

## WHAT IS YOUR DIAGNOSIS?



Fig. 1



Fig. 2



Fig. 3

45 year old male, cooly by profession, presented with the complaints of discolouration of skin, thickened localised skin lesions of palms and pain in the large joints and back, of 4 years duration. His mother, suffered from similar troubles ; died 20 years ago. He is the only child of his parents, not married.

Examination showed severe sun burn appearance of skin (Fig. 1) on exposed parts of his body, bluish black infiltrated areas of both ears, linear bluish black keratotic plaques along the borders of the palms (Fig. 2) and soles and pigmentation of the conjuction near the inner canthi of both eyes (Fig. 3). He also showed difficulty in bending and pain in the knees and shoulders.

## Differential Diagnosis

1. Arsenical pigmentation and Keratosis.
2. Generalised melanosis and secondary deposits in the spine due to malignant melanoma.
3. Alkaptonuria (Ochronosis)
4. Senile keratosis and elastosis with severe sun burn.

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Alkaptonuria is a rare metabolic disorder of Tyrosine, inherited as a recessive trait, due to absence of homogentisate oxidase which is necessary for oxidation of Homogentisic acid. As a result, Homogentisic acid accumulates in cartilage, intervertebral discs, skin, conjunctiva and many other tissues and is excreted in the urine. Homogentisic acid polymerizes to form brown to dark pigment in urine and tissues. No satisfactory treatment is available.

## Alkaptonuria (Ochronosis).

### Final Diagnosis

Urine turned dark, on standing for sometime and also on treatment with Benedict's solution.

X-rays showed decalcification of bones and narrowing of intervertebral spaces with dense calcification of intervertebral discs in the dorsal and lumbar region. Mild osteo-arthritis changes are noted in the hip, knee and shoulder joints. The cartilage of right ear showed dense calcification.

Biopsy of the palmar lesion showed marked hyperkeratosis and flattened epidermis. Upper dermis showed severe degeneration of collagen with deposits of homogenuous clumps of brownish pigment.