Case 15244

Eurorad

Kallmann’s syndrome: MR features of an atypical cause of anosmia

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Section: Neuroradiology
Area of Interest: Neuroradiology brain Head and neck
Procedure: Diagnostic procedure
Imaging Technique: MR
Special Focus: Congenital Acute Case Type: Clinical Cases
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Patient: 19 years, male

Clinical History:

A 19-year-old male patient presented at the endocrinology department with pubertal delay. He had no previous clinical history. On physical examination, hypogonadal phenotype was observed. He had prepubertal genital hair without scrotal pigmentation and small testes located in scrotal bags. In addition, he reported anosmia. Analysis showed hypogonatropic hypogonadism.

Imaging Findings:

A magnetic resonance was performed in order to complete the patient’s study. It revealed hypoplastic olfactory grooves, above all in left side, (figure1, panel A, blue arrows) and absence of olfactory bulbs and bands (figure 1, panel A, orange arrows/ figure 2). All these findings are typical of Kallmann’s syndrome. Furthermore, this patient had a sort of empty sella (figure 3), which is one of the anomalies that are associated to this syndrome. In sagital scan MR, anterior pituitary gland appears compressed by this empty sella (panel A, figure 3). Nevertheless, in coronal scan MR (panel B, figure 3) anterior pituitary is better seen.
Panel B in figure 1 shows normal anatomy in other patient (orange arrows: olfactory bulb/blue arrows: normal olfactory grooves).
Because of diagnosing Kallmann’s syndrome, the patient was undergone to hormone replacement therapy with steroids.

Discussion:

Smell is perceived in the gray matter which covers the lateral and medial stretchmarks of the olfactory gyrus. The lateral stretchmark runs from the inferomedial portion of the temporal lobe. The medial stretchmark runs from the medial and inferior sides of the frontal lobe to the olfactory trigone. This olfactory trigone extends anteriorly like olfactory bulb and olfactory girdle. Olfactory bulb and girdle are located below the olfactory grooves which separate the straight convolution of the medial fronto-orbital.

From the olfactory bulbs, the sensitive nerves pass through the crib sheet and divide into neurosensitive cells distributed in the nasal epithelium. [1]

Anosmia (loss of sense of smell) may be congenital (choanal atresia, holoprosencephaly, septo-optic dysplasia,

frontoethmoidal meningocoele, Down, Turner or Kallmann syndromes) or acquired (secondary to trauma, tumours or chronic nasosinusal inflammatory disease).

The association between anosmia and hypogonadism hypogonadotropic is very typical of Kallmann syndrome. On physical examination, patients present gynaecomastia, delayed puberty, eunuchoidism, obesity, cryptorchidism and other alterations (for example: diaphragmatic eventration, nystagmus, renal agenesis…) [2].

Pathophysiology of Kallmann syndrome consists of anormal migration of cells which produces luteinising hormone releasing factor (LHRH) from the olfactory plate to the hypothalamus [3].

The main morphological feature of Kallmann syndrome is the absence/poor development of olfactory bulbs and girdles. The olfactory grooves are absent in their anterior portion with variable development in the posterior region.

Magnetic resonance is the main image test performed to evaluate the olfactory system and helps to diagnose Kallmann syndrome earlier. The main finding is the absence of olfactory bulb (Fig. 1, panel A/Fig. 2) [4]. Other typical features are bulb hypoplasia, hypoplastic olfactory grooves and absence/hypoplasia of olfactory girdle. Another frequent finding is the presence of hypoplasia of the right gyrus and flattening of the fovea ethmoidalis in the anterior cranial fossa.

Furthermore, there are several anomalies which are associated with Kallmann Syndrome such as defects of the middle line, cleft lip, deafness, increase in the size of ethmoidal cells, septo-optic dysplasia, empty sella and aplasia/hypoplasia of the adenohypophysis.

Hormone replacement therapy can correct abnormalities of the endocrine system.

In summary, despite being a not frequent cause of anosmia, if a patient presents with anosmia, hypogonadism and absence/hypoplasia of olfactory bulbs and girdles, a Kallmann syndrome must be discarded.
Differential Diagnosis List: Kallmann syndrome, Meningioma of cribriform lamina, Traumatic lesions, Nasosinusal masses, Extra-axial collections

Final Diagnosis: Kallmann syndrome

References:

