages [3].

Immunohistochemical confirmation of the tumor for neural origin in our case was also done. The pigmented larger cells remained pigmented and showed focal positivity on HMB45 clone HMB45, DBS and synaptophysin clone GR007/SYPP, DBS (Fig. 3). Immunohistochemical analysis was also done with pancytokeratin, CD45 (clone 2b11/pd7-26, DBS), WT1 (clone EP122, DBS), CD99 (clone HO36-1.1, DBS), Ki67 (clone SP6, DBS), S100(clone EP-32, DBS), Desmin(clone GM007, DBS), SMA(clone IA4, DBS) and FLI 1(clone NX2/294, DBS), which showed clear negative result. Negative result with CD99 and FLI-1 ruled out Ewings sarcoma/PNET and positive result for cytokeratin and HMB-45 ruled out Rhabdomyosarcoma and neuroblastoma which also lacks biphasic pattern of tumor cell arrangement and pigmented epithelioid cells. Negative result with CD45 rules out lymphoma. Ki 67 labeling index was found to be 35%. This histopathologic and immunohistochemistry examination of the tissue confirmed the diagnosis of *MNTI*.

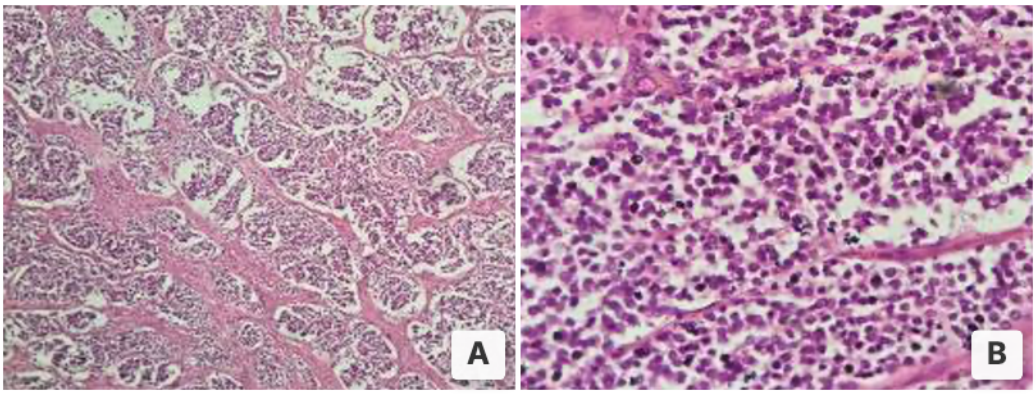

Figure 2: (A) Biphasic pattern of cell distribution with smaller round cells with scant cytoplasm and hyperchromatic nuclei in a fibrillar & vascularized fibrocollagenous background. (B) Cell population consisting of larger cells with vesicular nuclei and eosinophilic cytoplasm containing brownish pigment which clustered in alveolar and pseudoglandular patterns.

Left sided hemimandibulectomy was done and submandibular lymph nodes were removed under general anesthesia. The infant tolerated the surgical procedures well and was discharged on the 7th postoperative day. The removed mass was sent

