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CASE REPORT

Gorlin – Goltz syndrome-a rare presentation

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ABSTRACT

The Golin-Goltz syndrome is a rare, multisystemic and autosomal dominant disease. We are describing here, a case with an unusual presentation of purulent discharge into the mouth from infected cysts in the jaw, in a 22 year old male patient. The case was associated with some of the classical radiological features described in the literature for this syndrome.

Key Message: A simple case of purulent discharge in the mouth may turn out to be a rare genetic syndrome.

Key Words: Golin-Goltz, Multiple cysts, major and minor criteria.

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Introduction

The Gorlin-Goltz syndrome is an autosomal dominant, rare, hereditary disease which involves multiple body systems [1]**.** It is also known by many names like basal cell nevus syndrome, nevoid basal cell carcinoma syndrome [NBCCS], even by a complicated name of "multiple basal epithelioma, jaw cysts and bifid rib syndrome" [2],[3]. This syndrome is caused by mutations in the tumour suppressor gene called PTCH (Patched) gene found on the chromosome arm 9q [4]**.** However, there exists a highly complex variability of symptoms in individuals showing comparable molecular alterations. This variability of phenotype is due to the interactions of genetic and environmental factors. [5],[6]. Its prevalence varies according to the population studied and the generally accepted prevalence is 1 in 60.000 inhabitants [7].

Though the identification of the mutation in the PTCH-1 gene is diagnostic of the disease, it is not done regularly. The Gorlin – Goltz syndrome is still diagnosed by a constellation of clinical and radiological signs and symptoms which can be grouped into major and minor criteria. The presence of two major criteria or one major and two minor criteria is considered to be diagnostic of the Gorlin - Goltz syndrome. The most important major criteria include

basocellular carcinomas, odontogenic keratocysts, palmar and/or plantar pits,bilamellar calcifications of falx cerebri, bifid, fused or markedly splayed ribs and first-degree relatives with Golin-Goltz syndrome [1]. Along with these major features, more than 100 minor criteria have been described. The list includes macrocephaly, congenital malformations (eg, cleft lip or palate, frontal bossing, coarse face, hypertelorism), other skeletal abnormalities (eg, Sprengel deformity, marked pectus deformity, or syndactyly of the digits), radiological abnormalities (eg, bridging of the sella turcica), vertebral anomalies (eg, hemivertebrae, fusion or elongation of the vertebral bodies), modeling defects of the hands and feet or flame-shaped lucencies of the hands or feet, ovarian fibroma, medulloblastoma, hypertelorism and mandibular prognathia [1].

Case report

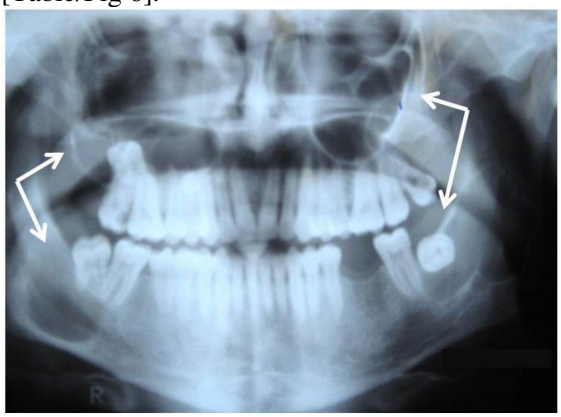
History and Clinical Presentation

A 22 years old male patient presented with the complaint of purulent discharge in the mouth without any history of trauma, tooth extraction, sinusitis, fever or pain. On examination, multiple hard swellings were felt over both the jaws. The swellings were slightly tender on the right mandible. The purulent discharge was seen oozing into the buccal cavity from the mandible on the left side. Two small nevi, one over the interscapular region and the other on the right side of the face, were seen. Other significant findings on examination were bilateral syndactyly of the second and third toes, syndactyly of the fourth and fifth phalanges and local gigantism of the second and third fingers of the left hand. Mild hypertelorism and prognathism were seen.

