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Case Report

Gorlin-Goltz syndrome: A rare case report

Mubeen Khan [a], *K.R. Vijayalakshmi* [b], *Preeti Rajguru* [c,*]

[a] *Professor and Head, Department of Oral Medicine and Radiology, Govt. Dental College and Research Institute, Bangalore, India*

[b] *Associate Professor, Department of Oral Medicine and Radiology, Govt. Dental College and Research Institute, Bangalore, India*

[c] *Post Graduate Student, Department of Oral Medicine and Radiology, Govt. Dental College and Research Institute, Bangalore, India*

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ABSTRACT

Gorlin-Goltz syndrome (GGS) is a rare genetic disease that is transmitted as an autosomal-dominant trait showing high level of penetrance and varying expressivity affecting multiple systems of the body. Characteristic clinical manifestations include the presence of multiple basal cell carcinomas, odontogenic keratocysts of the jaws, palmar/plantar pits and calcification of falx cerebri. Early diagnosis of GGS is of great importance due to susceptibility of affected individuals to multiple neoplasms of skin and brain (medulloblastoma) in an early age; life expectancy in GGS is not significantly altered, but morbidity from complications can be substantial. Dentist plays a crucial role in early diagnosis, which prevents recurrence and provides better survival rates from the existent diseases.

We are reporting a rare case of GGS in a 14-year-old girl who visited our institution with characteristic clinical, radiological and histological features.

1. Introduction[b]

Gorlin-Goltz syndrome (GGS) is an uncommon autosomal dominant inherited disorder with a high level of penetrance and variable expressivity due to a genetic defect or mutation in human homolog of patched (PTCH) gene,[1,2] which is a tumour suppressor gene responsible for growth, and development of normal tissue is located on long arm of chromosome no 9q 22.1-3-1,[3] but a significant fraction of cases are sporadic with no previous family history (30%).[2] This syndrome presents with a variable prevalence of 1 in 57,000 to 1 in 2,56,000 amongst general population.[1,2,4] It appears early in life after 5 years of age with equal predilection for either sex. It is characterised by a wide range of developmental abnormalities and a predisposition to neoplasms.[1,2]

* *Corresponding author.*
E-mail address: preetirajguru1@gmail.com (P. Rajguru).

[b] *Synonymous*: Basal cell nevus syndrome, Gorlin syndrome, Multiple nevoid basal cell epithelioma, Jaw cyst bifid rib syndrome, Multiple nevoid basal cell carcinomas (BCC) syndrome, Fifth phacomatosis, Hereditary cutaneomandibular polyoncosis, Epitheliomatose multiple generalisee.

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In the year 1894, Jarish and White reported the first case of this syndrome, and later on in 1960, Robert J. Gorlin and Robort W. Goltz together described the spectrum of features of this syndrome.[3]

The role of dental surgeon is very important in the diagnosis of this rare syndrome, as the early manifestations in majority of cases are multiple asymptomatic keratocysts of the jaws. Early detection results in better prognosis and prevents further cerebral complications.

2. Case report

A 14-year-old girl reported to our department with a chief complaint of swelling in the lower front jaw since 6 months. The swelling was slowly progressive in size not associated with any other symptoms and trauma. The prenatal, natal and postnatal course of development of the patient was uneventful. Past medical, dental and family history was non-contributory. There was no consanguineous marriage between the parents.

Examination of the extremities revealed multiple brownish pits of 1–2 mm in size on the palmar and plantar surfaces (as shown in Fig. 1).

On head and neck examination, hypertelorism, broad nasal bridge, everted and bulky lips were noted giving a coarse facial appearance (as shown in Fig. 2). Gross facial asymmetry noted due to swelling in the lower one third of the face.

Extra orally, a diffuse smooth surfaced, non-erythematous swelling measuring approximately 4 cm × 5 cm in its greatest dimension extending from the symphysis of the mandible to left parasymphysis region was noted. The skin over the swelling appeared normal with no secondary changes. Intraorally, buccal vestibular obliteration from 35 to 42 region with bluish translucent hue of the overlying mucosa is noted. Horizontally displaced 33 with Grade I mobility of 41, 42, 34, 35 and grade II mobility of 31, 32 and 33 (as shown in Fig. 3) and bilaterally retained maxillary deciduous canines with high arched palate (as shown in Fig. 4) is noted. On palpation, swelling was non-tender, soft, with signs of fluctuancy causing expansion of labial and lingual cortical plates. There was no paresthesia or lymphadenopathy associated with the swelling.

