Case Series

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Diagnostic Challenges of Uncommon Paediatric Head and Neck Masses- A Case Series

Pathology Section

KARTHIK SIGAMANI[1], PREETHI SRINIVASAN[2]

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ABSTRACT

Paediatric head and neck masses pose diagnostic challenge to the clinicians owing to the wide spectrum of lesions including congenital (developmental), inflammatory and neoplastic lesions. In the present case series, uncommon paediatric head and neck mass lesions that were diagnosed in a tertiary care hospital over a period of three years from January 2017 to December 2019, had been included, emphasising on the diagnostic challenges encountered. First case was of Cervical Chondrocutaneous Branchial Remnant (CCBR) that was misdiagnosed as branchial cyst clinically. Second case was a lymphoepithelial cyst that masqueraded as an acute suppurative lymphadenitis in Fine Needle Aspiration Cytology (FNAC) due to florid inflammation. Third case was an aggressive poorly differentiated malignant neoplasm of left nasal cavity namely Nuclear protein in Testis (NUT) carcinoma that had to be differentiated from several other small round blue cell tumours. Fourth was a case of lipoblastoma which could be mistaken for myxoid liposarcoma. The last two cases were vascular tumours, one of them was juvenile capillary hemangioma with a benign clinical course while the other was an intermediate grade tumour namely kaposiform hemangioendothelioma. A wholesome clinical, radiological and pathological evaluation will help to solve the diagnostic dilemmas in this group of lesions.

Keywords: Branchial cleft, Children, Congenital, Cyst, Tumour

INTRODUCTION

Paediatric head and neck masses are commonly encountered in clinical practice that pose serious diagnostic challenge to the clinicians owing to the wide spectrum of lesions that can occur in this region in children. Broadly paediatric head and neck lesions are classified into three categories namely congenital (developmental), inflammatory and neoplastic lesions based on aetiology [1]. Accurate diagnosis is possible only based on combination of proper clinical examination, radiological correlation and histopathological examination. The difficulty lies not only in diagnosis but also in handling parental anxiety and careful management of these lesions without any damage to vital structures in this region of the body of children. Any misdiagnosis can adversely affect the management and prognosis of this wide group of paediatric head and neck lesions. In the present case series, uncommon paediatric head and neck mass lesions that were diagnosed in a tertiary care hospital over a period of three years from January 2017 to December 2019 had been included, emphasising on the diagnostic challenges encountered and their unique features [Table/Fig-1].

CASE SERIES

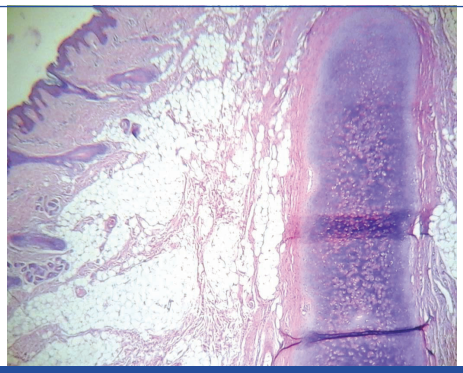
Case 1

A two-year-old male child reported to the Department of General Surgery with a chief complaint of painless mass of size 2×2 cm in the right lateral side of neck since birth. There were no other significant complaints and there was no family history of similar swelling in the family members. On clinical examination, the swelling was non tender and soft in consistency. Ultrasonogram (USG) neck impression was branchial cyst following which the lesion was excised and sent for histopathological examination. Grossly, skin covered soft tissue mass measured 3×2.5×1.5 cm with a grey white cut surface. Microscopic examination revealed stratified squamous lining epithelium with a central solid core of mature cartilaginous tissue extending down to the muscles. The mature cartilage was surrounded by lobules of mature adipocytes and skin adnexal structures. Final diagnosis of Cervical Chondrocutaneous Branchial Remnant (CCBR) was made [Table/Fig-1,2]. The follow-up period was uneventful.

