Content available at: https://www.ipinnovative.com/open-access-journals

IP Indian Journal of Anatomy and Surgery of Head, Neck and Brain

Journal homepage: https://www.ijashnb.org/

Case Report

# Audiological findings in Wilderwanck syndrome -A case report

**Balambigai N**[1], **Iyswarya Lakshmi**[1,*]

[1]*National Institute for Empowerment of Persons with Multiple Disabilities (Divyangjan)(NIEPMD), Muttukadu, Tamil Nadu, India*

ARTICLE INFO

*Article history:*
Received 04-10-2022
Accepted 11-10-2022
Available online 21-10-2022

*Keywords:*
Wilderwanck Syndrome
Mixed Hearing loss
Conductive hearing loss
Mondini's anamoly

ABSTRACT

Wilderwanck syndrome is a rare congenital disorder involving the neck, eyes and the hearing. It is characterised by a triad of symptoms, including Duane retraction syndrome, Klippel-Feil syndrome and Hearing loss. The original description of WS included sensori neural hearing loss. We describe a case of a 24-year-old female with Klippel-feil anamoly, Duane syndrome type 3 and mixed hearing loss, with unilateral congenital malformation of the inner ear.



## 1. Introduction

Wilderwanck syndrome (WS) is a rare congenital disorder. Symptoms of Wilderwanck syndrome include hearing loss, Klippel-Feil syndrome, and Duane retraction syndrome. The condition was first described by Wilderwanck in 1952,[1] and he further presented a series of 21 patients[2] with WS also known as Cervico-Oculo Acoustic syndrome. The occurrence of the condition is sporadic and is more frequently seen in females, hence increasing the possibility of X-linked inheritance, and a lethality in hemizygous males.[3] Individuals with WS exhibit a normal karyotyping.[4]

Klippel Feil anomaly is characterised by deformity of the cervical spine ie. fusion of cervical vertebrae resulting in short neck, Limited mobility of the neck, low hair line and spina bifida. Duane retraction syndrome also known as eye ball retraction syndrome was first described by Duane, in 1905.[5] The syndrome is characterised by lateral rectus palsy and retracted bulb resulting in limited adduction of the eye, palpebral fissure adduction during adduction and widening during abduction and strabismus.

Although Wilderwanck's definition[6] of syndrome included only sensori neural hearing loss, cases of conductive and mixed hearing loss have also been reported.[4,7,8] Similarly, though WS usually present with deformities of the inner ear, deformities of the middle ear and inner ear has also been reported.[9] WS can cause underdevelopment of membranous and bony labrynith thus resulting in underdeveloped cochlea and varying degree of inner ear abnormality.[10] On the other hand middle ear anomalies such as Ossicular chain discontinuity, fixation, congenital stapes fixation, and fusion has also been reported.[4,9]

This case report describes a subject with Wilderwanck syndrome with particular emphasis on the audiological findings.

## 2. Case Report

A 24-year-old girl born to non-consanguineous parents was referred to the department of Speech, Hearing and Communication with the complaint of reduced hearing in both ears. She has had a known hearing loss since childhood and has been using hearing aids bilaterally. And she is currently attending a vocational training program.

* *Corresponding author*.
*E-mail address*: ishuaashi315@gmail.com (I. Lakshmi).

https://doi.org/10.18231/j.ijashnb.2022.023

There was no family history of hearing loss. She was born of a full-term delivery and there were no prenatal and perinatal complications. Postnatally she presented with occipital encephalocele and meningitis at birth. She underwent surgery for excision of occipital encephalocele at the age of 3 months and as a complication she developed mild obstructive Hydrocephalus. Ventriculoperitoneal (VP) shunting was done and it was removed at 6 months of age. Developmental milestones including Speech and motor milestones were delayed. Karyotyping revealed a normal pattern (46, XX).

On ophthalmic evaluation, she exhibited limited abduction, Palpebral fissure widening on abduction and narrowing on adduction, left esotropia with the compensatory head movement to left. Based on the clinical features she was diagnosed as Duane syndrome type 3. She was recommended to use spectacles to correct the refractive error in both the eyes.

On physical examination, she presented with a short stature, short neck with limited movement of the neck, and low posterior hair line. Orthopaedic examination based on X-ray revealed multiple errors in segmentation of upper dorsal cervical spine, bilateral highly placed scapulae, kyphosis of dorsal spine and blocked vertebrae of cervical spine. And based on the findings she was diagnosed as Klippel feil syndrome.

