VAN DIE REDAKSIE : EDITORIAL

HYALINOSIS CUTIS ET MUCOSAE

Dit wil voorkom asof sekere oorgeërfde degeneratiewe siektes taamlik dikwels in Suid-Afrika aangetref word. Dit is byvoorbeeld 'n bekende feit dat porfirie en pseudoxanthoma elasticum hier meer dikwels voorkom as in ander lande. Elders in hierdie uitgawe van die *Tydskrif* plaas ons 'n artikel waarin Scott en Findlay[1] die aandag vestig op 'n ongewone metaboliese famieliesiekte, hyalinosis cutis et mucosae (lipoïde proteïenose). Hierdie skrywers het nie minder nie as 27 Suid-Afrikaanse gevalle opgespoor. Al hierdie gevalle was Blanke pasiënte behalwe 2 wat Kleurlinge was. Die 25 gevalle het in 18 Blanke families voorgekom waarvan 17 Afrikaanssprekend is. Die voorkoms van die toestand is betreklik hoog vir die klein Blanke bevolking van Suid-Afrika, aangesien altesaam minder as 80 gevalle in die mediese literatuur aangeteken is sedert Siebenmann hierdie siekte die eerste keer beskryf het in 1908. Dit lyk asof daar geen gevalle van hierdie siekte by Bantoepasiënte aangeteken is nie.

Die eerste twee Suid-Afrikaanse gevalle is in 1948 deur Loewenthal, Murray en Ruskin[2] gerapporteer. Sedertdien is ander gevalle deur dermatoloë ontdek en op dermatologiese byeenkomste gedemonstreer.

Die eerste gevalle van hierdie toestand is uit Duitsland gerapporteer en dit wil voorkom of die meeste gevalle van die siekte vandag in daardie land aangetref word. Dit is twyfelagtig of enige gevalle van hierdie siekte ooit teengekom is in Engeland en Holland—die twee lande van herkoms van die meeste Blankes in Suid-Afrika.

Die oupagrootjie van die twee Kleurlingpasiënte was 'n Duitser en sommige van die Blanke pasiënte het ook Duitse voorouers gehad. Soos die genoemde skrywers aantoon, is hierdie anomaliese toestand heel waarskynlik in 'n vroeë stadium van die Blanke nedersetting in Suid-Afrika uit Duitsland uit hier ingevoer.

Die meeste Afrikaanssprekendes behoort tot 'n min of meer beperkte aantal families en dit is moontlik dat familiestamboomstudies van hierdie gevalle interessante genetiese feite aangaande die oorsprong van die siekte mag aantoon, soos die geval byvoorbeeld ook is met die nou reeds bekende studies van Dean en Barnes[3,4] oor die genealogiese oorsprong van porfirie in Suid-Afrika.

Die skrywers vestig die aandag van algemene praktisyns en van oor-, neus- en keelspesialiste op die diagnostiese belang van onverklaarde heesheid by jong kinders as 'n moontlike simptoom van hyalinosis cutis et mucosae. Ander manifestasies van die siekte wat van belang mag wees in ander vertakkings van die medisyne sluit in verkalkings binne die skedel, epilepsie en geestesversteurings, abnormaliteite van die tande, abnormale groei van die hare, abnormaliteite van die oogagtergrond, diabetes, ens. Die skrywers verwys ook na 'n verband met aangebore hartsiekte en kwaadaardige bloedarmoede wat nog nie tot dusver gerapporteer is nie. En, alhoewel dit nie die primêre oorsaak is nie, kan sonlig ook as 'n presipiterende faktor geld aangesien die mees uitgebreide velverskynsels in hierdie toestand aangetref word op blootgestelde dele van die liggaam.

Dit lyk asof daar 'n taamlike uitgebreide versteuring is van koolhidraat-, lipoïde- en proteïen-metabolisme; en die interessante werk van Eberhartinger en andere[5,6] oor die alfa en beta globuliene en die Sf. lipoproteïene moet nog bevestig word in hierdie land. Verdere biochemiese studies mag ook meer lig werp op die rol van dieet in die ontwikkeling van die siekte.

In Suid-Afrika is daar baie materiaal vir die verdere studie van hierdie interessante anomalie en biochemiese en histochemiese navorsing mag lei tot verheldering van die patogenese van die toestand.