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Ph Rombaux, T. Duprez, T. Hummel (2009) Olfactory bulb volume in the clinical assessmentof olfactory dysfunction. Rhinology 47: 3-9 (PMID: 19382487)
Truwit CL, Barkovich AJ, Grumbach MM, Martini JJ (1993) MR imaging of Kallmann syndrome, a genetic disorder of neuronal migration affecting the olfactory and genital systems. AJNR 14: 827-838 (PMID: 8352153)
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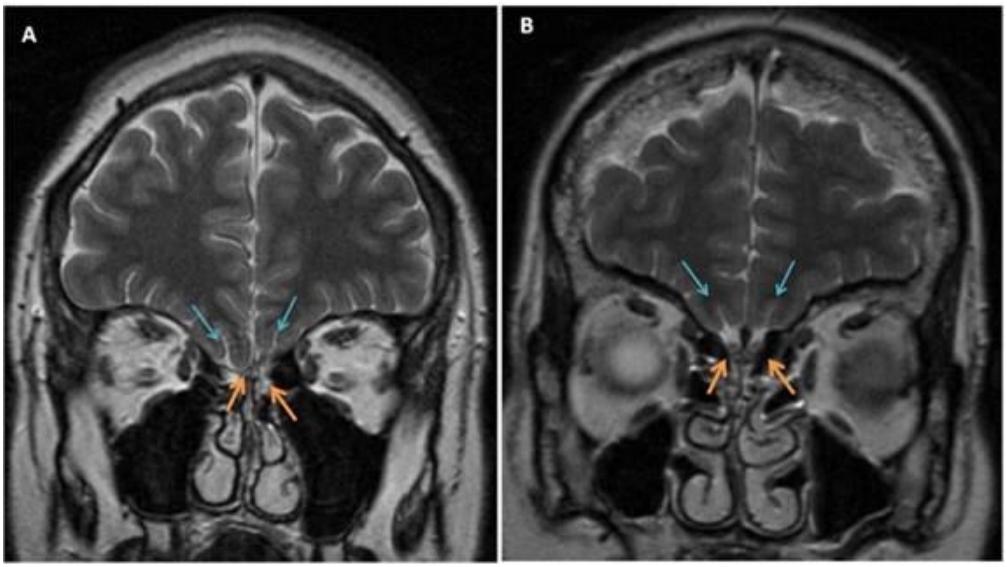
Figure 1

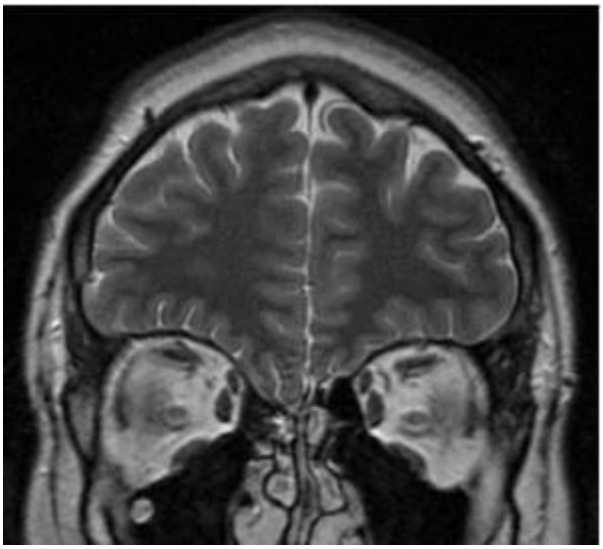
a



Description: Panel A shows hypoplastic olfactory grooves (blue arrows) and aplastic olfactory bulbs and bands (orange arrows). Panel B reveals normal anatomy in another patient with normal olfactory grooves (blue arrows) and bulbs (orange arrows). **Origin:** Department of Radiology, Clinical Hospital, Valladolid, Spain.

Figure 2

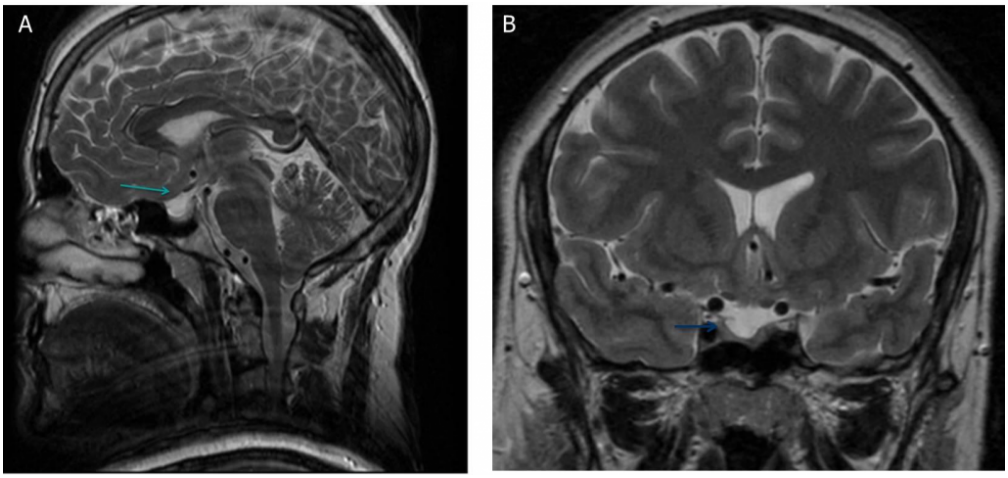
a



Description: This image continues showing the hypoplastic olfactory grooves (all above in left side) which were presented in our patient. The deep measure of these olfactory grooves was less than 8 mm (normal value). **Origin:** Radiology Department, Clinical Hospital, Valladolid, Spain

Figure 3

a



Description: This image shows a feature associated with Kallmann syndrome: a sort of empty sella (arrow in panel A) which is compressing anterior pituitary gland. Gland is better seen in coronal scan (arrow panel B). **Origin:** Radiology Department, Clinical Hospital, Valladolid, Spain