Acute Porphyria in a Bantu Male

H. D. Barnes, M.Sc., Ph.D., A.R.I.C., South African Institute for Medical Research, Johannesburg,

E. C. Neser, M.A., F.R.C.S., Klerksdorp, and E. N. Popper, M.D., M.R.C.P., Non-European Hospital, Johannesburg

Porphyria with cutaneous manifestations is by no means rare in Bantu subjects in South Africa and these patients do not seem to be liable to the episodes of abdominal pain and paralysis that are frequently seen in White porphyrias in this country. Acute porphyria without cutaneous lesions is not unknown in the Bantu but is encountered so rarely that reports of cases are justifiable as a reminder that they exist and should be fully investigated, for there are still several unanswered questions about aetiology, progress, and their relationship to other forms of porphyria.

Case Record

The present patient, M.S., an intelligent Chwana male aged 33 years, was admitted to Klerksdorp Hospital on 25 September 1958 for investigation. He had been an underground labourer and had spent the preceding week in the mine sick-bay with a febrile illness. His symptoms were non-specific, with no particular dyspnoea or abdominal pain. Examination was essentially negative, though he appeared to have lost weight. His skin was thin and melastatic but showed no ulcers or scars. The urine was normal to urinalysis, and porphyrins and porphobilinogen (PBG) assays by the procedure of Mauzerall and Granick1 were as follows:

**REFERENCES**


Skin and muscle biopsies on 23 October were strongly suggestive of a collagen disease, which was then thought to account for the entire picture, and he was discharged to the mine without further treatment. An interesting feature was that the biopsy wound, which was not infected, dehisced on removal of the stitches on the 7th day.

The girdle paralysis abated completely though gradually, but he never looked or felt well and did not return to underground work.

On 20 April 1959 he was seen again with dependent oedema, considerable dyspnoea, and elevated jugular venous pressure. Conjunctival pallor was noted. The urine contained no albumen but was of a darkish colour, which became somewhat more intense on standing. A specimen sent to Johannesburg was found to contain moderate amounts of porphyrin and porphobilinogen, whereupon the patient was transferred to the Non-European Hospital, Johannesburg, on 5 May for further investigation.

He here complained of a ‘ball in his stomach’ (right upper abdominal quadrant) for the past 7 weeks but there was no associated pain and he had not vomited. The variable facial oedema had sometimes made vision difficult. He had been coughing a good deal and producing sputum for 3 weeks. There was dyspnoea on effort but no chest pain. Physical examination indicated congestive cardiac failure but there were no clinical signs or symptoms attributable to porphyria. He later developed thrombophlebitis in his right leg, and on 16 May pulmonary embolism occurred and anticoagulant therapy was instituted. Though sputum was consistently negative for *M. tuberculosis*, X-ray again showed mililiary motting and anti-tuberculous treatment was resumed.

He developed a queer mental state progressing to dementia, became extremely weak and emaciated, and died on 27 July. Autopsy revealed the presence of generalized tuberculosis.

Stools had been analysed for porphyrins on 12 and 29 May. These contained 68 and 78 μg. of coproporphyrin and 47 and 70 μg. of uroporphyrin respectively per g. dry weight. Small amounts of porphyrin were found on spectroscopic examination in a number of specimens of urine, and aminolaevic acid (ALA) and porphobilinogen (PBG) assays by the procedure of Mauzerall and Granick1 were as follows:

**SUMMARY**

Artificial insemination with a donor’s semen (AID) is discussed from the point of view of a gynaecologist.

The historical aspect of the subject is surveyed, beginning with references to ancient Jewish and Arab writings.

The technique of AID is described, with mention of the medical and legal precautions to be taken.

Reference is made to some of the moral and legal objections to the practice of AID.

In drawing his conclusions the author feels that, all things considered, the practice of AID is undesirable, but that if it is not legally prohibited greater protection should be given to the individuals directly concerned.

**REFERENCES**

It was impossible, unfortunately, to continue these examinations, but urine obtained at autopsy contained but a trace of porphyrin and gave a negative Watson-Schwart test for porphobilinogen. The screen test on a fragment of rectal contents indicated a slight excess of porphyrin. Liver tissue contained 17.7 μg. per g. of ether-soluble porphyrin and 29 μg. per g. of porphyrin that was not soluble in ether.

