

Melanotic Neuroectodermal Tumour of Infancy - A Case Report and Review of Literature

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Abstract

Rationale: Melanotic neuroectodermal tumour of infancy (MNTI) is universally described as a rare, benign, pigmented lesion which most frequently involves the maxilla. Its origin is well established to be in the neural crest cells. Due to the high recurrence rate and aggressive behaviour mimicking malignancy, it poses a great challenge in their diagnosis, treatment planning, and prognosis. **Patient Concern:** Two-year-old female with no known comorbidities was brought in with the chief complaint of a growing swelling in the upper lip region. **Diagnosis and Treatment:** She was taken up for resection of the tumour under general anaesthesia. The specimen was subjected to histological and immunological examination confirming the diagnosis of MNTI. **Outcome:** The postoperative period was uneventful. After regular follow-up, the patient showed satisfactory healing with no signs of recurrence. **Take-Away Lessons:** Based on our experience, we feel that the diagnosis of MNTI is mainly clinical. Early conservative surgical excision and regular follow-up provide an excellent result with good prognosis.

Keywords: Melanotic neuroectodermal tumour of infancy, neural crest, resection, vanillylmandelic acid

INTRODUCTION

Melanotic neuroectodermal tumour of infancy (MNTI) is a rare, benign, pigmented lesion most frequently involving the maxilla but has also been reported in other anatomic locations distant from the premaxilla. Krompecherl was the first to describe the lesion.^[1] Historically, synonyms include melanotic progonoma, melanotic epithelial odontoma, and congenital melanocarcinoma.^[2] Although the plethora of various terminologies used creates a challenge in determining the exact cell of origin of the tumour, neural crest cells being the cell of origin is widely accepted.^[3] The high recurrence rates and its aggressive behaviour may give the impression of malignancy.^[4]

CASE REPORT

This 2-year-old female with no known comorbidities was brought in with the chief complaint of a growing swelling in her upper lip region. It rapidly increased in size leading to difficulty in mastication and deglutition with mild pain leading to facial asymmetry. On palpation, there was a single solitary diffuse swelling measuring 3 cm × 1.5 cm situated in the upper lip region extending superiorly from

the base of the nose inferiorly up to the vermilion border of the upper lip and either side to the corner of the mouth. The swelling was spherical in shape with ill defined margins and indistinct edges [Figure 1a and b]. On palpation, it was hard in consistency, fixed to the underlying tissues, no local rise in temperature, noncompressible, nonreducible, nonfluctuant, and no visible discharge. Intraorally, the dome-shaped sessile mass was present in the right anterior maxillary alveolar ridge extending from the lower border of the upper lip to the distal aspect of 61 and 51 with distinct spacing between them. A fine-needle aspiration biopsy was performed which revealed the presence of small, uniform basophilic cells with round nuclei and scanty cytoplasm [Figure 2]. Contrast-enhanced computed tomography revealed a well-defined expansile

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osteolytic lesion measuring 4 cm × 3 cm × 3 cm in the right maxillary alveolar ridge region and anterior part of the hard palate, crossing the midline [Figure 3a and b]. The lesion appeared sclerotic with a mixed chondroid matrix. Based on the clinical and radiological evaluation, a provisional diagnosis of a fibro-osseous lesion with respect to the anterior maxilla was made.

The patient was taken under general anaesthesia after normal findings in haematological and biochemical examination. An incision was made in the mucosa surrounding the extension of the tumour. The mucosa was reflected along the incision to expose the sound bony margins. The tumour was resected along with the embedded tooth germs [Figure 4a-c]. The postoperative recovery was uneventful. The patient was reviewed at intervals of 1, 2, 3, 6, and 12 months postoperatively, where she showed satisfactory healing and no signs of recurrence [Figures 5-7]. The specimen obtained during the excisional biopsy revealed macroscopic bluish-black gelatinous deposits. Sections revealed a biphasic population of cells arranged in nests consisting of larger epithelioid cells having intracytoplasmic melanin toward the periphery along with occasional nests of smaller primitive-looking small round neuroblasts. The larger cells were polygonal to cuboidal with abundant eosinophilic cytoplasm with melanin granules. Smaller primitive cells had a high N:C ratio and scant cytoplasm. The tumour was seen infiltrating into the bony trabeculae with some of the neuroblasts being differentiated to form ganglion cells. On immunohistochemistry, HMB-45 highlighted the larger epithelioid cells. Vimentin and neuron-specific enolase (NSE) stains were positive in both epithelioid and small round blue cells whereas synaptophysin stain was positive only in small round blue cells, therefore,

both histological and immunochemistry studies confirmed a diagnosis of MNTI [Figure 8].

