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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, anomalías 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 fétido 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.

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Introduction

Nevoid Basal Cell Carcinoma (NBCCS), also called Gorlin's syndrome is an autosomal dominant disorder.[1] Up to one-third of cases do not have any family history. The characteristics of NBCCS are multiple basal cell carcinomas, odontogenic keratocysts, palmar and plantar pits, dural calcifications, cranio-facial anomalies, intracranial tumors, ovarian fibromas, and variable mental retardation.[2] Characteristic imaging findings of NBCCS include: odontogenic keratocysts of the mandible and maxillae; calcification of the falx cerebri; exaggerated mandible length; macrocephaly with frontal bossing; cleft lip and/or palate; large paranasal sinuses; vertebral anomalies including kyphoscoliosis and abnormal segmentation; rib abnormalities, including fusion or splaying, sclerotic bone lesions, and a short fourth metacarpal bone.[3]

Case presentation

A thirteen year-old male presented with new onset malodorous serosanguineous oral drainage bilaterally, mandibular enlargement, mild frontal bossing and hypertelorism. The patient was afebrile and in no distress. Clinical suspicion of NBCC was high, due to

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ABSTRACT

Introduction: The nevoid basal cell syndrome (Gorlin's Syndrome) is a dominant autosomic disorder, characterized by multiple basal cell carcinomas, odontegenic keratocysts, dural calcifications, bone and face malformations, tumors including meduloblastoma and ovaric fibromas, as well as different degrees of mental retardation.
Characteristic imagine findings of the Gorlin's Syndrome are odontogenic keratocysts in the jaw and jawbone, prognathism, cleft lip and palate, macrocephalia, prominent paranasal cavities, inter-hemispheric bone calcifications, vertebrae malformations (cifoescoliosis and abnormal segmentation), ribs merging, short forth metacarpian and sclerotic bone lesions.
Material and methods: A case of a 13-year old male patient is presented, with family background of Gorlin's Syndrome who presented mouth fetid serum-bloody drain, prognathism and hypertelorism. Imaging studies showed bi-lateral cystic lesions on the jaw's angle and maxillary antra. MRI with T2 and T1 powered images with Gadolinium showed multiple cystic lesions with lobe-like contours, which had their peripheral contour highlighted after the administration of Gadolinium, some with a secondary liquid level at hemorrhagic component. Brain MRI showed minimum slimming of the callous body and a small prominence of the ventricle system for his age. The bone series did not show any alterations different from those already described.
Discusion: Even though the CT is useful in diagnosing face abnormalities related to Gorlin's Syndrome, MRI is better in it capacity to show the internal composition and the structures of odontogenic keratocysts.

Key words: Carcinoma, magnetic resonance imaging, odontogenic, computed tomography.

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[4], MR is superior in demonstrating the internal composition and structure of the odontogenic keratocysts commonly seen in this syndrome[5-7] 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

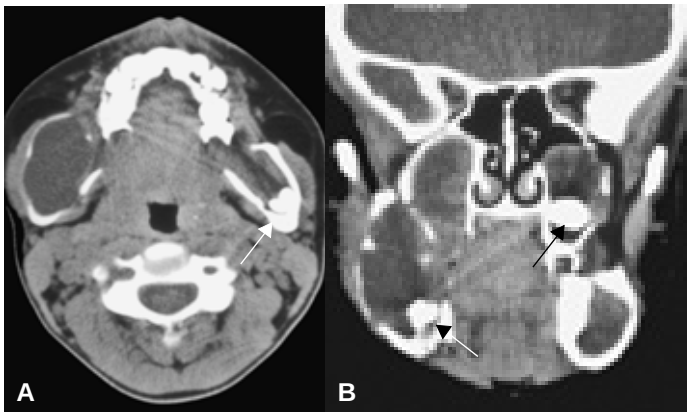

Figure 1. Axial **(A)** and coronal **(B)** CTs with bone windows show the expansile bilateral mandibular and maxillary keratocysts (arrows). Note the teeth within the cysts.

