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Case Report

Neuro–Otological Manifestations in Patients with a Hemifacial Port–Wine Stain: A Report of 2 Cases

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A port-wine stain (PWS) or nevus flammeus is a congenital capillary malformation that often affects the skin of the head and neck region. Little is known about neuro-otological manifestations associated with this birthmark. We describe 2 patients with a hemifacial PWS and sensorineural hearing loss, caused by involvement of the internal auditory canal (IAC) and inner ear structures. Case one had a history of sudden vestibular hypofunction, followed by high-frequency sensorineural hearing loss 2 years later. In the second case, the exact onset of the high-frequency hearing loss could not be determined. In both patients, magnetic resonance imaging (MRI) showed dural enhancement of the IAC and a loss of T2 signal intensity of the ipsilateral labyrinth. This report shows that a PWS of the head and neck region may be associated with dural thickening within the IAC and secondary inner ear dysfunction. In patients with this capillary malformation suffering from hearing impairment or balance problems, MRI is warranted to detect involvement of the ipsilateral dura and labyrinth.

KEYWORDS: Port-wine stain, magnetic resonance imaging, sensorineural hearing loss, dura mater

INTRODUCTION

A port-wine stain (PWS) or nevus flammeus is a common congenital capillary malformation affecting 0.3%-0.5% of newborns.[1] It presents as a pink or red birthmark which becomes darker and more raised over time. The majority of PWS are located in the head and neck region, especially in the trigeminal dermatomal distribution.[1] Histologically, a PWS consists of dilated capillaries and post-capillary venules. The exact pathophysiology still remains unclear. Disturbed innervation of dermal blood vessels and dysregulation of angiogenic factors leading to a decreased vascular tone and dilatation are the dominant theories.[1] A PWS may present as an isolated malformation or may be associated with other vascular anomalies such as Sturge–Weber syndrome (SWS). This neurocutaneous syndrome, also known as encephalotrigeminal angiomatosis, is characterized by a facial port-wine birthmark in the distribution of the trigeminal nerve, leptomeningeal angiomatosis, and vascular malformation of the eye.[2] The clinical presentation of SWS is very heterogeneous. Neurological symptoms are caused by impaired blood flow, are mostly progressive, and include seizures, mental retardation, hemiparesis, and headaches.[3,4] Hemispheric leptomeningeal enhancement on contrast-enhanced T1-weighted magnetic resonance imaging (MRI) is a hallmark of SWS. Other features on MRI include brain atrophy and prominent choroidal vessels.[3] Computed tomography may show cortical and subcortical calcifications.[5] Both isolated PWS and SWS are caused by a somatic mosaic activating variant in the *GNAQ* gene, which codes for the guanine nucleotide-binding protein G(q) subunit alpha.[6,7] It is assumed that this variant occurs early in embryologic development.[6]

Little is known about neuro-otological manifestations in patients with an isolated PWS or SWS. We present here 2 cases of patients with a facial capillary malformation associated with ipsilateral dural thickening of the internal auditory canal (IAC) and inner ear dysfunction.

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CASE PRESENTATIONS

Patient 1

A 31-year-old man presented at the outpatient clinic of the AZ Sint-Lucas Hospital with sudden high-frequency sensorineural hearing loss and tinnitus on the left side. Pure tone average (PTA) for the frequencies 0.5-4 kHz was 35.0 dB Hearing Level (HL) (Figure 1A). Two years before the sudden hearing loss, he had consulted another ENT specialist because of an acute vestibular syndrome causing rotatory vertigo that lasted for a couple of weeks. At that time, caloric testing showed complete vestibular areflexia and a decreased cVEMP amplitude on the left side, which did not recover. He had a congenital PWS on the left facial hemisphere, involving V3, C2, and C3, and a congenital absence of the left forearm. There was no history of glaucoma or seizures. Magnetic resonance imaging showed prominent pachymeningeal enhancement in the left-sided middle and posterior cranial fossa (Figure 2A) and IAC on T1-weighted images with Gadolinium (Figure 2B). In addition, a decreased T2-signal of the left inner ear structures (Figure 2C) was visible. Extensive neurological investigations, including blood tests, lumbar puncture, and positron emission tomography–computed tomography scan, were all negative. Oral corticosteroids were started, but no improvement was noted during follow-up. Verbal informed consent was obtained from the patient for publication of this case report.

Patient 2

A 46-year-old man came to the outpatient clinicof the AZ Sint-Lucas Hospital with complaints of left-sided tinnitus. He had a port-wine birthmark in the left head and neck region involving V2, V3, C2-C4. There was no history of glaucoma. Otomicroscopy showed the extension of the capillary hemangioma into the external ear canal and on the tympanic membrane. Audiometric testing revealed a high-frequency hearing loss with a PTA (0.5-4 kHz) of 60.0 dB HL (Figure 1B). A review of previous audiograms demonstrated that this hearing loss was already noticed 7 years before. Videonystagmography showed normal caloric responses on both sides. Clinical neurological examination was normal. There was no history of glaucoma or seizures. Magnetic resonance imaging showed pachymeningeal enhancement in the left-sided middle and posterior cranial fossa (Figure 3A) and IAC (Figure 3B) on T1-weighted images with Gadolinium and a signal decrease of the inner ear structures on T2 (Figure 3C). Further follow-up was scheduled. Verbal informed consent was obtained from the patient for publication of this case report.