Psychological evaluation using Binet-Kamat Test (BKT) showed her IQ was 8 years 8 months indicating a Mild Intellectual Disability. Based on the clinical triad of symptoms ie. occulo-cerviculo auricular symptoms, with normal karyotyping she was diagnosed as Wilderwanck syndrome by the Physician.

She had attended an inclusive school till $8^{th}$ grade and following that she attended a special school, and completed graduate education through Open University, which offered supported learning.

On oral examination the lip structures were normal at rest, and functions were adequate. There was no cleft of the lip or palate. She had Class III malocclusion with a wide arched hard palate. Tongue structure was normal but the range and rate of motion was restricted. She could communicate in phrases and sentences, though her speech intelligibility was limited. Language evaluation using Assessment of Language Development (ALD),[11] a formal language testing tool showed her receptive language age to be 6-7 years and expressive language age to be 4.6 years to 5 years. She exhibited several distortions and omissions in connected speech.

She reported of a history of recurrent middle ear infection during childhood. At the age of 9 years, she had an active ear discharge with central perforation, granular polypoidal middle ear mucosa, and was diagnosed as CSOM in right ear with Acute suppurative otitis media in the left ear. High resolution computerised tomography at the age of 10 years revealed mondini anomaly in left and mucosal thickening in both mastoid air cells with surrounding sclerosis with a Chronic osteomastoiditis on right and Chronic mastoiditis on left. She has had several episodes of ear discharge in both the ears till 12 years of age. At the age of 18 years during a routine follow up examination she was found to have a bilateral intact tympanic membrane.

A series of audiograms between 9 years to 12 years showed a moderate to moderately severe mixed hearing loss in right with severe mixed hearing loss in the left ear. Routine audiological evaluations performed annually till adulthood showed similar findings.

Currently she did not have any ear complaints except for hearing loss. ENT examination revealed a deviated nasal septum with bilateral retracted ear drum. There were no anomalies of the external ear. There were no reports of Vertigo and Tinnitus.

Tuning fork test showed a bilateral Rinne negative with Weber lateralizing towards the right ear. Her PTA averages were 53.3 dBHL in right ear, 85 dBHL in left ear with speech recognition threshold at 50 dBHL in the right ear, and 80 dBHL in the left ear; and WRS of 87.5% in the right ear and 75% in the left ear. Her tympanometric findings revealed bilateral“A” type tympanogram with low resonant frequency of 350Hz in right ear and 400Hz in left ear. Multi-component tympanometry findings showed a 3B1G pattern in both the ears. Bilateral ipsilateral and contra-lateral reflexes were absent. DPOAE were also absent in both the ears. VEMP couldn’t be done due to her occular and cervical limitations. And she was diagnosed as having moderate mixed hearing loss in right ear and severe mixed hearing loss in left ear. She was recommended to continue to use hearing aids.

## 3. Discussion

Wilderwanck syndrome is a rare developmental disorder affecting the hearing, eye movements and the bones in the neck. It is one of the congenital disorders associated with hearing loss. Though Wilderwanck’s original description included sensorineural hearing loss, cases of conductive and mixed hearing loss have also been reported.[12] The subject reported here presented with mixed hearing loss. The conductive component in the patient can be explained by Eustachian tube dysfunction during childhood, which in turn could have caused recurrent otitis media. Cases of eustachian tube dysfunction and recurrent otitis media have also been described earlier.[13] The low resonant frequency with a normal admittance and tympanometric width indicates an increase in mass and reduction in stiffness suggesting tympanosclerosis.[14] Long term middle ear infection, with perforation may have led to tympanosclerosis. In addition, she had normal ossicular chain, and no congenital anomalies of middle ear.Hence, it is also possible for patients to present with conductive

hearing loss with normal ossicular chain.[15] In addition, close to one –third of patients with Klippel feil anamoly present with either a conductive or a mixed hearing loss.[16]

Congenital anomalies of the inner ear in WS include small and underdeveloped cochlea and Mondini malformation and missing semicircular canals.[17] Our patient interestingly presented with unilateral monidini deformity. Similar to our report, West[9] had reported of a patient with unilateral mondini deformity with severe conductive hearing loss, and symmetrical hearing loss in both the ears. The findings can be explained by widely varying inner ear anamolies in WS, and the variability in terminology in Mondini anomaly. The original description of monidini included a triad of features including a normal basal turn and cystic cochlear apex, and a dilated vestibule and vestibular aqueduct with an enlarged endolymphatic duct. The case described here could have used the terminology to define a milder degree of the anomaly. It is also possible for the patient to have some residual hearing inspite of the mondini deformity, given the differences in severity.[18]

Hence, like typically developing school age children, children with syndromes are also prone to middle ear infections. It is important to identify them early, and provide prompt treatment, in order to avoid permanent hearing loss.

