PEUTZ-JEGHERS SYNDROME IN CHILDHOOD

Report of a case

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Peutz-Jeghers Syndrome (Peutz 1921, Jeghers *et al.* 1949) is a condition characterised by intestinal polyposis associated with typical pigmented spots around the mouth and on the oral mucosa. The syndrome is rare and to date about 300 cases have been reported in the world literature. It is much rarer in children than in adults.

Case Summary

The patient, an 8-year old boy, was admitted to the Paediatric Ward, Kettering General Hospital, with a 12 months history of recurrent episodes of anorexia, abdominal pain and vomiting. The pain was colicky and localised to the epigastrium, usually coming on a few minutes after meals. It was followed after a variable interval by projectile vomiting which was often bile-stained and which relieved the pain slightly. During these attacks, which lasted for a few days, the boy's appetite was generally poor and he looked miserable. The bowels were regular throughout and the stools were never noticed to be blood-stained or 'tarry'. Over the month before admission, the pain had occurred almost every day and had also become constant, losing any relation to meals. The boy had lost about a stone in weight over a period of 4 months. The only other complaint was recurrent oral ulceration. The patient's parents were both well as was an elder sister. The mother had facial freckles.

On examination the boy looked pale and thin, weighing 41 lbs. (on the 3rd. percentile). He had numerous freckles on the face, especially on the nose and cheeks ('butterfly' distribution) and around the mouth *(Fig. 1)*. The pigmentation extended over the vermilion of both lips and there were everal bluish-black pigmented spots on the labial and buccal mucosa *(Fig. 2)*. He also had scattered freckles on the dorsal aspects of both hands and feet and some 'beaking' of the fingernails, but no obvious clubbing. The abdomen was scaphoid and a sausage-shaped, smooth, slightly tender mass was easily palpable in the left iliac fossa. The bowel sounds were augmented and on rectal examination there was some tenderness on the left side. There was no blood on the examining finger.

