CASE OF THE MONTH

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HISTORY

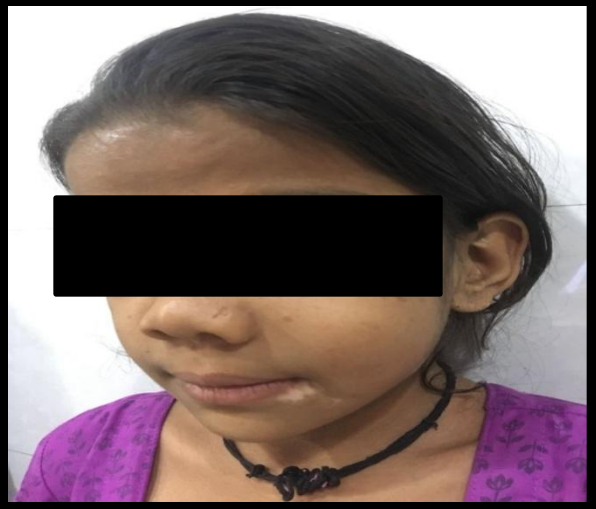
- A 13-year-old female visited our hospital with a chief complaint of swelling on lower anterior region of the jaw since 10 days. Swelling was approximately 2 cm × 1 cm in size, soft to firm in consistency and tender on palpation. Intraoral examination revealed his deciduous teeth were still present and few permanent teeth were missing.

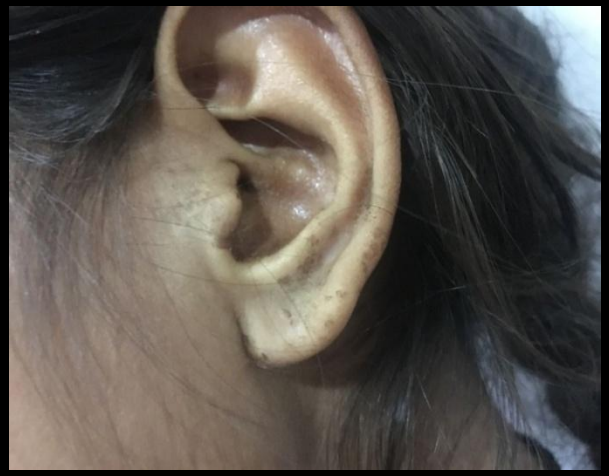
The orthopantomogram was advised, which revealed three cystic lesions in the mandible and two involving the maxillary sinus on either side with the displaced permanent teeth. Owing to the presence of multiple cysts like lesions in the jaw, GGS was suspected and further investigations were carried out.

Cont…

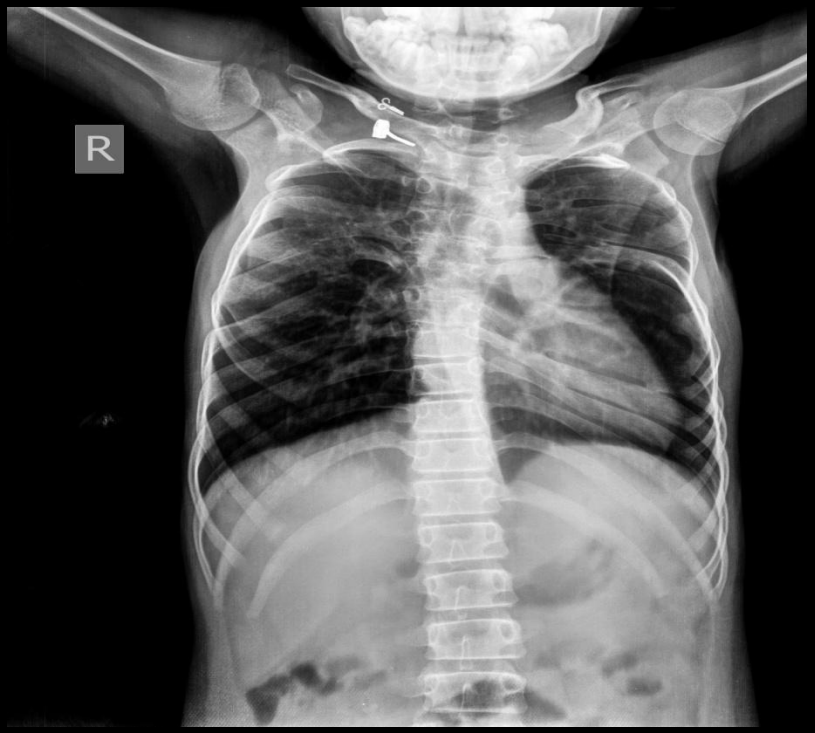
•The radiograph of the skull showed bilamellar calcification of the falx cerebri . Physical examination revealed macrocephaly with a head circumference of 92 cm (normal for a 13-year-old female is 54-57 cm), frontal bossing, depressed nasal bridge, hypertelorism and mandibular prognathism . Palmar pits were brown coloured and measuring 1-3 mm in diameter . No other anomalies of the skeletal, cardiovascular, or central nervous system were present. On the basis of clinical findings, diagnosis of GGS was made. Prior to the surgical procedure, an arch bar was placed on the lower jaw to prevent fracture of the mandible as well as to splint the unsupported permanent teeth. The cyst enucleation was done under general anesthesia via intraoral approach. After the cystic lesions enucleated, large areas of bone loss were seen and the displaced permanent teeth were visible on the floor of the cystic cavity . The enucleated cystic lining was sent for histopathological examination.

