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Case Report

Imaging Features in Nevoid Basal Cell Carcinoma Syndrome: A Case Study

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Abstract

Background: Nevoid basal cell carcinoma syndrome also known as Gorlin syndrome, an autosomal dominant condition, was defined by Gorlin and Goltz in 1960. It is a rare phakomatosis and the main characteristic feature is the presence of odontogenic keratocysts. Apart from the presence of keratocytes, it also comprises other skeletal abnormalities. These patients are also more prone to develop basal cell carcinoma. Although this is an autosomal dominant condition, 60% of patients with Gorlin–Goltz syndrome have no affected family members, therefore this condition can also occur spontaneously. Other than odontogenic keratocysts, patients also develop skeletal abnormalities. 5–10% of patients may also develop malignancies such as medulloblastoma which may be a potential cause of early death.

Key words: Gorlin, Nevoid basal cell carcinoma syndrome, Odontogenic keratocyst, Skeletal abnormalities

INTRODUCTION

Gorlin–Golz syndrome is an autosomal dominant condition which is most characteristically characterized by cutaneous basal cell carcinoma, odontogenic keratocysts (OKCs), and skeletal abnormalities.[4]

This is an autosomal dominant condition thought in 60% of patients there is no family history. The estimated prevalence of this condition is 1 in 60,000 population with equal preponderance in both males and females. The main pathogenesis in this condition is the loss of the PTCH1 gene which is located on chromosome 9. The common neoplasms that are reported in this condition include medulloblastoma, meningioma, ovarian, and cardiac fibromas.[2]

The clinical diagnosis is made using major and minor imaging criteria.[1]

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Major Criteria

- Basal cell carcinoma: >2 or 1 under the age of 20
- OKC
- Palmar pits: Three or more
- Bilamellar calcification of the falx cerebri
- Rib anomalies: Bifid rib (see image), fused, splayed
- First-degree relative with Gorlin syndrome.

Minor Criteria

- Macrocephaly
- Frontal bossing, cleft lip, or hypertelorism
- Sprengel deformity, pectus excavatum or pectus carinatum, syndactyly
- Bridging of the sella turcica, hemivertebrae, flame-shaped osseous radiolucencies
- Ovarian fibroma
- Medulloblastoma.

To make a diagnosis patient should have either two major or one major and two minor criteria.

CASE REPORT

Reporting a case of 25-year-old male who had come with complaints of facial pains and delayed tooth eruption. No other significant history was noted. No significant family history.

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Physical Examination

- Head and face: Broadened nasal root and mild hypertelorism
- Ophthalmological examination: Strabismus was noted
- Cutaneous: No evidence of basal cell carcinomas or other skin manifestation
- Skeletal manifestations: Polydactyly of hands and hallux valgus.

Imaging Studies

- Dental panoramic radiograph: Multiloculated lesion of the mandible for which further evaluation was advised
- X-ray paranasal sinuses [Figure 1]-PA view: Frontal sinus appeared normal. The nasal septum was intact. Dense radio-opaque opacifications were noted in both the maxillary sinuses and in the right half of the mandible. Curvilinear calcification extending to the maxilla. CT PNS was suggested for the same
- Chest X-ray PA view: Splaying of 7th and 8th ribs. No other significant abnormality was noted. Bilateral lung fields were clear. Cardiac size was normal. The rest of the visualized bones appeared normal
- Ultrasound abdomen: No significant abnormality
- CT PNS.

Right Side

- A large uni-locular expansile cystic lesion measuring 3.5 × 3.0 × 3.7 cm (AP × ML × CC) with heterogenous internal contents (fluid, soft-tissue densities-? debri, and alcifications) is seen almost entirely filling the maxillary sinus and contiguous with the alveolar process of maxilla [Figure 2a and b]
- An unerupted tooth (third molar) is seen at the superior aspect of the cystic lesion. The cyst appears to be pericoronal to the tooth

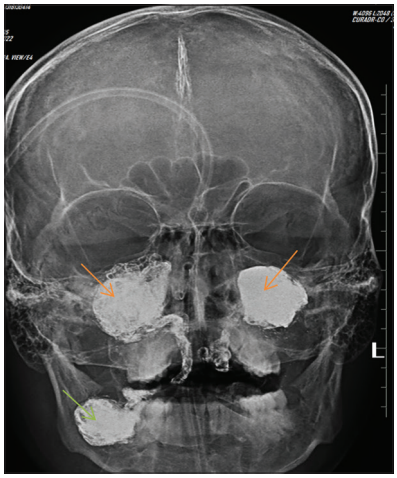

Figure 1: Dense radiopaque opacifications noted in both the maxillary sinuses (as indicated by orange arrows) and in the right half of the mandible (as indicated by green arrow) for which computed tomography paranasal sinus was suggested

- A focal cortical breach was noted at the inferolateral aspect with no soft tissue extension
- Mild maxillary sinus mucosal thickening was noted. Another well-defined cystic lesion measuring 3.0 × 1.2 × 2.0 cm is noted at the angle of the right mandible with an impacted tooth (3rd molar) within. The cyst appears to be pericoronal to the tooth [Figure 3]. The frontal sinus and frontal sinus drainage pathway (FSDP) are normal osteomeatal unit is patent.