Fine needle aspiration yielded yellow cheesy aspirate with blood tinged.

Based on the history, clinical examination and fine needle aspiration a provisional diagnosis of odontogenic keratocyst (OKC) of anterior mandible was established, considering the

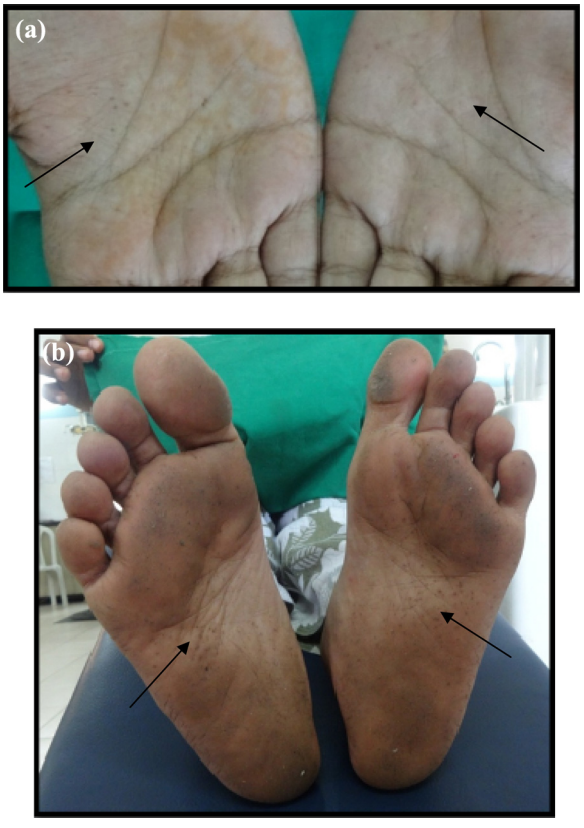

Fig. 1 – (a) Palmar and (b) plantar pits.

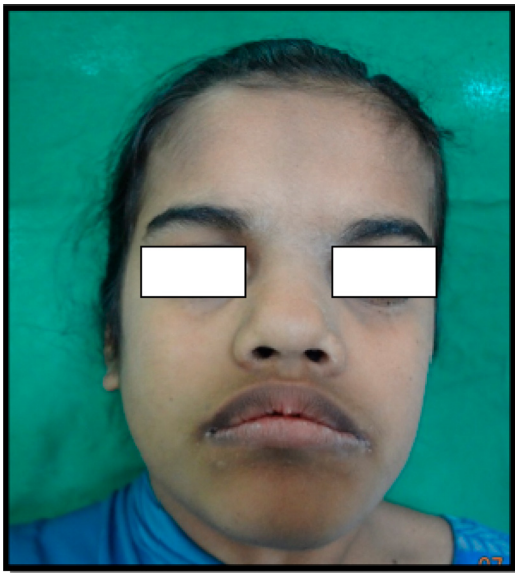

Fig. 2 – Extra oral photograph showing coarse facial features.

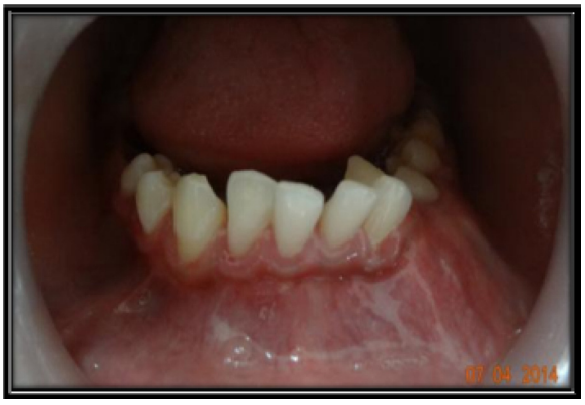

Fig. 3 – Intraoral photograph showing obliteration of vestibule from 42 to 35 regions and horizontally displaced 33.