Case Report

Wildervanck syndrome with hypoplastic frontal sinus: A rare case presentation

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We report a case of Wildervanck syndrome exhibiting Klippel–Feil anomaly, Duane's retraction syndrome and congenital deafness. Since the first case was reported in 1952, there have been more reports describing this triad either complete or incomplete. Our case has a complete triad of the syndrome along with frontal sinus hypoplasia. Our case is unique as the triad was associated with frontal sinus hypoplasia, which is very rare association.

Key words: Duane's syndrome, frontal sinus hypoplasia, Klippel Klippel–Feil deformity, Wildervanck syndrome

Introduction

In 1952, Wildervanck, described a cervico-oculo-acoustic syndrome consisting of Klippel–Feil deformity, abducens palsy with globe retraction (Duane's retraction), and congenital hearing loss.[1] Etiology is unknown. Majority of individuals have been females. Cases with complete as well incomplete triad have been described in literature. This report describes a case with hypoplastic frontal sinus along with triad of Wildervanck syndrome. This presentation has been rarely described in literature.

Case Report

The patient is a 9-year-old female child admitted to AVBR Hospital for deformity of an ear, short and deviated neck since birth. She was born of nonconsanguineous marriage. Antenatal history was uneventful. There was no history of any drugs or radiological exposure. Birth history was also uneventful without any complications during intrapartum as well postpartum period. Developmentally, she was normal. Anthropometric parameters were suggestive of short stature (height - 119 cm; <3rd percentile). On examination, her vitals were stable. Right ear was malformed without visible vestibular canal. Left ear was having discharge. The neck was short with deviation to right side [Figure 1]. Furthermore, there was kyphoscoliosis of the thoracic vertebrae [Figure 2]. Her right eye was smaller than the left eye with an intermittent involuntary decrease in size. The systemic examinations showed abducens and right facial nerve palsy. Ophthalmic evaluation was done for small size of the right eye. It was suggestive of right eyeball retraction.

All above findings were suggestive of Wildervanck syndrome. Child was investigated for various deformities. Her blood reports were normal. Audiometry revealed profound hearing loss on right side and moderate sensorineural hearing loss on the left side. Brainstem evoked response audiometry was also suggestive of sensorineural hearing loss on the left side. Computed tomography (CT) spine was showing of fusion of the thoracic vertebrae, suggestive of Klippel–Feil deformity. CT paranasal sinuses were suggestive of the hypoplastic frontal sinus [Figure 3]. CT brain was suggestive of right holesteatoma [Figure 4].This was rare association with Wildervanck syndrome. The patient was planned for surgery for cholesteatoma. As she was also having short neck, anesthetist gave high-risk fitness. Hence,

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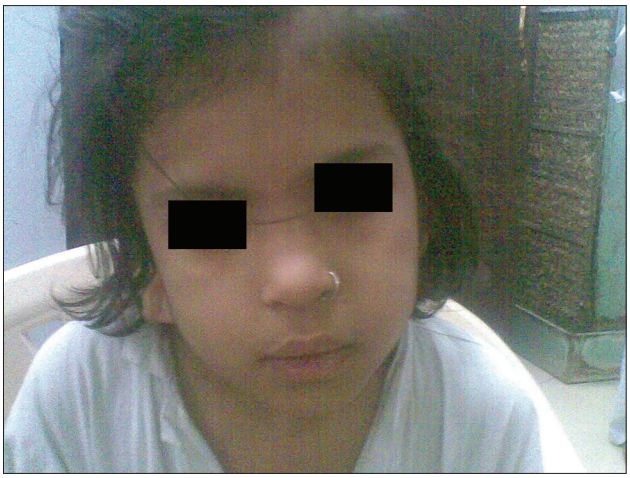

