HEREDITARY HAEMORRHAGIC TELANGIECTASIA

BY

HUGH G. GARLAND, T.D., M.D., F.R.C.P.

AND

S. T. ANNING, M.A., M.D., M.R.C.P.

(From the General Infirmary at Leeds)

In another paper (Garland and Anning, 1950) we have fully reviewed the literature relating to hereditary haemorrhagic telangiectasia, considered the genetics of the 244 affected families already recorded, and added 20 families examined personally. The purpose of this communication is to add three more families, also previously unreported, and to stress again that the condition is not very rare though frequently undiagnosed. Of our 23 families, 22 are from the West Riding of Yorkshire. The condition is usually not recognized as a cause of nose-bleeding. Arteriovenous aneurysm of the lung, which was present in two of the families reported here, is not uncommonly diagnosed as congenital heart disease, the presence of telangiectasia of the skin or mucous membrane being unobserved or its significance being ignored, and a family history of telangiectasia not elicited.

Family 21

● = Telangiectasia. ○ = Epistaxis only. X = Examined.

I, 2.—Examined aged 72; had suffered from frequent nose-bleeding during childhood and adolescence. No telangiectasia was seen except a diffuse type over the cheeks.

I, 3.—Died aged 68, but had suffered from persistent nose-bleeding during the first two decades. His three daughters live in Canada, and no information about them is available.

II, 1.—Has suffered from nose-bleeding all his life.

II, 3.—Died aged 36 from pulmonary tuberculosis, but is said to have had "spots" on the face like her sister's (II, 4) and nose-bleeding all her life.

II, 4.—Examined aged 33; has often had nose-bleeding from the age of 8 years, and has menorrhagia. The telangiectasia was not noticed until the time of her son's operation, when it was observed by Mr. P. R. Allison. She is pallid and has pinhead-sized angiomas scattered over the cheeks, with a few on the lips, tongue, and palate.

III, 2.—Was admitted at the age of 6 years to the thoracic surgical department of the General Infirmary at Leeds (Mr. Allison) with a history of occasional nose-bleeding and increasing cyanosis since the age of 3. On examination he showed considerable cyanosis, with finger-clubbing, and a spider naevus was present on the right cheek. There were no abnormal signs in the heart or lungs apart from a systolic murmur in the right axilla. Radiography showed the cardiac outline to be normal, but there was no opacity in the right middle lobe. Blood examination showed: red cells, 7,100,000 per c.mm.; haemoglobin, 136%; haematocrit reading, 69%. A diagnosis

of haemangioma of the right middle lobe was made and the chest was opened. The right middle lobe was of normal size and its surface was covered with dilated vessels. Right middle lobectomy was performed, following which his cyanosis immediately improved. He made an uneventful recovery.

Family 22

I, 1.—Died aged about 25; no other information about him was obtainable. His wife, who died aged 84, is said to have been healthy.

I, 5.—Suffered from occasional nose-bleeding all his life.

II, 1.—Died aged 50; had intermittent but severe nose-bleeding all his life. He had "spots" on his cheeks and had to be very careful while shaving, as they bled profusely if cut.

III, 1.—Examined aged 9 years; she had suffered from frequent nose-bleeding for several years. About four pinhead-sized telangiectatic spots were seen on her cheeks.

III, 2.—Examined aged 7 and 8; she was admitted under Mr. Allison when 7 years old. She had never been as strong as her sisters, and was smaller than her sister three years younger. For one year her mother had noticed blueness of her finger-nails. She had occasional nose-bleeding. On examination she was seen to be pale, with cyanosis of the cheeks, lips, and nail-beds. On the left cheek and nose there were several pinhead-sized telangiectases. There was moderate finger-clubbing. The heart sounds were normal, but a murmur was audible over the upper part of the chest on the left side. Radiography revealed an opacity in the left upper lobe. Blood examination showed: red cells, 5,200,000 per c.mm.; haemoglobin, 108%. A diagnosis of haemangioma of the left upper lobe was made, and at operation a haemangioma the size of a normal gall-bladder was found lying in the fissure at the root of the lingula, partly embedded in the upper lobe but largely projecting. Local excision of the haemangioma was carried out and she rapidly lost her cyanosis. She made an uneventful recovery, and when examined again eight months later was a good colour and very well. Finger-clubbing was still present.

III, 3.—Examined aged 5 years; she had suffered from occasional nose-bleeding for a few years, but showed no telangiectasia of the skin or buccal mucosa.

Family 23

I, 1.—Died aged 82, but had suffered all his life from occasional nose-bleeding and had "spots" on his face and lips.

II, 3.—The second husband of II, 2; suffered from recurrent nose-bleeding and had "spots" on his face like his sons. His brother (II, 5) was similarly affected, but no information could be discovered about the brother's children (III, 5 and 6) nor about his other brothers (II, 6, 7, and 8) and his sister (II, 4).

