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A female, 22-year-old patient presented with a soft tissue lesion adjacent to the calvaria. Skull radiography was performed as part of the preoperative investigation for the lesion excision (Figure 1). In order to supplement the evaluation of incidental radiographic findings, the patient was submitted to computed tomography (CT) of the facial sinuses (Figures 2 and 3).

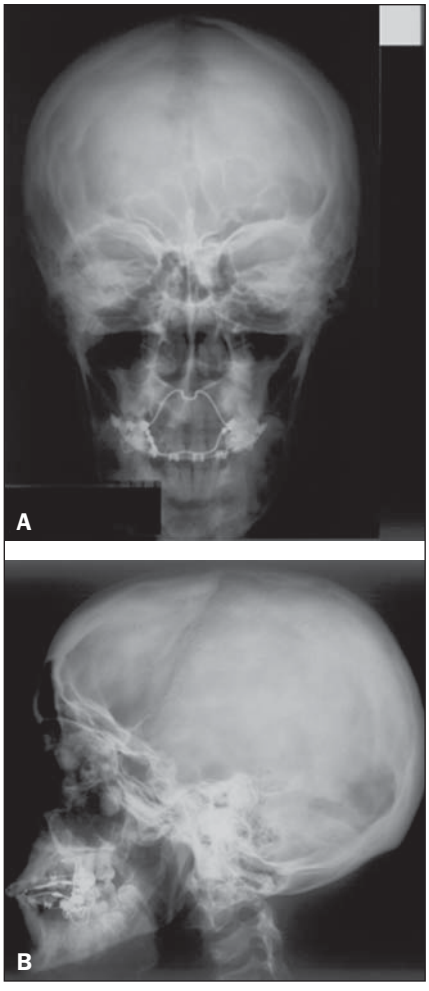

Figure 1. Skull radiography. Posteroanterior (**A**) and lateral (**B**) views.

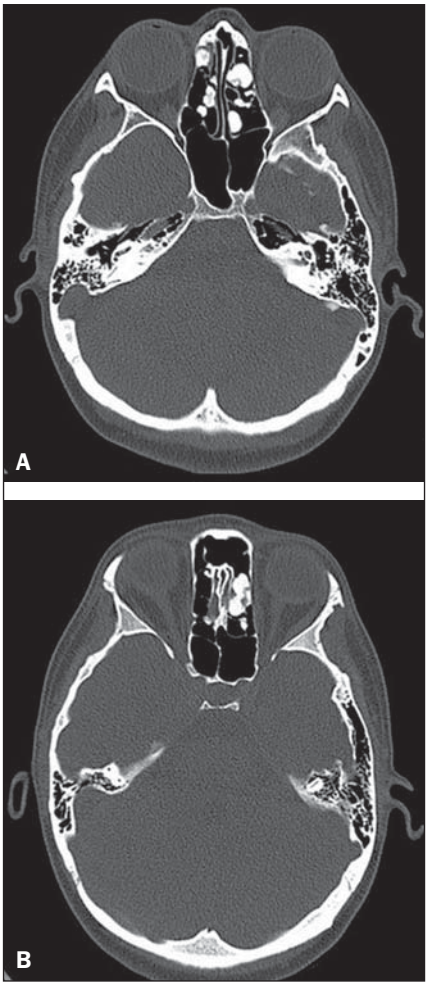

Figure 2. Non-contrast-enhanced CT, bone window.

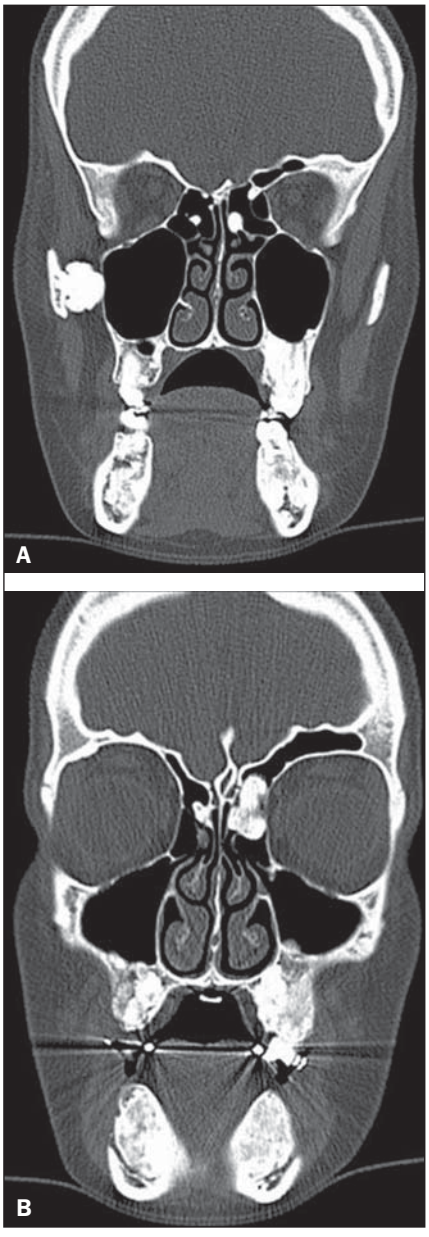

Figure 3. Coronal CT sections, bone window.

Images Description

Figure 1. Skull radiography, posteroanterior and lateral views demonstrating incidentally found osteoblastic lesion with projection into the facial sinuses and mandible.

Figure 2. Non-contrast-enhanced axial CT sections, bone window, confirming the presence of multiple osteoblastic lesions in the facial sinuses. Such lesions project into the air spaces of the paranasal sinuses.

Figure 3. Coronal CT sections, bone window, demonstrating multiple osteoblastic lesions in the facial sinuses and mandible. The lesion on the mandible shows exophytic growth. The findings are suggestive of multiple osteomas.

Diagnosis: Gardner's syndrome.

COMMENTS

Gardner's syndrome is a variant of familial adenomatous polyposis which is a dominant autosomal disease characterized by the presence of multiple adenomatous polyps on the intestinal mucosa, particularly in the colon, with high potential for malignant transformation[1].

Gardner's syndrome was originally described as a triad comprising: 1) adenomatous polyposis of the colon; 2) skull and mandible osteomas; 3) epidermoid cysts. However, from the original description by Garder in 1953, the triad has been expanded to include other soft tissue abnormalities such as desmoid tumors, sebaceous cysts, lipomas, fibromas and sarcomas[2]. The incidence of this syndrome is estimated to be 1:14.000 births[3].

Osteomas are benign, slow-growing bone tumors, corresponding to the most common benign neoplasms of the nose and facial sinuses. Generally, they are asymptomatic and incidentally found at imaging studies, although they may run their course with symptoms, depending on their size and location. Typically, the growth of these bone tumors occurs towards inside the air space of a facial sinus or simply exophytically in relation to the bone surface. In Gardner's syndrome, osteomas presentation is varied, generally preceding the onset of colon polyps[4,5].

The genetic basis of this syndrome is related to a mutation in the gene of colon adenomatous polyposis located in the long arm of the chromosome 5. The loss of the functions of this gene, which is a tumor suppressor, is considered to be the initial event in the development of adenomas[6,7].

Differential diagnoses include pure familial adenomatous polyposis, Turcot syndrome (an association of colon adenomas and encephalic tumors such as glioblastoma multiforme and medulloblastoma), Peutz-Jegher syndrome (consisting in multiple hamartomatous polyps) and hereditary nonpolyposis colorectal cancer[6].

The management of these patients includes total colectomy, since the malignant transformation of adenomas occurs in all of the cases. Familial screening is indicated[1–3].

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