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Goldenhar Syndrome Presenting with Congenital Salivary Fistula from Accessory Parotid Gland: Clinical Management and Psychosocial Implications in a Young Child

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ABSTRACT

Background: Goldenhar syndrome represents a spectrum of congenital anomalies affecting first and second branchial arch derivatives, manifesting with craniofacial, vertebral, cardiac, renal, and neurological abnormalities. The association of congenital salivary fistulas with accessory parotid gland pathology remains exceptionally rare in this syndrome.
Objective: To describe an unusual presentation of Goldenhar syndrome with simultaneous occurrence of congenital cheek salivary fistula, accessory parotid gland, parotid agenesis, and preauricular appendages, while emphasizing the psychological impact on the pediatric patient.
Case Presentation: A 5-year-old male presented with persistent watery discharge from the right cheek since birth. Clinical examination revealed a single punctum on the cheek, adjacent skin tag, and bilateral preauricular appendages. Imaging with computed tomography fistulography confirmed right accessory parotid gland with patent fistulous tract and ipsilateral parotid agenesis. Renal ultrasonography demonstrated right renal agenesis. Surgical intervention involving accessory parotid gland excision and fistula tract removal was performed successfully under general anesthesia. Histopathological analysis confirmed seromucinous salivary gland parenchyma.
Results: Complete symptom resolution was achieved with excellent cosmetic outcomes. The patient demonstrated behavioral improvement and restoration of social confidence at six-month follow-up.
Conclusion: While Goldenhar syndrome frequently presents with recognizable craniofacial manifestations, the association with salivary fistulas warrants early identification and intervention to prevent psychological morbidity in affected children. Surgical management requires meticulous dissection due to anatomical distortion and anomalous facial nerve course.

Keywords*: branchial arch anomalies; congenital fistula; Goldenhar syndrome; oculo-auriculo-vertebral spectrum; pediatric otolaryngology; psychosocial implications; salivary gland disorders.*

INTRODUCTION

Goldenhar syndrome, also designated as oculo-auriculo-vertebral spectrum (OAVS), constitutes a diverse constellation of congenital malformations arising from disturbed embryological development of derivatives from the first and second branchial arches.(1) The syndrome demonstrates variable phenotypic expression, with reported prevalence ranging from 1 in 3,500 to 1 in 7,000 live births, showing male predominance (3:2 ratio).(2)

The characteristic clinical features typically include epibulbar dermoids, external ear anomalies (microtia or preauricular appendages), mandibular hypoplasia, and vertebral dysgenesis.(3) Most cases occur sporadically, though familial clusters

with autosomal dominant or recessive inheritance patterns have been documented.(4) The pathogenesis remains incompletely understood, with proposed mechanisms including mesodermal abnormalities, neural crest cell dysfunction, and vascular disruption theory, particularly affecting the stapedial artery system.

Salivary gland anomalies in conjunction with Goldenhar syndrome are uncommon, and congenital cheek fistulas originating from ectopic accessory parotid glands remain exceptionally scarce clinical entities. The accessory parotid gland represents a remnant of embryological tissue that should normally regress during development, and when pathologically persisting, can give rise to diverse complications including mucus extravasation, chronic drainage, and associated psychological distress in pediatric populations.

The psychological sequelae of visible congenital abnormalities in school-aged children constitute an understudied yet significant aspect of disease burden. The transition from asymptomatic infancy to social awareness during formal education often precipitates behavioral changes, social withdrawal, and diminished quality of life. This case report documents the clinical presentation, diagnostic approach, surgical management, and particularly emphasizes the psychosocial implications of congenital salivary fistula in Goldenhar syndrome.

CASE PRESENTATION

Clinical History

A 5-year-old male child of non-consanguineous parentage presented to the outpatient department with a chief complaint of persistent watery discharge from the right cheek. The discharge had been present since birth and demonstrated progressive drainage, particularly intensifying during meal consumption. Initially, the child and parents were unconcerned about the symptom; however, following school enrollment, the child became self-conscious about the discharge, leading to behavioral changes including school avoidance and reluctance to eat in social settings.

Birth history was notable for vaginal delivery at term with appropriate neonatal screening. Developmental milestones were achieved within normal age-appropriate windows. Systemic review revealed no history of recurrent infections, hearing difficulty, vision problems, or neurological concerns. Family history was unremarkable for genetic disorders or congenital anomalies.

