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Case Report

Gorlin–Goltz syndrome: A clinico radiological illustrative case report

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ARTICLE INFO

Article history:
Received 11 August 2012
Accepted 16 May 2013

Keywords:
Keratocystic odontogenic tumor
Basal cell nevi
Bifid ribs
Bifid tongue
Partial ankyloglossia

ABSTRACT

Gorlin–Goltz syndrome is an infrequent multisystemic disease that is inherited in an autosomal dominant way showing a high level of penetrance & variable expressiveness. It is about a multisystemic process that is characterized by the presence of odontogenic keratocysts in the jaws, multiple basal cell nevi or carcinomas, palmar and/or plantar pits & calcification of falx cerebri. Diagnosis of Gorlin–Goltz syndrome is made by having two major criteria or one major & two minor criteria. Here, we report a case of a 11-year-old girl suffering from this syndrome presenting with two new entities namely bifid tongue & partial ankyloglossia.

1. Introduction

On April 2, 1967 Calvin Wells wrote a letter to Merton I. Satinoff at the University of Turin: 'Have you met any dentigerous cysts yet? If you do find any, be sure to look for bifid ribs in the same body: it would be a splendid find if you discovered the dual anomaly!' On 16 April Satinoff replied: 'You remember mentioning in your letter about the dentigerous cyst-bifid ribs syndrome, well I don't know if you believe in ancient Egyptian miracles or not but about 3/4 h before I received your letter, I was in fact examining such a case. You can imagine my excitement when I read what you wrote. It is rather like the curse of Tutankhamun in reverse!' (Tutankhamun was the emperor of dynastic period who ruled Egypt for 10 years & died at the age of 19 years because of congenital flaws.) The Paleorecord shows that those were the skeletons of two Egyptians of Dynastic Period. This syndrome existed during Dynastic Egyptian times in mummies dating back to 1000 B.C.[1,2]

Gorlin–Goltz syndrome is an infrequent multisystemic disease that is characterized by a predisposition to neoplasms & other developmental abnormalities.[3] It is a hereditary condition inherited in a dominant autosomal way & caused by mutations in the PTCH1(Patched1) gene which is mapped to the long arm of chromosome 9q22.3–q31.[3–6] The syndrome has received several names throughout the time such as Nevoid basal cell carcinoma syndrome, Gorlin–syndrome,

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0975-962X/$ – see front matter © 2013 Indian Journal of Dentistry. All rights reserved.
http://dx.doi.org/10.1016/j.ijd.2013.05.007

Basal cell nevus syndrome, Fifth phacomatosis, Multiple basilioma syndrome, Hereditary cutaneomandibular polyoncosis & the most complex one "Jaw-cyst-basal-cell-nevus, bifid rib syndrome."[3] Diagnosis of Gorlin–Goltz syndrome is made by having two major criteria or one major & two minor criteria. The major criteria includes odontogenic keratocysts of the jaws, more than two sites of basal cell carcinomas or one site of basal cell carcinoma in persons younger than 20 years, three or more palmar or plantar pits, calcification of falx cerebri, bifid or fused ribs, first degree relative with Gorlin–Goltz syndrome. The minor criteria includes macrocephaly, congenital malformations such as cleft lip or palate, frontal bossing, hypertelorism, skeletal abnormalities, vertebral anomalies, ovarian fibroma or medulloblastoma.[3,7,8] During the past few years very important advances have been taking place in the knowledge of the genetic characteristics of this syndrome, existent clinicopathologic variants & its different manifestations.[3] This case report proposes two new minor criteria to this syndrome, bifid tongue & partial ankyloglossia.

2. Case report

A 11-year-old female patient along with her parents had come to the Department of Oral Medicine Diagnosis and Radiology, with a chief complaint of swelling on right cheek region since 1 month [Fig. 1a] which was associated with pain. Past medical history revealed presence of extra fingers & toes attached to the little ones which were surgically excised in her early childhood. Family history was unremarkable with the absence of consanguinity. General physical examination revealed surgical scars of excised digits on little fingers & toes [Fig. 1b and c], palmar pits [Fig. 1d], sprengel deformity of the shoulders[Fig. 2a]. Extraoral examination revealed hypertelorism, wide nasal bridge [Fig. 1a] and flesh-colored brownish pin point papules in the right & left periorbital region, forearm & chest [Fig. 2b–d]. A diffuse swelling was present on the right cheek region which was firm to hard in consistency and was mildly tender on palpation [Fig. 1a]. Bilateral submandibular lymph nodes were palpable & non tender. Intraoral examination revealed a diffuse swelling extending mesiodistally from 12 to 16 teeth region and buccopalatally from buccal vestibule to mid palatine raphae [Fig. 3a]. The overlying mucosa was firm to hard in consistency and was tender on palpation. The tongue was bifid and partially ankylosed [Fig. 3b and c] along with a high arched palate [Fig. 3a].

