Case Report

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Gardner's Syndrome - A Case Report

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ABSTRACT

Background: Gardner's syndrome, a hereditary dominant condition, comprises of multiple osteomas, cutaneous and soft tissue tumors and polyposis coli. In 1951, Gardner reported the association between surface tumors and colonic polyps that are prone to malignant degeneration. It follows dominant hereditary pattern of multiple osteoma associated with colonic polyposis. The most commonly affected bones are the mandible and maxilla, followed by the frontal, sphenoid and ethmoid bones. The most common cutaneous finding in patients with Gardner's syndrome is epidermoid cysts, which tend to be numerous and are present in the multiple forms in 50 – 65% of the patients.

Keywords: Gardner's Syndrome, Hereditary dominant.

INTRODUCTION

Gardner's syndrome, a hereditary dominant condition, comprises multiple osteomas, cutaneous and soft tissue tumors and polyposis coli[1].

In 1951, Gardner reported the association between surface tumors and colonic polyps that are prone to malignant degeneration[2]. In 1952, Gardner and Plenk described the dominant hereditary pattern of multiple osteoma associated with colonic polyposis[2] and in 1953 the report by Gardner and Richards of the association of multiple cutaneous and subcutaneous lesions with hereditary colonic polyposis and osteomatosis completed the description of the clinical syndrome that has come to bear Gardner's name[2].

Less common features include hypertrophy of the pigment layer of the retina, thyroid tumors and liver tumors. The osteomas are largely confined to the skull bones. When long bones are involved, they show cortical thickening of their ends and are sometimes shortened and deformed. Rarely do osteomas arise from the phalanges.

The most commonly affected bones are the mandible and maxilla, followed by the frontal, sphenoid and ethmoid bones[3]. Hence, most patients present to dental surgeons with problems of dentition and for aesthetic considerations.

The most common cutaneous finding in patients with Gardner's syndrome is epidermoid cysts, which tend to be numerous and are present in the multiple forms in 50 – 65% of the patients. They may occur on the extremities, face, and scalp and may occur prior to or after the diagnosis of Familial Adenomatous Polyposis (FAP) or Gardner's syndrome.

Several factors differentiate cutaneous cysts associated with Gardner's syndrome from ordinary cysts. Firstly, these lesions occur at an earlier age than ordinary cysts, which occur around puberty. They also appear in less common locations, such as the face, the scalp and the extremities. Similar to epidermal inclusion cysts, the cysts in Gardner's syndrome are usually asymptomatic, however, they may become purulent and/or inflamed and may rupture[4].

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Fig 1: Showing swelling in in left lower orbital region.

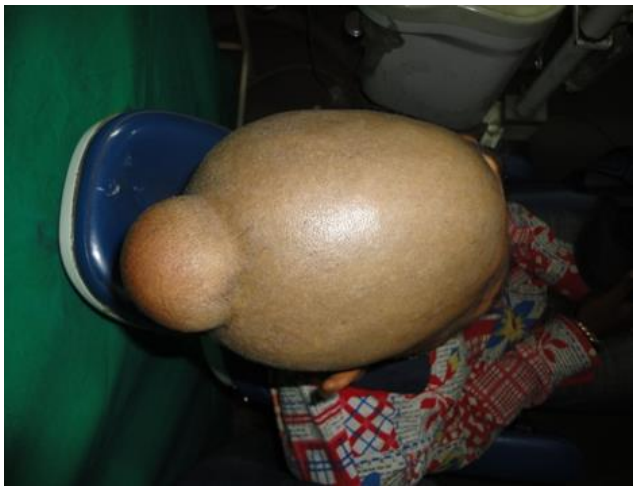

Fig 2: Showing swelling in left high parietal region.

Fig 3: Two firm small swelling of size 2x2 cm on right forearm.

