DOI: 10.7860/JCDR/2026/89054.24573

Bilateral Cleft Lip and Palate with Facial Nerve Palsy in Goldenhar Syndrome: A Rare and Severe Phenotypic Expression

SAMIKSHSA TIDKE[1], RANJIT KAMBLE[2], TEJASWI KAMBLE[3]

BY-NC-ND

Keywords: Craniofacial anomalies, Hemifacial microsomia, Oculo-auriculo-vertebral spectrum, Syndromic clefting

Goldenhar syndrome, also known as Oculo-Auriculo-Vertebral Spectrum (OAVS), is a rare congenital craniofacial disorder resulting from abnormal development of the first and second branchial arches during embryogenesis. The condition is classically characterised by a triad of hemifacial microsomia, auricular anomalies, and ocular defects, with variable vertebral and systemic involvement [1]. Although most cases occur sporadically, familial occurrence and autosomal dominant, autosomal recessive, and multifactorial inheritance patterns have been reported. Recent studies have identified variants in genes such as SF3B2, MYT1, PAX1, EYA3, and ZIC3, along with chromosomal abnormalities involving chromosomes 5, 12, 14, and 22. These alterations are believed to disrupt neural crest cell migration, vascular development, and branchial arch formation, contributing to the craniofacial anomalies characteristic of Goldenhar syndrome [2,3].

While the syndrome demonstrates considerable phenotypic variability, bilateral cleft lip and palate and concurrent facial nerve palsy represent particularly uncommon and severe manifestations, underscoring a more widespread developmental disruption. This paper reports a rare case in which all these features co-exist, with a detailed clinical and multimodal radiographic documentation.

An 18-year-old male reported to the Department of Orthodontics with a complaint of facial asymmetry. He was born to non-consanguineous parents, with no family history of congenital anomalies. The patient had undergone bilateral cleft lip repair at six months of age and cleft palate repair at one year of age.

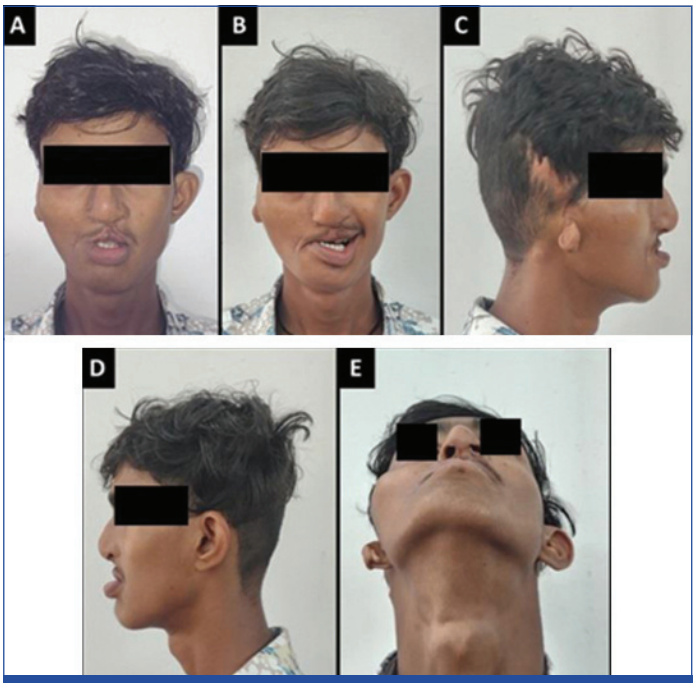
Informed consent was obtained from the patient for clinical examination, treatment planning, and publication of clinical images for academic purposes.

Extraoral clinical photographs revealed marked facial asymmetry attributable to right-sided mandibular hypoplasia. Postsurgical scars from repaired bilateral cleft lip were visible, with a tight upper lip, reduced vermillion display, and whistle deformity [Table/Fig-1].

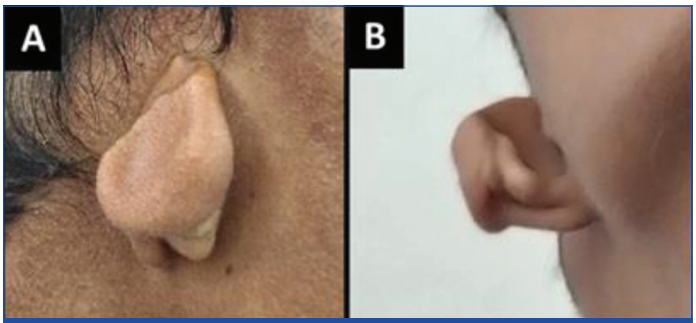
The right ear exhibited grade III microtia with an absent external auditory canal and auricular stenosis [Table/Fig-2].

