

Received: 2020.01.16

Accepted: 2020.02.13

Available online: 2020.02.27

Published: 2020.04.13

Vestibular Function Correlates with Radiologic Findings in a Gymnast with 22q11.2DS

Authors' Contribution:

Study Design A

Data Collection B

Statistical Analysis C

Data Interpretation D

Manuscript Preparation E

Literature Search F

Funds Collection G

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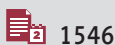
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Patient: Female, 14-year-old
Final Diagnosis: 22q11.2DS with vestibular dysfunction
Symptoms: Balance problems
Medication: —
Clinical Procedure: Comprehensive balance assessment
Specialty: Otolaryngology

Objective: Challenging differential diagnosis**Background:** The 22q11.2 deletion syndrome (22q11.2DS) is the most common identified microdeletion in humans. Anomalies of the vestibular system can occur with great frequency and are reported in the radiology literature. Fewer reports exist regarding vestibular function or its clinical features.**Case Report:** We present a case report of a competitive gymnast with 22q11.2DS who was noted to be having specific issues related to balance under conditions of competition, specifically on the balance beam. Comprehensive balance assessment provided evidence of the absence of lateral semicircular canal function, correlating with computed tomography findings and her symptoms. Counselling and targeted training greatly improved her performance.**Conclusions:** Comprehensive balance testing correlated with clinical and radiographic findings in a competitive gymnast with 22q11.2DS. Results demonstrated the functional aspect of this anomaly but also displayed the extent to which the complex interactions of all components of balance can work together to overcome balance issues under intense vestibular stress.**MeSH Keywords:** DiGeorge Syndrome • Neurotology • Vestibular Function Tests**Full-text PDF:** <https://www.amjcaserep.com/abstract/index/idArt/922908>

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Background

The 22q11.2 deletion syndrome (22q11.2DS) is the most common identified chromosomal microdeletion, occurring in approximately 1: 1000 fetuses [1]. Initially described eponymously as DiGeorge syndrome (after being described by DiGeorge in 1965), it was expanded on by Shprintzen in 1978 and thus described as Velocardiofacial syndrome. Today it has been established that microdeletions on the 22nd chromosome at the locus q11.2 can lead to heterogenous clinical presentations involving many organ systems and is referred to as 22q11.2DS.

Otologic and audiologic abnormalities are frequently identified as being associated with 22q11.2DS [2]. Weir et al. [3] describes a 60% incidence of hearing loss in these patients. With increasing use of computed tomography (CT) scanning to identify and characterize underlying anomalies of the temporal bone in the context of hearing loss, it has become apparent that certain anatomic malformations occur with increased frequency in patients with 22q11.2DS. In particular, abnormal lateral semicircular canals are noted in almost a third of ears scanned [4]. Despite this being a common finding in this patient population, a literature search identified only one article describing balance testing in 22q11.2DS patients. This article [5] confirmed that vestibular dysfunction is frequent in this patient population with 90% of patients exhibiting at least one abnormality on testing.

Case Report

A 14-year-old female known to have 22q11.2DS presented to the Pediatric Otolaryngology Clinic because of very specific issues she was encountering during gymnastics [6]. Specifically, she was having issues transitioning between elements such as jumps and flips on the balance beam. When she would complete one element and then land on the beam, she would often require a second or two to regain composure before attempting the next element. Although not necessarily unusual, it appeared to both her family and her coaches that she needed to steady herself before tackling the next element.

She was well known to our practice having been seen during her first few weeks of life. She had been born at 38 weeks to a G2P1 35-year-old healthy female. Antenatal ultrasound had shown a tetralogy of Fallot and she underwent her first cardiac procedure before discharge. At her initial surgery she was noted by Otolaryngology to have mild subglottic narrowing (Myer-Cotton Grade 1) on bronchoscopy requested by the Anesthesia Service due to difficulty intubating. This required only expectant management. During her subsequent years of follow-up, she required 3 sets of ventilation tubes because of chronic otitis media with effusion. She had normal hearing

thresholds when her ears were clear-there was no evidence of any sensorineural hearing loss.

As she got older, she began to develop her skills as a gymnast. Over the course of several years, she improved rapidly and by her presentation to the clinic at age 14, she was a national-level competitive gymnast. Her performance on the balance beam was where it was noticed that there was something a little different. During training, as elements are combined with footfalls on the beam linking them, she seemed to require a momentary pause where she would describe feeling unsteady prior to linking it to the next element in the combination. At times she would even pause long enough to tap her hand to her forehead before beginning the next phase of the routine. Despite this slightly unusual behavior she was performing well, although it was enough that the coaches were asking if there was something wrong.

Her examination was quite unremarkable. Her general physical examination and vitals were normal, and her ear, nose, and throat examination was normal. Her cranial nerve and cerebellar testing appeared to be normal, as well. General clinical determinants of balance were all within normal limits.

A review of her past medical imaging was undertaken, and it was discovered that she had had a computed tomography (CT) scan of her spine in 2014. Review of these images revealed that slices had been obtained as high as the temporal bone and that those images (although not of the typical quality one would require for a detailed temporal bone CT) showed evidence of an absent lateral semicircular canal on the right and a malformed one on the left (Figure 1). On the basis of the history, physical examination, and imaging, she was sent to the Vancouver General Hospital Neurotology Unit for Comprehensive Balance Testing. This included: electronystagmography (ENG), posturography, vestibular evoked myogenic potential (VEMP) testing, and head impulse testing (HIT).