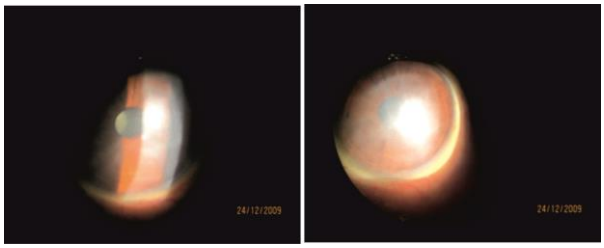

Fig 4: Media hazy, Corneal opacity, Lenticular opacity and Parapapillary atrophy.

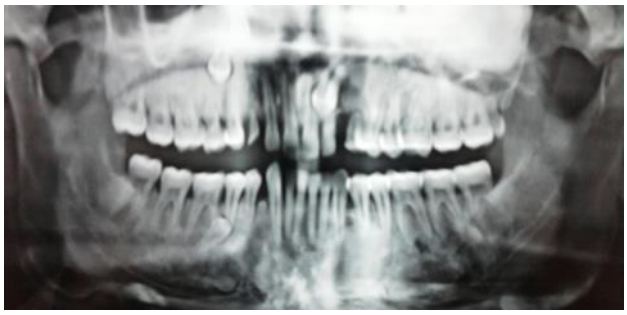

Fig 5: Dental Orthopantamogram.

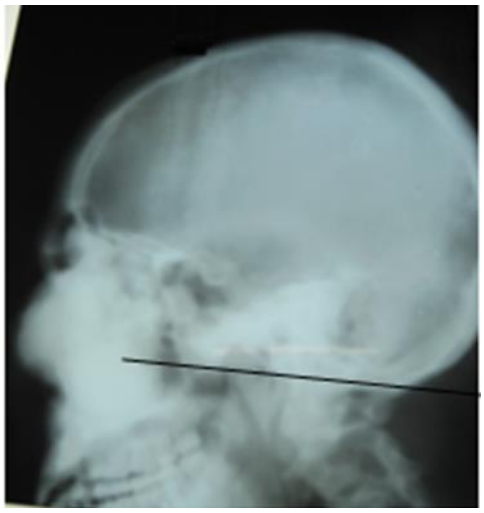

Fig 6: Showing Dense lobulated opacity over left paraorbital region.

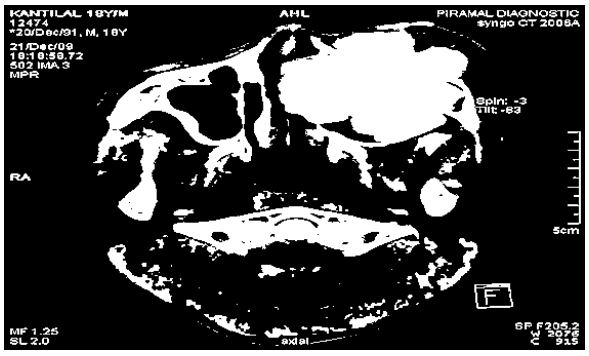

Fig 7: Multilobulated osseous lesion in left maxillary sinus protruding in left nasal cavity.

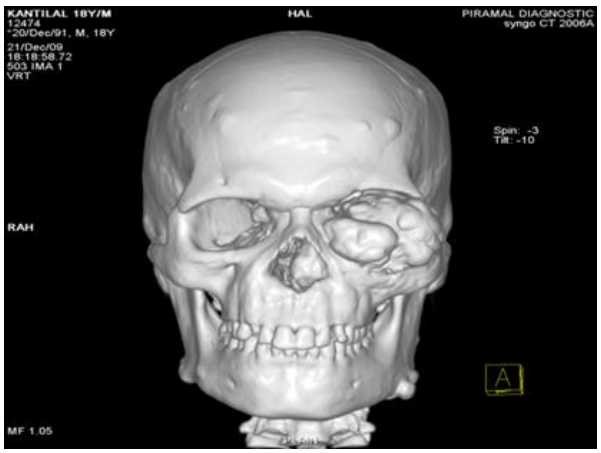

Fig 8: 3D CT Face showed large multilobulated mass seen along with left medial, inferior and lateral orbital wall.

