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CLINICAL IMAGE

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"De novo" familial adenomatous polyposis (FAP) presenting as rectal cancer

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Abstract

Familial adenomatous polyposis is an autosomal dominant disorder with familial predisposition. 25%-30% cases arise "de novo," without any clinical or genetic evidence.

KEYWORDS

de novo mutations, familial adenomatous polyposis (FAP), germline mutation

1 | CASE SUMMARY

1.1 | Surgical quiz

All of the following are true regarding "de novo" familial adenomatous polyposis (FAP) except.

a. Incidence is 11%-25%
b. Diagnosed by next generation (NGS) sequencing in parent offspring trios
c. It is a germline meiotic event
d. It is a somatic mitotic event

1.2 | Answer

d) It is a somatic mitotic event.

A 41-year-old woman presented with rectal bleeding for 10 years. Clinical, radiological, and endoscopic examination revealed a proliferative lesion (3.6 cm × 2.4 cm × 3.3 cm) 2 cm from anal verge with numerous polyps of varying size from anal canal to caecum. [Figure 1] There was no family history. Biopsy revealed adeno-carcinoma and benign tubular adenoma from the lesion and polyp, respectively.

Patient was diagnosed as "de novo" FAP, and total proctocolectomy with permanent ileostomy (TPI) was done. Postoperative specimen showed multiple polyps [Figure 2]. Histopathology confirmed poorly differentiated mucin secreting adeno-carcinoma. Colonoscopy and biopsy of her children revealed multiple benign polyps in the entire colon. Children have been advised annual colonoscopy.

2 | DISCUSSION

Familial adenomatous polyposis is an autosomal dominant disorder caused by germline mutation in APC gene in chromosome 5q21. A total of 11 to 25% arise "de novo" by novel genetic mutation.[1] Typical characteristic of de novo FAP which differentiates from other "de novo" mutations includes no evidence of paternal bias, no paternal age effect, atypical loci, nonrandom errors in DNA replication, no founder effect, and early age of onset. Next generation sequencing approach in parent-offspring trios can accurately calculate the number of "de novo" mutations throughout the genome. Presentation, diagnosis, management, and surveillance are similar to FAP.[2]

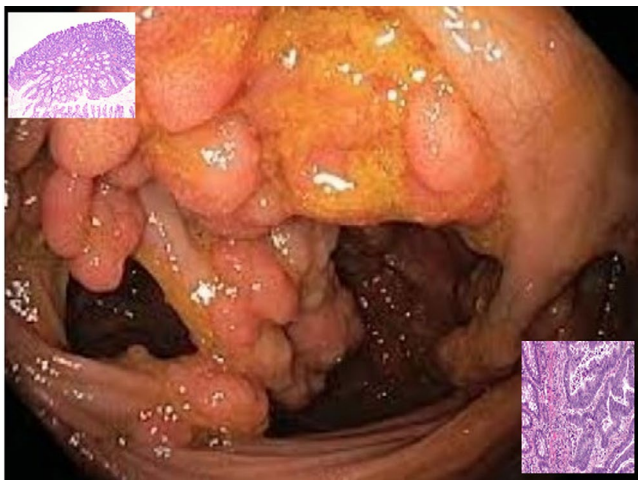


FIGURE 1 Colonoscopy showing colonic polyps, inset HPE (Left upper—benign adenoma, Right bottom—adeno-carcinoma)

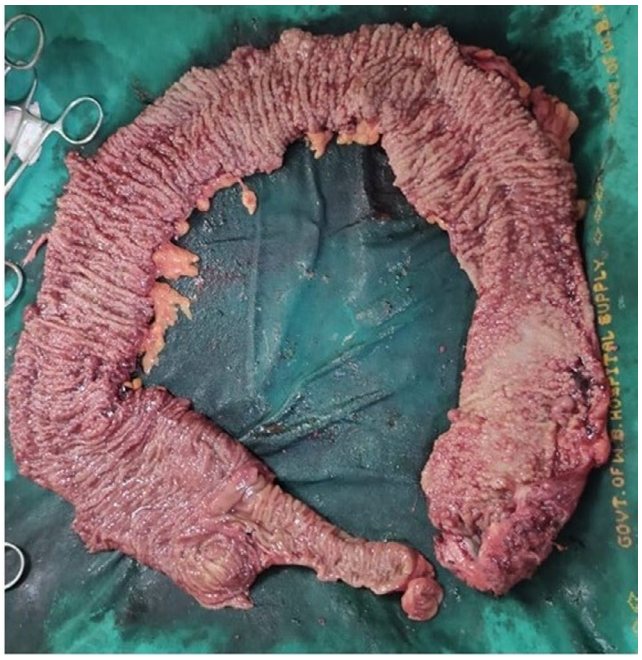


FIGURE 2 Specimen of total proctocolectomy showing numerous polyps

ACKNOWLEDGMENTS

None. Published with written consent of the patient.

CONFLICT OF INTEREST

None.

AUTHOR CONTRIBUTIONS

RA: conceived the presented idea. UD and RA: wrote the manuscript.

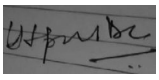
ETHICAL APPROVAL

Hereby, I, Dr Utpal De, consciously assure that for the manuscript "de novo" Familial adenomatous polyposis (FAP) presenting as rectal cancer" the following is fulfilled:

1. This material is the authors' own original work, which has not been previously published elsewhere.
2. The paper is not currently being considered for publication elsewhere.
3. The paper reflects the authors' own research and analysis in a truthful and complete manner.
4. The paper properly credits the meaningful contributions of co-authors and co-researchers.
5. The results are appropriately placed in the context of prior and existing research.
6. All citations used are properly disclosed.
7. All authors have been personally and actively involved in substantial work leading to the paper, and will take public responsibility for its content.

The violation of the Ethical Statement rules may result in severe consequences.

I agree with the above statements and declare that this submission follows the policies of as outlined in the Guide for Authors and in the Ethical Statement.



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