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CASE REPORT

A RARE CASE OF CYSTIC LESIONS OF JAW (ODONTOGENIC KERATOCYSTIC LESIONS) - GORLIN GOLTZ SYNDROME

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ABSTRACT

Gorlin-Goltz syndrome is a rare autosomal dominant inherited disorder identified by the presence of multiple odontogenic keratocysts, alongside a range of cutaneous, dental, osseous, ophthalmic, genital and neurological abnormalities. Prompt diagnosis is crucial as it can advance to aggressive basal cell carcinomas and neoplasias. Instances of Gorlin-Goltz syndrome in India are rare. Timely diagnosis and intervention play a critical role in mitigating the severity of long-term complications associated with this syndrome.(1)

INTRODUCTION

Gorlin-Goltz syndrome is a rare genetic disorder inherited in an autosomal dominant manner, resulting in multi-organ dysfunction. Its prevalence ranges from approximately 1 in 50,000 to 1 in 150,000, though this can vary based on regional and ethnic factors. (2).First described in 1894 by Jarish and White, it was later named Gorlin-Goltz syndrome after the comprehensive observations made by Gorlin and Goltz. Clinical manifestations of this syndrome include odontogenic keratocystic lesions in the jaw, which typically manifest within the first 30 years of life, basal cell carcinomas (BCC) appearing from early childhood, palmar and plantar pits, calcification of the falx cerebri, frontal bossing, macrocephaly, broad nasal bridge, mild mandibular prognathism, vertebral anomalies, cleft palate, cleft lip, highly arched palate, eye abnormalities, and tumors such as medulloblastoma and fibromas in the ovaries and heart. This condition, exhibiting high penetrance and, is attributed to mutations in the "protein patched homolog" (PTCH) gene.(3)

CASE REPORT

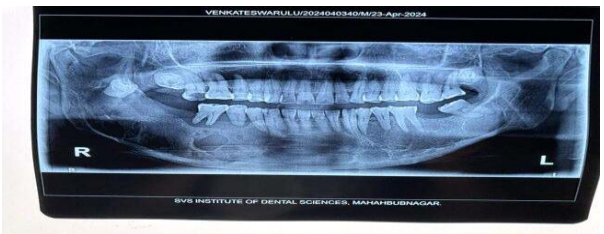
A 32 year old male presented with pain and swellings in the jaw since childhood with occasional difficulty in jaw movements while chewing.

On clinical examination, patient had restricted jaw movements with swellings in the jaw.The patient was marfanoid built who was slender and had elongated arms.The patient had multiple epidermal cysts on the face.

Imaging features

OPG

Multiple smooth, unilocular, radiolucent variable sized lesions with well corticated borders noted in the mandible and maxilla.