## 4. Conclusion

Individuals with Wilderwanck syndrome can present with unilateral congenital ear malformations. It is also possible for them to present with conductive and sensorineural hearing loss of varying degree. Hence it is important for the hearing loss to be identified at the earliest in order to prevent further impact on speech and language development and educational, social and emotional domains.

## 5. Source of Funding

None.

## 6. Conflict of Interest

The authors declare no conflict of interest.

## References

1. Wildervanck LS. A case of Klippel-Feil's syndrome with abducens paralysis; retraction of the eyeball and deaf-mutism. *Nederlands tijdschrift voor geneeskunde*. 1952;96(44):2752–8.
2. Wildervanck LS. A cervico-oculo-acoustic nerve syndrome. *Nederlands tijdschrift voor geneeskunde*. 1960;104:2600–5.
3. Abu-Amero KK, Kondkar AA, Alorainy IA, Khan AO, Al-Enazy LA, Oystreck DT. 3 microdeletion in a male with Wildervanck Syndrome. *Ophthalmic genetics*. 2014;35(1):18–24.
4. Eisemann MI, Sharma GK. The Wildervanck Syndrome: Cervico-Oculo-Acoustic dysplasia. *Otolaryngol Head Neck Surg*. 1979;87(6):892–7.
5. Duane A. Congenital deficiency of abduction, associated with impairment of adduction, retraction movements, contraction of the palpebral fissure and oblique movements of the eye. *Arch Ophthal*. 1905;114(10):1255–61.
6. Wildervanck LS, Bruyn GW, Myrianthopoulous NC. Congenital Malformations of the Spine and Spinal Cord. *Vinken PJ*. 1978;32(1):123–30.
7. Balcı S, Oguz KK, Fırat MM, Boduroglua K. Cervical diastematomyelia in cervico-oculo-acoustic Wildervanck syndrome: MRI findings. *Clin Dysmorphol*. 2002;11(2):125–33.
8. Cremers CW, Hoogland GA, Kuypers W. Hearing loss in the cervico-oculo-acoustic (Wildervanck) syndrome. *Arch Otolaryngol*. 1984;110(1):54–61.
9. West PD, Gholkar A, Ramsden RT. Wildervanck's syndrome–unilateral Mondini dysplasia identified by computed tomography. *J Laryngology Otol*. 1989;103(4):408–19.
10. Wildervanck LS, Hoeksema PE, Penning L. Radiological examination of the inner ear of deaf-mutes presenting the cervico-oculo-acusticus syndrome. *Acta oto-laryngologica*. 1966;61(5):445–53.
11. Lakkana S, Venkatesh K, Bhat JS, Karuppali S. Assessment of Language Development. A Manipal Manual 2nd ed. Manipal: Manipal Universal Press; 2021. Available from: https://www.amazon.in/Assessment-Language-Development-Manipal-Manual/dp/8195279791#detailBullets_feature_div.
12. Gupte G, Mahajan P, Shreenivas VK, Kher A, Bharucha BA. Wildervanck syndrome (cervico-oculo-acoustic syndrome). *J Postgrad Med*. 1992;38(4):180–2.
13. Jaffee IS. Congenital shoulder-neck-auditory anomalies. *Laryngoscope*. 1968;78(12):2119–39.
14. Katz J, Chasin M, Hood LJ, English K, Tillery KL. Handbook of Clinical Audiology 7th ed. Philadelphia, PA: Wolters Kluwer Health; 2015. p. 1–4.
15. Singh SP, Rock EH, Shulman A. Klippel-Feil syndrome with unexplained apparent conductive hearing loss. A case report. *Laryngoscope*. 1969;79(1):113–20.
16. Jarvis JF, Sellars SL. Klippel-Feil deformity associated with congenital conductive deafness. *J Laryngol Otol*. 1974;88(3):285–90.
17. Schuknecht HF. Mondini dysplasia A clinical and pathological study. *Rhinol Laryngol*. 1980;89(1):3–23.
18. Slack RW, Phelps PD. Familial mixed deafness with branchial arch defects (earpits-deafness syndrome). *Clinical Otolaryngol Allied Sci*. 1985;10(5):271–8.

## Author biography

**Balambigai N,** Assistant Professor https://orcid.org/0000-0002-8782-4422

**Iyswarya Lakshmi,** Undergraduate Student https://orcid.org/0000-0002-3982-3587

**Cite this article:** Balambigai N, Lakshmi I. Audiological findings in Wilderwanck syndrome -A case report. *IP Indian J Anat Surg Head, Neck Brain* 2022;8(3):98-100.