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EXPLORING THE UNCHARTED: A CASE SERIES OF RARE HEAD AND NECK SOFT TISSUE NEOPLASM

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ABSTRACT:

Background:

Head and neck sarcomas are a rare group of malignancies that pose diagnostic and therapeutic challenges and mimic carcinomas. In this case series, we present seven unique cases of head and neck soft tissue tumours, highlighting the diverse anatomical locations and clinical features encountered. The cases include fibromatosis of the maxilla, fibromatosis of the right Masseter muscle, tonsillar myofibroblastic tumour, spindle cell neoplasm of the tongue, Rhabdomyosarcoma nose, Leiomyosarcoma and synovial sarcomas of the larynx. Through this series, we aim to contribute to the limited existing literature on these uncommon tumours and provide insights into their management strategies and potential causes of recurrence in head and neck soft tissue tumours.

Materials and Methods:

This case series aims to present and discuss seven cases of rare head and neck sarcomas. The study design involves retrospective analysis of medical records, radiological imaging, and histopathological reports of the included cases. Ethical approval was obtained from the relevant institutional review board.

Case Selection:

The cases included in this series were identified from the institutional database of head and neck sarcomas. A thorough review of the available medical records and imaging studies was performed to select cases that met the inclusion criteria.

Inclusion criteria :

Patients diagnosed with head and neck soft tissue tumours confirmed by IHC

All operable cases were included

Exclusion criteria :

All cases whose final biopsy and IHC didn't suggest soft tissue neoplam were excluded

All patient who didn't give consent were excluded

Conclusion:

The materials and methods of this case series involved the retrospective analysis of medical records, imaging studies, and histopathological reports to present and discuss seven cases of rare head and neck sarcomas of which three had recurrence. The data collected provided insights into the clinical presentation, diagnostic evaluations, treatment modalities ,reason for recurrence and long-term outcomes of these sarcomas. The findings contribute to the existing knowledge on the management of these rare head and neck sarcomas and provide a basis for further research and investigation.

Introduction:

Head and neck sarcomas are rare tumours with a broad spectrum of histological subtypes, each presenting distinct diagnostic and therapeutic challenges. Despite their rarity, it is crucial to recognize these malignancies promptly as it mimics common tumours of head and neck like squamous cell carcinoma and adenocarcinoma , further establish an appropriate management plan. In this case series, we describe seven unique cases of head and neck sarcomas involving different anatomical sites, including the maxilla, masseter muscle, tonsil, nose, tongue, and larynx.

Case 1: Fibromatosis of the Maxilla

Abstract: A 52-year-old male presented with a painless swelling (figure 1a) and facial deformity. Radiographic imaging revealed an expansile lesion involving the maxilla left side (figure 1b) with no evidence of distant metastasis. Biopsy confirmed the diagnosis of fibromatosis with bland cells having mild cellularity and minimal atypia and beta catenin positivity in IHC. The patient underwent surgical resection with one centimetre margin (figure 2) with radial artery free flap reconstruction, no neck dissection was done, final biopsy confirmed diagnosis and TNM stage pT2b with adequate margins and achieved satisfactory functional and cosmetic outcomes. Regular follow-up of one year has shown no evidence of recurrence hence chemotherapy was not indicated.

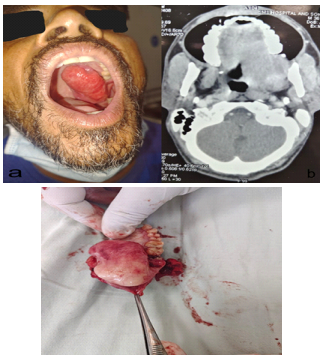

Figure 2
Wide local excison of tumour with adequate margins

Case 2: Fibromatosis of the Right Masseter Muscle

Abstract: A 57-year-old female presented with a progressively enlarging mass in the right masseter region. Imaging studies revealed a well-defined lesion involving the masseter muscle with no distant metastasis. Biopsy (figure 3) and IHC confirmed the diagnosis of fibromatosis with positivity for Beta catenin, SMA & H-caldesmon. Patient underwent wide local excision of masseter muscle with 1cm margins of tumour and reconstruction was done with PMMC flap with preservation of the surrounding structures. Post operative biopsy confirmed the diagnosis with pT2b TNM stage and superior margin was close (0.3 cm), she was not willing for reexcision hence sent for chemotherapy. Follow-up demonstrated evidence of recurrence after 9 months of primary surgery by CT and biopsy , for which she underwent reexcision and a free flap reconstruction.