DISCUSSION

The MNTI mainly arises in the craniofacial region, predominantly in the maxilla followed by the skull and mandible. The other sites include the brain, epididymis, ovaries, uterus, and mediastinum.^[4] Infants usually present with a painless, nonulcerative bluish-black gingival mass that is often confused with an eruption cyst.^[5] In our case, it was a 2-year-old female who presented with a dome-shaped sessile mass present in the right anterior maxillary alveolar ridge.

Although vanillylmandelic acid level in a 24-h urine collection, is elevated in 10%–15% of MNTI,^[5,6] it is of low diagnostic value. Clinically, MNTI appears as a rapidly expanding, nonulcerated mass. The tumour usually displaces the primary maxillary incisors on the side affected but seldom crosses the midline. Radiologically, the tumour shows irregular resorption of bone in the

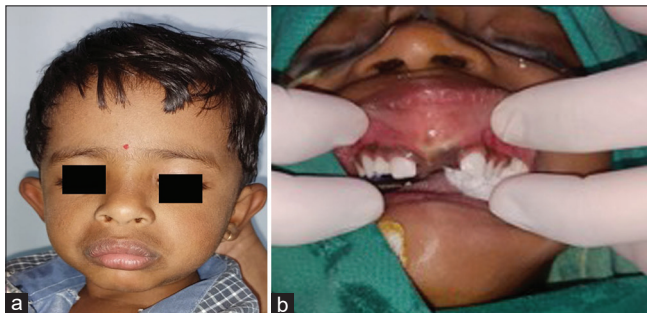


Figure 1: (a and b) Preoperative photograph

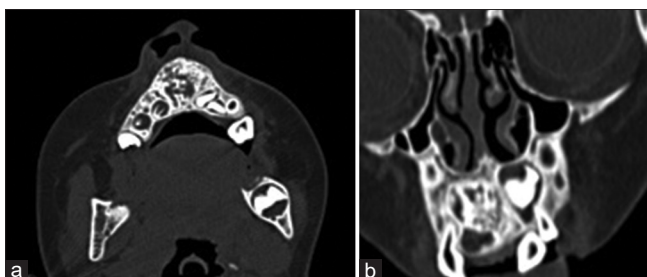


Figure 3: (a and b) Preoperative radiograph

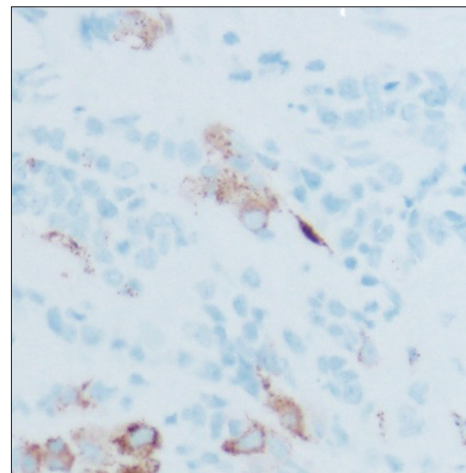


Figure 2: FNAC smear. FNAC: Fine-needle aspiration cytology

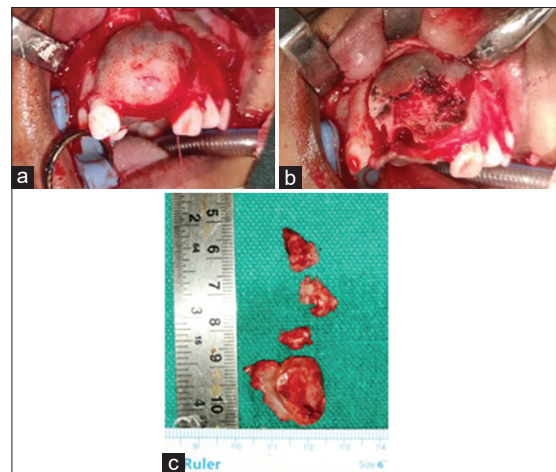


Figure 4: (a and b) Intraoperative photograph of the lesion, (c) Excisional biopsy specimen



