Familial craniofacial osteomas: a diagnostic challenge

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ABSTRACT

Background: Osteomas are benign, slow-growing bony tumors that usually occur as solitary, incidental findings. When multiple craniofacial osteomas are present, Gardner syndrome (GS), an Adenomatous Polyposis Coli-associated hereditary disorder, must be considered. Craniofacial osteomas may precede intestinal polyposis by several years, making them important early diagnostic indicators. However, attenuated GS and sporadic familial osteomas can present with similar features, creating a diagnostic dilemma. Early identification, genetic counselling, and surveillance are therefore essential to exclude delayed gastrointestinal or extracolonic manifestations.

Case presentation: We report two first-degree male relatives with multiple craniofacial osteomas but no systemic features. The proband, a 15-year-old boy, presented with mandibular, palatal, and paranasal swellings that were radiologically and histopathologically confirmed as compact osteomas. His 52-year-old father also exhibited multiple craniofacial osteomas, including mandibular and sinus involvement, but remained asymptomatic. Comprehensive systemic evaluation, including colonoscopy, abdominal imaging, dermatologic, and ophthalmologic examinations, was negative for intestinal polyposis or extracolonic manifestations.

Conclusion: These familial cases highlight the diagnostic dilemma between attenuated GS and sporadic familial osteomas. Genetic testing, counselling, and long-term surveillance are essential to exclude delayed gastrointestinal or systemic involvement.

Keywords: Craniofacial osteomas, Gardner syndrome, attenuated phenotype, familial osteomas.

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Introduction

Case presentation

Case 1

A 15-year-old male presented with a painless swelling in the left lower jaw, first noticed seven months earlier. The swelling was asymptomatic and gradually increased in size, with no history of trauma, gastrointestinal symptoms, or systemic illness. Extraoral examination revealed a diffuse swelling over the left mandibular angle region, resulting in mild facial asymmetry (Figure 1a,b).

On palpation, the swelling was bony hard, non-tender, non-compressible, and immobile. No regional lymphadenopathy was detected. Mouth opening was adequate and within normal limits.

Intraoral examination revealed no obliteration of the buccal vestibule in the left mandibular region, and the overlying mucosa appeared normal. A partially impacted 37 was noted. In the maxillary arch, a well-defined solitary bony swelling was present in the rugae–palatal region adjacent to teeth 14 and 15, measuring approximately 1.5 × 1.5 cm. The swelling did not cross the midline. It was bony hard and non-tender on palpation, with intact mucosa and no ulceration or discharge (Figure 1c-e).

Imaging

A panoramic radiograph revealed an irregular, predominantly radiopaque focus at the left mandibular angle, apical to tooth 37. Multiple irregularly shaped radiopaque masses were also visible within the right and left mandibular body, abutting the roots of teeth 46, 34, 35, and 36, as well as the erupting 46. Additionally, similar radiopaque foci were observed superimposed over the condylar and coronoid processes. All lesions appeared homogeneous, dense, and sharply demarcated from the surrounding bone, with no evidence of cortical destruction, periosteal

