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Case report

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Melanotic Neuroectodermal Tumour of Infancy - A Case Report of a Rapidly Enlarging Maxillary Mass

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Abstract

Introduction: The melanotic neuroectodermal tumor of infancy (MNTI) is a rare benign neoplasm that primarily affects the maxilla of infants during their first year of life. It is a rapidly growing lesion, clinically aggressive and surgery is the mainstay of treatment with proven efficacy.

Case Presentation: A case of maxillary MNTI in a 5-month-old male infant treated by radical surgery is reported here. Both extraoral and intraoral findings showed a massive swelling with multiple ulcerations of the overlying mucoas. Radiographs showed an ill defined mixed radiolucent radiopaque lesion. Histopathological examination showed biphasic microscopical presentation consisting of aggregates of small neuroblastic cells surrounded by the larger pigmented cells.

Conclusions: MNTI is a rare, rapidly progressive pigmented lesion that commonly affects the anterior maxilla. Other differentials should be carefully considered in arriving at a diagnosis. Wide surgical excision with margin of normal tissue is the mainstay of treatment.

Key Words: Melanotic neuroectodermal tumor of infancy, Surgery

Introduction

Melanotic neuroectodermal tumour of infancy (MNTI) is a rare connective tissue neoplasm of neural crest origin, comprising pigment producing large polygonal cells and small neuroblastic cells [1-5].

The origin of MNTI is uncertain, it is believed to arise from the retinal anlage, odontogenic origin, melanocarcinoma from epithelial rests or Jacobson vomeronasal organ [1-5]. Since the time MNTI was first reported in 1918, five major

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systematic reviews of English literature have identified more than 476 cases with no clear gender predilection [1-5]. It predominantly occurs in infants, involving the craniofacial site specifically the premaxilla/maxilla area although other sites have been reported [2, 3] Cases involving the mandible and skull have been reported [4-5]

It is characterized by rapid growth, bluish-black discoloration, displacement of the involved tooth buds, and high recurrence rate with a propensity for malignant transformation [1-5].

MNTI could pose diagnostic and treatment challenges to clinicians. Complete surgical excision is generally considered as the primary treatment option [6], but protocols combining local surgery and adjuvant chemotherapy have also been proposed for recurrent tumours [7, 8].

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Figure 1: Clinical photograph showing intraoral tumor

The severe adverse effects associated with chemotherapy in young children remain a matter of debate. A recent investigation showed BRAFV600E mutation in a case of MNTI, which might give rise to the use of targeted therapy in the management of MNTI [10].

The purpose of this report is to present a case of MNTI in a five-month-old boy who was treated by complete surgical excision. The clinical, imaging, and histological characteristics of this case are also discussed. To the best of our knowledge, no previous Nigerian case has been documented.

Case Report

A five-month-old boy was referred to the Oral and Maxillofacial Surgery Department of Federal Teaching Hospital, Gombe, Nigeria, with a 5month history of upper left jaw swelling. The swelling, noticed at birth was painless, rapidly increasing in size with associated difficulty in feeding and weight loss. There was no associated epistaxis or spontaneous bleeding from the lesion. He otherwise appeared healthy with remarkable medical or family history. On extraoral examination, the baby was emaciated with grossly enlarged mass of the left maxilla with extension out of the mouth. The swelling measured 8cm by 6cm in diameter, oval, firm, non-tender, with blackish colouration of the overlying mucosa. The nose was deviated to the right side (Figure 1). Extra-oral plain radiographs



Figure 2: Resected specimen

revealed ill defined mixed radiolucent radiopaque presentation.

Intra- oral examinations revealed a large expansile mass with well demarcated margins. There are multiple areas of ulceration of the alveolar mucosa labially and palatally. The two mandibular central incisors were the only teeth present. Excisional biopsy under general anesthesia was decided upon based on the age of the patient and the size of the lesion. The patient was worked up for surgery. The tumour was surgically excised with wide margin of normal tissue via bucco-gingival incision. Local buccal flap was used for closure of the defect. Grossly the tumor appeared pigmented (Figure 2). Microscopically, lesion consisted of small neuroblastic cells and larger polygonal cells which were pigmented with melanin granules. The small neuroblastic cells were arranged in nest surrounded by the larger pigmented cells (Figures 3 & 4). His postoperative recovery was uneventful (Figure 5) and patient was considered fit to go home a week later. The patient was subsequently lost to follow-up.

