ISSN: 2581-5989
PubMed - National Library of Medicine - ID: 101738774

International Journal of Dental Science and Innovative Research (IJDSIR)
IJDSIR : Dental Publication Service
Available Online at:www.ijdsir.com
Volume – 9, Issue – 4, August – 2026, Page No. : 01 – 06

A clinical Spectrum of Goltz – Goltz Syndrome – Detailed Presentation of Two Cases

1Dr. Jha Sachin, Resident, M.D.S., Department of Oral & Maxillofacial Surgery, Government College of Dentistry, Indore.

[2]Dr. Shakya Neelam, Associate Professor, M.D.S., Department of Oral & Maxillofacial Surgery, Government College of Dentistry, Indore.

[3]Dr. Baheti Nishita, Resident, Department of Oral & Maxillofacial Surgery, Government College of Dentistry, Indore, Madhya Pradesh, India.

[4]Dr. Chouhan Meenal, Resident, Department of Oral & Maxillofacial Surgery, Government College of Dentistry, Indore, Madhya Pradesh, India

Corresponding Author: Dr. Jha Sachin, Resident, M.D.S., Department of Oral & Maxillofacial Surgery, Government College of Dentistry, Indore.

Citation of this Article: Dr. Jha Sachin, Dr. Shakya Neelam, Dr. Baheti Nishita, Dr. Chouhan Meenal, "A clinical Spectrum of Goltz – Goltz Syndrome – Detailed Presentation of Two Cases", IJDSIR – August– 2026, Volume –9, Issue – 4, P. No. 01 – 06.

Type of Publication: Case Series

Conflicts of Interest: Nil

Abstract

Gorlin-Goltz syndrome (GGS), also known as nevoid basal cell carcinoma syndrome, is a rare, auto somal dominant condition characterized by multiple odontogenic keratocysts (OKCs), pal mar and plantar pits, calcification of the falx cerebri, and skeletal abnormalities like bifid ribs. Early diagnosis is crucial to minimize complications and recurrence. This article presents two detailed cases of GGS managed at our institution, emphasizing the clinical and radiological findings, surgical interventions, and long-term follow-up. Both patients demonstrated classic features of the syndrome and responded well to conservative surgical approaches, highlighting the importance of multi-disciplinary evaluation and personalized treatment planning.

Keywords: Basal Cell Nevus syndrome, odontogenic cysts, cone-beam computed tomography,marsupialisation

Introduction

Gorlin-Goltz syndrome (GGS) is a multisystem disorder with high phenotypic variability and incomplete penetrance. It was first described by Gorlin and Goltz in 1960,[1] and involves developmental anomalies and a predisposition to neo plasms. Diagnostic criteria were initially defined by Evans et al. (Table 1)[2] and later modified by Kim on is et al. (Table 2)[3] to include both major and minor criteria. Diagnosis requires at least two major and one minor or one major and three minor

criteria. Common manifestations include multiple OKCs (up to 90% of cases), palmar/plantar pits, bifid ribs, and intracranial calcifications. Although basal cell carcinoma is considered a hallmark, it is less commonly reported in the Indian population. Here, we present two patients diagnosed with GGS, highlighting their clinical presentation, imaging findings, histopathology, management, and follow-up.

Table 1: Diagnostic criteria by Evans et al, 1991

Major Criteria	Minor Criteria
More than 2 BCCs, one BCC in patients younger than 30 years of age or more than 10 basal cell nevi	Congenital skeletal anomaly (eg. Bifid, splayed, fused or missing ribs, or bifid wedged or fused vertebrae)
any OKC (proven by histology) or polyostotic bone cyst	Occipital-frontal circumference greater than the ninety-seventh percentile, with fontal bossing
Three or more palmer or plantar pits	Cardiac or ovarian fibromas
Ectopic calcification in patients younger than 20 years of age (lamellar or early falx cerebri calcification)	Medulloblastoma
a positive family history of NBCC	Lymphomesenteric cysts
	Congenital malformations such as cleft lip/palate, polydactylism or eye anomaly (e.g. Cataract, coloboma or microphthalmos)

