

# Gardner's Syndrome? - It is Osteomas and Intestinal Polyposis: A Case Report

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## Abstract

The Gardner's syndrome is inherited as an autosomal dominant mode with 100% penetrance of colonic manifestation familial intestinal polyposis and variable expression of non-colonic manifestations. The early non-colonic manifestations are mainly expressed in and around dental and facial region, predominantly multiple osteomas of mandible, supernumerary teeth, and multiple impacted teeth. The purpose of this study was to report an 18-year-old female who presented with osteoma and multiple radiopaque lesions of the mandible that opened the window to recognize other disease complex of intestinal polyposis. Her family members were also subjected to colonoscopic examination but were found to be free from the disease. A radiographic examination of the patient's father, mother, and her brother showed no pathology in the jaw. To our surprise, there was no evidence of familial inheritance. This case seems to represent sporadic mutation in the presence of a negative family history.

**Key words:** Adenomatous polyposis coli gene, Dentigerous cyst, Familial intestinal polyposis, Gardner's syndrome, Hypercementosis, Osteomas

## INTRODUCTION

The disease complex of Gardner's syndrome was thought to include a triad of familial intestinal polyposis (FAP), soft tissue tumors, and osteomas of the mandible and skull when it was first described by Gardner's in 1951.<sup>1</sup> In view of the finding of a mutation in gene adenomatous polyposis coli (APC) located on the long arm of chromosome 5 in both FAP and Gardner's syndrome,<sup>2,3</sup> the latter was considered as a variant of FAP.<sup>2,4</sup> It is called Gardner's syndrome only when both (colonic and non-colonic) are present.<sup>4,5</sup> The non-colonic manifestations include osteomas (76–93%), skin cysts (53%), desmoids tumors (4–38%), congenital pigmentation of retinal epithelium (65%), and dental anomalies.<sup>2</sup> Of these non-colonic manifestations, dental anomalies are expected to be present in 70% of the affected individuals.

Although FAP was recognized as components of the disease complex described by Gardner's 1951, according to Merg *et al.* 2005, Devic and Busy, in 1912, Cobat, in 1935, and Fitzgerald, in 1943, have all described the association of colonic and non-colonic manifestations. Further, the propensity to malignant transformation of adenomas to carcinomas was recognized as early as 1887 and was histologically proved in 1890 by Smith and Handford, respectively. Since non-colonic manifestations precede many years before the development of FAP; dental surgeons have a high chance of recognizing the disease complex of Gardner's syndrome, which are crucial for the early surveillance of potential malignancy.<sup>6,7</sup> Although colonic polyps can be found in other syndromes, the presence of osteomas in a disease complex is regarded as the defining feature of Gardner's syndrome.<sup>2,8,9</sup>

## CASE REPORT

An 18-year-old healthy female presented with the chief complaint of a painless hard mass over the right side of the lower jaw over the posterior region. She was worried about the unesthetic appearance of the mass. Her dental and medical history was not contributory. The general

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**Month of Submission :** 03-2016  
**Month of Peer Review :** 04-2016  
**Month of Acceptance :** 05-2016  
**Month of Publishing :** 05-2016

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examination revealed no lymph node involvement in the neck. Extra-oral examination revealed a localized painless bony swelling over the right angle of the mandible without any signs of inflammation. Intraoral examination revealed no obvious pathology except for retained deciduous teeth (C, A, K & R). Retained deciduous tooth A was found to be carious. Basic investigations were done.

#### Panoramic Radiograph

It revealed retained deciduous teeth, multiple unerupted or impacted teeth, supernumerary, and third molar tooth germs. A discrete radio-opaque mass was noted at the right angle of the mandible. Diffuse but irregular radio-opaque masses were also found bilaterally in the mandible, extending between the lower border and the alveolar crest. The radio-opaque mass obliterated the lamina dura and displaced the inferior alveolar canal. Similar radio-opaque masses were also evident in the maxilla (Figure 1).

Although the osteomas, in the present case, were solitary, there were multiple radiopaque lesions in both the maxilla



**Figure 1:** OPG reveals multiple unerupted teeth, an enlarging osteoma seen in the angle of mandible, other multiple osteomas and other multiple irregular radiopacities



**Figure 2:** Swelling seen in the angle of the mandible due to an enlarging osteoma

and mandible, which was widespread in the mandible but sparing the ramus region.

Hematological investigation and serum chemistry reports were within normal limits. In view of osteomas and multiple unerupted or impacted teeth, she was referred to a gastroenterologist to exclude intestinal polyposis as part of Gardner's syndrome. Colonoscopic examination showed numerous benign intestinal polyps.

