INTESTINAL POLYPOSIS ASSOCIATED WITH MELANOSIS ORIS

BY

LESLIE G. ANDREWS

From the Paediatric Unit, Whipps Cross Hospital

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Weber (1919) described a case of acute intussusception in one of a pair of twins originally noticed by Hutchinson (1896) to have oral pigmentation. Peutz (1921) recorded a family of seven, and Jeghers, McKusick and Katz (1949) reported 10 cases of melanosis of the lips and buccal mucosa together with polyposis of the gastro-intestinal tract. The latter's report included a review of the literature. Since then 45 cases have been recorded in the world literature, 15 of these in England (Foster, 1944; Tanner, 1951; Wolff, 1952; Kitchin, 1953; Hunter and Wilson, 1953; Smith, 1954; Savage, 1954; Young, 1954; Walker-Brash, 1954; Crone and Light, 1954). The case recorded here is probably an example of this syndrome although not yet proven.

Case Report

The patient, a boy with a fair complexion, was born in August, 1951. He began vomiting almost from birth and on the third day produced 'coffee grounds'. In October, 1951, he was admitted to hospital on account of an exacerbation of the vomiting. Pyloric stenosis was diagnosed and the child was treated medically, but the vomiting persisted. A barium meal examination in December showed no abnormality. In March, 1952, he again vomited brownish material and a second barium meal examination then suggested a small hiatus hernia. In June, 1952, the vomiting was worse for a short period and in December a third barium meal showed no change. He was readmitted to hospital in March, 1953, with vomiting and attacks of colicky abdominal pain; no cause was found. After a symptom-free period of one year, he again began vomiting with attacks of colicky abdominal pain and, for the first time, passed bright red blood in the stools for two days. A barium meal with a follow-through examination and two sigmoidoscopies were normal. The haemoglobin was 86 g. %; the urine showed no melanin or melanogen, and the test for occult blood in his stools was negative. A week later he had a further bout of colicky abdominal pain and more fresh blood was passed per rectum. Sigmoidoscopy to 14 cm. again showed no polyposis. In view of the child's age it was not felt justified to proceed with further air-and-barium contrast or sigmoidoscopic studies.

His mother was the first to notice the inky to brown pigmentation on the lower lip at 5 months of age. It is now visible on the gums between the right lower pre-molars, on the lower lip, the upper lip, the right temporal region, the right side of the neck, and it has a butterfly distribution over the nasal bridge (Fig. 1). The

FIG. 1.—Pigmented lips.

mother, a State Registered nurse and a reliable witness, is convinced that the pigmentation increases during a bout of symptoms, and an attack at the time of writing would appear to confirm this. No pigmentation is present on the extremities, around the penis, in the fundi, ears, conjunctivae or nose.

His elder brother has a butterfly pigmentation of the nasal bridge and a pigmented patch in the right axilla together with a large area over the left deltoid. He has never had any symptoms. A grandmother had died of malignant changes in papillomatosis of the bladder.

Discussion

The syndrome, having an equal sex distribution and often familial, is believed to be genetically determined by a simple Mendelian dominant, the factors being transmitted equally by men and women. Sporadic cases, however, do occur as in the patients of Perry and Zuska (1950), of Schaffer and Sachs (1952) and of Smith (1954). It has been recorded in white people of many countries, in an American negro (Jeghers *et al.*, 1949), and in an Indian woman, many members of whose family only had the pigmentation (Basu, 1952). Tanner (1951) stated that all cases had a dark complexion but since then Kitchin's (1953) and the above case are reported as of light complexions. The pigmentation, shown by biopsy to be due to melanin, varies from a brown to a deep bluish-black and has been recorded on the lips, gums, palate, tongue, face, conjunctivae, inside the nose and rectum, on the extremities including the nail beds, and in the umbilical region. The normal pigment sites show no increase of pigmentation nor has any been recorded in the natural body folds. In particular the buccal lesions appear essential to the syndrome to exclude the diagnosis of ephelides (freckles). An interesting feature of the case here recorded is the waxing and waning of pigmentation with the symptoms. The facial pigmentation regresses with age (Peutz, 1921) although it first appears very early and is sometimes present at birth (van Dijk and Oudendal, 1925; Jeghers *et al.*, 1949; Perry and Zuska, 1950). The buccal mucosal and labial pigmentation, however, is said to be constant.

Polyposis in this syndrome occurs chiefly in the jejunal part of the small intestine, but also throughout the whole gastro-intestinal tract including the rectum and very often the stomach (Jeghers *et al.*, 1949; Tanner, 1951; Bruwer, Bergen and Kierland, 1954), the latter accounting for haematemesis as a rare presenting symptom. Polyps have also been recorded in the nose and bladder (Peutz, 1921). Polyps can occur without pigmentation (Ravitch, 1948), just as pigmentation can occur without symptoms or evidence of polyposis (Touraine and Couder, 1946; Jeghers *et al.*, 1949), but these cases may not belong to this syndrome. Oldfield (1954) has described polyposis in three members of a family associated with multiple sebaceous cysts, this syndrome also being inherited as a Mendelian dominant.

The gastro-intestinal signs and symptoms of this syndrome include colicky abdominal pain, vomiting, haematemesis, passing blood per rectum, rectal prolapse and anaemia. In confirming the diagnosis, radiographs taken after a contrast air-and-barium enema may show up polypi not seen by ordinary barium radiography (Bruwer *et al.*, 1954), although most cases have only been diagnosed after laparotomy. A diagnosis of intussusception can be made with confidence when there are symptoms of intestinal obstruction and when the bucca are pigmented (Crone and Light, 1954).

Apart from the adenocarcinomatous changes recorded in six patients (Peutz, 1921; Foster, 1944; Jeghers *et al.*, 1949; Basu, 1952; Smith, 1954), most of the 45 patients suffered from repeated intussusception and many died before or after operation. Thus the long-term prognosis is poor.

Summary

A case is described of melanosis oris associated with some of the symptoms of acute intussusception. A review of the literature discloses a definite syndrome known as the Peutz-Jeghers syndrome consisting of melanosis of the body, especially of the buccal cavity and lips associated with gastro-intestinal polyposis. The patients usually present with small bowel obstruction due to one of the polyps causing an intussusception. Although not conclusively proven, this child's disease would appear to be a further example of this rare syndrome. It is believed that this patient is the youngest on record, and the case is of special interest because the diagnosis was made without operation.

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