

**Case report****Gorlin-Goltz syndrome – An unusual case report**Thayumanavan B<sup>1</sup>, Jeyanthikumari T<sup>2</sup>, Meghalapriya P<sup>3</sup>

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**ABSTRACT**

Gorlin-Goltz syndrome is an uncommon autosomal dominant disorder manifesting as a multisystemic disease. Keratocystic odontogenic tumor (KCOT) is considered as the most consistent feature of this syndrome. Dentists play a key role in making early diagnosis of this syndrome. Here we present a case of Gorlin-Goltz syndrome identified by multiple multilocular radiolucencies in the mandible. A review of literature of different diagnostic criteria for Gorlin-Goltz syndrome is also discussed.

**Keywords:** Gorlin-Goltz syndrome, nevoid basal cell carcinoma syndrome, keratocystic odontogenic tumor, multilocular radiolucency

**Introduction**

Gorlin-Goltz syndrome was first described by Jarisch and White in 1894. It is characterized by numerous basal cell carcinomas, keratocysts and musculoskeletal malformations and is inherited in an autosomal dominant pattern. [1] The syndrome is caused due to mutations in the tumor suppressor gene, PTCH (patched) mapped to chromosome 9q21-23. Defect in the Hedgehog (Hh) signaling pathway has also been proposed as an underlying mechanism. [1] When two mutagenic hits occur, both the alleles of PTCH gene are inactivated leading to tumor growth. Malformations and their variability in the patients may be ascribed to single germ cell hit. [2] This article describes a case of Gorlin-Goltz syndrome and gives a brief account of various diagnostic criteria.

**Case report**

An 18-year old patient reported to Sathyabama University, Dental College and Hospital with the chief complaint of pain in the right and left lower back teeth region since one year. On clinical examination, multiple nevi were found on the chest and shoulders (Fig 2).

OPG revealed multiple multilocular radiolucencies in the mandible (Fig 3). Chest X-ray showed bifid ribs (Fig 4).

Radiolucent lesion in the mandible was curetted and histopathologically examined.

Microscopically, parakeratinized stratified squamous epithelium of uniform thickness consisting of 6-10 layers with flat epithelial-connective tissue interface and corrugated parakeratin surface was seen. Epithelium showed features of moderate dysplasia. Basal layer of epithelium

showed polarized palisading nuclei with tombstone appearance. Connective tissue revealed multiple daughter cysts (Fig 5). Therefore, histopathological findings were consistent with keratocystic odontogenic tumor (KCOT).

Based on the clinical, radiographic and histological findings, diagnosis of Gorlin-Goltz syndrome was made.



Fig.1 Profile view of the patient showing a nevus in the face



Fig.2 Multiple nevi found on the chest and shoulders

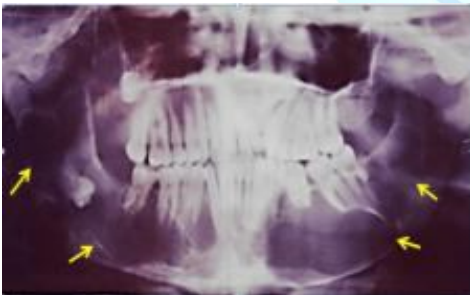


Fig.3 OPG revealed multiple multilocular radiolucencies in the mandible



Fig.4 Chest X-ray showed bifid ribs

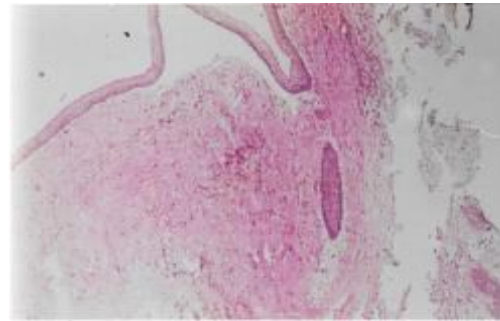


Fig.5 Histopathology showing features of KCOT with daughter cysts

### Discussion

The classic triad consisting of multiple basal cell carcinoma, keratocystic odontogenic tumors (KCOTs) in the jaws and bifid ribs which are the characteristic features of this syndrome was described by Gorlin and Goltz.<sup>[2]</sup> Other names suggested are nevoid basal cell carcinoma syndrome (NBCCS), hereditary cutaneomandibular polyoncosis and bifid rib syndrome.<sup>[3]</sup> Calcification of the falx cerebri, spine and rib anomalies, palmar and plantar epidermal pits, relative macrocephaly, facial milia, frontal bossing, ocular malformation, medulloblastomas, cleft lip and/or palate, and developmental malformations are other reported features of the syndrome.<sup>[2]</sup>

Numerous diagnostic criteria have been proposed for the diagnosis of Gorlin-Goltz syndrome. This article presents a review of literature of various diagnostic criteria.