Left Side

- A large uni-locular expansile cystic lesion measuring 2.9 × 2.7 × 3.6 cm (AP × ML × CC) with heterogenous internal contents (fluid, soft-tissue densities-? debris, and calcifications) is seen almost entirely filling the maxillary sinus and contiguous with the alveolar process of maxilla [Figure 2a and b]
- An unerupted tooth (third molar) is seen at the posterior aspect of the cystic lesion. The cyst appears to be pericoronal in relation to the tooth [Figure 2c].
- Focal cortical breaches were noted at the inferolateral aspect with no soft-tissue extension.
- Mild maxillary sinus mucosal thickening was noted
- Frontal sinus and FSDP are normal
- Osteomeatal unit is patent
- Mild mucosal thickening was noted in the ethmoid and sphenoid sinuses. No significant anatomic variants were noted. Extensive dural calcifications were noted along the falx cerebri, tentorium cerebelli, and bilateral cerebral convexities [Figure 4a and b].

Impression

Based on the above imaging features the following differential diagnosis was given:

- Multiple odontogenic keratocyst in association with Gorlin syndrome
- Multiple dentigerous cysts

Management Approach

The patient then underwent curettage of the OKC. The sample was then sent for histopathological analysis. Due to the familial tendency, all his family members were also subjected to clinical and radiological examinations. However, they did not show any relevant abnormalities.

Follow-up

Histopathology: Right maxilla, left maxilla, right mandible: The given sections show a cystic lumen lined by corrugated parakeratinized stratified squamous epithelium with a palisading basal layer. The fibrous capsule contains collagen fibers and plenty of inflammatory cell infiltration. Areas of daughter cysts and epithelial remnants are seen. Bony trabeculae are also presented.

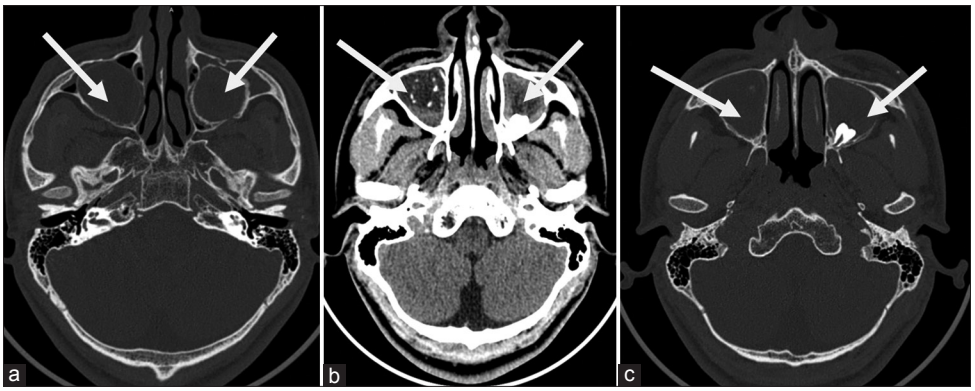

Figure 2: (a) CT PNS bone window showing bilateral large uni-locular expansile cystic lesion. (b) CT PNS bone window showing an unerupted tooth (third molar) at the posterior aspect of the cystic lesion within the left maxillary sinus(as indicated by arrow). The cyst appears to be pericoronal in relation to the tooth. (c) CT PNS soft tissue window showing heterogeneous internal contents of uni-locular expansile cystic lesions (as indicated by arrows) which is seen almost entirely filling the maxillary sinus

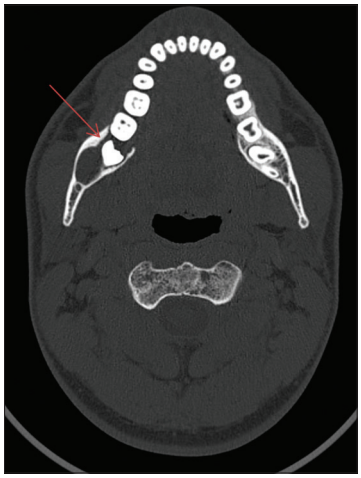

Figure 3: Computed tomography paranasal sinus bone window showing well-defined cystic lesion at the angle of right mandible with an impacted tooth (3RD molar) within, the cyst appears to be pericoronal in relation to the tooth (as indicated by red arrow)