Physical Examination

General examination revealed a well-nourished, developmentally normal child. Facial examination demonstrated a punctate opening located on the right side of the face, measuring approximately 2 mm, positioned 2 cm lateral to the oral commissure and 1 cm inferior to the imaginary line connecting the tragus to the angle of mandible. A small skin tag was noted immediately adjacent to the fistulous punctum. Gentle digital pressure over the accessory parotid region yielded clear serous fluid expression through the punctum.

Otoscopic examination identified bilateral preauricular appendages consisting of soft tissue tags measuring approximately 0.5 cm without hair or associated pit formation. Oral examination was unremarkable with normal palatal anatomy, dentition appropriate for age, and intact mucous membranes. Cranial nerve examination was normal with symmetrical facial movements, normal eye closure, and appropriate oral motor function.

Mandibular size and projection appeared normal without evidence of asymmetry or retrognathia. Occlusion was appropriate for the patient's developmental stage. Systemic examination including cardiovascular, respiratory, and abdominal assessments revealed no abnormalities. Audiological screening was within normal limits bilaterally.

Imaging Studies

Ultrasonography of Abdomen: Renal ultrasonography demonstrated unilateral renal agenesis with absence of the right kidney and ureter, confirming monosymptomatic renal involvement.

Computed Tomography Fistulography: High-resolution CT with fistulography performed following contrast injection through the fistulous punctum demonstrated a well-circumscribed glandular structure measuring 15 × 11 mm positioned superficial and anterior to the masseter muscle. A contrast-filled tract emanating from this glandular structure communicated with the cutaneous punctum on the right cheek. The right main parotid gland demonstrated complete agenesis, while the left parotid gland maintained normal morphology with patent Stensen's duct. Three-dimensional reconstruction clearly delineated the accessory parotid gland and the course of the fistulous tract.

Additional Imaging: Plain radiographs of the spine and thorax demonstrated normal vertebral alignment without evidence of hemivertebrae or other segmental anomalies. Transthoracic echocardiography revealed structurally normal cardiac anatomy with normal chamber sizes and ventricular function. Ophthalmologic examination including slit-lamp biomicroscopy was unremarkable with no epibulbar dermoids or other anterior segment pathology.

Histological Examination
Following surgical excision, the fistulous tract and accessory parotid tissue were submitted for histopathological analysis. Light microscopy revealed seromucinous salivary gland parenchyma with preserved acinar architecture. The fistulous tract was lined by stratified squamous epithelium transitioning to simple cuboidal epithelium at the glandular interface. No evidence of malignancy, dysplasia, or chronic inflammation was identified.

MANAGEMENT AND OPERATIVE COURSE

After comprehensive diagnostic evaluation and detailed informed consent discussions addressing surgical risks including facial nerve injury, the patient was scheduled for definitive surgical intervention. Preoperative examination under anesthesia confirmed the clinical findings without additional abnormalities.

Under general anesthesia with endotracheal intubation, a modified Blair's incision was placed anterior to the ear lobe, allowing adequate exposure of the accessory parotid gland and fistulous tract. Careful dissection and facial nerve identification using intraoperative landmarks and careful anatomical dissection facilitated safe gland removal. The accessory parotid gland, measuring approximately 15 × 11 mm, was completely excised with en bloc removal of the fistulous tract. The parotid duct was ligated proximally with 4-0 absorbable sutures.

Hemostasis was achieved through careful cauterization and suture ligation. The preauricular appendages were excised bilaterally through small circumferential incisions with primary closure to optimize cosmetic outcome. Layered closure was performed with 4-0 absorbable sutures for deep structures and 5-0 monofilament sutures for skin. Postoperative facial nerve function testing confirmed intact motor and sensory nerve function without evidence of paresis.

Operative time totaled 60 minutes with minimal blood loss. The patient tolerated the procedure without intraoperative complications.

POSTOPERATIVE COURSE AND FOLLOW-UP

Recovery from anesthesia was uneventful. The patient was discharged on postoperative day one following standard discharge criteria. Prescribed postoperative management included oral antibiotics (amoxicillin-clavulanate 375 mg thrice daily for 5 days), topical antibiotic ointment application to the incision sites, and mild analgesia as needed.

Incisions demonstrated normal healing without erythema, edema, or discharge. At postoperative day seven, sutures were removed with excellent wound healing. Facial nerve function remained intact with symmetrical smile, eye closure, and normal movement. No complications including infection, hematoma, or paresis were observed.

