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GIANT, MULTIPLE EPIDERMOID CYSTS IN GARDNER SYNDROME: CASE REPORT
N. A. Malungo, BScHB, MBChB, MMed-PRAS, FCS-ECSA, Plastic surgery, Moi Teaching and Referral Hospital, Kenya, ORCHID number 0000-0001-6168-3819 and **B. Wabwire**, MBChB, MMed, Fellow PRAS, Senior Consultant , Kenyatta National Hospital, Nairobi, Kenya, ORCHID number 0009-0005-2674-9540

Corresponding Author: Dr. N.A Mulongo, Plastic surgery, Moi Teaching and Referral Hospital and Kenyatta National Hospital, Nairobi, ORCHID number 0000-0001-6168-3819

GIANT, MULTIPLE EPIDERMOID CYSTS IN GARDNER SYNDROME: CASE REPORT

N. A. MALUNGO and B. WABWIRE

SUMMARY

Gardner syndrome is a rare genetic disorder within the familial adenomatous polyposis spectrum, characterized by intestinal polyposis, osteomas, and cutaneous tumours. Epidermoid cysts, its most frequent skin manifestation, often emerge early in life, serving as vital diagnostic clues.

We present a case of a 38-year-old male with a 30-year history of progressively enlarging skin swellings, lower back pain, dental issues, and blood-stained stool. The diagnostic workup was comprehensive. Clinical evaluation and histopathology were primarily used to assess the cutaneous lesions. To investigate gastrointestinal symptoms, a colonoscopy with multiple targeted tissue biopsies was performed. Extensive radiological imaging mapped skeletal anomalies, utilizing panoramic X-rays for the jaw, 3D CT reconstructions of the skull, an abdominal-pelvic CT scan, and radiographs of the long bones. Baseline laboratory tests, an abdominal ultrasound, and ophthalmoscopy were additionally conducted to rule out other extracolonic manifestations.

Histopathological analysis confirmed the skin lesions as giant epidermoid cysts, including a debilitating 15x10cm cyst on the lower back. Colonoscopy identified over 30 benign adenomatous polyps spanning the colon. Imaging revealed multiple cranial osteomas, mandibular sclerosis, and hypodontia. Blood tests, thyroid ultrasound, and retinal exams returned normal results. Interventions included staged surgical excisions of the cysts, psychosocial counselling, and a referral to general surgery for a prophylactic laparoscopic pan-colectomy.

In conclusion, multiple epidermoid cysts are crucial early indicators of Gardner syndrome. Prompt recognition enables vital gastroenterological interventions—such as prophylactic colectomy—to prevent colonic malignancy, while surgical management of the cysts directly addresses the patient's physical and psychosocial burdens.

INTRODUCTION

Epidermoid cysts are benign encapsulated, keratin-filled lesions occurring as sub-epidermal nodules (1). They are generally slow-growing and rarely undergo malignant transformation, with the most common location being on the trunk, face, neck, and genitals (2). However, cysts in the CNS and abdomen have been reported (3)

Epidermoid cysts can complicate by becoming infected, rupturing, or may exert pressure on underlying structures(1). A giant epidermoid cyst is defined as being more than 5cm in any dimension(1). Multiple epidermoid cysts are highly suggestive of a genetic cause. Syndromes associated with multiple epidermoid cysts include Gardner syndrome, basal cell nevus syndrome, Lowe syndrome, and pachyonychia congenital (1).

Gardner, in 1953, described a syndrome consisting; of intestinal polyposis, osteomas, and multiple cutaneous and subcutaneous tumors (4). Patients may also present with dental and retinal pigment epithelium hypertrophy (5).

We report a rare case of Gardner syndrome in a 38-year-old male who presented to the Kenyatta National Hospital (KNH) plastic surgery clinic with giant, multiple epidermoid cysts, numerous intestinal polyps, and multiple craniofacial osteomas.

