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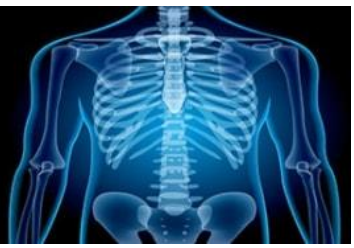

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Dr. Nikhil R Mahajan
Senior Resident, Department of Radiology, LTMMC & LTMGH, Sion, Mumbai, Maharashtra, India

Dr. Anagha R Joshi
Professor and Head, Department of Radiology, LTMMC & LTMGH, Sion, Mumbai, Maharashtra, India

Dr. Chinmayee Chitnis
Fellow, Department of Radiology, LTMMC & LTMGH, Sion, Mumbai, Maharashtra, India

Corresponding Author:
Dr. Nikhil R Mahajan
Senior Resident, Department of Radiology, LTMMC & LTMGH, Sion, Mumbai, Maharashtra, India

Wildervanck syndrome: A rare case of congenital hearing loss with cystic cochleovestibular malformation and cervical diastematomyelia

Nikhil R Mahajan, Anagha R Joshi and Chinmayee Chitnis

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Abstract
Wildervanck syndrome is a rare genetic disorder, infrequently seen in the general population. The disorder affects ocular movements, hearing, and is associated with cervical spine anomalies. Along with these clinical manifestations, the affected population have a characteristic appearance on radiological imaging which aids in the diagnosis of this syndrome.
We present clinico-radiological findings in a 4-year-old child with a classical triad of Wildervanck syndrome associated with cervical diastematomyelia.

Keywords: Klippel feil syndrome, magnetic resonance imaging, cervical spine, Wildervanck

Introduction
Wildervanck syndrome also called the "cervico-oculo-acoustic syndrome", is a rare congenital disorder characterized by a triad of congenital sensorineural deafness, Klippel Feil's syndrome, and Duane's retraction syndrome [1]. The pathognomonic features of this syndrome were first described by Wildervanck in 1960 [2].
Here we describe the case of a 4-year-old female patient with sensorineural hearing loss, abnormal ocular motility, and radiological findings suggestive of cervical vertebral fusion anomalies in association with cervical diastematomyelia.

Case
A 4-year-old female child, first by birth order, born out of a 3rd-degree consanguineous marriage, delivered by lower segment caesarean section because of severe oligohydramnios, was brought to our tertiary care centre for severely decreased response to sound since birth. The antenatal period progressed smoothly without any notable events and complications, while the immediate post-natal history was unremarkable. There was no positive family history of any genetic abnormality. She had delayed developmental milestones and no speech development. At the age of 4 months, Brain Evoked Response Audiometry (BERA) was performed, which revealed bilateral profound sensorineural hearing loss. The child was given a behind-the-ear hearing aid, however, no improvement in the hearing was noted. On thorough physical examination, the child had a height of 99 cm (between -1 to -2 standard deviation) and a weight of 13.3 kg (between -1 to -2 standard deviation). She also had a short and webbed neck with limitation of neck movements (Figure 1) and a low posterior hairline. Bilateral external auditory canals and tympanic membranes were normal. Cardiovascular, and respiratory systems examination was within normal limits. Abdominal ultrasound revealed no significant abnormality.
On ophthalmological evaluation, the child had limitation of abduction involving the right eye with widening of palpebral fissure on attempted abduction and globe retraction on adduction, suggestive of right sided Duane retraction syndrome.
High-resolution computed tomography of the temporal bone and computed tomography of the cervical spine was conducted on a Toshiba Aquilon Prime 160 slice CT machine with 0.5 mm thin sections for the temporal bone and 1 mm thin sections for the cervical spine.
The temporal bone computed tomography revealed cystic dilatation of bilateral cochlea and vestibules giving a 'figure of 8' appearance. There was an absence of turns of the cochlea and modiolus (Figure 2).

Bilateral lateral semicircular canals were short, while the rest of the semicircular canals were normal (Figure 3). Bilateral internal auditory canals were short with widened cochlear aperture (Figure 2). The middle ear with its ossicles and the external auditory canal were normal.

Cervical spine computed tomography showed various fusion anomalies involving cervical vertebral bodies and their posterior elements (Figure 4). Visualized upper dorsal vertebral bodies and their posterior elements also showed fusion anomalies. There was the fusion of shafts of right 3rd to 5th ribs and left 3rd-4th ribs (Figure 4). The foramen magnum was capacious (Figure 4). Focal cervical diastematomyelia was noted (Figure 5).

Multiplanar multi-echo MRI of the cochlea was performed on a Philips Ingenia 3T MRI machine with axial and sagittal highly T2-weighted (3D drive) sequences. MRI cochleography confirmed the above CT findings of cystic cochleo-vestibular malformation and cervical diastematomyelia (Figures 2 and 5). There was prominence of pre-pontine cisterns.

Written informed consent was obtained from the patient for publication of this case report and identifiable clinical images, as visualization of the eyes was essential for clinical relevance.

Figures

Fig 1: A frontal profile photograph of the patient demonstrates a short neck with a head tilt.

