



# Laparoscopic-assisted enteroscopy for intractable bleeding in a pediatric patient with Coats Plus Syndrome

Pedro E. Ruiz-Medina, MD, MPH; Jaime Aponte-Ortiz, MD; Juan Cruz-Lopez, MD; Antonio Del Valle, MD, FAAP; Jorge Zequeira, MD, FACS, FAAP  
University of Puerto Rico – Medical Sciences Campus  
School of Medicine- Department of Surgery



## Introduction

- Very rare genetic disorder with findings of cerebroretinal microangiopathy, gastrointestinal vascular anomalies, leukodystrophy, brain cysts, intracranial calcifications, and osteopenia.
- An autosomal recessive disorder, results from a genetic mutation in the CTC1 gene.
- It has been differentiated from other telomere biology disorders, in part, by recurrent gastrointestinal (GI) hemorrhage
- Our purpose is to inform on a very rare condition, and how we dealt with one of its dreadful manifestations, the gastrointestinal bleeding.

## Clinical Presentation

- 20 y/o female patient with Coats plus syndrome consulted to the pediatric surgery service due to presumed gastrointestinal bleeding of unknown origin that was diagnosed over a year ago.
- Suffered from significant anemia requiring PRBCs transfusions every 2 to 3 weeks.
- **Physical exam:** Unremarkable.
- **Previous studies:** Colonoscopy – anal fissures Upper endoscopies/Capsule endoscopy – esophagitis /mild gastritis.
- **Occult blood test** – consistently positive.
- **Tagged RBC scan** – blush in the left upper quadrant.

## Case Report

- Patient taken to OR for laparoscopic-assisted enteroscopy.
- Surgical findings:
  - No evidence of externally identifiable small bowel abnormalities after running the small bowel.
  - Enterotomy performed midway along the bowel through which the endoscope was pushed both proximally up to the ligament of Treitz and distally to the ileocecal valve.
  - Unremarkable distal small bowel with the exception of presence of melena.
  - Proximal jejunum had an area of ~5cm with obvious fresh blood clots and abundant blood. The bowel proximal to this region looked completely normal. There was also a small area about 10cm distal to the identified area with bleeding with a very mild mucosal bleed. No obviously identifiable bleeding vascular malformation or telangiectasia.
  - Enterectomy performed of area with obvious bleed and side to side, stapled anastomosis performed. Internal staple line verified to ensure absence of bleeding prior to enterotomy closure.
- Pathology report of the removed segment showed “small bowel with focal hemorrhages and vascular ectasia”
- Seen at clinics for follow up, hemoglobin levels have remained stable, repeat episodes of bleeding and transfusions, not been sufficiently frequent as to impair her quality of life as it had in the past.

## Discussion

- Recurrent gastrointestinal hemorrhage due to vascular ectasias in the stomach, small intestine, and liver is a major cause of morbidity and mortality.
- Conservative management strategies have proven ineffective.
- Multiple diagnostic modalities have been employed, ranging from standard endoscopy, capsule endoscopy and push enteroscopies.
- Standardized surgical management has not been established for this seldomly encountered condition with cases reported in the single digits.
- The infrequency and variability of symptoms makes tailored recommendations difficult.
- When diagnosis still unclear, and condition becomes life-altering or life-threatening, more aggressive management is warranted.
- With reports of fatal gastrointestinal bleeding, we conclude that laparoscopic-assisted enteroscopy is a viable diagnostic and palliative strategy that should be offered if conservative management fails.

## References

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