Hearing Loss in Klippel Feil Syndrome: A single case study

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DOI: 10.29322/IJSRP.14.06.2024.p15021
10.29322/IJSRP.14.06.2023.p15021

Paper Received Date: 06th May 2024
Paper Acceptance Date: 08th June 2024
Paper Publication Date: 15th June 2024

Abstract- Klippel-Feil syndrome (KFS) is a congenital anatomical defect that occurs due to the failure of fusion of two or more cervical vertebrae in the neck region. The physical features of the condition include a short neck, low hairline at the back of the head, and restricted movement of the upper spine. Otologic anomalies have been reported to be seen in 60% of patients with Klippel Feil syndrome which includes unilateral, bilateral and external, middle & inner ear pathologies. This case study focuses on a 4-year-old female child diagnosed with Klippel-Feil syndrome presenting with bilateral hearing impairment, combined vestibulo-cochlear-semicircular canal dysplasia along with delayed speech and language development. A detailed audiological evaluation was done and the results are further discussed.

Keywords – *Klippel Feil Syndrome (KFS), Hearing Impairment, Vestibulo-cochlear dysplasia, Cytomegalovirus (CMV)*

I. *Introduction*

The vertebral column is the arrangement of bony structures which is also known as the spinal column. It acts as a central axis of the skeleton, supports the body and provides attachment to the muscles and protects the spinal cord. Klippel-Feil syndrome (KFS) is a rare skeletal disorder that is primarily characterized by abnormal union or fusion of two or more bones of the spinal column (vertebrae) within the neck (cervical vertebrae). Some of the affected individuals have an abnormally short neck, restricted movement of the head and neck, and a low posterior hairline at the back of the head which are also known as triad features of KFS. The Klippel Feil syndrome was first described by French physicians Maurice Klippel and Andre Feil in 1912. The syndrome is also referred to as cervical vertebral fusion, congenital dystrophia brevicollis, congenital cervical synostosis, Klippel Feil anomaly and KFS. The condition is congenital in nature, but certain mild cases may go undiagnosed until later during life when symptoms worsen or first become apparent. In many individuals with KFS, the condition appears to occur randomly for unknown reasons (sporadically). In other cases, they are also inherited as autosomal dominant or autosomal recessive traits and are associated with mutation of the GDF6, GDF3 and MEOX1 genes. Due to the mutation, these genes decrease the production the proteins which are responsible for proper bone development, particularly the formation of the vertebrae. Several studies have hypothesized vascular disruption, global fetal insult and primary neural tube complications. Faulty segmentation occurs during gestational weeks between 3 to 8 of embryo development, resulting in a failure of normal segmentation or formation of the cervical somites.

Two classification systems exist for KFS, the original classification of KFS subtypes (I, II and III) as described by Maurice Klippel and Andre Feil based on the degree of cervical fusion and other classification of KFS Classes (KF1-4) as described by Clarke and colleagues based on patterns of inheritance, common associated anomalies and the axial level of the most anterior fusion. The original classification is, KFS type I is characterized by extensive fusion of vertebrae of the neck (cervical vertebrae) and the upper back; type II is characterized by fusion at one or two cervical or thoracic vertebrae; type III is characterized by fusion of vertebrae of the neck as well as the lower thoracic or lumbar vertebrae. According to the Clarke classification, KF1 is the only class presenting with C1-2 fusion, with very short neck and recessive inheritance of type I, II or III fusion patterns; KF2 is autosomal dominant with the most anterior fusion at C2-3 in association with type I, II or III fusion patterns; KF3 is recessive or has reduced penetrance of isolated fusions between any of the cervical vertebrae except C1- 2; KF4 includes cases of Wildervank and Duane syndrome.

Incidence and Prevalence

The exact incidence of Klippel-Feil syndrome is unknown. Most estimates have suggested that 1 in 42,000 to 50,000 people have KFS. The incidence of the condition varies with the type of Klippel-Feil syndrome. Females are affected more often than males, to be more specific approximately 65 percent affected individuals are girls. According to the medical literature, KFS type II appears to be most common form of the

condition. Females appear to be more frequently affected by the Type 1 and Type 3, but there is an equal gender incidence in Type 2. KFS can be associated with a variety of additional symptoms and physical abnormalities which includes abnormal curvature of the spine (scoliosis), spina bifida occulta, Sprengel's deformity, torticollis, rib defects and other skeletal abnormalities including malformations of the ear, nose, mouth and larynx including cleft palate and malformations of the craniofacial area; anomalies of the urinary tract and/or kidney including horse-shoe kidney, hypoplasia or agenesis of one or both kidneys, abnormal renal rotation or placement (ectopia), hydronephrosis due to blockage or narrowing of the ureters; structural abnormalities of the heart such as congenital heart defects particularly ventricular septal defects; and Synkinesia, webbing of the digits and digital hypoplasia, eye abnormalities such as cross-eye or convergent strabismus, nystagmus, ptosis or colobomas; audiological impairments and developmental delay.

KFS may occur as an isolated abnormality (Klippel-Feil anomaly) or as a syndrome with associated anomalies. The syndrome is typically confirmed through clinical examination, symptoms and imaging studies (X-rays, MRI or CT scan). Some people with KFS have few or no symptoms, and are diagnosed by chance after having imaging studies for some other reason. Thus, radiological evaluation plays a major role in diagnosing the syndrome. Management of this syndrome generally depends on the presenting symptoms. The team of medical professionals involved includes Neurologist, Neurosurgeon, Orthopedists, ophthalmologists, surgeon, Pain management specialist, Physical therapist, Audiologist, Cardiologist, etc. depending on the presenting features.