Features of right facial nerve palsy were evident, including loss of forehead wrinkling, incomplete eye closure, asymmetric facial muscle tone, flattening of the nasolabial fold, and deviation of the oral commissure to the contralateral side during smiling [Table/Fig-3]. Facial nerve involvement corresponded to House-Brackmann Grade IV (moderately severe dysfunction) [4].

Intraoral photographs showed a well-healed palatal scar, severe maxillary constriction with a V-shaped arch form, anterior and posterior crossbite, missing 12,15,22,25,45 and over-retained 55, 65,85 [Table/Fig-4]. The status of the third molars (18, 28, 38, and 48) could not be determined clinically because these teeth often exhibit delayed eruption, impaction, or developmental variation at this age.



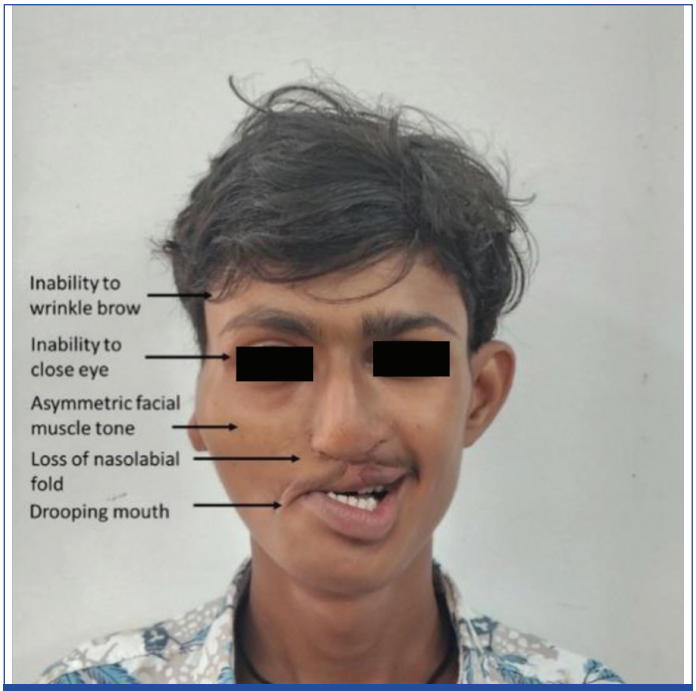
[Table/Fig-1]: Extraoral clinical photographs demonstrating craniofacial asymmetry associated with Goldenhar syndrome: (A) Frontal facial view showing facial asymmetry and repaired bilateral cleft lip scars; (B) Smiling view demonstrating whistle deformity, reduced vermillion display, and deviation of the oral commissure; (C) Right lateral profile; and (D) Left lateral profile showing mandibular deficiency and hemifacial microsomia; (E) Submental view demonstrating mandibular asymmetry and deviation of the chin toward the affected side.



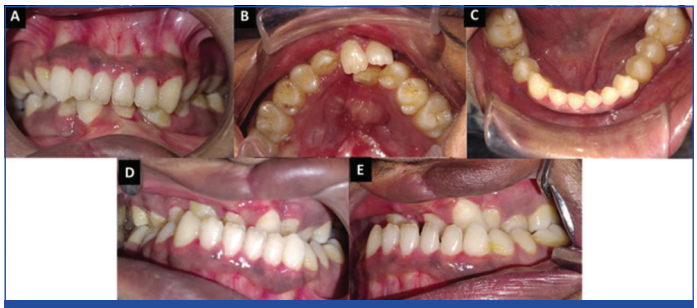
[Table/Fig-2]: Photograph showing grade III microtia with an absent external auditory canal.

Radiographic evaluation was performed using an Orthopantomogram (OPG). The OPG confirmed the absence of teeth 12, 15, 22, 25 and 45, corresponding to the clinical findings, along with retained deciduous teeth 55, 65, and 85. Additionally, the third molars 18, 28, 38, and 48 were absent radiographically. Tooth 85 showed carious involvement with associated root resorption. Analysis of the condyle and mandibular body demonstrated gross asymmetry.

Posteroanterior cephalometric analysis demonstrated deviation of Menton (Me) toward the right side of the facial midline, indicating mandibular asymmetry with chin deviation to the right. Bilateral