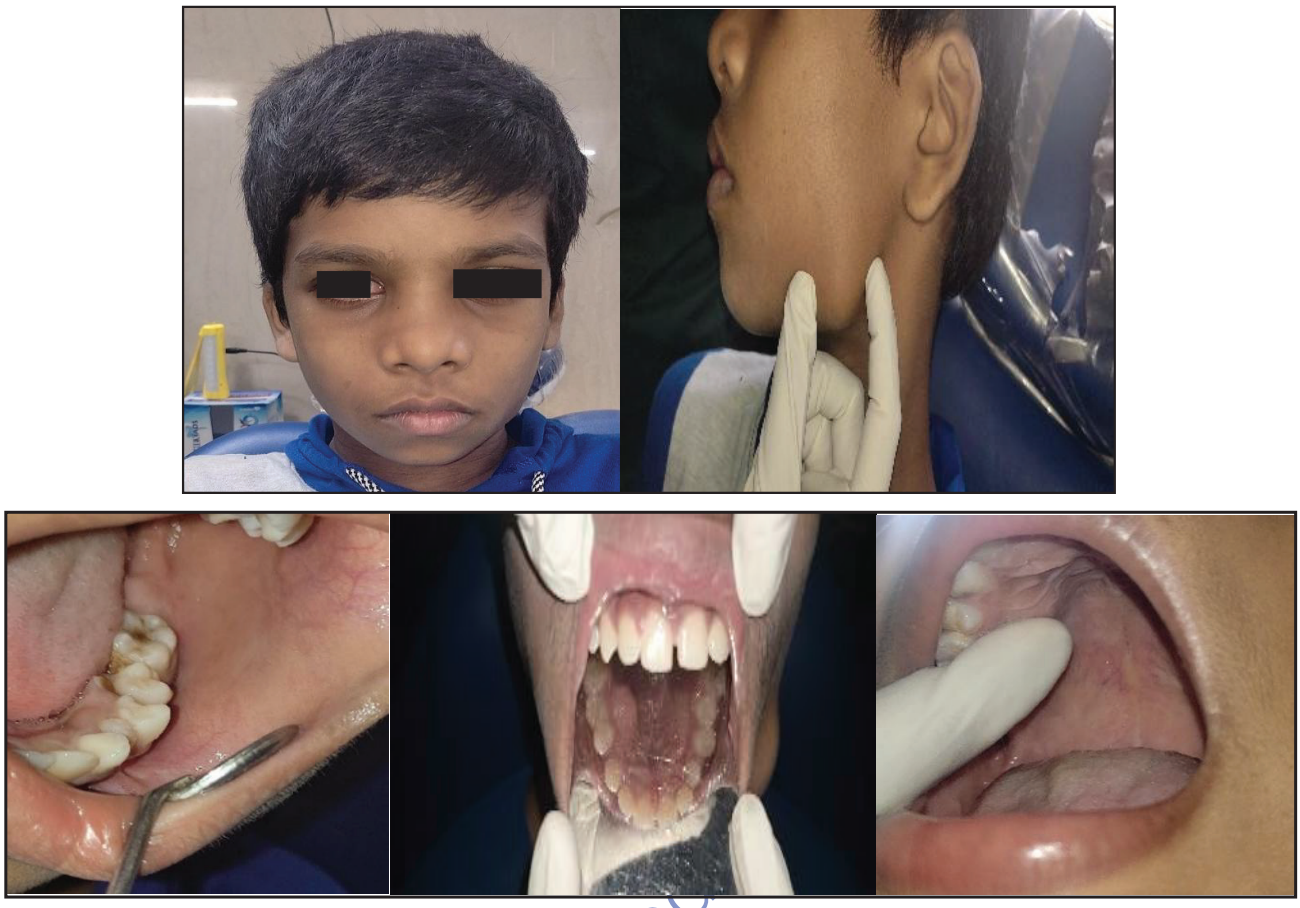

Figure 1. *(a) Frontal view showing diffuse swelling over the left mandibular angle causing facial asymmetry. (b) Lateral view showing the extent of swelling in the left mandibular angle region. (c) Intraoral photograph showing a solitary, well-defined bony swelling in the rugae–palatal region adjacent to teeth 14 and 15. (d) Closer intraoral view illustrating the smooth surface and intact overlying mucosa of the palatal bony swelling. (e) Intraoral occlusal view highlighting the size, boundaries, and localization of the palatal osteoma.*

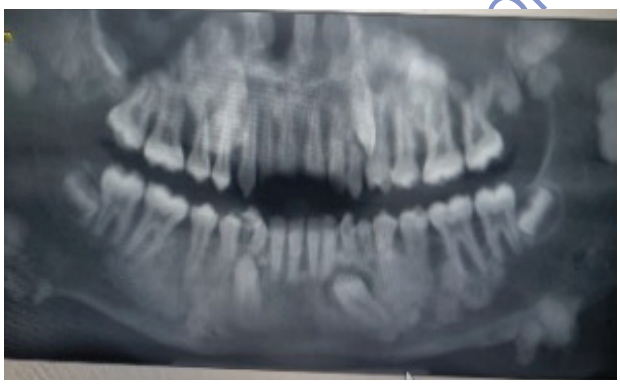

Figure 2. *Panoramic radiograph shows multiple radiopaque foci.*

reaction, or root resorption. The periodontal ligament spaces and lamina dura of adjacent teeth were intact (Figure 2).

Advanced imaging of case 1

Cone beam computed tomography (CBCT) revealed multiple well-circumscribed, lobulated osseous outgrowths attached to the mandibular cortical plate, showing homogeneous hyperdense internal architecture consistent with compact bone. The lesions maintained cortical continuity with the underlying mandible. Additional similar osseous proliferations were noted in the posterior palatal region and the ipsilateral paranasal sinus. CBCT clearly demonstrated the buccolingual extent and three-dimensional morphology of all lesions, with no cortical breach, medullary involvement, or soft-tissue extension (Figure 3a–c).