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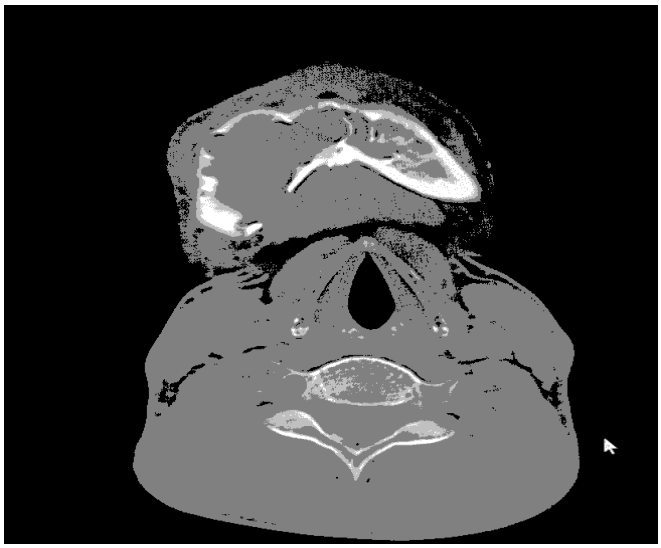
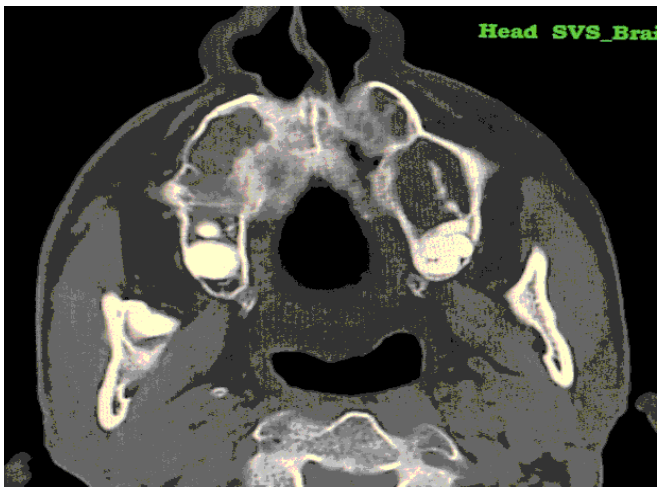
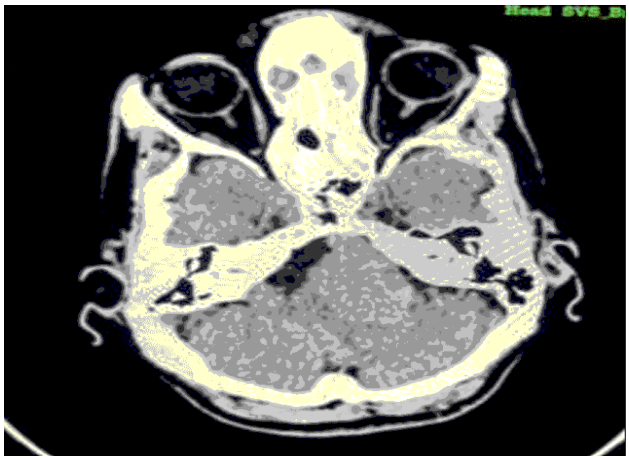
CT Skull with Brain

Multiple,expansile cystic lesions with well corticated borders noted in the marrow of mandible and maxilla with endosteal scalloping and multifocal cortical erosions.- Odontogenic keratocysts.





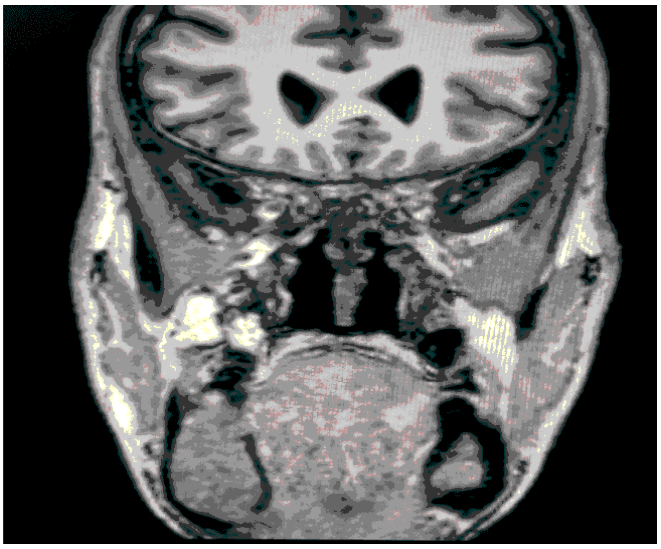
CT brain shows a single well defined dural based heterogeneously hyperdense lesion in left cerebello-pontine angle causing mass effect on pontomedullary junction.- suggestive of meningioma.

