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Rare and Unusual Tumors of the Head and Neck: Case Reports and Review of the Literature

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Abstract

We report three highly unusual primary head and neck malignancies: primary leiomyosarcoma of the sinonasal passages and cavities, Primary Cutaneous Mucinous Carcinoma (PMC) and primary cutaneous endocrine mucin-producing sweat gland carcinoma of the head and neck are extremely rare neoplasm's and one must rule out that these lesions represent metastatic lesions rather than primary lesions; a task that can be very difficult to do with absolute certainty.

Keywords: PMC; Leiomyosarcoma; PMSC; WHO

Introduction

There are numerous primary tumors of the head and neck that encompass a diverse spectrum of tissue types that clinicians and pathologists encounter in their everyday practice of medicine. Adding to this complex differential diagnoses physicians should always keep in mind that the lesion may not be primary and may represent a metastatic lesion. Primary leiomyosarcoma of the sinonasal passages and cavities, Primary Cutaneous Mucinous carcinoma (PMC) and Endocrine Mucin-Producing Sweat Gland Carcinoma (EMPSGC) of the head and neck are extremely rare neoplasm's and one must rule out that these lesions represent metastatic lesions rather than primary lesions; a task that can be very difficult to do with absolute certainty.

Case Presentation

Case 1

A 35 years old female who presented to the emergency room with epistasis and exam revealed a large fungating mass in the maxillary sinus. Biopsies revealed a high grade epithelioid and spindle cell neoplasm and the initial differential diagnoses were melanoma, sarcomatoid carcinoma, leiomyosarcoma and angiosarcoma (Figure 1). The tumor very much resembled leiomyosarcoma of the uterus however melanoma and sarcomatoid carcinoma are vastly more common. Immunohistochemistry demonstrated areas in which it was strongly positive for smooth muscle actin and negative for epithelial markers pankeratin, EMA and cytokeratin 5/6 (Cam 5/6) (Figure 2 and 3). It was also negative for melanoma markers and vascular markers. This workup revealed it to be most consistent with leiomyosarcoma.

Discussion: There have been approximately 100 cases of Primary Leiomyosarcoma of the sinonasal tract (PL) reported, including this case report. Agaimy reported that they represent 0.01% of all sinonasal neoplasm's [1]. They arise from the nasal cavities, ethmoid, frontal, orbital and maxillary sinuses. Predisposing factors include the clinical history of retinoblastoma, radiation and chemotherapy [2]. In the hereditary form of retinoblastoma, treated patients have a 20 fold increase in secondary malignancies compared to patients with the nonhereditary form of retinoblastoma [3]. The most recent WHO classification of tumors lists over 60 primary neoplasm's including several newly described lesions [4]. Included in the differential diagnosis of PL is primary carcinoma, melanoma, angiosarcoma and the newly described entity: biphenotypic sinonasal sarcoma which the authors have previously encountered. The latter neoplasm is a locally aggressive tumor with no reports of metastasis. It is comprised of medium sized, moderately pleomorphic, moderately hyperchromatic spindle shaped cells. Biphenotypic sinonsasal sarcoma is positive for smooth muscle actin and S100. In our experience with BSNS, the lesion had been completely excised and the patient tumor free for 36 months. In the present case of PL the tumor cells were extremely pleomorphic and very aggressive locally. It was strongly positive for smooth muscle actin and focally

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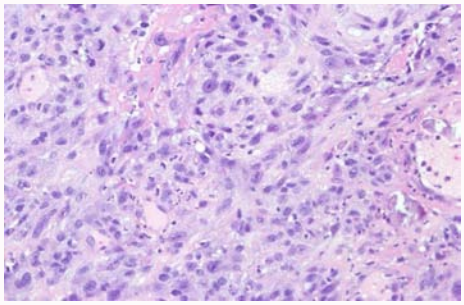

Figure 1: Malignant spindle cell neoplasm.

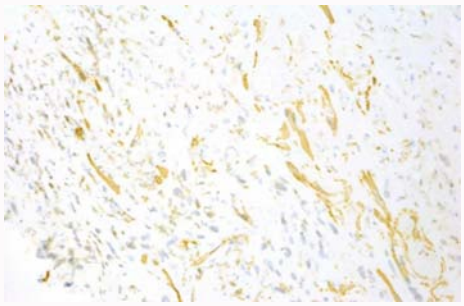

Figure 2: IHC for positive smooth muscle actin.

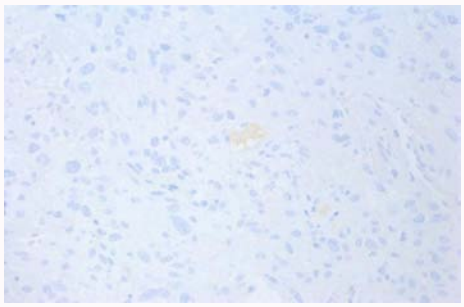

Figure 3: IHC for pankeratin, negative ruling out carcinoma.

