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Multiple odontogenic keratocysts in siblings with Gorlin-Goltz Syndrome: A two case series

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Abstract

Gorlin-Goltz syndrome (GGS) is a rare hereditary disorder characterized by multisystem developmental abnormalities involving the skin, skeleton, and jaws, most associated with pathogenic variants in the *PTCH1* gene. We report a familial occurrence of multiple odontogenic keratocysts (OKCs) in two paediatric siblings from South America. A 9-year-old boy and his 7-year-old sister presented with multiple cystic jaw lesions detected incidentally. Imaging revealed multiple hypodense lesions involving the jaws, with calcification of the falx cerebri in Case 1 and bifid ribs in Case 2. Histopathological analysis confirmed the diagnosis of OKC. Management consisted of a staged conservative surgical approach, including decompression followed by enucleation and curettage. Satisfactory healing was observed, with no recurrence of treated lesions during a minimum follow-up period of 24 months. This report highlights the importance of early diagnosis, multidisciplinary evaluation, and conservative management to preserve function and craniofacial development in paediatric patients.

Keywords: Gorlin-Goltz syndrome, nevoid basal cell carcinoma syndrome, odontogenic keratocysts, paediatric patient, case report

Introduction

Gorlin-Goltz syndrome (GGS), also known as nevoid basal cell carcinoma syndrome, is a rare autosomal dominant disorder, with an estimated incidence ranging from 1 in 50,000 to 1 in 150,000 individuals, characterized by multisystem developmental abnormalities, most associated with pathogenic variants in the *PTCH1* gene [1]. The syndrome is defined by a combination of major and minor diagnostic criteria, including multiple odontogenic keratocysts (OKCs), basal cell carcinomas, calcification of the falx cerebri, and skeletal anomalies such as bifid ribs [1, 2].

OKC is a potentially aggressive odontogenic lesion believed to arise from intraosseous dental lamina remnants. Its locally aggressive behaviour has led to debate regarding its biological nature and classification as a cyst or tumour [3]. The treatment options for OKC are marsupialization, decompression, enucleation and resection [4]. Treatment planning should consider the association with GGS, as syndromic OKCs have been reported to show high recurrence rates [5].

The present report describes an early-onset familial presentation of GGS in two paediatric siblings, both presenting with multiple OKCs and characteristic syndromic features. This case highlights the diagnostic value of clinic radiologic-histopathologic correlation in low-resource settings and supports the effectiveness of conservative management strategies in paediatric patients with syndromic OKCs

Case Report

Case 1

A 9-year-old boy from Brazil presented to the Oral and Maxillofacial Surgery Department after referral from a dental service due to delayed eruption of the permanent maxillary lateral incisor and canine (teeth 12 and 13). As part of the diagnostic investigation, panoramic radiographic examination was performed, revealing multiple cystic lesions involving the jaws.

The patient was asymptomatic, and a painless facial swelling had been noticed for approximately one month. There was no history of trauma, infection, or previous surgical interventions. Based on these findings, a facial CT scan (Fig. 1) was subsequently performed, demonstrating multiple well-defined hypodense lesions with a cystic appearance, associated with bone expansion, cortical thinning, and internal septations. Given the multiplicity and imaging characteristics of the lesions, a syndromic condition was suspected, and the patient was referred for comprehensive clinical, radiologic, and histopathologic evaluation.

Case 2

The patient's 7-year-old sister presented to the same department after incidental identification of mandibular alterations during routine dental imaging. She was asymptomatic, and no pain or signs of infection were reported. Based on the initial radiographic findings, a facial CT scan was requested for further characterization, which demonstrated multiple well-defined hypodense lesions involving the mandible (Fig. 2), raising suspicion for OKCs.