CASE PRESENTATION

A 38-year-old African male presented to the plastic surgery outpatient clinic with complaints of multiple body swellings for 30 years, associated with lower back pain for 7 years, loss of multiple teeth, and lower jaw discomfort for 8 years. He also reported altered bowel habits, which included constipation and occasionally blood-stained stool.

The first skin lesion was noted on the scalp at the age of 9 at the occiput as a single swelling. It was non-tender, soft, progressively increasing in size, not associated with any discharge, non-itchy, and was mobile. This was treated with surgical excision. Histology was not available from the excised specimen.

By the age of 13, more similar swellings developed over the rest of the scalp and trunk. The swellings on the trunk were more prominent and were associated with lower back pain that he described as dull, non-radiating, worse at night, and was relieved by analgesics.

He had been to the dentist severally for his loose teeth that had an irregular alignment. He had previous extractions for impacted teeth. He was also treated for periodontal disease.

He had no known chronic illness, and a review of his history revealed no history of significant skin, colon, or thyroid tumors in the family.

He was unemployed at presentation, having lost his job 3 years prior. He had low self-esteem due to the huge swellings that made formal dressing a challenge.

Clinical evaluation noted the multiple skin lesions, later confirmed with histopathology to be epidermoid cysts. Five cysts were noted on the scalp, the largest measuring 3 x 5cm. Lesions were present on the anterior and posterior trunk, as shown below.

Figure 1: Multiple epidermoid cysts on the posterior trunk. Arrows highlight the puncta over the smaller lesions.

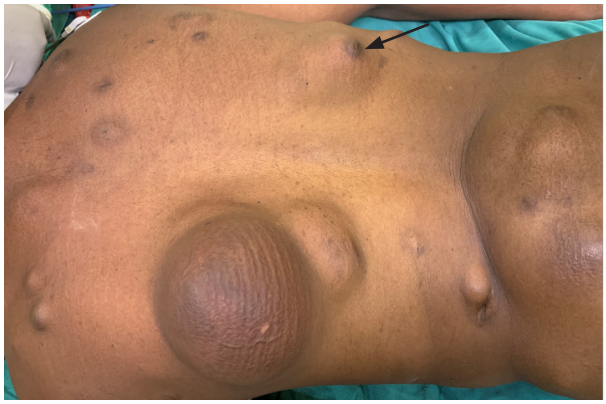


Figure 2: Posterior trunk epidermoid cysts on Posterior and lateral view. Appreciate the projection and size of the giant cysts

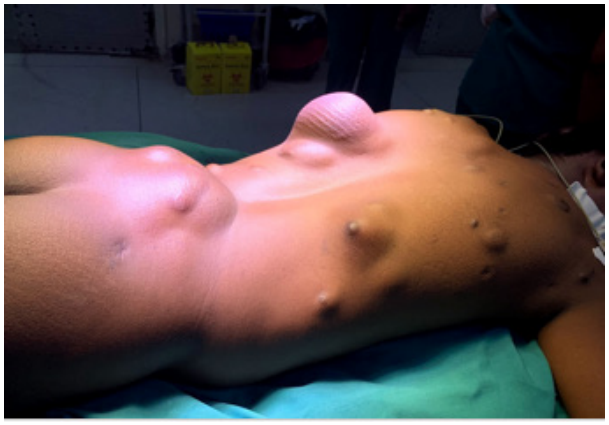
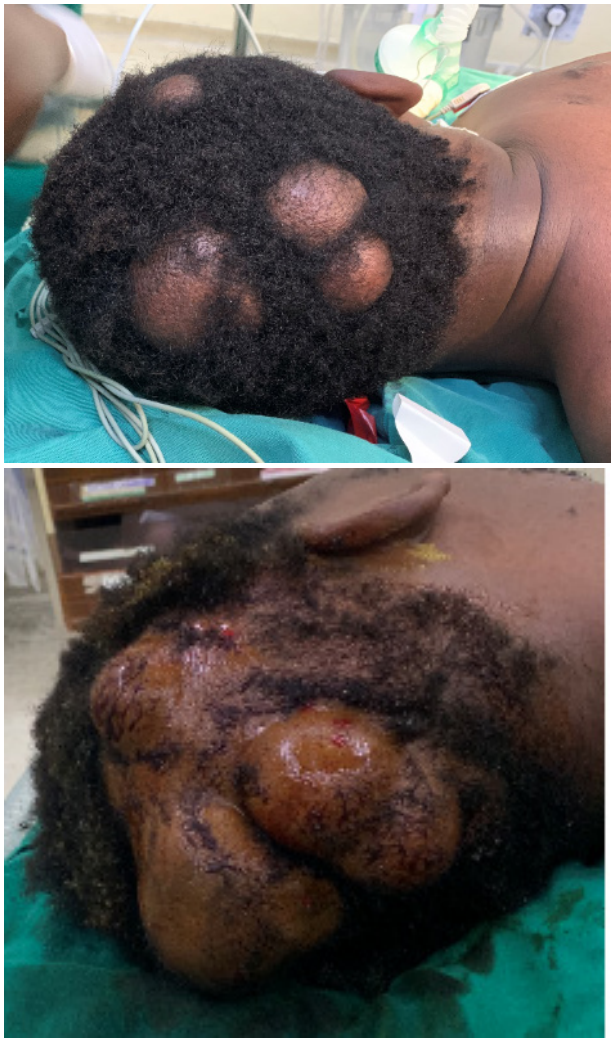