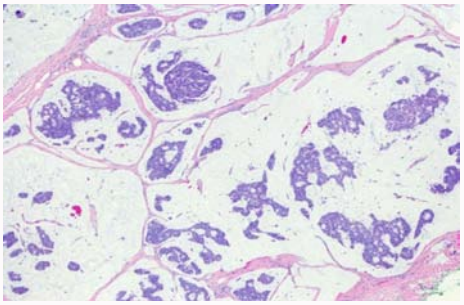

Figure 4: Malignant glands with abundant mucin.

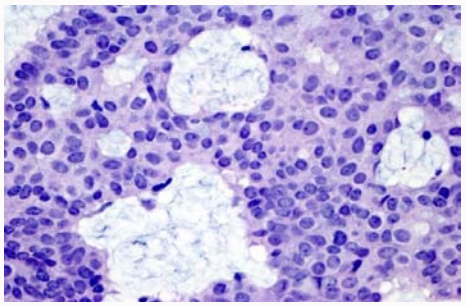

Figure 5: Bland, malignant glands.

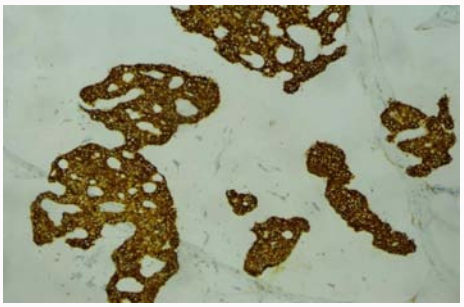

Figure 6: IHC for cytokeratin 7.

positive for caldesmon which stains cells that are myofibroblasts and does not stain epithelial cells. The tumor was negative for S100 and other melanoma markers, ruling out BSNS and melanoma. A complete work up did not reveal any other known primary. Also in the differential is a sarcomatoid carcinoma. All of the epithelial markers were negative in this case except that a consultant offered the suggestion that p63 may be focally positive and therefore remotely possible that this was a sarcomatoid carcinoma. It was felt that this interpretation was to afford the patient some form of treatment option. Generally, surgery is the treatment of choice with 5 year survival rates of 80% for surgery with wide margins, 59% with positive margins and 0% without surgery [5]. In the present case the tumor was extremely aggressive locally and surgery not an option and the tumor were described as growing is size by 50% in one week. Chemotherapy had been attempted but there was no response. As the tumor may have been carcinoma radiation could therefore be attempted, however it also had no significant effect on the tumor. The patient was admitted to hospice and presumably died of disease very soon after wards.

Case 2

The second patient is a 68 years old female diagnosed with breast cancer in 1979 status post right mastectomy and adjuvant chemotherapy and radiation. She presented to the otolaryngology clinic with a painless swelling of the left cheek. Biopsy demonstrated a mucinous adenocarcinoma (Figure 4 and 5). She has had no recurrence of the breast carcinoma after 40 years and has been followed with CT scans for evaluation of PET (+) left axillary LNs which she had biopsied on 5/14/2019 and was consistent with reactive lymph nodes, no malignancy noted. It was felt that the cheek lesion was most likely primary but of course this needed to be proved. The tumor cells were positive for CK7 and Gross Cystic Disease Fluid Protein however this does not rule out breast but does rule out GI origin (Figure 6 and 7). The most convincing evidence is the p53 stain which stains the basement membrane demonstrating of an in-situ component (p63 stain) (Figure 8).

Discussion: Primary cutaneous mucinous carcinoma is an unusual neoplasm with less than 200 cases reported [6]. Kazakov reported on 31 cases of pure mucinous carcinoma of the skin and all but one were located on the head and neck with 30% involving the eyelid [7,8]. It is comprised of rather bland cells that seem to be floating in pools of mucin. It is a locally aggressive tumor with recurrence rates nearly 30% and may metastasize to local nodes but rarely disseminates [9]. In general, the treatment is surgical with

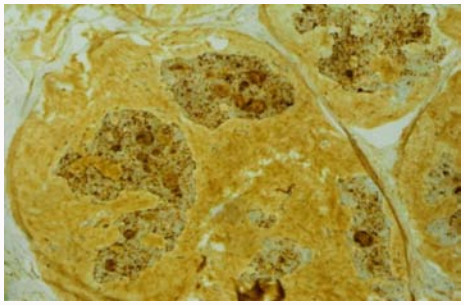

Figure 7: IHC for gross fluid disease cystic protein.

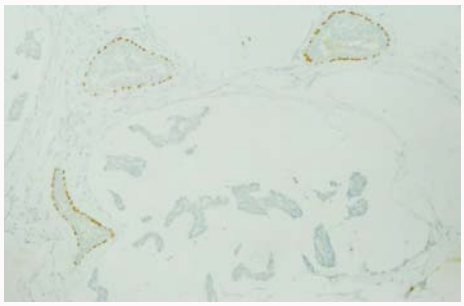

Figure 8: IHC for gross fluid disease cystic protein.