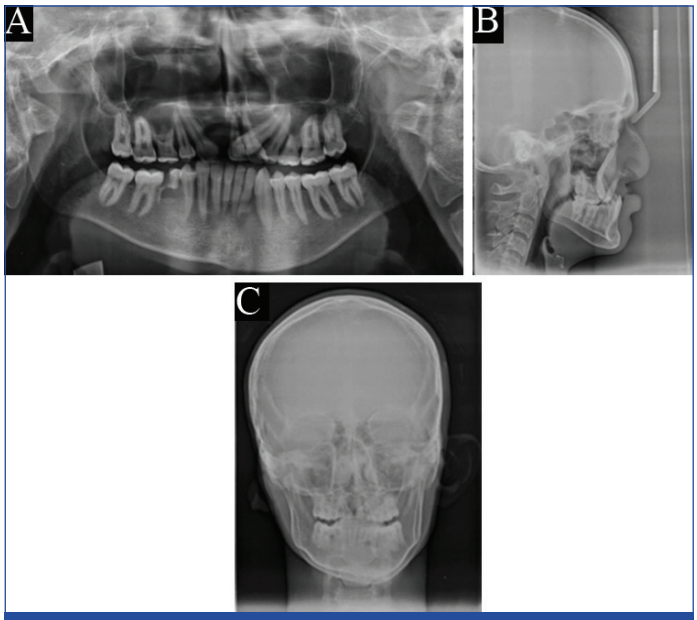


[Table/Fig-3]: Facial expressions illustrating signs consistent with House-Brackmann Grade IV dysfunction.



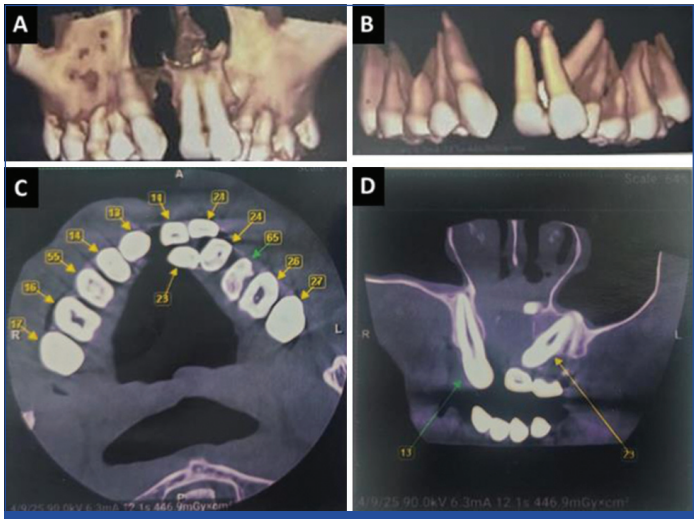
[Table/Fig-4]: Intraoral view: A) Frontal; B) Maxillary arch; C) Mandibular arch; D) Right lateral profile; E) Left lateral profile.

measurements revealed unequal Co–Ag–Me triangular dimensions, with a relatively larger mandibular body on the left and reduced ramus height on the right. These findings suggest unilateral mandibular growth disturbance [Table/Fig-5].



[Table/Fig-5]: Radiographic evaluation: A) Orthopantomogram (OPG); B) Lateral cephalogram; C) Posteroanterior cephalogram.

Cone-beam computed tomography demonstrated a defect in the alveolus in the 11-13 and 21-23 regions [Table/Fig-6a,b]. Palatally and obliquely placed 23 with apex lying in close proximity to the junction of the nasal and sinus floor in the 24 region, condylar and ramal hypoplasia on the right side, canting of the occlusal plane, dental agenesis, and delayed eruption [Table/Fig-6c,d].



[Table/Fig-6]: CBCT imaging showing alveolar defects, oblique displacement of 23, condylar/ramal hypoplasia, and occlusal plane canting: A) Axial CBCT section demonstrating an alveolar cleft defect in the maxillary anterior region; B) oblique displacement of 23; C) Occlusal view showing palatally placed 23; D) Coronal view demonstrating occlusal plane canting and mandibular asymmetry.

For the differential diagnosis, Treacher Collins syndrome was considered due to mandibulofacial dysostosis, but was excluded because it typically presents with bilateral symmetrical involvement and lacks facial nerve palsy [5]. Isolated hemifacial microsomia was ruled out, as it does not usually present with extensive auricular anomalies or cleft lip and palate [6]. Pierre Robin sequence was excluded because it lacks auricular malformations and facial nerve involvement [7]. Nager syndrome was considered but ruled out due to the absence of limb anomalies [8]. CHARGE syndrome was excluded based on the absence of characteristic systemic features and diagnostic criteria [9].

The co-existence of auricular anomalies, hemifacial microsomia, facial nerve palsy, and bilateral cleft lip and palate strongly supports the diagnosis of Goldenhar syndrome [1]. Genetic testing was not performed and the diagnosis was established clinically and radiographically based on characteristic features of Goldenhar syndrome. In addition, financial constraints and limited availability of advanced genetic investigations precluded further molecular evaluation. However, given the severity and breadth of the phenotypic expression, chromosomal microarray analysis and targeted gene panel testing are recommended to identify potential pathogenic variants and facilitate appropriate genetic counseling for the patient and family.