Variables	Case 1	Case 2	Case 3	Case 4	Case 5	Case 6
Age	2 years	11 years	10 years	5 years	11 years	8 years
Gender	Male	Female	Female	Female	Female	Male
Location of the head and neck mass	Right lateral side of neck	Right lateral side of neck	Left nasal cavity	Left lateral side of neck	Right lateral side of neck	Right lateral side of neck
Size of the head and neck mass	2×2 cm	3×2 cm	1.5 cm diameter	2×1 cm	3×2 cm	4×2 cm
Associated clinical features	Painless and soft	Soft and painful	Nasal obstruction and painless	Painless, soft and mobile	Reddish, soft and painless	Reddish, soft and painless
Clinical diagnosis	Branchial cyst	Acute suppurative lymphadenitis	Nasal polyp	Cervical lymphadenopathy	Vascular malformation	Haemangioma
Radiological impression	Branchial cyst	Abscess	Nasal polyp	Lymphadenopathy	Haemangioma	Haemangioma
Final histopathological diagnosis	Cervical chondrocutaneous branchial remnant	Lymphoepithelial cyst (Branchial cyst)	NUT carcinoma	Lipoblastoma	Juvenile capillary Haemangioma	Kaposiform haemangioendothelioma
Immunohistochemistry	-	-	CK-AE1/AE3 + CD99 – Synaptophysin- Chromogranin-	-	CD31+ CD34+	CD31+ CD34+

Category of lesion	Developmental	Developmental	Neoplastic (Malignant)	Neoplastic (Benign)	Neoplastic (Benign)	Neoplastic (Intermediate)
Diagnostic challenges encountered	Cystic lesion mistaken for branchial cyst clinically. Histopathological examination conclusive	Misdiagnosed as acute suppurative lymphadenitis in FNAC due to marked inflammation	Resembled nasal polyp clinically. Histopathological examination along with IHC conclusive	Uncommon site and mistaken for lymphadenopathy clinically. Histopathological examination conclusive	Mistaken for vascular malformation clinically. Histopathological examination along with IHC conclusive	Mistaken for hemangioma clinically. Histopathological examination along with IHC conclusive
Unique features	Cystic change and fistula can occur	- Extensive inflammation masks the diagnostic features in FNAC	- Resembles other small round blue cell tumours - Abrupt keratinisation present - Aggressive tumours	- Only 14% cases in head and neck - Resembles myxoid liposarcoma - Lobulation and fibrous septa are unique	- Head and neck common site - Two clinical phases: Early proliferative and late involutional phases	- Glomeruloid nodules present microscopically - Regression not common - Poor response to treatment

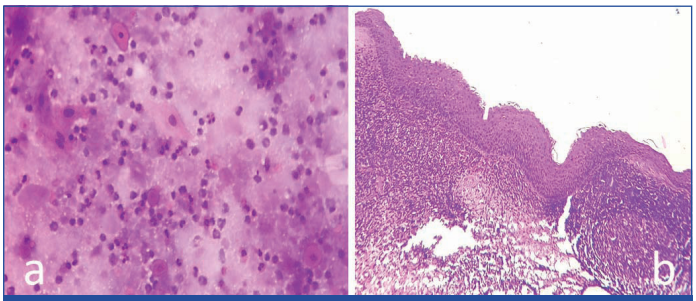
[Table/Fig-1]: Clinico-pathological features and diagnostic challenges of the cases.



[Table/Fig-2]: Cervical chondrocutaneous branchial remnant showing stratified squamous epithelium with underlying mature cartilage, adnexal structures and adipocytes (H&E, 10X).

Case 2

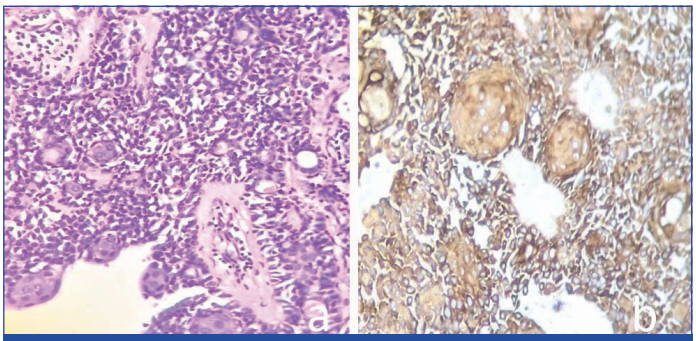
A 11-year-old female presented in the Department of General Surgery with a swelling of size 3×2 cm in the right lateral side of neck for six months duration associated with pain for past one month. Based on the radiological (ultrasound of neck) impression of abscess and initial Fine Needle Aspiration Cytology (FNAC) report of acute suppurative lymphadenitis, the patient was treated with a course of antibiotics following which pain subsided however there was no reduction in the size of the swelling. Hence, repeat FNAC was done and it showed cellular smears with singly scattered anucleate squames along with mature squamous epithelial cells and sheets of mixed inflammatory infiltrate. Based on these features, a revised diagnosis of branchial cyst was given in FNAC following which excision biopsy was carried out. On gross examination, skin covered soft tissue mass measured 3×1.5×0.7 cm and the cut section revealed a cystic cavity filled with pultaceous material. Microscopic examination showed a cyst lined by stratified squamous epithelium with underlying fibrocollagenous stroma containing diffuse infiltrate of lymphocytes forming follicles with prominent germinal centers. Histopathological features were consistent with lymphoepithelial cyst (branchial cyst) [Table/Fig-1, 3a,b]. The patient came for follow-up after two months of surgery and there were no complications or recurrence in the follow-up period.



