Neurological Sciences (2019) 40:2419–2421
https://doi.org/10.1007/s10072-019-03928-x

LETTER TO THE EDITOR

Combination of olfactory aplasia and congenital ocular motor apraxia: a previously unreported association

Hee Kyung Yang [1] · Jae Hyoung Kim [2] · Jeong-Min Hwang [1]

Received: 9 October 2018 / Accepted: 9 May 2019 / Published online: 25 May 2019

Dear Editor,

Congenital ocular motor apraxia is characterized by impaired horizontal voluntary saccades associated with compensatory head thrust [1]. Olfactory bulb hypoplasia is known to be associated with many syndromes such as Kallmann syndrome or CHARGE syndrome (Coloboma of the eye, Heart defects, Atresia of the choanae, Retardation of growth and/or development, Genital and/or urinary abnormalities, Ear anomalies and/or deafness) [2, 3]. We present two cases with a yet unreported association of congenital ocular motor apraxia and olfactory bulb aplasia.

Case report

Case 1

A 7-month-old boy presented with poor eye contact and poor grip of subjects. He was born at the age of 38 weeks with a body weight of 2.6 kg. He was diagnosed with hypothyroidism. Neurologic examination and electroencephalogram showed no other abnormal findings.

On ophthalmic examination, he fixed and followed a 5-in. object at near with either eye. He had esotropia of 15 prism diopters at near in the primary position measured with the Krimsky method. He showed head thrusts on attempted horizontal gazes. Cycloplegic refraction was + 4.25 Dsph −5.00 Dcyl x 180A OD and + 3.75 Dsph −2.00 Dcyl x 180A OS. Handheld slit lamp examination of the anterior segment and fundus examination showed no abnormal findings.

Axial T2-weighted magnetic resonance (MR) imaging showed inferior vermian hypoplasia (Fig. 1a, b). Thin-section coronal T2-weighted images showed bilateral absence of the olfactory bulbs and tracts within the shallow olfactory fossa (Fig. 1c–f).

Case 2

A 24-month-old boy presented with face turn to either side since early infancy. He was born at the age of 36 weeks with a body weight of 2.8 kg.

On ophthalmic examination, he fixed and followed a 5-in. object at near with either eye. He showed orthotropia at near in the primary position and head thrusts on attempted horizontal gazes. Cycloplegic refraction was + 4.75 Dsph OU. Handheld slit lamp examination of the anterior segment and fundus examination showed no abnormal findings.

Axial T2-weighted MR images showed inferior vermian hypoplasia (Fig. 2a). On thin-section coronal T2-weighted images, the olfactory bulbs and tracts within the shallow olfactory fossa were not identified on both sides (Fig. 2b).

Congenital ocular motor apraxia has been reported to be most frequently associated with structural abnormalities of the cerebellum, the corpus callosum, and the fourth ventricle. [3] In addition to the well-known cerebellar vermian hypoplasia, our two cases showed olfactory bulb aplasia or severe hypoplasia, which has not been previously reported in association with congenital ocular motor apraxia.

Blustajn et al. [2] reported that T2-weighted coronal images are the most sensitive sequence to identify abnormalities

✉ Jae Hyoung Kim
jaehkim@snubh.org

✉ Jeong-Min Hwang
hjm@snubh.org

Hee Kyung Yang
eye@snubh.org

[1] Department of Ophthalmology, Seoul National University Bundang Hospital, Seoul National University College of Medicine, 300 Gumi-dong, Bundang-gu, Seongnam, Gyeonggi-do 463-707, South Korea

[2] Department of Radiology, Seoul National University Bundang Hospital, Seoul National University College of Medicine, 300 Gumi-dong, Bundang-gu, Seongnam, Gyeonggi-do 463-707, South Korea

