

A Case of Erythropoietic Protoporphyrria

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Summary

A case of erythropoietic protoporphyria in a Malay male presenting with a history of photosensitivity was investigated and had increased fecal porphyrria and urine coproporphyrin levels. Blood film showed presence of fluorecytes. This is the first case of EPP reported in Malaysia.

Key words – Erythropoietic protoporphyria photosensitivity, coproporphyrin fluorecytes.

Case History

A 39-year old Malay male presented with a history of photosensitivity following exposure to sunlight. The skin became itchy for about 20 minutes and was followed by blister formation. The affected skin subsequently showed signs of peeling off. He has had similar experience since the age of 12 years and has learned to avoid unnecessary exposure to sunlight since then. Examination showed areas of hyperpigmentation and thickening of skin over metacarpo-phalangeal and proximal interphalangeal joints. There were no other abnormalities. His parents were not related and neither had any history of photosensitivity. There was no family history of photosensitivity. He gave no history of any drug medication.

Laboratory investigations showed a normal blood film with haemoglobin of 12 g/dl, normal blood lead level of 0.8 umol/l (reference range up to 1.8 umol/l), normal 24 hour urine output of lead of 0.11 umol (reference range up to 0.5) and delta aminolevulinic acid (ALA) of 20 umol (reference range up to 40). His serum alanine aminotransferase (ALT) activity was elevated at 247 IU/L (reference range up to 40) which declined to 68 IU/L 4 weeks after exposure to light.

In view of the history of photosensitivity, ultraviolet light examination of his teeth was carried out and showed no abnormality. Urine, faeces and heparinised blood were collected for porphyrin study. Urine showed presence of urobilinogen¹ and absence of porphobilinogen. Porphyrin screening test² showed marked increase of porphyrin in the faeces and trace amounts of porphyrin in the urine. Thin layer chromatography of methyl esters of porphyrins³ in faecal and urine extracts along with protoporphyrin and coproporphyrin standards showed marked increase of protoporphyrin in the faeces, and trace of coproporphyrin in the urine. The faecal protoporphyrin and coproporphyrin concentrations were 343 nmol/g (reference range up to 53 nmol/g) and 4 nmol/g (reference range up to 31 nmol/g) of dried faeces respectively. Urine coproporphyrin concentration was elevated at 336 nmol/l (reference range up to 115 nmol/l).

Examination of blood film prepared from the heparinised blood showed the presence of fluorecytes (Fig. 1) which underwent photoinactivation within 45 seconds in ultraviolet light and no significant changes in birefringence of the red cells was noted when examined in compensated polarised light.⁴ The erythrocyte protoporphyrin level⁵ was 12,200 nmol/dl of erythrocytes (range up to 280). Transmission electromicroscopy (using Phillip CM 10 transmission electron-microscope, Eindhoven, The Netherlands) of ultra-thin section of red cell concentrate (fixed in 4% glutaraldehyde and 1% osmium tetroxide embedded in epoxy resin and stained with uranyl acetate and lead citrate) showed intracellular aggregates of electron dense reticulated rods (Fig. 2).

**All correspondence to S. L. Ch'ng.*

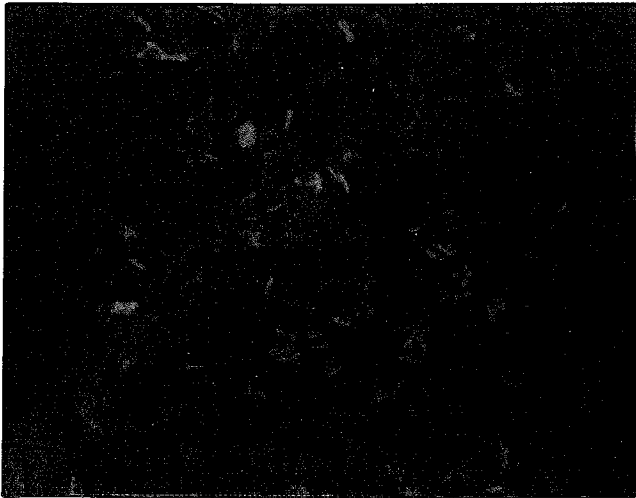


Fig. 1
Showing fluorecytes in blood film
examined in ultraviolet light.

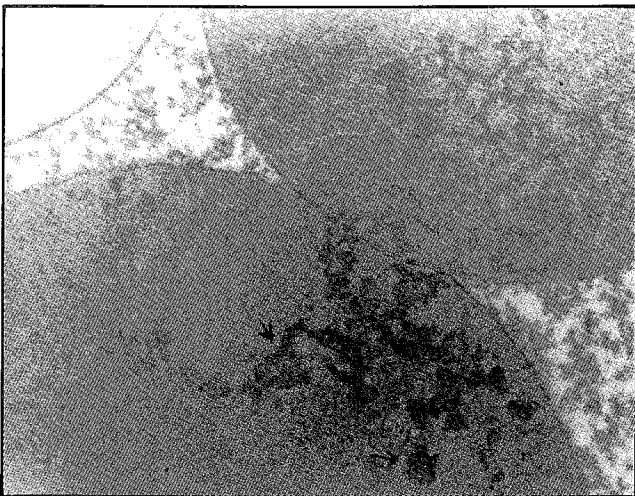


Fig. 2
Showing electron dense aggregates
(marked) within erythrocytes stained
with uranyl acetate and lead citrate
(total magnification was 36000 x)

Other investigations included ultrasound scan of the gall bladder which contained no stone. In view of the above findings a diagnosis of erythropoietic protoporphyria was established. He was treated with beta-carotene and advised to avoid exposure to sunlight. No further episodes of photosensitivity was noted. The fluorecytes persisted after treatment. His liver function test was monitored regularly and the subsequent clinical course was uneventful. Repeated examination of th heparinised blood (stored at 4C in the dark) after one month and wet blood film covered with glass cover (similarly stored) after 3 days still showed the presence of fluorecytes.

Discussion

This patient showed the classical clinical and chemical pathological features of erythropoietic protoporphyria (EPP) with the presence of labile fluorecytes (which indicated protoporphyrin rather than coproporphyrin⁶), elevated red cell and faecal protoporphyrin levels. The normal blood lead level, normal urine lead and dela- ALA output, normal faecal and increased urine coprophorphyrin levels excluded lead poisoning and congenital erythropoietic porphyria as the cause of the clinical features. The increased serum ALT, presence of urobilinogen and coproporphyrin in the urine indicated possible liver involvement. The electron dense aggregates most probably was due to protoporphyrin which chelated with the electron dense lead ions.

We believe this is the first documented case of EPP in Malaysia. It is one of the inborn errors of metabolism which is thought to be rare in this country. Our recent study⁸ using simple laboratory techniques and optimised test strategy showed that these conditions (such as phenylketonuria, maple syrup urine disease, methylmalonic aciduria, mixed carboxylase deficiency, galactosaemia and others) do exist in our local population and a majority of them are treatable. The laboratory techniques used in the investigation of this case are unsophisticated (except fluorescence spectrometry, fluorescence and electron microscopy) and can be readily performed in most peripheral hospitals. As the erythrocyte porphyrin is stable if kept in the dark at 4°C, the blood specimen can be transported to the central laboratories for further investigations. This along with other simple techniques could help in the initial assessment of the prevalence of inborn metabolic disorders.

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