URINE AND STOOL INVESTIGATIONS IN THE DIFFERENTIATION OF THE PORPHYRIAS AS SEEN IN THE THREE RACIAL GROUPS IN CAPE TOWN*

L. EALES, M.D., M.R.C.P.

In the delineation of disorders of porphyrin metabolism the determination of faecal porphyrin is just as important as the estimation of urinary porphyrin. For a complete appreciation urinary δ-aminolaevulic acid (ALA) and porphobilinogen (PBG), determinations may also be required.

The investigation reported here includes estimations of urinary and faecal porphyrin in a series of normal subjects, in a group of patients with various disease states and, finally, in various porphyric disorders as seen in the 3 main racial groups—the White, the Cape Coloured and the Bantu.

1. *Normal Subjects*

A series of 70 apparently normal subjects of the 3 main racial groups in Cape Town was investigated. Twenty-four-hour urine collections and faecal samples obtained during or just after the collection period were analysed. Urinary uroporphyrin and coproporphyrin, and faecal coproporphyrin and protoporphyrin were determined in all subjects while ALA and PBG were determined in several of them.

Urine. In the White subjects average coproporphyrin and uroporphyrin concentrations were 42·5 and 7·4 μg./l. respectively; in the Cape Coloured, 33·1 and 6·5 μg./l.; and in the Bantu, 35·1 and 5·4 μg./l. The uroporphyrin fraction did not usually exceed 20 μg./l.

Faeces. Faecal determinations were expressed per g. dry weight of faeces. In White subjects the total faecal porphyrin averaged 41·0, the coproporphyrin 8·8 and the protoporphyrin 30·2 μg./g. dried weight. In the Cape Coloured the figures were 23·9, 6·3 and 20·1, and in the Bantu 44·9, 14·6 and 34·4.

It is believed that the often quoted upper limit of normal total porphyrin (50 μg./g. dried weight) is too low. The range for total porphyrin in all subjects studied is 0·2 – 137·6. The majority of determinations fall below 50 μg./g., yet there are several values above 100 μg./g. Protoporphyrin accounts largely for the total porphyrin. In Bantu subjects in the metabolism ward on the 'basic Bantu diet' stool porphyrins are very low, yet the addition of meat in one instance failed to increase faecal porphyrin.

2. *Subjects with Various Disease States*

Twenty-six cases with cardiac, renal hepatic, gastro-intestinal or endocrine disorders were studied. In 3 cases of cirrhosis of the liver there was a low faecal porphyrin concentration, but the urinary coproporphyrin level was markedly increased; other cases of hepatic disease had normal levels as a rule, but an abnormal amount of urinary uroporphyrin was found in 1 case each of primary and secondary carcinoma of the liver.

The results in cases of cardiac, renal, and gastro-intestinal disease were usually within normal limits, but high values for urinary uroporphyrin were encountered in a case of congestive heart failure due to aortic stenosis, and in another patient with cor pulmonale. In both patients the faecal porphyrin concentration was within normal limits.

3. *Cases of Porphyria*

The clinical presentation in 122 cases was reported (67 White, 35 Coloured and 20 Bantu). All had uroporphyrinuria. The

* Abstract of paper presented at Research Forum, University of Cape Town, 4 August 1959.

clinical presentation was denoted as acute, cutaneous, and mixed. (Mixed referred to cases with both cutaneous and acute symptoms.) In the White race the 3 clinical forms were encountered with more or less equal frequency, while almost all of the Bantu cases were examples of the cutaneous form. The Cape Coloured patients studied fell between these two extremes; cutaneous porphyria predominated, but acute and mixed forms were encountered.

Urinary and faecal porphyrin determinations were carried out in 30 White, 16 Cape Coloured, and 10 Bantu porphyrics. In addition urinary ALA and PBG were determined in many of these patients.

White porphyrics. Of 30 closely studied porphyrics 25 had markedly raised stool porphyrin concentrations (211 – 2303 μg./g.). These were all South African-born porphyrics and in the majority there was a positive family history. The remaining 5 patients had stool concentrations of 136 μg./g. or less and in each case there was a possible cause for this. Two had cirrhosis of the liver; 2 were not South African born—a Russian and a Norwegian—and the 5th was a case of cutaneous porphyria in a child.

Coloured porphyrics. Of 16 cases 9 had normal stool porphyrins and the remaining 7 had markedly increased stool porphyrins (181 – 8135 μg./g. dried weight), and in some of these cases a positive family history was obtained.

Bantu porphyrics. Of 10 Bantu cases 8 had normal faecal porphyrin concentrations (19·9 – 93·5 μg./g.). All except 1 had gross uroporphyrinuria. Urinary coproporphyrin concentration was also markedly increased but to a lesser degree. In 1 Bantu patient with acute porphyria the stool porphyrin concentration was very greatly increased.

Thus the Bantu patients had mainly increased porphyrin in the urine (and not in the faeces), and cutaneous forms of the disease predominated; in the White South African patients faecal porphyrin concentration was usually markedly increased, all forms of the disease occurred, and affected families were common. The Cape Coloured patients seemed to fall between these two groups both as regards excessive excretion or porphyrin in the stool and as regards clinical presentation.

The Swedish form of the disease which is rarely seen here has increased amounts of ALA and PBG in the urine most of the time. In South African genetic porphyria these substances occur in the urine in excessive amounts only during the acute attacks, viz., in a fatal case of fulminating porphyria the urinary ALA excretion reached 316 mg. per 24 hours. In the few Bantu subjects with cutaneous porphyria so far investigated no significant increase has been encountered.

Other means of differentiation (ALA loading and electrophoresis of urine porphyrins) are under investigation.

Further experience with a faecal screening test was also reported (red fluorescence induced by exposure to U.V. light of an acetic acid, ether, and amyl alcohol extract of faeces. The test was graded as follows: O yellow blue-green; ± doubtful tinge of red; + definite red fluorescence; and ++ brilliant red fluorescence). 610 stools from 582 patients with miscellaneous disorders were examined: O 416; ± 99; + 76; ++ 19. Of the 76 with a positive reaction 2 had genetic porphyria; this was also the case in no less than 14 out of 19 patients whose stools had brilliant fluorescence. If anything the test was too sensitive. The results of the test correlated fairly well with the faecal porphyrin concentration (U.V. spectrophotometric method).

The help of Drs. S. J. Saunders and E. B. Dowdle; the support of the South African Council for Scientific and Industrial Research and the Staff Research Fund of the University of Cape Town is gratefully acknowledged.