A CASE OF HAEMATOPORPHYRIA CONGENITA (CONGENITAL PORPHYRINURIA).

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THE child to be described first came under notice at the age of 12 months, and the reason for the child being sent to the hospital was the dark red colour of the teeth. The case was then thoroughly investigated, and proved to be an example of haematoporphyrin congenita. Very few of the cases have as yet been recognized and recorded so early in life. Sir A. E. Garrod and Mackey¹ report one similar case, and I am indebted to the former's book, *Inborn Errors of Metabolism*,² for help.

The child was apparently born healthy and had had no illness of note. She had been easy to "bring up." All her life, however, and even directly after birth, she had passed urine which was red in colour and stained the napkins red. The teeth were cut at the usual time and were dark red in colour. The parents took her away to the seaside at the age of 4 months; the weather was then very hot and sunny. She came out in a rash on the face and back of the hands, which the mother described as looking like chicken-pox.

FIG. 1.—Showing the pigmentation (red) of the milk teeth.

Condition on Admission.

The child, aged 2½ years, is well grown and apparently healthy. She has been under observation for a year and a half, and the urine has been of the characteristic port-wine or red colour every time it has been examined. It contains no albumin or blood corpuscles. I am much indebted to Dr. A. Sellars for the spectroscopical examination of the urine and faeces. For this purpose the acid extract from the urine and the alkaline extract from the faeces were used.

Spectroscopic Examination.—*Acid Urine:* Position of two bands, mean central position at 6,200 and 5,640 (Angström units). *Alkaline Faeces:* Position of four bands at 6,480, 6,200, 5,640, 5,280 (Angström units). The position of the bands was ascertained by plotting against the spectral lines of lithium, sodium, thallium, and strontium.

The blood shows no abnormal condition and there is no enlargement of the spleen. A most striking feature of the case is the marked red pigmentation of the milk teeth. She cut her first tooth soon after the age of 6 months, and it came through red in colour. Dr. Wigoder ground a tooth, and the colour was as deep as the tooth could be ground, showing that the pigmentation is not superficial only. The child was demonstrated at a meeting of the British Dental Association, and no one had seen or heard of a similar case. It will be interesting to examine the milk teeth further, when they are shed, and also to see the condition of the permanent teeth. Only very rarely has the pigmentation of the teeth been noted. Curiously enough the bones do not show pigmentation when transilluminated by an electric bulb, nor do they show any changes on x-ray examination. The fundi of the eyes also show no abnormal pigmentation.

The child was again taken to the seaside this year during a spell of hot weather, and had to be brought home again on account of large blisters appearing on the face, back of the hands, and front of the knees. This condition is called hydroa vacciniforme. Her face had become markedly pigmented from the sun and there were large bullae on the affected parts. The blisters, some of which had burst, contained clear fluid of a reddish colour. Scarring has been left after the blisters have healed. The hair and the clothes give complete protection. The nails of the hands were also affected and had become dark red, as if they had all received a blow causing bruising. It is certain that great care will have to be taken in the future against exposure to the sun to prevent serious disfigurement.

FIG. 2.—Hydroa vacciniforme showing the large bullae, some of which have burst.

Sir A. E. Garrod concludes that the outlook for these cases is serious and that there is a liability to tuberculosis. This patient is, however, in good health; and, as long as precaution against excessive sunlight is taken, there is at present no sign that the child should not grow up and remain well. The family history of the child is negative. Most cases of porphyrinuria are the result of taking sulphonal or an allied drug for a considerable period.

The cause of the condition is unknown, but it is probable that the porphyrin is an intermediate product in the conversion of haemoglobin into bilirubin, as suggested by Hans Fisher. Whatever the exact cause may be, it is an inborn error of metabolism.

Haematoporphyrin is a most remarkable sensitizer.³ A small quantity of this injected into white guinea-pigs renders them so sensitive that they die from exposure to light. Meyer Betz made himself very ill by exposure to very moderate light, after injection of a very small quantity into himself, and remained hypersensitive for weeks.

REFERENCES.

¹ *Quart. Journ. of Med.*, 1922, xv, 319. ² Second edition, 1923, Henry Frowde, and Hodder and Stoughton. ³ Leonard Hill: *Sunshine and Open Air*, Edward Arnold and Co., 1924.

FATAL CASE OF ANEURYSM OF THE SPLENIC ARTERY.

BY

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It is well recognized that aneurysms are the risk of the strenuous life led by men and are uncommon in women; they are to be expected at that time in life when the tissues are beginning to weaken but the vital forces and feelings which impel violent action still remain at their height. They can be caused directly or indirectly by trauma which weakens the vessel wall, while certain diseases, by sapping the strength of the vessel wall, predispose the individual to the condition. Among the commonest of these diseases are atheroma and syphilis, while a less known cause is tubercle. A nodule of tubercle settling in the vessel wall may quite well be the cause of an aneurysm in those positions where a compensating fibrosis is unable to reinforce the strength of the vessel. For this reason aneurysms are sometimes met with in the brains of children—one such case is within my own recollection. There is also a case in which an important verdict depended on the result of the *post-mortem* examination, where a man had been seen to strike a boy upon the head and the boy shortly afterwards died, it was supposed, from the blow. Examination showed that a tuberculous aneurysm had ruptured in the ventricles of his brain.

These being the well recognized factors in the production of aneurysms, when we find that all these are set aside and we meet with an aneurysm in a woman of the age of 27, engaged in a light occupation, without any other sign of disease, and the aneurysm is not single but one of three, it seems to be a case that is worth recording: more especially as the symptoms resembled those of a ruptured gastric ulcer or an ectopic pregnancy in many of their features.

The case was as follows:

Jean O'C., aged 27, single, employed as kitchenmaid at a good address in London, was admitted to St. Mary's Hospital, Paddington, on November 27th, 1923, at 4 p.m.

History.—Sudden intense pain had come on with great tenderness in the pit of the stomach at 2 p.m., soon after food, with nothing whatever to account for it, and she was sent in by her doctor as probably a ruptured gastric ulcer.

On admission the patient was in a state of extreme collapse from shock; she was pale, cold, and clammy, and in great pain. On inquiry she had never had any indigestion pointing to a gastric or duodenal ulcer. Her blanched condition made one seek for symptoms pointing to an ectopic pregnancy, but there was nothing to indicate this. Light touch over the abdomen was not painful, but any pressure caused great pain. Her condition was very doubtful, and it was decided to leave her for some two hours to see if the shock would pass off. The respirations were 33, temperature 98°, and pulse 130.

At the end of one and half hours the shock was better, but the symptoms pointed still more strongly to haemorrhage, as she was still more blanched, and her pulse weaker, more rapid, and she was inclined to be restless.

At 6.30 operation was decided upon without any definite diagnosis being made. She was opened above the umbilicus, when the