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Gorlin-Goltz syndrome: Case report

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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. A 30-year-old man reported 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: Gorlin-Goltz, NBCCS, tumor suppressor gene

Introduction
Gorlin-Goltz syndrome, which is commonly known as Nevoid basal cell carcinoma syndrome (NBCCS) is a rare multisystemic disease that is inherited as an autosomal dominant trait. This disorder shows very high level of penetrance and variable expressivity. The syndrome is also known as multiple basal cell carcinoma syndrome (NBCCS), hereditary cutaneomandibular polyonocosis, multiple nevoid basal cell epithelioma-jaw cysts, or bifid rib syndrome. Pathogenesis of this syndrome is attributed to mutations or micro deletion of PTCH-1 gene located in the long arm of chromosome- 9. The prevalence of this syndrome is 1 in 50000 to 150000 in general populations though it may vary region wise. Male and female are equally affected.

Case Description
A 30-year-old male patient reported with 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.

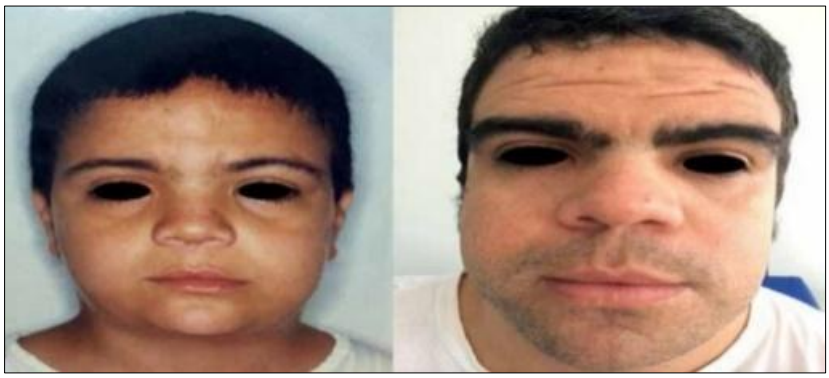

Fig 1: Physical features

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.

Fig 2: Basocellular carcinomas

A thorax X-ray detected bifid ribs and a panoramic radiograph of teeth revealed odontogenic cysts. Histopathology confirmed the presence of BCCs, which were removed by excision.

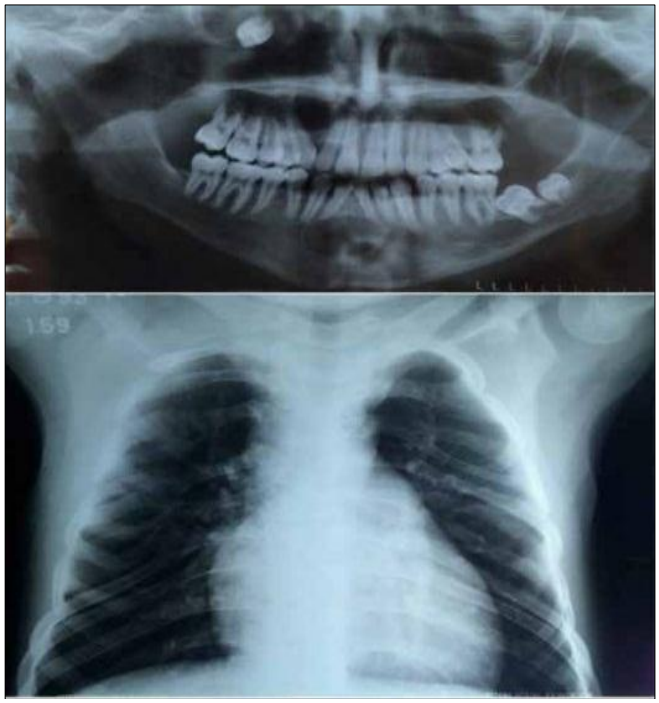

Fig 3: Odontogenic Cysts in teeth & Bifid Ribs

The patient continues to be monitored by multidisciplinary follow-up.

Discussion

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

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 bonecysts); pointed sella turcica; ovarian fibroma; medulloblastoma.

Conclusion

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. 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. As a rule, the prognosis depends on the behavior of the skin tumor.

Conflict of Interest

None

Funding

None

Consent for publication

Informed consent was obtained from the parents of the patients to publish this case in medical journal.

References

1. Sumi M, Ichikawa Y, Katayama I, Tashiro S, Nakamura T. Diffusion-weighted MR imaging of ameloblastomas and keratocystic odontogenic tumors: Differentiation by apparent diffusion coefficients of cystic lesions. AJNR Am J Neuroradiol, 2008.
2. Srinivasan K, Bhalla AS, Sharma R, Kumar A, Roycho udhury A, Bhutia O. Diffusion-weighted imaging in the evaluation of odontogenic cysts and tumours. Br J Radiol, 2012.
3. Han Y, Fan X, Su L, Wang Z. Diffusion-weighted MR imaging of unicystic odontogenic tumors for differentiation of unicystic ameloblastomas from keratocystic odontogenic tumors. Korean J Radiol. 2018.
4. Borghesi A, Nardi C, Giannitto C, Tironi A, Maroldi R, Di Bartolomeo F, *et al*. Odontogenic keratocyst: Imaging features of a benign lesion with an aggressive behaviour. Insights Imaging, 2018.