

WHAT IS YOUR DIAGNOSIS ?

A forty year old male patient complained of scaly lesions over the elbows, hands, knees and feet since the age of 2 years. He was born to consanguinous parents. There was no family



Fig. 1



Fig. 2

history of similar skin disease. He gave history of having lost his teeth early in life.

On examination patient was found to have uniform hyperkeratosis of the palms and soles with rough scaling extending on to the dorsa of hands and feet. Circumscribed scaly patches were seen on elbows, buttocks and knees. Hair, nails and mucous membranes were normal. Patient was found to be edentulous.

- Differential diagnosis :**
1. Mal de meleda
 2. Papillon-Lefèvre syndrome
 3. Hidrotic ectodermal dysplasia
 4. Gottron's syndrome

Final diagnosis : Papillon Lefèvre syndrome

The absence of family history and the presence of parental consanguinity suggested a recessively inherited disease. Thus Gottron's syndrome and hidrotic ectodermal dysplasia were considered unlikely. Further marked teeth abnormality is not a recognised feature of either of the above. Mal de Meleda is not associated with any specific defects of teeth whereas nail changes are frequent in this condition.

Thus this patient whose main features are marked palmoplantar hyperkeratosis, circumscribed areas of scaling on elbows, knees, buttocks and loss of teeth early in life is suggestive of papillon Lefèvre syndrome, acquired as a recessive trait from consanguinous parents. X-ray of the skull showed no abnormal calcification.