CLINICAL QUIZ

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2-year-old female child was brought with the complaints of blackish staining of wet nappy on drying since birth (Fig-1). She was the only child born of a nonconsanguineous marriage and was thriving well.

Fig. 1 : Stained garment of patient

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Her developmental milestones were normal. Clinical examination revealed no abnormality. Silver nitrate and Benedict's test were performed on the urine sample and found to be positive (Fig-2).

Fig. 2 : Urine test with control (L to R) Control, Urine on exposure, Benedict's test, and silver nitrate test

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Answer to Clinical Quiz

A CASE OF ALCAPTONURIA

Diagnosis: Alcaptonuria.

Alcaptonuria is a rare inborn error of metabolism of the amino acid tyrosine with an autosomal recessive inheritance. Its incidence is <1:500,000 [1]. In a study of inborn errors of metabolism in north India it was found to be among the four commonest disorders of amino acid metabolism and the frequency pattern of various amino acid disorders was found to be remarkably different from that observed in the West [2].

The disease results from a deficiency of homogentisic acid oxidase. Tyrosine is degraded to homogentisic acid (HGA) which is further metabolized by HGA oxidase to malylacetoacetic acid. Deficiency of HGA oxidase leads to accumulation of HGA which is excreted in urine imparting a black colour to the urine if allowed to stand and undergo oxidation. In addition retained oxidized HGA accumulates in collagen tissues (ochronosis) resulting in pigmentation of sclera, conjunctiva and skin [3]. It may also lead to ochrono-

tic arthritis in the 4th decade and rarely pigment stones of kidney and prostate or degenerative cardiovascular disease [4]. Confirmation of diagnosis may be done by urinary chromatography/enzymatic assay [5]. There is no specific treatment. Vit C was found to be useful in-vitro studies only.

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