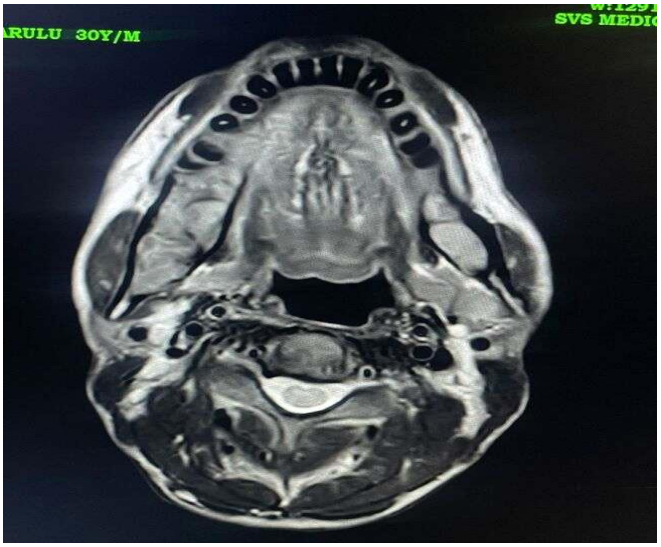


On MRI

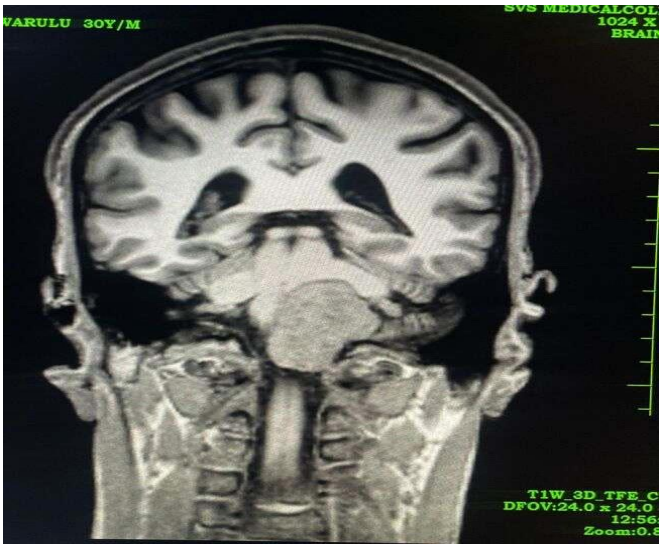
Multiple, T1isointense and T2 slightly hyperintense lobulated and expansile lesions noted in maxilla and mandible.

These lesions do not show restriction diffusion on DWI and no bloomings on GRE sequence.

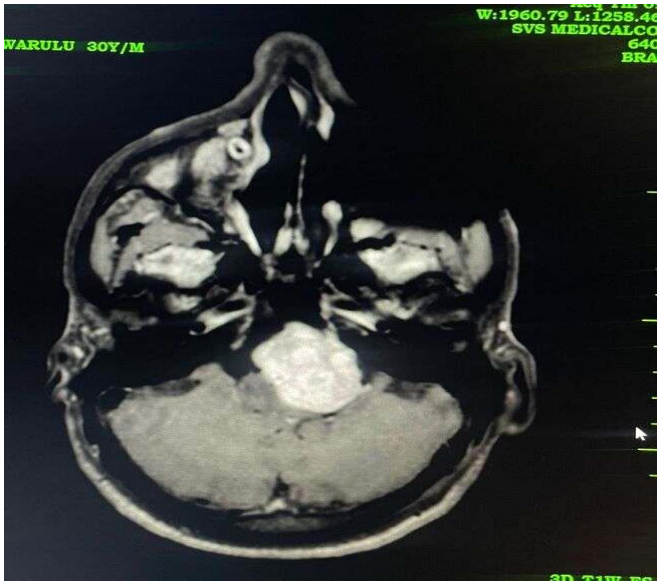




A well defined T1isointense, T2 slightly hyperintense lobulated lesion noted in the left cerebello-pontine cistern not showing restricted diffusion on DWI.