Table 2: Diagnostic criteria by Kimonis et al, 1997

Major Criteria	Minor Criteria
More than 2 BCCs or one BCC in patients younger then 20 years of age	Macrocephaly
OKCs of the jaw (proven by histologic analysis)	Congenital malformations (eg cleft lip, palate, frontal bossing, coarse faces, hyper telorism)
Three or more palmer or plantar pits	Other skeletal abnormalities (e.g. Sprengel deformity, marked pectus deformity and marked syndactyly of digits)
Bilamellar calcification of the falx cerebri	Radiological abnormalities (e.g. bridging of Sella turcica, vertebral anomalies, modelling defects of the hands and feet, or flame- shaped lucencies of the hands and feet)
Bifid, fused or markedly splayed ribs	Ovarian fibroma or medulloblastoma
A first degree relative with NBCCS	

Case Reports

Case 1

A 17-year-old male presented with pus discharge from the upper front-tooth region for 1.5 months, without pain, swelling, or fever, and with no relevant medical or family history. Extra orally, the face was symmetrical with a flattened nasal bridge, high-arched eyebrows, and hyper telorism (inter canthal distance 42 mm). Intra orally, no swelling, cortical expansion, or paresthesia was noted; sub mandibular nodes were non-tender. Grade I mobility was present in 31, 32, 41, 42; teeth 23, 28, 38, 43 were missing, with mandibular anterior displacement. OPG and CT revealed multiple well-defined unilocular radio lucencies with corticated margins and impacted 38 and 43, raising suspicion of GGS. No basal cell nevi were present; plantar keratosis, spinal kyphosis, right-sided bifid ribs, and falx cerebri calcification were noted. Under GA, three cysts were separately enucleated with peripheral ostectomy and 5 - fluorouracil cauterization. Histopathology confirmed odontogenic keratocysts. The patient remains recurrence-free over 8-month follow-up (Fig. 1).

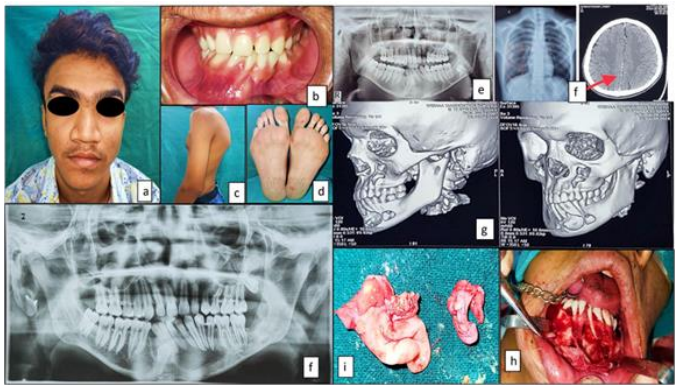

Fig 1: (a) Clinical Extra oral Presentation, (b) intraoral presentation, (c) pre-operative presentation, (d) CT face, (e) Plantar keratosis, (f) kyphosis of the spine, (g) Chest X-ray, (h) CT Brain,(g) Intra-Operative (h) specimen (i) post - orthopantomo graphy (after 8 month).

Case 2

A 14-year-old female with the complain of pain and swelling on her cheeks and gums for 4 months which was associated with pus discharge since last 5-7 days. There was no significant medical or family history. On extra oral examination, mild diffuse swelling was present extending from body to angle region of mandible antero - posteriorly; intra orally swelling extended from 45 to 47 region. Buccal cortical expansion was appreciable. Right sub mandibular lymph node was tender on palpation. An orthopantomo graphy revealed multiple, well defined, unilocular radio lucencies with well corticated margins and impacted 33, 42 and 43. CT scan of face revealed multiple, variable sized lesions involving both maxilla and mandible. CT scan of brain revealed calcification of falx and tentorium cerebelli, while chest x ray revealed presence of bifid ribs on left side. USG of pelvis revealed bilateral polycystic ovarian morphology. There were no basal cell lesions or skin lesions on face, neck or chest regions, neither was any palmer or plantar keratosis. All the above findings lead towards a diagnosis of GGS.

