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A Case Study of Audiological Test Battery in a Paediatric Patient with Klippel Feil Syndrome

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ABSTRACT

The Klippel-Feil syndrome is a congenital anatomical defect which occurs due to the failure of fusion of two or more cervical vertebrae at the neck region. Features include a short neck, low hairline at the back of the head, and restricted movement of the upper spine. This case study includes audiological test battery of a 3 years old with Klippel-Feil syndrome with features of hearing impairment, severely dysplastic internal acoustic meatus [IAM] along with delayed speech and language development. A detailed audiological evaluation was done and the management options were been further discussed.

Keywords: Klippel-Feil syndrome, hearing loss, CI, ABI

INTRODUCTION

The vertebral column is 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. The Klippel-Feil syndrome is a congenital condition affecting the development of the bones in the spine [1].People with Klippel-Feil syndrome are born with abnormal fusion of 2 or more vertebrae at the neck region, hence they are also defined as the short neck structure with decreased movement and low posterior hairline at the back of the head [1][2]. Radiologically, there is a failure of segmentation of the cervical vertebrae [2]. Over time, individuals with Klippel-Feil syndrome can develop a narrowing of the spinal canal {spinal stenosis} in the neck which can compress and damage the spinal cord [3].

The Klippel-Feil syndrome's cause are not well understood, most cases of isolated Klippel-Feil syndrome they are sporadic in nature [1]. They are also caused by the 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 [3]. This syndrome is been classified into three types, they are; Type 1: extensive fusion of most or all of the cervical spine, Type 2: fusion of 1 or 2 vertebrae in the cervical spine, Type 3: fusion exists in part of the thoracic and/or lumbar spine [4]. The exact incidence of the Klippel-Feil syndrome is unknown. Most estimates suggest that 1 in 40,000 to 1 in

42,000 people have Klippel-Feil syndrome [1].The incidence of the condition varies with the type of Klippel-Feil syndrome. Girls are appeared to be more frequently affected by the type 1 and type 3, but there is an equal sex incidence in type 2 [2]. This syndrome is associated with other anomalies including Scoliosis, Sprengel shoulder ,cleft palate, cardiac problems, renal anomalies, audiological impairments and develop-mental delay [1][2].

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 plays a major role in diagnosing the syndrome .The Genetic Testing Registry (GTR) gives in details about the genetical conditions. Management of this syndrome generally depends on the symptoms being presented. The medical professionals involved Neurologist, Neurosurgeon, Orthopaedic surgeon, Pain management specialist, Physical therapist, Audiologist, Cardiologist, etc depending on the management of features present.[4][5]

Audiological abnormalities are also seem to be most common. Some studies report that Klippel Feil syndrome has the association with hearing loss up to 50%. Hearing loss includes all the types of hearing loss and the most commonly seen is bilateral sensorineural hearing loss and then the conductive hearing loss. Otological abnormalities are also been associated with this syndrome that can range from non-specific external ear abnormalities to severe inner ear abnormalities. External ear anomalies include narrow external auditory meati (EAM), preauricular skin tags, and small ears (microtia). The inner ear anomalies include absence of vestibules and semi-circular canals. Cochlear anomalies could be total absence of the cochlea, decreased number of coils, and a Mondini deformity (one and a half coils of the cochlea is only present). Abnormalities of the middle ear are also described which includes deformed or absent ossicles, malformed or fixed stapes, fusion of components of the ossicular chain and non-specific hyperostotic changes of the entire footplate have been recorded in certain cases. [5]

The management of hearing loss generally depends on the radiological and audiological evaluation reports. Radiological evaluation includes the CT and MRI that reveals the structure of the inner and middle ear. Otoscopic evaluation is used to check the structures abnormalities of the ear canal and tympanic membrane. Pure tone audiometry- in case of child behavioural observational audiometry can be done. It is a subjective test used to assess the hearing. A sound is presented and the child's responses are noted. Responses can be eye blinking, crying, smiling, startle responses, etc. and it varies depending on degree of hearing loss. Immittance audiometry assesses the function of the middle ear and the findings help us to know the intact function of middle ear. Oto-acoustic emission assesses the function of outer hair cells. Auditory brainstem responses are a physiological measure used for threshold estimation and to assess retro cochlear pathology. Radiological test is to check the structures of inner ear and brain.

The management of hearing loss depends on the severity of hearing loss, treatment options include the use of hearing aids, cochlear implantation (CI) and Auditory brainstem implant (ABI).

In case of External ear anomalies and middle ear anomalies treatment options could include the Reconstruction surgical options, hearing aids and bone conduction devices.

