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DOI: 10.2478/rjr-2023-0007

Romanian Journal of Rhinology, Volume 13, No. 49, January - March 2023

CASE REPORT

Wildervanck syndrome: Case report

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ABSTRACT

BACKGROUND. Wildervanck syndrome was described for the first time in 1952. This rare entity is composed by the oculo-cervico-acoustic triad, which may be more or less complete.

MATERIAL AND METHODS. We present a 5-year-old girl diagnosed with Wildervanck syndrome, exhibiting: Klippel-Feil anomaly (fused cervical vertebrae), Duane Stilling Turk syndrome (bilateral abducens palsy) and congenital bilateral cophosis indicated by ABR.

RESULTS. The CT scan revealed major malformations of the brain and calvarium including the petrous part of both temporal bones. Despite all these brain malformations, the psychological development of the child was nearly normal. As it was previously reported in other cases, the child also presented spina bifida cervicalis and cleft palate. The cause of deafness was bilateral lack of cochleae, making impossible the insertion of the cochlear electrode for hearing rehabilitation. When the patients are discovered late (after 3 years of age), the management of hearing loss is by being taught sign language and lip reading.

CONCLUSION. Any cranio-facial malformation and any delay in speech production impose early attention of the otorhinolaryngologist or audiologist for timely diagnosis and rehabilitation of hearing loss. The imaging examination, especially the high-resolution temporal bone CT-scan, is of paramount importance to be performed in all profound congenital deaf children to evaluate the candidacy to cochlear implantation.

KEYWORDS: deafness, inner ear malformation, Wildervanck syndrome.

INTRODUCTION

In 1952, Wildervanck described a new nosologic entity that comprises multiple congenital anomalies, including: fused cervical vertebrae, abducens paralysis and deafness. He called this triad cervico–oculo–acoustic syndrome. The incidence rate of this disease was not yet been reported, but most of the investigators agree that the morbidity is low, the syndrome being seen seldom in the general population[1]. In the US, this disease is estimated to be fewer than 1000 cases[2]. Some other authors have reported even a lower prevalence, with fewer than 100 cases in US since 1952[3]. The range of people who may have this disease is an estimate and it is based on the available incidence/prevalence data for this disease and the population in the USA.

There is not yet any genetic test to confirm the Wildervanck syndrome. The diagnosis is made based on the clinical findings of neurologists, ophthalmologists, otorhinolaryngologists and from radiological evaluation. Within that process, the audiologist helps to determine the presence, magnitude and nature of hearing loss in children with cervico-oculo-acoustic syndrome.

In addition, the presence of other specialists, in the field of cardiology[4] or nephrology[5] is necessary, since the different reports associate this syndrome with heart or kidney malformations.

The present report brings attention over the severe craniocerebral malformations that include inner ear agenesis, which makes hearing rehabilitation very difficult.

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Received for publication: December 7, 2022 / **Accepted:** January 10, 2023

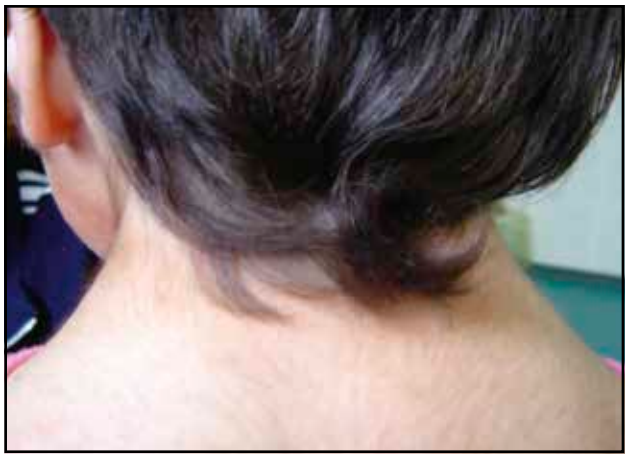

Figure 1. Low hairline in the posterior neck.

