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Degenerated Peutz-Jeghers Syndrome with Peritoneal Carcinomatosis: A Case Report and Overview of a Rare Disease

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1. Abstract

Peutz-Jeghers syndrome (PJS) is a genetic disease with dominant autosomic transmission, characterized by the development of hamartomatous polyps in the digestive tract, associated with muco-cutaneous pigmentation. PJS is a serious condition that can be life threatening due to the significant increase of cancer risk. We report the case of a 44-year-old man diagnosed with PJS at age 3 that was revealed by bowel obstruction for which the patient underwent surgery with grelic resection. The patient presented with peritoneal carcinosis with grade 2 ascitis secondary to an unknown primitive cancer, most likely of gastro-intestinal origin due to a malignant transformation of one of more of the polyps with a suspicious macroscopic aspect during endoscopy. Evolution was unfavorable, with the patient dying after bowel obstruction caused by obstructive polyps in the jejunum. Management of patients with PJS require a multidisciplinary approach, aiming at preventing and managing the associated complications; most notably bowel obstruction, intestinal invagination, and cancer.

2. Introduction

Peutz-Jeghers syndrome is a relatively rare genetic disease with dominant autosomal transmission. It is most often due to germ-line mutations in the STK11 (LKB1) gene on chromosome 19p13.3 and is characterized by gastrointestinal polyposis and mucocutaneous pigmented macules, particularly on the vermillion border of the lips) [1].

It was first described in 1921 by the Dutch physician Jan Peutz, then further detailed by the American internist Harold Jeghers, who demonstrated the associated risk of invasive cancers in patients who carry the aforementioned genetic mutation [2].

Polyps in the digestive tract are a cardinal sign of this syndrome and can reveal the disease with their complications such as hemorrhage and bowel obstruction. These polyps are found predominantly in the jejuno-ileon (90%), in the stomach (24%) and in the colon (9%). There is no correlation between the size of the polyps and the risk of carcinomatous transformation. It is estimated that around 2/75 of polyps can become malignant [2].

Some patients with Peutz-Jeghers syndrome develop gastro-intestinal lesions during their childhood. These patients require continuous medical care up until their adult age and suffer from sometimes serious complications that significantly alter their quality of life [3].

The scarcity and variability of the available literature data regarding this disease makes it difficult to reliably assess the tumoral risk and thus to establish guidelines on the monitoring protocol for patients who carry the STK11 mutation, whether they are symptomatic or not at diagnosis [4].

3. Observation

We report the case of Mr. R.D, aged 44, diagnosed with Peutz Jeghers syndrome at age 3 that was revealed by bowel obstruction for which the patient underwent surgery with grelic resection. He was operated two more times, at age 16 and 18, following the same clinical presentation.

The patient had no other reported history of medical problems. His son, aged 5, has the same syndrome for which he is regularly seen by pediatricians.

He was admitted to the hospital for an oedemato-ascitic syndrome that appeared a month before, with no accompanying signs aside from a marked alteration of the general state of the patient.

Clinical examination found grade 2 ascitis and oedema of the lower limbs reaching the upper part of the thighs.

Diagnostic paracentesis was performed with the following findings: Yellowish color, with leucocyte count at 320/mm3 and total protein at 34g/l. Cytological examination of the ascites found atypical cells, that appeared probably carcinomatous when immunohistochemistry was performed.

Abdomino-pelvic CT scan revealed a mild but diffuse submucosal swelling in the stomach, the small bowel and the colon. This swelling was nodular in places, iso-dense, with homogeneous enhancement after contrast agent injection.

Endoscopic investigations were performed. EGD revealed multiple polyps of different dimensions with a suspicious macroscopic aspect, lining the antro-fundic and duodenal mucosa. The same aspect was observed during colonoscopy. Multiple biopsies were done (Figure 1).

Histological examination of the gastric and colonic biopsic fragments showed polypoid formations with glandular distorsion, a hamartomatous aspect and ramified bundles of smooth muscle. This aspect is compatible with Peutz Jeghers polyps. No signs of dysplasia or malignancy were found.

The patient's state was aggravated by the onset of a sub-occlusive syndrome. Clinical examination found generalized abdominal guarding. An abdominal and pelvic CT scan was quickly performed and found multiples polyps in the lumen of the digestive tract, one of which, located in the jejunum, was particularly voluminous at 67 x 44 mm and obstructive, responsible of a dilation of the upstream loops. In addition, cloistered grade 3 ascitis was observed, with peritoneal implants associated to a diffuse infiltration of the mesenteric adipose tissue, suggesting peritoneal carcinosis.

The patient left the hospital against medical advice, turning down medical care. He died home, five days later (Figure 2 and 3).

