Classical presentation of Gardner's syndrome in an Indian patient: A case report

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Abstract

Gardner's syndrome is an autosomal dominant disease characterized by the presence of colonic polyposis, osteomas, and a multitude of soft-tissue tumors. Dental anomalies are present in estimated 30% of all affected individuals of Gardner's syndrome, so dental professionals play an important role in determining the early signs of the syndrome. The intestinal polyps have a 100% risk of undergoing malignant transformation if not treated thus, early diagnosis and regular surveillance are important. In this report, we describe classical presentation of Gardner's syndrome in a patient who presented with bilateral swellings on palate along with multiple impacted teeth.

Keywords: Gardner's syndrome, intestinal polyps, osteomas

Introduction

Gardner and Richard in their landmark article described a syndrome consisting of hereditary intestinal polyposis, with osteomas and multiple cutaneous and subcutaneous lesions.[1] After that other dental findings, skin and soft-tissue tumors have been added in the description of this syndrome.

The documented frequency of Gardner's syndrome is in between 1 in 1400 and 12000 live births.[2] The inheritance pattern is autosomal dominant with complete penetrance. However, approximately 20% of cases represent spontaneous mutations, with no family history reported. Mutation in the adenomatous polyposis coli gene which is present on chromosome 5 leads to this condition.[3] We report a case of Gardner's syndrome in an Indian patient which is representing the full continuum of dental, colonic, and extracolonic manifestations.

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Access this article online	
Quick Response Code:	Website: www.contempclindent.org
	DOI: 10.4103/0976-237X.183063

Case Report

A 52-year-old male patient reported to the outpatient department with a chief complaint of swelling on the right side of the palate for the past 5 years. The patient had undergone surgical treatment for a bony swelling at lower border of the mandible about 30 years back. After that, he was apparently normal until 5 years back when he noticed a small swelling on the palate which was very slowly increasing in size. The past medical history revealed chronic bowel upset in the form of abdominal cramps and diarrhea. Family history revealed that both sons of the patient had bony swellings of the jaw and both of them were reluctant to get their screening done for Gardner's syndrome.

On examination, multiple small nodular swellings were seen on the forehead. The nasolabial folds were obliterated bilaterally [Figure 1a]. The overlying skin appeared normal. On palpation, all the swellings were bony hard in consistency, nontender and were fixed to the underlying bone.

Intraoral examination revealed swellings on the right and left sides of the palate. On the right side, buccal vestibule was obliterated in 14 and 15 region and palatal swelling was extended from the mesial aspect of 16 to the distal aspect of tooth 17 anteroposteriorly. On the left side, the palatal swelling extended from mesial aspect of tooth 25 to the mesial aspect of 28 anteroposteriorly [Figure 1b]. A diffuse swelling was seen on mandible on the left side, causing buccal vestibular obliteration in 34 and 35 region. The overlying

How to cite this article: Verma P, Surya V, Kadam S, Umarji HR. Classical presentation of Gardner's syndrome in an Indian patient: A case report. Contemp Clin Dent 2016;7:277-80.