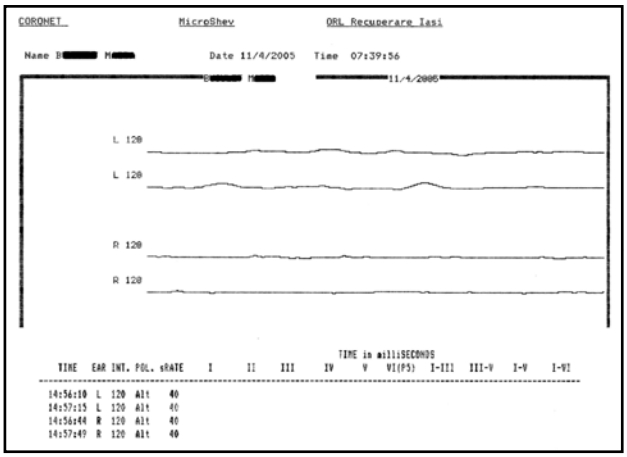

Figure 2. ABR – No response on 120 dB SPL with click stimulus – bilateral.

MATERIAL AND METHODS

The aim of this paper is to present a rare case of a 5-year-old girl diagnosed with Wildervanck syndrome by a clinical geneticist from the Paediatric Hospital of the "Grigore T. Popa" University of Medicine and Pharmacy, Iasi, and referred to our ENT Clinic for hearing assessment.

The evaluation of hearing thresholds was conducted in the most appropriate manner for the child's developmental level. The investigations started with behavioural testing procedures that are typically used with older children, but the lack of responses determined us to choose electrophysiological methods to identify the hearing thresholds. The tests were done with the child awake, without sedation. For identifying thresholds, the auditory brainstem responses (ABR) were recorded. Once the hearing loss was diagnosed, a radiological evaluation of the inner ear and the cochleovestibular nerve was considered: a cranio-cerebral CT scan was performed to establish the appropriate treatment.

Ophthalmologic examination, psychological evaluation and X-rays of the neck were also performed.

RESULTS AND DISCUSSIONS

The child was admitted in our clinic for a delay in speech and language acquisition. Family history of deafness or other anomalies was negative. From the child's history, we note an Apgar score of six and surgery for cleft palate.

Wildervanck syndrome is typically characterized by three primary findings (triad). These include: abnormal union or fusion of two or more bones of the spinal column (vertebrae) within the neck (Klippel-Feil syndrome); impairment or absence of certain eye (ocular) movements (Duane syndrome); hearing impairment that is present at birth. However, according to some reports, this triad may be incomplete in some individuals with Wildervanck syndrome. In addition, in some cases, additional physical findings or intellectual disability may be present[2].

The cause is presumed to be genetic, although, so far, no genetic anomaly was found. X-linked dominant inheritance has been suggested due to the high prevalence of affected females[2].

In our patient, the characteristic triad was entirely present.

