Case Report

Recto-sigmoid neurofibroma presenting as rectal prolapse in an adult patient with neurofibromatosis - a rare presentation

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Abstract

Neurofibroma occurs most frequently in the stomach and jejunum, but the colon may also be involved. Its association with rectal prolapse has not been reported yet. We are reporting here such a rare case report.

Case presentation

We report a case of NF leading to rectal prolapse. A 65-year-old man with neurofibromatosis (NF), type I presented with mass protruding from anus since 8 years and bleeding per rectum since 1 year. Physical exam revealed multiple cafe-u-lait spots all over the body. Local examination revealed Complete rectal prolapse, on per rectal examination and proctoscopy grade 1 haemorrhoids were found, no other gross abnormality was found. Colonoscopy was done which did not reveal any abnormality. Per abdomen was soft, non-tender, non distended, with normal bowel sounds. Laboratory studies were normal and the patient remained afebrile. Abdominal rectopexy was planned, but laparotomy revealed presence of a mass appx. 4 x 5 cm in the recto sigmoid region which was firm in consistency and had smooth surface. It was resected with 2cm margins from the rectum and 5cm from sigmoid colon. Diverting colostomy was done(Figure 3). Histopathological examination of the mass revealed a neurofibroma. Colostomy was closed 6 weeks later and colo rectal anastamoses was done. Patient on follow up is doing well.

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Discussion

Rectal prolapse refers to a circumferential, full-thickness protrusion of the rectum through the anus and has also been called "first-degree" prolapse, "complete" prolapse, or procidentia. In adults, this condition is far more common among women, with a female:male ratio of 6:1. Prolapse becomes more prevalent with age in women and peaks in the seventh decade of life. In men, prevalence is unrelated to age. Neurofibromatosis (NF) involves the gastrointestinal tract one third of the time, 5% of which are symptomatic. Neurofibroma occurs most frequently in the stomach and jejunum, but the colon may also be involved. This condition is characterized by multiple submucosal neurofibromas of the gastrointestinal tract and café au lait pigmentation, bony abnormalities, and neurofibromas of both central and peripheral nerves. The lesions consist of an overgrowth of neural tissue along with other mesenchymal elements. Gastrointestinal neurofibromas may cause occult bleeding, luminal obstruction, or intussusception. Malignant transformation into neurofibrosarcoma is rare. Lesions are essentially confined to the stomach and the jejunum. The symptoms usually include occult bleeding and obstruction. NF is a hereditary disorder associated with café-au-lait lesions, neurofibromas and involvement of multiple organs. The occurrence is 1 in 3500 to 4000 individuals. There are rare reports of NF involvement of the colon presenting as pseudo-obstruction, megacolon and colonic tumor. An extensive literature search failed to reveal any prior cases of rectal prolapse caused by a neurofibroma. In this patient with neurofibromatosis, we identified a recto-sigmoid neurofibroma as the tumor that led to rectal prolapse requiring surgery.

Conclusion- We report that the possibility of the rectosigmoid neurofibroma should be kept in mind in a patient of neurofibromatosis presenting with rectal prolapse.

References