HEREDITARY HAEMORRHAGIC TELANGIECTASIA WITH HEPATOSPLENOMEGALY AND ASCITES

BY

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Since Osler (1901) distinguished hereditary haemorrhagic telangiectasia from other hereditary bleeding diatheses, it has often been described, and has been found to be associated with familial epistaxis, melaena (Gambill, 1946), haematemesis, haematuria, pulmonary arteriovenous fistula (Moyer and Ackerman, 1948), profound anaemia, and splenic or liver enlargement (Roles, 1928; Fitz-Hugh, 1931; van Bogaert and Scherer, 1935). Anning, in a personal communication, states that hepatomegaly has not been noted in any of the cases of Garland and Anning (1950a, 1950b) and that the splenic enlargement is usually thought to accompany severe anaemia. None of 56 patients recorded by Garland in 1936 had evidence of liver disorder. Nora Schuster (1937) published a case of telangiectasia with splenomegaly in which there were multiple aneurysms of the splenic artery. She refers to five complete necropsies, in four of which the spleen was enlarged but not the liver. In two cases of Fitz-Hugh there was atrophic necrosis of the liver after transfusion intolerance. Another showed fatty change, haemorrhage, and biliary dilatation, and her own case showed venous congestion and patchy periportal and peribiliary fibrosis, the patient having died with heart failure. The necropsy on van Bogaert and Scherer's case was the only one in which a true cirrhosis was present. They described the case of a patient with familial telangiectasia and a true Laennec's cirrhosis whose mother also had hepatomegaly. Saksela *et al.* (1949) stated that Rooschütz (1937) was the first to diagnose atypical liver cirrhosis at necropsy, and that Johnson and Nordenson (1942) were able to find some sort of functional disorder of the liver in at least one test in all cases. Parkes Weber (1936) thought that the liver changes might be the result of dysbiotrophy. He referred to two further necropsies without detail.

The present report is of a patient with the Osler-Weber-Rendu syndrome, severe anaemia, nose-bleeding and intestinal bleeding, together with hepatosplenomegaly and ascites. The latter is assumed to be the result of cirrhosis of the liver as described by van Bogaert and Scherer. Ascites is not commonly noted, but has been described by Gurney Yates (Hurst and Plummer, 1932).

Case Report

A woman aged 63 was admitted to hospital on April 5, 1950, with gross anaemia, telangiectasia, ascites, and hepatosplenomegaly. Neither her two older sisters nor her parents were similarly affected. Her telangiectases did not begin until she was 30. At first a single naevus appeared on the right cheek, and in the course of years developed on the face, ears, and tongue, and finally on the hands at the age of 61. From the time the naevi appeared she suffered from 8 to 10 nose-bleedings a year, so severe that towels were soaked with the blood.

Sixteen years before admission she became breathless on walking uphill, and was admitted to a nursing-home, where

iron-deficiency anaemia with achlorhydria was diagnosed. Iron was given, and she felt better until 1942, when breathlessness again occurred. In 1948 she had melaena, followed by loss of consciousness, and on being admitted to hospital 5 pints (2.8 litres) of blood was transfused. Subsequently folic acid and iron were given. She improved steadily, but had a severe bleeding from an angioma on the tongue in April, 1949; this ceased after cauterization, and a further 2 pints (1.14 litres) of blood was transfused. In February, 1950, her abdomen began to swell and her liver was enlarged. She was admitted with a provisional diagnosis of Osler-Weber-Rendu syndrome and cirrhosis of the liver.

On examination she was extremely pale; multiple angiomas were seen on the face, lips, and inside of the mouth, and scattered on the trunk; there was gross ascites, with enlargement of the liver and spleen (Figs. 1 and 2). Her blood pressure was 170/90. A haemic murmur was heard over the base of the praecordium.

Paracentesis was performed, and 7½ pints (4.3 litres) of clear amber fluid with a protein content of 1.2% and a lymphocytic cellular content was withdrawn.

FIG. 1.—Showing enlarged liver and spleen after tapping of ascites.

FIG. 2.—Showing telangiectases of tongue and lips.

A blood count showed: red cells, 2,450,000; haemoglobin, 42%; colour index, 0.86; white cells, 3,050; platelets, 346,000 per c.mm.; cell volume, 21.4%; M.C.V., 95.6 μ^3 ; M.C.H.C., 28%; bleeding time, six minutes; coagulation time, initial clotting five minutes; complete clotting, eight minutes. The sternal marrow film showed increased haemopoiesis with normoblastic erythropoiesis. There were some deformed iron-deficiency normoblasts.

Tests for occult blood in the stools were repeatedly positive. The fractional test meal demonstrated a histamine-fast achlorhydria and blood in the test specimens. The blood urea was 34 mg., and the plasma fibrinogen 250 mg. Liver-function tests on April 5 showed: serum albumin, 3.3 g.; serum globulin, 1.8 g.; serum total cholesterol, 190 mg. per 100 ml.; serum alkaline phosphatase, 20 units; van den Bergh, direct and indirect reaction negative; cephalin cholesterol, negative; thymol turbidity, 0.5 unit; colloidal gold, 33210. The serum albumin was relatively low, and the serum alkaline phosphatase was raised.

The patient's blood was found to be Rhesus-negative, and she was transfused with 3 pints (1.7 litres) of packed cells from Rhesus-negative blood. After this her red cell count rose to 3,390,000 per c.mm., and the haemoglobin to 60%. On April 28 she was discharged on choline, 4 g. a day, and "rutin," 60 mg. a day.

