Case Report

Peutz-Jeguers syndrome: case report and literature review

Juvenal da Rocha Torres Neto[1], Rodrigo R. Santiago[2], Ana Carolina Lisboa Prudente[2], Dan Rodrigues Mariano[2], Felipe Augusto do Prado Torres[3] , Júlio Augusto do Prado Torres[3], Fernanda Mendonça Ramos[3], Raquel Matos de Santana[3]

[1]Doctor and Professor and Head of Coloproctology Service at the Universidade Federal de Sergipe (UFS) – Aracaju (SE), Brazil. [2]Resident in Coloproctology at UFS – Aracaju (SE), Brazil. [3]Residents in Coloproctology at UFS – Aracaju (SE), Brazil. [3]Academicians, Medical Sciences at UFS – Aracaju (SE), Brazil.

Torres Neto JR , Santiago RR, Prudente ACL, Mariano DR, Torres FAP, Torres JAP, Ramos FM, Santana RM. Peutz-Jeguers syndrome: case report and literature review. **J Coloproctol**, 2012;32(1):76-9.

ABSTRACT: The Peutz-Jeghers syndrome is a rare disease characterized by the presence of mucocutaneous melanic pigmentation of the lips, oral mucosa and perioral region, associated with hamartomatous intestinal polyposis. Malignization of the polyps and association with other types of cancer are also usual. Case report: 32-year-old patient, female, white, who had an intestinal occlusion by invagination, discovered during laparotomy, when an intestinal tumor was found as well. The material was sent to anotomopathological analysis. However, the results did not allow to identify the tumor nature due to tumor necrosis. Then, the patient was sent to our service because of the intestinal polyps, and during the interview, the characteristic melanic pigmentation was observed. Videocolonoscopy was performed, with excision of two rectal polyps, identified in the anatomopathological exam as hamartomatous polyps. The patient reported anal imperforation at birth, just like her brother. He had unexplained death. The authors found no correlation of the Peutz-Jeghers syndrome with anal imperforation in the literature and asked the patient if her brother also had the syndrome.

Keywords: Peutz-Jeghers syndrome; polyposis; hamartomatous intestinal; anus, imperforate; hyperpigmentation.

RESUMO: A síndrome de Peutz-Jeghers é uma doença rara que tem como características a pigmentação melânica mucocutânea de lábios, regiões perioral e de mucosa bucal associada à polipose hamartomatosa do trato intestinal, com possibilidade de malignização dos pólipos digestivos e associação com outros tipos de câncer. Relato de Caso: Paciente de 32 anos, de gênero feminino, branca, apresentou um quadro de oclusão intestinal por uma invaginação, evidenciada durante laparotomia exploradora, constatando-se, ainda, a presença de uma tumoração intestinal. O material foi encaminhado para exame anatomopatológico; porém, foi inconclusivo para a natureza da tumoração em decorrência da necrose. Em função do pólipo intestinal, a paciente foi encaminhada ao nosso serviço, quando percebemos a presença de pigmentação melânica característica. Realizamos videocolonoscopia com achado de dois pólipos de reto cujo resultado do exame anatomopatológico foi de pólipo hamartomatoso. A paciente relatou ainda ter nascido com imperfuração anal e possuir irmão que também nascera com a mesma imperfuração e que evoluiu para óbito não esclarecido. Não encontramos relato na literatura de associação da síndrome de Peutz-Jeghers com imperfuração anal e interrogamos se o irmão teria também a síndrome.

Palavras-chave: síndrome de Peutz-Jeghers; polipose; hamartomatosa intestinal; ânus imperfurado; hiperpigmentação.

INTRODUCTION

Peutz-Jeghers syndrome (SPJ) is a rare disease[1-3], characterized by presence of mucocutaneous melanic pigmentation of the lips, oral mucosa and perioral region[4,5], associated with hamartomatous polyposis in all gastrointestinal tract, affecting the small bowel with more frequency. The literature has reports of extraintestinal polyps associated with this syndrome[3]. In addition, the most usual complications that have been observed are: invagination of intestinal wall, bowel obstruction and bleeding[4,6]. The literature also shows cases associating this syndrome with gynecological carcinomatous manifestations in the digestive tract[1,2,7] and lungs[2,7], despite the rare polyp malignization.

Study carried out at the Coloproctology Service at the Hospital Universitário de Sergipe (HU-SE) and Torres Centro Médico.
Funding source: none.
Conflict of interests: nothing to declare.

Submitted on: 19/06/2009
Approved on: 24/07/2009

On the occasion of the first description of this malformation, laparotomy and surgical interventions were the standard treatment methods; however, new methods have also proved to be efficient and less invasive, with patients' morbidity reduction, such as upper endoscopy and videocolonoscopy, aiming at polypectomy and, more recently, *double*-balloon enteroscopy. Despite the variety of techniques, all therapeutic methods aim at the excision fo polyps and resolving complications that may occur[4,8,9].

