Odontogenic keratocysts in nevoid basal cell syndrome (Gorlin’s Syndrome). CT and MR evaluation

RESUMEN

Introducción: El síndrome de carcinoma basocelular de tipo nevoide (SCBCN o S. de Gorlin) es un desorden autosómico dominante, caracterizado por múltiples carcinomas basocelulares, queratoquistes odontogénicos, calcificaciones durales, deformidades óseas y faciales, tumores que incluyen meduloblastoma y fibromas ováricos y grados variables de retardo mental. Los hallazgos imagenológicos característicos del SCBCN son queratoquistes odontogénicos de la mandíbula y del maxilar, prognatismo, labio-paladar hendido, macrocefalia, cavidades paranasales prominentes, calcificaciones de la hoz interhemisférica, anormalías vertebrales (cifoescoliosis y segmentación anormal), fusión de cuerpos costales, cuarto metacarpiano corto y lesiones óseas escleróticas.

Material y métodos: Presentamos un caso de un paciente masculino de 13 años de edad, con antecedentes familiares de SCBCN, quien presentó drenaje serosanguinolento fetido bucal, prognatismo e hipertelorismo. Los estudios imagenológicos mostraron lesiones quísticas bilaterales en el ángulo de la mandíbula y antros maxilares. La RM con imágenes potenciadas en T2 y T1 con Gadolinium demostró múltiples lesiones quísticas de contornos lobulados, con realce periférico luego de la administración del Gadolinium, algunas con nivel líquido secundario a un componente hemorrágico. La RM cerebral demostró mínimo adelgazamiento del cuerpo calloso y discreta prominencia del sistema ventricular para la edad. La serie ósea no mostró alteraciones diferentes a las ya descritas.

Discusión: A pesar de que la TC es útil en el diagnóstico de las anomalías faciales asociadas al SCBCN; la RM es superior por su capacidad de demostrar la composición interna y estructuras de la queratosis odontogénica.

Palabras clave: Carcinoma, imágenes de resonancia magnética, odontogénico, tomografía computarizada.

continúa en la pág. 24
ABSTRACT

Introduction: The nevoid basal cell syndrome (Gorlin’s Syndrome) is a dominant autosomic disorder, characterized by multiple basal cell carcinomas, odontogenic keratocysts, dural calcifications, bone and face malformations, tumors including medulloblastoma and ovarian fibromas, as well as different degrees of mental retardation. Characteristic image findings of the Gorlin’s Syndrome are odontogenic keratocysts in the jaw and jawbone, prognathism, cleft lip and palate, macrocephalia, prominent paranasal cavities, the documented presence of this syndrome in the patient’s family.

The initial imaging workup included plain radiographs and Computed Tomography (CT) of the facial area and mandible, revealing large expansile cystic changes in the body and angle of the mandible, bilaterally (Figure 1). The expansile lesions were clearly intramedullary with erosion, thinning and scalloping of the endosteal cortical bone, without evidence of periosteal reaction. In general, the internal contents of the lesions were of low density and homogeneous, with a few scattered central calcifications within the mandibular cyst on the left side. No associated soft tissue mass was identified. Additional cysts were seen in the maxillary sinus, bilaterally and were larger on the right with bone remodeling and septation. There were non-erupted teeth within the cystic lesions in the maxilla on the left, as well as in the mandible on the right. Intracranially dural calcifications were noted in the falx.

Magnetic Resonance Imaging (MRI) was performed for further assessment of the cystic lesions and to rule out intracranial abnormalities. T1 Weighted (T1W) and T2 Weighted (T2W) sequences and a post-contrast T1W sequence were obtained (Figure 2). Multiple expansile cystic lesions were again identified involving the mandibular angle and ramus bilaterally. Cystic lesions were also present in the maxillary sinus bilaterally, with greater involvement on the right, showing septation. The cystic contents were of low signal intensity on T1W sequences, and of high signal intensity on the T2W sequences, with some heterogeneous signal intensity, probably related to hemorrhagic components and a thin rim enhancement following Gadolinium administration.

MR images of the brain demonstrated minimal thinning of the body of the corpus callosum. The lateral ventricles were slightly enlarged for the patient’s age. No abnormal enhancing brain parenchymal lesions, heterotopic gray matter, or brain tumor were identified. A plain film skeletal survey, including views of the scapulae, ribs, hands and lumbar spine were performed. All appeared unremarkable.

Discussion

Although CT is valuable in elucidating osseous craniofacial anomalies associated with NBCCS, MR is superior in demonstrating the internal composition and structure of the odontogenic keratocysts commonly seen in this syndrome. CT imaging defined the cystic osseous expansion, septation and wall thinning. Aside from tooth primordia within the cysts, the contents appeared homogenous on CT, except for a few small densities, probably representing calcifications in the mandibular cyst on the left. MR illustrated the hyperintensity of the lesions on the T2W images indicating the cystic nature and contrast enhancement of the cystic lining the post-contrast T1W images.

The imaging findings in the mandible and maxillae are consistent with odontogenic keratocysts, characteristic
lesions of NBCCS. Odontogenic keratocysts are dentic-
gerous or primordial in origin and lined with keratinized
epithelium and unerupted teeth.8
The oral drainage was the first complication of the disease
in our patient and that prompted a CT study of the facial
area to determine the nature and extent of the lesions. An
MR examination was performed, not only to further cha-
racterize the nature of the mandibular and maxillary lesio-
ns, but also to rule out intracranial abnormalities.
NBCCS is an autosomal dominant disorder with a pre-
valence of about 1 per 60,000, showing complete pene-
trance, but variable expressivity.9
Our patient is a third generation member of a family
with genetically documented NBCCS. The variable ex-
pressivity of NBCCS is dramatically demonstrated in
the patient's affected relatives (Figure 3). The patient's
mother and grandmother demonstrate only minimal
features of NBCCS, including hypertelorism, mandibular
enlargement, and palmar and plantar pitting. His three uncles, however, had more severe manifestations: one had multiple recurrent basal cell car-
cinomas requiring frequent surgical removal and an-
other had a fatal adenoid cystic carcinoma of the hard palate. The third uncle had a fatal posterior fossa ependymoma.
References