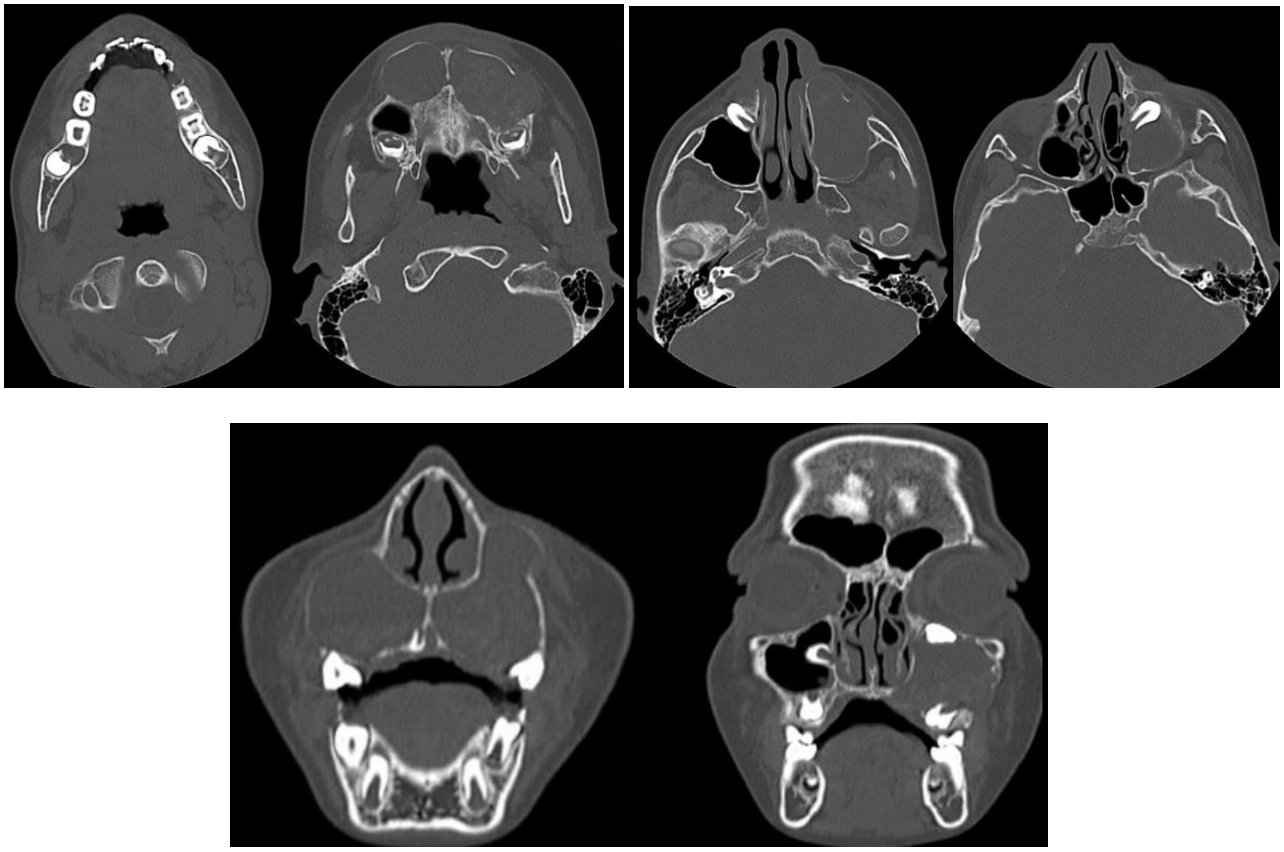

Fig 1: Facial CT images demonstrating multiple well-circumscribed hypodense lesions with a cystic appearance and smooth corticated margins, predominantly involving the maxilla and right side of the mandible

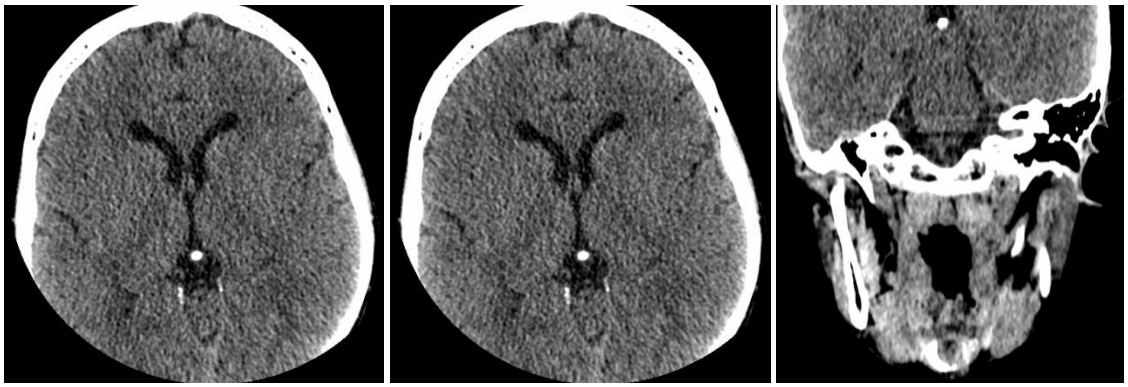

Fig 2: CT scan demonstrating multiple well-defined hypodense lesions involving the bilateral mandibular angles and symphyseal region, with features suggestive of OKCs

