J Surg Med. 2020;4(10):906-908.
DOI: 10.28982/josam.729936

Case report
Olgu sunumu

Journal of Surgery and Medicine
e-ISSN: 2602-2079

Infantile juvenile polyposis syndrome: A rare cause of protein-losing enteropathy

İnfantil juvenil poliposis sendromu: Protein kaybettiren enteropatinin nadir bir nedeni

Didem Gülcü Taşkın [1], Ömer Faruk Beser [2], Nuray Kepil [3], Sibel Erdamar Çetin [3], Fügen Çullu Çokuğraş [2], Tülay Erkan [2], Tufan Kutlu [2]

[1] Department of Pediatric Gastroenterology, Adana City Training and Research Hospital, Adana, Turkey
[2] Pediatric Gastroenterology, Istanbul University, Cerrahpasa Medical Faculty, İstanbul, Turkey
[3] Department of Pathology, Istanbul University, Cerrahpasa Medical Faculty, İstanbul, Turkey

ORCID ID of the author(s)

DGT: 0000-0002-2746-3799
OFB: 0000-0003-1927-7256
NK: 0000-0001-5494-6422
SEC: 0000-0001-7470-8835
FCC: 0000-0003-0886-1422
TE: 0000-0002-8924-2799
TK: 0000-0001-8396-4048

Corresponding author / Sorumlu yazar:
Didem Gülcü Taşkın
Address / Adres: Kışla Mahallesi, Dr. Mithat Özsan Bulvarı, 4522. Sokak No:1, Yüreğir, Adana, Türkiye
E-mail: dgulcu@gmail.com

Informed Consent: The authors stated that the written consent was obtained from the parents of the patient presented with images in the study.
Hasta Onamı: Yazarlar çalışmada görüntüleri ile sunulan hastanın ebevynlerinden yazılı onam alındığını ifade etmiştir.

Conflict of Interest: No conflict of interest was declared by the authors.
Çıkar Çatışması: Yazarlar çıkar çatışması bildirmemişlerdir.

Financial Disclosure: The authors declared that this study has received no financial support.
Finansal Destek: Yazarlar bu çalışma için finansal destek almadıklarını beyan etmişlerdir.

Published: 10/28/2020
Yayın Tarihi: 28.10.2020

Abstract

Juvenile Polyposis Syndrome (JPS) is a rare autosomal dominant hereditary syndrome affecting 1:100000-160000 individuals. JPS most presents with rectal bleeding, anemia, abdominal pain, obstruction and rarely with rectal prolapsus of the polyp. In this case, we diagnosed Infantile Juvenile Polyposis Syndrome due to protein losing enteropathy, rectal bleeding and extraintestinal manifestations of the syndrome. A 2-year-old male infant was referred to the hospital due to complaints of painless rectal bleeding and rectal prolapsus of the polyp which occurred at 15 months of age. Pathological examination revealed that it was a juvenile polyp. After a short while, the patient was hospitalized due to rectal bleeding, paleness, swelling in both legs and periorbital edema. Physical examination of the patient revealed +3 pretibial, scrotal, periorbital edema, clubbing and pale appearance. The patient had macrocephaly, hypotonicity and neuromotor retardation. Laboratory test results revealed low immunoglobulin levels, hypoalbuminemia, anemia, and electrolyte imbalance. We diagnosed the case with JPS due to protein losing enteropathy with extraintestinal manifestations of the syndrome. Gastroscopic and colonoscopic examinations revealed multiple polyps through the antrum and colon. JPS diagnosis is based on the detection of polyps which are histopathologically defined as juvenile polyps. One of the most common causes of painless, rectal bleeding in children are colorectal polyps. We wanted to emphasize that the sporadic juvenile polyp diagnosis should be made by pathological examination of polypectomy material and clinical exclusion of JPS. In our patient, there were extraintestinal system manifestations such as macrocephaly, congenital heart disease and clubbing, accompanied with protein-losing enteropathy. Awareness of these clinical findings is necessary for the differential diagnosis of protein-losing enteropathy and polyposis syndrome. We would also like to draw attention to the importance of a multidisciplinary approach, early recognition of the syndrome and appropriate referral of the patient.