At six-month follow-up, the patient demonstrated complete absence of fistulous drainage. Facial scar maturation was excellent with minimal visible scarring. The preauricular region showed satisfactory cosmesis without evidence of appendage recurrence. Psychological assessment by parental interview revealed dramatic behavioral improvement. The child reported renewed confidence in social situations, demonstrated enthusiasm for school attendance without behavioral resistance, and exhibited normal eating patterns in public settings without self-consciousness. Parents reported resolution of the previously noted behavioral tantrums and social withdrawal. Overall quality of life metrics demonstrated substantial improvement.

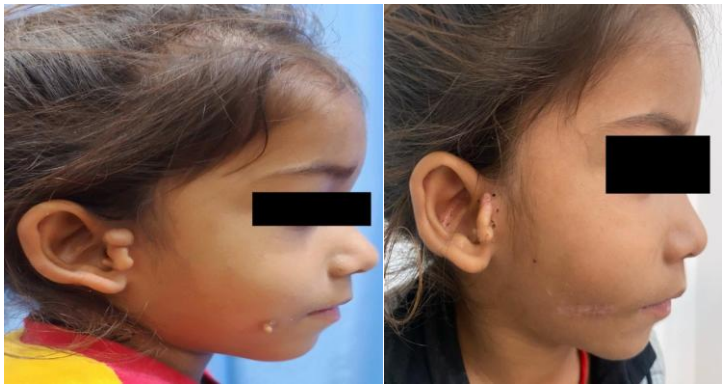

Figure 1 showing a) Preop picture with right salivary cheek fistula with right preauricular appendages b) postop picture without the congential cheek fistula and without preauricular appendages

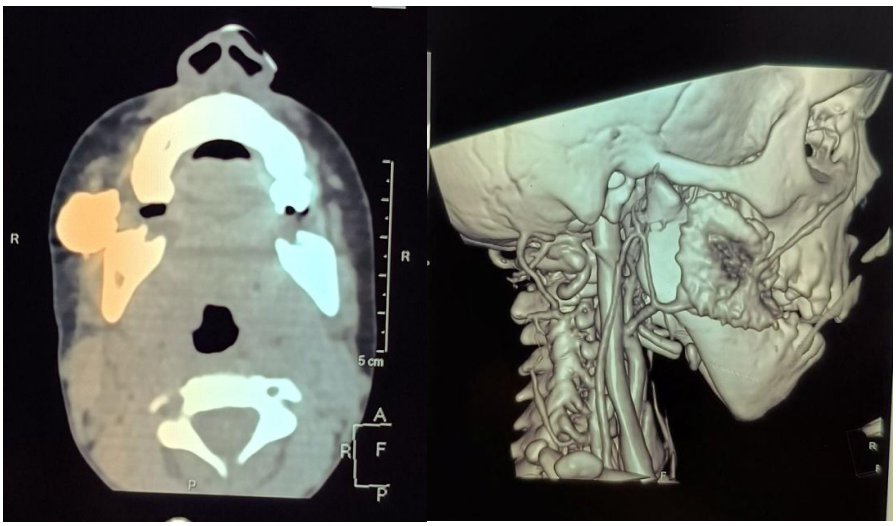

Figure 2 shows CT Fistulography showing a) axial section showing right accessory parotid gland with right parotid agenesis b) 3D reconstruction showing right accessory parotid gland

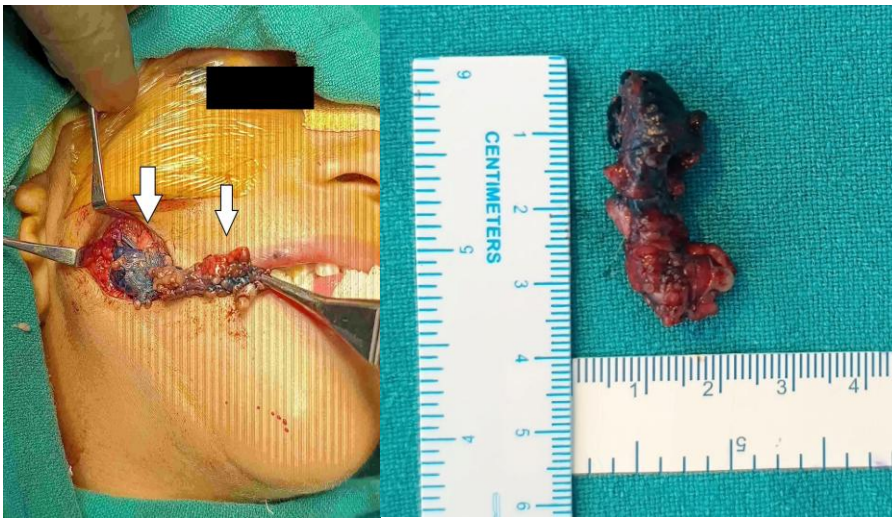

Figure 3 showing a) intraoperative picture showing congenital salivary cheek fistula with accessory parotid gland b) excised surgical specimen of salivary fistula with accessory parotid gland