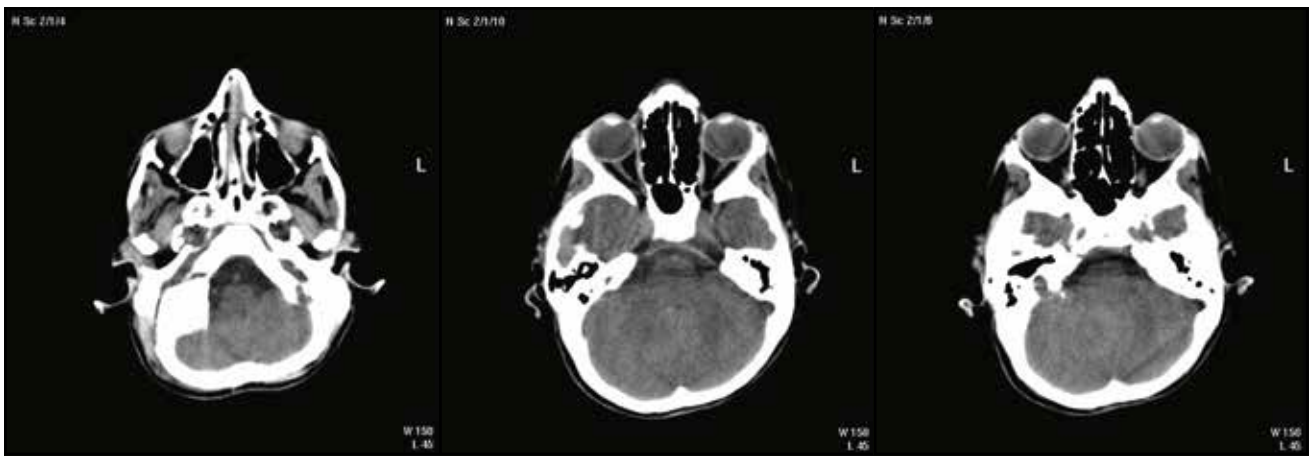

Figure 3. Temporal bone CT, axial slices: cochleae and internal auditory meatus could not be identified (absence of cochleae and internal auditory meatus).

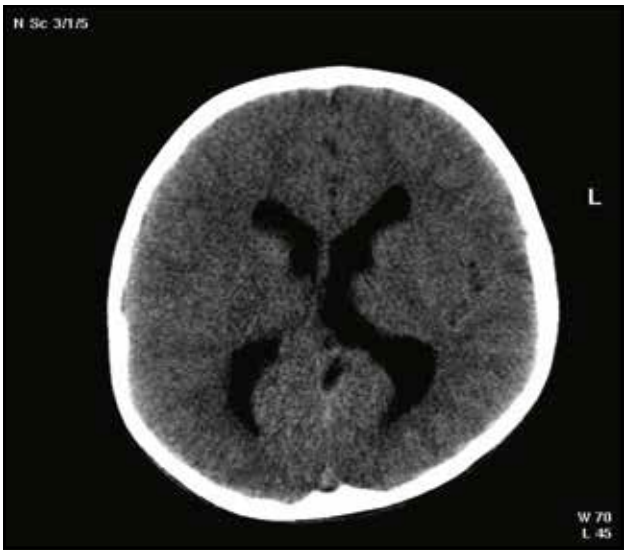

Figure 4. Hydrocephalus of the lateral ventricles which are malformed, possibly a supernumerary ventricle.

Clinical examination revealed low implantation of the external ears and of the hairline at the neck (Figure 1) and a short neck. The child had obvious strabismus confirmed by the ophthalmologist.

The psychological examination was required mainly because of the lack of any answers to the behavioural testing procedures regularly used with children of this age. The conclusion of the psychological examination was that the child was normally developed for her age. Bearing in mind that 2/3 of the patients with Wildervanck syndrome might exhibit deafness[6], we decided to find the hearing threshold using electrophysiological measurements – ABR – which, in this case, seemed to offer the best opportunity to diagnose the hearing loss. In Wildervanck syndrome, the hearing loss may be either sensorineural, conductive, or mixed, uni- or bilateral, and of different degree of gravity. In our patient, deafness was bilateral and complete on ABR (Figure 2). This result explains why the child had no response to behavioural tests.

The next step was to perform a temporal bone CT-scan. The purpose of this exam was to determine the morphology of the inner ears and of the internal auditory meatus. The CT-scan revealed an asymmetry of the cerebellar fossa with areas of the right occipital bone agenesis, and a complex malformation of the petrous part of the temporal bone with unidentifiable internal auditory meatus, and with aplasia of both cochleae (Figure 3). The CT-scan exam indicated also that the patient presented internal hydrocephalus of the lateral ventricles, which appeared malformed and, possibly, a supernumerary ventricle (Figure 4).

Once the CT scan and MRI explorations became available in many services and necessary for different reasons; more authors described in the patients with Wildervanck syndrome severe malformations of the inner ear and of the internal auditory meatus[1,7], major cerebral anomalies[8] like: vermian hypoplasia, tonsillar herniatum, triventricular hydrocephalus[9], abnormal cerebellum[10], brainstem hypoplasia[8], extensive malformation of the cochlea and vestibular parts of the inner ear[11], aplasia of the inner ear[12], Mondini dysplasia[13], absence of the vestibulo-cochlear nerve[11].

At X-rays of the cervical spine, we found not only fused cervical vertebrae, but also the description of spina bifida cervicalis (Figure 5), which was also reported by some authors in patients with Wildervanck syndrome[9,14].

Despite the presence of cerebral malformation and of congenital bilateral cophosis, the child was normally psychologically and cognitively developed.