Combined CECT (Face + Abdomen) of case 1

Contrast-enhanced CT (CECT) of the face and abdomen demonstrated multiple hyperdense, well-defined osseous lesions involving the left mandibular angle, posterior palate, and adjacent paranasal sinuses, with density equivalent to cortical bone and no post-contrast enhancement, confirming the absence of any soft-tissue component. All lesions showed smooth borders, intact cortices, and no periosteal reaction or local invasion (Figure 4a-c).

CECT of the abdomen revealed no gastrointestinal polyposis, visceral abnormalities, or other stigmata of Gardner syndrome (GS), with normal bowel morphology in both axial and coronal sections (Figure 4d,e). Abdominal ultrasound and clinical gastrointestinal assessment were also unremarkable.

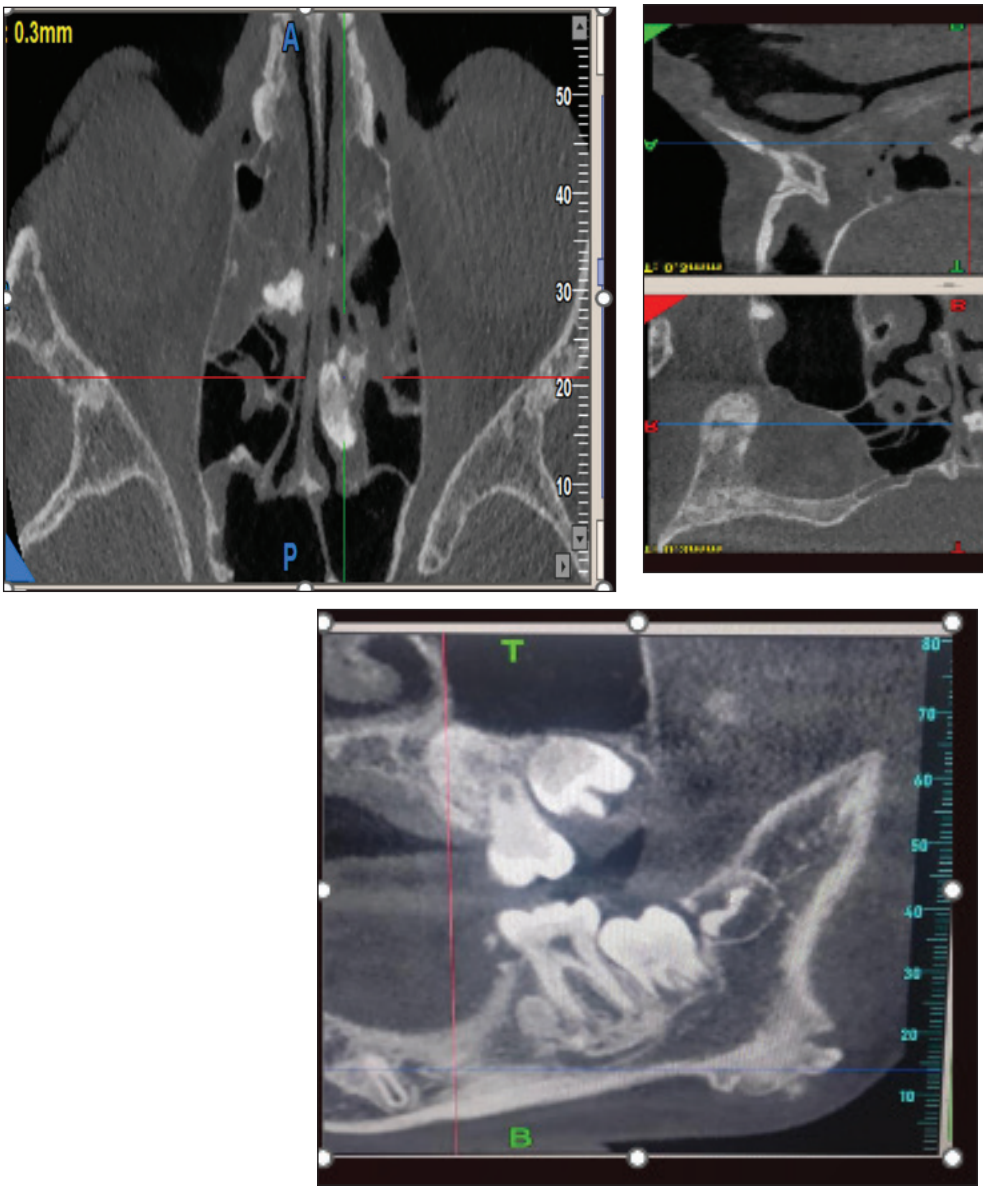

Figure 3. *(a) Axial section of CBCT. (b) coronal/sagittal section of CBCT. (c) Multiplanar view CBCT showing hyperdense structure at angle region. (d) CECT face axial contrast CT abdomen. (e) Coronal contrast CT abdomen.*

