

Case Report

A case report of Gorlin–Goltz syndrome as a rare hereditary disorder

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Abstract

Gorlin–Goltz syndrome is an autosomal dominant and a rare hereditary disease. Diagnosis of this syndrome is based on major and minor criteria. We report a Gorlin–Goltz syndrome in a 25-year-old male who was presented with progressive pain of maxilla and mandible over 5 years. The pain was diffuse and compatible with expansile cyst in alveolar ridges on panoramic radiography. In physical examination, he had coarse face and prognathism. Computer tomography of face revealed two expansile maxillary and one mandibular cyst. Calcification of entire length in falx and tentorium were detected in bone window.

KEYWORDS: Keratogenic Cyst, Falx, Tentorium, Gorlin-Goltz Syndrome.

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Gorlin–Goltz syndrome is a rare hereditary disorder. Patients have multiple manifestations that include basal cell carcinoma (BCC), Odontogenic cyst, skeletal anomalies, prognathism, calcification of tentorium and falx. These patients are predisposed to various neoplasms, such as medulloblastoma, meningioma, fibrosarcoma, cardiac fibroma. We will report a case of Gorlin–Goltz syndrome.

Case report

A 25-year-old male was presented with progressive pain of alveolar ridge of maxilla and mandible over 5 years. The pain began 5 years ago and has been aggravated during the last 10 months. The pain was diffuse and compatible with expansile cyst in maxillary and mandibular ridges on panoramic radiography. He was admitted 5 times to Azahra hospital for removal of maxillary and mandibular odontogenic cysts during the previous year. He was referred to our institution for taking 3-Dimensional face multislice CT-scan. In physical examination he had coarse face and prognathism. Non-enhanced Computer tomogra-

phy of face in axial section revealed two expansile maxillary and one mandibular cyst (Figure 1, 2, 4). In coronal and sagittal sections of bone window, calcification in entire length of falx and tentorium were detected (Figure 3, 4). In axial section, abnormal bony excrescence arising from left aspect of upper cervical vertebra was also observed (Figure 5). At surgery, three expansile jaw cysts were removed and the histological examination revealed that they were Keratogenic cysts.

Discussion

Gorlin–Goltz syndrome or nevoid basal cell carcinoma (NBCCS) is an autosomal dominant disorder and a rare hereditary disease.¹ It was first explained in the 1950s and 1960s by Gorlin and Goltz.² It is seen in males and females equally and in whites more than blacks.³

The estimated prevalence varies from 1/57000 to 1/256000.⁴ This syndrome is due to tumor suppressor gene mutation in the long arm of chromosome 9 (q22.3-q31).¹ Approximately, 35-50% of cases are due to new mutations.² Patients have different type of signs and symptoms that include basal cell carcinoma

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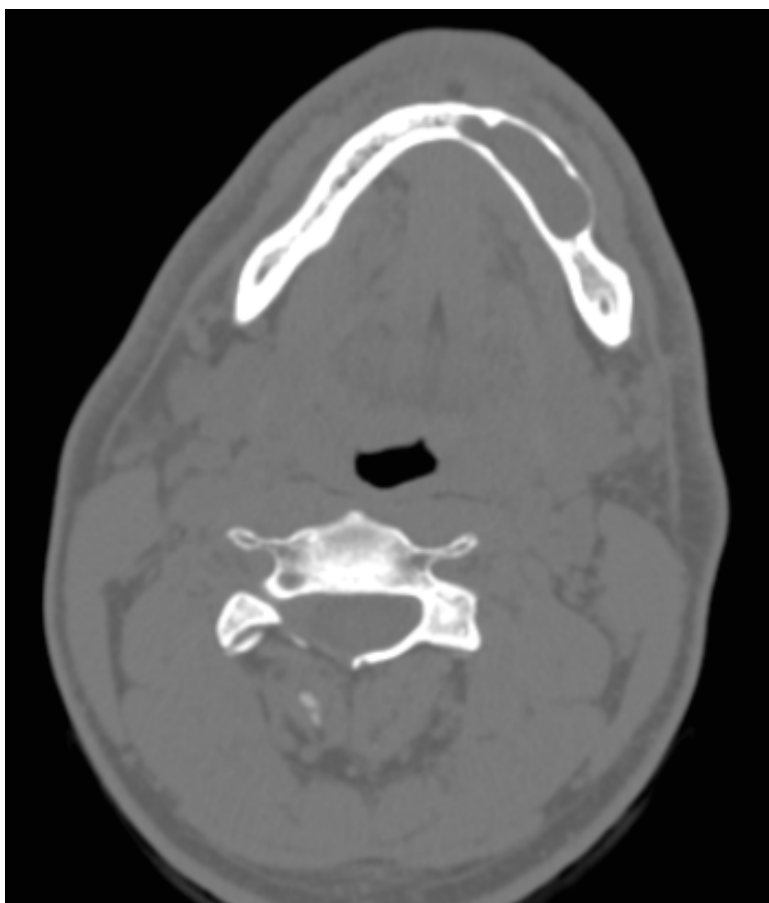