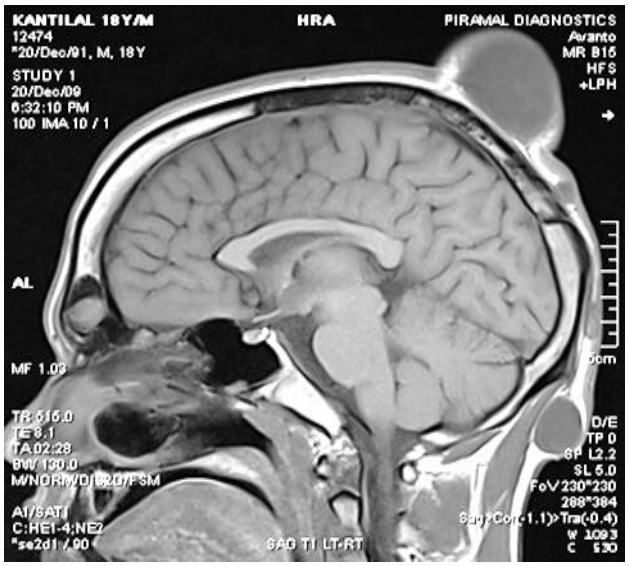

Fig 9: MRI head showed multiple focal well delineated cystic scalp lesions.

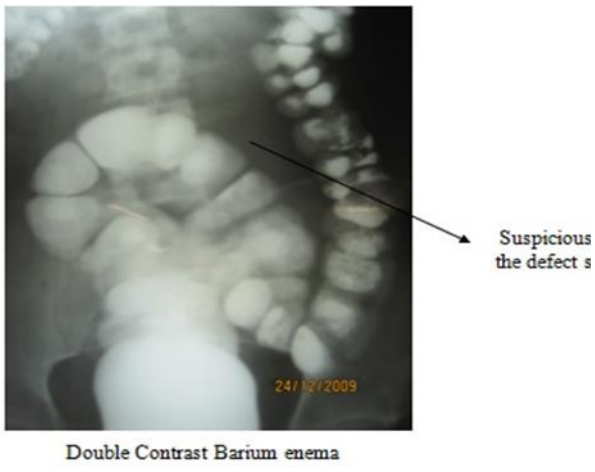

Fig 10: Barium Enema showed multiple filling defects in Sigmoid & Splenic flexure.

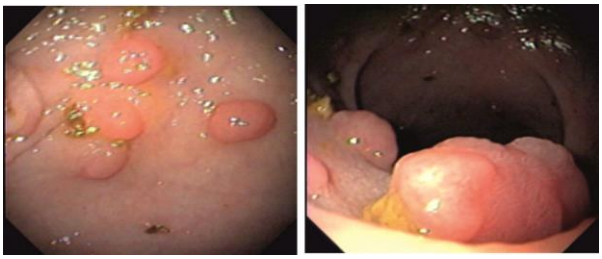

Fig 11: Multiple polyps of varying size throughout large bowel.

Other skin signs include presence of fibromas, lipomas, leiomyomas, neurofibromas, or pigmented skin lesions. Gastric fundic gland polyps occur in approximately 90% of affected individuals. Most of these lesions are hyperplastic and carry no malignant potential. However, adenomatous polyps and their progression to gastric cancer have been observed but is extremely rare[5].

Duodenal polyps occur in up to 90% of all affected individuals. If the polyps become very large, they may cause intestinal bleeding, intussusception or intestinal blockage. Otherwise, they may not cause any symptoms. Duodenal polyps have a predilection for the periampullary region and are premalignant lesions for periampullary carcinoma. The mortality rate for periampullary carcinoma in patients with Gardner's syndrome is approximately 20 – 25%[6]. The risk is 300 times higher compared with the general population[7]. Several cases of cholangiocarcinoma and FAP have been reported, which carries a more severe prognosis. Patients with periampullary lesions may present with abdominal pain, emesis, bleeding and gastric or biliary obstruction manifesting as jaundice. Duodenal polyps in persons with Gardner's syndrome have also been associated with pancreatitis secondary to obstructing the ampulla of Vater by the polyps. The intestinal polyps usually appear by the early to mid-teenage years. By age 35, almost all with this disease will have polyps.

