Syndrome in question: Gorlin-Goltz syndrome*

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CASE REPORT

A 30-year-old male patient presented a nodular cystic lesion on the knee when he was 5 years old and had been submitted to surgery at the time. At 10 years of age appeared mandible keratocystic odontogenic tumors (confirmed by histopathology) of recurrent character and he has had over 10 corrective surgeries since then. Three years after the onset of nodular lesions on hands and feet multiple surgeries were required to solve the lesions. The physical examination revealed coarse facies, hypertelorism, basocellular carcinomas (BCC), three on the face and eight on the upper part of the thorax, besides punctiform pits and cysts on palms (Figures 1 and 2). A thorax X-ray detected bifid ribs and a panoramic radiograph of teeth revealed odontogenic cysts (Figure 3). Histopathology confirmed the presence of BCCs, which were removed by exeresis. The patient continues to be monitored by multidisciplinary follow-up.

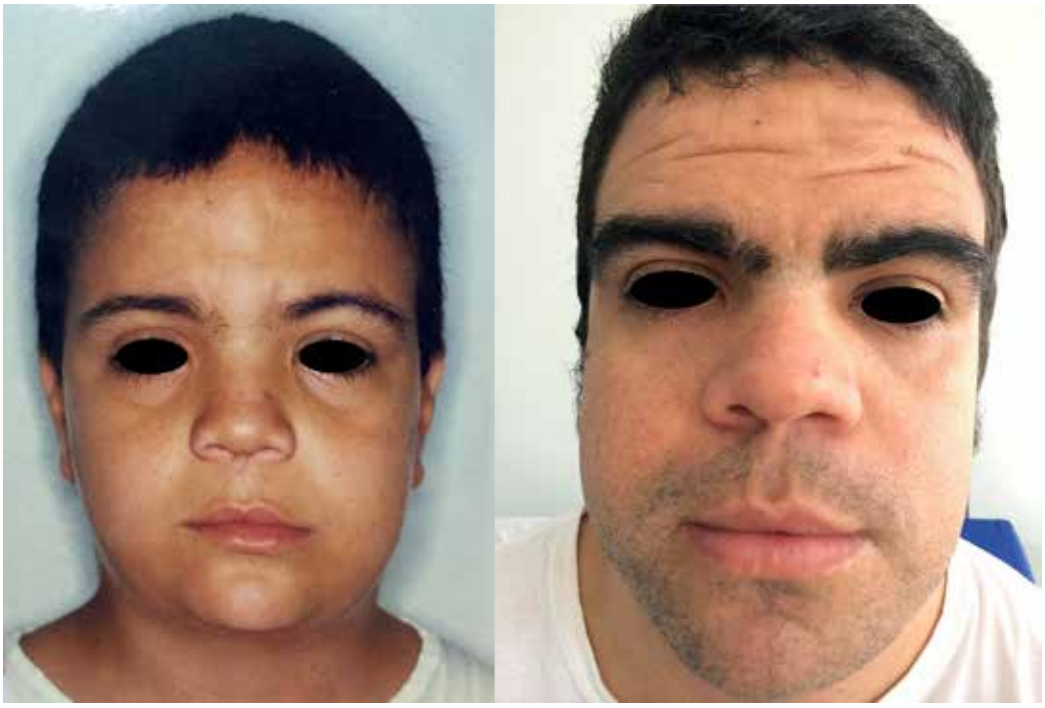

FIGURE 1: Comparing imaging in childhood and at 30 years of age: the coarse facies and hypertelorism can be noted, signs that were intensified with time

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Figure 2: In the photograph on the left, the presence of BCCs is noted at the upper portion of the trunk (red arrows) and, in the detail, a pigmented BCC. In the photograph on the right, it is possible to identify bone cysts on both hands

