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CASE REPORT

Congenital anosmia: a case report

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ABSTRACT

Congenital anosmia, isolated or as a symptom of Kallmann or Klinefelter syndrome, is a rare condition found in young patients and children. Anosmia is detected during childhood, being reported by the patient or by his/her family. Besides the clinical examination and olfactometric evaluation, imaging is mandatory for the olfactory pathways investigation. Multidisciplinary approach is needed for these patients in order to determine the etiology of the smell loss. In the current paper, we are presenting the case of an 11-year-old child diagnosed in our ENT Department with congenital anosmia.

KEYWORDS: anosmia, congenital, Kallmann, Klinefelter, brain MRI.

INTRODUCTION

Congenital anosmia is a rare condition, discovered since birth or in early childhood. It might be encountered as an isolated, congenital anosmia with no other abnormalities or as a symptom in endocrinological syndromes, such as Kallmann or Klinefelter syndrome[1].

Kallmann syndrome is known as a combination between hypogonadotropic hypogonadism and anosmia[2]. Klinefelter syndrome (47,XXY) is the most encountered sex chromosomal disorder and affects approximately 1/660 new-born boys[3].

According to the literature, congenital anosmia is more frequently found as an isolated pathology than within a chromosomal syndrome[4].

The diagnosis is based on a detailed patient history, ENT clinical evaluation, nasal endoscopic examination, olfactometric assessment, but the most relevant investigation is the brain MRI which assesses the olfactory bulb, the olfactory tract and the olfactory sulcus.

CASE REPORT

We present the case of an 11-year-old boy, who was brought by his parents to the ENT&HNS Department of "Sfanta Maria" Hospital, Bucharest, in July 2018. He had been accusing hyposmia for about two years. According to the boy's mother, the child underwent bilateral amygdalectomy at the age of 9, at which time the hyposmia was present and reported to the doctor. But, after a detailed discussion with the patient, we found out that he had never had a normal smell function.

The ENT clinical examination together with the nasal endoscopy did not find any significant anomalies of the respiratory and olfactory mucosa.

Fungal and bacteriological examination of the nasal secretion was performed with negative results.

After complete otorhinolaryngological examination, the subjective olfactometric assessment was performed. The dynamic olfactometry using n-Butanol and also the Snap and Sniff Test were suggestive for subjective anosmia (Figures 1,2).

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Measurement	Place	Clinica ORL - Spitalul Clinc "Sfanta Maria"
	Time of measurement	30.07.2018 14:34:15 - 30.07.2018 14:46:12
	Temperature of odour room	
	Olfactometer	TO8
	Last Calibration	
	Pre-Dilution	none

Measurement result

$Z_{ite,pan}$	0
c_{od}	0 OU_E/m^3 (*1)

Panel members	Round 1	ΔZ	Round 2	ΔZ	Round 3	ΔZ	Round 4	ΔZ
VIA	0	0,0	0	0,0	0	0,0	0	0,0

Panel members	Err. ref. air	Err. blanks
VIA	2	0 / 2

Figure 1 Dynamic olfactometry result showing subjective anosmia (no answer registered independent of the n-Butanol concentration presented to the patient).

