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Gorlin-Goltz Syndrome: A case report

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Abstract: Gorlin -Goltz syndrome or Jaw cyst-basal cell nevus-bifid rib syndrome involves multi organ systems. The most common findings are multiple odontogenic keratocysts in the jaws and basal cell nevi on the skin that have an early age of onset. These multiple odontogenic keratocysts require aggressive treatment at the earliest because of the damage associated with them and the high recurrence rate. Several cases of ameloblastoma have been reported in these cysts. Recurrence of these lesions should be taken into consideration while explaining the prognosis to the patient. We present a case report of Gorlin-Goltz syndrome characterized by multiple odontogenic keratocysts involving both the jaws.

Keywords: Gorlin-Goltz syndrome, basal cell nevus, odontogenic keratocysts, palmar pits, Carnoy's solution.

Introduction: Gorlin-Goltz syndrome also known as Jaw cyst-basal cell nevus-bifid rib syndrome is a rare hereditary condition with Autosomal Dominant inheritance with almost 100% penetrance. This was first described by Binkely and Johnson in 1951 and later reviewed by Gorlin and co-workers. Patients with this syndrome have several organ anomalies, many of which are subtle. This syndrome affects skeletal, skin, eye, reproductive and neural system[1]. Patients tend to develop multiple neoplasms , including basal cell carcinomas and medulloblastomas; and are extremely sensitive to ionizing radiation, including sunlight. Although all these features are rarely observed in a single patient, often these patients visit a dental hospital with the chief complaint of jaw swelling. In this report, a similar case of Gorlin syndrome was diagnosed, treated and followed at this hospital.

Case Report: An 18 year old male patient reported to the Department of Oral and Maxillofacial Surgery, Government Dental College, Kottayam on 21/01/08 with the chief complaint of an extra oral painless swelling on the lower right cheek region (Fig 1a). The gait was normal. The duration of the swelling was six weeks. There was no history of any such previous occurrence. On examination, extraorally a bony hard, non-tender swelling was present extending from the anterior region of the mandible to the angle . Intra orally, the buccal vestibule was obliterated on the right side from the region of 44 extending posteriorly along the body of the mandible (Fig 1b). The dental status of the patient - missing 17, 27. The general examination revealed palmar pits (Fig 1c), localized palmar keratosis, mild mandibular prognathism, frontal bossing and hypertelorism with wide nasal bridge (Fig 1a).

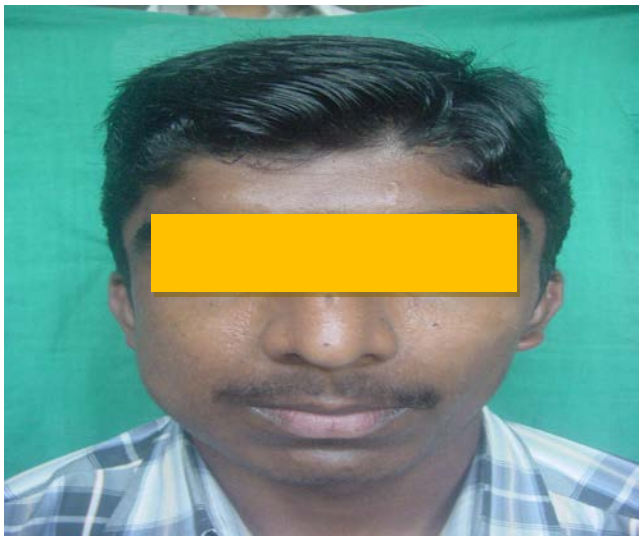

Fig. 1a: Frontal view of the patient showing mendibular prognathism and hypertelorism

