KLIPPEL-FEIL SYNDROME AND HEARING LOSS

(Received 5 June, 1996)

A. Koç, M.D.* / A. Tutkun, M.D.** / C. Batman, M.D.****
C. Üneri, M.D.* / M. A. Şehitoğlu, M.D.*

* *Professor, Department of Otorhinolaryngology, Faculty of Medicine, Marmara University, İstanbul, Turkey.*
** *Associate Professor, Department of Otorhinolaryngology, Faculty of Medicine, Marmara University, İstanbul, Turkey.*
*** *Chief Resident, Department of Otorhinolaryngology, Faculty of Medicine, Marmara University, İstanbul, Turkey.*

ABSTRACT

A wide variety of anomalies of the middle and inner ear has been reported in association with congenital deafness in Klippel-Feil syndrome. This syndrome is usually associated with sensorineural hearing loss. Conductive or mixed type hearing losses are rare. In this article this rare clinical entity and our case is discussed.

Key Words: Klippel-Feil, Hearing loss, Sensorineural.

INTRODUCTION

Klippel-Feil syndrome is characterized by fusion of two or more cervical vertebrae and,in its most severe form, consists of massive cervical fusion, short neck, limitation of head movement and low posterior hairline. Over 300 papers have been written on the subject, approximately 350 patients have been reported to date.

Klippel and Feil described the clinical picture in 1912, and Feil published 13 cases in 1919. The cause of this syndrome is unknown. Animal studies suggested the possibility of Klippel-Feil syndrome being the end result of teratogens in humans.

Associated anomalies of this syndrome include cleft palate, frontonasal malformations, scoliosis, hemiplegia, congenital heart defects, spina bifida, meningocoele, torticollis, renal anomalies and chorioretinal atrophy (1-3).

Wildervanck syndrome consists of Klippel-Feil anomaly, abducens paralysis with retracted bulb, and sensorineural or conductive deafness. Many reports describe the associated deafness as being sensorineural and reports of conductive losses are rare (4-9).

The purpose of this paper is to present a case of Klippel-Feil syndrome with sensorineural hearing loss and discuss the syndrome.

CASE REPORT

A 5-year-old girl was referred to our clinic for hearing aid. Her history revealed hearing loss suspected by her family since she was two years old. She was followed by different clinics. She was the only child of the family and the pregnancy, delivery and postnatal period were reported as uneventful. Otoscopic examination showed normal tympanic membranes and well-aerated middle ears. Short neck was noted during this examination, and radiographic examination showed deficiency of cervical vertebraes and fusion of lower cervical vertebraes. There was no abducens palsy. Findings at general physical examination revealed diminished mobility and webbing of the neck, and a low posterior hairline (Fig. 1-2). She was noted to have a heart murmur and it was diagnosed as a combined aortic stenosis with slight mitral insufficiency. Computed tomographic examination of temporal bones showed normal structures, including normal internal auditory canals. In audiological examination the responses of the child to speech and to warble tones were tested in the free field by CAVR technique. Speech responses were recorded at 80 dB level. Warble tones were recorded only at 250 and 500 Hz at high tone levels. Auditory brainstem response revealed no response. These findings suggested bilateral sensorineural hearing loss.

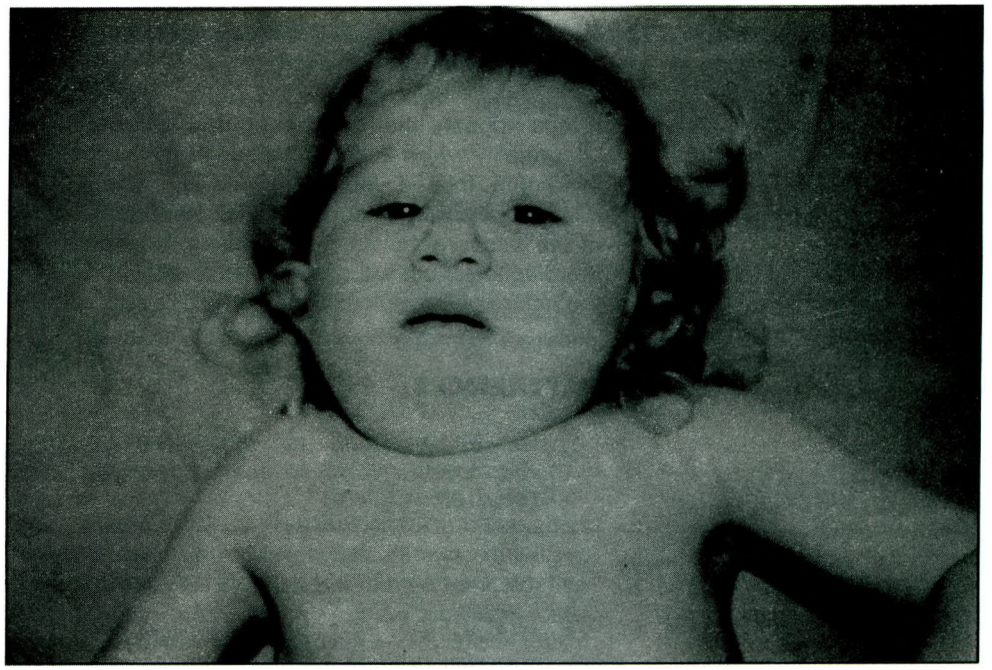

Fig. 1: *Klippel - Feil Syndrome*

Fig. 2: *Klippel - Feil Syndrome*

DISCUSSION

The hearing loss associated with Klippel-Feil syndrome is usually sensorineural and reports of conductive losses are rare. Windle-Taylor et al. described ten cases with hearing loss where only two of them were conductive (10).