Figure 1. Axial section shows one expansile odontogenic cyst in body of mandible



Figure 2. Axial section shows two expansile odontogenic cyst in maxillary alveolar ridge

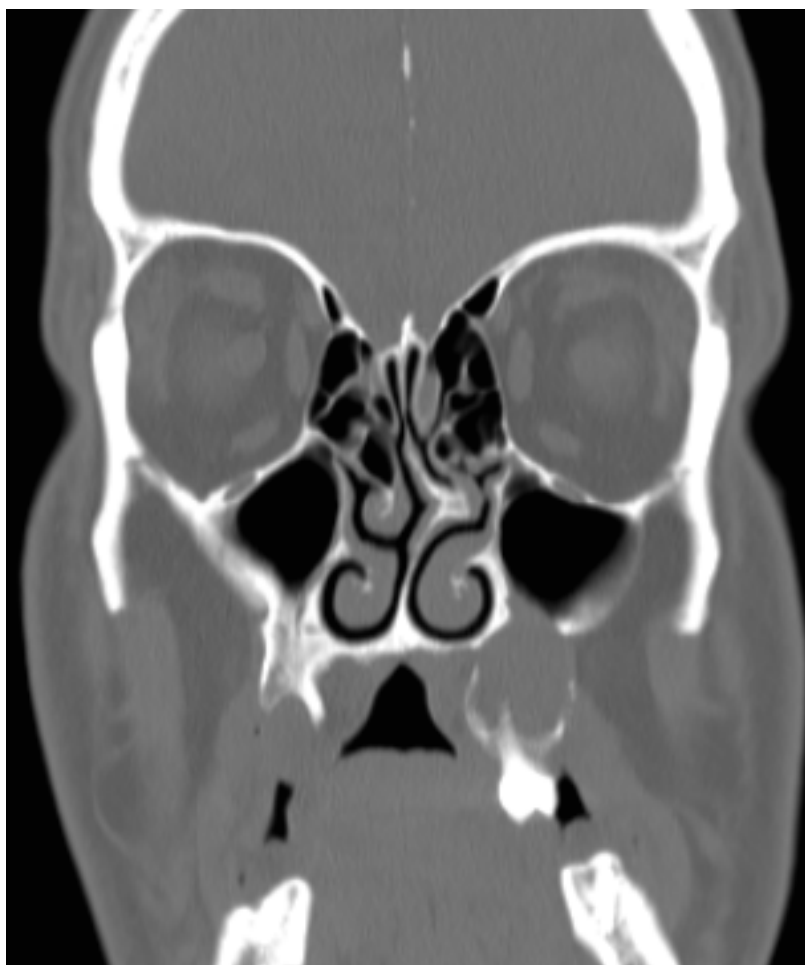


Figure 3. Coronal section shows calcification of falx cerebri and one expansile maxillary odontogenic cyst

