Case Report

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# Gardner's Syndrome with Significant Maxillofacial Cosmetic Deformity: A Rare Case Report

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**Abstract**

Gardners Syndrome, a common variant of familial adenomatosis polyposis affects one in approximately 8000 births. This syndrome is identified due to a distinctive triad of familial intestinal polyposis, surface tumours of hard tissue particularly osteomas in the skull and surface tumours of the soft tissue. The intestinal polyps have a 100% risk of undergoing malignant transformation. Hence, early detection of the disease is critical. This article discusses the surgical management of an unesthetic orbital osteoma in a particularly undiagnosed case of Gardners Syndrome.

**Keywords:** Gardners syndrome: Autosomal dominant; Polyposis; Osteomas; Soft tissue tumours.

## Introduction

Gardner's syndrome (GS), also known as 'familial colorectal polyposis,[1] is an autosomal dominant disorder localised to a small region on the long arm of chromosome 5(5q21-22).[2] It is characterised by adenomatous polyps of the gastrointestinal tract, desmoid tumors, osteomas (largely confined to skull bones), epidermoid cysts, lipomas, periampullary carcinoma and dental abnormalities. Less common features include hypertrophy of the pigment layer of retina, thyroid tumors and liver tumors.

Menzel first described adenomatosis of colon in 1721, and in 1863, Cripps discovered the heredity of colon polyposis and termed it familial adenomatosis.[3] Devic and Bussy first described GS in 1912 as a triad of intestinal polyposis, various soft tissue tumors such as fibromas, lipomas, neurofibromas, and epidermoid cysts, and multiple osteomas especially of the skull.[4] In 1951, Gardner reported the association between surface tumors and colonic polyps that are prone to malignant degeneration. In 1952, Gardner and Plenk described the dominant hereditary pattern of multiple osteomas associated with colonic polyposis. In 1953, the report by Gardner and Richards described the association of the multiple cutaneous and subcutaneous polyposis and osteomatosis. This completed the description of the clinical syndrome that has come to bear Gardner's name. In 1962 Gardner discovered the dental abnormalities and skeletal alterations in these patients.[5]

Dental annomalies include impacted or unerupted teeth, congenitally missing teeth, hypercementosis, dentigerous cysts, fused roots of the first and second molars, long tapered roots of posterior teeth, and multiple caries.[6] Osteomas are pathognomic of GS. The significance of this disease to the dentist is that the colonic polyps usually undergo malignant change by the fourth decade and the extra intestinal lesions may appear long before the bowel lesions. Thus, early detection of multiple jaw osteomas and/or multiple sebaceous cysts (particularly on scalp) may aid

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**Fig 1: Facial clinical picture showing exophthalmos of left eye.**

an early referral to a colorectal surgeon and might be life saving.

*Case Report*

A 22 year old male patient reported to the dental OPD in College of Dentistry, Indore in October 2009. He was referred for detailed evaluation to the oral and maxillofacial department. This patient reported with the chief complaint of severe pain and discharge from the left eye since last 15 days. He reported of forward displacement of the left eye since last 5 years and swelling in the left lower lid since last 10 years (Fig 1). The patient had no gastrointestinal complaints. However, the patient revealed that his father died of some unknown abdominal disease which was not properly investigated.

On further clinical examination, hard swelling of size 4x5 cm was observed in the left lower orbital region. A soft fluctuant cyst like swelling of size 6x7 cm was observed in the left high parietal region. Additionally, two firm small swellings of size 1x1 cm at right parietal region and two firm swellings of size