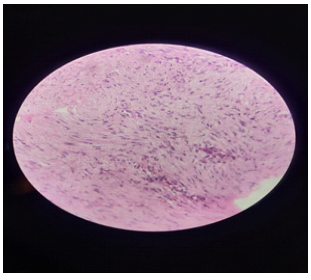

Figure 3
Histopathological picture of Fibromatosis

Case 3: Tonsillar Myofibroblastic Tumour

Abstract: 74 year old male smoker presented with a 2-month history difficulty swallowing. On examination, the patient had a mass in the left tonsillar fossa. Fibro optic laryngoscopy and CT confirmed a 3X 3cm mass arising from left tonsillar fossa with no distant metastasis .Biopsy of the mass revealed a spindle cell neoplasm with positive immunohistochemistry staining for smooth muscle actin and Vimentin and negative staining for S-100, Desmin ,P63, CD34, Cd 23, EMA and ALK . which suggested a mesenchymal neoplasm favoring low to intermediate grade Myofibroblastic origin. Tumour excision done with 1 cm margin (figure 4), no neck dissection was done , all margin clear in final biopsy with TNM stage of pT1b. No post operative chemotherapy was given as it was T1 lesion , Regular follow-up has shown no evidence of recurrence.

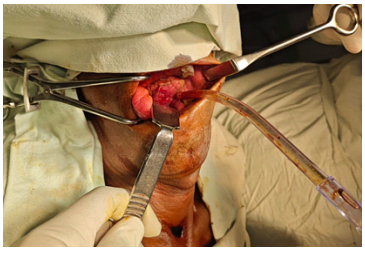

Figure 4

Case 4: Spindle cell neoplasm of the Tongue

Abstract: A 52-year-old male presented with a painless mass on the right lateral aspect of the tongue. Imaging studies revealed a well-circumscribed lesion(figure 5). Histopathological analysis gave the diagnosis of spindle cell neoplasm. The patient underwent partial glossectomy with 1 cm margin and a radial artery free flap reconstruction. Final biopsy confirmed spindle cell type sarcomatous stroma with large oval vesiculated nuclei with TNM stage of pT2a with margin negative no adjuvant treatment was done , and he has been recurrence free on 1 year follow up.

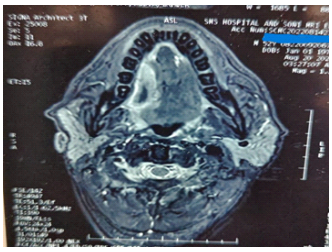

Figure 5
Ct film showing right tongue mass

Case 5: Leiomyosarcoma of the Larynx

Abstract: A 70-year-old male presented with progressive hoarseness and difficulty in breathing. Laryngoscopy and CT revealed an exophytic lesion(figure 6) involving the glottis and left aryepiglottic fold and left pyriform sinus . Biopsy and IHC demonstrated leiomyosarcoma with Desmin , SMA and vimentin positivity. The patient was not willing for operative procedure hence started on adjuvant radiotherapy with added chemotherapy . patient expired after 3 weeks of diagnosis due to aspiration pneumonia .

