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Kallmann's Syndrome: Rare Affection with Typical MRI Findings

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ABSTRACT

Kallmann's Syndrome (KS) is an inherited disorder distinguished by an association of hypogonadotropic hypogonadism and anosmia or hyposmia. This altered sense of smell is due to the absence or poor development of olfactory bulbs and girdles; the olfactory grooves are absent in their anterior portion with variable development in the posterior region. In this article, we report a case of Kallmann's syndrome in ten years old male patient who presented at the endocrinology department with pubertal delay and clinical findings suspecting Kallmann's syndrome.With hormonal assays, magnetic resonance imaging was performed as a useful adjunct to the diagnosis in the hierarchy of tests, which shows in our case characteristic findings of KS.The treatment is primarily aimed at restoring, normal pubertal development, and, in some cases, normal fertility, but unfortunately, no cure is available concerning olfactory disorders.

Introduction

Kallmann syndrome is a developmental genetic disorder characterized by delayed or absent puberty, due to gonadotrophin-releasing-hormone (GnRH) deficiency, and an impaired sense of smell (with hypoplasia or aplasia of the olfactory bulbs)[1].

Our patient has delayed puberty, cryptorchidism, and anosmia. His brain MRI reveals the typical findings which confirm the diagnosis and aims to restore the treatment.

Observation

A 10-years-old male patient presented at the endocrinology department with pubertal delay. He had no previous clinical history. On physical examination, the hypogonadal phenotype was observed. He had prepubertal genital hair without scrotal pigmentation, micropenis with small testes, and cryptorchidism. Besides, he reported anosmia.

The analysis showed hypogonadotropic hypogonadism.

MRI was performed to complete the patient's study. It revealed hypoplastic olfactory sulci, above all in the left side (The deep measure of these olfactory grooves was less than 8 mm (normal value)) with a pseudo fusion of rectus and medial orbital gyri forming a single thick gyrus (figure1, panel A, blue arrows), absence of olfactory bulbs and bands, and flattening of the fovea ethmoidalis in the anterior cranial fossa. (figure 1). All these findings are typical and frequent on Kallmann's syndrome. Furthermore, this patient had a sort of empty sella illustrated by a widening of the sella turcica mostly occupied by cephalosporin fluid (CSF) signal with flattening of the pituitary gland (the hight inferior to 2mm) and dipping of the stalk inside the sella below the level of the clinoid processes (figure 2)

Because of diagnosing Kallmann's syndrome, the patient was undergone to hormone replacement therapy with steroids.

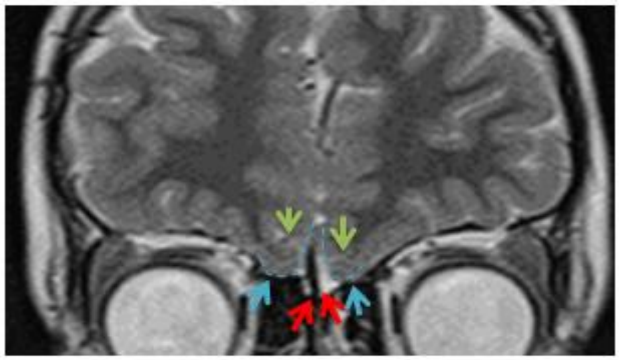

Figure 1. T2WI coronal image denoting the absence of olfactory bulbs and bands (red arrows) with hypoplastic olfactory sulci (green arrows), and a pseudo fusion of rectus and medial orbital gyri forming a single thick gyrus (blue dashes), we also note the flattening of the fovea ethmoidalis in the anterior cranial fossa (blue arrows)

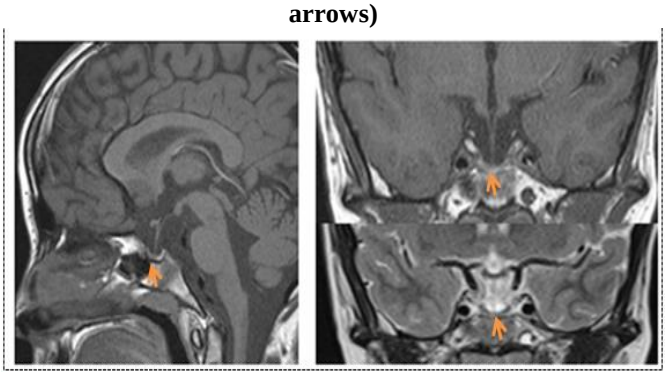

Figure 2. T1WI sagittal (a), coronal (b) images, and a T2WI coronal image (c) A widening of the sella turcica mostly occupied by the CSF signal with flattening of the pituitary gland (orange arrows) and dipping of the stalk inside the sella below the level of the clinoid processes (green dashes).

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Discussion

The association of anomalies in the olfactory system with hypogonadism was first observed by Maestre de San Juan in 1956[2].

In 1944, the American psychiatrist and geneticist Franz Josef Kallmann individualized the syndrome in 3 families and hypothesized that it was hereditary[3].

In the 1950s, the Swiss pathologist De Morsier deepened his knowledge of the syndrome by describing several cases of hypoplasia or anaplasia of the olfactory bulbs and bands in male patients with hypogonadism[4] [5] [6]. He called this syndrome "olfacto-genital dysplasia." He was the first to suggest the hypothalamic origin of hypogonadism.

Finally, in 1971, the hypothalamic origin of hypogonadism was demonstrated, secondary to a deficiency in gonadoliberin (GnRH)[7].

Normally, GnRH cells from olfactory placode migrate along the olfactory vomeronasal axons, through the holes in the cribriform plate, and into the forebrain. In Kallmann's syndrome, they are stuck in the dura at the plate[8]

Magnetic resonance is the main image test performed to evaluate the olfactory system and helps to diagnose Kallmann's syndrome earlier. High resolution coronal fast spin-echo T2- and T1-weighted images are the preferred sequences for morphologic evaluation of the olfactory system.

The principal finding is the absence of olfactory bulb with other typical features such as hypoplastic olfactory grooves and flattening of the fovea ethmoidalis.

Moreover, there are several anomalies associated with Kallmann Syndrome such as the increase in the size of ethmoidal cells, septo-optic dysplasia, and empty sella

The treatment is used to restore normal pubertal development and, in some cases, normal fertility by using exogenous sex steroids, and also a pulsed gonadotrophin-releasing hormone can be administrated for those who desire fertility[9].

Competing interests

The authors declare no competing interest.

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