# A Rare Case of Colonic Duplication Cyst in an Adult with Down Syndrome

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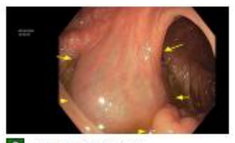
## Introduction

Alimentary duplications are cystic or tubular embryogenic anomalies occurring in 1 in 4500 individuals, often identified in children [1,2]. They are most commonly found in the small bowel followed by the Esophagus, with Colonic duplication cysts accounting for only 6.8% of the occurrence [3]. We present an extremely rare case of an adult with Down Syndrome with a Colonic Duplication cyst on Colonoscopy.

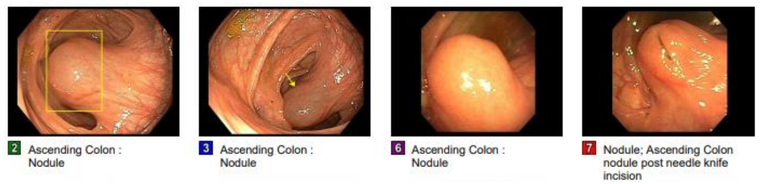
## Case Presentation

Our case is a 47-year-old female with a significant medical history of Down syndrome (complete trisomy 21) who exhibited ongoing weight loss (drop of 161 lbs to 155 lbs in one month). Initial evaluation revealed an elevated D-dimer level during assessment for syncope, with normal results from CT chest, CTA, and CT head. Abdominal CT revealed a large pedunculated lesion in the ascending colon, measuring approximately 5x3x3 cm, initially suspected to be a malignant polypoid colonic neoplasm. A standard colonoscopy identified a 50mm polypoid submucosal lesion in the ascending colon, which appeared cystic on further examination.



2 Ascending Colon

Biopsy during colonoscopy warranted additional sampling and a lower EUS Colonoscopy was performed which showed a round intramural (subepithelial) multi-cystic lesion of 26 mm thickness.




Biopsy results revealed a benign fragment of benign colonic mucosa with separate tissue fragments composed of increased small thin-walled vessels with smooth muscle with endothelial cells positive for CD31 and negative for CKAE1/3.
The diagnosis of colonic duplication cyst was confirmed. The risks of malignant transformation were discussed with the patient's mother, who preferred yearly colonoscopy with intra-cystic biopsies over surgical resection. Subsequent colonoscopy and biopsy results remained unchanged, and unfortunately the patient was lost to follow-up.

## Discussion

Colonic duplication cysts are a rare congenital anomaly that can be asymptomatic or present with symptoms such as abdominal pain, GI bleed, altered bowel habits, or weight loss as in our case.

While Down Syndrome patients are prone to enteric anomalies, no documentation of colonic duplication cysts exists in literature with two case reports on duodenal duplication cysts in this population[4,5]. Our study highlights the significance of this finding and calls for further research to explore a potential link between Down Syndrome and colonic duplication cysts. The cysts could consist of mucosa, submucosa, and muscularis propria elucidated with EUS Colonoscopy, as in this case, making this an effective diagnostic modality[6]. Though rare, the propensity of malignant transformation has been reported [7–9]. Surgical excision is the definitive treatment to avoid complications such as malignant transformation, intussusception, and hemorrhage [10].

## Conclusion

While colonic duplication cysts are rare and can be found incidentally or in symptomatic patients. Our study underscores the importance of considering the diagnosis in patients with Down Syndrome. It also highlights the use of effective diagnostic modalities and appropriate management strategies to prevent complications including malignant transformation, thereby improving patient outcomes.

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