INTESTINAL POLYPOSIS ASSOCIATED WITH FACIAL PIGMENTATION

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Two examples of this unusual syndrome have been encountered, each presenting interesting features which are worth recording.

CASE 1

On 12 August 1952 a 19-year-old European girl was admitted to hospital as an emergency, complaining of acute abdominal pain of 6 hours' duration. The pain was centrally situated, colicky in nature, and of gradually increasing severity. She felt nauseous and vomited once. No flatus or faeces had been passed since the onset of the attack.

For 2 years before admission the patient had been unwell and had been treated by several practitioners for anaemia. During this period she noticed that her stools were dark in colour on several occasions.

Examination. She was obviously anaemic and ill. An unusual pigmentation of the lips, nose, eyelids and interior of the mouth was noticed (Fig. 1), but no special significance was attached to it at the time. On abdominal examination a sausage-shaped mass which was slightly tender, was palpable immediately below the umbilicus. Moderate hyperperistalis was noted on auscultation of the abdomen. Rectal examination was negative.

Haemoglobin 10·02 g.%; red blood-cells 3·6 million per c.mm.; white blood-cells 14,000 per c.mm., with 89% polymorphs.

A diagnosis of acute intestinal obstruction, probably due to intussusception, was made. The anaemia was not explained.

Operation. 1,000 c.c. of blood was given and laparotomy performed (Prof. J. K. Bremer). An intussusception of the proximal jejunum was found. As this could not be reduced resection and anastomosis were performed. After completion of the anastomosis several polypi were palpated in the jejunum above the anastomosis, but in view of the patient's condition these were left and the abdomen closed. Examination of the resected bowel revealed a polyp as the cause of the intussusception. Histological examination showed this to be a simple adenoma with no evidence of malignancy.

The patient made an uneventful recovery. Subsequent X-ray examination with a barium meal and barium enema revealed no abnormality within the gastro-intestinal tract. Six weeks after the first operation laparotomy was again performed and 3 pedunculated polypi removed by local excision from the proximal jejunum (Fig. 2). No more polypi were found in the remainder of the intestinal canal.

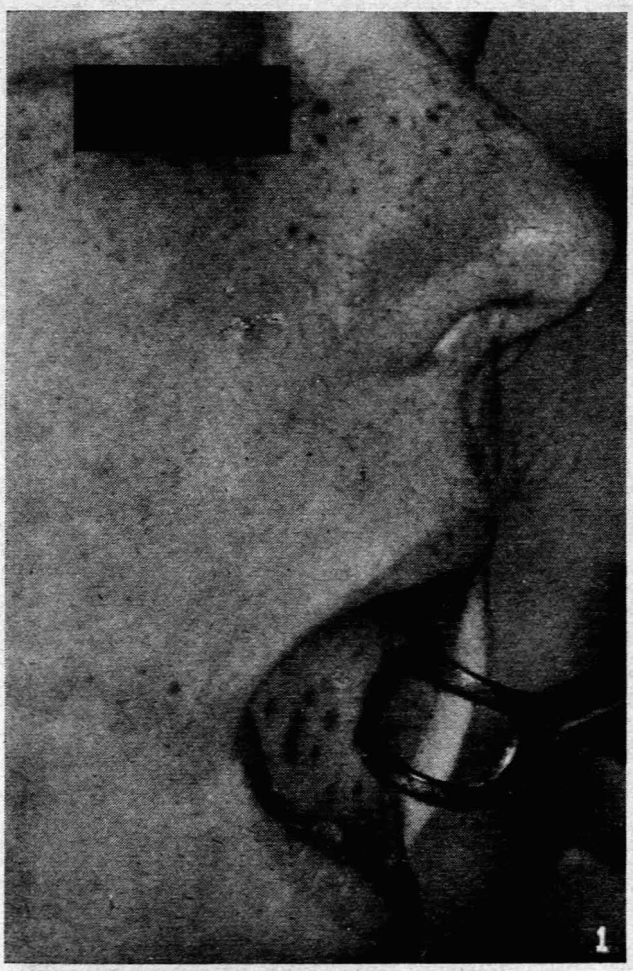

Fig. 1.

