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Gardner's Syndrome Represents by Abdominal Mass: A Case Report

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ABSTRACT

Gardner's syndrome is an autosomal dominant form of polyposis characterized by the presence of multiple polyps in the colon together with extra-colonic manifestations like multiple osteomas, soft tissue tumors, and desmoid tumors (which are benign neoplasia arising from musculoaponeurotic structures throughout the body, with local aggressive nature and can cause painful abdominal mass, intestinal and ureteric obstruction leading to hydronephrosis, hemorrhage, and fistula) that occur in the mesentery and the abdominal wall.

In our case, we will represent a case of 37-year-old male presents with abdominal pain on Clinical Examination we noticed epigastric mass diagnosed by core biopsy as Desmoid tumors, on physical examination, there is skull osteoma confirmed by skull X-ray and Colonoscopy did show multiple dysplastic polyps. From all the above we confirm the diagnosis of Gardner's Syndrome.

KEYWORDS

Gardner's syndrome; Desmoid tumors; Familial adenomatous polyposis

1. INTRODUCTION

In the early 1950s, Gardner described kindred with intestinal characteristics of familial adenomatous polyposis (FAP), but also with some extra-colonic growths, including osteomas, epidermal cysts, and fibromas [1].

Gardner's syndrome is more of a historical subdivision of FAP, associated with either upper gastrointestinal tract polyps, congenital hypertrophy of the retinal pigment epithelium, desmoid tumors, dental anomalies, soft tissue tumors, and other extra-colonic malignancies. FAP is an autosomal-dominant colorectal cancer syndrome, caused by a germline mutation in the adenomatous polyposis coli (APC) gene, on chromosome 5q21 [2].

It is characterized by hundreds of adenomatous colorectal polyps, with an almost inevitable progression to colorectal cancer at an average age of 35 years to 40 years [2]. It became apparent that both FAP and Gardner's syndrome arose from the same APC gene mutations [3].

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Other specified variants include Turcot syndrome, which was described by Turcot in 1959 as the occurrence of central nervous system malignancies like medulloblastoma in conjunction with colorectal polyps and FAP [4].

2. CASE REPORT

A 37-year-old male worker, chronic smoker, neither diabetic nor hypertensive, Irrelevant past medical history, he is the oldest of two brothers, no family history of diseases except diabetes in his father. His complaint started 2 months ago with persistent dragging abdominal pain mainly epigastric referred to the flanks, exaggerated after meals, and relieved by fasting. The patient also had intermittent vomiting and diarrhea for 3 months (loose stools of 3 motions/day). He sought medical advice and received treatment for gastritis with no improvement. Previous abdominal and pelvic ultrasound were done a few months ago and were normal. Clinical examination revealed a painless epigastric mass about (8 cm × 8 cm) with rounded borders and a smooth surface that is not fixated to the skin (amalgamated intestinal loops). Head examination showed multiple hard pea-sized swellings; he reports that he had these swellings since birth. It was confirmed by plain X-ray as multiple small osteomata (Figure 1 and Figure 2). CT abdomen report revealed diffuse scattered ill-defined confluent hypodense mass-like lesions involving mesentery with smudged related fat plains, most probably due to either sclerosing mesenteritis or lymphoma (Figure 3). Tumor markers CA19-9 & carcinogenic embryonic antigen (CEA) were done to exclude abdominal malignancy and the results were within normal. Colonoscopy revealed multiple minute small polyps (less than 5 mm) with biopsies showing dysplastic changes. Ultrasound-guided core biopsy from the mesenteric mass revealed desmoid fibromatosis, smears rich in atypical lymphoid cells suspicious of coexistence of lymphoma. The patient was admitted to the internal medicine department to determine the cause of mesenteritis, Flow cytometry was done that showed mainly reactive T lymphocytes with no immuno-phenotypic evidence of infiltration by leukemic or lymphomatous proliferations cells, and hence lymphoma was excluded.

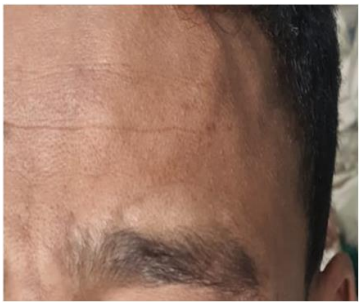

Figure 1: Osteoma in the forehead.