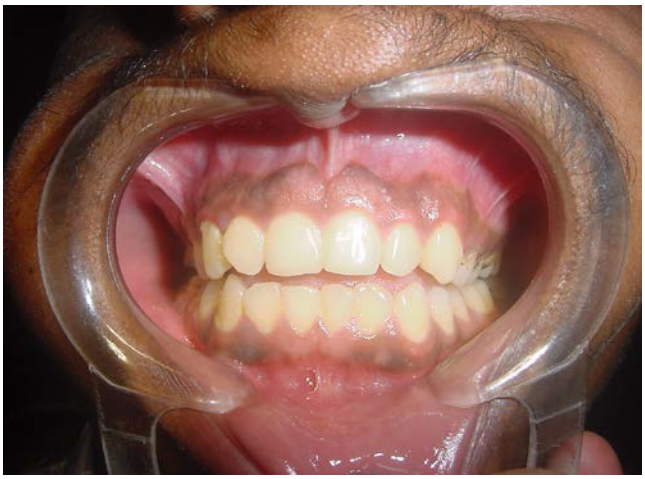

Fig 1b: Intra oral view

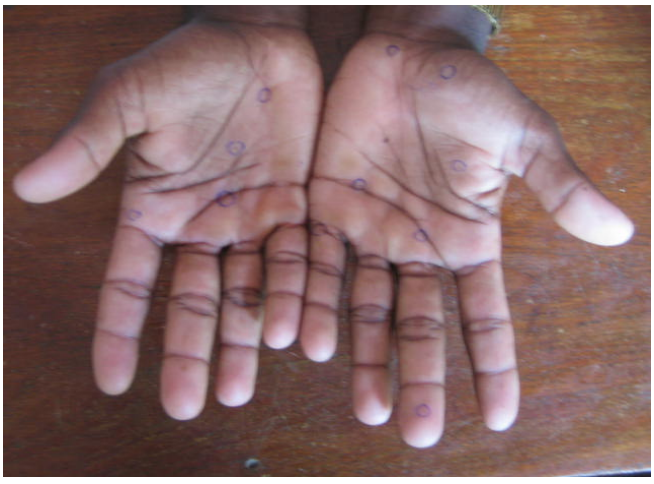

Fig 1c: Photograph showing pits on the palm (blue circles)

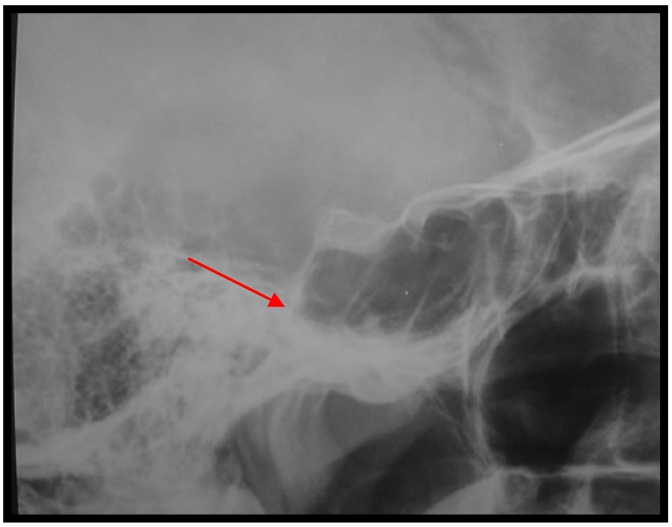

Fig 2a: Lateral view skull showing septae in sphenoid sinuses (red arrow)

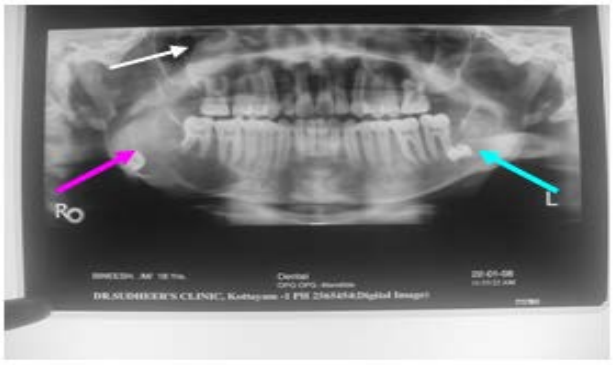

Fig. 2b: Orthopantomogram showing multiple cystic lesions

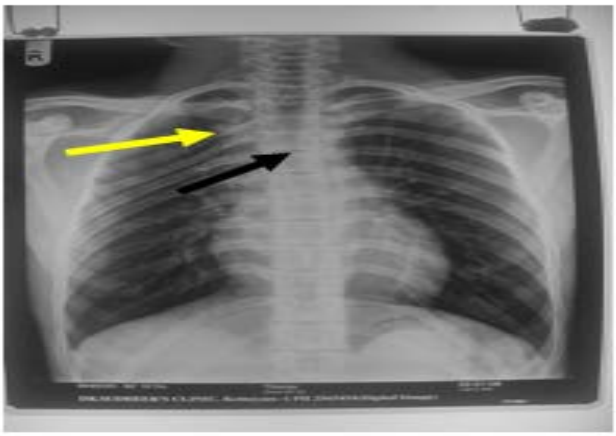

Fig. 2c: PA View of the chest showing the fused ribs

The orthopantomogram showed multiloculated medium and large size cysts involving the right maxilla(white