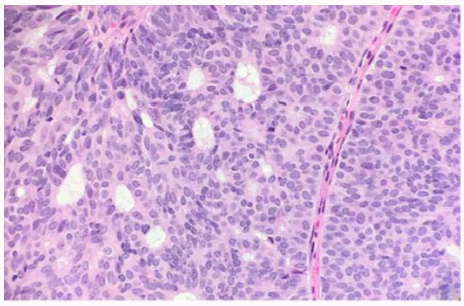

Figure 9: Malignant cribriform glands reminiscent of breast, no significant mucin in this field.

tumor free margins of 10 mms however this may not be possible as these lesions occur on the face near the orbit. Mohs microsurgery may be an option [10].

The most important and obvious neoplasm to consider, in a female, is metastatic breast carcinoma. In our experience with breast cancer the clinical history of breast cancer is generally known. Occult breast cancer accounts for 0.1% to 0.8% and usually presents as axillary adenopathy. In the present case the patient had a history of breast cancer nearly 40 years ago however there had been no recurrences. The typical IHC performed for CK7, EMA, and MUC 1 are positive in both breast and PMSC. 15 of 25 PMSC are also positive for GCDPF-15 and so IHC and histology may not be helpful. In the present case there was an in-situ component to the tumor demonstrated by p63 staining of myoepithelial layer in cytologically malignant in-situ glands adjacent to invasive mucinous glands, therefore the tumor was primary (Figure 8).

Case 3

The final case is that of a 54-years-old female presenting with a right cheek lesion. The biopsy of the lesion revealed a dermal-based nodular proliferation with unremarkable interspersed dermis,

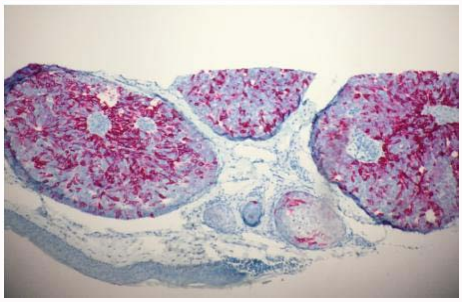

Figure 10: IHC for neuron-specific enolase (neuroendocrine marker).

underlying an uninvolved epidermis. These well-defined nodules displayed a cribriform pattern with focal intraluminal mucin (Figure 9). The morphology was very reminiscent of a ductal carcinoma of the breast. The neoplastic cells were uniformly round-to-oval with salt-and-pepper chromatin. Occasional mitotic figures were identified. Tumor necrosis was absent. Immunohistochemically, the tumor cells were strongly and diffusely positive for cytokeratin-7, neuron-specific enolase, and partially positive for synaptophysin demonstrating a neuroendocrine carcinoma. The patient had no relevant medical history and no breast lesions were found (Figure 10). The lack of a known breast primary combined with the histomorphological and immunostaining pattern strongly supported a diagnosis of a primary endocrine mucin-producing sweat gland carcinoma.

Discussion: Endocrine Mucin-Producing Sweat Gland Carcinoma (EMPSGC) is a rare cutaneous adnexal tumor possibly originating from the apocrine glands of Moll, accounting for <0.2% of cutaneous adnexal malignancies and less than 60 cases have been reported. The mean age at diagnosis is 70 years, without gender predilection, commonly seen in the face, particularly the eyelids. EMPSGC usually presents as a slow growing lesion. Morphologically and immunohistochemically, EMPSGC is analogous to endocrine ductal carcinoma in-situ and solid papillary carcinoma of the breast. EMPSGC displays variable architectural patterns, including solid, cystic, papillary, and cribriform. In contrast to the previous case which demonstrated malignant glands in pools of mucin, mucin production may be inconspicuous, such as seen in this last case. EMPSGC are often GATA3, ER and PR positive, posing a differential diagnostic challenge with breast carcinoma [11]. A thorough clinical examination ruled out the possibility of a metastatic papillary breast carcinoma, the most likely origin of a metastatic lesion [12]. Most EMPSGC behave in a benign fashion but the prediction of the clinical course is limited by the rarity of the tumor.

Conclusion

There are numerous neoplasms of the head and neck with new lesions being discovered and defined daily, each of these having different treatment modalities. Physicians need to be familiar with primary lesions, and should always be aware of the possibility of a metastasis as these may or may not have different treatment options. A lesion that is determined to be metastatic does not necessarily mean that surgery may not be a treatment option, however surgery does decrease tumor burden and may be the only treatment option available. In the three cases described here, a metastatic lesion was most likely however they were determined to be primary. In this scenario, surgery is the mainstay of treatment and will probably remain the primary treatment modality even though, most likely, some form of biologic/immunologic therapy will be discovered in

the future. Finally, metastatic tumor does generally imply a worse prognosis for the patient and so this determination is critical.

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