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

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 NBCCS was high, due to...
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 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, vertebræ 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 callosus 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.

Discussion: 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.
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

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 manifestations: one uncle experienced multiple recurrences of basal cell carcinoma that required frequent surgical removal. Another uncle died of adenoid cystic carcinoma of the hard palate, and the third uncle died of ependymoma. (Circles represent females, and squares represent males. Shaded shapes represent affected family members, and unshaded shapes represent unaffected members. Diagonal lines represent deceased members.)
References