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Vestibular Dysfunction in Turner Syndrome: A Case Report

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Abstract

Objective—Turner syndrome is a well-known cause of sensorineural hearing loss, and the lack of estrogen has been implicated in cochlear dysfunction. It has never been associated with vestibular dysfunction. We report a case of a patient with Turner syndrome who was found to have bilateral vestibular dysfunction based on video-oculography (VOG) testing.

Patient—A single patient with a history of Turner syndrome who was found to have significant bilateral vestibular dysfunction.

Intervention—After noticing a deficit in the vestibulo-ocular reflexes on qualitative horizontal head impulse examination, the patient underwent VOG testing.

Main Outcome Measure—VOG testing quantitatively measures angular vestibulo-ocular reflex (AVOR) gain in the horizontal semicircular canal plane. AVOR gain represents the eye movement response to a head movement; in normal individuals the eye movement is fully compensatory and gain values are close to unity.

Results—VOG results showed AVOR gains of 0.29 and 0.36 on the right and left sides, respectively.

Conclusion—We have presented a case of a woman with Turner syndrome with asymptomatic vestibular dysfunction demonstrated with VOG testing. Although there is a documented relationship between Turner syndrome and sensorineural hearing loss, there are no previous studies or case reports linking Turner syndrome and vestibular dysfunction. Additional research and added vigilance in monitoring Turner syndrome patients may be warranted.

Keywords

Turner syndrome; Vestibular dysfunction; Video head impulse testing

Turner syndrome has long been associated with both conductive and sensorineural hearing loss (1,2), but it has not previously been reported to have associations with vertigo or vestibular dysfunction. In 1 cohort of Swedish women with Turner syndrome, only 13% of women age 40 and older had normal hearing thresholds (3). Although the conductive hearing loss has been attributed to recurrent otitis media in childhood, the sensorineural

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hearing loss is imperfectly understood, with both cochlear and central processing components hypothesized to play a role. Estrogen has been implicated as integral to both and its presence is considered hearing protective (1,4). Women typically have shorter auditory brainstem responses than men and significantly less hearing loss than men of the same age, and variation in occupational exposures fail to explain these differences (1). Additionally, human fetal and adult inner ear specimens stain for estrogen receptor α and β (ER α and ER β) (1). Turner syndrome, characterized by gonadal dysgenesis and the absence of estrogen, may lead to both earlier presbycusis and longer auditory brainstem responses (5). Despite the documented association between hearing loss and Turner syndrome, to our knowledge, an association between Turner syndrome and vestibular dysfunction has never been reported. We present a case of a woman with Turner syndrome who has vestibular dysfunction ascertained by video-oculography (VOG) recordings.

CASE REPORT

SL is a 46-year-old Korean woman with a history of 45,X/46,XX mosaic Turner syndrome who presents for a history of bilateral tinnitus worse in her right ear that started about a month and a half ago. She is a professional musician and notes that the tinnitus seems to be progressing. She denied any symptoms of vertigo or dysequilibrium. An audiogram demonstrated normal hearing sensitivities bilaterally with speech discrimination scores of 100% bilaterally. She was first diagnosed with Turner syndrome at age 18 after presenting with primary amenorrhea. A horseshoe kidney seen on intravenous pyelogram was another notable complication of her Turner syndrome.

On physical exam, SL had a short stature and broad neck characteristic of Turner syndrome. Otoscopy and tuning fork testing were unremarkable. During clinical horizontal head impulse testing, she was noted to have gaze dysmetria bilaterally (i.e., evidence of refixation saccades) that was more significant on head rotation to the right. Video head impulse testing using the EyeSeeCam VOG system (Munich, Germany) was performed to further quantify the patient's horizontal semicircular canal function (6). VOG results showed an eye to head velocity ratio, or mean angular vestibulo-ocular reflex (AVOR) gain, of 0.29 and 0.36 in the right and left horizontal semicircular canal planes, respectively (Fig. 1A). A healthy control's AVOR gain is typically equal to 1 (Fig. 1B).