Clinical Features

Audiological abnormalities are also most commonly associated with KFS. Studies have reported that 80% of cases with Klippel Feil syndrome are associated with hearing loss. Hearing loss includes all the types and most commonly observed is bilateral sensorineural hearing loss followed by conductive hearing loss. Otological abnormalities are also associated with this syndrome ranging from nonspecific external ear abnormalities to severe inner ear abnormalities. External ear anomalies include narrow external auditory meati (EAM), preauricular skin tags, and small pinna (microtia). Middle ear anomalies include deformed or absent ossicles, malformed or fixed stapes, fusion of components of the ossicular chain, and non-specific hyperstatic changes of the entire foot plate. Inner ear anomalies commonly seen include absence of vestibules and semi-circular canals. Cochlear anomalies could be total absence of the cochlea, decreased number of coils, and Mondini deformity.

Management of hearing loss is usually based on the radiological and audiological evaluation results. A detailed audiological evaluation should be done prior to decding the appropriate management option. The management of hearing loss depends on the severity and treatment options include the use of hearing aids, cochlear implantation (CI) and Auditory brainstem implant (ABI). In cases with external and middle ear anomalies treatment options also include reconstruction surgery, hearing aids and bone conduction devices. Cochlear Implant (CI) is considered as a management option in cases of inner ear anomalies with accessible cochlea and in other conditions auditory brainstem implant (ABI) is the only option for restoring hearing.

Case Report

Background Information

A 4-year-old child had come to the department of audiology accompanied by her parents with the complaint of not responding to sounds and name call since birth. The child was also reported to have a delay in speech and language development.

Birth History

The child's mother is a known case of hypothyroidism and was affected with Cytomegalovirus [CMV] and Toxoplasmosis infection during pregnancy and was under medication for the same. The child was born 2 days prior to the due date in a normal delivery with a birth weight of 2 kgs and birth cry was reported to be immediate. The child also had respiratory distress & neonatal jaundice and was kept in NICU for 11 days.

Medical History

The child was exposed to cytomegalovirus [CMV] and toxoplasmosis in utero during the first trimester and hence developed multiple congenital anomalies, which is common with TORCH infection during pregnancy. The child was diagnosed with Klippel Feil syndrome- Type I Associated with (?) VACTERL Association. The child also has atrial septal defect and bifid aortic valve. She also had left kidney hydronephrosis with multiple cysts which were large and non-functional and surgery was done to remove the left kidney at the age of 2 years.

Radiological Findings

The MRI of mid-brain revealed normal study. The HRCT of temporal bone shows Mild soft tissue opacification in both middle ear cavities and combined mixed vestibulo-cochlear-semicircular canal dysplasia with mild dilation of Internal acoustic meatus in right ear and hypoplastic internal acoustic meatus in left ear.

Features of Klippel Feil syndrome

The child shows typical triad symptoms of KFS such as fusion of bones in the neck, short neck and low hairline and other signs like torticollis and ptosis. Audiological abnormalities include pre auricular tags and dysplastic vestibulo-cochlear-semicircular canals.

Speech And Language Evaluation

The child was reported to use gestures predominantly to communicate her needs. Formal evaluation was carried out using REELS [Receptive & Expressive Emergent Language Scale] and SECS [Scales of early communication skills] and the results

revealed a delay in receptive and expressive language skills.

Audiological Evaluation

Test battery approach was used for the detailed audiological assessment and the tests include otoscopic examination, Behavioural Observation Audiometry [BOA], Immittance Audiometry, Otoacoustic Emission [OAE], Auditory Brainstem Response [ABR] and Hearing aid trial.

Instruments Used

The instruments were calibrated before the testing. Behavioural observational audiometry was done using Piano Inventis audiometer in free field modality using speakers. Immittance audiometry was done using GSI Tympstar Pro using earphones. Distortion Product Otoacoustic emissions screening was done using Interacoustics Titan with inserts. Auditory brainstem responses were done using Intelligence hearing system (HIS) using ER-3 inserts. Hearing aid trial was done using the Piano Inventis audiometer in free field modality with appropriate amplification device.

Results

Otoscopic Examination

Right ear: Cone of light was not visible due to wax
Left ear: Cone of light was present

Behavioral Observational Audiometry

Behavioral responses observed include Startle response, auroplabebral reflex and localisation only at higher intensities at all frequencies.

Tympanometry & Reflexometry

'B' type tympanogram was observed in both ears with absent ipsilateral and contralateral reflexes indicating the presence of a middle ear pathology.

Otoacoustic Emissions

DPOAE'S were absent in both ears indicative of Outer Hair Cells Dysfunction.

Auditory Brainstem Response

Results revealed Severe to Profound hearing loss in both ears.

Hearing Aid Trial

The Aided Responses were observed to be out of speech spectrum using high power hearing aids in both ears.

Conclusion

The patient presented in this case study underwent an audiological test battery that included behavioral, physiological, electrophysiological tests and radiological evaluations. All the test results indicate the diagnosis of hearing loss associated with the hall mark features Klippel-Feil syndrome. Audiological management options for this unique condition remains challenging because of the co-morbid conditions of the syndrome.

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