This is an extremely rare case when both the inner ear and internal auditory meatus are malformed. In such cases, a good imaging evaluation of the inner ear and internal auditory meatus are important in planning the appropriate ther-

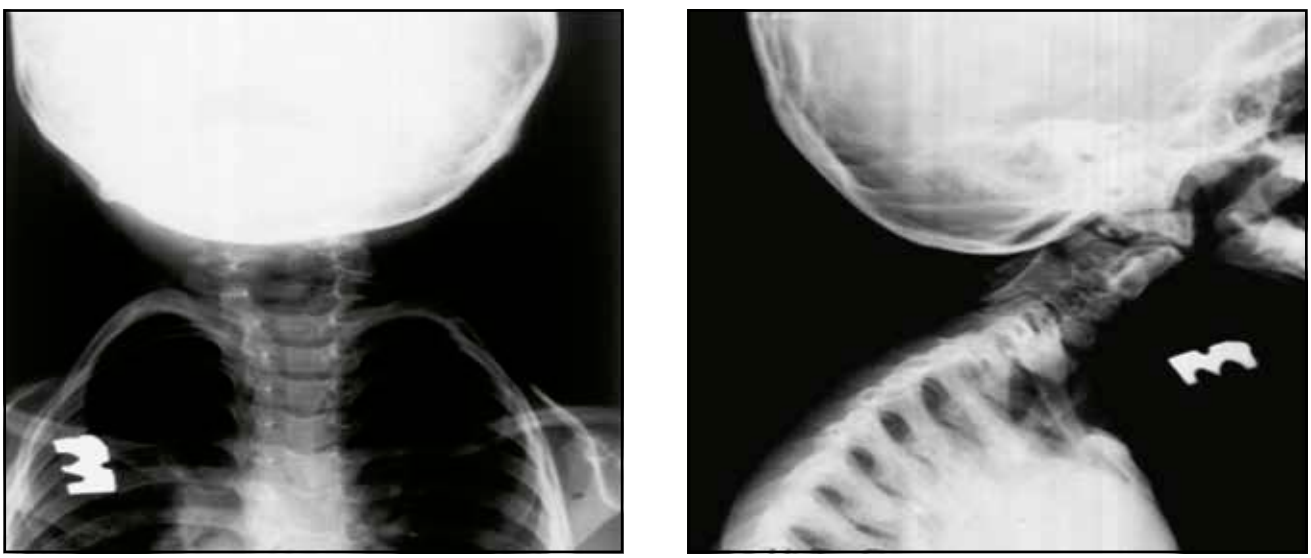

Figure 5. X-ray of the cervical spine revealed fused neck vertebrae and spina bifida cervicalis (face and profile).

apy for hearing rehabilitation. Due to the bilateral lack of cochlea, cochlear implant surgery and hearing rehabilitation is impossible. The possibility of brainstem stimulation could be discussed, but several factors must be considered: age of the patient at the diagnosis, surgical anatomy, surgical risks, minimal hearing benefit/quality of life and not at least the motivation of the family. In this case, the treatment possibilities are very limited, the most important limiting factor being the age of the child. She was already five years old at the diagnosis. A brainstem implant would have been a solution if the patient would have been addressed earlier, but, at this age, the expected benefit from a brainstem implant is insignificant in relation to the risks[15]. The only recommendation for the patients in similar situation, and as well for our patient, is to learn lip-reading and sign language, to follow the special school for deaf and to fit in the Deaf Society.

CONCLUSIONS

Any cranio-facial malformation and any delay in speech production impose early attention of the otorhinolaryngologist for timely diagnosis and treatment of hearing loss. In cases of deaf children older than 3 years, where cochlear implantation is not possible due to inner ear or eighth nerve absence, hearing rehabilitation is not possible with the technology available and has to do with the development, plasticity and maturation of the auditory pathway[13]. The wisest way to manage these cases is to recommend learning lip reading and sign language and attending a special school for the deaf[15]. Genetic counselling of the families of affected people should be considered.

Conflict of interest: The authors have no conflict of interest.

Contribution of authors: All the authors have equally contributed to this work.

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