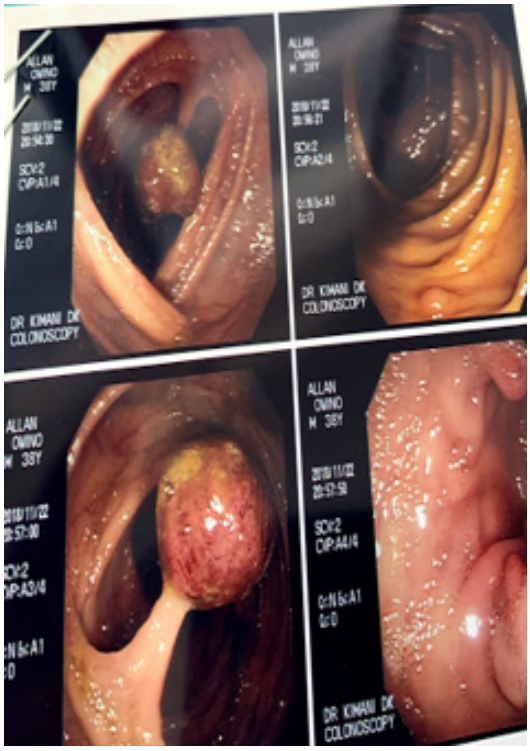
Figure 3: Scalp cysts predominantly in the occipital region



The oral cavity had poor oral hygiene, loose teeth with hypodontia, and malocclusion class. General and systemic examination was unremarkable. However, a colonoscopy revealed multiple polyps (>30 polyps)

extending from the ascending colon to the rectum, and biopsies were taken. Histopathology showed benign adenomatous polyps.

Figure 4: Multiple polyps in the colon at various stages of development



Laboratory blood works- Full hemogram, Kidney function, and liver function tests were all normal. The abdominal ultrasound and radiographs of the long bones were unremarkable.

Figure 5: Panoramic image shows relatively well-circumscribed multiple opacities in both the maxilla and mandible. Diffuse sclerosis of the mandible was seen.