arrow) and bilateral mandible(pink&green arrows) associated with unerupted 38, 48 displacing them from their original positions (Fig 2b).The chest radiograph (Fig 2c) showed bilateral fused ribs, expanded right 2nd rib(yellow arrow) and fusion of 3rd and 4th thoracic vertebrae on posterior aspect(black arrow). The lateral skull X-ray (Fig 2a) showed a small pituitary fossa with calcification of interclinoid ligament and sphenoid sinuses with several septations(red arrow). The clinical and radiological correlation confirmed Gorlin-Goltz syndrome. No skin lesions in the form of basal cell nevus were present. No other systemic features associated with this syndrome were present in this patient. The parents and the siblings of the patient did not show any characteristics of this condition.

The enucleation of all the odontogenic keratocysts followed by chemical cauterisation with Carnoy's solution [only for mandibular cysts] was planned for this patient[3]. High recurrence rate of these cysts was explained to the patient. Under general anesthesia all the cysts in the mandible and maxilla were enucleated along with the removal of the involved teeth.. The contents of the cysts were found to be thick and cheesy. This was followed by the application of Carnoy's solution in each cystic cavity and primary closure. The cystic lining was sent for histopathologic examination and the report confirmed the cysts to be odontogenic keratocysts. The patient has been on regular follow up since then.

Discussion

Gorlin-Goltz syndrome was described for the first time in 1894 from Jarisch and White. The disease is an autosomal dominant disorder characterized mainly by the presence of multiple basal cell carcinomas, odontogenic keratocysts of the jaw and palmar-plantar pits. In 1960, Robert James Gorlin and William Goltz gave a complete description of the syndrome[2]. This syndrome is associated with a wide spectrum of developmental anomalies and neoplasms. Skeletal malformations are expressed by frontal bossing, prominent supraorbital ridge giving the eyes a characteristic appearance, broad nasal root, hypertelorism, mild mandibular prognathism. Other skeletal anomalies include bifid/fused ribs, involving more than one rib unilaterally or bilaterally, kyphoscoliosis, fusion of vertebrae, spina bifida and polydactyly. The prevalence of this condition is about 1 in 60 000. Gorlin syndrome is associated with multiple keratocysts in patients in the second decade of their life. In the present case one of the first signs were multiple cystic lesions involving the maxilla and mandible, which have been histopathologically diagnosed as odontogenic keratocysts. The association with odontogenic keratocysts appears in more than 90 of the cases. All the other disorders are less frequent.

Despite the name of the syndrome, multiple basal cell carcinomas occur only in 50% of the cases. Basal cell carcinomas most often involve face and non-exposed areas such as the chest and back[4]. Rarely, they involve waist and extremities. They can vary from flesh colored papules to ulcerated plaques and may be mistaken for nevi, skin tags or hemangiomas. In the present case there was no evidence of basal cell carcinomas of the skin and the case is being followed for the occurrence of such lesion later. With cases of basal cell carcinomas radiation therapy should be avoided because it causes invasion of basal cell carcinomas years later. Evans and colleagues proposed major and minor diagnostic criteria for Gorlin-Goltz Syndrome:

Major Criteria

- Two or more basal cell carcinomas in persons less than 20 years of age.
- Odontogenic keratocysts of jaw.
- Three or more palmar pits.

- Bilamellar calcification of falx cerebri.
- Bifid/fused ribs.
- First degree relative with Gorlin Syndrome

Minor Criteria

- Macrocephaly
- Congenital malformation[CL/CP,frontal bossing ,hypertelorism]
- Marked pectus deformity or syndactyly.

There should be at least two major criteria or one major and two minor criteria for the diagnosis. In the present case three of the major criteria were covered namely, odontogenic keratocysts of jaw; 3 or more palmar-plantar pits and fused ribs. The minor criteria that were covered include frontal bossing and hypertelorism.

Conclusion

It is important to make an early diagnosis and a proper management of Gorlin-Goltz syndrome, which has malignant predisposition. The guidelines for diagnosis include a family history, careful oral and skin examinations, chest and skull radiographs, panoramic radiographs of the jaw, magnetic resonance imaging of the brain. The genealogical analysis is important for the determination of the genetic risk and the prognosis.

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