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A Comprehensive Case Review of Peutz-Jeghers Syndrome: Clinical and Pathological Perspectives

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Abstract **Case Report**

Peutz-Jeghers syndrome is an autosomal dominant disorder characterized by multiple hamartomatous polyps in the stomach, small intestine and colon, together with typical pigmented skin lesions. Skin lesions may appear in early childhood. They are located in the orifices: nose, lips, hands and feet, and in the anal and genital region. Polyps progressively increase in size, and can lead to occlusive complications through invagination, or to severe haemorrhaging when necrosis occurs. Carcinomas have been reported, probably developing from foci of adenomas. Other tumors are observed after the age of 50: testicular cancer, ovarian cysts, breast cancer, sometimes bilateral from the start, pancreatic and biliary cancers. The gene has not yet been identified. Treatment is endoscopic or surgical. Surveillance of subjects at risk is difficult. We report the case of a young patient aged 32, followed since 2018 for a Peutz Jeghers syndrome revealed during a workup of medium-abundance rectal discharge evolving in a context of altered general condition.

Keywords: Peutz Jeghers Syndrome, Hamartomatous Polyps, Oral Lentiginosis, Degeneration, Monitoring.

INTRODUCTION

Peutz-Jeghers syndrome is an uncommon genetic disorder characterized by autosomal dominant inheritance. The syndrome is the result of a germline mutation in the STK11 (LKB1) gene, and manifests itself as hamartomatous polyps in the gastrointestinal tract, accompanied by mucocutaneous pigmentation, particularly on the vermilion border of the lips [1]. First described in 1921 by the Dutch physician Jan Peutz, this syndrome was later studied in detail in 1949 by the American Harold Jeghers, who highlighted the increased risk of invasive cancers in patients carrying the genetic mutation. It is essential to take environmental factors into account when studying interactions between animal species [2]. Digestive polyps are a cardinal sign of this syndrome, and may reveal the disease when they are immediately manifested by complications such as digestive haemorrhage and obstruction. These polyps are located in the jejuno-ileum (90% of cases), colon (9% of cases) or stomach (24% of cases). There is no relationship between polyp size and degenerative potential. It is estimated that 2/75 polyps can degenerate [2]. Some individuals suffering from Peutz-Jeghers syndrome develop gastrointestinal lesions as early as childhood, requiring regular medical follow-up throughout adulthood. These patients can sometimes experience severe complications that have a significant impact on their quality of life [3]. The scarcity and variability of data available in the literature concerning this syndrome illustrate the complexity of precise tumor risk assessment, and consequently the difficulty of establishing recommendations for monitoring patients with a mutation in STK11, whether or not they are symptomatic at diagnosis. It is essential to take environmental factors into account when planning a construction project [4].

OBSERVATION

We report the case of Mr E.K, 32 years old, with no particular pathological history, followed since 2018 for Peutz Jeghers syndrome, revealed during a picture of chronic rectorrhagia of low to medium abundance evolving since with no notion of melena or haematemesis, no rectal syndrome, no notion of proctalgia or abdominal pain or transit disorders, no abdominal distension and no other associated digestive or Extradigestive signs. The patient was hemodynamically stable, with apyrexia and asthenia.

Clinical examination revealed a conscious, stable patient, very malnourished and dehydrated, with oral lentiginosis (Figure 1) and a blood-stained finger

pad on rectal examination. The rest of the clinical examination was unremarkable. The workup showed microcytic hypochromic anemia (Hb 6g/dl) with iron deficiency (ferritinemia 6) and hydroelectrolytic disorders (hyponatremia 123 and hypokalemia 2.5).

The patient underwent an endoscopic work-up. FOGD revealed an antrofundial and duodenal mucosa lined with polyps of varying caliber and suspicious appearance (Figure 2). Similarly, colonoscopy revealed a mucosa lined with polypoid formations, some of which were suspicious in appearance.