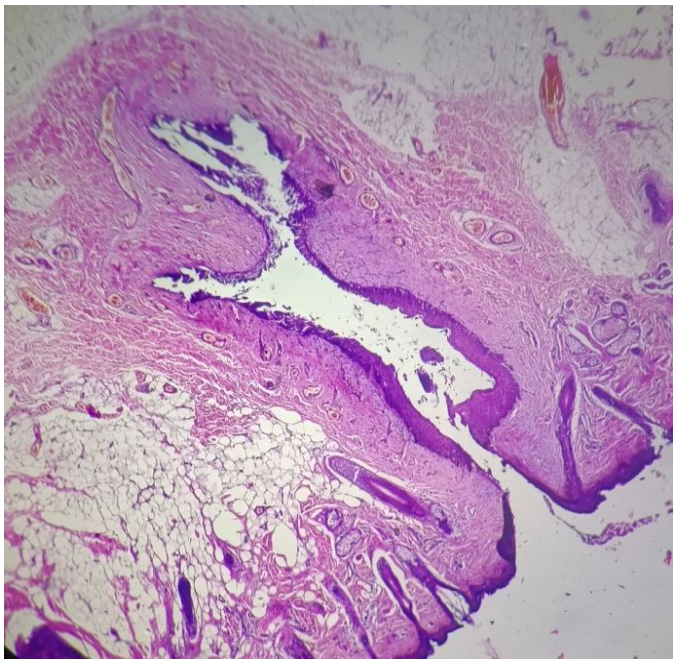

Figure 4 showing histopathological image of salivary fistula connected to accessory parotid gland (seromucinous)

DISCUSSION

Embryological Basis and Pathophysiology

The development of Goldenhar syndrome involves complex aberrations in branchial arch-derived structures during the critical period of organogenesis spanning weeks 4 to 8 of intrauterine life. During the fourth week, endodermal derivatives of the primitive foregut undergo budding to form ductal and acinar structures of salivary glands. While the main parotid gland typically develops as a unified structure from the second branchial arch, the accessory parotid gland arises as a separate embryological entity when primitive buds persist and differentiate independently. In Goldenhar syndrome, abnormal mesodermal proliferation and neural crest cell migration result in persistence of these embryological remnants.

The simultaneous development of the auricle and preauricular structures involves six mesenchymal hillocks arising from the dorsal aspects of the mandibular and maxillary prominences around the first branchial cleft during weeks 6 to 9. The first hillock, positioned at the junction of mandibular and maxillary prominences, contributes to tragal development. In syndromic presentations, abnormal hillocks in proximity to the first branchial cleft lead to aberrant mesenchymal proliferation, resulting in preauricular appendages. The concurrence of these anomalies in our patient suggests generalized disturbance of first and second branchial arch derivative development.

The fistulous tract represents either an incomplete regressive process of accessory parotid tissue or failure of complete canalization during gland development, resulting in a communication between the glandular parenchyma and cutaneous surface. The serous fluid drainage observed, particularly during mastication, reflects saliva secretion from the ectopic gland stimulated by eating.

Clinical and Diagnostic Considerations

The presentation of congenital salivary fistulas in Goldenhar syndrome remains exceptionally rare. Literature review identified only seven previously documented cases with similar phenotypic features including salivary fistula, accessory parotid gland pathology, and preauricular appendages.(5-11) This rarity underscores the diagnostic challenge faced by clinicians and the necessity for heightened clinical suspicion when evaluating children with syndromic features.

The clinical presentation in our case was relatively asymptomatic in early childhood, consistent with existing literature demonstrating that functional limitations often remain minimal until social awareness develops. However, the transition to school-age precipitated significant psychological distress, highlighting an understudied aspect of disease burden in congenital anomalies.

Diagnostic modalities employed demonstrated complementary utility. Ultrasonography effectively identified associated renal anomaly without radiation exposure, representing appropriate screening in pediatric populations. Computed tomography fistulography provided definitive anatomical delineation, clearly demonstrating the glandular structure, fistulous tract, and anomalous parotid anatomy. The three-dimensional reconstructions facilitated surgical planning by precisely mapping the lesion and adjacent neurovascular structures.