**Fig 2: Additional swelling**

**Fig 3: Ophthalmologic examination showing lenticular opacity and parapapillary Atrophy**

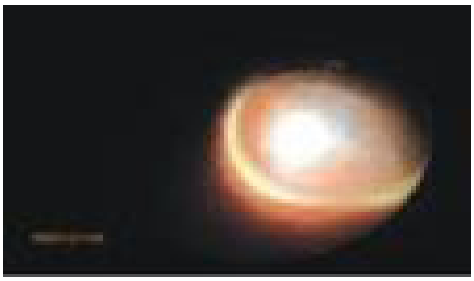


**Fig 4: Cropped Dental Panoramic Tomogram with Multiple unerupted teeth and cyst**

**Fig 5: CT face showing multiloculated osseous lesions in left maxillary sinus**

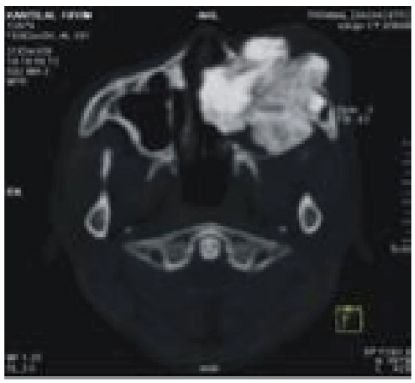


**Fig 6: MRI Head showing cystic scalp lesions**

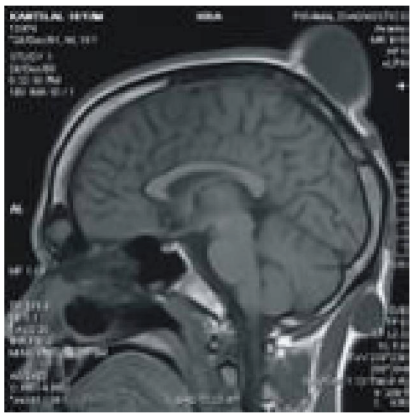


**Fig 7: Colonoscopy: multiple polyps**

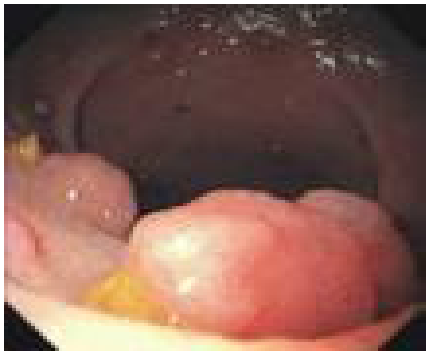


**Fig 8: Incision**

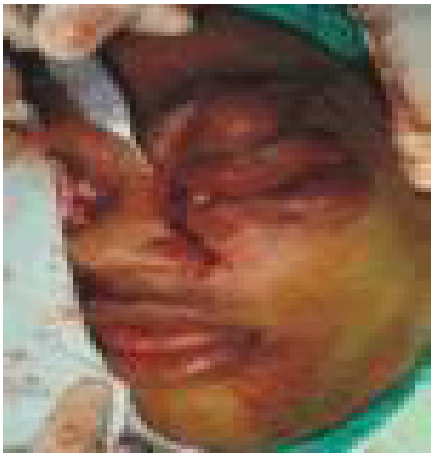
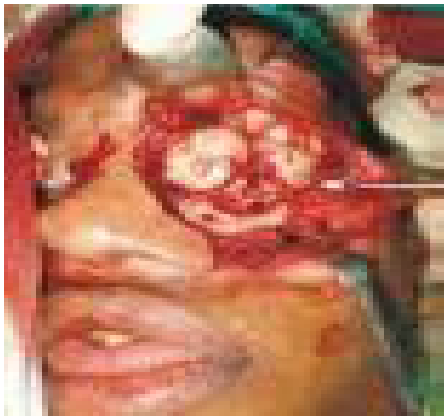


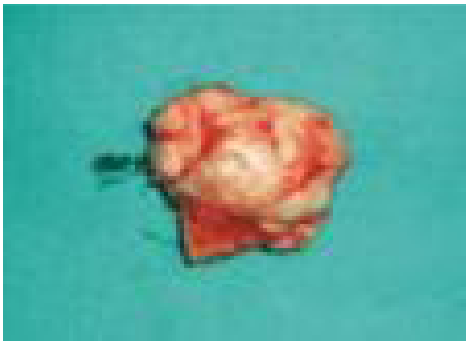
**Fig 9: Surgical Exposure of Osteoma**



2x2 cm were found on the right forearm (Fig 2). Per abdomen and per rectal examination were not significant. Opthalmological examination showed media hazy corneal opacity, lenticular opacity, para papillary atrophy (Fig 3).

The dental panoramic tomogram of this case showed multiple supernumerary and unerupted teeth associated with dentigerous

**Fig 10: Osteomatous mass**



cysts (Fig 4). Lateral view of the skull showed dense lobulated opacity over the left paraorbital region. The standard computed tomography (CT) of the face clearly showed multilobulated osseous lesion in left maxillary sinus protruding in left nasal cavity with a size of 6.5x6x5 cm. Multiple smaller osteomas were seen in left frontal sinus and bilateral ethmoid sinuses (Fig 5). The three dimentional reconstruction of CT showed large multilobulated mass seen along with the medial, inferior and lateral orbital wall of the left eye.