CASE REPORT

A 22 years old male reported with chief complaint of severe pain and discharge from left eye from 15 days, swelling in left lower lid from 10 years, forward displacement of left eye from 5 years (Figure 1). No abdominal complaint was present. On examination a hard swelling of size 4x5 cm in left lower orbital region, cystic swelling of size

6x7 cm in left high parietal region was present (Figure 2). Two firm small swellings of size 1x1 cm in right paraorbital region and two firm small swellings of size 2x2 cm on right forearm were present (Figure 3). No abnormality was detected in per abdomen examination and per rectal examination.

Opthalmologic examination showed media hazy, corneal opacity, lenticular opacity, parapapillary atrophy (Figure 4). Dental OPG showed no involvement of tooth (Figure 5). Radiograph lateral view showed dense lobulated opacity over left paraorbital region (Figure 6).

CT scan of face showed multilobulated osseous lesion in left maxillary sinus protruding in left nasal cavity of size 6-5x6x5 cm. Mulitiple smaller osteomas were seen in left frontal sinus & bilateral ethmoid sinus (Figure 7).

3D CT scan of face showed large multilobulated mass seen along with left medial, inferior and lateral orbital wall (Figure 8). MRI head showed multiple focal well delineated cystic scalp lesions. Largest was in right high parietal region sized 7x6 cm (Figure 9). Barium enema showed multiple filling defects in sigmoid & splenic flexure (Figure 10). Colonoscopy showed multiple polyps of varying size throughout large bowel (Figure 11).

Treatment

Treatment planning for orbital osteomas was directed towards the complete excision of osteomas to give the patient a satisfactory esthetic results 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 osteomas. A complete surgical exposure of the orbital osteomas was achieved after thorough dissection. A complete excision of the osteomatous mass was done with the help of chisels, burs and osteotomes. Satisfactory esthetic results were obtained with no ophthalmologic complications postoperatively (Figures 12-16).

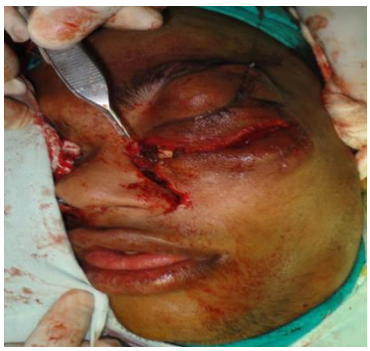

Fig 12: Incision Placed.

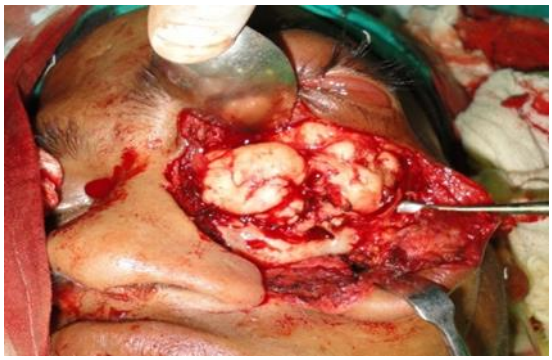

Fig 13: Dissection done to reach osteomas.

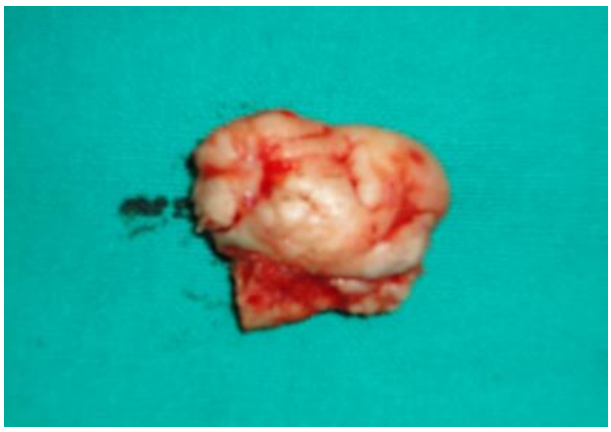

Fig 14: Osteomas removed.