Oculomotor testing revealed no evidence of cerebellar pathology. Calorics were absent. Sensory organization testing by posturography was normal. She performed particularly well on conditions 5 and 6 (these provide maximal stress to the vestibular system). HIT testing was carried out in all 3 planes. Vestibulo-ocular reflex (VOR) gains were borderline abnormal and there was also some evidence of covert (“catch-up”) saccades seen occasionally.

After gathering all the radiographic and vestibular testing information, the patient and her parents returned for follow-up consultation. Better understanding of her underlying condition greatly helped the patient, her family, and her coaches understand why she had issues under very specific conditions. Tailoring her practices to target this, along with her improved

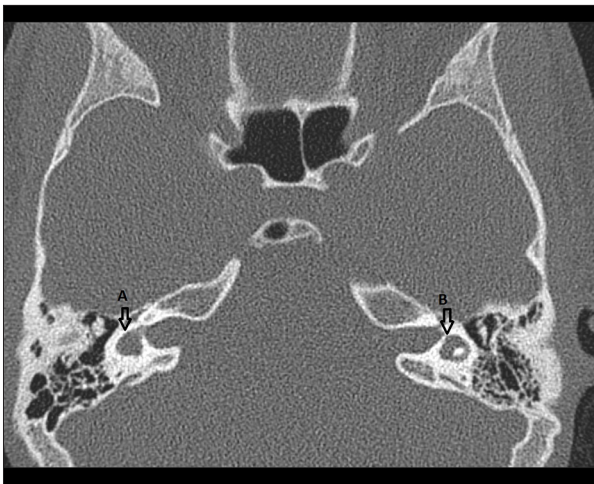


Figure 1. Computed tomography axial temporal bone scan displaying dysplastic right lateral semicircular canal and small bony-island abnormality of left vestibular system. A) Aplastic right lateral semicircular canal, B) hypoplastic left semicircular canal with some preservation of abnormally small bony island.

understanding, allowed for slow, sustained improvement in performance. Although it is impossible for her to regain function which does not exist due to a congenital absence, it is possible to better adapt to it. A deeper understanding for all parties involved was crucial to success in this case, highlighting that the plasticity of the vestibular system is tremendous.

Discussion

Previously known as DiGeorge or Velo-Cardio-Facial Syndrome, 22q11.2DS is a heterogenous condition associated with a variety of microdeletions at the specific gene locus ranging from 0.7–3 million base pairs. Most are *de novo* deletions that are not present in either parent, although it is inheritable, and with improved survival this mode of transmission is expected to increase. Clinical findings can include multi-organ dysfunction, cardiac anomalies, palatal abnormalities, scoliosis, immune differences, endocrine, genitourinary, and cognitive or neuropsychiatric illnesses. It is the commonest cause of palatal issues and the second commonest cause of congenital heart disease as well as developmental delays [1].

A systematic review [7] showed a wide range of hearing loss from studies of patients with 22q11.2DS concluding that it was a frequent finding and warranted close audiometric and otologic follow-up in these patients. Anatomic malformations of the inner ear relating to the audiologic findings have been noted [4]. Common CT findings suggestive of association with hearing loss include dense stapes (36%) and incomplete partition type 2 (23%). Whereas findings which may contribute

to balance dysfunction included malformations of the lateral semicircular canal in 33% and fusion of the lateral canal to the vestibule in 29% – suggesting that perhaps over half of patients with 22q11.2DS could harbor abnormalities of their vestibular system.

When looking at the measurable abnormalities of balance function in 22q11.2DS patients, Willaert et al. [5] found that 55% of patients had unilateral caloric hypofunction, absent or inconclusive c-VEMP response in 15% and 33% respectively, and abnormal posturography results in 68%. Perhaps more remarkably, 90% of the 22q11.2DS patients had weak caloric responses, independent of symmetry.

Patients with 22q11.2DS are at risk of a range of vestibular malformations and thus, altered vestibular physiology. Counselling patients and family members regarding the potential issues which could be encountered seems reasonable. As an example, an occupation where a fully functioning balance system could be critical (such as high building construction) would prompt detailed counselling and possibly further investigations. Gait abnormalities or delays in ambulatory motor skills in smaller children with 22q11.2DS should also prompt questions as to whether it is a manifestation of an underlying balance deficit. Any patient displaying issues with balance could have both radiologic and vestibular investigations tailored to their functional issues and balance needs.

We have described a patient who is a national-level competitive gymnast known to have 22q11.2DS. As she has advanced in her skill and ability, it has become apparent that she could have momentary instances of dysequilibrium associated with rapid spinning or flipping motions on the balance beam. These were most pronounced as she returned to upright posture just after landing on the beam. Her Comprehensive Balance Assessment was in agreement with her complaints as well as correlating with her CT imaging which showed severe malformation of her lateral semicircular canals bilaterally. She has learned to successfully compensate for her congenital balance deficit which shows the plasticity and complexity of the entirety of the balance system working together.

Conclusions

Absence or severe malformation of lateral semicircular canals is one of several known anomalies of the temporal bone associated with the 22q11.2 deletion syndrome. In this case report we have presented a competitive gymnast who had comprehensive balance assessment findings clearly demonstrating the functional aspect of this anomaly but also demonstrating the extent to which the complex interactions of all components of balance can work together to overcome this under intense

vestibular stress. All patients with 22q11.2DS should be considered to be at some risk for vestibular dysfunction and patient and parental counselling regarding this issue is recommended. Further radiologic or vestibular investigations should be tailored to the individual's needs but can be helpful to define and provide explanations to families.

Department and Institution where work was done

Work was done at both BC Children's Hospital (Lindy M.R. Moxham) and Vancouver General Hospital (Arthur I. Mallinson).

Conflict of interest

None.

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