Systemic evaluation included a plain chest radiograph and a pelvic radiograph, both of which were unremarkable of case 1 (Figure 5a,b).

Laboratory investigations demonstrated a mildly elevated serum alkaline phosphatase level (325 U/l).

An incisional biopsy was performed from the palatal swelling. Histopathological examination of hematoxylin and eosin–stained sections revealed dense, mature lamellar bone with well-organized haversian systems and osteocytes within lacunae. The trabeculae were separated by scant fibrofatty marrow spaces, with no evidence of cellular atypia, abnormal mitoses, or necrosis. These findings were consistent with a diagnosis of osteoma, supporting the clinical and radiological impression.

Family history: Screening of first-degree relatives revealed that the patient's 52-year-old father also had craniofacial swellings. He was asymptomatic, with no pain, intraoral swelling, or functional limitation.

Case 2

On extraoral examination, a solitary, firm bony swelling measuring approximately 2 × 2 cm was present on the forehead. The swelling was non-tender, immobile, and covered by normal skin (Figure 6a,b). Intraoral examination revealed no abnormality, except for a clinically missing canine (43).

Systemic evaluation—including a plain chest radiograph showing clear lung fields and normal bony structures. (Figure 7b) and contrast-enhanced CT of the abdomen in axial and coronal sections (Figure 9a,b), along with dermatologic and ophthalmologic screening—was unremarkable. No biopsy or surgical intervention was