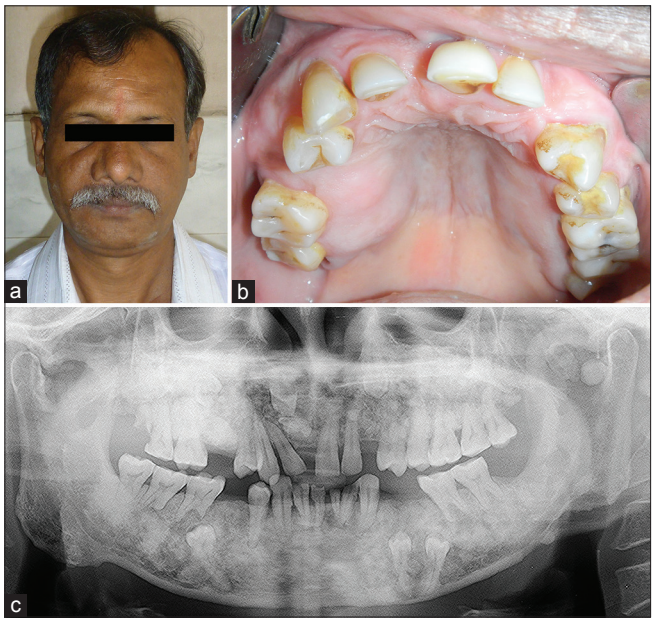

Figure 1: (a) Extraoral photograph shows fullness of nasolabial folds on both sides with small bony swellings seen on the right temple region. (b) Bony swellings seen on the right and left sides of the palate. On the right side, buccal vestibule is obliterated in 14 and 15 region and palatal swelling is extending from the mesial aspect of 16 to the distal aspect of tooth 17 anteroposteriorly. On the left side, the palatal swelling is extending from mesial aspect of tooth 25 to the mesial aspect of 28 anteroposteriorly. Teeth missing is 11, 15, and 23. (c) Orthopantomograph shows impacted teeth w.r.t. 11, 15, 23, 34, 35, and 45. Resorption of the roots w.r.t. 46 and 47 is noted. Dense radio-opacities are seen on the right and left maxilla in premolar-molar region. Maxilla and mandible show diffuse radiopaque areas suggestive of dense bone islands or enostoses. Surgical defect seen on the left lower border of mandible and a well-defined round radio-opacity is seen on the left sigmoid notch

mucosa was intact, with no ulceration or sinus discharge. On palpation, all the swellings were bony hard in consistency and nontender. There were clinically missing teeth 11, 15, 23, 34, 35, and 45.

To see the exact extent of the lesion, a panoramic radiograph was obtained which revealed impacted teeth w.r.t. 11, 15, 23, 34, 35, and 45. Diffuse radiopaque areas were seen in the maxilla and mandible. Surgical defect was noted on the left lower border of the mandible and a well-defined round radio-opacity was seen on the left sigmoid notch [Figure 1c].

Computed tomography (CT) scan showed multiple dense bony islands within the facial and skull bones. The obliteration of the right and left maxillary sinuses was noted. Diffuse radiopaque areas were seen causing expansion of maxilla and zygomatic bone. Dense bony exostoses were seen projecting from the palatal surface. Marked mediolateral expansion was seen on the mandible [Figure 2a-c]. Three-dimensional

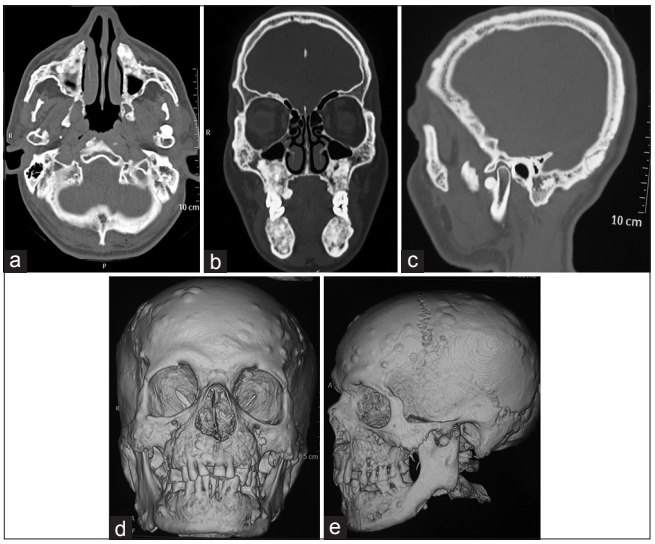

Figure 2: (a) Axial section shows near total obliteration of the right and partial obliteration of left maxillary sinus. Diffuse radiopaque areas are seen in the maxilla and zygomatic bones. Bilateral pterygoid plates show expansion. (b) Coronal section shows obliteration of the right and left maxillary sinuses. Diffuse radiopaque areas are seen which have caused expansion of maxilla and zygomatic bone. Dense bony exostoses are seen projecting from the palatal surface. Marked mediolateral expansion seen of the mandible is seen. (c) Sagittal sections of computed tomography scan show dense radio-opacities in the frontal and occipital bones. A well-defined dense round bony exostosis is seen attached to the left condyle. (d and e) Three-dimensional reconstruction on computed tomography scan showing multiple osteomas of frontal and parietal bones along with diffuse bony swelling of the maxilla and mandible. A well-demarcated osteoma of left sigmoid notch and a surgical defect on the left lower border of mandible is appreciated

CT images showed multiple small nodular swellings on the frontal, parietal, occipital, temporal, and sphenoid bones. Diffuse bony swellings of the maxilla and mandible were appreciated [Figure 2d and e].

Although the patient had no family history of major illnesses or disorders, based on our observations of multiple osteomas and the dental findings, he was advised to be evaluated by a gastrointestinal specialist for intestinal polyps to rule out Gardner's syndrome.