Keywords: Rectal bleeding, Infantile juvenile polyposis syndrome, Protein losing enteropathy

Öz

Juvenil Polipozis Sendromu (JPS), çok nadir görülen, otozomal dominant geçişli, 1/100000 ile 1/160000 insanı etkileyen, herediter bir sendromdur. JPS sıklıkla rektal kanama, anemi, karın ağrısı, tıkanma ve nadiren polipin rektal prolapsusu ile bulgu verir. Bu olgu ile protein kaybettiren enteropati, rektal kanama ve ekstraintestinal manifestasyonları olan hastamıza JPS tanısı koyduk. 2 yaş erkek hasta ağrısız rektal kanama nedeniyle hastaneye başvurdu. 15 aylık iken rektal polip prolapsusu olmuş ve bu polipin patolojik değerlendirmesi juvenil polip ile uyumlu imiş. Çok kısa süre sonra hasta rektal kanama, solukluk, ayaklarda şişlik ve göz çevresi ödem ile başvurdu. Fizik muayenesinde pretibial +3 ödem, skrotal ödem, göz çevresinde ödem, parmaklarda çomaklaşma ve soluk görünümü mevcut idi. Makrosefali, hipotoni ve nöromotor retardasyonu vardı. Laboratuvar testlerinde: immunglobulinler ve albumin düşük, anemisi mevcut ve elektrolit dengesizliği vardı. JPS tanısı protein kaybettiren enteropatiye eşlik eden ekstraintestinal system bulgularının varlığı ile konuldu. Yapılan gastroskopik ve kolonoskopik incelemesinde antrumdan başlayarak tüm kolon mukozası boyunca yaygın polipler mevcut idi. Histopatolojik incelemesi juvenil polip ile uyumlu idi. Çocuklarda ağrısız rektal kanamaların en sık nedeni kolorektal poliplerdir. Sporadik juvenil polip tanısı, patolojik olarak polipektomi materyalinin incelenmesi ve klinik olarak JPS'nun dışlanması ile konulmalıdır, bunu vurgulamak istedik. Bizim hastamızdaki gibi makrosefali, konjenital kalp hastalığı ve çomak parmak gibi ekstraintestinal bulguların farkındalığı ve protein kaybettiren enteropati ayırıcı tanısında polipozis sendromunun akılda tutulması gerektiğini vurgulamak istedik. Multidisipliner yaklaşım ile erken tanı prognozu iyileştirebilmektedir.

Anahtar kelimeler: Rektal kanama, İnfantil juvenil polipozis sendromu, Protein kaybettiren enteropati

Introduction

Juvenile Polyposis Syndrome (JPS) is a rare autosomal dominant hereditary syndrome affecting 1:100000-160000 individuals [1]. It is diagnosed according to the following criteria: a) At least 3 polyps detected on colonoscopy, b) Juvenile polyps in the entire digestive tract (stomach, small bowel, etc.), c) In cases of positive family history of the disease, any number of juvenile polyps [2]. Three subtypes of Juvenile Polyposis Syndrome were defined: 1. Juvenile polyposis coli (JPC), 2. Generalized juvenile polyposis (GJP), and 3. Juvenile polyposis of infancy (JPI) [3]. The first two subtypes are distinguished by the location and extent of polyps along the gastrointestinal tract with polyps located in the colon only in JPC and in the colon and upper gastrointestinal tract in GJP [4].

How to cite / Atıf için: Taşkın DG, Beser ÖF, Kepil N, Çetin SE, Çokuğraş KÇ, Erkan T, Kutlu T. Infantile juvenile polyposis syndrome: A rare cause of protein-losing enteropathy. J Surg Med. 2020;4(10):906-908.

JPS most presents with rectal bleeding, anemia, abdominal pain, obstruction and rarely, with rectal prolapsus of the polyp [5]. It is estimated that over 20% patients with JPS have macrocephaly, congenital heart diseases and urogenital system anomalies [6]. Germline mutations which causie JPS in 40–60% of patients have been identified in bone morphogenetic protein receptor type 1A (BMPR1A) and SMAD4 [7,8].

In this case, we diagnosed as Infantile Juvenile Polyposis Syndrome based on protein-losing enteropathy, rectal bleeding and the extraintestinal manifestations of the syndrome.

Case presentation

A 2-year-old male infant was referred to the hospital with complaints of painless rectal bleeding and rectal prolapsus of a polyp, which occurred when was 15 months old. Pathological examination revealed that it was a juvenile polyp. After a short while, the patient was hospitalized due to rectal bleeding, paleness, swelling in both legs and periorbital edema. Physical examination of the patient revealed +3 pretibial, scrotal, periorbital edema, clubbing and pale appearance. The patient had macrocephaly, hypotonicity and neuromotor retardation. Liver and spleen were subcostally palpable at 4 and 2 cm, respectively. We observed the prolapsus of the polyp during rectal examination (Figure 1).