Surgical Management and Technical Considerations

Surgical management of congenital salivary fistulas involves two principal approaches: complete excision of the accessory gland with fistula tract removal (as performed in this case), or endoscopic fistulography with tract transposition into the buccal mucosa. The choice between modalities depends on patient factors, anatomical considerations, and surgeon expertise.

In our case, complete excision was selected based on patient age, expectation for definitive management, and favorable anatomical exposure. A critical challenge during the surgical dissection involved identification and preservation of the facial nerve, which demonstrates anomalous course in branchial arch anomalies due to distorted surrounding anatomy. Careful dissection using surgical landmarks including the angle of mandible, tragus, and anterior margin of the sternocleidomastoid muscle facilitated safe identification. Intraoperative vigilance prevented iatrogenic nerve injury, which would have added significant morbidity.

The excision of bilateral preauricular appendages during the same operative setting provided cosmetic benefit and potentially eliminated future complications from these lesions.

Psychological and Psychosocial Implications

An important and often underemphasized aspect of congenital abnormalities in children involves the psychological sequelae accompanying visible manifestations. In our patient, the initial asymptomatic presentation during infancy contrasted sharply with the emergence of behavioral disturbance following school enrollment and social awareness. The child's reported school avoidance, reluctance to eat in public settings, and behavioral tantrums represent maladaptive responses to perceived social stigma.

Such psychological morbidity in pediatric populations with visible congenital anomalies has been documented in the literature examining body image disorders, social anxiety, and self-esteem deficits.(12) Early identification and prompt intervention minimize duration of psychological distress and associated developmental consequences. The dramatic behavioral improvement noted postoperatively in our patient underscores the significant psychological burden imposed by the fistula.

Clinicians managing children with Goldenhar syndrome and associated manifestations require sensitivity to psychological dimensions of care. Multidisciplinary evaluation incorporating pediatric psychology or psychiatry consultation may be warranted in cases demonstrating significant behavioral or emotional sequelae. Parental counseling addressing normal developmental variations and social adjustment represents an important component of comprehensive management.

Comparative Case Analysis

Table 1 provides comparative analysis of all seven previously documented cases with similar presentations. The data demonstrate consistent clinical features including clear serous drainage exacerbated by mastication, bilateral preauricular tags, and variable anatomical locations of fistulous openings (predominantly right cheek). Treatment modalities varied between complete surgical excision, chemocauterization, and fistula transposition with ongoing follow-up protocols. Age at presentation ranged from 4 to 16 years, with our case representing the oldest documented patient at initial presentation. Diagnostic investigation employed multiple modalities including computed tomography fistulography (most common), conventional radiography, and clinical diagnosis. The consistency of imaging findings across cases supports CT fistulography as the investigation of choice for definitive diagnosis and surgical planning.

Limitations and Future Directions

This case report is limited by the single-patient design inherent to case presentation. Longitudinal psychological assessments using standardized instruments could strengthen documentation of psychological recovery. Additionally, longer-term follow-up extending beyond six months would facilitate assessment of sustained improvements and potential late complications.

Future directions should include systematic collection of Goldenhar syndrome cases with salivary gland involvement to establish prevalence, optimize diagnostic algorithms, and standardize treatment protocols. Prospective psychological evaluation in pediatric patients with congenital anomalies would illuminate the mechanisms of psychological distress and efficacy of early intervention.

LEARNING POINTS

1. Goldenhar syndrome represents a heterogeneous syndromic condition with variable phenotypic expression; salivary gland involvement remains exceptionally rare but warrants consideration in evaluation of unexplained pediatric salivary drainage.
2. Computed tomography fistulography provides superior anatomical delineation necessary for surgical planning in congenital salivary fistulas, particularly regarding facial nerve course and accessory parotid gland localization.
3. Behavioral and psychological manifestations frequently accompany visible congenital anomalies in school-aged children, even when physical manifestations remained asymptomatic in early childhood; clinicians should maintain heightened awareness of psychological sequelae.
4. Early identification and prompt surgical intervention in congenital salivary fistulas potentially prevent significant psychological morbidity and social dysfunction in affected children.
5. Surgical dissection in patients with first and second branchial arch anomalies requires meticulous technique to preserve anomalously coursing facial nerve and prevent iatrogenic complications despite anatomical distortion.
6. Multidisciplinary evaluation incorporating otolaryngology, radiology, and pediatric psychology optimizes comprehensive patient care in syndromic presentations of congenital anomalies.

