CLINICAL IMAGES

A Case of Hypertrichosis

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This 15 year boy with history vesicular skin eruptions on exposed parts for last five years was referred to the Department of Endocrinology for excessive hair on the body. There was also history of passing high colored urine which turned burgundy red on exposure to light. Examination revealed Hypertrichosis on face, forehead and neck, pinched nose, sclerodermoid facies, and splenomegaly. There were numerous skin lesions in the form of vesicles and bullae on hand, feet and face, and hyper- and hypo-pigmented spots over hands and feet with whitish nails and onycholysis. Investigations revealed high serum LDH and evidence of haemolysis. His urine tested positive for Porphyrin as well as Uroporphyrin /

FIGURE 1 & 2: Hypertrichosis of face

FIGURE 3 & 4: Hyper- and hypo-pigmented spots over hands and feet with whitish nails and onycholysis

FIGURE 5: Patient's urine turning burgundy red in color on exposure to light

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Coproporphyrin. Stool also showed Coproporphyrin.

From history examination and investigations a provisional diagnosis of Congenital erythropoietic porphyria (CEP) is made with a differential diagnosis of Hepatoerythropoietic porphyria (HEP) and Porphyria cutanea tarda. For final diagnosis measurement of Erythrocyte porphyrin levels by high performance Liquid chromatography is needed.