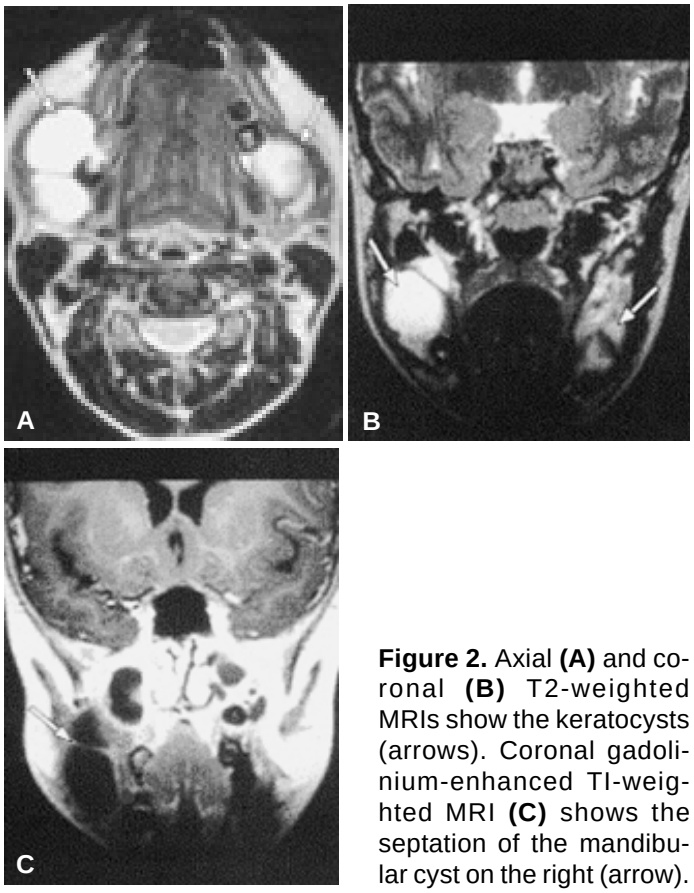

Figure 2. Axial **(A)** and coronal **(B)** T2-weighted MRIs show the keratocysts (arrows). Coronal gadolinium-enhanced TI-weighted MRI **(C)** shows the septation of the mandibular cyst on the right (arrow).

lesions of NBCCS. Odontogenic keratocysts are dentigerous 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

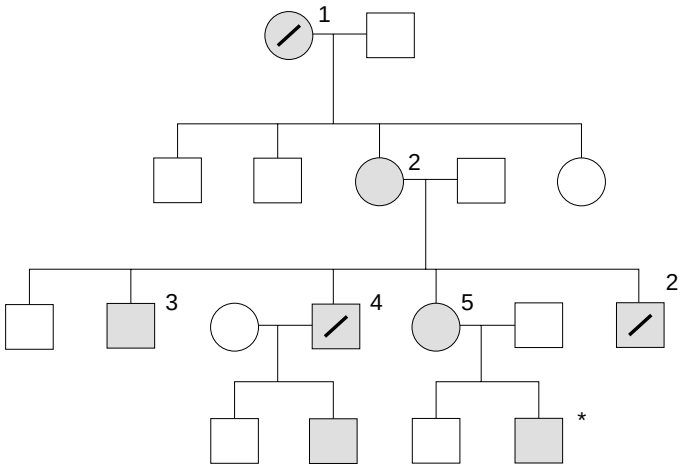

Figure 3. This pedigree illustrates the incidence Of NBCCS in our patient's (*) family. His maternal great-grandmother[1] developed basal cell carcinoma, but the status of her PTCH gene alleles was unknown. Documentation of PTCH mutations began with the patient's maternal grandmother[2]. Both she and the patient's mother[5] manifested only minimal features of NBCCS (hypertelorism, mandibular enlargement, and palmar and plantar pitting). His three uncles, however, had more severe manifestations. One uncle[3] experienced multiple recurrences of basal cell carcinoma that required frequent surgical removal. Another uncle[4] died of adenoid cystic carcinoma of the hard palate, and the third uncle[6] died of ependymoma[6]. (Circles represent females, and squares represent males. Shaded shapes represent affected family members, and unshaded shapes represent unaffected members. Diagonal lines represent deceased members.)

MR examination was performed, not only to further characterize the nature of the mandibular and maxillary lesions, but also to rule out intracranial abnormalities.

NBCCS is an autosomal dominant disorder with a prevalence of about 1 per 60,000, showing complete penetrance, but variable expressivity.[9]

Our patient is a third generation member of a family with genetically documented NBCCS. The variable expressivity 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 disease manifestations: one had multiple recurrent basal cell carcinomas requiring frequent surgical removal and another had a fatal adenoid cystic carcinoma of the hard palate. The third uncle had a fatal posterior fossa ependymoma.

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