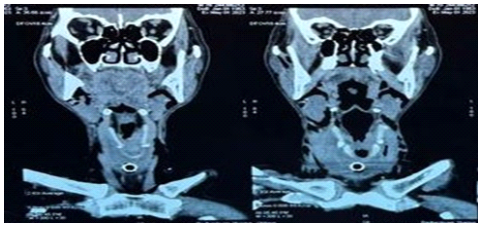

Figure 6
Ct film showing exophytic growth arising from left aryepiglotiic fold and left pyriform sinus

Case 6: Synovial Sarcoma of the Larynx

Abstract:25 year old male patient presented with mass over left side of neck , on CT it showed large mass in left parapharyngeal space and distant metastasis was ruled out doing a PET CT scan . Endoscopy and biopsy confirmed synovial sarcoma with IHC expression of Vimentin , TLE-1, and bcl2 . He underwent sarcoma excision with 1 cm margin , pharyngeal mucosal repair and tracheostomy final biopsy of TNM stage pT2b and had medial margin close of 1 mm . Patient had post operative chemotherapy with Doxorubicin and ifosfomide . But patient presented with recurrence after 7 months with large neck mass(figure 7) and total laryngectomy was planned .

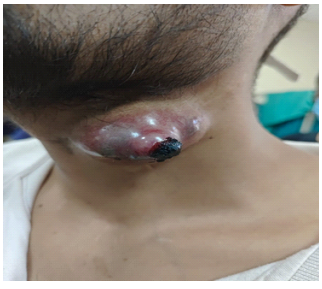

Figure 7
Clinical picture of patient showing recurrence after excision

Case 7: Rhabdomyosarcoma of the nose

3 year old male patient presented with a ulcerated mass over nose(figure 8) , MRI, biopsy and IHC confirmed diagnosis of Rhabdomyosarcoma nose . Patient underwent excision of tumour with 1 cm margin was done and final biopsy confirmed Rhabdomyosarcoma embryonal type with margin negative, stage of pT1a, Desmin and myogenin positive and high Ki67 index and adjuvant chemotherapy with vincristine was given but he had recurrence after 4months of onset of chemotherapy .

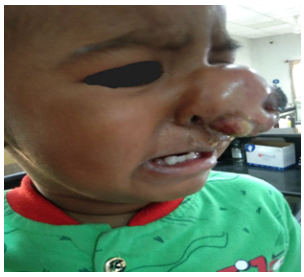

Figure 8
Clinical picture of patient having Rhabdomyosarcoma nose

Discussion:

Head and neck sarcomas are rare tumours accounting 1% of all head and neck tumours[1] that pose significant challenges in diagnosis and management. The cases presented in this series highlight the diverse anatomical locations and clinical presentations of these tumours, emphasizing the need for a multidisciplinary approach in their evaluation and treatment.

Accurate diagnosis of head and neck sarcomas is crucial, as they can mimic benign lesions or other more common malignancies like adenocarcinomas and squamous cell carcinomas . Histopathological examination, aided by immunohistochemistry and molecular studies, plays a critical role in establishing the precise histological subtype[2], which guides treatment decisions. In our series, the diagnoses included fibromatosis , synovial sarcoma, leimyosarcoma, spindle cell neoplasm and myofibroblastic tumour, highlighting the importance of considering a wide range of differential diagnoses for head and neck sarcoma.

According to WHO classification of soft tissue tumours of head and neck [3] they are further divided into malignant , borderline and benign soft tissue tumours . malignant including fibrosarcoma, leiomyosarcoma, rhabdomyosarcoma, angiosarcoma , synovial sarcoma and undifferentiated pleomorphic sarcoma. Borderline includes desmoid like fibromatosis, solitary fibrous tumours , glomangiopericytoma , haemangioendotheliomas etc, benign includes leiomyoma , haemangioma , schwannoma and neurofibroma.

Fibromatosis , also known as desmoid tumour or aggressive fibromatosis, is a rare entity characterized by the proliferation of fibroblasts within the connective tissue[4]. It represents a locally aggressive, but non-metastasizing, fibroblastic neoplasm. The exact etiology of this tumour remains unknown, but it has been associated with trauma, hormonal factors, and genetic predisposition in some cases[5].