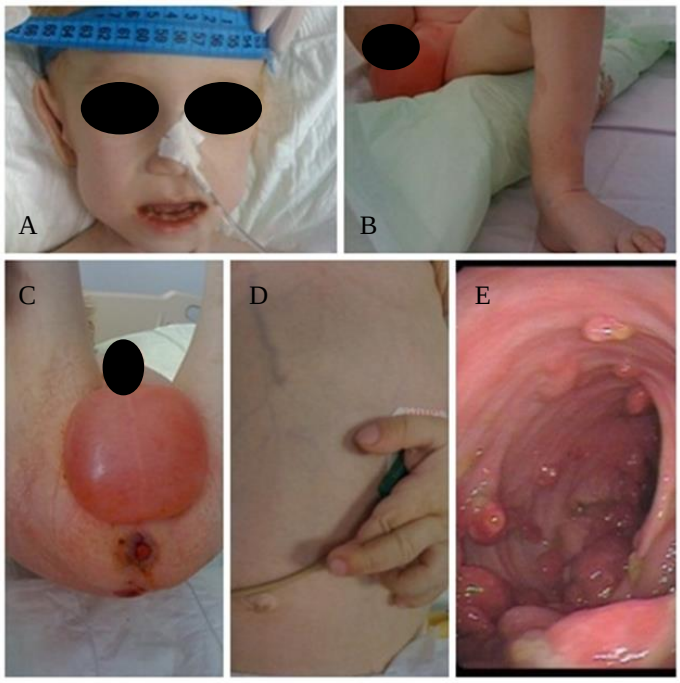

Figure 1: Physical examination of the patient. (A: Macrocephaly, B: Pretibial edema, C: Prolapsus of the polyp and scrotal edema during rectal examination, D: Clubbing, E: Macroscopic appearance of the polyps during colonoscopy)

Cardiovascular examination revealed a 1/6 heart murmur. Laboratory test results were as follows: Immunoglobulin (Ig) G: 266 IU/L, Ig A: 69 IU/ml, Ig M: 43 IU/ml, Ig E: 16 IU/ml, Total protein: 2.5 g/dl, Albumin: 1.4 g/dl, Hemoglobin: 6 g/dl, Hematocrit: 18%, Blood Sodium: 125 mmol/L, Potassium: 2.5 mmol/L. Echocardiography reported mitral valve prolapse and mild mitral valve deficiency. Urinary system ultrasonography and cranial magnetic resonance imaging were normal, along with the past medical history of the patient. Two grandfathers had died due to lung cancer and colon cancer. We considered polyposis syndrome in the differential diagnosis of protein-losing enteropathy and diagnosed the case with JPS based on the existence of protein-losing enteropathy with extraintestinal manifestations of the syndrome. Infantile juvenile polyposis syndrome is considered because the symptoms occurred during the first 2 years of age. Gastroscopic examination through the antrum revealed multiple (more than 10), spherical and lobulated, 5-20 cm-sized, pedunculated/sessile polyps. Colonoscopic examination revealed multiple polyps through the colon. JPS diagnosis must be based on histopathological findings coherent with juvenile polyps (Figure 2). We obtained written consents forms from the patient's primary caretakers for this case presentation.

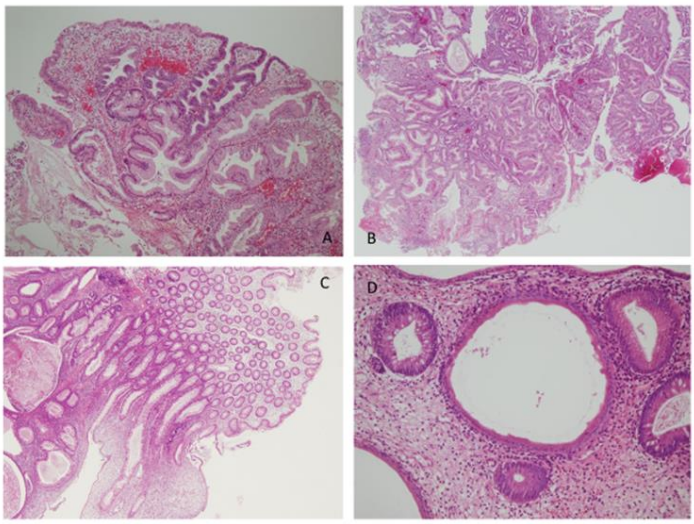

Figure 2: Mucosal pathology (A: Gastric mucosa juvenile poly, Hematoxylin&eosin x40, B: Gastric mucosa juvenile poly, Hematoxylin&eosin x100, C: Colon mucosa juvenile poly Hematoxylin&eosin x40, D: Focal low grade dysplasia, Hematoxylin&eosin x100