On June 8 she was readmitted. She felt better, but had had two further nose-bleeds and still had a pronounced pallor. The liver and spleen were unchanged in size. A blood count showed: red cells, 2,480,000 per c.mm.; haemoglobin, 36%; cell volume, 19.7 c.cm.; M.C.V., 79.4 μ^3 ; M.C.H.C., 25.4%; bleeding time, three minutes; coagulation time, initial clotting, eight minutes; complete clotting, 11 minutes; platelets, 275,000 per c.mm.; prothrombin time, 14 seconds. Tests for occult blood in stools were still strongly positive. Sigmoidoscopy failed to show any angiomas of the rectum and distal sigmoid colon; she was not submitted to gastroscopy. A further 3 pints of Rhesus-negative blood was given. She had no further nose-bleeds while in hospital, and was discharged on June 22.

On July 15 she was readmitted, the ascites having re-accumulated, but no further nose-bleeds having occurred. A blood count on July 16 was: red cells, 2,260,000; haemoglobin, 44%; colour index, 0.97; white cells, 4,050; platelets, 283,000 per c.mm. A further 12 pints (6.8 litres) of ascitic fluid was removed and she was discharged on July 17.

The patient's son, aged 33, had had angiomas of the cheek since he was 20. For five years he had had frequent nose-bleeds, up to three or four a week, and had required cauterization of the bleeding patches up to once a year. The daughter, aged 27, had had bleeding from both nostrils intermittently for 10 years, severe for seven years. There was one angioma on her left forefinger; this had only recently appeared. Neither had hepatosplenomegaly.

Comment

The above case illustrates the dictum that the true frequency of the condition would be realized if every physician, dermatologist, or rhinologist seeing cases of recurring epistaxis or multiple telangiectases of the face would inquire into the family history. The patient's daughter, although undergoing cauterization of the nose, was not previously considered to have hereditary haemorrhagic telangiectasia. All members of the family developed angiomas or nose bleeds after the age of 17. This is interesting, and recalls the work of Bean *et al.* (1949) on the angiomas of pregnancy, suggesting an endocrinological basis. The patient's severe anaemia was considered to be due to repeated blood loss in the stools, all tests for occult blood being repeatedly positive. Whether the bleeding arose from oesophageal varices as the result of cirrhosis of the liver, or from angiomas of the stomach or bowel, remains unproved, but Kushlan (1946) has described

cherry angiomas and ecchymotic telangiectases, together with a distended oozing vein on the angulus observed through the gastroscope. The evidence for suspecting that the pathology is that of cirrhosis depends largely upon the presence of hepatosplenomegaly with ascites, the low value of serum albumin and raised serum alkaline phosphatase being of themselves inconclusive.

Treatment with rutin has been disappointing in this case, the anaemia relapsing within a few weeks of transfusion, and the nose-bleeds persisting despite doses of 60 mg. a day. This confirms the observation of Petch (1948).

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REFERENCES

- Bean, W. B., *et al.* (1949). *Surg. Gynec. Obstet.*, **88**, 739.
 Fitz-Hugh, T. (1931). *Amer. J. med. Sci.*, **181**, 261.
 Gambill, E. E. (1946). *Proc. Mayo Clin.*, **21**, 157.
 Garland, H. G. (1936). *Lancet*, **1**, 390.
 — and Anning, S. T. (1950a). *Brit. J. Derm.*, **62**, 289.
 — (1950b). *British Medical Journal*, **1**, 700.
 Hurst, A. F., and Plummer, N. S. (1932). *Guy's Hosp. Rep.*, **82**, 81.
 Johnson, S. R., and Nordenson, N. G. (1942). *Svenska Läkartidn.*, **39**, 981.
 Kushlan, S. D. (1946). *Gastroenterology*, **7**, 199.
 Moyer, J. H., and Ackerman, A. J. (1948). *Ann. intern. Med.*, **29**, 775.
 Osler, W. (1901). *Bull. Johns Hopk. Hosp.*, **12**, 333.
 Petch, C. P. (1948). *British Medical Journal*, **2**, 785.
 Rendu, R. (1916). *Rev. gén. Clin.*, **30**, 351.
 Renshaw, R. J. F. (1939). *Cleveland Clin. Quart.*, **6**, 226.
 Roles, F. C. (1928). *St. Bart's Hosp. J.*, **36**, 19.
 Rooschütz (1937). *Verh. Ges. Kreisforsch.*, **10**, 262.
 Saksela, N., *et al.* (1949). *Ann. Med. int. Fenn.*, **38**, 132.
 Schuster, N. H. (1937). *J. Path. Bact.*, **44**, 29.
 Van Bogaert, L., and Scherer, J. H. (1935). *Ann. Méd.*, **38**, 290.
 Weber, F. Parkes (1936). *Lancet*, **1**, 305.

AN INTERESTING CASE OF POLYMASTIA OBSERVED IN PREGNANCY

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The occurrence of supernumerary nipples (polythelia) and of supernumerary breasts (polymastia) is well known. As students we meet first with these anomalies of development in the anatomy and embryology classes. In later days, in hospital or general practice, it is not uncommon to see patients in whom there are one or more supernumerary nipples; less frequently we may see a patient in whom supernumerary breasts are present. By the very nature of the examinations required, the hypertrophic and secretory changes which take place in the breasts, and the interest involved in breast-feeding, it is probable that most cases of polymastia are seen in pregnant women.

Case Report

A 2-gravida, aged 20, reported to the antenatal clinic of the Eastern District Hospital, Glasgow, on April 25, 1949. Her last menstrual period began on September 17, 1948, and her expected date of confinement was June 24, 1949. She had been pregnant on one occasion previously, and had delivered herself normally of a live male child weighing 6 lb. 8 oz. (2.9 kg.) in 1946. The baby was bottle-fed.

Her urine was clear of abnormal constituents, her blood pressure was 120/50 mm. Hg, and her weight was 9 st. 6 lb. (59.9 kg.). She made no complaint. She was of short