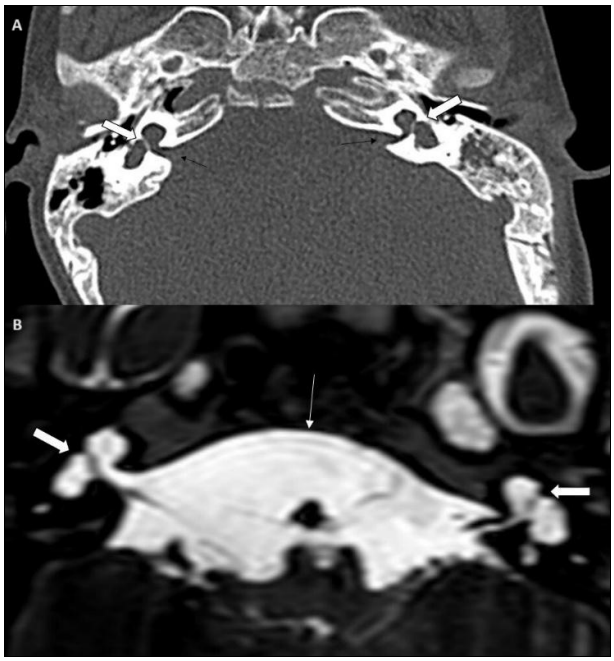

Fig 2: Image A is an axial computed tomography image of the temporal bone, and image B is an axial section of a highly T2-weighted (3D drive) sequence showing bilateral cystic dilatation of the cochlea and vestibule (marked by white block arrow) with absent turns of the cochlea and modiolus. A bilateral short internal auditory canal with a wide cochlear aperture is seen (marked by a black line arrow). Image B also shows the same findings with a widened prepontine cistern (marked by a white line arrow)

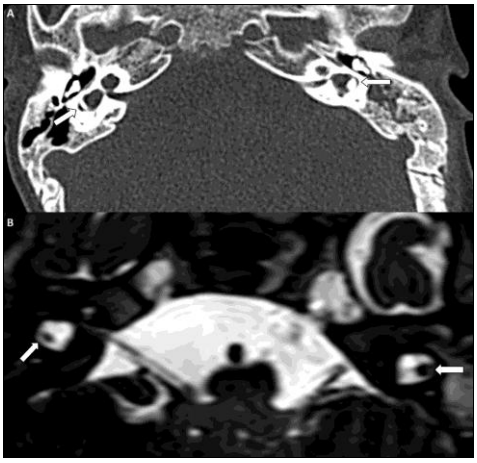

Fig 3: Image A is an axial computed tomography image of the temporal bone, and Image B is an axial section of a highly T2-weighted 3D drive sequence, which show short bilateral lateral semicircular canals (marked by white block arrow).

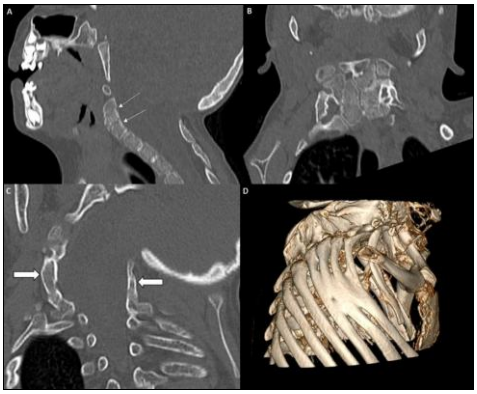

Fig 4: Image A is a sagittal MPR reconstruction of the CT cervical spine bone window, and Image B is a coronal MPR reconstruction CT cervical spine bone window showing fusion anomalies involving all cervical and upper dorsal vertebral bodies (marked by white line arrow). Image A also shows a capacious foramen magnum. Image C is a coronal MPR reconstruction CT cervical spine bone window showing fused bilateral cervical facet joints (marked by a white block arrow). Image D is a 3D CT volume rendering technique (VRT) sagittal view showing fused shafts of the 3rd to 6th ribs.

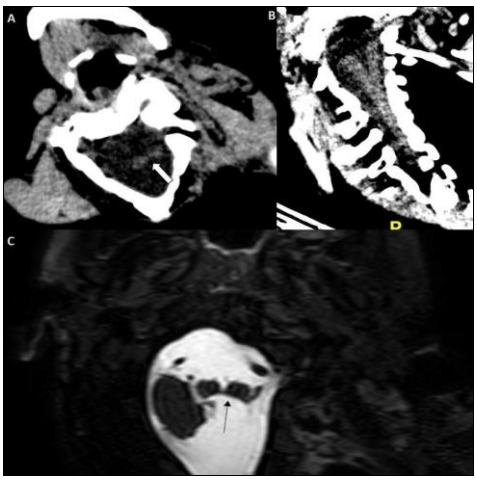

Fig 5: Image A is the axial section of the CT cervical spine and Image B is the coronal MPR reconstruction of the CT cervical spine showing splitting of the high cervical cord (marked by a white block arrow) suggestive of cervical diastematomyelia. Image C is the T2W axial section showing the splitting of the cervical cord (as marked by a black line arrow).

