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Spiral ganglion deficiency in adult-onset deafness-dystonia syndrome

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Introduction

Deafness-dystonia syndromes are rare group of syndromes that present with deafness and dystonia as the dominant symptoms and are increasingly being recognized for their clinical and genetic heterogeneity (1). Most reported cases are due to X-linked deafness-dystonia-optic neuropathy (DDON) or Mohr-Tranebjaerg syndrome (MTS); autosomal recessive Woodhouse-Sakati syndrome; succinate-coenzyme A (CoA) ligase, ADP-forming, b subunit (SUCLA2)-related mitochondrial DNA depletion syndrome; or organic acidurias (1). With respect to general phenotype, these individuals are characterized by pre- or post-lingual progressive auditory neuropathy (AN), dystonias that have a variable age of onset, association with optic atrophy in some cases (DDON in particular), and an association with psychosis, dementia and mental retardation in the more severe cases/syndromes (1,2). While previously most cases of deafness-dystonia syndrome have been associated with the onset of deafness in childhood, there is a small group of cases reporting onset of deafness at later ages into adulthood (1). Merchant and colleagues demonstrated the near-total loss of spiral ganglion neurons in patients with DDON despite an intact organ of Corti (3). Brookes and colleagues reported only marginal cochlear implant performance after 2 years in a child with DDON who underwent cochlear implantation (4). This patient exhibited continued poor scores in standardized speech, language, and audiometric tests and required higher than normal current for the most-comfortable-level implant setting (4). While the histopathology of this entity has been documented in patients with childhood-onset deafness, the histopathology of patients with deafness-dystonia syndrome who present with adult-onset deafness has not been previously reported.

Case Report

A 67 year-old male presented with worsening bilateral hearing loss and difficulties with balance. The patient noted a history of left arm and hand paralysis at the age of 42 years and noted that this had occurred prior to the initial onset of bilateral sensorineural hearing loss (SNHL) at the age of 52 years. Subsequently, the patient developed unexplained double vision. Just prior to his presentation, the patient described an episode associated with loss of residual hearing, worsening balance and double vision, and a paralysis involving his right biceps that occurred overnight. Exam audiogram revealed a profound bilateral SNHL with no response to caloric irrigation on electronystagmogram (ENG). A nerve conduction study of the right arm and bilateral lower extremities done as part of the patient's workup at the time revealed a moderately slowed nerve conduction velocity of 36 meters per second (normal >40–45 m/sec) in the right peroneal nerve.

Histopathology

Temporal bone histopathology of the cochlea at the level of the modiolus demonstrates intact organ of Corti with a near-total degeneration of the spiral ganglion cells of the cochlear nerve (Figure 1). The typical cellularity of the nerve is lost in both the cochlear and vestibular nerves, while the facial nerve is typically spared. This contrast in cellularity can be seen at the level of the superior vestibular nerve, where the facial nerve exhibits the normal fiber density of a nerve, in contrast to that of the superior vestibular nerve, which exhibits near-total loss of the normal fiber density (Figure 2A). Three remaining spiral ganglion cells remain (lowermost cell is marked by an asterisk). Some cochlear efferent fibers that remain are visualized in close proximity to the hook region of the basal turn of the cochlea (Figure 2B).

Discussion

Patient with deafness-dystonia syndrome may present with adult-onset deafness with an auditory neuropathy-like picture (1). Our patient presented with adult onset bilateral profound SNHL and exhibited near-total loss of spiral ganglion cells and severely degenerated architecture of the cochlear and vestibular nerves, despite normal appearing cochlear organ of Corti architecture. Furthermore, the patient's dystonia symptoms were mild and segmental. It has been previously reported that cochlear implant performance in deafness-dystonia optic neuronopathy (DDON) is marginal (4). It is possible that adult patients with a similar adult-onset auditory neuropathy-like presentation may possess a similar histological picture. Deafness-dystonia syndromes should be considered in the differential diagnosis of adults presenting with an auditory neuropathy-like picture. Furthermore, consideration of the potential marginal cochlear implant performance in these patients should be considered, but should not deter attempts at cochlear implantation. Marginal cochlear implant performance may in the future prompt consideration of auditory brainstem implantation in this population of patients.

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References

1. Kojovic M, Pareés I, Lampreia T, Pienczk-Reclawowicz K, Xiromerisiou G, Rubio-Agusti I, Kramberger M, Carecchio M, Alazami AN, Brancati F, Slawek J, Pirtosek Z, Valente EM, Alkuraya FS, Edwards MJ, Bhatia KP. The syndrome of deafness-dystonia: clinical and genetic heterogeneity. *Mov Disord*. 2013 Feb 15. [Epub ahead of print]. 10.1002/mds.25394
2. Tranebjaerg L, Hamel BCJ, Gabreels FJM, Renier WO, Van Ghelue M. A *de novo* missense mutation in a critical domain of the X-linked *DDP* gene causes the typical deafness-dystonia-optic atrophy syndrome. *Eur J Hum Genet*. 2000; 8:464–7. [PubMed: 10878669]
3. Merchant SN, McKenna MJ, Nadol JB Jr, Kristiansen AG, Tropitzsch A, Lindal S, Tranebjaerg L. Temporal bone histopathologic and genetic studies in Mohr-Tranebjaerg syndrome (DFN-1). *Otol Neurotol*. 2001; 22:506–511. [PubMed: 11449109]
4. Brookes JT, Kanis AB, Tan LY, Tranebjaerg L, Vore A, Smith RJH. Cochlear implantation in deafness-dystonia-optic neuronopathy (DDON) syndrome. *Int J Ped Otorhinolaryngol*. 2008; 72:121–6.

Figure 1. Mid-modiolar axial cross-section of the cochlea demonstrating normal organ of Corti structure (*short arrow*) with near total degeneration of the cochlear nerve (*long arrow*) (hematoxylin and eosin; original magnification $\times 4$).

Figure 2.

Neural structure histopathology in adult-onset deafness-dystonia. 2A, Axial cross-section of the cochlea at the level of the superior vestibular nerve (hematoxylin and eosin, original magnification x20). Note the normal fiber density of the facial nerve (left) in contrast to the near-total loss of the normal fiber density (right). A few remaining spiral ganglion cells remain (*asterix*). 2B, Axial cross-section of the cochlea at the level of the hook region of the basal turn of the cochlea demonstrates the presence of cochlear efferent fibers (*arrowheads*, hematoxylin and eosin, original magnification x10).