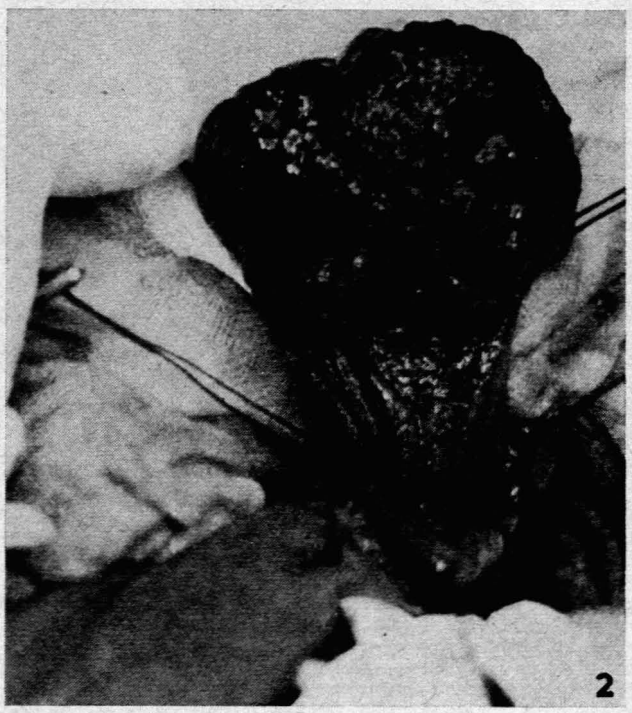

Fig. 2.

Three years later the patient was well and free from abdominal symptoms.

Family History. The patient was one of a family of 3. Her 2 brothers and her parents were alive and well and none showed

evidence of abnormal pigmentation. No member gave a history of malaena or of any acute abdominal emergency.

CASE 2

On 8 August 1955 the 16-year-old son of a medical practitioner was referred for surgical treatment. Two months previously he had complained of excessive fatigue and appeared completely worn out on return from school in the afternoons. Any physical effort produced dyspnoea and he became progressively less inclined to exert himself. During the July holidays he spent as much time as possible resting at home, complained of dizziness when standing erect, and was noticeably pale. At no time was there any complaint of pain or abdominal discomfort, nor was any alteration in the colour of the stools apparent. Blood investigation showed the haemoglobin content to be 50%, with the characteristic features of secondary hypochromic anaemia. The patient was seen by a physician, who recognized the significance of the very marked facial and buccal pigmentation. This is seen in Fig. 3, which also shows pigmented spots on the hands, especially over the knuckles. There were also brown spots on the hard palate. The physician referred the patient for a gastro-intestinal barium series; the radiologist reported as follows:

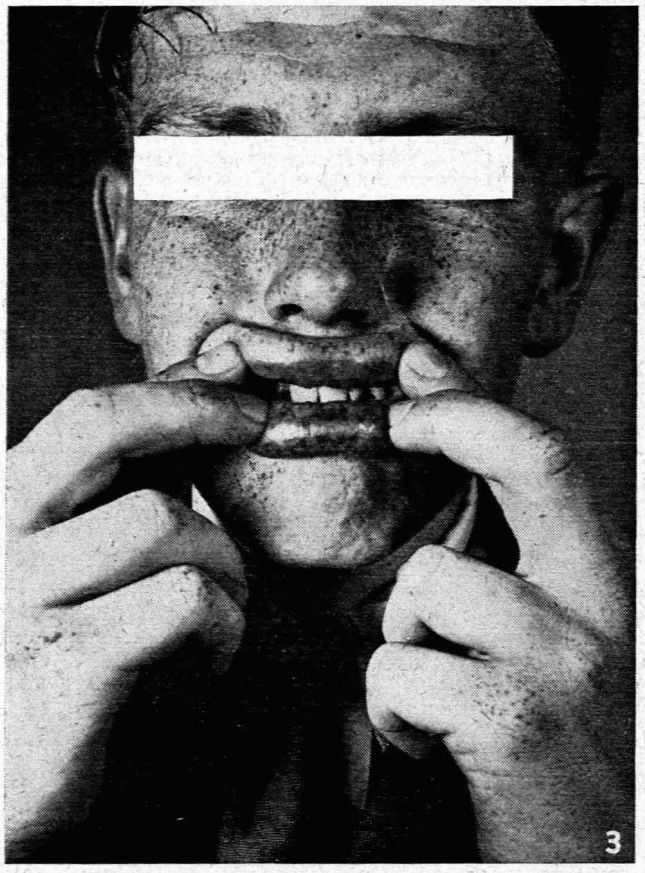

Fig. 3.

'Oesophagus and stomach normal. The second part of the duodenum shows a large irregular filling defect occupying chiefly the distal part of the loop. Note, however, that the proximal part of the duodenal loop appears also affected. Over the involved segment the lumen of the duodenum is unusually wide and the mucosal pattern abnormal. Some views show further rounded clearly-defined filling defects in the proximal loops of jejunum. The radiological appearances are consistant with a diagnosis of intestinal polyposis'.