MRI of the head of the patient showed significant multiple focal well delineated cystic scalp lesions. Largest amongst them was in the right high parietal region of the size 7x6 cm (Fig 6). In order to exclude significant bowel pathology, he was subjected to x-ray and ultra sonography of abdomen which revealed no abnormal pathology. However, barium enema revealed multiple filling defects in sigmoid and splenic flexure. He was then referred for an urgent colorectal opinion. A colonoscopy was arranged which showed the

**Fig 11: Immediate post operative picture with no ophthamolgic complications**

**Fig 12: Six month post operative picture**

presence of multiple polyps of varying size throughout the large bowel (Fig 7).

The huge orbital osteoma was planned to be excised to give the patient a satisfactory esthetic result followed by management for polyposis coli. A Weber fergusson incision terminating inferiorly at ala nasi with infraorbital extension was used for direct approach to the orbital osteoma (Fig 8). A complete surgical exposure of orbital osteoma was achieved after thorough dissection (Fig 9). The complete excision of osteomatous mass was done with the help of chisels, burs and osteotomes (Fig 10).

Satisfactory esthetic results were obtained with no opthalmologic complications post operatively (Fig 11). Patient was then transferred to the department of general surgery for total proctocolectomy with ileoanal anastomosis. The patient was kept under close observation for any change in the head and neck lesions as well as bowel complaints. The six month post operative followup showed no recurrence at the operated site (Fig 12).

## Discussion

Gardner's syndrome, also known as familial colorectal polyposis is inherited as an autosomal dominant disorder with an average incidence of about 1 in 8000.[7] It is known to be caused by a mutation in the adenomatous polyposis coli (APC) gene located at band q21 on chromosome 5. Approximately, one third cases occur spontaneously and appear to represent new gene mutation.[8] It is characterised by colorectal adenomas, multiple osteomas especially of the skull, and various soft tissue tumors. The clinical and radiological features in the maxillofacial region such as osteomas, skin cysts, atypical skin pigmentation, and abnormal dental findings or radiographic lesions can preceed the asymptomatic adenomas which typically occur prior to puberty and turn malignant by the fourth decade.[9] Hence, dentists play a significant role in early diagnosis, treatment of mandibular or other facial osteomas, and management of impacted or unerupted teeth and cysts of jaw or face. Soft tissue lesions such as fibromas, neurofiromas, keloids, sebaceous cysts, leiomyomas and lipomas are common in this syndrome.[5]

Approximately 75% of the patients have congenital hypertrophy of retinal pigment epithelium (CHRPE), which is easily detected on opthalmoscopy. It is rare for normal individuals to have more than three of these lesions. Thus, if more than three such lesions are found, this is highly suggestive of GS.[10]

The clinical diagnosis of GS is difficult because of the great variation in the dental, bony, and cutaneous features. Some patients have only one or two abnormalities, whereas other patients display all or many of the characteristic features. Osteomas, the most benign major feature of GS are fortunately the first sign to be noticed by the patients or by the parents of an affected child. Dental abnormalities are present in 30%-75% of the cases and osteomas in 68%-82% of GS patients. [11] Dental findings may include impacted or unerupted teeth, congenitally missing teeth, supernumerary teeth, hypercementosis, dentigerous cysts, long tapered or fused molar roots, hypodontia, compound odontomas and multiple carious broken teeth.[4,9]

Our case is reported to illustrate the role of an oral and maxillofacial surgeon in the diagnosis of GS. The remarkable feature of this

patient was the unusually huge orbital osteoma of the left eye and early manifestation of head and neck lesions. Unlike most patients of GS who present with gastrointestinal symptoms such as bloody diarrhoea and pain in abdomen, our patient presented with dental and facial cosmetic symptoms. This patient could not reveal any relevant familial history, suggesting possible gene mutation in this case. Panoramic radiography showed the presence of multiple supernumerary and unerupted teeth in association with dentigerous cysts. Ophthalmologic examination showed corneal and lenticular opacity. All these symptoms in this patient were suggestive of GS which was confirmed in colonoscopy, that showed the presence of innumerable polyps of varying sizes throughout the large bowel.

Management of GS is a challenging task, especially the management of polyposis. It is suggested that, as soon as the diagnosis of colonic polyposis has been made, a total or partial colectomy should be performed with an ileoproctoscopy and if necessary, fulguration of the developing polyp may be the treatment of choice. Osteomas and soft tissue tumors may require excision if they are severly deforming or they interfere with function.

Complete excision of the huge orbital osteoma of the left eye was planned in our department as the patient was complaining of continuous pain and discharge from the left eye and it was interfering with the cosmesis of the face.

## Conclusion

In conclusion this patient was a typical case of GS suggesting possible gene mutation on chromosome 5 showing the characteristic oral and maxillofacial features in addition to polyposis coli. The patient showed no signs of recurrence of orbital osteomas or intestinal polyps during the regular follow up visits.

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