CASE REPORT

Gorlin Goltz syndrome - a rare case report diagnosed radiographically

Dr. Niddhi Poptani ,[1] Dr. Shachi, Parikh,[1] Dr. Mitesh Padhiyar,[1] Dr. Nilesh Raval[2]

Postgraduate student, Department of Oral Medicine & Radiology, Uvarsad, Gandhinagar, India.[1] Professor & HOD, Department of Oral Medicine & Radiology, Uvarsad, Gandhinagar, India.[2]

Abstract: Gorlin Goltz syndrome(Nevoid Basal Cell Carcinoma)is an uncommon autosomal dominant condition caused by mutation in PTCH-1(patched) gene causing a wide range of developmental anomalies like cutaneous basal cell carcinoma, multiple keratocystic odontogenic tumors, palmar plantar pits and skeletal anomalies. Keratocystic odontogenic tumors being the first manifestation, dentists play a key role in diagnosing the syndrome. Thus enabling early diagnosis and treatment which reduces the severity of the long term sequale of the syndrome. We report a case of Gorlin-Goltz syndrome in a 24 years old male patient which was incidentally radiographically diagnosed in the patient.

Keywords: Basal cell carcinoma, KCOT, Falx cerebri

Introduction

Gorlin Goltz syndrome (GGS) is an autosomal dominant disorder characterized by a classical triad composed of multiple basal cell carcinomas, keratocystic odontogenic tumours (KCOTs) in the jaws and bifid ribs predisposition to neoplasms and other developmental abnormalities.[1]

The first description of this syndrome was given by Jarisch and White in 1894. In 1960 Gorlin and Goltz together, established a classical triad of basal cell carcinoma, odontogenic keratocyst and bifid ribs, that characterizes the diagnosis of this syndrome. Later Rayner et al modified this and suggested that for establishing the diagnosis, at least cysts had to appear in combination with calcification of the falx cerebri or palmar and planter pits.[2]

The prevalence of this syndrome is estimated to be 1 in 50,000-1,50,000 in the general population, varying by region.[3] Males and females are equally affected. This syndrome probably presents itself in all ethnic groups, although a few cases have been published in certain human races.[1]

NBCCS(Nevoid Basal Cell Carcinoma Syndrome) is caused by mutations in a tumor suppressor

gene PTCH (human homologue of a Drosophila segment polarity gene Ptch) located in chromosome 9q22.3. This protein can be found in the Hedgehog signaling pathway. PTCH in the absence of its ligand, it acts as a cell-cycle regulator, normally inhibiting expression of downstream genes that control cell fate, patterning and growth.[4]

A multidisciplinary approach various specialities like dentists, oral and maxillofacial surgeons, dermatologists and neurologists is required for the diagnosis and management of this syndrome. In this syndrome, management is about adequate treatment of cysts and removal of tumours.[5]

Case Report

A 24 year old male patient who reported to the Department of Oral Medicine and radiology with the complaint of difficulty in chewing from the left side. Patient had difficulty in chewing from last 5 years as he had undergone extraction of lower left back teeth as they were carious. He had no relevant past medical history. Patient had also undergone root canal treatment of upper right central incisors and canine before 5 years.Mild hypertelorism and frontal bossing was observed.On intraoral examination, lower left second and third molar, upper both lateral incisors were missing.

Radiographic examination of Orthopantomogram showed multiple radiolucent lesions in both left and right sides of lower jaw, out of which right one was associated with unerupted tooth. On left side radiolucent lesion extending from left first molar to neck of left condyle and coronoid process. Thinning of cortex was present on left side.

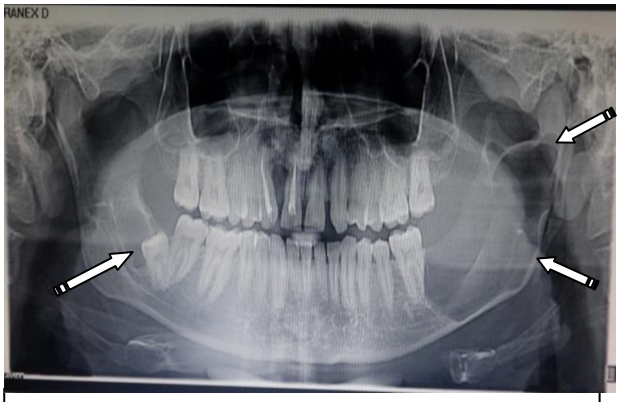

Fig. 1: OPG showed bilateral radiolucent lesions of mandible

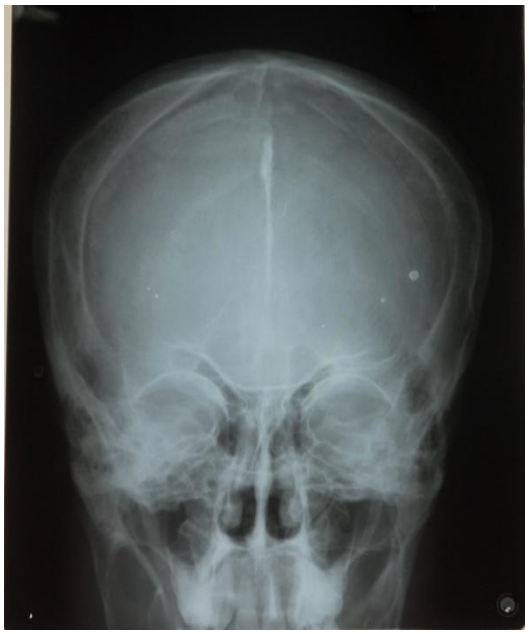

Fig. 2: Calcification of falx cerebri

Calcification of falx cerebri was observed on PA skull radiograph as a thin radio opaque line. Radiographic examination of Hand-wrist was none significant.There were no abnormalities detected on the chest X-ray.

