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Congenital anosmia: Our experience of eleven patients with aplasia or hypoplasia of the olfactory tract.

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Congenital anosmia: Our experience of eleven patients with aplasia or hypoplasia of the olfactory tract

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Introduction

Anosmia is present in approximately 1% of the population and can have a significant impact on quality of life [1]. In contrast to acquired anosmia, congenital anosmia is a relatively rare condition characterized by a complete lack of olfactory perception and the aplasia or hypoplasia of the olfactory bulb. Kallmann syndrome (congenital hypogonadotropic hypogonadism and anosmia) is the best known condition assoicated with congenital anosmia [2,3]. Kallmann Syndrome has a prevalence of around 1 in 8000, and is five times more common in men than women [2,3].

Methods

Ethical considerations

The study was registered with the Newcastle upon Tyne Hospitals NHS Foundation Trust Audit Department.

Case note and radiology review

We retrospectively reviewed the notes and scans of patients identified as having the diagnosis of congenital anosmia over the last 10 years (2006 – 2016). Patients were identified through rigorous searching of radiology and medical coding data. All patients presented to the Department of Otolaryngology-Head and Neck Surgery Newcastle upon Tyne. Radiology images were all reported by the same specialist head and neck consultant radiologist.

Results

Case note review

We identified 230 patients during the 10-year period with clinician diagnosed olfactory dysfunction as a primary complaint, and who underwent MR scanning to investigate this. Of those 11 (5%) patients had a clinical history and radiological evidence confirming congenital anosmia (6 male and 5 female). In the congenital anosmia cohort the median age at presentation was 19 years (range 7 – 42 years). Only 3/11 patients presented to Otolaryngology in childhood (16 years or younger). Only one patient was found to have Kallmann syndrome, the rest of the cohort (10/11) had no associated co-morbidities. All patients described anosmia since early memory. A thorough medical history, physical examination and nasal endoscopy was performed in all cases. All patients were sent for an MRI as the primary imaging modality.

Radiology review

All patients underwent an MRI at the Freeman Hospital, Newcastle upon Tyne on either a 1.5T Avanto or Symphony machine (Siemens, Erlangen, Germany). The standard protocol for scanning at our hospital includes 5mm axial T2 images through the whole brain to exclude any abnormality (such as meningioma or post traumatic gliosis) in the anterior cranial fossa and 3mm coronal T1, T2 and FLAIR (fluid attenuated inversion recovery) sequences to specifically look at the olfactory bulbs and tracts and sinonasal tract. Volumetric studies such as VIBE (volume interpolated brain examination) and DRIVE (driven equilibrium) sequences allow the acquistion of thinner slices but because of longer scanning times are sometimes prone to movement artefact and not used routinely in our practice.

MRI demonstrated complete absence of the olfactory sulci, bulbs and tracts in 8 patients (see figure 1 and 2) and hypoplasia in 3 others. Vestigial olfactory sulci were always present in patients with hypoplastic olfactory bulbs and tracts and generally easier to identify than the olfactory bulbs and tracts. It is therefore recommended that the olfactory sulci should be the first structures to be identified when interpreting MR images. Olfactory sulci can be identified on both the axial and coronal images but the olfactory bulbs and tracts are best seen on the coronal images.

[Insert figure 1 – Coronal FLAIR (Fluid Attenuated Inversion Recovery) magnetic resonance image demonstrating absent olfactory bulbs]

[Insert figure 2 –Coronal FLAIR magnetic resonance image showing a normal olfactory bulb for comparison.]

Discussion

Synopsis of key findings

Congenital anosmia is a rare condition making up only 5% (11/230) of cases of olfactory dysfunction in our unit over a 10 year period. Congenital anosmia was found to be an isolated diagnosis in most patients, only 1/11 patients in our case series had Kallmann syndrome. We also demonstrated that most patients with congenital anosmia presented to ENT services in adulthood with a long history of anosmia.

Strengths and weaknesses of the study

This is a relatively small case series, however given the rarity of this condition this is to be expected. It is possible that some cases were missed due to coding errors, however we rigorously searched both clinical coding records and radiology coding records to minimise this. None of the patients in our cohort had objective tests for anosmia. In keeping with the majority of UK ENT departments we do not routinely perform psychophysical assessments of olfaction in our unit. However, all patients gave a clear history of anosmia present since early memory and had imaging confirming olfactory tract abnormalities, therefore we feel this is diagnostic and fits in with clinical practice in the majority of units.

Comparisons with other studies

Several other small case series have been described of congenital anosmia [4 5 6 7]. Given the rarity of this condition the addition of further retrospective data is required to allow future pooling of the clinical data and analysis.

Clinical applicability of the study

This study demonstrates the experiences of congenital anosmia in one large teaching hospital in the UK over a 10-year period. We present the clinical and radiological findings one would expect in these patients to aid clinical identification and appropriate imaging in patients presenting with suspected congenital anosmia. Currently there is no cure available for anosmia, in such cases however patients should be counseled to ensure safety with smoke and gas alarms, as well as ensuring that perishable foods are labeled to minimise risks of illness from ingestion of spoiled food.

Key points

- Congenital anosmia is a rare condition, only 5% of cases of olfactory dysfunction seen in our unit over a 10 year period were congenital.
- Most cases of congenital anosmia in our series presented as an isolated phenomenon in adulthood.
- While there is no available cure identification of these patients is important to ensure appropriate counselling.
- All cases of anosmia reported since birth should have an MRI scan performed to identify an absent or hypoplastic olfactory tract.

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Conflict of interest

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