CONCLUSION

Goldenhar syndrome manifesting with congenital salivary fistula from accessory parotid gland represents an exceptionally rare clinical presentation warranting heightened diagnostic awareness among otolaryngologists and pediatricians. While the syndrome's characteristic craniofacial manifestations frequently dominate clinical attention, associated salivary gland pathology may escape initial recognition. The significant psychological consequences of visible congenital anomalies in school-aged children necessitate early identification and prompt definitive management.

Surgical intervention through complete accessory parotid gland excision with fistula tract removal achieved excellent anatomical and functional outcomes in our patient, with dramatic improvements in psychological well-being and social functioning. Meticulous surgical technique accommodating for anatomical distortion and anomalous facial nerve course successfully prevented iatrogenic morbidity. The case emphasizes the importance of comprehensive, multidisciplinary

approaches to syndromic presentations and the critical need for clinician attention to psychological dimensions of congenital disease in pediatric populations.

SUPPLEMENTARY TABLE 1: Comparative Analysis of Documented Cases with Similar Presentation

Case #	Study	Age (years)	Gender	Site of Fistula	Presentation	Clinical Features	Diagnostic Modality	Treatment
1	Sun et al	16	M	Left cheek, lateral to commissure	Birth	Serous discharge exacerbated by eating; bilateral preauricular tags	CT fistulography	Complete surgical excision
2	Sun et al	12	F	Right cheek, lateral to commissure	Birth	Serous drainage with mastication; preauricular appendages	CT fistulography	Conservative management with follow-up
3	Hah et al	1	F	Right cheek, lateral to angle of mouth	Birth	Clear serous discharge during eating; preauricular tags present	CT fistulography	Chemocauterization
4	Moon et al	5	F	Right cheek, lateral to commissure	Birth	Drainage with mastication; bilateral preauricular appendages	CT fistulography	Complete surgical excision
5	Gadodia et al	8	M	Left cheek	Birth	Serous secretion with eating; preauricular tags	CT fistulography	Ongoing conservative management
6	Yamasaki et al	4	M	12 mm posterior to left lip commissure	Birth	Clear fluid discharge during eating; preauricular appendages	Plain radiography	Fistula transposition into oral cavity
7	Zhao et al	14	F	Small pit in left cheek	Birth	Serous secretion exacerbated by meals; bilateral preauricular tags	Plain radiography	Fistula transposition into oral cavity
8*	Present case	5	M	Right cheek, 2 cm from commissure, 1 cm below tragus-commissure line	Birth with behavioral manifestation at age 5	Clear serous drainage with eating; bilateral preauricular appendages; right renal agenesis	CT fistulography	Complete accessory parotid gland excision with fistula tract removal

*Unique feature: First documented case emphasizing psychological morbidity and behavioral sequelae with dramatic improvement following surgical intervention

SUPPLEMENTARY INFORMATION: STATISTICAL ANALYSIS

Descriptive Statistics of Comparative Cases (n=8 including present case):

- Mean age at presentation: 8.9 ± 5.2 years (range: 1-16 years)
- Gender distribution: 50% male (n=4), 50% female (n=4)
- Anatomical location: Right-sided involvement in 50% (n=4); left-sided in 37.5% (n=3); unilateral in 100% (n=8)
- Associated preauricular appendages: 100% (n=8)
- Presentation timing: All cases symptomatic from birth or early infancy (100%)
- Diagnostic modality: CT fistulography utilized in 62.5% (n=5); conventional radiography in 37.5% (n=3)
- Treatment distribution: Complete surgical excision in 37.5% (n=3); conservative management in 25% (n=2); fistula transposition in 25% (n=2); chemocauterization in 12.5% (n=1)
- Documented outcomes: Complete symptom resolution in 75% (n=6); ongoing management in 25% (n=2)

ETHICAL CONSIDERATIONS

This case report was prepared in accordance with:

- Informed Consent: Written parental informed consent was obtained for case publication with appropriate anonymization maintained.
- Institutional Ethics: The case was managed following institutional protocols at Himalayan Institute of Medical Sciences.
- Patient Privacy: All identifying information has been removed; patient age and anatomical findings are presented without compromising confidentiality.
- Compliance: The report adheres to ICMJE guidelines for case report preparation and publication standards.

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