Histological examination of biopsy fragments from the stomach, as well as polyps removed from the colon, showed gastric and colonic polypoid lesions whose morphological appearance may be consistent with peutz jeghers polyps, but without signs of dysplasia or malignancy.

An abdominal CT scan revealed discrete diffuse submucosal thickening in the stomach, coves and colon. This thickening was nodular in places, iso-dense and homogeneously enhanced after contrast.The patient's management consisted of close surveillance with annual endoscopic monitoring and screening for cancers of systems other than the gastrointestinal tract.

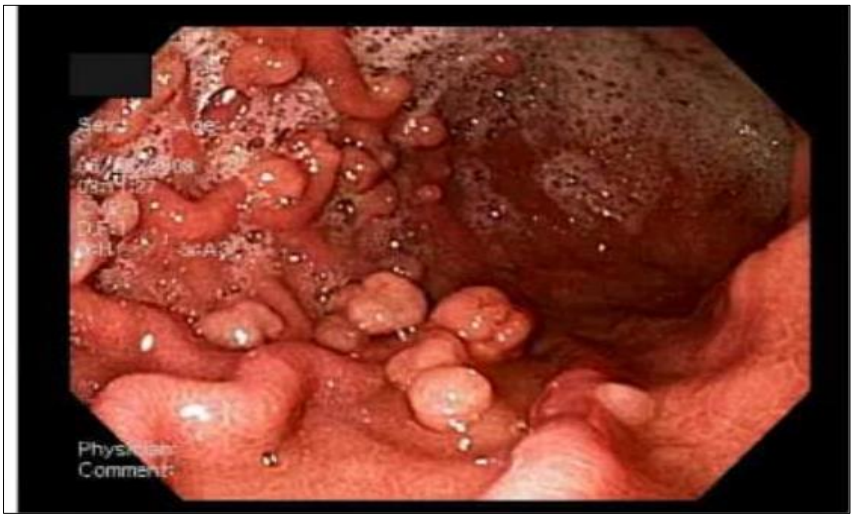

Figure 1: Oral lentiginosis in our patient

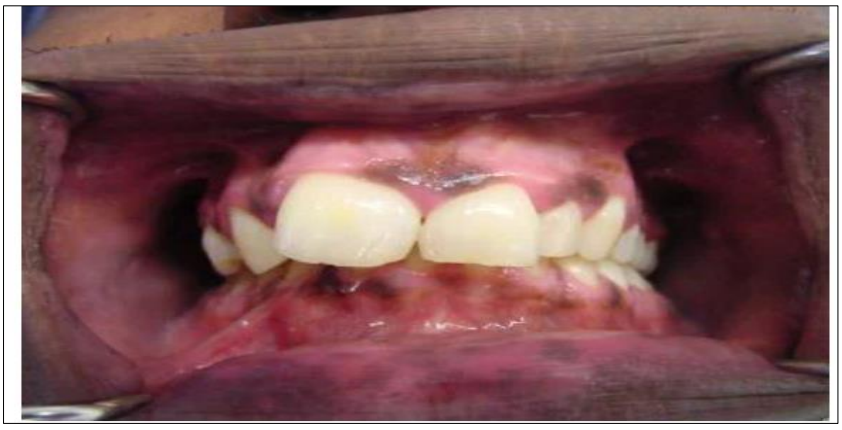

Figure 2: Multiple gastric polyps at our patient's FOGD

DISCUSSION

Peutz-Jeghers syndrome is a rare disorder, with an estimated incidence of around 1 case per 25,000 to 300,000 births. The syndrome can occur in all ethnic groups and affects males and females equally [5]. The Peutz-Jeghers syndrome locus is identified on the short arm of chromosome 19, between chromosome bands 34 and 36, and contains the STK11 gene encoding a serine-threonine kinase. This gene acts as a tumor suppressor, and any genetic alteration at its level induces anarchic cell proliferation, favoring the formation of hamartomas and malignant tumors. Peutz-Jeghers syndrome can present in hereditary or sporadic patterns, with considerable clinical diversity [6].

