Surgical aspects of intussusception secondary to Peutz-Jeghers syndrome

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Resumen

El síndrome de Peutz-Jeghers es una condición familiar caracterizada por la presencia de manchas cutáneas hiperpigmentadas y poliposis intestinal. La invaginación intestinal es la complicación abdominal más frecuente, pero es muy rara. Este es el segundo reporte que se comunica en nuestro país. En este artículo presentamos cuatro pacientes que sufrieron el síndrome, que se complicó con invaginación intestinal. Todas fueron del sexo femenino e intervenidas quirúrgicamente. La invaginación estuvo localizada en el ileon y el colon. No hay más de 20 casos de esta asociación referidos en la literatura mundial. Los niños que sufren el síndrome de Peutz-Jeghers tienen altas posibilidades de sufrir una invaginación ileo-ileoal o jejuno-ileoal, que con mucha frecuencia requerirá de un procedimiento quirúrgico especial.

Palabras clave: invaginación, niños, síndrome de Peutz-Jeghers, poliposis intestinal, abdomen agudo.

Summary

Peutz-Jeghers syndrome (PJS) is a familial condition characterized by the presence of pigmented mucocutaneous spots and intestinal polyposis. Intussusception is the most frequent abdominal complication, but it is very rare. In our country, this is the second known report. In this article we present four cases of PJS associated with intussusception. All the patients were female and all underwent a surgical procedure. The intussusception was located in the ileum and colon. There are no more than 20 reported cases in the world where these two entities are associated. Children with PJS have a high risk of suffering from ileo-ileoal or jejuno-ileoal intussusception that will frequently require a special surgical procedure.

Key words: intussusception, children, Peutz-Jeghers syndrome, intestinal polyposis, acute abdomen.

Introduction

Intussusception is a disease of unknown origin, is more frequent between 3 to 9 months of age, in a seasonal manner, predominantly in colder months, and is accompanied by an intestinal obstruction syndrome which has as a main clinical component abdominal pain and the presence of muco-hematic evacuations that resemble “currant jelly.” It is one of the diseases that, according to the masterly and almost unchanged description made by Potts almost 40 years ago,1 due to its very predictable and constant clinical expression, clinical diagnosis must be made with high precision, even by telephone. In general, it appears in eutrophic children, but this does not mean that undernourished children are exempt.2 In the physical exam in the first hours of initiation of the disease, the presence of an elongated sausage-shaped tumor and displacement of the right colon and terminal ileum towards the origin of the traction are well known, being known as the Dance sign.3 Regarding frequency, this shows seasonal elevations and varies from region to region. In our country it depends on the level of attention exhibited by the particular hospital, but we see an average of 32 cases per year.

Peutz-Jeghers syndrome (PJS) is a condition with a well-known hereditary component and which is characteristic because children who suffer from it have gastrointestinal tract polyposis and “café au lait” spots distributed in skin and
mucosa. Until 1983, 124 cases had been reported in patients < 17 years of age. The association between intussusception and this syndrome is a very predictable but unusual complication, and to date no more than 15 pediatric cases have been reported in the literature worldwide. In our country there is only one previous report.

The objective of this report is to analyze the clinical course for four patients whom we treated, to show the casuistry of more recollected cases in the world, and as a main objective, to emphasize the surgical findings.

Clinical cases

This is a retrospective study that consisted of the analysis of 256 cases of intussusception in children <15 years of age, observed during a period of 8 years. Because there are relatively few cases, two were reported previously in which there was the association of intussusception and PJS. We will describe each of the patients.

Case 1

A 15-year-old female patient arrived at the emergency room with sudden onset abdominal pain accompanied by cutaneous paleness. She was immediately sent from her school to our unit. According to her familial history, her maternal great grandparents and her mother had PJS.

During physical exam the patient was pale and diaphoretic but conscious. She had about 20 hyperchromatic spots in the mucosa of the mouth and lips, and their diameter oscillated between 3 and 5 mm (Figure 1). The abdomen was flat, soft, compressible and with pain at superficial and deep palpation. No masses were palpated. An abdominal ultrasound was requested reporting the classic image of intestinal intussusception for which the patient was sent to the operating room where an exploratory laparotomy was done. Scarce fetid hemopurulent liquid was found. The viscera were exposed and there was a segment of necrotic small bowel and a jejuno-ileal intussusception. It was necessary to remove 20 cm of ileum and construct a double-barrel ileostomy.

The patient’s evolution was satisfactory and she was released to her home 6 days postoperatively. She was re-hospitalized 6 weeks later for restoration of intestinal permeability, a surgery that was performed without problem. The pathologist reported that in the interior of the intussusception there were polyps to which the complication was attributed.

