

A Rare Case of Klippel-Feil Syndrome with Cholesteatoma, Thrice Operated with Final Outcome of Cochlear Implant

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ABSTRACT

Klippel-Feil syndrome (KF) is defined as the congenital fusion of two or more vertebrae. Due to skeletal abnormalities, patients with KF syndrome may potentially present with hearing loss. To the best of our knowledge, this is a rare case of an adolescent patient with underlying KF syndrome and cholesteatoma. Our patient underwent three successive mastoid operations, primarily due to middle ear disease, with cochlear implantation for hearing rehabilitation.

INTRODUCTION

Klippel-Feil syndrome is defined as the congenital fusion of two or more vertebrae. Otorhinolaryngological manifestations in Klippel-Feil (KF) syndrome are hearing loss (52% conductive hearing loss, 40% sensory neural hearing loss, as well as mixed hearing loss), dysphagia (39%) and airway issues (i.e., sleep disorder breathing, vocal cord problems, and/or acute or chronic rhinosinusitis).¹ Klippel-Feil syndrome with cholesteatoma is a rare condition. Herein we report a rare case of an adolescent patient with Klippel-Feil syndrome and cholesteatoma.

CASE REPORT

A 16-year-old boy with Klippel-Feil syndrome and a history of generalized dystonia, scoliosis and tracheoesophageal fistula repair presented to our clinic with left ear otorrhea for the past four years. He has been using bilateral hearing aids since the age of 5, due to pre-existing moderate-to-severe hearing loss. His first mastoidectomy was performed after the initial presentation of left ear discharge. Unfortunately, post-operatively, he continued to have persistent ear discharge. A high-resolution computed tomography (HRCT) scan of temporal bones reported as cholesteatoma of the left ear,

for which he underwent left modified radical mastoidectomy. Intra-operative findings revealed left external ear canal stenosis, filled with mucopus, a sclerotic mastoid, an anteriorly placed sigmoid sinus, and cholesteatoma filling the entire antrum. Only the remnant of incus seen over the epitympanum. The malleolus head, short process and remnant of incus was removed to facilitate the surgery. Post-operatively, he still complained of left scanty intermittent serous, foul smelling ear discharge. There was neither otalgia, blood stain otorrhea, vertigo, tinnitus nor any sign to suggest intracranial complications. As predicted, his hearing loss progressively worsened. Left otoscopy showed evidence of cholesteatoma with keratin debris at the neotympanum and the mastoid bowl with a high facial ridge.

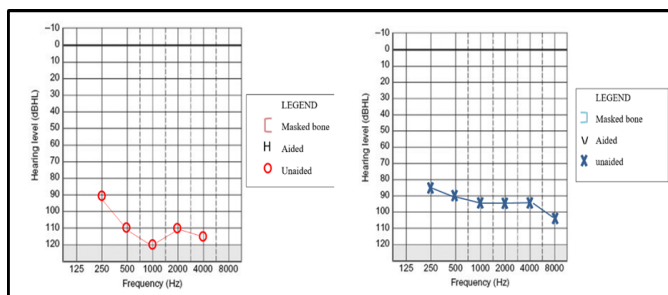


Figure 1: Pre-operative pure tone audiometry shows bilateral profound hearing loss.

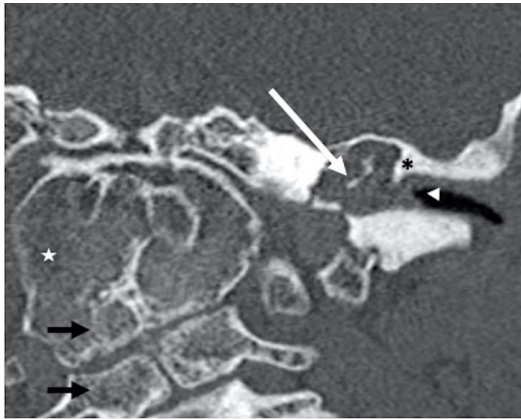


Figure 2: Coronal reformatted view of HRCT of left temporal bone and upper cervical spine shows soft tissue density within the middle ear cavity (*arrow*) with involvement of the epitympanum and extension to the external ear canal (*arrowhead*). Erosion of the scutum (*dark asterisk*) and ossicular chain are present. Note also the abnormal occipito-atlanto and C1/C2 vertebral fusions (*star*) with C2 and C3 hemivertebrae (*dark arrows*).



Figure 3: Axial view of HRCT left temporal bone (A) and (B) show soft tissue density within the middle ear cavity (*arrow*) and the external ear canal (*arrowhead*). The tympanic membrane is not visualized with loss of normal configuration of the ossicular chain, likely due to erosion. The soft tissue density extends to the mastoid cavity (*asterisk*), which appears to coalesce with the middle ear. Bony inner ear structures are normal.

Pure tone audiometry revealed bilateral profound hearing loss (Fig. 1). The repeated HRCT temporal bones reported as residual of cholesteatoma that involved the epitympanum, extension to external ear canal, erosion of the scutum, and the ossicular chain (Figs. 2 and 3). He underwent left redo modified radical mastoidectomy. Intra-operative findings revealed cholesteatoma in the mastoid bowl. There was tegmen mastoideum dehiscence, and the cholesteatoma sac was occupying the mastoid cavity, antrum, epitympanum, and mesotympanum. Post-operatively, there were no complications, and he went on to have an uneventful recovery. He then became a cochlear implant candidate.

DISCUSSION

Klippel-Feil syndrome (KFS) was first described by Maurice Klippel and Andre Feil (1912)¹. KFS usually has clinical a triad of short neck, limited neck movement and

low posterior hairline. Less than 50% of patients have a classical triad.² The prevalence of KFS was one in 40,000 newborns worldwide, with female predominance (60%).² KFS can has multiple otorhinolaryngology manifestations. The most common abnormalities are hearing loss, which is found in about one third of KFS patients¹. Some studies reported 65% of KFS having some form of hearing loss, and 60% having associated anomalies of the external, middle and/or inner ear^{3,4}.

Kenna et al. (2018) reported that only two out of 95 KFS patients presented with cholesteatoma¹. There was a reported case of a 21-year-old female with underlying KFS, who presented with cholesteatoma, profound hearing loss and required twice mastoid surgery. Similarly, our 16-year-old patient had profound hearing loss, but underwent three mastoid surgeries. Canal wall down (CWD) surgery may be challenging due to difficulty in positioning the patient's head intra-operatively. This is because the patient had limited neck movement. The recurrence rate of cholesteatoma is between 0-7% after the CWD approach, due to either insufficient lowering of the facial ridge or inadequate meatoplasty⁶. Our patient had a high facial ridge, which may produce a large volume of middle ear and the formation of a retraction pocket. The CWD approach is a primary choice of treatment for tympano-mastoid cholesteatoma with a very low rate (7.7%) of recurrent or residual after follow-ups over 10 years⁷.

Cochlear implant is a hearing rehabilitation of choice for patients with bilateral profound hearing loss. The patient's ear post-CWD surgery and disease-free, can still be implanted⁸. Our patient will be implanted with a cochlear implant in the future.

CONCLUSION

Otitis media with cholesteatoma in a KF syndrome child should be treated vigorously. The treatment of cholesteatoma is to prevent progression and complication of disease. This patient requires cochlear implantation on the contralateral ear for hearing rehabilitation after the affected ear remains dry.

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