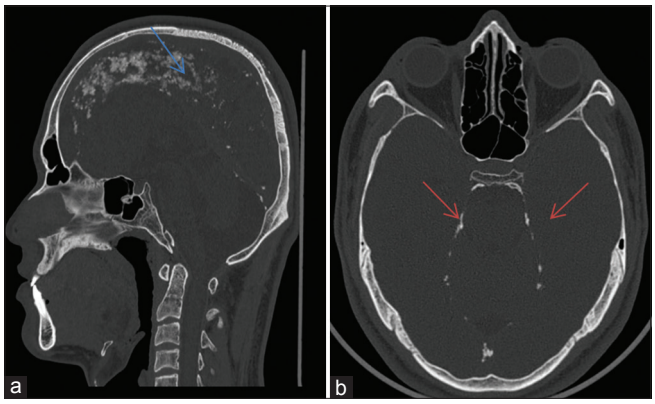

Figure 4: (a and b) Computed tomography paranasal sinus bone window showing extensive dural calcifications along the falxcerebri (as indicated by blue arrow) and tentorium cerebelli (as indicated by red arrow)

- Hard tissues: The given section shows normal lamellar bone tissues
- Impression: Right maxilla: Inflammatory OKC
- Left maxilla: Inflammatory OKC
- Right mandible: Inflammatory OKC.

Advised to rule out Gorlin–Goltz syndrome. Following the histopathological report, a diagnosis of Gorlin syndrome was made.

Long-term Monitoring: The patient was asked to follow-up.

DISCUSSION

The case presented involves a 25-year-old male patient who presented with complaints of facial pain and delayed tooth eruption. The patient then underwent X-ray PNS which showed dense radiopaque opacifications noted in both the maxillary sinuses and in the right half of the mandible. Curvilinear calcification extending to the maxilla for which CT PNS was suggested. CT PNS of the patient showed. Multiple odontogenic keratocyst along with falx calcification. The above imaging features had raised suspicion of Gorlins syndrome for which he underwent excision and curettage of odontogenic keratocytes. The histopathological report confirmed the diagnosis as OKC. Based on these a diagnosis of Gorlins syndrome was made.

Gorlin syndrome also known as nevoid basal cell carcinoma syndrome, basal cell nevus syndrome, syndrome of jaw cysts, and jaw cyst-basal cell nevus-bifid rib syndrome is a rare autosomal dominant disorder.[3] These patients are more prone to develop nevoid basal cell carcinoma syndrome; however, all affected adults did not have basal cell carcinomas.[4] The condition is known as Gorlin syndrome because of Gorlin's contributions to the understanding of the condition. Clinical features of Gorlin–Goltz syndrome most commonly arise in the first, second, or third decade.

Table 1: Major and minor criteria for the diagnosis of Gorlin syndrome

Major criteria	Minor criteria
1. Basal cell carcinoma: >2 or 1 under the age 20	1. Macrocephaly
2. Odontogenic keratocysts	2. Frontal bossing, cleft lip or hypertelorism
3. Palmar pits: 3 or more	3. Sprengel deformity, pectus excavatum or pectus carinatum, syndactyly
4. Bilamellar calcification of the falx cerebri	4. Bridging of the sella turcica, hemivertebrae, flame-shaped osseous radiolucencies
5. Rib anomalies: bifid rib (see image), fused, splayed	5. Ovarian fibroma
6. First degree relative with Gorlin syndrome	6. Medulloblastoma

In the present case, the features were identified in the second decade and no significant abnormality was noted in the family members. Although autosomal dominant, it can also arise spontaneously or can have a variable phenotypic penetration.[5] A similar pattern of inheritance was observed in our cases. Gorlin and Goltz in 1960 gave a complete description of the syndrome.[1] This syndrome is associated with a wide spectrum of developmental anomalies and neoplasms. The most common and significant features are summarized in Table 1.

CONCLUSION

The clinical presentation, imaging findings on X-ray PNS and CT PNS, and subsequent histopathological evaluation confirmed the diagnosis of OKC in association with Gorlins syndrome in this patient.

The present case highlights the importance of awareness of this rare syndrome, especially in young patients.

To arrive at an early diagnosis of the syndrome, specialists should carry out clinical, radiographic testing in early ages of life.

Guidelines for follow-up have been established and include the following: Neurological examination twice yearly, cerebral MRI once in a year for 1–7 years of age, orthopantomogram every 12–18 months starting at the age of 8 years, yearly skin examination and cardiologic examination according to the signs and symptoms.

The families of the patients with NBCCS should be examined and genetic counseling should be offered, as it is inherited as an autosomal dominant disorder.

Early diagnosis of NBCCS is crucial for the patients and their families, considering the risk of developing malignancies such as medulloblastoma and aggressive skin cancers. It is very important to screen for medulloblastoma in the early years of life in patients with NBCCS as it may be a potential cause of early death and early diagnosis is important to give adequate genetic advice.

A negative family history could hamper the early clinical recognition of patients with NBCCS: Nonetheless, patients can be diagnosed during early childhood if the clinician is also aware of the minor clinical signs of the disease.

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