Answers

Case 1

DIAGNOSIS

Multiple Endocrine Neoplasias type 2B (MEN2B).

Thyroid gland needs to be investigated for malignancy.

BACKGROUND

Multiple endocrine neoplasias type 2B (MEN2B) is an inherited disorder characterized by the development of endocrine malignancies. It results from germline mutations in the RET proto-oncogene and is transmitted in an autosomal dominant fashion. It is characterized by following features:

- Aggressive and penetrant Medullary Thyroid Carcinoma (occurring in 100% of cases)
- Pheochromocytoma (50%)
- Mucosal neuromas (95%-98%)
- Intestinal ganglion neuromas (40%)
- Nearly all patients have a distinct marfanoid habitus

OUR PATIENT

Our patient exhibited a distinct marfanoid habitus with multiple oral mucosal swellings (Fig 1&2) . The biopsy proved these swellings to be neuromas(fig 3). Evaluation of upper GI revealed similar swellings which were not biopsied but are likely to be same. Biopsy of thyroid confirmed a subclinical medullary thyroid carcinoma in early stages. Thyroidectomy was performed with complete cure of the malignancy. RET gene analysis confirmed the mutation. The patient was put on life time surveillance for development of further malignancies.

Cases courtesy:

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Case 2

DIAGNOSIS

Multiple Myeloma with Paraneoplastic Addisonian Hyperpigmentation.

BACKGROUND

Paraneoplastic Dermatoses are cutaneous reaction patterns of internal malignancy which are due to different substances secreted by the tumor and resemble a number of dermatoses. There is no actual involvement of the skin with the tumor. Recognition of these changes can be very helpful in early diagnosis internal malignancy or may be the earliest symptom of relapse of a previous cancer. Classical Addisonian pigmentation is seen in Addison's disease and comprises of generalized black brown pigmentation which is accentuated in sun exposed areas, over pressure points and palmar creases. In addition there is nail and oral mucosal pigmentation. This type of pigmentation may also be seen as a paraneoplastic phenomenon in bronchogenic carcinoma as result of secretion of melanogenic substances by the tumor.

OUR PATIENT

Our patient presented to Dermatology Department for evaluation of Addisonian type black brown pigmentation with accentuation over sun exposed sites, oral mucosa, nails and palmar creases (Fig 1). The patient was investigated for Addison's disease. Morning and evening serum cortisol levels were within normal limits. Plain X-rays abdomen did not reveal calcification of adrenal glands. Instead permeative pattern of bone destruction was seen in the lower ribs bilaterally. Their were Multiple lytic lesions in the ribs and vertebrae. X-rays showed lytic lesions in the skull (Fig 2). Diagnosis of multiple myeloma was confirmed on serum protein electrophoresis (Fig 3) and bone marrow. The patient was referred to oncologist for further management

This case was initially suspected to be suffering from Addison's disease on the bases of the characteristic pigmentation but during investigations was found to be suffering from undiagnosed multiple myeloma and there was no evidence of adrenal dysfunction thus highlighting the importance of suspecting para neoplastic dermatoses.