**Family history.** The patient referred to his paralytic episode as rheumatism and stated that his father had been similarly affected. On examination the father proved to be a very well preserved Native apparently over 70 years of age. Examination of his central nervous, cardiovascular and respiratory systems revealed no abnormality. The skin of his face and forearms was very dark, in contrast to the medium reddish-brown of protected areas, but no ulceration or scarring were observed and there was no temporal hirsutism. He gave no history of abdominal pain but stated that he had had 'rheumatism' in his legs which was disabling, and that he could not, on this account, do hard work. He was cheerful and loquacious, referring several times to urinary disability but denying ever passing red urine. No porphyrin was detected on spectrophotometric examination of a freshly passed specimen. The Watson-Schwart test showed a slight excess of urobilinogen and a faint pink tinge remained in the aqueous phase. Next morning, however, this specimen contained 2-3 mg. of aminolaevulic acid and less than 1 mg. of porphobilinogen per litre. His wife, mother of the patient, and sister, both aged 19 years, referred to their work in the neighbourhood, and the patient also had 2 children, but none of these was available for examination.

**DISCUSSION**

Salient points in this history are the episode of paralysis, the observation some months later of dark urine the colour of which deepened appreciably on standing, and the finding within the next few weeks of porphyrin together with moderate increases of aminolaevulic acid and porphobilinogen in several specimens of urine. Had these been found at one and the same time acute porphyria could scarcely have been in doubt, and even when spread over half a year they are strongly suggestive of this diagnosis. The low chloride in the spinal fluid at the time of the paralysis also fits into the picture, since there was no clinical evidence of tuberculous meningitis at that time. The tuberculosis, though it was responsible for the fatal termination, seems not to have aggravated the porphyria, for the urine obtained at autopsy did not suggest an exacerbation of the porphyrin condition.

Several cases of acute porphyria have been seen in past years in Bantu patients but this is the first to be studied in detail by methods now available. None of them showed evidence of skin eruptions, which suggests that they may fall into a different group from the variegate porphyria in White South Africans, where cutaneous lesions are not uncommon. In this respect the Bantu patients conform to the classical intermittent acute porphyria reported from other parts of the world, notably Sweden. The biochemical findings in the present patient likewise conform in that urinary excretion of aminolaevulic acid and porphobilinogen was still significantly elevated during a clinical remission some months after the paralytic episode. The stool porphyrins, though slightly above normal, were well below the level commonly found in patients with variegate porphyria.

Waldenström has established beyond doubt that intermittent acute porphyria in Sweden follows the Mendelian dominant rule of inheritance. There is, thus far, no satisfactory evidence of heredity in the cutaneous form of porphyria which is common in South African Bantu. No indications of porphyria were found in our patient's father and the meagre information available about his mother does not suggest that her death could be attributed to this condition. This aspect of the case remains incomplete, because several immediate relations eluded examination.

Several other cases of acute porphyria in African Natives have been reported in some detail. It is hoped that more instances will become available for detailed investigation by methods now at our disposal so that answers may be found to the obvious questions.

**SUMMARY**

A case of acute porphyria in a Bantu male is presented. The findings differed in several respects from those in White South African patients with variegate porphyria but conformed to the pattern of the intermittent acute form. No evidence of heredity was obtained but not all members of the family were available for study.

We have to thank the Director of the South African Institute for Medical Research for providing facilities for the laboratory studies and Dr. P. Keen, Superintendent of the Non-European Hospital, Johannesburg, for admitting the patient for closer observation.

**REFERENCES**


**TRIAL OF AN ORAL DIURETIC, HYDROCHLOROTHIAZIDE**

**J. V. O. Reid, B.M., M.R.C.P., and G. Newell**

From the Departments of Medicine and Pathology, University of Natal, and King Edward VIII Hospital, Durban

Hydrochlorothiazide has been introduced as an oral diuretic agent with a potency considered greater than that of any other oral agent, and has been submitted to clinical trial by various observers. In general its efficacy has been confirmed, although there is disagreement concerning the loss of potassium which it induces, and its potency compared with other diuretics. In the present study the effect of a relatively large single oral dose, 250 mg., is compared with that of 2 c.c. of a parenteral mercurial, mersalyl, in the same patients. This oral dose was chosen after preliminary trials with single doses increasing from 50 to 250 mg. had indicated increasing effect until at the higher figure a water diuresis equivalent to that of 2 c.c. of mersalyl was obtained.