South African Medical Journal : Suid-Afrikaanse Tydskrif vir Geneeskunde

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PORPHYRIA IN SOUTH AFRICA

Until a few years ago porphyria was considered to be a very rare disease, although it had been reported from many parts of the world. Congenital or erythropoietic porphyria,[1] which causes pink staining of the teeth and bones and marked light sensitivity from birth, is inherited as a Mendelian recessive trait. Only 36 authenticated cases have been reported in the world literature.[2,3] In cattle it is known as pink-tooth disease.

Waldenstrom, of the University of Lund, who carried out pioneer work on porphyria, described a type of porphyria that is common in the North and comparatively rare in the South of Sweden. His suggestion that the general incidence of porphyria in Sweden is 1·5 per 100,000 inhabitants, is perhaps an underestimate.[4] This type of acute intermittent porphyria is inherited as a non sex-linked Mendelian dominant trait. There is no increased skin sensitivity to sunlight or injury, but an acute attack of porphyria may be precipitated by the use of certain drugs, particularly barbiturates and sulphonamides. The acute attack is characterized by abdominal and muscle pain and by marked emotional disturbance; it may proceed to generalized peripheral neuritis, and perhaps the death of the patient. During the quiescent phase there is, in this type of porphyria in adults, a great excess of porphobilinogen and delta-aminolaevulic acid, precursors of porphyrin in the urine, and the Watson-Schwartz test with Ehrlich's aldehyde is positive. There is little or no increase in the faecal porphyrin.

Porphyria is very common in the White population of South Africa, among whom it is also inherited as a Mendelian dominant trait.[5-7] It also occurs among the Coloured people. Porphyria, which is relatively common in South Africa, was introduced by an early settler from Holland, whose descendants have multiplied extremely rapidly under ideal conditions. Today, one million of the three million White people in this country bear the names of 40 original settlers, and it is interesting to contemplate that all the male members of these families, illegitimacy apart, inherit their Y chromosome from 40 original free burghers. Dean[8] claims that he has traced 38 large groups of porphyric families to one original ancestor, Gerrit, the son of Jan, who married at the Cape in 1688, and he considers that they have inherited porphyria from Gerrit or his wife Ariaantje.

The Mendelian dominant type of porphyria, which is common in South Africans, is a different genetic disorder from the acute intermittent porphyria described by Waldenstrom.[9] The South African type, porphyria variegata, is characterized by acute attacks precipitated by drugs, and often by light sensitivity of the exposed skin. South African porphyria may be symptomless except for a high excretion of porphyrin in the faeces and some increase in urinary porphyrin. Acute attacks occur more frequently in women, and the skin sensitivity is usually more marked in men. When there are no sores or scars on the back of the hands, the skin may still abrade easily if scratched with the finger nail—a useful clinical test. In the acute attacks the urine is often port wine in colour, and the Watson-Schwartz test for porphobilinogen is always positive. In the quiescent phase the urine is normal in colour, the Watson-Schwartz test is negative, and there may be only a slight increase in urinary porphyrin. However, in both the acute and the quiescent phases there is a great excess of porphyrin in the faeces, unlike that in acute intermittent porphyria.

The easiest way to screen the faeces for excess porphyrin is to prepare a solution of a small fragment in 2 ml. of a solvent consisting of equal parts of glacial acetic acid, amyl alcohol, and ether. When this brown solution is examined in ultraviolet light with a Wood's filter, its colour is normally green or grey. A brilliant pink fluorescence suggests the possibility of porphyria, and the patient and the family should be investigated clinically, and a quantitative analysis of the porphyrin excretion in the urine and faeces should be undertaken. A great excess of chlorophyll sometimes gives a pink fluorescence. Excess of porphyrin in one specimen of stool does not necessarily mean that the patient is a porphyric; further investigation will confirm or refute the diagnosis.

