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LETTER TO THE EDITOR

Absent cochlear and abducens nerves in a patient with Duane retraction syndrome

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Dear Editor,

Duane retraction syndrome (DRS) is characterized by congenital abduction or adduction limitation, palpebral fissure narrowing, and upshoots or downshoots on adduction. DRS is a congenital cranial dysinnervation disorder (CCDD) which represents various neurodevelopmental disorders of the cranial nerve nuclei and their axonal connections in the brainstem [1]. Aberrant innervation occurs when other cranial nerve fibers innervate the primarily dysinnervated muscles [1]. Kim and Hwang reported that the abducens nerve is absent in type 1 and in part of type 3 DRS [1, 2]. Congenital hearing loss has also been reported in patients with DRS; however, the etiology of hearing loss has not been clearly elucidated [3]. Herein, we found a patient with DRS and coexistent hearing loss, whose cochlear nerve and abducens nerve were both absent on magnetic resonance imaging (MRI).

A 2-year-old boy presented with limited abduction of the left eye since infancy. He was born full term with no significant perinatal history. Hearing loss was found on the left side during a newborn screening test. Computed tomogram revealed an incomplete turn of the left cochlea and prominent left utricle dilatation, suggesting hypoplasia of the left cochlear nerve.

On ophthalmologic examination, he fixed and followed with each eye. He showed orthotropia at distance and at near with alternate prism and cover test. Ductions and versions were normal in the right eye, and showed limitation of abduction, fissure narrowing, and upshoots on adduction in the left eye (Fig. 1a). His pupils were round and isocoric. Dilated fundus examinations were normal. Cycloplegic refraction showed − 1.50 diopters of myopia in both eyes. There were no physical findings related to Klippel-Feil anomaly, radial aplasia, thenar hypoplasia, or absent thumbs. Developmental milestones were reached within normal limits.

Using a 3-T MRI system (Ingenia CX, Philips, Best, the Netherlands), 0.7-mm-thick transverse T2-weighted imaging for cranial nerves in the basal cistern, and 1-mm-thick coronal T2-weighted imaging for the orbit were performed. The left abducens nerve and cochlear nerve were not found (Fig. 1b, c), and the left cochlea was hypoplastic (Fig. 1d). The right abducens nerve, right cochlear nerve, and both vestibular nerves were normally identified. All other cranial nerves including the oculomotor, trochlear, and facial nerves were normally identified, and extraocular muscles were normal in size and shape on both sides (Fig. 1e).

The development of high-resolution thin–section MRI has facilitated the diagnosis of CCDD. In this study, concomitant absence of the abducens nerve and cochlear nerve was found in a patient with congenital hearing loss and DRS.

There are many syndromes presenting with congenital hearing loss and DRS. Wildervanck's cervico-oculo-acusticus syndrome is characterized by Klippel-Feil anomaly, congenital hearing loss, and DRS [4, 5]. Okihiro syndrome encompasses bilateral radial aplasia with absent thumbs, unilateral sensorineural hearing loss, and bilateral DRS [4, 5]. Bosley-Salih-Alorainy syndrome features bilateral DRS type 3, congenital hearing loss, delayed motor milestones, cognitive abnormalities, and cerebrovascular malformations [4, 5]. Our patient did not show any of these findings other than DRS and congenital hearing loss.

The incidence of hearing loss in patients with DRS ranges from 1 to 72%, depending on appropriate audiologic and otologic examinations [4, 5]. However, the etiology of hearing loss in DRS patients has not been clearly documented. Hearing loss in Wildervanck syndrome has been reported to be caused by cochlear bony malformation [4, 5]. In our case, absence of the cochlear nerve was found on high-resolution

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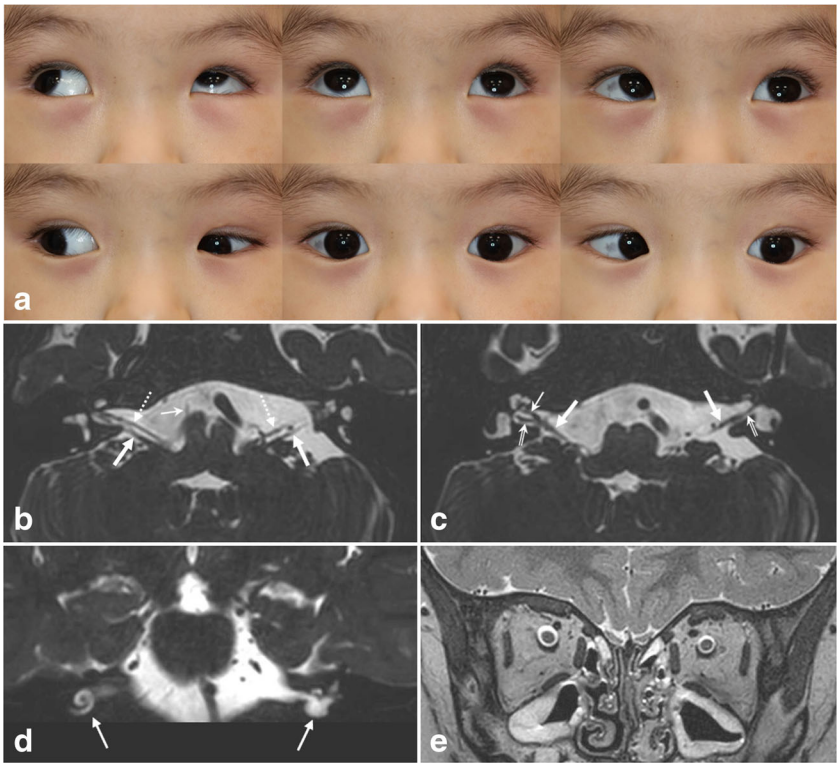

Fig. 1 **a** Ocular versions show a marked limitation of abduction and upshoot on adduction in the left eye. **b** On transverse T2-weighted imaging, the right abducens nerve is identified (thin arrow), but the left one is not. The facial nerves (dotted arrows) and acoustic nerves (thick arrows) are well identified bilaterally. **c** On transverse T2-weighted imaging obtained at a slightly lower level than (**b**), the right acoustic nerves (thick arrow) divide normally into the cochlear nerve (thin arrow) anteriorly, and the vestibular nerve (double arrow) posteriorly. The left acoustic nerve (thick arrow) courses laterally continuing to the vestibular nerve (double arrow), without branching into the cochlear nerve. **d** On reformatted coronal T2-weighted imaging, the right cochlea (arrow) has the normal spiral shape. The left cochlea appears round suggesting hypoplasia (arrow). **e** On coronal T2-weighted imaging, all extraocular muscles on both sides are normal in size and shape

MRI, which extends our understanding of the etiology of hearing loss in patients with DRS.

Abducens nerve agenesis in DRS may be associated with other cranial nerve abnormalities including the trochlear nerve and facial nerve [6–8]. This is the first report which documents the combined absence of the abducens and cochlear nerves on MRI.

In conclusion, combined absence of abducens and cochlear nerves in a patient with coexistent DRS and hearing loss implies that both conditions are manifestations of CCDD.

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Compliance with ethical standards

Conflict of interest statement The authors declare that they have no conflict of interest.

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