Laboratory investigation gave the following results: Hb. 5G/100ml., P.C.V. 22%,

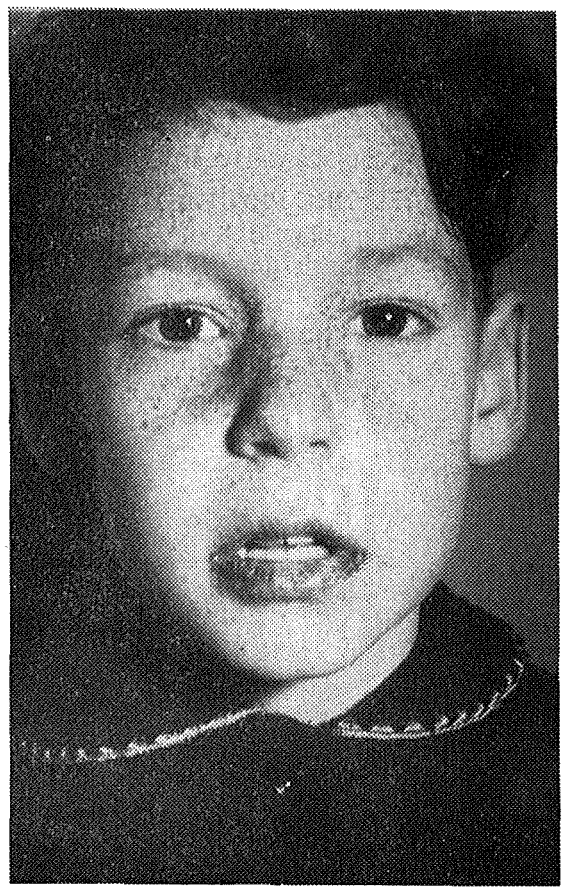

Fig. 1

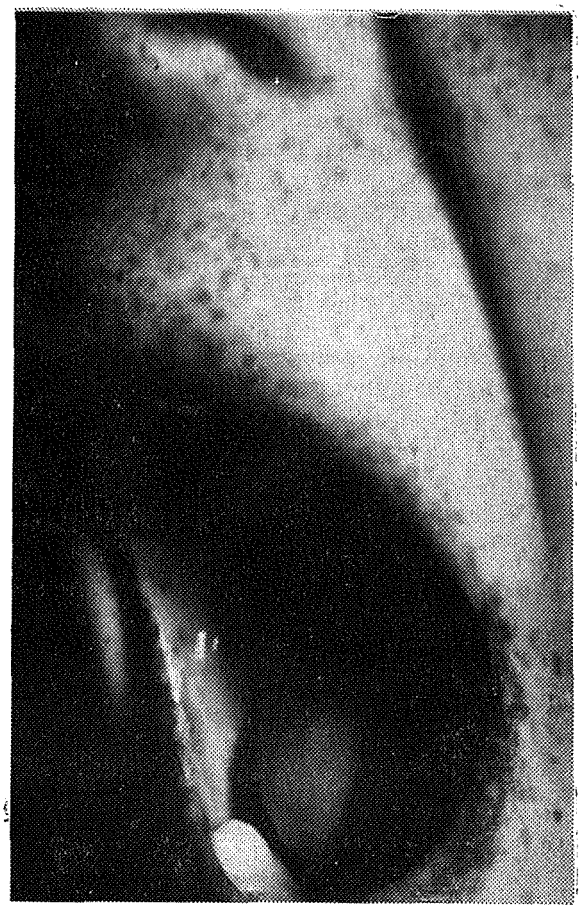

Fig. 2

M.C.H.C. 23%, M.C.V. 58cuμ. The peripheral blood film showed anisocytosis, poikilocytosis, microcytosis and hypochromia. Reticulocytes: 2.1%, E.S.R. 24mm in 1hr (Westergren). The W.B.C. count, Urinalysis, Blood Urea and serum Proteins were normal. The serum Electrolytes were: Na 127mEq/lit., Cl 93mEq/lit., K 3.2mEq/lit. The Heaf test was negative. Stool examination for occult blood was positive on three occasions. A plain X-Ray of the abdomen was normal. On barium study there was no evidence of hiatus hernia or oesophageal reflux and no ulcers could be seen in the stomach or duodenum. Folow-through for small intestine showed a large dilated loop of jejunum. The rate of transit of the contrast medium was normal. There were numerous, small filling defects in the ileum which were very suggestive of small polypi and two other large ones in the jejunum. The colon was normal. The overall impression was that of intermittent intussusception (although no actual apex for this could be seen) and multiple small intestine polyps.

During the first week in hospital, the boy had several episodes of abdominal pain with vomiting and examination on different occasions showed visible peristalsis, tenderness in the epigastrium and an evanescent abdominal mass which shifted in position from day to day. The bowel sounds were increased. The motions were normal. In view of the decision to operate, the anaemia was corrected by blood transfusion.

Sigmoidoscopy up to 15cms. was normal. At laparotomy (Mr J. H. C. Phillips) there was no small bowel dilatation but six polyps were found in the upper jejunum *(Fig. 3)*. Approximately 25cms. of involved jejunum were excised and an end-to-end anastomosis performed.