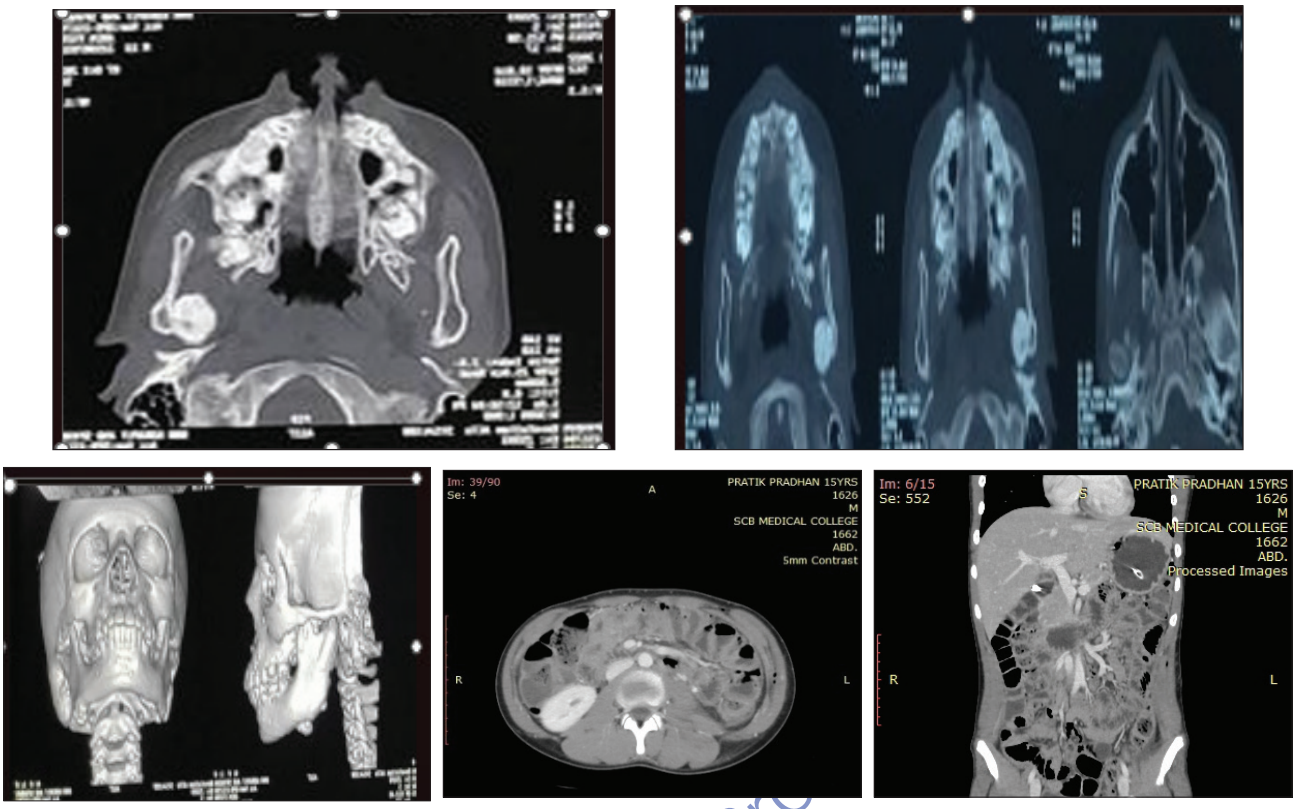

Figure 4. *(a) Axial CECT image showing a well-defined hyperdense osseous lesion involving the left mandibular angle and adjacent paranasal sinus region, with no soft-tissue component. (b) Serial section of axial the images demonstrate well-defined, homogeneously radiopaque osseous masses involving the maxilla mandible and adjacent paranasal sinus regions. The lesions are smoothly marginated, causing expansion and partial sinus obliteration, but without evidence of cortical destruction or soft tissue invasion. (c) CECT face. (d) Axial contrast CT abdomen. (e) coronal contrast CT abdomen.*

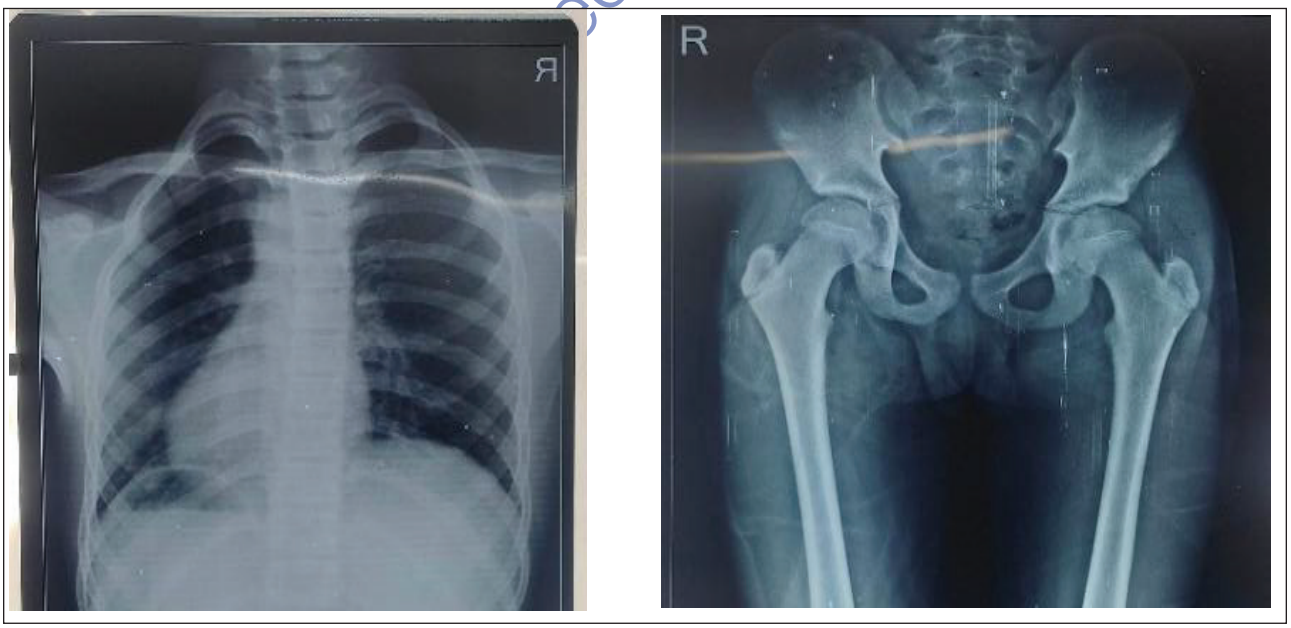

Figure 5. *Plain chest and pelvic x-ray.*

performed due to the asymptomatic nature of the lesions, and the patient was placed on periodic follow-up.

