

Inaugural Degeneration of a Polyp during Peutz-Jeghers Syndrome: A Case Report and Literature Review.

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Summary:

Peutz-Jeghers Syndrome (PJS) is a rare autosomal dominant disease, defined by the association of a lentiginosis and lesion of the digestive, pulmonary and reproductive organs.

The PJS gene has been located on chromosome 19p13.13. The skin signs are visible and constitute a telltale sign of the disease, they are type of lentigines, They sit most often on the lips, the oral mucosa.

Keywords: Peutz -Jeghers syndrome, Lentiginosis, Polyposis, Invagination.

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I. Introduction:

In 1949, an American doctor named Harold Joseph Jeghers published a detailed description of patients with intestinal polyposis and skin pigmentation, which led to the identification of the syndrome named after the two doctors (**1, 2, 3**).

The prevalence is estimated at 1 / 200,000, with no predominance of sex or race, the average age of diagnosis is 22 years out of a review of 75 cases (**4**).

The inaugural degeneration of polyps in the framework of Peutz-Jeghers syndrome has been proven histologically by the demonstration of the hamartoma-dysplasia-cancer parentage, and STK11 which is a tumor suppressor gene when it is inactivated by the allelic imbalance (**56**), leads to the development of the hamartomatous polyp and the occurrence of a somatic mutation which is at the origin of its degeneration.

II. Observation:

Patient R. M, 21 years old, without any previous history, who consulted an emergency for severe abdominal pain with incoercible vomiting, without fever and whose clinical examination showed mucosal cutaneous pallor with agitation, moreover there is a physical sign Particular to the inspection in the case of perioral lentiginosis (**Photo 1**), the pain is of epigastric site without significant distension, the digital rectal examination is without particularity and the hernial orifices were free.

The ASP (**Photo2**) showed hydro aeric levels of small intestine and the abdominal ultrasound had objectified an acute intestinal intussusception without detectable causes, we decided to intervene immediately.

The acute intestinal invagination was caused by a degenerated intestinal polyposis at the level of the first jejunal loop (**Photos3**). The patient underwent a jejunal resection (20cm) taking away 3 polyps including one degenerate (adenocarcinoma) restoration of digestive continuity. The follow-up operations were interspersed with an acute intestinal obstruction that required resumption of surgery where the dissection was laborious and the hemodynamic state precarious which forced us to perform a double ileostomy.

This intervention was complicated by acute peritonitis generalized by anastomotic release and six months later, we carried out the restoration of digestive continuity, after investigations, and anatomopathological study this polyposis was part of the Peutz-Jeghers syndrome.



**Photo. 1 perioral
lentiginosis**



**Photo. 2 hydro
aeric levels**



**Photo. 3 The acute
intes tinal
invagination**

III. Discussion:

The Peutz-Jeghers polyps have their own histological characteristics which make it possible to distinguish them from other hamartomatous polyps, and in particular juvenile polyps. They can be the seat of adenomatous contingents which would explain the risk of possible degeneration. They are the second cardinal sign of this syndrome and they can reveal the disease when they manifest themselves at the outset by their complications such as digestive hemorrhages and intestinal obstructions (7-8). People with PJS are at increased risk of cancer, the cumulative risk of cancer is estimated at 3% **Tb1**, The organs most frequently affected are the gastrointestinal tract (esophagus, stomach, small intestine, colon, rectum and pancreas) (4), lung (9), prostate (10), breast and reproductive organs (3-11).

Table 1: The Cumulative Risk of Cancers:

<i>Organs</i>	<i>Cumulative Risk (%)</i>	<i>Relative risk and Confidence inter</i>
<i>Esophagus</i>	0,5	57 (2-557)
<i>Stomach</i>	29	96(96- 368)
<i>small intestine</i>	13	520(230-1306)
<i>Colon</i>	39	84 (41-137)
<i>Pancreás</i>	32	64 (71-132)
<i>Lung</i>	36	132 (44-216)
<i>Breast</i>	15	17 (5-39)
<i>Uterus</i>	54	15 (8- 27)
<i>Ovary</i>	9	16 (2 - 56)
	21	27 (7- 68)

The management of PJS is mainly based on surveillance and the treatment of hamartomatous polyps. There is no standardized treatment for skin and mucosa pigmentation. Treatment involves cryosurgery, dermabrasion and laser (3).

Recommendations: Monitoring: R.J. Fingerote, emedecine 2005

- Annual clinical examination (breasts, abdomen, pelvis, and testicles)
- NFS (3-8)

-Resection of any polyp > 5mm

-Upper endoscopy, colonoscopy, and Video Capsule Endoscopy / 2 years - Smear / 3 years.

These digestive cancers can develop not only in the small intestine, but also in the stomach and colon rectum, sometimes at a very young age. When it comes to monitoring the small intestine, the CT scan and endoscopic capsule are the exams of choice that have overtaken barium transit. Entero-MRI may have a place in the future. It is currently recommended to carry out, from the age of 16, an examination by endoscopic capsule and / or enteroscanner every 2 years. Laparotomy with intra operative enteroscopy and removal of the maximum number of small bowel polyps is the standard treatment in order to avoid acute complications and limit the number of surgical procedures.

Now, double balloon enteroscopy allows the excision of certain polyps, as long as they are accessible by high or low route.

In practice, the removal of small bowel polyps by double balloon enteroscopy is increasingly performed and it avoids too frequent recourse to surgery. Tumor risks justify the implementation of a systematic screening strategy for digestive, but also pancreatic, gynecological and mammary tumors. In our young patient, the diagnostic argument, namely the degenerative polyp invagination associated with a peri-oral lentiginous were in favor of Peutz-jeghers syndrome, in her case, the follow-up is based on expert recommendations. Colonoscopies must be performed every 2 to 3 years, and be associated with an esogastroduodenal endoscopy every 3 years from the age of 25 years, associating in this context the lifelong research of digestive and gynecological cancers as well as the detection of the degeneration of polyps.

IV. Conclusion:

The degeneration of polyps in the framework of Peutz Jeghers syndrome is currently accepted on the basis of our observation and data from the literature, the surveillance of polyps has a double objective consisting in watching for complications as well as cancer.

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