On the basis of clinical as well as radiographic findings diagnosis of Gorlin-Goltz syndrome was established.Surgical removal of the cystic lesion followed by histopathological evaluation was advised. Clinical-radiographic diagnosis was confirmed by histopathological report.

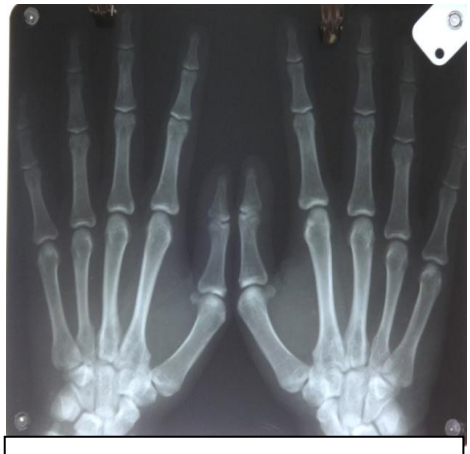

Fig. 3: Hand-Wrist radiograph

Discussion

Gorlin Goltz syndrome is also known as several names such as nevoid basal cell carcinoma syndrome, Gorlin syndrome, basal cell nevus syndrome, fifth phacomatosis, multiple basilioma syndrome, hereditary cutaneomandibular polyoncosis and the most complex one 'Jaw-cyst-basal-cell-nevus, bifid rib syndrome.'[6]Keratocysts appear in 75% of patients affected by this syndrome and are normally the first symptoms,[3] which usually appear in the early stages of life. Generally they are multiple, and are asymptomatic, although they can

spark off infections and disturbances in the dental eruption.[2] They are more recurrent during treatment and are more aggressive in behaviour than the keratocysts of non-syndrome patients. OKCs are more common in the adult life, the peak incidence being the third decade of life.[7] However, in the GGS, OKC occurs at a much younger age. In our case also we found multiple KCOT in young male patients. There was no presence of the skin lesions.

Diagnostic protocols in NBCCS [8]

Family history, Past medical and dental history are very important.

Clinical examinations of the following regions: Oral, skin, CNS, head circumference, inter-pupillary distance, eyes, genitourinary system, cardiovascular system, respiratory system, skeletal system, genetic testing.

X-ray: Chest A. P. and lateral skull, OPG, cervical and thoracic spine-A. P. and lateral, hands (for pseudo cysts), pelvic (female), ovarian ultrasound (female) for ovarian fibroma, Echocardiogram (children) for cardiac fibroma.

Diagnosis of Gorlin-Goltz syndrome can be made in the presence of: a) 2 major criteria, b) 1 major criteria and molecular confirmation or c) 1 major and 2 minor criteria. [9,10]

Major criteria

1. Excessive numbers of basal cell carcinomas out of proportion with prior sun exposure and skin type or < 20 yrs of age
2. Odontogenic keratocysts of the jaws prior to 20 yrs of age
3. Palmar or plantar pitting
4. Lamellar calcification of the falx cerebri
5. Medulloblastoma, typically desmoplastic
6. 1st degree relative with Gorlin-Goltz syndrome

Minor criteria

1. Rib anomalies
2. Other specific skeletal malformations and radiologic changes (i.e. vertebral anomalies, kyphoscoliosis, short 4th metacarpals, postaxial polydactyly)
3. Macrocephaly
4. Cleft lip and/or palate
5. Ovarian/cardiac fibroma
6. Lymphomesenteric cysts
7. Ocular abnormalities (i.e. strabismus, hypertelorism, congenital cataracts, glaucoma, coloboma)

References

1. Casaroto et al.: Early diagnosis of Gorlin-Goltz syndrome: case report. Head & Face Medicine 2011 7:2
2. Kiran NK, Tilak Raj TN, Mukunda KS, Reddy VR. Nevoid basal cell carcinoma syndrome (Gorlin-Goltz syndrome). Contemp Clin Dent 2012;3:514-8.
3. Pol CA, Ghige SK, Kalaskar RR, Gosavi SR. Gorlin-Goltz syndrome: A rare case report. Contemp Clin Dent 2013;4:547-50.
4. Mohan RPS, Verma S, Agarwal N, et al. BMJ Case Rep doi:10.1136/ bcr-2013-010409
5. Guruprasad Y, Prabhu PR. Gorlin-Goltz syndrome with situs oppositus. Natl J Maxillofac Surg [serial online] 2010 [cited 2016 Jan 5];1:58-62
6. Mohan RPS, Verma S, Agarwal N, Singh U. Gorlin-Goltz syndrome: rare case report. BMJ Case Reports. 2013
7. Mehta D, Raval N, Patadiya H, Tarsariya V. Gorlin-Goltz Syndrome. Annals of Medical and Health Sciences Research. 2014;4(2):279-282.
8. Chandran S, Marudhamuthu K, Riaz R, Balasubramaniam S. Odontogenic keratocysts in Gorlin–Goltz syndrome: A case report. J Int Oral Health 2015;7(Suppl 1):76-79.
9. Kimonis VE, Goldstein AM, Pastakia B et al. Clinical manifestations in 105 persons with nevoid basal cell carcinoma syndrome. Am J Med Genet 1997;69:299-308.
10. Evans DG, Ladusans EJ, Rimmer S et al. Complications of the naevoid basal cell carcinoma syndrome: results of a population based study. J Med Genet 1993;30:460-4.

Corresponding author: Dr. Niddhi Poptani, Postgraduate student, Department of Oral Medicine & Radiology, Uvarsad, Gandhinagar, India.
Email: nidhipoptani88@gmail.com
Confilct of interest:No,Financial Support: No