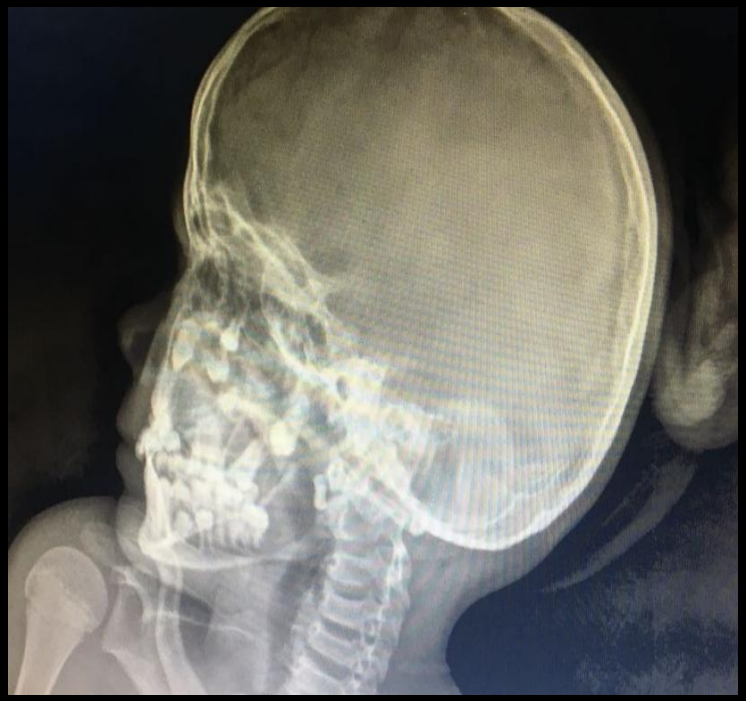
CLINICAL IMAGE





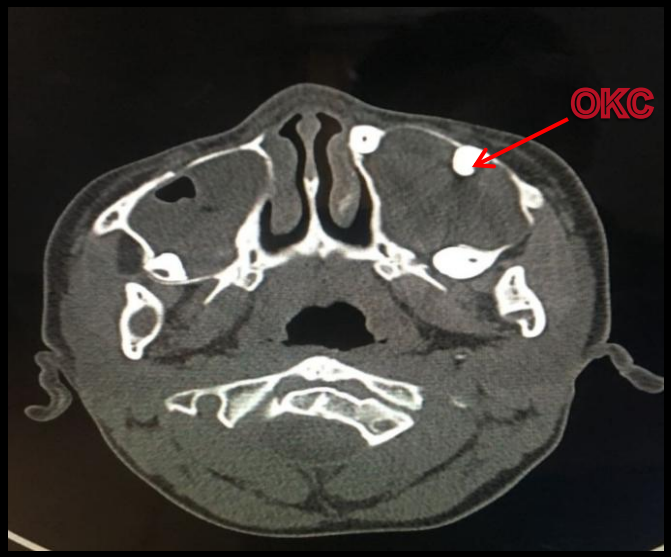
X-RAY IMAGE

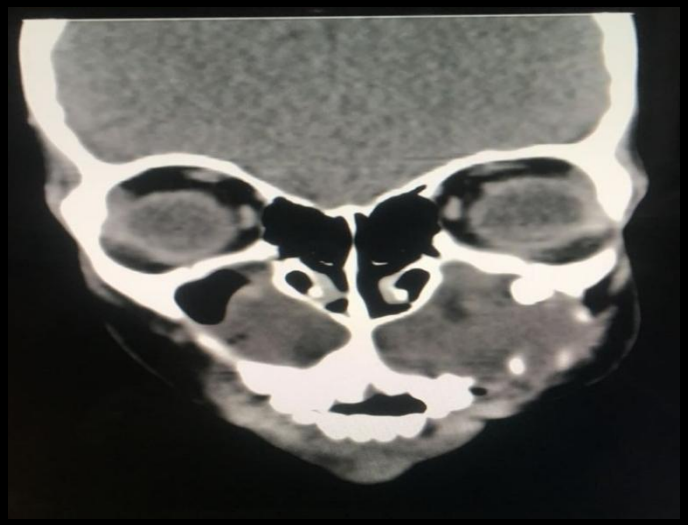


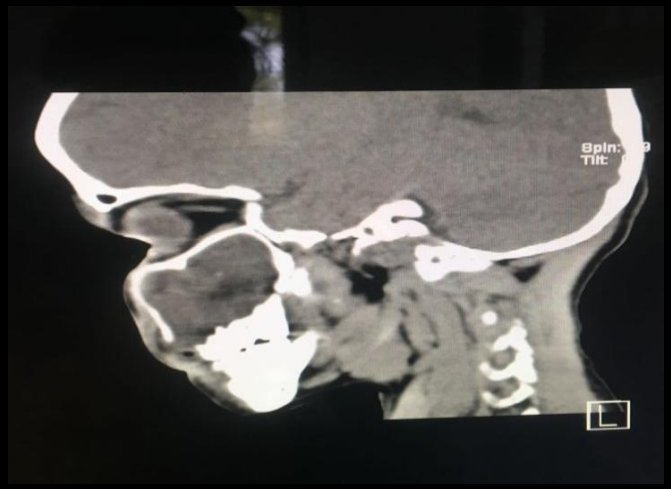


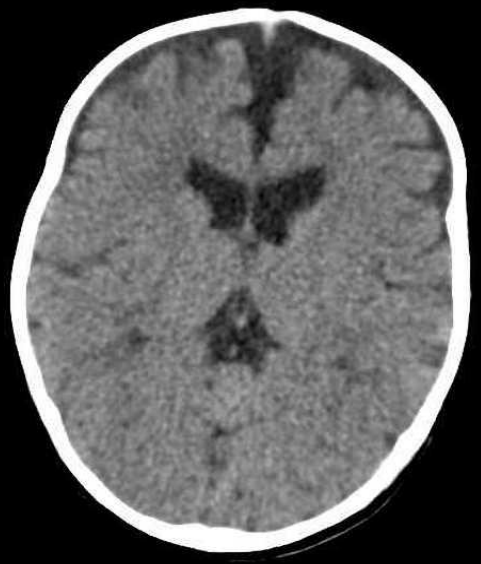
Chest X-Ray on left side shows multiple bifid as well As fused ribs on left side. On lateral skull (right side) falx calcification is present.

CT IMAGES









DIAGNOSIS

Gorlin-Goltz syndrome

DISCUSSION

•Gorlin-Goltz syndrome (GGS) also known as nevoid basal cell carcinoma syndrome (NBCCS) is an autosomal dominant inherited disorder.

• The incidence of this disorder is estimated to be 1 in 50,000-1,50,000 in the general population, varying by region.It appears in all ethnic groups, but most often in whites; males and females are equally affected .

• The pathogenesis of GGS is attributed to abnormalities linked to the long arm of chromosome 9 (q22.3-q31). It has been reported that loss of human patched gene (PTCH1 gene), which is a tumor suppressor gene, could be the molecular origin of the syndrome.Human patched gene (PTCH1 gene) is significant for embryonic structuring and cellular cycle and thus its mutation comprises a key event for the development of this syndrome.

GGS is characterized mainly by the presence of multiple basal cell carcinomas (BCC), odontogenic keratocysts (OKCs) of the jaw, palmar pits and ectopic calcifications of the falx cerebri. More than 100 minor criteria have been described.

• GGS has rarely been reported from India. We report here one such patient diagnosed at our hospital.

The major criteria are-

- Multiple BCC 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 NBCCS.

The Minor diagnnostic 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 syndactily 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.

• In general, OKCs are more common in the adult life, the peak incidence being the third decade of life. However, in the GGS, OKC occurs at a much younger age Lo Muzio *et al*. observation showed that OKCs were often the first sign of GGS in 78% of the cases and they could be detected in patients younger than 10 years of age.

•OKCs, which are relatively common in GGS, are diagnosed with dental panoramic radiography. Keratocysts may show a uni - or multilocular pattern and the cystic spaces may have a smooth or scalloped border. and found significant differences between syndrome keratocysts and non-syndromic keratocysts. Syndrome keratocysts were found to have a markedly increased number of satellite cysts, solid islands of epithelial proliferation, odontogenic rests within the capsule, and mitotic figures in the epithelial lining of the main cavity. In our patient, the lining of the OKCs revealed the presence of parakeratinized uniform squamous epithelial lining with multiple satellite and daughter cysts in the connective tissue wall, thus indicating the association with GGS.

CONCLUSION

- The GGS is a well-known syndrome with a variety of findings in and outside the head and neck region. The OKC is the most common manifestation of this syndrome.

- Early diagnosis of this syndrome is important to reduce the associated complications, which are life-threatening and also to provide genetic counselling to the parents.

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