Both patients were otherwise healthy, with no significant systemic conditions or previous surgical history. Family history revealed that a paternal aunt reportedly had a history of multiple jaw cysts, although no formal clinical or genetic evaluation had been performed. This familial pattern supported a possible autosomal dominant mode of inheritance.

The presence of multiple cystic jaw lesions in both siblings, together with the positive family history, supported the clinical suspicion of hereditary GGS within this family.

Clinical Findings

Case 1

Extraoral examination

Revealed localized swelling involving the bilateral maxillary region, with effacement of the nasolabial sulcus. The swelling was firm on palpation, with overlying skin of normal colour

Intraoral examination

Showed expansion of the cortical plates with normal overlying mucosa of typical colour and texture. Delayed eruption of teeth 12 and 13 was observed. The occlusal plane exhibited an anterior open bite.

General and syndromic features

The patient presented typical craniofacial features of GGS, including frontal bossing, hypertelorism, mild macrocephaly and, falx cerebri calcification. No cutaneous lesions were clinically evident.

Case 2

Extraoral examination

Revealed mild bilateral mandibular expansion, more prominent at the left angle. The swelling was firm on palpation, with no signs of inflammation.

Intraoral examination

Revealed subtle expansion of the buccal and lingual cortical plates, covered by normal-coloured mucosa. No draining sinus tracts or ulcerations were noted.

General and syndromic features

The patient exhibited hypertelorism and bifid ribs, consistent with the phenotype of GGS. No other systemic or cutaneous abnormalities were observed on clinical examination.

The clinical course and management of both cases are summarized in Table 1, according to CARE guidelines [6].

Table 1: Timeline of clinical course and management

Time period	Clinical course
June 2023	Incidental radiographic identification of multiple cystic jaw lesions in Case 1, prompting suspicion of a syndromic condition
June-August 2023	Comprehensive clinical, radiologic, and histopathologic evaluation confirmed the diagnosis of OKC and supported a clinical diagnosis of GGS
August-December 2023	Staged conservative surgical management, including decompression followed by enucleation and curettage, was performed in both patients
July - 2024	Periodic clinical and radiographic follow-up demonstrated satisfactory healing of treated sites, with no recurrence of previously managed lesions
February 2025	Follow-up imaging revealed the development of new cystic lesions in Case 1, while Case 2 remained asymptomatic with no evidence of recurrence or new lesions
March 2026	Ongoing clinical and radiographic follow-up showed no evidence of recurrence or new cystic lesions in either patient

Diagnostic Assessment

Computed tomography (CT) imaging demonstrated multiple hypodense cystic lesions involving the maxilla in Case 1 and the mandible in Case 2. Calcification of the falx cerebri was observed in Case 1 (Fig. 3), while bifid ribs were identified in Case 2 (Fig. 4). The presence of multiple jaw lesions associated with characteristic craniofacial and skeletal findings was consistent with a syndromic presentation.

