CASE REPORT

External manifestations of Gardner's syndrome as the presenting clinical entity

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SUMMARY

Gardner's syndrome is an autosomal dominant disease characterised by the presence of colonic polyposis, osteomas and a multitude of soft tissue tumours. Pathological features such as osteomas of the mandible, skull and facial skeleton are unaesthetic as well as incapacitating. We present the case of a 22-year-old man with pain and discharge from the left eye and a firm swelling in the left infraorbital region leading to proptosis of the left eye. A detailed examination of the patient led to the presence of a large osteoma in the left orbital region, multiple cystic lesion, corneal opacity and parapapillary atrophy in the left eye. Radiography revealed the presence of multiple unerupted supernumerary teeth and osteomas. Colonoscopic findings showed the presence of multiple polyps. Thus, external manifestations of the patient's facial region led to the establishment of the diagnosis of Gardner's syndrome. The importance of our case highlights the necessity of maintaining a high vigilance with regard to the occurrence of such an entity.

BACKGROUND

Gardner's syndrome (GS) is well studied but an uncommonly encountered entity in surgical practice. It is supposed to be a clinical variant of familial adenomatous polyposis (FAP)¹ comprising multiple intestinal polyps, dental and soft tissue

abnormalities,²⁻⁴ bony pathological changes along with debilitating ophthalmological involvement. An almost 100% malignant transformation of its intestinal lesions is well known which, if left untreated, can prove to be fatal. In addition, its other pathological features such as osteomas of the mandible, skull, facial skeleton, sebaceous and epidermoid cysts of the back and neck are unaesthetic as well as incapacitating. Most debilitating manifestation of this condition is the visually handicapping lesion which is not only a cosmetic blemish but also a perimetric hindrance for the patient. As the bony and dental signs of GS usually precede the gastrointestinal symptoms, the dentist may play an important role in the diagnosis of FAP. In lieu of this, we report such an uncommon instance where a young adult male suffering from this condition reported to us, after 15 years of its onset.

CASE PRESENTATION

A 22-year-old illiterate farmer who was psychologically subdued due to social ridicule reported to our hospital in January 2010 with reports of severe pain and discharge from his left eye since the past 15 days. Further detailed history revealed a progressively increasing hard swelling in his left lower lid since 10 years associated with proptosis, since 5 years.

Figure 1 Preoperative photographs. (A) Large osteoma on the left side causing displacement of the eyeball, (B) sebaceous cyst at lateral canthus of the right eye and (C) bird's eye view showing proptosis of the left eye.

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Figure 2 (A and B) Slit lamp examination showing corneal opacity.

On general examination an irregular, hard lesion of 4×5 cm was present in the left inferior orbital region (figure 1A–C), a cystic swelling of 6×7 cm in the left high parietal region and another similar one of 3×4 cm in the lower lumbar region. In addition, two firm swellings of 1×1 cm in the right paraorbital region and a 2×2 cm in the right forearm were concomitantly present.

On ophthalmological examination, corneal opacity and papillary atrophy was present (figure 2A, B). On the basis of these findings, a presumptive diagnosis of GS was made. Moving on the same track the per abdominal examination was found to be normal and on repeated probing no abnormality was detected apart from a history of his father's death due to some unknown abdominal condition.

Figure 3 Panoramic image showing multiple unerupted and impacted supernumerary teeth.

INVESTIGATIONS

In panoramic view, findings of multiple unerupted and supernumerary teeth were found (figure 3). Three-dimensional CT of the face revealed multilobulated osseous lesions in the left orbital and infraorbital region (figures 4A, B). Multiple smaller osteomas were seen in the left frontal sinus and bilateral ethmoid sinuses. MRI findings demonstrated multiple focal well-delineated scalp lesions predominantly appearing cystic. The largest one was seen in the high parietal region and other smaller similar scalp lesions were seen in the right temporal left half of the midline occipital region and over the right parieto-occipital region (figure 5).

Barium enema showed multiple filling defects in the sigmoid and splenic flexure (figure 6). Colonoscopic findings presented with multiple polyps of varying size throughout the large bowel along with a few bleeding polyps as well (figure 6A).

TREATMENT

Treatments planning for orbital osteomas were directed towards the complete excision of osteomas. Satisfactory aesthetic results were obtained with no incidence of recurrence (figures 7A–C). The patient was then referred to the department of general surgery for the management of the polyposis coli and is maintained on regular follow-ups.