Under all aseptic conditions and antibiotic prophylaxis, hypo tensive general anesthesia. Full length intraoral vestibular incision was made second molar to second molar in both maxillary and mandibular arches, muco periosteal flaps were raised and all four cysts were addressed in similar manner Enucleation was performed followed by peripheral ostectomy with an acrylic trimming bur under copious irrigation with normal saline. This was followed by chemical cauterization of 5-flurouracil. The enucleated tissue was sent for Histopathological examination. All three lesions were diagnosed as Odontogenic Keratocysts. Clinical features were consistent with prior Indian case series.[7]

The patient is on regular follow up in the last 6 months now and shows no signs of recurrence. (Fig. 2)

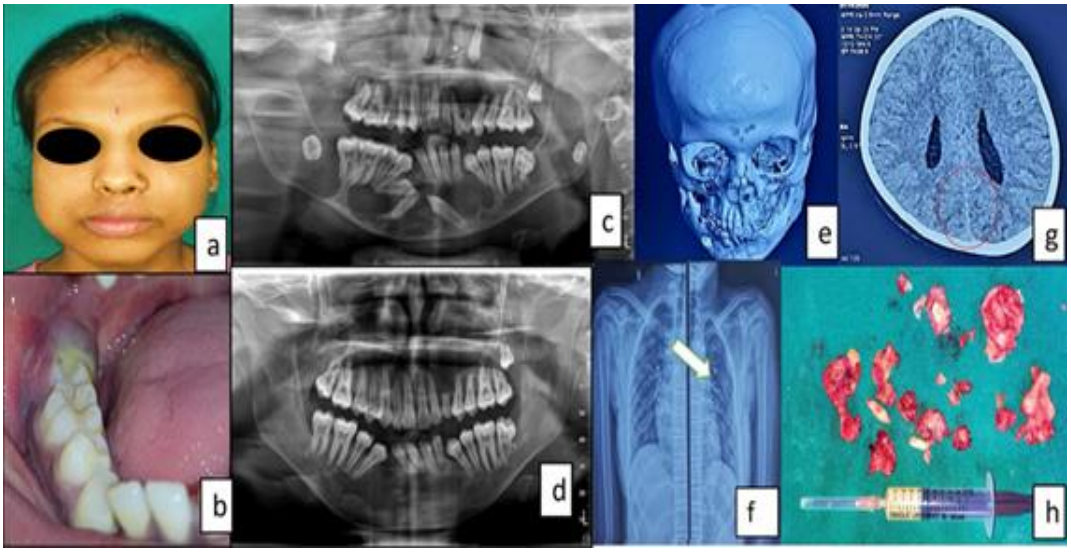

Fig 2: (a) Clinical Extra oral Presentation, (b) Intraoral Presentation, (c) Pre- Orthopantomography, (d) Post-Orthopantomography (after 6 month), (e) CT Face (f) Chest X-ray, (g) CT Brain Case 2 (h) biopsy specimen.

Table 3: Summary Table

Parameter	Case 1	Case 2
Age/Sex	17/M	14/F
Presenting Complaint	Pus discharge	Maxillary and mandibular swelling with pain and pus discharge
Number of Cysts	3 (2 Mandible + 1Maxilla)	4 (2Mandible + 2 Maxilla)
Palmar Pits	Absent	Absent
Plantar Pits/ keratosis	Present	Not observed
Frontal Bossing and hyper telorism	Present	Present
Macro cephaly	Absent	Absent
Falx Cerebri Calcified	Present	Present
Rib Anomaly	Bifid ribs	bifid ribs
Histopathology	Odontogenic Keratocyte	Odontogenic Keratocyte
Treatment	Enucleation with peripheral ostectomy and usage of 5-fluorouracil	enucleation with peripheral ostectomy and usage of 5-fluorouracil
Follow-Up Duration	8 months	6 months
Recurrence	No recurrence	No recurrence

Discussion

Gorlin–Goltz syndrome (GGS), or Nevoid basal cell carcinoma syndrome (NBCCS), is a rare auto somal dominant disorder caused mainly by PTCH1 gene mutations on chromosome 9q22.3, with about 70% familial and 20–30% de novo cases. Patients exhibit variable expressivity and age - related penetrance. In many cases, OKCs may be the initial manifestation, as

seen in our patient, making oral findings pivotal in early diagnosis.

