ORIGINAL RESEARCH PAPER
Volume - 13 | Issue - 12 | December - 2024 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

GARDNER SYNDROME IN DISGUISE-DESMOID TUMOUR AS AN INITIAL PRESENTATION OF FAMILIAL ADENOMATOUS POLYPOSIS

General Surgery

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ABSTRACT

Desmoid tumours are rare, fibromatous lesions which are non-metastasizing and locally aggressive. They have a high rate of recurrence even after complete resection. It is the second most common tumour to be associated with Familial Adenomatous Polyposis (FAP). A desmoid tumour being an initial presentation of FAP is rare. Gardner syndrome is a phenotypic variant of FAP characterized by numerous adenomatous polyps lining the intestinal mucosal surface along with extra colonic manifestations like desmoids, osteomas and epidermoid cysts. This case report of a 30 year female presenting with an innocuous tumour hiding a more sinister diagnosis is being presented for its rarity.

KEYWORDS

INTRODUCTION:

The term "desmoid" means a tendon or bark in Greek. It was first used by the German physiologist and comparative anatomist Johannes Müller in 1838. Desmoid tumors (DTs) form a minor 0.03% of all tumors. Predisposing factors to DT include female gender, prior abdominal trauma or surgery, and a family history of fibromatoses. Primary or sporadic DTs are extremely rare and originate from the proliferation of fibroblasts-myofibroblasts with the formation of benign stromal neoplasms. Secondary DTs are commonly associated with hereditary polyposis syndromes with 30% of FAP patients developing intra-abdominal desmoids disease, including tumors, nodules, and sheet-like lesions. (1) They are usually present in familial adenomatous polyposis (FAP) syndrome as extra- and intra-abdominal manifestations (**2**). Extra-abdominal DTs originate from muscle-aponeurotic structures, usually part of the abdominal wall, while intra-abdominal DTs develop in mesenteric folds or in retroperitoneal tissue.(3)

CASE REPORT:

A 30 year old mother of 3 children with no known comorbidities presented with a hypogastric lump for 2 years which was insidious in onset, progressive in size and intermittently painful. On examination, she had a 6x4 cm firm lump to the right of the LSCS Pfanensteil scar. She was provisionally worked up as rectus sheath hematoma. She also had a swelling in the right hemi mandible.

Imaging revealed heterogeneously enhancing soft tissue density lesion arising from the right rectus muscle, 7.2x6.5x3.7 cm, HU-43

Core needle biopsy Histopathological Examination: Desmoid fibromatosis

Swelling in the right hemi mandible was found to be an osteoma

Colonoscopy revealed multiple polypoid lesions in the cecum. Multiple small diminutive polyps of size 5x5mm-dispersed from splenic flexure to 10cm from anal verge

HPE: Tubular adenoma with low grade dysplasia

Procedure done: Total colectomy with ileorectal anastomosis, excision of desmoid, anterior component separation and onlay mesh repair

Post op histopathological examination also showed the presence of adenomatous polyps with low grade dysplasia.

The patient is under follow up with annual colonoscopies. Her family members have been counselled regarding the nature of the disease and the importance of screening,

DISCUSSION:

2% of cases of desmoid tumours are associated with FAP [4]. Whereas the incidence of desmoid tumours in the general population is 2–4 per million per year, the incidence in FAP is 10–20%, meaning that a patient with FAP is at an 852-fold increased risk of developing a desmoid tumours (5). While sporadic desmoid tumours show a slight female preponderance, there is an equal incidence in males and females in FAP patients [6]. Familial aggregation is seen in FAP desmoid tumours, but not in sporadic cases. FAP is an autosomal dominant condition caused by a germline mutation in the adenomatous polyposis coli (APC) gene, on chromosome 5q21 (7).FAP is characterized by hundreds of adenomatous colorectal polyps, with an almost inevitable progression to colorectal cancer at an average age of 35–40 years (7) Biallelic mutations of the APC gene induces desmoid tumour formation, hence the association between these two disorders. Gardner's syndrome is also caused by a mutation in the APC gene. It includes extracolonic manifestations, both benign, such as osteomas, skin cysts, congenital hypertrophy of the retinal pigmented epithelium and desmoid tumours and malignant, such as duodenal, thyroid, pancreatic, liver and central nervous system cancers. Previously, Gardner's syndrome used to be considered as a separate entity, but it is now considered a different phenotypic presentation of FAP.

CONCLUSION:

Desmoid tumours are rare tumours with limited malignant or metastatic potential but locally aggressive. They are the second most common tumour associated with FAP, but an FAP presenting initially with a desmoid is rare

A head to foot clinical survey is essential in all patients since there may be syndromic associations as was in this case.

A close surveillance of such patients is necessary and screening of family members is crucial to detect the disease earlier on, if present.

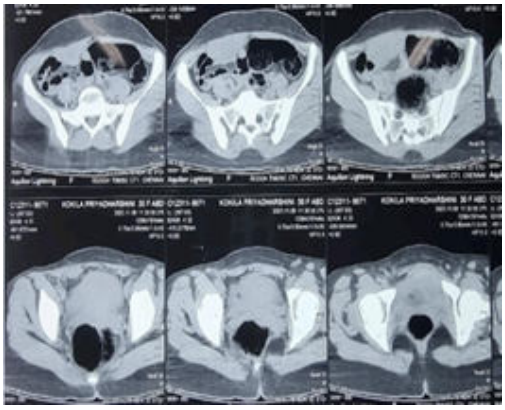

Figure 1: Contrast Enhaced CT Scan Showing Desmoid Tumour

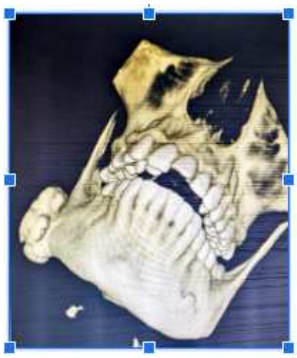

Figure 2: Cone Beam Computed Tomography Showing Osteoma Of Right Hemimandible

Figure 2: Resected Colon Showing Multiple Polyps

Figure 3: Onlay Mesh Repair Post Desmoid Excision

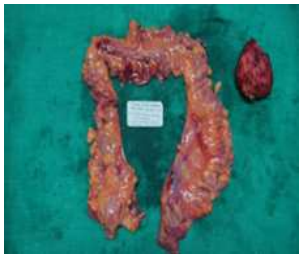

Figure 4: Postoperative Specimen Of Resected Bowel And Desmoid

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