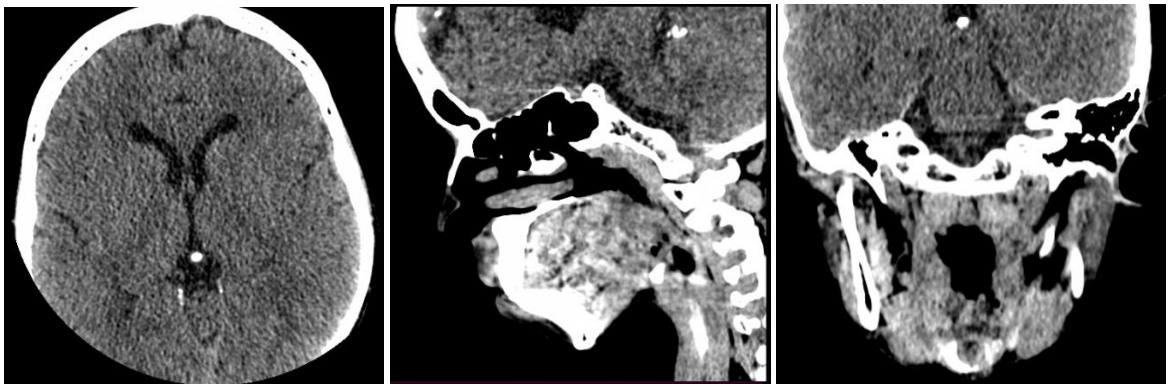

Fig 3: Non-contrast CT on brain window, demonstrating linear hyperdensity along the midline, consistent with calcification of the falx cerebri

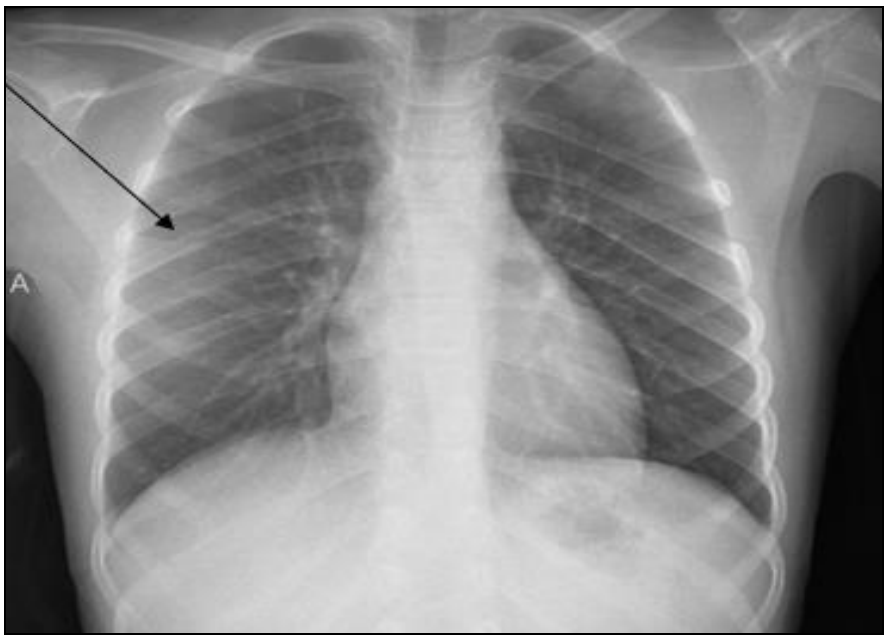

Fig 4: Frontal chest X-ray demonstrating a bifid rib, at the level of the right third rib

Histopathological examination showed parakeratinized stratified squamous epithelium exhibiting epithelial ridges at the epithelial-connective tissue interface, with a moderate inflammatory infiltrate in the underlying connective tissue on Case 1 (Fig. 5). In Case 2, histopathological examination showed a cystic lining of parakeratinized stratified squamous epithelium with a corrugated luminal surface and a palisaded hyperchromatic basal layer (Fig. 6). The epithelial-connective tissue interface was relatively flat, and no significant inflammatory infiltrate was evident in the underlying connective tissue, supporting the diagnosis of OKC.