Case 2

An 11-year-old male patient was referred from another hospital unit to which he had entered with a history of being operated on 72 h before because of a manually corrected intussusception. Upon his arrival, we found hyperpigmented spots in the face, lips and oral mucosa. He was conscious, without abnormal movements, and well hydrated. The abdomen was distended and tense, with signs of peritoneal irritation. Surgical scar was normal. There was no peristalsis. For these reasons, a re-intussusception was suspected for which, after the corresponding laboratory and imaging studies were performed (Figure 2), he was again taken to the operating room, finding the intestine folded within itself. The intussusception was ileocecal and was so tight that it required an en bloc extirpation of the affected segment and construction of an enterostomy. The patient’s post-surgery evolution was favorable; however, at the third day after surgery he presented protrusion of various polyps through the stoma, the reason for which he was re-explored, finding an intestinal intussusception for the third time that was corrected manually.

Figure 1. Plain radiology study of the abdomen in a horizontal position that shows segmental dilation of the small intestine and some disseminated bubbles in the remainder of the abdomen that suggest peritonitis.
Twelve weeks later he was admitted to give continuity to the exposed intestine, an operation that was accomplished without complications. Pathology reported intestinal necrosis of 40 cm in length, as well as a polyp, as the cause of the intussusception.

Case 3

A 5-year-old female from Mexico City was admitted to the emergency room with a clinical picture of 10 h and with a history that on a previous occasion she had a similar crisis characterized by intermittent abdominal pain and distension, accompanied by biliary vomiting. The presence of hyperchromatic spots in lips and oral mucosa were noted. The abdomen was dilated and painful. There was struggle peristalsis and a 12-cm-long tumor was palpated in the right hemi-abdomen. The radiology study showed an image suggesting mechanic intestinal blockage (Figure 3).

In the operating room, an ileo-colic intussusception was found that produced necrosis of an intestinal segment, for which it was necessary to remove the affected segment en bloc. A termino-terminal anastomosis was performed with a satisfactory subsequent evolution. The histological study reported hamartomatous polyps.

Case 4

An 8-year-old female was admitted to our hospital unit with a history that indicated her illness began many weeks earlier, with vomiting, abdominal distension and pain. The discomfort was so intense that the patient required hospitalization on many previous occasions. The girl lived in Mexico City but was originally from Veracruz. She was an adopted daughter.

During the physical exam negroid features and hyperpigmented spots on the lips and oral mucosa were found. She was hospitalized with the diagnosis of PJS and intestinal subocclusion. After 3 days she abruptly presented exacerbation of the symptoms, and an intestinal intussusception was suspected. After demonstration by the clinical exam and the imaging studies, she was sent to the operating room where a triple intussusception was found: two were jejuno-jejunal and one jejuno-ileal. The first two were 45 and 60 cm from the angle of Treitz and the other one 20 cm from the ileo-cecal valve. In the latter, removal of the affected segment was carried out with a termino-terminal anastomosis. The other two were corrected without difficult maneuvers. A longitudinal jejunotomy was performed through which four polyps were extracted of approximately 6 cm each, and it was repaired in a transverse manner. Fifteen days after she was released from the hospital, she was re-admitted with a similar symptomatic picture for which she was re-operated on with a diagnosis of intestinal re-intussusception. On this second occasion a tight ileo-ileal...
intussusception was found for which it was necessary to carry out a resection and termino-terminal anastomosis. The patient was released from the hospital in good conditions. Histological study reported the presence of an acute inflammatory infiltrate and hamartomatous polyps (Figure 4).

Discussion

There are many conditions in which a disease with a genetic or systemic basis is a determinant factor for the presence of acute and surgically natured complications such as in the case of hemophilia, Henoch-Schönlein purpura, and PJS. These three can determine the presence of abdominal pain as an isolated symptom as a consequence of the formation of hematomas in the intestinal wall and mesentery, or through the introduction of a portion of the intestine within itself, respectively. The latter is the reason for this presentation. It is the best known, by virtue of having recollected four unusual cases, and because it is the disease that we have seen most frequently in our practice.