A panoramic radiograph revealed multiple well-defined radiopaque masses of varying sizes (0.5-2.5 cm) involving the mandibular body, angle, and condyle bilaterally (Figure 7a).

CBCT (Figure 8a-d) confirmed multiple hyperdense osseous growths arising from the cortical surfaces of the mandible, frontal sinus, and ethmoid sinus, with the frontal lesion corresponding to the forehead swelling. These appeared sessile showed no cortical breach, periosteal

Figure 6. *Case 2 showing extraoral swelling in frontal region, no intraoral abnormalities.*

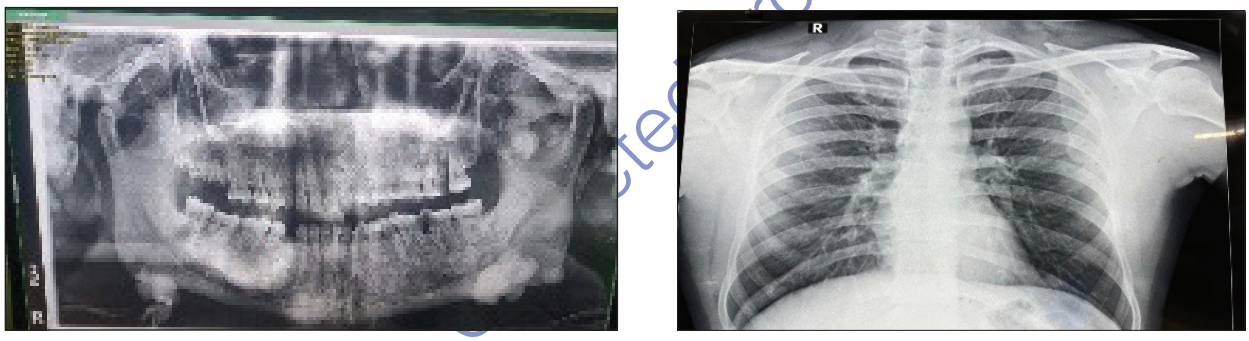

Figure 7. *(a) Panoramic. (b) Plain chest Xray chest.*

reaction, or soft tissue component, consistent with osteomas.

Discussion

Osteomas are benign, slow-growing bony proliferations that are usually solitary and incidental [1,2]. However, the presence of multiple craniofacial osteomas frequently raises concern for GS, an Adenomatous Polyposis Coli-associated variant of familial adenomatous polyposis characterized by intestinal polyposis and diverse extracolonic lesions [3,4]. Importantly, craniofacial osteomas may precede intestinal features by several years and, therefore, warrant careful evaluation [4,5].

In the present report, two first-degree male relatives exhibited multiple craniofacial osteomas without gastrointestinal, cutaneous, or extracranial skeletal abnormalities. This clinical pattern presents two major diagnostic possibilities: (i) attenuated GS, in which osteomas may occur with delayed or minimal polyposis, and (ii) sporadic familial osteomas, a rare non-syndromic condition characterized by isolated craniofacial involvement.

Our cases lacked the extracranial skeletal lesions reported by Putro et al. [6] and also differed from multi-generational osseous–cutaneous variants described by Fuhrmann et al. [7], suggesting a distinct presentation. Recent genetic evidence provides a biological basis for sporadic osteomas: Baumhoer et al [8]. demonstrated that somatic CTNNB1 (β-catenin) mutations can drive osteoma formation via aberrant WNT/β-catenin signaling independent of APC alterations. Additional reports, such as Castelino et al. [9], similarly describe isolated multiple osteomas without syndromic associations. Conversely, APC-related GS continues to show significant genotypic heterogeneity; for example, Zeng et al. [10] identified a novel APC mutation presenting with odontogenic lesions as early markers. These findings collectively reinforce the need for molecular evaluation before GS can be excluded [11].