Discussion

MNTI is a rare tumor; we report the first Nigerian case in a 5-month old infant. Camevale *et al.*, [9] earlier reported a case in West Africa. The clinical presentation of a rapidly enlarging nonulcerative pseudoencapsulated and solitary discolored



Figure 3: Photomicrograph showing tumor composed of small neuroblastic cells and larger polygonal cells (H & E x40)

maxillary mass in an infant is highly suggestive of MNTI as majority of cases previously reported have similar presentations. The peak age of occurrence has been reported to be 4 months [2]. Clinical differential diagnosis of a rapidly enlarging mass of the upper jaw includes rhabdomyosarcoma (RMS), Ewing family of tumours (EFT) and Burkitt lymphoma (BL) were considered but were excluded based on clinical and histologic grounds.

African endemic BL may present with other abdominal symptoms which were absent in this present case. Also, BL is not known to present with this typical discoloration. Histologically, BL does not present with a biphasic cellular pattern of small neuroblastic cells and large polygonal melanin producing cells. The typical morphologic starry-sky appearance in dense lymphoid cells caused by tingible body macrophages seen in BL was not present in our case. Phenotypically and genetically, BL should be positive for CD45 (leucocyte common antigen) and isolated MYC/8q24 rearrangement.[11].

RMS is also not known to present with this typical discoloration. Histologically, the morphologic presentation of RMS depends on the particular subtype: alveolar, embryonal, spindle-sclerosing and pleomorphic. RMS should be positive for desmin phenotypically [11].



Figure 4: Photomicrograph showing tumor cells laden with intracellular melanin. The small neuroblastic cells were arranged in nest surrounded by the larger pigmented cells

EFT, when it presents in the jaw does not present with discoloration. Histologically, the morphologic presentation of EFT is that of small round blue cells. EFT is positive for CD99 and EWSR1 rearrangement[11]. In MNTI, the small positive neuroblastic cells for are neuroendocrine markers such as Chromogranin, synaptophysin, and neuron-specific enolase. The large polygonal cells which are usually pigmented with granules of melanin pigment are positive for Human melanoma black-45 and cytokeratin [11]. Furthermore, patients with MNTI, although not all, show positivity to urinary3-methoxy 4hydromandelic acid (VMA) [2].

Computerized tomography (CT) scan and magnetic resonance imaging (MRI) are of benefit



Figure 5: Postoperative photograph showing recovery.

in preoperative assessment and treatment planning. However, only extraoral plain radiographs were used in this case because the patient could not afford the cost of these advanced imaging techniques. This is a common challenge seen in the management of this tumour in a resource poor setting.

Treatment is by wide surgical excision although recurrences have been reported usually within the first few months of surgery. Since the patient did not return after surgical excision, we could not ascertain the possibility of recurrence. Long term follow up is mandatory because the course of the disease cannot be predicted by morphological findings alone. However, the age at presentation is an important prognostic predictive marker of recurrence with younger patients more likely to develop such [12]. Previous studies have reported positive results following surgical treatment [7, 8]. Wide surgical excision with margin of normal tissue provides the best efficacy for local tumour clearance and cure [13]. Radiotherapy and chemotherapy have been found to be ineffective in the treatment of MNTI [2].

Conclusions

MNTI is a rare, rapidly progressive pigmented lesion that commonly affects the anterior maxilla. Other differentials should be carefully considered in arriving at a diagnosis. Wide surgical excision with margin of normal tissue is the mainstay of treatment.

Conflict of interest

The authors declare that there are no conflicts of interests

Authors' contribution

AOB was responsible for conception, design, data acquisition, drafting/revision of the article and final approval.

OOK was responsible for design, data acquisition/analysis/interpretation, drafting/revision of the article and final approval.

SOO made substantial contribution to design, drafting/revision of the article and final approval.

PUH made substantial contribution to design, drafting/revision of the article and final approval.

AYM made substantial contribution to design, drafting/revision of the article and final approval.

Ethical consideration

The written informed consent was taken from the next of kin of the patient and is available with the authors.

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