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\(^4\) 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 into the genealogical origin of porphyria in South Africa.

Our authors draw the attention of general practitioners and oto- laryngologists to the diagnostic importance of unexplained hoarseness in young children as a symptom of hyalnosis 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, lipid and protein metabolism, and the interesting work of Eberhartinger and others 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.

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, 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.

RUAPETi OF THE STOMACH IN THE NEWBORN

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This condition was first reported by Siebold in 1825, since when 68 cases have been reported. The first case treated successfully by surgery was reported by Legar et al. in 1950. Vargas et al. reported 11 cases in 1955, 8 of which had been operated upon, with 2 survivals. At that time a total of 55 cases had been reported. In 1959 Linkner and Benson reported 13 cases operated upon, with 6 survivals.

Following is a report, one of 2 cases which have presented at the King Edward VIII Hospital, Durban, within the last 5 years.

The other one is to be published shortly.

CASE REPORT

B.S., an African female infant, was born in April 1959. The pregnancy was normal and she was delivered at term by a pupil-midwife without any difficulty. At birth the infant weighed 6 lb, 6 oz. and no abnormalities were found on examination at this time.

It was noticed 48 hours after birth that the infant had a markedly distended abdomen, and up to that time had passed no meconium. There had been no vomiting. The infant was found to be listless, apathetic and rather ill. The abdomen was grossly distended and the subcutaneous veins coursing over its surface gave it an unhealthy marbled appearance. The flanks were markedly distended. On palpation the abdomen was soft although the infant obviously resented this procedure. A fluid thrill was elicited from flank to flank. On rectal examination a finger could not be introduced more than 3-4 cm., nor could a lubricated catheter be passed per rectum and guided into the sigmoid, but only with difficulty. The rest of the bowel was narrow and collapsed. There was an incomplete rotation of the bowel but no volvulus. The small bowel, and most of the large bowel, were suspended from the posterior abdominal wall by a very short mesentery. A thickened band of tissue crossed the 2nd part of the duodenum but otherwise the duodenum was patent. The small and flimsy greater omentum had milk curds and pieces of fibrin adherent to it.

The stomach, which was about 2½ inches long, was almost completely everted through a linear rupture 1½ inches long, which extended parallel and adjacent to the greater curvature from the fundus down to the pyloric antrum. The edges of the tear were thin and transparent and did not bleed, and the surrounding stomach showed no evidence of induration, ulceration or necrosis.

A straight X-ray of the abdomen revealed some free gas beneath the diaphragm, but this was small in amount compared to the large amount of fluid, which gave the rest of the abdomen a typical ground-glass appearance. There was no visible gas bubble in the stomach. A film in the inverted position showed free gas in the pelvis (Fig. 1).

A blood drip was set-up and shortly afterwards laparotomy was carried out under general anaesthesia.

A right paramedian muscle-splitting incision was made. When the peritoneal cavity was opened, gas and a large amount of turbid fluid escaped. The free fluid was aspirated and the abdominal contents examined. The rectum was not atretic, but it was very narrow and collapsed. A catheter could be passed per rectum and guided into the sigmoid, but only with difficulty. The rest of the bowel was narrow and collapsed. There was an incomplete rotation of the bowel but no volvulus. The small bowel, and most of the large bowel, were suspended from the posterior abdominal wall by a very short mesentery. A thickened band of tissue crossed the 2nd part of the duodenum but otherwise the duodenum was patent. The small and flimsy greater omentum had milk curds and pieces of fibrin adherent to it.

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