[Table/Fig-3]: a) FNAC of branchial cyst showing mature squamous cells in an inflammatory background (H&E, 40X); b) Histopathology of branchial cyst showing stratified squamous epithelial lining with underlying reactive lymphoid tissue with germinal centers (H&E, 40X).

Case 3

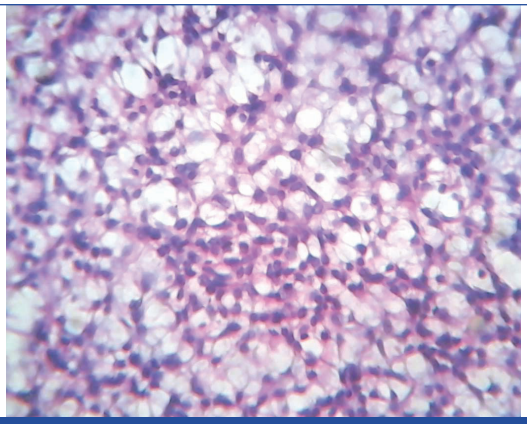
A 10-year-old female child presented in the Department of Otorhinolaryngology with a slowly growing painless mass of size 1.5 cm in diameter in the left nasal cavity associated with nasal obstruction for three months duration. Clinically and radiologically (computed tomography scan), the mass was diagnosed as nasal polyp following which excision of the mass was performed. On gross examination, multiple grey white soft tissue fragments altogether measured 0.5 cc in aggregate. Microscopic examination of the excisional biopsy showed a neoplasm composed of small uniform round cells arranged in diffuse sheets with scant cytoplasm, round nuclei and coarse chromatin. Focal areas showed characteristic abrupt keratinisation. All these findings were suggestive of a small round blue cell tumour with the differentials of Nuclear protein in Testis (NUT) carcinoma, Ewing's sarcoma/Primitive Neuro-Ectodermal Tumour (PNET), olfactory neuroblastoma and sinonasal undifferentiated carcinoma. Immunohistochemistry (IHC) showed positivity for pancytokeratin (CK- AE1/AE3). All other markers namely synaptophysin, chromogranin and CD99 were negative. So, on correlating with histopathological features of characteristic abrupt keratinisation along with cytokeratin positivity in IHC, final diagnosis of NUT carcinoma was made [Table/Fig-1, 4a,b]. The patient was referred to higher centre for further management.



[Table/Fig-4]: a) NUT carcinoma composed of sheets of small to medium sized cells with abrupt keratinisation (H&E, 40X); b) NUT carcinoma showing CK-AE1/AE3 positivity in the tumour cells (IHC, 40X).

Case 4

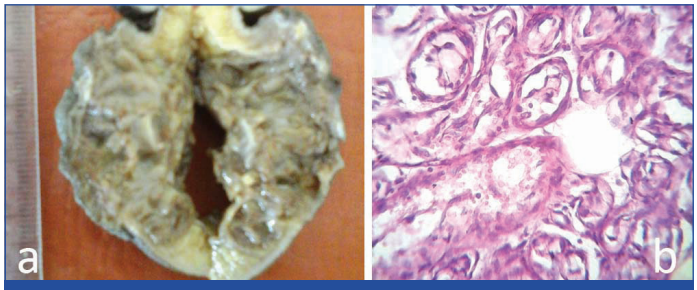
A five-year-old female child presented in the Department of General Surgery with a painless, soft and mobile swelling of size 2×1 cm in the left lateral side of neck for one year duration. Excision of the swelling was done with a clinical and radiological (USG neck) diagnosis of cervical lymphadenopathy. Grossly, the soft tissue mass measured 1.5×1×0.5 cm with yellowish cut surface. Histopathological examination showed a tumour composed of mature and immature adipocytes arranged in lobular pattern in a background of myxoid stroma with small number of capillaries. There was no nuclear atypia or increased mitotic activity. Therefore, the diagnosis was consistent with lipoblastoma [Table/Fig-1,5]. Patient was completely normal in the follow-up period after surgery with no recurrence of the swelling.



[Table/Fig-5]: Lipoblastoma showing lipoblasts, mature adipocytes and spindle cells (H&E, 40X).