The questionnaire was asked to the family members and it was found to be completely negative. Further clinical, colonoscopic and radiological examination for any sebaceous cyst, abdominal pathologies, osteomas and impacted teeth were also found to be found negative in the family members. Ophthalmological examination was normal in both the patient and family members.

Based on the clinical and radiographic findings, the patient was provisionally diagnosed with multiple osteomas



**Figure 3:** Intraoperative view of the maxillary arch shows a retained deciduous canine C



**Figure 4:** Intraoperative view of the mandibular arch shows a retained deciduous mandibular molar K and deciduous canine R

associated with multiple impacted and supernumerary teeth.

### Treatment

The solitary osteoma was removed surgically for cosmetic reasons and the mass was found to be histologically consistent with osteoma. The other radiopaque lesions were left untreated. The patient was explained about the need for routine bi-annual follow-up (Figures 1-8).

## DISCUSSION

Osteomas are rare benign bone-forming by compact or cancellous bone. Depending on their location, they are called as endosteal tumors characterized and periosteal osteomas.<sup>10</sup> They are generally solitary when unassociated with syndrome, especially Gardner's syndrome. The latter osteomas are usually multiple and can affect any bone but have a predilection for the mandible, mainly the condyle.<sup>10</sup>



**Figure 5:** Intraoperative view of the maxillary arch shows a retained deciduous maxillary molar A which is carious



**Figure 6:** Intraoperative view of the osteoma during removal

They share both higher frequency of 76% to 93% and a lower frequency of 13.5% in Gardner's syndrome patients.<sup>2,11</sup> This case was brought to attention because of an enlarging osteoma. Interestingly, the osteomas develop during puberty and increase in number through adolescence.<sup>3</sup> The radiopaque lesions were not described by Gardner's, in 1953, in his seminal paper but a number of authors have drawn attention to their occurrence in patients with Gardner's syndrome.<sup>5,11,12</sup> These radiopaque lesions may begin from as early as 9 years to 55 years of age and may precede the development of intestinal polyps.<sup>12</sup> It occurs in 80% of cases and is of two types; a focal (62.2%) and diffuse types (13.5%).<sup>11</sup>

### Differential Diagnosis

Isolated radiopaque lesions may lead to confusion with florid osseous dysplasia or chronic sclerosing osteomyelitis, but negative signs of inflammation in conjunction with other features of Gardner's syndrome exclude these possibilities.<sup>5</sup>

The diagnosis of Gardner's syndrome is usually made when there is a positive family history or endoscopic evidence



**Figure 7:** The operated site on closure with sutures



**Figure 8:** Intestinal polyposis seen on colonoscopy

of intestinal polyposis.<sup>3</sup> The frequency of occurrence of familial adenomatous polyposis in a congenital mendelian dominant fashion was found to be 80% on an average while a remaining 20% of the cases represented spontaneous mutations, with no familial history.<sup>13</sup>

The main risk factor for developing Gardner's syndrome is having at least one of the parents with this condition. The wholesome management of the condition involves prescribing cyclo-oxygenase 2 inhibitor, close monitoring to make sure that the polyps do not become malignant and surgical intervention may be essential when these lesions become malignant. A protocol of proper nutrition, exercise and stress reduction therapy complements the treatment. The prognosis varies as there are high chances of colon cancer as they age and without the surgical treatment almost all the people with APC gene mutation are prone to colon cancer by the age of 40.

Although jaw manifestations in Gardner's syndrome are not in itself life threatening, the associated benign adenomas and their subsequent propensity for malignant transformation are a cause for concern. In this case, the adenomas were benign and were not treated at present but insisted on close monitoring of the patient.

## CONCLUSION

The present case was illustrated to emphasize the role of a dental care provider, who may be the first person to come across patients with the jaw lesions described here, to promptly refer the patient for subsequent clinical consultation to establish the correct diagnosis rather than to treat the patient for their jaw pathology alone. The early

diagnosis and intervention at multidisciplinary levels may serve a life-saving purpose to the patient as these jaw lesions serve as a tip of an iceberg of an underlying pathology.

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**How to cite this article:** Padmanaban SA, Saravanan R, Kavitha PS, Subramanian VA. Gardner's Syndrome? - It is Osteomas and Intestinal Polyposis: A Case Report. Int J Sci Stud 2016;4(2):278-281.

**Source of Support:** Nil, **Conflict of Interest:** None declared.