Abstract:
Variegate porphyria is a rare autosomal domi-
nant disorder which occurs due to a deficiency of
the enzyme protoporphyrinogen oxidase (1,2).
The name variegate was first coined by Dean and
Barnes (3). This disease was first described in
south African white population. The disease fre-
quency is 3 cases per 1000 persons. The disease
shows a varied presentation which includes neu-
rovisceral, cutaneous manifestations or both and
hence its name.

Case report:
A 22 Year old female presented with complaints
of recurrent skin blisters over the face and both
hands for the past 1 year. These blisters were
painful. The blisters were noted more when ex-
posed to sunlight. These blisters healed sponta-
neously with scarring. She had no history of ab-
dominal pain, convulsions or paralysis. She was
married and has one child. Her child was perfect-
ly normal. No other family members including
her parents, grandparents and cousins were
affected.

On examination, she was moderately built and
nourished. Her systemic examination did not re-
veal any abnormalities.
Multiple erythematous maculopapular rashes were seen over her face, the extensors of the forearms and over the dorsum of both the hands. There were also areas of hypo and hyper pigmentations. Nails, teeth and hair were normal.

Erythematous maculopapular rashes over the dorsum of both the hands

Both qualitative and quantitative tests for porphyrins were carried out. Urine was dark brown in colour and showed reddish brown discolouration when exposed to sunlight. Examination of urine by woods lamp showed reddish fluorescence due to increased porphyrins. Direct examination of the stool sample with woods lamp showed reddish fluorescence. Fluorescence emission spectrometry of plasma showed an emission peak at 625 nm unique to variegate porphyria.

Reddish fluorescence of urine in dark with woods lamp

TEST FOR PORPHOBILINOGEN IN URINE

1. Ehrlich’s aldehyde test (Hoesch test)

Reagents - 2% para-dimethyl amino benzaldehyde in 2N HCl.

Procedure - Add 1 ml of ehrlich’s reagent to 5 ml of fresh urine. Wait for 10 mins.

Reddish brown discolouration of patient’s urine when compared with normal urine

Routine investigations including complete blood picture, liver and renal function tests, serum magnesium and serum cholesterol were within normal limits.

An Initiative of The Tamil Nadu Dr M.G.R. Medical University
A Pink colour develops when urobilinogen or porphobilinogen is present in the urine. The two substances are distinguished by adding 5 cc of chloroform and shaking. Pink colour produced by porphobilinogen is not extracted in the chloroform layer.

Observation- No pink colour developed in the patients sample.

Inference- Absence of porphobilinogen in the patients urine.

2. Watson-Schwartz test

Watson Schwartz test is used as a simple screening test for the presence of elevated urinary porphobilinogen.

Reagents

a. Ehrlich’s aldehyde reagent.

Mix 0.7gm p-dimethyl aminobenzaldehyde in 150ml of conc Hcl and 100ml of distilled water.

b. Saturated aqueous sodium acetate.

Add 150gm of sodium acetate to 100 ml of distilled water. Dissolve by heating gently and then cool.

c. Chloroform

d. n-butanol

Procedure-Mix about 2.5 ml of freshly voided urine to 2.5 ml of Ehrlich’s reagent. Shake well and allow to stand for few minutes. Then add 5 ml of saturated solution of sodium acetate. Mix well.

- A Pink colour developing immediately after the addition of Ehrlich’s reagent implies the presence of porphobilinogen.

- With urobilinogen, pink colour develops after the addition of sodium acetate.

- If no pink colour develops, porphobilinogen is absent.

If pink colour develops, add a few ml of chloroform and shake well. Two layers result. chloroform and water are immiscible and the chloroform being heavier sinks to the bottom. If porphobilinogen is present, it remains in the top aqueous layer. In case of urobilinogen, the bottom chloroform layer is red in colour.

Observation- No pink colour appeared in the patients sample.

Inference- This confirms the absence of porphobilinogen in the patients urine.

URINE SCREENING FOR UROPORPHYRINS AND COPROPORPHYRINS.
Reagents-Mix 1 part of amyl alcohol, 1 part of glacial acetic acid and 1 part of ether.

Procedure-To 10 ml of fresh urine in a test tube, add one ml of the solvent. Mix well. Allow the test tube to stand for a few minutes, so that the solvent forms the top layer. Examine in a dark room under uv light. The ether solution on the top shows a reddish fluorescence if coproporphyrin is present.

Observation- A red coloured fluorescence was observed in the top layer of the patients sample.

Inference-This shows the presence of coproporphyrin in the patients urine.

Red coloured fluorescence developed in the top layer of the patients sample confirming coproporphyrin in the patients urine.