DISCUSSION

Pachymeningeal or dural enhancement is a radiological feature that can have various etiologies, including inflammatory, infectious, neoplastic, and idiopathic processes.[8] In the above cases, these possible underlying conditions could be ruled out. In both patients, the area of the PWS (overlapping V3, C2, and C3) was similar, as well as the affected pachymeninges (middle and posterior cranial fossa and IAC). To our knowledge, only 1 comparable case can be found in the literature.[9] This concerned a patient with SWS who had a bilateral facial hemangioma of V2-V3, bilateral glaucoma, and a history of seizures. This woman had a left vestibular paresis and developed a progressive high-frequency sensorineural hearing loss over a period of 2 years. Apart from cortico-subcortical atrophy, pial angiomatosis, and gyral calcifications—which are typical signs of SWS—a similar

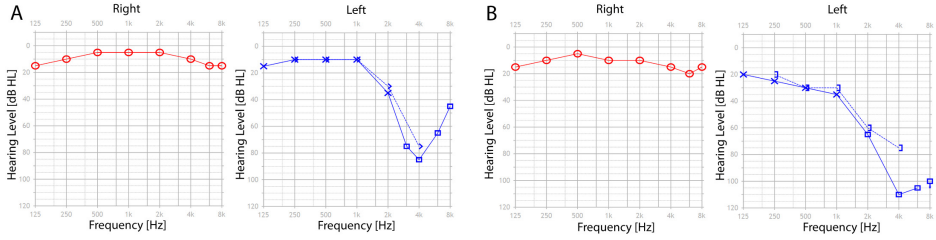

Figure 1. Audiograms. (A) High-frequency sensorineural hearing loss (PTA0,5-4 kHz: 35.0 dB HL) on the left side in patient 1. Normal hearing thresholds at the right side. (B) High-frequency sensorineural hearing loss (PTA 0,5-4 kHz: 60.0 dB HL) on the left side in patient 2. Normal hearing thresholds at the right side. PTA, pure tone average.

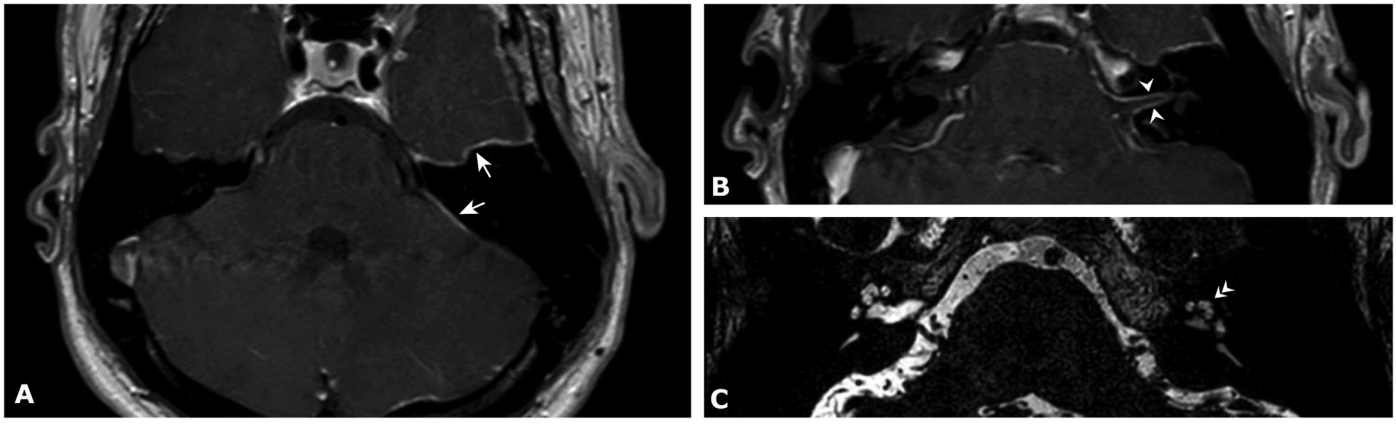

Figure 2. Magnetic resonance imaging in patient 1. (A) Axial T1-weighted image after administration of Gadolinium demonstrates prominent dural enhancement in the left-sided middle and posterior cranial fossa (arrows). (B) Similar dural enhancement is seen along the internal auditory meatus (arrow heads). (C) Axial T2-weighted 3D cisternography shows a decreased signal of the entire inner ear structures on the left side (double arrow head).