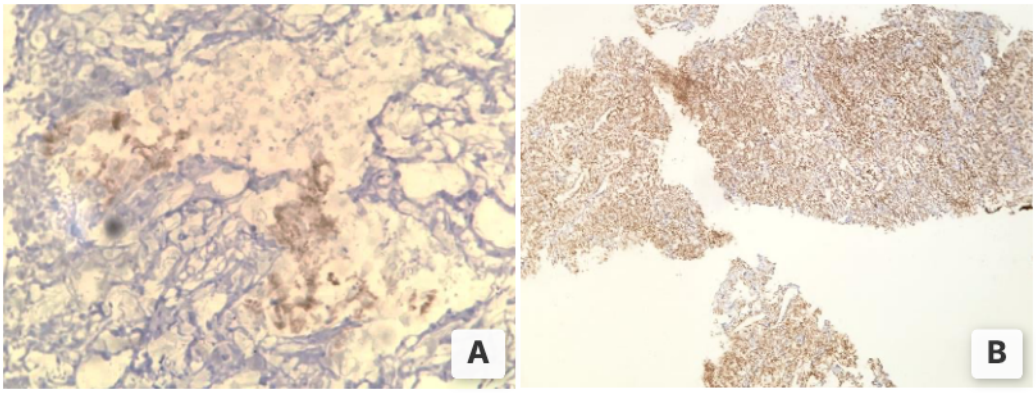

Figure 3: Immunohistochemistry depicting pigmented larger cells remained pigmented and showed focal positivity on HMB45 (A) and synaptophysin (B).

for histopathological evaluation which showed the same histologic findings.

The postoperative course was uneventful. The patient was re-evaluated after 3 months and no re-growth was noted. The patient is on regular follow-up subsequently with no recurrence reported till date.

Discussion

Melanotic Neuroectodermal Tumor of Infancy, also named Pigmented Neuroectodermal Tumor of Infancy, is clinically benign neoplasm of neuroectodermal origin [2]. It was first described as 'congenital Melanocarcinoma' by Krompecher in 1918. After confirming the neuroectodermal origin, Borello and Gorlin coined the term *MNTI* [1, 2].

The tumor commonly occurs in ages less than one year with no variation in sex [2]. It is typically bluish in color due to presence of melanin. There are studies suggesting release of VMA in urine indicative of neural crest origin [4]. In spite of being a benign tumor it has 2 % chance of converting into a malignant lesion. It is locally aggressive in nature [5].

The tumor is nonencapsulated with tumor cells arranged in tubular and alveolar pattern which is very useful to rule out other tumors like neuroblastoma and alveolar rhabdomyosarcoma [2]. The most common differential diagnosis of *MNTI* sharing a common histological and immunophenotypic expression are immature teratoma, malignant melanoma, metastatic neuroblastoma, and alveolar rhabdomyosarcoma. *MNTI* however does not show S-100 positivity as seen in our case [4]. Other markers, such as HMB45, Melan A, cytokeratin, and neuroblastic markers, such as synaptophysin and neuron-specific enolase, can help with the diagnosis [4]. In the current case, melanin-producing epithelial cells were positive for HMB-45. These findings are supported by Barrett et al [6] Cui et al [7] and Krishnamurthy et al [8]. Few studies found predominance of primitive cells, mitotic rate of $\geq$2/10 hpf, Ki-67 labeling index $>$25% and CD99 positivity as predictors of aggressive behaviour. However, there are no radiological or histopathological criteria to differentiate benign and malignant neoplasms [9].

MNTI has a high risk of recurrence reaching 10 to 15%, especially in those where the tumor most probably was not excised completely. Recurrence is reported after 3 months of the initial surgery in orbital *MNTI* in one case and after 4 months of the initial resection in mandibular tumor in another case. Primary malignant *MNTI* is typically seen associated with immature teratomas or in tumors located in the thigh [2].

MNTI not only poses a diagnostic challenge but is difficult to manage by surgeons. Total excision including up to 1 cm free margin is recommended. Chemotherapy or radiotherapy is commonly not required [2]. Radiotherapy and combination chemotherapy including vinblastine, ifosphamide, etoposide, cyclophosphamide, doxorubicin and dactinomycin has been advocated for inoperable recurrence or margin-positive resection [5]. A strong suspicion is required to diagnose this tumor and close follow-up is important to detect recurrence.