III, 2.—Aged 60 and married but childless; has suffered from nose-bleeding only rarely throughout his life. He is said to have "spots" on his face just like his brothers.

III, 3.—Examined aged 57, a single man; has suffered from severe nose-bleeding since childhood. The spots on his face appeared during the second decade, but have been more prominent during the last ten years. On examination he was very pallid, with multiple telangiectases of about 2 mm. diameter on the lips, several similar lesions on the tongue and soft palate, a few pinhead-sized telangiectases on the cheeks and ears, and diffuse telangiectasia over the tip of his nose.

III, 4.—Aged 55, also single; was said to have had occasional nose-bleeding all his life and spots on his face like his brother (III, 3).

Summary

Three more families with hereditary haemorrhagic telangiectasia from the West Riding are reported. They illustrate again that epistaxis is usual in this condition, that arteriovenous aneurysm of the lung may be present, and that the defect is inherited as a simple dominant.

We are grateful to Mr. P. R. Allison for so kindly placing his records at our disposal, and to Dr. John Savage, Dr. J. P. Peel, Dr. John Rushton, and Dr. Bennet Smallhorn for allowing us to examine their patients.

REFERENCE

Garland, H. G., and Anning, S. T. (1950). *Brit. J. Derm.* In press.

NUTRITIONAL DYSTROPHY AMONG CHILDREN IN MADRAS

BY

S. T. ACHAR, M.D., F.R.C.P. Canada

Professor of Paediatrics, Madras Medical College

Quite a large number of infants and children with oedema of varying degrees not associated with albuminuria have recently been admitted to the children's wards of the General Hospital and of the Women and Children's Hospital, Madras (South India). A good proportion of these cases had skin changes of a peculiar nature and of varying pattern in different patients and even in the same child (Fig. 1). In some, large areas of skin were discoloured, while in other areas the skin was actually peeling off. Cracks and fissures at the flexures were a frequent feature. Large areas of depigmentation with some hyperpigmented areas were often present.

The term "crazy-pavement skin" or "mosaic skin" serves to describe the majority of skin lesions. An occasional case, however, showed a fine scaling with pigmentation characteristic of pellagrous skin, though the distribution of this scaly lesion was not always characteristic of pellagra. Some cases had keratomalacia, while others showed cheilosis, angular stomatitis, and the orogenital syndrome. The common feature of all was a pronounced debility and apparent low vitality. The child lost its liveliness, looked ill (but not the toxic look

of an infectious disease), and seemed to be slipping away from life quietly and rather quickly.

For purposes of this review a series of 78 cases that were admitted to the above-mentioned hospitals in 1948 (March–December) have been considered.

Investigation of a Series of Cases

Age Incidence and Social Status.—One child in the series was 8 months old; the rest were distributed as shown in Table I, the majority being 1 and 2 years. Most came from very poor families, though an occasional one was encountered among the middle and even the rich classes.

Dietetic History and Previous Illness.—Almost all the children had been fed very unsatisfactorily, usually because of the absence of cow's milk and milk products but sometimes from ignorance. Their diet consisted mainly of rice, very little of other cereals, and vegetables—apart from the scanty mother's milk the younger children were able to get. More than half gave a history of dysentery or diarrhoea lasting for a while—often recurrent bouts of diarrhoea rather than a single episode—at some time preceding the onset of oedema. In some instances the lack of liveliness and vigour as well as the oedema appeared to follow an attack of diarrhoea or dysentery.

TABLE I.—Age Incidence and Economic Background

Age	No.	Social Status	No.
Under 1 year	4 (5%)	Very poor	40
1 and 2 years	48 (62%)	Poor	31
3 " 4 "	18 (23%)	Middle	6
5 to 10 "	8 (10%)	Rich	1

Clinical Features.—Apart from the oedema and absence of liveliness in all cases, nearly half of them showed varying types of skin changes, with and without alteration in the colour and texture of the hair. Eye changes indicative of vitamin-A deficiency were fairly prevalent, leading in some cases to blindness, and signs of riboflavin deficiency were present in many. There were surprisingly few with clinical evidence of active rickets or scurvy in spite of the poor dietetic history. The main clinical features are shown in Table II.

Course of Illness

The mortality of the untreated cases is very high and the morbidity among those who survive considerable. There were 12 deaths in this series (mostly in the earlier cases, when the technique of repeated blood and concentrated serum transfusions was being developed). Most of these deaths seemed to be attributable to acute parenchymatous hepatic failure, the child sinking into stupor and dying quickly in this stuporous state. Septic complications were common; otitis media, abscesses, pyelitis,

FIG. 1.—Severe dermatoses.