The patient was counseled that the tinnitus may reflect subclinical cochlear dysfunction, and the association between sensorineural hearing loss and Turner syndrome was reviewed with the patient. Hearing protection and serial audiometry were recommended. Her physiologic evidence of vestibular dysfunction was also reviewed with her, and potential clinical sequelae including increasing gaze instability with head movement (oscillopsia) and the potential benefits of balance-enhancing activities such as Tai Chi were discussed.

DISCUSSION

This patient clearly demonstrated impaired vestibuloocular reflexes in the plane of the horizontal semicircular canal quantified by VOG. The patient had not appreciated any vestibular symptoms during daily life but did notice her field of vision slip as her head was

passively rotated rapidly during the physical exam. She likely had compensated well to this point using visual cues and proprioception to assist dysfunctional vestibular reflexes. The degree to which her vestibular dysfunction is caused by or even associated with Turner syndrome is unclear. Although there is literature related to Turner syndrome and deficits in cochlear function and central auditory processing, to our knowledge, there are no previous studies related to Turner syndrome and vestibular dysfunction.

Turner syndrome is characterized by gonadal dysgenesis, primary amenorrhea, and short stature. Congenital features include webbed neck, nail dysplasia, high palate, and short fourth metacarpal, whereas in adulthood, hearing loss, hypothyroidism and abnormal liver function tests become more prevalent. More than half of those with Turner syndrome, including our patient, experience a mosaic form in which they have a chromosomal complement of 45,X and 46,XX cells. Ear and hearing dysfunction are common in Turner syndrome, and recurrent otitis media is often the first presenting complication of Turner syndrome in young girls (7). The lack of estrogen characteristic of Turner syndrome is believed to have a role in the increased rates of sensorineural hearing loss (3).

The evidence that estrogen, or its absence, may play a role in inner ear dysfunction comes from immunohistochemical staining for estrogen receptors in mice and humans. In mice, the utricle stained for both ER α and the utricle and the ampulla stained for ER β (1). A “Turner mouse” model has been developed that showed higher rates of otitis media and hearing loss than controls but similar patterns of ER α and ER β expression (5). In humans, these receptors have also been identified within the cochlea, but no studies have investigated the presence of these receptors within the human vestibule. Additionally, the impact of activating and antagonizing these estrogen receptors in the cochlea and vestibule on hearing or balance has not been well characterized.

Further supporting a role of estrogen in modulating inner ear function is the observation that some women with Ménière’s disease experience a worsening of symptoms during the premenstrual phase when estrogen levels are high (8). Estrogen’s role in extravasation of intravascular fluid to extravascular space has been implicated. Although distinct processes, Turner syndrome related sensorineural hearing loss and premenstrual worsening of Ménière’s disease may indicate that there is a healthy range of estrogen that optimizes inner ear function. Further research will be required to elucidate the role of estrogen in vestibular function.

CONCLUSION

We have presented a case of a woman with Turner syndrome with asymptomatic vestibular dysfunction demonstrated with VOG testing, which provided a quantitative measure of horizontal AVOR gain. Although there is a documented relationship between Turner syndrome and sensorineural hearing loss, no previous studies or case reports could be found linking Turner syndrome and vestibular dysfunction. The biological mechanism may involve the loss of inner ear protection provided by the presence of estrogen. Mouse and human cochlea and mouse vestibular apparatus have stained positive for estrogen receptors. Further research to investigate the impact of estrogen on the vestibule is warranted, especially in the

context of Turner syndrome. Added vigilance by providers in testing for vestibular dysfunction in women with Turner syndrome is also reasonable.

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FIG. 1.

Video-oculography recordings for our patient with Turner syndrome (*A*) and a subject with normal vestibular function (*B*). Head movement velocities are recorded in red, eye movement velocities in blue. Mean horizontal AVOR gains for the Turner syndrome patient were 0.29 and 0.36 on the right and left sides, respectively (*A*), compared with mean AVOR gains of 0.99 and 1.00 on the right and left sides, respectively, in a control subject (*B*). Note the prominent refixation saccades made by the Turner syndrome patient at the end of the head movement.