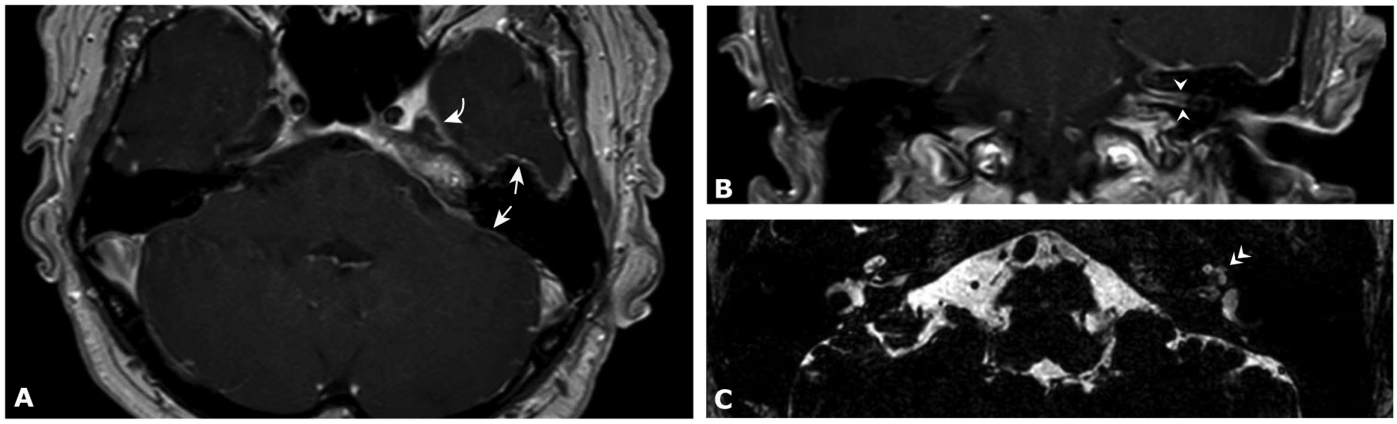

Figure 3. Magnetic resonance imaging in patient 2. (A) Axial T1-weighted image after administration of Gadolinium shows prominent dural enhancement in the left-sided middle and posterior cranial fossa (arrows). A similar dural thickening is seen around Meckle's cave (curved arrow). (B) Coronal T1-weighted image after administration of Gadolinium shows dural enhancement along the internal auditory meatus (arrow heads). (C) Axial T2-weighted 3D cisternography reveals a decreased signal of the entire inner ear structures on the left side (double arrow head).

meningeal enhancement of the IAC on T1-weighted images with Gadolinium was reported. Based on these findings, it seems that pachymeningeal enhancement of the IAC can occur in patients with an isolated PWS of the head and neck as well as in patients with SWS. It has been shown that a somatic mosaic activating variant in *GNAQ* can be found in the vast majority of PWS. This variant is also present in the brain tissue of patients with SWS.[6] It is quite possible that the dural thickening in the above cases shares similar histopathologic features to the dermal capillary malformations. Unfortunately, tissue samples of the affected skin or hypertrophic dura of our patients were not available. Further research is needed to better understand the underlying genetic conditions, the pathophysiology, and the progression over time.

Literature on audio-vestibular dysfunction in patients with PWS is scarce. One patient has been reported with a hemifacial PWS caused by a variant in *GNAQ*, and a bilateral large vestibular aqueduct due to a variant in *SLC26A4*.[10] Though it remains unclear if this concerns a real association or rather a coincidental combination of 2 unrelated entities.

A remarkable finding in the presented patients is the signal loss of the inner ear structures on the T2-weighted 3D cisternographic images. This signal loss was not limited to the cochlea but could be observed in the entire labyrinth. Similar intermediate labyrinthine fluid signal intensity occurs in a subset of patients with a vestibular schwannoma and has been linked to elevated protein levels.[11] Ototoxic or neurotoxic secretions of schwannomas passing through the fundus or ischemia due to compression of the labyrinthine artery are possible theories that have been proposed.[12,13] In patients with pachymeningeal thickening of the IAC, a similar pathophysiological mechanism might be the cause of increased protein content of the inner ear fluids. A clear correlation between a decreased T2 signal intensity of the inner ear structures and hearing loss or vestibular dysfunction has not been found.[11] Why the above patients presented with sensorineural hearing loss at the higher frequencies and why one patient also had vestibular areflexia still remains unclear.

In patients with an isolated PWS or SWS suffering from hearing loss, both pure-tone audiometry and videonystagmography should be conducted for adequate assessment. An MRI scan is needed to screen for involvement of the IAC and the ipsilateral labyrinth.

CONCLUSION

This article presents 2 cases of an isolated PWS associated with sensorineural hearing loss caused by involvement of the IAC and labyrinth. In patients with auditory or vestibular complaints and a PWS of the head and neck, clinical suspicion for this diagnosis should be raised, and an MRI is warranted. Further research is needed to understand the underlying mechanisms.

Availability of Data and Materials: The data that support the findings of this study are available on request from the corresponding author.

Ethics Committee Approval: N/A.

Informed Consent: Verbal informed consent was obtained from the patients who participated in this study.

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Declaration of Interests: The authors have no conflicts of interest to declare.

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