A Case of Congenital Anosmia
Um Caso de Anósmia Congênita

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An 11-year-old girl is referred to an Allergology consultation for rhinorrhea, frequent sneezing, nasal pruritus and diminished sense of smell, following an influenza-like syndrome. However, after detailed anamnesis, we assessed that anosmia had, in all likelihood, been present from early childhood.

Allergy screening tests were positive for dust mites, but despite adequate treatment with antihistaminic and nasal corticosteroids, the anosmia persisted. Further evaluation by a pediatric otorhinolaryngologist was normal.

She had a history of headaches and had previously undergone a head computed tomography (CT) scan revealing no abnormalities.

Suspecting congenital anosmia (CA) a brain magnetic resonance imaging (MRI) (field strength of 1.5 Tesla) with evaluation of the olfactory system was requested. The olfactory bulb was absent bilaterally, with hypoplastic olfactory sulci and flattened olfactory fossae, with a reduced depth of 3 mm (Keros classification type I). No other alterations were identified. These results were consistent with CA.

Although rare, (1:10 000),1 CA should always be considered if an impaired sense of smell is identified and a plausible alternative explanation is not found. If available, diagnosis should be confirmed by MRI,2 allowing adequate evaluation of the olfactory bulbs and sulci.

CA can exist as an isolated entity or part of a genetic syndrome, which should be ruled out, particularly Kallmann syndrome (KS): a rare cause of hypogonadotropic hypogonadism, usually occurring with hypoplastic olfactory bulbs,3 and requiring lifelong hormone replacement therapy. Normal pubertal development and additional testing were able to exclude KS in this case.

Despite the lack of curative treatment for CA, patients usually develop coping mechanisms4. They should nonetheless be advised

Figure 1. (A) Coronal T1 and (B) Coronal T2 MRI show absence of the olfactory bulbs in olfactory (arrowheads) with hypoplastic olfactory sulci (arrows).
about day-to-day precautions like installing home gas detectors and frequently checking expiration dates on dairy products.

Due to these coping mechanisms and lack of complaints in early childhood, CA often goes unnoticed until puberty. Suspected patients should be always be supervised and screened for delayed or incomplete puberty.

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References / Referências