CASE REPORT

Anosmia and Isolated Olfactory Bulbs Absence

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Abstract

Congenital anosmia is frequently associated with some developmental abnormalities. Its occurrence as an isolated defect is extremely rare. In children, olfactory conditions can be difficult to determine due to their mild symptomatology. The diagnosis is often delayed. The authors describe and discuss the case report of a 15-year-old-girl with congenital anosmia and isolated olfactory bulbs agenesis.

Keywords: Adolescent; Olfactory Bulb/abnormalities; Olfaction Disorders/congenital; Olfaction Disorders/diagnosis

Introduction

Olfactory dysfunction is a very common condition with a reported prevalence between 4% and 25% of the general population. In the pediatric ages, conditions associated with potential smell disorders, namely enlarged adenoids, allergic rhinitis, nasal septum deviation, or juvenile angiofibroma, are frequent[1,2]; however, the complete loss of smell (anosmia) is rarely seen in children and adolescents.

Individuals suffering from anosmia most often have an acquired condition resulting from head trauma, upper respiratory tract infection, sinonasal disease, endocrine disorders, brain tumors, or neurodegenerative illnesses, such as Parkinson's disease or Alzheimer's disease.[3-5] A small number of individuals present congenital anosmia, normally in association with other anomalies, where Kallmann syndrome is the best described or, occasionally, the anosmia can be isolated.[3,6]

Isolated congenital anosmia is a diagnosis of exclusion. The patients have never been able to smell, and no other underlying condition can be identified. It affects approximately 1% of the anosmic children. Head magnetic resonance imaging (MRI) is the gold standard test to detect the abnormalities of the olfactory bulbs, tracts, and sulci.[4]

Case Report

A 15 year-old-girl was referred to our department because of her lack of smell starting approximately two years before. The mother suspected that her daughter suffered from anosmia for finding her alone at home while there was food burning, without her smelling it. Her mother returned and found smoke everywhere, but no fire. Until that moment, no one had perceived the girl's inability to smell.

The patient did not have any relevant family history of smell disturbances, and her physical and psychological development was perfectly normal. Her mother denied prenatal alcohol or drug exposure; the adolescent denied having taken drugs too. She also denied respiratory complaints, visual impairment, hearing loss, and other relevant symptoms. There was no prior head trauma.

The physical examination, anterior and posterior rhinoscopy, complemented with nasopharyngolaryngoscopy, did not reveal any intranasal structural changes. The tympanic membrane was bilaterally intact, and we also did not find midline cranio-facial defects, as a cleft lip or palate.

To supplement the presented data, an olfactory test was performed (with smell stimuli) by checking the scents of alcohol, cinnamon, lemon, and coffee. The girl did not just recognize any of the fragrances and was not aware of their presence.

In order to assess other neurological dysfunctions, we tested the remaining cranial nerves, particularly, the optic and auditory ones. The optic nerve was evaluated by confrontation visual field testing and the hearing level was estimated using Rinne and Weber tests. All neurological responses were perfectly normal, with no further neurological deficits aside from absence of smell.

A head MRI was run and demonstrated the bilateral absence of olfactory bulbs and hypoplasia of the left olfactory tract (Fig. 1). Accordingly, we excluded possible meningiomas along the cribriform plate, posttraumatic injury, extra-axial collections, and other sinonasal masses that could affect olfactory function.

We requested a pediatric endocrinology evaluation to rule out hypogonadotropic hypogonadism, as it is found in Kallman syndrome. Her serum gonadotropin levels

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were within the normal limits: follicle-stimulating hormone (FSH) (3.5 mUI/mL), luteinizing hormone (LH) (4.3 mUI/mL), estradiol (140 ng/L) and her pubertal maturation was suitable for her age according to Tanner classification (P5). The pediatrician requested an abdominal ultrasound to assess renal structures and a pelvic ultrasound to determine the size and shape of the uterus. These exams did not reveal additional abnormalities, which supported the diagnosis of isolated olfactory bulbs agenesis.

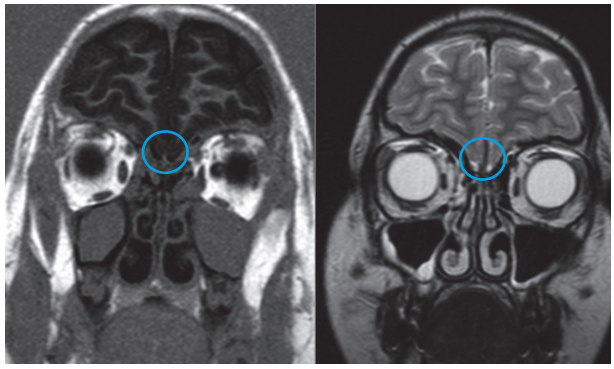

Figure 1. Coronal planes weighted in T1 and T2: bilateral bulbs absence.