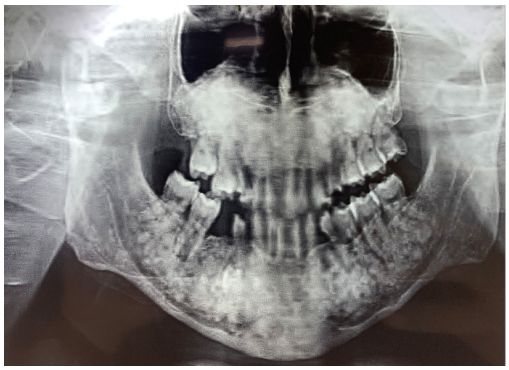
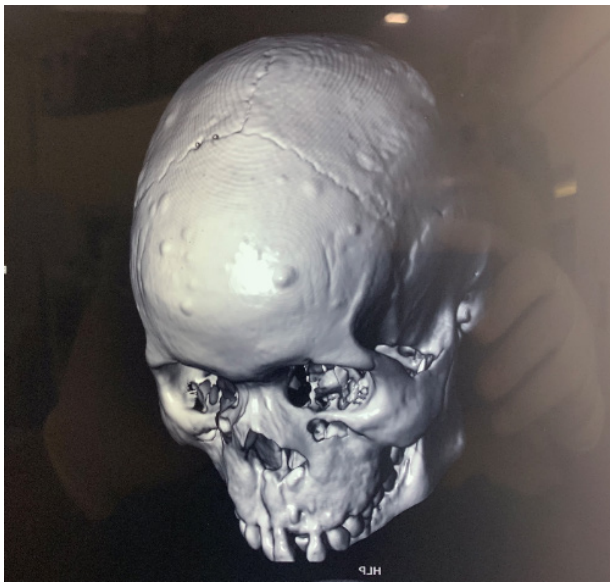
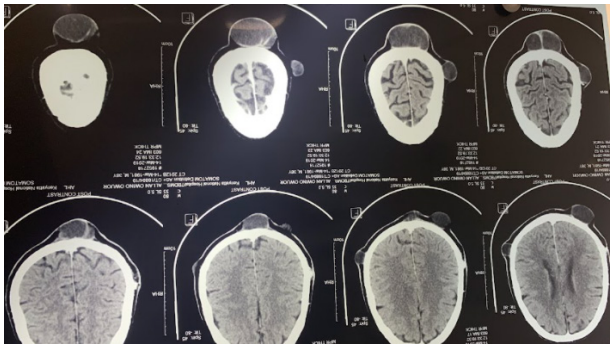


Figure 6: Multiple skull osteomas on CT 3D reconstruct





Ophthalmoscopy was normal.
Additional tests CEA, abdominal pelvic CT scan, and thyroid ultrasound, were otherwise normal.

Treatment
The cysts on the anterior and posterior trunk were excised. Scalp cysts were excised in subsequent operations. The patient was referred to the general surgeons for Laparoscopic pan-colectomy and end ileostomy. Psychosocial counseling was given. Follow-up for screening of other family members is planned as one of his sons has a soft tissue lesion on his trunk.

Figure 7: Residual pedunculated epidermoid cyst on the anterior trunk after selective excision on the posterior trunk

