

PEUTZ-JEGHER SYNDROME CASE REPORT AND REVIEW OF LITERATURE

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Peutz-jeghers syndrome is characterised mainly by gastrointestinal polyposis and mucocutaneous pigmentation. It is an inherited condition defined by the presence of hamartomatous polyposis of gastrointestinal tract and melanin spots on the lips & buccal mucosa. The Polyps are mostly found in the small bowel and less frequently seen in the stomach and colon. A case of peutz-jeghers syndrome is presented with a complain of severe abdominal pain; small bowel intussusception was diagnosed and an operation was performed.

Key words: Peutz-Jeghers syndrome

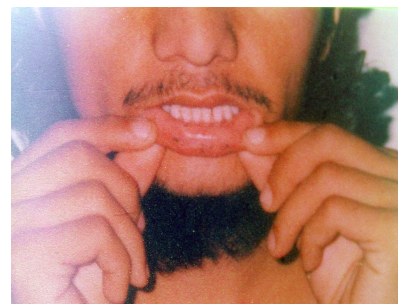
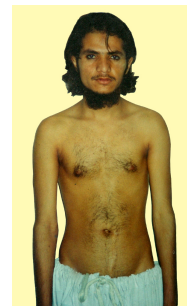
INTRODUCTION

Peutz-jeghers syndrome is characterised mainly by gastrointestinal polyposis and mucocutaneous pigmentation. It is an inherited condition defined by the presence of hamartomatous polyposis of gastrointestinal tract and melanin spots on the lips & buccal mucosa. The hamartomas are found most frequently in the small bowel, but are only slightly less frequent in the stomach & colon. The hamartomas have been described occasionally in other mucosal surfaces, including the upper respiratory tract & urinary tract. The polyps of Peutz-Jeghers Syndrome are composed of normal elements indigenous to the site in which they arise, but in which the architecture is markedly abnormal. Histologically, the lesions usually have an arborizing pattern with fronds covered by normal epithelium that incorporated all the cell types usually located in the site of origin. Endocrine cells are present in the polyps if all sites. Smooth muscle form an important component of the lesions & extends into the branching fronds of the polyps. Benign glands may be surrounded by smooth muscle & may extend into the submucosa. Carcinoma appears to be an infrequent complication of Peutz-Jeghers syndrome. However Linos co-workers followed 48 Patients with Peutz-Jeghers syndrome for a period of 33 years no patient with the complete syndrome developed of Gastric carcinoma.

CASE REPORT

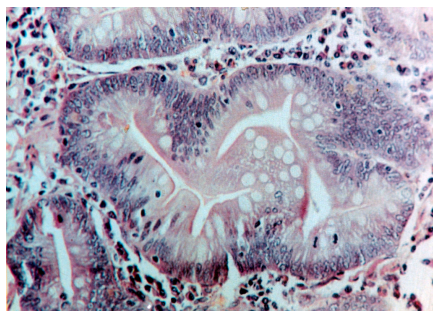
The patient, a young man of 22 years presented at the Jinnah Postgraduate Medical centre, Karachi with a complaint of severe abdominal pain; small bowel intussusception was diagnosed and an operation was performed. Many polyps about 9 were

detected by barium meal and removed. The post operative course was uneventful. He had a history of laprotomy done at the age of 8 years for intussusception and polyps were removed since then patient complained constipation off and on. Barium meal was done, polyps were seen in large intestine, sigmoid colon.



Family History is being looked into, as the patient says his younger sister is having similar complained but not investigated. Laprotomy was done and polyps were removed from duodenum and sigmoid colon. Laboratory data on admission were normal, carcinoembryonic antigen was not done.

Pathological findings; Of the 9 polyps resected, all were hamartomatous. No malignant change was found. Largest polyp measures 6x5 cms and smallest measures 1x0.8 cms cut surface of the polyps revealed papillary gray white growth. No obvious stalk seen.



DISCUSSION

Petuz-jeghers syndrome with its polyps and rather distinctive skin and oral mucosal pigmentation was described by peutz in 1921 and become more widely recognised after the article by jeghers and coworkers in 1949.

Petuz-jeghers syndrome is inherited as an autosomal dominant characteristic and comprises multiple polyps of the stomach and bowel leading to transient intussusception and a brownish pigmentation peppered around the mouth and lips. The melanin spots of PJS are present in over 90 % of patients^{2,3} and occur most commonly on the lips and buccal mucosa, but are found also on circumoral and fascial skin, on the palm and soles and on the digits. The melanotic macules on the skin may not develop until after the intestinal polyps and may fade with age^{4,5}.

In a recent review of Utsunomiya et al (1975), 189 of their 222 patients developed polyps. At some stage in the colon or rectum. The polyps apparently continue to appear and may either grow or remain static for long periods⁵.

The general opinion has been that these polyps have little or no precancerous distribution. However there has been a slow and steady flow of reports of carcinoma associated with this syndrome. Reid (1974) suggested that 2-3% of patients with peutz - jeghers syndrome developed gastrointestinal carcinoma, with duodenal and antral carcinoma being most frequent^{6,7}. Because there are now at least 19 reports of cancer arising in individual with PJS who were less than 40 years of age, it is difficult not to think that the cancers are in some way related to the syndrome^{1,8,9}.

It is usually thought of in connection with the small intestine since at the site of polyps more

readily cause intussusception. The polyps however are distributed throughout the gastrointestinal tract.

Extra cutaneous lesions most often involve female patients with peutz - jeghers syndrome appear to have a definite tendency to develop bilateral breast cancers.

An unusual ovarian tumor first described by scully in (1970) and given the name sex cord tumor with anular tubules can be identified in almost all female patients with peutz-jeghers syndrome³. If the ovaries are examined carefully, well differentiated "adenocarcinoma cervix (Adenoma magnum)" has been seen in patients with peutz-jeghers syndrome. Six of approximately 20 reported cases of this tumor have occurred in such patients. Male patients are not spared the gonadal tumors of pjs⁶; cantu and colleagues reported a 6- year old boy with a feminizing sertoli cell tumor of the testis. Female patients with PJS appear to have a definite tendency to develop bilateral Breast cancers as a number of recent reports testify many other neoplasms have been described in individuals with PJS but it is difficult to ascertain whether they occur in this population with an increased frequency, because they include tumors that are common in general population⁹.

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