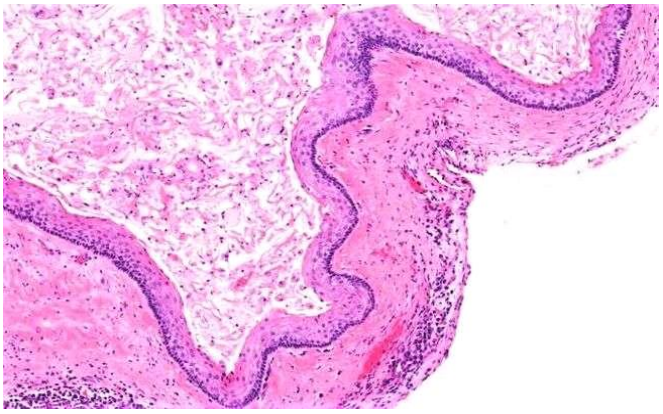


The lesion is showing avid enhancement on postcontrast studies.



Biopsy of the cystic lesions in mandible was done and sent for Histopathological examination which showed:

Cyst wall lined by bites of uniform epithelial lining consisting of 6-8 layered cells lacking rete ridges with focal clefting between epithelium and the underlying fibroconnective tissue with dense inflammatory infiltrates. Epithelium charectarised by palisaded hyperchromatic basal cell layer comprised of cuboidal - columnar cells. Lumen showing abundant areas of keratinaceous debris. At places show few osteoid areas.



Taking in consideration multiple OKC in the jaw, meningioma, epidermal cysts and histopathological findings, the diagnosis of Gorlin-goltz was made.

DISCUSSION

Gorlin-Goltz syndrome is a relatively a rare autosomal dominant inherited disorder, with prevalence rates ranging from 1 in 57,000 to 1 in 256,000 and a balanced male-to-female ratio. It displays high penetrance and variable expressivity. The syndrome is caused by mutations in the PTCH1 gene located at chromosome 9q22.3-q31. Interestingly, up to 60% of cases occur without any family history of the condition. PTCH1 serves as a tumor suppressor gene, crucial in regulating the cell cycle and embryonic development. Mutations in this gene often result in a truncation of its C-terminal, leading to the loss of its suppressor function.(4)

The primary clinical feature linked with Gorlin-Goltz syndrome (GGS) is basal cell carcinoma (BCC), which can appear at even 2 years of age. However, more commonly, the patients present with multiple odontogenic keratocysts (OKCs) during the 2nd to 3rd decade of life. If any of the major criteria are detected, even incidentally, it should prompt a comprehensive search for other criteria. Radiologists play a pivotal role in GGS diagnosis. Patients with GGS benefit from regular follow-up to facilitate early detection of skin malignancies, up to 80% of the white population and 38% of blacks develop BCCs.(4)

Common imaging features of this condition include radiolucent jaw lesions (odontogenic keratocysts) in approximately 90% of cases, along with calcifications observed in the falx cerebri (70–85%), tentorium cerebelli (20–40%), and diaphragma sellae (60–80%). Other prevalent findings include macrocephaly (70%) and rib anomalies such as bifid, fused, or splayed ribs (30–60%). Less frequent imaging findings encompass cleft lip and cleft palate (5%) and even vertebral anomalies, such as malformations in the craniovertebral junction and fusion of cervical and upper thoracic vertebrae (10–40%). Additionally, several other neoplastic conditions may occur, albeit at a lower frequency, including medulloblastoma (1–2%), meningioma (3–5%), and ovarian fibroma (25–50% of all female patients).(5)

While multiple odontogenic keratocysts (OKCs) are nearly ubiquitous in Gorlin-Goltz syndrome (GGS), other conditions also exhibit this feature. These conditions include Ehlers-Danlos syndrome, Noonan syndrome, and oral-facial-digital syndrome. Additionally, there are rare instances of nonsyndromic multiple OKCs being reported.

CONCLUSION

This case underscores the significance of maintaining awareness and a high level of suspicion for this uncommon syndrome in young individuals, particularly those lacking skin lesions. Identifying a major criterion, even if discovered incidentally, should trigger an investigation for additional associations. This proactive approach can facilitate the early detection of malignancies, consequently diminishing both mortality and morbidity rates.(6).

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