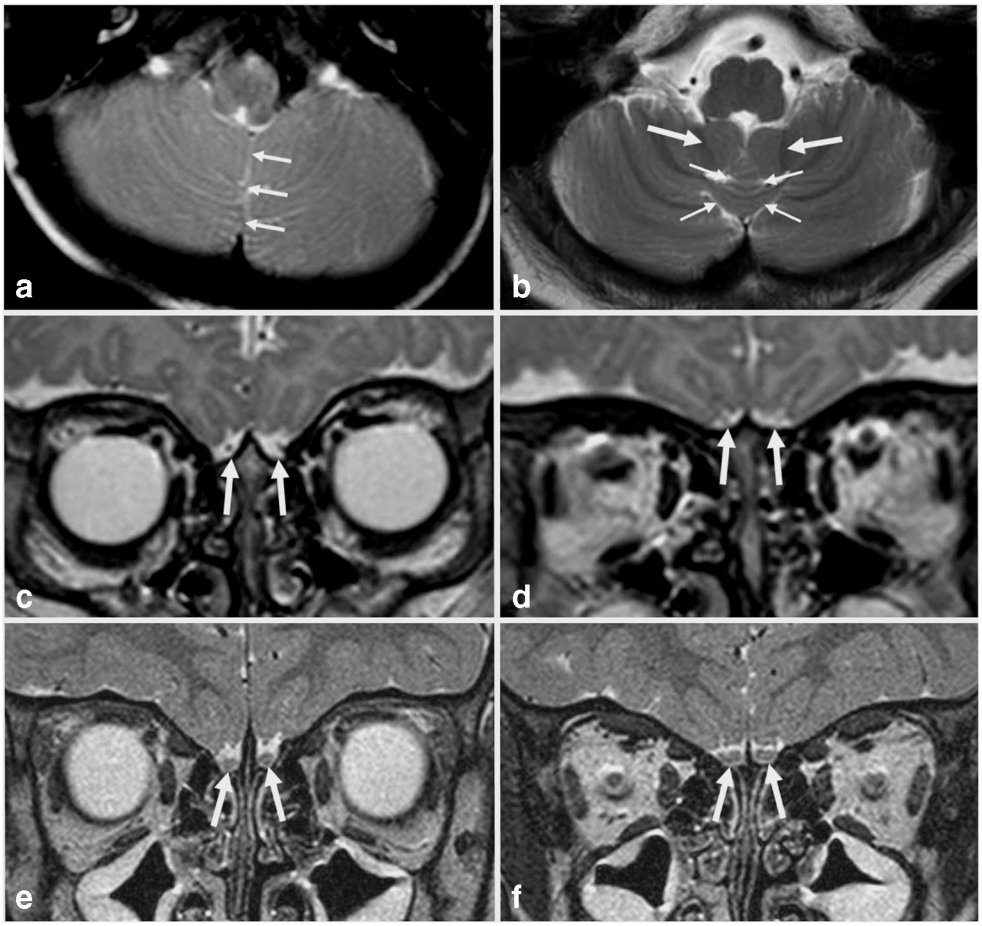

Fig. 1 Case 1. **a** Axial T2-weighted image shows broad apposition of the right and left inferior cerebellum without the intervening inferior vermis (arrows), suggestive of inferior vermian hypoplasia. Compare with the normal MR image in Fig. 1b. **b** MR image of a normal control shows the ovoid shape of both cerebellar tonsils (large arrows) and the intervening inferior vermis (small arrows) without broad apposition of both cerebellums. **c**, **d** Thin-section coronal T2-weighted images show bilateral absence of the olfactory bulbs within the olfactory fossa (arrows). Small dot-like dark signals are normal vessels in the olfactory fossa. Compare with normal MR image in Fig. 1e. **e**, **f** Olfactory bulbs (arrows) are well identified bilaterally within the olfactory fossa in a normal control

of the olfactory bulb owing to the bright signal intensity of the surrounding cerebrospinal fluid. In our institute, thin-section coronal T2-weighted imaging is a routine protocol incorporated in orbital MR imaging. Since the olfactory fossa containing the olfactory bulb is located in the anterior skull base just medial to the orbit, we can easily evaluate the olfactory bulb during orbital MR imaging.

Anosmia is known to be present in 1% of the general population [4]; however, congenital anosmia is a rare condition characterized by the lack of olfactory perception associated with hypoplasia of the olfactory bulb. We could not test olfaction in our study because the patients were both young children. The relevance of olfactory hypoplasia and congenital ocular motor apraxia should be demonstrated in future studies concerning a large number of patients. Meanwhile, Kallmann syndrome is one of the most frequent causes of congenital anosmia which is also accompanied by hypogonadotropic hypogonadism. This was not found in both cases. Booth and Rollins [4] have reported the clinical and radiological findings of children with olfactory aplasia. Eight patients (20%) had clinical findings indicating optic pathway pathology, including optic nerve hypoplasia and/or nystagmus. Vermian hypoplasia was found in three out of 41 cases with olfactory aplasia; however, whether these children had congenital ocular motor apraxia is not described in detail. The clinical significance of the association of vermian hypoplasia and

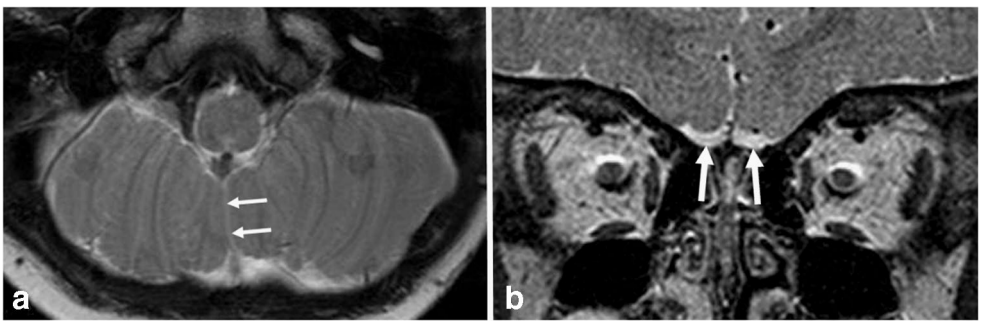

Fig. 2 Case 2. **a** Axial T2-weighted image shows broad apposition of the right and left inferior cerebellum without the intervening inferior vermis (arrows), suggestive of inferior vermian hypoplasia. **b** Thin-section coronal T2-weighted image shows bilateral absence of the olfactory bulbs within the olfactory fossa (arrows)

olfactory aplasia is not well understood, yet advances in neuroimaging and genetics are revealing various disorders that affect the cerebellar structures and function leading to neurodevelopmental dysfunction.

In addition to impaired horizontal saccades, congenital ocular motor apraxia may encompass widespread abnormalities such as developmental delay, speech delay, learning difficulties, and mild mental retardation [3, 5, 6]. Findings of our cases support that congenital ocular motor apraxia may be a more expanded disease.

In conclusion, aplasia of the olfactory bulb may be associated with congenital ocular motor apraxia.

Compliance with ethical standards The authors declare that the study has been performed in accordance with ethical standards laid down in the 1964 Declaration of Helsinki.

Conflict of interest The authors declare that they have no conflicts of interest.

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