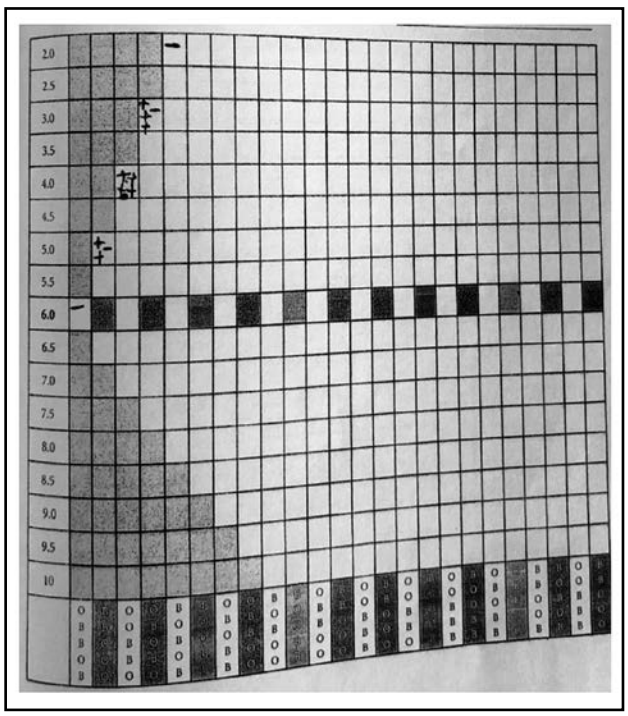

Figure 2 Snap and Sniff test result showing subjective anosmia.

Given the fact that we were in front of a child with a long-time impaired smell function, with subjective anosmia found after olfactometric examination, our diagnosis suspicion was congenital anosmia, so we did not perform the olfactory evoked potentials. We preferred to send the patient first to the Radiology and Imaging Service in order to have a magnetic resonance image of the brain (the technique of choice for congenital anosmia diagnosis[5]), with focus on the area of the olfactory bulb. The result described the lack of the olfactory bulbs and of the olfactory tracts; narrow olfactory fissures with fluid content and a vascular loop at the anterior limit of each of them; superficial, unevenly sketched, bilateral olfactory sulcus (Figure 3 a,b).

The diagnosis was congenital anosmia and we referred the patient to the Endocrinology Department in order to establish if the anosmia is isolated or if it is a symptom within a chromosomal syndrome (Kallmann or Klinefelter). The endocrinological examination showed no anomalies suggestive for genetic impairment, so the final diagnosis was isolated congenital anosmia.