Discussion

The Wildervanck syndrome comprises a combination of fused cervical vertebrae (Klippel-Feil anomaly), congenital deafness, and palsy of the abducens nerve with retracted eyeballs (Duane syndrome) [3]. In 1952, Wildervanck reported a case with a combination of the above-described clinical features and collectively called these combinations of findings cervico-oculo-acoustic syndrome [2]. The male-to-female ratio of this syndrome is 1:10 [1]. The incidence of this syndrome is 1 in 40,000 cases [4]. The cause for female preponderance has been attributed to an X-linked dominant inheritance pattern associated with a high fatality rate among males in a hemizygous state [5]. Although over the recent years, no light has been thrown over specific etiology for Wildervanck syndrome, however various genes that are proposed to be associated include the FGF13 gene (Fibroblast growth factor 13) due to microdeletion of Xp26.3 region or GDF3 and GDF6 genes (Growth differentiation factor 3, 6) [5]. Due to resource limitations, genetic analysis was not feasible with our patient.

Klippel-Feil syndrome, which is a component of this syndrome, includes the congenital fusion of at least two cervical vertebrae along with the absence of intervertebral discs and occurs as a result of abnormal segmentation and fusion processes involving cervical somites [6]. Our case had fusion anomalies involving multiple cervical as well as dorsal vertebrae and rib anomalies in the form of fused ribs.

Duane retraction syndrome, another component of this syndrome, is an ocular movement disorder in which there is limited abduction and/or adduction of the affected eye. This occurs due to the absence of or partial development of the abducens nerve, causing aberrant innervation of the lateral rectus muscle [4]. Our case had absent abduction and globe retraction on adduction with narrow palpebral fissures.

Schild *et al*. were the first to show CT imaging of this syndrome and reported bilateral cystic cochlea, absence of modioli, deficiency of bony septum between the cochlea and the vestibule, and dilated vestibule [2]. Similar imaging findings of bilateral cystic cochleo-vestibular malformation (also called incomplete partition type 1) on CT/MRI, along with profound sensorineural hearing loss on BERA, were seen in our patient.

Diastematomyelia is an uncommon congenital anomaly characterized by incomplete or complete longitudinal splitting of the cord. The occurrence of cervical diastematomyelia is rare and is usually seen in association with craniovertebral junction anomalies, like Klippel-Feil syndrome and Chiari malformations [7]. In our case, there was a splitting of the high cervical cord with cervico-dorsal spine fusion anomalies, however, our patient did not exhibit any symptoms. Brodsky *et al* reported a case of Wildervanck syndrome with associated enlarged prepontine cistern, ectatic foramen magnum, and diffuse brainstem hypoplasia. Our case also demonstrated a widened prepontine cistern and a capacious foramen magnum. However, the brain stem appeared normal. In patients with a combination of Klippel–Feil anomaly and deafness, the closest differential to Wildervanck syndrome is Goldenhar syndrome, which shows an absence of Duane syndrome and is associated with lipodermoids, epibulbar dermoids, preauricular appendages, and aural fistulas [8]. In a resource-limited setting, radiological evaluation in correlation with clinical findings from otorhinolaryngologists, ophthalmologists, and neurologists is relied upon to make

the diagnosis. In all children with profound congenital deafness, a high-resolution CT scan of the temporal bone is necessary to evaluate the ear anomalies and select candidates for cochlear implantation. A detailed cardiac, musculoskeletal, otorhinolaryngeal, and ophthalmic evaluation is of utmost importance in the patient with Klippel-Feil anomaly to rule out Wildervanck syndrome.

The majority of patients with cochlea-vestibular malformation benefit from cochlear implantation [9]. However, the last resort in patients with profound hearing loss who fail to respond to cochlear implantation is the auditory brainstem implant. [10]. Our patient who had no improvement with the hearing aid was advised cochlear implantation, however the patient was subsequently lost to follow-up. Auditory brain stem implants are indicated in patients with dysplastic or absent cochleae or patients where the cochlea is unsuitable for implantation [10]. Patients not eligible for the above procedures to improve hearing are advised to continue using hearing aids and learn sign language or lip reading [5]. Duane retraction syndrome can be non-surgically treated with spectacles or lenses for refractive error and standard treatment of amblyopia. Corrective spectacles were prescribed to this patient for refractive error management. Early diagnosis followed by early intervention will help improve patients' quality of life.

Conclusion

Wildervanck syndrome is a rare congenital disorder marked by the triad of Klippel-Feil anomaly, Duane retraction syndrome, and sensorineural hearing loss. This case highlights the radiological and clinical complexity of the syndrome, further complicated by the rare finding of cervical diastematomyelia. High-resolution CT and MRI played a crucial role in diagnosis. Early recognition, multidisciplinary evaluation, and timely interventions like cochlear implantation and vision correction are essential for improving patient outcomes. This report underscores the value of detailed imaging and clinical correlation in diagnosing and managing complex congenital syndromes.

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Dr. Anagha R Joshi, Professor and Head, Department of Radiology, Lokmanya Tilak Municipal Medical College, Sion, Mumbai-22, India.

Conflict of interest

The authors have no conflict of interest.

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