Although genetic testing was not performed in our patients, the absence of polyposis, cutaneous lesions, and extracranial skeletal findings currently supports a provisional diagnosis of sporadic familial

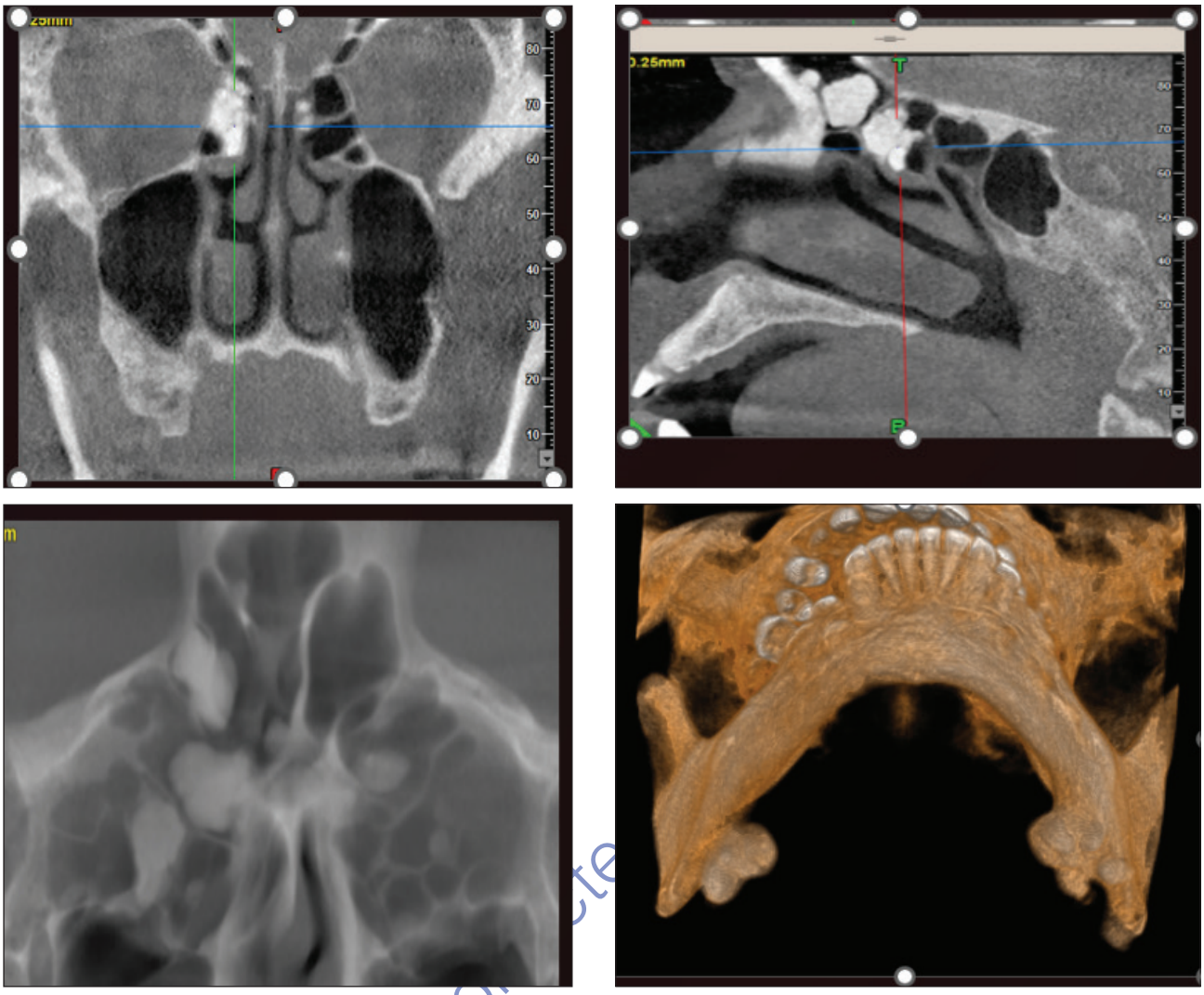

Figure 8. *(a) Coronal CBCT. (b) Sagittal CBCT. (c) Multiplanar of CBCT. (d) 3D CBCT.*

