

SEVERE SCOLIOSIS, TORTICOLLIS AND SHORT STATURE IN A WOMAN WITH WILDERVANCK SYNDROME (WS)

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Abstract

Wildervanck syndrome (WS) combines features of Klippel-Feil syndrome (KFS), sixth nerve palsy, and deafness. This is a case of a 23 year old woman, diagnosed with KFS (a triad of short neck, low posterior hairline and restricted neck movements) at the age of 20 days. The manifestations of the WS in this patient are severe: she has torticollis, and an extremely severe scoliosis. In addition, she is short (-3 SD; parental target height + 0.8SD) and has mixed sensorineural and conductive deafness. She also has ptosis, strabismus and a high myopia. Radiologically, there are multiple coalitions of cervical vertebrae. Intelligence is unaffected (IQ 95), but deafness, strabismus and high myopia forced her early out of school. Karyotype is 46, XX. In brief, this is a patient with severe WS and additional anomalies. Short and/or reduced parental target height is a part of WS.

Key words: Wildervanck syndrome, short stature, Duane retraction syndrome, Klippel Feil anomaly, deafness, torticollis, severe scoliosis.

Introduction

Wildervanck syndrome (WS) comprises Klippel Feil's (KF) anomaly (congenitally fused cervical vertebrae), deafness and Duane's retraction syndrome (deficient abduction with retraction on adduction) [1]. Reports described this triad, either completely or incompletely.

KFS is a developmental disorder presenting with congenital fusion of at least 2 cervical vertebrae. KFS can be associated with congenital anomalies of the cardiovascular, genitourinary tract, of the auditory system and the spinal cord.

Duane retraction syndrome is a congenital eye movement disorder characterized by a failure of cranial nerve VI (the abducens nerve) to develop normally. This results in restriction or absence of abduction, adduction, or both [2, 3]. The palpebral fissures are narrow; the globe is retracted on attempted adduction.

This is a description of a particularly severe form of a WV in a woman followed from newborn period to her 23rd year of life.

Case presentation

This 23 year old woman was diagnosed with KFS at the age of 20 days. She was the third child of healthy parents, born after an uneventful pregnancy of 40 weeks, with normal birth weight and birth length. Nevertheless, her father's side has an increased incidence of mental retardation and various disabilities. Not all data concerning those defects could be obtained, but mental retardation was found in at least 3 members of the family. The girl has a particularly severe form of KFS: torticollis, extremely pronounced scoliosis, short stature (-3SD, target height was +0.8SD [4] (fig. 1, 2). Audiogram revealed a mixed sensorineural and conductive deafness.



Figure 1 – The back: extreme scoliosis



Figure 2 – X-ray of the back, scoliosis

Ocular examination showed normal anterior and posterior segment findings. Ptosis, strabismus (6th nerve palsy) and high myopia further limit her life quality. An additional limitation of abduction in both eyes with widening of palpebral fissure on attempted abduction and globe retraction on adduction was also noted.

Cognitive development was normal but she was not able to attend school due to deaf-

ness and high myopia. MPS IV was excluded by enzyme analysis. Cardiovascular and respiratory systems' examination was within normal limits. Ultrasound of the kidneys and the internal genitals is uneventful. Karyotype is normal female, 46, XX.

Vertebral fusions were assessed via spinal radiography and MRI, which revealed multiple coalitions of cervical vertebrae. Fusion of C2-Th1 was found.

Discussion

WS is rarely associated with short stature [5–7]. We would further support this observation by another patient with WS and short stature. In addition, we believe (not all publications state the target height) that even the patients without short stature have a reduced predicted adult height (PAH), e.g. they fail to attain the target (genetic, parental) height as a consequence to their vertebral shortening.

WS is a rare condition, with less than 50 patients reported [7]. Several modes of transmission have been proposed: sex-linked dominance with lethality in the hemizygous male, multifactorial inheritance, polygenic inheritance with limitation to females [1, 8, 9].

One-third of the patients with WS have been described as having hearing loss, but WS may be responsible for ~1% of deafness among females. The deafness in WS is perceptive [3, 9–11], sensorineural or both, but only one-third of the patients with WS have hearing loss. It is of note that our patient has the rarely observed mixed, sensorineural deafness.

Most patients have unilateral Duane retraction syndrome (congenital eye movement disorder characterized by a failure of the abducens nerve to develop normally) [9]. It is of note that delayed diagnosis in children can lead to a permanent uncorrectable loss of vision.

Occasionally other congenital anomalies have been described in WS: mental retardation, facial asymmetry, cleft palate [5, 7], absent-deformed auricles, severe inner ear anomalies, cardiac anomalies, brainstem hypoplasia, Dandy Walker syndrome, vertebral artery dissection.

Recently Abu-Amarpo et al. (2014) detected an approximately 3-kb deletion in chromosome Xq26.3. Further genetic investigations are needed to precise the genetic defect(s) in WV.

Conclusions

So far, it remains a matter of controversy whether WV is a discrete entity, or if it is just one part of the spectrum of congenital spinal deformities. This patient with severe WS and additional anomalies needed a comprehensive evaluation in the search for additional accompanying anomalies. Team approach and early treatment of skeletal and extrasketelal problems in KFS patients is needed. A reduction of adult height seems to be a regular feature, short stature an occasional one. In addition, we believe (not all publications state the target height) that even the patients without short stature have a parental target height reduced (predicted adult height), e.g. they fail to attain the target (genetic, parental) height as a consequence to their vertebral shortening.

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Резиме

ИЗРАЗЕНА СКОЛИОЗА, ТОРТИКОЛИС И НИЗОК РАСТ КАЈ ЖЕНА СО WILDERVANCK SYNDROME (СИНДРОМ НА ВИЛДЕРВАНК)

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Wildervanck syndrome (WS) ги комбинира карактеристиките на Klippel-Feil syndrome (KFS), здружени со парализа на шестиот кранијален нерв и глувост. Станува збор за 23-годишна жена кај која на возраст од 20 дена била поставена дијагноза на синдромот на KFS (тријада на кус врат, ниска задна линија на косата и ограничени движења на вратот). Манифестациите на WS кај оваа пациентка се тешки: тортиколис и екстремно тешка сколиоза. Исто така, таа има низок раст (-3СД, при што родителската целна висина е +0.85 СД. Нејзината глувост е од мешан сензориневрален и кондуктивен тип. Забележана е и птоза, страбизам и висока миопија. Рендгенолошкиот наод се мултипли коалиции на цервикалниот рбет. Интелигенцијата е нормална (IQ 95), но глувоста, страбизмот и високата миопија ја натерале да прекине со школување. Кариотипот е 46, XX. Накусо, станува збор за пациентка со тешка форма на WS и додатни аномалии. Низок раст, помал од родителската целна висина, е дел на синдромот.

Клучни зборови: Wildervanck syndrome, низок раст, Duane ретракионен синдром, Klippel Feil аномија, глувост, тортиколис, тешка сколиоза.