Case 5

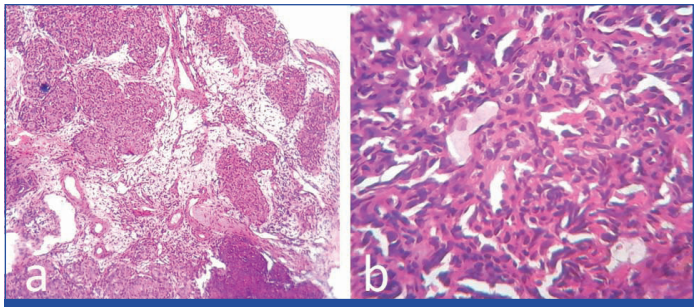
A 11-year-old female child presented in the Department of General Surgery with a reddish, soft and painless swelling in the right lateral side of neck since birth. The swelling was gradually increasing in size for the past two years and attained the present size of 3×2 cm. Clinically, the lesion was provisionally diagnosed as vascular malformation and the patient was subjected to radiological (ultrasound neck) investigation. The radiological impression was haemangioma following which the swelling was excised and sent for histopathological examination. On gross examination, skin covered soft tissue mass measured 4×3×1.5 cm and the cut surface showed a highly vascular reddish circumscribed mass measuring 3×3×2 cm in the dermis. Microscopic examination showed stratified squamous epithelium with underlying vascular lesion arranged in lobules and composed of numerous capillary sized blood vessels lined by plump endothelial cells with inconspicuous lumens. Histopathological features were consistent with that of juvenile capillary haemangioma. IHC showed CD31 and CD34 positivity thereby confirming the histopathological diagnosis [Table/Fig-1,6 a,b]. The follow-up period was uneventful.



[Table/Fig-6]: a) Gross- Vascular reddish circumscribed mass measuring 3×3×2 cm in the dermis; b) Juvenile capillary haemangioma showing proliferating capillaries lined by plump endothelium with occluded lumens (H&E, 40X).

Case 6

An eight-year-old male child presented in the Department of General Surgery with a violaceous to reddish, soft and painless swelling of size 4×2 cm in the right lateral side of neck for one year duration. The swelling was not present at birth and it was gradually increasing in size. A provisional clinical diagnosis of haemangioma was made on correlating with radiological findings (ultrasound neck) following which excision of the mass was done. On gross examination, globular skin covered soft tissue mass measured 4×2.5×2 cm with a grey brown cut surface. Microscopic examination revealed a tumour composed of fascicles and nodular pattern of spindle shaped cells with slit like spaces containing Red Blood Cells (RBCs) along with capillary sized vessels separated by myxoid stroma. Immunohistochemistry showed positivity for CD31 and CD34. Final diagnosis of kaposiform haemangioendothelioma was made [Table/Fig-1,7 a,b]. The follow-up period was uneventful and there was no recurrence of the swelling after three months of surgery.



[Table/Fig-7]: a) Kaposiform haemangioendothelioma showing nodular pattern with intervening myxoid areas (H&E, 10X); b) Kaposiform haemangioendothelioma showing glomeruloid aggregates of capillaries within the nodules (H&E, 40X).

DISCUSSION

Paediatric head and neck mass lesions are unique and diagnostically challenging, compared to that in adult population. A wide spectrum of lesions ranging from non neoplastic to neoplastic lesions can be encountered with low incidence of malignancy in the head and neck region of children unlike that in adults [1].

In the present case series, four cases (66%) were reported in females and only two cases (34%) in males with the mean age group of 7.8 years. All the masses were present in the lateral side of neck with a single case involving the nasal cavity and their mean size was 2.6 cm. In a study of 281 paediatric neck masses by Gov-Ari E and Hopewell BL, 55% were females, the mean age group was 7.4 years and the mean size of the mass was 2.6 cm [1]. All these findings, were in concordance with the present study. Out of the 281 cases in the study of Gov-Ari E and Hopewell BL, from United States of America, a wide variety of histopathological distribution of paediatric head and neck lesions were encountered with the most common being congenital lesions followed by inflammatory and neoplastic lesions respectively. The common congenital lesion, in their study was, thyroglossal cyst followed by branchial cyst [1]. In a study of 35 paediatric neck masses by Osifo OD and Ugiagbe EE, from Nigeria, thyroglossal cyst was the most frequent non neoplastic lesion while hodgkin lymphoma was the commonest neoplastic lesion [2]. In another study of neck masses in 207 children from China by Xia X et al., thyroglossal cyst was the most common lesion accounting for 31% of the cases [3]. In all the above studies by Gov-Ari E and Hopewell BL, [1], Osifo OD and Ugiagbe EE, [2] and Xia X et al., [3], there were no cases of CCBR, juvenile capillary haemangioma, kaposiform haemangioendothelioma, NUT carcinoma or lipoblastoma reported. This highlights the rarity of such paediatric head and neck mass lesions.