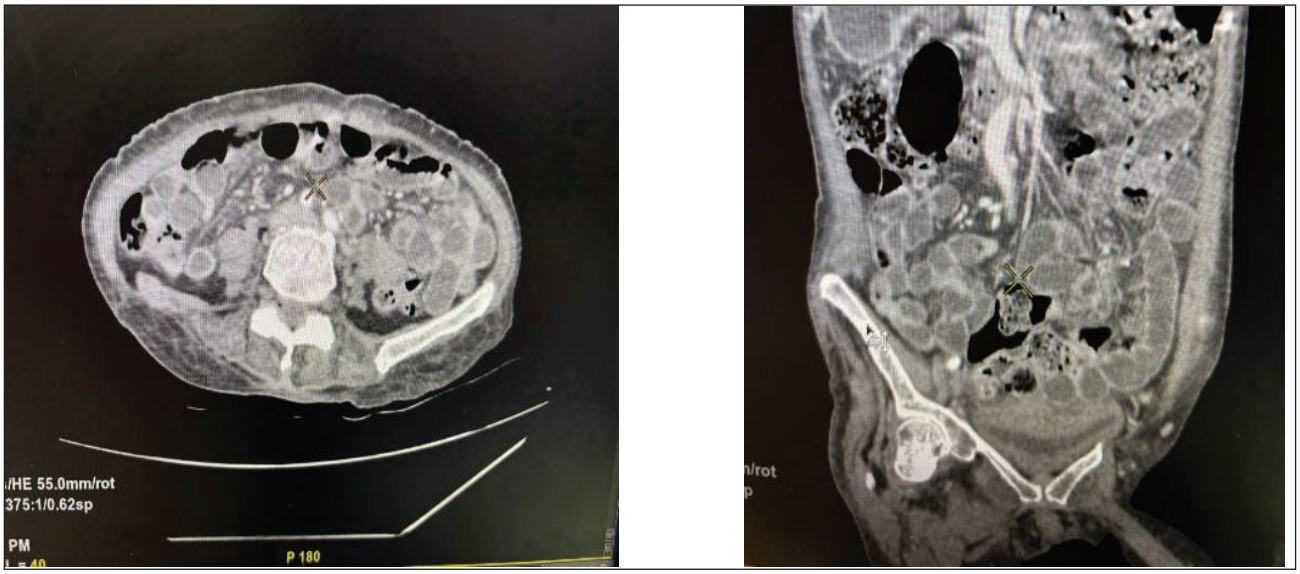

Figure 9. *(a) Contrast CT axial abdomen axial. (b) Contrast CT sagittal abdomen.*

osteomas. Nevertheless, because GS manifestations may emerge later, long-term surveillance remains critical. Recommended monitoring includes periodic colonoscopy every 3-5 years, baseline and interval dermatologic

evaluation, and ongoing clinical review for new skeletal or soft-tissue lesions.

Management of craniofacial osteomas depends on symptoms. Asymptomatic lesions may be observed, whereas surgical removal is indicated for functional impairment or cosmetic concerns. Consistent with reports by Tarsitano et al. [11], selective excision in our proband yielded favorable functional and aesthetic outcomes.

Conclusion

Familial multiple craniofacial osteomas present a diagnostic dilemma between attenuated GS and sporadic familial osteomas. Despite negative systemic screening in our patients, genetic testing and counselling are essential to accurately differentiate APC-related disease from non-syndromic variants. Continued gastrointestinal surveillance and long-term follow-up every 2-3 years are recommended, as phenotypic evolution may occur over time. Early recognition and appropriate monitoring are crucial to prevent delayed diagnosis of potential APC-associated malignant transformation.

What is new?

- Two first-degree relatives presented with multiple craniofacial osteomas without intestinal or extracolonic features, creating a true diagnostic dilemma between attenuated GS and sporadic familial osteomas.
- The cases reinforce emerging evidence that some craniofacial osteomas may arise independent of APC mutations, possibly via CTNNB1-related pathways.
- The report highlights the importance of genetic testing and structured long-term surveillance, even when initial systemic screening is normal.
- Practical follow-up recommendations—periodic colonoscopy and dermatologic evaluation—are emphasized for early detection of late-onset features.
- This familial presentation adds to the very limited literature describing multiple craniofacial osteomas without polyposis.

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Abbreviations

APC	**Adenomatous polyposis coli**
CBCT	**Cone-beam computed tomography**
CT	**Computed tomography**
GS	**Gardner syndrome**

Conflict of interest

The authors declare that there is no conflict of interest regarding the publication of this manuscript.

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Author contributions

Author 1: Conceptualization, clinical diagnosis, data acquisition, manuscript drafting.
Author 2: Radiologic interpretation, literature review, preparation of figure panels.
Author 3: Critical revision, final approval of the manuscript.
All authors reviewed and approved the final version.

Availability of data and materials

All relevant data are included within the manuscript.

Informed consent

Written informed consent for publication of clinical details and images was obtained from both patients.

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Summary of the case

1	Patient (gender, age)	15 year male, 52 year male
2	Final diagnosis	Familial craniofacial osteoma
3	Symptoms	Painless swelling with Facial asymmetry of left mandible region
4	Medications	NA
5	Clinical procedure	Oral and systemic evaluation with biopsy
6	Specialty	Oral Medicine and Radiology