Rectal adenocarcinoma following imperforate anus: A rare case report

Sarah Khan[1], Hina Abdul Qayoom Khan[1*], Jan Muhammad Agha[2], Khursheed Ahmed Samo[1], Mujeeb Ur Rehman Abbasi[1], Amjad Siraj Memon[1]

European Journal of Medical Case Reports
Volume 4(8):252–254

https://doi.org/10.24911/ejmcr/173-1582698308

ABSTRACT

Background: Anorectal malformations are a rare presentation. The standard treatment modality for imperforate anus is surgery. The abdominoperineal pull-through procedure was popular in the past for this malformation, but, with recent advancement, the posterior sagittal anorectoplasty and colostomy as a staged procedure are other options. Chronic constipation following pull-through procedures for imperforate anus may also lead to degeneration of the anorectal segment.

Case Presentation: A 28-year-old male presented with the complaint of on and off constipation and painful defecation since childhood. The patient had a history of imperforate anus congenitally, for which a pull-through procedure was done after birth at another hospital. He underwent multiple surgeries for constipation. During this time period, the patient had constant complaints of difficulty in defecation and constipation, for which he used to use Hager's dilators to dilate the anal canal. He also used per rectal enema and suppositories. His digital rectal examination showed narrowed anal opening. Colonoscopy was planned, which showed ulcerated and necrotic area at 6–8 cm from anal verge. Biopsy came out to be moderate-to-poor differentiated adenocarcinoma with signet ring cell differentiation. His computed tomography scan was performed, which showed circumferential mural thickening starting from the anal verge to the rectosigmoid junction with local infiltration to prostate, bilateral levator ani, and posterior sacral space. The patient was given a diversion colostomy and was referred for neoadjuvant chemoradiotherapy.

Conclusion: All patients undergoing pull-through procedures should be closely followed in a multidisciplinary unit with interval colonoscopy for the early detection and prompt management of rectal cancer.

Keywords: Rectal carcinoma, anorectal malformation, pull-through, case report.

Received: 03 April 2020 **Accepted:** 10 June 2020

Type of Article: CASE REPORT **Specialty:** General Surgery

Funding: None.

Declaration of conflicting interests: The authors declare that there is no conflict of interest regarding the publication of this article.

Correspondence to: Hina Abdul Qayoom Khan
*Surgical Unit 3, Dow University of Health Sciences, Karachi, Pakistan.
Email: hina.khan@duhs.edu.pk
Full list of author information is available at the end of the article.

Background

Anorectal malformations are a rare presentation. About 1 in every 10,000 children is born with imperforate anus [1]. In the past, the abdominoperineal pull-through procedure was used to correct these malformations. However, with recent advancement, the posterior sagittal anorectoplasty has been used with better results to correct this deformity and was described for the first time by Levitt et al. in 2012 [2]. A colostomy is an add-on treatment modality that is considered when the patient has co-existing rectobulbar urethral fistula, rectal atresia, rectobladder neck fistula, and imperforate anus without fistula. However, the risk associated with surgery results in long-term morbidity and post-operative sequelae, which includes fecal soiling, constipation, and fecal incontinence. There has also been extremely rare report of malignancy following pull-through procedures within the anorectal segment as well [3–6].

Colorectal carcinoma is associated with multiple predisposing factors. These include genetic, dietary, and environmental factors. The chronic exposure of the rectal mucosa to carcinogens following a chronic constipation is associated with distal colonic and rectal cancer.

Case Presentation

A 28-year-old male presented with the complaints of on and off constipation and painful defecation since childhood. The patient had a history of imperforate anus congenitally, for which a pull-through procedure was done after birth. At 8 years of age, the patient had an ileostomy made along with another anal procedure. This ileostomy was reversed after 6 months. At 15 years of age, the patient had another ileostomy made for absolute constipation also reversed after 6 months. After the closure of stoma, the patient had constant complaints of difficulty in defecation and constipation, for which he used to use Hager's dilators to dilate the anal canal and also used per rectal enema and suppositories. A family history was not significant for colorectal cancer.

There was a narrowed anal opening on the digital rectal examination.

Before 2 years, his first colonoscopy showed non-obstructing narrowing at anal verge with hard and fibrotic mucosa. A biopsy was unremarkable. We planned for another colonoscopy that showed ulcerated and necrotic area at 6–8 cm from the anal verge. The biopsy came out to be moderate-to-poor differentiated adenocarcinoma with signet ring cell differentiation.