One of the most distinctive and often earliest features of the syndrome is the presence of odontogenic keratocysts, typically appearing during the first or second decade of life. These cysts predominantly affect the posterior mandible, are often bilateral or multiple, and carry a high recurrence rate following surgical removal. Basal cell carcinomas (BCCs) represent another hallmark of the syndrome, often developing during adolescence or early adulthood. Unlike sporadic BCCs, which are usually sun-induced and arise later in life, those associated with NBCCS may occur on sun-protected areas and tend to be numerous. Additional radiological findings, such as bilamellar calcification of the falx cerebri, bridging of the sella turcica, and rib anomalies (e.g., bifid or fused ribs), provide further diagnostic clues and support the importance of comprehensive imaging.

Management of GGS requires a multidisciplinary team, including oral and maxillofacial surgeons, dermato logists, radiologists, geneticists, and paediatricians. Management of odontogenic keratocysts (OKCs) in Gorlin-Goltz Syndrome poses a significant challenge due to their tendency to be multiple, recurrent, and aggressive. Long-term follow-up is essential, particularly to monitor for recurrence of OKCs and development of other associated neo plasms. Surgical enucleation with peripheral ostectomy is the most commonly employed treatment, aiming to completely remove the cystic lining and reduce recurrence. Recurrence rates of OKC in GGS are reported to be higher than non-syndromic cases.[8,9] Adjunctive use of chemical cauterization like Carnoy's solution has been widely used to minimize recurrence[10] also 5-flurouracil is often recommended.

Marsupialization or decompression may be considered in large cysts, especially in younger patients, to reduce lesion size and preserve vital structures before definitive enucleation. However, due to the high recurrence rate—up to 60% in syndromic cases—long-term radiographic surveillance using panoramic imaging or CBCT at regular intervals (every 6–12 months) is essential. Preventive dental evaluation and genetic counselling are also important to identify new lesions early and guide family members at risk.

Conclusion

Gorlin–Goltz syndrome is a multisystem disorder, with odontogenic keratocysts being early, distinctive signs. Dentists often first detect these lesions through routine examinations, and their vigilance in identifying multiple or recurrent cysts and craniofacial features enables early diagnosis, referral, complication prevention, and improved outcomes through multidisciplinary care.[5,6,9,10]

References

1. Gorlin RJ, Goltz RW. Multiple nevoid basal-cell epithelioma, jaw cysts and bifid rib: a syndrome. N Engl J Med. 1960;262:908-12.

2. EvansDG, Ladusans EJ, Rimmer S, Burnell LD, Thakker N, Farndon PA. Complications of the nevoid basal cell carcinoma syndrome: results of a population-based study. J Med Genet. 1993;30(6):460-4.

3. Kimonis VE, Goldstein AM, Pastakia B, Yang ML, Kase R, DiGiovanna JJ, et al. Clinical manifestations in 105 persons with nevoid basal cell carcinoma syndrome. Am J Med Genet. 1997;69(3):299-308.

4. Al-Jarboua MN, Al-Husayni AH, Al-Mgran M, Al-Omar AF, Al-Omar A. Gorlin-Goltz syndrome: A case report and literature review. Cureus. 2019 Jan 8;11(1).

5. Spadari F, Pulicari F, Pellegrini M, Scribante A, Garagiola U. Multidisciplinary approach to Gorlin-Goltz syndrome: from diagnosis to surgical treatment of jaw bones. Maxillofacial Plastic and Reconstructive Surgery. 2022 Jul 18; 44(1):25.

6. Joshi PS, Deshmukh V, Golgire S. Gorlin-Goltz syndrome. Dent Res J (Isfahan). 2012;9(1):100-6.

7. Lata J, Verma N, Kaur A. Gorlin-Goltz syndrome: a case series of 5 patients in North Indian population with comparative analysis of literature. Contemp Clin Dent. 2015;6(Suppl 1):S192-S201.

8. Sivanmalai S, Kandhasamy K, Prabu N, Prince CN, Prabu CS. Carnoy's solution in the management of odontogenic keratocyst. J Pharm Bioallied Sci. 2012;4(Suppl 2):S183-5.

9. Agrawal A, Murari A, Vutukuri S, Singh A. Gorlin-Goltz syndrome: case report of a rare hereditary disorder. Case Rep Dent. 2012; 2012:475439.

10. Narang A, Maheshwari C, Aggarwal V, Bansal P, Singh P. Gorlin-Goltz syndrome with intracranial meningioma: case report and review of literature. World Neurosurg. 2020; 133:324-30.