In reviewing the findings in the literature, it can be concluded that there is a very wide spectrum of ossicular chain anomalies (1,6-10). In some cases there were no ossicles at all and this was combined with aplasia of the oval window niche.

Sakai et al. described one case in which a tympanostomy was performed. Findings included an atrophic long prosess of the incus and a fixed stapes footplate. Stapedectomy was succesful (5).

Jarvis et al. reported one case of bilateral conductive loss in whom tympanotomies were carried out (7). The right middle ear contained a rudimentary

malleus, absent incus and diminutive stapes. The facial nerve was dehiscent. A homograft incus interposition was performed with a small gain in hearing. Left tympanotomy showed fixation of the stapes and an abnormally narrow external auditory meatus. Again there was a dehiscent facial nerve. A stapedectomy was successfully performed.

Livingtone and Delahunty reported findings in four ears (8). In one of them, the incudostapedial joint was absent because the long process of the incus was too short, but the stapes was mobile. Reconstruction of the ossicular chain with an allograft incus was successful.

Van Rijn reported 2 ears with fixed stapes which stapedectomy and teflon interposition was performed and the hearing level returned to the preoperative values (9).

It appears that the absence of the incudostapedial joint is an ideal indication for reconstruction of the ossicular chain. Successful surgery can be expected when the stapes is mobile. But the results of surgery are unpredictable. The risk of a perilymph gusher is high in these patients and may result in profund sensorineural loss.

Singh et al. reported one case of conductive deafness on whom tympanotomy was performed (6). No abnormalities were found. They proposed that the deficit in sound transmission is at the cochlear fluid level or a defect at the transmission level of the inner ear.

Schild et al. reported patients with the Wildervanck syndrome that showed bilateral Mondini deformities, with cystic cochlea, absent modioli and deficient septa between cochlea and vestibules, which were dilated (4).

Radiological investigation of patients with Wildervanck syndrome has revealed a variety of otological malformations. Wildervanck emphasized a notable feature that is common in most cases as the abnormality of the vestibular labyrinth on tomographic examination (11). Gross abnormalities of the inner ear and internal auditory meatus were reported by Kirkham in one of two patients (12), by Wildervanck in two out of three (11), and by Regenbogen and Godel in one patient (13). In their report Windle-Taylor et al. have stated that 4 of their 11 patients that showed dysplasia with a short fat cochlea, large vestibule and wide, small or missing semicircular canals and immature sensorineural structures. The other 7 patients had non-specific inner ear dysplasias affecting the sensorineural canals. Additional 3 patients showed a complete absence of internal auditory meatus, and 5 showed a reduction in the size of the same structure (10).

It thus appears that there is no one with otological abnormality which is characteristic of the Klippel-Feil syndrome. Indeed, more than one abnormality may coexist in the same patient. Radiological examination of the temporal bones may not help in establishing the diagnosis, but will assist considerably in defining the nature of the otological defect.

REFERENCES

1. *Stewart EJ, O'Reilly BF. Klippel-Feil syndrome and conductive deafness. J Laryngol Otol 1989:103;947-949.*
2. *West PDB, Gholkar A, Ramsden RT. Wildervanck's syndrome-unilateral Mondini dysplasia identified by computed tomography. J Laryngol Otol 1989:103;408-411.*
3. *McBride WZ. Klippel-Feil syndrome. Am Fam Physician 1992:45;633-635.*
4. *Schild JA, Mcfee MF, Miller MF. Wildervanck syndrome - The external appearance and radiological findings. Inter J Pediatr Otorhinolaryngol 1984:7;305-310.*
5. *Sakai M, Hirosater M, Shinkawa A, Komatsu N. Klippel-Feil syndrome with conductive deafness and histological findings of removed stapes. Ann Otol Rhinol Laryngol 1983:92;202-205.*
6. *Singh SP, Rock EM, Shulman A. Klippel-Feil syndrome with unexplained apparent conductive hearing loss. A case report. Laryngoscope 1969:79;113-117.*
7. *Jarvis JF, Sellars SL. Klippel-Feil deformity associated with congenital conductive deafness. J Laryngol Otol 1974:88;285-289.*
8. *Livingstone G, Delahunty JE. Malformations of the ear associated with congenital ophthalmic and other conditions. J Laryngol Otol 1968:82;495-504.*
9. *Van Rijn PM, Cremers CW. Surgery for congenital conductive deafness in Klippel-Feil syndrome. Ann Otol Rhinol Laryngol 1988:97;347-351.*
10. *Windle-Taylor PC- Emery PJ, Phelps PD. Ear deformities associated with the Klippel-Feil syndrome. Ann Otol Rhinol Laryngol 1981:90;210-216.*
11. *Wildervanck LS, Hoeksema PE, Penning L. Radiological examination of the inner ear of deaf-mutes presenting the Cervico-occulo-acoustic syndrome. Acta Otolaryngol (Stockh), 1966:61;445-453.*
12. *Kirkham TH. Cervico-occulo-acousticus syndrome with pseudopapilloedema. Arch Dis Child 1969:44;445-508.*
13. *Regenbogen L, Godel V. Cervico-occulo-acoustic syndrome. Opht Paediatr Gen 1985:6;183-187.*