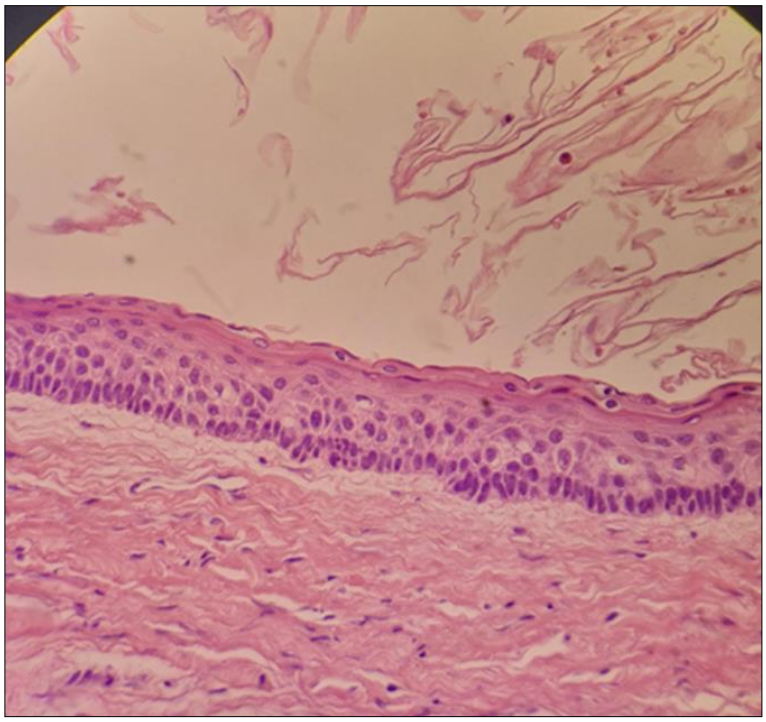

Fig 5: Parakeratinized stratified squamous epithelium with epithelial ridges and moderate inflammatory infiltrate. (H&E, ×40)

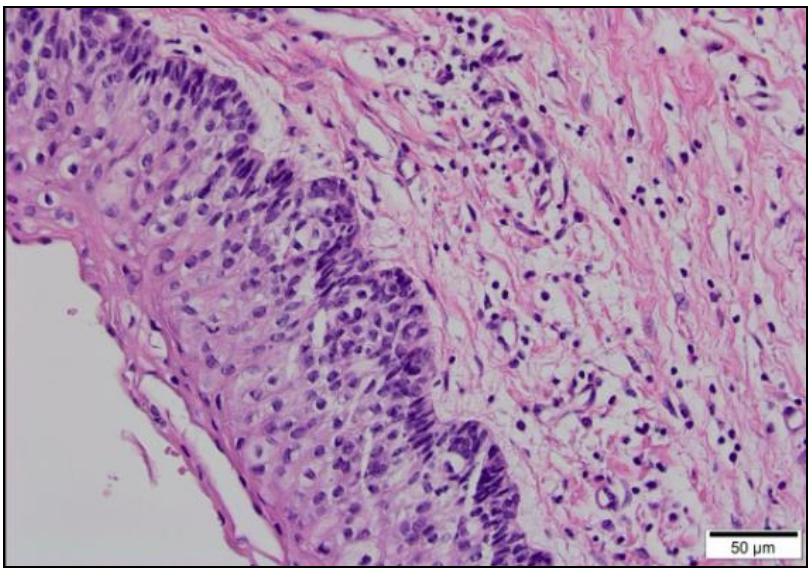

Fig 6: Parakeratinized stratified squamous epithelium with palisaded basal cells and absence of inflammatory infiltrate. (H&E, ×40)

Family history further supported a hereditary pattern, as a close paternal relative reportedly had a history of multiple jaw cysts and was undergoing clinical evaluation for GGS at the time of diagnosis.

Based on the presence of multiple OKCs and additional major diagnostic features, namely calcification of the falx cerebri and bifid ribs, both patients fulfilled at least two major diagnostic criteria for GGS, according to the diagnostic guidelines proposed by Evans *et al*. and later refined by Kimonis *et al*. Molecular genetic testing for *PTCH1* mutations was not performed due to its unavailability within the public healthcare system.

Consequently, the diagnosis was established on clinical, radiologic, and histopathological grounds.

Treatment and follow-up

Both patients were managed using a staged conservative surgical approach. Initially, decompression of the cystic cavities was performed with placement and maintenance of a drainage device, and the patients' legal guardian was

instructed on daily irrigation and cleaning of the drains using sterile saline solution.

After histopathological examination confirmed the diagnosis of OKC, definitive surgical management was carried out through enucleation and curettage of the lesions. Teeth directly involved with the cystic cavities were extracted in both cases.

Postoperative recovery was uneventful, and both patients were enrolled in periodic clinical and radiographic follow-up, with no evidence of recurrence as of March 2026 (Fig. 7 and 8). Antibiotic therapy with amoxicillin-clavulanate was administered during hospitalization and continued for seven days postoperatively.