The syndrome was described in 1942 by Peutz, a German physician. The original report describes seven children who constituted the problem family: five had many dark spots on the face, lips and oral mucosa; three of them, one girl and two boys, had intermittent abdominal pain crises and rectal bleeding. There was an ileal intussusception due to polyps in the small and large intestine in two cases, and in one of the surgical specimens malignant degeneration was demonstrated. The father of this family had no symptoms but had hyperchromatic spots on the oral mucosa. Two sisters who had similar pigmentations died due to intestinal obstruction at ages 11 and 20, respectively. Analysis of the disease and the conclusions to which Peutz came to led him to define the disease as an autosomal dominant condition. The German family originally described was seen by Wilk for almost 30 years, and he published his observations in 1950. Today we know the gene responsible for PJS, and it codifies the serine-threonine kinase LKB1 or STK11. Seeing the fundamental genetic defect of the disease, a mutation in the PJS gene has been found. The mutation consists of the insertion of a T in hexon 1 (codon 66) of the LKB1 gene, which in turn originates the codon (162) of the hexon 4 (4).

The diagnosis of the syndrome is easy because the stigmas are in general evident, and identifying it is a mere procedure, the hyperpigmented spots lead us to the diagnosis. Those distinctive clinical macules usually measure between 1 and 5 mm are distributed around the nostrils, eyes and mouth, and in the peri-anal region. They can also be found on the toes and fingers, and less frequently on oral and rectal mucosa. The total-depth biopsy of the spots is characterized by an increase in the number of melanocytes in the dermo-epidermic union and the melanine in the basal cells.

In relation to the polyposis, generally there are hundreds that can be counted, and have a main distribution in the interior of the small intestine, followed by the large intestine and the stomach. The ones found in the small and large intestine have a tendency to be pediculated, and the ones in the stomach to be broad-based and histologically they are hamartomatous. An additional peculiarity of the syndrome is that it is usually accompanied by malignant neoplasms. In his study, Tovar et al.,11 until 1983, observed that among 70 children who had PJS, 7.14% suffered from some type of cancer, with a frequency of gonadal tumors.

It is not known how many cases are reported in the literature because there are no reports that are exclusively about this theme. However, to the point where we could investigate, there are only seven children < 15 years of age.11-13 Of these, none was < 4 years, and taking as a reference point the number of cases of intussusception that are seen each year in a hospital such as ours, we have calculated that PJS appears in 1.4% of the total children who suffer from the disease.

The association between the syndrome and intussusception is a totally predictable association because of the fact that the polyps at the edge of their nature can become the factor that produces the introduction of the intestine within itself. We do not know why it appears in older children, but probably the dimension and weight of the polyps, as well as the standing position, are factors that are an influence so the larger polyps can lead an intestinal segment to the interior of the immediate distal segment. With respect to the differential diagnosis, there are other hereditary diseases that also manifest as hyperchromatic maculae and that are accompanied, such as in the case of multiple familial neurofibromatosis, by neurofibromas on the trunk and extremities. Other conditions that

Figure 4. Hamartomatous polyp of the small intestine. At low magnification the characteristic architectural arrangement is visible, with a mucosa composed of glands supported by interdigitating muscular bundles.
can simulate PJS are familial polyposis, Gardner syndrome and juvenile polyposis.12

The presence of polyps in PJS is not an indication for surgical intervention. These measures are necessary when there is intestinal obstruction, prolapse of the polyps, coexistence of a neoplasm or considerable gastrointestinal hemorrhage. Nonetheless, it is important to mention that the quantity of existing polyps in the interior, particularly of the small intestine, is usually so large that the majority of children who have the disease have recurrent crisis of intussusception that disappear as fast as they appear. It is probable that a large number of polyps present in a short segment of the intestine makes an intussusception repeat itself two or more times, and the child may need multiple surgeries.

Once a laparotomy has been performed for any of the above-mentioned reasons, multiple enterostomies and individualized removal of as many possible units of polyps is recommended.15 Under other circumstances, it is sometimes necessary to perform an en bloc removal of an ample length of intestinal segment, but when it is located near the Vater ampulla, these maneuvers are difficult.11 Retrograde and antegrade endoscopy, trans-operative or not, can be very useful.

When there is coexistence of PJS and intussusception, the surgical procedure that we use depends on various circumstances. We have considered that in children whose evolution is < 24 h and who have segmental necrosis or an impossibility to correct it manually, en bloc removal of the affected intestinal segment and a termino-terminal anastomosis could be the procedure of choice, despite the pre-anastomotic high-pressure zone that the polyps generate. When there are signs suggesting regional ischemia and peritonitis, we suggest that an enterostomy be carried out preferentially. Once the derivation is planned, multiple enterostomies and trans-operative endoscopy must be utilized to extract the greatest number of polyps as possible.15 When the disease produces recurrent intussusceptions, it is recommended in the first surgeries to additionally carry out the Noble procedure.16

With relation to the management as outpatients, aside from having periodic control of the polyps’ behavior, it is necessary to take into account that these types of patients have a known risk for malignant neoplasms, particularly ovarian or testicular tumors, for which there must be a strict regimen of oncologic surveillance.

References