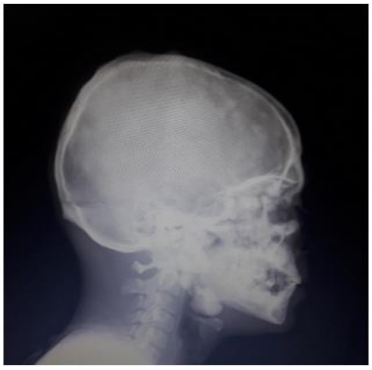

Figure 2: Multiple osteomas in skull X-ray.

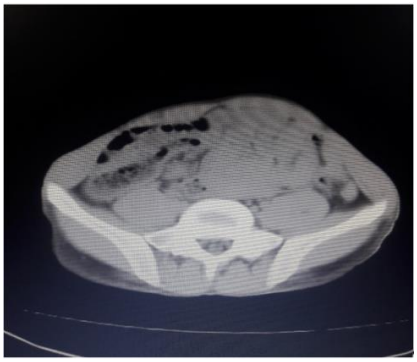

Figure 3: CT of the abdomen showing amalgamated intestinal loops.

3. DISCUSSION

Gardner's syndrome is a genetic disease with autosomal dominant inheritance. It is a variant of FAP characterized by colonic polyps, and multiple osteomas. Colonic polyps begin to form in puberty, but the average age of diagnosis is 22-years-old. Cutaneous findings of the

disease include epidermoid cysts, desmoid tumors, and other benign tumors [2].

Osteoma is one of the extra-colonic manifestations of Gardner's syndrome, found in 20% of families with FAP. It is a benign bone growth that is mostly found in the skull and mandible, with variation in size and number but no malignant transformation [5].

Desmoid tumors (DT) is a rare, locally invasive fibromatosis, was first described by Johannes Mueller in 1838 as benign neoplasia arising from musculoaponeurotic structures throughout the body, with locally aggressive nature, incidences in the mesentery are about (50% - 75%) and in the abdominal wall is about (25% - 50%).

DT occurs in about 10% to 25% of FAP patients, commonly found in women, and is one of the causes of complications after colonic surgery. It is usually asymptomatic but may cause pain and bowel or ureteric obstruction. DT may be the first manifestation of GS [6].

Although they are considered benign but can be life-threatening through progressive growth that threatens the intra-abdominal organs, nerves, and vessels, they also, can arise in the mesentery or abdominal wall. There is currently no standard treatment for DT associated with FAP [6].

Surgical excision carries the risk of bleeding and short bowel syndrome, with a high rate of recurrence up to 45%, Non- steroids anti-inflammatory (sulindac) and antiestrogens have been used in management but only less than third of the tumors stabilize or regress [7].

Gardner syndrome is caused by mutations in the APC gene, which is called a "tumor suppressor". Polyps have a 100% risk of undergoing malignant transformation, consequently, early identification of Gardner's syndrome is critical, one person per million population is diagnosed with Gardner syndrome. FAP accounts for 1% of the cases of inherited colorectal cancer [8].

The main risk factor for developing Gardner's syndrome is having at least one parent with the condition. A spontaneous mutation in the APC gene is a much less common occurrence.

A blood test to check for Gardner's syndrome if multiple colon polyps are detected during lower GI tract endoscopy and blood test reveals if there is an APC gene mutation. Gardner's syndrome is inherited, there is no way to prevent it, CEA testing, liver function tests, and thyroid function tests to evaluate for possible metastasis.

In our case, the patient presented initially with abdominal swelling diagnosed as a desmoid tumor on physical examination, there is skull osteoma confirmed by skull X-ray and colonoscopy showing multiple dysplastic polyps. All of the above confirms the diagnosis of Gardner's syndrome.

We informed the patient about colectomy, and high incidence of malignant transformation, and explained to him that all guidelines advise surgery at age of 30 years as the majority of the FAP patients will develop cancer at age of 40 years. Prophylactic surgery is the only curative treatment; proctocolectomy with ileal pouch-anal anastomosis or total colectomy with ileorectal anastomosis [9].

Sulindac, a long-acting derivative of indomethacin, has been shown to cause regression of rectal polyps in 80% of cases of FAP after the patient has undergone total colectomy [10].

4. CONCLUSION

Diagnosis and management of extracolonic features of Gardner's syndrome are the major challenges in each case. Follow up and screening for patients and his family, endoscopic screening of FAP to reduce the occurrence of

colorectal cancer, Colectomy remains the optimal prophylactic treatment. For desmoids tumors surgical excision is not recommended, because of the high recurrence rate, surgery is reserved for life-threatening complications. Drugs like sulindac and antiestrogen have been used but only with limited effects.

5. CONFLICT OF INTEREST

There is no conflict of interest.

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