CASE REPORT

DACS, 32-year-old female patient, white, presented in 2007 a condition of intestinal occlusion. During laparotomy, intestinal invagination was observed, with loop necrosis and a polypoid tumor also necrosed. The anatomopathological analysis. However, the results could not identify the polyp histology due to the presence of necrosis. The patient was sent to the coloproctologist to be submitted to colonoscopy. During the interview, the characteristic melanic pigmentation was observed (Figures 1 to 3), suggestive of SPJ. The patient also reported anal imperforation at birth, just like her brother, who had died when he was a child. Videocolonoscopy was performed in September 2007, with the excision of two rectal polyps, identified in the anatomopathological exam as hamartomatous polyps (Figure 4). Some intestinal polyps were also found during endoscopy. In addition, we verified the intestinal flow, which did not show any other lesion (Figure 5). Now, the patient has no symptoms and she is receiving follow-up care by the Coloproctology Service.

DISCUSSION

SPJ syndrome is an autosomal dominant disorder, described for the first time in 1896 by Jonathan Hutchinson[10], when he observed the classic perioral pigmentation in female twins, without knowing the etiology, but already suspecting of a rare disease. In 1919, when one of the twins died of intestinal intussusception, his suspicion of the syndrome was confirmed. Peutz, in 1921[11,12], when studying the entity in a family, related mucocutaneous melanic spots with intestinal polyposis to the syndrome. Finally, in 1949, Jeghers et al.[13] complemented the early description, based on a study that confirmed its autosomal dominant and, therefore, hereditary characteristic.