Figure 1: Left abducens nerve palsy with short neck and malformed right ear

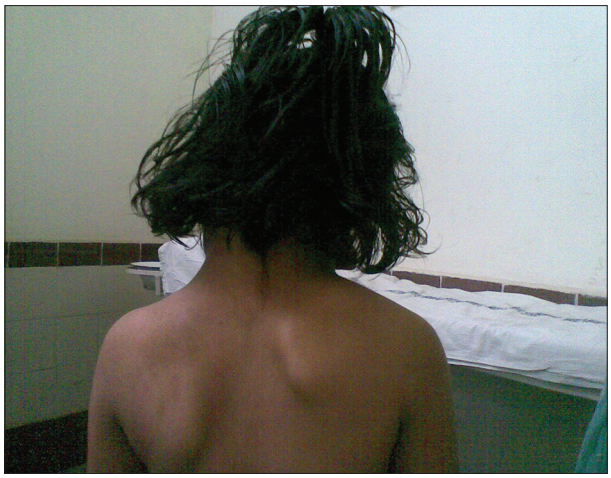

Figure 2: Klippel–Feil deformity

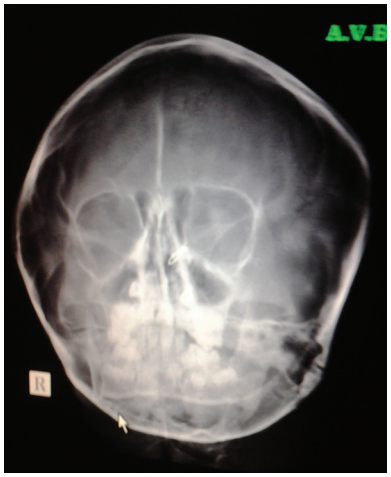

Figure 3: Right frontal sinus hypoplasia

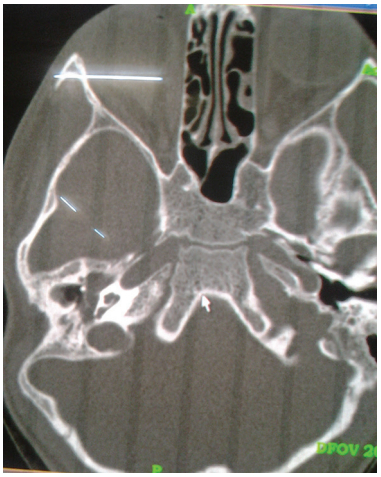

Figure 4: Cholesteatoma

patient's relatives refused to opt for surgery; hence, child was referred to higher center with better expertise management for surgery.

Discussion

Wildervanck syndrome comprises of the triad of Klippel–Feil deformity (fusion of ≥ 1 cervical vertebra), Duane retraction syndrome, and hearing loss.[2] The neck is short, thick, webbed, and immobile.[2] The head seems to sit directly on the trunk. Other spinal deformities (spina bifida occulta, sprengel deformity, and hemivertebrae, fusion of the ribs, absent ribs, kyphosis, scoliosis, and basilar impression) may coexist. Hearing loss in patients with the Wildervanck syndrome may be sensorineural, conductive, or mixed and may be accompanied by malformations of the external ear, external acoustic meatus, ossicles, and bony labyrinth.[2] Our case also had a malformed right ear. Intelligence may be mildly to severely reduced.[3]

There is a consensus about the mode of inheritance of Wildervanck syndrome, but all agree that genetic factors are involved. Autosomal dominant inheritance with incomplete penetrance and variable expressivity has been suggested.[4,5] Further, the gene would be partly sex limited acting on a polygenic background, which is modified by sex, rendering females more susceptible than males to action of the gene.[6] An environmental etiology, due to a vascular disruption sequence during embryonic development has been noted in Klippel–Feil anomalies as in Moebius and Poland sequences. A combination of defects (Kiippel–Feil and Moebius) could induce the more complex phenotype observed in Wildervanck syndrome.[7]

Wildervanck stated that deafness should be sensorineural in type; cases with conductive or mixed losses have also been reported.[8,9] Only one-third of the patients with Wildervanck have been described as having hearing loss; although, audiometry in our patient revealed a moderate degree of sensorineural deafness.

Conclusion

Wildervanck syndrome with frontal sinus hypoplasia and cholesteatoma is very rare association, which has not yet been described in literature. Furthermore, surgical intervention is necessary for the drainage of cholesteatoma. An expert team work is required for the surgery as intubation may be difficult because of Klippel–Feil deformity. The possibility of Wildervanck should be kept in mind, while evaluating a case of Klippel–Feil deformity.

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