Diagnosing fibromatosis of the head and neck region can be challenging due to its rarity and overlapping clinical and radiographic features with other benign and malignant maxillofacial tumours. The initial evaluation includes a thorough clinical examination, imaging studies such as computed tomography (CT) or magnetic resonance imaging (MRI), and a biopsy for histopathological analysis. The histological examination reveals a cellular proliferation of fibroblasts with a low mitotic index, infiltrating between the normal collagen bundles. Immunohistochemistry can aid in confirming the diagnosis by demonstrating positive staining for markers such as beta-catenin and vimentin[6]. The management of fibromatosis requires a multidisciplinary. The primary goal of treatment is achieving complete surgical resection while preserving the aesthetics and function of the affected region. However, due to the infiltrative nature of the tumour, achieving negative surgical margins can be challenging, and local recurrence rates remain relatively high.

In cases where complete resection is not feasible or would result in significant morbidity, alternative treatment options may be considered. These can include adjuvant therapies such as radiation therapy or systemic therapies such as nonsteroidal anti-inflammatory drugs (NSAIDs), selective estrogen receptor modulators (SERMs), or targeted agents[7].

Spindle cell neoplasms of the tongue encompass a diverse group of tumours characterized by the presence of spindle-shaped cells within the tongue tissue.

The differential diagnosis of spindle cell neoplasms of the tongue includes various benign and malignant entities such as fibromatosis, schwannoma, leiomyoma, fibrosarcoma, and spindle cell carcinoma[8]. Histopathological examination plays a crucial role in establishing the diagnosis. The tumour is characterized by proliferation of spindle-shaped cells with elongated nuclei, arranged in interlacing fascicles. Immunohistochemistry is often employed to further characterize the tumour, with markers such as smooth muscle actin, S100, and desmin[9] being useful in differentiating between various spindle cell neoplasms.

Surgical excision with clear margins remains the mainstay of treatment, with adjuvant therapies considered in select cases. Continued research and collaboration are necessary to further understand the molecular mechanisms underlying these neoplasms and develop targeted treatment strategies.

Spindle cell neoplasms of the tonsil are rare, accounting for less than 1% of all tonsillar tumours [10]. Myofibroblasts are type of contractile fibroblast that play a crucial role in wound healing and tissue repair[11] , myofibroblastic tumours are usually treated by excision but Low grade myofibroblastic tumours can be treated with radiotherapy [12]. Recent research suggests ALK 1 positive cases treatment with crizotinib may be considered [13].

Synovial sarcoma is a rare soft tissue malignancy that typically arises in the extremities of young adults[14]. However, its occurrence in the larynx is exceedingly rare, representing a diagnostic and therapeutic challenge.

Histologically, synovial sarcoma of the larynx demonstrates characteristic biphasic or monophasic patterns[15]. Biphasic tumours consist of both epithelial and spindle cell components, whereas monophasic tumours predominantly exhibit spindle cells. Immunohistochemistry can aid in confirming the diagnosis, with markers such as cytokeratin, epithelial membrane antigen (EMA), and Bcl-2 [16]being commonly expressed in synovial sarcoma.

A multidisciplinary approach, including surgery, radiation therapy, and systemic therapy when appropriate, is necessary for optimal management .Adjuvant radiation therapy is often considered to reduce the risk of local recurrence[17], especially in cases with high-grade tumours, close or positive margins, or evidence of perineural or lymphovascular invasion. Systemic therapy, such as chemotherapy, may be administered in advanced or metastatic cases, Doxorubicin and Ifosfomide are the commonly used agents .

Rhabdomyosarcoma is a malignant tumour that arises from immature skeletal muscle cells[18]. While it most commonly affects the head and neck region in children, rhabdomyosarcoma arising in the nose is relatively rare.

Histologically, rhabdomyosarcoma is characterized by the presence

of primitive round or spindle-shaped cells with eosinophilic cytoplasm. The tumour cells often display cross-striations or show positivity for muscle-specific markers, such as desmin and myogenin[19], on immunohistochemical analysis. Subtyping of rhabdomyosarcoma into embryonal, alveolar, or pleomorphic[20] types is important as it can guide treatment decisions and provide prognostic information.