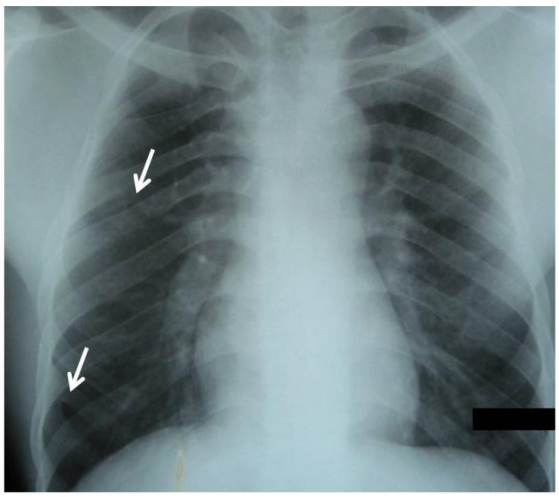
Radiographic Investigations

Orthopantomography [OPG] showed multiple, well defined, mildly expansile, odontogenic cystic lesions with well defined sclerotic margins, involving the mandible and maxillary bone symmetrically [Table/Fig 1]. The cysts in the left mandible and the right maxilla were associated with respective uninterrupted third molars [Table/Fig 1]. The right mandibular cyst was in close vicinity of the roots of the second and third molars, but without encompassing it [Table/Fig 1] . The left maxillary cyst was not associated with any tooth or tooth primordium [Table/Fig 1]. A limited skeletal survey revealed:

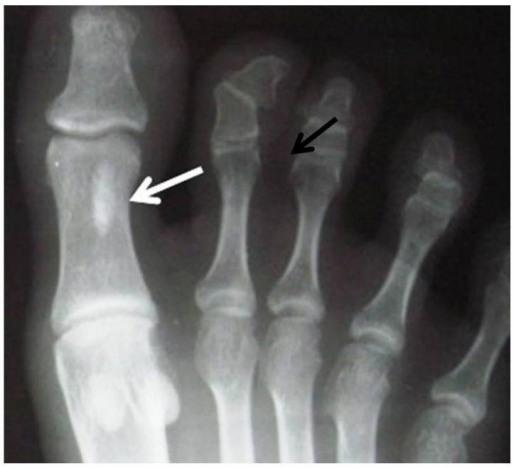
1. Chest X-Ray [PA]- Spina-bifida occulta of C7 and T1, fusion and forking of the third and fourth ribs, anterior forking of the seventh rib on the right side and forking of the fifth rib on the left side [Table/Figure 2] .
2. X-Ray of feet [AP] – Proximal soft tissue syndactyly of the right second and third toes, the bone island over the proximal phalanx of the right big toe. [Table/Figure 3].
3. X-Ray of hands [AP] – Local gigantism of the second and third digits with flexion deformity at the proximal metacarpo-phalangeal joints in the left hand. Soft tissue syndactyly of the fourth and fifth fingers with fusion of the terminal phalanges [Table/Fig 4]. Alignment deformity of the distal interphalangeal joints of the second and third fingers was seen on the right side.
4. Computed tomography (CT) scan of the jaws ~~D~~ demonstrated the cortical break in the infected left mandibular cyst and established the source of purulent discharge in the mouth~~,~~ which otherwise was not established by OPG[Table/Fig 5].
5. CT scan also showed the all important bilamellar calcification of the falx cerebri [Table/Fig 6].



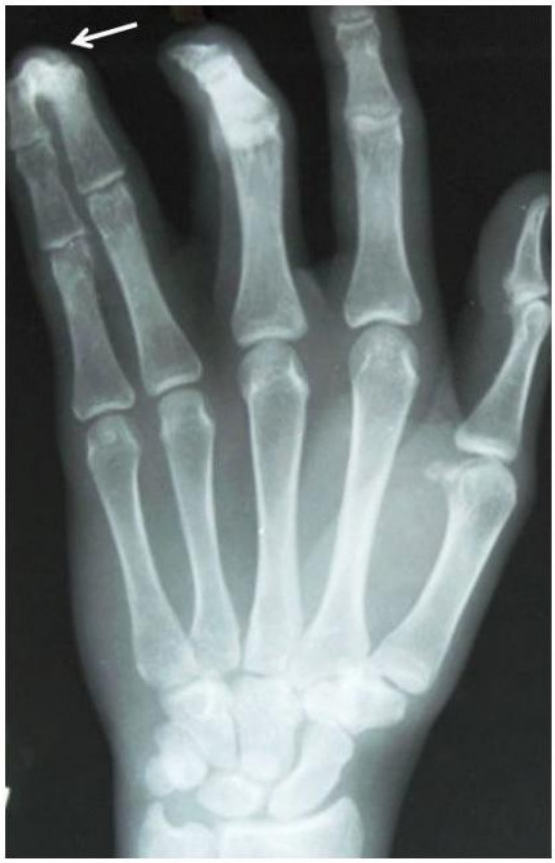
(Table/Fig 1)Odontogenic cystic lesions with well defined sclerotic margins, involving mandible and maxillary bone symmetrically.