His computed tomography scan and magnetic resonance imaging were performed, which showed circumferential mural thickening starting from the anal verge to the rectosigmoid junction with local infiltration to prostate, bilateral levator ani and posterior sacral space, and stage III rectal carcinoma (Figure 1).

The patient was given a diversion colostomy and was referred for neoadjuvant chemoradiotherapy.

Discussion

We presented the case of a 28-year-old patient with no family history of colorectal cancer presenting to us with signet ring cell adenocarcinoma of the rectum following a pull-through procedure. About 0.1%–2.4% of cases of all colorectal cancers present with primary signet ring cell differentiation [7]. The 5-year survival rate is found to be 9.1% in a study conducted by Messerini et al. [8]. In a study conducted by Anthony et al., 72.4% of patients with primary signet ring cell carcinoma present with distant metastasis or nodal involvement at the time of presentation, of which 75.8% died of advanced disease in an average time of 18 months [7]. This highlights the tendency of signet ring cell adenocarcinoma to involve retroperitoneal lymph nodes being a major contributory factor to failure in response to therapy.

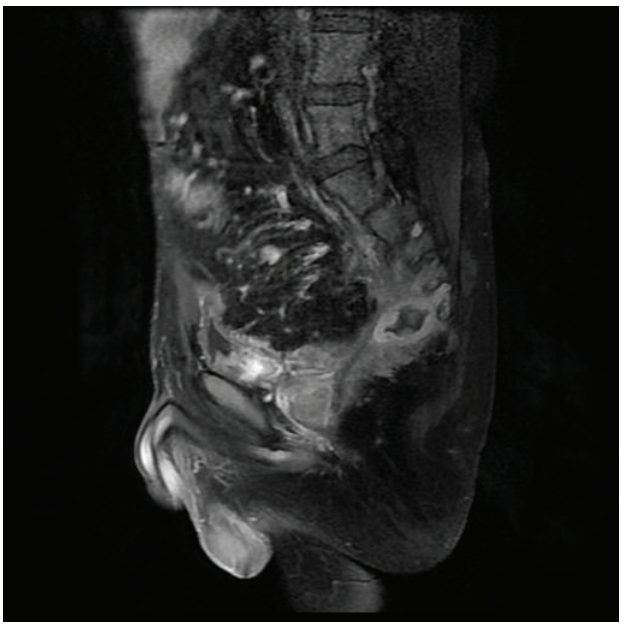

Figure 1. *MRI of Pelvis showing circumferential mural thickening from the anal verge to the rectosigmoid junction with local infiltration to prostate, bilateral levator ani and posterior sacral space.*

A review of articles has reported that overall 12 such cases have been reported [9]. In one study conducted by Midrio et al. [10] in 2016, two cases of imperforate anus presented with mucinous adenocarcinoma of the rectum at a young age following a pull-through procedure in the past. However, these cases had rectourethral and rectobulbar fistula at presentation. Similarly, Mukawa et al. and Symons et al. [3,4], in their case report, documented the finding of adenocarcinoma of the rectum following a pull-through procedure for rectobulbar and rectourethral fistula.

Posey et al. [11] reported a case of signet ring cell adenocarcinoma as a consequence of chronic exposure of urine as an irritant to rectal mucosa in a patient of imperforate anus with coexisting neurogenic bladder.

Symons et al. and Gupta et al. [4,6] reported the occurrence of para-neorectal adenocarcinoma, suggesting that it arises most likely from remnants of rectal mucosa left outside the neorectum following pull-through procedures. Another case study done by Mukawa et al. [3] stated that fecal incontinence as a complication to pull through procedure leads to the chronic source of irritation to rectal mucosa, hence leading to the development of anorectal carcinoma.

In this study, the patient had a long history of constipation following the pull-through procedure. The development of rectal adenocarcinoma, in this case, is likely a result of the chronic exposure of the rectal mucosa to fecal content due to increased transit time. This might be a predisposing factor leading to the malignant degeneration of the already compromised pulled through segment of the anorectum.

However, evidence is not conclusive, and more studies are required to evaluate the exact pathophysiology behind the occurrence of adenocarcinoma of anorectum following pull-through procedures.

