A CASE OF HAEMATOPORPHYRIA CONGENITA (CONGENITAL PORPHYRINURIA).

BY

HUGH T. ASHBY, B.A., M.D.CAMB., F.R.C.P.,

VISITING PHYSICIAN TO THE ROYAL MANCHESTER CHILDREN'S HOSPITAL, ETC.

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

DUNCAN C. L. FITZWILLIAMS, C.M.G., M.D.,
CH.M., F.R.C.S.

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

abdominal cavity was found full of blood. A dresser at once prepared and gave her intravenous saline. Blaming myself for believing a single woman's history about her menstrual periods, and thinking that it was obviously a case of ectopic pregnancy, an incision was rapidly made above the pubes, only to expose a perfectly normal set of pelvic organs. On mopping out the blood and clots the whole of the peritoneum of the posterior abdominal wall was found to be one huge tense haematoma, almost filling the abdominal cavity and approaching the anterior abdominal wall. Blood had found its way into the mesentery of the small intestine and to a certain extent into the omentum. The lesser sac was full of blood, and was emptied in the hope of finding the main bleeding point, but the whole area seemed to ooze blood and no bleeding

vessel was seen. As the patient was in a critical state and nothing was to be gained by prolonging the operation, the abdominal cavity was filled with saline solution and the wound closed; she was sent back to bed, and the foot of the bed raised. She died at 10.30 that night.

At the *post-mortem* examination the splenic artery was found tortuous and there were three aneurysmal dilatations along its course. The largest of these was the size of a walnut, and situated near the hilum of the spleen. It had calcified walls, which showed that it was not of recent formation. This had ruptured and was the cause of the bleeding into the tissues behind the peritoneum. The aorta and the arteries appeared healthy and there was no sign of the origin of the condition.

British Medical Association.

PROCEEDINGS OF SECTIONS AT THE ANNUAL MEETING, BRADFORD, 1924.

SECTION OF LARYNGOLOGY AND OTOTOLOGY.

W. JOBSON HORNE, M.D., President.

PRESIDENT'S OPENING REMARKS.

THE PRESIDENT, in opening the proceedings of the Section, said that, while he had no intention of inflicting a presidential address upon the Section, he could not refrain from expressing his appreciation of the honour conferred upon him. The officers of the Section, after fully considering the choice of several subjects suggested for general discussion, had unanimously decided upon the two subjects now submitted—namely, "Chronic tonsillitis: indications for treatment and results obtained," and "The importance of early treatment of acute otitis." The officers, in making this selection, were fully alive to the fact that the subjects were time-worn, but it was felt that there were points which would bear repeating if not emphasizing, and, moreover, that the matters to be discussed were of daily occurrence, and therefore of more practical value than other subjects less commonplace but more academic. Dr. Jobson Horne reminded them that the Section met on two days only, but it was his hope, with the help and support of the large number present, to get three days' work into two days.

DISCUSSION ON CHRONIC TONSILLITIS: INDICATIONS FOR AND RESULT OF TREATMENT.

OPENING PAPERS.

I.—A. BROWN KELLY, M.D., F.R.F.P.S.,
Surgeon for Diseases of Throat and Nose, Victoria Infirmary,
Glasgow.

THE term "chronic tonsillitis" designates a long-continued, mild inflammation of the tonsil; oftener, however, it is used to imply a state of the tonsil which is not manifestly inflammatory but which predisposes to acute attacks. The two conditions of the tonsil which are mainly accountable for chronic inflammation and recurrent exacerbations are hypertrophy and sepsis; these are commonly associated.

In discussing the indications for treatment of chronic tonsillitis, we have to deal, not only with the disease itself, but also with its distant effects; further, in making choice of a method of treatment it may be necessary to take into account special circumstances—for example, the patient's age, general health, or occupation.

The commonest affection of the tonsils with which we have to deal is that of enlargement in the young. This may give rise to various symptoms which experience has taught will pass off when the tonsils are removed; consequently we advise operation, and in the great majority of cases our recommendation is accepted or even welcomed by the parents, the benefit generally obtained being a matter of common knowledge. On the other hand, various objections to operation

may be advanced on the patient's behalf or may arise in our minds. First among these is the natural disinclination to the removal of an organ or tissue not obviously diseased. It should be remembered, however, that an enlarged tonsil, although apparently healthy on probing, squeezing, etc., if examined microscopically after enucleation will be found to be diseased. According to Dietrich,¹ the tonsils, even in earliest childhood, are attacked by acute inflammatory changes of all degrees, without signs or symptoms, in the course of various diseases of the upper air passages (measles, bronchopneumonia, etc.). Similarly, Howarth and Gloyne² found evidence of bacterial infection in every tonsil examined, their cases ranging from 2 to 15 years of age; while D. J. Davis doubts the existence of a normal tonsil after the first few days of life. From the bacteriological standpoint every tonsil is therefore a diseased tonsil. It is obvious that we cannot utilize this criterion, so that it becomes necessary to base our opinion mainly on the clinical history, concomitant signs, and local aspect.

Another question which occasionally perturbs parent and doctor alike, especially since tonsillectomy has become *de rigueur*, is whether this operation may not abrogate or destroy some important function from the deprivation of which the patient will suffer. This is conceivable in the young child and will be referred to immediately; but our everyday experience proves the contrary, so that we may cease to be discomposed by physiological misgivings. For the present it should suffice to remember that the tonsil is made up of numerous nodules of lymphadenoid tissue precisely similar to those found, not only in the pharyngeal and lingual tonsils, but also scattered singly or in small groups beneath the mucous membrane of the pharynx, and that the faucial tonsil differs in no way from any solitary nodule excepting in that it consists of an immense collection of such nodules. Schlemmer³ has recently emphasized these facts, and maintains that there is no anatomical or physiological reason for assigning the tonsil a separate position or special function.

If, then, the tonsillar function is shared by the lymphadenoid tissue throughout the pharynx and nasopharynx, is it not probable that when the tonsil is removed its functions are carried on vicariously by the lymphadenoid tissue elsewhere? For, as already stated, no deleterious effect on the constitution has been traced to the mere absence of the tonsils after operation. This view gains support from a sequence of events occasionally noted. In a young child with large adenoids and small tonsils, if the former are removed and the latter left, at a not distant date the tonsils are found to have enlarged considerably; moreover, in children whose tonsils and adenoids are completely removed large granulations sometimes appear on the posterior or lateral wall of the pharynx and become inflamed from time to time.

In very young subjects the age is often advanced as a contraindication. There need, however, be no hesitation in removing enlarged or diseased tonsils from a child of 2 years, or even of 18 months, if healthy and vigorous. When younger, in order to lessen the bleeding and shock it is advisable to operate on the adenoids first, and after an interval of weeks or months to deal with the tonsils. It may be imperative to remove adenoids from a suckling, but it is rarely necessary to operate on tonsils before the completion of the first year of life.

Does the removal of tonsils increase or diminish the susceptibility to contagious diseases? In young children when the tonsils are at the height of their physiological