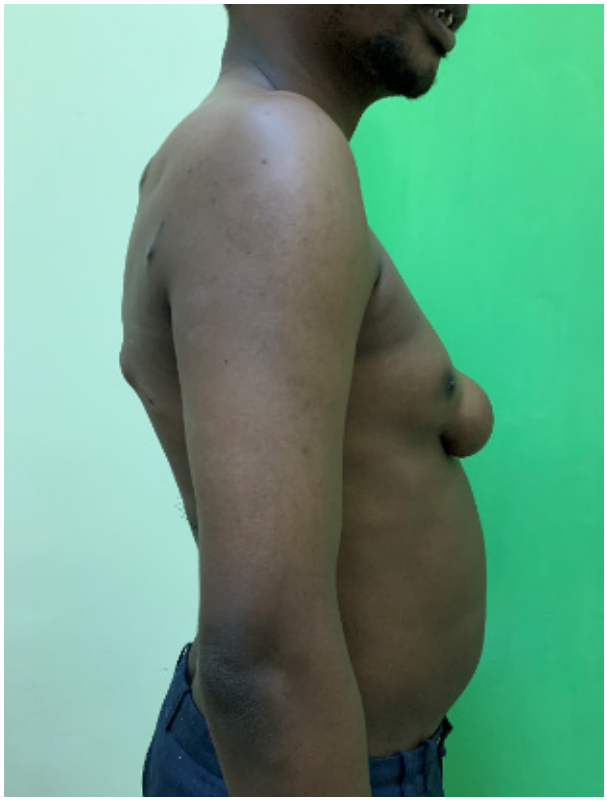
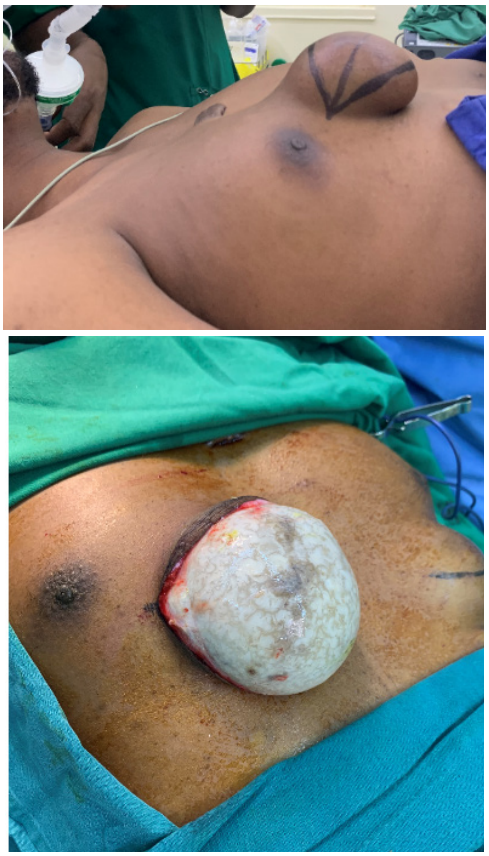


Figure 8: Before and after excision on the anterior trunk



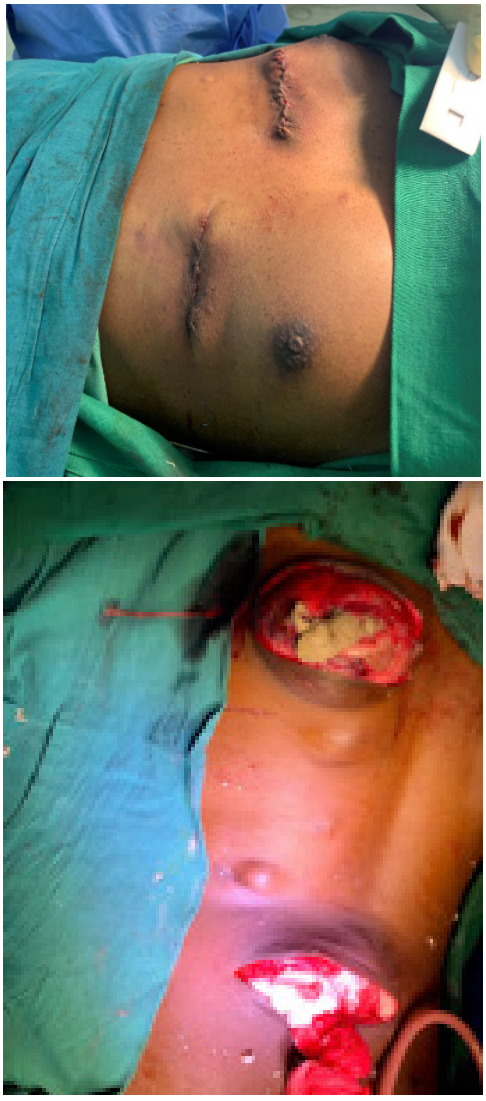
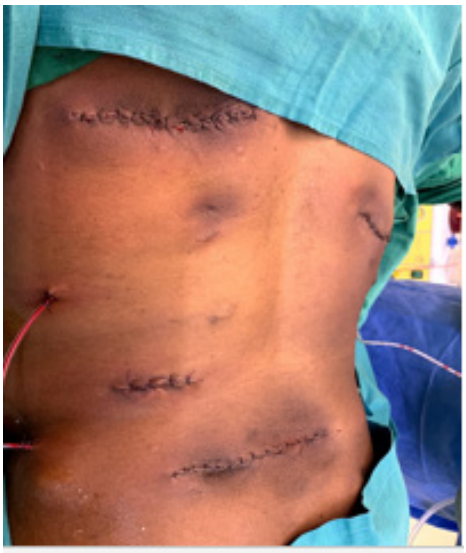