During the past six months an interesting pilot experiment in routine testing for porphyria has been carried out in Port Elizabeth. All patients admitted to hospital in Port Elizabeth have been subjected to a routine test for porphyria before the administration of pentothal, other barbiturates, or sulphonamides. An account of this experiment will be given at the South African Medical Congress in East London towards the end of this month.

Barnes[10] describes a disturbance of porphyrin metabolism with light sensitivity which is common among the Bantu. There is no evidence that this is a hereditary disorder; it would appear to be due to a disturbance of liver function, often secondary to malnutrition and the use or abuse of adulterated alcoholic beverages. This type of porphyrin disorder is accompanied by a high excretion of porphyrin in the urine, but little or no increase in the faeces. It would fit into the group described as porphyria cutanea tarda symptomatica. As Garrod[11] defined porphyria as an 'inborn error of porphyrin metabolism', it is a question of semantics whether a disorder of porphyrin metabolism that is not inborn should be called porphyria. In this type of por-

phyrin disorder skin lesions may be severe, but attacks of acute porphyria do not occur.

Acute porphyria is an extremely serious illness and a threat to the patient's life. The correct diagnosis is often missed and, when it is made, treatment necessitates a very high level of medical and nursing attention and is not always successful. Prevention of the acute attack will save much suffering. Prevention depends on considering the possibility of porphyria and excluding it before prescribing barbiturates and sulphonamides. In Dean's view, patients in South Africa should be screened for porphyria as a routine, especially before the administration of a pentothal anaesthetic. When porphyria is diagnosed, all the relatives who can be traced should also be screened for the disorder. Porphyrics should be warned of the extreme danger of taking certain drugs, and should be given a letter describing the evidence for the diagnosis. This letter should be shown by the patient to any doctor whom he or she may consult.

1. Watson, C. J. *et al.* (1951): Trans. Assoc. Amer. Phycns., **64**, 345.
2. Schmid, R., Schwartz, S. and Sundberg, R. D. (1955): Blood, **10**, 416.
3. Townsend-Coles, W. F. and Barnes, H. D. (1957): Lancet, **2**, 271.
4. Waldenstrom, J. (1957): Amer. J. Med., **22**, 758.
5. Barnes, H. D. (1951): S. Afr. J. Clin. Sci., **2**, 117.
6. Dean, G. (1953): Brit. Med. J., **2**, 1291.
7. Dean, G. and Barnes, H. D. (1955): *Ibid.*, **2**, 89.
8. Personal communication.
9. Dean, G. and Barnes, H. D. (1959): S. Afr. Med. J., **33**, 246.
10. Barnes, H. D. (1959): *Ibid.*, 33, 274.
11. Garrod, A. E. (1923): *Inborn Errors of Metabolism*, 2nd ed. London: Frowde & Hodder & Stoughton.

DIE VERENIGING VIR GEESTESGESONDHEID

Gedurende die afgelope aantal weke het die *Tydskrif* eksemplare ontvang van die jaarverslae van al die verskillende takke van die Vereniging vir Geestesgesondheid. Die groot en belangrike werk wat hierdie Vereniging dwarsdeur die land doen word duidelik in hierdie verslae weerspieël, en almal wat in die werksaamhede van die Vereniging belangstel word aangeraai om met die oog op verdere besonderhede in verbinding te tree met die Sekretaris van die Suid-Afrikaanse Nasionale Raad vir Geestesgesondheid.*

Die Vereniging vir Geestesgesondheid bestaan al sedert 1918 en is soos volg saamgestel: In die eerste plaas is daar die algemene koördinerende en beleidvormende liggaam: die Suid-Afrikaanse Nasionale Raad vir Geestesgesondheid. Verder bestaan die Vereniging uit 10 takke of susterverenigings wat versprei is in die groot dorpe en stede van ons land.