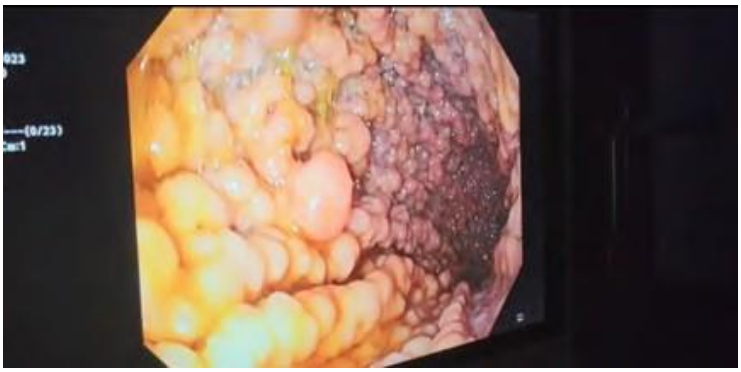

Figure 1: Multiple gastric polyps on endoscopy

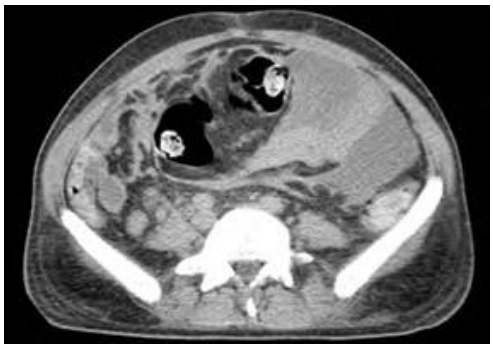

Figure 2: Multiples polyps in the lumen of the digestive tract

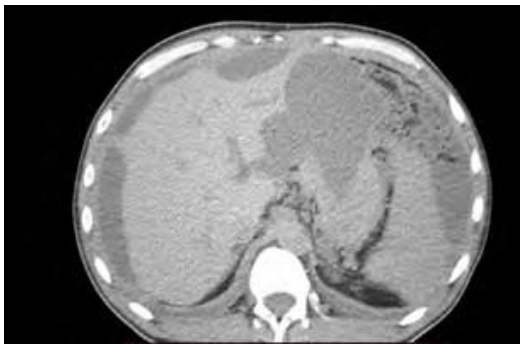

Figure 3: Cloistered grade 3 ascitis

4. Discussion

Peutz-Jeghers syndrome is a rare disease whose incidence is estimated at 1/25000 to 1/300 000 births. It can occur in every ethnicity, and affects men as much as women [5].

The gene responsible for Peutz-Jeghers syndrome is located on the 19p34-p36 chromosome and codes for the serine/threonine kinase (STK11), which corresponds to a tumor suppressing gene. A mutation of this gene causes an unrestrained cellular proliferation, which leads to the development of hamartomas and cancers [6].

Peutz-Jeghers syndrome can manifest in a hereditary or sporadic fashion, with a significantly heterogenous clinical presentation [6]. In our case, we could not find parents that had the syndrome in the patient's family. However, literature data suggests that about 50% of cases are sporadic and correspond to new mutations, which was probably the case with our patient; who subsequently transmitted the disease to his 5-year-old son. A genetic study could not be performed.

Diagnosis is suspected in patients having at least two Peutz-Jeghers polyps; any number of polyps if they have a family history of PJS; muco-cutaneous pigmentation with a family history of PJS; or any number of Peutz-Jeghers polyps associated with muco-cutaneous pigmentation [6].

In our patient's case, gastric, duodenal and colonic polyps were found during endoscopy and on the CT scan, with histological examination identifying them as hamartomas. There was no muco-cutaneous hyperpigmentation. However, literature data suggests that not all patients present the entire clinical spectrum of the disease. Around 95% of patients with PJS have muco-cutaneous hyperpigmentation [6,7].

PJS is a serious disease that can be life-threatening due to the significant increase in cancer risk (cumulated risk estimated at 89%) [4]. Mean age of cancer occurrence is 40 [8]. Digestive cancers are the most frequent, with a cumulated incidence of 55% (colo-rectal cancer: 39%, small bowel cancer: 13%, pancreatic cancer: 11 to 36%). There is an increase in the risk of extra-digestive cancers as well, most notably breast cancer, whose risk is similar to that of patients who carry the BRCA1 or BRCA2 mutation (cumulated incidence of 45%); as well as gynecological and gonadic cancers: cervical cancer with specific histological characteristics (adenoma malignum), ovarian cancer, testicular cancer… Lung cancer risk is increased as well [4].

In our patient's case, he had peritoneal carcinosis due to an unknown primary cancer, most likely of digestive origin caused by malignant transformation of one or some of the digestive tract polyps.

Polyp-induced complications include abdominal pain, hemorrhage and bowel obstruction due to the invagination or the obstruction of the gastro-intestinal lumen. They can appear as early as the first year of life; or much later during the fifth decade. At 10 years old, 30% of patients with PJS have already underwent laparotomy. In case of symptomatic or voluminous (diameter>1.5cm) polyps, laparotomy with enteroscopy are recommended.

Around half of patients had at least two laparotomies with intestinal resection, which explains a relatively high prevalence of short bowel syndrome in this population [9].

Similarly, our patient had his first complication at age 3. At age 18, he had already undergone 3 laparotomies.

Management for patients with PJS requires a multidisciplinary approach for life. It includes monitoring and prevention of disease manifestations as well as treatment of eventual complications [10]. The current guidelines include a screening EGD starting from the age of 12; which should be repeated every year in the presence of polyps, and every two to three years in their absence. Screening colonoscopy is also recommended from the age of 12 and earlier in case of symptomatic disease; and should be performed annually in the presence of polyps and every one to three years in their absence. Magnetic resonance cholangiopancreatography (MRCP) or endoscopic ultra-sound (EUS) are recommended from age 25 to 30 and should be done every one to two years.

Clinical examination of the breasts should be done bi-annually starting from the age of 25. Mammography from age 25, and annual Papanicolaou with annual transvaginal ultrasound from age 18. For men, an annual testicular examination is recommended, and ultrasound is indicated if the patient is symptomatic from birth [1-5-6-11].

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