In our case, we were unable to find any relatives with Peutz-Jeghers syndrome in the patient's family. However, according to the literature, around 50% of cases are sporadic and represent new mutations, as is the

case in our patient. The genetic study was not carried out in the patient.

The diagnosis is considered in patients with two or more Peutz-Jeghers polyps, any number of polyps with a family history of Peutz-Jeghers syndrome, mucocutaneous pigmentations with a family history of Peutz-Jeghers syndrome, or any number of Peutz-Jeghers polyps with mucocutaneous pigmentations [6]. In our case study, gastric, duodenal and colonic polyps of the small intestine and stomach were found on endoscopic examinations and confirmed histologically as hamartomas. We found no mucocutaneous hyperpigmentation, but according to the literature, some patients do not present the full spectrum of the disease, and around 95% of patients with Peutz-Jeghers syndrome have mucocutaneous pigmentation [6, 7].

Peutz-Jeghers syndrome is a serious, life-threatening condition that significantly increases the lifetime risk of cancer (cumulative risk 89%) [4]. The average age of cancer sufferers is 40 [8]. At the top of the list are digestive cancers, with a cumulative incidence of 55% of gastrointestinal cancers, including 39% of colorectal cancers, 13% of small intestine cancers and between 11% and 36% of pancreatic cancers. There is also an increased risk of extradigestive cancers, most notably breast cancer, whose risk is almost identical to that of patients with a deleterious BRCA1 or BRCA2 mutation (cumulative incidence 45%). There is also a risk of tumors of the gynecological and gonadal spheres: cancers of the uterine cervix with particular histological characteristics (adenoma malignum), ovarian sex cord tumors with ringed tubules (SCATs) and testicular tumors with calcified Sertoli cells. The risk of lung cancer is also increased [4].

Complications associated with polyps include colicky abdominal pain, bleeding, and intestinal obstruction caused by invagination or obstruction of the gastrointestinal lumen by polyps. They can appear as early as the first year of life, or by the age of 40. By the age of ten, 30% of individuals suffering from Peutz-Jeghers syndrome have already undergone a laparotomy. In the case of symptoms or large polyps (diameter greater than 1.5 cm), laparotomy with enteroscopy is recommended. Almost half of patients underwent two or more laparotomies, leading to a significant percentage of patients developing short bowel syndrome following repeated bowel resections [9].

Management of patients with Peutz-Jeghers syndrome requires a lifelong, multidisciplinary approach. It includes monitoring, prevention of disease manifestations and treatment of complications [10]. Current recommendations include basic FOGD screening from the age of 12, to be repeated every year if polyps are present, and every 2 to 3 years in their absence. Screening colonoscopy is performed from the age of 12, or earlier if the patient is symptomatic, and is repeated every year if polyps are present, and at intervals of 1 to 3 years in their absence. Magnetic resonance cholangiopancreatography (MRCP) and/or endoscopic ultrasound is recommended from the age of 25 to 30, and repeated every 1 to 2 years. Clinical breast examination is performed every 6 months from the age of 25. Mammography is recommended from the age of 25, and an annual Pap smear and transvaginal ultrasound from the age of 18. For men, an annual testicular examination is recommended, and ultrasound is indicated if the patient is symptomatic from birth [1-11].

CONCLUSION

Peutz-Jeghers syndrome is an autosomal dominant genetic disorder characterized by the development of hamartomatous polyps in the gastrointestinal tract, associated with mucocutaneous pigmentation. Peutz-Jeghers syndrome is a serious, life-threatening condition with a significantly increased lifetime risk of cancer. Care of patients with Peutz-Jeghers syndrome requires a lifelong multidisciplinary approach to cancer surveillance, given the significantly elevated cancer risk.

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