Ons het al by verskillende vorige geleenthede[1-3] gewys op die onbevredigende toestande wat hier geld ten opsigte van die versorging en behandeling van geestesongesteldes. Omdat ons in Suid-Afrika nie beskik oor genoegsame doeltreffende kliniese dienste op die gebied van die psigiatrie nie, is die dienste wat die Vereniging vir Geestesgesondheid lewer van des te groter belang.

Die volgende is 'n baie beknopte beeld van wat hierdie Vereniging doen op al die plekke waar hy takke het: Die Vereniging reël en organiseer gereelde klinieke op 'n buitepasiënte-basis vir persone wat aan enige vorm van ligte of ernstige geestesversteuring ly. Ook reël hy klinieke vir verstandelik vertraagde en swaksinnige kinders en persone, en kinderleidingklinieke. Deur middel van spanne maatskaplike werksters word gevallewerk en gesinsorg op groot skaal onderneem. Die mediese professionele werk in die klinieke word waargeneem deur mediese beamptes van die Departement Gesondheid—beamptes wat psigiatriese werk doen in die verskillende hospitale vir geestesongesteldes.

Die bronne waaruit hierdie Vereniging put om sy administratiewe en finansiële verpligtinge na te kom, is soos volg: Elke tak van die Vereniging ontvang 'n jaarlikse subsidie van die Nasionale Raad, wat op sy beurt weer gelde ontvang uit openbare bronne sowel as 'n aansienlike toelaag van die Departement Uniegesondheid. Gedurende die afgelope jaar was daar sprake dat die Departement Gesondheid besig is om die verdere toesegging van dié toelaag in heroorweging te neem. Ons wil hier graag die geleentheid gebruik om die Departement—wat die Vereniging in elke geval baie goedgesind is—dringend te versoek om, indien enigsins moontlik, daardie toelaag nie terug te trek nie, maar eerder te vergroot. Dit sou 'n ernstige slag beteken vir verskeie takke van die Vereniging as hulle nie meer kan reken op daardie bydrae nie.

Elke tak van die Vereniging ontvang ook ondersteuning uit verskillende openbare bronne uit die gebiede waar hulle werksaam is. In die belang van die toekoms van die dienste wat op hierdie gebied gelewer word, is dit egter uiters noodsaaklik dat die samestellende dele van die Vereniging probeer om finansieel so onafhanklik moontlik te word.

Ons leef in 'n tyd waarin daar 'n groeiende bewustheid is van die groot rol wat geestesgesondheid, en die beginsels waarop dit berus, speel in die nastrewe van beter aanpassing en 'n gesonder en gelukkiger wêreld in die algemeen. Hierdie groeiende bewustheid van die betekenis van geestesgesondheid is nie net waar te neem in ons eie land nie, maar dwarsoor die hele beskaafde wêreld. Die Wêreld Federasie vir Geestesgesondheid, waarvan die Suid-Afrikaanse Nasionale Raad vir Geestesgesondheid 'n stigterslid is, het juis die jaar 1960 as Wêreldgeestesgesondheidsjaar aangedui. Die doel met hierdie voorgenome plan is om nuwe belangstelling te wek vir die werksaamhede van die internasionale en nasionale organisasies wat dit vir hulself ten doel gestel het om 'n groter mate van geestesgesondheid vir soveel mense as moontlik na te streef. Om hierdie rede is dit dus ook van pas dat ons nou al navraag doen na waar ons ten opsigte van hierdie verpligting staan—wat betref alle moontlike dienste soos wat gelewer word deur die Vereniging vir Geestesgesondheid, die Unie- en Provinsiale Gesondheidsdepartemente, plaaslike hospitale, private psigiaters en geneeshere, en die algemene publiek.

1. Van die Redaksie (1958): S. Afr. T. Geneesk., 32, 652.
2. *Idem* (1958): *Ibid.*, **32**, 996.
3. *Idem* (1959): *Ibid.*, 33, 269.

*Posbus 2587, Johannesburg.