The management of rhabdomyosarcoma of the nose requires a multidisciplinary approach involving surgeons, radiation oncologists, and medical oncologists. Surgical resection plays a role in the management of rhabdomyosarcoma of the nose, aiming to achieve complete tumour removal while preserving functional and cosmetic outcomes. However, the extensive local invasion and proximity to critical structures in the nose can make achieving negative surgical margins challenging. Therefore, adjuvant therapies are often employed to enhance local control. Radiation therapy may be utilized preoperatively, postoperatively, or as definitive treatment for unresectable or metastatic disease. Chemotherapy agents like vincristine is typically administered as part of multimodal therapy, often in conjunction with radiation therapy, to target micrometastatic disease and improve overall survival rates.

Surgical resection remains the cornerstone of treatment for localized head and neck sarcomas. The extent of resection depends on various factors, including tumour size, location, and invasion of adjacent structures. In cases involving the maxilla, tongue, and temporal muscle, achieving negative surgical margins while preserving functional and cosmetic outcomes can be challenging. The use of advanced surgical techniques, such as microvascular reconstruction and functional organ preservation, can help optimize outcomes in these cases. Adjuvant therapies, including radiotherapy and chemotherapy, may be employed in certain cases to improve local control and reduce the risk of recurrence.

Long-term follow-up is essential for patients with head and neck sarcomas to monitor for potential recurrences, metastases, or treatment-related complications. Regular surveillance imaging, clinical examinations, and functional assessments are necessary to detect any signs of disease progression or late complications. The duration and frequency of follow-up visits should be tailored to each patient's specific risk profile.

Among the seven cases presented in this series, three patients experienced recurrence of their soft tissue tumours [21] and one patient expired. The recurrence occurred after varying periods following the initial treatment, ranging from months to years. Understanding the recurrence patterns can provide insights into the behaviour of these rare soft tissue tumours in the head and neck region.

Several factors may contribute to the recurrence of head and neck sarcomas. Inadequate surgical margins [22], defined as the presence of tumour cells at or close to the resection margin, have been identified as a significant risk factor for local recurrence. The infiltrative nature of these tumours and their proximity to critical structures can make achieving clear margins challenging. In cases where complete resection is not feasible due to cosmetic challenge, residual microscopic disease may persist and lead to local recurrence.

Histological factors, such as high-grade tumours or the presence of aggressive histological subtypes, may also increase the risk of recurrence. Additionally, tumour size, depth of invasion, lymphovascular invasion, and perineural invasion have been implicated as potential predictors of recurrence in head and neck sarcomas.

Adjuvant therapies, such as radiation therapy and chemotherapy, may be employed in cases of recurrent disease to enhance local control and mitigate the risk of further recurrence or distant metastasis.

The aggressiveness of the recurrent disease, potential for distant metastasis, and limited treatment options in cases of refractory or recurrent disease contribute to the challenges faced in achieving long-term disease control.

Due to the rarity of head and neck soft tissue tumours, there is limited evidence available to guide treatment decisions and prognostic assessments. Collaborative efforts, including multi-institutional studies and international registries, are crucial to accumulate more data and improve our understanding of these tumours. The development of targeted therapies and personalized treatment approaches based on specific molecular alterations holds promise for the future management.

In conclusion, this case series highlights the complexity and challenges associated with head and neck sarcomas especially recurrence due to compromising margin due to cosmetic also due to tumours lying proximity to vital structures in Head and neck region. The cases presented underscore the importance of accurate diagnosis, multidisciplinary collaboration, and individualized treatment strategies. Further research and collaborative efforts are needed to advance our knowledge and improve outcomes for patients with head and neck soft tissue tumours.

Abbreviations:

CT: Computed Tomography
MRI: Magnetic Resonance Imaging
IHC: Immunohistochemistry
TNM: Tumor, Node, Metastasis (staging system)
WHO: World Health Organization
NSAIDs: Nonsteroidal Anti-Inflammatory Drugs
SERMs: Selective Estrogen Receptor Modulators
EMA: Epithelial Membrane Antigen
ALK: Anaplastic Lymphoma Kinase
PET CT: Positron Emission Tomography Computed Tomography
PMMC: Pectoralis Major Myocutaneous Flap
SMA: Smooth Muscle Actin
H- caldesmon: Heavy Caldesmon
HPV: Human Papillomavirus
SCC: Squamous Cell Carcinoma

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