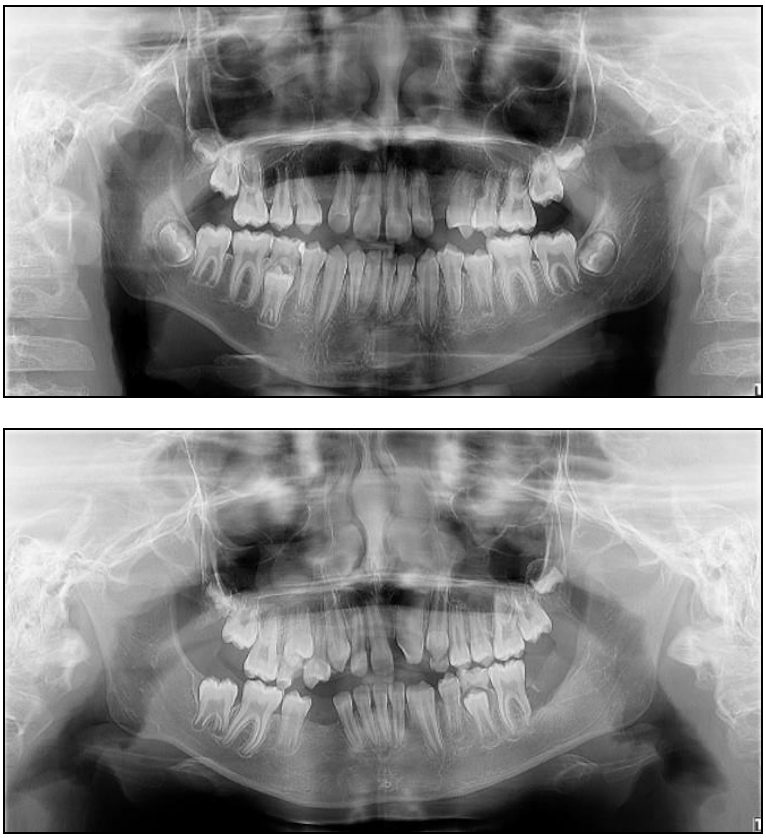

Fig 7 and 8: Follow-up panoramic radiograph from Case 1 and 2, demonstrating mixed dentition and appropriate dental development, with no evident radiographic signs of lesion progression or recurrence

Discussion

The high recurrence rate reported for syndromic OKC has led some authors to advocate resective surgical approaches [7]. However, in paediatric patients, conservative management strategies, such as decompression followed by enucleation and curettage, have been reported to provide favourable outcomes, allowing preservation of bone integrity, developing dentition, and craniofacial growth [5].

In the present cases, this staged conservative approach resulted in satisfactory healing and preservation of function, supporting its use in growing children affected by syndromic OKCs. In addition to preserving craniofacial development, conservative approaches may reduce functional impairment and psychosocial burden associated with more aggressive surgical interventions in paediatric patients.

According to Kimonis *et al*., the majority of patients with GGS develop OKC during the second decade of life, while basal cell carcinomas typically arise after puberty. However, in the present case, both patients developed lesions at a considerably younger age (9 and 7 years old), highlighting the variability in phenotypic expression and reinforcing the importance of early surveillance in paediatric populations [8]

From a histopathological perspective, syndromic OKC differ from solitary lesions by exhibiting increased epithelial proliferation and the presence of satellite and daughter cysts, features associated with a higher risk of recurrence [9]. These familial cases highlight the hereditary and developmental nature of GGS and demonstrate that conservative surgical management, combined with careful clinical, radiologic, and histopathologic evaluation, can be effective even in resource-limited healthcare settings. Long-term multidisciplinary follow-up remains fundamental due to the lifelong risk of new lesion development.

Ethical Approval

This study was conducted in accordance with the principles of the Declaration of Helsinki. This is an observational case report. The institutional Research Ethics Committee confirmed that ethical approval was not required. Informed consent was obtained from the patients' legal guardian for the publication of this report and the accompanying images. Clinical trial number: not applicable.

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Declaration of generative AI and AI-assisted technologies in the manuscript preparation process

During the preparation of this work, the authors used ChatGPT in order to assist with language revision and translation editing. After using this tool, the authors reviewed and edited the content as needed and take full responsibility for the content of the published article.

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