Isolated olfactory nerve agenesis

Introduction

The absence of sense of smell (anosmia) is often underestimated (McNeill et al, 2007). Certain professions, such as sommeliers and chefs, are highly skilled olfactory-practitioners. However, every individual needs to be able to detect things such as rotten food and smoke for self-preservation.

Smell perception requires the normal anatomy and physiology of the nasal cavity (sometimes referred to as the conductive element, analogous to hearing), the presence of sensory receptors and the normal functioning neural pathways.

Isolated olfactory nerve agenesis is rare, as reported by Di Rienzo (2002). Hypoplasia or agenesis occurs in combination with other neurological malformations, most often associated with Kallman syndrome, with no other causative genetic link known (Feldmesser et al, 2007).

Discussion

Clinical testing of olfactory function has been debated in peer-reviewed otolaryngology publications (McNeill et al, 2007). It requires good compliance, which is often difficult when dealing with children. In this instance, the child and parents were keen to establish the cause of her problem and whether anything needed to be done. Magnetic resonance imaging enabled us to diagnose isolated congenital anosmia. Importantly, we were able to give this diagnosis, and explain that this is a rare problem and that she will need to take precautions for the rest of her life to ensure her anosmia does not endanger her. BJHM

Di Rienzo L (2002) Isolated congenital agenesis of the olfactory bulbs and tracts in a child without Kallman's syndrome. *Ann Otol Rhinol Laryngol* **111:** 657–60

Feldmesser E, Bercovich D, Avidan N et al (2007) Mutations in olfactory signal transduction genes are not a major cause of human congenital general anosmia. *Chem Senses* **32**(1): 21–30

McNeill E, Ramakrishnan Y, Carrie S (2007) Diagnosis and management of olfactory disorders: survey of UK-based consultants and literature review. *J Laryngol Otol* **121:** 713–20

Wani MK, Ruckenstein MJ, Parikh S (2001) Magnetic resonance imaging of the paranasal sinuses: incidental abnormalities and their relationship to patient symptoms. *J Otolaryngol* **30**(5): 257–62

Figure 1. Coronal section through the anterior skull base of the index case.

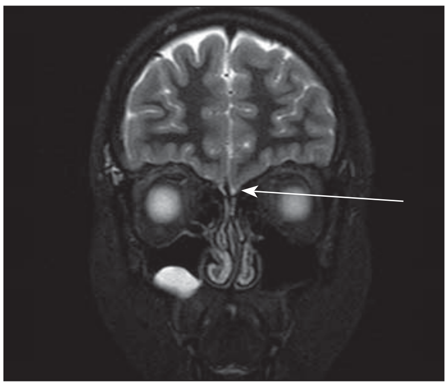


Figure 2. Coronal section through the anterior skull base of the index case, further posteriorly.

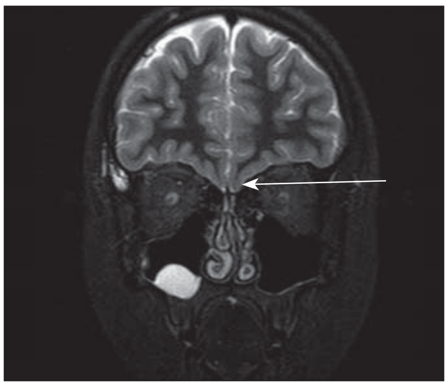


Figure 3. Coronal section through the anterior skull base of the index case, again moving posteriorly.

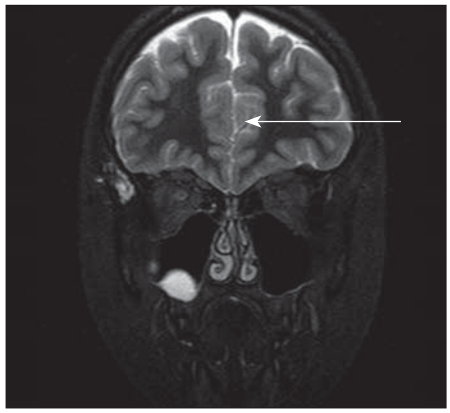
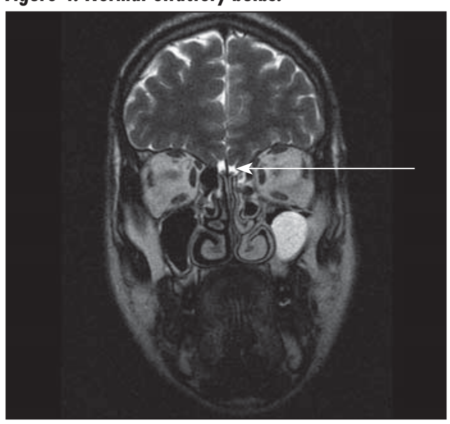


Figure 4. Normal olfactory bulbs.



Case Report

An otherwise fit and well 12-year-old girl presented to the otolaryngology clinic with lifelong anosmia. Although she had suffered a nasal injury when she was 4 years old, she was adamant that the deficit pre-dated her injury. Interestingly, her maternal grandfather also complained of this problem.

Clinical examination was entirely normal, except for her anosmia. There was no intranasal pathology such as obstructive deviations of the nasal septum or polyposis and cranial nerve examination, including trigeminal stimulation, only confirmed the absence of olfaction.

Subsequent imaging with the magnetic resonance scanner revealed a complete absence of the olfactory bulbs and nerves (see *Figures 1–3* and compare to the normal image in *Figure 4*). T2-weighted coronal sections through the anterior cranial fossa were performed. As demonstrated in the normal image (*Figure 4*), one can expect to see high signal in the olfactory sulcus along the base of the frontal lobe. This was entirely absent in this case, with no other significant pathology apparent. There is a high signal return in the right maxillary sinus of the subject, which is incidental and not related to smell perception. Also, *Figure 4* is in a patient with a cyst in the left maxillary sinus. Such incidental, asymptomatic 'sinusitis' on magnetic resonance scans is present in 30–40% of cases (Wani et al, 2001).

Both the patient and her mother were counselled about the risks of environmental hazards. No further investigation was deemed necessary, or indeed desired by the patient.

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