Figure 5: Postoperative photograph (12-month follow-up)



Figure 6: Postoperative radiograph (12-month follow-up)



Figure 7: Postoperative radiograph with good healing and no recurrence

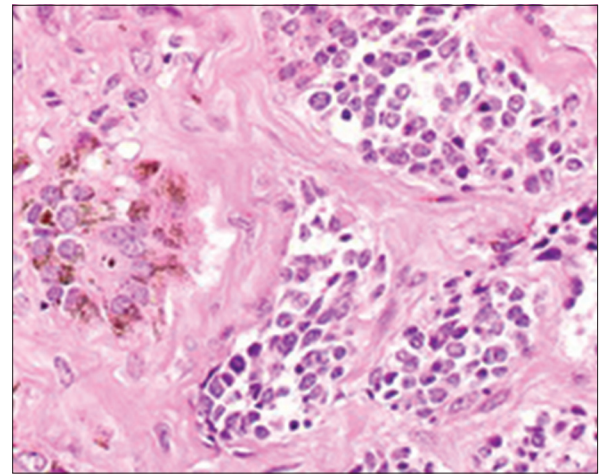


Figure 8: Histological picture

localised region with displacement of tooth buds.^[6] In our case, the swelling was evident in the region of the central incisor not crossing the midline. The differential diagnosis of MNTI includes neuroblastoma, Ewing's sarcoma, rhabdomyosarcoma, peripheral neuroepithelioma, desmoplastic small round cell tumour, and malignant melanoma and lymphoma.^[3,6] Microscopically, MNTIs are biphasic tumours, characterised by larger polygonal epithelioid cells resembling melanocytes, with variable deposits of melanin, and smaller neuroblast-like round cells. Reported results of immunohistochemical staining in MNTIs seem rather inhomogeneous.^[3] The small neuroblast-like cells mainly expressed CD56 and synaptophysin. In most of the cases, both cell populations were positive for NSE. Expression of Ki-67/CD99 in MNTI, which is quite uncommon, might be correlated with more aggressive growth of the tumour.^[3,7] In our reported case, the histopathological examination revealed a noncapsulated infiltrating tumour mass arranged in an alveolar pattern with fibroblasts dispersed in the cellular connective stroma. The alveolar spaces had a biphasic cell population and peripheral irregular, large, melanin-stained layer of cuboidal cells.

The central cells were small, round and had neuroblast-like hyperchromatic nuclei and scant cytoplasm. Based on these features, a diagnosis of MNTI was made consistent with the reported literature.^[8,9] MNTIs have rapidly progressed and augmented with high growth potential.^[7]

Surgical excision is the typical treatment for MNTI. There are reports of wide excision, subtotal maxillectomy, and use of titanium miniplates for reconstruction in the literature but conservative treatment is widely advocated.^[8] Cases of MNTI with malignant changes may be augmented/substituted with other modes of treatment including only chemotherapy, a combination of chemotherapy with radiotherapy before or after the surgical treatment,^[9] but these were not required in our patient. The recurrence rate in both genders after a 5-year postoperative period was similar and the overall incidence of local recurrence is 15%–27%.^[5,7] Infants who manifested within the first 2 months of birth were associated with a high risk of recurrence which generally occurred within 6 months from treatment.^[3,7]

The strengths of our surgical technique included the conservative approach by removing the tumour mass in toto without disrupting the uninvolved periosteum and surrounding structures which would not hamper the functional and aesthetic aspects at the same time, preventing recurrence, but the limitation was the removal of the embedded tooth buds which later will require prosthodontic rehabilitation to further prevent facial asymmetry.

CONCLUSION

Based on our experience, we feel that the diagnosis of MNTI is mainly clinical. It is well established that the neural crest is the cell of origin of MNTI. In neuroblastomas, the expression of NSE is a poor prognostic feature, but this was not seen to apply to MNTI in our study. We conclude that early diagnosis and surgical management through excision as supported by various literatures provide an excellent result with good prognosis in the future management of individuals with MNTI.

Declaration of patient consent

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

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Conflicts of interest

There are no conflicts of interest.

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