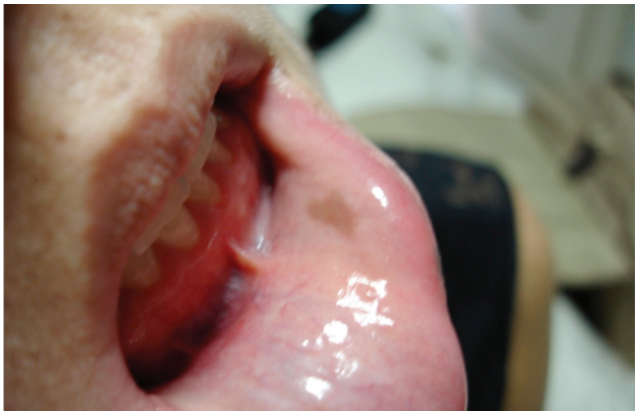

Figure 1. *Characteristic mucocutaneous melanic pigmentation.*

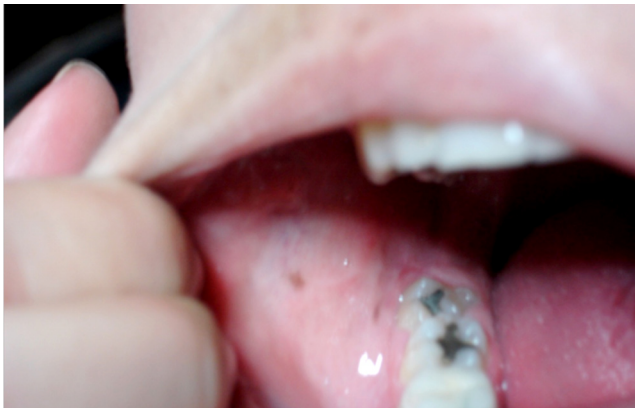

Figure 2. *Characteristic mucocutaneous melanic pigmentation.*

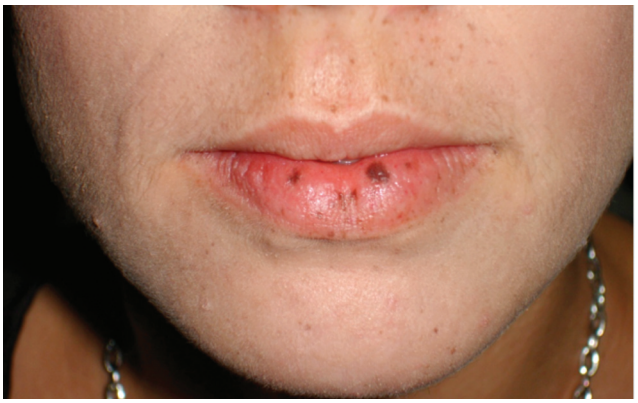

Figure 3. *Characteristic mucocutaneous melanic pigmentation.*

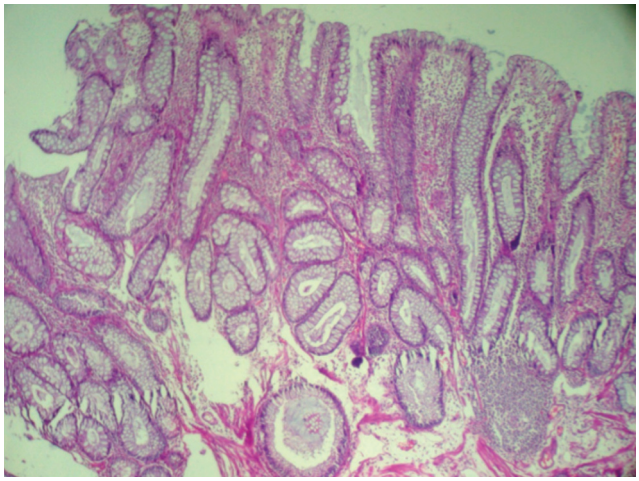

Figure 4. *Arboriform aspect of the hamartomatous polyp.*

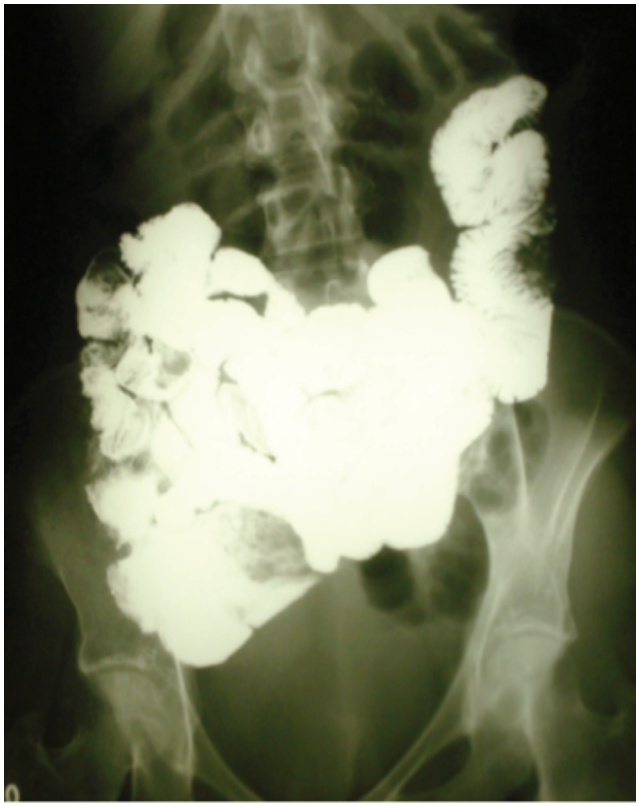

Figure 5. *Intestinal flow, not showing other lesion.*

The clinical entity has global prevalence of around 1 case in 200,000 people, with variable manifestation; some may present melanic pigmentation only, while others may show concomitant polyps. The first polyps appear in around 60% of the cases after 11 years of age, although the literature reports manifestations beginning at birth. There is no relation with ethnic group, but the disease affects more females[14-16].

As mentioned above, the great number of hamartomatous polyps across the entire gastrointestinal tract and melanic pigmentation characterize the syndrome, which are essential for the diagnosis. These polyps are the result of an excessive growth of native cells of the tissue where they occur and are characterized by their arboriform aspect at the anatomopathological exam, due to complex protrusions of the smooth muscle tissue in the lamina propria involved by hyperplastic glands. The mucocutaneous pigmentation usually affects the lips, the perioral region and the oral mucosa, but it can also occur on the eyelids, fingers, hands, back, sole of the foot and, less frequently, on lumbosacral and perineal regions. Such pigmentation is the most important aspect for the clinical diagnosis[4,5,13,15].

Hamartomatous polyposis syndromes represent a small, but important, number of hereditary intestinal syndromes predisposed to cancer, mainly in colon and pancreas. The literature also has reports of malignization of polyps and other carcinomatous presentations related to the syndrome, such as gynecological, testicle, breast, lung and bile duct manifestations. In this study, two hamartomatous polyps were found in the rectum, but without suspicion of cancer[1,2,7,14-16].

Usual complications of the SPJ are tract obstruction, loop invagination and gastrointestinal wall bleeding. Tract obstruction occurs especially due to polyps in the small bowel and may result, due to poor absorption of nutrients, in anemia or poor protein nutrition. Extensive excision of polyps may also cause difficult absorption of nutrients due to intestinal wall lesion. As described in this case, invagination of intestinal wall may lead to loop necrosis caused by ischemic lesions[4].