Discussion

Congenital absence of smell can be divided into two major groups[5]:

- One that comprises approximately 12% of patients, associated with major congenital abnormalities, including hypogonadotropic hypogonadism like Kallmann syndrome;
- The other, the majority (88%), comprises patients with isolated anosmia.

Kallmann syndrome is a rare genetic disorder with an estimated prevalence of one in 10,000 males and one in 50,000 females.[1] The associated anosmia is due to the failure of development of the olfactory bulbs and tracts while the hypogonadism is caused by gonadotropin hormone deficiency. Alongside the olfactory changes, this syndrome may be associated with other abnormalities, such as hearing loss, color blindness, cleft lip and palate or renal agenesis. Kallmann syndrome was reported in eight of 25 patients with congenital anosmia in one study.[7] Other authors evaluated 41 patients with hypogonadotropic hypogonadism and, according to their report, 25 patients were affected by Kallmann syndrome and 16 were normosmic.[8]

In this specific case, despite the correct physical development of the patient, we decided for a careful observation by a pediatrician. After normal hormone values and ultrasound studies, Kallmann syndrome was excluded.

The approach to smell disorders includes smell testing, physical examination, and nasal endoscopy, however they cannot determine the anatomic cause of congenital olfactory loss. A head MRI is the gold standard to study the first cranial nerve pair, providing an excellent view of the olfactory bulbs, tracts, and all this system within the brain.[9]

The main olfactory epithelium is located at the apex of the nasal cavity, below the cribriform plate. An odor needs to activate the receptors, surrounded by nasal mucous membrane, in order to stimulate the olfactory system. The axons from the olfactory receptors release small nerve bundles, which pass through the cribriform plate of the ethmoid bone to the olfactory bulb. Within the olfactory bulb, the nerve terminals synapse with mitral cells whose axons (as olfactory tract) project directly to the olfactory cortex.[2,10]

When we test olfactory impairment, it is important to remember that we have not only the olfactory nerve cells in the nasal cavity, but also the trigeminal nerve fibers that are sensitive to irritating substances and temperature.[11]

Although most odorants activate both, the olfactory and the trigeminal systems, a couple of them present selective stimulation. For instance, carbon dioxide activates the trigeminal system without concomitant olfactory stimulation. In contrast, vanillin only works in the olfactory nerves, with little impact on the trigeminal nerve.[11-13] In this context, some studies have admitted that the odorant mixtures and individual odorants are recognized differently in the human brain. Odorant mixtures do not simply recruit more brain areas but also activate specific regions, specialized in mixture processing, in particular, piriform cortex, orbitofrontal cortex, and rostral insula.[10-12] Thus, these are the main areas of interaction between the trigeminal and olfactory system.

It is interesting to note that patients with olfactory dysfunction exhibit lower trigeminal sensitivity compared with individuals with normal olfactory function. This is true only for acquired anosmia since in congenital anosmia trigeminal sensitivity seems unaffected.[13-15]

The recent research focuses in plasticity in the sense of smell, since the olfactory training improves olfactory function in patients with neurodegenerative diseases or trauma injuries. It will be important to understand the effect of continuous trigeminal stimulation in the olfactory perception, mostly in congenital anosmia, as a possible treatment in the future.[14,15]

This case report is particularly interesting since the trigger to the perception of anosmia was burned food, which is a scary situation to an adolescent girl. The management of the patient is difficult and, for now, there is nothing to offer except for daily life, social life, eating, and security counselling.

Interdisciplinarity between medical specialties is important to achieve the diagnosis and the best patient management.

WHAT THIS CASE REPORT ADDS

- Isolated congenital anosmia occurring as a single-family case is extremely rare, and its diagnosis should be based on a careful evaluation.
- The olfactory loss has a negative impact in human quality of life, modulating important behaviors, mainly in eating, detecting of harmful smoke, and to some extent in social situations.
- Most odorant molecules have the propensity to simultaneously stimulate olfactory and trigeminal systems in the nasal cavity. Both systems' interactions are complex, occurring at peripheral and central levels.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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Anosmia e Ausência Isolada de Bulbos Olfativos

Resumo:
A anosmia congénita associa-se frequentemente a anomalias do desenvolvimento. A sua ocorrência como um defeito isolado é extremamente rara. Nas crianças, as patologias do trato olfativo podem ser difíceis de diagnosticar devido à limitação daquelas para expressar os sintomas. O diagnóstico é frequentemente tardio. Os autores descrevem e discutem o caso clínico de uma adolescente de 15 anos de idade com anosmia e ausência isolada dos bulbos olfativos.

Palavras-Chave: Adolescente; Bulbo Olfativo/anomalias congénitas; Perturbações do Olfato/congénito; Perturbações do Olfato/diagnóstico