Taking into consideration the above findings, the patient was subjected to radiographic investigations. Orthopantomograph revealed multiple radiolucencies present in the right maxillary posterior region, right mandibular ramus region surrounding the tooth bud of 48 and right parasymphyseal region involving horizontally impacted 43 [Fig. 3d]. Dorsal spine PA view revealed 3rd, 4th & 6th bifid ribs on the right side & 3rd bifid ribs on the left side [Fig. 4a]. Dorsal spine AP view revealed scoliosis and decreased intervertebral space between C5 and C6 [Fig. 4b]. Both wrist PA view revealed no change in metacarpals. Coronal and axial CT images show the presence of multiple cystic radiolucent lesions involving maxilla and mandible [Fig. 4c and d].

Based on clinical history & radiographic findings the case was provisionally diagnosed as Gorlin–Goltz syndrome. The

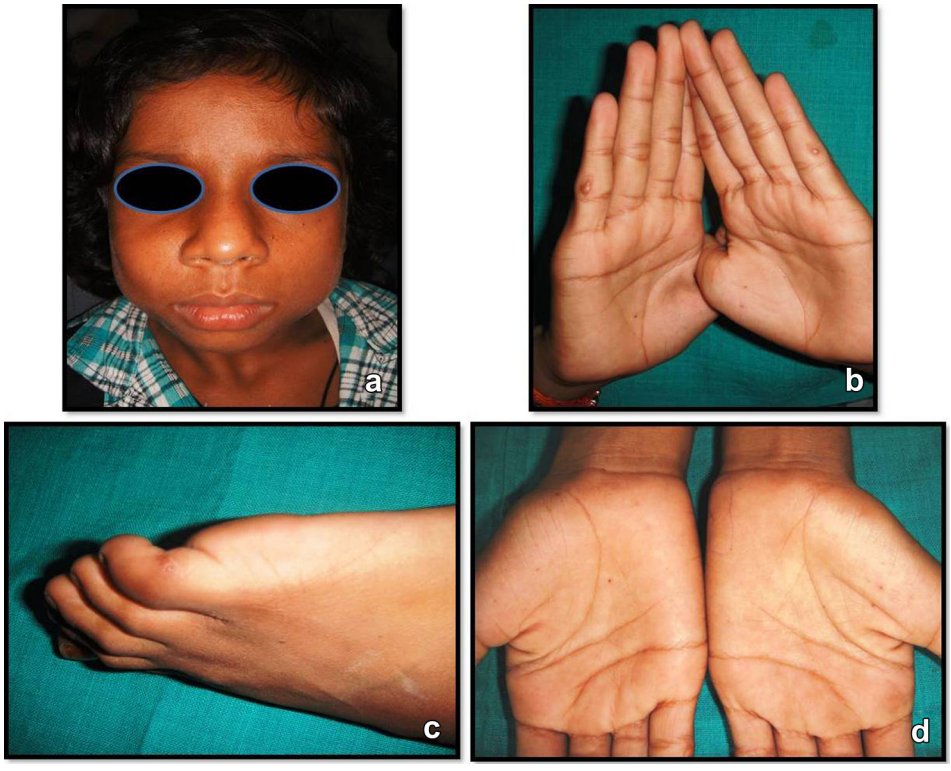

Fig. 1 – a: Profile photograph showing hypertelorism, wide nasal bridge with swelling in the right cheek region. b: Photograph of hands revealing surgical scars of excised digits on little fingers. c: Photograph of foot showing surgical scar of excised digit on little toe. d: Photograph of palms showing multiple palmar pits.

Please cite this article in press as: Mohan RPS, et al., Gorlin–Goltz syndrome: A clinico radiological illustrative case report, Indian Journal of Dentistry (2013), http://dx.doi.org/10.1016/j.ijd.2013.05.007