

Gardener Syndrome: A Rare Case Report

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Abstract

Gardner syndrome (GS) is a group of diseases manifested as familial adenomatous polyposis accompanied by characteristic jaw lesions. The documented prevalence of Gardner syndrome varies from 1: 8,300 to 1: 16,000 live births in varying literatures [1]. The syndrome associated intestinal polyps have a 100% risk of malignant transformation [2]. Early identification and surgical intervention of the disease are important to prolong the life of the patient. Facial abnormalities like familial adenomatous polyposis, osteomatous jaw, ocular lesions and several dental abnormalities, and abdominal desmoid tumors are the characteristic features of this disease [1]. We elaborate a case of Gardner syndrome emphasizing on the diagnostic aspect and the management of the syndrome.

Keywords: Gardner syndrome; Osteoma; Intestinal polyposis

Introduction

First described in 1951, Gardner's syndrome is a fairly rare condition sharing similarity with polyposis coli. It is characterized by surface tumors, multiple polyposis, and bone tumors, and is estimated to affect one in every 1,000,000 people [3].

Familial adenomatous coli, (FAC), genetically an autosomal dominant syndrome, are characterized by the presence of large numbers of adenomatous polyps in the colon with a marked tendency to malignant transformation. It is accompanied by malignant lesions in the upper digestive tract and other organs. FAC with osteomas and benign tumors of the soft tissues is referred to as Gardner's syndrome [4].

Gardner's syndrome is generally caused by a mutation of the APC (Adenomatous Polyposis Coli) gene, located on chromosome 5. About 30% of individuals with FAC have a new dominant mutation, being the first affected member of their family. Early diagnosis in at-risk individuals is usually performed by colonoscopy for detection of intestinal polyposis [5].

Polyps arising after the age of 20 years may affect the whole gastrointestinal tract and have a high potential for malignant transformation, which usually occurs around the fourth decade. Osteomas are mainly located in the mandible but can occur in any bone of the skull and even in long bones; their presence is necessary for the diagnosis of Gardner syndrome. Osteomas usually precede the intestinal polyposis by often as much as 10 years so their detection may lead to appropriate further investigations for the early diagnosis of the syndrome [6].

Case report

A 64 year old patient reported to Department of Oral and Maxillofacial surgery with a chief complain of swelling on the right

lower part of face for past 6 years and in front of right ear region for past 3 months. History of presenting swelling was that swellings were small in size and had gradually increased in size to attain the present size. The swelling was painless upon palpation. Patient also complained of another swelling in right temple region. Facial swellings were not associated with pain or difficulty in mouth opening (Figure 1a).

Extra oral examination showed round swelling with well-defined margins measuring approximately 2.5 x 2.5 cms in the right mandibular molar region. Skin over the swelling was normal in color without any secondary changes. Another oval swelling with well-defined margin measuring approximately 3 x 2 cms seen in the right pre-auricular region with normal skin color and no secondary changes (Figure 1b). Both swellings were bony hard, fixed to underlying bone and non-tender upon palpation.



Figure 1: A) Clinical picture shows ovoid swellings on frontal and left mandibular body region, B) Clinical picture shows obliteration of buccal sulcus in third quadrant and an extra-oral swelling on left mandibular region

Intra Oral Examination revealed two oval well circumscribed swellings measuring approximately 2.5 x 2.5cms and approximately 1.5 x 1.5 cms, seen on the buccal aspect of 46, 47 and 35, 36 with obliteration of the buccal sulcus. Overlying mucosa was normal in color and texture (Figure 1b).

Orthopantomograph revealed three well defined dense radiopaque masses at right condyle region, right body of the mandible and in the region of lower border of the mandible on left side, measuring approximately 2 x 1.5 cms, 2.5 x 2 cms, and 0.3 x 0.3 cms respectively (Figure 2). Water's view, obtained to inspect other facial regions, revealed a radio-opaque mass in the left frontal sinus measuring approximately 0.2 x 0.2 cms (Figure 3). Colonoscopy showed sessile polyps in the rectum and descending colon (Figure 4a and 4b). Biopsy revealed benign growths and no features of malignancy.

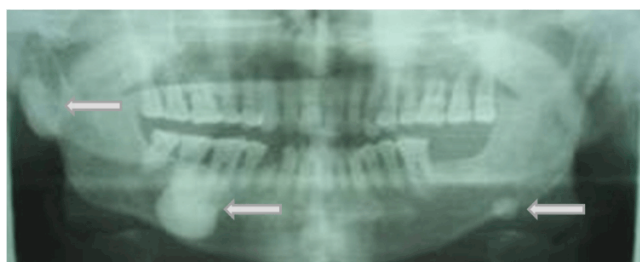


Figure 2: OPG reveals radiopaque masses in right condyle and body of mandible and left lower border of the mandible



Figure 3: Submento-vertex view reveals radiopaque mass in frontal sinus

With the above clinical, radiological and colonoscopy report we arrived at diagnosis of Gardner's Syndrome. As the facial swellings were benign and non-interfering with the function, patient was not willing for the surgical intervention for the Osteomas of mandible. Patient underwent prophylactic colectomy as the polyps have highest

potential for malignant transformation. Patient on regular follow up of two years showed no recurrence of intestinal polyps.

