Muir-Torre syndrome – a case report and review of literature

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Citation

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Abstract

Muir-Torre syndrome (MTS) is a rare autosomal dominant condition in which one or more primary malignancies occur together with a sebaceous gland tumor. We report a case of this rare syndrome who presented with a sebaceous epithelioma occurring in association with invasive ductal carcinoma breast (No special type-NST) in a 63 years old female patient. Implications for diagnosis, management and screening of the family members are discussed.

INTRODUCTION

Muir-Torre syndrome is defined by the combined occurrence of at least one sebaceous skin tumor and one internal malignancy in the same patient first described by Muir (1967) [1] . The cutaneous lesions most commonly seen are sebaceous adenoma, epithelioma or carcinoma and keratoacanthoma (KA). Among the visceral malignancies, colorectal carcinoma is most commonly seen [12] .

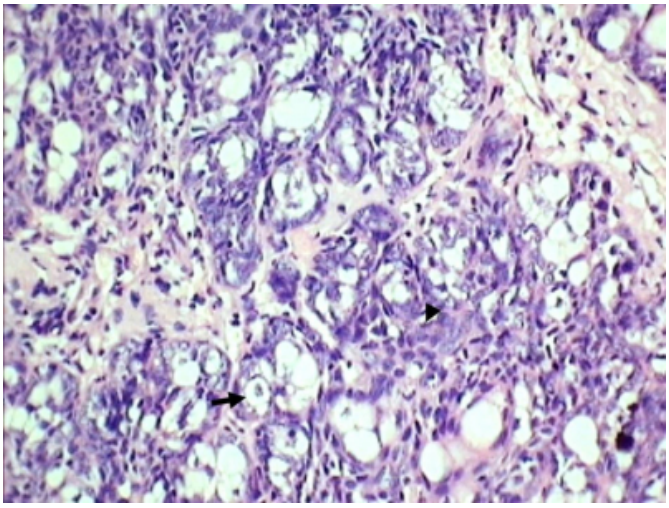
CASE REPORT

A 63 years old female presented with a nodule on the forehead since last four years. Three years earlier the patient underwent modified radical mastectomy for invasive ductal carcinoma breast (NST). The lesion on the forehead was not excised at that time. Since last few months she was having mild pain in the nodular swelling so excision biopsy was carried out.

Gross examination of the resected specimen revealed a nodular mass measuring 1.5x1x1 cms. Overlying bit of skin measured 1x0.5 cms .Cut section was solid with grey white to grey yellow areas. Microscopic examination showed multiple well circumscribed nodules located in the dermis with overlying atrophied epidermis. These nodules were composed of sheets and nests of undifferentiated basaloid cells with aggregates of mature sebaceous cells interspersed in between. Basaloid cells accounted for 70-80% of tumor cells while the rest were mature sebaceous cells (Figure 1).

Figure 1

Figure 1- Photomicrograph showing tumor with sheets of basaloid cells (Arrowhead) admixed with mature sebaceous cells (Arrow) (H&E, x 400).



DISCUSSION

MTS represents the association of multiple sebaceous tumors along with one or more visceral malignancy occurring in the same individual in the absence of other precipitating factors such as radiotherapy or AIDS [13] . The cutaneous lesions often precede the first manifestation of internal malignancies [4] .

Typical skin tumors associated with this syndrome include sebaceous adenomas, epitheliomas and carcinomas. Keratoacanthomas and basal cell carcinomas with sebaceous differentiation also occur. All these sebaceous gland tumors are rare in the general population; the finding of such a

tumour is a marker for MTS and should prompt a search for occult malignancy [12].

Colorectal cancer is the commonest visceral neoplasm to occur in MTS, and the most frequent initial cancer [3], although a wide tumor spectrum exists including carcinoma of the genito-urinary tract, breast, head and neck, small intestine and lymphomas [34]. In the present case, sebaceous epithelioma was seen in association with primary invasive ductal carcinoma (NST) in the breast. Many a times, these skin lesions are missed due to lesser importance in comparison to the associated malignancies. Every patient with multiple KA should be evaluated for the presence of sebaceous neoplasms, the absence of which still requires consideration of this syndrome [4]. Sebaceous gland tumors are rare and the diagnosis of such a tumor should suggest the possibility of the syndrome and prompt a search for associated malignancies [34].

The genetic disorder in MTS is an autosomal dominant inherited germline mutation in one of the DNA mismatch repair genes, most commonly hMSH2 [5]. It is inherited with a high degree of penetrance and variable expression. The male:female ratio is 3:2. Children of an MTS individual, therefore, have a 50% risk of inheriting the cancer predisposition. In families where the germline mutation can be identified, those individuals who have inherited the mutation should be offered regular screening examinations [6].

CONCLUSION

Muir –Torre syndrome is very rare. The management should be multidisciplinary including genetic counseling, regular dermatology follow-up and relevant cancer screening. Early recognition of the syndrome in patients with sebaceous gland tumours should facilitate early detection of subsequent visceral malignancies if the patient is entered into appropriate screening programmes.

References

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