DISCUSSION

Gardner⁵ in 1951 reported the significant correlation between the external osseous and cystic tumours and polyposis coli, which has high malignant propensity. Subsequently, in the next year Gardner and Plenk⁶ reported that the occurrence of multiple osteomas along with polyposis coli has a dominant hereditary pattern of inheritance. In 1953 Gardner and Richards⁷ gave the final description of GS, that is, hereditary colonic polyposis associated with osteomatosis and multiple cutaneous and sebaceous tumours.

Mutation of the APC (adenomatous polyposis coli locus) gene located on band q21 on chromosome 5 (5q21) causes GS and another more common disease predisposing to colon cancer—FAP. More than 1400 different mutations of this gene have been reported. Owing to this variable expression of APC gene mutations associated with GS and FAP, a wide range of phenotypes are observed clinically.

GS is a disease that affects multiple systems. Symptoms are usually presented by the end of second decade of life, but they may present anytime between 2 months and 70 years. In our case, the patient reported in the early third decade of his life but history revealed the presence of progressively increasing firm swelling in the left eye since 12 years of his life. In total 68–82% of FAP patients^{8–11} have osteomas present as one of the manifestations. The epidermoid cysts are considered to be the most common cutaneous findings in GS and may occur anywhere on the face and scalp and less commonly on the extremities. In total 50–65% patients have multiple cysts. Cysts are asymptomatic usually resembling ordinary epidermal cyst. On occasions they may be inflamed, may be pruritic and may rupture. Pathological findings in these cysts are usually similar to those in non-GS cysts. However, some may present with pilomatricoma-like changes.¹² Other cutaneous findings include fibromas, lipomas, neurofibromas or leiomyomas. Fundus may present with multifocal pigmented lesions in 80% of patients, manifesting shortly after birth. Thus, these may be the first sign

Figure 4 (A) CT scan showing large osseous lesion measuring about $6.5 \times 6 \times 5$ cm in the left maxillary sinus and displacement of left nasal cavity. (B) Three-dimensional reconstructed CT scan showing large osteoma filling the left orbital content.

of the disease. The ophthalmological finding includes hypertrophy of the pigmented layer of the retina. In 30–75% of GS patients dental abnormalities are present.^{8,9} Dental findings include multiple supernumeraries, malerupted and unerupted teeth sometimes associated with cystic lesions as well. Simultaneous presence of dental abnormalities with an osteoma is highly suggestive of the underlying FAP-GS. So in this manner a dental surgeon could be the first person to scrutinise the case

of GS and could lead to relevant investigations and the management of FAP. Our case presented with all these pathognomonic signs of GS and essentially a large osteoma with left orbital region that demanded earliest surgical correction.

Treatment of GS is essentially a multidisciplinary effort. Prophylactic abdominal colectomy with ileorectal anastomosis may be a treatment option for polyposis. Proctosigmoidoscopy followed by complete excision and fulguration of polyps may be

Figure 5 (A) MRI of the brain showing large multilobulated lesion in the left maxillary sinus protruding into the left orbit causing severe proptosis. (B) Large cystic lesions in the parietal region measuring 7×6 cm and small lesion in the supraoccipital scalp region.

Figure 6 (A and B) Endoscopic examination revealed the presence of multiple intestinal polyps.

Figure 7 (A) Extended infraorbital incision. (B) Multilobulated large osteoma exposed. (C) The patient on the seventh postoperative day with restored eyeball position. (D) The patient at 2-month follow-up.

an alternative. Serial colonoscopy and upper endoscopy should be done for at least 1–2 year to exclude recurrence. Osteomas may be treated by peripheral ostectomy and osteoplasty for functional as well as cosmetic repair. Desmoids tumours treated by antioestrogens and anthracycline-containing regimens show favourable radiological response in some patients. Odontectomy

of supernumerary teeth to allow eruption of permanent teeth combined with orthodontic treatment for establishing functionally and aesthetically acceptable occlusion is the treatment of choice to treat dental problems. Serial orthopantomograms are observed for 6–12 months to monitor osteomatous change of mandible, if any.

Learning points

- ▶ Gardner's syndrome is well studied but uncommonly encountered in surgical practice, comprising multiple intestinal polyps, dental and soft tissue abnormalities, bony pathological changes along with debilitating ophthalmological involvement.
- ▶ However, only ophthalmological involvement as an external manifestation, leading subsequently to the diagnosis of Gardner's syndrome, is a rare occurrence.
- ▶ The importance of our case highlights the necessity of maintaining a high vigilance with regard to the occurrence of such an entity, and subsequently guarded management for the same.

Competing interests None.

Patient consent Obtained.

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