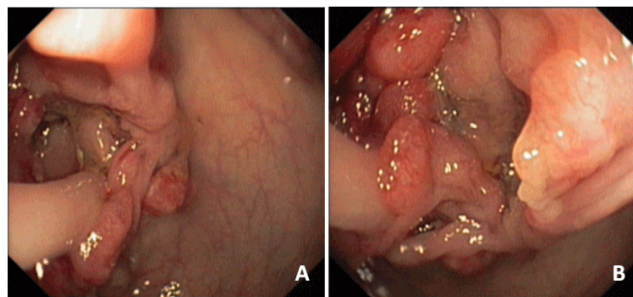


Figure 4a,b: Colonoscopy shows multiple sessile and pedunculated masses in intestine

Discussion

Familial adenomatous polyposis (FAP) is an autosomal dominant colon cancer syndrome characterized by the presence of polyps in the colon and rectum. Gardner syndrome (GS) is a variant of FAP showing extracolonic manifestations such as osteomas, dental anomalies, epidermoid cysts and congenital hypertrophy of the retinal pigmented epithelium.

The intestinal polyps carry as high as 100% potential of malignant change, and early diagnosis is therefore essential. As the extra-colonic manifestations of the disorder often precede the colonic polyps, they may facilitate early diagnosis of pathology allow timely intervention. Several of these manifestations occur in the oral and maxillofacial region and may be discovered during routine dental examination [7].

Tooth extraction in patient with Gardner syndrome (GS) has been reported to be likely difficult. Radiological studies attribute such difficulty to a thickening of interdental cortical bone. Studies suggest that periodontal tissues in few cases show almost complete absence of the periodontal space caused by an extensive hypercementosis. Dental anomalies are characterized by multiple impacted permanent teeth, supernumerary teeth and root anomalies. Odontomas and cementomas are also frequent Furthermore, tooth extraction in patients with GS has been reported to be very difficult likely because of the extremely dense nature of the alveolar bone and to the almost complete absence of the periodontal space caused by hypercementosis [5].

Skeletal abnormalities, the most common of which are Osteomas, are an essential component of Gardner syndrome. These benign tumors are characterized by slow, continuous growth, and occur most frequently in the mandible, the outer cortex of the skull and the paranasal sinuses as seen in our case at mandible regions and also in frontal sinus. The angle of the mandible is a particularly diagnostic site and is frequently associated with facial deformity [6]. The osteomas are usually peripheral exostoses which are clinically identifiable while enostoses are detectable only radiographically. The radiographic appearance of either type is a localized radiopaque lesion with a sharp border. Another type of lesion has been described, which appears as a large and diffuse radiopaque cotton-wool-like area in either jaw, and is referred to as a widespread radiopaque lesion [7].

The limitation mouth opening caused by the osteoma of the condyle is a significant and unusual finding. Limited opening has also been previously associated to an osteomas located in the maxilla in close proximity to the ascending ramus, the external body of the mandible, the coronoid process, and the inferior border of zygomatic arch.

Epidermoid cysts precede the intestinal polyposis and differ by common skin cysts for age and location of occurrence. In fact, those arising with puberty are generally multiple and asymptomatic and may be found on limbs, face, and scalp. Congenital hypertrophy of the retinal-pigmented epithelium (CHRPE) is reported in 80% of patients with GS and occurs early after birth so it may represent the first detected sign of the syndrome. However, it seems controversial, considering the presence of CHRPE as an early marker for FAP in at-risk individuals because of interfamilial difference in predisposition to CHRPE [5].

Desmoid tumours, although histologically benign, may behave aggressively and can cause death in some patients. Several other neoplasms are also associated with Gardner syndrome [7]. Other reported lesions include hepatocellular carcinoma, desmoid tumor, fibromas, lipomas, leiomyomas, neurofibromata, and pigmented skin lesions. In addition, several studies have reported an association between GS and endocrine disorders like Cushing's syndrome, thyroid gland nodules, craniopharyngioma, and thyroid cancer [5].

Prophylactic colectomy is usually recommended. Removal of jaw osteomas and epidermoid cysts for cosmetic reasons sometimes may be indicated, but the long term prognosis depends on the behavior of the bowl adenocarcinomas.

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