1. Scott, F. P. en Findlay, G. H. (1960): S. Afr. T. Geneesk., **34**, 189.
2. Loewenthal, L. J. A., Murray, F. J. en Ruskin, H. D. (1948): Clin. Proc., **7**, 97.
3. Dean, G. en Barnes, H. D. (1955): Brit. Med. J., **2**, 89.
4. Dean, G. (1956): S. Afr. T. Geneesk., **30**, 377.
5. Eberhartinger, C. en Reinhardt, F. (1958): Hautarzt, **9**, 503.
6. Eberhartinger, C. en Niebauer, G. (1959): *Ibid.*, **10**, 54.

HYALINOSIS CUTIS ET MUCOSAE

Certain inherited degenerative diseases seem to occur fairly frequently in South Africa. It is a well-known fact that porphyria and pseudoxanthoma elasticum are seen here much more often than in other countries. In this issue of the *Journal* Scott and Findlay[1] draw attention to a rare familial metabolic disease, hyalinosis cutis et mucosae (lipoid proteinosis). They have collected no less than 27 South African cases. All these were in White patients except for the 2 cases in a Coloured family. The other 25 cases occurred in 18 White families, of which 17 are Afrikaans speaking. This is a relatively high incidence for the small White population of South Africa, for since the disease was first described by Siebenmann, in 1908, less than 80 cases have been recorded in the world literature. No cases seem to have been recorded in the Bantu.

The first two South African cases were reported by Loewenthal, Murray and Ruskin[2] in 1948. Since then other cases have been discovered by dermatologists and demonstrated at dermatological meetings.

The first cases were reported from Germany and this seems to be the country where most cases are seen today. In England and Holland, the two preponderant countries of origin of the White people of South Africa, it is doubtful whether any cases of the disease have ever been seen. The great-grandfather of the two Coloured patients was a German and some of the White patients also had German forefathers. As our authors point out, this anomaly was most probably imported from Germany at an early stage of the White settlement in South Africa.

Most Afrikaans-speaking persons belong to a somewhat limited number of families and it is likely that family-tree studies of these cases would reveal interesting genetic facts about the origin of the disease, such as have been disclosed by Dean and Barnes's now well-known researches[3,4] into the genealogical origin of porphyria in South Africa.

Our authors draw the attention of general practitioners and otolaryngologists to the diagnostic importance of unexplained hoarseness in young children as a symptom of hyalinosis cutis et mucosae. Other manifestations of the disease which will be of interest in other branches of medicine include intracranial calcifications, epilepsy and mental disturbances, dental abnormalities, abnormal hair growth, abnormalities of the eye fundus, diabetes, etc. Our authors refer to an association with congenital heart disease and pernicious anaemia not hitherto reported. Sunlight, although not the primary cause, acts as a precipitating agent, for the most extensive skin anomalies are seen on the exposed parts of the body.

There seems to be a very extensive disturbance of carbohydrate, lipoid and protein metabolism, and the interesting work of Eberhartinger and others[5,6] on the alpha and beta globulins and the Sf. lipoproteins awaits confirmation in this country. Further biochemical studies might also throw more light on the role of the diet on the development of this disease.

South Africa offers enough material for further study of this interesting anomaly and biochemical and histochemical research that might clarify its pathogenesis.

1. Scott, F. P. and Findlay, G. H. (1960): S. Afr. Med. J., **34**, 189.
2. Loewenthal, L. J. A., Murray, F. J. and Ruskin, H. D. (1948): Clin. Proc., **7**, 97.
3. Dean, G. and Barnes, H. D. (1955): Brit. Med. J., **2**, 89.
4. Dean, G. (1956): S. Afr. Med. J., **30**, 377.
5. Eberhartinger, C. and Reinhardt, F. (1958): Hautarzt, **9**, 503.
6. Eberhartinger, C. and Niebauer, G. (1959): *Ibid.*, **10**, 54.

THE NATIONAL CANCER ASSOCIATION OF SOUTH AFRICA : EXPERT PANEL

The National Cancer Association of South Africa announces the establishment of an Expert Panel for the histological investigation of suspected malignant tumours.

The Panel consists of experts from different centres of the Union acting in an honorary capacity.

Any practitioner who requires an opinion from the panel in doubtful cases must request the pathologist employed by him to forward clinical particulars, original blocks and sections and the specimen (if still available) to the National Secretary, National Cancer Association of South Africa, P.O. Box 2000, Johannesburg.

The opinion of the Expert Panel will be forwarded to the original reporting pathologist and to the practitioner.

No fees will be charged for this service.

Practitioners are advised that specimens for investigation will only be received by the Expert Panel *via* the original reporting pathologist.

Further particulars may be obtained from the National Secretary, National Cancer Association of South Africa, P.O. Box 2000, Johannesburg.