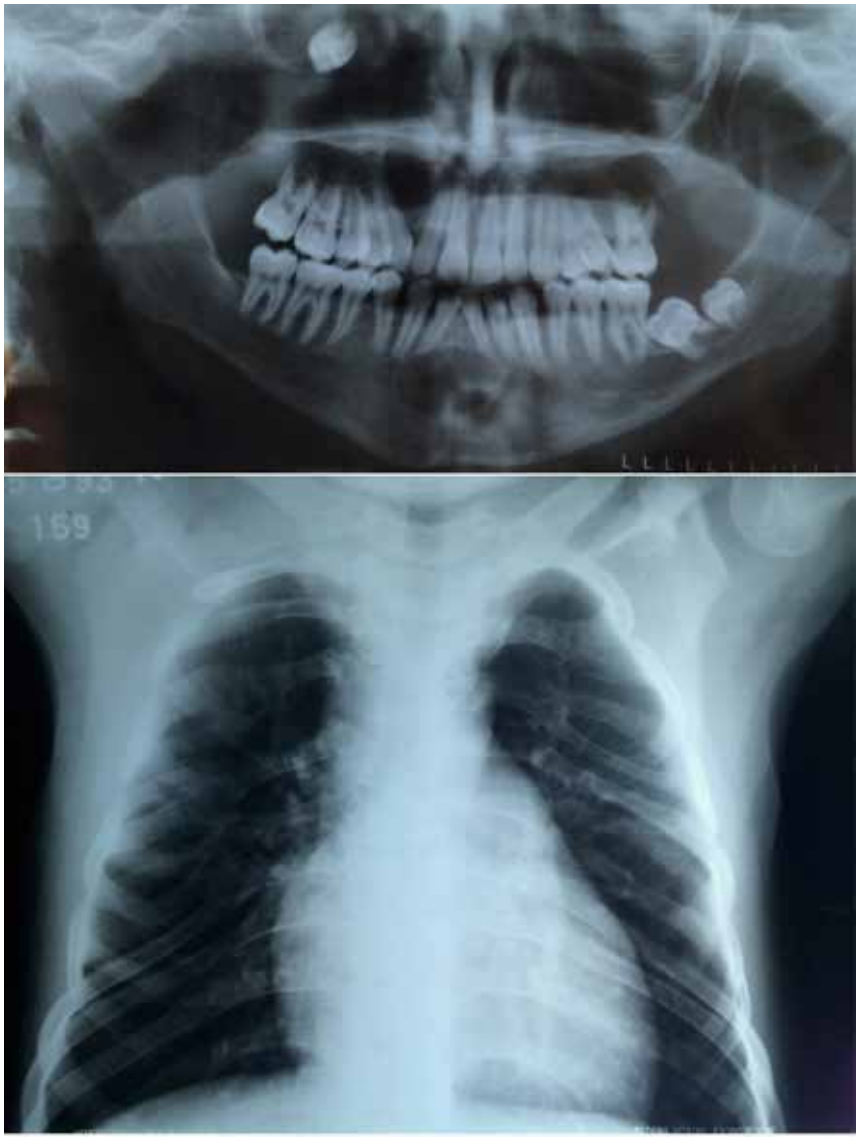

Figure 3:
In the panoramic X-ray, well-delimited and unilocular radiolucent images can be seen in the maxillary region on the right and in the mandibular body, compatible with odontogenic keratocysts. In the radiograph of the thorax, multiple bifid ribs and scoliosis can be observed

DISCUSSION

Gorlin-Goltz syndrome or nevoid basal cell carcinoma syndrome (NBCCS) was first described in 1894 by Jarish and White, but better defined by Gorlin and Goltz in 1960.[1,2] It is a rare disorder due to mutation in the patched tumor suppressor gene PTCH, located in chromosome 9q22.3q31, which functions as a component of the Hedgehog signaling pathway.[3,4] NBCCS has autosomal dominant inheritance with complete penetrance, although variable phenotypic expression.[5] It affects males and females equally but predominates in white people.[3] The prevalence is 1/57.000 and the incidence varies from 1/164.000 to 1/256.000 in the literature.[3,6,7]

The diagnosis is based on clinical findings and confirmed by the presence of two major criteria or one major associated with two minor ones.[3,8]

The major criteria are: more than 2 BCCs or 1 before 20 years of age; odontogenic keratocysts confirmed by histology; one or more palmoplantar pits; bilamellar calcification of cerebral falx; fused or flattened bifid ribs; 1st degree relative affected.

The minor criteria are: macrocephaly; congenital malformations (cleft lip or palate, frontal bossing, coarse facies, hypertelorism); skeletal alterations (Sprengel deformity, deformed chest, hemivertebrae, fusion or lengthening of vertebral bodies, anomalies in hands and feet, syndactyly, candle-flame shaped hand bone cysts); pointed sella turcica; ovarian fibroma; medulloblastoma.

The BCCs are the main findings and their onset is between puberty and the age of 35 years.[3] Their number varies from a few to hundreds, in any area of the body, mainly photoexposed skin. Any clinical type may be present, however both the invasive process and metastases are rare. [1,4,5]

In view of the wide clinical spectrum of this syndrome, the management of its modalities is not standardized. It is recommended that yearly radiographs be taken to detect skeletal anomalies, as well as a panoramic jaw X-ray to provide adequate diagnosis and approach to keratocysts, which should be duly removed on account of their aggressive bone resorption potential. As a general rule, radiotherapy is avoided due to the intense sensitivity of these individuals to ionizing radiation. The best conduct is exeresis and, for extensive areas with many BCCs, photodynamic therapy with *5-aminolevulinic* acid is an option.[9,10] Recommendations such as photoprotection and regular visits to the dermatologist are necessary in the surveillance and management of skin cancer; this routine should begin in adolescence. Children considered high risck should have magnetic resonance tomography to scan for medulloblastomas.[9] As a rule, the prognosis depends on the behavior of the skin tumor.[3,6] ❑

Abstract: The Nevoid Basal Cell Carcinoma Syndrome (NBCCS) is an uncommon disorder caused by a mutation in Patched, tumor suppressor gene. It is mainly characterized by numerous early onset basal cell carcinomas, odontogenic cysts of jaw and skeletal abnormalities. Due to the wide clinical spectrum, treatment and management of its modalities are not standardized and should be individualized and monitored by a multidisciplinary team. We report a typical case in a 30-year-old man with multiple basal cell carcinomas, keratotic pits of palmar creases and bifid ribs, with a history of several corrective surgeries for keratocystic odontogenic tumors, among other lesions characteristic of the syndrome.
Keywords: Basal cell nevus syndrome; Carcinoma, basal cell; Odontogenic cysts

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