Cervical Chondrocutaneous Branchial Remnants (CCBR) are uncommon paediatric developmental lesions. In a study, by Woo HY and Kim HS, which included 1096 patients with branchial cleft anomalies, CCBR accounted for only 0.4% of cases [4]. The CCBRs, are most commonly present in the cervical region along the anterior border of the sternocleidomastoid muscle as cysts, fistulae or sinuses [4]. Such lesions should be differentiated from other developmental lesions like thyroglossal cyst, thymic cyst, branchial cleft cyst and hamartoma [5]. Histologically, CCBR is characterised by skin with underlying central core of mature cartilage surrounded by fat and skin appendages [4]. The CCBRs can be external markers of other serious internal congenital anomalies involving cardiovascular system, auditory system, central nervous system, respiratory system etc. Simple surgical excision is curative for this developmental lesion [4].

Branchial cyst (Lymphoepithelial cyst), though a common developmental branchial cleft abnormality in head and neck region of children, it can be mistaken for inflammatory lesions in FNAC, due to florid inflammatory infiltrate [6] similar to the present study. About 90% of branchial cysts are lined by squamous epithelium with the presence of subepithelial lymphoid tissue. Sometimes, the epithelium overlying the reactive lymphoid tissue is attenuated or absent

causing diagnostic difficulties [7]. Complete surgical excision of the cyst is curative in most cases with a recurrence rate of 5% due to incomplete surgical cyst excision [7].

Lipoblastoma is a benign soft tissue neoplasm of infants and children that commonly affects extremities with only 14% cases occurring in head and neck region [8]. These tumours exhibit characteristic lobular pattern with spectrum of adipocytic maturation including lipoblasts and mature adipocytes along with spindle to stellate cells in a myxoid matrix resembling myxoid liposarcoma. However, younger age group, presence of lobular pattern and fibrous septa will help in excluding myxoid liposarcoma [9]. Complete surgical excision is often curative for lipoblastoma. However, recurrence rates of 27% for head and neck lipoblastomas have been reported [9].

Vascular lesions of children include several entities including vascular malformations, various types of haemangiomas, haemangioendotheliomas and very rarely angiosarcomas. Juvenile capillary haemangiomas, belong to benign proliferative lesions of vascular origin [10]. They are clinically characterised by early proliferative and late involutional stages [11]. Kaposiform haemangioendothelioma is an intermediate grade vascular tumour of infancy and childhood [12]. It has to be differentiated from juvenile haemangiomas, since, it doesn't show remission and exhibits poor response to treatment. Histologically, characteristic nodules with glomeruloid aggregates of vascular channels lined by endothelial cells are seen in kaposiform haemangioendothelioma. All vascular lesions express CD31 and CD34 in immunohistochemical study [13].

The NUT Carcinoma is an aggressive poorly differentiated tumour accounting for 18% of the poorly differentiated carcinomas of the sinonasal region [14]. Histologically, the tumour consists of sheets of monotonous population of medium-sized cells having round to oval nuclei along with foci of squamous differentiation in the form of "abrupt" keratinisation [14]. This uncommon tumour poses diagnostic dilemma by resembling sinonasal undifferentiated carcinoma, Ewing's sarcoma/PNET and olfactory neuroblastoma. IHC will help in arriving at the correct diagnosis with NUT1 being the specific marker. The BRD4-NUT fusion is detected by molecular studies in around 67% of cases. The overall prognosis is poor and it is a highly lethal malignancy in children [14].

CONCLUSION(S)

There is a great variability in the clinico-histopathological profile of paediatric head and neck mass lesions. In addition to the common lesions, it is imperative to be aware of the uncommon lesions, that, can be encountered in this region which will aid in proper diagnosis and treatment. A multi-modality approach that includes clinical examination, radiological evaluation and histopathological confirmation is required for the management of this challenging group of mass lesions in children.

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PARTICULARS OF CONTRIBUTORS:
1. Associate Professor, Department of Pathology, Karpaga Vinayaga Institute of Medical Sciences and Research Centre, Chengalpattu, Tamil Nadu, India.
2. Senior Resident, Department of Pathology, Karpaga Vinayaga Institute of Medical Sciences and Research Centre, Chengalpattu, Tamil Nadu, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Preethi Srinivasan,
Om Nivas, A-21/B, Alagesa Nagar, Chengalpattu-603001, Tamil Nadu, India.
E-mail: om.lovely93@gmail.com

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