Extra ocular Sebaceous Carcinoma

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Abstract

Sebaceous carcinomas are relatively rare cutaneous tumours. There are fewer than 600 cases reported in literature. They most commonly arise within the meibomian gland of the eyelid. Extra-ocular sebaceous carcinoma is an extremely rare tumour; less than 150 cases reported in literature are extra ocular. Of these 75% cases involve head and neck. Other reported locations of extra-ocular sebaceous carcinoma include external genitalia, breast, parotid and oral mucosa. These tumours have a high incidence of local recurrence and regional metastasis.

We present a case of an extra-ocular sebaceous carcinoma at the back of neck in 48 years old male.

The relationship of sebaceous tumors and visceral malignancy is well established in literature. The diagnosis of extra-ocular sebaceous carcinoma should suggest the possibility of Muir-Torre syndrome and should prompt a search for an internal malignancy, genomic mutations and screening of family members. Investigations like USG abdomen, CT scan & colonoscopy did not reveal any positive findings, hence Muir-Torre syndrome was ruled out in our case. Surgery with wide surgical margins is the standard treatment, which was done in our case.

KEYWORDS: Extra ocular, sebaceous carcinoma, Muir – Torre Syndrome.

INTRODUCTION

Sebaceous gland carcinomas are rare tumors1. The ocular sebaceous Carcinomas (SC) occur most frequently on the eyelids and it accounts for 1-5% of malignant ophthalmic tumors. Extra ocular sebaceous carcinoma is an extremely rare tumor and of these, 75% involved head and neck2. Other reported locations include external genitalia3, external auditory meatus, nasal vestibule4, axilla, breast5 and pinna6. Cystic presentation of sebaceous neoplasm is rare. Recently, cystic sebaceous neoplasia (CSN) has been characterized as a marker lesion of Muir-Torre Syndrome (MTS)7. We present one such rare case of extra ocular sebaceous carcinoma in adult male.

CASE REPORT

A forty Eight year old male presented with swelling on the nape of neck since one year.
It started as a painless nodule one year back and increased progressively to 5 x 4 cm size. No submandibular, cervical or axillary lymph nodes were palpable. A provisional diagnosis of squamous cell carcinoma was made and wide local excision was done.

**GROSS FINDINGS:**
On gross evaluation, it was seen as multilobulated mass of 5 x 4 cm with skin at the base of the mass. No ulceration was presented (Fig 1). On cut section tumor had yellowish hue in fresh state (Fig 2).

**HISTOLOGICAL FINDINGS:**
Section from the tumor show moderately differentiated infiltrative carcinoma involving dermis and subcutaneous fat. Tumor cells have vacuolated cytoplasm are arranged in nests (Fig 3). The peripheral cells have a basaloid appearance and very high mitotic activity of 3-5 mitoses per high power field (Fig 4). The vacuolated cells are positive for oil red O and Sudan V (Fig 5). The tumor cells are positive for cytokeratin and isolated tumors cells show S100 positivity.

A diagnosis of sebaceous carcinoma was made.

Complete ophthalmic examination was done to rule out primary ocular sebaceous carcinoma. Work up for other primary malignancies included sonography and tomography of the chest, abdomen and pelvis was done to look for associated visceral malignancies.

**DISCUSSION:**
Carcinomas originating from sebaceous glands are ocular and extra ocular type. The ocular type most frequently occurs on eyelids and account for approximately 0.2% of all eye lesions and 1-5% of ophthalmic malignant tumors. Extra ocular sebaceous carcinoma is rare tumor with fewer than 120 cases reported. Of these, 75% involved the head and neck. Other reported locations of extra ocular SC include external genitalia, breast parotid and oral mucosa. The age distribution of extra ocular is similar to periocular SC with average age of 63 years. Few cases of extra ocular SC are reported in children and young adults.

The present case of extra ocular sebaceous carcinoma is in forty eight old male patient at the nape of neck.

Grossly sebaceous carcinoma present as nodules or cauliflower like masses. Cystic lesions are present in patients with Muir-Torre syndrome. The present lesion on cut section is solid with no cystic areas.

Histologically SC reveals asymmetric poorly circumscribed dermal tumor with variably sized lobules separated by fibro vascular stroma. The cells show variable sebaceous differentiation characterized by finely vacuolated or foamy cytoplasm. The vacuolated cell demonstrate abundant lipid. The nuclei are round to oval with vesicular chromatin and prominent nucleoli. Ocular SC show pagetoid involvement but is rarely present in extra ocular SC. Well differentiated tumors shows abundant cytoplasm, vesicular nuclei and less mitosis in contrast to poorly differentiated SC where cells are more pleomorphic with high N:C ratio and have brisk mitosis. SC may exhibit basaloid, squamous or carcinoid like growth pattern.

The tumor is arranged in sheets and lobules in this case and most of the cells show sebaceous differentiation with few basaloid cells. The mitosis is 3-5 per high power field. Hence according to classification by Font, the present case is moderately
differentiated extra ocular sebaceous carcinoma. Complete ophthalmic examination was done to rule out primary ocular sebaceous carcinoma.

For equivocal cases immunohistochemistry may be employed. The tumor cells stain positive for epithelial membrane antigen (EMA) and androgen receptors but not for CEA and S100 protein. On immunohistochemistry, the tumor cells in our case show cytokeratin positivity and are negative for S100.

A minority of individuals with extra ocular SC has Muir-Torre syndrome (MTS). Recently, cystic sebaceous neoplasia (CSN) has been characterized as a marker lesion of Muir-Torre Syndrome (MTS). In subset of MTS families, the disease is due to an underlying DNA-mismatch repair defect and about half of these patients are affected by colorectal and genitourinary malignancies. Genomic mutations of genes Hmsh2 and hMLH1 are well documented in patients with MTS. The investigations like USG abdomen, CT scan abdomen and colonoscopy do not reveal any internal malignancy in our case hence Muir-Torre Syndrome is ruled out. Screening of family members for sebaceous neoplasm and Muir-Torre Syndrome do not reveal any positive findings.

The primary treatment of SC is complete surgical excision. Sebaceous carcinoma is relatively radio resistant and chemotherapy has got a very limited role in the management of this carcinoma. In this case, wide local excision was done.

Diminished retinoid receptors (RXR- Beta and RXR-gamma) expression is thought to be related to development of SC; hence combination of Interferon with retinoids (Isotretinoin) at the dose of 0.8m/kg/day seem to be of promise in preventing tumor development in MTS.

Patients with SC should be followed up for detection of possible metastasis. There is no recurrence or metastasis in the case presented after two years follow-up.

CONCLUSION:

Extra ocular sebaceous carcinomas are rare. The diagnosis of extra ocular sebaceous carcinoma should suggest the possibility of MTRS and should prompt a search for associated internal malignancies and genomic mutations. Family members should be screened to detect the sebaceous neoplasms in early stage and should be screened for internal malignancies to rule out Muir-Torre syndrome.

BIBLIOGRAPHY

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Fig 1: Cauliflower like tumour.

Fig 2: Cut surface of tumour.
Fig 3: Tumor cells with vacuolated cytoplasm and sebaceous differentiation. (H& E stain 40 X)

Fig 4: Tumor cells with vacuolated cytoplasm and sebaceous differentiation. (H& E stain 40 X)