WHAT IS YOUR DIAGNOSIS?

Three year old female child was admitted with history of excessive heat all over the body, sparse scalp and body hair and abnormal dentiton. She was born to consanguinous parents as the last of 4 children. An elder brother has similar problem. Patient was noticed to have club foot at the time of birth. She developed chronic ear discharge in infancy and suffered from frequent upper respitory infection and occasional diarrhoea. There was no personal or family history of atopy.

Examination showed an active alert child of normal stature. Her facies was characterised by saddle nose, evertid lips and prominent supraorbital ridges with sparse eyebrow hairs. Dry crusting was present in the nostrils and pus discharge in both ears. Skin was generally ichthyotic, atrichotic and anhidrotic. Scalp hair was sparse, scraggly, brownish, short and dry. 2 peg shaped upper molars were the only teeth present. Eyes were normal. Salivation was normal. Gums were normal. Palms and soles were tylotic. Examination of the affected male sibling showed identical facies and skin. Both parents were normal.

Differential diagnosis

1. Congenital anhidrotic ectodermal dysplasia
2. Congenital syphilis
3. Cretinism
4. Block-sulzbager syndrome

Final diagnosis : Congenital anhidrotic ectodermal dysplasia

Facies and symptoms were characteristic of the above diagnosis. Biopsy of the skin showed marked hypoplasia of eccrine sweat glands, sebaceous glands and hair follicles. The child probably has the rarer form of autosomal recessive type of the disease in which affected males and females present with a picture identical to that of males with the more common x-linked form of the disease. Inability to tolerate common bacterial and viral infections as seen in this child is a recognised feature of this disease. The ichthyosis and hyperkeratosis of skin of palms and soles as observed in this patient and her sibling are unusual in anhidrotic ectodermal dysplasia.

ANSWERS

A. The patient was most probably suffering from mycosis fungoides. A small degree of infiltrate would tend to suggest that chronic superficial dermatitis and parapsoriasis en plaque are unlikely. The lack of loss of sensation and the absence of thickening of nerves made the diagnosis of borderline leprosy untenable. The skin involvement was out of proportion to lymphnode involvement and hence was not in favour of diagnosis of Hodgkin's disease. Asymptomatic large plaques made the diagnosis of mastocytosis difficult, despite positive Dariers' sign.

B. Despite a large number of mast cells, the abundance of histiocytes in the infiltrate suggested a diagnosis of mycosis fungoides. The presence of mast cells were misleading but has been reported in patients with mycosis fungoides[1]. The absence of AFB confirmed that the patient did not have leprosy.

With the diagnosis of infiltrated and nodular form of mycosis fungoides, the best line of treatment seemed to be a combination of cyclophosphamide and prednisolone. The patient improved remarkably but a recurrence cannot be excluded. There is no immediate danger to life, though some of these patients behave unpredictably.

Mikhail G R and Milinska A M : Mast cell population in human skin, J Invest Dermatol, 43 : 249, 1964.

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