In case of inner pathologies where the cochlear could be accessible or at least half of it is developed then cochlear implant could be an option. Cases where CI could not be done auditory brainstem implant (ABI) is the only option for restoring hearing.

CASE STUDY

Background Information:

A Child of 3years old was accompanied with his parents to the Department of Audiology with a concern of not responding to loud sounds and name calls. The problem was been noticed since birth and is reported to be unchanged from the time of notice till present. The child was also reported to have a delay in speech and language development.

Birth History:

Pre-Natal History:

The mother was reported to have gestational diabetes during the last trimester.

Peri-Natal History:

This reveals that the child was born out of post-term caesarean delivery, was reported to have a birth weight of 2.86kg with a normal birth cry.

Post-Natal History:

No significant post-natal history was been reported.

Previous Medical Intervention:

The child was been diagnosed with VSD [ventricular septal defect] and has done meningeal repair at the back of the neck [C2-C3].

Radiological Findings:

The MRI scan findings reveal severely dysplastic internal acoustic meatus [IAM] bilaterally which is greater at the left side compared to the right.

A mild dilation of the lateral ventricles with the post-operative changes of the occipital region.

Developmental History:

The child's developmental history was reported to be normal.

Speech And Language Evaluation:

The child was reported to use gestures predominantly to communicate On formal evaluation with GA (Gestural assessment) and SECS (Scales of early communication skills) test , the results revealed a delay in receptive and expressive language skills.

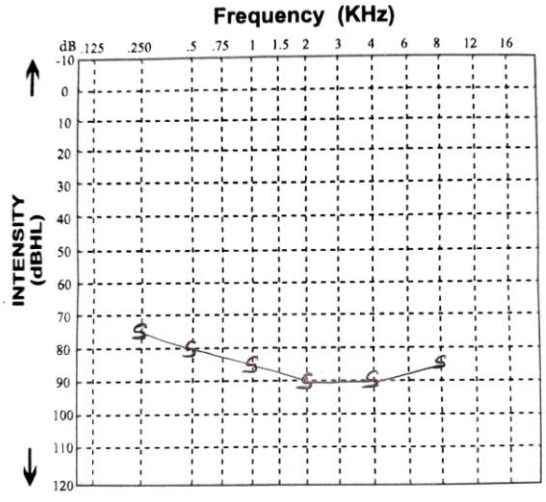
Audiological Evaluation:

Test battery approach was used for the audiological evaluation of the child. The test battery was used for the detailed audiological assessment purpose and the tests include Behavioural observational audiometry (BOA), Immittance audiometry, Otoacoustic emission, Auditory brainstem response and Hearing aid trial.

Instruments used:

Before testing the instruments were calibrated to check the output and the calibrated values were corrected in the reports accordingly. The behavioural observational audiometry was done using piano inventis audiometry in free field modality using speakers. Immittance audiometry was done using clarinet inventis using earphones. Otoacoustic emission-DPOAE screening was done using Natus echoport screener with inserts. Auditory brainstem responses were done using Intelligence hearing system (IHS DP-OAE 4.70) using ER-3 inserts. Auditory Hearing aid trail was done using the piano inventis audiometry in free field modality with respected amplification device.

Behavioural Observational Audiometry:



With respect to the test performed, the child was reported to have Severe to Profound Hearing Loss.

The behaviours observed were:

- Auropalpebral reflex
- Searching for sound

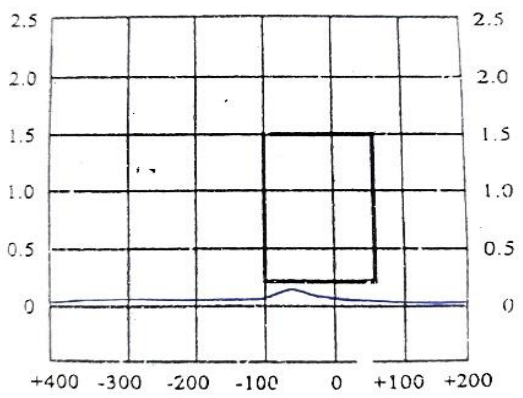
- Crying

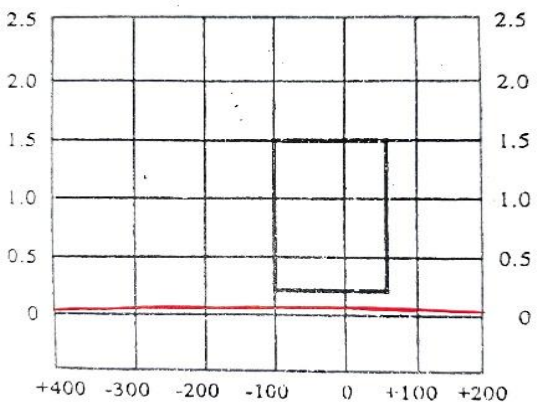
Provisional Diagnosis: Severe to Profound Hearing Loss.