Ophthalmological assessment revealed no ocular abnormalities, including epibulbar dermoids, eyelid coloboma, or Duane syndrome. Clinical examination did not reveal cervical deformity, restricted neck movement, or other signs suggestive of vertebral involvement; therefore, additional spinal imaging was not performed. Audiological and otolaryngological evaluation revealed hearing impairment on the affected side, associated with grade III microtia and absence of the external auditory canal; however, detailed audiometric records were unavailable at the time of reporting. The patient was planned for multidisciplinary management involving orthodontic correction, maxillofacial surgical evaluation for facial asymmetry and mandibular hypoplasia, prosthodontic rehabilitation for missing teeth, and otolaryngology consultation for auricular deformity and hearing assessment.

The patient and guardians were informed about the nature of the condition, associated craniofacial anomalies, treatment options,

expected outcomes, therapy limitations, and the need for long-term multidisciplinary care.

Facial nerve palsy has been reported in a minority of cases and significantly affects facial expression, ocular protection, and psychosocial well-being. In the present case, House-Brackmann Grade IV palsy contributed to both functional impairment and aesthetic compromise [10].

Clinical and radiographic images are invaluable in illustrating the extent of craniofacial deformity and in facilitating diagnosis and treatment planning. Management of such cases necessitates a multidisciplinary approach involving plastic surgery, orthodontics, oral and maxillofacial surgery, otolaryngology, ophthalmology, speech therapy, and psychological support [1].

This case illustrates a rare and severe manifestation of Goldenhar syndrome characterised by bilateral cleft lip and palate, facial nerve palsy, microtia, and mandibular hypoplasia. High-quality clinical and radiographic images play a pivotal role in recognising such complex presentations and emphasise the importance of comprehensive multidisciplinary care.

REFERENCES

[1] Martelli H, Miranda RT, Fernandes CM, Bonan PRF, Paranaíba LMR, Graner E, et al. Goldenhar syndrome: Clinical features with orofacial emphasis. J Appl Oral Sci. 2010;18(6):646-49.
[2] Chamarthi VS, Chamarthi S, Tripathy K. Oculo-Auriculo-Vertebral Spectrum (Goldenhar Syndrome). In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2026.
[3] Strömland K, Miller M, Sjögreen L, Johansson M, Joelsson BME, Billstedt E, et al. Oculo-auriculo-vertebral spectrum: Associated anomalies, functional deficits and possible developmental risk factors. Am J Med Genet A. 2007;143A(12):1317-25.
[4] House JW, Brackmann DE. Facial nerve grading system. Otolaryngol Head Neck Surg. 1985;93(2):146-47.
[5] Marszałek-Kruk BA, Wójcicki P, Dowgierd K, Śmigiel R. Treacher Collins syndrome: Genetics, clinical features and management. Genes (Basel). 2021;12(9):1392.
[6] Young A, Winters R, Spinner A. Hemifacial Microsomia. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2026.
[7] Baxter D, Shanks AL. Pierre Robin Syndrome. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2026.
[8] Marszałek-Kruk BA, Myśliwiec A, Lipowicz A, Wolański W, Kulesa-Mrowiecka M, Dowgierd K. Children with rare Nager syndrome: Literature review, clinical and physiotherapeutic management. Genes (Basel). 2024;15(1):29.
[9] Usman N, Sur M. CHARGE Syndrome. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2026.
[10] Misra I, Rath B, Rath I, Mondal H. Predictors of recovery in facial nerve palsy: Insights from an observational study in Odisha, India. Cureus. 2024;16(4):e58949.

PARTICULARS OF CONTRIBUTORS:
1. Postgraduate Student, Department of Orthodontics, Sharad Pawar Dental College, Wardha, Maharashtra, India.
2. Professor and Head, Department of Orthodontics, Sharad Pawar Dental College, Wardha, Maharashtra, India.
3. Postgraduate Student, Department of Orthodontics, Sharad Pawar Dental College, Wardha, Maharashtra, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Samikshsa Tidke,
Postgraduate Student, Department of Orthodontics, Sharad Pawar Dental College and Hospital, Datta Meghe Institute of Higher Education and Research, Sawangi (Meghe), Wardha-442001, Maharashtra, India.
E-mail: samitidke@gmail.com

PLAGIARISM CHECKING METHODS: [Jain H et al.]
- Plagiarism X-checker: Apr 04, 2026
- Manual Googling: Aug 06, 2026
- iThenticate Software: Aug 08, 2026 (1%)

ETYMOLOGY: Author Origin

EMENDATIONS: 7

AUTHOR DECLARATION:
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Mar 18, 2026**
Date of Peer Review: **May 26, 2026**
Date of Acceptance: **Aug 10, 2026**
Date of Publishing: **Nov 01, 2026**