Clinical Images and Case Reports Journal

Clinical Image | Vol 3 Iss 3

Juvenile Polyposis Syndrome: A New Case in a 7 Month Old Female

Jose Carlos Salazar[1*], Justo Valverde[1], María Rubio[1], Alejandro Rodríguez[1], Beatriz Espín[1], María José Moya[2] and Rocío Granero[2]

[1]Unit of Gastroenterology, Hepatology and Pediatric Nutrition, HHUU Virgen del Rocío, Sevilla, Spain

[2]UGC Pediatric Surgery, HHUU Virgen del Rocío, Sevilla, Spain

[*]**Corresponding author:** Jose Carlos Salazar, Unit of Gastroenterology, Hepatology and Pediatric Nutrition, HHUU Virgen del Rocío, Sevilla, Spain, Tel: 654755602; E-mail: josesolrac@hotmail.com

Received: February 10, 2021; **Accepted:** February 16, 2021; **Published:** March 25, 2021

Clinical Image

A 7-months-old girl consulted for recurrent rectal prolapse (Figure 1) associated with episodes of bloody stools. She underwent esophagogastroduodenoscopy and colonoscopy with significant findings of 6 duodenal polyps and more than 50 polyps in the colon (Figure 2). Histopathology revealed juveniles polyps in all cases, without adenomatous transformation. Genetic testing and array bases comparative genomic hybridization demonstrated a de novo deletion at 10q23.21q23.31, encompassing the PTEN and BMPR1A genes.

Clinically, juvenile polyposis syndrome is defined by the presence of 5 or more juvenile polyps in the colorectum, juveniles polyps throughout the gastrointestinal tract or any number of juvenile polyps and a positive family history of juvenile polyposis [1,2]. A germline mutation in the SMAD4 or BMPR1A gene is found in about 50-60% of JPS [3,4]. These infants suffer from diarrhoea, haemorrhage, malnutrition and intussusception. Death usually occurs at an early age [5]. Management is mainly based in expert opinion. Prophylactic surgery is considered in patients with >50-100 polyps, severe gastrointestinal bleeding or diarrhoea, juvenile polyps with dysplasia or a strong family history of colorectal cancer. [1,2,5].

In our case it is interesting the unusual presentation of the syndrome, with the combination of rectal prolapse and bloody stools in an infant. As far as we know, it has not been reported so far.

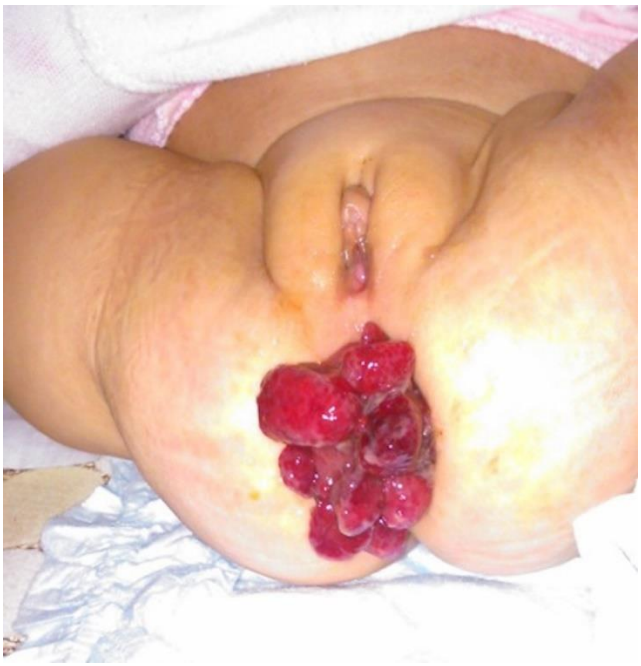

Figure 1: Rectal prolapse in an infant.

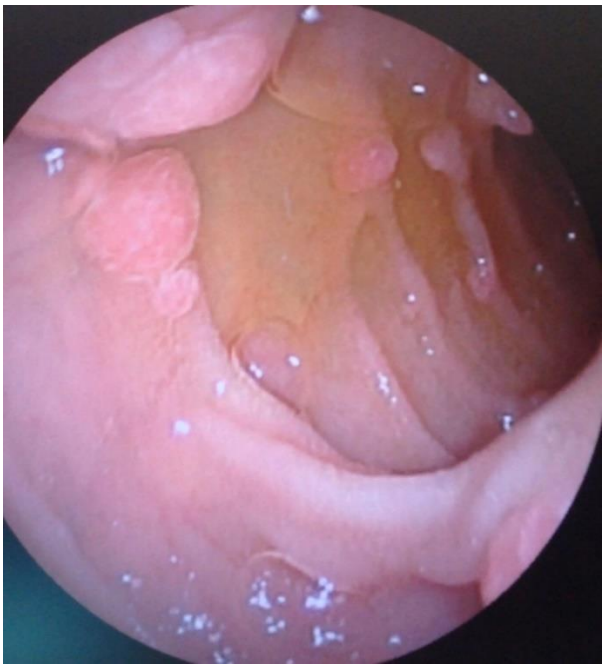

Figure 2: Various colonics polyps.

REFERENCES

1. Brosens LA, Langeveld D, van Hattem WA, et al. Juvenile polyposis syndrome. World J Gastroenterol. 2011; 17: 4839-4844.
2. Latchford AR, Neale K, Phillips RK, et al. Juvenile polyposis syndrome: A study of genotype, phenotype, and long-term outcome. Dis Colon Rectum. 2012; 55: 1038-1043.
3. Septer S, Zhang L, Lawson CE, et al. Aggressive juvenile polyposis in children with chromosome 10q23 deletion. World J Gastroenterol. 2013; 19: 2286-2292.
4. Dahdaleh FS, Carr JC, Calva D, et al. Juvenile polyposis and other intestinal polyposis syndromes with microdeletions of chromosome 10q22-23. Clin Genet. 2012; 81: 110-116.
5. Oliveira PH, Cunha C, Almeida S, et al. Juvenile polyposis of infancy in a child with deletion of BMPR1A and PTEN genes: Surgical approach. J Pediatr Surg. 2013; 48: e33-37.