Immittance Evaluation:

Findings:

EAR	TYMPANOMETRIC TYPE
RIGHT EAR	TYPE B
LEFT EAR	TYPE As





The test results revealed that the child had Bilateral Indication of Middle Ear Pathology. The Reflexes were also reported to be absent.

Impression:

Both ears: Indication Of Middle Ear Pathology.

Reflex Audiometry:

Both ears: Absent

Otoacoustic Emissions:

OAEs	Right	Left
DPOAEs	Absent	Absent

The Frequencies tested are: 250Hz,500Hz,1KHz,2KHz,4KHz,6KHz,8KHz.

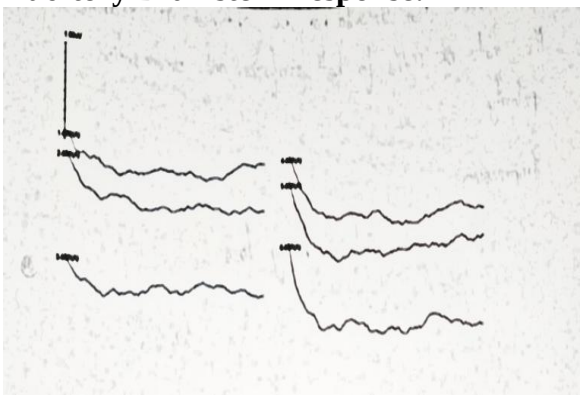
Findings

Both Ears: DPOAE'S absent

Impression

Both Ears: Suggestive Outer hair cell Dysfunction In Both Ears.

Auditory Brainstem Response:



Findings:

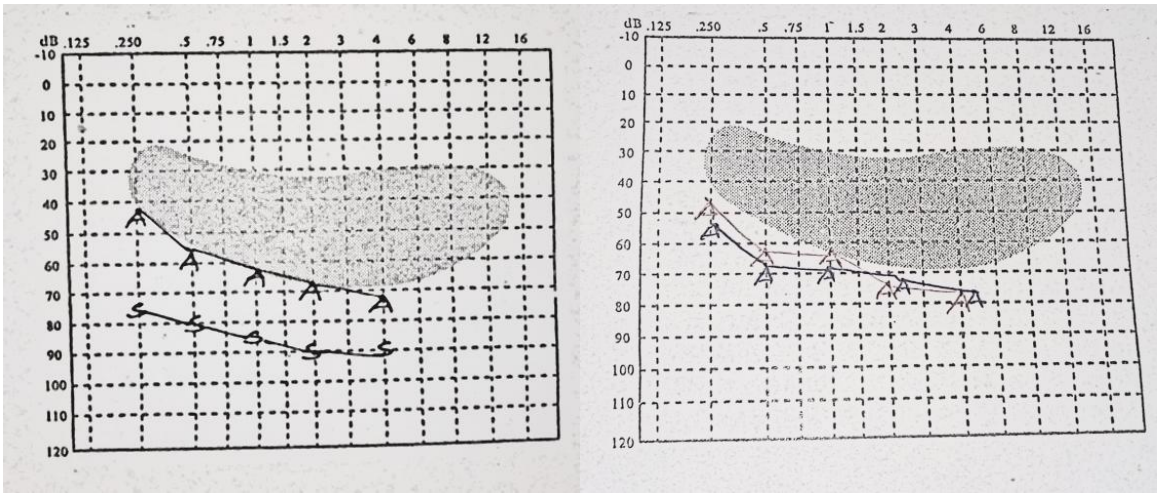
Both Ears: The test reported that in both the ears ABR peak V could not be obtained at 90 dBnHL using both click and tone burst stimulus at the rate of 11.1/secs in rarefaction, condensation and alternating polarities.

Impression:

Both Ears: Severe To Profound Hearing Loss

HEARING AID TRIAL:

	250	500	1k	2k	4k
Unaided	75	80	85	90	90
Aided Rt	50	65	65	75	80
Aided Lt	55	70	70	75	80
Both	45	60	65	70	75



The test revealed that the aided responses were improved and obtained at the lower border of the speech spectrum.

Impression: The Aided Responses Are At The Lower Border Of The Speech Spectrum.

RECOMMENDATIONS:

- Counselled regarding ABI
- Psychological evaluation
- Auditory Verbal Therapy
- Speech and Language Therapy
- ENT Review
- Follow up

For this child based on the radiological and audiological results ABI was the opted recommendation.

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Conflict of Interest: None

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