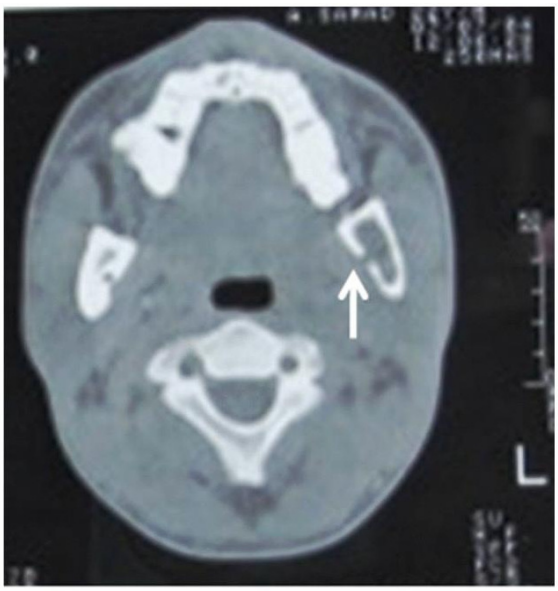
(Table/Fig 2) Fusion and forking of the third and fourth ribs, anterior forking of the seventh rib, on the right side.



(Table/Fig 3)Bone Island over proximal phalanx of right big toe [White arrow] and proximal soft tissue syndactyly of second and third toes [black arrow].

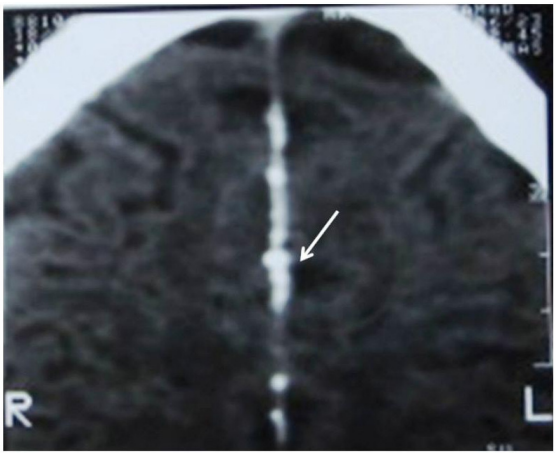


(Table/Fig 4)Soft tissue syndactyly of fourth and fifth fingers with fusion of terminal phalanges.



(Table/Fig 5)Computed tomography (CT) scan of

jaws demonstrated the cortical break in the infected left mandibular cyst.



(Table/Fig 6)Bilamellar calcification of falx cerebri

Discussion

Diagnosis was made in the present patient, based on the presence of the above criteria. Odontogenic cysts, calcification of the falx cerebri and forking of the ribs were the major features identified and the minor features present were hypertelorism, prognathism of the jaw and syndactyly of the toes and digits. The first degree relatives could not be examined. CT scan of the jaws established the cortical break in the dentigerous cyst as the source of purulent discharge into the mouth.

In Gorlin-Goltz syndromes, early diagnosis is very important because of serious complications like the malignancy of skin and brain and to give genetic advice. First degree relatives should be examined in order to make early diagnosis. Along with skeletal survey, pelvic ultrasonogrphies should be done in women [1]. All patients, especially those with odontogenic keratocysts must be examined every year [3]. These patients are susceptible to the development of basocellular carcinomas on exposure to UV light and other forms of ionizing radiations [8]. Hence, these patients must be protected from sunlight and exposure to diagnostic ionizing radiations should be limited to a minimum.

In conclusion, it is emphasized that all patients with suspicious symptoms must be investigated in detail to make an early diagnosis of the disease, in view of the severity of the complications. The patients should be encouraged to get their first degree relatives medically examined, as the Gorlin-Goltz syndrome is an autosomal dominant disease. All patients with this syndrome should undergo yearly check-ups.

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