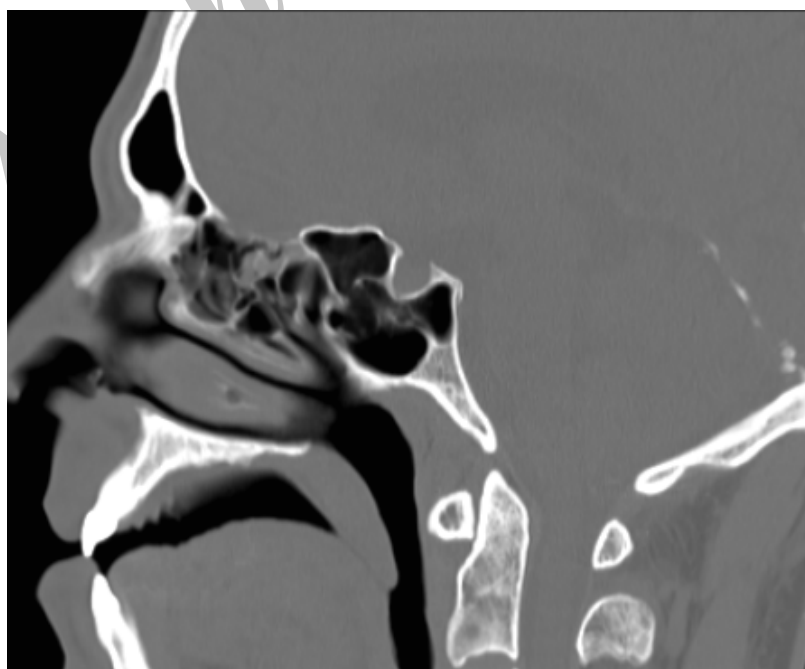


Figure 4. Sagittal section shows tentorial calcification



Figure 5. Axial section shows bony excrescence arising from body of upper cervical vertebra.

(BCC), odontogenic cyst, skeletal anomalies, prognathism, calcification of tentorium and falx.¹ Because of the mutation of tumor suppressor gene, patients are predisposed to various neoplasm, such as medulloblastoma, meningioma, fibrosarcoma and cardiac fibroma. Therefore, correct diagnosis is necessary to prevent fatal complications. Evans et al⁵ first described major and minor criteria for diagnosis of this syndrome, then were modified by Kimonis et al.⁶

Major and minor criteria are

Major criteria: more than two basal cell carcinomas or one appearing in patient < 20 years old; odontogenic cyst of the jaw that confirmed by histopathology; three or more palmar or plantar pits; calcifications of falx cerebri and tentorium; first-degree relative with Gorlin-Goltz syndrome.

Minor: macrocephaly, congenital and skeletal anomalies (frontal bossing, coarse facies, prog-

nathism; moderate or severe hypertelorism, cleft lip and palate, vertebral anomalies); other skeletal anomalies (Sprengel deformity, marked pectus deformity, and marked syndactyly of the digits); bridging of the sella turcica, ovarian fibroma or medulloblastoma; neurological anomalies (dysgenesis of corpus callosum, mental retardation).

Two major criteria or 1 major and 2 minor criteria are necessary to diagnose Gorlin-Goltz syndrome.⁶ In our case, two major criteria (keratogenic cyst that was approved with histological exam, calcification of falx cerebri and tentorium) and two minor criteria (vertebral anomaly and coarse face) were seen.

BCCs are seen in 50- 97% of these cases.⁷ Face and neck in first and chest in second are affected. BCC causes local invasion and brain metastases. Odontogenic cysts are often multiple and presented in about 75% of patients, recurrent operation and removal is necessary.² Fine needle aspiration cytology (FNAC) is also

useful in preoperative diagnosis of odontogenic keratocyst.⁸ Medulloblastoma is detected in 5-10% of cases, and is a fatal complication.¹ Early recognition of this syndrome is necessary for appropriate management. Sun ray protection and vitamin A may have protective effect against skin cancer growth.⁹ Repeated surgical excision of odontogenic cysts is necessary. Periodic abdominopelvic ultrasound and brain MRI are recommended for early detection of malignancies.

Conflict of Interests

Authors have no conflict of interests.

Authors' Contributions

Authors NT and MS carried out the study; prepared and wrote the manuscript. All authors have read and approved the content of the manuscript.

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Conclusion

After spinal anesthesia, the painless period is approximately 2 to 3 hours and then LBP and RLP appear. The use of EAT may have a protective effect on late adhesive irritating scar tissue and when impregnated with bupivacaine or methylprednisolone acetate could increase the duration of painless interval. According to our data bupivacaine, was effective on LBP relief and steroid was effective on RLP relief during the first 12 hours after surgery.