The treatment of this clinical entity typically uses laparotomy and enterotomy, and, more recently, the association of endoscopic methods – high endoscopy and videocolonoscopy – less invasive techniques aiming at polyp excision and intestinal clearing, which also prevent resection of bowel loops. The literature also shows a new non-surgical therapeutic method: double-balloon enteroscopy.

This is a therapeutic and diagnostic method, a safe and reliable technique that can revolutionize the polyp excision procedure[8].

This study points out the simultaneous occurrence of anal imperforation with the syndrome, once the literature has no cases reporting such occurrence related to the PJS. Anal imperforation, for occurring in both the studied patient and her brother, shows the genetic and hereditary aspect of this disorder and, as the PJS is also hereditary, it raises the suspicion of a correlation. Due to the early death of the patient's brother, it was not possible to diagnose the PJS in him and, consequently, associate the information.

This study reported a case of PJS, pointing out the fact that, when diagnosing small bowel polyps or melanic pigmentation, special attention should be dedicated to the simultaneous occurrence of the syndrome, whose report is important due to its low incidence.

REFERENCES

1. Erdas E, Licheri S, Pisano G, Pomata M, Daniele GM. Peutz-Jeghers syndrome: an account of 3 cases in the same family and a review of the literature. Chir Ital 2005;57(4):425-36.
2. Capasso L, Lombari P, Scarano MI, Izzo P, D'Ambrosio R, Iannucci A, et al. Peutz-Jeghers syndrome: case report and update on diagnosis and treatment. Minerva Chir 2001;56(6):643-7.
3. Vogel T, Schumacher V, Saleh A, Trojan J, Möslein G. Extraintestinal polyps in Peutz-Jeghers syndrome: presentation of four cases and review of the literature. Int J Colorectal Dis 2000;15(2):118-23.
4. Gama-Rodrigues J, Silva JH, Aisaka AA, Jureidini R, Falci RJr, Maluf FF, et al. Intestinal intussusception and occlusion caused by small bowel polyps in the Peutz- Jeghers syndrome. Management by combined intraoperative enteroscopy and ressection through minimal enterostomy: Case Report. Rev Hosp Clin 2000;55(6):219-24.
5. Westerman AM, Wilson JH. Peutz-Jeghers syndrome: risks of a hereditary condition. Scand J Gastroenterol Suppl 1999;230:64-70.
6. Rubio Tapia A, Ramírez Arias F, Angeles Angeles A, Uscanga L. Peutz-Jeghers syndrome. Rev Gastroenterol Mex 2005;70(3):291-5.
7. Miyaki M. Peutz-Jeghers syndrome. Nippon Rinsho 2000;58(4):1400-4.
8. Plum N, May AD, Manner H, Ell C. Peutz-jeghers syndrome: endoscopic detection and treatment of small bowel polyps by double-balloon enteroscopy. Z Gastroenterol 2007;45(10):1049-55.
9. Edwards DP, Khosraviani K, Stafferton R, Phillips RK. Long-term results of polyp clearance by intraoperative enteroscopy in the Peutz-Jeghers syndrome. Dis Colon Rectum 2003;46(1):48-50.
10. Hutchinson J, Pigmentatons of lips and mouth. Arch Surg 1896;290.
11. Webber FP. Patches of deep pigmentation of the oral mucous membrane not connected with Addinson's disease. Quart J Med 1919;12:404-8.
12. Peutz JLA. Over Een Zeer Merkwaardige, Gecombineerde Familiaire polyposis Van De Slijmvliesen Van De Neuskeelholte En Gepaard Met Eigenaardig Pigmentaties Van Huit-En Slijmvliesen. Nederl M Geneesk 1921;10:134-46.
13. Jeghers H, Mckusick VA, Katz KH. Generalized intestinal polyposis and melanin spots of the oral mucosa, lips and digits. New Eng J Med 1949;241:993-1005.
14. Costa JHG, Azevedo IF, França MAV, Maia HP, Gama RC, Primo CC, et al. Síndrome de Peutz-Jeghers: apresentação de um caso. Rev bras Coloproct 1987;7(4):153-8.
15. Mallman ACM, Diniz FF, Carvalho LP, Balbinot GL, Rolim JHH, Matos TC. Síndrome de Peutz-Jeghers com malignização: relato de caso. Rev Bras Cir 1995;15(4):182-4.
16. Schreibman IR, Baker M, Amos C, McGarrity TJ. The Hamartomatous Polyposis Syndromes: A clinical and molecular review. Am J Gastroenterol 2005;100(2):476-90.

Correspondence to:
Juvenal da Rocha Torres Neto
Rua Ananias Azevedo, 100, apto. 902 – Praia 13 de Julho
CEP: 49020-080 – Aracaju (SE), Brazil
E-mail: jtorres@infonet.com.br