Within a week after our recommendations, Barium enema was done [Figure 3] which showed multiple colonic polyps and suspicious short segment narrowing in the sigmoid colon with irregular margins, which needed additional evaluation with colonoscopy. Colonoscopy was done and showed multiple sessile polyps all over the large intestine from rectum to ileocecal junction. To rule out malignancy, biopsy was taken from the polyp which revealed normal colonic mucosa with moderately dense chronic inflammatory infiltrates in the lamina propria. Based on this clinical, radiographic,

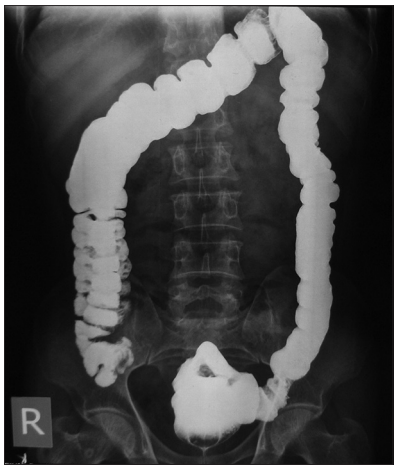

Figure 3: Barium enema shows enhancement of the entire large intestine with narrowing in the sigmoid colon with irregular margins

and histopathological evidence, a diagnosis of Gardner's syndrome was given.

Discussion

Gardner's syndrome belongs to spectrum of familial adenomatous polyposis. Intestinal polyposis is the most serious characteristic feature of this syndrome which may undergo malignant transformation. The polyps generally occur before puberty and around the second decade become generalized. The polyps are multiple in numbers and scattered in distribution, occur particularly in the distal colon, but may involve any location in the GIT. Passage of blood or mucosa, diarrhea, and cramp-like abdominal pain are the common presenting symptoms.[4] The present case also had a complaint of abdominal cramps.

One of the essential components of Gardner's syndrome is osteomas which vary from slight thickening to large masses and may affect all parts of the skeleton. Commonly affected sites include angle of the mandible, skull, and the paranasal sinuses. Clinical and radiographic evidence of colonic polyposis or Gardner's syndrome may be preceded by osteomas; therefore, they serve as an indicator for the disease.[3] In the present case, multiple osteomas of the skull and facial bones were appreciated.

Thirty percent of patients with Gardner's syndrome show various dental anomalies.[5] Commonly seen abnormalities are congenitally missing teeth, impacted teeth, hypercementosis, and supernumerary teeth. Other defects such as fused roots of the first and second molars, long and tapered roots of posterior teeth, dentigerous cyst, and multiple caries are also seen. Multiple impacted teeth were noted in our case.

Epidermoid cysts are the most common benign cutaneous lesions observed in this syndrome. They may occur on the face, extremities, and scalp and commonly seen around puberty. These cysts are usually asymptomatic; however, they may become inflamed. Adrenal adenoma, papillary carcinoma, hepatocellular carcinoma, adenocarcinoma, and osteosarcoma have also been documented.[6] No cutaneous manifestations were present in our case.

Desmoid tumors are locally aggressive but commonly do not metastasize. They account for 3.5% of all fibrous tissue tumors and 0.03% of all neoplasms.[7] The present case showed no evidence of desmoid tumors.

Incidence of multiple congenital hypertrophy of retinal pigment epithelium lesions has been associated with the presence and development of polyposis in Gardner's syndrome. This finding was not present in our patient.[8]

Management is mainly symptomatic with long-term follow-up. Colonoscopy should be advised to rule out development of malignancy in intestinal polyps. Surgical excision can opt for osteomas which are interfering with normal function or causing severe deformation. Dental managements may include extraction of the impacted teeth and enucleation or marsupialization of the jaw cysts. The patient may be referred to an ophthalmologist for evaluation of retinal anomalies. For esthetic concern, our patient was treated by surgical recontouring of the osteomas. Removable partial denture was fabricated for all the missing teeth and the patient is under regular follow-up for colonoscopy.

Conclusion

This case represents a classical picture of Gardner's syndrome which is usually overlooked by many medical and dental professionals. This can be very unfortunate as intestinal polyps in this condition have 100% chance of malignant transformation. Timely detection of dental, colonic, and extracolonic manifestations may help in halting this dreaded disease from further progression. After the diagnosis of Gardner's syndrome, the patients must be aggressively followed-up since there is a constant threat to their lives at any age.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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