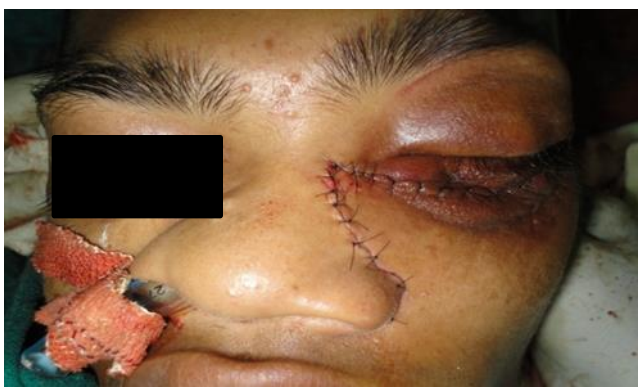

Fig 15: Tissue sutured.

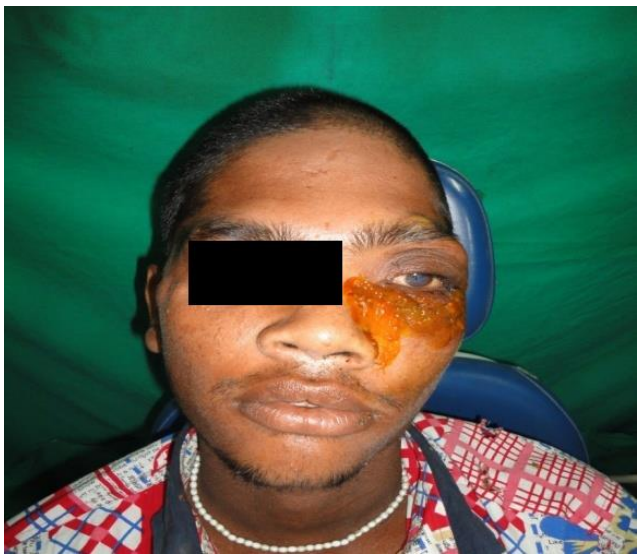

Fig 16: Five days post operative.

DISCUSSION

Osteomas, the most benign major feature of Gardner's syndrome are fortunately the first sign to be noticed by the patient or by the parents of an affected child. Gardner syndrome is known to be caused by a mutation in the adenomatous polyposis coli gene located at band q21 on chromosome 5. This is the same gene that is mutated in FAP, and GS is considered to be a clinical variation of FAP, with the presence of FAP being a marker for GS[8].

The clinical diagnosis of GS is difficult because of the great variation in the dental, bony and cutaneous features. Some patients have only 1 or 2 abnormalities, whereas other patients display all or many of the characteristic features. Dental abnormalities are present in 30%-75% of Gardner syndrome patients. An osteoma has been diagnosed in 68%-82% of FAP patients. Simultaneous presence of dental abnormalities with an osteoma is highly suggestive of underlying FAP–Gardner syndrome. Early detection of jaw osteomas and/or dental abnormalities by dentists could lead to further investigations and treatments of FAP and this could save patients' lives.

In Gardner syndrome intestinal polyps are predominantly adenomas and have a 100% potential for malignant change, which usually occurs in subjects who are 20-40 years old. Polyposis may be fatal if not treated.

SUMMARY

Management of Gardner's Syndrome is a challenging task, specially the management of intestinal polyposis. It is suggested that when the diagnosis of colonic polyposis has been made, a colectomy should be performed; total or a partial with an ileoproctostomy and if necessary, fulguration of the developing polyps may be the treatment of choices. Osteomas and soft tissue tumours may be excised without any danger of recurrence.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

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