**Diagnostic criteria proposed by Evans et al (1993)<sup>[4]</sup>:**

Diagnosis requires the presence of two major or one major and two minor criteria.

#### Major criteria:

- Multiple basal cell carcinoma or one occurring under the age of 20 years.
- Histologically proven OKCs of the jaws.
- Palmar or plantar pits (three or more).
- Bilamellar calcification of the falx cerebri.

- Bifid, fused or markedly splayed ribs.
- First-degree relative with Nevroid Basal Cell Carcinoma syndrome.

**Minor criteria:**

- Macrocephaly (adjusted for height).
- Congenital malformation: cleft lip or palate, frontal bossing, coarse face, moderate or severe hypertelorism.
- Other skeletal abnormalities: Sprengel deformity, marked pectus deformity, marked syndactyly of the digits.
- Radiological abnormalities: bridging of the sella turcica, vertebral anomalies such as hemivertebrae, fusion or elongation of the vertebral bodies, modeling defects of the hands and feet or flame shaped hands or feet.
- Ovarian fibroma.
- Medulloblastoma.

**Evan's diagnostic criteria modified by Kimonis et al (1997)<sup>[5]</sup>:**

Diagnosis requires the presence of two major or one major and two minor criteria.

**Major criteria:**

- More than two BCCs or one BCC at younger than 30 years of age or more than 10 basal cell nevi.
- Any odontogenic keratocyst (proven on histology) or polyostotic bone cyst.
- Three or more palmar or plantar pits (present in about 65% of patients).
- Ectopic calcification: Lamellar or early at younger than 20 years of age.
- Falx cerebri calcification.
- Positive family history of nevoid BCC.

**Minor criteria:**

- Congenital skeletal anomalies; fused, splayed, missing, or bifid ribs, wedged or fused vertebrae.
- Occipital–frontal circumference more than 97%.
- Cardiac or ovarian fibroma.
- Medulloblastoma.
- Lymphomesenteric cysts.

- Congenital malformations such as cleft lip or palate, polydactylism or eye anomalies (cataract, coloboma, microphthalmus).

**Diagnostic criteria proposed by Manfredi et al (2004)<sup>[6]</sup>:**

Presence of two major, or one major and two minor criteria is required to establish the diagnosis of Gorlin-Goltz syndrome.

**Major criteria:**

- Presence of more than two basal cell carcinomas or one under the age of 20 years
- Histologically-proven KCOT of the jaw
- Cutaneous palmar or plantar pits
- Bifid, fused or markedly splayed ribs

**Minor criteria:**

- Orofacial congenital malformations (cleft lip or palate, frontal bossing or moderate or severe hypertelorism)
- Skeletal and radiological abnormalities (bridging of the sella turcica and vertebral anomalies)
- Ovarian fibroma
- Medulloblastoma

**Diagnostic criteria modified by Bree et al (2011)<sup>[7]</sup>:**

Diagnosis of Gorlin-Goltz syndrome can be made in the presence of: a) 2 major criteria, b) 1 major criteria and molecular confirmation or c) 1 major and 2 minor criteria.

**Major criteria:**

- Excessive numbers of basal cell carcinomas out of proportion with prior sun exposure and skin type or < 20 yrs of age
- Odontogenic keratocysts of the jaws prior to 20 yrs of age
- Palmar or plantar pitting
- Lamellar calcification of the falx cerebri
- Medulloblastoma, typically desmoplastic
- 1st degree relative with Gorlin-Goltz syndrome

**Minor criteria:**

- Rib anomalies
- Other specific skeletal malformations and radiologic changes (i.e. vertebral anomalies, kyphoscoliosis, short 4th metacarpals, postaxial polydactyly)
- Macrocephaly
- Cleft lip and/or palate
- Ovarian/cardiac fibroma
- Lymphomesenteric cysts
- Ocular abnormalities (i.e. strabismus, hypertelorism, congenital cataracts, glaucoma, coloboma)

In our case, two major criteria viz. histologically proven KCOT and bifid ribs were fulfilled supporting the diagnosis of Gorlin-Goltz syndrome.

**Conclusion**

When multiple multilocular radiolucencies are encountered in jaws, the dental surgeon must consider Gorlin Goltz syndrome in the differential diagnosis and must take necessary steps to check for other features. In our case, multiple multilocular radiolucencies in the mandible and multiple nevi prompted us to proceed for chest X-ray.

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