INTESTINAL POLYPOSIS WITH MUCOCUTANEOUS PIGMENTATION:

THE PEUTZ-JEGHERS SYNDROME

REPORT OF A CASE


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THE Peutz-Jeghers Syndrome is an unusual familial defect in which polyps occur in the stomach and intestines together with a macular melanotic pigmentation of the skin and certain mucous membranes. The polyps are hamartomas, not true neoplasms. The colour of the pigmentation varies from light brown to bluish black. It always affects the buccal mucosa, frequently the lips and less often the areas around the eyes and nostrils and the skin of the hands and feet. It is most striking in children and tends to fade after puberty.

The entity was first described by Peutz,1 a Dutch physician, in 1921. Jeghers, McKusick and Katz2 in 1949 published an account of 10 cases and this drew general attention to the syndrome. An investigation of 21 patients from five families was then reported by Dormandy.3 Recently, Bartholomew, Moore, Dahlín and Waugh added 8 new cases and in an exhaustive review collected 174 from the literature.4 Although the disease has been described in many countries and in all races, previously it has not been reported in Ireland.

REPORT OF A CASE

Case No. A7437, a 58 year old unmarried, male store keeper was admitted to our hospital on the 29th November, 1961. He gave a two year history of pain in the right buttock which radiated into the right leg. It was associated with numbness of this limb. No history of trauma, malaena, or haematemesis was obtained.

Past History disclosed a single episode of colicky abdominal pain in 1951 which subsided spontaneously. The patient had been known to be hypertensive for some years' but no investigations were carried out and no treatment prescribed.

Family History showed that his father died when aged 57 years from intestinal obstruction caused by a bowel tumour. Neither operation nor autopsy was performed. Except for the father there was no evidence even remotely suggesting Peutz-Jeghers syndrome in any member of the family. We are aware of 50 members (including the patient) in five generations and propose to make a detailed study of these and report on them in the future.

Examination revealed a co-operative grey-eyed dark-haired moderately obese man. The pulse and temperature were normal, but the blood pressure was 210/115 millimeters of mercury.

On the lips and in the mucosa of the oral vestibule were many circumscribed macular areas of bluish-black pigmentation. (See fig. 1) Similar, though lighter coloured and more sparsely distributed spots were observed on the anterior and posterior aspects of the hands and fingers.

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Neurological examination disclosed a sensory defect with absent touch discrimination on the right side below the eighth dorsal segment. It included the right leg and was associated with loss of temperature appreciation. There was no motor defect.

Despite the hypertension no cardiac, renal or retinal abnormalities were found.

Investigation showed glycosuria and a glucose tolerance curve of the diabetic type. Radiological examination of the gastro-intestinal tract revealed discrete polyps in the duodenum, small bowel and colon. (See fig. 2) There was evidence of spondylosis from T.8 to T.10 and from L.3 to L.4 on roentgenological examination of the spine. The following investigations gave normal results, full blood count, blood urea estimation, cerebro-spinal fluid examination, disagnex blue test, sigmoidoscopy, radiological investigation of the chest, hips, pelvis and urinary tract, and peroral biopsy of the jejunal mucosa.

Progress. The diabetes was stabilised with insulin and control was maintained by oral administration of tolbutamide. Rest and sedation quickly brought the blood pressure into the normal range. Despite analgesics and physiotherapy the limb pain remained troublesome and on March 4th a left hemi-cordotomy was performed. (Mr. J. P. Lanigan). It gave immediate relief. Unfortunately this was temporary and the pair returned.

Fig. 1.—Photograph of lips and vestibule of the mouth. Note the dark stippled pigmentation on both lips and the lighter coloured spots on the oral mucosa.

Fig. 2.—Polyps illustrated in radiographs taken using a special technique for outlining the small bowel. (a) Note the cluster of micropolyps which are a feature of the Peutz-Jeghers Syndrome. (b) Polyps of a larger size. (Full size).