Figure 9: Before and after selective excision of cysts on the posterior trunk



DISCUSSION

Gardner syndrome is a relatively rare genetic disorder with a reported prevalence of one in a million and an incidence of 1 in 8000 (6). Current consensus recognizes Gardner syndrome as one end of the spectrum of Familial adenomatous polyposis (FAP) disorder(6)

The presence of multiple colonic polyps in Gardner syndrome makes early diagnosis important because of malignant change in the polyps by the fourth decade(3). In this report, the patient developed intestinal symptoms by the third decade of life. A colonoscopy was requested based on the clinical presentation of multiple epidermoid cysts and his vague abdominal symptoms. A prophylactic colectomy is indicated in these patients once the diagnosis is made. Patient counseling for colectomy and family genetic screening should ideally be done.

The frequency of extracolonic symptoms varies. Epidermoid cysts are the main cutaneous symptom with a frequency of about 65%. These usually arise around puberty, as reported in this case(4). Though they have little malignant potential, they can undergo spontaneous rupture, infection, or cause distortion due to pressure or displacement of local tissues(2). Since the cysts present relatively earlier in life, they are important in the early diagnosis of the condition. In the present case, the patient presented with lower back pain due to a large giant cyst of about 15x10cm lying over the mid-lower trunk and sacrum. In addition to these pressure symptoms, social embarrassment and loss of employment were a cause of distress since the cysts were continually growing in size.

Desmoid tumors exist in less than 10% of patients (7). These are benign, slow-growing fibromatoses lying in the deeper tissues. No desmoid tumors were noted in this patient. Similar to epidermoid cysts, desmoid tumors rarely undergo malignant change but have the potential to cause local destruction. Fibromas, lipomas, leiomyomas, pigmented lesions, and basal cell carcinoma can occur in Gardner syndrome(4)

Osteomas occur in 68-82% of patients, the commonest site being the mandible. Approximately a third of patients will report dental anomalies such as absent or supernumerary teeth, as is the present case (4,5). The osteomas may be clinically evident as exostoses (peripheral osteomas) or only detectable radiologically as enostoses when they develop on the endosteal surface of the cortex into the cancellous bone(8). In the present case, the patient had exostoses on the cranial bones, predominantly enostoses in the mandible and maxilla, and dental abnormalities. Though not detected in the present case, Traboulsi *et al*. reported up to 85% of patients to have congenital hypertrophy of the retinal pigmented epithelium (CHRPE)(3).

Gardner syndrome has no known cure. Surgery is the primary mode of management for Colonic polyps and soft tissue tumors. A healthy diet and certain non-steroidal anti-inflammatory drugs can reduce the growth rate of intestinal polyps. Other syndromes like Turcot syndrome may mimic Garner syndrome, but the skin lesions in the former are predominantly café-au-lait spots(6).

CONCLUSION

Epidermoid cysts are the commonest cutaneous manifestation of Gardner syndrome. They are slow-growing, keratin-filled lesions with very low malignant potential. Epidermoid cysts and other extracolonic symptoms often precede colonic symptoms making early diagnosis of the condition possible. Patients presenting with these lesions should be referred for further gastroenterological evaluation and counseled for prophylactic colectomy due to the high risk of colon adenocarcinoma. Despite the perceived benign nature, multiple epidermoid cysts in these patients can complicate and cause social isolation and loss of income.

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