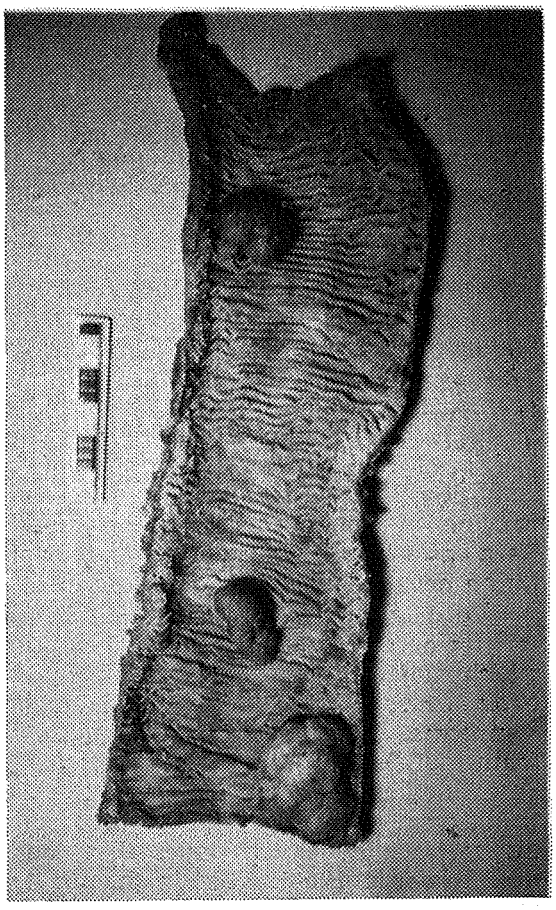

Fig. 3

The pathologist's report (Dr. P. S. Andrews) was as follows: "23 cms of small intestine with six polyps. The largest is sessile 3.2 x 2cms in size; others, ranging in size from 2.5 x 1.2 cms down to 0.2 x 0.1cms. No glands were found in the pedicle. Histology showed benign jejunal adenomatous polypi with abundant collections of histiocytes in parts of the stroma". The histology of a freckle excised from the skin was normal except for some irregularity of the melanin pigmentation in the basal-cell layer.

The post-operative period was uneventful and the patient was discharged home twelve days after operation with a Haemoglobin of 11 G %. He has been followed up as an out-patient and has up to now remained well and symptom-free.

Comments

Peutz-Jeghers Syndrome can present at any age but reports of it in the paediatric literature are sparse (Massimo 1959, Wenzl *et al.* 1961). Cases occurring in childhood usually come to light after the condition has been discovered in one of the patient's parents or older siblings.

The condition is inherited as an autosomal dominant of high penetrance and the majority of cases have therefore been found to involve several members of one family (Christiaens *et al.* 1959, Bartholomew *et al.* 1962). Sporadic cases do however occur and the case here reported is probably one of these. The boy's mother had facial freckles at first suggestive of the condition, but she was symptom-free and had no pigmentation inside the mouth. Rare cases where either polypi or pigmentation occur alone have been described.

In diagnosis, it is usually the characteristic peri-oral and buccal pigmentation which suggests intestinal polyps as the basic cause of the patient's abdominal symptoms. A peculiar and often puzzling feature of the syndrome is the presence of an evanescent palpable mass which shifts to different positions in the abdomen during separate colicky episodes (Dormandy and Edwards 1956). This is due to the self-reducing intussusception and, as happened in this case, it may be mistaken initially for a faecal mass and therefore thought to be of no significance.

The condition can closely mimic the syndrome of cyclical vomiting, which is very much more common and is not associated with buccal pigmentation.

The polyps are notoriously difficult to visualise radiologically and barium studies are very often negative (Dormandy 1958). In most cases therefore, the absolute diagnosis can only be made at laparotomy when multiple polyps are found in the intestine, usually in the jejunum and ileum.

The question of possible malignant change occurring in these polyps is still somewhat controversial. In a review of 67 published cases, as high an incidence of malignancy as 19% was recorded (Bailey 1957). Dormandy attributed this to misinterpretation of the histological findings and pointed out that clear-cut cases of metastases had never been reported (Dormandy 1958). A definite case of Peutz-Jeghers syndrome with metastases in a 52-year old woman has however been subsequently recorded (Williams and Knudsen 1965). Nevertheless, most authorities seem to agree that such a malignant change is extremely rare and histologically the polyps are usually hamartomatous malformations (Dormandy 1958, Bartholomew *et al.* 1962). Because of this, most surgeons prefer conservative treatment and perform multiple enterotomies and polypectomies rather than gut resections. In the case here reported it was thought justifiable to resect the involved part because the polyps were localised to a small stretch of jejunum.

The prognosis of the condition is, on the whole, good though these patients are prone to develop anaemia from intestinal blood loss and have repeated episodes of intussusception from further polyps which tend to crop up in spurts over the years and which often require further surgical treatment.

Acknowledgements

I would like to thank Dr. R. Wigglesworth, Consultant Paediatrician in the Kettering and Northampton Areas for permission to publish this case.

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