Operation. After the transfusion of 1,000 c.c. of whole blood the haemoglobin level rose to 86% and on 18 August laparotomy was undertaken by one of us (A.M.G.). A large sponge-like polyp was found arising from the postero-medial wall of the second part of the duodenum just below the ampulla of Vater and occupying the bulk of the duodenal lumen. The growth had a short broad stalk, and local excision and suture through a trans-duodenal approach proved a relatively simple procedure.

On careful examination of the whole intestine 3 more cherry-like pedunculated polypi were found, situated 18 inches, 2 feet and 5 feet beyond the duodeno-jejunal junction. These were also treated by local excision and suture. No growths were found in the large gut.

Convalescence was entirely uneventful.

Pathological Report. Simple adenomatous polyps of the intestine.

Family History. This boy is the only one of a family of 3 sons who shows any abnormal pigmentation. Both parents are normal and healthy and neither gives any family history of pigmentation or of polyposis. Photographs taken during childhood show no evidence whatever of facial pigmentation at the age of 9 months but at 6 years the aggregation of pigmented spots around the mouth and on the lips is obvious. The mother is of the opinion that the spots appeared gradually between 2 and 6 years.

DISCUSSION

The syndrome of intestinal polyposis associated with facial pigmentation was first described by Peutz,[5] of The Hague, in 1921. The condition attracted very little attention in the English literature until Jeghers *et al.*[1] described several cases in 1949. Since that time sporadic reports of cases have appeared.[2,3,4,6,7] Essentially the syndrome consists of intestinal polyposis, an unusual pigmentation of the face and fingers, and a strong family history which, however, as in the 2 cases here described, is not always obtained.

Intestinal Polyposis. In all reported cases, multiple polypi have been present in the jejunum, usually predominating in the upper jejunum although no part is immune. Polypi may also be present in the stomach, duodenum, ileum, and colon. They vary in size from a few millimetres in diameter to quite large tumours. They may be sessile or pedunculated. They are reddish brown in colour and are very friable, so that haemorrhage is common. Histologically they are adenomatous; malignancy has been reported in 2 cases by Peutz[5] and in 1 case by Jeghers *et al.*[1] The benign tumours should probably be regarded as pre-malignant.

Pigmentation is most obvious on the face in the region of the nose, eyelids and mouth. On the lips and within the mouth it is typical of this syndrome. This is in sharp contrast with freckles, which do not occur on the inside of the mouth. Pigmentation also occurs on the dorsum of the fingers, especially in the region of the terminal interphalangeal joints. The pigment consists of melanin and is deposited in numerous small sharply defined spots. As far as can be ascertained the spots are present from a very early age and do not tend to fade in later years. The distribution and the character of the pigmentation is typical of the syndrome and, if borne in mind, is a most useful aid in diagnosis.

A family history of pigmentation or polyposis can often be ascertained. Jeghers stressed the importance of this, but cases have, however, been reported in which no family history was obtained, as in the 2 cases here reported.

The diagnosis has usually been made after operation for acute intestinal obstruction caused by the intussusception of an intestinal polyp. Bleeding may be an

important symptom, although this is not particularly stressed in the literature. Both our cases presented initially with severe anaemia. In case 1 the patient was treated for anaemia for 2 years before the correct diagnosis was made after operation for acute intussusception. In case 2 the patient also presented with anaemia, but the examining physician was aware of the syndrome and the diagnosis was promptly established. Bleeding from the tumour may also cause obvious malaena, as in case 1.

Treatment of the polyposis is surgical—usually an emergency operation for acute intestinal obstruction. At operation it is most important to examine the whole intestinal tract after reduction or resection of the intussusception so that any additional polypi can be removed at the same time if the patient's condition permits. If not, a subsequent operation is essential. If the diagnosis is suspected in the absence of intestinal symptoms, the patient should be thoroughly examined by barium series, the radiologist being informed of the possible diagnosis so that particular attention may be paid to the upper jejunal loops.

Pre-operative sterilization of the bowl is performed.

The operation consists of laparotomy, and complete examination of the intestinal tract from stomach to rectum. It may be extremely difficult to feel small polypi and the utmost care should be taken in this examination. Trans-illumination against the operating light tilted towards the surgeon is most useful for revealing small tumours which cannot be felt by the examining fingers. If numerous polypi are present in a small segment of bowl a resection and end-to-end anastomosis should be performed. Usually, however, it is sufficient to open the bowel by enterotomy or colotomy and perform local excision of individual lesions. This is quite easy since these tumours are usually pedunculated.

SUMMARY

1. Two cases of intestinal polyposis associated with pigmentation of the face are described.
2. The clinical features and diagnosis are discussed. The diagnostic importance of anaemia associated with the typical facial pigmentation is stressed.
3. The operative management is described.

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