The classic histologic findings and the supportive positive immunohistochemistry tests for synaptophysin and HMB-45 confirm the diagnosis. Follow-up of patients is important as the Recurrence rate is around 20% [10]. Recurrence increases the morbidity and complicates further surgical resection. Some employ the measurement of Urinary vanillylmandelic acid (VMA) levels. Urinary VMA levels can be negative in recurrent cases and therefore is not of much use in follow up cases. CT scan is advocated to identify recurrences [3]. The above measures of urinary VMA measurement and repeat imaging may not be possible especially when the patient is from a remote area. The primary care givers must be explained about the signs like swelling, any neurological symptom, disturbed visual acuity. If in case any such condition is faced patient must be brought to the hospital and further investigations must be done.

Conclusion

Melanotic neuroectodermal tumor of infancy (*MNTI*) must be differentiated from other round cell tumors. Histopathology and Immunohistochemistry can aid in the correct diagnosis and differentiating it from other tumors of same category. The main emphasis must be on its locally aggressive nature and decision for further management depends on the same. After hemimandibulectomy / wide-margin excision, patient must be kept on follow up for possible recurrences and malignant transformation. Early diagnosis is important because it has high potential to cause deformities in surrounding structures.

Acknowledgements: Dr. Ajay Malik

Funding: NA

Competing Interests: NIL

Declaration of patient consent The authors certify that they have obtained all appropriate consent from the guardian of patient. In the form, the legal guardian has given his consent for images and other clinical information to be reported in the journal.

Ethical approval The study is approved by the ethical committee of the hospital.

References

1. Agarwal P, Agarwal I, Raina VK. Melanotic neuroectodermal tumor of infancy: case report of an unusual tumor. Indian J Plast Surg. 2008;41(2):214-216. doi:10.4103/0970-0358.44942
2. Asefa M, Hailu T. Melanotic neuroectodermal tumor of infancy: a case report. J Med Case Rep. 2024;18:230. doi:10.1186/s13256-024-04550-y
3. Rachidi S, Sood AJ, Patel KG, Nguyen SA, Hamilton H, Neville BW, et al. Melanotic neuroectodermal tumor of infancy: a systematic review. J Oral Maxillofac Surg. 2015;73(10):1946–1956. doi: 10.1016/j.joms.2015.03.061
4. Surana A, Magalhaes MHCG, Pires FR, de Almeida OP. Clinical and immunohistochemical study of melanotic neuroectodermal tumor of infancy in the maxilla. Einstein (Sao Paulo). 2018;16(2):Erc4205. Doi:10.1590/S1679-45082018rc4205
5. Sailukar M, Bhagwat R, Seth T. Melanocytic neuroectodermal tumor of infancy. J Indian Assoc Pediatr Surg. 2007;12(3):178-179. doi:10.4103/0971-9261.35942
6. Barrett AW, Morgan M, Ramsay AD, Farthing PM, Newman L, Speight PM. A clinicopathologic and immunohistochemical analysis of melanotic neuroectodermal tumor of infancy. Oral Surg Oral Med Oral Pathol Oral Radiol Endod. 2002;93(6):688–698. doi: 10.1067/moe.2002.124000.
7. Cui Y, Mao Z, Liao C. Melanotic neuroectodermal tumor of infancy: a case report and review of the surgical treatment. Oncol Lett. 2015;9(1):29–34. doi: 10.3892/ol.2014.2665.
8. Krishnamurthy A, Vaidhyanathan A, Majhi U. Malignant melanotic neuroectodermal tumor of infancy arising in the mandible. J Cancer Res Ther. 2011;7(3):368–372. doi: 10.4103/0973-1482.87018
9. Dhal I, Lali BS, Chowdhury Z, Saha S. Melanotic neuroectodermal tumor of infancy: a case series and comprehensive review of this rare pediatric neoplasm. Asian Pac J Cancer Biol. 2023;8(3):309-311. doi:10.31557/APJCB.2023.8.3.309-311
10. Tiwari A, Yadav ML. Melanotic Neuroectodermal Tumor of Infancy: A Rare Case Report. Cureus [Internet]. 2019. January 14, 2023 at: https://www.cureus.com/articles/26329-melanotic-neuroectodermal-tumor-of-infancy-a-rare-case-report