

Case W15

Clinical Presentation

An 11-year-old boy with disfiguring mandibular lesions presents for evaluation.



Pearl

- Facial MR may assist in evaluating the soft tissue extent of OKCs into subgingival tissues.
- Brain MR is indicated to screen for intracranial neoplasia.

Pitfall

- In some cases, the presence of maxillary and mandibular cysts and CSF spread of tumor to the infundibulum may mimic Langerhans' cell histiocytosis.

Radiologic Findings

Coronal non-contrast CT scans through the mandible and paranasal sinuses (Figs. A and B) photographed with bone windows demonstrate bilateral expansile lesions of the mandibular rami as well as the body of the right mandible. Note that the crowns of several teeth are associated with these lesions. Expansile cysts are also present in the maxilla bilaterally, encroaching on the maxillary sinuses. These lesions appear cystic on an axial image photographed in soft tissue window (Fig. C). A frontal skull film (Fig. D) reveals prominent midline falx calcification (*arrows*).

Diagnosis

Basal cell nevus syndrome (also known as Gorlin's syndrome or nevoid basal cell carcinoma syndrome)

Differential Diagnosis (of lucent jaw lesions)

- Metastases (cortical destruction, mixed lytic-sclerotic pattern, often irregularly marginated)
- Myeloma (often a permeative pattern)
- Langerhans' cell histiocytosis (indistinguishable in some cases though usually more destructive; has frequent involvement of skull, spine, or long bones and may be associated with diabetes insipidus and visceral organ involvement)
- Non-syndromic odontogenic cyst (usually isolated rather than multiple)
- Ameloblastoma (often mixed lytic-sclerotic pattern, often a significant soft tissue component)

Discussion

Background

Basal cell nevus syndrome is a rare, autosomal dominant syndrome that is characterized by multiple cutaneous basal cell carcinomas, odontogenic keratocysts (OKCs) of the jaw, lamellar falx calcifications, and dyskeratotic pitting of the palms and soles of the feet. Multiple other associations have been reported including macrocephaly, bifid ribs, vertebral anomalies, and an increased incidence of soft tissue tumors. Patients are also at increased risk of CNS tumors including medulloblastoma, meningioma, astrocytoma, and craniopharyngioma.

Etiology

Gorlin's syndrome has been associated with a mutation on the q arm of chromosome 9, which is the locus of a tumor suppression gene.

Clinical Findings

Eighty percent of patients are diagnosed within the first two decades of life. The mean age of presentation for jaw cysts is 15.5 years; the mean age for presentation of basal cell carcinoma is 20.3 years. Symptoms related to odontogenic keratocysts include facial swelling, abnormal taste sensation, visual disturbance, facial paresthesias, and discharge from the mouth.

Pathology

Gross

- Usually large, multilocular cysts containing variable amounts of keratin

Microscopic

- Cysts are lined with a thin stratified squamous epithelium with a prominent basal cell layer

Imaging Findings

CT

- OKCs are well-circumscribed cystic lesions most commonly found in the posterior body and ramus of the mandible
- The cysts may be next to or displace the crown of an unerupted tooth, giving the radiographic appearance of a “floating tooth”
- Maxillary cysts can extend into and fill the paranasal sinuses

MR

- Usually multiple well-circumscribed expansile lesions which demonstrate T1 and T2 prolongation
- Peripheral gadolinium enhancement may be seen

Treatment

Aggressive complete curettage of the OKCs

Prognosis

- Cysts are often recurrent
- Overall prognosis is most dependent on the presence or absence of CNS neoplasia

Suggested Readings

Gorlin RJ, Goltz RW. Multiple nevoid basal cell epitheliomas, jaw cysts and bifid ribs. *N Engl J Med* 262:908–912, 1960.

Lovin JD, Talarico CL, Wegert SL. Gorlin’s syndrome with associated odontogenic cysts. *Pediatr Radiol* 21:584–587, 1991.

Shanley S, Ratcliffe J, Hockey A, et al. Nevoid basal cell carcinoma syndrome: review of 118 affected individuals. *Amer J Med Genetics* 50:282–290, 1994.