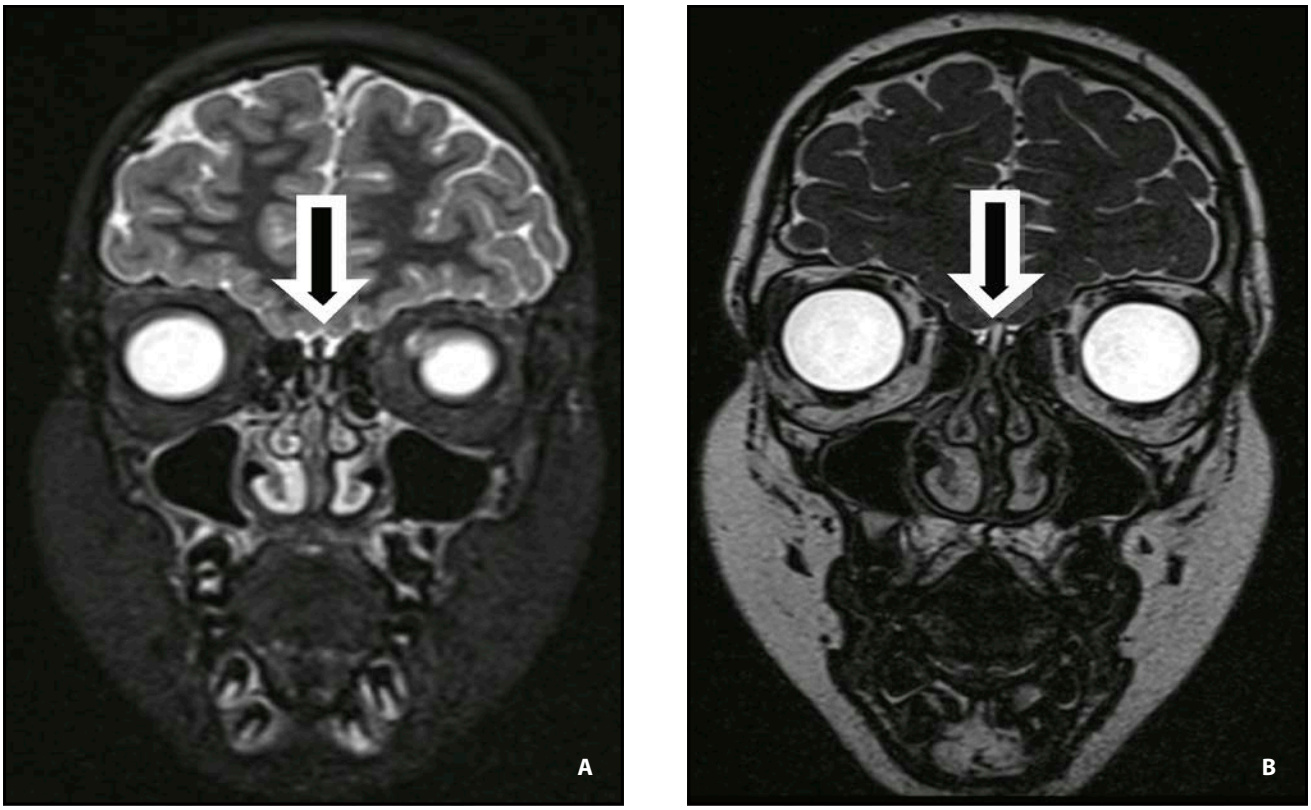

Figure 3 A,B. MRI images showing the absence of the olfactory bulbs – rows showing the area where normal olfactory bulbs should be seen.

DISCUSSIONS

The first diagnosis suspicion in young patients complaining about smell function impairment, with no significant personal history, should be congenital anosmia, even though it is a very rare condition (prevalence 1 in 10,000[6]).

It appears in patients with vestigial or complete absence of the olfactory pathway and the main diagnosis method is the brain MRI. Even if this is a rare condition, it has been studied at large scale, being a subject of interest not only for otorhinolaryngologists, but also for endocrinologists and geneticists. It is known that congenital anosmia can exist as single entity or as a component of a chromosomal disorder, such as Kallmann or Klinefelter syndrome.

Patients with isolated congenital anosmia (ICA) do not have any additional symptoms, and there is no underlying disease-causing condition identified[7]. According to the literature, this condition may be discovered isolated in a family, or more family members might be affected[8,9] and the underlying genetic architecture of ICA is barely known. In 1918, Glaser et al.[10] first reported isolated congenital anosmia as a potentially hereditary smell disfunction, and, over the time, there have been few genes identified as the potential genetic cause for isolated congenital anosmia[11,12]. In what concerns the inheritance, most reported anosmia cases are transmitted through autosomal dominant patterns[8]; however, in some instances, it is an X-linked trait[10,12].

According to the literature, the evaluation of patients accusing anosmia or hyposmia since always must include, besides the complete ENT clinical examination, nasal endoscopy and olfactometric assessment, imaging examination. The MRI focused on the olfactory pathway is pathognomonic for the diagnosis, as it can evaluate the olfactory bulb and sulcus, which can be hypoplastic or aplastic[13].

In what concerns the quality of life in these patients, there is no doubt that it is affected, but in contrast to people with acquired olfactory impairment, patients with congenital anosmia have developed coping strategies[14].

CONCLUSIONS

Congenital anosmia is a rare condition encountered the most frequently in early-aged patients and the most important diagnosis tool is the brain MRI, which evaluates the olfactory bulb and tract

(presence / absence of the olfactory bulb and tract, olfactory bulb volume if present), and also the olfactory sulcus depth.

An endocrinological examination should establish if the smell loss is isolated or if it is part of a chromosomal syndrome, so a multidisciplinary approach is needed.

Unfortunately, there is no treatment for smell regeneration in congenital anosmia.

Conflict of interest: The author has no conflict of interest to declare.

Contribution of authors: All authors have equally contributed to this work.

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