Conclusion and Rationale

We recommend that all patients undergoing pull-through procedures should be closely followed in a multidisciplinary unit with interval colonoscopy for the early detection and prompt management of rectal cancer.

What is new?

Anorectal canal malformations are a rare presentation. About 1 in every 10000 children is born with Imperforate anus. Therefore, information and knowledge is required for the proper management. This is a intresting case report of a patient with carcinoma rectum due to surgical procedure done in childhood for anorectal malformation.

Consent for publication

A written consent was taken from the patient for publication.

Ethical approval

Ethical approval is not required at the institution to publish an anonymous case report.

Author details

Sarah Khan[1], Hina Abdul Qayoom Khan[1], Jan Muhammad Agha[2], Khursheed Ahmed Samo[1], Mujeeb Ur Rehman Abbasi[1], Amjad Siraj Memon[1]

1. Surgical unit 3, Dow University of Health Sciences, Karachi, Pakistan
2. Surgical Unit 3, Dr. Ruth K.M. Pfau Civil Hospital Karachi, Pakistan

References

1. Taipale P, Rovamo L, Hiilesmaa V. First-trimester diagnosis of imperforate anus. Ultrasound Obstet Gynecol. 2005;25(2):187–8. https://doi.org/10.1002/uog.1832
2. Samuk I, Bischoff A, Hall J, Levitt M, Peña A. Anorectal malformation with rectobladder neck fistula: a distinct and challenging malformation. J Pediatr Surg. 2016;51(10):1592–6. https://doi.org/10.1016/j.jpedsurg.2016.06.001
3. Mukawa K, Kurabayashi Y, Nakamura T, Nakano G, Nagamachi Y. Adenocarcinoma arising at the perineal wound after pull-through procedure for imperforate anus. Jpn J Surg. 1988;18(2):203–7. https://doi.org/10.1007/BF02471431
4. Symons NRA, Guenther T, Gupta A, Northover JMA. Para-neorectal mucinous adenocarcinoma following childhood pull-through procedure for imperforate anus. Colorectal Dis. 2010;12(3):262–3. https://doi.org/10.1111/j.1463-1318.2008.01762.x
5. Ahmed SN, Evans MD, Bose P, Drew P, Beynon J, Davies M. Rectal cancer following abdomino-perineal pull-through for imperforate anus. Colorectal Dis. 2012;14:e363–4. https://doi.org/10.1111/j.1463-1318.2012.02876.x .
6. Gupta RA, Nagral S, Mangukiya D, Nagral A. Rectal adenocarcinoma in an adult with imperforate anus at birth. Case Rep. 2012;2012:bcr2012006385. https://doi.org/10.1136/bcr-2012-006385
7. Anthony T, George R, Rodriguez-Bigas M, Petrelli NJ. Primary signet-ring cell carcinoma of the colon and rectum. Ann Surg Oncol. 1996;3:344–8. https://doi.org/10.1007/bf02305663
8. Messerini L, Palomba A, Zampi G. Primary signet-ring cell carcinoma of the colon and rectum. Dis Colon Rectum. 1995;38:1189–92. https://doi.org/10.1007/bf02048335
9. Ou J, Jao S, Kang J, Lai H, Hsiao C. Anorectal adenocarcinoma after pull-through procedure for imperforate anus. J Med Sci. 2007;27:137–40. https://doi.org/10.6136/JMS.2007.27(3).137
10. Midrio P, Battaglia S, Urso E, Castagnetti M, Gamba P. Rectal adenocarcinoma in patients with anorectal malformations: report of two cases and a review of the literature. Springerplus 2016;5(1):1623. https://doi.org/10.1186/s40064-016-3263-5
11. Posey JT, Neulander EZ, Soloway MS, Civantos F. Signet ring cell carcinoma of a pulled-through sigmoid colon mimicking a primary invasive bladder tumor: case report and review of the literature. Urology 2000;55(6):949. https://doi.org/10.1016/s0090-4295(00)00477-5

Summary of the case

1	**Patient (gender, age)**	Male, 28 year old
2	**Final diagnosis**	Colorectal Cancer as a sequel of pull through procedure done for imperforate anus
3	**Symptoms**	Severe constipation
4	**Medications**	Stool softeners, ispaghol husk, hagers dilator
5	**Clinical procedure**	Pull – through procedure, multiple ileaostomy due to constipation
6	**Specialty**	General Surgery