Discussion

There are several reasons of rectal bleeding, including infections, medications, and inflammatory bowel disease [9]. One of the most common causes of painless, rectal bleeding in children is colorectal polyps, most of which are sporadic, usually isolated, colorectal juvenile polyps. Sporadic isolated colorectal juvenile polyps are present in as many as 2% of symptomatic children [10]. We want to emphasize that the sporadic juvenile polyp diagnosis should be based on pathological examination of the polyps and clinical exclusion of JPS.

In our patient, extraintestinal system manifestations such as macrocephaly, congenital heart disease and clubbing accompanied protein losing enteropathy. Awareness of these clinical findings is necessary for the diagnosis of polyposis syndrome as the differential diagnosis of protein losing enteropathy. We mentioned the importance of a multidisciplinary approach, early recognition of the syndrome due to the elevated risk of mortality and malignancy development at later stages, and appropriate referral of the patient. This provides the best outcome for patients affected by polyposis syndrome. JPS carries an increased risk of gastrointestinal malignancy (38 % to 68 %) [11].

Conclusion

Our case highlights the importance of combined clinical findings of JPS with extraintestinal system manifestations and endoscopic examination for diagnosis. It is a rare syndrome, requiring more studies in which patients are monitored

prospectively to reach a comprehensive understanding of JPS and make its early diagnosis possible.

References

1. Howe JR, Sayed MG, Ahmed AF, Ringold J, Larsen-Haidle J, Merg A, et al. The prevalence of MADH4 and BMPR1A mutations in juvenile polyposis and absence of BMPR2, BMPR1B, and ACVR1 mutations. J Med Genet. 2004;41(7):484–91.
2. Syngal S, Brand RE, Church JM, Giardiello FM, Hampel HL, Burt RW. ACG clinical guideline: Genetic testing and management of hereditary gastrointestinal cancer syndromes. Am J Gastroenterol. 2015;110(2):223-62.
3. Sachatello CR, Griffen WO Jr. Hereditary polypoid diseases of the gastrointestinal tract: a working classification. Am J Surg. 1975;129:198–03.
4. Dahdaleh FS1, Carr JC, Calva D, Howe JR. Juvenile polyposis and other intestinal polyposis syndromes with microdeletions of chromosome 10q22-23. Clin Genet. 2012;81(2):110–6.
5. Schreibman IR1, Baker M, Amos C, McGarrity TJ. The hamartomatous polyposis syndromes: a clinical and molecular review. Am J. Gastroenterol. 2005;100:476-90.
6. Stojcev Z, Borun P, Hermann J, Krokowicz P, Cichy W, Kubaszewski L, et al. Hamartomatous polyposis syndromes. Hereditary Cancer in Clinical Practice. 2013 Jun 1;11(1):4.
7. Calva CD, Chinnathambi S, Pechman B, Bair J, Larsen HJ, Howe JR. The rate of germline mutation and large deletions of SMAD4 and BMPR1A in juvenile polyposis. Clin Genet. 2009;75:79-85.
8. Joy LH, James RH. Juvenile Polyposis Syndrome. Gene Reviews. 2003 May 13;1-19.
9. Peker A, Yarkıcı H, Akar H. Gastrointestinal bleeding secondary to use of high-dose methotrexate: A case report. J Surg Med. 2018;2(2):151-3.
10. Attard TM, Young RJ. Diagnosis and management of gastrointestinal polyps: pediatric considerations. Gastroenterol Nurs. 2006 Jan-Feb;29(1):16-22;quiz 23-4.
11. Cohen S, Hyer W, Mas E, Auth M, Attard TM, Spalinger J, et al. Management of Juvenile Polyposis Syndrome in Children and Adolescents: A Position Paper From the ESPGHAN Polyposis Working Group. J Pediatr Gastroenterol Nutr. 2019 Mar;68(3):453-62.

This paper has been checked for language accuracy by JOSAM editors.
The National Library of Medicine (NLM) citation style guide has been used in this paper.