

CASE REPORT

Isolated Congenital Anosmia

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ABSTRACT

Congenital anosmia is commonly described in conjunction with various developmental abnormalities and has been reported to be familial. Congenital anosmia as an isolated defect in a single-family member is extremely rare. We described a case of a 5-year-old child with isolated congenital anosmia.

Keywords: Congenital anosmia, Isolated congenital anosmia, Pediatric anosmia.

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INTRODUCTION

Anosmia simply means the absence of sense of smell. Anosmia can be either congenital or acquired. Acquired causes include head trauma, tumor, surgery, and inflammation.¹ In head trauma, anosmia occurs as a result of shearing or contusion of the olfactory nerves as they pass through the cribriform plate, or may also be due to meningeal reaction or fibrous scarring that develops after injury. Olfactory groove meningioma is an example of olfactory tract neoplasm that can cause anosmia. In inflammation, anosmia can occur due to obstruction of nasal cavity from sinus disease and nasal polyposis.² Congenital anosmia is a rare occurrence, and isolated congenital anosmia is even extremely rare.

CASE REPORT

A 5-year-old girl presented with an inability to smell. Besides that she also has reduced taste sensation. She did not have nasal obstruction, rhinorrhea, or history of trauma. She was born at term via spontaneous vaginal delivery.

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On examination, she was not syndromic; naso-endoscopic examination showed normal turbinates, no nasal polyp or mass obstructing the nasal cavity, and no deviated nasal septum. Smell test also did not show any positive findings.

Magnetic resonance imaging (MRI) findings showed the absence of olfactory bulbs (Fig. 1). Both olfactory sulcus and tracts were also not well visualized. Based on imaging, she was diagnosed as having agenesis of olfactory bulbs.

DISCUSSION

Congenital anosmia is occasionally reported as familial, and to the best of our knowledge, nine cases of isolated congenital anosmia have been reported.^{2,3} Abnormal embryological development of the olfactory system that causes anosmia may be due to replacement of olfactory epithelium by respiratory epithelium³ or aplasia of the olfactory bulbs, sulci, and tract.²

Congenital anosmia is commonly associated with various abnormalities and familial disorder. The most common association is hypogonadotropic hypogonadism, which was described by Kallmann et al.⁴ Kallmann syndrome may be associated with several abnormalities, such as mental retardation, audiological abnormalities, cleft lip and palate, cardiac and intracranial abnormalities. Our patient did not have any of those features.

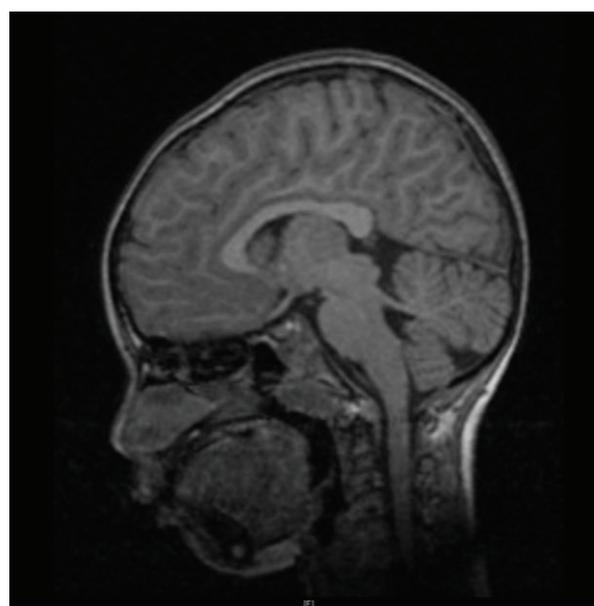


Fig. 1: Magnetic resonance imaging showed the absence of olfactory bulbs

Familial anosmia was first reported in 1918.⁵ In our case, she is the only one who has anosmia; none of the other family members have similar problem.

Magnetic resonance imaging is the investigation of choice in congenital anosmia. Smell testing, physical examination, and nasoendoscopy are not able to determine the anatomic cause of congenital olfactory loss. Initially, imaging of the olfactory system was done with computed tomography (CT) scan.⁶ However, visualization of olfactory system is limited in CT due to the beam-hardening artifacts at the skull base and not in MRI. Magnetic resonance imaging precludes the use of more invasive nasal biopsy as a diagnostic tool.

In terms of management, there is little to offer for patients with congenital anosmia.

CONCLUSION

Isolated congenital anosmia is extremely rare, and this case is not frequently reported. Patients with congenital anosmia should be fully investigated in order to exclude

more serious causes and the more commonly associated Kallmann syndrome before making a conclusion of isolated case of congenital anosmia.

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